# The Journey to Diagnosis
# for People Living with Rare Diseases

A Rare Barometer Survey

**Target population:**

- Patients living with a rare disease
- People not yet diagnosed but living with a disease considered to be rare
- Their family member (parents or close relatives)

**March 17 - June 15, 2022**

**13300** respondents worldwide and

**10486** in Europe

**27** languages

**107** countries

**1900+** diseases represented

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A EURORDIS INITIATIVE

**EURORDIS**
RARE DISEASES EUROPE

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

# DASHBOARD FOR EUROPE

## HOW TO USE THIS DASHBOARD

In this dashboard, you will find results for every question of the Rare Barometer survey on the journey to diagnosis for people living with a rare disease.

Please do not use results of questions for which there are less than 30 respondents.

**Please refer to Rare Barometer or add the Rare Barometer logo when using the results.**

## LANGUAGES

You can change the language at the bottom left of this page, and have access to the questions and modalities as they appeared to respondents in the 27 languages of the survey.

Translation is not available for new variables that were calculated after the questionnaire was closed and for some comments added in this dashboard.

## INFORMATION

For more information
- contact the Rare Barometer team at rare.barometer@eurordis.org
- or visit the Rare Barometer website at eurordis.org/rare-barometer

## LIST OF CONTENT

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Sample size 10486 responses

# Chapter 1.

## Sample description

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Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

## Number of respondents

# 10.486

### Are you a...

| | N |
|---|---|
| Patient | 6.772 |
| Former or recovering patient (e.g. cancer survivor) | 247 |
| Parent of a person living with a rare disease | 3.078 |
| Grandparent of a person living with a rare disease | 40 |
| Spouse of a person living with a rare disease | 186 |
| Uncle/aunt of a person living with a rare disease | 23 |
| Sibling of a person living with a rare disease | 48 |
| Other, specify... | 92 |
| **TOTAL** | **10.486** |

### Are you a...

- Patient — %65
- Former or recovering patient (e.g. cancer survivor) — %2
- Parent of a person living with a rare disease — %29
- Grandparent of a person living with a rare disease — %0
- Spouse of a person living with a rare disease — %2
- Uncle/aunt of a person living with a rare disease — %0
- Sibling of a person living with a rare disease — %0
- Other, specify... — %1
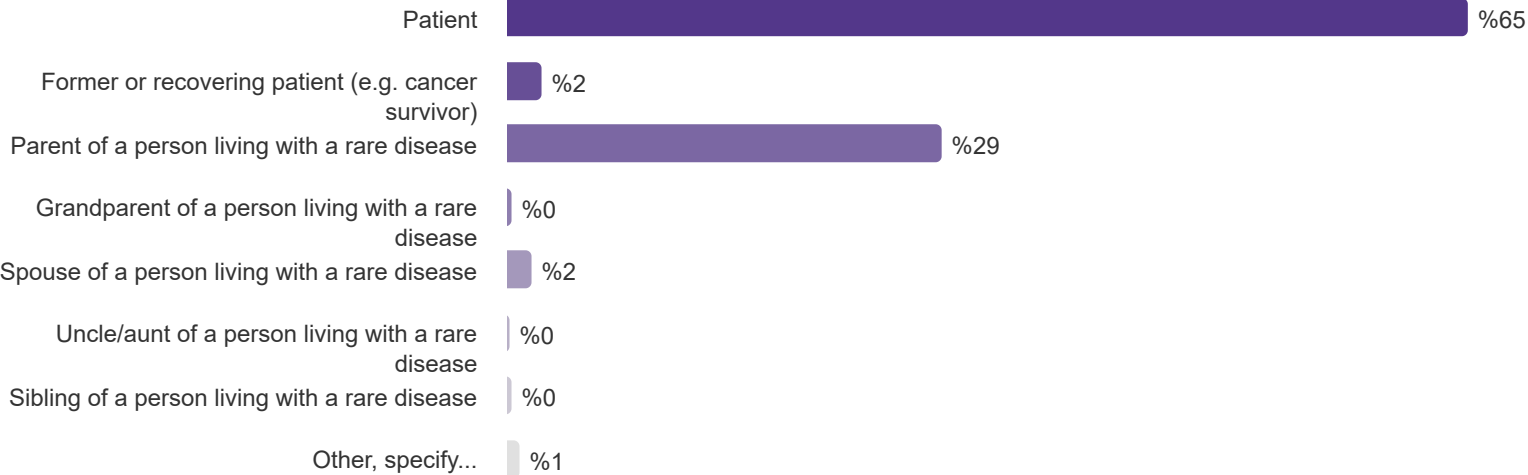
**Respondents can be:**

**- the person directly affected** by the rare disease

**- or family members of the person affected** (parents, grand-parents, spouses, uncles/aunts, siblings or other family member).
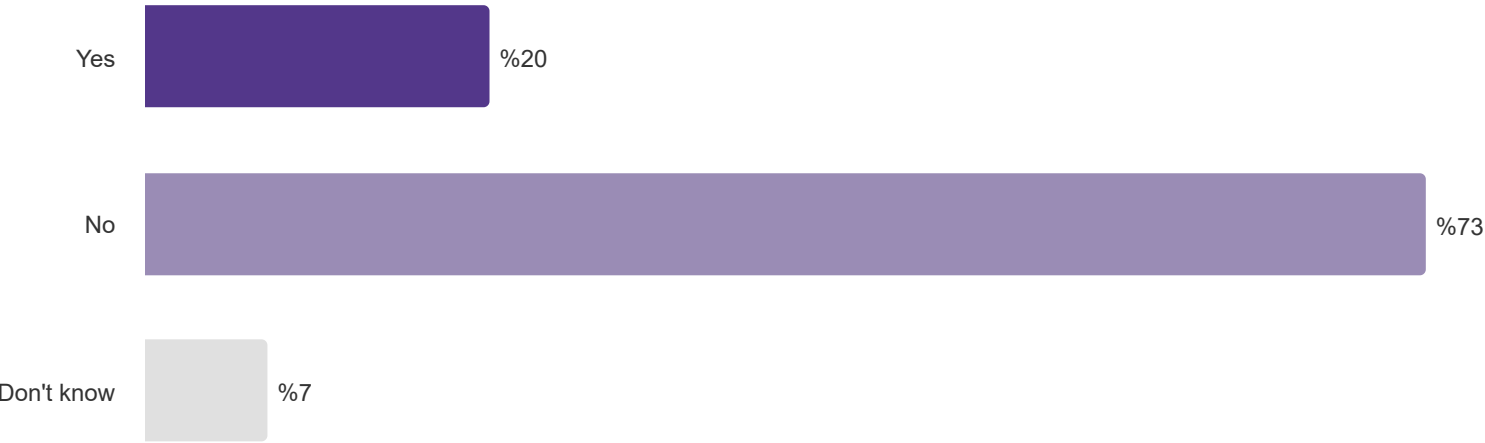
### Are you a...

- Person affected by the rare disease — %68
- Family member — %32

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Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

## Are you a patient representative, i.e. involved in policy activities to support the cause of rare diseases?

| | N |
|---|---|
| Yes | 2.073 |
| No | 7.666 |
| Don't know | 747 |
| **TOTAL** % | **10.486** |

## Are you a patient representative, i.e. involved in policy activities to support the cause of rare diseases?

Yes %20

No %73

Don't know %7

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

**Age of the respondent when the questionnaire was filled in**
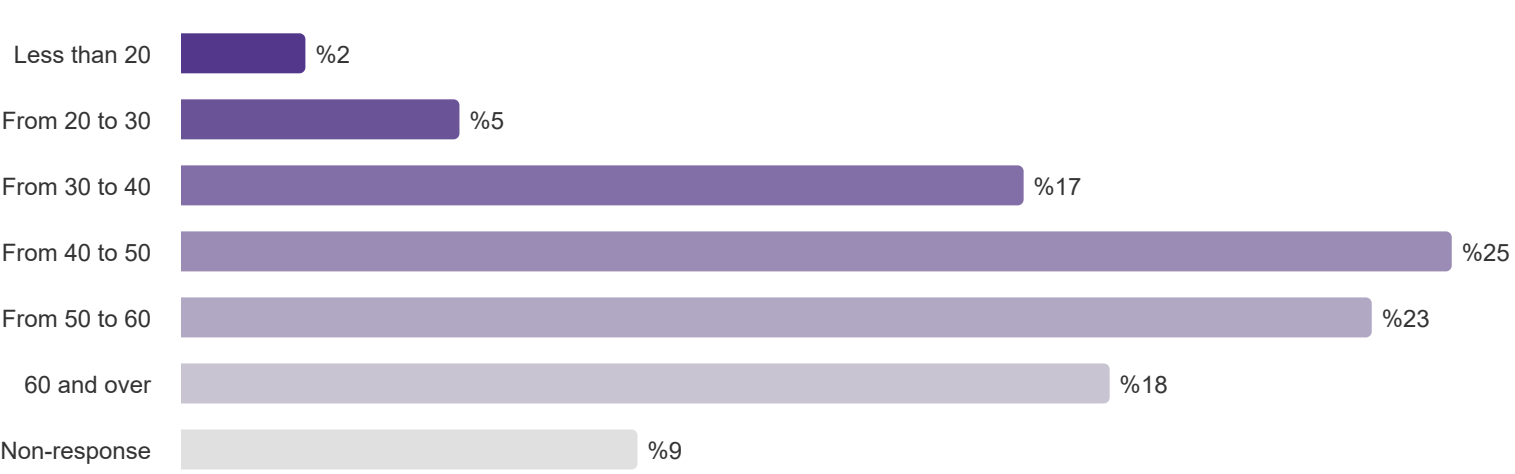
Calculated based on:

- the date of birth of the respondent: "What is your month and year of birth?"

- the date when questionnaire was filled (automatically saved by the software)

## Age of the respondent when filling the questionnaire

| | N |
|---|---|
| Less than 20 | 259 |
| From 20 to 30 | 575 |
| From 30 to 40 | 1.734 |
| From 40 to 50 | 2.614 |
| From 50 to 60 | 2.451 |
| 60 and over | 1.913 |
| Non-response | 940 |
| **TOTAL** | **10.486** |

## Age of the respondent when filling the questionnaire

| Category | Percentage |
|---|---|
| Less than 20 | %2 |
| From 20 to 30 | %5 |
| From 30 to 40 | %17 |
| From 40 to 50 | %25 |
| From 50 to 60 | %23 |
| 60 and over | %18 |
| Non-response | %9 |

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Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

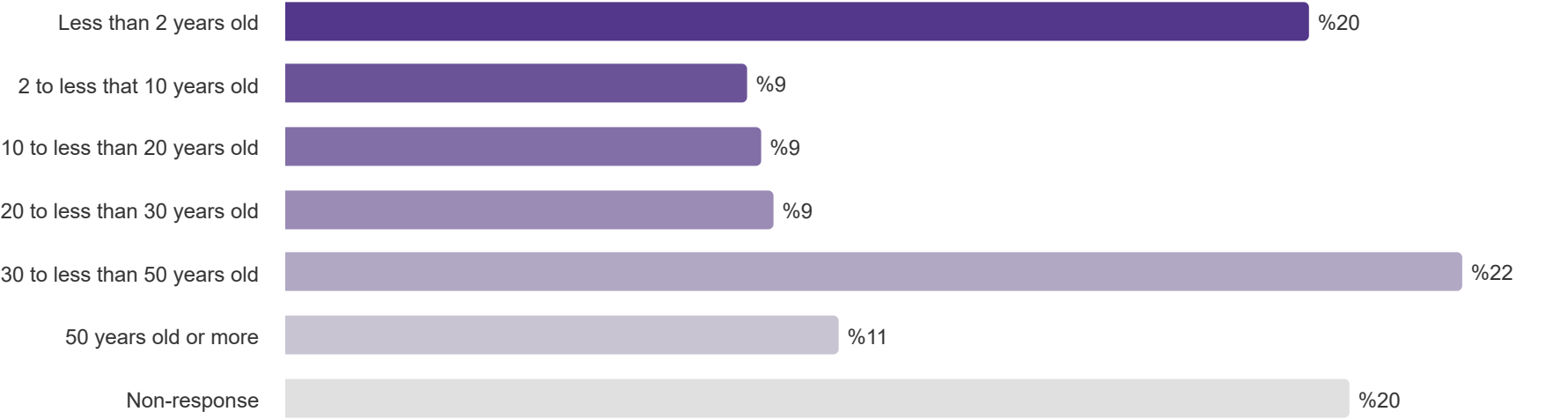## Age of the person affected by the rare disease when the first symptoms were noticed

Calculated based on:

- date of birth of the respondents who are patients themselves: "What is your month and year of birth?"

- date of birth of the patient when respondents are family members of the person affected by the rare disease: "What is the month and year of birth of the person affected by the rare disease?"

- date when first symptoms were noticed: "As far as you remember, when did you or a healthcare professional first notice the symptoms of the rare disease or think that something was wrong?"

### Age of the person affected by the rare disease when first symptoms were noticed

|  | N |
|---|---|
| Less than 2 years old | 2.045 |
| 2 to less that 10 years old | 925 |
| 10 to less than 20 years old | 952 |
| 20 to less than 30 years old | 978 |
| 30 to less than 50 years old | 2.353 |
| 50 years old or more | 1.107 |
| Non-response | 2.126 |
| **TOTAL** | **10.486** |

### Age of the person affected by the rare disease when first symptoms were noticed



- Less than 2 years old: %20
- 2 to less that 10 years old: %9
- 10 to less than 20 years old: %9
- 20 to less than 30 years old: %9
- 30 to less than 50 years old: %22
- 50 years old or more: %11
- Non-response: %20

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Gender of the respondent

### Are you:

| | N |
|---|---|
| Female | 7.930 |
| Male | 1.807 |
| Other | 56 |
| Non-response | 693 |
| **TOTAL** | **10.486** |

### Are you:

| | |
|---|---|
| Female | %76 |
| Male | %17 |
| Other | %1 |
| Non-response | %7 |
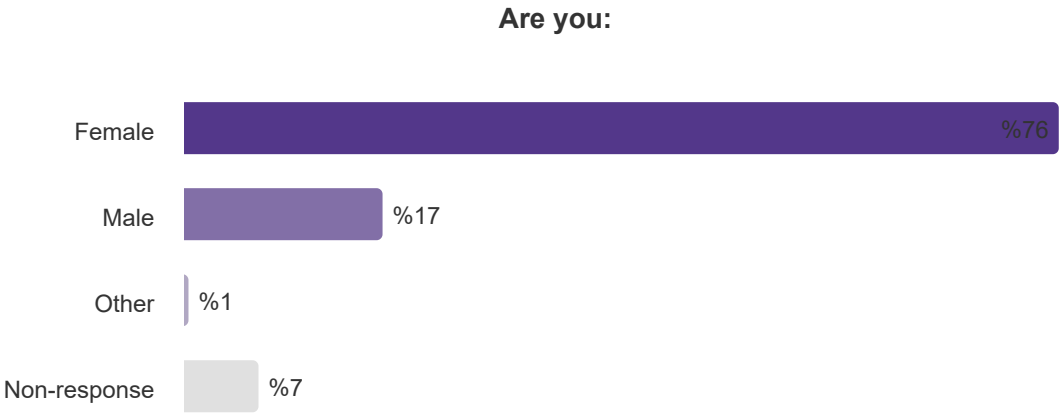
## Gender of the person affected by the rare disease

### Gender of the person affected by the rare disease

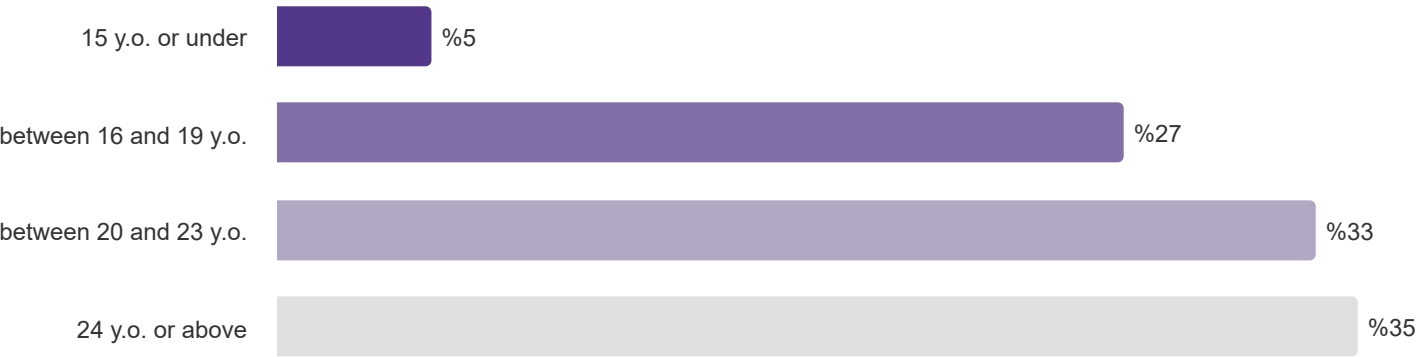| | N |
|---|---|
| Female | 6.659 |
| Male | 2.810 |
| Other | 101 |
| Non-response | 916 |
| **TOTAL** | **10.486** |

### Gender of the person affected by the rare disease

| | |
|---|---|
| Female | %64 |
| Male | %27 |
| Other | %1 |
| Non-response | %9 |
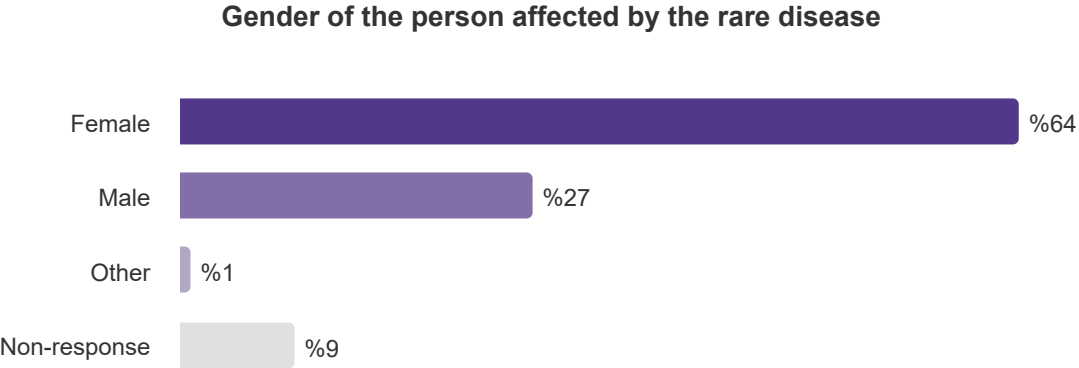
**Education of the respondent**

**How old were you when you stopped full-time education?**

| | N |
|---|---|
| 15 y.o. or under | 455 |
| between 16 and 19 y.o. | 2.464 |
| between 20 and 23 y.o. | 3.022 |
| 24 y.o. or above | 3.145 |
| **TOTAL** | **9.086** |

**How old were you when you stopped full-time education?**

| | |
|---|---|
| 15 y.o. or under | %5 |
| between 16 and 19 y.o. | %27 |
| between 20 and 23 y.o. | %33 |
| 24 y.o. or above | %35 |

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Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

## How would you best describe yourself?

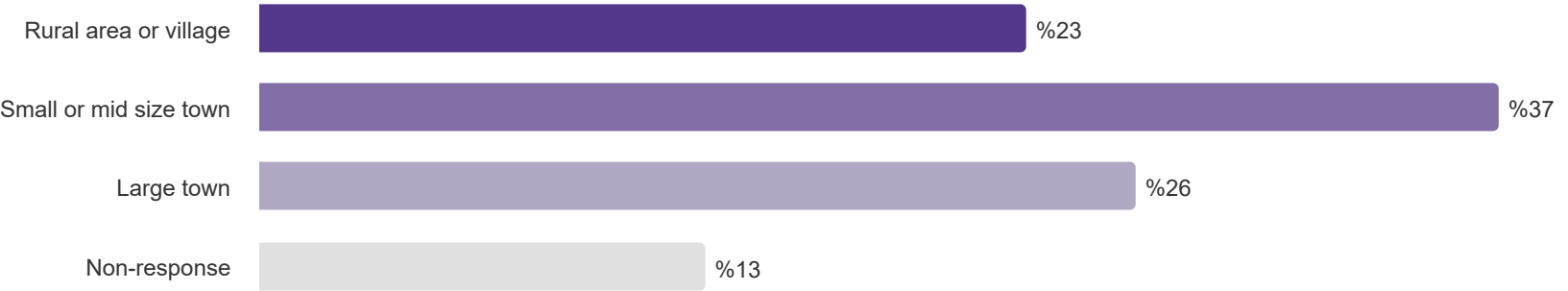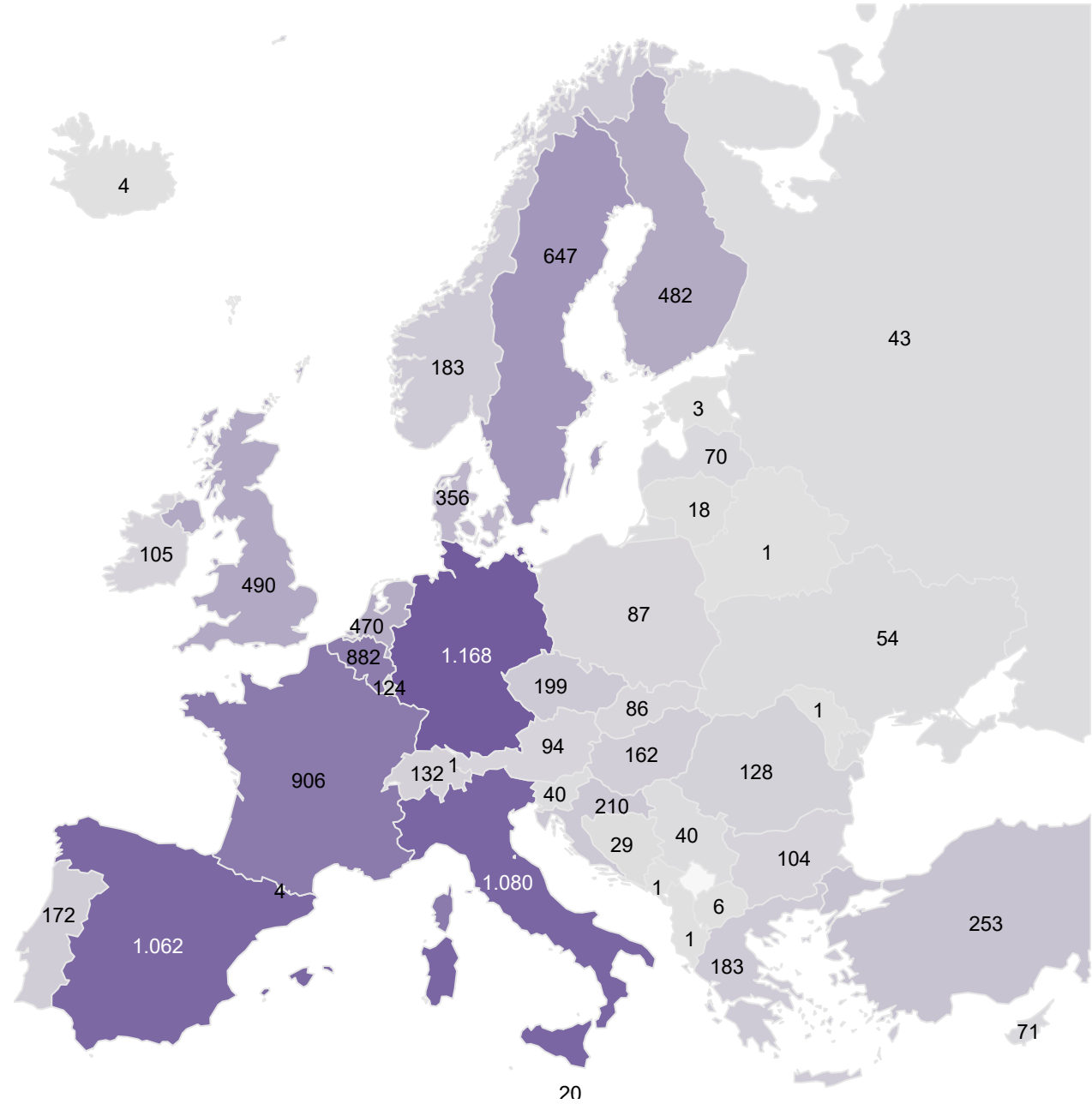| | N |
|---|---|
| I belong to the ethnic majority in the country where I live | 7.125 |
| I am part of an ethnic minority in the country where I live | 465 |
| Other, specify... | 337 |
| Non-response | 1.653 |
| **TOTAL** | **9.580** |

## How would you best describe yourself?

| | |
|---|---|
| I belong to the ethnic majority in the country where I live | %74 |
| I am part of an ethnic minority in the country where I live | %5 |
| Other, specify... | %4 |
| Non-response | %17 |
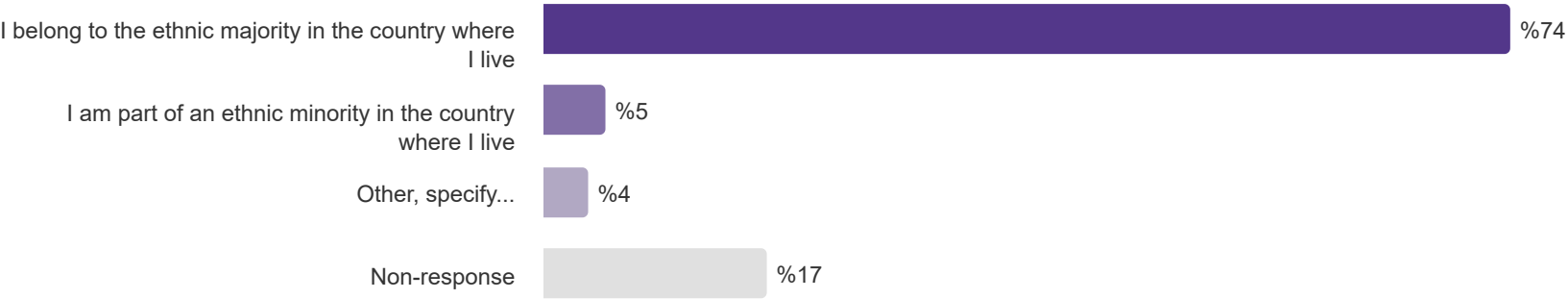
## Would you say that you, or the person you care for, live in a:

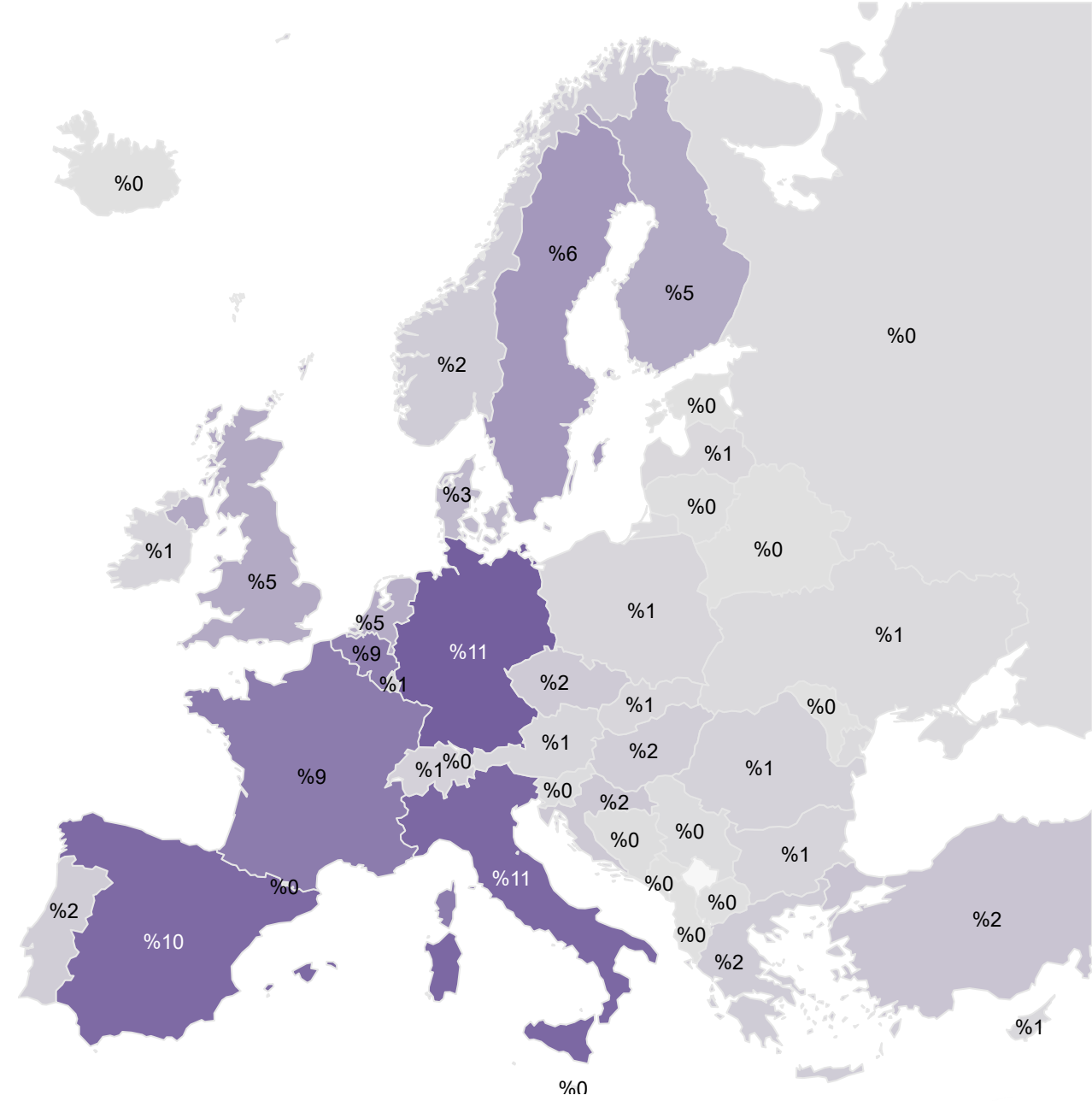| | N |
|---|---|
| Rural area or village | 2.416 |
| Small or mid size town | 3.901 |
| Large town | 2.760 |
| Non-response | 1.409 |
| **TOTAL** | **10.486** |

## Would you say that you, or the person you care for, live in a:

| | |
|---|---|
| Rural area or village | %23 |
| Small or mid size town | %37 |
| Large town | %26 |
| Non-response | %13 |

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## In which country do you live?



## In which country do you live?

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Questions as they appear in the questionnaire:

### Please select the sentence that best describes your situation or the situation of the person you care for:

| | N | % |
|---|---|---|
| I know the NAME of the rare disease, syndrome or malformation and it has been CONFIRMED by appropriate genetic, clinical, medical imaging, molecular or biochemical tests (e.g biopsy, blood or urine test) | 9.048 | %86 |
| I know the NAME of the rare disease, syndrome or malformation but it has NOT yet been confirmed by appropriate genetic, clinical, medical imaging, molecular or biochemical tests | 760 | %7 |
| I only have PARTIAL information on the name of the rare disease or the gene involved or the type of disease | 306 | %3 |
| I know that the disease is rare but the name or the cause have NOT BEEN IDENTIFIED | 348 | %3 |
| Other, specify... | 24 | %0 |
| **TOTAL** | **10.486** | **%100** |

### Simplified items corresponding to the questions above:

**Please select the sentence that best describes your situation or the situation of the person you care for:**

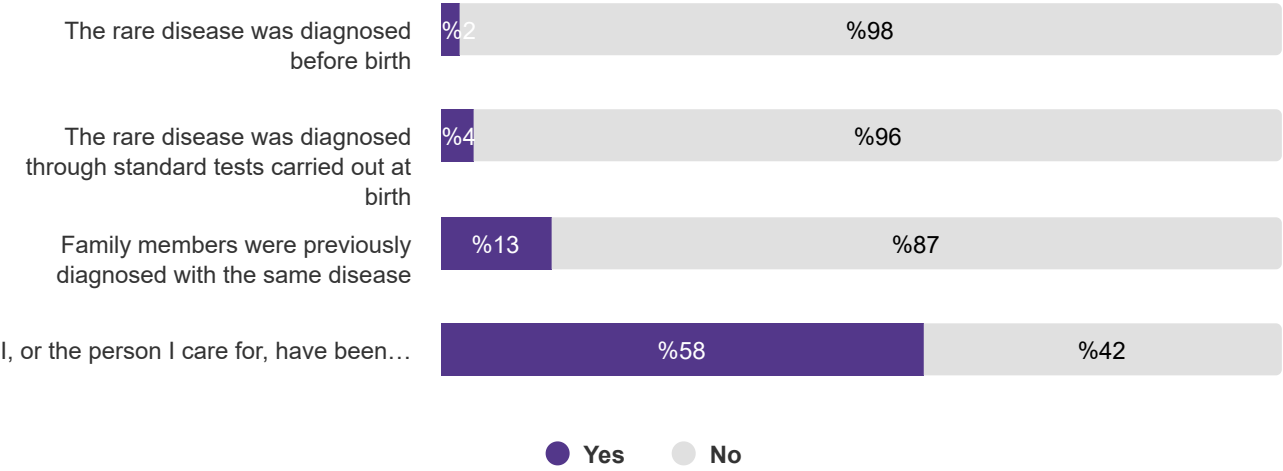| | N |
|---|---|
| Confirmed diagnosis | 9.048 |
| Initial diagnosis | 760 |
| Partial diagnosis | 306 |
| Unsolved case | 372 |
| **TOTAL** | **10.486** |

**Please select the sentence that best describes your situation or the situation of the person you care for:**

| | % |
|---|---|
| Confirmed diagnosis | %86 |
| Initial diagnosis | %7 |
| Partial diagnosis | %3 |
| Unsolved case | %4 |

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

## Do the following sentences apply to your situation?

| | YES | NO | TOTAL |
|---|---|---|---|
| The rare disease was diagnosed before birth | 222 | 9.513 | 9.735 |
| The rare disease was diagnosed through standard tests carried out at birth | 396 | 9.139 | 9.535 |
| Family members were previously diagnosed with the same disease | 1.309 | 8.426 | 9.735 |
| I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases | 5.998 | 4.415 | 10.413 |
| TOTAL | 7.925 | 31.493 | 39.418 |

## Do the following sentences apply to your situation?

The rare disease was diagnosed before birth: %2 Yes / %98 No

The rare disease was diagnosed through standard tests carried out at birth: %4 Yes / %96 No

Family members were previously diagnosed with the same disease: %13 Yes / %87 No

I, or the person I care for, have been…: %58 Yes / %42 No

● Yes    ● No

**Respondent living with a rare disease that is currently part of the NBS programme of the country they live in Source: ISNS**

## Newborn screening
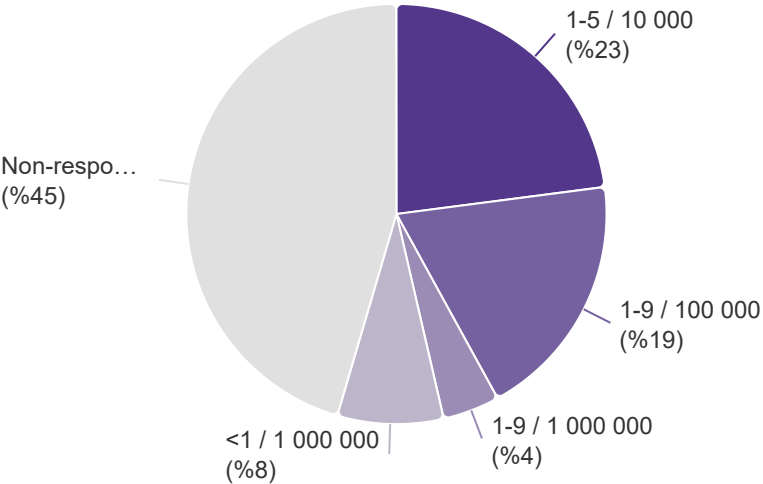
Respondents living with a disease that is being screened as part of a compulsory newborn screening programme in their country AND who answered "yes" to the question "The rare disease was diagnosed through standard tests carried out at birth" (see previous page).

Source: ISNS list of diseases screened per country.

https://membership.isns-neoscreening.org/public/screening-panels?export=0&name=&disorder=&region=2&country=&province=&pp=200

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*▼ Sample information : NBS_recod among "Yes"*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

Variables calculated based on the name of respondents' disease and Orphanet data

orphadata.org

**Calculation point prevalence**

**Calculation point prevalence**
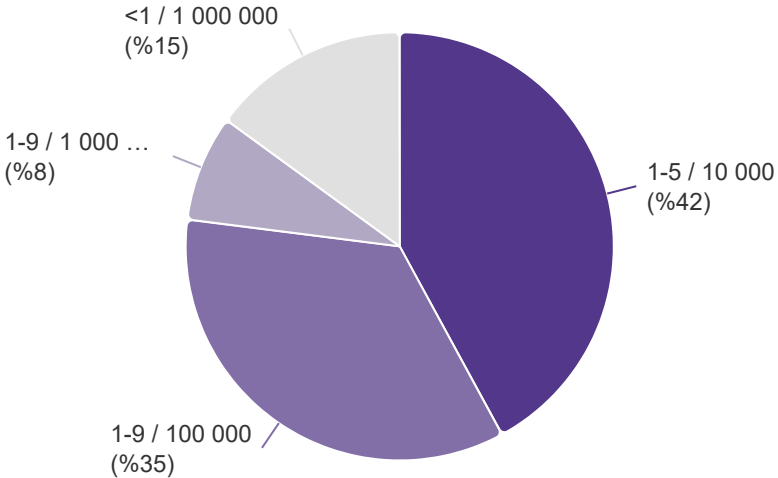
### Calculation of point prevalence 2 modalities

| | N | % |
|---|---|---|
| 1-5 / 10 000 | 2.407 | %23 |
| 1-9 / 100 000 | 1.999 | %19 |
| 1-9 / 1 000 000 | 459 | %4 |
| <1 / 1 000 000 | 856 | %8 |
| Non-response | 4.765 | %45 |
| **TOTAL** | **10.486** | **%100** |



1-5 / 10 000 (%23)

1-9 / 100 000 (%19)

1-9 / 1 000 000 (%4)

<1 / 1 000 000 (%8)

Non-respo... (%45)



<1 / 1 000 000 (%15)

1-9 / 1 000 ... (%8)

1-5 / 10 000 (%42)

1-9 / 100 000 (%35)

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

Variables calculated based on the name of respondents' disease and Orphanet data

orphadata.org

## Genetic diseases

|  | N |
|---|---|
| Genetic diseases | 5.447 |
| Non Genetic diseases | 2.627 |
| Non-response | 2.412 |
| **TOTAL** | **10.486** |

### Genetic diseases

| %52 | %25 | %23 |
|---|---|---|

- Genetic diseases
- Non Genetic diseases
- Non-response

## Transmission mode of the disease

|  | N |
|---|---|
| Autosomal dominant | 3.165 |
| Autosomal recessive | 1.147 |
| Mitochondrial inheritance | 25 |
| Multigenic/multifactorial | 882 |
| No data available | 15 |
| Not applicable | 1.486 |
| Unknown | 907 |
| X-linked dominant | 0 |
| X-linked recessive | 0 |
| Non-response | 2.858 |
| **TOTAL** | **10.485** |

## Transmission mode of the disease

Autosomal dominant (%30)

Autosomal recessive (%11)

Multigenic/multifactorial (%8)

Not applicable (%14)

Unknown (%9)

Non-response (%27)

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*
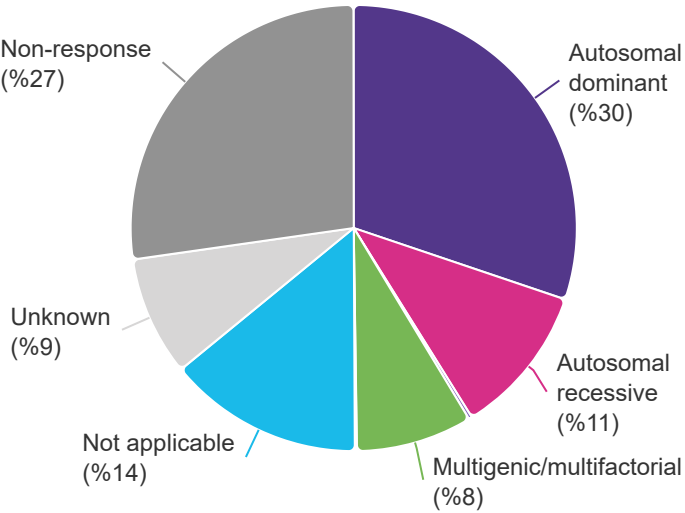
## orphacode

# 1.679

### Orphacode associated nomenclature (english)

| | N | % |
|---|---|---|
| Hereditary hemorrhagic telangiectasia | 458 | %5 |
| Hypermobile Ehlers-Danlos syndrome | 317 | %4 |
| Sarcoidosis | 170 | %2 |
| Classical Ehlers-Danlos syndrome | 137 | %2 |
| Williams syndrome | 136 | %2 |
| Cystic fibrosis | 128 | %2 |
| Myasthenia gravis | 120 | %1 |
| Systemic sclerosis | 107 | %1 |
| Tuberous sclerosis complex | 98 | %1 |
| Neurofibromatosis type 1 | 92 | %1 |
| Interstitial cystitis | 74 | %1 |
| Addison disease | 73 | %1 |
| 22q11.2 deletion syndrome | 68 | %1 |
| Chronic inflammatory demyelinating polyneuropathy | 65 | %1 |
| Perineural cyst | 63 | %1 |
| Acute inflammatory demyelinating polyradiculoneuropathy | 62 | %1 |
| Rett syndrome | 60 | %1 |
| Marfan syndrome | 52 | %1 |
| Fragile X syndrome | 49 | %1 |
| Behçet disease | 47 | %1 |
| Primary sclerosing cholangitis | 46 | %1 |
| Primary lymphedema | 43 | %1 |
| Granulomatosis with polyangiitis | 42 | %0 |

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

## EURORDIS European Federations

| | N |
|---|---|
| HHT Europe | 458 |
| Federation of European Scleroderma Associations | 200 |
| Sarcoidosis | 178 |
| Lupus Europe | 150 |
| European Myasthenia Gravis Association | 139 |
| European Federation of Williams Syndrome | 136 |
| CF Europe | 128 |
| NF Patients United | 125 |
| European Tuberous Sclerosis Complex Association | 98 |
| PHA Europe (Pulmonary Arterial Hypertension) | 86 |
| 22Q11 Europe | 80 |
| Multinational Interstitial Cystitis Association | 74 |
| Marfan Europe Network | 72 |
| Rett Syndrome Europe | 65 |
| Perineural cyst | 63 |
| European Federation for Hereditary Spastic Paraplegia | 52 |

## EURORDIS European Federations

| | N |
|---|---|
| European Fragile X Network | 49 |
| Sclerosing Cholangitis | 46 |
| European Society for Phenylketonuria | 45 |
| OIFE - Osteogenesis Imperfecta Federation Europe | 43 |
| Albi France | 41 |
| Duchenne Muscular Dystrophy | 41 |
| European Federation of Associations of Patients with Haemochromatosis | 41 |
| SMA Europe | 35 |
| MPS Europe | 34 |
| European Idiopathic Pulmonary Fibrosis & Related Disorders Federation | 32 |

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Orphanet_classification

Variables calculated based on the name of respondents' disease and Orphanet data

[orphadata.org](orphadata.org)

| | N | % |
|---|---|---|
| Abdominal surgical diseases | 239 | %3 |
| Allergic diseases | 3 | %0 |
| Bone diseases | 799 | %9 |
| Cardiac diseases | 660 | %8 |
| Cardiac malformations | 295 | %3 |
| Circulatory system diseases | 1.351 | %16 |
| Developmental anomalies during embryogenesis | 3.347 | %40 |
| Diseases due to toxic effects | 3 | %0 |
| Endocrine diseases | 995 | %12 |
| Gastroenterological diseases | 305 | %4 |
| Genetic diseases | 5.447 | %65 |
| Gynecologic/obstetric diseases | 284 | %3 |
| Hematological diseases | 412 | %5 |
| Hepatic diseases | 891 | %11 |
| Immunological diseases | 286 | %3 |
| Inborn errors of metabolism | 774 | %9 |
| Infectious diseases | 17 | %0 |
| Infertility | 410 | %5 |
| Neoplastic diseases | 870 | %10 |
| Neurological diseases | 4.169 | %49 |
| Odontological diseases | 222 | %3 |
| Ophthalmic diseases | 1.784 | %21 |
| Ophthalmic disorders | 7 | %0 |

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

# Chapter 2.

## Diagnosis journey

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Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

# Combined analysis

| Question | | | | | | |
|---|---|---|---|---|---|---|
| As far as you remember, when did you or a healthcare professional first notice the symptoms of the rare disease or think that something was wrong? | %5 | %7 | %12 | %22 | %43 | %11 |
| ...did you first seek MEDICAL HELP regarding the symptoms of the rare disease? | %4 | %6 | %12 | %22 | %43 | %12 |
| ...were you first referred to a HOSPITAL unit specialised in the rare disease or group of rare diseases, as part of the diagnosis or care of the rare disease? | %2 | %3 | %8 | %20 | %49 | %18 |
| As far as you remember, when did you first receive treatments, surgery or care to help you manage the symptoms associated with the rare disease or caused by this disease? | %3 | %4 | %8 | %20 | %46 | %18 |
| ...did you first know the NAME of the rare disease, syndrome or malformation? | %3 | %4 | %9 | %19 | %46 | %19 |
| ...was the name of the rare disease, syndrome or malformation CONFIRMED by appropriate genetic, clinical, medical imaging, molecular or biochemical tests (e.g biopsy, blood or urine test)? | %2 | %3 | %8 | %19 | %48 | %21 |

- ● Before the 01/01/1980
- ● From 01/01/1980 to 01/01/1990
- ● From 01/01/1990 to 01/01/2000
- ● From 01/01/2000 to 01/01/2010
- ● From 01/01/2010 to 01/01/2020
- ● After the 01/01/2020

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# Average diagnosis journey

**If number of years is negative, it means that on average the step of the diagnosis journey happened before first symptoms were noticed**

| | MEAN | LOWER QUARTILE | MEDIAN | UPPER QUARTILE | MINIMUM | MAXIMUM | FREQUENCY |
|---|---|---|---|---|---|---|---|
| Time between first symptoms and first medical contact, in years | 0,5 | 0,0 | 0,0 | 0,3 | -58,9 | 78,1 | 7.820 |
| Time between first symptom and first symptomatic treatment, in years | 3,5 | 0,0 | 0,5 | 3,6 | -56,9 | 62,7 | 7.322 |
| Time between first symptoms and first referral to a Centre of Expertise, in years | 3,9 | 0,0 | 0,4 | 3,4 | -51,9 | 70,0 | 4.335 |
| Time between first symptoms and initial diagnosis (first hearing the name of the disease), in years | 3,6 | 0,0 | 0,4 | 3,8 | -54,6 | 70,0 | 7.843 |
| Time between first symptoms and confirmed diagnosis, in years | 4,7 | 0,1 | 0,8 | 5,0 | -54,6 | 71,2 | 6.507 |
| Time from first symptom to when the questionnaire was filled, for undiagnosed respondents (unsolved cases and "other"), in years | 13,8 | 4,2 | 9,5 | 20,6 | 0,0 | 63,6 | 378 |

**First symptoms** = when they, or a healthcare professional, first noticed the symptoms of the rare disease or thought that something was wrong.

**First medical contact** = when they first seeked medical help regarding the symptoms of the rare disease

**First symptomatic treatment** = when they first receive treatments, surgery or care to help them manage the symptoms associated with the rare disease or caused by this disease.

**The initial diagnosis** = the first time they heard the name of the rare disease, syndrome or malformation.

**The first referral to a centre of expertise** (CoE) = when they were first referred to a hospital unit specialised in the rare disease or group of rare diseases, as part of the diagnosis or care of the rare disease (only for respondents who said they were referred to a centre of expertise).

**Confirmed diagnosis** = when the name of the rare disease, syndrome or malformation was confirmed by appropriate genetic, clinical, medical imaging, molecular or biochemical tests (e.g biopsy, blood or urine test).

| Average number of years | Number of years it took for **25%** of the sample to get to this step of their diagnosis journey | Number of years it took for **50%** of the sample to get to this step of their diagnosis journey | Number of years it took for **75%** of the sample to get to this step of their diagnosis journey | Lowest number of years | Highest number of years | Number of respondents for which we have the number of years to this step of the diagnosis journey |
|---|---|---|---|---|---|---|

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EURORDIS
RARE DISEASES EUROPE

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses



Time between first symptoms and confirmed diagnosis, in years

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Multiple Cross

| Gender of the person affected by the rare disease | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| Female | 0,6 | 5.053 | 4,1 | 4.750 | 4,6 | 2.787 | 4,2 | 5.050 | 5,4 | 4.193 |
| Male | 0,3 | 2.113 | 2,5 | 1.976 | 2,4 | 1.198 | 2,5 | 2.186 | 3,7 | 1.839 |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is weakly significant. p-value= 0,1 ; Fisher= 2,4.*

*Inter variance= 107,9. Intra variance= 45,4.*

**Mean** = average time, in number of years

**N** = number of respondents for which we have the average time

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Multiple Cross**

| How old were you when you stopped full-time education? | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| 15 y.o. or under | 0,6 | 292 | 3,7 | 298 | 4,1 | 170 | 3,9 | 312 | 4,9 | 253 |
| between 16 and 19 y.o. | 0,6 | 1.807 | 3,6 | 1.677 | 3,9 | 951 | 3,4 | 1.835 | 4,9 | 1.495 |
| between 20 and 23 y.o. | 0,5 | 2.340 | 3,5 | 2.163 | 3,9 | 1.323 | 3,6 | 2.345 | 4,7 | 1.987 |
| 24 y.o. or above | 0,4 | 2.435 | 3,5 | 2.303 | 3,8 | 1.372 | 3,7 | 2.454 | 4,7 | 2.067 |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is not significant. p-value= 0,8 ; Fisher= 0,3.*

*Inter variance= 13,5. Intra variance= 45,2.*

**Mean** = average time, in number of years

**N** = number of respondents for which we have the average time

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

## Multiple Cross

| How would you best describe yourself? | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| I belong to the ethnic majority in the country where I live | 0,3 | 5.468 | 3,5 | 5.124 | 3,7 | 2.962 | 3,5 | 5.494 | 4,7 | 4.605 |
| I am part of an ethnic minority in the country where I live | 0,8 | 324 | 3,3 | 305 | 3,9 | 156 | 3,6 | 323 | 4,2 | 268 |
| Other, specify... | 0,7 | 243 | 3,3 | 222 | 4,0 | 103 | 4,6 | 233 | 5,1 | 196 |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is not significant. p-value= 0,3 ; Fisher= 1,2.*

*Inter variance= 51,2. Intra variance= 44,3.*

**Mean** = average time, in number of years

**N** = number of respondents for which we have the average time

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

## Multiple Cross

| Would you say that you, or the person you care for, live in a: | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| Rural area or village | 0,4 | 1.841 | 3,5 | 1.732 | 4,1 | 1.019 | 3,7 | 1.847 | 4,8 | 1.520 |
| Small or mid size town | 0,4 | 2.974 | 3,7 | 2.749 | 3,8 | 1.656 | 3,5 | 2.981 | 4,8 | 2.500 |
| Large town | 0,6 | 2.051 | 3,4 | 1.955 | 3,8 | 1.138 | 3,5 | 2.111 | 4,8 | 1.776 |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is not significant. p-value= 0,4 ; Fisher= 0,8.*

*Inter variance= 36,5. Intra variance= 45,1.*

**Mean** = average time, in number of years

**N** = number of respondents for which we have the average time

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

| Please select the sentence that best describes your situation or the situation of the person you care for: | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| I know the NAME of the rare disease, syndrome or malformation and it has been CONFIRMED by appropriate genetic, clinical, medical imaging, molecular or biochemical tests (e.g biopsy, blood or urine test) | 0,5 | 6.818 | 3,4 | 6.506 | 3,6 | 3.876 | 3,3 | 7.135 | 4,7 | 6.501 |
| I know the NAME of the rare disease, syndrome or malformation but it has NOT yet been confirmed by appropriate genetic, clinical, medical imaging, molecular or biochemical tests | 0,8 | 566 | 5,2 | 475 | 6,1 | 228 | 6,6 | 539 | -6,0 | 3 |
| I only have PARTIAL information on the name of the rare disease or the gene involved or the type of disease | -0,2 | 190 | 3,1 | 167 | 6,5 | 95 | 6,2 | 167 | -0,7 | 2 |
| I know that the disease is rare but the name or the cause have NOT BEEN IDENTIFIED | 0,7 | 241 | 2,7 | 168 | 4,7 | 134 | 2,5 | 2 | 0,1 | 1 |
| Other, specify... | 0,9 | 5 | 9,7 | 6 | 11,0 | 2 | | 0 | | 0 |

■ *Under-represented elements*　　■ *Over-represented elements*

*The relationship is not significant. p-value= 0,5 ; Fisher= 0,9.*

*Inter variance= 40,1. Intra variance= 46,0.*

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Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

## Multiple Cross

| I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| Yes | 0,5 | 4.422 | 3,3 | 4.272 | 3,9 | 4.334 | 3,0 | 4.491 | 4,3 | 3.875 |
| No | 0,5 | 3.345 | 3,8 | 2.999 | 33,0 | 1 | 4,4 | 3.299 | 5,4 | 2.594 |

■ *Under-represented elements*　　■ *Over-represented elements*

*The relationship is not significant. p-value= 0,7 ; Fisher= 0,1.*

*Inter variance= 5,6. Intra variance= 46,2.*

**Mean** = average time, in number of years

**N** = number of respondents for which we have the average time

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

## Multiple Cross

| Family members were previously diagnosed with the same disease | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| Yes | 1,9 | 785 | 5,7 | 757 | 5,9 | 527 | 1,3 | 834 | 7,1 | 707 |
| No | 0,3 | 6.552 | 3,2 | 6.175 | 3,4 | 3.543 | 3,8 | 6.840 | 4,4 | 5.797 |

🟪 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Fisher= 40,1.*

*Inter variance= 1.832,5. Intra variance= 45,7.*

**Mean** = average time, in number of years

**N** = number of respondents for which we have the average time

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Multiple Cross

| Point prevalence of the rare disease | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| 1-5 / 10 000 | 0,8 | 1.802 | **4,4** | 1.753 | **4,9** | 1.087 | 3,4 | 1.925 | **5,7** | 1.674 |
| 1-9 / 100 000 | 0,3 | 1.544 | **3,2** | 1.486 | **3,1** | 929 | 3,1 | 1.640 | **4,0** | 1.417 |
| 1-9 / 1 000 000 | 0,1 | 352 | 3,8 | 330 | 3,7 | 188 | 4,1 | 379 | 5,0 | 335 |
| <1 / 1 000 000 | 0,3 | 638 | 3,4 | 567 | 3,8 | 326 | **4,9** | 654 | 5,5 | 528 |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is weakly significant. p-value= 0,1 ; Fisher= 2,1.*
*Inter variance= 98,0. Intra variance= 46,3.*

**Mean** = average time, in number of years

**N** = number of respondents for which we have the average time

**Disease prevalence:**

- **very rare diseases**: less than 1 case for 100,000 people

- **less rare diseases**: from 2 cases for 5,000 people to 1 case for 100,000 people.

- **Non-response**: unsolved cases (undiagnosed respondents) or disease prevalence unknown.

Source: orpha.data

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*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Multiple Cross

| Genetic diseases | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| Genetic diseases | 0,6 | 4.017 | 4,1 | 3.700 | 4,6 | 2.368 | 4,2 | 4.276 | 5,9 | 3.632 |
| Non Genetic diseases | 0,1 | 2.154 | 2,4 | 2.161 | 2,3 | 1.142 | 2,2 | 2.247 | 2,6 | 1.888 |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Fisher= 8,5.*

*Inter variance= 352,6. Intra variance= 41,6.*

**Mean** = average time, in number of years

**N** = number of respondents for which we have the average time

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

# Chapter 3.

## Age of the person affected when first symptoms were noticed

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Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

## Multiple Cross

| Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable) | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| Less than 2 years old | 1,1 | 1.699 | 3,2 | 1.576 | 3,4 | 989 | 4,4 | 1.762 | 5,0 | 1.529 |
| 2 to less that 10 years old | 1,8 | 758 | 6,5 | 705 | 7,7 | 417 | 7,0 | 789 | 8,8 | 666 |
| 10 to less than 20 years old | 3,1 | 819 | 8,3 | 767 | 9,7 | 438 | 8,0 | 815 | 10,4 | 629 |
| 20 to less than 30 years old | 0,6 | 841 | 4,2 | 773 | 4,3 | 451 | 3,8 | 841 | 5,5 | 691 |
| 30 to less than 50 years old | -0,7 | 2.062 | 2,3 | 1.904 | 2,2 | 1.120 | 1,6 | 2.005 | 2,7 | 1.671 |
| 50 years old or more | -1,5 | 941 | 0,3 | 943 | 0,6 | 549 | 0,0 | 972 | 0,6 | 807 |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Fisher= 64,8.*

*Inter variance= 2.821,1. Intra variance= 43,5.*

⚠️ **If number of years is negative, it means that on average the step of the diagnosis journey happened before first symptoms were noticed**

**Mean** = average time, in number of years

**N** = number of respondents for which we have the average time

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## Cross: Gender of the person affected by the rare disease / Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable)

| GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE | AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | | | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | LESS THAN 2 YEARS OLD | | 2 TO LESS THAT 10 YEARS OLD | | 10 TO LESS THAN 20 YEARS OLD | | 20 TO LESS THAN 30 YEARS OLD | | 30 TO LESS THAN 50 YEARS OLD | | 50 YEARS OLD OR MORE | | | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Female | 1.069 | %18 | 550 | %10 | 750 | %13 | 800 | %14 | 1.882 | %33 | 735 | %13 | 5.786 | %100 |
| Male | 939 | %38 | 360 | %14 | 188 | %8 | 174 | %7 | 461 | %19 | 369 | %15 | 2.491 | %100 |
| Other | 37 | %45 | 15 | %18 | 14 | %17 | 4 | %5 | 10 | %12 | 3 | %4 | 83 | %100 |
| TOTAL | 2.045 | %24 | 925 | %11 | 952 | %11 | 978 | %12 | 2.353 | %28 | 1.107 | %13 | 8.360 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 580,2 ; dof= 10.*

## Cross: How old were you when you stopped full-time education? / Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable)

| HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION? | AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | | | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | LESS THAN 2 YEARS OLD | | 2 TO LESS THAT 10 YEARS OLD | | 10 TO LESS THAN 20 YEARS OLD | | 20 TO LESS THAN 30 YEARS OLD | | 30 TO LESS THAN 50 YEARS OLD | | 50 YEARS OLD OR MORE | | | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| 15 y.o. or under | 114 | %29 | 64 | %17 | 51 | %13 | 31 | %8 | 81 | %21 | 46 | %12 | 387 | %100 |
| between 16 and 19 y.o. | 434 | %20 | 230 | %11 | 236 | %11 | 243 | %11 | 659 | %31 | 353 | %16 | 2.155 | %100 |
| between 20 and 23 y.o. | 668 | %25 | 315 | %12 | 285 | %10 | 323 | %12 | 771 | %28 | 357 | %13 | 2.719 | %100 |
| 24 y.o. or above | 807 | %29 | 293 | %10 | 329 | %12 | 335 | %12 | 755 | %27 | 310 | %11 | 2.829 | %100 |
| TOTAL | 2.023 | %25 | 902 | %11 | 901 | %11 | 932 | %12 | 2.266 | %28 | 1.066 | %13 | 8.090 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 100,0 ; dof= 15.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Would you say that you, or the person you care for, live in a: / Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable)

| WOULD YOU SAY THAT YOU, OR THE PERSON YOU CARE FOR, LIVE IN A: | LESS THAN 2 YEARS OLD | | 2 TO LESS THAT 10 YEARS OLD | | 10 TO LESS THAN 20 YEARS OLD | | 20 TO LESS THAN 30 YEARS OLD | | 30 TO LESS THAN 50 YEARS OLD | | 50 YEARS OLD OR MORE | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Rural area or village | 536 | %25 | _215_ | _%10_ | 234 | %11 | _221_ | _%10_ | _642_ | _%30_ | 305 | %14 | **2.153** | **%100** |
| Small or mid size town | 828 | %24 | 396 | %11 | 392 | %11 | 425 | %12 | 971 | %28 | 450 | %13 | **3.462** | **%100** |
| Large town | _655_ | _%27_ | 291 | %12 | 273 | %11 | 286 | %12 | _651_ | _%26_ | 310 | %13 | **2.466** | **%100** |
| **TOTAL** | **2.019** | **%25** | **902** | **%11** | **899** | **%11** | **932** | **%12** | **2.264** | **%28** | **1.065** | **%13** | **8.081** | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is significant. p-value= 0,0 ; Chi2= 19,9 ; dof= 10.*

## Cross: Typology of countries based on size and welfare / Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable)

| TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE | LESS THAN 2 YEARS OLD | | 2 TO LESS THAT 10 YEARS OLD | | 10 TO LESS THAN 20 YEARS OLD | | 20 TO LESS THAN 30 YEARS OLD | | 30 TO LESS THAN 50 YEARS OLD | | 50 YEARS OLD OR MORE | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Group A ('Eastern Europe') | _454_ | _%35_ | _213_ | _%16_ | 145 | %11 | 142 | %11 | _272_ | _%21_ | _67_ | _%5_ | **1.293** | **%100** |
| Group B ('Western Europe') | _986_ | _%23_ | _445_ | _%10_ | 486 | %11 | _543_ | _%13_ | _1.253_ | _%29_ | 593 | %14 | **4.306** | **%100** |
| Group C ('Northern Europe') | _601_ | _%22_ | _264_ | _%10_ | 315 | %12 | _288_ | _%11_ | _817_ | _%30_ | _442_ | _%16_ | **2.727** | **%100** |
| **TOTAL** | **2.041** | **%25** | **922** | **%11** | **946** | **%11** | **973** | **%12** | **2.342** | **%28** | **1.102** | **%13** | **8.326** | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 228,7 ; dof= 10.*

## Cross: How would you best describe yourself? / Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable)

| | AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | | | | | | | | | | | | | |
| | LESS THAN 2 YEARS OLD | | 2 TO LESS THAT 10 YEARS OLD | | 10 TO LESS THAN 20 YEARS OLD | | 20 TO LESS THAN 30 YEARS OLD | | 30 TO LESS THAN 50 YEARS OLD | | 50 YEARS OLD OR MORE | | TOTAL | |
| HOW WOULD YOU BEST DESCRIBE YOURSELF? | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| I belong to the ethnic majority in the country where I live | 1.612 | %25 | 705 | %11 | 716 | %11 | 723 | %11 | 1.778 | %28 | 836 | %13 | 6.370 | %100 |
| I am part of an ethnic minority in the country where I live | 107 | %27 | 57 | %14 | 36 | %9 | 61 | %15 | 113 | %28 | 26 | %7 | 400 | %100 |
| Other, specify... | 73 | %25 | 28 | %10 | 20 | %7 | 27 | %9 | 92 | %32 | 49 | %17 | 289 | %100 |
| TOTAL | 1.792 | %25 | 790 | %11 | 772 | %11 | 811 | %11 | 1.983 | %28 | 911 | %13 | 7.059 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 34,8 ; dof= 10.*

## Cross: Genetic diseases / Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable)

| | AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | | | | | | | | | | | | | | | |
| | LESS THAN 2 YEARS OLD | | 2 TO LESS THAT 10 YEARS OLD | | 10 TO LESS THAN 20 YEARS OLD | | 20 TO LESS THAN 30 YEARS OLD | | 30 TO LESS THAN 50 YEARS OLD | | 50 YEARS OLD OR MORE | | NON-RESPONSE | | TOTAL | |
| GENETIC DISEASES | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| Genetic diseases | 1.656 | %30 | 675 | %12 | 554 | %10 | 465 | %9 | 886 | %16 | 324 | %6 | 887 | %16 | 5.447 | %100 |
| Non Genetic diseases | 119 | %5 | 123 | %5 | 199 | %8 | 320 | %12 | 988 | %38 | 594 | %23 | 284 | %11 | 2.627 | %100 |
| TOTAL | 1.775 | %22 | 798 | %10 | 753 | %9 | 785 | %10 | 1.874 | %23 | 918 | %11 | 1.171 | %15 | 8.074 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 1.500,5 ; dof= 6.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable) / Please select the sentence that best describes your situation or the situation of the person you care for:**

| AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | PLEASE SELECT THE SENTENCE THAT BEST DESCRIBES YOUR SITUATION OR THE SITUATION OF THE PERSON YOU CARE FOR: | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| | CONFIRMED DIAGNOSIS | | INITIAL DIAGNOSIS | | PARTIAL DIAGNOSIS | | UNSOLVED CASE | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % |
| Less than 2 years old | **1.838** | **%90** | **95** | **%5** | 48 | %2 | 64 | %3 | 2.045 | %100 |
| 2 to less that 10 years old | 814 | %88 | 54 | %6 | 27 | %3 | 30 | %3 | 925 | %100 |
| 10 to less than 20 years old | **789** | **%83** | **98** | **%10** | 27 | %3 | 38 | %4 | 952 | %100 |
| 20 to less than 30 years old | 850 | %87 | 79 | %8 | 21 | %2 | 28 | %3 | 978 | %100 |
| 30 to less than 50 years old | **2.009** | **%85** | **188** | **%8** | 69 | %3 | 87 | %4 | 2.353 | %100 |
| 50 years old or more | 973 | %88 | 79 | %7 | 21 | %2 | 34 | %3 | 1.107 | %100 |
| **TOTAL** | **7.273** | **%87** | **593** | **%7** | **213** | **%3** | **281** | **%3** | **8.360** | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 50,0 ; dof= 15.*

**Cross: Calculation of point prevalence 2 modalities / Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable)**

| CALCULATION OF POINT PREVALENCE 2 MODALITIES | AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | LESS THAN 2 YEARS OLD | | 2 TO LESS THAT 10 YEARS OLD | | 10 TO LESS THAN 20 YEARS OLD | | 20 TO LESS THAN 30 YEARS OLD | | 30 TO LESS THAN 50 YEARS OLD | | 50 YEARS OLD OR MORE | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Respondents with less rare diseases | **772** | **%20** | **418** | **%11** | **481** | **%13** | **473** | **%12** | **1.127** | **%29** | **572** | **%15** | 3.843 | %100 |
| Respondents with very rare diseases | **365** | **%33** | **170** | **%16** | **102** | **%9** | **107** | **%10** | **239** | **%22** | **111** | **%10** | 1.094 | %100 |
| **TOTAL** | **1.137** | **%23** | **588** | **%12** | **583** | **%12** | **580** | **%12** | **1.366** | **%28** | **683** | **%14** | **4.937** | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 123,7 ; dof= 5.*

**Cross: Disease complexity classified into five groups, based on the number of affected body parts. / Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable)**

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | LESS THAN 2 YEARS OLD | | 2 TO LESS THAT 10 YEARS OLD | | 10 TO LESS THAN 20 YEARS OLD | | 20 TO LESS THAN 30 YEARS OLD | | 30 TO LESS THAN 50 YEARS OLD | | 50 YEARS OLD OR MORE | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| 1-3 body parts | 1.120 | %23 | 527 | %11 | 532 | %11 | 570 | %12 | 1.350 | %28 | 737 | %15 | 4.836 | %100 |
| 4-7 body parts | 677 | %27 | 269 | %11 | 274 | %11 | 269 | %11 | 701 | %28 | 279 | %11 | 2.469 | %100 |
| 8-11 body parts | 181 | %24 | 87 | %11 | 98 | %13 | 105 | %14 | 214 | %28 | 78 | %10 | 763 | %100 |
| 12-15 body parts | 54 | %23 | 28 | %12 | 37 | %16 | 27 | %11 | 76 | %32 | 13 | %6 | 235 | %100 |
| 16 body parts or more | 13 | %23 | 14 | %25 | 11 | %19 | 7 | %12 | 12 | %21 | 0 | %0 | 57 | %100 |
| TOTAL | 2.045 | %24 | 925 | %11 | 952 | %11 | 978 | %12 | 2.353 | %28 | 1.107 | %13 | 8.360 | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 83,5 ; dof= 20.*

**Cross: …behavioural disorders that cause problems in school, at home or in social situations / Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable)**

| …BEHAVIOURAL DISORDERS THAT CAUSE PROBLEMS IN SCHOOL, AT HOME OR IN SOCIAL SITUATIONS | AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | LESS THAN 2 YEARS OLD | | 2 TO LESS THAT 10 YEARS OLD | | 10 TO LESS THAN 20 YEARS OLD | | 20 TO LESS THAN 30 YEARS OLD | | 30 TO LESS THAN 50 YEARS OLD | | 50 YEARS OLD OR MORE | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 726 | %31 | 342 | %15 | 274 | %12 | 219 | %9 | 539 | %23 | 214 | %9 | 2.314 | %100 |
| No | 1.243 | %22 | 557 | %10 | 644 | %11 | 717 | %13 | 1.705 | %30 | 851 | %15 | 5.717 | %100 |
| Don't know | 76 | %23 | 26 | %8 | 34 | %10 | 42 | %13 | 109 | %33 | 42 | %13 | 329 | %100 |
| TOTAL | 2.045 | %24 | 925 | %11 | 952 | %11 | 978 | %12 | 2.353 | %28 | 1.107 | %13 | 8.360 | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 185,7 ; dof= 10.*

## Cross: …intellectual disabilities or cognitive symptoms (i.e. problems with memory, language, thinking or judgement) / Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable)

| …INTELLECTUAL DISABILITIES OR COGNITIVE SYMPTOMS (I.E. PROBLEMS WITH MEMORY, LANGUAGE, THINKING OR JUDGEMENT) | AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | | | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | LESS THAN 2 YEARS OLD | | 2 TO LESS THAT 10 YEARS OLD | | 10 TO LESS THAN 20 YEARS OLD | | 20 TO LESS THAN 30 YEARS OLD | | 30 TO LESS THAN 50 YEARS OLD | | 50 YEARS OLD OR MORE | | | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 900 | %38 | 314 | %13 | 190 | %8 | 210 | %9 | 563 | %24 | 189 | %8 | 2.366 | %100 |
| No | 1.099 | %19 | 586 | %10 | 744 | %13 | 733 | %13 | 1.723 | %30 | 874 | %15 | 5.759 | %100 |
| Don't know | 46 | %20 | 25 | %11 | 18 | %8 | 35 | %15 | 67 | %29 | 44 | %19 | 235 | %100 |
| TOTAL | 2.045 | %24 | 925 | %11 | 952 | %11 | 978 | %12 | 2.353 | %28 | 1.107 | %13 | 8.360 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 418,1 ; dof= 10.*

## Cross: …clinical signs or symptoms that come and go / Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable)

| …CLINICAL SIGNS OR SYMPTOMS THAT COME AND GO | AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | | | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | LESS THAN 2 YEARS OLD | | 2 TO LESS THAT 10 YEARS OLD | | 10 TO LESS THAN 20 YEARS OLD | | 20 TO LESS THAN 30 YEARS OLD | | 30 TO LESS THAN 50 YEARS OLD | | 50 YEARS OLD OR MORE | | | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 963 | %20 | 506 | %10 | 600 | %12 | 651 | %14 | 1.488 | %31 | 612 | %13 | 4.820 | %100 |
| No | 908 | %30 | 359 | %12 | 295 | %10 | 282 | %9 | 723 | %24 | 413 | %14 | 2.980 | %100 |
| Don't know | 174 | %31 | 60 | %11 | 57 | %10 | 45 | %8 | 142 | %25 | 82 | %15 | 560 | %100 |
| TOTAL | 2.045 | %24 | 925 | %11 | 952 | %11 | 978 | %12 | 2.353 | %28 | 1.107 | %13 | 8.360 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 174,5 ; dof= 10.*

### Cross: …invisible symptoms such as pain, dizziness, headaches, fatigue, etc. / Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable)

| …INVISIBLE SYMPTOMS SUCH AS PAIN, DIZZINESS, HEADACHES, FATIGUE, ETC. | AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | | | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | LESS THAN 2 YEARS OLD | | 2 TO LESS THAT 10 YEARS OLD | | 10 TO LESS THAN 20 YEARS OLD | | 20 TO LESS THAN 30 YEARS OLD | | 30 TO LESS THAN 50 YEARS OLD | | 50 YEARS OLD OR MORE | | | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 821 | %14 | 555 | %10 | 752 | %13 | 795 | %14 | 1.939 | %34 | 804 | %14 | 5.666 | %100 |
| No | 966 | %42 | 307 | %14 | 176 | %8 | 170 | %7 | 379 | %17 | 276 | %12 | 2.274 | %100 |
| Don't know | 258 | %61 | 63 | %15 | 24 | %6 | 13 | %3 | 35 | %8 | 27 | %6 | 420 | %100 |
| TOTAL | 2.045 | %24 | 925 | %11 | 952 | %11 | 978 | %12 | 2.353 | %28 | 1.107 | %13 | 8.360 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 1.196,8 ; dof= 10.*

### Cross: …sudden onset symptoms requiring urgent care / Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable)

| …SUDDEN ONSET SYMPTOMS REQUIRING URGENT CARE | AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | | | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | LESS THAN 2 YEARS OLD | | 2 TO LESS THAT 10 YEARS OLD | | 10 TO LESS THAN 20 YEARS OLD | | 20 TO LESS THAN 30 YEARS OLD | | 30 TO LESS THAN 50 YEARS OLD | | 50 YEARS OLD OR MORE | | | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 891 | %24 | 397 | %11 | 435 | %12 | 491 | %13 | 1.072 | %29 | 458 | %12 | 3.744 | %100 |
| No | 1.040 | %25 | 479 | %11 | 476 | %11 | 440 | %11 | 1.150 | %28 | 594 | %14 | 4.179 | %100 |
| Don't know | 114 | %26 | 49 | %11 | 41 | %9 | 47 | %11 | 131 | %30 | 55 | %13 | 437 | %100 |
| TOTAL | 2.045 | %24 | 925 | %11 | 952 | %11 | 978 | %12 | 2.353 | %28 | 1.107 | %13 | 8.360 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 23,6 ; dof= 10.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Family members were previously diagnosed with the same disease / Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable)

| FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | LESS THAN 2 YEARS OLD | | 2 TO LESS THAT 10 YEARS OLD | | 10 TO LESS THAN 20 YEARS OLD | | 20 TO LESS THAN 30 YEARS OLD | | 30 TO LESS THAN 50 YEARS OLD | | 50 YEARS OLD OR MORE | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 134 | %14 | 89 | %9 | 144 | %15 | 164 | %17 | 297 | %31 | 119 | %13 | 947 | %100 |
| No | 1.795 | %26 | 775 | %11 | 737 | %11 | 758 | %11 | 1.873 | %27 | 926 | %13 | 6.864 | %100 |
| TOTAL | 1.929 | %25 | 864 | %11 | 881 | %11 | 922 | %12 | 2.170 | %28 | 1.045 | %13 | 7.811 | |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 99,2 ; dof= 5.*

## Cross: Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable) / I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases

| AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | | | | | |
|---|---|---|---|---|---|---|
| | YES | | NO | | TOTAL | |
| | N | % | N | % | N | % |
| Less than 2 years old | 1.219 | %60 | 817 | %40 | 2.036 | %100 |
| 2 to less that 10 years old | 540 | %59 | 380 | %41 | 920 | %100 |
| 10 to less than 20 years old | 537 | %57 | 413 | %43 | 950 | %100 |
| 20 to less than 30 years old | 535 | %55 | 441 | %45 | 976 | %100 |
| 30 to less than 50 years old | 1.312 | %56 | 1.028 | %44 | 2.340 | %100 |
| 50 years old or more | 644 | %58 | 460 | %42 | 1.104 | %100 |
| TOTAL | 4.787 | %57 | 3.539 | %43 | 8.326 | |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is weakly significant. p-value= 0,1 ; Chi2= 10,7 ; dof= 5.*

## Cross: Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable) / How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?

| AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | 0 | | 1 | | BETWEEN 2 AND 4 | | BETWEEN 5 AND 7 | | BETWEEN 8 AND 10 | | MORE THAN 10 | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Less than 2 years old | 46 | %2 | 270 | %13 | 848 | %41 | 379 | %19 | 138 | %7 | 364 | %18 | 2.045 | %100 |
| 2 to less that 10 years old | 11 | %1 | 70 | %8 | 381 | %41 | 194 | %21 | 74 | %8 | 195 | %21 | 925 | %100 |
| 10 to less than 20 years old | 9 | %1 | 70 | %7 | 346 | %36 | 182 | %19 | 86 | %9 | 259 | %27 | 952 | %100 |
| 20 to less than 30 years old | 6 | %1 | 93 | %10 | 424 | %43 | 185 | %19 | 79 | %8 | 191 | %20 | 978 | %100 |
| 30 to less than 50 years old | 17 | %1 | 217 | %9 | 1.062 | %45 | 530 | %23 | 197 | %8 | 330 | %14 | 2.353 | %100 |
| 50 years old or more | 12 | %1 | 138 | %12 | 605 | %55 | 193 | %17 | 68 | %6 | 91 | %8 | 1.107 | %100 |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 271,1 ; dof= 25.*

## Cross: Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable) / …wrongly attributed to another physical disease?

| AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | …WRONGLY ATTRIBUTED TO ANOTHER PHYSICAL DISEASE? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Less than 2 years old | 338 | %17 | 679 | %33 | 1.028 | %50 | 2.045 | %100 |
| 2 to less that 10 years old | 176 | %19 | 377 | %41 | 372 | %40 | 925 | %100 |
| 10 to less than 20 years old | 176 | %18 | 489 | %51 | 287 | %30 | 952 | %100 |
| 20 to less than 30 years old | 185 | %19 | 507 | %52 | 286 | %29 | 978 | %100 |
| 30 to less than 50 years old | 479 | %20 | 1.183 | %50 | 691 | %29 | 2.353 | %100 |
| 50 years old or more | 242 | %22 | 420 | %38 | 445 | %40 | 1.107 | %100 |
| TOTAL | 1.596 | %19 | 3.655 | %44 | 3.109 | %37 | 8.360 | %100 |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 293,4 ; dof= 10.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable) / …neglected, not taken seriously and/or considered as psychological?**

| AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | …NEGLECTED, NOT TAKEN SERIOUSLY AND/OR CONSIDERED AS PSYCHOLOGICAL? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Less than 2 years old | 206 | %10 | 717 | %35 | 1.122 | %55 | 2.045 | %100 |
| 2 to less that 10 years old | 91 | %10 | 458 | %50 | 376 | %41 | 925 | %100 |
| 10 to less than 20 years old | 96 | %10 | 597 | %63 | 259 | %27 | 952 | %100 |
| 20 to less than 30 years old | 115 | %12 | 561 | %57 | 302 | %31 | 978 | %100 |
| 30 to less than 50 years old | 334 | %14 | 1.249 | %53 | 770 | %33 | 2.353 | %100 |
| 50 years old or more | 139 | %13 | 420 | %38 | 548 | %50 | 1.107 | %100 |
| TOTAL | 981 | %12 | 4.002 | %48 | 3.377 | %40 | 8.360 | %100 |

🟪 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 417,5 ; dof= 10.*

**Cross: Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable) / Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed.**

| AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Less than 2 years old | 525 | %26 | 748 | %37 | 772 | %38 | 2.045 | %100 |
| 2 to less that 10 years old | 282 | %30 | 414 | %45 | 229 | %25 | 925 | %100 |
| 10 to less than 20 years old | 252 | %26 | 534 | %56 | 166 | %17 | 952 | %100 |
| 20 to less than 30 years old | 260 | %27 | 545 | %56 | 173 | %18 | 978 | %100 |
| 30 to less than 50 years old | 591 | %25 | 1.312 | %56 | 450 | %19 | 2.353 | %100 |
| 50 years old or more | 278 | %25 | 488 | %44 | 341 | %31 | 1.107 | %100 |
| TOTAL | | %26 | 4.041 | %48 | | %25 | | |

🟪 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 341,2 ; dof= 10.*

## Cross: Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable) / Genetic test(s) looking for genetic changes (also called mutations or variants)

| AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | GENETIC TEST(S) LOOKING FOR GENETIC CHANGES (ALSO CALLED MUTATIONS OR VARIANTS) | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW/DON'T REMEMBER | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Less than 2 years old | 1.648 | %81 | 332 | %16 | 65 | %3 | 2.045 | %100 |
| 2 to less that 10 years old | 657 | %71 | 218 | %24 | 50 | %5 | 925 | %100 |
| 10 to less than 20 years old | 484 | %51 | 388 | %41 | 80 | %8 | 952 | %100 |
| 20 to less than 30 years old | 425 | %43 | 461 | %47 | 92 | %9 | 978 | %100 |
| 30 to less than 50 years old | 863 | %37 | 1.304 | %55 | 186 | %8 | 2.353 | %100 |
| 50 years old or more | 300 | %27 | 681 | %62 | 126 | %11 | 1.107 | %100 |
| TOTAL | 4.377 | %52 | 3.384 | %40 | 599 | %7 | 8.360 | %100 |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 1.344,8 ; dof= 10.*

## Cross: Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable) / Other test(s) such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc

| AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | OTHER TEST(S) SUCH AS CLINICAL EXAMINATION(S), MEDICAL IMAGING (MRI, SCANS…), BIOPSY, BIOCHEMICAL TEST(S) (BLOOD OR URINE TESTS…), ETC | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW/DON'T REMEMBER | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Less than 2 years old | 1.788 | %87 | 190 | %9 | 67 | %3 | 2.045 | %100 |
| 2 to less that 10 years old | 830 | %90 | 73 | %8 | 22 | %2 | 925 | %100 |
| 10 to less than 20 years old | 894 | %94 | 37 | %4 | 21 | %2 | 952 | %100 |
| 20 to less than 30 years old | 919 | %94 | 49 | %5 | 10 | %1 | 978 | %100 |
| 30 to less than 50 years old | 2.192 | %93 | 123 | %5 | 38 | %2 | 2.353 | %100 |
| 50 years old or more | 1.038 | %94 | 49 | %4 | 20 | %2 | 1.107 | %100 |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 83,7 ; dof= 10.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable) / …you could not afford it?**

Have you ever needed a genetic test but could not access it because... …YOU COULD NOT AFFORD IT?

| AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | YES | | NO | | NOT RELEVANT | | TOTAL | |
|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % |
| Less than 2 years old | 199 | %10 | 1.565 | %77 | 281 | %14 | 2.045 | %100 |
| 2 to less that 10 years old | 111 | %12 | 679 | %73 | 135 | %15 | 925 | %100 |
| 10 to less than 20 years old | 110 | %12 | 639 | %67 | 203 | %21 | 952 | %100 |
| 20 to less than 30 years old | 112 | %11 | 638 | %65 | 228 | %23 | 978 | %100 |
| 30 to less than 50 years old | 249 | %11 | 1.509 | %64 | 595 | %25 | 2.353 | %100 |
| 50 years old or more | 56 | %5 | 681 | %62 | 370 | %33 | 1.107 | %100 |
| **TOTAL** | 837 | %10 | 5.711 | %68 | 1.812 | %22 | 8.360 | %100 |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 241,5 ; dof= 10.*

**Cross: Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable) / …it was not available in your country?**

Have you ever needed a genetic test but could not access it because... …IT WAS NOT AVAILABLE IN YOUR COUNTRY?

| AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | YES | | NO | | NOT RELEVANT | | TOTAL | |
|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % |
| Less than 2 years old | 279 | %14 | 1.482 | %72 | 284 | %14 | 2.045 | %100 |
| 2 to less that 10 years old | 124 | %13 | 667 | %72 | 134 | %14 | 925 | %100 |
| 10 to less than 20 years old | 131 | %14 | 597 | %63 | 224 | %24 | 952 | %100 |
| 20 to less than 30 years old | 98 | %10 | 626 | %64 | 254 | %26 | 978 | %100 |
| 30 to less than 50 years old | 213 | %9 | 1.467 | %62 | 673 | %29 | 2.353 | %100 |
| 50 years old or more | 74 | %7 | 628 | %57 | 405 | %37 | 1.107 | %100 |
| **TOTAL** | 919 | %11 | 5.467 | %65 | 1.974 | %24 | 8.360 | %100 |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 312,2 ; dof= 10.*

**Cross: Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable) / ...healthcare professionals were reluctant or not sufficiently informed?**

Have you ever needed a genetic test but could not access it because...

**...HEALTHCARE PROFESSIONALS WERE RELUCTANT OR NOT SUFFICIENTLY INFORMED?**

| AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | YES | | NO | | NOT RELEVANT | | TOTAL | |
|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % |
| Less than 2 years old | 543 | %27 | 1.254 | %61 | 248 | %12 | 2.045 | %100 |
| 2 to less that 10 years old | 272 | %29 | 535 | %58 | 118 | %13 | 925 | %100 |
| 10 to less than 20 years old | 321 | %34 | 453 | %48 | 178 | %19 | 952 | %100 |
| 20 to less than 30 years old | 295 | %30 | 478 | %49 | 205 | %21 | 978 | %100 |
| 30 to less than 50 years old | 608 | %26 | 1.141 | %48 | 604 | %26 | 2.353 | %100 |
| 50 years old or more | 183 | %17 | 556 | %50 | 368 | %33 | 1.107 | %100 |
| **TOTAL** | **2.222** | **%27** | **4.417** | **%53** | **1.721** | **%21** | **8.360** | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 334,3 ; dof= 10.*

**Cross: Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable) / To your knowledge, the genetic test(s) that were conducted targeted...**

**TO YOUR KNOWLEDGE, THE GENETIC TEST(S) THAT WERE CONDUCTED TARGETED...**

| AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | ONLY ONE GENE | | SEVERAL GENES AT THE SAME TIME (GENE PANEL SEQUENCING) | | THE WHOLE DNA (WHOLE GENOME SEQUENCING) | | ALL THE GENES (WHOLE EXOME SEQUENCING) | | A TUMOUR (GENETIC PROFILING OF A TUMOUR) | | OTHER (EPIGENOME, RNA, ETC.) | | DON'T KNOW | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Less than 2 years old | 456 | %28 | 532 | %32 | 341 | %21 | 269 | %16 | 17 | %1 | 41 | %2 | 369 | %22 | 1.648 | |
| 2 to less that 10 years old | 161 | %25 | 216 | %33 | 119 | %18 | 77 | %12 | 14 | %2 | 12 | %2 | 175 | %27 | 657 | |
| 10 to less than 20 years old | 113 | %23 | 170 | %35 | 48 | %10 | 33 | %7 | 13 | %3 | 8 | %2 | 154 | %32 | 484 | |
| 20 to less than 30 years old | 122 | %29 | 144 | %34 | 54 | %13 | 22 | %5 | 16 | %4 | 12 | %3 | 118 | %28 | 425 | |
| 30 to less than 50 years old | 233 | %27 | 277 | %32 | 98 | %11 | 56 | %6 | 43 | %5 | 15 | %2 | 260 | %30 | 863 | |
| 50 years old or more | 83 | %28 | 83 | %27 | 35 | %12 | 14 | %5 | 8 | %2 | 3 | %1 | 104 | %25 | 393 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 227,3 ; dof= 30.*

## Cross: Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable) / Did you ever request a private company or laboratory to conduct genetic testing to diagnose the disease?

| AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | DID YOU EVER REQUEST A PRIVATE COMPANY OR LABORATORY TO CONDUCT GENETIC TESTING TO DIAGNOSE THE DISEASE? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO, NEVER | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Less than 2 years old | 177 | %11 | 84 | %5 | 1.387 | %84 | 1.648 | %100 |
| 2 to less that 10 years old | 83 | %13 | 35 | %5 | 539 | %82 | 657 | %100 |
| 10 to less than 20 years old | 54 | %11 | 18 | %4 | 412 | %85 | 484 | %100 |
| 20 to less than 30 years old | 38 | %9 | 17 | %4 | 370 | %87 | 425 | %100 |
| 30 to less than 50 years old | 86 | %10 | 44 | %5 | 733 | %85 | 863 | %100 |
| 50 years old or more | 20 | %7 | 12 | %4 | 268 | %89 | 300 | %100 |
| TOTAL | 458 | %10 | 210 | %5 | 3.709 | %85 | 4.377 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is not significant. p-value= 0,2 ; Chi2= 13,2 ; dof= 10.*

## Cross: Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable) / In general, how satisfied are you with how the results of the GENETIC TESTS were given to you?

| AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | IN GENERAL, HOW SATISFIED ARE YOU WITH HOW THE RESULTS OF THE GENETIC TESTS WERE GIVEN TO YOU? | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | VERY DISSATISFIED | | DISSATISFIED | | NEITHER SATISFIED NOR DISSATISFIED | | SATISFIED | | VERY SATISFIED | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Less than 2 years old | 197 | %12 | 200 | %12 | 343 | %21 | 572 | %35 | 267 | %16 | 69 | %4 | 1.648 | %100 |
| 2 to less that 10 years old | 63 | %10 | 95 | %14 | 138 | %21 | 221 | %34 | 116 | %18 | 24 | %4 | 657 | %100 |
| 10 to less than 20 years old | 44 | %9 | 72 | %15 | 109 | %23 | 148 | %31 | 85 | %18 | 26 | %5 | 484 | %100 |
| 20 to less than 30 years old | 47 | %11 | 58 | %14 | 87 | %20 | 142 | %33 | 70 | %16 | 21 | %5 | 425 | %100 |
| 30 to less than 50 years old | 83 | %10 | 77 | %9 | 200 | %23 | 295 | %34 | 149 | %17 | 59 | %7 | 863 | %100 |
| 50 years old or more | 29 | %10 | 20 | %7 | 42 | %14 | 124 | %41 | 64 | %21 | 21 | %7 | 300 | %100 |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 60,5 ; dof= 25.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable) / After the tests were performed, were you offered genetic counselling (e.g. given information about how your genetic condition might affect you and your family)?

| AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | AFTER THE TESTS WERE PERFORMED, WERE YOU OFFERED GENETIC COUNSELLING (E.G. GIVEN INFORMATION ABOUT HOW YOUR GENETIC CONDITION MIGHT AFFECT YOU AND YOUR FAMILY)? | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| | YES, WITH A GENETIC COUNSELLOR OR CLINICAL GENETICIST | | YES, BY A HEALTHCARE PROFESSIONAL | | NO, I WASN'T OFFERED GENETIC COUNSELLING | | NOT SURE / DON'T REMEMBER | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % |
| Less than 2 years old | 793 | %48 | 322 | %20 | 413 | %25 | 120 | %7 | 1.648 | %100 |
| 2 to less that 10 years old | 252 | %38 | 140 | %21 | 222 | %34 | 43 | %7 | 657 | %100 |
| 10 to less than 20 years old | 157 | %32 | 108 | %22 | 176 | %36 | 43 | %9 | 484 | %100 |
| 20 to less than 30 years old | 134 | %32 | 89 | %21 | 179 | %42 | 23 | %5 | 425 | %100 |
| 30 to less than 50 years old | 272 | %32 | 210 | %24 | 318 | %37 | 63 | %7 | 863 | %100 |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 122,3 ; dof= 15.*

## Cross: Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable) / Genetic tests

| AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | GENETIC TESTS | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Less than 2 years old | 296 | %18 | 1.314 | %80 | 38 | %2 | 1.648 | %100 |
| 2 to less that 10 years old | 130 | %20 | 511 | %78 | 15 | %2 | 656 | %100 |
| 10 to less than 20 years old | 62 | %13 | 405 | %84 | 17 | %4 | 484 | %100 |
| 20 to less than 30 years old | 55 | %13 | 360 | %85 | 10 | %2 | 425 | %100 |
| 30 to less than 50 years old | 103 | %12 | 739 | %86 | 21 | %2 | 863 | %100 |
| 50 years old or more | 26 | %9 | 271 | %90 | 3 | %1 | 300 | %100 |
| **TOTAL** | 672 | %15 | 3.600 | %82 | 104 | %2 | 4.376 | %100 |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 46,6 ; dof= 10.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable) / Other diagnostic tests such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc.**

| AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | OTHER DIAGNOSTIC TESTS SUCH AS CLINICAL EXAMINATION(S), MEDICAL IMAGING (MRI, SCANS…), BIOPSY, BIOCHEMICAL TEST(S) (BLOOD OR URINE TESTS…), ETC. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Less than 2 years old | 280 | %16 | **1.470** | **%82** | 37 | %2 | **1.787** | %100 |
| 2 to less that 10 years old | **143** | **%17** | 677 | %82 | 10 | %1 | **830** | %100 |
| 10 to less than 20 years old | 127 | %14 | 754 | %84 | 13 | %1 | **894** | %100 |
| 20 to less than 30 years old | 144 | %16 | 758 | %82 | 17 | %2 | **919** | %100 |
| 30 to less than 50 years old | 294 | %13 | 1.861 | %85 | 37 | %2 | **2.192** | %100 |
| 50 years old or more | 133 | %13 | **896** | **%86** | **9** | **%1** | **1.038** | %100 |
| TOTAL | | | | | | | | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is significant. p-value= 0,0 ; Chi2= 20,1 ; dof= 10.*

**Cross: Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable) / Additional advice from a healthcare professional specialised in the rare disease (in person or virtually)**

| AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | ADDITIONAL ADVICE FROM A HEALTHCARE PROFESSIONAL SPECIALISED IN THE RARE DISEASE (IN PERSON OR VIRTUALLY) | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Less than 2 years old | **515** | **%25** | **1.485** | **%73** | **45** | **%2** | **2.045** | %100 |
| 2 to less that 10 years old | **230** | **%25** | **676** | **%73** | 19 | %2 | **925** | %100 |
| 10 to less than 20 years old | 183 | %19 | 753 | %79 | 16 | %2 | **952** | %100 |
| 20 to less than 30 years old | 185 | %19 | 777 | %79 | 16 | %2 | **978** | %100 |
| 30 to less than 50 years old | **414** | **%18** | **1.908** | **%81** | 31 | %1 | **2.353** | %100 |
| 50 years old or more | **145** | **%13** | **956** | **%86** | **6** | **%1** | **1.107** | %100 |
| TOTAL | | | | | | | | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 110,7 ; dof= 10.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable) / …psychological support**

| AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | …PSYCHOLOGICAL SUPPORT | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Less than 2 years old | 200 | %10 | 180 | %9 | 211 | %10 | 489 | %24 | 965 | %47 | 2.045 | %100 |
| 2 to less that 10 years old | 72 | %8 | 93 | %10 | 88 | %10 | 236 | %26 | 436 | %47 | 925 | %100 |
| 10 to less than 20 years old | 73 | %8 | 87 | %9 | 113 | %12 | 254 | %27 | 425 | %45 | 952 | %100 |
| 20 to less than 30 years old | 67 | %7 | 76 | %8 | 85 | %9 | 297 | %30 | 453 | %46 | 978 | %100 |
| 30 to less than 50 years old | 213 | %9 | 213 | %9 | 212 | %9 | 739 | %31 | 976 | %41 | 2.353 | %100 |
| 50 years old or more | 106 | %10 | 105 | %9 | 64 | %6 | 477 | %43 | 355 | %32 | 1.107 | %100 |
| TOTAL | 731 | %9 | 754 | %9 | 773 | %9 | 2.492 | %30 | 3.610 | %43 | 8.360 | %100 |

🟪 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 184,8 ; dof= 20.*

**Cross: Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable) / …care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc.**

| AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | …CARE COORDINATION SUPPORT SUCH AS HELP TO FIND THE NECESSARY INFORMATION ON THE DISEASE AND THE RIGHT PROFESSIONALS, ARRANGING APPOINTMENTS WITH DIFFERENT HEALTH PROVIDERS, ETC. | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Less than 2 years old | 471 | %23 | 66 | %3 | 341 | %17 | 216 | %11 | 951 | %47 | 2.045 | %100 |
| 2 to less that 10 years old | 199 | %22 | 36 | %4 | 141 | %15 | 99 | %11 | 450 | %49 | 925 | %100 |
| 10 to less than 20 years old | 146 | %15 | 26 | %3 | 123 | %13 | 135 | %14 | 522 | %55 | 952 | %100 |
| 20 to less than 30 years old | 153 | %16 | 27 | %3 | 124 | %13 | 159 | %16 | 515 | %53 | 978 | %100 |
| 30 to less than 50 years old | 422 | %18 | 78 | %3 | 313 | %13 | 397 | %17 | 1.143 | %49 | 2.353 | %100 |

🟪 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 168,0 ; dof= 20.*

Rare Barometer — A EURORDIS ℞ INITIATIVE

EURORDIS RARE DISEASES EUROPE

## Cross: Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable) / …financial support including social security benefits

| AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | …FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Less than 2 years old | 391 | %19 | 45 | %2 | 334 | %16 | 464 | %23 | 802 | %39 | 2.036 | %100 |
| 2 to less that 10 years old | 144 | %16 | 30 | %3 | 125 | %14 | 238 | %26 | 383 | %42 | 920 | %100 |
| 10 to less than 20 years old | 85 | %9 | 21 | %2 | 110 | %12 | 340 | %36 | 394 | %41 | 950 | %100 |
| 20 to less than 30 years old | 90 | %9 | 17 | %2 | 100 | %10 | 354 | %36 | 415 | %43 | 976 | %100 |
| 30 to less than 50 years old | 256 | %11 | 34 | %1 | 226 | %10 | 917 | %39 | 907 | %39 | 2.340 | %100 |
| 50 years old or more | 137 | %12 | 31 | %3 | 76 | %7 | 589 | %53 | 271 | %25 | 1.104 | %100 |
| TOTAL | 1.103 | %13 | 178 | %2 | 971 | %12 | 2.902 | %35 | 3.172 | %38 | 8.326 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 474,3 ; dof= 20.*

**Cross: Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable) / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?**

| AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY... | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Less than 2 years old | 1.135 | %56 | 998 | %49 | 113 | %6 | 41 | %2 | 235 | %11 | 65 | %3 | 84 | %4 | 2.045 | |
| 2 to less that 10 years old | 478 | %52 | 419 | %45 | 45 | %5 | 30 | %3 | 121 | %13 | 45 | %5 | 50 | %5 | 925 | |
| 10 to less than 20 years old | 477 | %50 | 515 | %54 | 52 | %5 | 11 | %1 | 104 | %11 | 49 | %5 | 48 | %5 | 952 | |
| 20 to less than 30 years old | 471 | %48 | 505 | %52 | 36 | %4 | 7 | %1 | 116 | %12 | 66 | %7 | 48 | %5 | 978 | |
| 30 to less than 50 years old | 1.228 | %52 | 1.167 | %50 | 76 | %3 | 30 | %1 | 270 | %11 | 101 | %4 | 115 | %5 | 2.353 | |
| 50 years old or more | 547 | %49 | 487 | %44 | 31 | %3 | 15 | %1 | 149 | %13 | 66 | %6 | 68 | %6 | 1.107 | |
| **TOTAL** | **4.336** | **%52** | **4.091** | **%49** | **353** | **%4** | **134** | **%2** | **995** | **%12** | **392** | **%5** | **413** | **%5** | **8.360** | |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 109,4 ; dof= 30.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable) / Access to the most adapted care, treatments or surgery…**

| AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | ACCESS TO THE MOST ADAPTED CARE, TREATMENTS OR SURGERY… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Less than 2 years old | <u>134</u> | <u>%7</u> | 908 | %47 | 717 | %37 | 96 | %5 | 72 | %4 | 1.927 | %100 |
| 2 to less that 10 years old | 91 | %11 | <u>432</u> | <u>%50</u> | <u>281</u> | <u>%33</u> | 30 | %3 | 28 | %3 | 862 | %100 |
| 10 to less than 20 years old | 97 | %11 | 420 | %48 | 303 | %35 | 32 | %4 | 24 | %3 | 876 | %100 |
| 20 to less than 30 years old | 88 | %10 | 407 | %44 | 353 | %38 | 40 | %4 | 29 | %3 | 917 | %100 |
| 30 to less than 50 years old | 223 | %10 | 963 | %45 | 808 | %37 | 99 | %5 | 70 | %3 | 2.163 | %100 |
| 50 years old or more | 115 | %11 | <u>410</u> | <u>%39</u> | 403 | %39 | <u>65</u> | <u>%6</u> | <u>50</u> | <u>%5</u> | 1.043 | %100 |
| **TOTAL** | **748** | **%10** | **3.540** | **%45** | **2.865** | **%37** | **362** | **%5** | **273** | **%4** | **7.788** | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 61,5 ; dof= 20.*

**Cross: Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable) / Understanding how the disease will progress…**

| AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | UNDERSTANDING HOW THE DISEASE WILL PROGRESS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Less than 2 years old | <u>114</u> | <u>%6</u> | <u>1.156</u> | <u>%60</u> | <u>506</u> | <u>%26</u> | 97 | %5 | <u>54</u> | <u>%3</u> | 1.927 | %100 |
| 2 to less that 10 years old | 64 | %7 | <u>523</u> | <u>%61</u> | <u>216</u> | <u>%25</u> | 46 | %5 | 13 | %2 | 862 | %100 |
| 10 to less than 20 years old | 78 | %9 | 516 | %59 | 237 | %27 | <u>31</u> | <u>%4</u> | 14 | %2 | 876 | %100 |
| 20 to less than 30 years old | 72 | %8 | 510 | %56 | 290 | %32 | 39 | %4 | <u>6</u> | <u>%1</u> | 917 | %100 |
| 30 to less than 50 years old | 172 | %8 | <u>1.161</u> | <u>%54</u> | <u>676</u> | <u>%31</u> | <u>129</u> | <u>%6</u> | <u>25</u> | <u>%1</u> | 2.163 | %100 |
| 50 years old or more | 84 | %8 | <u>533</u> | <u>%51</u> | <u>353</u> | <u>%34</u> | 55 | %5 | 18 | %2 | 1.043 | %100 |
| **TOTAL** | **584** | **%7** | **4.399** | **%56** | **2.278** | **%29** | **397** | **%5** | **130** | **%2** | **7.788** | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 83,9 ; dof= 20.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

## Cross: Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable) / Financial support including social security benefits…

| AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Less than 2 years old | 58 | %16 | 78 | %21 | 144 | %39 | 30 | %8 | 59 | %16 | 369 | %100 |
| 2 to less that 10 years old | 53 | %16 | 66 | %20 | 144 | %44 | 25 | %8 | 40 | %12 | 328 | %100 |
| 10 to less than 20 years old | 120 | %17 | 145 | %21 | 278 | %40 | 55 | %8 | 100 | %14 | 698 | %100 |
| 20 to less than 30 years old | 126 | %15 | 164 | %19 | 334 | %39 | 72 | %9 | 151 | %18 | 847 | %100 |
| 30 to less than 50 years old | 340 | %17 | 353 | %17 | 803 | %39 | 194 | %9 | 361 | %18 | 2.051 | %100 |
| 50 years old or more | 98 | %10 | 119 | %13 | 365 | %39 | 86 | %9 | 268 | %29 | 936 | %100 |
| TOTAL | 795 | %15 | 925 | %18 | 2.068 | %40 | 462 | %9 | 979 | %19 | 5.229 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 111,9 ; dof= 20.*

## Cross: Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable) / Integration at school…

| AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | INTEGRATION AT SCHOOL… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Less than 2 years old | 184 | %10 | 425 | %22 | 720 | %37 | 128 | %7 | 470 | %24 | 1.927 | %100 |
| 2 to less that 10 years old | 139 | %16 | 192 | %22 | 313 | %36 | 42 | %5 | 176 | %20 | 862 | %100 |
| 10 to less than 20 years old | 123 | %14 | 91 | %10 | 244 | %28 | 53 | %6 | 365 | %42 | 876 | %100 |
| 20 to less than 30 years old | 69 | %8 | 37 | %4 | 158 | %17 | 69 | %8 | 584 | %64 | 917 | %100 |
| 30 to less than 50 years old | 97 | %4 | 56 | %3 | 244 | %11 | 158 | %7 | 1.608 | %74 | 2.163 | %100 |
| 50 years old or more | 34 | %3 | 12 | %1 | 54 | %5 | 47 | %5 | 896 | %86 | 1.043 | %100 |
| TOTAL | 646 | %8 | 813 | %10 | 1.733 | %22 | 497 | %6 | 4.099 | %53 | 7.788 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 2.268,3 ; dof= 20.*

Rare Barometer
A EURORDIS ✖ INITIATIVE

EURORDIS
RARE DISEASES EUROPE

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable) / Integration at work…**

| AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | INTEGRATION AT WORK… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Less than 2 years old | 387 | %20 | 183 | %9 | 671 | %35 | 151 | %8 | 535 | %28 | 1.927 | %100 |
| 2 to less that 10 years old | 192 | %22 | 90 | %10 | 294 | %34 | 61 | %7 | 225 | %26 | 862 | %100 |
| 10 to less than 20 years old | 247 | %28 | 98 | %11 | 287 | %33 | 43 | %5 | 201 | %23 | 876 | %100 |
| 20 to less than 30 years old | 290 | %32 | 112 | %12 | 265 | %29 | 45 | %5 | 205 | %22 | 917 | %100 |
| 30 to less than 50 years old | 765 | %35 | 191 | %9 | 591 | %27 | 86 | %4 | 530 | %25 | 2.163 | %100 |
| 50 years old or more | 236 | %23 | 53 | %5 | 167 | %16 | 36 | %3 | 551 | %53 | 1.043 | %100 |
| TOTAL | 2.117 | %27 | 727 | %9 | 2.275 | %29 | 422 | %5 | 2.247 | %29 | 7.788 | |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 532,3 ; dof= 20.*

**Cross: Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable) / Access to social services (e.g. social worker support, household chores support)…**

| AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | ACCESS TO SOCIAL SERVICES (E.G. SOCIAL WORKER SUPPORT, HOUSEHOLD CHORES SUPPORT)… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Less than 2 years old | 230 | %12 | 345 | %18 | 726 | %38 | 221 | %11 | 405 | %21 | 1.927 | %100 |
| 2 to less that 10 years old | 100 | %12 | 137 | %16 | 345 | %40 | 86 | %10 | 194 | %23 | 862 | %100 |
| 10 to less than 20 years old | 114 | %13 | 89 | %10 | 301 | %34 | 105 | %12 | 267 | %30 | 876 | %100 |
| 20 to less than 30 years old | 121 | %13 | 76 | %8 | 279 | %30 | 106 | %12 | 335 | %37 | 917 | %100 |
| 30 to less than 50 years old | 280 | %13 | 183 | %8 | 648 | %30 | 265 | %12 | 787 | %36 | 2.163 | %100 |
| 50 years old or more | 111 | %11 | 71 | %7 | 241 | %23 | 99 | %9 | 521 | %50 | 1.043 | %100 |
| TOTAL | 956 | %12 | 901 | %12 | 2.540 | %33 | 882 | %11 | 2.509 | %32 | 7.788 | |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 425,7 ; dof= 20.*

## Cross: Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable) / Access to clinical trials…

| AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | ACCESS TO CLINICAL TRIALS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Less than 2 years old | 104 | %5 | 548 | %28 | 698 | %36 | 307 | %16 | 270 | %14 | 1.927 | %100 |
| 2 to less that 10 years old | 59 | %7 | 259 | %30 | 329 | %38 | 117 | %14 | 98 | %11 | 862 | %100 |
| 10 to less than 20 years old | 59 | %7 | 234 | %27 | 318 | %36 | 127 | %14 | 138 | %16 | 876 | %100 |
| 20 to less than 30 years old | 68 | %7 | 234 | %26 | 292 | %32 | 174 | %19 | 149 | %16 | 917 | %100 |
| 30 to less than 50 years old | 175 | %8 | 462 | %21 | 779 | %36 | 394 | %18 | 353 | %16 | 2.163 | %100 |
| 50 years old or more | 70 | %7 | 188 | %18 | 365 | %35 | 198 | %19 | 222 | %21 | 1.043 | %100 |
| TOTAL | 535 | %7 | 1.925 | %25 | 2.781 | %36 | 1.317 | %17 | 1.230 | %16 | 7.788 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 120,0 ; dof= 20.*

## Cross: Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable) / Access to financial products, such as loans, mortgages, insurance…

| AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | ACCESS TO FINANCIAL PRODUCTS, SUCH AS LOANS, MORTGAGES, INSURANCE… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Less than 2 years old | 264 | %14 | 86 | %4 | 601 | %31 | 384 | %20 | 592 | %31 | 1.927 | %100 |
| 2 to less that 10 years old | 168 | %19 | 31 | %4 | 250 | %29 | 173 | %20 | 240 | %28 | 862 | %100 |
| 10 to less than 20 years old | 212 | %24 | 14 | %2 | 253 | %29 | 152 | %17 | 245 | %28 | 876 | %100 |
| 20 to less than 30 years old | 244 | %27 | 9 | %1 | 231 | %25 | 155 | %17 | 278 | %30 | 917 | %100 |
| 30 to less than 50 years old | 490 | %23 | 13 | %1 | 566 | %26 | 369 | %17 | 725 | %34 | 2.163 | %100 |
| 50 years old or more | 114 | %11 | 21 | %2 | 161 | %15 | 157 | %15 | 590 | %57 | 1.043 | %100 |
| TOTAL | 1.492 | %19 | 174 | %2 | 2.062 | %26 | 1.390 | %18 | 2.670 | %34 | 7.788 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 468,1 ; dof= 20.*

**Cross: Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable) / Your social life…**

| AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | YOUR SOCIAL LIFE… | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Less than 2 years old | 791 | %41 | 175 | %9 | 765 | %40 | 57 | %3 | 139 | %7 | 1.927 | %100 |
| 2 to less that 10 years old | 384 | %45 | 78 | %9 | 334 | %39 | 14 | %2 | 52 | %6 | 862 | %100 |
| 10 to less than 20 years old | 413 | %47 | 93 | %11 | 325 | %37 | 14 | %2 | 31 | %4 | 876 | %100 |
| 20 to less than 30 years old | 482 | %53 | 91 | %10 | 300 | %33 | 9 | %1 | 35 | %4 | 917 | %100 |
| 30 to less than 50 years old | 1.289 | %60 | 128 | %6 | 627 | %29 | 37 | %2 | 82 | %4 | 2.163 | %100 |
| 50 years old or more | 621 | %60 | 48 | %5 | 306 | %29 | 11 | %1 | 57 | %5 | 1.043 | %100 |
| **TOTAL** | **3.980** | **%51** | **613** | **%8** | **2.657** | **%34** | **142** | **%2** | **396** | **%5** | **7.788** | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 240,3 ; dof= 20.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

# Chapter 4.

## Family members were already diagnosed with the same rare disease

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Questions asked only to respondents who are diagnosed

| Family members were previously diagnosed with the same disease | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| Yes | **1,9** | 785 | **5,7** | 757 | **5,9** | 527 | **1,3** | 834 | **7,1** | 707 |
| No | 0,3 | 6.552 | 3,2 | 6.175 | 3,4 | 3.543 | 3,8 | 6.840 | 4,4 | 5.797 |

🟪 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Fisher= 40,1.*

*Inter variance= 1.832,5. Intra variance= 45,7.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Questions asked only to respondents who are diagnosed**

### Cross: Family members were previously diagnosed with the same disease / Are you a patient representative, i.e. involved in policy activities to support the cause of rare diseases?

| FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | ARE YOU A PATIENT REPRESENTATIVE, I.E. INVOLVED IN POLICY ACTIVITIES TO SUPPORT THE CAUSE OF RARE DISEASES? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 257 | %20 | 964 | %74 | 88 | %7 | 1.309 | %100 |
| No | 1.701 | %20 | 6.137 | %73 | 584 | %7 | 8.422 | %100 |
| TOTAL | 1.958 | %20 | 7.101 | %73 | 672 | %7 | 9.731 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is not significant. p-value= 0,8 ; Chi2= 0,3 ; dof= 2.*

### Cross: Family members were previously diagnosed with the same disease / Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable)

| FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | LESS THAN 2 YEARS OLD | | 2 TO LESS THAT 10 YEARS OLD | | 10 TO LESS THAN 20 YEARS OLD | | 20 TO LESS THAN 30 YEARS OLD | | 30 TO LESS THAN 50 YEARS OLD | | 50 YEARS OLD OR MORE | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 134 | %14 | 89 | %9 | 144 | %15 | 164 | %17 | 297 | %31 | 119 | %13 | 947 | %100 |
| No | 1.795 | %26 | 775 | %11 | 737 | %11 | 758 | %11 | 1.873 | %27 | 926 | %13 | 6.864 | %100 |
| TOTAL | 1.929 | %25 | 864 | %11 | 881 | %11 | 922 | %12 | 2.170 | %28 | 1.045 | %13 | 7.811 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 99,2 ; dof= 5.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

**Questions asked only to respondents who are diagnosed**

## Cross: Gender of the person affected by the rare disease / Family members were previously diagnosed with the same disease

| GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE | FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | | | | | |
|---|---|---|---|---|---|---|
| | YES | | NO | | TOTAL | |
| | N | % | N | % | N | % |
| Female | 848 | %14 | 5.320 | %86 | 6.168 | %100 |
| Male | 348 | %13 | 2.305 | %87 | 2.653 | %100 |
| Other | 6 | %7 | 76 | %93 | 82 | %100 |
| TOTAL | 1.202 | %14 | 7.701 | %86 | 8.903 | %100 |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is not significant. p-value= 0,2 ; Chi2= 3,3 ; dof= 2.*

## Cross: How old were you when you stopped full-time education? / Family members were previously diagnosed with the same disease

| HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION? | FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | | | | | |
|---|---|---|---|---|---|---|
| | YES | | NO | | TOTAL | |
| | N | % | N | % | N | % |
| 15 y.o. or under | 55 | %13 | 358 | %87 | 413 | %100 |
| between 16 and 19 y.o. | 360 | %16 | 1.931 | %84 | 2.291 | %100 |
| between 20 and 23 y.o. | 371 | %13 | 2.460 | %87 | 2.831 | %100 |
| 24 y.o. or above | 359 | %12 | 2.558 | %88 | 2.917 | %100 |
| TOTAL | 1.145 | %14 | 7.307 | %86 | 8.452 | %100 |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 13,5 ; dof= 3.*

**Questions asked only to respondents who are diagnosed**

### Cross: How would you best describe yourself? / Family members were previously diagnosed with the same disease

| | FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | | | | | |
| --- | --- | --- | --- | --- | --- | --- |
| | YES | | NO | | TOTAL | |
| HOW WOULD YOU BEST DESCRIBE YOURSELF? | N | % | N | % | N | % |
| I belong to the ethnic majority in the country where I live | 866 | %13 | 5.779 | %87 | 6.645 | %100 |
| I am part of an ethnic minority in the country where I live | 78 | %19 | 343 | %81 | 421 | %100 |
| Other, specify... | 37 | %13 | 259 | %88 | 296 | %100 |
| TOTAL | 981 | %13 | 6.381 | %87 | 7.362 | |

🟪 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 10,5 ; dof= 2.*

### Cross: Typology of countries based on size and welfare / Family members were previously diagnosed with the same disease

| | FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | | | | | |
| --- | --- | --- | --- | --- | --- | --- |
| | YES | | NO | | TOTAL | |
| TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE | N | % | N | % | N | % |
| Group A ('Eastern Europe') | 140 | %9 | 1.472 | %91 | 1.612 | %100 |
| Group B ('Western Europe') | 660 | %14 | 4.157 | %86 | 4.817 | %100 |
| Group C ('Northern Europe') | 456 | %15 | 2.556 | %85 | 3.012 | %100 |
| TOTAL | 1.256 | %13 | 8.185 | %87 | 9.441 | %100 |

🟪 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 39,3 ; dof= 2.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Questions asked only to respondents who are diagnosed**

## Cross: Would you say that you, or the person you care for, live in a: / Family members were previously diagnosed with the same disease

| WOULD YOU SAY THAT YOU, OR THE PERSON YOU CARE FOR, LIVE IN A: | FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | | | | | |
|---|---|---|---|---|---|---|
| | YES | | NO | | TOTAL | |
| | N | % | N | % | N | % |
| Rural area or village | 314 | %14 | 1.931 | %86 | 2.245 | %100 |
| Small or mid size town | 503 | %14 | 3.135 | %86 | 3.638 | %100 |
| Large town | 328 | %13 | 2.233 | %87 | 2.561 | %100 |
| **TOTAL** | **1.145** | **%14** | **7.299** | **%86** | **8.444** | **%100** |

🟥 *Under-represented elements* 🟦 *Over-represented elements*

*The relationship is not significant. p-value= 0,4 ; Chi2= 1,8 ; dof= 2.*

## Cross: Point prevalence of the rare disease / Family members were previously diagnosed with the same disease

| POINT PREVALENCE OF THE RARE DISEASE | FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | | | | | |
|---|---|---|---|---|---|---|
| | YES | | NO | | TOTAL | |
| | N | % | N | % | N | % |
| 1-5 / 10 000 | 494 | %21 | 1.887 | %79 | 2.381 | %100 |
| 1-9 / 100 000 | 222 | %11 | 1.744 | %89 | 1.966 | %100 |
| 1-9 / 1 000 000 | 57 | %13 | 397 | %87 | 454 | %100 |
| <1 / 1 000 000 | 95 | %12 | 727 | %88 | 822 | %100 |
| **TOTAL** | **868** | **%15** | **4.755** | **%85** | **5.623** | |

🟥 *Under-represented elements* 🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 89,7 ; dof= 3.*

**Questions asked only to respondents who are diagnosed**

**Cross: Orphacode associated nomenclature (english) / Family members were previously diagnosed with the same disease**

| ORPHACODE ASSOCIATED NOMENCLATURE (ENGLISH) | FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | | | | | |
|---|---|---|---|---|---|---|
| | YES | | NO | | TOTAL | |
| | N | % | N | % | N | % |
| Hereditary hemorrhagic telangiectasia | 294 | %65 | 160 | %35 | 454 | %100 |
| Hypermobile Ehlers-Danlos syndrome | 43 | %14 | 268 | %86 | 311 | %100 |
| Sarcoidosis | 7 | %4 | 163 | %96 | 170 | %100 |
| Classical Ehlers-Danlos syndrome | 13 | %10 | 122 | %90 | 135 | %100 |
| Williams syndrome | 0 | %0 | 134 | %100 | 134 | %100 |
| Cystic fibrosis | 14 | %11 | 113 | %89 | 127 | %100 |
| Myasthenia gravis | 3 | %3 | 115 | %97 | 118 | %100 |
| Systemic sclerosis | 5 | %5 | 100 | %95 | 105 | %100 |
| Tuberous sclerosis complex | 8 | %8 | 90 | %92 | 98 | %100 |
| Neurofibromatosis type 1 | 19 | %21 | 73 | %79 | 92 | %100 |
| Interstitial cystitis | 2 | %3 | 72 | %97 | 74 | %100 |
| Addison disease | 5 | %7 | 68 | %93 | 73 | %100 |
| 22q11.2 deletion syndrome | 3 | %4 | 65 | %96 | 68 | %100 |
| Chronic inflammatory demyelinating polyneuropathy | 1 | %2 | 62 | %98 | 63 | %100 |
| Perineural cyst | 3 | %5 | 58 | %95 | 61 | %100 |
| Acute inflammatory demyelinating polyradiculoneuropathy | 1 | %2 | 61 | %98 | 62 | %100 |
| Rett syndrome | 1 | %2 | 59 | %98 | 60 | %100 |

🟪 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 3.382,7 ; dof= 1.629.*

Rare Barometer
A EURORDIS INITIATIVE

EURORDIS
RARE DISEASES EUROPE

**Questions asked only to respondents who are diagnosed**

## Cross: Orphanet classification of rare diseases (one disease can be classified in several categories) / Family members were previously diagnosed with the same disease

| ORPHANET CLASSIFICATION OF RARE DISEASES (ONE DISEASE CAN BE CLASSIFIED IN SEVERAL CATEGORIES) | FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | | | | | |
|---|---|---|---|---|---|---|
| | YES | | NO | | TOTAL | |
| | N | % | N | % | N | % |
| Abdominal surgical diseases | 14 | %6 | 221 | %94 | 235 | %100 |
| Allergic diseases | 0 | %0 | 3 | %100 | 3 | %100 |
| Bone diseases | 88 | %11 | 695 | %89 | 783 | %100 |
| Cardiac diseases | 77 | %12 | 577 | %88 | 654 | %100 |
| Cardiac malformations | 6 | %2 | 285 | %98 | 291 | %100 |
| Circulatory system diseases | 389 | %29 | 941 | %71 | 1.330 | %100 |
| Clinical sign | 0 | %0 | 0 | %0 | 0 | %100 |
| Developmental anomalies during embryogenesis | 600 | %18 | 2.678 | %82 | 3.278 | %100 |
| Diseases due to toxic effects | 0 | %0 | 3 | %100 | 3 | %100 |
| Endocrine diseases | 69 | %7 | 913 | %93 | 982 | %100 |
| Gastroenterological diseases | 37 | %12 | 262 | %88 | 299 | %100 |
| Genetic diseases | 952 | %18 | 4.362 | %82 | 5.314 | %100 |
| Gynecologic/obstetric diseases | 24 | %9 | 257 | %91 | 281 | %100 |
| Hematological diseases | 41 | %10 | 354 | %90 | 395 | %100 |
| Hepatic diseases | 345 | %39 | 540 | %61 | 885 | %100 |
| Immunological diseases | 25 | %9 | 249 | %91 | 274 | %100 |
| Inborn errors of metabolism | 90 | %12 | 667 | %88 | 757 | %100 |

🟪 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 2.168,5 ; dof= 34.*

**Questions asked only to respondents who are diagnosed**

**Cross: Family members were previously diagnosed with the same disease / How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?**

| FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | 0 | | 1 | | BETWEEN 2 AND 4 | | BETWEEN 5 AND 7 | | BETWEEN 8 AND 10 | | MORE THAN 10 | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 34 | %3 | 275 | %21 | 604 | %46 | 184 | %14 | 63 | %5 | 149 | %11 | 1.309 | %100 |
| No | 112 | %1 | 815 | %10 | 3.730 | %44 | 1.701 | %20 | 665 | %8 | 1.399 | %17 | 8.422 | %100 |
| TOTAL | 146 | %2 | 1.090 | %11 | 4.334 | %45 | 1.885 | %19 | 728 | %7 | 1.548 | %16 | 9.731 | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 198,8 ; dof= 5.*

**Cross: Family members were previously diagnosed with the same disease / I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases**

| FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | | | | | |
|---|---|---|---|---|---|---|
| | YES | | NO | | TOTAL | |
| | N | % | N | % | N | % |
| Yes | 858 | %66 | 442 | %34 | 1.300 | %100 |
| No | 4.717 | %56 | 3.647 | %44 | 8.364 | %100 |
| TOTAL | 5.575 | %58 | 4.089 | %42 | 9.664 | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 42,5 ; dof= 1.*

**Questions asked only to respondents who are diagnosed**

### Cross: Family members were previously diagnosed with the same disease / Genetic test(s) looking for genetic changes (also called mutations or variants)

| FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | GENETIC TEST(S) LOOKING FOR GENETIC CHANGES (ALSO CALLED MUTATIONS OR VARIANTS) | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW/DON'T REMEMBER | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 906 | %69 | 332 | %25 | 71 | %5 | 1.309 | %100 |
| No | 4.206 | %50 | 3.544 | %42 | 672 | %8 | 8.422 | %100 |
| TOTAL | 5.112 | %53 | 3.876 | %40 | 743 | %8 | 9.731 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 169,3 ; dof= 2.*

### Cross: Family members were previously diagnosed with the same disease / Other test(s) such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc

| FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | OTHER TEST(S) SUCH AS CLINICAL EXAMINATION(S), MEDICAL IMAGING (MRI, SCANS…), BIOPSY, BIOCHEMICAL TEST(S) (BLOOD OR URINE TESTS…), ETC | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW/DON'T REMEMBER | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 1.123 | %86 | 143 | %11 | 43 | %3 | 1.309 | %100 |
| No | 7.691 | %91 | 530 | %6 | 201 | %2 | 8.422 | %100 |
| TOTAL | 8.814 | %91 | 673 | %7 | 244 | %3 | 9.731 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 42,6 ; dof= 2.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

Questions asked only to respondents who are diagnosed

## Cross: Family members were previously diagnosed with the same disease / …you could not afford it?

Have you ever needed a genetic test but could not access it because…

…YOU COULD NOT AFFORD IT?

| FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | YES | | NO | | NOT RELEVANT | | TOTAL | |
|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % |
| Yes | 119 | %9 | 1.000 | %76 | 190 | %15 | 1.309 | %100 |
| No | 826 | %10 | 5.693 | %68 | 1.903 | %23 | 8.422 | %100 |
| TOTAL | 945 | %10 | 6.693 | %69 | 2.093 | %22 | 9.731 | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 47,7 ; dof= 2.*

## Cross: Family members were previously diagnosed with the same disease / …it was not available in your country?

Have you ever needed a genetic test but could not access it because…

…IT WAS NOT AVAILABLE IN YOUR COUNTRY?

| FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | YES | | NO | | NOT RELEVANT | | TOTAL | |
|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % |
| Yes | 131 | %10 | 956 | %73 | 222 | %17 | 1.309 | %100 |
| No | 924 | %11 | 5.448 | %65 | 2.050 | %24 | 8.422 | %100 |
| TOTAL | 1.055 | %11 | 6.404 | %66 | 2.272 | %23 | 9.731 | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 39,4 ; dof= 2.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

Questions asked only to respondents who are diagnosed

### Cross: Family members were previously diagnosed with the same disease / …healthcare professionals were reluctant or not sufficiently informed?

| FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | …HEALTHCARE PROFESSIONALS WERE RELUCTANT OR NOT SUFFICIENTLY INFORMED? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 337 | %26 | 795 | %61 | 177 | %14 | 1.309 | %100 |
| No | 2.121 | %25 | 4.485 | %53 | 1.816 | %22 | 8.422 | %100 |
| TOTAL | 2.458 | %25 | 5.280 | %54 | 1.993 | %20 | 9.731 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 47,6 ; dof= 2.*

### Cross: Family members were previously diagnosed with the same disease / To your knowledge, the genetic test(s) that were conducted targeted…

| FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | TO YOUR KNOWLEDGE, THE GENETIC TEST(S) THAT WERE CONDUCTED TARGETED… | | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | ONLY ONE GENE | | SEVERAL GENES AT THE SAME TIME (GENE PANEL SEQUENCING) | | THE WHOLE DNA (WHOLE GENOME SEQUENCING) | | ALL THE GENES (WHOLE EXOME SEQUENCING) | | A TUMOUR (GENETIC PROFILING OF A TUMOUR) | | OTHER (EPIGENOME, RNA, ETC.) | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 311 | %34 | 242 | %27 | 106 | %12 | 45 | %5 | 15 | %2 | 14 | %2 | 260 | %29 | 906 | |
| No | 1.079 | %26 | 1.354 | %32 | 685 | %16 | 441 | %10 | 107 | %3 | 92 | %2 | 1.148 | %27 | 4.206 | |
| TOTAL | 1.390 | %27 | 1.596 | %31 | 791 | %15 | 486 | %10 | 122 | %2 | 106 | %2 | 1.408 | %28 | 5.112 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 66,3 ; dof= 6.*

**Questions asked only to respondents who are diagnosed**

### Cross: Family members were previously diagnosed with the same disease / Did you ever request a private company or laboratory to conduct genetic testing to diagnose the disease?

| FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | DID YOU EVER REQUEST A PRIVATE COMPANY OR LABORATORY TO CONDUCT GENETIC TESTING TO DIAGNOSE THE DISEASE? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO, NEVER | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 67 | %7 | 25 | %3 | 814 | %90 | 906 | %100 |
| No | 471 | %11 | 213 | %5 | 3.522 | %84 | 4.206 | %100 |
| TOTAL | 538 | %11 | 238 | %5 | 4.336 | %85 | 5.112 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 22,0 ; dof= 2.*

### Cross: Family members were previously diagnosed with the same disease / In general, how satisfied are you with how the results of the GENETIC TESTS were given to you?

| FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | IN GENERAL, HOW SATISFIED ARE YOU WITH HOW THE RESULTS OF THE GENETIC TESTS WERE GIVEN TO YOU? | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | VERY DISSATISFIED | | DISSATISFIED | | NEITHER SATISFIED NOR DISSATISFIED | | SATISFIED | | VERY SATISFIED | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 86 | %9 | 68 | %8 | 174 | %19 | 332 | %37 | 203 | %22 | 43 | %5 | 906 | %100 |
| No | 439 | %10 | 481 | %11 | 879 | %21 | 1.518 | %36 | 699 | %17 | 190 | %5 | 4.206 | %100 |
| TOTAL | 525 | %10 | 549 | %11 | 1.053 | %21 | 1.850 | %36 | 902 | %18 | 233 | %5 | 5.112 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 26,7 ; dof= 5.*

Questions asked only to respondents who are diagnosed

**Cross: Family members were previously diagnosed with the same disease / After the tests were performed, were you offered genetic counselling (e.g. given information about how your genetic condition might affect you and your family)?**

| FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | AFTER THE TESTS WERE PERFORMED, WERE YOU OFFERED GENETIC COUNSELLING (E.G. GIVEN INFORMATION ABOUT HOW YOUR GENETIC CONDITION MIGHT AFFECT YOU AND YOUR FAMILY)? | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| | YES, WITH A GENETIC COUNSELLOR OR CLINICAL GENETICIST | | YES, BY A HEALTHCARE PROFESSIONAL | | NO, I WASN'T OFFERED GENETIC COUNSELLING | | NOT SURE / DON'T REMEMBER | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % |
| Yes | 403 | %44 | 218 | %24 | 222 | %25 | 63 | %7 | 906 | %100 |
| No | 1.626 | %39 | 922 | %22 | 1.363 | %32 | 295 | %7 | 4.206 | %100 |
| **TOTAL** | **2.029** | **%40** | **1.140** | **%22** | **1.585** | **%31** | **358** | **%7** | **5.112** | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 22,9 ; dof= 3.*

**Cross: Family members were previously diagnosed with the same disease / Genetic tests**

| FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | GENETIC TESTS | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 90 | %10 | 792 | %87 | 24 | %3 | 906 | %100 |
| No | 668 | %16 | 3.430 | %82 | 107 | %3 | 4.205 | %100 |
| **TOTAL** | **758** | **%15** | **4.222** | **%83** | **131** | **%3** | **5.111** | **%100** |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 20,9 ; dof= 2.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Questions asked only to respondents who are diagnosed**

### Cross: Family members were previously diagnosed with the same disease / Other diagnostic tests such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc.

| FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | OTHER DIAGNOSTIC TESTS SUCH AS CLINICAL EXAMINATION(S), MEDICAL IMAGING (MRI, SCANS…), BIOPSY, BIOCHEMICAL TEST(S) (BLOOD OR URINE TESTS…), ETC. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 147 | %13 | 955 | %85 | 21 | %2 | **1.123** | **%100** |
| No | 1.122 | %15 | 6.437 | %84 | 131 | %2 | **7.690** | **%100** |
| **TOTAL** | **1.269** | **%14** | **7.392** | **%84** | **152** | **%2** | **8.813** | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is not significant. p-value= 0,4 ; Chi2= 1,9 ; dof= 2.*

### Cross: Family members were previously diagnosed with the same disease / Additional advice from a healthcare professional specialised in the rare disease (in person or virtually)

| FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | ADDITIONAL ADVICE FROM A HEALTHCARE PROFESSIONAL SPECIALISED IN THE RARE DISEASE (IN PERSON OR VIRTUALLY) | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 219 | %17 | 1.057 | %81 | 33 | %3 | 1.309 | %100 |
| No | 1.700 | %20 | 6.575 | %78 | 147 | %2 | 8.422 | %100 |
| **TOTAL** | **1.919** | **%20** | **7.632** | **%78** | **180** | **%2** | **9.731** | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 11,6 ; dof= 2.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

**Questions asked only to respondents who are diagnosed**

## Cross: Family members were previously diagnosed with the same disease / …psychological support

| FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | …PSYCHOLOGICAL SUPPORT | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 97 | %7 | 116 | %9 | 94 | %7 | 557 | %43 | 445 | %34 | 1.309 | %100 |
| No | 746 | %9 | 778 | %9 | 758 | %9 | 2.439 | %29 | 3.701 | %44 | 8.422 | %100 |
| TOTAL | 843 | %9 | 894 | %9 | 852 | %9 | 2.996 | %31 | 4.146 | %43 | 9.731 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 101,5 ; dof= 4.*

## Cross: Family members were previously diagnosed with the same disease / …care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc.

| FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | …CARE COORDINATION SUPPORT SUCH AS HELP TO FIND THE NECESSARY INFORMATION ON THE DISEASE AND THE RIGHT PROFESSIONALS, ARRANGING APPOINTMENTS WITH DIFFERENT HEALTH PROVIDERS, ETC. | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 306 | %23 | 56 | %4 | 150 | %11 | 285 | %22 | 512 | %39 | 1.309 | %100 |
| No | 1.680 | %20 | 309 | %4 | 1.196 | %14 | 1.246 | %15 | 3.991 | %47 | 8.422 | %100 |
| TOTAL | 1.986 | %20 | 365 | %4 | 1.346 | %14 | 1.531 | %16 | 4.503 | %46 | 9.731 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 65,6 ; dof= 4.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

**Questions asked only to respondents who are diagnosed**

## Cross: Family members were previously diagnosed with the same disease / …financial support including social security benefits

| FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | …FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS | | | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 155 | %12 | 37 | %3 | 104 | %8 | 547 | %42 | 457 | %35 | 1.300 | %100 |
| No | 1.162 | %14 | 186 | %2 | 991 | %12 | 2.824 | %34 | 3.201 | %38 | 8.364 | %100 |
| TOTAL | 1.317 | %14 | 223 | %2 | 1.095 | %11 | 3.371 | %35 | 3.658 | %38 | 9.664 | |

🟪 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 45,0 ; dof= 4.*

## Cross: Family members were previously diagnosed with the same disease / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?

| FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | | | | | | | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY... | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 751 | %57 | 576 | %44 | 64 | %5 | 14 | %1 | 112 | %9 | 71 | %5 | 117 | %9 | 1.309 | |
| No | 4.333 | %51 | 4.142 | %49 | 348 | %4 | 143 | %2 | 963 | %11 | 429 | %5 | 354 | %4 | 8.422 | |
| TOTAL | 5.084 | %52 | 4.718 | %48 | 412 | %4 | 157 | %2 | 1.075 | %11 | 500 | %5 | 471 | %5 | 9.731 | |

🟪 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 79,4 ; dof= 6.*

Rare Barometer
A EURORDIS INITIATIVE

EURORDIS
RARE DISEASES EUROPE

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

# Chapter 4.

**Family members were already diagnosed with the**

## Which parts of the body does the rare disease impact?

| | N |
|---|---|
| Lungs | 2.386 |
| Kidneys | 1.365 |
| Liver | 1.405 |
| Heart | 2.198 |
| Skin | 2.929 |
| Bones | 2.016 |
| Muscles, joints (musculoskeletal diseases) | 4.164 |
| Eyes | 2.777 |
| Craniofacial and ENT diseases | 1.371 |
| Gastrointestinal tract (from the mouth to the anus) | 2.624 |
| Vascular system | 1.957 |
| Blood (haematological diseases) | 1.232 |
| Reproductive and/or urinary systems | 1.324 |
| Immune system (immunodeficiency, autoinflammatory and autoimmune diseases) | 2.892 |
| Nervous system (brain, spine, nerves) | 3.254 |
| Nerves and muscles (neuromuscular diseases) | 2.795 |
| Hormonal functions (endocrine diseases) | 1.393 |
| Metabolism | 1.301 |
| Don't know | 208 |
| Other, specify... | 529 |
| **TOTAL** | **10.486** |

## Which parts of the body does the rare disease impact?
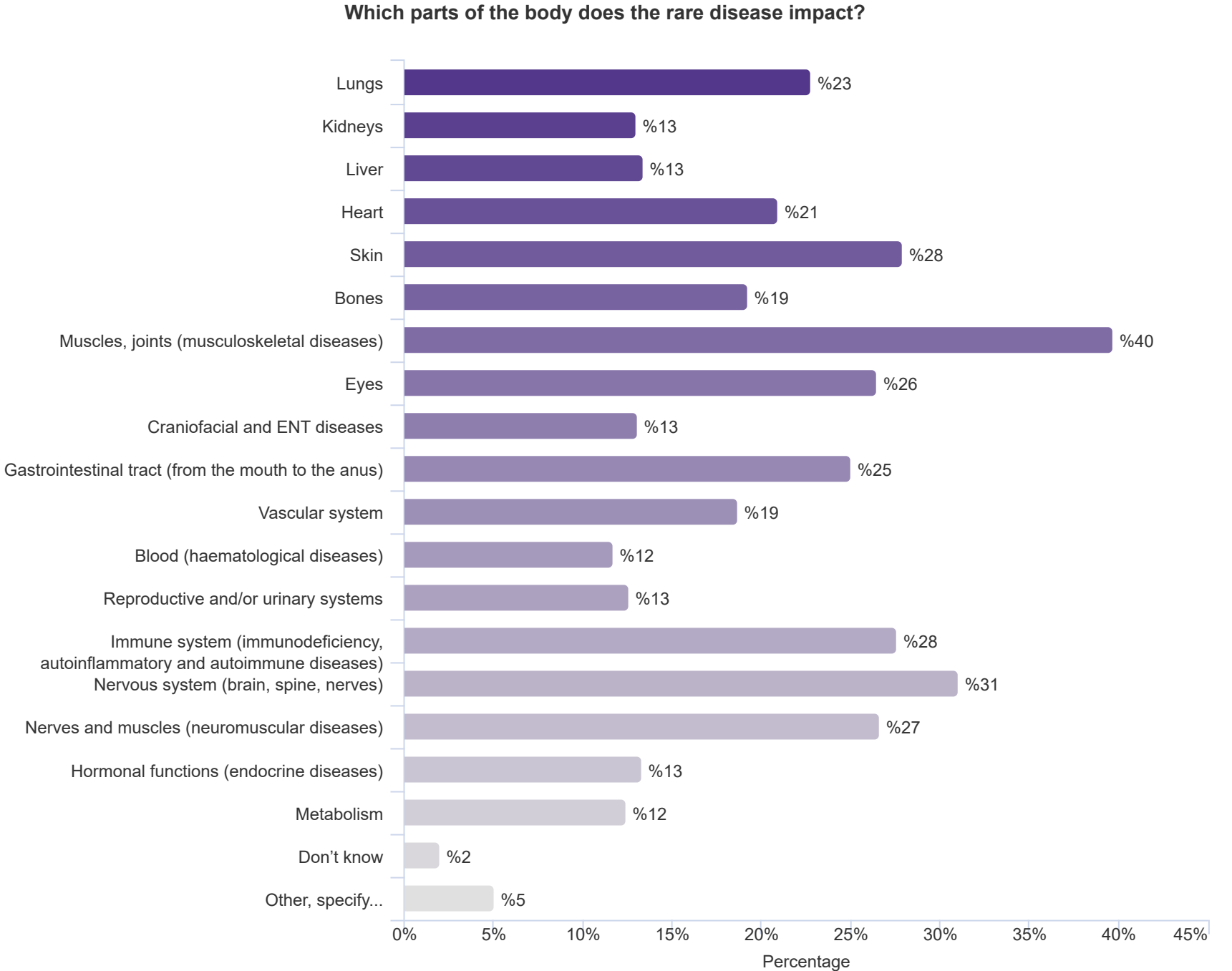
**Complexity of the rare disease:** number of body parts impacted by the rare disease

**Disease complexity classified into five groups, based on the number of affected body parts.**

|  | N |
|---|---|
| 1-3 body parts | 6.103 |
| 4-7 body parts | 3.081 |
| 8-11 body parts | 951 |
| 12-15 body parts | 286 |
| 16 body parts or more | 65 |
| **TOTAL** | **10.486** |

**Disease complexity classified into five groups, based on the number of affected body parts.**



| | |
|---|---|
| 1-3 body parts | %58 |
| 4-7 body parts | %29 |
| 8-11 body parts | %9 |
| 12-15 body parts | %3 |
| 16 body parts or more | %1 |

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*
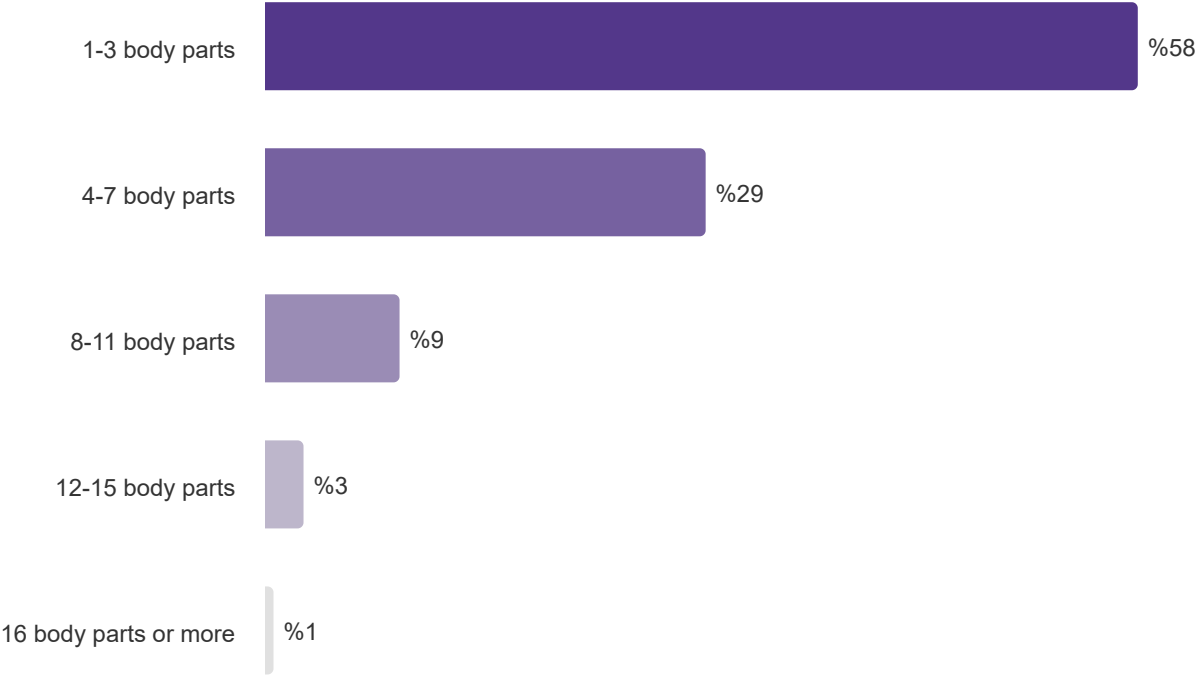
| Disease complexity classified into five groups, based on the number of affected body parts. | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| 1-3 body parts | 0,5 | 4.500 | 3,0 | 4.202 | 2,9 | 2.526 | 2,7 | 4.536 | 3,9 | 3.796 |
| 4-7 body parts | 0,6 | 2.312 | 3,7 | 2.227 | 4,4 | 1.286 | 3,9 | 2.321 | 5,1 | 1.937 |
| 8-11 body parts | 0,1 | 729 | 5,0 | 649 | 6,3 | 383 | 5,9 | 722 | 7,2 | 563 |
| 12-15 body parts | 0,0 | 228 | 5,3 | 201 | 8,1 | 109 | 8,3 | 217 | 9,2 | 172 |
| 16 body parts or more | 3,5 | 51 | 10,3 | 43 | 14,0 | 31 | 12,2 | 47 | 12,2 | 39 |

■ *Under-represented elements*　　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Fisher= 3,6.*

*Inter variance= 167,4. Intra variance= 45,9.*

**Mean** = average time, in number of years

**N** = number of respondents for which we have the average time

Rare Barometer
A EURORDIS INITIATIVE

EURORDIS
RARE DISEASES EUROPE

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Disease complexity classified into five groups, based on the number of affected body parts. / Are you a patient representative, i.e. involved in policy activities to support the cause of rare diseases?

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | ARE YOU A PATIENT REPRESENTATIVE, I.E. INVOLVED IN POLICY ACTIVITIES TO SUPPORT THE CAUSE OF RARE DISEASES? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 1-3 body parts | 1.149 | %19 | 4.525 | %74 | 429 | %7 | 6.103 | %100 |
| 4-7 body parts | 629 | %20 | 2.237 | %73 | 215 | %7 | 3.081 | %100 |
| 8-11 body parts | 207 | %22 | 674 | %71 | 70 | %7 | 951 | %100 |
| 12-15 body parts | 72 | %25 | 188 | %66 | 26 | %9 | 286 | %100 |
| 16 body parts or more | 16 | %25 | 42 | %65 | 7 | %11 | 65 | %100 |
| TOTAL | 2.073 | %20 | 7.666 | %73 | 747 | %7 | 10.486 | |

🔴 *Under-represented elements*　🔵 *Over-represented elements*

*The relationship is significant. p-value= 0,0 ; Chi2= 17,7 ; dof= 8.*

## Cross: Disease complexity classified into five groups, based on the number of affected body parts. / Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable)

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | LESS THAN 2 YEARS OLD | | 2 TO LESS THAT 10 YEARS OLD | | 10 TO LESS THAN 20 YEARS OLD | | 20 TO LESS THAN 30 YEARS OLD | | 30 TO LESS THAN 50 YEARS OLD | | 50 YEARS OLD OR MORE | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| 1-3 body parts | 1.120 | %23 | 527 | %11 | 532 | %11 | 570 | %12 | 1.350 | %28 | 737 | %15 | 4.836 | %100 |
| 4-7 body parts | 677 | %27 | 269 | %11 | 274 | %11 | 269 | %11 | 701 | %28 | 279 | %11 | 2.469 | %100 |
| 8-11 body parts | 181 | %24 | 87 | %11 | 98 | %13 | 105 | %14 | 214 | %28 | 78 | %10 | 763 | %100 |
| 12-15 body parts | 54 | %23 | 28 | %12 | 37 | %16 | 27 | %11 | 76 | %32 | 13 | %6 | 235 | %100 |
| 16 body parts or more | 13 | %23 | 14 | %25 | 11 | %19 | 7 | %12 | 12 | %21 | 0 | %0 | 57 | %100 |
| TOTAL | 2.045 | %24 | 925 | %11 | 952 | %11 | 978 | %12 | 2.353 | %28 | 1.107 | %13 | 8.360 | |

🔴 *Under-represented elements*　🔵 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 83,5 ; dof= 20.*

**Cross: Gender of the person affected by the rare disease / Disease complexity classified into five groups, based on the number of affected body parts.**

| GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE | DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | | | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | 1-3 BODY PARTS | | 4-7 BODY PARTS | | 8-11 BODY PARTS | | 12-15 BODY PARTS | | 16 BODY PARTS OR MORE | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Female | 3.647 | %55 | 2.033 | %31 | 707 | %11 | 216 | %3 | 56 | %1 | 6.659 | %100 |
| Male | 1.837 | %65 | 758 | %27 | 162 | %6 | 48 | %2 | 5 | %0 | 2.810 | %100 |
| Other | 62 | %61 | 26 | %26 | 9 | %9 | 2 | %2 | 2 | %2 | 101 | %100 |
| **TOTAL** | **5.546** | **%58** | **2.817** | **%29** | **878** | **%9** | **266** | **%3** | **63** | **%1** | **9.570** | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 131,1 ; dof= 8.*

**Cross: Typology of countries based on size and welfare / Disease complexity classified into five groups, based on the number of affected body parts.**

| TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE | DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | | | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | 1-3 BODY PARTS | | 4-7 BODY PARTS | | 8-11 BODY PARTS | | 12-15 BODY PARTS | | 16 BODY PARTS OR MORE | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Group A ('Eastern Europe') | 1.005 | %56 | 546 | %30 | 184 | %10 | 52 | %3 | 7 | %0 | 1.794 | %100 |
| Group B ('Western Europe') | 3.138 | %61 | 1.450 | %28 | 381 | %7 | 111 | %2 | 25 | %0 | 5.105 | %100 |
| Group C ('Northern Europe') | 1.764 | %54 | 998 | %30 | 362 | %11 | 116 | %4 | 33 | %1 | 3.273 | %100 |
| **TOTAL** | **5.907** | **%58** | **2.994** | **%29** | **927** | **%9** | **279** | **%3** | **65** | **%1** | **10.172** | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 80,7 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Would you say that you, or the person you care for, live in a: / Disease complexity classified into five groups, based on the number of affected body parts.**

| WOULD YOU SAY THAT YOU, OR THE PERSON YOU CARE FOR, LIVE IN A: | DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | | | | | |
|---|---|---|---|---|---|---|
| | 1-3 BODY PARTS | 4-7 BODY PARTS | 8-11 BODY PARTS | 12-15 BODY PARTS | 16 BODY PARTS OR MORE | TOTAL |
| Rural area or village | %59 | %29 | %9 | %3 | %1 | %100 |
| Small or mid size town | %59 | %29 | %9 | %3 | %1 | %100 |
| Large town | %57 | %30 | %9 | %3 | %1 | %100 |
| TOTAL | %58 | %29 | %9 | %3 | %1 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is not significant. p-value= 0,6 ; Chi2= 6,2 ; dof= 8.*

**Cross: Disease complexity classified into five groups, based on the number of affected body parts. / Please select the sentence that best describes your situation or the situation of the person you care for:**

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | PLEASE SELECT THE SENTENCE THAT BEST DESCRIBES YOUR SITUATION OR THE SITUATION OF THE PERSON YOU CARE FOR: | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|
| | I KNOW THE NAME OF THE RARE DISEASE, SYNDROME OR MALFORMATION AND IT HAS BEEN CONFIRMED BY APPROPRIATE GENETIC, CLINICAL, MEDICAL IMAGING, MOLECULAR OR BIOCHEMICAL TESTS (E.G BIOPSY, BLOOD OR URINE TEST) | | I KNOW THE NAME OF THE RARE DISEASE, SYNDROME OR MALFORMATION BUT IT HAS NOT YET BEEN CONFIRMED BY APPROPRIATE GENETIC, CLINICAL, MEDICAL IMAGING, MOLECULAR OR BIOCHEMICAL TESTS | | I ONLY HAVE PARTIAL INFORMATION ON THE NAME OF THE RARE DISEASE OR THE GENE INVOLVED OR THE TYPE OF DISEASE | | I KNOW THAT THE DISEASE IS RARE BUT THE NAME OR THE CAUSE HAVE NOT BEEN IDENTIFIED | | OTHER, SPECIFY... | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 1-3 body parts | 5.333 | %87 | 388 | %6 | 169 | %3 | 197 | %3 | 16 | %0 | 6.103 | %100 |
| 4-7 body parts | 2.652 | %86 | 225 | %7 | 90 | %3 | 109 | %4 | 5 | %0 | 3.081 | %100 |
| 8-11 body parts | 781 | %82 | 111 | %12 | 27 | %3 | 30 | %3 | 2 | %0 | 951 | %100 |
| 12-15 body parts | 230 | %80 | 29 | %10 | 17 | %6 | 9 | %3 | 1 | %0 | 286 | %100 |
| 16 body parts or more | 52 | %80 | 7 | %11 | 3 | %5 | 3 | %5 | 0 | %0 | 65 | %100 |
| TOTAL | 9.048 | %86 | 760 | %7 | 306 | %3 | 348 | %3 | 24 | %0 | 10.486 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 53,4 ; dof= 16.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Disease complexity classified into five groups, based on the number of affected body parts. / Genetic diseases**

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | GENETIC DISEASES | | | | | |
| --- | --- | --- | --- | --- | --- | --- |
| | GENETIC DISEASES | | NON GENETIC DISEASES | | TOTAL | |
| | N | % | N | % | N | % |
| 1-3 body parts | 2.995 | %64 | 1.665 | %36 | 4.660 | %100 |
| 4-7 body parts | 1.726 | %72 | 670 | %28 | 2.396 | %100 |
| 8-11 body parts | 527 | %70 | 223 | %30 | 750 | %100 |
| 12-15 body parts | 158 | %72 | 60 | %28 | 218 | %100 |
| 16 body parts or more | 41 | %82 | 9 | %18 | 50 | %100 |
| TOTAL | 5.447 | %67 | 2.627 | %33 | 8.074 | |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 54,5 ; dof= 4.*

**Cross: Disease complexity classified into five groups, based on the number of affected body parts. / Calculation of point prevalence 2 modalities**

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | CALCULATION OF POINT PREVALENCE 2 MODALITIES | | | | | |
| --- | --- | --- | --- | --- | --- | --- |
| | RESPONDENTS WITH LESS RARE DISEASES | | RESPONDENTS WITH VERY RARE DISEASES | | TOTAL | |
| | N | % | N | % | N | % |
| 1-3 body parts | 2.431 | %77 | 745 | %23 | 3.176 | %100 |
| 4-7 body parts | 1.357 | %77 | 412 | %23 | 1.769 | %100 |
| 8-11 body parts | 449 | %79 | 118 | %21 | 567 | %100 |
| 12-15 body parts | 135 | %80 | 33 | %20 | 168 | %100 |
| 16 body parts or more | 34 | %83 | 7 | %17 | 41 | %100 |
| TOTAL | 4.406 | %77 | 1.315 | %23 | 5.721 | |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is not significant. p-value= 0,4 ; Chi2= 3,9 ; dof= 4.*

**Cross: Disease complexity classified into five groups, based on the number of affected body parts. / The rare disease was diagnosed before birth**

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | THE RARE DISEASE WAS DIAGNOSED BEFORE BIRTH | | | | | |
|---|---|---|---|---|---|---|
| | YES | | NO | | TOTAL | |
| | N | % | N | % | N | % |
| 1-3 body parts | 147 | %3 | 5.539 | %97 | 5.686 | %100 |
| 4-7 body parts | 54 | %2 | 2.799 | %98 | 2.853 | %100 |
| 8-11 body parts | 15 | %2 | 867 | %98 | 882 | %100 |
| 12-15 body parts | 3 | %1 | 251 | %99 | 254 | %100 |
| 16 body parts or more | 3 | %5 | 53 | %95 | 56 | %100 |
| TOTAL | 222 | %2 | 9.509 | %98 | 9.731 | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is weakly significant. p-value= 0,1 ; Chi2= 9,4 ; dof= 4.*

**Cross: Disease complexity classified into five groups, based on the number of affected body parts. / The rare disease was diagnosed through standard tests carried out at birth**

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | THE RARE DISEASE WAS DIAGNOSED THROUGH STANDARD TESTS CARRIED OUT AT BIRTH | | | | | |
|---|---|---|---|---|---|---|
| | YES | | NO | | TOTAL | |
| | N | % | N | % | N | % |
| 1-3 body parts | 256 | %5 | 5.307 | %95 | 5.563 | %100 |
| 4-7 body parts | 111 | %4 | 2.678 | %96 | 2.789 | %100 |
| 8-11 body parts | 24 | %3 | 851 | %97 | 875 | %100 |
| 12-15 body parts | 2 | %1 | 250 | %99 | 252 | %100 |
| 16 body parts or more | 3 | %5 | 53 | %95 | 56 | %100 |
| TOTAL | 396 | %4 | 9.139 | %96 | 9.535 | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 14,7 ; dof= 4.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Disease complexity classified into five groups, based on the number of affected body parts. / How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?**

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | 0 | | 1 | | BETWEEN 2 AND 4 | | BETWEEN 5 AND 7 | | BETWEEN 8 AND 10 | | MORE THAN 10 | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| 1-3 body parts | 88 | %1 | 775 | %13 | 3.067 | %50 | 1.113 | %18 | 406 | %7 | 654 | %11 | 6.103 | %100 |
| 4-7 body parts | 41 | %1 | 304 | %10 | 1.171 | %38 | 700 | %23 | 258 | %8 | 607 | %20 | 3.081 | %100 |
| 8-11 body parts | 21 | %2 | 62 | %7 | 269 | %28 | 174 | %18 | 93 | %10 | 332 | %35 | 951 | %100 |
| 12-15 body parts | 5 | %2 | 7 | %2 | 54 | %19 | 38 | %13 | 27 | %9 | 155 | %54 | 286 | %100 |
| 16 body parts or more | 0 | %0 | 2 | %3 | 8 | %12 | 8 | %12 | 7 | %11 | 40 | %62 | 65 | %100 |
| TOTAL | 155 | %1 | 1.150 | %11 | 4.569 | %44 | 2.033 | %19 | 791 | %8 | 1.788 | %17 | 10.486 | |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 943,4 ; dof= 20.*

**Cross: Disease complexity classified into five groups, based on the number of affected body parts. / I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases**

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | | | | | |
|---|---|---|---|---|---|---|
| | YES | | NO | | TOTAL | |
| | N | % | N | % | N | % |
| 1-3 body parts | 3.515 | %58 | 2.544 | %42 | 6.059 | %100 |
| 4-7 body parts | 1.784 | %58 | 1.275 | %42 | 3.059 | %100 |
| 8-11 body parts | 514 | %54 | 432 | %46 | 946 | %100 |
| 12-15 body parts | 144 | %51 | 141 | %49 | 285 | %100 |
| 16 body parts or more | 41 | %64 | 23 | %36 | 64 | %100 |
| TOTAL | 5.998 | %58 | 4.415 | %42 | 10.413 | |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is significant. p-value= 0,0 ; Chi2= 12,1 ; dof= 4.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Disease complexity classified into five groups, based on the number of affected body parts. / ...wrongly attributed to another physical disease?

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | ...WRONGLY ATTRIBUTED TO ANOTHER PHYSICAL DISEASE? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 1-3 body parts | 1.230 | %20 | 2.152 | %35 | 2.721 | %45 | 6.103 | %100 |
| 4-7 body parts | 552 | %18 | 1.513 | %49 | 1.016 | %33 | 3.081 | %100 |
| 8-11 body parts | 135 | %14 | 587 | %62 | 229 | %24 | 951 | %100 |
| 12-15 body parts | 30 | %10 | 213 | %74 | 43 | %15 | 286 | %100 |
| 16 body parts or more | 3 | %5 | 55 | %85 | 7 | %11 | 65 | %100 |
| TOTAL | 1.950 | %19 | 4.520 | %43 | 4.016 | %38 | 10.486 | |

🟥 *Under-represented elements* 🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 505,4 ; dof= 8.*

## Cross: Disease complexity classified into five groups, based on the number of affected body parts. / ...neglected, not taken seriously and/or considered as psychological?

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | ...NEGLECTED, NOT TAKEN SERIOUSLY AND/OR CONSIDERED AS PSYCHOLOGICAL? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 1-3 body parts | 759 | %12 | 2.405 | %39 | 2.939 | %48 | 6.103 | %100 |
| 4-7 body parts | 385 | %12 | 1.592 | %52 | 1.104 | %36 | 3.081 | %100 |
| 8-11 body parts | 79 | %8 | 652 | %69 | 220 | %23 | 951 | %100 |
| 12-15 body parts | 17 | %6 | 231 | %81 | 38 | %13 | 286 | %100 |
| 16 body parts or more | 6 | %9 | 54 | %83 | 5 | %8 | 65 | %100 |
| TOTAL | 1.246 | %12 | 4.934 | %47 | 4.306 | %41 | 10.486 | |

🟥 *Under-represented elements* 🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 533,1 ; dof= 8.*

**Cross: Disease complexity classified into five groups, based on the number of affected body parts. / Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed.**

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | | | | | | | |
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
|---|---|---|---|---|---|---|---|---|
| 1-3 body parts | 1.656 | %27 | 2.467 | %40 | 1.980 | %32 | 6.103 | %100 |
| 4-7 body parts | 774 | %25 | 1.661 | %54 | 646 | %21 | 3.081 | %100 |
| 8-11 body parts | 203 | %21 | 614 | %65 | 134 | %14 | 951 | %100 |
| 12-15 body parts | 46 | %16 | 219 | %77 | 21 | %7 | 286 | %100 |
| 16 body parts or more | 4 | %6 | 57 | %88 | 4 | %6 | 65 | %100 |
| TOTAL | 2.683 | %26 | 5.018 | %48 | 2.785 | %27 | 10.486 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 474,2 ; dof= 8.*

**Cross: Disease complexity classified into five groups, based on the number of affected body parts. / Genetic test(s) looking for genetic changes (also called mutations or variants)**

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | GENETIC TEST(S) LOOKING FOR GENETIC CHANGES (ALSO CALLED MUTATIONS OR VARIANTS) | | | | | | | |
| | YES | | NO | | DON'T KNOW/DON'T REMEMBER | | TOTAL | |
| | N | % | N | % | N | % | N | % |
|---|---|---|---|---|---|---|---|---|
| 1-3 body parts | 3.054 | %50 | 2.561 | %42 | 488 | %8 | 6.103 | %100 |
| 4-7 body parts | 1.741 | %57 | 1.098 | %36 | 242 | %8 | 3.081 | %100 |
| 8-11 body parts | 506 | %53 | 372 | %39 | 73 | %8 | 951 | %100 |
| 12-15 body parts | 151 | %53 | 115 | %40 | 20 | %7 | 286 | %100 |
| 16 body parts or more | 38 | %58 | 25 | %38 | 2 | %3 | 65 | %100 |
| TOTAL | 5.490 | %52 | 4.171 | %40 | 825 | %8 | 10.486 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 40,1 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Disease complexity classified into five groups, based on the number of affected body parts. / Other test(s) such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | OTHER TEST(S) SUCH AS CLINICAL EXAMINATION(S), MEDICAL IMAGING (MRI, SCANS…), BIOPSY, BIOCHEMICAL TEST(S) (BLOOD OR URINE TESTS…), ETC | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW/DON'T REMEMBER | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 1-3 body parts | 5.458 | %89 | 459 | %8 | 186 | %3 | 6.103 | %100 |
| 4-7 body parts | 2.832 | %92 | 191 | %6 | 58 | %2 | 3.081 | %100 |
| 8-11 body parts | 870 | %91 | 62 | %7 | 19 | %2 | 951 | %100 |
| 12-15 body parts | 263 | %92 | 17 | %6 | 6 | %2 | 286 | %100 |
| 16 body parts or more | 59 | %91 | 4 | %6 | 2 | %3 | 65 | %100 |
| TOTAL | 9.482 | %90 | 733 | %7 | 271 | %3 | 10.486 | %100 |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 20,2 ; dof= 8.*

## Cross: Disease complexity classified into five groups, based on the number of affected body parts. / …you could not afford it?

Have you ever needed a genetic test but could not access it because...

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | …YOU COULD NOT AFFORD IT? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 1-3 body parts | 492 | %8 | 4.261 | %70 | 1.350 | %22 | 6.103 | %100 |
| 4-7 body parts | 347 | %11 | 2.115 | %69 | 619 | %20 | 3.081 | %100 |
| 8-11 body parts | 172 | %18 | 574 | %60 | 205 | %22 | 951 | %100 |
| 12-15 body parts | 84 | %29 | 133 | %47 | 69 | %24 | 286 | %100 |
| 16 body parts or more | 22 | %34 | 33 | %51 | 10 | %15 | 65 | %100 |
| TOTAL | 1.117 | %11 | 7.116 | %68 | 2.253 | %21 | 10.486 | %100 |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 255,4 ; dof= 8.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

**Cross: Disease complexity classified into five groups, based on the number of affected body parts. / …it was not available in your country?**

Have you ever needed a genetic test but could not access it because...
IT WAS NOT AVAILABLE IN YOUR COUNTRY?

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | YES | | NO | | NOT RELEVANT | | TOTAL | |
|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % |
| 1-3 body parts | 569 | %9 | 4.056 | %66 | 1.478 | %24 | 6.103 | %100 |
| 4-7 body parts | 362 | %12 | 2.025 | %66 | 694 | %23 | 3.081 | %100 |
| 8-11 body parts | 177 | %19 | 569 | %60 | 205 | %22 | 951 | %100 |
| 12-15 body parts | 74 | %26 | 142 | %50 | 70 | %24 | 286 | %100 |
| 16 body parts or more | 15 | %23 | 36 | %55 | 14 | %22 | 65 | %100 |
| TOTAL | 1.197 | %11 | 6.828 | %65 | 2.461 | %23 | 10.486 | %100 |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 148,7 ; dof= 8.*

**Cross: Disease complexity classified into five groups, based on the number of affected body parts. / …healthcare professionals were reluctant or not sufficiently informed?**

Have you ever needed a genetic test but could not access it because...
HEALTHCARE PROFESSIONALS WERE RELUCTANT OR NOT SUFFICIENTLY INFORMED?

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | YES | | NO | | NOT RELEVANT | | TOTAL | |
|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % |
| 1-3 body parts | 1.310 | %21 | 3.458 | %57 | 1.335 | %22 | 6.103 | %100 |
| 4-7 body parts | 900 | %29 | 1.600 | %52 | 581 | %19 | 3.081 | %100 |
| 8-11 body parts | 388 | %41 | 405 | %43 | 158 | %17 | 951 | %100 |
| 12-15 body parts | 165 | %58 | 77 | %27 | 44 | %15 | 286 | %100 |
| 16 body parts or more | 42 | %65 | 16 | %25 | 7 | %11 | 65 | %100 |
| TOTAL | 2.805 | %27 | 5.556 | %53 | 2.125 | %20 | 10.486 | %100 |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 383,6 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

### Cross: Disease complexity classified into five groups, based on the number of affected body parts. / To your knowledge, the genetic test(s) that were conducted targeted…

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | TO YOUR KNOWLEDGE, THE GENETIC TEST(S) THAT WERE CONDUCTED TARGETED… | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | ONLY ONE GENE | | SEVERAL GENES AT THE SAME TIME (GENE PANEL SEQUENCING) | | THE WHOLE DNA (WHOLE GENOME SEQUENCING) | | ALL THE GENES (WHOLE EXOME SEQUENCING) | | A TUMOUR (GENETIC PROFILING OF A TUMOUR) | | OTHER (EPIGENOME, RNA, ETC.) | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| 1-3 body parts | 820 | %27 | 903 | %30 | 475 | %16 | 299 | %10 | 87 | %3 | 56 | %2 | 857 | %28 | 3.054 | |
| 4-7 body parts | 449 | %26 | 572 | %33 | 307 | %18 | 195 | %11 | 29 | %2 | 39 | %2 | 474 | %27 | 1.741 | |
| 8-11 body parts | 140 | %28 | 191 | %38 | 68 | %13 | 51 | %10 | 11 | %2 | 7 | %1 | 127 | %25 | 506 | |
| 12-15 body parts | 41 | %27 | 53 | %35 | 24 | %16 | 15 | %10 | 5 | %3 | 13 | %9 | 40 | %26 | 151 | |
| 16 body parts or more | 10 | %26 | 12 | %32 | 6 | %16 | 7 | %18 | 3 | %8 | 2 | %5 | 13 | %34 | 38 | |
| TOTAL | 1.460 | %27 | 1.731 | %32 | 880 | %16 | 567 | %10 | 135 | %2 | 117 | %2 | 1.511 | %28 | 5.490 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 70,0 ; dof= 24.*

### Cross: Disease complexity classified into five groups, based on the number of affected body parts. / Did you ever request a private company or laboratory to conduct genetic testing to diagnose the disease?

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | DID YOU EVER REQUEST A PRIVATE COMPANY OR LABORATORY TO CONDUCT GENETIC TESTING TO DIAGNOSE THE DISEASE? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO, NEVER | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 1-3 body parts | 311 | %10 | 127 | %4 | 2.616 | %86 | 3.054 | %100 |
| 4-7 body parts | 192 | %11 | 83 | %5 | 1.466 | %84 | 1.741 | %100 |
| 8-11 body parts | 66 | %13 | 28 | %6 | 412 | %81 | 506 | %100 |
| 12-15 body parts | 18 | %12 | 20 | %13 | 113 | %75 | 151 | %100 |
| 16 body parts or more | 3 | %8 | 8 | %21 | 27 | %71 | 38 | %100 |
| TOTAL | 590 | %11 | 266 | %5 | 4.634 | %84 | 5.490 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 53,6 ; dof= 8.*

**Cross: Disease complexity classified into five groups, based on the number of affected body parts. / In general, how satisfied are you with how the results of the GENETIC TESTS were given to you?**

### IN GENERAL, HOW SATISFIED ARE YOU WITH HOW THE RESULTS OF THE GENETIC TESTS WERE GIVEN TO YOU?

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | VERY DISSATISFIED | | DISSATISFIED | | NEITHER SATISFIED NOR DISSATISFIED | | SATISFIED | | VERY SATISFIED | | DON'T KNOW | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| 1-3 body parts | 296 | %10 | 303 | %10 | 607 | %20 | 1.138 | %37 | 548 | %18 | 162 | %5 | 3.054 | %100 |
| 4-7 body parts | 183 | %11 | 206 | %12 | 402 | %23 | 586 | %34 | 280 | %16 | 84 | %5 | 1.741 | %100 |
| 8-11 body parts | 54 | %11 | 84 | %17 | 102 | %20 | 166 | %33 | 69 | %14 | 31 | %6 | 506 | %100 |
| 12-15 body parts | 27 | %18 | 26 | %17 | 39 | %26 | 32 | %21 | 23 | %15 | 4 | %3 | 151 | %100 |
| 16 body parts or more | 12 | %32 | 4 | %11 | 9 | %24 | 8 | %21 | 3 | %8 | 2 | %5 | 38 | %100 |
| TOTAL | 572 | %10 | 623 | %11 | 1.159 | %21 | 1.930 | %35 | 923 | %17 | 283 | %5 | 5.490 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 83,6 ; dof= 20.*

**Cross: Disease complexity classified into five groups, based on the number of affected body parts. / After the tests were performed, were you offered genetic counselling (e.g. given information about how your genetic condition might affect you and your family)?**

### AFTER THE TESTS WERE PERFORMED, WERE YOU OFFERED GENETIC COUNSELLING (E.G. GIVEN INFORMATION ABOUT HOW YOUR GENETIC CONDITION MIGHT AFFECT YOU AND YOUR FAMILY)?

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | YES, WITH A GENETIC COUNSELLOR OR CLINICAL GENETICIST | | YES, BY A HEALTHCARE PROFESSIONAL | | NO, I WASN'T OFFERED GENETIC COUNSELLING | | NOT SURE / DON'T REMEMBER | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % | N | % |
| 1-3 body parts | 1.214 | %40 | 695 | %23 | 900 | %29 | 245 | %8 | 3.054 | %100 |
| 4-7 body parts | 693 | %40 | 374 | %21 | 555 | %32 | 119 | %7 | 1.741 | %100 |
| 8-11 body parts | 175 | %35 | 80 | %16 | 220 | %43 | 31 | %6 | 506 | %100 |
| 12-15 body parts | 48 | %32 | 27 | %18 | 68 | %45 | 8 | %5 | 151 | %100 |
| 16 body parts or more | 7 | %18 | 3 | %8 | 27 | %71 | 1 | %3 | 38 | %100 |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 81,6 ; dof= 12.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Disease complexity classified into five groups, based on the number of affected body parts. / Genetic tests

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | GENETIC TESTS | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 1-3 body parts | 465 | %15 | 2.505 | %82 | 83 | %3 | 3.053 | %100 |
| 4-7 body parts | 262 | %15 | 1.439 | %83 | 40 | %2 | 1.741 | %100 |
| 8-11 body parts | 74 | %15 | 416 | %82 | 16 | %3 | 506 | %100 |
| 12-15 body parts | 24 | %16 | 123 | %81 | 4 | %3 | 151 | %100 |
| 16 body parts or more | 6 | %16 | 32 | %84 | 0 | %0 | 38 | %100 |
| TOTAL | 831 | %15 | 4.515 | %82 | 143 | %3 | 5.489 | |

■ *Under-represented elements* ■ *Over-represented elements*

*The relationship is not significant. p-value= 1,0 ; Chi2= 2,6 ; dof= 8.*

## Cross: Disease complexity classified into five groups, based on the number of affected body parts. / Other diagnostic tests such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc.

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | OTHER DIAGNOSTIC TESTS SUCH AS CLINICAL EXAMINATION(S), MEDICAL IMAGING (MRI, SCANS…), BIOPSY, BIOCHEMICAL TEST(S) (BLOOD OR URINE TESTS…), ETC. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 1-3 body parts | 767 | %14 | 4.580 | %84 | 111 | %2 | 5.458 | %100 |
| 4-7 body parts | 426 | %15 | 2.361 | %83 | 44 | %2 | 2.831 | %100 |
| 8-11 body parts | 142 | %16 | 716 | %82 | 12 | %1 | 870 | %100 |
| 12-15 body parts | 55 | %21 | 206 | %78 | 2 | %1 | 263 | %100 |
| 16 body parts or more | 13 | %22 | 45 | %76 | 1 | %2 | 59 | %100 |
| TOTAL | 1.403 | %15 | 7.908 | %83 | 170 | %2 | 9.481 | %100 |

■ *Under-represented elements* ■ *Over-represented elements*

*The relationship is significant. p-value= 0,0 ; Chi2= 18,8 ; dof= 8.*

**Cross: Disease complexity classified into five groups, based on the number of affected body parts. / Additional advice from a healthcare professional specialised in the rare disease (in person or virtually)**

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | ADDITIONAL ADVICE FROM A HEALTHCARE PROFESSIONAL SPECIALISED IN THE RARE DISEASE (IN PERSON OR VIRTUALLY) | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 1-3 body parts | 1.109 | %18 | 4.858 | %80 | 136 | %2 | 6.103 | %100 |
| 4-7 body parts | 652 | %21 | 2.382 | %77 | 47 | %2 | 3.081 | %100 |
| 8-11 body parts | 223 | %23 | 707 | %74 | 21 | %2 | 951 | %100 |
| 12-15 body parts | 84 | %29 | 197 | %69 | 5 | %2 | 286 | %100 |
| 16 body parts or more | 15 | %23 | 50 | %77 | 0 | %0 | 65 | %100 |
| TOTAL | 2.083 | %20 | 8.194 | %78 | 209 | %2 | 10.486 | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 44,4 ; dof= 8.*

**Cross: Disease complexity classified into five groups, based on the number of affected body parts. / …psychological support**

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | …PSYCHOLOGICAL SUPPORT | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 1-3 body parts | 544 | %9 | 599 | %10 | 458 | %8 | 2.009 | %33 | 2.493 | %41 | 6.103 | %100 |
| 4-7 body parts | 281 | %9 | 242 | %8 | 317 | %10 | 864 | %28 | 1.377 | %45 | 3.081 | %100 |
| 8-11 body parts | 66 | %7 | 80 | %8 | 129 | %14 | 226 | %24 | 450 | %47 | 951 | %100 |
| 12-15 body parts | 24 | %8 | 26 | %9 | 38 | %13 | 56 | %20 | 142 | %50 | 286 | %100 |
| 16 body parts or more | 7 | %11 | 8 | %12 | 10 | %15 | 10 | %15 | 30 | %46 | 65 | %100 |
| TOTAL | 922 | %9 | 955 | %9 | 952 | %9 | 3.165 | %30 | 4.492 | %43 | 10.486 | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 129,6 ; dof= 16.*

**Cross: Disease complexity classified into five groups, based on the number of affected body parts. / …care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc.**

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | …CARE COORDINATION SUPPORT SUCH AS HELP TO FIND THE NECESSARY INFORMATION ON THE DISEASE AND THE RIGHT PROFESSIONALS, ARRANGING APPOINTMENTS WITH DIFFERENT HEALTH PROVIDERS, ETC. | | | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 1-3 body parts | 1.316 | %22 | 266 | %4 | 764 | %13 | 1.113 | %18 | 2.644 | %43 | 6.103 | %100 |
| 4-7 body parts | 597 | %19 | 97 | %3 | 486 | %16 | 387 | %13 | 1.514 | %49 | 3.081 | %100 |
| 8-11 body parts | 136 | %14 | 20 | %2 | 156 | %16 | 115 | %12 | 524 | %55 | 951 | %100 |
| 12-15 body parts | 30 | %10 | 4 | %1 | 41 | %14 | 12 | %4 | 199 | %70 | 286 | %100 |
| 16 body parts or more | 4 | %6 | 4 | %6 | 16 | %25 | 0 | %0 | 41 | %63 | 65 | %100 |
| **TOTAL** | **2.083** | **%20** | **391** | **%4** | **1.463** | **%14** | **1.627** | **%16** | **4.922** | **%47** | **10.486** | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 245,7 ; dof= 16.*

**Cross: Disease complexity classified into five groups, based on the number of affected body parts. / …financial support including social security benefits**

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | …FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS | | | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 1-3 body parts | 855 | %14 | 169 | %3 | 630 | %10 | 2.368 | %39 | 2.037 | %34 | 6.059 | %100 |
| 4-7 body parts | 428 | %14 | 55 | %2 | 408 | %13 | 931 | %30 | 1.237 | %40 | 3.059 | %100 |
| 8-11 body parts | 98 | %10 | 16 | %2 | 140 | %15 | 201 | %21 | 491 | %52 | 946 | %100 |
| 12-15 body parts | 23 | %8 | 1 | %0 | 40 | %14 | 40 | %14 | 181 | %64 | 285 | %100 |
| 16 body parts or more | 1 | %2 | 2 | %3 | 14 | %22 | 4 | %6 | 43 | %67 | 64 | %100 |
| **TOTAL** | **1.405** | **%13** | **243** | **%2** | **1.232** | **%12** | **3.544** | **%34** | **3.989** | **%38** | **10.413** | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 363,7 ; dof= 16.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

## Did the first symptoms include…

| | YES | NO | DON'T KNOW | TOTAL |
|---|---|---|---|---|
| …behavioural disorders that cause problems in school, at home or in social situations | 2.957 | 7.085 | 444 | 10.486 |
| …intellectual disabilities or cognitive symptoms (i.e. problems with memory, language, thinking or judgement) | 2.936 | 7.236 | 314 | 10.486 |
| …clinical signs or symptoms that come and go | 5.940 | 3.788 | 758 | 10.486 |
| …invisible symptoms such as pain, dizziness, headaches, fatigue, etc. | 7.020 | 2.916 | 550 | 10.486 |
| …sudden onset symptoms requiring urgent care | 4.648 | 5.251 | 587 | 10.486 |

## Did the first symptoms include…



- Yes
- No
- Don't know
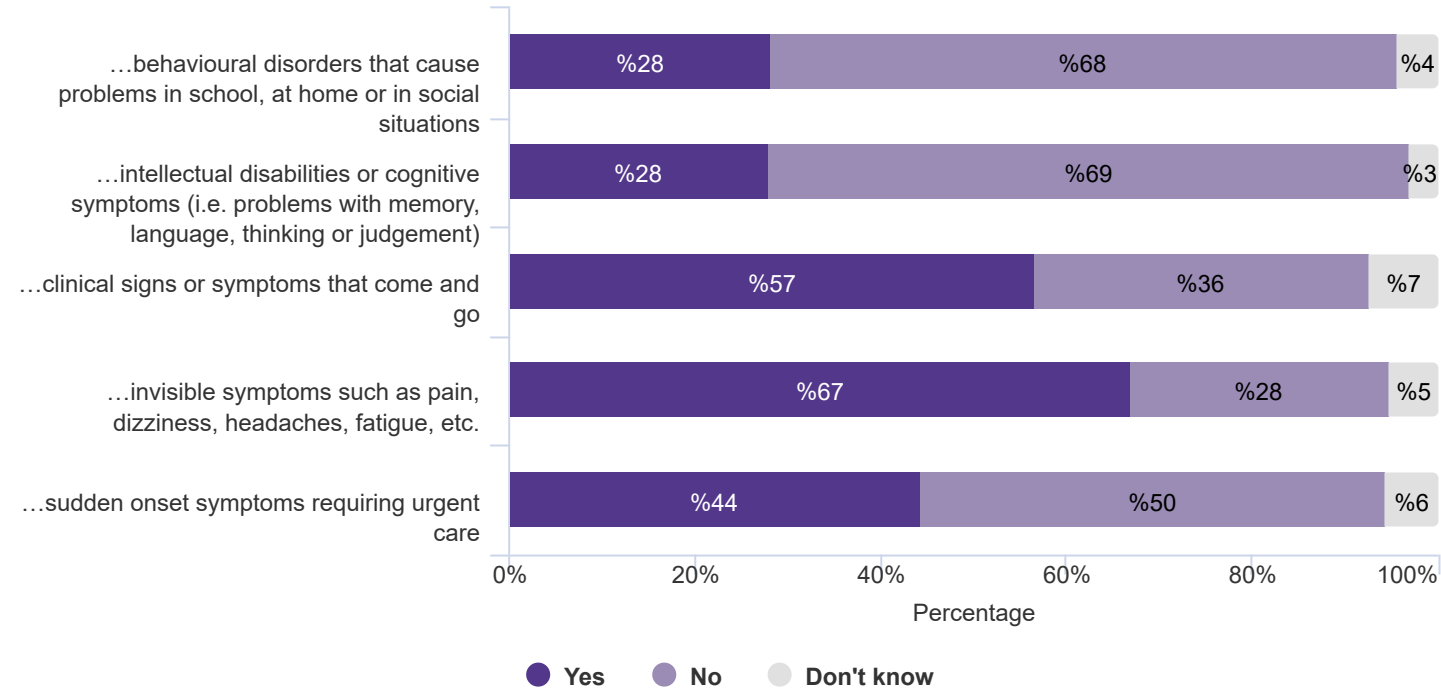
## And the rare disease causes:

| | N |
|---|---|
| Epilepsy | 997 |
| Congenital malformations, developmental anomalies or intellectual disabilities | 2.424 |
| Predisposition to the development of tumours | 877 |
| Solid tumours | 409 |
| Blood cancers (including leukaemia, lymphoma and myeloma) | 365 |
| A need for organ transplantation | 766 |
| None of the above | 4.064 |
| Other, specify... | 2.133 |
| **TOTAL** | **9.693** |

## And the rare disease causes:

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

# Chapter 4.

## Family members were already diagnosed with the --

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

## Did the first symptoms include…

| | YES | NO | DON'T KNOW | TOTAL |
|---|---|---|---|---|
| …behavioural disorders that cause problems in school, at home or in social situations | 2.957 | 7.085 | 444 | **10.486** |
| …intellectual disabilities or cognitive symptoms (i.e. problems with memory, language, thinking or judgement) | 2.936 | 7.236 | 314 | **10.486** |
| …clinical signs or symptoms that come and go | 5.940 | 3.788 | 758 | **10.486** |
| …invisible symptoms such as pain, dizziness, headaches, fatigue, etc. | 7.020 | 2.916 | 550 | **10.486** |
| …sudden onset symptoms requiring urgent care | 4.648 | 5.251 | 587 | **10.486** |
| **TOTAL** | **23.501** | **26.276** | **2.653** | **52.430** |

## Did the first symptoms include…



- Yes
- No
- Don't know

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses
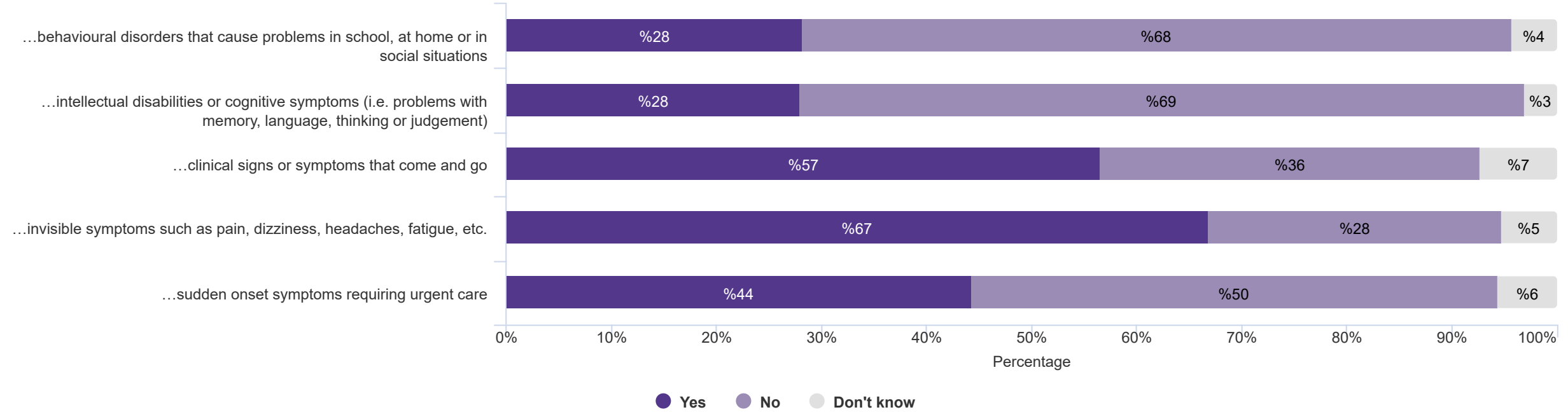
**Did the first symptoms include...**

## Multiple Cross

| ...behavioural disorders that cause problems in school, at home or in social situations | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| Yes | 0,8 | 2.226 | 3,6 | 2.003 | 3,8 | 1.172 | 3,9 | 2.190 | 5,0 | 1.783 |
| No | 0,4 | 5.320 | 3,5 | 5.071 | 3,8 | 3.032 | 3,5 | 5.370 | 4,6 | 4.501 |
| Don't know | -0,4 | 274 | 2,9 | 248 | 4,8 | 131 | 3,8 | 283 | 5,2 | 223 |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Fisher= 5,0.*

*Inter variance= 230,0. Intra variance= 46,0.*

**Mean** = average time, in number of years

**N** = number of respondents for which we have the average time

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Did the first symptoms include...**

**Multiple Cross**

| ...intellectual disabilities or cognitive symptoms (i.e. problems with memory, language, thinking or judgement) | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | **MEAN** | **N** | **MEAN** | **N** | **MEAN** | **N** | **MEAN** | **N** | **MEAN** | **N** |
| Yes | 0,3 | 2.254 | *3,0* | 1.967 | 3,4 | 1.147 | 3,8 | 2.220 | 4,5 | 1.830 |
| No | 0,6 | 5.358 | 3,7 | 5.172 | 4,0 | 3.075 | 3,5 | 5.431 | 4,8 | 4.517 |
| Don't know | -0,2 | 208 | 2,4 | 183 | 4,8 | 113 | 3,4 | 192 | 4,9 | 160 |

🟣 *Under-represented elements*  🔵 *Over-represented elements*

*The relationship is significant. p-value= 0,0 ; Fisher= 3,3.*

*Inter variance= 150,6. Intra variance= 46,0.*

**Mean** = average time, in number of years

**N** = number of respondents for which we have the average time

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

Did the first symptoms include...

## Multiple Cross

| ...clinical signs or symptoms that come and go | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| Yes | 0,4 | 4.586 | 4,0 | 4.324 | 4,4 | 2.467 | 4,1 | 4.488 | 5,4 | 3.680 |
| No | 0,6 | 2.744 | 2,8 | 2.547 | 3,0 | 1.600 | 2,8 | 2.858 | 3,8 | 2.417 |
| Don't know | 0,3 | 490 | 2,7 | 451 | 3,7 | 268 | 3,4 | 497 | 4,1 | 410 |

■ *Under-represented elements*　　■ *Over-represented elements*

*The relationship is not significant. p-value= 0,6 ; Fisher= 0,6.*

*Inter variance= 26,9. Intra variance= 46,0.*

**Mean** = average time, in number of years

**N** = number of respondents for which we have the average time

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

**Did the first symptoms include...**

## Multiple Cross

| ...invisible symptoms such as pain, dizziness, headaches, fatigue, etc. | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| Yes | 0,4 | 5.372 | 3,9 | 5.099 | 4,5 | 2.939 | 4,2 | 5.271 | 5,1 | 4.309 |
| No | 0,6 | 2.071 | 2,6 | 1.902 | 2,6 | 1.197 | 2,4 | 2.191 | 3,9 | 1.877 |
| Don't know | 0,5 | 377 | 1,8 | 321 | 2,6 | 199 | 2,7 | 381 | 3,8 | 321 |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is not significant. p-value= 0,7 ; Fisher= 0,4.*

*Inter variance= 18,8. Intra variance= 46,0.*

**Mean** = average time, in number of years

**N** = number of respondents for which we have the average time

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Did the first symptoms include...**

## Multiple Cross

| ...sudden onset symptoms requiring urgent care | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| Yes | 0,2 | 3.601 | **3,1** | 3.462 | 3,8 | 1.947 | 3,5 | 3.547 | 4,8 | 2.940 |
| No | 0,7 | 3.836 | 3,8 | 3.526 | 3,7 | 2.180 | 3,6 | 3.917 | 4,6 | 3.280 |
| Don't know | 0,7 | 383 | 4,3 | 334 | 5,4 | 208 | 4,5 | 379 | 5,1 | 287 |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is significant. p-value= 0,0 ; Fisher= 4,1.*

*Inter variance= 190,5. Intra variance= 46,0.*

**Mean** = average time, in number of years

**N** = number of respondents for which we have the average time

## Cross: Gender of the person affected by the rare disease / …behavioural disorders that cause problems in school, at home or in social situations

| GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE | …BEHAVIOURAL DISORDERS THAT CAUSE PROBLEMS IN SCHOOL, AT HO SOCIAL SITUATIONS | | | | | | TO |
|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | |
| | N | % | N | % | N | % | N |
| Female | 1.702 | %26 | 4.661 | %70 | 296 | %4 | 6.659 |
| Male | 912 | %32 | 1.795 | %64 | 103 | %4 | 2.810 |
| Other | 35 | %35 | 57 | %56 | 9 | %9 | 101 |
| TOTAL | 2.649 | %28 | 6.513 | %68 | 408 | %4 | 9.570 |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 56,6 ; dof= 4.*

## Cross: Gender of the person affected by the rare disease / …intellectual disabilities or cognitive symptoms (i.e. problems with memory, language, thinking or judgement)

| GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE | …INTELLECTUAL DISABILITIES OR COGNITIVE SYMPTOMS (I.E. PROBLEM MEMORY, LANGUAGE, THINKING OR JUDGEMENT) | | | | | | TO |
|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | |
| | N | % | N | % | N | % | N |
| Female | 1.735 | %26 | 4.721 | %71 | 203 | %3 | 6.659 |
| Male | 873 | %31 | 1.858 | %66 | 79 | %3 | 2.810 |
| Other | 37 | %37 | 58 | %57 | 6 | %6 | 101 |
| TOTAL | 2.645 | %28 | 6.637 | %69 | 288 | %3 | 9.570 |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 32,8 ; dof= 4.*

## Cross: Gender of the person affected by the rare disease / …clinical signs or symptoms that come and go

| GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE | …CLINICAL SIGNS OR SYMPTOMS THAT COME AND GO | | | | | | TO |
|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | |
| | N | % | N | % | N | % | N |
| Female | 4.062 | %61 | 2.137 | %32 | 460 | %7 | 6.659 |
| Male | 1.363 | %49 | 1.239 | %44 | 208 | %7 | 2.810 |
| Other | 50 | %50 | 41 | %41 | 10 | %10 | 101 |
| TOTAL | 5.475 | %57 | 3.417 | %36 | 678 | %7 | 9.570 |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 137,2 ; dof= 4.*

## Cross: Gender of the person affected by the rare disease / …invisible symptoms such as pain, dizziness, headaches, fatigue, etc.

| GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE | …INVISIBLE SYMPTOMS SUCH AS PAIN, DIZZINESS, HEADACHES, FATIGU | | | | | | TO |
|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | |
| | N | % | N | % | N | % | N |
| Female | 4.869 | %73 | 1.506 | %23 | 284 | %4 | 6.659 |
| Male | 1.542 | %55 | 1.067 | %38 | 201 | %7 | 2.810 |
| Other | 57 | %56 | 34 | %34 | 10 | %10 | 101 |
| TOTAL | 6.468 | %68 | 2.607 | %27 | 495 | %5 | 9.570 |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 308,1 ; dof= 4.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Gender of the person affected by the rare disease / …sudden onset symptoms requiring urgent care

| GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE | …SUDDEN ONSET SYMPTOMS REQUIRING URGENT CARE | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Female | 3.016 | %45 | 3.242 | %49 | 401 | %6 | 6.659 | %100 |
| Male | 1.202 | %43 | 1.479 | %53 | 129 | %5 | 2.810 | %100 |
| Other | 41 | %41 | 50 | %50 | 10 | %10 | 101 | %100 |
| TOTAL | 4.259 | %45 | 4.771 | %50 | 540 | %6 | 9.570 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 19,8 ; dof= 4.*

## Cross: How old were you when you stopped full-time education? / …behavioural disorders that cause problems in school, at home or in social situations

| HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION? | …BEHAVIOURAL DISORDERS THAT CAUSE PROBLEMS IN SCHOOL, AT HOME OR IN SOCIAL SITUATIONS | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 15 y.o. or under | 176 | %39 | 252 | %55 | 27 | %6 | 455 | %100 |
| between 16 and 19 y.o. | 737 | %30 | 1.585 | %64 | 142 | %6 | 2.464 | %100 |
| between 20 and 23 y.o. | 814 | %27 | 2.090 | %69 | 118 | %4 | 3.022 | %100 |
| 24 y.o. or above | 809 | %26 | 2.234 | %71 | 102 | %3 | 3.145 | %100 |
| TOTAL | 2.536 | %28 | 6.161 | %68 | 389 | %4 | 9.086 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 73,7 ; dof= 6.*

## Cross: How old were you when you stopped full-time education? / …intellectual disabilities or cognitive symptoms (i.e. problems with memory, language, thinking or judgement)

| HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION? | …INTELLECTUAL DISABILITIES OR COGNITIVE SYMPTOMS (I.E. PROBLEMS WITH MEMORY, LANGUAGE, THINKING OR JUDGEMENT) | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 15 y.o. or under | 149 | %33 | 282 | %62 | 24 | %5 | 455 | %100 |
| between 16 and 19 y.o. | 719 | %29 | 1.662 | %67 | 83 | %3 | 2.464 | %100 |
| between 20 and 23 y.o. | 814 | %27 | 2.124 | %70 | 84 | %3 | 3.022 | %100 |
| 24 y.o. or above | 868 | %28 | 2.197 | %70 | 80 | %3 | 3.145 | %100 |
| TOTAL | 2.550 | %28 | 6.265 | %69 | 271 | %3 | 9.086 | %100 |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 23,1 ; dof= 6.*

## Cross: How old were you when you stopped full-time education? / …clinical signs or symptoms that come and go

| HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION? | …CLINICAL SIGNS OR SYMPTOMS THAT COME AND GO | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 15 y.o. or under | 249 | %55 | 160 | %35 | 46 | %10 | 455 | %100 |
| between 16 and 19 y.o. | 1.377 | %56 | 889 | %36 | 198 | %8 | 2.464 | %100 |
| between 20 and 23 y.o. | 1.740 | %58 | 1.079 | %36 | 203 | %7 | 3.022 | %100 |
| 24 y.o. or above | 1.810 | %58 | 1.136 | %36 | 199 | %6 | 3.145 | %100 |
| TOTAL | 5.176 | %57 | 3.264 | %36 | 646 | %7 | 9.086 | %100 |

🟣 *Under-represented elements*  🔵 *Over-represented elements*

*The relationship is significant. p-value= 0,0 ; Chi2= 13,6 ; dof= 6.*

## Cross: How old were you when you stopped full-time education? / …invisible symptoms such as pain, dizziness, headaches, fatigue, etc.

| HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION? | …INVISIBLE SYMPTOMS SUCH AS PAIN, DIZZINESS, HEADACHES, FATIGUE, ETC. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 15 y.o. or under | 289 | %64 | 135 | %30 | 31 | %7 | 455 | %100 |
| between 16 and 19 y.o. | 1.705 | %69 | 639 | %26 | 120 | %5 | 2.464 | %100 |
| between 20 and 23 y.o. | 2.033 | %67 | 816 | %27 | 173 | %6 | 3.022 | %100 |
| 24 y.o. or above | 2.040 | %65 | 940 | %30 | 165 | %5 | 3.145 | %100 |
| TOTAL | 6.067 | %67 | 2.530 | %28 | 489 | %5 | 9.086 | %100 |

🟣 *Under-represented elements*  🔵 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 17,7 ; dof= 6.*

## Cross: How old were you when you stopped full-time education? / …sudden onset symptoms requiring urgent care

| HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION? | …SUDDEN ONSET SYMPTOMS REQUIRING URGENT CARE | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 15 y.o. or under | 224 | %49 | 204 | %45 | 27 | %6 | 455 | %100 |
| between 16 and 19 y.o. | 1.165 | %47 | 1.141 | %46 | 158 | %6 | 2.464 | %100 |
| between 20 and 23 y.o. | 1.303 | %43 | 1.542 | %51 | 177 | %6 | 3.022 | %100 |
| 24 y.o. or above | 1.337 | %43 | 1.659 | %53 | 149 | %5 | 3.145 | %100 |
| TOTAL | 4.029 | %44 | 4.546 | %50 | 511 | %6 | 9.086 | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 32,7 ; dof= 6.*

## Cross: …behavioural disorders that cause problems in school, at home or in social situations / How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?

| …BEHAVIOURAL DISORDERS THAT CAUSE PROBLEMS IN SCHOOL, AT HOME OR IN SOCIAL SITUATIONS | HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | 0 | | 1 | | BETWEEN 2 AND 4 | | BETWEEN 5 AND 7 | | BETWEEN 8 AND 10 | | MORE THAN 10 | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 45 | %2 | 248 | %8 | 1.161 | %39 | 642 | %22 | 253 | %9 | 608 | %21 | 2.957 | %100 |
| No | 96 | %1 | 852 | %12 | 3.218 | %45 | 1.314 | %19 | 507 | %7 | 1.098 | %15 | 7.085 | %100 |
| Don't know | 14 | %3 | 50 | %11 | 190 | %43 | 77 | %17 | 31 | %7 | 82 | %18 | 444 | %100 |
| TOTAL | 155 | %1 | 1.150 | %11 | 4.569 | %44 | 2.033 | %19 | 791 | %8 | 1.788 | %17 | 10.486 | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 101,9 ; dof= 10.*

## Cross: …intellectual disabilities or cognitive symptoms (i.e. problems with memory, language, thinking or judgement) / How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?

| …INTELLECTUAL DISABILITIES OR COGNITIVE SYMPTOMS (I.E. PROBLEMS WITH MEMORY, LANGUAGE, THINKING OR JUDGEMENT) | HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | 0 | | 1 | | BETWEEN 2 AND 4 | | BETWEEN 5 AND 7 | | BETWEEN 8 AND 10 | | MORE THAN 10 | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 47 | %2 | 242 | %8 | 1.104 | %38 | 634 | %22 | 274 | %9 | 635 | %22 | 2.936 | %100 |
| No | 102 | %1 | 880 | %12 | 3.335 | %46 | 1.334 | %18 | 494 | %7 | 1.091 | %15 | 7.236 | %100 |
| Don't know | 6 | %2 | 28 | %9 | 130 | %41 | 65 | %21 | 23 | %7 | 62 | %20 | 314 | %100 |
| TOTAL | 155 | %1 | 1.150 | %11 | 4.569 | %44 | 2.033 | %19 | 791 | %8 | 1.788 | %17 | 10.486 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 148,7 ; dof= 10.*

## Cross: …clinical signs or symptoms that come and go / How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?

| …CLINICAL SIGNS OR SYMPTOMS THAT COME AND GO | HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | 0 | | 1 | | BETWEEN 2 AND 4 | | BETWEEN 5 AND 7 | | BETWEEN 8 AND 10 | | MORE THAN 10 | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 66 | %1 | 503 | %8 | 2.378 | %40 | 1.230 | %21 | 533 | %9 | 1.230 | %21 | 5.940 | %100 |
| No | 69 | %2 | 555 | %15 | 1.834 | %48 | 666 | %18 | 214 | %6 | 450 | %12 | 3.788 | %100 |
| Don't know | 20 | %3 | 92 | %12 | 357 | %47 | 137 | %18 | 44 | %6 | 108 | %14 | 758 | %100 |
| TOTAL | 155 | %1 | 1.150 | %11 | 4.569 | %44 | 2.033 | %19 | 791 | %8 | 1.788 | %17 | 10.486 | %100 |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 295,6 ; dof= 10.*

## Cross: …invisible symptoms such as pain, dizziness, headaches, fatigue, etc. / How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?

| ...INVISIBLE SYMPTOMS SUCH AS PAIN, DIZZINESS, HEADACHES, FATIGUE, ETC. | HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | 0 | | 1 | | BETWEEN 2 AND 4 | | BETWEEN 5 AND 7 | | BETWEEN 8 AND 10 | | MORE THAN 10 | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 65 | %1 | 619 | %9 | 2.878 | %41 | 1.432 | %20 | 608 | %9 | 1.418 | %20 | 7.020 | %100 |
| No | 77 | %3 | 468 | %16 | 1.454 | %50 | 478 | %16 | 148 | %5 | 291 | %10 | 2.916 | %100 |
| Don't know | 13 | %2 | 63 | %11 | 237 | %43 | 123 | %22 | 35 | %6 | 79 | %14 | 550 | %100 |
| TOTAL | 155 | %1 | 1.150 | %11 | 4.569 | %44 | 2.033 | %19 | 791 | %8 | 1.788 | %17 | 10.486 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 364,2 ; dof= 10.*

## Cross: …sudden onset symptoms requiring urgent care / How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?

| ...SUDDEN ONSET SYMPTOMS REQUIRING URGENT CARE | HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | 0 | | 1 | | BETWEEN 2 AND 4 | | BETWEEN 5 AND 7 | | BETWEEN 8 AND 10 | | MORE THAN 10 | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 65 | %1 | 419 | %9 | 1.857 | %40 | 957 | %21 | 398 | %9 | 952 | %20 | 4.648 | %100 |
| No | 77 | %1 | 674 | %13 | 2.469 | %47 | 949 | %18 | 353 | %7 | 729 | %14 | 5.251 | %100 |
| Don't know | 13 | %2 | 57 | %10 | 243 | %41 | 127 | %22 | 40 | %7 | 107 | %18 | 587 | %100 |
| TOTAL | 155 | %1 | 1.150 | %11 | 4.569 | %44 | 2.033 | %19 | 791 | %8 | 1.788 | %17 | 10.486 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 149,7 ; dof= 10.*

## Cross: …behavioural disorders that cause problems in school, at home or in social situations / I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases

| …BEHAVIOURAL DISORDERS THAT CAUSE PROBLEMS IN SCHOOL, AT HOME OR IN SOCIAL SITUATIONS | I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | | | | | |
|---|---|---|---|---|---|---|
| | YES | | NO | | TOTAL | |
| | N | % | N | % | N | % |
| Yes | 1.669 | %57 | 1.259 | %43 | 2.928 | %100 |
| No | 4.110 | %58 | 2.933 | %42 | 7.043 | %100 |
| Don't know | 219 | %50 | 223 | %50 | 442 | %100 |
| **TOTAL** | **5.998** | **%58** | **4.415** | **%42** | **10.413** | **%100** |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 13,8 ; dof= 2.*

## Cross: …intellectual disabilities or cognitive symptoms (i.e. problems with memory, language, thinking or judgement) / I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases

| …INTELLECTUAL DISABILITIES OR COGNITIVE SYMPTOMS (I.E. PROBLEMS WITH MEMORY, LANGUAGE, THINKING OR JUDGEMENT) | I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | | | | | |
|---|---|---|---|---|---|---|
| | YES | | NO | | TOTAL | |
| | N | % | N | % | N | % |
| Yes | 1.564 | %54 | 1.349 | %46 | 2.913 | %100 |
| No | 4.260 | %59 | 2.927 | %41 | 7.187 | %100 |
| Don't know | 174 | %56 | 139 | %44 | 313 | %100 |
| **TOTAL** | **5.998** | **%58** | **4.415** | **%42** | **10.413** | **%100** |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 27,0 ; dof= 2.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: …clinical signs or symptoms that come and go / I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases**

| …CLINICAL SIGNS OR SYMPTOMS THAT COME AND GO | I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | | | | | |
| | YES | | NO | | TOTAL | |
| | N | % | N | % | N | % |
| Yes | **3.342** | **%57** | **2.555** | **%43** | 5.897 | %100 |
| No | **2.218** | **%59** | **1.542** | **%41** | 3.760 | %100 |
| Don't know | 438 | %58 | 318 | %42 | 756 | %100 |
| **TOTAL** | **5.998** | **%58** | **4.415** | **%42** | **10.413** | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is weakly significant. p-value= 0,1 ; Chi2= 5,1 ; dof= 2.*

**Cross: …invisible symptoms such as pain, dizziness, headaches, fatigue, etc. / I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases**

| …INVISIBLE SYMPTOMS SUCH AS PAIN, DIZZINESS, HEADACHES, FATIGUE, ETC. | I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | | | | | |
| | YES | | NO | | TOTAL | |
| | N | % | N | % | N | % |
| Yes | 3.972 | %57 | 2.993 | %43 | 6.965 | %100 |
| No | **1.721** | **%59** | **1.177** | **%41** | 2.898 | %100 |
| Don't know | 305 | %55 | 245 | %45 | 550 | %100 |
| **TOTAL** | **5.998** | **%58** | **4.415** | **%42** | **10.413** | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is weakly significant. p-value= 0,1 ; Chi2= 5,8 ; dof= 2.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

**Cross: …sudden onset symptoms requiring urgent care / I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases**

| …SUDDEN ONSET SYMPTOMS REQUIRING URGENT CARE | I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | | | | | |
|---|---|---|---|---|---|---|
| | YES | | NO | | TOTAL | |
| | N | % | N | % | N | % |
| Yes | 2.674 | %58 | 1.932 | %42 | 4.606 | %100 |
| No | 3.005 | %58 | 2.216 | %42 | 5.221 | %100 |
| Don't know | 319 | %54 | 267 | %46 | 586 | %100 |
| TOTAL | 5.998 | %58 | 4.415 | %42 | 10.413 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is not significant. p-value= 0,2 ; Chi2= 2,8 ; dof= 2.*

Rare Barometer
A EURORDIS INITIATIVE

EURORDIS
RARE DISEASES EUROPE

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: …behavioural disorders that cause problems in school, at home or in social situations / … wrongly attributed to another physical disease?**

| … BEHAVIOUR… DISORDERS THAT CAUSE PROBLEMS IN SCHOOL, AT HOME OR IN SOCIAL SITUATIONS | …WRONGLY ATTRIBUTED TO ANOTHER PHYSICAL DISEASE? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 527 | %18 | 1.382 | %47 | 1.048 | %35 | 2.957 | %100 |
| No | 1.351 | %19 | 2.935 | %41 | 2.799 | %40 | 7.085 | %100 |
| Don't know | 72 | %16 | 203 | %46 | 169 | %38 | 444 | %100 |
| TOTAL | 1.950 | %19 | 4.520 | %43 | 4.016 | %38 | 10.486 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 26,5 ; dof= 4.*

**Cross: …behavioural disorders that cause problems in school, at home or in social situations / … neglected, not taken seriously and/or considered as psychological?**

| … BEHAVIOURAL DISORDERS THAT CAUSE PROBLEMS IN SCHOOL, AT HOME OR IN SOCIAL SITUATIONS | …NEGLECTED, NOT TAKEN SERIOUSLY AND/OR CONSIDERED AS PSYCHOLOGICAL? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 338 | %11 | 1.624 | %55 | 995 | %34 | 2.957 | %100 |
| No | 863 | %12 | 3.076 | %43 | 3.146 | %44 | 7.085 | %100 |
| Don't know | 45 | %10 | 234 | %53 | 165 | %37 | 444 | %100 |
| TOTAL | 1.246 | %12 | 4.934 | %47 | 4.306 | %41 | 10.486 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 124,5 ; dof= 4.*

**Cross: …behavioural disorders that cause problems in school, at home or in social situations / Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed.**

| …BEHAVIOURAL DISORDERS THAT CAUSE PROBLEMS IN SCHOOL, AT HOME OR IN SOCIAL SITUATIONS | HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 776 | %26 | 1.525 | %52 | 656 | %22 | 2.957 | %100 |
| No | 1.797 | %25 | 3.271 | %46 | 2.017 | %28 | 7.085 | %100 |
| Don't know | 110 | %25 | 222 | %50 | 112 | %25 | 444 | %100 |
| TOTAL | 2.683 | %26 | 5.018 | %48 | 2.785 | %27 | 10.486 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 45,3 ; dof= 4.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

## Cross: …intellectual disabilities or cognitive symptoms (i.e. problems with memory, language, thinking or judgement) / …wrongly attributed to another physical disease?

| …INTELLECT… DISABILITIES OR COGNITIVE SYMPTOMS (I.E. PROBLEMS WITH MEMORY, LANGUAGE, THINKING OR JUDGEMENT) | …WRONGLY ATTRIBUTED TO ANOTHER PHYSICAL DISEASE? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 514 | %18 | 1.368 | %47 | 1.054 | %36 | 2.936 | %100 |
| No | 1.383 | %19 | 3.010 | %42 | 2.843 | %39 | 7.236 | %100 |
| Don't know | 53 | %17 | 142 | %45 | 119 | %38 | 314 | %100 |
| TOTAL | 1.950 | %19 | 4.520 | %43 | 4.016 | %38 | 10.486 | |

■ Under-represented elements    ■ Over-represented elements

*The relationship is very significant. p-value= < 0,01 ; Chi2= 22,1 ; dof= 4.*

## Cross: …intellectual disabilities or cognitive symptoms (i.e. problems with memory, language, thinking or judgement) / …neglected, not taken seriously and/or considered as psychological?

| …INTELLECTUAL DISABILITIES OR COGNITIVE SYMPTOMS (I.E. PROBLEMS WITH MEMORY, LANGUAGE, THINKING OR JUDGEMENT) | …NEGLECTED, NOT TAKEN SERIOUSLY AND/OR CONSIDERED AS PSYCHOLOGICAL? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 327 | %11 | 1.602 | %55 | 1.007 | %34 | 2.936 | %100 |
| No | 876 | %12 | 3.161 | %44 | 3.199 | %44 | 7.236 | %100 |
| Don't know | 43 | %14 | 171 | %54 | 100 | %32 | 314 | %100 |
| TOTAL | 1.246 | %12 | 4.934 | %47 | 4.306 | %41 | 10.486 | |

■ Under-represented elements    ■ Over-represented elements

*The relationship is very significant. p-value= < 0,01 ; Chi2= 115,5 ; dof= 4.*

## Cross: …intellectual disabilities or cognitive symptoms (i.e. problems with memory, language, thinking or judgement) / Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed.

| …INTELLECTUAL DISABILITIES OR COGNITIVE SYMPTOMS (I.E. PROBLEMS WITH MEMORY, LANGUAGE, THINKING OR JUDGEMENT) | HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 787 | %27 | 1.493 | %51 | 656 | %22 | 2.936 | %100 |
| No | 1.817 | %25 | 3.366 | %47 | 2.053 | %28 | 7.236 | %100 |
| Don't know | 79 | %25 | 159 | %51 | 76 | %24 | 314 | %100 |
| TOTAL | 2.683 | %26 | 5.018 | %48 | 2.785 | %27 | 10.486 | |

■ Under-represented elements    ■ Over-represented elements

*The relationship is very significant. p-value= < 0,01 ; Chi2= 40,3 ; dof= 4.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: …clinical signs or symptoms that come and go / …wrongly attributed to another physical disease?

| …CLINICAL SIGNS OR SYMPTOMS THAT COME AND GO | …WRONGLY ATTRIBUTED TO ANOTHER PHYSICAL DISEASE? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 1.062 | %18 | 3.070 | %52 | 1.808 | %30 | 5.940 | %100 |
| No | 745 | %20 | 1.171 | %31 | 1.872 | %49 | 3.788 | %100 |
| Don't know | 143 | %19 | 279 | %37 | 336 | %44 | 758 | %100 |
| TOTAL | 1.950 | %19 | 4.520 | %43 | 4.016 | %38 | 10.486 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 468,4 ; dof= 4.*

## Cross: …clinical signs or symptoms that come and go / …neglected, not taken seriously and/or considered as psychological?

| …CLINICAL SIGNS OR SYMPTOMS THAT COME AND GO | …NEGLECTED, NOT TAKEN SERIOUSLY AND/OR CONSIDERED AS PSYCHOLOGICAL? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 703 | %12 | 3.352 | %56 | 1.885 | %32 | 5.940 | %100 |
| No | 447 | %12 | 1.264 | %33 | 2.077 | %55 | 3.788 | %100 |
| Don't know | 96 | %13 | 318 | %42 | 344 | %45 | 758 | %100 |
| TOTAL | 1.246 | %12 | 4.934 | %47 | 4.306 | %41 | 10.486 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 570,6 ; dof= 4.*

## Cross: …clinical signs or symptoms that come and go / Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed.

| …CLINICAL SIGNS OR SYMPTOMS THAT COME AND GO | HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 1.485 | %25 | 3.340 | %56 | 1.115 | %19 | 5.940 | %100 |
| No | 987 | %26 | 1.364 | %36 | 1.437 | %38 | 3.788 | %100 |
| Don't know | 211 | %28 | 314 | %41 | 233 | %31 | 758 | %100 |
| TOTAL | 2.683 | %26 | 5.018 | %48 | 2.785 | %27 | 10.486 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 532,5 ; dof= 4.*

**Cross: …invisible symptoms such as pain, dizziness, headaches, fatigue, etc. / …wrongly attributed to another physical disease?**

| …INVISIBLE SYMPTOMS SUCH AS PAIN, DIZZINESS, HEADACHES, FATIGUE, ETC. | …WRONGLY ATTRIBUTED TO ANOTHER PHYSICAL DISEASE? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 1.330 | %19 | 3.580 | %51 | 2.110 | %30 | 7.020 | %100 |
| No | 533 | %18 | 775 | %27 | 1.608 | %55 | 2.916 | %100 |
| Don't know | 87 | %16 | 165 | %30 | 298 | %54 | 550 | %100 |
| TOTAL | 1.950 | %19 | 4.520 | %43 | 4.016 | %38 | 10.486 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 687,8 ; dof= 4.*

**Cross: …invisible symptoms such as pain, dizziness, headaches, fatigue, etc. / …neglected, not taken seriously and/or considered as psychological?**

| …INVISIBLE SYMPTOMS SUCH AS PAIN, DIZZINESS, HEADACHES, FATIGUE, ETC. | …NEGLECTED, NOT TAKEN SERIOUSLY AND/OR CONSIDERED AS PSYCHOLOGICAL? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 850 | %12 | 3.966 | %56 | 2.204 | %31 | 7.020 | %100 |
| No | 332 | %11 | 780 | %27 | 1.804 | %62 | 2.916 | %100 |
| Don't know | 64 | %12 | 188 | %34 | 298 | %54 | 550 | %100 |
| TOTAL | 1.246 | %12 | 4.934 | %47 | 4.306 | %41 | 10.486 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 898,9 ; dof= 4.*

**Cross: …invisible symptoms such as pain, dizziness, headaches, fatigue, etc. / Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed.**

| …INVISIBLE SYMPTOMS SUCH AS PAIN, DIZZINESS, HEADACHES, FATIGUE, ETC. | HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 1.793 | %26 | 3.932 | %56 | 1.295 | %18 | 7.020 | %100 |
| No | 743 | %25 | 899 | %31 | 1.274 | %44 | 2.916 | %100 |
| Don't know | 147 | %27 | 187 | %34 | 216 | %39 | 550 | %100 |
| TOTAL | 2.683 | %26 | 5.018 | %48 | 2.785 | %27 | 10.486 | %100 |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 826,2 ; dof= 4.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: …sudden onset symptoms requiring urgent care / …wrongly attributed to another physical disease?

| …SUDDEN ONSET SYMPTOMS REQUIRING URGENT CARE | …WRONGLY ATTRIBUTED TO ANOTHER PHYSICAL DISEASE? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 852 | %18 | 2.315 | %50 | 1.481 | %32 | 4.648 | %100 |
| No | 1.001 | %19 | 1.925 | %37 | 2.325 | %44 | 5.251 | %100 |
| Don't know | 97 | %17 | 280 | %48 | 210 | %36 | 587 | %100 |
| TOTAL | 1.950 | %19 | 4.520 | %43 | 4.016 | %38 | 10.486 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 204,3 ; dof= 4.*

## Cross: …sudden onset symptoms requiring urgent care / …neglected, not taken seriously and/or considered as psychological?

| …SUDDEN ONSET SYMPTOMS REQUIRING URGENT CARE | …NEGLECTED, NOT TAKEN SERIOUSLY AND/OR CONSIDERED AS PSYCHOLOGICAL? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 517 | %11 | 2.443 | %53 | 1.688 | %36 | 4.648 | %100 |
| No | 651 | %12 | 2.180 | %42 | 2.420 | %46 | 5.251 | %100 |
| Don't know | 78 | %13 | 311 | %53 | 198 | %34 | 587 | %100 |
| TOTAL | 1.246 | %12 | 4.934 | %47 | 4.306 | %41 | 10.486 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 138,4 ; dof= 4.*

## Cross: …sudden onset symptoms requiring urgent care / Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed.

| …SUDDEN ONSET SYMPTOMS REQUIRING URGENT CARE | HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 1.118 | %24 | 2.531 | %54 | 999 | %21 | 4.648 | %100 |
| No | 1.406 | %27 | 2.187 | %42 | 1.658 | %32 | 5.251 | %100 |
| Don't know | 159 | %27 | 300 | %51 | 128 | %22 | 587 | %100 |
| TOTAL | 2.683 | %26 | 5.018 | %48 | 2.785 | %27 | 10.486 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 193,2 ; dof= 4.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

# Chapter 4.

## Prevention

Rare
Barometer

A EURORDIS INITIATIVE

EURORDIS
RARE DISEASES EUROPE

**Only respondents living with a diagnosed rare disease**

## Cross: Typology of countries based on size and welfare / The rare disease was diagnosed before birth

| TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE | THE RARE DISEASE WAS DIAGNOSED BEFORE BIRTH | | | | | |
| | YES | | NO | | TOTAL | |
| | N | % | N | % | N | % |
|---|---|---|---|---|---|---|
| Group A ('Eastern Europe') | 23 | %1 | 1.589 | %99 | 1.612 | %100 |
| Group B ('Western Europe') | 153 | %3 | 4.664 | %97 | 4.817 | %100 |
| Group C ('Northern Europe') | 39 | %1 | 2.973 | %99 | 3.012 | %100 |
| TOTAL | 215 | %2 | 9.226 | %98 | 9.441 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 35,8 ; dof= 2.*

## Cross: Typology of countries based on size and welfare / The rare disease was diagnosed through standard tests carried out at birth

| TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE | THE RARE DISEASE WAS DIAGNOSED THROUGH STANDARD TESTS CARRIED OUT AT BIRTH | | | | | |
| | YES | | NO | | TOTAL | |
| | N | % | N | % | N | % |
|---|---|---|---|---|---|---|
| Group A ('Eastern Europe') | 155 | %10 | 1.457 | %90 | 1.612 | %100 |
| Group B ('Western Europe') | 336 | %7 | 4.481 | %93 | 4.817 | %100 |
| Group C ('Northern Europe') | 93 | %3 | 2.919 | %97 | 3.012 | %100 |
| TOTAL | 584 | %6 | 8.857 | %94 | 9.441 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 87,7 ; dof= 2.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Only respondents living with a diagnosed rare disease**

### Cross: Family members were previously diagnosed with the same disease / The rare disease was diagnosed before birth

| FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | THE RARE DISEASE WAS DIAGNOSED BEFORE BIRTH | | | | | |
| --- | --- | --- | --- | --- | --- | --- |
| | YES | | NO | | TOTAL | |
| | N | % | N | % | N | % |
| Yes | 39 | %3 | 1.270 | %97 | 1.309 | %100 |
| No | 183 | %2 | 8.239 | %98 | 8.422 | %100 |
| TOTAL | 222 | %2 | 9.509 | %98 | 9.731 | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is weakly significant. p-value= 0,1 ; Chi2= 3,3 ; dof= 1.*

### Cross: Family members were previously diagnosed with the same disease / The rare disease was diagnosed through standard tests carried out at birth

| FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | THE RARE DISEASE WAS DIAGNOSED THROUGH STANDARD TESTS CARRIED OUT AT BIRTH | | | | | |
| --- | --- | --- | --- | --- | --- | --- |
| | YES | | NO | | TOTAL | |
| | N | % | N | % | N | % |
| Yes | 38 | %3 | 1.249 | %97 | 1.287 | %100 |
| No | 358 | %4 | 7.887 | %96 | 8.245 | %100 |
| TOTAL | 396 | %4 | 9.136 | %96 | 9.532 | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is significant. p-value= 0,0 ; Chi2= 5,4 ; dof= 1.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Only respondents living with a diagnosed rare disease**

### Cross: The rare disease was diagnosed before birth / How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?

| THE RARE DISEASE WAS DIAGNOSED BEFORE BIRTH | HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | | | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | 0 | | 1 | | BETWEEN 2 AND 4 | | BETWEEN 5 AND 7 | | BETWEEN 8 AND 10 | | MORE THAN 10 | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 5 | %2 | 34 | %15 | 112 | %50 | 39 | %18 | 11 | %5 | 21 | %9 | 222 | %100 |
| No | 141 | %1 | 1.056 | %11 | 4.222 | %44 | 1.846 | %19 | 717 | %8 | 1.527 | %16 | 9.509 | %100 |
| TOTAL | 146 | %2 | 1.090 | %11 | 4.334 | %45 | 1.885 | %19 | 728 | %7 | 1.548 | %16 | 9.731 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is significant. p-value= 0,0 ; Chi2= 14,3 ; dof= 5.*

### Cross: The rare disease was diagnosed through standard tests carried out at birth / How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?

| THE RARE DISEASE WAS DIAGNOSED THROUGH STANDARD TESTS CARRIED OUT AT BIRTH | HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | | | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | 0 | | 1 | | BETWEEN 2 AND 4 | | BETWEEN 5 AND 7 | | BETWEEN 8 AND 10 | | MORE THAN 10 | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 22 | %6 | 91 | %23 | 191 | %48 | 47 | %12 | 10 | %3 | 35 | %9 | 396 | %100 |
| No | 116 | %1 | 964 | %11 | 4.050 | %44 | 1.808 | %20 | 712 | %8 | 1.489 | %16 | 9.139 | %100 |
| TOTAL | 138 | %1 | 1.055 | %11 | 4.241 | %44 | 1.855 | %19 | 722 | %8 | 1.524 | %16 | 9.535 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 141,8 ; dof= 5.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

**Only respondents living with a diagnosed rare disease**

## Cross: The rare disease was diagnosed before birth / I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases

| THE RARE DISEASE WAS DIAGNOSED BEFORE BIRTH | I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | | | | | |
|---|---|---|---|---|---|---|
| | YES | | NO | | TOTAL | |
| | N | % | N | % | N | % |
| Yes | 146 | %66 | 75 | %34 | 221 | %100 |
| No | 5.429 | %57 | 4.014 | %43 | 9.443 | %100 |
| TOTAL | 5.575 | %58 | 4.089 | %42 | 9.664 | |

🟥 *Under-represented elements* 🟦 *Over-represented elements*

*The relationship is significant. p-value= 0,0 ; Chi2= 6,5 ; dof= 1.*

## Cross: The rare disease was diagnosed through standard tests carried out at birth / I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases

| THE RARE DISEASE WAS DIAGNOSED THROUGH STANDARD TESTS CARRIED OUT AT BIRTH | I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | | | | | |
|---|---|---|---|---|---|---|
| | YES | | NO | | TOTAL | |
| | N | % | N | % | N | % |
| Yes | 271 | %69 | 120 | %31 | 391 | %100 |
| No | 5.185 | %57 | 3.893 | %43 | 9.078 | %100 |
| TOTAL | 5.456 | %58 | 4.013 | %42 | 9.469 | |

🟥 *Under-represented elements* 🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 22,8 ; dof= 1.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Only respondents living with a diagnosed rare disease**

## Cross: The rare disease was diagnosed before birth / …psychological support

| THE RARE DISEASE WAS DIAGNOSED BEFORE BIRTH | …PSYCHOLOGICAL SUPPORT | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 27 | %12 | 21 | %9 | 31 | %14 | 47 | %21 | 96 | %43 | 222 | %100 |
| No | 816 | %9 | 873 | %9 | 821 | %9 | 2.949 | %31 | 4.050 | %43 | 9.509 | %100 |
| TOTAL | 843 | %9 | 894 | %9 | 852 | %9 | 2.996 | %31 | 4.146 | %43 | 9.731 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 17,1 ; dof= 4.*

## Cross: The rare disease was diagnosed through standard tests carried out at birth / …psychological support

| THE RARE DISEASE WAS DIAGNOSED THROUGH STANDARD TESTS CARRIED OUT AT BIRTH | …PSYCHOLOGICAL SUPPORT | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 54 | %14 | 51 | %13 | 44 | %11 | 90 | %23 | 157 | %40 | 396 | %100 |
| No | 762 | %8 | 829 | %9 | 783 | %9 | 2.869 | %31 | 3.896 | %43 | 9.139 | %100 |
| TOTAL | 816 | %9 | 880 | %9 | 827 | %9 | 2.959 | %31 | 4.053 | %43 | 9.535 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 31,2 ; dof= 4.*

**Only respondents living with a diagnosed rare disease**

### Cross: The rare disease was diagnosed before birth / …care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc.

| THE RARE DISEASE WAS DIAGNOSED BEFORE BIRTH | …CARE COORDINATION SUPPORT SUCH AS HELP TO FIND THE NECESSARY INFORMATION ON THE DISEASE AND THE RIGHT PROFESSIONALS, ARRANGING APPOINTMENTS WITH DIFFERENT HEALTH PROVIDERS, ETC. | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 59 | %27 | 11 | %5 | 37 | %17 | 29 | %13 | 86 | %39 | 222 | %100 |
| No | 1.927 | %20 | 354 | %4 | 1.309 | %14 | 1.502 | %16 | 4.417 | %46 | 9.509 | %100 |
| **TOTAL** | **1.986** | **%20** | **365** | **%4** | **1.346** | **%14** | **1.531** | **%16** | **4.503** | **%46** | **9.731** | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is significant. p-value= 0,0 ; Chi2= 10,2 ; dof= 4.*

### Cross: The rare disease was diagnosed through standard tests carried out at birth / …care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc.

| THE RARE DISEASE WAS DIAGNOSED THROUGH STANDARD TESTS CARRIED OUT AT BIRTH | …CARE COORDINATION SUPPORT SUCH AS HELP TO FIND THE NECESSARY INFORMATION ON THE DISEASE AND THE RIGHT PROFESSIONALS, ARRANGING APPOINTMENTS WITH DIFFERENT HEALTH PROVIDERS, ETC. | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 129 | %33 | 19 | %5 | 61 | %15 | 48 | %12 | 139 | %35 | 396 | %100 |
| No | 1.805 | %20 | 333 | %4 | 1.250 | %14 | 1.458 | %16 | 4.293 | %47 | 9.139 | %100 |
| **TOTAL** | **1.934** | **%20** | **352** | **%4** | **1.311** | **%14** | **1.506** | **%16** | **4.432** | **%46** | **9.535** | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 48,0 ; dof= 4.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

**Only respondents living with a diagnosed rare disease**

## Cross: The rare disease was diagnosed before birth / …financial support including social security benefits

| THE RARE DISEASE WAS DIAGNOSED BEFORE BIRTH | …FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 39 | %18 | 7 | %3 | 37 | %17 | 61 | %28 | 77 | %35 | **221** | **%100** |
| No | 1.278 | %14 | 216 | %2 | 1.058 | %11 | 3.310 | %35 | 3.581 | %38 | **9.443** | **%100** |
| **TOTAL** | **1.317** | **%14** | **223** | **%2** | **1.095** | **%11** | **3.371** | **%35** | **3.658** | **%38** | **9.664** | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is significant. p-value= 0,0 ; Chi2= 13,2 ; dof= 4.*

## Cross: The rare disease was diagnosed through standard tests carried out at birth / …financial support including social security benefits

| THE RARE DISEASE WAS DIAGNOSED THROUGH STANDARD TESTS CARRIED OUT AT BIRTH | …FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 88 | %23 | 21 | %5 | 54 | %14 | 93 | %24 | 135 | %35 | **391** | **%100** |
| No | 1.202 | %13 | 193 | %2 | 1.016 | %11 | 3.237 | %36 | 3.430 | %38 | **9.078** | **%100** |
| **TOTAL** | **1.290** | **%14** | **214** | **%2** | **1.070** | **%11** | **3.330** | **%35** | **3.565** | **%38** | **9.469** | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 59,4 ; dof= 4.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Only respondents living with a diagnosed rare disease**

**Cross: The rare disease was diagnosed before birth / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?**

| THE RARE DISEASE WAS DIAGNOSED BEFORE BIRTH | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY... | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 131 | %59 | 95 | %43 | 6 | %3 | 2 | %1 | 23 | %10 | 15 | %7 | 7 | %3 | **222** | |
| No | 4.953 | %52 | 4.623 | %49 | 406 | %4 | 155 | %2 | 1.052 | %11 | 485 | %5 | 464 | %5 | **9.509** | |
| **TOTAL** | **5.084** | **%52** | **4.718** | **%48** | **412** | **%4** | **157** | **%2** | **1.075** | **%11** | **500** | **%5** | **471** | **%5** | **9.731** | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is not significant. p-value= 0,2 ; Chi2= 8,1 ; dof= 6.*

**Cross: The rare disease was diagnosed through standard tests carried out at birth / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?**

| THE RARE DISEASE WAS DIAGNOSED THROUGH STANDARD TESTS CARRIED OUT AT BIRTH | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY... | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 243 | %61 | 166 | %42 | 18 | %5 | 5 | %1 | 33 | %8 | 19 | %5 | 16 | %4 | **396** | |
| No | 4.736 | %52 | 4.473 | %49 | 383 | %4 | 148 | %2 | 1.013 | %11 | 469 | %5 | 445 | %5 | **9.139** | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

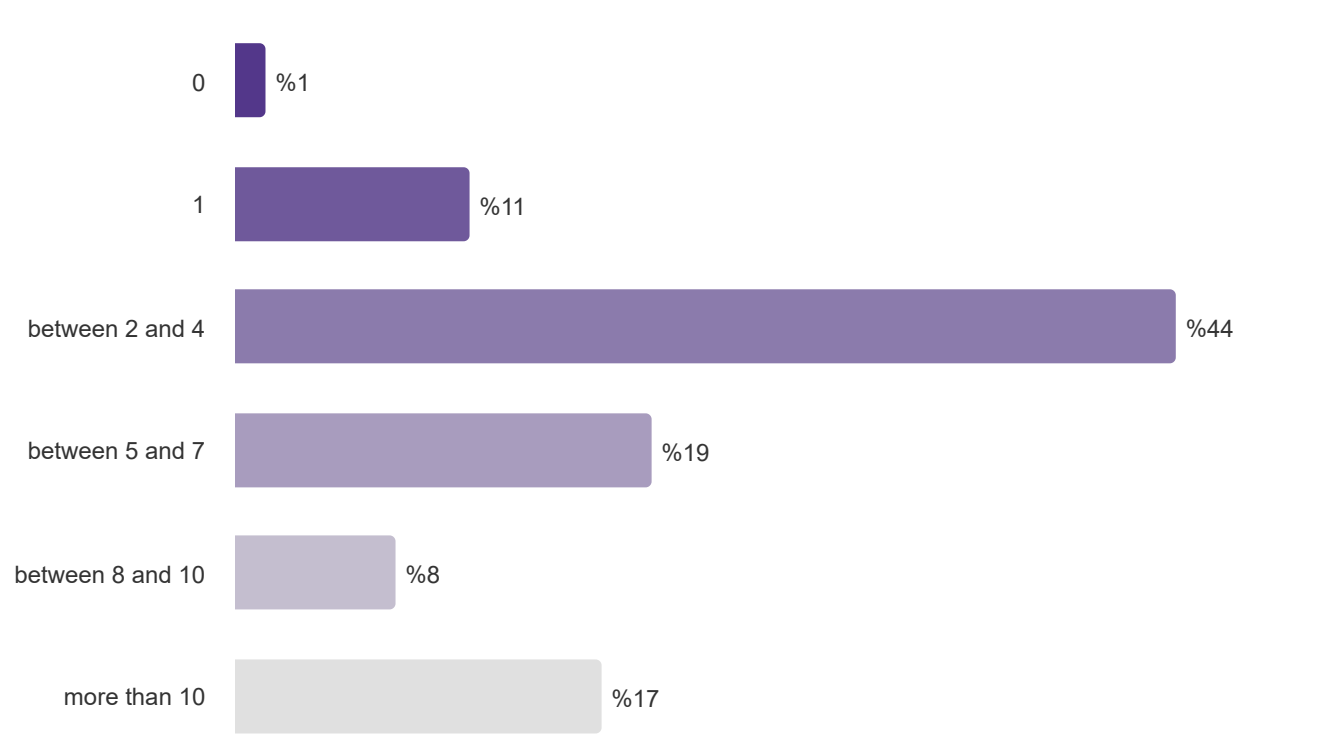*The relationship is significant. p-value= 0,0 ; Chi2= 14,1 ; dof= 6.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

# Chapter 4.

## Prevention

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

## How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?

|  | N |
| --- | --- |
| 0 | 155 |
| 1 | 1.150 |
| between 2 and 4 | 4.569 |
| between 5 and 7 | 2.033 |
| between 8 and 10 | 791 |
| more than 10 | 1.788 |
| **TOTAL** | **10.486** |

## How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?

| | |
| --- | --- |
| 0 | %1 |
| 1 | %11 |
| between 2 and 4 | %44 |
| between 5 and 7 | %19 |
| between 8 and 10 | %8 |
| more than 10 | %17 |

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Multiple Cross**

| How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis? | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | **MEAN** | **N** | **MEAN** | **N** | **MEAN** | **N** | **MEAN** | **N** | **MEAN** | **N** |
| 0-1 | 1,3 | 846 | 2,4 | 815 | 3,1 | 532 | 1,0 | 924 | 2,5 | 824 |
| between 2 and 4 | 0,7 | 3.394 | 2,5 | 3.213 | 2,5 | 2.042 | 1,8 | 3.478 | 3,1 | 2.956 |
| between 5 and 7 | 0,3 | 1.600 | 3,1 | 1.501 | 3,7 | 806 | 3,3 | 1.563 | 4,4 | 1.276 |
| between 8 and 10 | 0,4 | 597 | 4,4 | 575 | 4,8 | 304 | 5,5 | 592 | 6,9 | 455 |
| more than 10 | -0,3 | 1.383 | 6,9 | 1.218 | 8,6 | 651 | 9,7 | 1.286 | 10,9 | 996 |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Fisher= 7,9.*

*Inter variance= 362,7. Intra variance= 45,8.*

**Mean** = average time, in number of years

**N** = number of respondents for which we have the average time

**Cross: Gender of the person affected by the rare disease / How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?**

| GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE | HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | 0-1 | | BETWEEN 2 AND 4 | | BETWEEN 5 AND 7 | | BETWEEN 8 AND 10 | | MORE THAN 10 | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Female | 747 | %11 | 2.821 | %42 | 1.315 | %20 | 532 | %8 | 1.244 | %19 | 6.659 | %100 |
| Male | 393 | %14 | 1.324 | %47 | 538 | %19 | 189 | %7 | 366 | %13 | 2.810 | %100 |
| Other | 16 | %16 | 45 | %45 | 15 | %15 | 6 | %6 | 19 | %19 | 101 | %100 |
| TOTAL | 1.156 | %12 | 4.190 | %44 | 1.868 | %20 | 727 | %8 | 1.629 | %17 | 9.570 | |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 67,3 ; dof= 8.*

**Cross: Point prevalence of the rare disease / How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?**

| POINT PREVALENCE OF THE RARE DISEASE | HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | 0-1 | | BETWEEN 2 AND 4 | | BETWEEN 5 AND 7 | | BETWEEN 8 AND 10 | | MORE THAN 10 | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 1-5 / 10 000 | 338 | %14 | 1.102 | %46 | 409 | %17 | 178 | %7 | 380 | %16 | 2.407 | %100 |
| 1-9 / 100 000 | 271 | %14 | 934 | %47 | 399 | %20 | 117 | %6 | 278 | %14 | 1.999 | %100 |
| 1-9 / 1 000 000 | 54 | %12 | 191 | %42 | 103 | %22 | 43 | %9 | 68 | %15 | 459 | %100 |
| <1 / 1 000 000 | 89 | %10 | 334 | %39 | 170 | %20 | 76 | %9 | 187 | %22 | 856 | %100 |
| TOTAL | 752 | %13 | 2.561 | %45 | 1.081 | %19 | 414 | %7 | 913 | %16 | 5.721 | |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 61,9 ; dof= 12.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable) / How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?

| AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | 0-1 | | BETWEEN 2 AND 4 | | BETWEEN 5 AND 7 | | BETWEEN 8 AND 10 | | MORE THAN 10 | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Less than 2 years old | 316 | %15 | 848 | %41 | 379 | %19 | 138 | %7 | 364 | %18 | 2.045 | %100 |
| 2 to less that 10 years old | 81 | %9 | 381 | %41 | 194 | %21 | 74 | %8 | 195 | %21 | 925 | %100 |
| 10 to less than 20 years old | 79 | %8 | 346 | %36 | 182 | %19 | 86 | %9 | 259 | %27 | 952 | %100 |
| 20 to less than 30 years old | 99 | %10 | 424 | %43 | 185 | %19 | 79 | %8 | 191 | %20 | 978 | %100 |
| 30 to less than 50 years old | 234 | %10 | 1.062 | %45 | 530 | %23 | 197 | %8 | 330 | %14 | 2.353 | %100 |
| 50 years old or more | 150 | %14 | 605 | %55 | 193 | %17 | 68 | %6 | 91 | %8 | 1.107 | %100 |
| **TOTAL** | 959 | %11 | 3.666 | %44 | 1.663 | %20 | 642 | %8 | 1.430 | %17 | 8.360 | |

🟣 *Under-represented elements*    🔵 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 257,8 ; dof= 20.*

## Cross: Genetic diseases / How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?

| GENETIC DISEASES | HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | 0-1 | | BETWEEN 2 AND 4 | | BETWEEN 5 AND 7 | | BETWEEN 8 AND 10 | | MORE THAN 10 | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Genetic diseases | 775 | %14 | 2.278 | %42 | 1.003 | %18 | 399 | %7 | 992 | %18 | 5.447 | %100 |
| Non Genetic diseases | 244 | %9 | 1.315 | %50 | 560 | %21 | 210 | %8 | 298 | %11 | 2.627 | %100 |
| **TOTAL** | 1.019 | %13 | 3.593 | %45 | 1.563 | %19 | 609 | %8 | 1.290 | %16 | 8.074 | |

🟣 *Under-represented elements*    🔵 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 122,4 ; dof= 4.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases / How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?**

| I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | 0-1 | | BETWEEN 2 AND 4 | | BETWEEN 5 AND 7 | | BETWEEN 8 AND 10 | | MORE THAN 10 | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 802 | %13 | 2.812 | %47 | 1.085 | %18 | 408 | %7 | 891 | %15 | 5.998 | %100 |
| No | 497 | %11 | 1.731 | %39 | 933 | %21 | 372 | %8 | 882 | %20 | 4.415 | %100 |
| Non-response | 6 | %8 | 26 | %36 | 15 | %21 | 11 | %15 | 15 | %21 | 73 | %100 |
| TOTAL | 1.305 | %12 | 4.569 | %44 | 2.033 | %19 | 791 | %8 | 1.788 | %17 | 10.486 | |

🟥 *Under-represented elements* 🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 111,9 ; dof= 8.*

**Cross: Family members were previously diagnosed with the same disease / How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?**

| FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | 0-1 | | BETWEEN 2 AND 4 | | BETWEEN 5 AND 7 | | BETWEEN 8 AND 10 | | MORE THAN 10 | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 309 | %24 | 604 | %46 | 184 | %14 | 63 | %5 | 149 | %11 | 1.309 | %100 |
| No | 927 | %11 | 3.730 | %44 | 1.701 | %20 | 665 | %8 | 1.399 | %17 | 8.422 | %100 |
| Non-response | 11 | %14 | 24 | %31 | 18 | %23 | 2 | %3 | 22 | %29 | 77 | %100 |
| TOTAL | 1.247 | %13 | 4.358 | %44 | 1.903 | %19 | 730 | %7 | 1.570 | %16 | 9.808 | |

🟥 *Under-represented elements* 🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 212,1 ; dof= 8.*

**Cross: …wrongly attributed to another physical disease? / How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?**

| …WRONGLY ATTRIBUTED TO ANOTHER PHYSICAL DISEASE? | HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | 0-1 | | BETWEEN 2 AND 4 | | BETWEEN 5 AND 7 | | BETWEEN 8 AND 10 | | MORE THAN 10 | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, one time | 181 | %9 | 1.092 | %56 | 398 | %20 | 134 | %7 | 145 | %7 | 1.950 | %100 |
| YES, several times | 186 | %4 | 1.386 | %31 | 1.084 | %24 | 504 | %11 | 1.360 | %30 | 4.520 | %100 |
| NO | 938 | %23 | 2.091 | %52 | 551 | %14 | 153 | %4 | 283 | %7 | 4.016 | %100 |
| TOTAL | 1.305 | %12 | 4.569 | %44 | 2.033 | %19 | 791 | %8 | 1.788 | %17 | 10.486 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 2.022,5 ; dof= 8.*

**Cross: …neglected, not taken seriously and/or considered as psychological? / How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?**

| …NEGLECTED, NOT TAKEN SERIOUSLY AND/OR CONSIDERED AS PSYCHOLOGICAL? | HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | 0-1 | | BETWEEN 2 AND 4 | | BETWEEN 5 AND 7 | | BETWEEN 8 AND 10 | | MORE THAN 10 | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, one time | 116 | %9 | 660 | %53 | 276 | %22 | 83 | %7 | 111 | %9 | 1.246 | %100 |
| YES, several times | 277 | %6 | 1.600 | %32 | 1.142 | %23 | 551 | %11 | 1.364 | %28 | 4.934 | %100 |
| NO | 912 | %21 | 2.309 | %54 | 615 | %14 | 157 | %4 | 313 | %7 | 4.306 | %100 |
| TOTAL | 1.305 | %12 | 4.569 | %44 | 2.033 | %19 | 791 | %8 | 1.788 | %17 | 10.486 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 1.612,0 ; dof= 8.*

**Cross: Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed. / How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?**

| HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | 0-1 | | BETWEEN 2 AND 4 | | BETWEEN 5 AND 7 | | BETWEEN 8 AND 10 | | MORE THAN 10 | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, one time | **304** | **%11** | **1.402** | **%52** | 529 | %20 | 186 | %7 | **262** | **%10** | 2.683 | %100 |
| YES, several times | **229** | **%5** | **1.683** | **%34** | **1.184** | **%24** | **529** | **%11** | **1.393** | **%28** | 5.018 | %100 |
| NO | **772** | **%28** | **1.484** | **%53** | **320** | **%11** | **76** | **%3** | **133** | **%5** | 2.785 | %100 |
| TOTAL | 1.305 | %12 | 4.569 | %44 | 2.033 | %19 | 791 | %8 | 1.788 | %17 | 10.486 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 1.947,3 ; dof= 8.*

**Cross: How old were you when you stopped full-time education? / How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?**

| HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION? | HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | 0-1 | | BETWEEN 2 AND 4 | | BETWEEN 5 AND 7 | | BETWEEN 8 AND 10 | | MORE THAN 10 | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 15 y.o. or under | 50 | %11 | 214 | %47 | 77 | %17 | 33 | %7 | 81 | %18 | 455 | %100 |
| between 16 and 19 y.o. | 312 | %13 | **1.135** | **%46** | 449 | %18 | 181 | %7 | 387 | %16 | 2.464 | %100 |
| between 20 and 23 y.o. | 362 | %12 | 1.337 | %44 | **627** | **%21** | 227 | %8 | **469** | **%16** | 3.022 | %100 |
| 24 y.o. or above | 374 | %12 | **1.303** | **%41** | 622 | %20 | 246 | %8 | **600** | **%19** | 3.145 | %100 |
| TOTAL | 1.098 | %12 | 3.989 | %44 | 1.775 | %20 | 687 | %8 | 1.537 | %17 | 9.086 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 30,5 ; dof= 12.*

## Cross: How would you best describe yourself? / How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?

| HOW WOULD YOU BEST DESCRIBE YOURSELF? | 0-1 | | BETWEEN 2 AND 4 | | BETWEEN 5 AND 7 | | BETWEEN 8 AND 10 | | MORE THAN 10 | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % | N | % | N | % |
| I belong to the ethnic majority in the country where I live | 835 | %12 | 3.062 | %43 | 1.419 | %20 | 550 | %8 | 1.259 | %18 | 7.125 | %100 |
| I am part of an ethnic minority in the country where I live | 57 | %12 | 207 | %45 | 82 | %18 | 31 | %7 | 88 | %19 | 465 | %100 |
| Other, specify... | 43 | %13 | 143 | %42 | 68 | %20 | 25 | %7 | 58 | %17 | 337 | %100 |
| TOTAL | 935 | %12 | 3.412 | %43 | 1.569 | %20 | 606 | %8 | 1.405 | %18 | 7.927 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is not significant. p-value= 0,9 ; Chi2= 2,9 ; dof= 8.*

## Cross: Disease complexity classified into five groups, based on the number of affected body parts. / How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | 0-1 | | BETWEEN 2 AND 4 | | BETWEEN 5 AND 7 | | BETWEEN 8 AND 10 | | MORE THAN 10 | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 1-3 body parts | 863 | %14 | 3.067 | %50 | 1.113 | %18 | 406 | %7 | 654 | %11 | 6.103 | %100 |
| 4-7 body parts | 345 | %11 | 1.171 | %38 | 700 | %23 | 258 | %8 | 607 | %20 | 3.081 | %100 |
| 8-11 body parts | 83 | %9 | 269 | %28 | 174 | %18 | 93 | %10 | 332 | %35 | 951 | %100 |
| 12-15 body parts | 12 | %4 | 54 | %19 | 38 | %13 | 27 | %9 | 155 | %54 | 286 | %100 |
| 16 body parts or more | 2 | %3 | 8 | %12 | 8 | %12 | 7 | %11 | 40 | %62 | 65 | %100 |
| TOTAL | 1.305 | %12 | 4.569 | %44 | 2.033 | %19 | 791 | %8 | 1.788 | %17 | 10.486 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 927,2 ; dof= 16.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Please select the sentence that best describes your situation or the situation of the person you care for: / How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?**

| PLEASE SELECT THE SENTENCE THAT BEST DESCRIBES YOUR SITUATION OR THE SITUATION OF THE PERSON YOU CARE FOR: | HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | 0-1 | | BETWEEN 2 AND 4 | | BETWEEN 5 AND 7 | | BETWEEN 8 AND 10 | | MORE THAN 10 | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| I know the NAME of the rare disease, syndrome or malformation and it has been CONFIRMED by appropriate genetic, clinical, medical imaging, molecular or biochemical tests (e.g biopsy, blood or urine test) | 1.182 | %13 | 4.122 | %46 | 1.731 | %19 | 645 | %7 | 1.368 | %15 | 9.048 | %100 |
| I know the NAME of the rare disease, syndrome or malformation but it has NOT yet been confirmed by appropriate genetic, clinical, medical imaging, molecular or biochemical tests | 65 | %9 | 236 | %31 | 172 | %23 | 85 | %11 | 202 | %27 | 760 | %100 |
| I only have PARTIAL information on the name of the rare disease or the gene involved or the type of disease | 25 | %8 | 94 | %31 | 61 | %20 | 28 | %9 | 98 | %32 | 306 | %100 |
| I know that the disease is rare but the name or the cause have NOT BEEN IDENTIFIED | 27 | %8 | 109 | %31 | 68 | %20 | 30 | %9 | 114 | %33 | 348 | %100 |
| Other, specify... | 6 | %25 | 8 | %33 | 1 | %4 | 3 | %13 | 6 | %25 | 24 | %100 |
| **TOTAL** | **1.305** | **%12** | **4.569** | **%44** | **2.033** | **%19** | **791** | **%8** | **1.788** | **%17** | **10.486** | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 262,3 ; dof= 16.*

**Cross: Orphacode associated nomenclature (english) / How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?**

| ORPHACODE ASSOCIATED NOMENCLATURE (ENGLISH) | HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | | | | | | | | | | | |
| | 0-1 | | BETWEEN 2 AND 4 | | BETWEEN 5 AND 7 | | BETWEEN 8 AND 10 | | MORE THAN 10 | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Hereditary hemorrhagic telangiectasia | 118 | %26 | 230 | %50 | 53 | %12 | 22 | %5 | 35 | %8 | 458 | %100 |
| Hypermobile Ehlers-Danlos syndrome | 7 | %2 | 44 | %14 | 55 | %17 | 39 | %12 | 172 | %54 | 317 | %100 |
| Sarcoidosis | 20 | %12 | 86 | %51 | 37 | %22 | 10 | %6 | 17 | %10 | 170 | %100 |
| Classical Ehlers-Danlos syndrome | 5 | %4 | 33 | %24 | 25 | %18 | 11 | %8 | 63 | %46 | 137 | %100 |
| Williams syndrome | 35 | %26 | 57 | %42 | 27 | %20 | 5 | %4 | 12 | %9 | 136 | %100 |
| Cystic fibrosis | 34 | %27 | 62 | %48 | 22 | %17 | 3 | %2 | 7 | %5 | 128 | %100 |
| Myasthenia gravis | 10 | %8 | 67 | %56 | 20 | %17 | 8 | %7 | 15 | %13 | 120 | %100 |
| Systemic sclerosis | 13 | %12 | 66 | %62 | 17 | %16 | 6 | %6 | 5 | %5 | 107 | %100 |
| Tuberous sclerosis complex | 19 | %19 | 50 | %51 | 19 | %19 | 4 | %4 | 6 | %6 | 98 | %100 |
| Neurofibromatosis type 1 | 15 | %16 | 49 | %53 | 15 | %16 | 6 | %7 | 7 | %8 | 92 | %100 |
| Interstitial cystitis | 3 | %4 | 22 | %30 | 25 | %34 | 12 | %16 | 12 | %16 | 74 | %100 |
| Addison disease | 6 | %8 | 35 | %48 | 18 | %25 | 5 | %7 | 9 | %12 | 73 | %100 |
| 22q11.2 deletion syndrome | 17 | %25 | 26 | %38 | 13 | %19 | 6 | %9 | 6 | %9 | 68 | %100 |
| Chronic inflammatory demyelinating polyneuropathy | 9 | %14 | 34 | %52 | 11 | %17 | 3 | %5 | 8 | %12 | 65 | %100 |
| Perineural cyst | 5 | %8 | 22 | %35 | 18 | %29 | 6 | %10 | 12 | %19 | 63 | %100 |
| Acute inflammatory demyelinating polyradiculoneuropathy | 2 | %3 | 36 | %58 | 14 | %23 | 4 | %6 | 6 | %10 | 62 | %100 |
| Rett syndrome | 4 | %7 | 26 | %43 | 12 | %20 | 5 | %8 | 13 | %22 | 60 | %100 |
| Marfan syndrome | 8 | %15 | 16 | %31 | 10 | %19 | 7 | %13 | 11 | %21 | 52 | %100 |
| Fragile X syndrome | 4 | %8 | 23 | %47 | 12 | %24 | 6 | %12 | 4 | %8 | 49 | %100 |
| BehÃ§et disease | 1 | %2 | 14 | %30 | 15 | %32 | 5 | %11 | 12 | %26 | 47 | %100 |

🟥 *Under-represented elements*　　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 8.482,9 ; dof= 6.700.*

**Cross: Orphanet classification of rare diseases (one disease can be classified in several categories) / How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?**

| ORPHANET CLASSIFICATION OF RARE DISEASES (ONE DISEASE CAN BE CLASSIFIED IN SEVERAL CATEGORIES) | HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | 0-1 | | BETWEEN 2 AND 4 | | BETWEEN 5 AND 7 | | BETWEEN 8 AND 10 | | MORE THAN 10 | | | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Abdominal surgical diseases | 27 | %11 | 77 | %32 | 39 | %16 | 19 | %8 | 77 | %32 | 239 | %100 |
| Allergic diseases | 0 | %0 | 1 | %33 | 2 | %67 | 0 | %0 | 0 | %0 | 3 | %100 |
| Bone diseases | 140 | %18 | 310 | %39 | 145 | %18 | 59 | %7 | 145 | %18 | 799 | %100 |
| Cardiac diseases | 88 | %13 | 348 | %53 | 119 | %18 | 34 | %5 | 71 | %11 | 660 | %100 |
| Cardiac malformations | 63 | %21 | 128 | %43 | 49 | %17 | 18 | %6 | 37 | %13 | 295 | %100 |
| Circulatory system diseases | 230 | %17 | 616 | %46 | 239 | %18 | 102 | %8 | 164 | %12 | 1.351 | %100 |
| Clinical sign | 0 | %0 | 0 | %0 | 0 | %0 | 0 | %0 | 0 | %0 | 0 | %100 |
| Developmental anomalies during embryogenesis | 479 | %14 | 1.310 | %39 | 602 | %18 | 265 | %8 | 691 | %21 | 3.347 | %100 |
| Diseases due to toxic effects | 0 | %0 | 1 | %33 | 1 | %33 | 0 | %0 | 1 | %33 | 3 | %100 |
| Endocrine diseases | 124 | %12 | 455 | %46 | 217 | %22 | 75 | %8 | 124 | %12 | 995 | %100 |
| Gastroenterological diseases | 57 | %19 | 147 | %48 | 50 | %16 | 20 | %7 | 31 | %10 | 305 | %100 |
| Genetic diseases | 775 | %14 | 2.278 | %42 | 1.003 | %18 | 399 | %7 | 992 | %18 | 5.447 | %100 |
| Gynecologic/obstetric diseases | 49 | %17 | 112 | %39 | 55 | %19 | 24 | %8 | 44 | %15 | 284 | %100 |
| Hematological diseases | 70 | %17 | 190 | %46 | 77 | %19 | 31 | %8 | 44 | %11 | 412 | %100 |
| Hepatic diseases | 207 | %23 | 446 | %50 | 124 | %14 | 39 | %4 | 75 | %8 | 891 | %100 |
| Immunological diseases | 33 | %12 | 106 | %37 | 51 | %18 | 25 | %9 | 71 | %25 | 286 | %100 |
| Inborn errors of metabolism | 110 | %14 | 338 | %44 | 150 | %19 | 53 | %7 | 123 | %16 | 774 | %100 |
| Infectious diseases | 3 | %18 | 5 | %29 | 4 | %24 | 1 | %6 | 4 | %24 | 17 | %100 |
| Infertility | 76 | %19 | 183 | %45 | 76 | %19 | 25 | %6 | 50 | %12 | 410 | %100 |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 1.122,8 ; dof= 136.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: …behavioural disorders that cause problems in school, at home or in social situations / How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?**

| | HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| …BEHAVIOURAL DISORDERS THAT CAUSE PROBLEMS IN SCHOOL, AT HOME OR IN SOCIAL SITUATIONS | 0-1 | | BETWEEN 2 AND 4 | | BETWEEN 5 AND 7 | | BETWEEN 8 AND 10 | | MORE THAN 10 | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 293 | %10 | 1.161 | %39 | 642 | %22 | 253 | %9 | 608 | %21 | 2.957 | %100 |
| No | 948 | %13 | 3.218 | %45 | 1.314 | %19 | 507 | %7 | 1.098 | %15 | 7.085 | %100 |
| Don't know | 64 | %14 | 190 | %43 | 77 | %17 | 31 | %7 | 82 | %18 | 444 | %100 |
| TOTAL | 1.305 | %12 | 4.569 | %44 | 2.033 | %19 | 791 | %8 | 1.788 | %17 | 10.486 | |

🟪 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 89,2 ; dof= 8.*

**Cross: …intellectual disabilities or cognitive symptoms (i.e. problems with memory, language, thinking or judgement) / How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?**

| | HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| …INTELLECTUAL DISABILITIES OR COGNITIVE SYMPTOMS (I.E. PROBLEMS WITH MEMORY, LANGUAGE, THINKING OR JUDGEMENT) | 0-1 | | BETWEEN 2 AND 4 | | BETWEEN 5 AND 7 | | BETWEEN 8 AND 10 | | MORE THAN 10 | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 289 | %10 | 1.104 | %38 | 634 | %22 | 274 | %9 | 635 | %22 | 2.936 | %100 |
| No | 982 | %14 | 3.335 | %46 | 1.334 | %18 | 494 | %7 | 1.091 | %15 | 7.236 | %100 |
| Don't know | 34 | %11 | 130 | %41 | 65 | %21 | 23 | %7 | 62 | %20 | 314 | %100 |
| TOTAL | 1.305 | %12 | 4.569 | %44 | 2.033 | %19 | 791 | %8 | 1.788 | %17 | 10.486 | |

🟪 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 141,2 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: …clinical signs or symptoms that come and go / How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?

| …CLINICAL SIGNS OR SYMPTOMS THAT COME AND GO | HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | 0-1 | | BETWEEN 2 AND 4 | | BETWEEN 5 AND 7 | | BETWEEN 8 AND 10 | | MORE THAN 10 | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 569 | %10 | 2.378 | %40 | 1.230 | %21 | 533 | %9 | 1.230 | %21 | 5.940 | %100 |
| No | 624 | %16 | 1.834 | %48 | 666 | %18 | 214 | %6 | 450 | %12 | 3.788 | %100 |
| Don't know | 112 | %15 | 357 | %47 | 137 | %18 | 44 | %6 | 108 | %14 | 758 | %100 |
| TOTAL | 1.305 | %12 | 4.569 | %44 | 2.033 | %19 | 791 | %8 | 1.788 | %17 | 10.486 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 290,5 ; dof= 8.*

## Cross: …invisible symptoms such as pain, dizziness, headaches, fatigue, etc. / How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?

| …INVISIBLE SYMPTOMS SUCH AS PAIN, DIZZINESS, HEADACHES, FATIGUE, ETC. | HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | 0-1 | | BETWEEN 2 AND 4 | | BETWEEN 5 AND 7 | | BETWEEN 8 AND 10 | | MORE THAN 10 | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 684 | %10 | 2.878 | %41 | 1.432 | %20 | 608 | %9 | 1.418 | %20 | 7.020 | %100 |
| No | 545 | %19 | 1.454 | %50 | 478 | %16 | 148 | %5 | 291 | %10 | 2.916 | %100 |
| Don't know | 76 | %14 | 237 | %43 | 123 | %22 | 35 | %6 | 79 | %14 | 550 | %100 |
| TOTAL | 1.305 | %12 | 4.569 | %44 | 2.033 | %19 | 791 | %8 | 1.788 | %17 | 10.486 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 355,1 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

### Cross: …sudden onset symptoms requiring urgent care / How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?

| …SUDDEN ONSET SYMPTOMS REQUIRING URGENT CARE | HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | 0-1 | | BETWEEN 2 AND 4 | | BETWEEN 5 AND 7 | | BETWEEN 8 AND 10 | | MORE THAN 10 | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 484 | %10 | 1.857 | %40 | 957 | %21 | 398 | %9 | 952 | %20 | 4.648 | %100 |
| No | 751 | %14 | 2.469 | %47 | 949 | %18 | 353 | %7 | 729 | %14 | 5.251 | %100 |
| Don't know | 70 | %12 | 243 | %41 | 127 | %22 | 40 | %7 | 107 | %18 | 587 | %100 |
| TOTAL | 1.305 | %12 | 4.569 | %44 | 2.033 | %19 | 791 | %8 | 1.788 | %17 | 10.486 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 143,7 ; dof= 8.*

### Cross: …healthcare professionals were reluctant or not sufficiently informed? / How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?

Have you ever needed a genetic test but could not access it because...

| …HEALTHCARE PROFESSIONALS WERE RELUCTANT OR NOT SUFFICIENTLY INFORMED? | HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | 0-1 | | BETWEEN 2 AND 4 | | BETWEEN 5 AND 7 | | BETWEEN 8 AND 10 | | MORE THAN 10 | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 155 | %6 | 853 | %30 | 611 | %22 | 287 | %10 | 899 | %32 | 2.805 | %100 |
| No | 896 | %16 | 2.702 | %49 | 1.012 | %18 | 338 | %6 | 608 | %11 | 5.556 | %100 |
| Not relevant | 254 | %12 | 1.014 | %48 | 410 | %19 | 166 | %8 | 281 | %13 | 2.125 | %100 |
| TOTAL | 1.305 | %12 | 4.569 | %44 | 2.033 | %19 | 791 | %8 | 1.788 | %17 | 10.486 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 886,3 ; dof= 8.*

**Cross: Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease? / How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?**

| ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | 0-1 | | BETWEEN 2 AND 4 | | BETWEEN 5 AND 7 | | BETWEEN 8 AND 10 | | MORE THAN 10 | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, through a patient organisation | 712 | %13 | 2.342 | %44 | 1.024 | %19 | 397 | %7 | 851 | %16 | 5.326 | %100 |
| YES, through online communities | 551 | %11 | 2.011 | %40 | 1.010 | %20 | 419 | %8 | 1.001 | %20 | 4.992 | %100 |
| YES, through local networks (e.g. schools) | 47 | %11 | 180 | %41 | 78 | %18 | 39 | %9 | 92 | %21 | 436 | %100 |
| NO, because of accessibility issues (e.g. language or technical barriers) | 21 | %11 | 77 | %41 | 35 | %18 | 15 | %8 | 42 | %22 | 190 | %100 |
| NO, because I have not been able to find other people with the same disease | 141 | %11 | 558 | %43 | 274 | %21 | 90 | %7 | 247 | %19 | 1.310 | %100 |
| NO, because I don't want to | 89 | %16 | 284 | %52 | 72 | %13 | 38 | %7 | 64 | %12 | 547 | %100 |
| Other, specify... | 81 | %16 | 225 | %44 | 93 | %18 | 40 | %8 | 75 | %15 | 514 | %100 |
| **TOTAL** | **1.305** | **%12** | **4.569** | **%44** | **2.033** | **%19** | **791** | **%8** | **1.788** | **%17** | **10.486** | |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 122,4 ; dof= 24.*

Rare Barometer
A EURORDIS INITIATIVE

EURORDIS
RARE DISEASES EUROPE

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Would you say that you, or the person you care for, live in a: / How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?**

| WOULD YOU SAY THAT YOU, OR THE PERSON YOU CARE FOR, LIVE IN A: | HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | 0-1 | | BETWEEN 2 AND 4 | | BETWEEN 5 AND 7 | | BETWEEN 8 AND 10 | | MORE THAN 10 | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Rural area or village | 287 | %12 | 1.102 | %46 | 476 | %20 | 179 | %7 | 372 | %15 | 2.416 | %100 |
| Small or mid size town | 458 | %12 | 1.730 | %44 | 770 | %20 | 288 | %7 | 655 | %17 | 3.901 | %100 |
| Large town | 353 | %13 | 1.151 | %42 | 528 | %19 | 219 | %8 | 509 | %18 | 2.760 | %100 |
| TOTAL | 1.098 | %12 | 3.983 | %44 | 1.774 | %20 | 686 | %8 | 1.536 | %17 | 9.077 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is weakly significant. p-value= 0,1 ; Chi2= 14,7 ; dof= 8.*

**Cross: Typology of countries based on size and welfare / How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?**

| TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE | HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | 0-1 | | BETWEEN 2 AND 4 | | BETWEEN 5 AND 7 | | BETWEEN 8 AND 10 | | MORE THAN 10 | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Group A ('Eastern Europe') | 184 | %10 | 813 | %45 | 356 | %20 | 134 | %7 | 307 | %17 | 1.794 | %100 |
| Group B ('Western Europe') | 682 | %13 | 2.323 | %46 | 988 | %19 | 377 | %7 | 735 | %14 | 5.105 | %100 |
| Group C ('Northern Europe') | 394 | %12 | 1.297 | %40 | 631 | %19 | 253 | %8 | 698 | %21 | 3.273 | %100 |
| TOTAL | 1.260 | %12 | 4.433 | %44 | 1.975 | %19 | 764 | %8 | 1.740 | %17 | 10.172 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 84,6 ; dof= 8.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

# Chapter 4.

## Prevention

Rare Barometer

A EURORDIS INITIATIVE

EURORDIS
RARE DISEASES EUROPE

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Gender of the person affected by the rare disease / I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases

| | I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | | | | | |
| | YES | | NO | | TOTAL | |
| **GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE** | N | % | N | % | N | % |
|---|---|---|---|---|---|---|
| Female | 3.729 | %56 | 2.904 | %44 | 6.633 | %100 |
| Male | 1.674 | %60 | 1.124 | %40 | 2.798 | %100 |
| Other | 59 | %58 | 42 | %42 | 101 | %100 |
| **TOTAL** | 5.462 | %57 | 4.070 | %43 | 9.532 | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

The relationship is very significant. p-value= < 0,01 ; Chi2= 10,5 ; dof= 2.

## Cross: How old were you when you stopped full-time education? / I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases

| | I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | | | | | |
| | YES | | NO | | TOTAL | |
| **HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION?** | N | % | N | % | N | % |
|---|---|---|---|---|---|---|
| 15 y.o. or under | 283 | %63 | 168 | %37 | 451 | %100 |
| between 16 and 19 y.o. | 1.368 | %56 | 1.092 | %44 | 2.460 | %100 |
| between 20 and 23 y.o. | 1.756 | %58 | 1.248 | %42 | 3.004 | %100 |
| 24 y.o. or above | 1.771 | %57 | 1.363 | %43 | 3.134 | %100 |
| **TOTAL** | 5.178 | %57 | 3.871 | %43 | 9.049 | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

The relationship is significant. p-value= 0,0 ; Chi2= 10,8 ; dof= 3.

**Cross: How would you best describe yourself? / I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases**

| | I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | | | | | |
| | YES | | NO | | TOTAL | |
| HOW WOULD YOU BEST DESCRIBE YOURSELF? | N | % | N | % | N | % |
|---|---|---|---|---|---|---|
| I belong to the ethnic majority in the country where I live | 3.966 | %56 | 3.123 | %44 | 7.089 | %100 |
| I am part of an ethnic minority in the country where I live | 263 | %57 | 201 | %43 | 464 | %100 |
| Other, specify... | 179 | %53 | 158 | %47 | 337 | %100 |
| TOTAL | 4.408 | %56 | 3.482 | %44 | 7.890 | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is not significant. p-value= 0,6 ; Chi2= 1,2 ; dof= 2.*

**Cross: Would you say that you, or the person you care for, live in a: / I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases**

| | I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | | | | | |
| | YES | | NO | | TOTAL | |
| WOULD YOU SAY THAT YOU, OR THE PERSON YOU CARE FOR, LIVE IN A: | N | % | N | % | N | % |
|---|---|---|---|---|---|---|
| Rural area or village | 1.378 | %57 | 1.031 | %43 | 2.409 | %100 |
| Small or mid size town | 2.253 | %58 | 1.628 | %42 | 3.881 | %100 |
| Large town | 1.543 | %56 | 1.207 | %44 | 2.750 | %100 |
| TOTAL | 5.174 | %57 | 3.866 | %43 | 9.040 | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is not significant. p-value= 0,3 ; Chi2= 2,5 ; dof= 2.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

### Cross: Typology of countries based on size and welfare / I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases

| TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE | I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | | | | | |
|---|---|---|---|---|---|---|
| | YES | | NO | | TOTAL | |
| | N | % | N | % | N | % |
| Group A ('Eastern Europe') | 960 | %54 | 833 | %46 | 1.793 | %100 |
| Group B ('Western Europe') | 2.863 | %56 | 2.207 | %44 | 5.070 | %100 |
| Group C ('Northern Europe') | 2.003 | %61 | 1.269 | %39 | 3.272 | %100 |
| TOTAL | 5.826 | %57 | 4.309 | %43 | 10.135 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 32,2 ; dof= 2.*

### Cross: Point prevalence of the rare disease / I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases

| POINT PREVALENCE OF THE RARE DISEASE | I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | | | | | |
|---|---|---|---|---|---|---|
| | YES | | NO | | TOTAL | |
| | N | % | N | % | N | % |
| 1-5 / 10 000 | 1.464 | %61 | 934 | %39 | 2.398 | %100 |
| 1-9 / 100 000 | 1.220 | %61 | 772 | %39 | 1.992 | %100 |
| 1-9 / 1 000 000 | 253 | %56 | 200 | %44 | 453 | %100 |
| <1 / 1 000 000 | 437 | %51 | 414 | %49 | 851 | %100 |
| TOTAL | 3.374 | %59 | 2.320 | %41 | 5.694 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 30,7 ; dof= 3.*

## Cross: Orphacode associated nomenclature (english) / I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases

| ORPHACODE ASSOCIATED NOMENCLATURE (ENGLISH) | I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | | | | | |
|---|---|---|---|---|---|---|
| | YES | | NO | | TOTAL | |
| | N | % | N | % | N | % |
| Hereditary hemorrhagic telangiectasia | 369 | %81 | 86 | %19 | 455 | %100 |
| Hypermobile Ehlers-Danlos syndrome | 146 | %46 | 170 | %54 | 316 | %100 |
| Sarcoidosis | 79 | %47 | 90 | %53 | 169 | %100 |
| Classical Ehlers-Danlos syndrome | 67 | %50 | 68 | %50 | 135 | %100 |
| Williams syndrome | 79 | %58 | 57 | %42 | 136 | %100 |
| Cystic fibrosis | 111 | %87 | 17 | %13 | 128 | %100 |
| Myasthenia gravis | 70 | %58 | 50 | %42 | 120 | %100 |
| Systemic sclerosis | 65 | %61 | 42 | %39 | 107 | %100 |
| Tuberous sclerosis complex | 62 | %64 | 35 | %36 | 97 | %100 |
| Neurofibromatosis type 1 | 68 | %74 | 24 | %26 | 92 | %100 |
| Interstitial cystitis | 36 | %49 | 38 | %51 | 74 | %100 |
| Addison disease | 35 | %48 | 38 | %52 | 73 | %100 |
| 22q11.2 deletion syndrome | 38 | %56 | 30 | %44 | 68 | %100 |
| Chronic inflammatory demyelinating polyneuropathy | 35 | %54 | 30 | %46 | 65 | %100 |
| Perineural cyst | 7 | %11 | 56 | %89 | 63 | %100 |
| Acute inflammatory demyelinating polyradiculoneuropathy | 29 | %47 | 33 | %53 | 62 | %100 |
| Rett syndrome | 37 | %62 | 23 | %38 | 60 | %100 |
| Marfan syndrome | 24 | %50 | 24 | %50 | 48 | %100 |
| Fragile X syndrome | 26 | %53 | 23 | %47 | 49 | %100 |
| Behçet disease | 30 | %64 | 17 | %36 | 47 | %100 |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 2.217,6 ; dof= 1.672.*

**Cross: Orphanet classification of rare diseases (one disease can be classified in several categories) / I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases**

| ORPHANET CLASSIFICATION OF RARE DISEASES (ONE DISEASE CAN BE CLASSIFIED IN SEVERAL CATEGORIES) | I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | | | | | |
|---|---|---|---|---|---|---|
| | YES | | NO | | TOTAL | |
| | N | % | N | % | N | % |
| Abdominal surgical diseases | 135 | %57 | 102 | %43 | 237 | %100 |
| Allergic diseases | 1 | %33 | 2 | %67 | 3 | %100 |
| Bone diseases | 441 | %56 | 351 | %44 | 792 | %100 |
| Cardiac diseases | 399 | %61 | 260 | %39 | 659 | %100 |
| Cardiac malformations | 181 | %61 | 114 | %39 | 295 | %100 |
| Circulatory system diseases | 904 | %67 | 436 | %33 | 1.340 | %100 |
| Clinical sign | 0 | %0 | 0 | %0 | 0 | %100 |
| Developmental anomalies during embryogenesis | 1.931 | %58 | 1.395 | %42 | 3.326 | %100 |
| Diseases due to toxic effects | 2 | %67 | 1 | %33 | 3 | %100 |
| Endocrine diseases | 551 | %56 | 435 | %44 | 986 | %100 |
| Gastroenterological diseases | 201 | %66 | 104 | %34 | 305 | %100 |
| Genetic diseases | 3.237 | %60 | 2.181 | %40 | 5.418 | %100 |
| Gynecologic/obstetric diseases | 161 | %58 | 118 | %42 | 279 | %100 |
| Hematological diseases | 259 | %63 | 151 | %37 | 410 | %100 |
| Hepatic diseases | 645 | %73 | 243 | %27 | 888 | %100 |
| Immunological diseases | 173 | %62 | 108 | %38 | 281 | %100 |
| Inborn errors of metabolism | 477 | %62 | 295 | %38 | 772 | %100 |
| Infectious diseases | 5 | %29 | 12 | %71 | 17 | %100 |
| Infertility | 292 | %72 | 116 | %28 | 408 | %100 |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 437,3 ; dof= 34.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Please select the sentence that best describes your situation or the situation of the person you care for: / I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases**

| PLEASE SELECT THE SENTENCE THAT BEST DESCRIBES YOUR SITUATION OR THE SITUATION OF THE PERSON YOU CARE FOR: | I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | | | | | |
|---|---|---|---|---|---|---|
| | YES | | NO | | TOTAL | |
| | N | % | N | % | N | % |
| I know the NAME of the rare disease, syndrome or malformation and it has been CONFIRMED by appropriate genetic, clinical, medical imaging, molecular or biochemical tests (e.g biopsy, blood or urine test) | 5.298 | %59 | 3.692 | %41 | 8.990 | %100 |
| I know the NAME of the rare disease, syndrome or malformation but it has NOT yet been confirmed by appropriate genetic, clinical, medical imaging, molecular or biochemical tests | 322 | %43 | 429 | %57 | 751 | %100 |
| I only have PARTIAL information on the name of the rare disease or the gene involved or the type of disease | 159 | %52 | 146 | %48 | 305 | %100 |
| I know that the disease is rare but the name or the cause have NOT BEEN IDENTIFIED | 206 | %60 | 139 | %40 | 345 | %100 |
| Other, specify... | 13 | %59 | 9 | %41 | 22 | %100 |
| TOTAL | 5.998 | %58 | 4.415 | %42 | 10.413 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 77,6 ; dof= 4.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Genetic diseases / I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases**

| GENETIC DISEASES | I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | | | | | |
| | YES | | NO | | TOTAL | |
| | N | % | N | % | N | % |
| Genetic diseases | 3.237 | %60 | 2.181 | %40 | 5.418 | %100 |
| Non Genetic diseases | 1.465 | %56 | 1.148 | %44 | 2.613 | %100 |
| **TOTAL** | **4.702** | **%59** | **3.329** | **%41** | **8.031** | **%100** |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 9,8 ; dof= 1.*

**Cross: Disease complexity classified into five groups, based on the number of affected body parts. / I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases**

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | | | | | |
| | YES | | NO | | TOTAL | |
| | N | % | N | % | N | % |
| 1-3 body parts | 3.515 | %58 | 2.544 | %42 | **6.059** | **%100** |
| 4-7 body parts | 1.784 | %58 | 1.275 | %42 | **3.059** | **%100** |
| 8-11 body parts | 514 | %54 | 432 | %46 | **946** | **%100** |
| 12-15 body parts | 144 | %51 | 141 | %49 | **285** | **%100** |
| 16 body parts or more | 41 | %64 | 23 | %36 | **64** | **%100** |
| **TOTAL** | **5.998** | **%58** | **4.415** | **%42** | **10.413** | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is significant. p-value= 0,0 ; Chi2= 12,1 ; dof= 4.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases / Genetic test(s) looking for genetic changes (also called mutations or variants)

| I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | GENETIC TEST(S) LOOKING FOR GENETIC CHANGES (ALSO CALLED MUTATIONS OR VARIANTS) | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW/DON'T REMEMBER | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 3.458 | %58 | 2.038 | %34 | 502 | %8 | 5.998 | %100 |
| No | 1.998 | %45 | 2.097 | %47 | 320 | %7 | 4.415 | %100 |
| TOTAL | 5.456 | %52 | 4.135 | %40 | 822 | %8 | 10.413 | |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 195,7 ; dof= 2.*

## Cross: I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases / Other test(s) such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc

| I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | OTHER TEST(S) SUCH AS CLINICAL EXAMINATION(S), MEDICAL IMAGING (MRI, SCANS…), BIOPSY, BIOCHEMICAL TEST(S) (BLOOD OR URINE TESTS…), ETC | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW/DON'T REMEMBER | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 5.513 | %92 | 348 | %6 | 137 | %2 | 5.998 | %100 |
| No | 3.906 | %88 | 381 | %9 | 128 | %3 | 4.415 | %100 |
| TOTAL | 9.419 | %90 | 729 | %7 | 265 | %3 | 10.413 | |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 36,2 ; dof= 2.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

**Cross: I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases / ...you could not afford it?**

**Have you ever needed a genetic test but could not access it because...**
**...YOU COULD NOT AFFORD IT?**

| I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | YES | | NO | | NOT RELEVANT | | TOTAL | |
|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % |
| Yes | 507 | %8 | 4.292 | %72 | 1.199 | %20 | 5.998 | %100 |
| No | 587 | %13 | 2.780 | %63 | 1.048 | %24 | 4.415 | %100 |
| TOTAL | 1.094 | %11 | 7.072 | %68 | 2.247 | %22 | 10.413 | |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 100,9 ; dof= 2.*

**Cross: I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases / ...it was not available in your country?**

**Have you ever needed a genetic test but could not access it because...**
**...IT WAS NOT AVAILABLE IN YOUR COUNTRY?**

| I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | YES | | NO | | NOT RELEVANT | | TOTAL | |
|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % |
| Yes | 607 | %10 | 4.045 | %67 | 1.346 | %22 | 5.998 | %100 |
| No | 578 | %13 | 2.738 | %62 | 1.099 | %25 | 4.415 | %100 |
| TOTAL | 1.185 | %11 | 6.783 | %65 | 2.445 | %23 | 10.413 | |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 37,7 ; dof= 2.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases / …healthcare professionals were reluctant or not sufficiently informed?**

Have you ever needed a genetic test but could not access it because...

**…HEALTHCARE PROFESSIONALS WERE RELUCTANT OR NOT SUFFICIENTLY INFORMED?**

| I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | YES | | NO | | NOT RELEVANT | | TOTAL | |
|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % |
| Yes | 1.372 | %23 | 3.451 | %58 | 1.175 | %20 | 5.998 | %100 |
| No | 1.408 | %32 | 2.064 | %47 | 943 | %21 | 4.415 | %100 |
| TOTAL | 2.780 | %27 | 5.515 | %53 | 2.118 | %20 | 10.413 | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 137,2 ; dof= 2.*

**Cross: I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases / To your knowledge, the genetic test(s) that were conducted targeted…**

**TO YOUR KNOWLEDGE, THE GENETIC TEST(S) THAT WERE CONDUCTED TARGETED…**

| I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | ONLY ONE GENE | | SEVERAL GENES AT THE SAME TIME (GENE PANEL SEQUENCING) | | THE WHOLE DNA (WHOLE GENOME SEQUENCING) | | ALL THE GENES (WHOLE EXOME SEQUENCING) | | A TUMOUR (GENETIC PROFILING OF A TUMOUR) | | OTHER (EPIGENOME, RNA, ETC.) | | DON'T KNOW | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 945 | %27 | 1.079 | %31 | 562 | %16 | 330 | %10 | 84 | %2 | 70 | %2 | 960 | %28 | 3.458 | |
| No | 498 | %25 | 648 | %32 | 315 | %16 | 231 | %12 | 51 | %3 | 47 | %2 | 540 | %27 | 1.998 | |
| TOTAL | 1.443 | %26 | 1.727 | %32 | 877 | %16 | 561 | %10 | 135 | %2 | 117 | %2 | 1.500 | %27 | 5.456 | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is weakly significant. p-value= 0,1 ; Chi2= 9,5 ; dof= 6.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

**Cross: I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases / Did you ever request a private company or laboratory to conduct genetic testing to diagnose the disease?**

| I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | DID YOU EVER REQUEST A PRIVATE COMPANY OR LABORATORY TO CONDUCT GENETIC TESTING TO DIAGNOSE THE DISEASE? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO, NEVER | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 325 | %9 | 143 | %4 | 2.990 | %86 | 3.458 | %100 |
| No | 258 | %13 | 119 | %6 | 1.621 | %81 | 1.998 | %100 |
| TOTAL | 583 | %11 | 262 | %5 | 4.611 | %85 | 5.456 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 27,6 ; dof= 2.*

**Cross: I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases / In general, how satisfied are you with how the results of the GENETIC TESTS were given to you?**

| I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | IN GENERAL, HOW SATISFIED ARE YOU WITH HOW THE RESULTS OF THE GENETIC TESTS WERE GIVEN TO YOU? | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | VERY DISSATISFIED | | DISSATISFIED | | NEITHER SATISFIED NOR DISSATISFIED | | SATISFIED | | VERY SATISFIED | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 351 | %10 | 353 | %10 | 690 | %20 | 1.232 | %36 | 656 | %19 | 176 | %5 | 3.458 | %100 |
| No | 216 | %11 | 269 | %13 | 459 | %23 | 685 | %34 | 264 | %13 | 105 | %5 | 1.998 | %100 |
| TOTAL | 567 | %10 | 622 | %11 | 1.149 | %21 | 1.917 | %35 | 920 | %17 | 281 | %5 | 5.456 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 43,4 ; dof= 5.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases / After the tests were performed, were you offered genetic counselling (e.g. given information about how your genetic condition might affect you and your family)?

| I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | AFTER THE TESTS WERE PERFORMED, WERE YOU OFFERED GENETIC COUNSELLING (E.G. GIVEN INFORMATION ABOUT HOW YOUR GENETIC CONDITION MIGHT AFFECT YOU AND YOUR FAMILY)? | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| | YES, WITH A GENETIC COUNSELLOR OR CLINICAL GENETICIST | | YES, BY A HEALTHCARE PROFESSIONAL | | NO, I WASN'T OFFERED GENETIC COUNSELLING | | NOT SURE / DON'T REMEMBER | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % |
| Yes | 1.398 | %40 | 827 | %24 | 972 | %28 | 261 | %8 | 3.458 | %100 |
| No | 733 | %37 | 340 | %17 | 785 | %39 | 140 | %7 | 1.998 | %100 |
| TOTAL | 2.131 | %39 | 1.167 | %21 | 1.757 | %32 | 401 | %7 | 5.456 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 82,4 ; dof= 3.*

## Cross: I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases / Genetic tests

| I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | GENETIC TESTS | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 490 | %14 | 2.871 | %83 | 96 | %3 | 3.457 | %100 |
| No | 337 | %17 | 1.615 | %81 | 46 | %2 | 1.998 | %100 |
| TOTAL | 827 | %15 | 4.486 | %82 | 142 | %3 | 5.455 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is significant. p-value= 0,0 ; Chi2= 7,9 ; dof= 2.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

**Cross: I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases / Other diagnostic tests such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc.**

| I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | OTHER DIAGNOSTIC TESTS SUCH AS CLINICAL EXAMINATION(S), MEDICAL IMAGING (MRI, SCANS…), BIOPSY, BIOCHEMICAL TEST(S) (BLOOD OR URINE TESTS…), ETC. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 754 | %14 | 4.652 | %84 | 107 | %2 | 5.513 | %100 |
| No | 644 | %16 | 3.199 | %82 | 62 | %2 | 3.905 | %100 |
| TOTAL | 1.398 | %15 | 7.851 | %83 | 169 | %2 | 9.418 | |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 15,5 ; dof= 2.*

**Cross: I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases / Additional advice from a healthcare professional specialised in the rare disease (in person or virtually)**

| I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | ADDITIONAL ADVICE FROM A HEALTHCARE PROFESSIONAL SPECIALISED IN THE RARE DISEASE (IN PERSON OR VIRTUALLY) | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 1.150 | %19 | 4.720 | %79 | 128 | %2 | 5.998 | %100 |
| No | 920 | %21 | 3.417 | %77 | 78 | %2 | 4.415 | %100 |
| TOTAL | 2.070 | %20 | 8.137 | %78 | 206 | %2 | 10.413 | |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is weakly significant. p-value= 0,1 ; Chi2= 5,8 ; dof= 2.*

# Chapter 10.

## Misdiagnosis

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

| …wrongly attributed to another physical disease? | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| YES, one time | 0,3 | 1.542 | 2,6 | 1.448 | 2,4 | 838 | 2,7 | 1.506 | 3,5 | 1.274 |
| YES, several times | 0,4 | 3.471 | 4,8 | 3.203 | 5,7 | 1.753 | 5,9 | 3.389 | 6,9 | 2.720 |
| NO | 0,7 | 2.807 | 2,4 | 2.671 | 2,8 | 1.744 | 1,4 | 2.948 | 3,0 | 2.513 |

■ *Under-represented elements*　　■ *Over-represented elements*

*The relationship is weakly significant. p-value= 0,1 ; Fisher= 2,3.*

*Inter variance= 105,6. Intra variance= 46,0.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

| ...neglected, not taken seriously and/or considered as psychological? | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| YES, one time | 0,3 | 958 | 2,4 | 899 | 2,3 | 521 | 2,4 | 951 | 3,0 | 805 |
| YES, several times | 0,5 | 3.785 | 5,1 | 3.486 | 6,0 | 1.867 | 5,7 | 3.691 | 6,9 | 2.927 |
| NO | 0,5 | 3.077 | 2,0 | 2.937 | 2,2 | 1.947 | 1,5 | 3.201 | 3,0 | 2.775 |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is not significant. p-value= 0,6 ; Fisher= 0,5.*

*Inter variance= 23,0. Intra variance= 46,0.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

| Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed. | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| YES, one time | 0,5 | 2.058 | 3,2 | 1.917 | 3,4 | 1.095 | 3,0 | 2.055 | 4,2 | 1.704 |
| YES, several times | 0,4 | 3.867 | 4,6 | 3.570 | 5,3 | 1.972 | 5,6 | 3.764 | 6,5 | 3.052 |
| NO | 0,7 | 1.895 | 1,8 | 1.835 | 2,1 | 1.268 | 0,6 | 2.024 | 2,2 | 1.751 |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is not significant. p-value= 0,3 ; Fisher= 1,2.*

*Inter variance= 53,4. Intra variance= 46,0.*

Rare Barometer
A EURORDIS 🧬 INITIATIVE

EURORDIS
RARE DISEASES EUROPE

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Gender of the person affected by the rare disease / …wrongly attributed to another physical disease?

| GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE | …WRONGLY ATTRIBUTED TO ANOTHER PHYSICAL DISEASE? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | NO | | YES, ONE TIME | | YES, SEVERAL TIMES | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Female | _2.291_ | _%34_ | 1.215 | %18 | _3.153_ | _%47_ | 6.659 | %100 |
| Male | _1.275_ | _%45_ | 555 | %20 | _980_ | _%35_ | 2.810 | %100 |
| Other | 45 | %45 | 14 | %14 | 42 | %42 | 101 | %100 |
| TOTAL | 3.611 | %38 | 1.784 | %19 | 4.175 | %44 | 9.570 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 138,5 ; dof= 4.*

## Cross: Gender of the person affected by the rare disease / …neglected, not taken seriously and/or considered as psychological?

| GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE | …NEGLECTED, NOT TAKEN SERIOUSLY AND/OR CONSIDERED AS PSYCHOLOGICAL? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | NO | | YES, ONE TIME | | YES, SEVERAL TIMES | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Female | _2.382_ | _%36_ | 765 | %11 | _3.512_ | _%53_ | 6.659 | %100 |
| Male | _1.444_ | _%51_ | 348 | %12 | _1.018_ | _%36_ | 2.810 | %100 |
| Other | 45 | %45 | 12 | %12 | 44 | %44 | 101 | %100 |
| TOTAL | 3.871 | %40 | 1.125 | %12 | 4.574 | %48 | 9.570 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 234,0 ; dof= 4.*

## Cross: Gender of the person affected by the rare disease / Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed.

| GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE | HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Female | 1.706 | %26 | _3.453_ | _%52_ | _1.500_ | _%23_ | 6.659 | %100 |
| Male | 747 | %27 | _1.119_ | _%40_ | _944_ | _%34_ | 2.810 | %100 |
| Other | 21 | %21 | 45 | %45 | _35_ | _%35_ | 101 | %100 |
| TOTAL | 2.474 | %26 | 4.617 | %48 | 2.479 | %26 | 9.570 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 157,8 ; dof= 4.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: How old were you when you stopped full-time education? / …wrongly attributed to another physical disease?

| HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION? | ...WRONGLY ATTRIBUTED TO ANOTHER PHYSICAL DISEASE? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | NO | | YES, ONE TIME | | YES, SEVERAL TIMES | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 15 y.o. or under | 190 | %42 | 99 | %22 | 166 | %36 | 455 | %100 |
| between 16 and 19 y.o. | 924 | %38 | 447 | %18 | 1.093 | %44 | 2.464 | %100 |
| between 20 and 23 y.o. | 1.212 | %40 | 532 | %18 | 1.278 | %42 | 3.022 | %100 |
| 24 y.o. or above | 1.144 | %36 | 594 | %19 | 1.407 | %45 | 3.145 | %100 |
| TOTAL | 3.470 | %38 | 1.672 | %18 | 3.944 | %43 | 9.086 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 19,5 ; dof= 6.*

## Cross: How old were you when you stopped full-time education? / …neglected, not taken seriously and/or considered as psychological?

| HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION? | ...NEGLECTED, NOT TAKEN SERIOUSLY AND/OR CONSIDERED AS PSYCHOLOGICAL? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | NO | | YES, ONE TIME | | YES, SEVERAL TIMES | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 15 y.o. or under | 189 | %42 | 56 | %12 | 210 | %46 | 455 | %100 |
| between 16 and 19 y.o. | 991 | %40 | 298 | %12 | 1.175 | %48 | 2.464 | %100 |
| between 20 and 23 y.o. | 1.247 | %41 | 363 | %12 | 1.412 | %47 | 3.022 | %100 |
| 24 y.o. or above | 1.267 | %40 | 346 | %11 | 1.532 | %49 | 3.145 | %100 |
| TOTAL | 3.694 | %41 | 1.063 | %12 | 4.329 | %48 | 9.086 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is not significant. p-value= 0,7 ; Chi2= 4,1 ; dof= 6.*

## Cross: How old were you when you stopped full-time education? / Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed.

| HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION? | HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 15 y.o. or under | 146 | %32 | 188 | %41 | 121 | %27 | 455 | %100 |
| between 16 and 19 y.o. | 582 | %24 | 1.220 | %50 | 662 | %27 | 2.464 | %100 |
| between 20 and 23 y.o. | 798 | %26 | 1.408 | %47 | 816 | %27 | 3.022 | %100 |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 22,3 ; dof= 6.*

## Cross: How would you best describe yourself? / …wrongly attributed to another physical disease?

| HOW WOULD YOU BEST DESCRIBE YOURSELF? | …WRONGLY ATTRIBUTED TO ANOTHER PHYSICAL DISEASE? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | NO | | YES, ONE TIME | | YES, SEVERAL TIMES | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| I belong to the ethnic majority in the country where I live | 2.637 | %37 | 1.287 | %18 | 3.201 | %45 | 7.125 | %100 |
| I am part of an ethnic minority in the country where I live | 184 | %40 | 93 | %20 | 188 | %40 | 465 | %100 |
| Other, specify... | 127 | %38 | 74 | %22 | 136 | %40 | 337 | %100 |
| TOTAL | 2.948 | %37 | 1.454 | %18 | 3.525 | %44 | 7.927 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is weakly significant. p-value= 0,1 ; Chi2= 7,5 ; dof= 4.*

## Cross: How would you best describe yourself? / …neglected, not taken seriously and/or considered as psychological?

| HOW WOULD YOU BEST DESCRIBE YOURSELF? | …NEGLECTED, NOT TAKEN SERIOUSLY AND/OR CONSIDERED AS PSYCHOLOGICAL? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | NO | | YES, ONE TIME | | YES, SEVERAL TIMES | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| I belong to the ethnic majority in the country where I live | 2.858 | %40 | 837 | %12 | 3.430 | %48 | 7.125 | %100 |
| I am part of an ethnic minority in the country where I live | 188 | %40 | 57 | %12 | 220 | %47 | 465 | %100 |
| Other, specify... | 138 | %41 | 42 | %12 | 157 | %47 | 337 | %100 |
| TOTAL | 3.184 | %40 | 936 | %12 | 3.807 | %48 | 7.927 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is not significant. p-value= 1,0 ; Chi2= 0,5 ; dof= 4.*

## Cross: How would you best describe yourself? / Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed.

| HOW WOULD YOU BEST DESCRIBE YOURSELF? | HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| I belong to the ethnic majority in the country where I live | 1.775 | %25 | 3.530 | %50 | 1.820 | %26 | 7.125 | %100 |
| I am part of an ethnic minority in the country where I live | 126 | %27 | 217 | %47 | 122 | %26 | 465 | %100 |
| Other, specify... | 84 | %25 | 158 | %47 | 95 | %28 | 337 | %100 |
| TOTAL | 1.985 | %25 | 3.905 | %49 | 2.037 | %26 | 7.927 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Typology of countries based on size and welfare / …wrongly attributed to another physical disease?

| TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE | ...WRONGLY ATTRIBUTED TO ANOTHER PHYSICAL DISEASE? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | NO | | YES, ONE TIME | | YES, SEVERAL TIMES | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Group A ('Eastern Europe') | 675 | %38 | 363 | %20 | 756 | %42 | 1.794 | %100 |
| Group B ('Western Europe') | 1.887 | %37 | 1.025 | %20 | 2.193 | %43 | 5.105 | %100 |
| Group C ('Northern Europe') | 1.325 | %40 | 503 | %15 | 1.445 | %44 | 3.273 | %100 |
| TOTAL | 3.887 | %38 | 1.891 | %19 | 4.394 | %43 | 10.172 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 34,8 ; dof= 4.*

## Cross: Typology of countries based on size and welfare / …neglected, not taken seriously and/or considered as psychological?

| TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE | ...NEGLECTED, NOT TAKEN SERIOUSLY AND/OR CONSIDERED AS PSYCHOLOGICAL? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | NO | | YES, ONE TIME | | YES, SEVERAL TIMES | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Group A ('Eastern Europe') | 806 | %45 | 250 | %14 | 738 | %41 | 1.794 | %100 |
| Group B ('Western Europe') | 2.031 | %40 | 636 | %12 | 2.438 | %48 | 5.105 | %100 |
| Group C ('Northern Europe') | 1.344 | %41 | 316 | %10 | 1.613 | %49 | 3.273 | %100 |
| TOTAL | 4.181 | %41 | 1.202 | %12 | 4.789 | %47 | 10.172 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 47,4 ; dof= 4.*

## Cross: Typology of countries based on size and welfare / Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed.

| TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE | HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Group A ('Eastern Europe') | 431 | %24 | 861 | %48 | 502 | %28 | 1.794 | %100 |
| Group B ('Western Europe') | 1.399 | %27 | 2.442 | %48 | 1.264 | %25 | 5.105 | %100 |
| Group C ('Northern Europe') | 784 | %24 | 1.567 | %48 | 922 | %28 | 3.273 | %100 |
| TOTAL | 2.614 | %26 | 4.870 | %48 | 2.688 | %26 | 10.172 | |

■ *Under-represented elements*   ■ *Over-represented elements*

## Cross: Orphacode associated nomenclature (english) / …wrongly attributed to another physical disease?

| ORPHACODE ASSOCIATED NOMENCLATURE (ENGLISH) | …WRONGLY ATTRIBUTED TO ANOTHER PHYSICAL DISEASE? | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|
| | NO | | YES, ONE TIME | | YES, SEVERAL TIMES | | | |
| | N | % | N | % | N | % | N | % |
| Hereditary hemorrhagic telangiectasia | 265 | %58 | 61 | %13 | 132 | %29 | 458 | %100 |
| Hypermobile Ehlers-Danlos syndrome | 35 | %11 | 31 | %10 | 251 | %79 | 317 | %100 |
| Sarcoidosis | 36 | %21 | 51 | %30 | 83 | %49 | 170 | %100 |
| Classical Ehlers-Danlos syndrome | 18 | %13 | 16 | %12 | 103 | %75 | 137 | %100 |
| Williams syndrome | 76 | %56 | 24 | %18 | 36 | %26 | 136 | %100 |
| Cystic fibrosis | 67 | %52 | 19 | %15 | 42 | %33 | 128 | %100 |
| Myasthenia gravis | 38 | %32 | 37 | %31 | 45 | %38 | 120 | %100 |
| Systemic sclerosis | 44 | %41 | 25 | %23 | 38 | %36 | 107 | %100 |
| Tuberous sclerosis complex | 63 | %64 | 16 | %16 | 19 | %19 | 98 | %100 |
| Neurofibromatosis type 1 | 58 | %63 | 14 | %15 | 20 | %22 | 92 | %100 |
| Interstitial cystitis | 9 | %12 | 16 | %22 | 49 | %66 | 74 | %100 |
| Addison disease | 25 | %34 | 17 | %23 | 31 | %42 | 73 | %100 |
| 22q11.2 deletion syndrome | 37 | %54 | 11 | %16 | 20 | %29 | 68 | %100 |
| Chronic inflammatory demyelinating polyneuropathy | 24 | %37 | 18 | %28 | 23 | %35 | 65 | %100 |
| Perineural cyst | 9 | %14 | 7 | %11 | 47 | %75 | 63 | %100 |
| Acute inflammatory demyelinating polyradiculoneuropathy | 23 | %37 | 22 | %35 | 17 | %27 | 62 | %100 |
| Rett syndrome | 25 | %42 | 10 | %17 | 25 | %42 | 60 | %100 |
| Marfan syndrome | 26 | %50 | 6 | %12 | 20 | %38 | 52 | %100 |
| Fragile X syndrome | 29 | %59 | 10 | %20 | 10 | %20 | 49 | %100 |
| Behçet disease | 3 | %6 | 10 | %21 | 34 | %72 | 47 | %100 |
| Primary sclerosing cholangitis | 25 | %54 | 10 | %22 | 11 | %24 | 46 | %100 |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 4.655,5 ; dof= 3.350.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Orphacode associated nomenclature (english) / …neglected, not taken seriously and/or considered as psychological?

| ORPHACODE ASSOCIATED NOMENCLATURE (ENGLISH) | …NEGLECTED, NOT TAKEN SERIOUSLY AND/OR CONSIDERED AS PSYCHOLOGICAL? | | | |
| --- | --- | --- | --- | --- |
| | YES, ONE TIME | YES, SEVERAL TIMES | NO | TOTAL |
| Hereditary hemorrhagic telangiectasia | %7 | %39 | %53 | %100 |
| Hypermobile Ehlers-Danlos syndrome | %5 | %89 | %6 | %100 |
| Sarcoidosis | %14 | %49 | %37 | %100 |
| Classical Ehlers-Danlos syndrome | %5 | %85 | %10 | %100 |
| Williams syndrome | %13 | %32 | %56 | %100 |
| Cystic fibrosis | %12 | %27 | %62 | %100 |
| Myasthenia gravis | %19 | %43 | %38 | %100 |
| Systemic sclerosis | %14 | %35 | %51 | %100 |
| Tuberous sclerosis complex | %12 | %28 | %60 | %100 |
| Neurofibromatosis type 1 | %17 | %37 | %46 | %100 |
| Interstitial cystitis | %5 | %82 | %12 | %100 |
| Addison disease | %14 | %64 | %22 | %100 |
| 22q11.2 deletion syndrome | %4 | %41 | %54 | %100 |
| Chronic inflammatory demyelinating polyneuropathy | %23 | %28 | %49 | %100 |
| Perineural cyst | %10 | %83 | %8 | %100 |
| Acute inflammatory demyelinating polyradiculoneuropathy | %27 | %27 | %45 | %100 |
| Rett syndrome | %10 | %50 | %40 | %100 |
| Marfan syndrome | %15 | %46 | %38 | %100 |
| Fragile X syndrome | %12 | %53 | %35 | %100 |
| Behçet disease | %13 | %77 | %11 | %100 |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 4.664,6 ; dof= 3.350.*

**Cross: Orphacode associated nomenclature (english) / Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed.**

| ORPHACODE ASSOCIATED NOMENCLATURE (ENGLISH) | HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Hereditary hemorrhagic telangiectasia | 124 | %27 | 149 | %33 | 185 | %40 | 458 | %100 |
| Hypermobile Ehlers-Danlos syndrome | 55 | %17 | 253 | %80 | 9 | %3 | 317 | %100 |
| Sarcoidosis | 47 | %28 | 97 | %57 | 26 | %15 | 170 | %100 |
| Classical Ehlers-Danlos syndrome | 24 | %18 | 105 | %77 | 8 | %6 | 137 | %100 |
| Williams syndrome | 35 | %26 | 43 | %32 | 58 | %43 | 136 | %100 |
| Cystic fibrosis | 20 | %16 | 48 | %38 | 60 | %47 | 128 | %100 |
| Myasthenia gravis | 29 | %24 | 60 | %50 | 31 | %26 | 120 | %100 |
| Systemic sclerosis | 29 | %27 | 42 | %39 | 36 | %34 | 107 | %100 |
| Tuberous sclerosis complex | 30 | %31 | 22 | %22 | 46 | %47 | 98 | %100 |
| Neurofibromatosis type 1 | 32 | %35 | 24 | %26 | 36 | %39 | 92 | %100 |
| Interstitial cystitis | 20 | %27 | 52 | %70 | 2 | %3 | 74 | %100 |
| Addison disease | 28 | %38 | 36 | %49 | 9 | %12 | 73 | %100 |
| 22q11.2 deletion syndrome | 18 | %26 | 21 | %31 | 29 | %43 | 68 | %100 |
| Chronic inflammatory demyelinating polyneuropathy | 15 | %23 | 31 | %48 | 19 | %29 | 65 | %100 |
| Perineural cyst | 13 | %21 | 48 | %76 | 2 | %3 | 63 | %100 |
| Acute inflammatory demyelinating polyradiculoneuropathy | 21 | %34 | 26 | %42 | 15 | %24 | 62 | %100 |
| Rett syndrome | 19 | %32 | 27 | %45 | 14 | %23 | 60 | %100 |
| Marfan syndrome | 14 | %27 | 22 | %42 | 16 | %31 | 52 | %100 |
| Fragile X syndrome | 23 | %47 | 13 | %27 | 13 | %27 | 49 | %100 |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 4.559,4 ; dof= 3.350.*

## Cross: Genetic diseases / …wrongly attributed to another physical disease?

| GENETIC DISEASES | …WRONGLY ATTRIBUTED TO ANOTHER PHYSICAL DISEASE? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | NO | | YES, ONE TIME | | YES, SEVERAL TIMES | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Genetic diseases | 2.311 | %42 | 909 | %17 | 2.227 | %41 | 5.447 | %100 |
| Non Genetic diseases | 855 | %33 | 602 | %23 | 1.170 | %45 | 2.627 | %100 |
| TOTAL | 3.166 | %39 | 1.511 | %19 | 3.397 | %42 | 8.074 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 86,5 ; dof= 2.*

## Cross: Genetic diseases / …neglected, not taken seriously and/or considered as psychological?

| GENETIC DISEASES | …NEGLECTED, NOT TAKEN SERIOUSLY AND/OR CONSIDERED AS PSYCHOLOGICAL? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Genetic diseases | 579 | %11 | 2.463 | %45 | 2.405 | %44 | 5.447 | %100 |
| Non Genetic diseases | 386 | %15 | 1.234 | %47 | 1.007 | %38 | 2.627 | %100 |
| TOTAL | 965 | %12 | 3.697 | %46 | 3.412 | %42 | 8.074 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 39,9 ; dof= 2.*

## Cross: Genetic diseases / Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed.

| GENETIC DISEASES | HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Genetic diseases | 1.390 | %26 | 2.434 | %45 | 1.623 | %30 | 5.447 | %100 |
| Non Genetic diseases | 690 | %26 | 1.340 | %51 | 597 | %23 | 2.627 | %100 |
| TOTAL | 2.080 | %26 | 3.774 | %47 | 2.220 | %27 | 8.074 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 47,8 ; dof= 2.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Point prevalence of the rare disease / …wrongly attributed to another physical disease?

| POINT PREVALENCE OF THE RARE DISEASE | …WRONGLY ATTRIBUTED TO ANOTHER PHYSICAL DISEASE? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | NO | | YES, ONE TIME | | YES, SEVERAL TIMES | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 1-5 / 10 000 | 1.008 | %42 | 418 | %17 | 981 | %41 | 2.407 | %100 |
| 1-9 / 100 000 | 778 | %39 | 409 | %20 | 812 | %41 | 1.999 | %100 |
| 1-9 / 1 000 000 | 166 | %36 | 95 | %21 | 198 | %43 | 459 | %100 |
| <1 / 1 000 000 | 335 | %39 | 124 | %14 | 397 | %46 | 856 | %100 |
| TOTAL | 2.287 | %40 | 1.046 | %18 | 2.388 | %42 | 5.721 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 24,8 ; dof= 6.*

## Cross: Point prevalence of the rare disease / …neglected, not taken seriously and/or considered as psychological?

| POINT PREVALENCE OF THE RARE DISEASE | …NEGLECTED, NOT TAKEN SERIOUSLY AND/OR CONSIDERED AS PSYCHOLOGICAL? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 1-5 / 10 000 | 256 | %11 | 1.184 | %49 | 967 | %40 | 2.407 | %100 |
| 1-9 / 100 000 | 260 | %13 | 845 | %42 | 894 | %45 | 1.999 | %100 |
| 1-9 / 1 000 000 | 47 | %10 | 215 | %47 | 197 | %43 | 459 | %100 |
| <1 / 1 000 000 | 111 | %13 | 403 | %47 | 342 | %40 | 856 | %100 |
| TOTAL | 674 | %12 | 2.647 | %46 | 2.400 | %42 | 5.721 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 25,1 ; dof= 6.*

## Cross: Calculation point prevalence / The variable computes the number of times respondents were misdiagnosed an classifies them accordingly.

| POINT PREVALENCE OF THE RARE DISEASE | HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 1-5 / 10 000 | 640 | %27 | 1.089 | %45 | 678 | %28 | 2.407 | %100 |
| 1-9 / 100 000 | 503 | %25 | 925 | %46 | 571 | %29 | 1.999 | %100 |
| 1-9 / 1 000 000 | 127 | %28 | 220 | %48 | 112 | %24 | 459 | %100 |
| <1 / 1 000 000 | 203 | %24 | 431 | %50 | 222 | %26 | 856 | %100 |
| TOTAL | 1.473 | %26 | 2.665 | %47 | 1.583 | %28 | 5.721 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

## Cross: Disease complexity classified into five groups, based on the number of affected body parts. / …wrongly attributed to another physical disease?

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | ...WRONGLY ATTRIBUTED TO ANOTHER PHYSICAL DISEASE? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | NO | | YES, ONE TIME | | YES, SEVERAL TIMES | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 1-3 body parts | 2.721 | %45 | 1.230 | %20 | 2.152 | %35 | 6.103 | %1 |
| 4-7 body parts | 1.016 | %33 | 552 | %18 | 1.513 | %49 | 3.081 | % |
| 8-11 body parts | 229 | %24 | 135 | %14 | 587 | %62 | 951 | %1 |
| 12-15 body parts | 43 | %15 | 30 | %10 | 213 | %74 | 286 | % |
| 16 body parts or more | 7 | %11 | 3 | %5 | 55 | %85 | 65 | %1 |
| TOTAL | 4.016 | %38 | 1.950 | %19 | 4.520 | %43 | 10.486 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 505,4 ; dof= 8.*

## Cross: Disease complexity classified into five groups, based on the number of affected body parts. / …neglected, not taken seriously and/or considered as psychological?

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | ...NEGLECTED, NOT TAKEN SERIOUSLY AND/OR CONSIDERED AS PSYCHOLOGICAL? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 1-3 body parts | 759 | %12 | 2.405 | %39 | 2.939 | %48 | 6.103 | % |
| 4-7 body parts | 385 | %12 | 1.592 | %52 | 1.104 | %36 | 3.081 | % |
| 8-11 body parts | 79 | %8 | 652 | %69 | 220 | %23 | 951 | % |
| 12-15 body parts | 17 | %6 | 231 | %81 | 38 | %13 | 286 | % |
| 16 body parts or more | 6 | %9 | 54 | %83 | 5 | %8 | 65 | % |
| TOTAL | 1.246 | %12 | 4.934 | %47 | 4.306 | %41 | 10.486 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 533,1 ; dof= 8.*

## Cross: Disease complexity classified into five groups, based on the number of affected body parts. / Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed.

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 1-3 body parts | 1.656 | %27 | 2.467 | %40 | 1.980 | %32 | 6.103 | %100 |
| 4-7 body parts | 774 | %25 | 1.661 | %54 | 646 | %21 | 3.081 | %100 |
| 8-11 body parts | 203 | %21 | 614 | %65 | 134 | %14 | 951 | %100 |
| 12-15 body parts | 46 | %16 | 219 | %77 | 21 | %7 | 286 | %100 |
| 16 body parts or more | 4 | %6 | 57 | %88 | 4 | %6 | 65 | %100 |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 474,2 ; dof= 8.*

Rare Barometer · A EURORDIS Ⓡ INITIATIVE    EURORDIS RARE DISEASES EUROPE

## Cross: Family members were previously diagnosed with the same disease / …wrongly attributed to another physical disease?

| FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | …WRONGLY ATTRIBUTED TO ANOTHER PHYSICAL DISEASE? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | NO | | YES, ONE TIME | | YES, SEVERAL TIMES | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 697 | %53 | 166 | %13 | 446 | %34 | 1.309 | %100 |
| No | 3.104 | %37 | 1.639 | %19 | 3.679 | %44 | 8.422 | %100 |
| TOTAL | 3.801 | %39 | 1.805 | %19 | 4.125 | %42 | 9.731 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 130,7 ; dof= 2.*

## Cross: Family members were previously diagnosed with the same disease / …neglected, not taken seriously and/or considered as psychological?

| FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | …NEGLECTED, NOT TAKEN SERIOUSLY AND/OR CONSIDERED AS PSYCHOLOGICAL? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 125 | %10 | 534 | %41 | 650 | %50 | 1.309 | %100 |
| No | 1.027 | %12 | 3.986 | %47 | 3.409 | %40 | 8.422 | %100 |
| TOTAL | 1.152 | %12 | 4.520 | %46 | 4.059 | %42 | 9.731 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 40,0 ; dof= 2.*

## Cross: Family members were previously diagnosed with the same disease / Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed.

| FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 323 | %25 | 486 | %37 | 500 | %38 | 1.309 | %100 |
| No | 2.190 | %26 | 4.093 | %49 | 2.139 | %25 | 8.422 | %100 |
| TOTAL | 2.513 | %26 | 4.579 | %47 | 2.639 | %27 | 9.731 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 100,9 ; dof= 2.*

## Cross: How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis? / ...wrongly attributed to another physical disease?

| HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | ...WRONGLY ATTRIBUTED TO ANOTHER PHYSICAL DISEASE? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | NO | | YES, ONE TIME | | YES, SEVERAL TIMES | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 0-1 | 938 | %72 | 181 | %14 | 186 | %14 | 1.305 | %100 |
| between 2 and 4 | 2.091 | %46 | 1.092 | %24 | 1.386 | %30 | 4.569 | %100 |
| between 5 and 7 | 551 | %27 | 398 | %20 | 1.084 | %53 | 2.033 | %100 |
| between 8 and 10 | 153 | %19 | 134 | %17 | 504 | %64 | 791 | %100 |
| more than 10 | 283 | %16 | 145 | %8 | 1.360 | %76 | 1.788 | %100 |
| TOTAL | 4.016 | %38 | 1.950 | %19 | 4.520 | %43 | 10.486 | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 2.022,5 ; dof= 8.*

## Cross: How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis? / ...neglected, not taken seriously and/or considered as psychological?

| HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | ...NEGLECTED, NOT TAKEN SERIOUSLY AND/OR CONSIDERED AS PSYCHOLOGICAL? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | NO | | YES, ONE TIME | | YES, SEVERAL TIMES | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 0-1 | 912 | %70 | 116 | %9 | 277 | %21 | 1.305 | %100 |
| between 2 and 4 | 2.309 | %51 | 660 | %14 | 1.600 | %35 | 4.569 | %100 |
| between 5 and 7 | 615 | %30 | 276 | %14 | 1.142 | %56 | 2.033 | %100 |
| between 8 and 10 | 157 | %20 | 83 | %10 | 551 | %70 | 791 | %100 |
| more than 10 | 313 | %18 | 111 | %6 | 1.364 | %76 | 1.788 | %100 |
| TOTAL | 4.306 | %41 | 1.246 | %12 | 4.934 | %47 | 10.486 | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 1.612,0 ; dof= 8.*

Rare Barometer
A EURORDIS INITIATIVE

EURORDIS
RARE DISEASES EUROPE

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed. / …neglected, not taken seriously and/or considered as psychological?**

| HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | …NEGLECTED, NOT TAKEN SERIOUSLY AND/OR CONSIDERED AS PSYCHOLOGICAL? | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | NO | | YES, ONE TIME | | YES, SEVERAL TIMES | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| YES, one time | 791 | %29 | 337 | %13 | 1.555 | %58 | 2.683 | %100 |
| YES, several times | 730 | %15 | 909 | %18 | 3.379 | %67 | 5.018 | %100 |
| NO | 2.785 | %100 | 0 | %0 | 0 | %0 | 2.785 | %100 |
| TOTAL | 4.306 | %41 | 1.246 | %12 | 4.934 | %47 | 10.486 | |

🟥 *Under-represented elements* 🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 5.615,6 ; dof= 4.*

**Cross: I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases / …wrongly attributed to another physical disease?**

| I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | …WRONGLY ATTRIBUTED TO ANOTHER PHYSICAL DISEASE? | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | NO | | YES, ONE TIME | | YES, SEVERAL TIMES | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 2.555 | %43 | 1.119 | %19 | 2.324 | %39 | 5.998 | %100 |
| No | 1.445 | %33 | 813 | %18 | 2.157 | %49 | 4.415 | %100 |
| TOTAL | 4.000 | %38 | 1.932 | %19 | 4.481 | %43 | 10.413 | %100 |

🟥 *Under-represented elements* 🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 125,0 ; dof= 2.*

**Cross: I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases / …neglected, not taken seriously and/or considered as psychological?**

| I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | …NEGLECTED, NOT TAKEN SERIOUSLY AND/OR CONSIDERED AS PSYCHOLOGICAL? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | NO | | YES, ONE TIME | | YES, SEVERAL TIMES | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 2.772 | %46 | 705 | %12 | 2.521 | %42 | 5.998 | %100 |
| No | 1.509 | %34 | 527 | %12 | 2.379 | %54 | 4.415 | %100 |
| TOTAL | 4.281 | %41 | 1.232 | %12 | 4.900 | %47 | 10.413 | |

🟥 *Under-represented elements* 🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 165,6 ; dof= 2.*

**Cross: I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases / Has the person affected by the rare disease already been misdiagnosed?**
**Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed.**

| I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 1.512 | %25 | 2.613 | %44 | 1.873 | %31 | 5.998 | %100 |
| No | 1.157 | %26 | 2.359 | %53 | 899 | %20 | 4.415 | %100 |
| TOTAL | 2.669 | %26 | 4.972 | %48 | 2.772 | %27 | 10.413 | |

🟥 *Under-represented elements* 🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 165,6 ; dof= 2.*

**Only respondents who said that the rare disease has already been misdiagnosed**

### As a consequence of the misdiagnosis, please tell us if you or the person you care for…

| | DEFINITELY | PROBABLY | PROBABLY NOT | DEFINITELY NOT | DON'T KNOW/NOT APPLICABLE | TOTAL |
|---|---|---|---|---|---|---|
| …could not access all the information needed to make informed reproductive choices such as planning whether or not to have children, or deciding whether or not to conduct prenatal tests | 1.264 | 956 | 810 | 1.621 | 3.050 | **7.701** |
| …received inappropriate care, treatment or surgery | 2.400 | 1.647 | 1.033 | 1.709 | 912 | **7.701** |
| …had a delayed access to the most appropriate care, treatment or surgery | 3.380 | 1.883 | 787 | 973 | 678 | **7.701** |
| …could not access appropriate care, treatment or surgery | 2.858 | 1.786 | 991 | 1.297 | 769 | **7.701** |
| …experienced worsening of the symptoms | 3.967 | 1.634 | 856 | 724 | 520 | **7.701** |
| **TOTAL** | **13.869** | **7.906** | **4.477** | **6.324** | **5.929** | **38.505** |

### As a consequence of the misdiagnosis, please tell us if you or the person you care for…
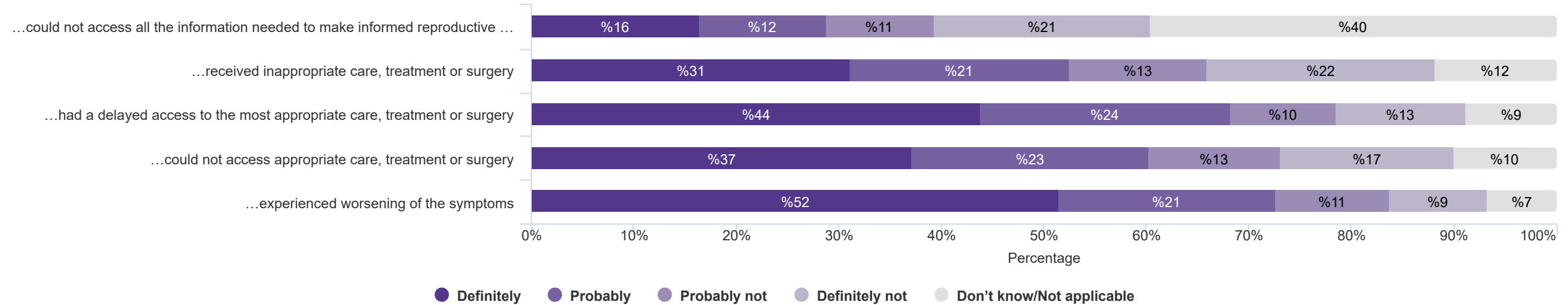
| | Definitely | Probably | Probably not | Definitely not | Don't know/Not applicable |
|---|---|---|---|---|---|
| …could not access all the information needed to make informed reproductive … | %16 | %12 | %11 | %21 | %40 |
| …received inappropriate care, treatment or surgery | %31 | %21 | %13 | %22 | %12 |
| …had a delayed access to the most appropriate care, treatment or surgery | %44 | %24 | %10 | %13 | %9 |
| …could not access appropriate care, treatment or surgery | %37 | %23 | %13 | %17 | %10 |
| …experienced worsening of the symptoms | %52 | %21 | %11 | %9 | %7 |

Percentage

● Definitely  ● Probably  ● Probably not  ● Definitely not  ● Don't know/Not applicable

Rare Barometer
A EURORDIS INITIATIVE

EURORDIS
RARE DISEASES EUROPE

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Only respondents who said that the rare disease has already been misdiagnosed

### Cross: …wrongly attributed to another physical disease? / …could not access all the information needed to make informed reproductive choices such as planning whether or not to have children, or deciding whether or not to conduct prenatal tests

| …WRONGLY ATTRIBUTED TO ANOTHER PHYSICAL DISEASE? | …COULD NOT ACCESS ALL THE INFORMATION NEEDED TO MAKE INFORMED REPRODUCTIVE CHOICES SUCH AS PLANNING WHETHER OR NOT TO HAVE CHILDREN, OR DECIDING WHETHER OR NOT TO CONDUCT PRENATAL TESTS | | | | | | | | | |
| | DEFINITELY | | PROBABLY | | PROBABLY NOT | | DEFINITELY NOT | | DON'T KNOW/NOT APPLICABLE | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, one time | 232 | %12 | 227 | %12 | 231 | %12 | 497 | %25 | 763 | %39 | 1.950 | %100 |
| YES, several times | 885 | %20 | 605 | %13 | 455 | %10 | 824 | %18 | 1.751 | %39 | 4.520 | %100 |
| NO | 147 | %12 | 124 | %10 | 124 | %10 | 300 | %24 | 536 | %44 | 1.231 | %100 |
| TOTAL | 1.264 | %16 | 956 | %12 | 810 | %11 | 1.621 | %21 | 3.050 | %40 | 7.701 | %100 |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 128,6 ; dof= 8.*

### Cross: …wrongly attributed to another physical disease? / …received inappropriate care, treatment or surgery

| …WRONGLY ATTRIBUTED TO ANOTHER PHYSICAL DISEASE? | …RECEIVED INAPPROPRIATE CARE, TREATMENT OR SURGERY | | | | | | | | | |
| | DEFINITELY | | PROBABLY | | PROBABLY NOT | | DEFINITELY NOT | | DON'T KNOW/NOT APPLICABLE | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, one time | 397 | %20 | 377 | %19 | 288 | %15 | 637 | %33 | 251 | %13 | 1.950 | %100 |
| YES, several times | 1.756 | %39 | 1.074 | %24 | 556 | %12 | 707 | %16 | 427 | %9 | 4.520 | %100 |
| NO | 247 | %20 | 196 | %16 | 189 | %15 | 365 | %30 | 234 | %19 | 1.231 | %100 |
| TOTAL | 2.400 | %31 | 1.647 | %21 | 1.033 | %13 | 1.709 | %22 | 912 | %12 | 7.701 | %100 |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 542,3 ; dof= 8.*

## Only respondents who said that the rare disease has already been misdiagnosed

### Cross: …wrongly attributed to another physical disease? / …had a delayed access to the most appropriate care, treatment or surgery

| …WRONGLY ATTRIBUTED TO ANOTHER PHYSICAL DISEASE? | …HAD A DELAYED ACCESS TO THE MOST APPROPRIATE CARE, TREATMENT OR SURGERY | | | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | DEFINITELY | | PROBABLY | | PROBABLY NOT | | DEFINITELY NOT | | DON'T KNOW/NOT APPLICABLE | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, one time | 592 | %30 | 498 | %26 | 273 | %14 | 394 | %20 | 193 | %10 | 1.950 | %100 |
| YES, several times | 2.381 | %53 | 1.103 | %24 | 364 | %8 | 369 | %8 | 303 | %7 | 4.520 | %100 |
| NO | 407 | %33 | 282 | %23 | 150 | %12 | 210 | %17 | 182 | %15 | 1.231 | %100 |
| TOTAL | 3.380 | %44 | 1.883 | %24 | 787 | %10 | 973 | %13 | 678 | %9 | 7.701 | %100 |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 502,9 ; dof= 8.*

### Cross: …wrongly attributed to another physical disease? / …received inappropriate care, treatment or surgery

| …WRONGLY ATTRIBUTED TO ANOTHER PHYSICAL DISEASE? | …RECEIVED INAPPROPRIATE CARE, TREATMENT OR SURGERY | | | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | DEFINITELY | | PROBABLY | | PROBABLY NOT | | DEFINITELY NOT | | DON'T KNOW/NOT APPLICABLE | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, one time | 397 | %20 | 377 | %19 | 288 | %15 | 637 | %33 | 251 | %13 | 1.950 | %100 |
| YES, several times | 1.756 | %39 | 1.074 | %24 | 556 | %12 | 707 | %16 | 427 | %9 | 4.520 | %100 |
| NO | 247 | %20 | 196 | %16 | 189 | %15 | 365 | %30 | 234 | %19 | 1.231 | %100 |
| TOTAL | 2.400 | %31 | 1.647 | %21 | 1.033 | %13 | 1.709 | %22 | 912 | %12 | 7.701 | %100 |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 542,3 ; dof= 8.*

**Only respondents who said that the rare disease has already been misdiagnosed**

### Cross: …wrongly attributed to another physical disease? / …experienced worsening of the symptoms

| …WRONGLY ATTRIBUTED TO ANOTHER PHYSICAL DISEASE? | …EXPERIENCED WORSENING OF THE SYMPTOMS | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | DEFINITELY | | PROBABLY | | PROBABLY NOT | | DEFINITELY NOT | | DON'T KNOW/NOT APPLICABLE | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, one time | 747 | %38 | 449 | %23 | 294 | %15 | 314 | %16 | 146 | %7 | 1.950 | %100 |
| YES, several times | 2.730 | %60 | 946 | %21 | 390 | %9 | 238 | %5 | 216 | %5 | 4.520 | %100 |
| NO | 490 | %40 | 239 | %19 | 172 | %14 | 172 | %14 | 158 | %13 | 1.231 | %100 |
| TOTAL | 3.967 | %52 | 1.634 | %21 | 856 | %11 | 724 | %9 | 520 | %7 | 7.701 | %100 |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 532,7 ; dof= 8.*

### Cross: …neglected, not taken seriously and/or considered as psychological? / …could not access all the information needed to make informed reproductive choices such as planning whether or not to have children, or deciding whether or not to conduct prenatal tests

| …NEGLECTED, NOT TAKEN SERIOUSLY AND/OR CONSIDERED AS PSYCHOLOGICAL? | …COULD NOT ACCESS ALL THE INFORMATION NEEDED TO MAKE INFORMED REPRODUCTIVE CHOICES SUCH AS PLANNING WHETHER OR NOT TO HAVE CHILDREN, OR DECIDING WHETHER OR NOT TO CONDUCT PRENATAL TESTS | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | DEFINITELY | | PROBABLY | | PROBABLY NOT | | DEFINITELY NOT | | DON'T KNOW/NOT APPLICABLE | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, one time | 133 | %11 | 133 | %11 | 141 | %11 | 313 | %25 | 526 | %42 | 1.246 | %100 |
| YES, several times | 1.005 | %20 | 665 | %13 | 490 | %10 | 870 | %18 | 1.904 | %39 | 4.934 | %100 |
| NO | 126 | %8 | 158 | %10 | 179 | %12 | 438 | %29 | 620 | %41 | 1.521 | %100 |
| TOTAL | 1.264 | %16 | 956 | %12 | 810 | %11 | 1.621 | %21 | 3.050 | %40 | 7.701 | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 235,0 ; dof= 8.*

**Only respondents who said that the rare disease has already been misdiagnosed**

## Cross: …neglected, not taken seriously and/or considered as psychological? / …received inappropriate care, treatment or surgery

| …NEGLECTED, NOT TAKEN SERIOUSLY AND/OR CONSIDERED AS PSYCHOLOGICAL? | …RECEIVED INAPPROPRIATE CARE, TREATMENT OR SURGERY | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | DEFINITELY | | PROBABLY | | PROBABLY NOT | | DEFINITELY NOT | | DON'T KNOW/NOT APPLICABLE | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, one time | 301 | %24 | 237 | %19 | 181 | %15 | 364 | %29 | 163 | %13 | 1.246 | %100 |
| YES, several times | 1.868 | %38 | 1.157 | %23 | 573 | %12 | 800 | %16 | 536 | %11 | 4.934 | %100 |
| NO | 231 | %15 | 253 | %17 | 279 | %18 | 545 | %36 | 213 | %14 | 1.521 | %100 |
| TOTAL | 2.400 | %31 | 1.647 | %21 | 1.033 | %13 | 1.709 | %22 | 912 | %12 | 7.701 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 531,2 ; dof= 8.*

## Cross: …neglected, not taken seriously and/or considered as psychological? / …had a delayed access to the most appropriate care, treatment or surgery

| …NEGLECTED, NOT TAKEN SERIOUSLY AND/OR CONSIDERED AS PSYCHOLOGICAL? | …HAD A DELAYED ACCESS TO THE MOST APPROPRIATE CARE, TREATMENT OR SURGERY | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | DEFINITELY | | PROBABLY | | PROBABLY NOT | | DEFINITELY NOT | | DON'T KNOW/NOT APPLICABLE | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, one time | 421 | %34 | 344 | %28 | 152 | %12 | 203 | %16 | 126 | %10 | 1.246 | %100 |
| YES, several times | 2.600 | %53 | 1.143 | %23 | 387 | %8 | 400 | %8 | 404 | %8 | 4.934 | %100 |
| NO | 359 | %24 | 396 | %26 | 248 | %16 | 370 | %24 | 148 | %10 | 1.521 | %100 |
| TOTAL | 3.380 | %44 | 1.883 | %24 | 787 | %10 | 973 | %13 | 678 | %9 | 7.701 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 619,7 ; dof= 8.*

## Only respondents who said that the rare disease has already been misdiagnosed

### Cross: …neglected, not taken seriously and/or considered as psychological? / …could not access appropriate care, treatment or surgery

| …NEGLECTED, NOT TAKEN SERIOUSLY AND/OR CONSIDERED AS PSYCHOLOGICAL? | …COULD NOT ACCESS APPROPRIATE CARE, TREATMENT OR SURGERY | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | DEFINITELY | | PROBABLY | | PROBABLY NOT | | DEFINITELY NOT | | DON'T KNOW/NOT APPLICABLE | | | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, one time | 319 | %26 | 301 | %24 | 170 | %14 | 300 | %24 | 156 | %13 | 1.246 | %100 |
| YES, several times | 2.280 | %46 | 1.193 | %24 | 523 | %11 | 519 | %11 | 419 | %8 | 4.934 | %100 |
| NO | 259 | %17 | 292 | %19 | 298 | %20 | 478 | %31 | 194 | %13 | 1.521 | %100 |
| TOTAL | 2.858 | %37 | 1.786 | %23 | 991 | %13 | 1.297 | %17 | 769 | %10 | 7.701 | |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 785,3 ; dof= 8.*

### Cross: …neglected, not taken seriously and/or considered as psychological? / …experienced worsening of the symptoms

| …NEGLECTED, NOT TAKEN SERIOUSLY AND/OR CONSIDERED AS PSYCHOLOGICAL? | …EXPERIENCED WORSENING OF THE SYMPTOMS | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | DEFINITELY | | PROBABLY | | PROBABLY NOT | | DEFINITELY NOT | | DON'T KNOW/NOT APPLICABLE | | | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, one time | 547 | %44 | 266 | %21 | 188 | %15 | 157 | %13 | 88 | %7 | 1.246 | %100 |
| YES, several times | 2.945 | %60 | 1.010 | %20 | 414 | %8 | 272 | %6 | 293 | %6 | 4.934 | %100 |
| NO | 475 | %31 | 358 | %24 | 254 | %17 | 295 | %19 | 139 | %9 | 1.521 | %100 |
| TOTAL | 3.967 | %52 | 1.634 | %21 | 856 | %11 | 724 | %9 | 520 | %7 | 7.701 | |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 570,3 ; dof= 8.*

**Only respondents who said that the rare disease has already been misdiagnosed**

**Cross: Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed. / … could not access all the information needed to make informed reproductive choices such as planning whether or not to have children, or deciding whether or not to conduct prenatal tests**

| HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | …COULD NOT ACCESS ALL THE INFORMATION NEEDED TO MAKE INFORMED REPRODUCTIVE CHOICES SUCH AS PLANNING WHETHER OR NOT TO HAVE CHILDREN, OR DECIDING WHETHER OR NOT TO CONDUCT PRENATAL TESTS | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | DEFINITELY | | PROBABLY | | PROBABLY NOT | | DEFINITELY NOT | | DON'T KNOW/NOT APPLICABLE | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, one time | 311 | %12 | 300 | %11 | 294 | %11 | 675 | %25 | 1.103 | %41 | 2.683 | %100 |
| YES, several times | 953 | %19 | 656 | %13 | 516 | %10 | 946 | %19 | 1.947 | %39 | 5.018 | %100 |
| NO | 0 | %0 | 0 | %0 | 0 | %0 | 0 | %0 | 0 | %0 | 0 | %100 |
| **TOTAL** | **1.264** | **%16** | **956** | **%12** | **810** | **%11** | **1.621** | **%21** | **3.050** | **%40** | **7.701** | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 99,5 ; dof= 4.*

**Cross: Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed. / … received inappropriate care, treatment or surgery**

| HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | …RECEIVED INAPPROPRIATE CARE, TREATMENT OR SURGERY | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | DEFINITELY | | PROBABLY | | PROBABLY NOT | | DEFINITELY NOT | | DON'T KNOW/NOT APPLICABLE | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, one time | 515 | %19 | 475 | %18 | 405 | %15 | 864 | %32 | 424 | %16 | 2.683 | %100 |
| YES, several times | 1.885 | %38 | 1.172 | %23 | 628 | %13 | 845 | %17 | 488 | %10 | 5.018 | %100 |
| NO | 0 | %0 | 0 | %0 | 0 | %0 | 0 | %0 | 0 | %0 | 0 | %100 |
| **TOTAL** | **2.400** | **%31** | **1.647** | **%21** | **1.033** | **%13** | **1.709** | **%22** | **912** | **%12** | **7.701** | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 464,6 ; dof= 4.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Only respondents who said that the rare disease has already been misdiagnosed**

**Cross: Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed. / …had a delayed access to the most appropriate care, treatment or surgery**

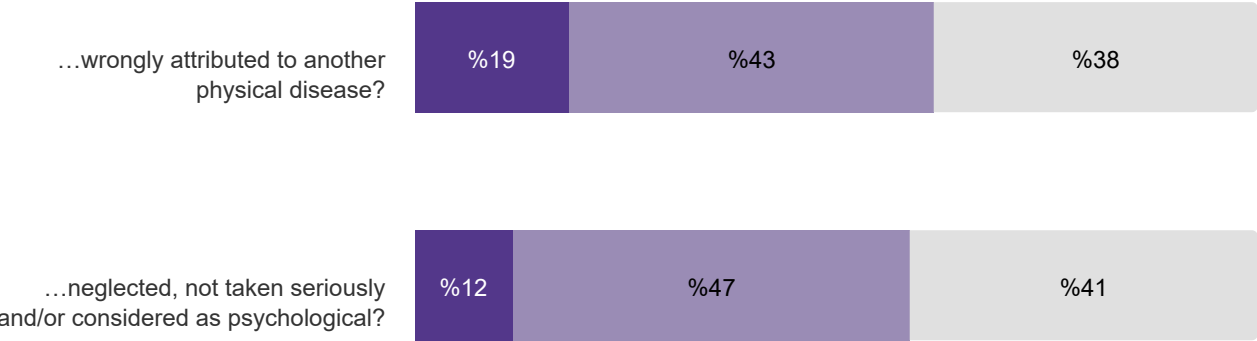| HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | …HAD A DELAYED ACCESS TO THE MOST APPROPRIATE CARE, TREATMENT OR SURGERY | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | DEFINITELY | | PROBABLY | | PROBABLY NOT | | DEFINITELY NOT | | DON'T KNOW/NOT APPLICABLE | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, one time | 821 | %31 | 655 | %24 | 357 | %13 | 517 | %19 | 333 | %12 | 2.683 | %100 |
| YES, several times | 2.559 | %51 | 1.228 | %24 | 430 | %9 | 456 | %9 | 345 | %7 | 5.018 | %100 |
| NO | 0 | %0 | 0 | %0 | 0 | %0 | 0 | %0 | 0 | %0 | 0 | %100 |
| TOTAL | 3.380 | %44 | 1.883 | %24 | 787 | %10 | 973 | %13 | 678 | %9 | 7.701 | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 408,4 ; dof= 4.*

**Cross: Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed. / …could not access appropriate care, treatment or surgery**
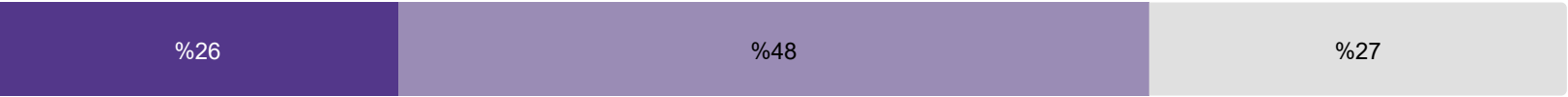
| HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | …COULD NOT ACCESS APPROPRIATE CARE, TREATMENT OR SURGERY | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | DEFINITELY | | PROBABLY | | PROBABLY NOT | | DEFINITELY NOT | | DON'T KNOW/NOT APPLICABLE | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, one time | 674 | %25 | 540 | %20 | 422 | %16 | 671 | %25 | 376 | %14 | 2.683 | %100 |
| YES, several times | 2.184 | %44 | 1.246 | %25 | 569 | %11 | 626 | %12 | 393 | %8 | 5.018 | %100 |
| NO | 0 | %0 | 0 | %0 | 0 | %0 | 0 | %0 | 0 | %0 | 0 | %100 |
| TOTAL | 2.858 | %37 | 1.786 | %23 | 991 | %13 | 1.297 | %17 | 769 | %10 | 7.701 | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 432,4 ; dof= 4.*

## Only respondents who said that the rare disease has already been misdiagnosed

**Cross: Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed. / …experienced worsening of the symptoms**

| HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | …EXPERIENCED WORSENING OF THE SYMPTOMS | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | DEFINITELY | | PROBABLY | | PROBABLY NOT | | DEFINITELY NOT | | DON'T KNOW/NOT APPLICABLE | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, one time | 1.022 | %38 | 583 | %22 | 370 | %14 | 426 | %16 | 282 | %11 | 2.683 | %100 |
| YES, several times | 2.945 | %59 | 1.051 | %21 | 486 | %10 | 298 | %6 | 238 | %5 | 5.018 | %100 |
| NO | 0 | %0 | 0 | %0 | 0 | %0 | 0 | %0 | 0 | %0 | 0 | %100 |
| TOTAL | 3.967 | %52 | 1.634 | %21 | 856 | %11 | 724 | %9 | 520 | %7 | 7.701 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 440,8 ; dof= 4.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

## Did it ever happen that the symptoms of the rare disease were…

| | YES, ONE TIME | YES, SEVERAL TIMES | NO | TOTAL |
|---|---|---|---|---|
| …wrongly attributed to another physical disease? | 1.950 | 4.520 | 4.016 | 10.486 |
| …neglected, not taken seriously and/or considered as psychological? | 1.246 | 4.934 | 4.306 | 10.486 |
| TOTAL | 3.196 | 9.454 | 8.322 | 20.972 |

## Did it ever happen that the symptoms of the rare disease were…



## Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed.

| | N |
|---|---|
| YES, one time | 2.683 |
| YES, several times | 5.018 |
| NO | 2.785 |
| TOTAL | 10.486 |

## Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed.

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Genetic test(s) looking for genetic changes (also called mutations or variants) / …wrongly attributed to another physical disease?**

| GENETIC TEST(S) LOOKING FOR GENETIC CHANGES (ALSO CALLED MUTATIONS OR VARIANTS) | …WRONGLY ATTRIBUTED TO ANOTHER PHYSICAL DISEASE? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | NO | | YES, ONE TIME | | YES, SEVERAL TIMES | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 2.330 | %42 | 969 | %18 | 2.191 | %40 | 5.490 | %100 |
| No | 1.369 | %33 | 812 | %19 | 1.990 | %48 | 4.171 | %100 |
| Don't know/don't remember | 317 | %38 | 169 | %20 | 339 | %41 | 825 | %100 |
| TOTAL | 4.016 | %38 | 1.950 | %19 | 4.520 | %43 | 10.486 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 97,5 ; dof= 4.*

**Cross: Genetic test(s) looking for genetic changes (also called mutations or variants) / …neglected, not taken seriously and/or considered as psychological?**

| GENETIC TEST(S) LOOKING FOR GENETIC CHANGES (ALSO CALLED MUTATIONS OR VARIANTS) | …NEGLECTED, NOT TAKEN SERIOUSLY AND/OR CONSIDERED AS PSYCHOLOGICAL? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | NO | | YES, ONE TIME | | YES, SEVERAL TIMES | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 2.503 | %46 | 606 | %11 | 2.381 | %43 | 5.490 | %100 |
| No | 1.453 | %35 | 529 | %13 | 2.189 | %52 | 4.171 | %100 |
| Don't know/don't remember | 350 | %42 | 111 | %13 | 364 | %44 | 825 | %100 |
| TOTAL | 4.306 | %41 | 1.246 | %12 | 4.934 | %47 | 10.486 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 117,9 ; dof= 4.*

**Cross: Genetic test(s) looking for genetic changes (also called mutations or variants) / Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed.**

| GENETIC TEST(S) LOOKING FOR GENETIC CHANGES (ALSO CALLED MUTATIONS OR VARIANTS) | HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 1.428 | %26 | 2.419 | %44 | 1.643 | %30 | 5.490 | %100 |
| No | 1.045 | %25 | 2.208 | %53 | 918 | %22 | 4.171 | %100 |
| Don't know/don't remember | 210 | %25 | 391 | %47 | 224 | %27 | 825 | %100 |
| TOTAL | 2.683 | %26 | 5.018 | %48 | 2.785 | %27 | 10.486 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 96,0 ; dof= 4.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Other test(s) such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc / …wrongly attributed to another physical disease?

| OTHER TEST(S) SUCH AS CLINICAL EXAMINATION(S), MEDICAL IMAGING (MRI, SCANS…), BIOPSY, BIOCHEMICAL TEST(S) (BLOOD OR URINE TESTS…), ETC | …WRONGLY ATTRIBUTED TO ANOTHER PHYSICAL DISEASE? | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | NO | | YES, ONE TIME | | YES, SEVERAL TIMES | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 3.539 | %37 | 1.771 | %19 | 4.172 | %44 | 9.482 | %100 |
| No | 345 | %47 | 136 | %19 | 252 | %34 | 733 | %100 |
| Don't know/don't remember | 132 | %49 | 43 | %16 | 96 | %35 | 271 | %100 |
| TOTAL | 4.016 | %38 | 1.950 | %19 | 4.520 | %43 | 10.486 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 44,3 ; dof= 4.*

## Cross: Other test(s) such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc / …neglected, not taken seriously and/or considered as psychological?

| OTHER TEST(S) SUCH AS CLINICAL EXAMINATION(S), MEDICAL IMAGING (MRI, SCANS…), BIOPSY, BIOCHEMICAL TEST(S) (BLOOD OR URINE TESTS…), ETC | …NEGLECTED, NOT TAKEN SERIOUSLY AND/OR CONSIDERED AS PSYCHOLOGICAL? | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | NO | | YES, ONE TIME | | YES, SEVERAL TIMES | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 3.848 | %41 | 1.139 | %12 | 4.495 | %47 | 9.482 | %100 |
| No | 329 | %45 | 83 | %11 | 321 | %44 | 733 | %100 |
| Don't know/don't remember | 129 | %48 | 24 | %9 | 118 | %44 | 271 | %100 |
| TOTAL | 4.306 | %41 | 1.246 | %12 | 4.934 | %47 | 10.486 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is significant. p-value= 0,0 ; Chi2= 11,0 ; dof= 4.*

## Cross: Other test(s) such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc / Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed.

| OTHER TEST(S) SUCH AS CLINICAL EXAMINATION(S), MEDICAL IMAGING (MRI, SCANS…), BIOPSY, BIOCHEMICAL TEST(S) (BLOOD OR URINE TESTS…), ETC | HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 2.421 | %26 | 4.619 | %49 | 2.442 | %26 | 9.482 | %100 |
| No | 196 | %27 | 290 | %40 | 247 | %34 | 733 | %100 |
| Don't know/don't remember | 66 | %24 | 109 | %40 | 96 | %35 | 271 | %100 |
| TOTAL | 2.683 | %26 | 5.018 | %48 | 2.785 | %27 | 10.486 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 40,2 ; dof= 4.*

## Cross: …you could not afford it? / …wrongly attributed to another physical disease?

Have you ever needed a genetic test but could not access it because…

| …YOU COULD NOT AFFORD IT? | …WRONGLY ATTRIBUTED TO ANOTHER PHYSICAL DISEASE? | | | | | | | |
| | NO | | YES, ONE TIME | | YES, SEVERAL TIMES | | TOTAL | |
| | N | % | N | % | N | % | N | % |
|---|---|---|---|---|---|---|---|---|
| Yes | 228 | %20 | 174 | %16 | 715 | %64 | 1.117 | %100 |
| No | 2.997 | %42 | 1.355 | %19 | 2.764 | %39 | 7.116 | %100 |
| Not relevant | 791 | %35 | 421 | %19 | 1.041 | %46 | 2.253 | %100 |
| TOTAL | 4.016 | %38 | 1.950 | %19 | 4.520 | %43 | 10.486 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 280,9 ; dof= 4.*

## Cross: …you could not afford it? / …neglected, not taken seriously and/or considered as psychological?

Have you ever needed a genetic test but could not access it because…

| …YOU COULD NOT AFFORD IT? | …NEGLECTED, NOT TAKEN SERIOUSLY AND/OR CONSIDERED AS PSYCHOLOGICAL? | | | | | | | |
| | NO | | YES, ONE TIME | | YES, SEVERAL TIMES | | TOTAL | |
| | N | % | N | % | N | % | N | % |
|---|---|---|---|---|---|---|---|---|
| Yes | 228 | %20 | 113 | %10 | 776 | %69 | 1.117 | %100 |
| No | 3.266 | %46 | 830 | %12 | 3.020 | %42 | 7.116 | %100 |
| Not relevant | 812 | %36 | 303 | %13 | 1.138 | %51 | 2.253 | %100 |
| TOTAL | 4.306 | %41 | 1.246 | %12 | 4.934 | %47 | 10.486 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 335,4 ; dof= 4.*

## Cross: …you could not afford it? / Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed.

Have you ever needed a genetic test but could not access it because…

| …YOU COULD NOT AFFORD IT? | HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | | | | | | | |
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
|---|---|---|---|---|---|---|---|---|
| Yes | 233 | %21 | 760 | %68 | 124 | %11 | 1.117 | %100 |
| No | 1.846 | %26 | 3.109 | %44 | 2.161 | %30 | 7.116 | %100 |
| Not relevant | 604 | %27 | 1.149 | %51 | 500 | %22 | 2.253 | %100 |
| TOTAL | 2.683 | %26 | 5.018 | %48 | 2.785 | %27 | 10.486 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 292,5 ; dof= 4.*

## Cross: …it was not available in your country? / …wrongly attributed to another physical disease?

Have you ever needed a genetic test but could not access it because…

| …IT WAS NOT AVAILABLE IN YOUR COUNTRY? | …WRONGLY ATTRIBUTED TO ANOTHER PHYSICAL DISEASE? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | NO | | YES, ONE TIME | | YES, SEVERAL TIMES | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 305 | %25 | 195 | %16 | 697 | %58 | 1.197 | %100 |
| No | 2.848 | %42 | 1.284 | %19 | 2.696 | %39 | 6.828 | %100 |
| Not relevant | 863 | %35 | 471 | %19 | 1.127 | %46 | 2.461 | %100 |
| TOTAL | 4.016 | %38 | 1.950 | %19 | 4.520 | %43 | 10.486 | |

■ *Under-represented elements*   ■ *Over-represented elements*

The relationship is very significant. p-value= < 0,01 ; Chi2= 171,2 ; dof= 4.

## Cross: …it was not available in your country? / …neglected, not taken seriously and/or considered as psychological?

Have you ever needed a genetic test but could not access it because…

| …IT WAS NOT AVAILABLE IN YOUR COUNTRY? | …NEGLECTED, NOT TAKEN SERIOUSLY AND/OR CONSIDERED AS PSYCHOLOGICAL? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | NO | | YES, ONE TIME | | YES, SEVERAL TIMES | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 348 | %29 | 135 | %11 | 714 | %60 | 1.197 | %100 |
| No | 3.053 | %45 | 788 | %12 | 2.987 | %44 | 6.828 | %100 |
| Not relevant | 905 | %37 | 323 | %13 | 1.233 | %50 | 2.461 | %100 |
| TOTAL | 4.306 | %41 | 1.246 | %12 | 4.934 | %47 | 10.486 | |

■ *Under-represented elements*   ■ *Over-represented elements*

The relationship is very significant. p-value= < 0,01 ; Chi2= 140,4 ; dof= 4.

## Cross: …it was not available in your country? / Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed.

Have you ever needed a genetic test but could not access it because…

| …IT WAS NOT AVAILABLE IN YOUR COUNTRY? | HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 252 | %21 | 756 | %63 | 189 | %16 | 1.197 | %100 |
| No | 1.769 | %26 | 3.016 | %44 | 2.043 | %30 | 6.828 | %100 |
| Not relevant | 662 | %27 | 1.246 | %51 | 553 | %22 | 2.461 | %100 |
| TOTAL | 2.683 | %26 | 5.018 | %48 | 2.785 | %27 | 10.486 | |

■ *Under-represented elements*   ■ *Over-represented elements*

The relationship is very significant. p-value= < 0,01 ; Chi2= 190,3 ; dof= 4.

## Cross: …healthcare professionals were reluctant or not sufficiently informed? / …wrongly attributed to another physical disease?

Have you ever needed a genetic test but could not access it because...

| …HEALTHCARE PROFESSIONALS WERE RELUCTANT OR NOT SUFFICIENTLY INFORMED? | …WRONGLY ATTRIBUTED TO ANOTHER PHYSICAL DISEASE? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | NO | | YES, ONE TIME | | YES, SEVERAL TIMES | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 594 | %21 | 463 | %17 | 1.748 | %62 | 2.805 | %100 |
| No | 2.641 | %48 | 1.069 | %19 | 1.846 | %33 | 5.556 | %100 |
| Not relevant | 781 | %37 | 418 | %20 | 926 | %44 | 2.125 | %100 |
| TOTAL | 4.016 | %38 | 1.950 | %19 | 4.520 | %43 | 10.486 | |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 715,0 ; dof= 4.*

## Cross: …healthcare professionals were reluctant or not sufficiently informed? / …neglected, not taken seriously and/or considered as psychological?

Have you ever needed a genetic test but could not access it because...

| …HEALTHCARE PROFESSIONA… WERE RELUCTANT OR NOT SUFFICIENTLY INFORMED? | …NEGLECTED, NOT TAKEN SERIOUSLY AND/OR CONSIDERED AS PSYCHOLOGICAL? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | NO | | YES, ONE TIME | | YES, SEVERAL TIMES | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 552 | %20 | 329 | %12 | 1.924 | %69 | 2.805 | %100 |
| No | 2.913 | %52 | 656 | %12 | 1.987 | %36 | 5.556 | %100 |
| Not relevant | 841 | %40 | 261 | %12 | 1.023 | %48 | 2.125 | %100 |
| TOTAL | 4.306 | %41 | 1.246 | %12 | 4.934 | %47 | 10.486 | |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 916,3 ; dof= 4.*

## Cross: …healthcare professionals were reluctant or not sufficiently informed? / Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed.

Have you ever needed a genetic test but could not access it because...

| …HEALTHCARE PROFESSIONALS WERE RELUCTANT OR NOT SUFFICIENTLY INFORMED? | HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 645 | %23 | 1.873 | %67 | 287 | %10 | 2.805 | %100 |
| No | 1.453 | %26 | 2.115 | %38 | 1.988 | %36 | 5.556 | %100 |
| Not relevant | 585 | %28 | 1.030 | %48 | 510 | %24 | 2.125 | %100 |
| TOTAL | 2.683 | %26 | 5.018 | %48 | 2.785 | %27 | 10.486 | |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 797,1 ; dof= 4.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: To your knowledge, the genetic test(s) that were conducted targeted… / …wrongly attributed to another physical disease?**

| TO YOUR KNOWLEDGE, THE GENETIC TEST(S) THAT WERE CONDUCTED TARGETED… | …WRONGLY ATTRIBUTED TO ANOTHER PHYSICAL DISEASE? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | NO | | YES, ONE TIME | | YES, SEVERAL TIMES | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Only one gene | 632 | %43 | 269 | %18 | 559 | %38 | 1.460 | %100 |
| Several genes at the same time (gene panel sequencing) | _641_ | _%37_ | 307 | %18 | _783_ | _%45_ | 1.731 | %100 |
| The whole DNA (Whole Genome Sequencing) | 398 | %45 | 157 | %18 | _325_ | _%37_ | 880 | %100 |
| All the genes (Whole Exome Sequencing) | 221 | %39 | 97 | %17 | _249_ | _%44_ | 567 | %100 |
| A tumour (genetic profiling of a tumour) | 48 | %36 | 32 | %24 | 55 | %41 | 135 | %100 |
| Other (epigenome, RNA, etc.) | 47 | %40 | 17 | %15 | 53 | %45 | 117 | %100 |
| Don't know | 667 | %44 | _235_ | _%16_ | 609 | %40 | 1.511 | %100 |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 44,3 ; dof= 12.*

**Cross: To your knowledge, the genetic test(s) that were conducted targeted… / …neglected, not taken seriously and/or considered as psychological?**

| TO YOUR KNOWLEDGE, THE GENETIC TEST(S) THAT WERE CONDUCTED TARGETED… | …NEGLECTED, NOT TAKEN SERIOUSLY AND/OR CONSIDERED AS PSYCHOLOGICAL? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | NO | | YES, ONE TIME | | YES, SEVERAL TIMES | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Only one gene | 670 | %46 | 153 | %10 | 637 | %44 | 1.460 | %100 |
| Several genes at the same time (gene panel sequencing) | _735_ | _%42_ | 177 | %10 | _819_ | _%47_ | 1.731 | %100 |
| The whole DNA (Whole Genome Sequencing) | _441_ | _%50_ | 110 | %13 | _329_ | _%37_ | 880 | %100 |
| All the genes (Whole Exome Sequencing) | 271 | %48 | 68 | %12 | 228 | %40 | 567 | %100 |
| A tumour (genetic profiling of a tumour) | 52 | %39 | 21 | %16 | 62 | %46 | 135 | %100 |
| Other (epigenome, RNA, etc.) | 43 | %37 | _20_ | _%17_ | 54 | %46 | 117 | %100 |
| Don't know | 696 | %46 | 169 | %11 | 646 | %43 | 1.511 | %100 |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 37,2 ; dof= 12.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: To your knowledge, the genetic test(s) that were conducted targeted… / Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed.**

| TO YOUR KNOWLEDGE, THE GENETIC TEST(S) THAT WERE CONDUCTED TARGETED… | HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Only one gene | 389 | %27 | 624 | %43 | 447 | %31 | 1.460 | %100 |
| Several genes at the same time (gene panel sequencing) | 449 | %26 | _844_ | _%49_ | **438** | **%25** | 1.731 | %100 |
| The whole DNA (Whole Genome Sequencing) | 225 | %26 | 363 | %41 | _292_ | _%33_ | 880 | %100 |
| All the genes (Whole Exome Sequencing) | 141 | %25 | 269 | %47 | 157 | %28 | 567 | %100 |
| A tumour (genetic profiling of a tumour) | 31 | %23 | 67 | %50 | 37 | %27 | 135 | %100 |
| Other (epigenome, RNA, etc.) | 32 | %27 | 58 | %50 | 27 | %23 | 117 | %100 |
| Don't know | **363** | **%24** | 672 | %44 | 476 | %32 | 1.511 | %100 |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 37,6 ; dof= 12.*

**Cross: Did you ever request a private company or laboratory to conduct genetic testing to diagnose the disease? / …wrongly attributed to another physical disease?**

| DID YOU EVER REQUEST A PRIVATE COMPANY OR LABORATORY TO CONDUCT GENETIC TESTING TO DIAGNOSE THE DISEASE? | …WRONGLY ATTRIBUTED TO ANOTHER PHYSICAL DISEASE? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| YES, one time | 121 | %21 | _260_ | _%44_ | **209** | **%35** | 590 | %100 |
| YES, several times | 40 | %15 | _152_ | _%57_ | **74** | **%28** | 266 | %100 |
| NO, never | 808 | %17 | **1.779** | **%38** | _2.047_ | _%44_ | 4.634 | %100 |
| **TOTAL** | **969** | **%18** | **2.191** | **%40** | **2.330** | **%42** | **5.490** | **%100** |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 52,4 ; dof= 4.*

## Cross: Did you ever request a private company or laboratory to conduct genetic testing to diagnose the disease? / …neglected, not taken seriously and/or considered as psychological?

| DID YOU EVER REQUEST A PRIVATE COMPANY OR LABORATORY TO CONDUCT GENETIC TESTING TO DIAGNOSE THE DISEASE? | …NEGLECTED, NOT TAKEN SERIOUSLY AND/OR CONSIDERED AS PSYCHOLOGICAL? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| YES, one time | 75 | %13 | 277 | %47 | 238 | %40 | 590 | %100 |
| YES, several times | 32 | %12 | 144 | %54 | 90 | %34 | 266 | %100 |
| NO, never | 499 | %11 | 1.960 | %42 | 2.175 | %47 | 4.634 | %100 |
| TOTAL | 606 | %11 | 2.381 | %43 | 2.503 | %46 | 5.490 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 25,6 ; dof= 4.*

## Cross: Did you ever request a private company or laboratory to conduct genetic testing to diagnose the disease? / Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed.

| DID YOU EVER REQUEST A PRIVATE COMPANY OR LABORATORY TO CONDUCT GENETIC TESTING TO DIAGNOSE THE DISEASE? | HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| YES, one time | 151 | %26 | 297 | %50 | 142 | %24 | 590 | %100 |
| YES, several times | 49 | %18 | 166 | %62 | 51 | %19 | 266 | %100 |
| NO, never | 1.228 | %26 | 1.956 | %42 | 1.450 | %31 | 4.634 | %100 |
| TOTAL | 1.428 | %26 | 2.419 | %44 | 1.643 | %30 | 5.490 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 55,5 ; dof= 4.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: In general, how satisfied are you with how the results of the GENETIC TESTS were given to you? / Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed.**

| IN GENERAL, HOW SATISFIED ARE YOU WITH HOW THE RESULTS OF THE GENETIC TESTS WERE GIVEN TO YOU? | HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Very Dissatisfied | 155 | %27 | 283 | %49 | 134 | %23 | 572 | %100 |
| Dissatisfied | 159 | %26 | 335 | %54 | 129 | %21 | 623 | %100 |
| Neither satisfied nor dissatisfied | 306 | %26 | 572 | %49 | 281 | %24 | 1.159 | %100 |
| Satisfied | 518 | %27 | 773 | %40 | 639 | %33 | 1.930 | %100 |
| Very Satisfied | 222 | %24 | 323 | %35 | 378 | %41 | 923 | %100 |
| Don't know | 68 | %24 | 133 | %47 | 82 | %29 | 283 | %100 |
| **TOTAL** | **1.428** | **%26** | **2.419** | **%44** | **1.643** | **%30** | **5.490** | |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 134,4 ; dof= 10.*

**Cross: After the tests were performed, were you offered genetic counselling (e.g. given information about how your genetic condition might affect you and your family)? / Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed.**

| AFTER THE TESTS WERE PERFORMED, WERE YOU OFFERED GENETIC COUNSELLING (E.G. GIVEN INFORMATION ABOUT HOW YOUR GENETIC CONDITION MIGHT AFFECT YOU AND YOUR FAMILY)? | HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| YES, with a genetic counsellor or clinical geneticist | 565 | %26 | 813 | %38 | 759 | %36 | 2.137 | %100 |
| YES, by a healthcare professional | 313 | %27 | 496 | %42 | 370 | %31 | 1.179 | %100 |
| NO, I wasn't offered genetic counselling | 452 | %26 | 946 | %53 | 372 | %21 | 1.770 | %100 |
| Not sure / Don't remember | 98 | %24 | 164 | %41 | 142 | %35 | 404 | %100 |
| **TOTAL** | **1.428** | **%26** | **2.419** | **%44** | **1.643** | **%30** | **5.490** | |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 129,8 ; dof= 6.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Genetic tests / Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed.**

| | HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | | | | | | | |
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| **GENETIC TESTS** | N | % | N | % | N | % | N | % |
|---|---|---|---|---|---|---|---|---|
| Yes | 219 | %26 | 397 | %48 | 215 | %26 | 831 | %100 |
| No | 1.171 | %26 | 1.967 | %44 | 1.377 | %30 | 4.515 | %100 |
| Don't know | 37 | %26 | 55 | %38 | 51 | %36 | 143 | %100 |
| **TOTAL** | **1.427** | **%26** | **2.419** | **%44** | **1.643** | **%30** | **5.489** | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is significant. p-value= 0,0 ; Chi2= 10,5 ; dof= 4.*

**Cross: Other diagnostic tests such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc. / Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed.**

| | HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | | | | | | | |
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| **OTHER DIAGNOSTIC TESTS SUCH AS CLINICAL EXAMINATION(S), MEDICAL IMAGING (MRI, SCANS…), BIOPSY, BIOCHEMICAL TEST(S) (BLOOD OR URINE TESTS…), ETC.** | N | % | N | % | N | % | N | % |
|---|---|---|---|---|---|---|---|---|
| Yes | 337 | %24 | 768 | %55 | 298 | %21 | 1.403 | %100 |
| No | 2.036 | %26 | 3.780 | %48 | 2.092 | %26 | 7.908 | %100 |
| Don't know | 48 | %28 | 71 | %42 | 51 | %30 | 170 | %100 |
| **TOTAL** | **2.421** | **%26** | **4.619** | **%49** | **2.441** | **%26** | **9.481** | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 29,2 ; dof= 4.*

**Cross: Additional advice from a healthcare professional specialised in the rare disease (in person or virtually) / Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed.**

| ADDITIONAL ADVICE FROM A HEALTHCARE PROFESSIONAL SPECIALISED IN THE RARE DISEASE (IN PERSON OR VIRTUALLY) | HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 529 | %25 | 1.086 | %52 | 468 | %22 | 2.083 | %100 |
| No | 2.095 | %26 | 3.856 | %47 | 2.243 | %27 | 8.194 | %100 |
| Don't know | 59 | %28 | 76 | %36 | 74 | %35 | 209 | %100 |
| **TOTAL** | **2.683** | **%26** | **5.018** | **%48** | **2.785** | **%27** | **10.486** | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 36,8 ; dof= 4.*

**Cross: Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed. / … psychological support**

| HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | …PSYCHOLOGICAL SUPPORT | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, one time | 232 | %9 | 237 | %9 | 205 | %8 | 802 | %30 | 1.207 | %45 | 2.683 | %100 |
| YES, several times | 358 | %7 | 376 | %7 | 586 | %12 | 1.271 | %25 | 2.427 | %48 | 5.018 | %100 |
| NO | 332 | %12 | 342 | %12 | 161 | %6 | 1.092 | %39 | 858 | %31 | 2.785 | %100 |
| **TOTAL** | **922** | **%9** | **955** | **%9** | **952** | **%9** | **3.165** | **%30** | **4.492** | **%43** | **10.486** | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 416,1 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: …care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc. / Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed.**

| …CARE COORDINATION SUPPORT SUCH AS HELP TO FIND THE NECESSARY INFORMATION ON THE DISEASE AND THE RIGHT PROFESSIONALS, ARRANGING APPOINTMENTS WITH DIFFERENT HEALTH PROVIDERS, ETC. | HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| YES and enough to meet my needs | 525 | %25 | 682 | %33 | 876 | %42 | 2.083 | %100 |
| YES but it is/was not needed | 106 | %27 | 127 | %32 | 158 | %40 | 391 | %100 |
| YES but NOT enough to meet my needs | 395 | %27 | 744 | %51 | 324 | %22 | 1.463 | %100 |
| NO but it is/was NOT needed | 423 | %26 | 556 | %34 | 648 | %40 | 1.627 | %100 |
| NO but it is/was needed | 1.234 | %25 | 2.909 | %59 | 779 | %16 | 4.922 | %100 |
| **TOTAL** | **2.683** | **%26** | **5.018** | **%48** | **2.785** | **%27** | **10.486** | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 866,1 ; dof= 8.*

**Cross: Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed. / …financial support including social security benefits**

| HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | …FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS | | | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, one time | 339 | %13 | 61 | %2 | 306 | %11 | 958 | %36 | 1.005 | %38 | 2.669 | %100 |
| YES, several times | 526 | %11 | 79 | %2 | 633 | %13 | 1.443 | %29 | 2.291 | %46 | 4.972 | %100 |
| NO | 540 | %19 | 103 | %4 | 293 | %11 | 1.143 | %41 | 693 | %25 | 2.772 | %100 |
| **TOTAL** | **1.405** | **%13** | **243** | **%2** | **1.232** | **%12** | **3.544** | **%34** | **3.989** | **%38** | **10.413** | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

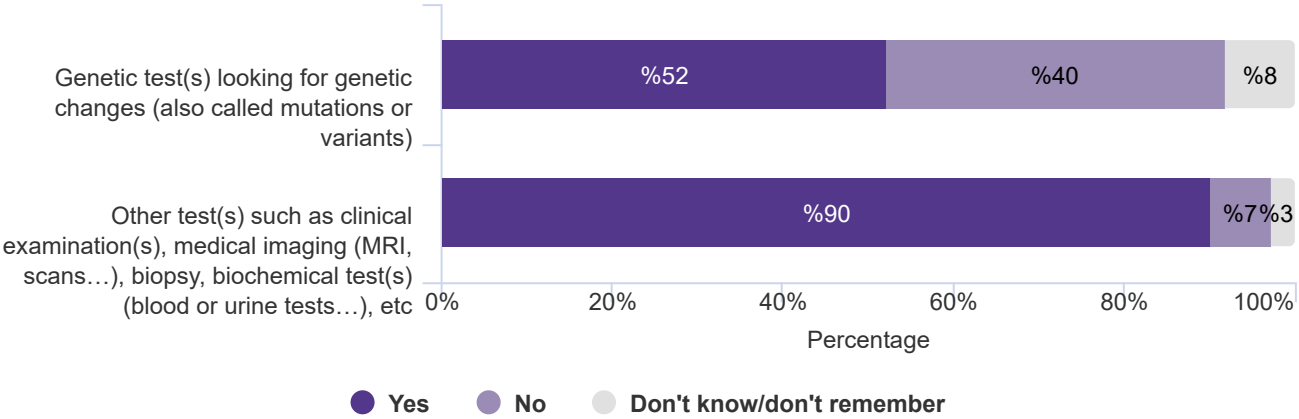*The relationship is very significant. p-value= < 0,01 ; Chi2= 436,6 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease? / Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed.**

| ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| YES, through a patient organisation | 1.389 | %26 | 2.465 | %46 | 1.472 | %28 | 5.326 | %100 |
| YES, through online communities | 1.264 | %25 | 2.602 | %52 | 1.126 | %23 | 4.992 | %100 |
| YES, through local networks (e.g. schools) | 103 | %24 | 224 | %51 | 109 | %25 | 436 | %100 |
| NO, because of accessibility issues (e.g. language or technical barriers) | 42 | %22 | 100 | %53 | 48 | %25 | 190 | %100 |
| NO, because I have not been able to find other people with the same disease | 325 | %25 | 634 | %48 | 351 | %27 | 1.310 | %100 |
| NO, because I don't want to | 142 | %26 | 215 | %39 | 190 | %35 | 547 | %100 |
| Other, specify... | 128 | %25 | 233 | %45 | 153 | %30 | 514 | %100 |
| **TOTAL** | **2.683** | **%26** | **5.018** | **%48** | **2.785** | **%27** | **10.486** | |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 84,3 ; dof= 12.*

# Chapter 11.

## Misdiagnosis

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Which tests were performed as part of the diagnosis of the rare disease?

| | YES | NO | DON'T KNOW/… REMEM… | TOTAL |
|---|---|---|---|---|
| Genetic test(s) looking for genetic changes (also called mutations or variants) | 5.490 | 4.171 | 825 | 10.486 |
| Other test(s) such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc | 9.482 | 733 | 271 | 10.486 |

## Which tests were performed as part of the diagnosis of the rare disease?



Genetic test(s) looking for genetic changes (also called mutations or variants): %52 / %40 / %8

Other test(s) such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc: %90 / %7 / %3

● Yes   ● No   ● Don't know/don't remember

## Cross: Genetic diseases / Genetic test(s) looking for genetic changes (also called mutations or variants)

| | GENETIC TEST(S) LOOKING FOR GENETIC CHANGES (ALSO CALLED MUTATIONS OR VARIANTS) | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW/DON'T REMEMBER | | TOTAL | |
| GENETIC DISEASES | N | % | N | % | N | % | N | % |
| Genetic diseases | 3.862 | %71 | 1.338 | %25 | 247 | %5 | 5.447 | %100 |
| Non Genetic diseases | 540 | %21 | 1.760 | %67 | 327 | %12 | 2.627 | %100 |
| **TOTAL** | **4.402** | **%55** | **3.098** | **%38** | **574** | **%7** | **8.074** | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 1.811,7 ; dof= 2.*

**Only respondents who had genetic tests**

## Have you ever needed a genetic test but could not access it because…

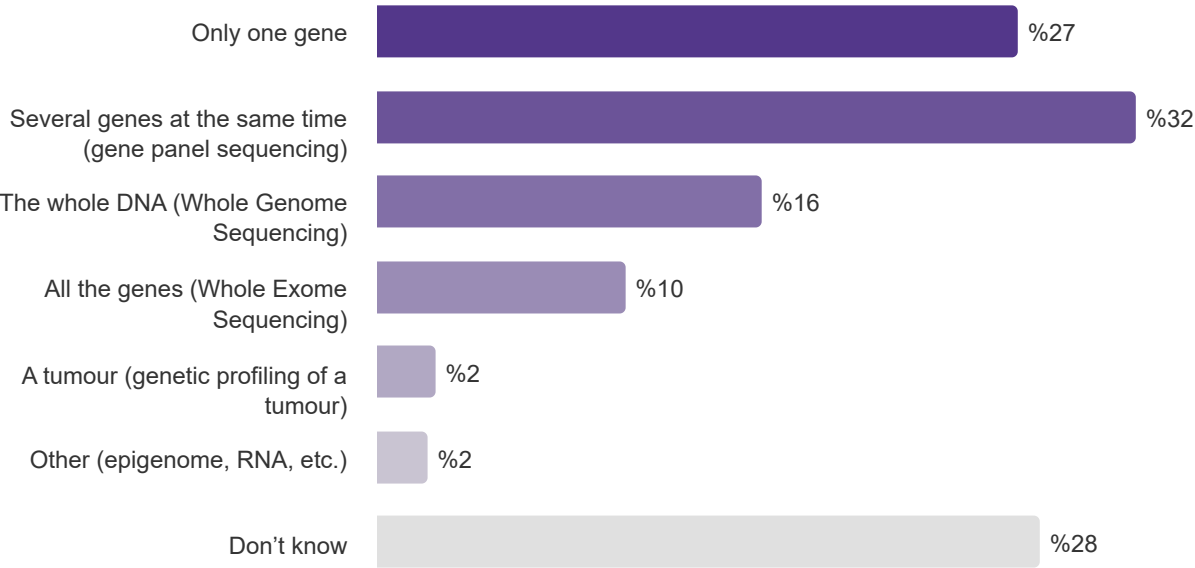| | YES | NO | NOT RELEVANT | TOTAL |
|---|---|---|---|---|
| …you could not afford it? | 1.117 | 7.116 | 2.253 | 10.486 |
| …it was not available in your country? | 1.197 | 6.828 | 2.461 | 10.486 |
| …healthcare professionals were reluctant or not sufficiently informed? | 2.805 | 5.556 | 2.125 | 10.486 |

## Have you ever needed a genetic test but could not access it because…

| …you could not afford it? | %11 | %68 | %21 |
|---|---|---|---|
| …it was not available in your country? | %11 | %65 | %23 |
| …healthcare professionals were reluctant or not sufficiently informed? | %27 | %53 | %20 |

● Yes  ● No  ● Not relevant
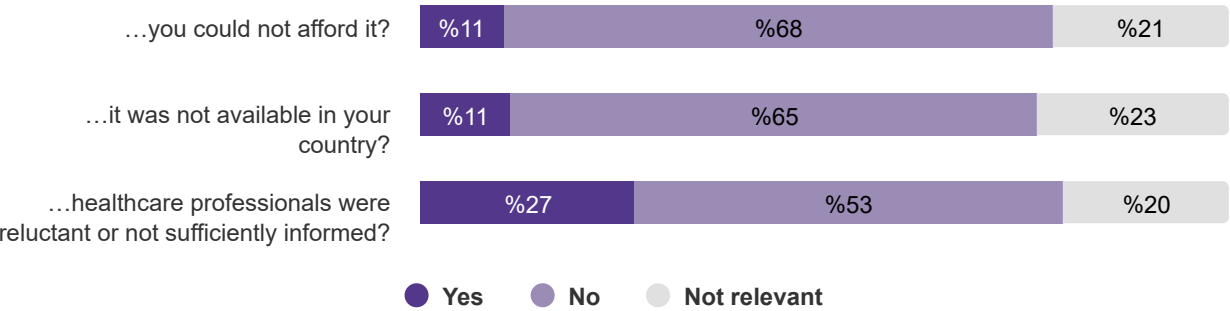
## To your knowledge, the genetic test(s) that were conducted targeted…

| | N |
|---|---|
| Only one gene | 1.460 |
| Several genes at the same time (gene panel sequencing) | 1.731 |
| The whole DNA (Whole Genome Sequencing) | 880 |
| All the genes (Whole Exome Sequencing) | 567 |
| A tumour (genetic profiling of a tumour) | 135 |
| Other (epigenome, RNA, etc.) | 117 |
| Don't know | 1.511 |
| **TOTAL** | **5.490** |

## To your knowledge, the genetic test(s) that were conducted targeted…

| | |
|---|---|
| Only one gene | %27 |
| Several genes at the same time (gene panel sequencing) | %32 |
| The whole DNA (Whole Genome Sequencing) | %16 |
| All the genes (Whole Exome Sequencing) | %10 |
| A tumour (genetic profiling of a tumour) | %2 |
| Other (epigenome, RNA, etc.) | %2 |
| Don't know | %28 |

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Only respondents who had genetic tests**

### Did you ever request a private company or laboratory to conduct genetic testing to diagnose the disease?

|  | N |
|---|---|
| YES, one time | 590 |
| YES, several times | 266 |
| NO, never | 4.634 |
| **TOTAL** | **5.490** |

### Did you ever request a private company or laboratory to conduct genetic testing to diagnose the disease?

| | |
|---|---|
| YES, one time | %11 |
| YES, several times | %5 |
| NO, never | %84 |

### In general, how satisfied are you with how the results of the GENETIC TESTS were given to you?

|  | N |
|---|---|
| Very Dissatisfied | 572 |
| Dissatisfied | 623 |
| Neither satisfied nor dissatisfied | 1.159 |
| Satisfied | 1.930 |
| Very Satisfied | 923 |
| Don't know | 283 |
| **TOTAL** | **5.490** |

### In general, how satisfied are you with how the results of the GENETIC TESTS were given to you?

| | |
|---|---|
| Very Dissatisfied | %10 |
| Dissatisfied | %11 |
| Neither satisfied nor dissatisfied | %21 |
| Satisfied | %35 |
| Very Satisfied | %17 |
| Don't know | %5 |
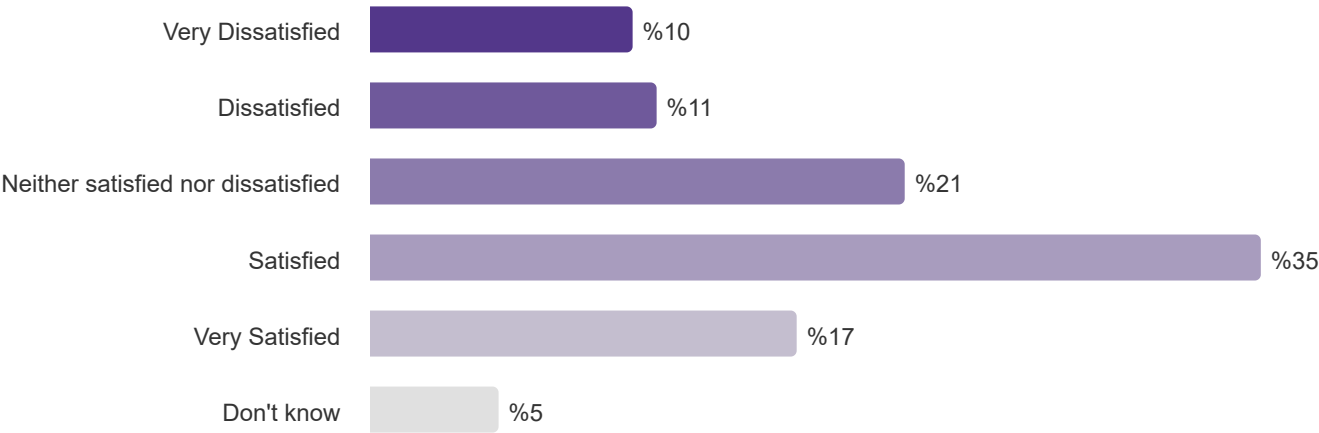
**Only respondents who had genetic tests**

**After the tests were performed, were you offered genetic counselling (e.g. given information about how your genetic condition might affect you and your family)?**

|  | N |
|---|---|
| YES, with a genetic counsellor or clinical geneticist | 2.137 |
| YES, by a healthcare professional | 1.179 |
| NO, I wasn't offered genetic counselling | 1.770 |
| Not sure / Don't remember | 404 |
| **TOTAL** | **5.490** |

**After the tests were performed, were you offered genetic counselling (e.g. given information about how your genetic condition might affect you and your family)?**

- YES, with a genetic counsellor or clinical geneticist — %39
- YES, by a healthcare professional — %21
- NO, I wasn't offered genetic counselling — %32
- Not sure / Don't remember — %7

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses
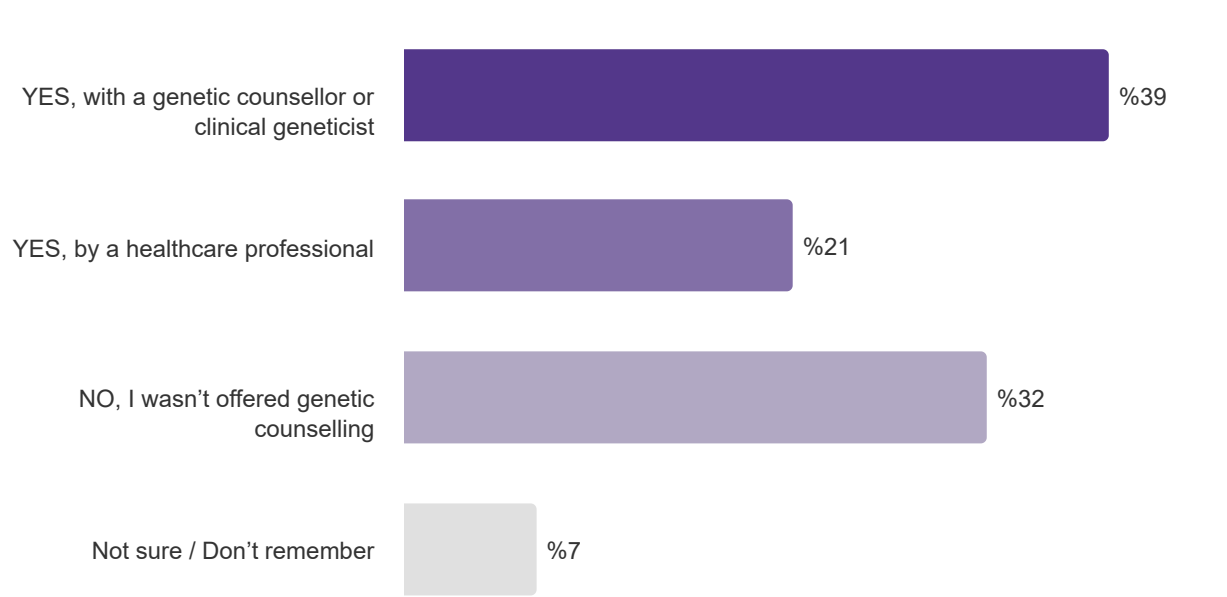
**Only respondents who had genetic tests**

| Genetic test(s) looking for genetic changes (also called mutations or variants) | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| Yes | 0,7 | 4.023 | 3,7 | 3.655 | 4,2 | 2.451 | 3,8 | 4.096 | 5,6 | 3.565 |
| No | 0,2 | 3.263 | 3,4 | 3.138 | 3,7 | 1.564 | 3,6 | 3.216 | 3,9 | 2.506 |
| Don't know/don't remember | 0,5 | 534 | 2,1 | 529 | 2,2 | 320 | 2,5 | 531 | 2,9 | 436 |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

| Have you ever needed a genetic test but could not access it because… ...you could not afford it? | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| Yes | 0,4 | 812 | 5,3 | 705 | 6,1 | 334 | 5,6 | 742 | 7,0 | 513 |
| No | 0,6 | 5.349 | 3,4 | 5.028 | 3,7 | 3.148 | 3,3 | 5.442 | 4,7 | 4.639 |
| Not relevant | 0,3 | 1.659 | 3,1 | 1.589 | 3,6 | 853 | 3,6 | 1.659 | 4,1 | 1.355 |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

## Have you ever needed a genetic test but could not access it because...

| …it was not available in your country? | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| Yes | 0,7 | 866 | 4,4 | 765 | 5,0 | 409 | 5,1 | 830 | 6,8 | 624 |
| No | 0,5 | 5.170 | 3,5 | 4.830 | 3,8 | 2.972 | 3,4 | 5.212 | 4,6 | 4.405 |
| Not relevant | 0,4 | 1.784 | 3,2 | 1.727 | 3,7 | 954 | 3,5 | 1.801 | 4,1 | 1.478 |

■ *Under-represented elements*　　■ *Over-represented elements*

## Have you ever needed a genetic test but could not access it because...

| …healthcare professionals were reluctant or not sufficiently informed? | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| Yes | 0,6 | 2.081 | 5,1 | 1.874 | 6,0 | 984 | 6,1 | 2.014 | 7,7 | 1.493 |
| No | 0,6 | 4.167 | 2,9 | 3.930 | 3,2 | 2.513 | 2,6 | 4.247 | 3,9 | 3.664 |
| Not relevant | 0,2 | 1.572 | 3,0 | 1.518 | 3,2 | 838 | 3,2 | 1.582 | 3,8 | 1.350 |

■ *Under-represented elements*　　■ *Over-represented elements*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

| To your knowledge, the genetic test(s) that were conducted targeted… | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| Only one gene | 0,7 | 1.078 | 4,1 | 973 | 4,3 | 682 | 3,3 | 1.097 | 5,4 | 995 |
| Several genes at the same time (gene panel sequencing) | 0,8 | 1.328 | 4,1 | 1.202 | **5,3** | 800 | 4,5 | 1.318 | **6,5** | 1.130 |
| The whole DNA (Whole Genome Sequencing) | 0,6 | 653 | 3,1 | 588 | **3,3** | 413 | 3,6 | 674 | 4,9 | 576 |
| All the genes (Whole Exome Sequencing) | 0,5 | 429 | **2,9** | 377 | **3,1** | 236 | 4,7 | 408 | 5,3 | 353 |
| A tumour (genetic profiling of a tumour) | 0,2 | 99 | 3,4 | 109 | 3,5 | 55 | 2,7 | 95 | **3,1** | 83 |
| Other (epigenome, RNA, etc.) | 0,7 | 85 | 5,1 | 73 | 5,2 | 48 | 5,9 | 86 | 6,3 | 67 |
| Don't know | 0,8 | 1.065 | 3,7 | 986 | 4,0 | 636 | 3,8 | 1.101 | 5,4 | 931 |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is not significant. p-value= 1,0 ; Fisher= 0,3.*

*Inter variance= 11,4. Intra variance= 43,7.*

Rare Barometer
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RARE DISEASES EUROPE

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

| Did you ever request a private company or laboratory to conduct genetic testing to diagnose the disease? | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| YES, one time | 0,7 | 437 | 3,6 | 372 | 4,0 | 215 | 3,9 | 434 | 4,9 | 373 |
| YES, several times | 0,2 | 196 | 3,9 | 170 | 4,3 | 91 | 3,2 | 189 | 4,7 | 162 |
| NO, never | 0,7 | 3.390 | 3,8 | 3.113 | 4,2 | 2.145 | 3,8 | 3.473 | 5,7 | 3.030 |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is not significant. p-value= 0,6 ; Fisher= 0,6.*

*Inter variance= 26,0. Intra variance= 46,9.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

| In general, how satisfied are you with how the results of the GENETIC TESTS were given to you? | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| Very Dissatisfied | 0,4 | 434 | 3,2 | 385 | 4,5 | 249 | 3,9 | 428 | 5,6 | 362 |
| Dissatisfied | 0,8 | 475 | 4,3 | 430 | 5,1 | 258 | 4,8 | 470 | 6,1 | 373 |
| Neither satisfied nor dissatisfied | 0,7 | 845 | 4,4 | 734 | 4,3 | 480 | 4,1 | 831 | 5,5 | 686 |
| Satisfied | 0,8 | 1.405 | 3,3 | 1.297 | 3,8 | 879 | 3,5 | 1.473 | 5,4 | 1.339 |
| Very Satisfied | 0,7 | 666 | 3,9 | 646 | 4,5 | 479 | 3,2 | 716 | 5,9 | 679 |
| Don't know | 0,3 | 198 | 3,5 | 163 | 3,3 | 106 | 3,8 | 178 | 4,0 | 126 |

■ *Under-represented elements*　　■ *Over-represented elements*

*The relationship is not significant. p-value= 0,9 ; Fisher= 0,3.*

*Inter variance= 14,9. Intra variance= 46,9.*

| After the tests were performed, were you offered genetic counselling (e.g. given information about how your genetic condition might affect you and your family)? | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| YES, with a genetic counsellor or clinical geneticist | 0,7 | 1.594 | 3,3 | 1.428 | 4,0 | 1.025 | 3,3 | 1.629 | 5,6 | 1.477 |
| YES, by a healthcare professional | 0,7 | 859 | 4,0 | 813 | 3,9 | 601 | 3,6 | 915 | 5,2 | 817 |
| NO, I wasn't offered genetic counselling | 0,7 | 1.312 | 4,2 | 1.176 | **5,2** | 674 | **4,4** | 1.286 | 5,9 | 1.064 |
| Not sure / Don't remember | 0,4 | 258 | 3,5 | 238 | 2,9 | 151 | 4,0 | 266 | 4,6 | 207 |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is not significant. p-value= 0,9 ; Fisher= 0,2.*

*Inter variance= 9,0. Intra variance= 46,9.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Gender of the person affected by the rare disease / Genetic test(s) looking for genetic changes (also called mutations or variants)**

| GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE | GENETIC TEST(S) LOOKING FOR GENETIC CHANGES (ALSO CALLED MUTATIONS OR VARIANTS) | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW/DON'T REMEMBER | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Female | 3.113 | %47 | 3.004 | %45 | 542 | %8 | 6.659 | %100 |
| Male | 1.801 | %64 | 820 | %29 | 189 | %7 | 2.810 | %100 |
| Other | 62 | %61 | 31 | %31 | 8 | %8 | 101 | %100 |
| TOTAL | 4.976 | %52 | 3.855 | %40 | 739 | %8 | 9.570 | %100 |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 248,0 ; dof= 4.*

**Cross: Gender of the person affected by the rare disease / Other test(s) such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc**

| GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE | OTHER TEST(S) SUCH AS CLINICAL EXAMINATION(S), MEDICAL IMAGING (MRI, SCANS…), BIOPSY, BIOCHEMICAL TEST(S) (BLOOD OR URINE TESTS…), ETC | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW/DON'T REMEMBER | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Female | 6.095 | %92 | 406 | %6 | 158 | %2 | 6.659 | %100 |
| Male | 2.506 | %89 | 229 | %8 | 75 | %3 | 2.810 | %100 |
| Other | 89 | %88 | 6 | %6 | 6 | %6 | 101 | %100 |
| TOTAL | 8.690 | %91 | 641 | %7 | 239 | %2 | 9.570 | %100 |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 19,3 ; dof= 4.*

# Have you ever needed a genetic test but could not access it because...

## Cross: Gender of the person affected by the rare disease / ...you could not afford it?

| GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE | ...YOU COULD NOT AFFORD IT? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Female | 721 | %11 | 4.376 | %66 | 1.562 | %23 | 6.659 | %100 |
| Male | 242 | %9 | 2.053 | %73 | 515 | %18 | 2.810 | %100 |
| Other | 21 | %21 | 54 | %53 | 26 | %26 | 101 | %100 |
| TOTAL | 984 | %10 | 6.483 | %68 | 2.103 | %22 | 9.570 | %100 |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 63,5 ; dof= 4.*

## Cross: Gender of the person affected by the rare disease / ...it was not available in your country?

| GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE | ...IT WAS NOT AVAILABLE IN YOUR COUNTRY? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Female | 726 | %11 | 4.225 | %63 | 1.708 | %26 | 6.659 | %100 |
| Male | 320 | %11 | 1.922 | %68 | 568 | %20 | 2.810 | %100 |
| Other | 13 | %13 | 61 | %60 | 27 | %27 | 101 | %100 |
| TOTAL | 1.059 | %11 | 6.208 | %65 | 2.303 | %24 | 9.570 | %100 |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 33,1 ; dof= 4.*

# Have you ever needed a genetic test but could not access it because...

## Cross: Gender of the person affected by the rare disease / ...healthcare professionals were reluctant or not sufficiently informed?

| GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE | ...HEALTHCARE PROFESSIONALS WERE RELUCTANT OR NOT SUFFICIENTLY INFORMED? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Female | 1.884 | %28 | 3.318 | %50 | 1.457 | %22 | 6.659 | %100 |
| Male | 645 | %23 | 1.657 | %59 | 508 | %18 | 2.810 | %100 |
| Other | 27 | %27 | 50 | %50 | 24 | %24 | 101 | %100 |
| TOTAL | 2.556 | %27 | 5.025 | %53 | 1.989 | %21 | 9.570 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 66,9 ; dof= 4.*

## Cross: Gender of the person affected by the rare disease / To your knowledge, the genetic test(s) that were conducted targeted...

| GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE | TO YOUR KNOWLEDGE, THE GENETIC TEST(S) THAT WERE CONDUCTED TARGETED... | | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | ONLY ONE GENE | | SEVERAL GENES AT THE SAME TIME (GENE PANEL SEQUENCING) | | THE WHOLE DNA (WHOLE GENOME SEQUENCING) | | ALL THE GENES (WHOLE EXOME SEQUENCING) | | A TUMOUR (GENETIC PROFILING OF A TUMOUR) | | OTHER (EPIGENOME, RNA, ETC.) | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Female | 820 | %26 | 991 | %32 | 458 | %15 | 279 | %9 | 86 | %3 | 73 | %2 | 899 | %29 | 3.113 | |
| Male | 504 | %28 | 575 | %32 | 314 | %17 | 227 | %13 | 38 | %2 | 30 | %2 | 434 | %24 | 1.801 | |
| Other | 12 | %19 | 20 | %32 | 7 | %11 | 10 | %16 | 2 | %3 | 0 | %0 | 20 | %32 | 62 | |
| TOTAL | 1.336 | %27 | 1.586 | %32 | 779 | %16 | 516 | %10 | 126 | %3 | 103 | %2 | 1.353 | %27 | 4.976 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 41,3 ; dof= 12.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Gender of the person affected by the rare disease / Did you ever request a private company or laboratory to conduct genetic testing to diagnose the disease?**

| GENERAL GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE | DID YOU EVER REQUEST A PRIVATE COMPANY OR LABORATORY TO CONDUCT GENETIC TESTING TO DIAGNOSE THE DISEASE? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO, NEVER | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Female | 322 | %10 | 141 | %5 | 2.650 | %85 | 3.113 | %100 |
| Male | 191 | %11 | 93 | %5 | 1.517 | %84 | 1.801 | %100 |
| Other | 8 | %13 | 5 | %8 | 49 | %79 | 62 | %100 |
| TOTAL | 521 | %10 | 239 | %5 | 4.216 | %85 | 4.976 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is not significant. p-value= 0,5 ; Chi2= 3,1 ; dof= 4.*

**Cross: Gender of the person affected by the rare disease / In general, how satisfied are you with how the results of the GENETIC TESTS were given to you?**

| GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE | IN GENERAL, HOW SATISFIED ARE YOU WITH HOW THE RESULTS OF THE GENETIC TESTS WERE GIVEN TO YOU? | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | VERY DISSATISFIED | | DISSATISFIED | | NEITHER SATISFIED NOR DISSATISFIED | | SATISFIED | | VERY SATISFIED | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Female | 329 | %11 | 380 | %12 | 656 | %21 | 1.056 | %34 | 509 | %16 | 183 | %6 | 3.113 | %100 |
| Male | 182 | %10 | 189 | %10 | 375 | %21 | 658 | %37 | 333 | %18 | 64 | %4 | 1.801 | %100 |
| Other | 6 | %10 | 7 | %11 | 18 | %29 | 16 | %26 | 8 | %13 | 7 | %11 | 62 | %100 |
| TOTAL | 517 | %10 | 576 | %12 | 1.049 | %21 | 1.730 | %35 | 850 | %17 | 254 | %5 | 4.976 | %100 |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 29,3 ; dof= 10.*

## Cross: Gender of the person affected by the rare disease / After the tests were performed, were you offered genetic counselling (e.g. given information about how your genetic condition might affect you and your family)?

| GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE | AFTER THE TESTS WERE PERFORMED, WERE YOU OFFERED GENETIC COUNSELLING (E.G. GIVEN INFORMATION ABOUT HOW YOUR GENETIC CONDITION MIGHT AFFECT YOU AND YOUR FAMILY)? | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| | YES, WITH A GENETIC COUNSELLOR OR CLINICAL GENETICIST | | YES, BY A HEALTHCARE PROFESSIONAL | | NO, I WASN'T OFFERED GENETIC COUNSELLING | | NOT SURE / DON'T REMEMBER | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % |
| Female | 1.168 | %38 | 623 | %20 | 1.071 | %34 | 251 | %8 | 3.113 | %100 |
| Male | 734 | %41 | 443 | %25 | 515 | %29 | 109 | %6 | 1.801 | %100 |
| Other | 26 | %42 | 8 | %13 | 19 | %31 | 9 | %15 | 62 | %100 |
| TOTAL | 1.928 | %39 | 1.074 | %22 | 1.605 | %32 | 369 | %7 | 4.976 | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 39,0 ; dof= 6.*

## Cross: How old were you when you stopped full-time education? / Genetic test(s) looking for genetic changes (also called mutations or variants)

| HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION? | GENETIC TEST(S) LOOKING FOR GENETIC CHANGES (ALSO CALLED MUTATIONS OR VARIANTS) | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW/DON'T REMEMBER | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 15 y.o. or under | 259 | %57 | 143 | %31 | 53 | %12 | 455 | %100 |
| between 16 and 19 y.o. | 1.217 | %49 | 1.003 | %41 | 244 | %10 | 2.464 | %100 |
| between 20 and 23 y.o. | 1.584 | %52 | 1.212 | %40 | 226 | %7 | 3.022 | %100 |
| 24 y.o. or above | 1.720 | %55 | 1.256 | %40 | 169 | %5 | 3.145 | %100 |
| TOTAL | 4.780 | %53 | 3.614 | %40 | 692 | %8 | 9.086 | %100 |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 65,1 ; dof= 6.*

### Cross: How old were you when you stopped full-time education? / Other test(s) such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc

| HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION? | OTHER TEST(S) SUCH AS CLINICAL EXAMINATION(S), MEDICAL IMAGING (MRI, SCANS…), BIOPSY, BIOCHEMICAL TEST(S) (BLOOD OR URINE TESTS…), ETC | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW/DON'T REMEMBER | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 15 y.o. or under | 406 | %89 | 30 | %7 | 19 | %4 | 455 | %100 |
| between 16 and 19 y.o. | 2.240 | %91 | 155 | %6 | 69 | %3 | 2.464 | %100 |
| between 20 and 23 y.o. | 2.749 | %91 | 199 | %7 | 74 | %2 | 3.022 | %100 |
| 24 y.o. or above | 2.858 | %91 | 224 | %7 | 63 | %2 | 3.145 | %100 |
| TOTAL | 8.253 | %91 | 608 | %7 | 225 | %2 | 9.086 | %100 |

🟥 *Under-represented elements* 🟦 *Over-represented elements*

*The relationship is weakly significant. p-value= 0,1 ; Chi2= 10,9 ; dof= 6.*

### Cross: How old were you when you stopped full-time education? / …you could not afford it?

Have you ever needed a genetic test but could not access it because…

| HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION? | …YOU COULD NOT AFFORD IT? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 15 y.o. or under | 61 | %13 | 297 | %65 | 97 | %21 | 455 | %100 |
| between 16 and 19 y.o. | 283 | %11 | 1.641 | %67 | 540 | %22 | 2.464 | %100 |
| between 20 and 23 y.o. | 295 | %10 | 2.054 | %68 | 673 | %22 | 3.022 | %100 |
| 24 y.o. or above | 287 | %9 | 2.171 | %69 | 687 | %22 | 3.145 | %100 |
| TOTAL | 926 | %10 | 6.163 | %68 | 1.997 | %22 | 9.086 | %100 |

🟥 *Under-represented elements* 🟦 *Over-represented elements*

*The relationship is significant. p-value= 0,0 ; Chi2= 14,6 ; dof= 6.*

## Cross: How old were you when you stopped full-time education? / …it was not available in your country?

Have you ever needed a genetic test but could not access it because…

…IT WAS NOT AVAILABLE IN YOUR COUNTRY?

| HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION? | YES | | NO | | NOT RELEVANT | | TOTAL | |
|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % |
| 15 y.o. or under | 60 | %13 | 287 | %63 | 108 | %24 | 455 | %100 |
| between 16 and 19 y.o. | 260 | %11 | 1.593 | %65 | 611 | %25 | 2.464 | %100 |
| between 20 and 23 y.o. | 334 | %11 | 1.953 | %65 | 735 | %24 | 3.022 | %100 |
| 24 y.o. or above | 355 | %11 | 2.069 | %66 | 721 | %23 | 3.145 | %100 |
| TOTAL | 1.009 | %11 | 5.902 | %65 | 2.175 | %24 | 9.086 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is not significant. p-value= 0,5 ; Chi2= 5,5 ; dof= 6.*

## Cross: How old were you when you stopped full-time education? / …healthcare professionals were reluctant or not sufficiently informed?

Have you ever needed a genetic test but could not access it because…

…HEALTHCARE PROFESSIONALS WERE RELUCTANT OR NOT SUFFICIENTLY INFORMED?

| HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION? | YES | | NO | | NOT RELEVANT | | TOTAL | |
|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % |
| 15 y.o. or under | 130 | %29 | 241 | %53 | 84 | %18 | 455 | %100 |
| between 16 and 19 y.o. | 654 | %27 | 1.287 | %52 | 523 | %21 | 2.464 | %100 |
| between 20 and 23 y.o. | _762_ | _%25_ | 1.610 | %53 | 650 | %22 | 3.022 | %100 |
| 24 y.o. or above | 863 | %27 | 1.648 | %52 | 634 | %20 | 3.145 | %100 |
| TOTAL | 2.409 | %27 | 4.786 | %53 | 1.891 | %21 | 9.086 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is not significant. p-value= 0,3 ; Chi2= 6,8 ; dof= 6.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

## Cross: How old were you when you stopped full-time education? / To your knowledge, the genetic test(s) that were conducted targeted…

| HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION? | TO YOUR KNOWLEDGE, THE GENETIC TEST(S) THAT WERE CONDUCTED TARGETED… | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | ONLY ONE GENE | | SEVERAL GENES AT THE SAME TIME (GENE PANEL SEQUENCING) | | THE WHOLE DNA (WHOLE GENOME SEQUENCING) | | ALL THE GENES (WHOLE EXOME SEQUENCING) | | A TUMOUR (GENETIC PROFILING OF A TUMOUR) | | OTHER (EPIGENOME, RNA, ETC.) | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| 15 y.o. or under | 63 | %24 | 71 | %27 | 40 | %15 | 32 | %12 | 8 | %3 | 6 | %2 | 75 | %29 | 259 | |
| between 16 and 19 y.o. | 303 | %25 | 347 | %29 | 213 | %18 | 101 | %8 | 35 | %3 | 20 | %2 | 378 | %31 | 1.217 | |
| between 20 and 23 y.o. | 410 | %26 | 517 | %33 | 245 | %15 | 162 | %10 | 39 | %2 | 36 | %2 | 431 | %27 | 1.584 | |
| 24 y.o. or above | 508 | %30 | 596 | %35 | 259 | %15 | 208 | %12 | 41 | %2 | 39 | %2 | 398 | %23 | 1.720 | |
| TOTAL | 1.284 | %27 | 1.531 | %32 | 757 | %16 | 503 | %11 | 123 | %3 | 101 | %2 | 1.282 | %27 | 4.780 | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 51,6 ; dof= 18.*

## Cross: How old were you when you stopped full-time education? / Did you ever request a private company or laboratory to conduct genetic testing to diagnose the disease?

| HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION? | DID YOU EVER REQUEST A PRIVATE COMPANY OR LABORATORY TO CONDUCT GENETIC TESTING TO DIAGNOSE THE DISEASE? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO, NEVER | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 15 y.o. or under | 26 | %10 | 18 | %7 | 215 | %83 | 259 | %100 |
| between 16 and 19 y.o. | 137 | %11 | 45 | %4 | 1.035 | %85 | 1.217 | %100 |
| between 20 and 23 y.o. | 160 | %10 | 77 | %5 | 1.347 | %85 | 1.584 | %100 |
| 24 y.o. or above | 176 | %10 | 89 | %5 | 1.455 | %85 | 1.720 | %100 |
| TOTAL | 499 | %10 | 229 | %5 | 4.052 | %85 | 4.780 | %100 |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is not significant. p-value= 0,3 ; Chi2= 7,3 ; dof= 6.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: How old were you when you stopped full-time education? / In general, how satisfied are you with how the results of the GENETIC TESTS were given to you?**

| HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION? | IN GENERAL, HOW SATISFIED ARE YOU WITH HOW THE RESULTS OF THE GENETIC TESTS WERE GIVEN TO YOU? | | | | | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | VERY DISSATISFIED | | DISSATISFIED | | NEITHER SATISFIED NOR DISSATISFIED | | SATISFIED | | VERY SATISFIED | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| 15 y.o. or under | 41 | %16 | 29 | %11 | 54 | %21 | 89 | %34 | 36 | %14 | 10 | %4 | 259 | %100 |
| between 16 and 19 y.o. | 143 | %12 | 124 | %10 | 259 | %21 | 412 | %34 | 208 | %17 | 71 | %6 | 1.217 | %100 |
| between 20 and 23 y.o. | 151 | %10 | 185 | %12 | 333 | %21 | 576 | %36 | 261 | %16 | 78 | %5 | 1.584 | %100 |
| 24 y.o. or above | 167 | %10 | 221 | %13 | 357 | %21 | 579 | %34 | 312 | %18 | 84 | %5 | 1.720 | %100 |
| TOTAL | 502 | %11 | 559 | %12 | 1.003 | %21 | 1.656 | %35 | 817 | %17 | 243 | %5 | 4.780 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is weakly significant. p-value= 0,1 ; Chi2= 23,1 ; dof= 15.*

**Cross: How old were you when you stopped full-time education? / After the tests were performed, were you offered genetic counselling (e.g. given information about how your genetic condition might affect you and your family)?**

| HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION? | AFTER THE TESTS WERE PERFORMED, WERE YOU OFFERED GENETIC COUNSELLING (E.G. GIVEN INFORMATION ABOUT HOW YOUR GENETIC CONDITION MIGHT AFFECT YOU AND YOUR FAMILY)? | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | YES, WITH A GENETIC COUNSELLOR OR CLINICAL GENETICIST | | YES, BY A HEALTHCARE PROFESSIONAL | | NO, I WASN'T OFFERED GENETIC COUNSELLING | | NOT SURE / DON'T REMEMBER | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % |
| 15 y.o. or under | 87 | %34 | 71 | %27 | 78 | %30 | 23 | %9 | 259 | %100 |
| between 16 and 19 y.o. | 459 | %38 | 246 | %20 | 413 | %34 | 99 | %8 | 1.217 | %100 |
| between 20 and 23 y.o. | 606 | %38 | 368 | %23 | 496 | %31 | 114 | %7 | 1.584 | %100 |
| 24 y.o. or above | 711 | %41 | 348 | %20 | 550 | %32 | 111 | %6 | 1.720 | %100 |
| TOTAL | 1.863 | %39 | 1.033 | %22 | 1.537 | %32 | 347 | %7 | 4.780 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is significant. p-value= 0,0 ; Chi2= 19,3 ; dof= 9.*

## Cross: How would you best describe yourself? / Genetic test(s) looking for genetic changes (also called mutations or variants)

| HOW WOULD YOU BEST DESCRIBE YOURSELF? | GENETIC TEST(S) LOOKING FOR GENETIC CHANGES (ALSO CALLED MUTATIONS OR VARIANTS) | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW/DON'T REMEMBER | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| I belong to the ethnic majority in the country where I live | 3.701 | %52 | 2.899 | %41 | 525 | %7 | 7.125 | %100 |
| I am part of an ethnic minority in the country where I live | 269 | %58 | 147 | %32 | 49 | %11 | 465 | %100 |
| Other, specify... | 169 | %50 | 131 | %39 | 37 | %11 | 337 | %100 |
| TOTAL | 4.139 | %52 | 3.177 | %40 | 611 | %8 | 7.927 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 22,9 ; dof= 4.*

## Cross: How would you best describe yourself? / Other test(s) such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc

| HOW WOULD YOU BEST DESCRIBE YOURSELF? | OTHER TEST(S) SUCH AS CLINICAL EXAMINATION(S), MEDICAL IMAGING (MRI, SCANS…), BIOPSY, BIOCHEMICAL TEST(S) (BLOOD OR URINE TESTS…), ETC | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW/DON'T REMEMBER | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| I belong to the ethnic majority in the country where I live | 6.483 | %91 | 472 | %7 | 170 | %2 | 7.125 | %100 |
| I am part of an ethnic minority in the country where I live | 417 | %90 | 35 | %8 | 13 | %3 | 465 | %100 |
| Other, specify... | 299 | %89 | 23 | %7 | 15 | %4 | 337 | %100 |
| TOTAL | 7.199 | %91 | 530 | %7 | 198 | %2 | 7.927 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is not significant. p-value= 0,2 ; Chi2= 6,5 ; dof= 4.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: How would you best describe yourself? / ...you could not afford it?**

Have you ever needed a genetic test but could not access it because...    ...YOU COULD NOT AFFORD IT?

| HOW WOULD YOU BEST DESCRIBE YOURSELF? | YES | | NO | | NOT RELEVANT | | TOTAL | |
|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % |
| I belong to the ethnic majority in the country where I live | 736 | %10 | 4.880 | %68 | 1.509 | %21 | 7.125 | %100 |
| I am part of an ethnic minority in the country where I live | 76 | %16 | 309 | %66 | 80 | %17 | 465 | %100 |
| Other, specify... | 45 | %13 | 202 | %60 | 90 | %27 | 337 | %100 |
| TOTAL | 857 | %11 | 5.391 | %68 | 1.679 | %21 | 7.927 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 28,7 ; dof= 4.*

**Cross: How would you best describe yourself? / ...it was not available in your country?**

Have you ever needed a genetic test but could not access it because...    ...IT WAS NOT AVAILABLE IN YOUR COUNTRY?

| HOW WOULD YOU BEST DESCRIBE YOURSELF? | YES | | NO | | NOT RELEVANT | | TOTAL | |
|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % |
| I belong to the ethnic majority in the country where I live | 809 | %11 | 4.691 | %66 | 1.625 | %23 | 7.125 | %100 |
| I am part of an ethnic minority in the country where I live | 67 | %14 | 300 | %65 | 98 | %21 | 465 | %100 |
| Other, specify... | 48 | %14 | 191 | %57 | 98 | %29 | 337 | %100 |
| TOTAL | 924 | %12 | 5.182 | %65 | 1.821 | %23 | 7.927 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 16,0 ; dof= 4.*

## Cross: How would you best describe yourself? / …healthcare professionals were reluctant or not sufficiently informed?

Have you ever needed a genetic test but could not access it because...

…HEALTHCARE PROFESSIONALS WERE RELUCTANT OR NOT SUFFICIENTLY INFORMED?

| HOW WOULD YOU BEST DESCRIBE YOURSELF? | YES | | NO | | NOT RELEVANT | | TOTAL | |
|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % |
| I belong to the ethnic majority in the country where I live | 1.913 | %27 | 3.778 | %53 | 1.434 | %20 | 7.125 | %100 |
| I am part of an ethnic minority in the country where I live | 150 | %32 | 236 | %51 | 79 | %17 | 465 | %100 |
| Other, specify... | 101 | %30 | 156 | %46 | 80 | %24 | 337 | %100 |
| TOTAL | 2.164 | %27 | 4.170 | %53 | 1.593 | %20 | 7.927 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is significant. p-value= 0,0 ; Chi2= 13,2 ; dof= 4.*

## Cross: How would you best describe yourself? / To your knowledge, the genetic test(s) that were conducted targeted…

TO YOUR KNOWLEDGE, THE GENETIC TEST(S) THAT WERE CONDUCTED TARGETED…

| HOW WOULD YOU BEST DESCRIBE YOURSELF? | ONLY ONE GENE | | SEVERAL GENES AT THE SAME TIME (GENE PANEL SEQUENCING) | | THE WHOLE DNA (WHOLE GENOME SEQUENCING) | | ALL THE GENES (WHOLE EXOME SEQUENCING) | | A TUMOUR (GENETIC PROFILING OF A TUMOUR) | | OTHER (EPIGENOME, RNA, ETC.) | | DON'T KNOW | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| I belong to the ethnic majority in the country where I live | 975 | %26 | 1.225 | %33 | 584 | %16 | 408 | %11 | 94 | %3 | 83 | %2 | 965 | %26 | 3.701 | |
| I am part of an ethnic minority in the country where I live | 64 | %24 | 86 | %32 | 53 | %20 | 29 | %11 | 7 | %3 | 6 | %2 | 83 | %31 | 269 | |
| Other, specify... | 54 | %32 | 35 | %21 | 32 | %19 | 15 | %9 | 7 | %4 | 4 | %2 | 59 | %35 | 169 | |
| TOTAL | 1.093 | %26 | 1.346 | %33 | 669 | %16 | 452 | %11 | 108 | %3 | 93 | %2 | 1.107 | %27 | 4.139 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is significant. p-value= 0,0 ; Chi2= 22,4 ; dof= 12.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: How would you best describe yourself? / Did you ever request a private company or laboratory to conduct genetic testing to diagnose the disease?**

| | DID YOU EVER REQUEST A PRIVATE COMPANY OR LABORATORY TO CONDUCT GENETIC TESTING TO DIAGNOSE THE DISEASE? | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO, NEVER | | TOTAL | |
| **HOW WOULD YOU BEST DESCRIBE YOURSELF?** | N | % | N | % | N | % | N | % |
| I belong to the ethnic majority in the country where I live | <u>401</u> | <u>%11</u> | 185 | %5 | <u>3.115</u> | <u>%84</u> | 3.701 | %100 |
| I am part of an ethnic minority in the country where I live | <u>43</u> | <u>%16</u> | 14 | %5 | 212 | %79 | 269 | %100 |
| Other, specify... | 20 | %12 | 12 | %7 | 137 | %81 | 169 | %100 |
| **TOTAL** | **464** | **%11** | **211** | **%5** | **3.464** | **%84** | **4.139** | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is weakly significant. p-value= 0,1 ; Chi2= 8,4 ; dof= 4.*

**Cross: How would you best describe yourself? / In general, how satisfied are you with how the results of the GENETIC TESTS were given to you?**

| | IN GENERAL, HOW SATISFIED ARE YOU WITH HOW THE RESULTS OF THE GENETIC TESTS WERE GIVEN TO YOU? | | | | | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | VERY DISSATISFIED | | DISSATISFIED | | NEITHER SATISFIED NOR DISSATISFIED | | SATISFIED | | VERY SATISFIED | | DON'T KNOW | | TOTAL | |
| **HOW WOULD YOU BEST DESCRIBE YOURSELF?** | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| I belong to the ethnic majority in the country where I live | <u>374</u> | <u>%10</u> | 431 | %12 | 776 | %21 | 1.295 | %35 | <u>657</u> | <u>%18</u> | <u>168</u> | <u>%5</u> | 3.701 | %100 |
| I am part of an ethnic minority in the country where I live | 33 | %12 | 29 | %11 | 65 | %24 | 92 | %34 | <u>32</u> | <u>%12</u> | 18 | %7 | 269 | %100 |
| Other, specify... | 25 | %15 | 22 | %13 | 37 | %22 | 51 | %30 | 22 | %13 | 12 | %7 | 169 | %100 |
| **TOTAL** | **432** | **%10** | **482** | **%12** | **878** | **%21** | **1.438** | **%35** | **711** | **%17** | **198** | **%5** | **4.139** | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is weakly significant. p-value= 0,1 ; Chi2= 18,3 ; dof= 10.*

## Cross: How would you best describe yourself? / After the tests were performed, were you offered genetic counselling (e.g. given information about how your genetic condition might affect you and your family)?

| | AFTER THE TESTS WERE PERFORMED, WERE YOU OFFERED GENETIC COUNSELLING (E.G. GIVEN INFORMATION ABOUT HOW YOUR GENETIC CONDITION MIGHT AFFECT YOU AND YOUR FAMILY)? | | | | | | | | | |
| | YES, WITH A GENETIC COUNSELLOR OR CLINICAL GENETICIST | | YES, BY A HEALTHCARE PROFESSIONAL | | NO, I WASN'T OFFERED GENETIC COUNSELLING | | NOT SURE / DON'T REMEMBER | | TOTAL | |
| HOW WOULD YOU BEST DESCRIBE YOURSELF? | N | % | N | % | N | % | N | % | N | % |
|---|---|---|---|---|---|---|---|---|---|---|
| I belong to the ethnic majority in the country where I live | 1.445 | %39 | 823 | %22 | 1.184 | %32 | 249 | %7 | 3.701 | %100 |
| I am part of an ethnic minority in the country where I live | 88 | %33 | 53 | %20 | 102 | %38 | 26 | %10 | 269 | %100 |
| Other, specify... | 58 | %34 | 20 | %12 | 71 | %42 | 20 | %12 | 169 | %100 |
| TOTAL | 1.591 | %38 | 896 | %22 | 1.357 | %33 | 295 | %7 | 4.139 | |

■ *Under-represented elements* ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 27,7 ; dof= 6.*

## Cross: Typology of countries based on size and welfare / Genetic test(s) looking for genetic changes (also called mutations or variants)

| | GENETIC TEST(S) LOOKING FOR GENETIC CHANGES (ALSO CALLED MUTATIONS OR VARIANTS) | | | | | | | |
| | YES | | NO | | DON'T KNOW/DON'T REMEMBER | | TOTAL | |
| TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE | N | % | N | % | N | % | N | % |
|---|---|---|---|---|---|---|---|---|
| Group A ('Eastern Europe') | 1.158 | %65 | 485 | %27 | 151 | %8 | 1.794 | %100 |
| Group B ('Western Europe') | 2.702 | %53 | 2.090 | %41 | 313 | %6 | 5.105 | %100 |
| Group C ('Northern Europe') | 1.470 | %45 | 1.476 | %45 | 327 | %10 | 3.273 | %100 |
| TOTAL | 5.330 | %52 | 4.051 | %40 | 791 | %8 | 10.172 | |

■ *Under-represented elements* ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 223,3 ; dof= 4.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Typology of countries based on size and welfare / Other test(s) such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc

| TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE | OTHER TEST(S) SUCH AS CLINICAL EXAMINATION(S), MEDICAL IMAGING (MRI, SCANS…), BIOPSY, BIOCHEMICAL TEST(S) (BLOOD OR URINE TESTS…), ETC | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW/DON'T REMEMBER | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Group A ('Eastern Europe') | 1.632 | %91 | 108 | %6 | 54 | %3 | 1.794 | %100 |
| Group B ('Western Europe') | 4.614 | %90 | 365 | %7 | 126 | %2 | 5.105 | %100 |
| Group C ('Northern Europe') | 2.951 | %90 | 239 | %7 | 83 | %3 | 3.273 | %100 |
| TOTAL | 9.197 | %90 | 712 | %7 | 263 | %3 | 10.172 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is not significant. p-value= 0,3 ; Chi2= 4,7 ; dof= 4.*

## Cross: Typology of countries based on size and welfare / …you could not afford it?

Have you ever needed a genetic test but could not access it because…

| TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE | …YOU COULD NOT AFFORD IT? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Group A ('Eastern Europe') | 389 | %22 | 1.174 | %65 | 231 | %13 | 1.794 | %100 |
| Group B ('Western Europe') | 473 | %9 | 3.507 | %69 | 1.125 | %22 | 5.105 | %100 |
| Group C ('Northern Europe') | 216 | %7 | 2.220 | %68 | 837 | %26 | 3.273 | %100 |
| TOTAL | 1.078 | %11 | 6.901 | %68 | 2.193 | %22 | 10.172 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 355,8 ; dof= 4.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Typology of countries based on size and welfare / …it was not available in your country?

Have you ever needed a genetic test but could not access it because…

…IT WAS NOT AVAILABLE IN YOUR COUNTRY?

| TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE | YES | | NO | | NOT RELEVANT | | TOTAL | |
|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % |
| Group A ('Eastern Europe') | 492 | %27 | 1.056 | %59 | 246 | %14 | 1.794 | %100 |
| Group B ('Western Europe') | 400 | %8 | 3.493 | %68 | 1.212 | %24 | 5.105 | %100 |
| Group C ('Northern Europe') | 277 | %8 | 2.060 | %63 | 936 | %29 | 3.273 | %100 |
| TOTAL | 1.169 | %11 | 6.609 | %65 | 2.394 | %24 | 10.172 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 612,8 ; dof= 4.*

## Cross: Typology of countries based on size and welfare / …healthcare professionals were reluctant or not sufficiently informed?

Have you ever needed a genetic test but could not access it because…

…HEALTHCARE PROFESSIONALS WERE RELUCTANT OR NOT SUFFICIENTLY INFORMED?

| TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE | YES | | NO | | NOT RELEVANT | | TOTAL | |
|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % |
| Group A ('Eastern Europe') | 614 | %34 | 948 | %53 | 232 | %13 | 1.794 | %100 |
| Group B ('Western Europe') | 1.273 | %25 | 2.779 | %54 | 1.053 | %21 | 5.105 | %100 |
| Group C ('Northern Europe') | 834 | %25 | 1.652 | %50 | 787 | %24 | 3.273 | %100 |
| TOTAL | 2.721 | %27 | 5.379 | %53 | 2.072 | %20 | 10.172 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 122,2 ; dof= 4.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Typology of countries based on size and welfare / To your knowledge, the genetic test(s) that were conducted targeted…

| TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE | TO YOUR KNOWLEDGE, THE GENETIC TEST(S) THAT WERE CONDUCTED TARGETED… | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | ONLY ONE GENE | | SEVERAL GENES AT THE SAME TIME (GENE PANEL SEQUENCING) | | THE WHOLE DNA (WHOLE GENOME SEQUENCING) | | ALL THE GENES (WHOLE EXOME SEQUENCING) | | A TUMOUR (GENETIC PROFILING OF A TUMOUR) | | OTHER (EPIGENOME, RNA, ETC.) | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Group A ('Eastern Europe') | 284 | %25 | 368 | %32 | 193 | %17 | 148 | %13 | 38 | %3 | 34 | %3 | 319 | %28 | 1.158 | |
| Group B ('Western Europe') | 761 | %28 | 904 | %33 | 387 | %14 | 258 | %10 | 49 | %2 | 49 | %2 | 695 | %26 | 2.702 | |
| Group C ('Northern Europe') | 370 | %25 | 416 | %28 | 282 | %19 | 149 | %10 | 46 | %3 | 31 | %2 | 442 | %30 | 1.470 | |
| **TOTAL** | **1.415** | **%27** | **1.688** | **%32** | **862** | **%16** | **555** | **%10** | **133** | **%2** | **114** | **%2** | **1.456** | **%27** | **5.330** | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 57,6 ; dof= 12.*

## Cross: Typology of countries based on size and welfare / Did you ever request a private company or laboratory to conduct genetic testing to diagnose the disease?

| TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE | DID YOU EVER REQUEST A PRIVATE COMPANY OR LABORATORY TO CONDUCT GENETIC TESTING TO DIAGNOSE THE DISEASE? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO, NEVER | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Group A ('Eastern Europe') | 261 | %23 | 139 | %12 | 758 | %65 | 1.158 | %100 |
| Group B ('Western Europe') | 235 | %9 | 89 | %3 | 2.378 | %88 | 2.702 | %100 |
| Group C ('Northern Europe') | 79 | %5 | 33 | %2 | 1.358 | %92 | 1.470 | %100 |
| **TOTAL** | **575** | **%11** | **261** | **%5** | **4.494** | **%84** | **5.330** | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 418,4 ; dof= 4.*

## Cross: Typology of countries based on size and welfare / In general, how satisfied are you with how the results of the GENETIC TESTS were given to you?

| TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE | IN GENERAL, HOW SATISFIED ARE YOU WITH HOW THE RESULTS OF THE GENETIC TESTS WERE GIVEN TO YOU? | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | VERY DISSATISFIED | | DISSATISFIED | | NEITHER SATISFIED NOR DISSATISFIED | | SATISFIED | | VERY SATISFIED | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Group A ('Eastern Europe') | 109 | %9 | 124 | %11 | 279 | %24 | 432 | %37 | 161 | %14 | 53 | %5 | 1.158 | %100 |
| Group B ('Western Europe') | 330 | %12 | 332 | %12 | 577 | %21 | 917 | %34 | 416 | %15 | 130 | %5 | 2.702 | %100 |
| Group C ('Northern Europe') | 118 | %8 | 149 | %10 | 270 | %18 | 526 | %36 | 320 | %22 | 87 | %6 | 1.470 | %100 |
| TOTAL | 557 | %10 | 605 | %11 | 1.126 | %21 | 1.875 | %35 | 897 | %17 | 270 | %5 | 5.330 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 68,4 ; dof= 10.*

## Cross: Typology of countries based on size and welfare / After the tests were performed, were you offered genetic counselling (e.g. given information about how your genetic condition might affect you and your family)?

| TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE | AFTER THE TESTS WERE PERFORMED, WERE YOU OFFERED GENETIC COUNSELLING (E.G. GIVEN INFORMATION ABOUT HOW YOUR GENETIC CONDITION MIGHT AFFECT YOU AND YOUR FAMILY)? | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| | YES, WITH A GENETIC COUNSELLOR OR CLINICAL GENETICIST | | YES, BY A HEALTHCARE PROFESSIONAL | | NO, I WASN'T OFFERED GENETIC COUNSELLING | | NOT SURE / DON'T REMEMBER | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % |
| Group A ('Eastern Europe') | 410 | %35 | 229 | %20 | 447 | %39 | 72 | %6 | 1.158 | %100 |
| Group B ('Western Europe') | 1.049 | %39 | 629 | %23 | 817 | %30 | 207 | %8 | 2.702 | %100 |
| Group C ('Northern Europe') | 617 | %42 | 283 | %19 | 458 | %31 | 112 | %8 | 1.470 | %100 |
| TOTAL | 2.076 | %39 | 1.141 | %21 | 1.722 | %32 | 391 | %7 | 5.330 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 37,2 ; dof= 6.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

## Cross: Would you say that you, or the person you care for, live in a: / Genetic test(s) looking for genetic changes (also called mutations or variants)

| WOULD YOU SAY THAT YOU, OR THE PERSON YOU CARE FOR, LIVE IN A: | GENETIC TEST(S) LOOKING FOR GENETIC CHANGES (ALSO CALLED MUTATIONS OR VARIANTS) | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW/DON'T REMEMBER | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Rural area or village | 1.245 | %52 | 968 | %40 | 203 | %8 | 2.416 | %100 |
| Small or mid size town | 2.065 | %53 | 1.546 | %40 | 290 | %7 | 3.901 | %100 |
| Large town | 1.468 | %53 | 1.094 | %40 | 198 | %7 | 2.760 | %100 |
| TOTAL | 4.778 | %53 | 3.608 | %40 | 691 | %8 | 9.077 | %100 |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is not significant. p-value= 0,4 ; Chi2= 3,7 ; dof= 4.*

## Cross: Would you say that you, or the person you care for, live in a: / Other test(s) such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc

| WOULD YOU SAY THAT YOU, OR THE PERSON YOU CARE FOR, LIVE IN A: | OTHER TEST(S) SUCH AS CLINICAL EXAMINATION(S), MEDICAL IMAGING (MRI, SCANS…), BIOPSY, BIOCHEMICAL TEST(S) (BLOOD OR URINE TESTS…), ETC | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW/DON'T REMEMBER | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Rural area or village | 2.212 | %92 | 147 | %6 | 57 | %2 | 2.416 | %100 |
| Small or mid size town | 3.520 | %90 | 273 | %7 | 108 | %3 | 3.901 | %100 |
| Large town | 2.515 | %91 | 185 | %7 | 60 | %2 | 2.760 | %100 |
| TOTAL | 8.247 | %91 | 605 | %7 | 225 | %2 | 9.077 | %100 |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is not significant. p-value= 0,3 ; Chi2= 4,7 ; dof= 4.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

# Have you ever needed a genetic test but could not access it because...

## Cross: Would you say that you, or the person you care for, live in a: / ...you could not afford it?

| WOULD YOU SAY THAT YOU, OR THE PERSON YOU CARE FOR, LIVE IN A: | ...YOU COULD NOT AFFORD IT? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Rural area or village | **201** | **%8** | 1.646 | %68 | **569** | **%24** | 2.416 | %100 |
| Small or mid size town | 400 | %10 | 2.665 | %68 | 836 | %21 | 3.901 | %100 |
| Large town | 323 | %12 | 1.847 | %67 | 590 | %21 | 2.760 | %100 |
| TOTAL | 924 | %10 | 6.158 | %68 | 1.995 | %22 | 9.077 | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

The relationship is very significant. p-value= < 0,01 ; Chi2= 18,7 ; dof= 4.

## Cross: Would you say that you, or the person you care for, live in a: / ...it was not available in your country?

| WOULD YOU SAY THAT YOU, OR THE PERSON YOU CARE FOR, LIVE IN A: | ...IT WAS NOT AVAILABLE IN YOUR COUNTRY? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Rural area or village | **230** | **%10** | 1.578 | %65 | 608 | %25 | 2.416 | %100 |
| Small or mid size town | **401** | **%10** | **2.584** | **%66** | 916 | %23 | 3.901 | %100 |
| Large town | **376** | **%14** | **1.736** | **%63** | 648 | %23 | 2.760 | %100 |
| TOTAL | 1.007 | %11 | 5.898 | %65 | 2.172 | %24 | 9.077 | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

The relationship is very significant. p-value= < 0,01 ; Chi2= 28,6 ; dof= 4.

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

Have you ever needed a genetic test but could not access it because...

**Cross: Would you say that you, or the person you care for, live in a: / ...healthcare professionals were reluctant or not sufficiently informed?**

| WOULD YOU SAY THAT YOU, OR THE PERSON YOU CARE FOR, LIVE IN A: | ...HEALTHCARE PROFESSIONALS WERE RELUCTANT OR NOT SUFFICIENTLY INFORMED? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Rural area or village | **589** | **%24** | 1.301 | %54 | 526 | %22 | **2.416** | **%100** |
| Small or mid size town | 1.052 | %27 | 2.053 | %53 | 796 | %20 | **3.901** | **%100** |
| Large town | 765 | %28 | 1.428 | %52 | 567 | %21 | **2.760** | **%100** |
| **TOTAL** | **2.406** | **%27** | **4.782** | **%53** | **1.889** | **%21** | **9.077** | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is weakly significant. p-value= 0,1 ; Chi2= 8,5 ; dof= 4.*

**Cross: Would you say that you, or the person you care for, live in a: / To your knowledge, the genetic test(s) that were conducted targeted...**

| WOULD YOU SAY THAT YOU, OR THE PERSON YOU CARE FOR, LIVE IN A: | TO YOUR KNOWLEDGE, THE GENETIC TEST(S) THAT WERE CONDUCTED TARGETED... | | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | ONLY ONE GENE | | SEVERAL GENES AT THE SAME TIME (GENE PANEL SEQUENCING) | | THE WHOLE DNA (WHOLE GENOME SEQUENCING) | | ALL THE GENES (WHOLE EXOME SEQUENCING) | | A TUMOUR (GENETIC PROFILING OF A TUMOUR) | | OTHER (EPIGENOME, RNA, ETC.) | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Rural area or village | 329 | %26 | **366** | **%29** | 200 | %16 | 133 | %11 | 34 | %3 | 24 | %2 | **368** | **%30** | 1.245 | |
| Small or mid size town | 565 | %27 | 678 | %33 | 341 | %17 | **193** | **%9** | 47 | %2 | 40 | %2 | 538 | %26 | 2.065 | |
| Large town | 390 | %27 | 485 | %33 | 216 | %15 | **177** | **%12** | 42 | %3 | 37 | %3 | 376 | %26 | 1.468 | |
| **TOTAL** | **1.284** | **%27** | **1.529** | **%32** | **757** | **%16** | **503** | **%11** | **123** | **%3** | **101** | **%2** | **1.282** | **%27** | **4.778** | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is weakly significant. p-value= 0,1 ; Chi2= 19,4 ; dof= 12.*

## Cross: Would you say that you, or the person you care for, live in a: / Did you ever request a private company or laboratory to conduct genetic testing to diagnose the disease?

| WOULD YOU SAY THAT YOU, OR THE PERSON YOU CARE FOR, LIVE IN A: | DID YOU EVER REQUEST A PRIVATE COMPANY OR LABORATORY TO CONDUCT GENETIC TESTING TO DIAGNOSE THE DISEASE? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO, NEVER | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Rural area or village | 108 | %9 | 38 | %3 | 1.099 | %88 | 1.245 | %100 |
| Small or mid size town | 195 | %9 | 83 | %4 | 1.787 | %87 | 2.065 | %100 |
| Large town | 196 | %13 | 108 | %7 | 1.164 | %79 | 1.468 | %100 |
| TOTAL | 499 | %10 | 229 | %5 | 4.050 | %85 | 4.778 | %100 |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 55,9 ; dof= 4.*

## Cross: Would you say that you, or the person you care for, live in a: / In general, how satisfied are you with how the results of the GENETIC TESTS were given to you?

| WOULD YOU SAY THAT YOU, OR THE PERSON YOU CARE FOR, LIVE IN A: | IN GENERAL, HOW SATISFIED ARE YOU WITH HOW THE RESULTS OF THE GENETIC TESTS WERE GIVEN TO YOU? | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | VERY DISSATISFIED | | DISSATISFIED | | NEITHER SATISFIED NOR DISSATISFIED | | SATISFIED | | VERY SATISFIED | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Rural area or village | 126 | %10 | 137 | %11 | 269 | %22 | 440 | %35 | 211 | %17 | 62 | %5 | 1.245 | %100 |
| Small or mid size town | 230 | %11 | 239 | %12 | 406 | %20 | 747 | %36 | 329 | %16 | 114 | %6 | 2.065 | %100 |
| Large town | 146 | %10 | 183 | %12 | 328 | %22 | 469 | %32 | 275 | %19 | 67 | %5 | 1.468 | %100 |
| TOTAL | 502 | %11 | 559 | %12 | 1.003 | %21 | 1.656 | %35 | 815 | %17 | 243 | %5 | 4.778 | %100 |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is weakly significant. p-value= 0,1 ; Chi2= 16,1 ; dof= 10.*

**Cross: Would you say that you, or the person you care for, live in a: / After the tests were performed, were you offered genetic counselling (e.g. given information about how your genetic condition might affect you and your family)?**

| WOULD YOU SAY THAT YOU, OR THE PERSON YOU CARE FOR, LIVE IN A: | AFTER THE TESTS WERE PERFORMED, WERE YOU OFFERED GENETIC COUNSELLING (E.G. GIVEN INFORMATION ABOUT HOW YOUR GENETIC CONDITION MIGHT AFFECT YOU AND YOUR FAMILY)? | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|
| | YES, WITH A GENETIC COUNSELLOR OR CLINICAL GENETICIST | | YES, BY A HEALTHCARE PROFESSIONAL | | NO, I WASN'T OFFERED GENETIC COUNSELLING | | NOT SURE / DON'T REMEMBER | | | |
| | N | % | N | % | N | % | N | % | N | % |
| Rural area or village | 466 | %37 | 280 | %22 | 401 | %32 | 98 | %8 | 1.245 | %100 |
| Small or mid size town | **839** | **%41** | 458 | %22 | **619** | **%30** | 149 | %7 | 2.065 | %100 |
| Large town | 557 | %38 | 295 | %20 | **516** | **%35** | 100 | %7 | 1.468 | %100 |
| **TOTAL** | **1.862** | **%39** | **1.033** | **%22** | **1.536** | **%32** | **347** | **%7** | **4.778** | **%100** |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is significant. p-value= 0,0 ; Chi2= 13,1 ; dof= 6.*

**Cross: Orphacode associated nomenclature (english) / To your knowledge, the genetic test(s) that were conducted targeted…**

| ORPHACODE ASSOCIATED NOMENCLATURE (ENGLISH) | ONLY ONE GENE | | SEVERAL GENES AT THE SAME TIME (GENE PANEL SEQUENCING) | | THE WHOLE DNA (WHOLE GENOME SEQUENCING) | | ALL THE GENES (WHOLE EXOME SEQUENCING) | | A TUMOUR (GENETIC PROFILING OF A TUMOUR) | | OTHER (EPIGENOME, RNA, ETC.) | | DON'T KNOW | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Hereditary hemorrhagic telangiectasia | 115 | %31 | 131 | %36 | 45 | %12 | 7 | %2 | 2 | %1 | 1 | %0 | 92 | %25 | 369 | |
| Hypermobile Ehlers-Danlos syndrome | 15 | %16 | 51 | %54 | 8 | %8 | 9 | %9 | 0 | %0 | 3 | %3 | 22 | %23 | 95 | |
| Sarcoidosis | 2 | %20 | 2 | %20 | 0 | %0 | 1 | %10 | 0 | %0 | 0 | %0 | 5 | %50 | 10 | |
| Classical Ehlers-Danlos syndrome | 6 | %9 | 36 | %56 | 7 | %11 | 3 | %5 | 1 | %2 | 2 | %3 | 16 | %25 | 64 | |
| Williams syndrome | 49 | %37 | 39 | %30 | 17 | %13 | 5 | %4 | 1 | %1 | 0 | %0 | 30 | %23 | 131 | |
| Cystic fibrosis | 47 | %39 | 42 | %35 | 13 | %11 | 6 | %5 | 0 | %0 | 0 | %0 | 22 | %18 | 121 | |
| Myasthenia gravis | 1 | %6 | 8 | %47 | 2 | %12 | 0 | %0 | 2 | %12 | 3 | %18 | 5 | %29 | 17 | |
| Systemic sclerosis | 0 | %0 | 0 | %0 | 1 | %25 | 0 | %0 | 0 | %0 | 0 | %0 | 3 | %75 | 4 | |
| Tuberous sclerosis complex | 17 | %19 | 33 | %37 | 12 | %13 | 3 | %3 | 1 | %1 | 4 | %4 | 26 | %29 | 89 | |
| Neurofibromatosis type 1 | 20 | %32 | 14 | %22 | 12 | %19 | 1 | %2 | 6 | %10 | 2 | %3 | 20 | %32 | 63 | |
| Interstitial cystitis | 0 | %0 | 0 | %0 | 1 | %25 | 0 | %0 | 1 | %25 | 1 | %25 | 2 | %50 | 4 | |
| Addison disease | 2 | %25 | 2 | %25 | 0 | %0 | 0 | %0 | 0 | %0 | 0 | %0 | 4 | %50 | 8 | |
| 22q11.2 deletion syndrome | 20 | %32 | 15 | %24 | 10 | %16 | 6 | %10 | 0 | %0 | 1 | %2 | 16 | %25 | 63 | |
| Chronic inflammatory demyelinating polyneuropathy | 4 | %25 | 3 | %19 | 1 | %6 | 0 | %0 | 0 | %0 | 1 | %6 | 8 | %50 | 16 | |
| Perineural cyst | 1 | %17 | 4 | %67 | 2 | %33 | 0 | %0 | 1 | %17 | 0 | %0 | 2 | %33 | 6 | |
| Acute inflammatory demyelinating polyradiculoneuropathy | 0 | %0 | 0 | %0 | 0 | %0 | 0 | %0 | 0 | %0 | 0 | %0 | 3 | %100 | 3 | |
| Rett syndrome | 27 | %48 | 19 | %34 | 10 | %18 | 10 | %18 | 0 | %0 | 0 | %0 | 11 | %20 | 56 | |
| Marfan syndrome | 13 | %31 | 13 | %31 | 3 | %7 | 1 | %2 | 0 | %0 | 0 | %0 | 14 | %33 | 42 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 9.611,8 ; dof= 7.314.*

**Cross: Please select the sentence that best describes your situation or the situation of the person you care for: / To your knowledge, the genetic test(s) that were conducted targeted…**

TO YOUR KNOWLEDGE, THE GENETIC TEST(S) THAT WERE CONDUCTED TARGETED…

| PLEASE SELECT THE SENTENCE THAT BEST DESCRIBES YOUR SITUATION OR THE SITUATION OF THE PERSON YOU CARE FOR: | ONLY ONE GENE | | SEVERAL GENES AT THE SAME TIME (GENE PANEL SEQUENCING) | | THE WHOLE DNA (WHOLE GENOME SEQUENCING) | | ALL THE GENES (WHOLE EXOME SEQUENCING) | | A TUMOUR (GENETIC PROFILING OF A TUMOUR) | | OTHER (EPIGENOME, RNA, ETC.) | | DON'T KNOW | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| I know the NAME of the rare disease, syndrome or malformation and it has been CONFIRMED by appropriate genetic, clinical, medical imaging, molecular or biochemical tests (e.g biopsy, blood or urine test) | 1.346 | %27 | 1.510 | %31 | 765 | %16 | 458 | %9 | 121 | %2 | 97 | %2 | 1.345 | %27 | 4.901 | |
| I know the NAME of the rare disease, syndrome or malformation but it has NOT yet been confirmed by appropriate genetic, clinical, medical imaging, molecular or biochemical tests | 49 | %21 | 93 | %39 | 31 | %13 | 30 | %13 | 5 | %2 | 10 | %4 | 75 | %31 | 239 | |
| I only have PARTIAL information on the name of the rare disease or the gene involved or the type of disease | 30 | %18 | 53 | %32 | 36 | %22 | 31 | %19 | 6 | %4 | 4 | %2 | 48 | %29 | 164 | |
| I know that the disease is rare but the name or the cause have NOT BEEN IDENTIFIED | 33 | %18 | 73 | %40 | 47 | %26 | 46 | %25 | 3 | %2 | 5 | %3 | 41 | %23 | 181 | |
| Other, specify... | 2 | %40 | 2 | %40 | 1 | %20 | 2 | %40 | 0 | %0 | 1 | %20 | 2 | %40 | 5 | |
| **TOTAL** | 1.460 | %27 | 1.731 | %32 | 880 | %16 | 567 | %10 | 135 | %2 | 117 | %2 | 1.511 | %28 | 5.490 | |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 120,1 ; dof= 24.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Point prevalence of the rare disease / Genetic test(s) looking for genetic changes (also called mutations or variants)**

| POINT PREVALENCE OF THE RARE DISEASE | GENETIC TEST(S) LOOKING FOR GENETIC CHANGES (ALSO CALLED MUTATIONS OR VARIANTS) | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW/DON'T REMEMBER | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 1-5 / 10 000 | 1.195 | %50 | 1.026 | %43 | 186 | %8 | 2.407 | %100 |
| 1-9 / 100 000 | 1.111 | %56 | 751 | %38 | 137 | %7 | 1.999 | %100 |
| 1-9 / 1 000 000 | 300 | %65 | 132 | %29 | 27 | %6 | 459 | %100 |
| <1 / 1 000 000 | 547 | %64 | 258 | %30 | 51 | %6 | 856 | %100 |
| TOTAL | 3.153 | %55 | 2.167 | %38 | 401 | %7 | 5.721 | |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 75,9 ; dof= 6.*

**Cross: I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases / Genetic test(s) looking for genetic changes (also called mutations or variants)**

| I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | GENETIC TEST(S) LOOKING FOR GENETIC CHANGES (ALSO CALLED MUTATIONS OR VARIANTS) | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW/DON'T REMEMBER | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 3.458 | %58 | 2.038 | %34 | 502 | %8 | 5.998 | %100 |
| No | 1.998 | %45 | 2.097 | %47 | 320 | %7 | 4.415 | %100 |
| TOTAL | 5.456 | %52 | 4.135 | %40 | 822 | %8 | 10.413 | %100 |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 195,7 ; dof= 2.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases / Other test(s) such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc**

| I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | OTHER TEST(S) SUCH AS CLINICAL EXAMINATION(S), MEDICAL IMAGING (MRI, SCANS…), BIOPSY, BIOCHEMICAL TEST(S) (BLOOD OR URINE TESTS…), ETC | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW/DON'T REMEMBER | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 5.513 | %92 | 348 | %6 | 137 | %2 | 5.998 | %100 |
| No | 3.906 | %88 | 381 | %9 | 128 | %3 | 4.415 | %100 |
| TOTAL | 9.419 | %90 | 729 | %7 | 265 | %3 | 10.413 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 36,2 ; dof= 2.*

**Cross: I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases / …you could not afford it?**

Have you ever needed a genetic test but could not access it because...

| I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | …YOU COULD NOT AFFORD IT? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 507 | %8 | 4.292 | %72 | 1.199 | %20 | 5.998 | %100 |
| No | 587 | %13 | 2.780 | %63 | 1.048 | %24 | 4.415 | %100 |
| TOTAL | 1.094 | %11 | 7.072 | %68 | 2.247 | %22 | 10.413 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 100,9 ; dof= 2.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases / …it was not available in your country?**

**Have you ever needed a genetic test but could not access it because...**
...IT WAS NOT AVAILABLE IN YOUR COUNTRY?

| I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | YES | | NO | | NOT RELEVANT | | TOTAL | |
|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % |
| Yes | 607 | %10 | 4.045 | %67 | 1.346 | %22 | 5.998 | %100 |
| No | 578 | %13 | 2.738 | %62 | 1.099 | %25 | 4.415 | %100 |
| TOTAL | 1.185 | %11 | 6.783 | %65 | 2.445 | %23 | 10.413 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 37,7 ; dof= 2.*

**Cross: I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases / …healthcare professionals were reluctant or not sufficiently informed?**

**Have you ever needed a genetic test but could not access it because...**
...HEALTHCARE PROFESSIONALS WERE RELUCTANT OR NOT SUFFICIENTLY INFORMED?

| I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | YES | | NO | | NOT RELEVANT | | TOTAL | |
|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % |
| Yes | 1.372 | %23 | 3.451 | %58 | 1.175 | %20 | 5.998 | %100 |
| No | 1.408 | %32 | 2.064 | %47 | 943 | %21 | 4.415 | %100 |
| TOTAL | 2.780 | %27 | 5.515 | %53 | 2.118 | %20 | 10.413 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 137,2 ; dof= 2.*

**Cross: I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases / To your knowledge, the genetic test(s) that were conducted targeted…**

| I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | TO YOUR KNOWLEDGE, THE GENETIC TEST(S) THAT WERE CONDUCTED TARGETED… | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | ONLY ONE GENE | | SEVERAL GENES AT THE SAME TIME (GENE PANEL SEQUENCING) | | THE WHOLE DNA (WHOLE GENOME SEQUENCING) | | ALL THE GENES (WHOLE EXOME SEQUENCING) | | A TUMOUR (GENETIC PROFILING OF A TUMOUR) | | OTHER (EPIGENOME, RNA, ETC.) | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 945 | %27 | 1.079 | %31 | 562 | %16 | 330 | %10 | 84 | %2 | 70 | %2 | 960 | %28 | 3.458 | |
| No | 498 | %25 | 648 | %32 | 315 | %16 | 231 | %12 | 51 | %3 | 47 | %2 | 540 | %27 | 1.998 | |
| TOTAL | 1.443 | %26 | 1.727 | %32 | 877 | %16 | 561 | %10 | 135 | %2 | 117 | %2 | 1.500 | %27 | 5.456 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is weakly significant. p-value= 0,1 ; Chi2= 9,5 ; dof= 6.*

**Cross: I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases / Did you ever request a private company or laboratory to conduct genetic testing to diagnose the disease?**

| I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | DID YOU EVER REQUEST A PRIVATE COMPANY OR LABORATORY TO CONDUCT GENETIC TESTING TO DIAGNOSE THE DISEASE? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO, NEVER | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 325 | %9 | 143 | %4 | 2.990 | %86 | 3.458 | %100 |
| No | 258 | %13 | 119 | %6 | 1.621 | %81 | 1.998 | %100 |
| TOTAL | 583 | %11 | 262 | %5 | 4.611 | %85 | 5.456 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 27,6 ; dof= 2.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases / In general, how satisfied are you with how the results of the GENETIC TESTS were given to you?**

| I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | IN GENERAL, HOW SATISFIED ARE YOU WITH HOW THE RESULTS OF THE GENETIC TESTS WERE GIVEN TO YOU? | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | VERY DISSATISFIED | | DISSATISFIED | | NEITHER SATISFIED NOR DISSATISFIED | | SATISFIED | | VERY SATISFIED | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 351 | %10 | 353 | %10 | 690 | %20 | 1.232 | %36 | 656 | %19 | 176 | %5 | 3.458 | %100 |
| No | 216 | %11 | 269 | %13 | 459 | %23 | 685 | %34 | 264 | %13 | 105 | %5 | 1.998 | %100 |
| TOTAL | 567 | %10 | 622 | %11 | 1.149 | %21 | 1.917 | %35 | 920 | %17 | 281 | %5 | 5.456 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 43,4 ; dof= 5.*

**Cross: I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases / After the tests were performed, were you offered genetic counselling (e.g. given information about how your genetic condition might affect you and your family)?**

| I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | AFTER THE TESTS WERE PERFORMED, WERE YOU OFFERED GENETIC COUNSELLING (E.G. GIVEN INFORMATION ABOUT HOW YOUR GENETIC CONDITION MIGHT AFFECT YOU AND YOUR FAMILY)? | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| | YES, WITH A GENETIC COUNSELLOR OR CLINICAL GENETICIST | | YES, BY A HEALTHCARE PROFESSIONAL | | NO, I WASN'T OFFERED GENETIC COUNSELLING | | NOT SURE / DON'T REMEMBER | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % |
| Yes | 1.398 | %40 | 827 | %24 | 972 | %28 | 261 | %8 | 3.458 | %100 |
| No | 733 | %37 | 340 | %17 | 785 | %39 | 140 | %7 | 1.998 | %100 |
| TOTAL | 2.131 | %39 | 1.167 | %21 | 1.757 | %32 | 401 | %7 | 5.456 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 82,4 ; dof= 3.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: …you could not afford it? / Did you ever request a private company or laboratory to conduct genetic testing to diagnose the disease?

**Have you ever needed a genetic test but could not access it because...**

| ...YOU COULD NOT AFFORD IT? | DID YOU EVER REQUEST A PRIVATE COMPANY OR LABORATORY TO CONDUCT GENETIC TESTING TO DIAGNOSE THE DISEASE? | | | | | | | |
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO, NEVER | | TOTAL | |
| | N | % | N | % | N | % | N | % |
|---|---|---|---|---|---|---|---|---|
| Yes | 136 | %24 | 83 | %15 | 353 | %62 | 572 | %100 |
| No | 398 | %9 | 164 | %4 | 3.750 | %87 | 4.312 | %100 |
| Not relevant | 56 | %9 | 19 | %3 | 531 | %88 | 606 | %100 |
| **TOTAL** | **590** | **%11** | **266** | **%5** | **4.634** | **%84** | **5.490** | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 263,5 ; dof= 4.*

## Cross: …it was not available in your country? / Did you ever request a private company or laboratory to conduct genetic testing to diagnose the disease?

**Have you ever needed a genetic test but could not access it because...**

| ...IT WAS NOT AVAILABLE IN YOUR COUNTRY? | DID YOU EVER REQUEST A PRIVATE COMPANY OR LABORATORY TO CONDUCT GENETIC TESTING TO DIAGNOSE THE DISEASE? | | | | | | | |
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO, NEVER | | TOTAL | |
| | N | % | N | % | N | % | N | % |
|---|---|---|---|---|---|---|---|---|
| Yes | 174 | %24 | 95 | %13 | 470 | %64 | 739 | %100 |
| No | 370 | %9 | 153 | %4 | 3.632 | %87 | 4.155 | %100 |
| Not relevant | 46 | %8 | 18 | %3 | 532 | %89 | 596 | %100 |
| **TOTAL** | **590** | **%11** | **266** | **%5** | **4.634** | **%84** | **5.490** | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 288,4 ; dof= 4.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: …healthcare professionals were reluctant or not sufficiently informed? / Did you ever request a private company or laboratory to conduct genetic testing to diagnose the disease?**

Have you ever needed a genetic test
but could not access it because…

| …HEALTHCARE PROFESSIONALS WERE RELUCTANT OR NOT SUFFICIENTLY INFORMED? | DID YOU EVER REQUEST A PRIVATE COMPANY OR LABORATORY TO CONDUCT GENETIC TESTING TO DIAGNOSE THE DISEASE? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES, ONE TIME | | YES, SEVERAL TIMES | | NO, NEVER | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 264 | %17 | 150 | %10 | 1.106 | %73 | 1.520 | %100 |
| No | 287 | %8 | 101 | %3 | 3.104 | %89 | 3.492 | %100 |
| Not relevant | 39 | %8 | 15 | %3 | 424 | %89 | 478 | %100 |
| **TOTAL** | **590** | **%11** | **266** | **%5** | **4.634** | **%84** | **5.490** | **%100** |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

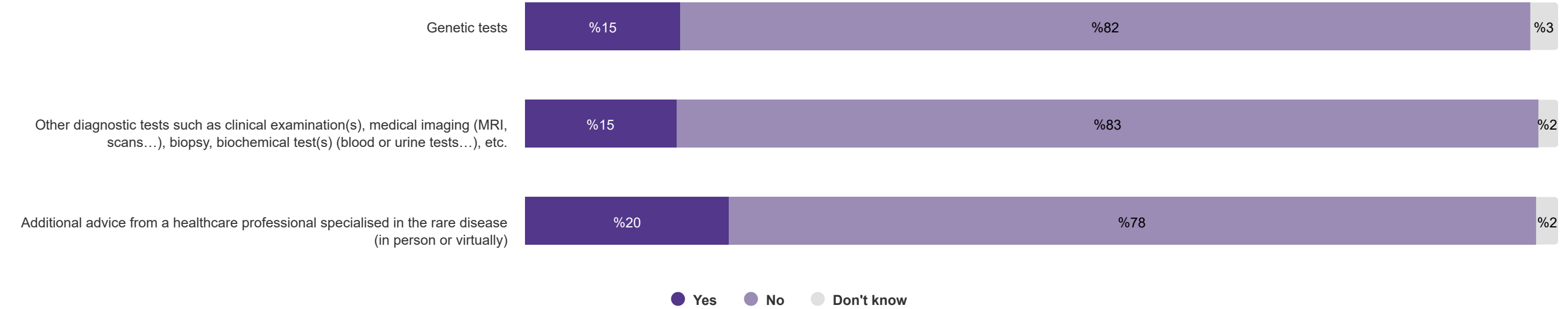*The relationship is very significant. p-value= < 0,01 ; Chi2= 229,0 ; dof= 4.*

**Cross: To your knowledge, the genetic test(s) that were conducted targeted… / After the tests were performed, were you offered genetic counselling (e.g. given information about how your genetic condition might affect you and your family)?**

| TO YOUR KNOWLEDGE, THE GENETIC TEST(S) THAT WERE CONDUCTED TARGETED… | AFTER THE TESTS WERE PERFORMED, WERE YOU OFFERED GENETIC COUNSELLING (E.G. GIVEN INFORMATION ABOUT HOW YOUR GENETIC CONDITION MIGHT AFFECT YOU AND YOUR FAMILY)? | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| | YES, WITH A GENETIC COUNSELLOR OR CLINICAL GENETICIST | | YES, BY A HEALTHCARE PROFESSIONAL | | NO, I WASN'T OFFERED GENETIC COUNSELLING | | NOT SURE / DON'T REMEMBER | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % |
| Only one gene | 590 | %40 | 324 | %22 | 471 | %32 | 75 | %5 | 1.460 | %100 |
| Several genes at the same time (gene panel sequencing) | 775 | %45 | 359 | %21 | 514 | %30 | 83 | %5 | 1.731 | %100 |
| The whole DNA (Whole Genome Sequencing) | 440 | %50 | 173 | %20 | 214 | %24 | 53 | %6 | 880 | %100 |
| All the genes (Whole Exome Sequencing) | 283 | %50 | 99 | %17 | 163 | %29 | 22 | %4 | 567 | %100 |
| A tumour (genetic profiling of a tumour) | 38 | %28 | 25 | %19 | 60 | %44 | 12 | %9 | 135 | %100 |
| Other (epigenome, RNA, etc.) | 37 | %32 | 26 | %22 | 42 | %36 | 12 | %10 | 117 | %100 |
| Don't know | 394 | %26 | 352 | %23 | 568 | %38 | 197 | %13 | 1.511 | %100 |
| **TOTAL** | **2.137** | **%39** | **1.179** | **%21** | **1.770** | **%32** | **404** | **%7** | **5.490** | **%100** |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 288,0 ; dof= 18.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Did you ever request a private company or laboratory to conduct genetic testing to diagnose the disease? / In general, how satisfied are you with how the results of the GENETIC TESTS were given to you?**

| DID YOU EVER REQUEST A PRIVATE COMPANY OR LABORATORY TO CONDUCT GENETIC TESTING TO DIAGNOSE THE DISEASE? | IN GENERAL, HOW SATISFIED ARE YOU WITH HOW THE RESULTS OF THE GENETIC TESTS WERE GIVEN TO YOU? | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | VERY DISSATISFIED | | DISSATISFIED | | NEITHER SATISFIED NOR DISSATISFIED | | SATISFIED | | VERY SATISFIED | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, one time | 63 | %11 | 80 | %14 | 149 | %25 | 193 | %33 | 85 | %14 | 20 | %3 | 590 | %100 |
| YES, several times | 44 | %17 | 40 | %15 | 68 | %26 | 74 | %28 | 32 | %12 | 8 | %3 | 266 | %100 |
| NO, never | 465 | %10 | 503 | %11 | 942 | %20 | 1.663 | %36 | 806 | %17 | 255 | %6 | 4.634 | %100 |
| **TOTAL** | **572** | **%10** | **623** | **%11** | **1.159** | **%21** | **1.930** | **%35** | **923** | **%17** | **283** | **%5** | **5.490** | |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 45,0 ; dof= 10.*

**Cross: Did you ever request a private company or laboratory to conduct genetic testing to diagnose the disease? / After the tests were performed, were you offered genetic counselling (e.g. given information about how your genetic condition might affect you and your family)?**

| DID YOU EVER REQUEST A PRIVATE COMPANY OR LABORATORY TO CONDUCT GENETIC TESTING TO DIAGNOSE THE DISEASE? | AFTER THE TESTS WERE PERFORMED, WERE YOU OFFERED GENETIC COUNSELLING (E.G. GIVEN INFORMATION ABOUT HOW YOUR GENETIC CONDITION MIGHT AFFECT YOU AND YOUR FAMILY)? | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| | YES, WITH A GENETIC COUNSELLOR OR CLINICAL GENETICIST | | YES, BY A HEALTHCARE PROFESSIONAL | | NO, I WASN'T OFFERED GENETIC COUNSELLING | | NOT SURE / DON'T REMEMBER | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % |
| YES, one time | 211 | %36 | 125 | %21 | 224 | %38 | 30 | %5 | 590 | %100 |
| YES, several times | 85 | %32 | 55 | %21 | 114 | %43 | 12 | %5 | 266 | %100 |
| NO, never | 1.841 | %40 | 999 | %22 | 1.432 | %31 | 362 | %8 | 4.634 | %100 |
| **TOTAL** | **2.137** | **%39** | **1.179** | **%21** | **1.770** | **%32** | **404** | **%7** | **5.490** | |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 32,0 ; dof= 6.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

# Chapter 12.

## Cross-border healthcare

Rare
Barometer

A EURORDIS INITIATIVE

EURORDIS
RARE DISEASES EUROPE

## Did you access any of the following services in another country?

| | YES | NO | DON'T KNOW | TOTAL |
|---|---|---|---|---|
| Genetic tests | 831 | 4.515 | 143 | 5.489 |
| Other diagnostic tests such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc. | 1.403 | 7.908 | 170 | 9.481 |
| Additional advice from a healthcare professional specialised in the rare disease (in person or virtually) | 2.083 | 8.194 | 209 | 10.486 |
| TOTAL | 4.317 | 20.617 | 522 | 25.456 |

## Did you access any of the following services in another country?



Genetic tests: %15 Yes, %82 No, %3 Don't know

Other diagnostic tests such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc.: %15 Yes, %83 No, %2 Don't know

Additional advice from a healthcare professional specialised in the rare disease (in person or virtually): %20 Yes, %78 No, %2 Don't know

● Yes  ● No  ● Don't know

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

| Genetic tests | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| Yes | <u>0,0</u> | 624 | 3,1 | 548 | 3,5 | 344 | 3,8 | 612 | 5,0 | 543 |
| No | 0,8 | 3.307 | 3,9 | 3.019 | 4,4 | 2.049 | 3,8 | 3.388 | 5,7 | 2.952 |
| Don't know | 1,2 | 91 | 3,5 | 87 | 3,7 | 57 | 2,6 | 95 | 4,6 | 69 |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is significant. p-value= 0,0 ; Fisher= 3,5.*

*Inter variance= 161,7. Intra variance= 46,8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

| Other diagnostic tests such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc. | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| Yes | 0,2 | 1.067 | 3,8 | 992 | 3,6 | 513 | **4,3** | 1.028 | 5,4 | 861 |
| No | 0,5 | 6.037 | 3,5 | 5.700 | 3,9 | 3.449 | 3,5 | 6.062 | 4,6 | 5.105 |
| Don't know | 0,6 | 101 | 2,6 | 103 | 2,8 | 65 | **1,5** | 106 | **2,5** | 88 |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is not significant. p-value= 0,3 ; Fisher= 1,2.*

*Inter variance= 52,0. Intra variance= 43,6.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

| Additional advice from a healthcare professional specialised in the rare disease (in person or virtually) | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| Yes | 0,1 | 1.591 | 3,3 | 1.484 | 3,7 | 787 | 4,0 | 1.558 | 4,9 | 1.291 |
| No | 0,6 | 6.113 | 3,6 | 5.723 | 3,9 | 3.475 | 3,5 | 6.165 | 4,7 | 5.119 |
| Don't know | 2,1 | 116 | 3,4 | 115 | 3,2 | 73 | 3,4 | 120 | 3,5 | 97 |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Fisher= 6,1.*

*Inter variance= 279,8. Intra variance= 45,9.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Gender of the person affected by the rare disease / Genetic tests

| GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE | GENETIC TESTS | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Female | 396 | %13 | 2.628 | %84 | 88 | %3 | 3.112 | %100 |
| Male | 320 | %18 | 1.450 | %81 | 31 | %2 | 1.801 | %100 |
| Other | 20 | %32 | 37 | %60 | 5 | %8 | 62 | %100 |
| TOTAL | 736 | %15 | 4.115 | %83 | 124 | %2 | 4.975 | |

🟥 *Under-represented elements* 🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 52,1 ; dof= 4.*

## Cross: Gender of the person affected by the rare disease / Other diagnostic tests such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc.

| GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE | OTHER DIAGNOSTIC TESTS SUCH AS CLINICAL EXAMINATION(S), MEDICAL IMAGING (MRI, SCANS…), BIOPSY, BIOCHEMICAL TEST(S) (BLOOD OR URINE TESTS…), ETC. | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Female | 853 | %14 | 5.131 | %84 | 111 | %2 | 6.095 | %100 |
| Male | 394 | %16 | 2.074 | %83 | 37 | %1 | 2.505 | %100 |
| Other | 20 | %22 | 65 | %73 | 4 | %4 | 89 | %100 |
| TOTAL | 1.267 | %15 | 7.270 | %84 | 152 | %2 | 8.689 | |

🟥 *Under-represented elements* 🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 14,2 ; dof= 4.*

## Cross: Gender of the person affected by the rare disease / Additional advice from a healthcare professional specialised in the rare disease (in person or virtually)

| GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE | ADDITIONAL ADVICE FROM A HEALTHCARE PROFESSIONAL SPECIALISED IN THE RARE DISEASE (IN PERSON OR VIRTUALLY) | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Female | 1.228 | %18 | 5.298 | %80 | 133 | %2 | 6.659 | %100 |
| Male | 643 | %23 | 2.130 | %76 | 37 | %1 | 2.810 | %100 |
| Other | 25 | %25 | 70 | %69 | 6 | %6 | 101 | %100 |
| TOTAL | 1.896 | %20 | 7.498 | %78 | 176 | %2 | 9.570 | |

🟥 *Under-represented elements* 🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 39,9 ; dof= 4.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: How old were you when you stopped full-time education? / Genetic tests

| HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION? | GENETIC TESTS | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 15 y.o. or under | 52 | %20 | 197 | %76 | 10 | %4 | 259 | %100 |
| between 16 and 19 y.o. | 161 | %13 | 1.021 | %84 | 35 | %3 | 1.217 | %100 |
| between 20 and 23 y.o. | 218 | %14 | 1.332 | %84 | 34 | %2 | 1.584 | %100 |
| 24 y.o. or above | 286 | %17 | 1.396 | %81 | 37 | %2 | 1.719 | %100 |
| TOTAL | 717 | %15 | 3.946 | %83 | 116 | %2 | 4.779 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 18,4 ; dof= 6.*

## Cross: How old were you when you stopped full-time education? / Other diagnostic tests such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc.

| HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION? | OTHER DIAGNOSTIC TESTS SUCH AS CLINICAL EXAMINATION(S), MEDICAL IMAGING (MRI, SCANS…), BIOPSY, BIOCHEMICAL TEST(S) (BLOOD OR URINE TESTS…), ETC. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 15 y.o. or under | 65 | %16 | 328 | %81 | 13 | %3 | 406 | %100 |
| between 16 and 19 y.o. | 269 | %12 | 1.931 | %86 | 40 | %2 | 2.240 | %100 |
| between 20 and 23 y.o. | 415 | %15 | 2.291 | %83 | 43 | %2 | 2.749 | %100 |
| 24 y.o. or above | 451 | %16 | 2.358 | %83 | 48 | %2 | 2.857 | %100 |
| TOTAL | 1.200 | %15 | 6.908 | %84 | 144 | %2 | 8.252 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 22,2 ; dof= 6.*

## Cross: How old were you when you stopped full-time education? / Additional advice from a healthcare professional specialised in the rare disease (in person or virtually)

| HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION? | ADDITIONAL ADVICE FROM A HEALTHCARE PROFESSIONAL SPECIALISED IN THE RARE DISEASE (IN PERSON OR VIRTUALLY) | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 15 y.o. or under | 103 | %23 | 338 | %74 | 14 | %3 | 455 | %100 |
| between 16 and 19 y.o. | 430 | %17 | 1.979 | %80 | 55 | %2 | 2.464 | %100 |
| between 20 and 23 y.o. | 601 | %20 | 2.376 | %79 | 45 | %1 | 3.022 | %100 |
| 24 y.o. or above | 659 | %21 | 2.436 | %77 | 50 | %2 | 3.145 | %100 |
| TOTAL | 1.793 | %20 | 7.129 | %78 | 164 | %2 | 9.086 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 22,4 ; dof= 6.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: How would you best describe yourself? / Genetic tests

| HOW WOULD YOU BEST DESCRIBE YOURSELF? | GENETIC TESTS | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| I belong to the ethnic majority in the country where I live | **561** | **%15** | **3.055** | **%83** | 84 | %2 | **3.700** | **%100** |
| I am part of an ethnic minority in the country where I live | **65** | **%24** | **201** | **%75** | 3 | %1 | **269** | **%100** |
| Other, specify... | 36 | %21 | 129 | %76 | 4 | %2 | **169** | **%100** |
| **TOTAL** | **662** | **%16** | **3.385** | **%82** | **91** | **%2** | **4.138** | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 19,9 ; dof= 4.*

## Cross: How would you best describe yourself? / Other diagnostic tests such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc.

| HOW WOULD YOU BEST DESCRIBE YOURSELF? | OTHER DIAGNOSTIC TESTS SUCH AS CLINICAL EXAMINATION(S), MEDICAL II (MRI, SCANS…), BIOPSY, BIOCHEMICAL TEST(S) (BLOOD OR URINE TESTS... | | | | | | |
|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTA |
| | N | % | N | % | N | % | N |
| I belong to the ethnic majority in the country where I live | **909** | **%14** | **5.471** | **%84** | 102 | %2 | **6.482** |
| I am part of an ethnic minority in the country where I live | **120** | **%29** | **295** | **%71** | 2 | %0 | **417** |
| Other, specify... | 56 | %19 | **231** | **%77** | **12** | **%4** | **299** |
| **TOTAL** | **1.085** | **%15** | **5.997** | **%83** | **116** | **%2** | **7.198** |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 83,6 ; dof= 4.*

## Cross: How would you best describe yourself? / Additional advice from a healthcare professional specialised in the rare disease (in person or virtually)

| HOW WOULD YOU BEST DESCRIBE YOURSELF? | ADDITIONAL ADVICE FROM A HEALTHCARE PROFESSIONAL SPECIALISED IN THE RARE DISEASE (IN PERSON OR VIRTUALLY) | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| I belong to the ethnic majority in the country where I live | **1.417** | **%20** | **5.598** | **%79** | **110** | **%2** | 7.125 | %100 |
| I am part of an ethnic minority in the country where I live | **132** | **%28** | **323** | **%69** | 10 | %2 | 465 | %100 |
| Other, specify... | 69 | %20 | 252 | %75 | **16** | **%5** | 337 | %100 |
| **TOTAL** | **1.618** | **%20** | **6.173** | **%78** | **136** | **%2** | 7.927 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 40,3 ; dof= 4.*

## Cross: Typology of countries based on size and welfare / Genetic tests

| TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE | GENETIC TESTS | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Group A ('Eastern Europe') | 341 | %29 | 776 | %67 | 41 | %4 | 1.158 | %100 |
| Group B ('Western Europe') | 234 | %9 | 2.419 | %90 | 49 | %2 | 2.702 | %100 |
| Group C ('Northern Europe') | 230 | %16 | 1.190 | %81 | 49 | %3 | 1.469 | %100 |
| TOTAL | 805 | %15 | 4.385 | %82 | 139 | %3 | 5.329 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 296,0 ; dof= 4.*

## Cross: Typology of countries based on size and welfare / Other diagnostic tests such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc.

| TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE | OTHER DIAGNOSTIC TESTS SUCH AS CLINICAL EXAMINATION(S), MEDICAL IMAGING (MRI, SCANS…), BIOPSY, BIOCHEMICAL TEST(S) (BLOOD OR URINE TESTS…), ETC. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Group A ('Eastern Europe') | 387 | %24 | 1.184 | %73 | 60 | %4 | 1.631 | %100 |
| Group B ('Western Europe') | 516 | %11 | 4.042 | %88 | 56 | %1 | 4.614 | %100 |
| Group C ('Northern Europe') | 453 | %15 | 2.449 | %83 | 49 | %2 | 2.951 | %100 |
| TOTAL | 1.356 | %15 | 7.675 | %83 | 165 | %2 | 9.196 | %100 |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 203,6 ; dof= 4.*

## Cross: Typology of countries based on size and welfare / Additional advice from a healthcare professional specialised in the rare disease (in person or virtually)

| TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE | ADDITIONAL ADVICE FROM A HEALTHCARE PROFESSIONAL SPECIALISED IN THE RARE DISEASE (IN PERSON OR VIRTUALLY) | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Group A ('Eastern Europe') | 576 | %32 | 1.150 | %64 | 68 | %4 | 1.794 | %100 |
| Group B ('Western Europe') | 848 | %17 | 4.190 | %82 | 67 | %1 | 5.105 | %100 |
| Group C ('Northern Europe') | 606 | %19 | 2.602 | %79 | 65 | %2 | 3.273 | %100 |
| TOTAL | 2.030 | %20 | 7.942 | %78 | 200 | %2 | 10.172 | %100 |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 262,4 ; dof= 4.*

## Cross: Orphacode associated nomenclature (english) / Genetic tests

| ORPHACODE ASSOCIATED NOMENCLATURE (ENGLISH) | GENETIC TESTS | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Hereditary hemorrhagic telangiectasia | 22 | %6 | 339 | %92 | 8 | %2 | 369 | %100 |
| Hypermobile Ehlers-Danlos syndrome | 6 | %6 | 88 | %93 | 1 | %1 | 95 | %100 |
| Sarcoidosis | 1 | %10 | 8 | %80 | 1 | %10 | 10 | %100 |
| Classical Ehlers-Danlos syndrome | 4 | %6 | 58 | %91 | 2 | %3 | 64 | %100 |
| Williams syndrome | 6 | %5 | 121 | %92 | 4 | %3 | 131 | %100 |
| Cystic fibrosis | 14 | %12 | 101 | %83 | 6 | %5 | 121 | %100 |
| Myasthenia gravis | 3 | %18 | 14 | %82 | 0 | %0 | 17 | %100 |
| Systemic sclerosis | 0 | %0 | 4 | %100 | 0 | %0 | 4 | %100 |
| Tuberous sclerosis complex | 16 | %18 | 73 | %82 | 0 | %0 | 89 | %100 |
| Neurofibromatosis type 1 | 6 | %10 | 54 | %86 | 3 | %5 | 63 | %100 |
| Interstitial cystitis | 0 | %0 | 4 | %100 | 0 | %0 | 4 | %100 |
| Addison disease | 3 | %38 | 5 | %63 | 0 | %0 | 8 | %100 |
| 22q11.2 deletion syndrome | 4 | %6 | 55 | %87 | 4 | %6 | 63 | %100 |
| Chronic inflammatory demyelinating polyneuropathy | 3 | %19 | 13 | %81 | 0 | %0 | 16 | %100 |
| Perineural cyst | 0 | %0 | 6 | %100 | 0 | %0 | 6 | %100 |
| Acute inflammatory demyelinating polyradiculoneuropathy | 0 | %0 | 2 | %67 | 1 | %33 | 3 | %100 |
| Rett syndrome | 8 | %14 | 46 | %82 | 2 | %4 | 56 | %100 |
| Marfan syndrome | 5 | %12 | 37 | %88 | 0 | %0 | 42 | %100 |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is not significant. p-value= 0,4 ; Chi2= 2.456,3 ; dof= 2.438.*

## Cross: Orphacode associated nomenclature (english) / Other diagnostic tests such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc.

| ORPHACODE ASSOCIATED NOMENCLATURE (ENGLISH) | OTHER DIAGNOSTIC TESTS SUCH AS CLINICAL EXAMINATION(S), MEDICAL IMAGING (MRI, SCANS…), BIOPSY, BIOCHEMICAL TEST(S) (BLOOD OR URINE TESTS…), ETC. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Hereditary hemorrhagic telangiectasia | 32 | %8 | 368 | %90 | 8 | %2 | 408 | %100 |
| Hypermobile Ehlers-Danlos syndrome | 46 | %16 | 235 | %83 | 2 | %1 | 283 | %100 |
| Sarcoidosis | 14 | %8 | 154 | %91 | 1 | %1 | 169 | %100 |
| Classical Ehlers-Danlos syndrome | 14 | %11 | 110 | %89 | 0 | %0 | 124 | %100 |
| Williams syndrome | 5 | %6 | 72 | %90 | 3 | %4 | 80 | %100 |
| Cystic fibrosis | 19 | %17 | 89 | %77 | 7 | %6 | 115 | %100 |
| Myasthenia gravis | 33 | %28 | 83 | %70 | 2 | %2 | 118 | %100 |
| Systemic sclerosis | 6 | %6 | 100 | %94 | 0 | %0 | 106 | %100 |
| Tuberous sclerosis complex | 5 | %5 | 91 | %95 | 0 | %0 | 96 | %100 |
| Neurofibromatosis type 1 | 14 | %16 | 70 | %81 | 2 | %2 | 86 | %100 |
| Interstitial cystitis | 7 | %10 | 65 | %90 | 0 | %0 | 72 | %100 |
| Addison disease | 9 | %13 | 60 | %83 | 3 | %4 | 72 | %100 |
| 22q11.2 deletion syndrome | 4 | %8 | 46 | %88 | 2 | %4 | 52 | %100 |
| Chronic inflammatory demyelinating polyneuropathy | 12 | %19 | 52 | %81 | 0 | %0 | 64 | %100 |
| Perineural cyst | 12 | %20 | 48 | %79 | 1 | %2 | 61 | %100 |
| Acute inflammatory demyelinating polyradiculoneuropathy | 5 | %8 | 55 | %90 | 1 | %2 | 61 | %100 |
| Rett syndrome | 6 | %12 | 41 | %84 | 2 | %4 | 49 | %100 |
| Marfan syndrome | 4 | %9 | 39 | %91 | 0 | %0 | 43 | %100 |
| Fragile X syndrome | 1 | %3 | 32 | %97 | 0 | %0 | 33 | %100 |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 3.368,8 ; dof= 3.164.*

**Cross: Orphacode associated nomenclature (english) / Additional advice from a healthcare professional specialised in the rare disease (in person or virtually)**

| ORPHACODE ASSOCIATED NOMENCLATURE (ENGLISH) | ADDITIONAL ADVICE FROM A HEALTHCARE PROFESSIONAL SPECIALISED IN THE RARE DISEASE (IN PERSON OR VIRTUALLY) | | | | | | | |
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
|---|---|---|---|---|---|---|---|---|
| Hereditary hemorrhagic telangiectasia | _59_ | _%13_ | _389_ | _%85_ | 10 | %2 | **458** | **%100** |
| Hypermobile Ehlers-Danlos syndrome | 59 | %19 | 255 | %80 | 3 | %1 | **317** | **%100** |
| Sarcoidosis | _14_ | _%8_ | 155 | 91 | 1 | %1 | **170** | **%100** |
| Classical Ehlers-Danlos syndrome | _17_ | _%12_ | 119 | %87 | 1 | %1 | **137** | **%100** |
| Williams syndrome | 18 | %13 | 116 | %85 | 2 | %1 | **136** | **%100** |
| Cystic fibrosis | 26 | %20 | 92 | %72 | _10_ | _%8_ | **128** | **%100** |
| Myasthenia gravis | 26 | %22 | 93 | %78 | 1 | %1 | **120** | **%100** |
| Systemic sclerosis | _8_ | _%7_ | 99 | %93 | 0 | %0 | **107** | **%100** |
| Tuberous sclerosis complex | 23 | %23 | 75 | %77 | 0 | %0 | **98** | **%100** |
| Neurofibromatosis type 1 | 15 | %16 | 75 | %82 | 2 | %2 | **92** | **%100** |
| Interstitial cystitis | 13 | %18 | 61 | %82 | 0 | %0 | **74** | **%100** |
| Addison disease | 12 | %16 | 57 | %78 | _4_ | _%5_ | **73** | **%100** |
| 22q11.2 deletion syndrome | 11 | %16 | 56 | %82 | 1 | %1 | **68** | **%100** |
| Chronic inflammatory demyelinating polyneuropathy | 13 | %20 | 51 | %78 | 1 | %2 | **65** | **%100** |
| Perineural cyst | _22_ | _%35_ | _40_ | _%63_ | 1 | %2 | **63** | **%100** |
| Acute inflammatory demyelinating polyradiculoneuropathy | 7 | %11 | 53 | %85 | 2 | %3 | **62** | **%100** |
| Rett syndrome | 10 | %17 | 48 | %80 | 2 | %3 | **60** | **%100** |
| Marfan syndrome | 10 | %19 | 41 | %79 | 1 | %2 | **52** | **%100** |
| Fragile X syndrome | 6 | %12 | 40 | %82 | _3_ | _%6_ | **49** | **%100** |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 3.696,6 ; dof= 3.350.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Orphanet classification of rare diseases (one disease can be classified in several categories) / Genetic tests**

| ORPHANET CLASSIFICATION OF RARE DISEASES (ONE DISEASE CAN BE CLASSIFIED IN SEVERAL CATEGORIES) | GENETIC TESTS | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Abdominal surgical diseases | 7 | %7 | 93 | %89 | 5 | %5 | 105 | %100 |
| Allergic diseases | 0 | %0 | 1 | %100 | 0 | %0 | 1 | %100 |
| Bone diseases | 78 | %14 | 482 | %84 | 16 | %3 | 576 | %100 |
| Cardiac diseases | 46 | %15 | 247 | %83 | 5 | %2 | 298 | %100 |
| Cardiac malformations | 19 | %7 | 223 | %88 | 12 | %5 | 254 | %100 |
| Circulatory system diseases | 82 | %9 | 823 | %89 | 20 | %2 | 925 | %100 |
| Clinical sign | 0 | %0 | 0 | %0 | 0 | %0 | 0 | %100 |
| Developmental anomalies during embryogenesis | 272 | %12 | 1.942 | %86 | 49 | %2 | 2.263 | %100 |
| Diseases due to toxic effects | 0 | %0 | 2 | %100 | 0 | %0 | 2 | %100 |
| Endocrine diseases | 70 | %14 | 409 | %83 | 15 | %3 | 494 | %100 |
| Gastroenterological diseases | 30 | %13 | 187 | %83 | 9 | %4 | 226 | %100 |
| Genetic diseases | 584 | %15 | 3.190 | %83 | 88 | %2 | 3.862 | %100 |
| Gynecologic/obstetric diseases | 31 | %18 | 139 | %79 | 6 | %3 | 176 | %100 |
| Hematological diseases | 32 | %15 | 181 | %84 | 3 | %1 | 216 | %100 |
| Hepatic diseases | 66 | %10 | 585 | %87 | 21 | %3 | 672 | %100 |
| Immunological diseases | 42 | %21 | 158 | %77 | 4 | %2 | 204 | %100 |
| Inborn errors of metabolism | 139 | %22 | 469 | %75 | 18 | %3 | 626 | %100 |
| Infectious diseases | 0 | %0 | 1 | %100 | 0 | %0 | 1 | %100 |
| Infertility | 41 | %13 | 250 | %82 | 13 | %4 | 304 | %100 |

■ *Under-represented elements*　　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 201,0 ; dof= 68.*

Rare Barometer — A EURORDIS 'K' INITIATIVE

EURORDIS RARE DISEASES EUROPE

**Cross: Orphanet classification of rare diseases (one disease can be classified in several categories) / Other diagnostic tests such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc.**

| ORPHANET CLASSIFICATION OF RARE DISEASES (ONE DISEASE CAN BE CLASSIFIED IN SEVERAL CATEGORIES) | OTHER DIAGNOSTIC TESTS SUCH AS CLINICAL EXAMINATION(S), MEDICAL IMAGING (MRI, SCANS…), BIOPSY, BIOCHEMICAL TEST(S) (BLOOD OR URINE TESTS…), ETC. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Abdominal surgical diseases | 26 | %12 | 191 | %87 | 3 | %1 | **220** | %100 |
| Allergic diseases | 0 | %0 | 3 | %100 | 0 | %0 | **3** | %100 |
| Bone diseases | 90 | %13 | 579 | %86 | 7 | %1 | **676** | %100 |
| Cardiac diseases | **65** | **%10** | **549** | **%89** | 6 | %1 | **620** | %100 |
| Cardiac malformations | 20 | %10 | 178 | %86 | **9** | **%4** | **207** | %100 |
| Circulatory system diseases | **128** | **%11** | **1.038** | **%87** | 21 | %2 | **1.187** | %100 |
| Clinical sign | 0 | %0 | 0 | %0 | 0 | %0 | **0** | %100 |
| Developmental anomalies during embryogenesis | **373** | **%13** | **2.488** | **%86** | 37 | %1 | **2.898** | %100 |
| Diseases due to toxic effects | 0 | %0 | 3 | %100 | 0 | %0 | **3** | %100 |
| Endocrine diseases | 115 | %13 | 778 | %86 | 14 | %2 | **907** | %100 |
| Gastroenterological diseases | 37 | %13 | 230 | %83 | **11** | **%4** | **278** | %100 |
| Genetic diseases | 684 | %14 | 4.025 | %84 | 81 | %2 | **4.790** | %100 |
| Gynecologic/obstetric diseases | 39 | %15 | 210 | %83 | 3 | %1 | **252** | %100 |
| Hematological diseases | 64 | %16 | 319 | %81 | 10 | %3 | **393** | %100 |
| Hepatic diseases | **95** | **%12** | 697 | %86 | 19 | %2 | **811** | %100 |
| Immunological diseases | 41 | %16 | 207 | %81 | 8 | %3 | **256** | %100 |
| Inborn errors of metabolism | **136** | **%19** | **563** | **%78** | **19** | **%3** | **718** | %100 |
| Infectious diseases | 3 | %18 | 14 | %82 | 0 | %0 | **17** | %100 |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 122,9 ; dof= 68.*

**Cross: Orphanet classification of rare diseases (one disease can be classified in several categories) / Additional advice from a healthcare professional specialised in the rare disease (in person or virtually)**

| ORPHANET CLASSIFICATION OF RARE DISEASES (ONE DISEASE CAN BE CLASSIFIED IN SEVERAL CATEGORIES) | ADDITIONAL ADVICE FROM A HEALTHCARE PROFESSIONAL SPECIALISED IN THE RARE DISEASE (IN PERSON OR VIRTUALLY) | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Abdominal surgical diseases | 43 | %18 | 193 | %81 | 3 | %1 | 239 | %100 |
| Allergic diseases | 1 | %33 | 2 | %67 | 0 | %0 | 3 | %100 |
| Bone diseases | 179 | %22 | 606 | %76 | 14 | %2 | 799 | %100 |
| Cardiac diseases | 111 | %17 | 542 | %82 | 7 | %1 | 660 | %100 |
| Cardiac malformations | 48 | %16 | 238 | %81 | 9 | %3 | 295 | %100 |
| Circulatory system diseases | 226 | %17 | 1.104 | %82 | 21 | %2 | 1.351 | %100 |
| Clinical sign | 0 | %0 | 0 | %0 | 0 | %0 | 0 | %100 |
| Developmental anomalies during embryogenesis | 664 | %20 | 2.626 | %78 | 57 | %2 | 3.347 | %100 |
| Diseases due to toxic effects | 1 | %33 | 2 | %67 | 0 | %0 | 3 | %100 |
| Endocrine diseases | 197 | %20 | 775 | %78 | 23 | %2 | 995 | %100 |
| Gastroenterological diseases | 55 | %18 | 234 | %77 | 16 | %5 | 305 | %100 |
| Genetic diseases | 1.135 | %21 | 4.205 | %77 | 107 | %2 | 5.447 | %100 |
| Gynecologic/obstetric diseases | 65 | %23 | 212 | %75 | 7 | %2 | 284 | %100 |
| Hematological diseases | 85 | %21 | 317 | %77 | 10 | %2 | 412 | %100 |
| Hepatic diseases | 133 | %15 | 730 | %82 | 28 | %3 | 891 | %100 |
| Immunological diseases | 58 | %20 | 221 | %77 | 7 | %2 | 286 | %100 |
| Inborn errors of metabolism | 188 | %24 | 560 | %72 | 26 | %3 | 774 | %100 |
| Infectious diseases | 3 | %18 | 14 | %82 | 0 | %0 | 17 | %100 |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 173,2 ; dof= 68.*

Rare Barometer
A EURORDIS INITIATIVE
EURORDIS RARE DISEASES EUROPE

**Cross: Please select the sentence that best describes your situation or the situation of the person you care for: / Genetic tests**

| PLEASE SELECT THE SENTENCE THAT BEST DESCRIBES YOUR SITUATION OR THE SITUATION OF THE PERSON YOU CARE FOR: | GENETIC TESTS | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| I know the NAME of the rare disease, syndrome or malformation and it has been CONFIRMED by appropriate genetic, clinical, medical imaging, molecular or biochemical tests (e.g biopsy, blood or urine test) | 726 | %15 | 4.053 | %83 | 122 | %2 | 4.901 | %100 |
| I know the NAME of the rare disease, syndrome or malformation but it has NOT yet been confirmed by appropriate genetic, clinical, medical imaging, molecular or biochemical tests | 37 | %16 | 191 | %80 | 10 | %4 | 238 | %100 |
| I only have PARTIAL information on the name of the rare disease or the gene involved or the type of disease | 29 | %18 | 130 | %79 | 5 | %3 | 164 | %100 |
| I know that the disease is rare but the name or the cause have NOT BEEN IDENTIFIED | 37 | %20 | 138 | %76 | 6 | %3 | 181 | %100 |
| Other, specify... | 2 | %40 | 3 | %60 | 0 | %0 | 5 | %100 |
| **TOTAL** | **831** | **%15** | **4.515** | **%82** | **143** | **%3** | **5.489** | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is not significant. p-value= 0,2 ; Chi2= 11,2 ; dof= 8.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

**Cross: Please select the sentence that best describes your situation or the situation of the person you care for: / Other diagnostic tests such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc.**

| PLEASE SELECT THE SENTENCE THAT BEST DESCRIBES YOUR SITUATION OR THE SITUATION OF THE PERSON YOU CARE FOR: | YES | | NO | | DON'T KNOW | | TOTAL | |
|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % |
| I know the NAME of the rare disease, syndrome or malformation and it has been CONFIRMED by appropriate genetic, clinical, medical imaging, molecular or biochemical tests (e.g biopsy, blood or urine test) | 1.183 | %14 | 6.942 | %84 | 146 | %2 | 8.271 | %100 |
| I know the NAME of the rare disease, syndrome or malformation but it has NOT yet been confirmed by appropriate genetic, clinical, medical imaging, molecular or biochemical tests | 96 | %16 | 502 | %83 | 9 | %1 | 607 | %100 |
| I only have PARTIAL information on the name of the rare disease or the gene involved or the type of disease | 62 | %23 | 206 | %76 | 4 | %1 | 272 | %100 |
| I know that the disease is rare but the name or the cause have NOT BEEN IDENTIFIED | 54 | %17 | 250 | %80 | 8 | %3 | 312 | %100 |
| Other, specify... | 8 | %42 | 8 | %42 | 3 | %16 | 19 | %100 |
| TOTAL | 1.403 | %15 | 7.908 | %83 | 170 | %2 | 9.481 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 53,3 ; dof= 8.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

**Cross: Please select the sentence that best describes your situation or the situation of the person you care for: / Additional advice from a healthcare professional specialised in the rare disease (in person or virtually)**

| PLEASE SELECT THE SENTENCE THAT BEST DESCRIBES YOUR SITUATION OR THE SITUATION OF THE PERSON YOU CARE FOR: | ADDITIONAL ADVICE FROM A HEALTHCARE PROFESSIONAL SPECIALISED IN THE RARE DISEASE (IN PERSON OR VIRTUALLY) | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| I know the NAME of the rare disease, syndrome or malformation and it has been CONFIRMED by appropriate genetic, clinical, medical imaging, molecular or biochemical tests (e.g biopsy, blood or urine test) | 1.795 | %20 | 7.079 | %78 | 174 | %2 | 9.048 | %100 |
| I know the NAME of the rare disease, syndrome or malformation but it has NOT yet been confirmed by appropriate genetic, clinical, medical imaging, molecular or biochemical tests | 141 | %19 | 609 | %80 | 10 | %1 | 760 | %100 |
| I only have PARTIAL information on the name of the rare disease or the gene involved or the type of disease | 73 | %24 | 221 | %72 | 12 | %4 | 306 | %100 |
| I know that the disease is rare but the name or the cause have NOT BEEN IDENTIFIED | 68 | %20 | 271 | %78 | 9 | %3 | 348 | %100 |
| Other, specify... | 6 | %25 | 14 | %58 | 4 | %17 | 24 | %100 |
| TOTAL | 2.083 | %20 | 8.194 | %78 | 209 | %2 | 10.486 | |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 40,7 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Point prevalence of the rare disease / Genetic tests

| POINT PREVALENCE OF THE RARE DISEASE | GENETIC TESTS | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 1-5 / 10 000 | 123 | %10 | 1.039 | %87 | 33 | %3 | 1.195 | %100 |
| 1-9 / 100 000 | 160 | %14 | 924 | %83 | 26 | %2 | 1.110 | %100 |
| 1-9 / 1 000 000 | 65 | %22 | 227 | %76 | 8 | %3 | 300 | %100 |
| <1 / 1 000 000 | 98 | %18 | 439 | %80 | 10 | %2 | 547 | %100 |
| TOTAL | 446 | %14 | 2.629 | %83 | 77 | %2 | 3.152 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 36,1 ; dof= 6.*

## Cross: Point prevalence of the rare disease / Other diagnostic tests such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc.

| POINT PREVALENCE OF THE RARE DISEASE | OTHER DIAGNOSTIC TESTS SUCH AS CLINICAL EXAMINATION(S), MEDICAL IMAGING (MRI, SCANS…), BIOPSY, BIOCHEMICAL TEST(S) (BLOOD OR URINE TESTS…), ETC. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 1-5 / 10 000 | 243 | %11 | 1.925 | %87 | 34 | %2 | 2.202 | %100 |
| 1-9 / 100 000 | 263 | %14 | 1.532 | %84 | 37 | %2 | 1.832 | %100 |
| 1-9 / 1 000 000 | 74 | %18 | 343 | %81 | 5 | %1 | 422 | %100 |
| <1 / 1 000 000 | 114 | %15 | 625 | %83 | 13 | %2 | 752 | %100 |
| TOTAL | 694 | %13 | 4.425 | %85 | 89 | %2 | 5.208 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 22,6 ; dof= 6.*

## Cross: Point prevalence of the rare disease / Additional advice from a healthcare professional specialised in the rare disease (in person or virtually)

| POINT PREVALENCE OF THE RARE DISEASE | ADDITIONAL ADVICE FROM A HEALTHCARE PROFESSIONAL SPECIALISED IN THE RARE DISEASE (IN PERSON OR VIRTUALLY) | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 1-5 / 10 000 | 375 | %16 | 1.989 | %83 | 43 | %2 | 2.407 | %100 |
| 1-9 / 100 000 | 392 | %20 | 1.565 | %78 | 42 | %2 | 1.999 | %100 |
| 1-9 / 1 000 000 | 104 | %23 | 342 | %75 | 13 | %3 | 459 | %100 |
| <1 / 1 000 000 | 196 | %23 | 645 | %75 | 15 | %2 | 856 | %100 |
| TOTAL | 1.067 | %19 | 4.541 | %79 | 113 | %2 | 5.721 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 34,5 ; dof= 6.*

### Cross: …behavioural disorders that cause problems in school, at home or in social situations / Genetic tests

| …BEHAVIOURAL DISORDERS THAT CAUSE PROBLEMS IN SCHOOL, AT HOME OR IN SOCIAL SITUATIONS | GENETIC TESTS | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 263 | %15 | 1.453 | %82 | 54 | %3 | 1.770 | %100 |
| No | 538 | %15 | 2.892 | %82 | 80 | %2 | 3.510 | %100 |
| Don't know | 30 | %14 | 170 | %81 | 9 | %4 | 209 | %100 |
| TOTAL | 831 | %15 | 4.515 | %82 | 143 | %3 | 5.489 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is not significant. p-value= 0,2 ; Chi2= 5,4 ; dof= 4.*

### Cross: …behavioural disorders that cause problems in school, at home or in social situations / Other diagnostic tests such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc.

| …BEHAVIOURAL DISORDERS THAT CAUSE PROBLEMS IN SCHOOL, AT HOME OR IN SOCIAL SITUATIONS | OTHER DIAGNOSTIC TESTS SUCH AS CLINICAL EXAMINATION(S), MEDICAL IMAGING (MRI, SCANS…), BIOPSY, BIOCHEMICAL TEST(S) (BLOOD OR URINE TESTS…), ETC. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 424 | %16 | 2.122 | %82 | 51 | %2 | 2.597 | %100 |
| No | 915 | %14 | 5.480 | %84 | 107 | %2 | 6.502 | %100 |
| Don't know | 64 | %17 | 306 | %80 | 12 | %3 | 382 | %100 |
| TOTAL | 1.403 | %15 | 7.908 | %83 | 170 | %2 | 9.481 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 14,5 ; dof= 4.*

### Cross: …behavioural disorders that cause problems in school, at home or in social situations / Additional advice from a healthcare professional specialised in the rare disease (in person or virtually)

| …BEHAVIOURAL DISORDERS THAT CAUSE PROBLEMS IN SCHOOL, AT HOME OR IN SOCIAL SITUATIONS | ADDITIONAL ADVICE FROM A HEALTHCARE PROFESSIONAL SPECIALISED IN THE RARE DISEASE (IN PERSON OR VIRTUALLY) | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 712 | %24 | 2.185 | %74 | 60 | %2 | 2.957 | %100 |
| No | 1.304 | %18 | 5.647 | %80 | 134 | %2 | 7.085 | %100 |
| Don't know | 67 | %15 | 362 | %82 | 15 | %3 | 444 | %100 |
| TOTAL | 2.083 | %20 | 8.194 | %78 | 209 | %2 | 10.486 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 53,5 ; dof= 4.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: …intellectual disabilities or cognitive symptoms (i.e. problems with memory, language, thinking or judgement) / Genetic tests

| …INTELLECTUAL DISABILITIES OR COGNITIVE SYMPTOMS (I.E. PROBLEMS WITH MEMORY, LANGUAGE, THINKING OR JUDGEMENT) | GENETIC TESTS | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 309 | %16 | 1.528 | %81 | 51 | %3 | 1.888 | %100 |
| No | 502 | %14 | 2.875 | %83 | 88 | %3 | 3.465 | %100 |
| Don't know | 20 | %15 | 112 | %82 | 4 | %3 | 136 | %100 |
| TOTAL | 831 | %15 | 4.515 | %82 | 143 | %3 | 5.489 | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is not significant. p-value= 0,5 ; Chi2= 3,7 ; dof= 4.*

## Cross: …intellectual disabilities or cognitive symptoms (i.e. problems with memory, language, thinking or judgement) / Other diagnostic tests such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc.

| …INTELLECTUAL DISABILITIES OR COGNITIVE SYMPTOMS (I.E. PROBLEMS WITH MEMORY, LANGUAGE, THINKING OR JUDGEMENT) | OTHER DIAGNOSTIC TESTS SUCH AS CLINICAL EXAMINATION(S), MEDICAL IMAGING (MRI, SCANS…), BIOPSY, BIOCHEMICAL TEST(S) (BLOOD OR URINE TESTS…), ETC. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 424 | %16 | 2.116 | %82 | 47 | %2 | 2.587 | %100 |
| No | 932 | %14 | 5.577 | %84 | 114 | %2 | 6.623 | %100 |
| Don't know | 47 | %17 | 215 | %79 | 9 | %3 | 271 | %100 |
| TOTAL | 1.403 | %15 | 7.908 | %83 | 170 | %2 | 9.481 | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 13,6 ; dof= 4.*

## Cross: …intellectual disabilities or cognitive symptoms (i.e. problems with memory, language, thinking or judgement) / Additional advice from a healthcare professional specialised in the rare disease (in person or virtually)

| …INTELLECTUAL DISABILITIES OR COGNITIVE SYMPTOMS (I.E. PROBLEMS WITH MEMORY, LANGUAGE, THINKING OR JUDGEMENT) | ADDITIONAL ADVICE FROM A HEALTHCARE PROFESSIONAL SPECIALISED IN THE RARE DISEASE (IN PERSON OR VIRTUALLY) | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 706 | %24 | 2.175 | %74 | 55 | %2 | 2.936 | %100 |
| No | 1.328 | %18 | 5.769 | %80 | 139 | %2 | 7.236 | %100 |
| Don't know | 49 | %16 | 250 | %80 | 15 | %5 | 314 | %100 |
| TOTAL | 2.083 | %20 | 8.194 | %78 | 209 | %2 | 10.486 | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 58,3 ; dof= 4.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: …clinical signs or symptoms that come and go / Genetic tests

| …CLINICAL SIGNS OR SYMPTOMS THAT COME AND GO | GENETIC TESTS | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 421 | %15 | 2.350 | %83 | 67 | %2 | 2.838 | %100 |
| No | 362 | %16 | 1.827 | %81 | 61 | %3 | 2.250 | %100 |
| Don't know | 48 | %12 | 338 | %84 | 15 | %4 | 401 | %100 |
| TOTAL | 831 | %15 | 4.515 | %82 | 143 | %3 | 5.489 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is weakly significant. p-value= 0,1 ; Chi2= 7,5 ; dof= 4.*

## Cross: …clinical signs or symptoms that come and go / Other diagnostic tests such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc.

| …CLINICAL SIGNS OR SYMPTOMS THAT COME AND GO | OTHER DIAGNOSTIC TESTS SUCH AS CLINICAL EXAMINATION(S), MEDICAL IMAGING (MRI, SCANS…), BIOPSY, BIOCHEMICAL TEST(S) (BLOOD OR URINE TESTS…), ETC. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 844 | %15 | 4.557 | %83 | 86 | %2 | 5.487 | %100 |
| No | 484 | %14 | 2.805 | %84 | 55 | %2 | 3.344 | %100 |
| Don't know | 75 | %12 | 546 | %84 | 29 | %4 | 650 | %100 |
| TOTAL | 1.403 | %15 | 7.908 | %83 | 170 | %2 | 9.481 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 34,2 ; dof= 4.*

## Cross: …clinical signs or symptoms that come and go / Additional advice from a healthcare professional specialised in the rare disease (in person or virtually)

| …CLINICAL SIGNS OR SYMPTOMS THAT COME AND GO | ADDITIONAL ADVICE FROM A HEALTHCARE PROFESSIONAL SPECIALISED IN THE RARE DISEASE (IN PERSON OR VIRTUALLY) | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 1.205 | %20 | 4.635 | %78 | 100 | %2 | 5.940 | %100 |
| No | 757 | %20 | 2.955 | %78 | 76 | %2 | 3.788 | %100 |
| Don't know | 121 | %16 | 604 | %80 | 33 | %4 | 758 | %100 |
| TOTAL | 2.083 | %20 | 8.194 | %78 | 209 | %2 | 10.486 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 30,7 ; dof= 4.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: …invisible symptoms such as pain, dizziness, headaches, fatigue, etc. / Genetic tests

| …INVISIBLE SYMPTOMS SUCH AS PAIN, DIZZINESS, HEADACHES, FATIGUE, ETC. | GENETIC TESTS | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 461 | %15 | 2.587 | %82 | 92 | %3 | **3.140** | **%100** |
| No | 313 | %16 | 1.592 | %82 | 40 | %2 | **1.945** | **%100** |
| Don't know | 57 | %14 | 336 | %83 | 11 | %3 | **404** | **%100** |
| **TOTAL** | **831** | **%15** | **4.515** | **%82** | **143** | **%3** | **5.489** | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is not significant. p-value= 0,2 ; Chi2= 5,5 ; dof= 4.*

## Cross: …invisible symptoms such as pain, dizziness, headaches, fatigue, etc. / Other diagnostic tests such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc.

| …INVISIBLE SYMPTOMS SUCH AS PAIN, DIZZINESS, HEADACHES, FATIGUE, ETC. | OTHER DIAGNOSTIC TESTS SUCH AS CLINICAL EXAMINATION(S), MEDICAL IMAGING (MRI, SCANS…), BIOPSY, BIOCHEMICAL TEST(S) (BLOOD OR URINE TESTS…), ETC. | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 994 | %15 | 5.412 | %83 | 114 | %2 | **6.520** | **%100** |
| No | 344 | %14 | 2.110 | %84 | 45 | %2 | **2.499** | **%100** |
| Don't know | 65 | %14 | 386 | %84 | 11 | %2 | **462** | **%100** |
| **TOTAL** | **1.403** | **%15** | **7.908** | **%83** | **170** | **%2** | **9.481** | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is not significant. p-value= 0,4 ; Chi2= 4,3 ; dof= 4.*

## Cross: …invisible symptoms such as pain, dizziness, headaches, fatigue, etc. / Additional advice from a healthcare professional specialised in the rare disease (in person or virtually)

| …INVISIBLE SYMPTOMS SUCH AS PAIN, DIZZINESS, HEADACHES, FATIGUE, ETC. | ADDITIONAL ADVICE FROM A HEALTHCARE PROFESSIONAL SPECIALISED IN THE RARE DISEASE (IN PERSON OR VIRTUALLY) | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 1.380 | %20 | 5.505 | %78 | 135 | %2 | **7.020** | **%100** |
| No | 593 | %20 | 2.266 | %78 | 57 | %2 | **2.916** | **%100** |
| Don't know | 110 | %20 | 423 | %77 | 17 | %3 | **550** | **%100** |
| **TOTAL** | **2.083** | **%20** | **8.194** | **%78** | **209** | **%2** | **10.486** | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is not significant. p-value= 0,4 ; Chi2= 4,2 ; dof= 4.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: …sudden onset symptoms requiring urgent care / Genetic tests

| …SUDDEN ONSET SYMPTOMS REQUIRING URGENT CARE | GENETIC TESTS | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 418 | %17 | 1.907 | %80 | 72 | %3 | 2.397 | %100 |
| No | 387 | %14 | 2.372 | %84 | 57 | %2 | 2.816 | %100 |
| Don't know | 26 | %9 | 236 | %86 | 14 | %5 | 276 | %100 |
| TOTAL | 831 | %15 | 4.515 | %82 | 143 | %3 | 5.489 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 33,3 ; dof= 4.*

## Cross: …sudden onset symptoms requiring urgent care / Other diagnostic tests such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc.

| …SUDDEN ONSET SYMPTOMS REQUIRING URGENT CARE | OTHER DIAGNOSTIC TESTS SUCH AS CLINICAL EXAMINATION(S), MEDICAL IMAGING (MRI, SCANS…), BIOPSY, BIOCHEMICAL TEST(S) (BLOOD OR URINE TESTS…), ETC. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 732 | %17 | 3.508 | %81 | 73 | %2 | 4.313 | %100 |
| No | 610 | %13 | 3.981 | %85 | 77 | %2 | 4.668 | %100 |
| Don't know | 61 | %12 | 419 | %84 | 20 | %4 | 500 | %100 |
| TOTAL | 1.403 | %15 | 7.908 | %83 | 170 | %2 | 9.481 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 44,1 ; dof= 4.*

## Cross: …sudden onset symptoms requiring urgent care / Additional advice from a healthcare professional specialised in the rare disease (in person or virtually)

| …SUDDEN ONSET SYMPTOMS REQUIRING URGENT CARE | ADDITIONAL ADVICE FROM A HEALTHCARE PROFESSIONAL SPECIALISED IN THE RARE DISEASE (IN PERSON OR VIRTUALLY) | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 1.052 | %23 | 3.514 | %76 | 82 | %2 | 4.648 | %100 |
| No | 932 | %18 | 4.221 | %80 | 98 | %2 | 5.251 | %100 |
| Don't know | 99 | %17 | 459 | %78 | 29 | %5 | 587 | %100 |
| TOTAL | 2.083 | %20 | 8.194 | %78 | 209 | %2 | 10.486 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 66,9 ; dof= 4.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis? / Genetic tests**

| HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | GENETIC TESTS | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 0-1 | 70 | %9 | 664 | %88 | 22 | %3 | 756 | %100 |
| between 2 and 4 | 303 | %13 | 1.935 | %84 | 56 | %2 | 2.294 | %100 |
| between 5 and 7 | 158 | %15 | 870 | %82 | 31 | %3 | 1.059 | %100 |
| between 8 and 10 | 76 | %19 | 312 | %79 | 9 | %2 | 397 | %100 |
| more than 10 | 224 | %23 | 734 | %75 | 25 | %3 | 983 | %100 |
| TOTAL | 831 | %15 | 4.515 | %82 | 143 | %3 | 5.489 | |

■ *Under-represented elements*　■ *Over-represented elements*

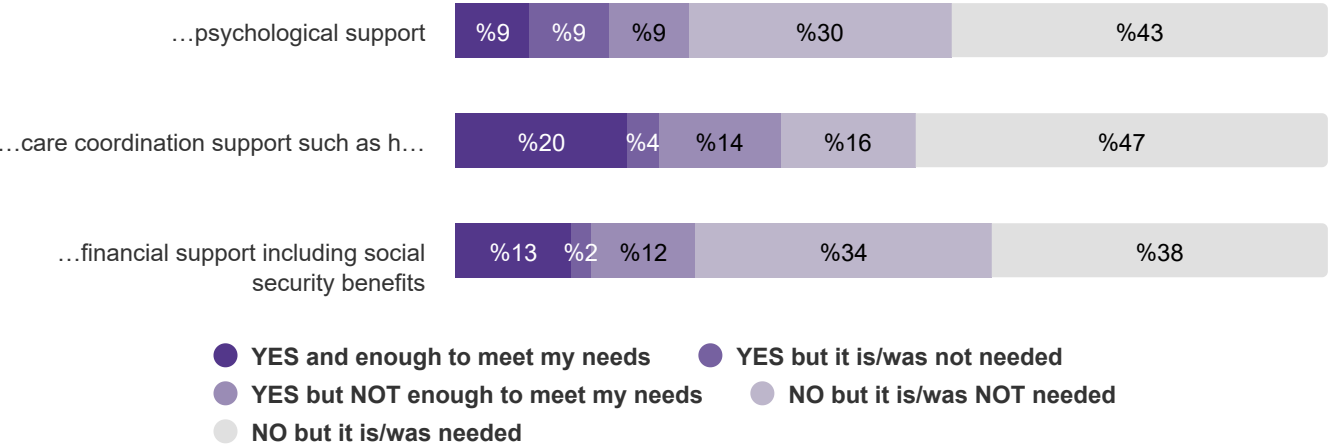*The relationship is very significant. p-value= < 0,01 ; Chi2= 77,9 ; dof= 8.*

**Cross: How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis? / Other diagnostic tests such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc.**

| HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | OTHER DIAGNOSTIC TESTS SUCH AS CLINICAL EXAMINATION(S), MEDICAL IMAGING (MRI, SCANS…), BIOPSY, BIOCHEMICAL TEST(S) (BLOOD OR URINE TESTS…), ETC. | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 0-1 | 121 | %11 | 939 | %86 | 27 | %2 | 1.087 | %100 |
| between 2 and 4 | 526 | %13 | 3.535 | %86 | 65 | %2 | 4.126 | %100 |
| between 5 and 7 | 257 | %14 | 1.582 | %84 | 39 | %2 | 1.878 | %100 |
| between 8 and 10 | 140 | %19 | 579 | %79 | 18 | %2 | 737 | %100 |
| more than 10 | 359 | %22 | 1.273 | %77 | 21 | %1 | 1.653 | %100 |
| TOTAL | 1.403 | %15 | 7.908 | %83 | 170 | %2 | 9.481 | |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 108,5 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis? / Additional advice from a healthcare professional specialised in the rare disease (in person or virtually)

| HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | ADDITIONAL ADVICE FROM A HEALTHCARE PROFESSIONAL SPECIALISED IN THE RARE DISEASE (IN PERSON OR VIRTUALLY) | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| 0-1 | **185** | **%14** | **1.083** | **%83** | **37** | **%3** | **1.305** | %100 |
| between 2 and 4 | **765** | **%17** | **3.728** | **%82** | **76** | **%2** | **4.569** | %100 |
| between 5 and 7 | 430 | %21 | 1.560 | %77 | 43 | %2 | **2.033** | %100 |
| between 8 and 10 | **210** | **%27** | **563** | **%71** | 18 | %2 | **791** | %100 |
| more than 10 | **493** | **%28** | **1.260** | **%70** | 35 | %2 | **1.788** | %100 |
| **TOTAL** | **2.083** | %20 | **8.194** | %78 | **209** | %2 | **10.486** | |

🟣 *Under-represented elements*   🔵 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 154,0 ; dof= 8.*

## Cross: I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases / Genetic tests

| I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | GENETIC TESTS | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | **490** | **%14** | **2.871** | **%83** | 96 | %3 | **3.457** | %100 |
| No | **337** | **%17** | **1.615** | **%81** | 46 | %2 | **1.998** | %100 |
| **TOTAL** | **827** | %15 | **4.486** | %82 | **142** | %3 | **5.455** | |

🟣 *Under-represented elements*   🔵 *Over-represented elements*

*The relationship is significant. p-value= 0,0 ; Chi2= 7,9 ; dof= 2.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases / Other diagnostic tests such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc.**

| I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | OTHER DIAGNOSTIC TESTS SUCH AS CLINICAL EXAMINATION(S), MEDICAL IMAGING (MRI, SCANS…), BIOPSY, BIOCHEMICAL TEST(S) (BLOOD OR URINE TESTS…), ETC. | | | | | | | |
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 754 | %14 | 4.652 | %84 | 107 | %2 | 5.513 | %100 |
| No | 644 | %16 | 3.199 | %82 | 62 | %2 | 3.905 | %100 |
| TOTAL | 1.398 | %15 | 7.851 | %83 | 169 | %2 | 9.418 | |

🟥 *Under-represented elements* 🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 15,5 ; dof= 2.*

**Cross: I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases / Additional advice from a healthcare professional specialised in the rare disease (in person or virtually)**

| I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | ADDITIONAL ADVICE FROM A HEALTHCARE PROFESSIONAL SPECIALISED IN THE RARE DISEASE (IN PERSON OR VIRTUALLY) | | | | | | | |
| | YES | | NO | | DON'T KNOW | | TOTAL | |
| | N | % | N | % | N | % | N | % |
| Yes | 1.150 | %19 | 4.720 | %79 | 128 | %2 | 5.998 | %100 |
| No | 920 | %21 | 3.417 | %77 | 78 | %2 | 4.415 | %100 |
| TOTAL | 2.070 | %20 | 8.137 | %78 | 206 | %2 | 10.413 | |

🟥 *Under-represented elements* 🟦 *Over-represented elements*

*The relationship is weakly significant. p-value= 0,1 ; Chi2= 5,8 ; dof= 2.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

**Cross: Genetic tests / After the tests were performed, were you offered genetic counselling (e.g. given information about how your genetic condition might affect you and your family)?**

| GENETIC TESTS | AFTER THE TESTS WERE PERFORMED, WERE YOU OFFERED GENETIC COUNSELLING (E.G. GIVEN INFORMATION ABOUT HOW YOUR GENETIC CONDITION MIGHT AFFECT YOU AND YOUR FAMILY)? | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| | YES, WITH A GENETIC COUNSELLOR OR CLINICAL GENETICIST | | YES, BY A HEALTHCARE PROFESSIONAL | | NO, I WASN'T OFFERED GENETIC COUNSELLING | | NOT SURE / DON'T REMEMBER | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % |
| Yes | 338 | %41 | 181 | %22 | 268 | %32 | 44 | %5 | 831 | %100 |
| No | 1.753 | %39 | 974 | %22 | 1.455 | %32 | 333 | %7 | 4.515 | %100 |
| Don't know | 46 | %32 | 23 | %16 | 47 | %33 | 27 | %19 | 143 | %100 |
| **TOTAL** | **2.137** | **%39** | **1.178** | **%21** | **1.770** | **%32** | **404** | **%7** | **5.489** | |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 35,0 ; dof= 6.*

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RARE DISEASES EUROPE

# Chapter 13.

## Support

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## During your search for a diagnosis, were you offered…

| | YES AND ENOUGH TO MEET MY NEEDS | YES BUT IT IS/WAS NOT NEEDED | YES BUT NOT ENOUGH TO MEET MY NEEDS | NO BUT IT IS/WAS NOT NEEDED | NO BUT IT IS/WAS NEEDED | TOTAL |
|---|---|---|---|---|---|---|
| …psychological support | 922 | 955 | 952 | 3.165 | 4.492 | **10.486** |
| …care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc. | 2.083 | 391 | 1.463 | 1.627 | 4.922 | **10.486** |
| …financial support including social security benefits | 1.405 | 243 | 1.232 | 3.544 | 3.989 | **10.413** |

**Needs met:** YES and enough to meet my needs + YES but it is/was not needed + NO but it is/was NOT needed.

**Needs unmet:** YES but NOT enough to meet my needs + NO but it is/was needed

## During your search for a diagnosis, were you offered…

| | | |
|---|---|---|
| …psychological support | %9 / %9 / %9 / %30 | %43 |
| …care coordination support such as h… | %20 / %4 / %14 / %16 | %47 |
| …financial support including social security benefits | %13 / %2 / %12 / %34 | %38 |

- ● YES and enough to meet my needs
- ● YES but it is/was not needed
- ● YES but NOT enough to meet my needs
- ● NO but it is/was NOT needed
- ● NO but it is/was needed

## During your search for a diagnosis, were you offered…

| | | |
|---|---|---|
| …psychological support | %48 | %52 |
| …care coordination support such as h… | %39 | %61 |
| …financial support including social security benefits | %50 | %50 |

- ● Needs met
- ● Needs unmet

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*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Multiple Cross

| …psychological support | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| YES and enough to meet my needs | -0,3 | 685 | 2,1 | 670 | 2,0 | 471 | 2,0 | 695 | 2,5 | 584 |
| YES but it is/was not needed | 0,3 | 688 | 3,1 | 664 | 3,6 | 461 | 3,4 | 717 | 4,0 | 615 |
| YES but NOT enough to meet my needs | 0,5 | 739 | 3,5 | 674 | 4,4 | 425 | 4,5 | 724 | 5,1 | 555 |
| NO but it is/was NOT needed | 0,8 | 2.255 | 3,9 | 2.166 | 3,9 | 1.280 | 3,1 | 2.330 | 4,9 | 1.965 |
| NO but it is/was needed | 0,5 | 3.453 | 3,6 | 3.148 | 4,3 | 1.698 | 4,1 | 3.377 | 5,2 | 2.788 |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Fisher= 3,5.*

*Inter variance= 159,1. Intra variance= 45,9.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

## Multiple Cross

| …care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc. | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| YES and enough to meet my needs | 0,4 | 1.551 | 2,3 | 1.535 | 2,4 | 1.117 | 1,7 | 1.619 | 2,8 | 1.425 |
| YES but it is/was not needed | 0,5 | 258 | 2,2 | 248 | 2,0 | 167 | 2,2 | 268 | 3,2 | 235 |
| YES but NOT enough to meet my needs | 0,5 | 1.109 | 3,4 | 1.028 | 3,6 | 678 | 3,7 | 1.100 | 4,7 | 921 |
| NO but it is/was NOT needed | 0,5 | 1.109 | 3,5 | 1.068 | 3,7 | 646 | 2,7 | 1.149 | 4,4 | 978 |
| NO but it is/was needed | 0,5 | 3.793 | 4,2 | 3.443 | 5,2 | 1.727 | 4,8 | 3.707 | 5,9 | 2.948 |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is not significant. p-value= 1,0 ; Fisher= 0,1.*

*Inter variance= 3,4. Intra variance= 46,0.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

## Multiple Cross

| …financial support including social security benefits | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| YES and enough to meet my needs | 0,4 | 1.050 | **2,6** | 1.012 | **2,7** | 701 | **2,6** | 1.084 | **3,2** | 932 |
| YES but it is/was not needed | 0,7 | 162 | 2,2 | 171 | 3,0 | 113 | **1,8** | 166 | **2,9** | 152 |
| YES but NOT enough to meet my needs | 0,3 | 903 | 3,2 | 815 | 3,8 | 505 | 3,9 | 869 | 4,6 | 717 |
| NO but it is/was NOT needed | 0,3 | 2.686 | 3,6 | 2.589 | 3,5 | 1.576 | **2,9** | 2.756 | 4,5 | 2.375 |
| NO but it is/was needed | 0,7 | 2.966 | **4,0** | 2.684 | **4,9** | 1.440 | **4,7** | 2.915 | **5,8** | 2.293 |

🟥 *Under-represented elements*      🟦 *Over-represented elements*

*The relationship is not significant. p-value= 0,2 ; Fisher= 1,5.*

*Inter variance= 67,9. Intra variance= 46,2.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Gender of the person affected by the rare disease / …psychological support

| GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE | …PSYCHOLOGICAL SUPPORT | | | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Female | 536 | %8 | 546 | %8 | 639 | %10 | 1.973 | %30 | 2.965 | %45 | 6.659 | %100 |
| Male | 280 | %10 | 301 | %11 | 226 | %8 | 921 | %33 | 1.082 | %39 | 2.810 | %100 |
| Other | 7 | %7 | 15 | %15 | 9 | %9 | 19 | %19 | 51 | %50 | 101 | %100 |
| TOTAL | 823 | %9 | 862 | %9 | 874 | %9 | 2.913 | %30 | 4.098 | %43 | 9.570 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 60,8 ; dof= 8.*

## Cross: Gender of the person affected by the rare disease / …care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc.

| GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE | …CARE COORDINATION SUPPORT SUCH AS HELP TO FIND THE NECESSARY INFORMATION ON THE DISEASE AND THE RIGHT PROFESSIONALS, ARRANGING APPOINTMENTS WITH DIFFERENT HEALTH PROVIDERS, ETC. | | | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Female | 1.165 | %17 | 201 | %3 | 902 | %14 | 1.040 | %16 | 3.351 | %50 | 6.659 | %100 |
| Male | 689 | %25 | 127 | %5 | 424 | %15 | 427 | %15 | 1.143 | %41 | 2.810 | %100 |
| Other | 19 | %19 | 4 | %4 | 10 | %10 | 14 | %14 | 54 | %53 | 101 | %100 |
| TOTAL | 1.873 | %20 | 332 | %3 | 1.336 | %14 | 1.481 | %15 | 4.548 | %48 | 9.570 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 107,2 ; dof= 8.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

**Cross: Gender of the person affected by the rare disease / …financial support including social security benefits**

| GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE | ...FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Female | 818 | %12 | 120 | %2 | 728 | %11 | 2.302 | %35 | 2.665 | %40 | 6.633 | %100 |
| Male | 421 | %15 | 89 | %3 | 368 | %13 | 985 | %35 | 935 | %33 | 2.798 | %100 |
| Other | 13 | %13 | 5 | %5 | 17 | %17 | 18 | %18 | 48 | %48 | 101 | %100 |
| TOTAL | 1.252 | %13 | 214 | %2 | 1.113 | %12 | 3.305 | %35 | 3.648 | %38 | 9.532 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 75,5 ; dof= 8.*

**Cross: How old were you when you stopped full-time education? / …psychological support**

| HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION? | ...PSYCHOLOGICAL SUPPORT | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 15 y.o. or under | 45 | %10 | 46 | %10 | 44 | %10 | 130 | %29 | 190 | %42 | 455 | %100 |
| between 16 and 19 y.o. | 228 | %9 | 225 | %9 | 202 | %8 | 761 | %31 | 1.048 | %43 | 2.464 | %100 |
| between 20 and 23 y.o. | 263 | %9 | 275 | %9 | 257 | %9 | 977 | %32 | 1.250 | %41 | 3.022 | %100 |
| 24 y.o. or above | 247 | %8 | 273 | %9 | 325 | %10 | 900 | %29 | 1.400 | %45 | 3.145 | %100 |
| TOTAL | 783 | %9 | 819 | %9 | 828 | %9 | 2.768 | %30 | 3.888 | %43 | 9.086 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is significant. p-value= 0,0 ; Chi2= 25,4 ; dof= 12.*

**Cross: How old were you when you stopped full-time education? / …care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc.**

| HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION? | …CARE COORDINATION SUPPORT SUCH AS HELP TO FIND THE NECESSARY INFORMATION ON THE DISEASE AND THE RIGHT PROFESSIONALS, ARRANGING APPOINTMENTS WITH DIFFERENT HEALTH PROVIDERS, ETC. | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 15 y.o. or under | 91 | %20 | 20 | %4 | 89 | %20 | 57 | %13 | 198 | %44 | 455 | %100 |
| between 16 and 19 y.o. | 498 | %20 | 96 | %4 | 333 | %14 | 357 | %14 | 1.180 | %48 | 2.464 | %100 |
| between 20 and 23 y.o. | 620 | %21 | 96 | %3 | 409 | %14 | 497 | %16 | 1.400 | %46 | 3.022 | %100 |
| 24 y.o. or above | 574 | %18 | 104 | %3 | 442 | %14 | 474 | %15 | 1.551 | %49 | 3.145 | %100 |
| TOTAL | 1.783 | %20 | 316 | %3 | 1.273 | %14 | 1.385 | %15 | 4.329 | %48 | 9.086 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 29,7 ; dof= 12.*

**Cross: How old were you when you stopped full-time education? / …financial support including social security benefits**

| HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION? | …FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 15 y.o. or under | 63 | %14 | 11 | %2 | 81 | %18 | 118 | %26 | 178 | %39 | 451 | %100 |
| between 16 and 19 y.o. | 299 | %12 | 43 | %2 | 284 | %12 | 831 | %34 | 1.003 | %41 | 2.460 | %100 |
| between 20 and 23 y.o. | 398 | %13 | 75 | %2 | 349 | %12 | 1.103 | %37 | 1.079 | %36 | 3.004 | %100 |
| 24 y.o. or above | 429 | %14 | 76 | %2 | 360 | %11 | 1.077 | %34 | 1.192 | %38 | 3.134 | %100 |
| TOTAL | 1.189 | %13 | 205 | %2 | 1.074 | %12 | 3.129 | %35 | 3.452 | %38 | 9.049 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 44,0 ; dof= 12.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: How would you best describe yourself? / …psychological support

| | …PSYCHOLOGICAL SUPPORT | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| HOW WOULD YOU BEST DESCRIBE YOURSELF? | N | % | N | % | N | % | N | % | N | % | N | % |
| I belong to the ethnic majority in the country where I live | **587** | **%8** | 617 | %9 | 658 | %9 | **2.185** | **%31** | 3.078 | %43 | **7.125** | **%100** |
| I am part of an ethnic minority in the country where I live | 47 | %10 | **55** | **%12** | 54 | %12 | 123 | %26 | 186 | %40 | **465** | **%100** |
| Other, specify... | **42** | **%12** | 27 | %8 | 27 | %8 | 90 | %27 | 151 | %45 | **337** | **%100** |
| **TOTAL** | **676** | **%9** | **699** | **%9** | **739** | **%9** | **2.398** | **%30** | **3.415** | **%43** | **7.927** | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 22,0 ; dof= 8.*

## Cross: How would you best describe yourself? / …care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc.

| | …CARE COORDINATION SUPPORT SUCH AS HELP TO FIND THE NECESSARY INFORMATION ON THE DISEASE AND THE RIGHT PROFESSIONALS, ARRANGING APPOINTMENTS WITH DIFFERENT HEALTH PROVIDERS, ETC. | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| HOW WOULD YOU BEST DESCRIBE YOURSELF? | N | % | N | % | N | % | N | % | N | % | N | % |
| I belong to the ethnic majority in the country where I live | 1.378 | %19 | 239 | %3 | 1.015 | %14 | 1.049 | %15 | 3.444 | %48 | **7.125** | **%100** |
| I am part of an ethnic minority in the country where I live | 96 | %21 | 18 | %4 | 68 | %15 | 62 | %13 | 221 | %48 | **465** | **%100** |
| Other, specify... | 65 | %19 | 14 | %4 | 49 | %15 | 47 | %14 | 162 | %48 | **337** | **%100** |
| **TOTAL** | **1.539** | **%19** | **271** | **%3** | **1.132** | **%14** | **1.158** | **%15** | **3.827** | **%48** | **7.927** | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is not significant. p-value= 1,0 ; Chi2= 2,1 ; dof= 8.*

## Cross: How would you best describe yourself? / …financial support including social security benefits

| HOW WOULD YOU BEST DESCRIBE YOURSELF? | ...FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS | | | | | | | | | | TOTAL | |
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| I belong to the ethnic majority in the country where I live | 898 | %13 | 155 | %2 | 857 | %12 | 2.494 | %35 | 2.685 | %38 | 7.089 | %100 |
| I am part of an ethnic minority in the country where I live | 60 | %13 | 14 | %3 | 68 | %15 | 118 | %25 | 204 | %44 | 464 | %100 |
| Other, specify... | 45 | %13 | 8 | %2 | 42 | %12 | 96 | %28 | 146 | %43 | 337 | %100 |
| **TOTAL** | **1.003** | **%13** | **177** | **%2** | **967** | **%12** | **2.708** | **%34** | **3.035** | **%38** | **7.890** | |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 25,8 ; dof= 8.*

## Cross: Typology of countries based on size and welfare / …psychological support

| TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE | ...PSYCHOLOGICAL SUPPORT | | | | | | | | | | TOTAL | |
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| Group A ('Eastern Europe') | 170 | %9 | 180 | %10 | 144 | %8 | 481 | %27 | 819 | %46 | 1.794 | %100 |
| Group B ('Western Europe') | 399 | %8 | 426 | %8 | 429 | %8 | 1.501 | %29 | 2.350 | %46 | 5.105 | %100 |
| Group C ('Northern Europe') | 313 | %10 | 306 | %9 | 352 | %11 | 1.097 | %34 | 1.205 | %37 | 3.273 | %100 |
| **TOTAL** | **882** | **%9** | **912** | **%9** | **925** | **%9** | **3.079** | **%30** | **4.374** | **%43** | **10.172** | |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 91,2 ; dof= 8.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

## Cross: Typology of countries based on size and welfare / …care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc.

| TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE | …CARE COORDINATION SUPPORT SUCH AS HELP TO FIND THE NECESSARY INFORMATION ON THE DISEASE AND THE RIGHT PROFESSIONALS, ARRANGING APPOINTMENTS WITH DIFFERENT HEALTH PROVIDERS, ETC. | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Group A ('Eastern Europe') | 352 | %20 | 88 | %5 | 277 | %15 | 234 | %13 | 843 | %47 | 1.794 | %100 |
| Group B ('Western Europe') | 1.026 | %20 | 160 | %3 | 740 | %14 | 763 | %15 | 2.416 | %47 | 5.105 | %100 |
| Group C ('Northern Europe') | 633 | %19 | 119 | %4 | 416 | %13 | 573 | %18 | 1.532 | %47 | 3.273 | %100 |
| TOTAL | 2.011 | %20 | 367 | %4 | 1.433 | %14 | 1.570 | %15 | 4.791 | %47 | 10.172 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 36,2 ; dof= 8.*

## Cross: Typology of countries based on size and welfare / …financial support including social security benefits

| TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE | …FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Group A ('Eastern Europe') | 237 | %13 | 47 | %3 | 326 | %18 | 336 | %19 | 847 | %47 | 1.793 | %100 |
| Group B ('Western Europe') | 627 | %12 | 107 | %2 | 526 | %10 | 1.873 | %37 | 1.937 | %38 | 5.070 | %100 |
| Group C ('Northern Europe') | 496 | %15 | 81 | %2 | 361 | %11 | 1.228 | %38 | 1.106 | %34 | 3.272 | %100 |
| TOTAL | 1.360 | %13 | 235 | %2 | 1.213 | %12 | 3.437 | %34 | 3.890 | %38 | 10.135 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 287,3 ; dof= 8.*

## Cross: Orphacode associated nomenclature (english) / …psychological support

| ORPHACODE ASSOCIATED NOMENCLATURE (ENGLISH) | …PSYCHOLOGICAL SUPPORT | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Hereditary hemorrhagic telangiectasia | **20** | **%4** | **28** | **%6** | **20** | **%4** | 250 | %55 | **140** | **%31** | 458 | %100 |
| Hypermobile Ehlers-Danlos syndrome | 28 | %9 | **40** | **%13** | **52** | **%16** | **69** | **%22** | 128 | %40 | 317 | %100 |
| Sarcoidosis | 11 | %6 | 10 | %6 | **5** | **%3** | **71** | **%42** | 73 | %43 | 170 | %100 |
| Classical Ehlers-Danlos syndrome | 7 | %5 | 16 | %12 | **21** | **%15** | 36 | %26 | 57 | %42 | 137 | %100 |
| Williams syndrome | 15 | %11 | 7 | %5 | 13 | %10 | **28** | **%21** | **73** | **%54** | 136 | %100 |
| Cystic fibrosis | 16 | %13 | **21** | **%16** | 14 | %11 | **25** | **%20** | 52 | %41 | 128 | %100 |
| Myasthenia gravis | 10 | %8 | **3** | **%3** | 6 | %5 | 40 | %33 | 61 | %51 | 120 | %100 |
| Systemic sclerosis | 6 | %6 | 8 | %7 | 7 | %7 | **43** | **%40** | 43 | %40 | 107 | %100 |
| Tuberous sclerosis complex | 13 | %13 | 9 | %9 | 7 | %7 | 26 | %27 | 43 | %44 | 98 | %100 |
| Neurofibromatosis type 1 | 9 | %10 | 8 | %9 | 9 | %10 | 30 | %33 | 36 | %39 | 92 | %100 |
| Interstitial cystitis | 6 | %8 | 6 | %8 | 2 | %3 | 15 | %20 | **45** | **%61** | 74 | %100 |
| Addison disease | 3 | %4 | 11 | %15 | **1** | **%1** | 25 | %34 | 33 | %45 | 73 | %100 |
| 22q11.2 deletion syndrome | 8 | %12 | 3 | %4 | 7 | %10 | 14 | %21 | 36 | %53 | 68 | %100 |
| Chronic inflammatory demyelinating polyneuropathy | 8 | %12 | 8 | %12 | 10 | %15 | 21 | %32 | **18** | **%28** | 65 | %100 |
| Perineural cyst | 7 | %11 | 9 | %14 | 5 | %8 | 16 | %25 | 26 | %41 | 63 | %100 |
| Acute inflammatory demyelinating polyradiculoneuropathy | 6 | %10 | **11** | **%18** | 6 | %10 | 17 | %27 | 22 | %35 | 62 | %100 |
| Rett syndrome | 4 | %7 | 6 | %10 | 4 | %7 | 14 | %23 | 32 | %53 | 60 | %100 |
| Marfan syndrome | 2 | %4 | 4 | %8 | 7 | %13 | 16 | %31 | 23 | %44 | 52 | %100 |
| Fragile X syndrome | 4 | %8 | 4 | %8 | 6 | %12 | 16 | %33 | 19 | %39 | 49 | %100 |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 7.481,1 ; dof= 6.700.*

**Cross: Orphacode associated nomenclature (english) / …care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc.**

| ORPHACODE ASSOCIATED NOMENCLATURE (ENGLISH) | …CARE COORDINATION SUPPORT SUCH AS HELP TO FIND THE NECESSARY INFORMATION ON THE DISEASE AND THE RIGHT PROFESSIONALS, ARRANGING APPOINTMENTS WITH DIFFERENT HEALTH PROVIDERS, ETC. | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Hereditary hemorrhagic telangiectasia | 133 | %29 | 10 | %2 | 53 | %12 | 117 | %26 | 145 | %32 | 458 | %100 |
| Hypermobile Ehlers-Danlos syndrome | 18 | %6 | 3 | %1 | 27 | %9 | 32 | %10 | 237 | %75 | 317 | %100 |
| Sarcoidosis | 39 | %23 | 9 | %5 | 15 | %9 | 30 | %18 | 77 | %45 | 170 | %100 |
| Classical Ehlers-Danlos syndrome | 8 | %6 | 4 | %3 | 15 | %11 | 22 | %16 | 88 | %64 | 137 | %100 |
| Williams syndrome | 36 | %26 | 5 | %4 | 22 | %16 | 13 | %10 | 60 | %44 | 136 | %100 |
| Cystic fibrosis | 50 | %39 | 8 | %6 | 17 | %13 | 15 | %12 | 38 | %30 | 128 | %100 |
| Myasthenia gravis | 18 | %15 | 4 | %3 | 20 | %17 | 19 | %16 | 59 | %49 | 120 | %100 |
| Systemic sclerosis | 33 | %31 | 9 | %8 | 20 | %19 | 8 | %7 | 37 | %35 | 107 | %100 |
| Tuberous sclerosis complex | 30 | %31 | 5 | %5 | 18 | %18 | 16 | %16 | 29 | %30 | 98 | %100 |
| Neurofibromatosis type 1 | 25 | %27 | 6 | %7 | 10 | %11 | 18 | %20 | 33 | %36 | 92 | %100 |
| Interstitial cystitis | 18 | %24 | 2 | %3 | 10 | %14 | 8 | %11 | 36 | %49 | 74 | %100 |
| Addison disease | 15 | %21 | 3 | %4 | 11 | %15 | 8 | %11 | 36 | %49 | 73 | %100 |
| 22q11.2 deletion syndrome | 13 | %19 | 3 | %4 | 18 | %26 | 9 | %13 | 25 | %37 | 68 | %100 |
| Chronic inflammatory demyelinating polyneuropathy | 16 | %25 | 1 | %2 | 7 | %11 | 13 | %20 | 28 | %43 | 65 | %100 |
| Perineural cyst | 4 | %6 | 4 | %6 | 4 | %6 | 5 | %8 | 46 | %73 | 63 | %100 |
| Acute inflammatory demyelinating polyradiculoneuropathy | 21 | %34 | 0 | %0 | 13 | %21 | 12 | %19 | 16 | %26 | 62 | %100 |
| Rett syndrome | 10 | %17 | 2 | %3 | 9 | %15 | 8 | %13 | 31 | %52 | 60 | %100 |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 7.163,3 ; dof= 6.700.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Orphacode associated nomenclature (english) / …financial support including social security benefits

| ORPHACODE ASSOCIATED NOMENCLATURE (ENGLISH) | …FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS | | | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Hereditary hemorrhagic telangiectasia | 69 | %15 | 9 | %2 | 19 | %4 | 235 | %52 | 123 | %27 | 455 | %100 |
| Hypermobile Ehlers-Danlos syndrome | 23 | %7 | 2 | %1 | 42 | %13 | 73 | %23 | 176 | %56 | 316 | %100 |
| Sarcoidosis | 11 | %7 | 1 | %1 | 7 | %4 | 97 | %57 | 53 | %31 | 169 | %100 |
| Classical Ehlers-Danlos syndrome | 8 | %6 | 1 | %1 | 20 | %15 | 33 | %24 | 73 | %54 | 135 | %100 |
| Williams syndrome | 28 | %21 | 0 | %0 | 17 | %13 | 39 | %29 | 52 | %38 | 136 | %100 |
| Cystic fibrosis | 38 | %30 | 3 | %2 | 26 | %20 | 20 | %16 | 41 | %32 | 128 | %100 |
| Myasthenia gravis | 12 | %10 | 1 | %1 | 20 | %17 | 34 | %28 | 53 | %44 | 120 | %100 |
| Systemic sclerosis | 13 | %12 | 8 | %7 | 7 | %7 | 54 | %50 | 25 | %23 | 107 | %100 |
| Tuberous sclerosis complex | 12 | %12 | 3 | %3 | 14 | %14 | 38 | %39 | 30 | %31 | 97 | %100 |
| Neurofibromatosis type 1 | 12 | %13 | 1 | %1 | 9 | %10 | 37 | %40 | 33 | %36 | 92 | %100 |
| Interstitial cystitis | 6 | %8 | 0 | %0 | 6 | %8 | 18 | %24 | 44 | %59 | 74 | %100 |
| Addison disease | 4 | %5 | 0 | %0 | 8 | %11 | 39 | %53 | 22 | %30 | 73 | %100 |
| 22q11.2 deletion syndrome | 14 | %21 | 3 | %4 | 14 | %21 | 12 | %18 | 25 | %37 | 68 | %100 |
| Chronic inflammatory demyelinating polyneuropathy | 9 | %14 | 1 | %2 | 5 | %8 | 26 | %40 | 24 | %37 | 65 | %100 |
| Perineural cyst | 6 | %10 | 1 | %2 | 9 | %14 | 16 | %25 | 31 | %49 | 63 | %100 |
| Acute inflammatory demyelinating polyradiculoneuropathy | 7 | %11 | 3 | %5 | 6 | %10 | 28 | %45 | 18 | %29 | 62 | %100 |
| Rett syndrome | 9 | %15 | 0 | %0 | 13 | %22 | 11 | %18 | 27 | %45 | 60 | %100 |
| Marfan syndrome | 2 | %4 | 2 | %4 | 7 | %15 | 18 | %38 | 19 | %40 | 48 | %100 |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 7.793,7 ; dof= 6.688.*

Rare Barometer
A EURORDIS ✕ INITIATIVE

EURORDIS
RARE DISEASES EUROPE

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Orphanet classification of rare diseases (one disease can be classified in several categories) / …psychological support**

| ORPHANET CLASSIFICATION OF RARE DISEASES (ONE DISEASE CAN BE CLASSIFIED IN SEVERAL CATEGORIES) | …PSYCHOLOGICAL SUPPORT | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Abdominal surgical diseases | 25 | %10 | 27 | %11 | **35** | **%15** | **55** | **%23** | 97 | %41 | 239 | %100 |
| Allergic diseases | 0 | %0 | 0 | %0 | 0 | %0 | 1 | %33 | 2 | %67 | 3 | %100 |
| Bone diseases | 62 | %8 | 66 | %8 | 76 | %10 | 231 | %29 | 364 | %46 | 799 | %100 |
| Cardiac diseases | 49 | %7 | 57 | %9 | 55 | %8 | **229** | **%35** | 270 | %41 | 660 | %100 |
| Cardiac malformations | **36** | **%12** | **17** | **%6** | 32 | %11 | **64** | **%22** | **146** | **%49** | 295 | %100 |
| Circulatory system diseases | **87** | **%6** | **89** | **%7** | **92** | **%7** | **533** | **%39** | **550** | **%41** | 1.351 | %100 |
| Clinical sign | 0 | %0 | 0 | %0 | 0 | %0 | 0 | %0 | 0 | %0 | 0 | %100 |
| Developmental anomalies during embryogenesis | **250** | **%7** | **269** | **%8** | **318** | **%10** | 1.041 | %31 | 1.469 | %44 | 3.347 | %100 |
| Diseases due to toxic effects | 0 | %0 | 0 | %0 | 0 | %0 | 1 | %33 | 2 | %67 | 3 | %100 |
| Endocrine diseases | 75 | %8 | 89 | %9 | 82 | %8 | 303 | %30 | 446 | %45 | 995 | %100 |
| Gastroenterological diseases | 32 | %10 | **44** | **%14** | 34 | %11 | **67** | **%22** | 128 | %42 | 305 | %100 |
| Genetic diseases | 450 | %8 | 498 | %9 | 497 | %9 | 1.637 | %30 | 2.365 | %43 | 5.447 | %100 |
| Gynecologic/obstetric diseases | 22 | %8 | 33 | %12 | 22 | %8 | **70** | **%25** | 137 | %48 | 284 | %100 |
| Hematological diseases | 40 | %10 | 36 | %9 | **48** | **%12** | 110 | %27 | 178 | %43 | 412 | %100 |
| Hepatic diseases | **58** | **%7** | 78 | %9 | **53** | **%6** | **408** | **%46** | **294** | **%33** | 891 | %100 |
| Immunological diseases | 28 | %10 | 21 | %7 | 27 | %9 | 77 | %27 | 133 | %47 | 286 | %100 |
| Inborn errors of metabolism | **79** | **%10** | 71 | %9 | 69 | %9 | 224 | %29 | 331 | %43 | 774 | %100 |
| Infectious diseases | 1 | %6 | 2 | %12 | 1 | %6 | 4 | %24 | 9 | %53 | 17 | %100 |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 478,4 ; dof= 136.*

**Cross: Orphanet classification of rare diseases (one disease can be classified in several categories) / …care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc.**

| ORPHANET CLASSIFICATION OF RARE DISEASES (ONE DISEASE CAN BE CLASSIFIED IN SEVERAL CATEGORIES) | …CARE COORDINATION SUPPORT SUCH AS HELP TO FIND THE NECESSARY INFORMATION ON THE DISEASE AND THE RIGHT PROFESSIONALS, ARRANGING APPOINTMENTS WITH DIFFERENT HEALTH PROVIDERS, ETC. | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Abdominal surgical diseases | 43 | %18 | 8 | %3 | 30 | %13 | 35 | %15 | 123 | %51 | 239 | %100 |
| Allergic diseases | 0 | %0 | 0 | %0 | 1 | %33 | 2 | %67 | 0 | %0 | 3 | %100 |
| Bone diseases | 165 | %21 | 30 | %4 | 127 | %16 | 114 | %14 | 363 | %45 | 799 | %100 |
| Cardiac diseases | 168 | %25 | 31 | %5 | 94 | %14 | 103 | %16 | 264 | %40 | 660 | %100 |
| Cardiac malformations | 71 | %24 | 11 | %4 | 51 | %17 | 37 | %13 | 125 | %42 | 295 | %100 |
| Circulatory system diseases | 315 | %23 | 44 | %3 | 194 | %14 | 256 | %19 | 542 | %40 | 1.351 | %100 |
| Clinical sign | 0 | %0 | 0 | %0 | 0 | %0 | 0 | %0 | 0 | %0 | 0 | %100 |
| Developmental anomalies during embryogenesis | 642 | %19 | 93 | %3 | 461 | %14 | 495 | %15 | 1.656 | %49 | 3.347 | %100 |
| Diseases due to toxic effects | 0 | %0 | 1 | %33 | 0 | %0 | 1 | %33 | 1 | %33 | 3 | %100 |
| Endocrine diseases | 204 | %21 | 36 | %4 | 147 | %15 | 150 | %15 | 458 | %46 | 995 | %100 |
| Gastroenterological diseases | 95 | %31 | 14 | %5 | 44 | %14 | 37 | %12 | 115 | %38 | 305 | %100 |
| Genetic diseases | 1.100 | %20 | 192 | %4 | 773 | %14 | 811 | %15 | 2.571 | %47 | 5.447 | %100 |
| Gynecologic/obstetric diseases | 67 | %24 | 11 | %4 | 32 | %11 | 39 | %14 | 135 | %48 | 284 | %100 |
| Hematological diseases | 93 | %23 | 21 | %5 | 51 | %12 | 64 | %16 | 183 | %44 | 412 | %100 |
| Hepatic diseases | 256 | %29 | 33 | %4 | 100 | %11 | 204 | %23 | 298 | %33 | 891 | %100 |
| Immunological diseases | 55 | %19 | 10 | %3 | 57 | %20 | 43 | %15 | 121 | %42 | 286 | %100 |
| Inborn errors of metabolism | 197 | %25 | 38 | %5 | 110 | %14 | 108 | %14 | 321 | %41 | 774 | %100 |
| Infectious diseases | 3 | %18 | 0 | %0 | 3 | %18 | 2 | %12 | 9 | %53 | 17 | %100 |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 611,4 ; dof= 136.*

Rare Barometer · EURORDIS RARE DISEASES EUROPE

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Orphanet classification of rare diseases (one disease can be classified in several categories) / …financial support including social security benefits

| ORPHANET CLASSIFICATION OF RARE DISEASES (ONE DISEASE CAN BE CLASSIFIED IN SEVERAL CATEGORIES) | …FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Abdominal surgical diseases | 31 | %13 | 6 | %3 | 30 | %13 | 68 | %29 | 102 | %43 | 237 | %100 |
| Allergic diseases | 1 | %33 | 0 | %0 | 0 | %0 | 2 | %67 | 0 | %0 | 3 | %100 |
| Bone diseases | 128 | %16 | 11 | %1 | 104 | %13 | 236 | %30 | 313 | %40 | 792 | %100 |
| Cardiac diseases | 89 | %14 | 18 | %3 | 50 | %8 | 280 | %42 | 222 | %34 | 659 | %100 |
| Cardiac malformations | 64 | %22 | 7 | %2 | 41 | %14 | 67 | %23 | 116 | %39 | 295 | %100 |
| Circulatory system diseases | 179 | %13 | 27 | %2 | 113 | %8 | 547 | %41 | 474 | %35 | 1.340 | %100 |
| Clinical sign | 0 | %0 | 0 | %0 | 0 | %0 | 0 | %0 | 0 | %0 | 0 | %100 |
| Developmental anomalies during embryogenesis | 459 | %14 | 64 | %2 | 410 | %12 | 1.027 | %31 | 1.366 | %41 | 3.326 | %100 |
| Diseases due to toxic effects | 0 | %0 | 0 | %0 | 0 | %0 | 1 | %33 | 2 | %67 | 3 | %100 |
| Endocrine diseases | 128 | %13 | 19 | %2 | 113 | %11 | 395 | %40 | 331 | %34 | 986 | %100 |
| Gastroenterological diseases | 59 | %19 | 8 | %3 | 52 | %17 | 79 | %26 | 107 | %35 | 305 | %100 |
| Genetic diseases | 771 | %14 | 115 | %2 | 688 | %13 | 1.751 | %32 | 2.093 | %39 | 5.418 | %100 |
| Gynecologic/obstetric diseases | 47 | %17 | 12 | %4 | 32 | %11 | 94 | %34 | 94 | %34 | 279 | %100 |
| Hematological diseases | 62 | %15 | 9 | %2 | 51 | %12 | 140 | %34 | 148 | %36 | 410 | %100 |
| Hepatic diseases | 153 | %17 | 20 | %2 | 73 | %8 | 417 | %47 | 225 | %25 | 888 | %100 |
| Immunological diseases | 42 | %15 | 7 | %2 | 50 | %18 | 68 | %24 | 114 | %41 | 281 | %100 |
| Inborn errors of metabolism | 123 | %16 | 25 | %3 | 100 | %13 | 250 | %32 | 274 | %35 | 772 | %100 |
| Infectious diseases | 1 | %6 | 2 | %12 | 2 | %12 | 3 | %18 | 9 | %53 | 17 | %100 |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 578,9 ; dof= 136.*

**Cross: Please select the sentence that best describes your situation or the situation of the person you care for: / …psychological support**

| PLEASE SELECT THE SENTENCE THAT BEST DESCRIBES YOUR SITUATION OR THE SITUATION OF THE PERSON YOU CARE FOR: | …PSYCHOLOGICAL SUPPORT | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| I know the NAME of the rare disease, syndrome or malformation and it has been CONFIRMED by appropriate genetic, clinical, medical imaging, molecular or biochemical tests (e.g biopsy, blood or urine test) | 790 | %9 | 835 | %9 | 773 | %9 | 2.827 | %31 | 3.823 | %42 | 9.048 | %100 |
| I know the NAME of the rare disease, syndrome or malformation but it has NOT yet been confirmed by appropriate genetic, clinical, medical imaging, molecular or biochemical tests | 61 | %8 | 66 | %9 | 89 | %12 | 185 | %24 | 359 | %47 | 760 | %100 |
| I only have PARTIAL information on the name of the rare disease or the gene involved or the type of disease | 25 | %8 | 22 | %7 | 54 | %18 | 69 | %23 | 136 | %44 | 306 | %100 |
| I know that the disease is rare but the name or the cause have NOT BEEN IDENTIFIED | 44 | %13 | 28 | %8 | 35 | %10 | 80 | %23 | 161 | %46 | 348 | %100 |
| Other, specify... | 2 | %8 | 4 | %17 | 1 | %4 | 4 | %17 | 13 | %54 | 24 | %100 |
| **TOTAL** | **922** | **%9** | **955** | **%9** | **952** | **%9** | **3.165** | **%30** | **4.492** | **%43** | **10.486** | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 75,7 ; dof= 16.*

**Cross: Please select the sentence that best describes your situation or the situation of the person you care for: / ...care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc.**

| PLEASE SELECT THE SENTENCE THAT BEST DESCRIBES YOUR SITUATION OR THE SITUATION OF THE PERSON YOU CARE FOR: | ...CARE COORDINATION SUPPORT SUCH AS HELP TO FIND THE NECESSARY INFORMATION ON THE DISEASE AND THE RIGHT PROFESSIONALS, ARRANGING APPOINTMENTS WITH DIFFERENT HEALTH PROVIDERS, ETC. | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| I know the NAME of the rare disease, syndrome or malformation and it has been CONFIRMED by appropriate genetic, clinical, medical imaging, molecular or biochemical tests (e.g biopsy, blood or urine test) | <u>1.935</u> | <u>%21</u> | 348 | %4 | 1.262 | %14 | <u>1.439</u> | <u>%16</u> | <u>4.064</u> | <u>%45</u> | 9.048 | %100 |
| I know the NAME of the rare disease, syndrome or malformation but it has NOT yet been confirmed by appropriate genetic, clinical, medical imaging, molecular or biochemical tests | <u>62</u> | <u>%8</u> | 19 | %3 | 97 | %13 | 102 | %13 | <u>480</u> | <u>%63</u> | 760 | %100 |
| I only have PARTIAL information on the name of the rare disease or the gene involved or the type of disease | <u>35</u> | <u>%11</u> | 15 | %5 | 45 | %15 | <u>31</u> | <u>%10</u> | <u>180</u> | <u>%59</u> | 306 | %100 |
| I know that the disease is rare but the name or the cause have NOT BEEN IDENTIFIED | <u>49</u> | <u>%14</u> | 8 | %2 | 56 | %16 | 48 | %14 | <u>187</u> | <u>%54</u> | 348 | %100 |
| Other, specify... | 2 | %8 | 1 | %4 | 3 | %13 | 7 | %29 | 11 | %46 | 24 | %100 |
| **TOTAL** | **2.083** | **%20** | **391** | **%4** | **1.463** | **%14** | **1.627** | **%16** | **4.922** | **%47** | **10.486** | |

■ *Under-represented elements* ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 165,3 ; dof= 16.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Please select the sentence that best describes your situation or the situation of the person you care for: / ...financial support including social security benefits

| PLEASE SELECT THE SENTENCE THAT BEST DESCRIBES YOUR SITUATION OR THE SITUATION OF THE PERSON YOU CARE FOR: | ...FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| I know the NAME of the rare disease, syndrome or malformation and it has been CONFIRMED by appropriate genetic, clinical, medical imaging, molecular or biochemical tests (e.g biopsy, blood or urine test) | 1.257 | %14 | 210 | %2 | 1.032 | %11 | 3.180 | %35 | 3.311 | %37 | 8.990 | %100 |
| I know the NAME of the rare disease, syndrome or malformation but it has NOT yet been confirmed by appropriate genetic, clinical, medical imaging, molecular or biochemical tests | 67 | %9 | 14 | %2 | 75 | %10 | 207 | %28 | 388 | %52 | 751 | %100 |
| I only have PARTIAL information on the name of the rare disease or the gene involved or the type of disease | 31 | %10 | 9 | %3 | 51 | %17 | 66 | %22 | 148 | %49 | 305 | %100 |
| I know that the disease is rare but the name or the cause have NOT BEEN IDENTIFIED | 47 | %14 | 9 | %3 | 66 | %19 | 86 | %25 | 137 | %40 | 345 | %100 |
| Other, specify... | 3 | %14 | 1 | %5 | 8 | %36 | 5 | %23 | 5 | %23 | 22 | %100 |
| **TOTAL** | **1.405** | **%13** | **243** | **%2** | **1.232** | **%12** | **3.544** | **%34** | **3.989** | **%38** | **10.413** | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 140,5 ; dof= 16.*

## Cross: Point prevalence of the rare disease / …psychological support

| POINT PREVALENCE OF THE RARE DISEASE | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 1-5 / 10 000 | 194 | %8 | **199** | **%8** | 192 | %8 | **823** | **%34** | 999 | %42 | **2.407** | %100 |
| 1-9 / 100 000 | 157 | %8 | **215** | **%11** | 174 | %9 | 609 | %30 | 844 | %42 | **1.999** | %100 |
| 1-9 / 1 000 000 | 38 | %8 | 51 | %11 | 36 | %8 | 132 | %29 | 202 | %44 | **459** | %100 |
| <1 / 1 000 000 | 81 | %9 | **62** | **%7** | **96** | **%11** | 256 | %30 | 361 | %42 | **856** | %100 |
| TOTAL | **470** | **%8** | **527** | **%9** | **498** | **%9** | **1.820** | **%32** | **2.406** | **%42** | **5.721** | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 31,3 ; dof= 12.*

## Cross: Point prevalence of the rare disease / …care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc.

| POINT PREVALENCE OF THE RARE DISEASE | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 1-5 / 10 000 | 525 | %22 | 85 | %4 | 328 | %14 | 379 | %16 | 1.090 | %45 | **2.407** | %100 |
| 1-9 / 100 000 | 422 | %21 | 82 | %4 | 276 | %14 | 325 | %16 | 894 | %45 | **1.999** | %100 |
| 1-9 / 1 000 000 | 98 | %21 | 14 | %3 | 67 | %15 | 66 | %14 | 214 | %47 | **459** | %100 |
| <1 / 1 000 000 | 170 | %20 | 32 | %4 | 126 | %15 | 129 | %15 | 399 | %47 | **856** | %100 |
| TOTAL | **1.215** | **%21** | **213** | **%4** | **797** | **%14** | **899** | **%16** | **2.597** | **%45** | **5.721** | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is not significant. p-value= 1,0 ; Chi2= 5,2 ; dof= 12.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Point prevalence of the rare disease / …financial support including social security benefits

| POINT PREVALENCE OF THE RARE DISEASE | …FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS | | | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 1-5 / 10 000 | 295 | %12 | 56 | %2 | 249 | %10 | 940 | %39 | 858 | %36 | 2.398 | %100 |
| 1-9 / 100 000 | 273 | %14 | 47 | %2 | 218 | %11 | 694 | %35 | 760 | %38 | 1.992 | %100 |
| 1-9 / 1 000 000 | 77 | %17 | 13 | %3 | 49 | %11 | 142 | %31 | 172 | %38 | 453 | %100 |
| <1 / 1 000 000 | 117 | %14 | 12 | %1 | 121 | %14 | 282 | %33 | 319 | %37 | 851 | %100 |
| TOTAL | 762 | %13 | 128 | %2 | 637 | %11 | 2.058 | %36 | 2.109 | %37 | 5.694 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 32,9 ; dof= 12.*

## Cross: Disease complexity classified into five groups, based on the number of affected body parts. / …psychological support

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | …PSYCHOLOGICAL SUPPORT | | | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 1-3 body parts | 544 | %9 | 599 | %10 | 458 | %8 | 2.009 | %33 | 2.493 | %41 | 6.103 | %100 |
| 4-7 body parts | 281 | %9 | 242 | %8 | 317 | %10 | 864 | %28 | 1.377 | %45 | 3.081 | %100 |
| 8-11 body parts | 66 | %7 | 80 | %8 | 129 | %14 | 226 | %24 | 450 | %47 | 951 | %100 |
| 12-15 body parts | 24 | %8 | 26 | %9 | 38 | %13 | 56 | %20 | 142 | %50 | 286 | %100 |
| 16 body parts or more | 7 | %11 | 8 | %12 | 10 | %15 | 10 | %15 | 30 | %46 | 65 | %100 |
| TOTAL | 922 | %9 | 955 | %9 | 952 | %9 | 3.165 | %30 | 4.492 | %43 | 10.486 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 129,6 ; dof= 16.*

**Cross: Disease complexity classified into five groups, based on the number of affected body parts. / …care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc.**

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | …CARE COORDINATION SUPPORT SUCH AS HELP TO FIND THE NECESSARY INFORMATION ON THE DISEASE AND THE RIGHT PROFESSIONALS, ARRANGING APPOINTMENTS WITH DIFFERENT HEALTH PROVIDERS, ETC. | | | | | | | | | |
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 1-3 body parts | 1.316 | %22 | 266 | %4 | 764 | %13 | 1.113 | %18 | 2.644 | %43 | 6.103 | %100 |
| 4-7 body parts | 597 | %19 | 97 | %3 | 486 | %16 | 387 | %13 | 1.514 | %49 | 3.081 | %100 |
| 8-11 body parts | 136 | %14 | 20 | %2 | 156 | %16 | 115 | %12 | 524 | %55 | 951 | %100 |
| 12-15 body parts | 30 | %10 | 4 | %1 | 41 | %14 | 12 | %4 | 199 | %70 | 286 | %100 |
| 16 body parts or more | 4 | %6 | 4 | %6 | 16 | %25 | 0 | %0 | 41 | %63 | 65 | %100 |
| TOTAL | 2.083 | %20 | 391 | %4 | 1.463 | %14 | 1.627 | %16 | 4.922 | %47 | 10.486 | %100 |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 245,7 ; dof= 16.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

### Cross: Disease complexity classified into five groups, based on the number of affected body parts. / …financial support including social security benefits

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | …FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 1-3 body parts | 855 | %14 | 169 | %3 | 630 | %10 | 2.368 | %39 | 2.037 | %34 | 6.059 | %100 |
| 4-7 body parts | 428 | %14 | 55 | %2 | 408 | %13 | 931 | %30 | 1.237 | %40 | 3.059 | %100 |
| 8-11 body parts | 98 | %10 | 16 | %2 | 140 | %15 | 201 | %21 | 491 | %52 | 946 | %100 |
| 12-15 body parts | 23 | %8 | 1 | %0 | 40 | %14 | 40 | %14 | 181 | %64 | 285 | %100 |
| 16 body parts or more | 1 | %2 | 2 | %3 | 14 | %22 | 4 | %6 | 43 | %67 | 64 | %100 |
| **TOTAL** | 1.405 | %13 | 243 | %2 | 1.232 | %12 | 3.544 | %34 | 3.989 | %38 | 10.413 | %100 |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 363,7 ; dof= 16.*

### Cross: …behavioural disorders that cause problems in school, at home or in social situations / …psychological support

| …BEHAVIOURAL DISORDERS THAT CAUSE PROBLEMS IN SCHOOL, AT HOME OR IN SOCIAL SITUATIONS | …PSYCHOLOGICAL SUPPORT | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 293 | %10 | 226 | %8 | 386 | %13 | 608 | %21 | 1.444 | %49 | 2.957 | %100 |
| No | 597 | %8 | 689 | %10 | 523 | %7 | 2.425 | %34 | 2.851 | %40 | 7.085 | %100 |
| Don't know | 32 | %7 | 40 | %9 | 43 | %10 | 132 | %30 | 197 | %44 | 444 | %100 |
| **TOTAL** | 922 | %9 | 955 | %9 | 952 | %9 | 3.165 | %30 | 4.492 | %43 | 10.486 | %100 |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 255,9 ; dof= 8.*

**Cross: …behavioural disorders that cause problems in school, at home or in social situations / …care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc.**

| …BEHAVIOURAL DISORDERS THAT CAUSE PROBLEMS IN SCHOOL, AT HOME OR IN SOCIAL SITUATIONS | …CARE COORDINATION SUPPORT SUCH AS HELP TO FIND THE NECESSARY INFORMATION ON THE DISEASE AND THE RIGHT PROFESSIONALS, ARRANGING APPOINTMENTS WITH DIFFERENT HEALTH PROVIDERS, ETC. | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 551 | %19 | 92 | %3 | 488 | %17 | 321 | %11 | 1.505 | %51 | 2.957 | %100 |
| No | 1.464 | %21 | 280 | %4 | 913 | %13 | 1.236 | %17 | 3.192 | %45 | 7.085 | %100 |
| Don't know | 68 | %15 | 19 | %4 | 62 | %14 | 70 | %16 | 225 | %51 | 444 | %100 |
| TOTAL | 2.083 | %20 | 391 | %4 | 1.463 | %14 | 1.627 | %16 | 4.922 | %47 | 10.486 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 108,0 ; dof= 8.*

**Cross: …behavioural disorders that cause problems in school, at home or in social situations / …financial support including social security benefits**

| …BEHAVIOURAL DISORDERS THAT CAUSE PROBLEMS IN SCHOOL, AT HOME OR IN SOCIAL SITUATIONS | …FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 412 | %14 | 53 | %2 | 470 | %16 | 696 | %24 | 1.297 | %44 | 2.928 | %100 |
| No | 958 | %14 | 182 | %3 | 708 | %10 | 2.719 | %39 | 2.476 | %35 | 7.043 | %100 |
| Don't know | 35 | %8 | 8 | %2 | 54 | %12 | 129 | %29 | 216 | %49 | 442 | %100 |
| TOTAL | 1.405 | %13 | 243 | %2 | 1.232 | %12 | 3.544 | %34 | 3.989 | %38 | 10.413 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 275,3 ; dof= 8.*

**Cross: …intellectual disabilities or cognitive symptoms (i.e. problems with memory, language, thinking or judgement) / …psychological support**

| …INTELLECTUAL DISABILITIES OR COGNITIVE SYMPTOMS (I.E. PROBLEMS WITH MEMORY, LANGUAGE, THINKING OR JUDGEMENT) | …PSYCHOLOGICAL SUPPORT | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 292 | %10 | 231 | %8 | 375 | %13 | 626 | %21 | 1.412 | %48 | 2.936 | %100 |
| No | 604 | %8 | 696 | %10 | 548 | %8 | 2.446 | %34 | 2.942 | %41 | 7.236 | %100 |
| Don't know | 26 | %8 | 28 | %9 | 29 | %9 | 93 | %30 | 138 | %44 | 314 | %100 |
| TOTAL | 922 | %9 | 955 | %9 | 952 | %9 | 3.165 | %30 | 4.492 | %43 | 10.486 | %100 |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 210,3 ; dof= 8.*

**Cross: …intellectual disabilities or cognitive symptoms (i.e. problems with memory, language, thinking or judgement) / …care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc.**

| …INTELLECTUAL DISABILITIES OR COGNITIVE SYMPTOMS (I.E. PROBLEMS WITH MEMORY, LANGUAGE, THINKING OR JUDGEMENT) | …CARE COORDINATION SUPPORT SUCH AS HELP TO FIND THE NECESSARY INFORMATION ON THE DISEASE AND THE RIGHT PROFESSIONALS, ARRANGING APPOINTMENTS WITH DIFFERENT HEALTH PROVIDERS, ETC. | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 502 | %17 | 84 | %3 | 501 | %17 | 281 | %10 | 1.568 | %53 | 2.936 | %100 |
| No | 1.517 | %21 | 297 | %4 | 918 | %13 | 1.301 | %18 | 3.203 | %44 | 7.236 | %100 |
| Don't know | 64 | %20 | 10 | %3 | 44 | %14 | 45 | %14 | 151 | %48 | 314 | %100 |
| TOTAL | 2.083 | %20 | 391 | %4 | 1.463 | %14 | 1.627 | %16 | 4.922 | %47 | 10.486 | %100 |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 186,1 ; dof= 8.*

## Cross: …intellectual disabilities or cognitive symptoms (i.e. problems with memory, language, thinking or judgement) / …financial support including social security benefits

| …INTELLECTUAL DISABILITIES OR COGNITIVE SYMPTOMS (I.E. PROBLEMS WITH MEMORY, LANGUAGE, THINKING OR JUDGEMENT) | …FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 430 | %15 | 50 | %2 | 503 | %17 | 626 | %21 | 1.304 | %45 | 2.913 | %100 |
| No | 943 | %13 | 182 | %3 | 681 | %9 | 2.822 | %39 | 2.559 | %36 | 7.187 | %100 |
| Don't know | 32 | %10 | 11 | %4 | 48 | %15 | 96 | %31 | 126 | %40 | 313 | %100 |
| **TOTAL** | **1.405** | **%13** | **243** | **%2** | **1.232** | **%12** | **3.544** | **%34** | **3.989** | **%38** | **10.413** | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 363,5 ; dof= 8.*

## Cross: …clinical signs or symptoms that come and go / …psychological support

| …CLINICAL SIGNS OR SYMPTOMS THAT COME AND GO | …PSYCHOLOGICAL SUPPORT | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 494 | %8 | 501 | %8 | 588 | %10 | 1.661 | %28 | 2.696 | %45 | 5.940 | %100 |
| No | 372 | %10 | 391 | %10 | 293 | %8 | 1.265 | %33 | 1.467 | %39 | 3.788 | %100 |
| Don't know | 56 | %7 | 63 | %8 | 71 | %9 | 239 | %32 | 329 | %43 | 758 | %100 |
| **TOTAL** | **922** | **%9** | **955** | **%9** | **952** | **%9** | **3.165** | **%30** | **4.492** | **%43** | **10.486** | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 76,5 ; dof= 8.*

**Cross: …clinical signs or symptoms that come and go / …care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc.**

| …CLINICAL SIGNS OR SYMPTOMS THAT COME AND GO | …CARE COORDINATION SUPPORT SUCH AS HELP TO FIND THE NECESSARY INFORMATION ON THE DISEASE AND THE RIGHT PROFESSIONALS, ARRANGING APPOINTMENTS WITH DIFFERENT HEALTH PROVIDERS, ETC. | | | | | | | | | | | |
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 1.048 | %18 | 184 | %3 | 862 | %15 | 812 | %14 | 3.034 | %51 | 5.940 | %100 |
| No | 864 | %23 | 174 | %5 | 487 | %13 | 718 | %19 | 1.545 | %41 | 3.788 | %100 |
| Don't know | 171 | %23 | 33 | %4 | 114 | %15 | 97 | %13 | 343 | %45 | 758 | %100 |
| **TOTAL** | 2.083 | %20 | 391 | %4 | 1.463 | %14 | 1.627 | %16 | 4.922 | %47 | 10.486 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 152,2 ; dof= 8.*

**Cross: …clinical signs or symptoms that come and go / …financial support including social security benefits**

| …CLINICAL SIGNS OR SYMPTOMS THAT COME AND GO | …FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS | | | | | | | | | | | |
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 705 | %12 | 111 | %2 | 721 | %12 | 1.941 | %33 | 2.419 | %41 | 5.897 | %100 |
| No | 584 | %16 | 107 | %3 | 408 | %11 | 1.363 | %36 | 1.298 | %35 | 3.760 | %100 |
| Don't know | 116 | %15 | 25 | %3 | 103 | %14 | 240 | %32 | 272 | %36 | 756 | %100 |
| **TOTAL** | 1.405 | %13 | 243 | %2 | 1.232 | %12 | 3.544 | %34 | 3.989 | %38 | 10.413 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 77,4 ; dof= 8.*

## Cross: …invisible symptoms such as pain, dizziness, headaches, fatigue, etc. / …psychological support

| …INVISIBLE SYMPTOMS SUCH AS PAIN, DIZZINESS, HEADACHES, FATIGUE, ETC. | …PSYCHOLOGICAL SUPPORT | | | | | | | | | | |
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 625 | %9 | 622 | %9 | 710 | %10 | 1.968 | %28 | 3.095 | %44 | 7.020 | %100 |
| No | 245 | %8 | 284 | %10 | 191 | %7 | 1.043 | %36 | 1.153 | %40 | 2.916 | %100 |
| Don't know | 52 | %9 | 49 | %9 | 51 | %9 | 154 | %28 | 244 | %44 | 550 | %100 |
| **TOTAL** | **922** | **%9** | **955** | **%9** | **952** | **%9** | **3.165** | **%30** | **4.492** | **%43** | **10.486** | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 83,5 ; dof= 8.*

## Cross: …invisible symptoms such as pain, dizziness, headaches, fatigue, etc. / …care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc.

| …INVISIBLE SYMPTOMS SUCH AS PAIN, DIZZINESS, HEADACHES, FATIGUE, ETC. | …CARE COORDINATION SUPPORT SUCH AS HELP TO FIND THE NECESSARY INFORMATION ON THE DISEASE AND THE RIGHT PROFESSIONALS, ARRANGING APPOINTMENTS WITH DIFFERENT HEALTH PROVIDERS, ETC. | | | | | | | | | | |
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 1.257 | %18 | 234 | %3 | 982 | %14 | 1.009 | %14 | 3.538 | %50 | 7.020 | %100 |
| No | 702 | %24 | 135 | %5 | 396 | %14 | 541 | %19 | 1.142 | %39 | 2.916 | %100 |
| Don't know | 124 | %23 | 22 | %4 | 85 | %15 | 77 | %14 | 242 | %44 | 550 | %100 |
| **TOTAL** | **2.083** | **%20** | **391** | **%4** | **1.463** | **%14** | **1.627** | **%16** | **4.922** | **%47** | **10.486** | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 132,7 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: …invisible symptoms such as pain, dizziness, headaches, fatigue, etc. / …financial support including social security benefits

| …INVISIBLE SYMPTOMS SUCH AS PAIN, DIZZINESS, HEADACHES, FATIGUE, ETC. | …FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS | | | | | | | | | | TOTAL | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 838 | %12 | 146 | %2 | 794 | %11 | 2.295 | %33 | 2.892 | %42 | 6.965 | %100 |
| No | 466 | %16 | 80 | %3 | 340 | %12 | 1.106 | %38 | 906 | %31 | 2.898 | %100 |
| Don't know | 101 | %18 | 17 | %3 | 98 | %18 | 143 | %26 | 191 | %35 | 550 | %100 |
| TOTAL | 1.405 | %13 | 243 | %2 | 1.232 | %12 | 3.544 | %34 | 3.989 | %38 | 10.413 | %100 |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 143,7 ; dof= 8.*

## Cross: …sudden onset symptoms requiring urgent care / …psychological support

| …SUDDEN ONSET SYMPTOMS REQUIRING URGENT CARE | …PSYCHOLOGICAL SUPPORT | | | | | | | | | | TOTAL | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 451 | %10 | 403 | %9 | 487 | %10 | 1.202 | %26 | 2.105 | %45 | 4.648 | %100 |
| No | 422 | %8 | 507 | %10 | 390 | %7 | 1.786 | %34 | 2.146 | %41 | 5.251 | %100 |
| Don't know | 49 | %8 | 45 | %8 | 75 | %13 | 177 | %30 | 241 | %41 | 587 | %100 |
| TOTAL | 922 | %9 | 955 | %9 | 952 | %9 | 3.165 | %30 | 4.492 | %43 | 10.486 | %100 |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 112,6 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: …sudden onset symptoms requiring urgent care / …care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc.

| …SUDDEN ONSET SYMPTOMS REQUIRING URGENT CARE | …CARE COORDINATION SUPPORT SUCH AS HELP TO FIND THE NECESSARY INFORMATION ON THE DISEASE AND THE RIGHT PROFESSIONALS, ARRANGING APPOINTMENTS WITH DIFFERENT HEALTH PROVIDERS, ETC. | | | | | | | | | |
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 907 | %20 | 148 | %3 | 691 | %15 | 592 | %13 | 2.310 | %50 | 4.648 | %100 |
| No | 1.067 | %20 | 218 | %4 | 673 | %13 | 956 | %18 | 2.337 | %45 | 5.251 | %100 |
| Don't know | 109 | %19 | 25 | %4 | 99 | %17 | 79 | %13 | 275 | %47 | 587 | %100 |
| **TOTAL** | **2.083** | **%20** | **391** | **%4** | **1.463** | **%14** | **1.627** | **%16** | **4.922** | **%47** | **10.486** | |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 82,6 ; dof= 8.*

## Cross: …sudden onset symptoms requiring urgent care / …financial support including social security benefits

| …SUDDEN ONSET SYMPTOMS REQUIRING URGENT CARE | …FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS | | | | | | | | | |
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 567 | %12 | 95 | %2 | 605 | %13 | 1.386 | %30 | 1.953 | %42 | 4.606 | %100 |
| No | 760 | %15 | 134 | %3 | 533 | %10 | 1.992 | %38 | 1.802 | %35 | 5.221 | %100 |
| Don't know | 78 | %13 | 14 | %2 | 94 | %16 | 166 | %28 | 234 | %40 | 586 | %100 |
| **TOTAL** | **1.405** | **%13** | **243** | **%2** | **1.232** | **%12** | **3.544** | **%34** | **3.989** | **%38** | **10.413** | |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 131,7 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis? / …psychological support**

| | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | **…PSYCHOLOGICAL SUPPORT** | | | | | | | | | | | | |
| **HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS?** | **YES AND ENOUGH TO MEET MY NEEDS** | | **YES BUT IT IS/WAS NOT NEEDED** | | **YES BUT NOT ENOUGH TO MEET MY NEEDS** | | **NO BUT IT IS/WAS NOT NEEDED** | | **NO BUT IT IS/WAS NEEDED** | | **TOTAL** | |
| | **N** | **%** | **N** | **%** | **N** | **%** | **N** | **%** | **N** | **%** | **N** | **%** |
| 0-1 | 125 | %10 | 146 | %11 | 66 | %5 | 538 | %41 | 430 | %33 | 1.305 | %100 |
| between 2 and 4 | 429 | %9 | 413 | %9 | 319 | %7 | 1.572 | %34 | 1.836 | %40 | 4.569 | %100 |
| between 5 and 7 | 173 | %9 | 184 | %9 | 220 | %11 | 532 | %26 | 924 | %45 | 2.033 | %100 |
| between 8 and 10 | 59 | %7 | 62 | %8 | 84 | %11 | 182 | %23 | 404 | %51 | 791 | %100 |
| more than 10 | 136 | %8 | 150 | %8 | 263 | %15 | 341 | %19 | 898 | %50 | 1.788 | %100 |
| **TOTAL** | **922** | **%9** | **955** | **%9** | **952** | **%9** | **3.165** | **%30** | **4.492** | **%43** | **10.486** | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 385,8 ; dof= 16.*

**Cross: How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis? / …care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc.**

| | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | **…CARE COORDINATION SUPPORT SUCH AS HELP TO FIND THE NECESSARY INFORMATION ON THE DISEASE AND THE RIGHT PROFESSIONALS, ARRANGING APPOINTMENTS WITH DIFFERENT HEALTH PROVIDERS, ETC.** | | | | | | | | | | | | |
| **HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS?** | **YES AND ENOUGH TO MEET MY NEEDS** | | **YES BUT IT IS/WAS NOT NEEDED** | | **YES BUT NOT ENOUGH TO MEET MY NEEDS** | | **NO BUT IT IS/WAS NOT NEEDED** | | **NO BUT IT IS/WAS NEEDED** | | **TOTAL** | |
| | **N** | **%** | **N** | **%** | **N** | **%** | **N** | **%** | **N** | **%** | **N** | **%** |
| 0-1 | 355 | %27 | 67 | %5 | 144 | %11 | 318 | %24 | 421 | %32 | 1.305 | %100 |
| between 2 and 4 | 1.114 | %24 | 213 | %5 | 604 | %13 | 839 | %18 | 1.799 | %39 | 4.569 | %100 |
| between 5 and 7 | 354 | %17 | 65 | %3 | 329 | %16 | 242 | %12 | 1.043 | %51 | 2.033 | %100 |
| between 8 and 10 | 109 | %14 | 14 | %2 | 123 | %16 | 85 | %11 | 460 | %58 | 791 | %100 |
| more than 10 | 151 | %8 | 32 | %2 | 263 | %15 | 143 | %8 | 1.199 | %67 | 1.788 | %100 |
| **TOTAL** | **2.083** | **%20** | **391** | **%4** | **1.463** | **%14** | **1.627** | **%16** | **4.922** | **%47** | **10.486** | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 767,9 ; dof= 16.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

## Cross: How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis? / …financial support including social security benefits

| HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS? | …FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 0-1 | 219 | %17 | 47 | %4 | 96 | %7 | 553 | %43 | 384 | %30 | 1.299 | %100 |
| between 2 and 4 | 691 | %15 | 127 | %3 | 458 | %10 | 1.815 | %40 | 1.452 | %32 | 4.543 | %100 |
| between 5 and 7 | 243 | %12 | 44 | %2 | 281 | %14 | 626 | %31 | 824 | %41 | 2.018 | %100 |
| between 8 and 10 | 89 | %11 | 12 | %2 | 93 | %12 | 213 | %27 | 373 | %48 | 780 | %100 |
| more than 10 | 163 | %9 | 13 | %1 | 304 | %17 | 337 | %19 | 956 | %54 | 1.773 | %100 |
| TOTAL | 1.405 | %13 | 243 | %2 | 1.232 | %12 | 3.544 | %34 | 3.989 | %38 | 10.413 | %100 |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 585,4 ; dof= 16.*

## Cross: I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases / …psychological support

| I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | …PSYCHOLOGICAL SUPPORT | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 643 | %11 | 653 | %11 | 567 | %9 | 1.812 | %30 | 2.323 | %39 | 5.998 | %100 |
| No | 276 | %6 | 300 | %7 | 376 | %9 | 1.338 | %30 | 2.125 | %48 | 4.415 | %100 |
| TOTAL | 919 | %9 | 953 | %9 | 943 | %9 | 3.150 | %30 | 4.448 | %43 | 10.413 | %100 |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 159,2 ; dof= 4.*

**Cross: I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases / …care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc.**

| I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | …CARE COORDINATION SUPPORT SUCH AS HELP TO FIND THE NECESSARY INFORMATION ON THE DISEASE AND THE RIGHT PROFESSIONALS, ARRANGING APPOINTMENTS WITH DIFFERENT HEALTH PROVIDERS, ETC. | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 1.519 | %25 | 268 | %4 | 902 | %15 | 951 | %16 | 2.358 | %39 | 5.998 | %100 |
| No | 550 | %12 | 120 | %3 | 547 | %12 | 668 | %15 | 2.530 | %57 | 4.415 | %100 |
| TOTAL | 2.069 | %20 | 388 | %4 | 1.449 | %14 | 1.619 | %16 | 4.888 | %47 | 10.413 | |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 421,9 ; dof= 4.*

**Cross: I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases / …financial support including social security benefits**

| I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | …FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 974 | %16 | 168 | %3 | 727 | %12 | 2.067 | %34 | 2.062 | %34 | 5.998 | %100 |
| No | 431 | %10 | 75 | %2 | 505 | %11 | 1.477 | %33 | 1.927 | %44 | 4.415 | %100 |
| TOTAL | 1.405 | %13 | 243 | %2 | 1.232 | %12 | 3.544 | %34 | 3.989 | %38 | 10.413 | |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 151,1 ; dof= 4.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: …wrongly attributed to another physical disease? / …psychological support

| …WRONGLY ATTRIBUTED TO ANOTHER PHYSICAL DISEASE? | …PSYCHOLOGICAL SUPPORT | | | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, one time | 168 | %9 | 183 | %9 | 157 | %8 | 601 | %31 | 841 | %43 | 1.950 | %100 |
| YES, several times | 321 | %7 | 329 | %7 | 533 | %12 | 1.117 | %25 | 2.220 | %49 | 4.520 | %100 |
| NO | 433 | %11 | 443 | %11 | 262 | %7 | 1.447 | %36 | 1.431 | %36 | 4.016 | %100 |
| TOTAL | 922 | %9 | 955 | %9 | 952 | %9 | 3.165 | %30 | 4.492 | %43 | 10.486 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 314,6 ; dof= 8.*

## Cross: …wrongly attributed to another physical disease? / …care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc.

| …WRONGLY ATTRIBUTED TO ANOTHER PHYSICAL DISEASE? | …CARE COORDINATION SUPPORT SUCH AS HELP TO FIND THE NECESSARY INFORMATION ON THE DISEASE AND THE RIGHT PROFESSIONALS, ARRANGING APPOINTMENTS WITH DIFFERENT HEALTH PROVIDERS, ETC. | | | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, one time | 446 | %23 | 90 | %5 | 296 | %15 | 289 | %15 | 829 | %43 | 1.950 | %100 |
| YES, several times | 559 | %12 | 104 | %2 | 667 | %15 | 478 | %11 | 2.712 | %60 | 4.520 | %100 |
| NO | 1.078 | %27 | 197 | %5 | 500 | %12 | 860 | %21 | 1.381 | %34 | 4.016 | %100 |
| TOTAL | 2.083 | %20 | 391 | %4 | 1.463 | %14 | 1.627 | %16 | 4.922 | %47 | 10.486 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 758,6 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: …wrongly attributed to another physical disease? / …financial support including social security benefits

| …WRONGLY ATTRIBUTED TO ANOTHER PHYSICAL DISEASE? | …FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS | | | | | | | | | | | |
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, one time | 269 | %14 | 48 | %2 | 220 | %11 | 699 | %36 | 696 | %36 | 1.932 | %100 |
| YES, several times | 448 | %10 | 63 | %1 | 582 | %13 | 1.259 | %28 | 2.129 | %48 | 4.481 | %100 |
| NO | 688 | %17 | 132 | %3 | 430 | %11 | 1.586 | %40 | 1.164 | %29 | 4.000 | %100 |
| TOTAL | 1.405 | %13 | 243 | %2 | 1.232 | %12 | 3.544 | %34 | 3.989 | %38 | 10.413 | %100 |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 400,0 ; dof= 8.*

## Cross: …neglected, not taken seriously and/or considered as psychological? / …psychological support

| …NEGLECTED, NOT TAKEN SERIOUSLY AND/OR CONSIDERED AS PSYCHOLOGICAL? | …PSYCHOLOGICAL SUPPORT | | | | | | | | | | | |
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, one time | 113 | %9 | 114 | %9 | 128 | %10 | 384 | %31 | 507 | %41 | 1.246 | %100 |
| YES, several times | 338 | %7 | 356 | %7 | 565 | %11 | 1.135 | %23 | 2.540 | %51 | 4.934 | %100 |
| NO | 471 | %11 | 485 | %11 | 259 | %6 | 1.646 | %38 | 1.445 | %34 | 4.306 | %100 |
| TOTAL | 922 | %9 | 955 | %9 | 952 | %9 | 3.165 | %30 | 4.492 | %43 | 10.486 | %100 |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 512,9 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: …neglected, not taken seriously and/or considered as psychological? / …care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc.**

| …NEGLECTED, NOT TAKEN SERIOUSLY AND/OR CONSIDERED AS PSYCHOLOGICAL? | …CARE COORDINATION SUPPORT SUCH AS HELP TO FIND THE NECESSARY INFORMATION ON THE DISEASE AND THE RIGHT PROFESSIONALS, ARRANGING APPOINTMENTS WITH DIFFERENT HEALTH PROVIDERS, ETC. | | | | | | | | | | |
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, one time | 253 | %20 | 48 | %4 | 203 | %16 | 212 | %17 | 530 | %43 | 1.246 | %100 |
| YES, several times | 567 | %11 | 108 | %2 | 693 | %14 | 503 | %10 | 3.063 | %62 | 4.934 | %100 |
| NO | 1.263 | %29 | 235 | %5 | 567 | %13 | 912 | %21 | 1.329 | %31 | 4.306 | %100 |
| TOTAL | 2.083 | %20 | 391 | %4 | 1.463 | %14 | 1.627 | %16 | 4.922 | %47 | 10.486 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 1.105,3 ; dof= 8.*

**Cross: …neglected, not taken seriously and/or considered as psychological? / …financial support including social security benefits**

| …NEGLECTED, NOT TAKEN SERIOUSLY AND/OR CONSIDERED AS PSYCHOLOGICAL? | …FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS | | | | | | | | | | |
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, one time | 175 | %14 | 32 | %3 | 141 | %11 | 444 | %36 | 440 | %36 | 1.232 | %100 |
| YES, several times | 433 | %9 | 67 | %1 | 618 | %13 | 1.381 | %28 | 2.401 | %49 | 4.900 | %100 |
| NO | 797 | %19 | 144 | %3 | 473 | %11 | 1.719 | %40 | 1.148 | %27 | 4.281 | %100 |
| TOTAL | 1.405 | %13 | 243 | %2 | 1.232 | %12 | 3.544 | %34 | 3.989 | %38 | 10.413 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 600,7 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed. / … psychological support**

| HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | ...PSYCHOLOGICAL SUPPORT | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, one time | 232 | %9 | 237 | %9 | 205 | %8 | 802 | %30 | 1.207 | %45 | 2.683 | %100 |
| YES, several times | 358 | %7 | 376 | %7 | 586 | %12 | 1.271 | %25 | 2.427 | %48 | 5.018 | %100 |
| NO | 332 | %12 | 342 | %12 | 161 | %6 | 1.092 | %39 | 858 | %31 | 2.785 | %100 |
| TOTAL | 922 | %9 | 955 | %9 | 952 | %9 | 3.165 | %30 | 4.492 | %43 | 10.486 | |

🟥 *Under-represented elements* 🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 416,1 ; dof= 8.*

**Cross: Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed. / …care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc.**

| HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | ...CARE COORDINATION SUPPORT SUCH AS HELP TO FIND THE NECESSARY INFORMATION ON THE DISEASE AND THE RIGHT PROFESSIONALS, ARRANGING APPOINTMENTS WITH DIFFERENT HEALTH PROVIDERS, ETC. | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, one time | 525 | %20 | 106 | %4 | 395 | %15 | 423 | %16 | 1.234 | %46 | 2.683 | %100 |
| YES, several times | 682 | %14 | 127 | %3 | 744 | %15 | 556 | %11 | 2.909 | %58 | 5.018 | %100 |
| NO | 876 | %31 | 158 | %6 | 324 | %12 | 648 | %23 | 779 | %28 | 2.785 | %100 |
| TOTAL | 2.083 | %20 | 391 | %4 | 1.463 | %14 | 1.627 | %16 | 4.922 | %47 | 10.486 | |

🟥 *Under-represented elements* 🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 866,1 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed. / …financial support including social security benefits**

| HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | …FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES AND ENOUGH TO MEET MY NEEDS | | YES BUT IT IS/WAS NOT NEEDED | | YES BUT NOT ENOUGH TO MEET MY NEEDS | | NO BUT IT IS/WAS NOT NEEDED | | NO BUT IT IS/WAS NEEDED | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, one time | 339 | %13 | 61 | %2 | 306 | %11 | 958 | %36 | 1.005 | %38 | 2.669 | %100 |
| YES, several times | 526 | %11 | 79 | %2 | 633 | %13 | 1.443 | %29 | 2.291 | %46 | 4.972 | %100 |
| NO | 540 | %19 | 103 | %4 | 293 | %11 | 1.143 | %41 | 693 | %25 | 2.772 | %100 |
| **TOTAL** | **1.405** | **%13** | **243** | **%2** | **1.232** | **%12** | **3.544** | **%34** | **3.989** | **%38** | **10.413** | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 436,6 ; dof= 8.*

# Chapter 13.

## Support

Rare Barometer
A EURORDIS INITIATIVE

EURORDIS
RARE DISEASES EUROPE
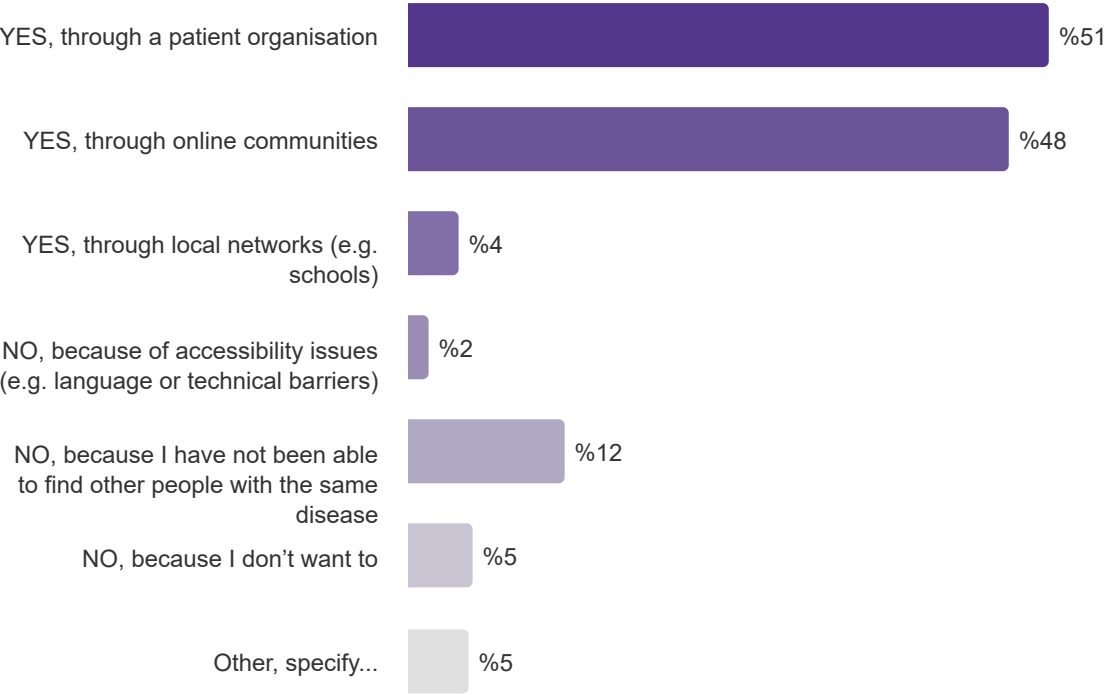
**Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?**

| | N |
|---|---|
| YES, through a patient organisation | 5.326 |
| YES, through online communities | 4.992 |
| YES, through local networks (e.g. schools) | 436 |
| NO, because of accessibility issues (e.g. language or technical barriers) | 190 |
| NO, because I have not been able to find other people with the same disease | 1.310 |
| NO, because I don't want to | 547 |
| Other, specify... | 514 |
| **TOTAL** | **10.486** |

**Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?**

| | |
|---|---|
| YES, through a patient organisation | %51 |
| YES, through online communities | %48 |
| YES, through local networks (e.g. schools) | %4 |
| NO, because of accessibility issues (e.g. language or technical barriers) | %2 |
| NO, because I have not been able to find other people with the same disease | %12 |
| NO, because I don't want to | %5 |
| Other, specify... | %5 |

**Respondents could choose several items**

Rare Barometer
A EURORDIS INITIATIVE

EURORDIS
RARE DISEASES EUROPE

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

| Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease? | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| YES, through a patient organisation | 0,5 | 4.097 | 3,7 | 3.923 | 4,1 | 2.416 | 3,6 | 4.165 | 5,0 | 3.558 |
| YES, through online communities | 0,3 | 3.852 | 3,9 | 3.657 | 4,0 | 2.100 | 4,0 | 3.887 | 4,9 | 3.213 |
| YES, through local networks (e.g. schools) | 0,1 | 321 | 4,4 | 298 | 4,8 | 175 | 4,7 | 334 | 5,3 | 274 |
| NO, because of accessibility issues (e.g. language or technical barriers) | 0,3 | 126 | 3,0 | 114 | 5,5 | 72 | 5,7 | 124 | **7,6** | 96 |
| NO, because I have not been able to find other people with the same disease | 0,6 | 923 | **2,6** | 811 | **3,1** | 468 | 3,9 | 848 | 4,4 | 664 |
| NO, because I don't want to | 1,0 | 368 | 2,9 | 339 | 2,9 | 222 | **2,4** | 361 | **3,9** | 299 |
| Other, specify... | 0,5 | 372 | 3,8 | 340 | 4,8 | 210 | 3,3 | 371 | 5,3 | 328 |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is not significant. p-value= 0,6 ; Fisher= 0,8.*

*Inter variance= 38,6. Intra variance= 48,2.*

**Cross: Are you a... / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?**

| ARE YOU A... | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | | | | | | | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY... | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Patient | 3.481 | %51 | 3.358 | %50 | 240 | %4 | 96 | %1 | 791 | %12 | 356 | %5 | 338 | %5 | 6.772 | |
| Former or recovering patient (e.g. cancer survivor) | 108 | %44 | 94 | %38 | 13 | %5 | 5 | %2 | 39 | %16 | 35 | %14 | 11 | %4 | 247 | |
| Parent of a person living with a rare disease | 1.560 | %51 | 1.416 | %46 | 160 | %5 | 74 | %2 | 410 | %13 | 121 | %4 | 136 | %4 | 3.078 | |
| Grandparent of a person living with a rare disease | 22 | %55 | 18 | %45 | 5 | %13 | 1 | %3 | 5 | %13 | 3 | %8 | 0 | %0 | 40 | |
| Spouse of a person living with a rare disease | 81 | %44 | 56 | %30 | 9 | %5 | 8 | %4 | 37 | %20 | 16 | %9 | 11 | %6 | 186 | |
| Uncle/aunt of a person living with a rare disease | 15 | %65 | 7 | %30 | 3 | %13 | 0 | %0 | 3 | %13 | 1 | %4 | 0 | %0 | 23 | |
| Sibling of a person living with a rare disease | 25 | %52 | 16 | %33 | 1 | %2 | 2 | %4 | 2 | %4 | 6 | %13 | 5 | %10 | 48 | |
| Other, specify... | 34 | %37 | 27 | %29 | 5 | %5 | 4 | %4 | 23 | %25 | 9 | %10 | 13 | %14 | 92 | |
| TOTAL | 5.326 | %51 | 4.992 | %48 | 436 | %4 | 190 | %2 | 1.310 | %12 | 547 | %5 | 514 | %5 | 10.486 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 208,5 ; dof= 42.*

**Respondents can be:**

**- patients themselves** (directly affected by the rare disease, or recovering from the rare disease).

**- or family members of patients** (parents, grand-parents, spouses, uncles/aunts, siblings or other family member).

**Cross: Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable) / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?**

| AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE) | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY... | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Less than 2 years old | 1.135 | %56 | 998 | %49 | 113 | %6 | 41 | %2 | 235 | %11 | 65 | %3 | 84 | %4 | 2.045 | |
| 2 to less that 10 years old | 478 | %52 | 419 | %45 | 45 | %5 | 30 | %3 | 121 | %13 | 45 | %5 | 50 | %5 | 925 | |
| 10 to less than 20 years old | 477 | %50 | 515 | %54 | 52 | %5 | 11 | %1 | 104 | %11 | 49 | %5 | 48 | %5 | 952 | |
| 20 to less than 30 years old | 471 | %48 | 505 | %52 | 36 | %4 | 7 | %1 | 116 | %12 | 66 | %7 | 48 | %5 | 978 | |
| 30 to less than 50 years old | 1.228 | %52 | 1.167 | %50 | 76 | %3 | 30 | %1 | 270 | %11 | 101 | %4 | 115 | %5 | 2.353 | |
| 50 years old or more | 547 | %49 | 487 | %44 | 31 | %3 | 15 | %1 | 149 | %13 | 66 | %6 | 68 | %6 | 1.107 | |
| **TOTAL** | **4.336** | **%52** | **4.091** | **%49** | **353** | **%4** | **134** | **%2** | **995** | **%12** | **392** | **%5** | **413** | **%5** | **8.360** | |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 109,4 ; dof= 30.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

**Cross: How old were you when you stopped full-time education? / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?**

| HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION? | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY... | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| 15 y.o. or under | 185 | %47 | 166 | %42 | 17 | %4 | 12 | %3 | 58 | %15 | 19 | %5 | 21 | %5 | **391** | |
| between 16 and 19 y.o. | 1.139 | %47 | 1.128 | %47 | 100 | %4 | 46 | %2 | 322 | %13 | 121 | %5 | 129 | %5 | **2.420** | |
| between 20 and 23 y.o. | 1.588 | %54 | 1.367 | %46 | 121 | %4 | 51 | %2 | 364 | %12 | 144 | %5 | 138 | %5 | **2.955** | |
| 24 y.o. or above | 1.547 | %55 | 1.429 | %51 | 121 | %4 | 36 | %1 | 311 | %11 | 127 | %4 | 153 | %5 | **2.827** | |
| still studying | 220 | %45 | 252 | %51 | 29 | %6 | 13 | %3 | 65 | %13 | 39 | %8 | 24 | %5 | **494** | |
| **TOTAL** | **4.679** | **%51** | **4.342** | **%48** | **388** | **%4** | **158** | **%2** | **1.120** | **%12** | **450** | **%5** | **465** | **%5** | **9.087** | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 67,3 ; dof= 24.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Are you: / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?**

| ARE YOU: | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | | | | | | | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY... | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Female | 3.891 | %49 | 3.952 | %50 | 317 | %4 | 137 | %2 | 985 | %12 | 381 | %5 | 390 | %5 | 7.930 | |
| Male | 1.111 | %61 | 725 | %40 | 92 | %5 | 33 | %2 | 207 | %11 | 112 | %6 | 86 | %5 | 1.807 | |
| Other | 28 | %50 | 29 | %52 | 2 | %4 | 1 | %2 | 6 | %11 | 3 | %5 | 5 | %9 | 56 | |
| TOTAL | 5.030 | %51 | 4.706 | %48 | 411 | %4 | 171 | %2 | 1.198 | %12 | 496 | %5 | 481 | %5 | 9.793 | |

■ *Under-represented elements* ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 86,4 ; dof= 12.*

**Cross: How old were you when you stopped full-time education? / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?**

| HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION? | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | | | | | | | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY... | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| 15 y.o. or under | 211 | %46 | 201 | %44 | 19 | %4 | 14 | %3 | 63 | %14 | 25 | %5 | 26 | %6 | 455 | |
| between 16 and 19 y.o. | 1.154 | %47 | 1.144 | %46 | 102 | %4 | 47 | %2 | 333 | %14 | 127 | %5 | 130 | %5 | 2.464 | |
| between 20 and 23 y.o. | 1.614 | %53 | 1.399 | %46 | 125 | %4 | 54 | %2 | 373 | %12 | 153 | %5 | 141 | %5 | 3.022 | |
| 24 y.o. or above | 1.700 | %54 | 1.598 | %51 | 142 | %5 | 43 | %1 | 350 | %11 | 145 | %5 | 168 | %5 | 3.145 | |
| TOTAL | 4.679 | %51 | 4.342 | %48 | 388 | %4 | 158 | %2 | 1.119 | %12 | 450 | %5 | 465 | %5 | 9.086 | |

■ *Under-represented elements* ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 47,4 ; dof= 18.*

**Cross: How would you best describe yourself? / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?**

| HOW WOULD YOU BEST DESCRIBE YOURSELF? | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY... | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| I belong to the ethnic majority in the country where I live | 3.714 | %52 | 3.533 | %50 | 307 | %4 | 111 | %2 | 825 | %12 | 316 | %4 | 352 | %5 | 7.125 | |
| I am part of an ethnic minority in the country where I live | 179 | %38 | 214 | %46 | 23 | %5 | 16 | %3 | 87 | %19 | 26 | %6 | 22 | %5 | 465 | |
| Other, specify... | 139 | %41 | 140 | %42 | 12 | %4 | 8 | %2 | 58 | %17 | 24 | %7 | 30 | %9 | 337 | |
| TOTAL | 4.032 | %51 | 3.887 | %49 | 342 | %4 | 135 | %2 | 970 | %12 | 366 | %5 | 404 | %5 | 7.927 | |

🟥 *Under-represented elements*　　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 79,7 ; dof= 12.*

**Cross: In which country do you live? / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?**

| IN WHICH COUNTRY DO YOU LIVE? | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY... | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Austria | 52 | %55 | 53 | %56 | 5 | %5 | 2 | %2 | 10 | %11 | 3 | %3 | 3 | %3 | 94 | |
| Belgium | 431 | %49 | 334 | %38 | 32 | %4 | 20 | %2 | 157 | %18 | 53 | %6 | 63 | %7 | 882 | |
| Bosnia and Herzegovina | 8 | %28 | 13 | %45 | 3 | %10 | 2 | %7 | 10 | %34 | 0 | %0 | 0 | %0 | 29 | |
| Bulgaria | 61 | %59 | 63 | %61 | 2 | %2 | 1 | %1 | 10 | %10 | 1 | %1 | 4 | %4 | 104 | |
| Croatia | 64 | %30 | 113 | %54 | 7 | %3 | 5 | %2 | 46 | %22 | 2 | %1 | 4 | %2 | 210 | |
| Cyprus | 25 | %35 | 31 | %44 | 2 | %3 | 4 | %6 | 23 | %32 | 5 | %7 | 1 | %1 | 71 | |
| Czech Republic | 64 | %32 | 103 | %52 | 4 | %2 | 11 | %6 | 38 | %19 | 11 | %6 | 7 | %4 | 199 | |
| Denmark | 189 | %53 | 189 | %53 | 20 | %6 | 3 | %1 | 43 | %12 | 17 | %5 | 24 | %7 | 356 | |
| Finland | 235 | %49 | 326 | %68 | 13 | %3 | 7 | %1 | 49 | %10 | 14 | %3 | 23 | %5 | 482 | |
| France | 544 | %60 | 331 | %37 | 33 | %4 | 15 | %2 | 113 | %12 | 63 | %7 | 51 | %6 | 906 | |
| Germany | 702 | %60 | 569 | %49 | 66 | %6 | 15 | %1 | 94 | %8 | 51 | %4 | 57 | %5 | 1.168 | |
| Greece | 83 | %45 | 96 | %52 | 5 | %3 | 9 | %5 | 24 | %13 | 6 | %3 | 8 | %4 | 183 | |
| Hungary | 75 | %46 | 94 | %58 | 3 | %2 | 3 | %2 | 10 | %6 | 8 | %5 | 3 | %2 | 162 | |
| Ireland | 32 | %30 | 75 | %71 | 2 | %2 | 1 | %1 | 13 | %12 | 5 | %5 | 1 | %1 | 105 | |
| Italy | 570 | %53 | 460 | %43 | 21 | %2 | 9 | %1 | 121 | %11 | 53 | %5 | 52 | %5 | 1.080 | |
| Latvia | 13 | %19 | 25 | %36 | 4 | %6 | 7 | %10 | 26 | %37 | 4 | %6 | 6 | %9 | 70 | |
| Luxembourg | 45 | %36 | 48 | %39 | 6 | %5 | 1 | %1 | 36 | %29 | 8 | %6 | 5 | %4 | 124 | |

🔴 *Under-represented elements*    🔵 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 1.071,6 ; dof= 192.*

Rare Barometer — A EURORDIS INITIATIVE

EURORDIS RARE DISEASES EUROPE

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Typology of countries based on size and welfare / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?**

| TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY... | | TOTAL |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Group A ('Eastern Europe') | 698 | %39 | 947 | %53 | 63 | %4 | 64 | %4 | 292 | %16 | 92 | %5 | 53 | %3 | 1.794 | |
| Group B ('Western Europe') | 2.857 | %56 | 2.217 | %43 | 230 | %5 | 70 | %1 | 550 | %11 | 253 | %5 | 235 | %5 | 5.105 | |
| Group C ('Northern Europe') | 1.632 | %50 | 1.704 | %52 | 130 | %4 | 50 | %2 | 422 | %13 | 177 | %5 | 206 | %6 | 3.273 | |
| **TOTAL** | 5.187 | %51 | 4.868 | %48 | 423 | %4 | 184 | %2 | 1.264 | %12 | 522 | %5 | 494 | %5 | 10.172 | |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 222,1 ; dof= 12.*

## Cross: Orphacode associated nomenclature (english) / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?

**ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE?**

| ORPHACODE ASSOCIATED NOMENCLATURE (ENGLISH) | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY... | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Hereditary hemorrhagic telangiectasia | 278 | %61 | 174 | %38 | 10 | %2 | 1 | %0 | 32 | %7 | 30 | %7 | 50 | %11 | 458 | |
| Hypermobile Ehlers-Danlos syndrome | 169 | %53 | 233 | %74 | 23 | %7 | 7 | %2 | 6 | %2 | 16 | %5 | 17 | %5 | 317 | |
| Sarcoidosis | 72 | %42 | 78 | %46 | 6 | %4 | 2 | %1 | 24 | %14 | 13 | %8 | 8 | %5 | 170 | |
| Classical Ehlers-Danlos syndrome | 62 | %45 | 88 | %64 | 9 | %7 | 2 | %1 | 8 | %6 | 10 | %7 | 7 | %5 | 137 | |
| Williams syndrome | 93 | %68 | 63 | %46 | 9 | %7 | 1 | %1 | 3 | %2 | 2 | %1 | 4 | %3 | 136 | |
| Cystic fibrosis | 80 | %63 | 78 | %61 | 10 | %8 | 1 | %1 | 2 | %2 | 10 | %8 | 4 | %3 | 128 | |
| Myasthenia gravis | 63 | %53 | 79 | %66 | 2 | %2 | 2 | %2 | 8 | %7 | 1 | %1 | 3 | %3 | 120 | |
| Systemic sclerosis | 60 | %56 | 68 | %64 | 3 | %3 | 1 | %1 | 8 | %7 | 5 | %5 | 5 | %5 | 107 | |
| Tuberous sclerosis complex | 50 | %51 | 37 | %38 | 3 | %3 | 2 | %2 | 12 | %12 | 7 | %7 | 3 | %3 | 98 | |
| Neurofibromatosis type 1 | 46 | %50 | 49 | %53 | 7 | %8 | 0 | %0 | 8 | %9 | 8 | %9 | 6 | %7 | 92 | |
| Interstitial cystitis | 48 | %65 | 22 | %30 | 2 | %3 | 1 | %1 | 8 | %11 | 3 | %4 | 6 | %8 | 74 | |
| Addison disease | 35 | %48 | 43 | %59 | 2 | %3 | 2 | %3 | 6 | %8 | 3 | %4 | 4 | %5 | 73 | |
| 22q11.2 deletion syndrome | 47 | %69 | 26 | %38 | 2 | %3 | 3 | %4 | 5 | %7 | 1 | %1 | 3 | %4 | 68 | |
| Chronic inflammatory demyelinating polyneuropathy | 33 | %51 | 31 | %48 | 4 | %6 | 0 | %0 | 8 | %12 | 6 | %9 | 4 | %6 | 65 | |
| Perineural cyst | 41 | %65 | 41 | %65 | 1 | %2 | 0 | %0 | 5 | %8 | 0 | %0 | 1 | %2 | 63 | |
| Acute inflammatory demyelinating polyradiculoneuropathy | 19 | %31 | 28 | %45 | 2 | %3 | 0 | %0 | 8 | %13 | 13 | %21 | 3 | %5 | 62 | |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 1.385,7 ; dof= 888.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Please select the sentence that best describes your situation or the situation of the person you care for: / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?**

| PLEASE SELECT THE SENTENCE THAT BEST DESCRIBES YOUR SITUATION OR THE SITUATION OF THE PERSON YOU CARE FOR: | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE | | | | | | | | | | | | | | TOTAL |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY... | | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N |
| I know the NAME of the rare disease, syndrome or malformation and it has been CONFIRMED by appropriate genetic, clinical, medical imaging, molecular or biochemical tests (e.g biopsy, blood or urine test) | 4.770 | %53 | 4.356 | %48 | 383 | %4 | 144 | %2 | 991 | %11 | 470 | %5 | 441 | %5 | 9.048 |
| I know the NAME of the rare disease, syndrome or malformation but it has NOT yet been confirmed by appropriate genetic, clinical, medical imaging, molecular or biochemical tests | 356 | %47 | 399 | %53 | 31 | %4 | 17 | %2 | 93 | %12 | 33 | %4 | 37 | %5 | 760 |
| I only have PARTIAL information on the name of the rare disease or the gene involved or the type of disease | 96 | %31 | 118 | %39 | 10 | %3 | 16 | %5 | 89 | %29 | 21 | %7 | 15 | %5 | 306 |
| I know that the disease is rare but the name or the cause have NOT BEEN IDENTIFIED | 98 | %28 | 111 | %32 | 11 | %3 | 12 | %3 | 133 | %38 | 20 | %6 | 16 | %5 | 348 |
| Other, specify... | 6 | %25 | 8 | %33 | 1 | %4 | 1 | %4 | 4 | %17 | 3 | %13 | 5 | %21 | 24 |
| **TOTAL** | 5.326 | %51 | 4.992 | %48 | 436 | %4 | 190 | %2 | 1.310 | %12 | 547 | %5 | 514 | %5 | 10.486 |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 416,1 ; dof= 24.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Genetic diseases / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?**

| GENETIC DISEASES | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY... | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Genetic diseases | 2.991 | %55 | 2.660 | %49 | 261 | %5 | 98 | %2 | 545 | %10 | 244 | %4 | 301 | %6 | 5.447 | |
| Non Genetic diseases | 1.290 | %49 | 1.274 | %48 | 81 | %3 | 40 | %2 | 327 | %12 | 158 | %6 | 105 | %4 | 2.627 | |
| **TOTAL** | **4.281** | **%53** | **3.934** | **%49** | **342** | **%4** | **138** | **%2** | **872** | **%11** | **402** | **%5** | **406** | **%5** | **8.074** | |

🟣 *Under-represented elements* 🔵 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 50,7 ; dof= 6.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Point prevalence of the rare disease / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?**

| POINT PREVALENCE OF THE RARE DISEASE | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY... | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| 1-5 / 10 000 | 1.376 | %57 | 1.196 | %50 | 108 | %4 | 27 | %1 | 186 | %8 | 124 | %5 | 150 | %6 | 2.407 | |
| 1-9 / 100 000 | 1.081 | %54 | 1.029 | %51 | 91 | %5 | 29 | %1 | 181 | %9 | 100 | %5 | 98 | %5 | 1.999 | |
| 1-9 / 1 000 000 | 246 | %54 | 228 | %50 | 17 | %4 | 11 | %2 | 51 | %11 | 20 | %4 | 23 | %5 | 459 | |
| <1 / 1 000 000 | 397 | %46 | 395 | %46 | 33 | %4 | 21 | %2 | 136 | %16 | 39 | %5 | 40 | %5 | 856 | |
| **TOTAL** | **3.100** | **%54** | **2.848** | **%50** | **249** | **%4** | **88** | **%2** | **554** | **%10** | **283** | **%5** | **311** | **%5** | **5.721** | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 79,0 ; dof= 18.*

**Cross: Disease complexity classified into five groups, based on the number of affected body parts. / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?**

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY... | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| 1-3 body parts | 3.045 | %50 | 2.656 | %44 | 225 | %4 | 94 | %2 | 836 | %14 | 375 | %6 | 306 | %5 | 6.103 | |
| 4-7 body parts | 1.639 | %53 | 1.517 | %49 | 142 | %5 | 66 | %2 | 349 | %11 | 128 | %4 | 141 | %5 | 3.081 | |
| 8-11 body parts | 476 | %50 | 581 | %61 | 45 | %5 | 23 | %2 | 97 | %10 | 30 | %3 | 43 | %5 | 951 | |
| 12-15 body parts | 135 | %47 | 194 | %68 | 18 | %6 | 5 | %2 | 22 | %8 | 9 | %3 | 20 | %7 | 286 | |
| 16 body parts or more | 31 | %48 | 44 | %68 | 6 | %9 | 2 | %3 | 6 | %9 | 5 | %8 | 4 | %6 | 65 | |
| TOTAL | 5.326 | %51 | 4.992 | %48 | 436 | %4 | 190 | %2 | 1.310 | %12 | 547 | %5 | 514 | %5 | 10.486 | |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 166,3 ; dof= 24.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: …behavioural disorders that cause problems in school, at home or in social situations / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?**

| …BEHAVIOURAL DISORDERS THAT CAUSE PROBLEMS IN SCHOOL, AT HOME OR IN SOCIAL SITUATIONS | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY... | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 1.546 | %52 | 1.329 | %45 | 156 | %5 | 81 | %3 | 350 | %12 | 124 | %4 | 160 | %5 | 2.957 | |
| No | 3.588 | %51 | 3.436 | %48 | 262 | %4 | 96 | %1 | 889 | %13 | 399 | %6 | 323 | %5 | 7.085 | |
| Don't know | 192 | %43 | 227 | %51 | 18 | %4 | 13 | %3 | 71 | %16 | 24 | %5 | 31 | %7 | 444 | |
| **TOTAL** | **5.326** | **%51** | **4.992** | **%48** | **436** | **%4** | **190** | **%2** | **1.310** | **%12** | **547** | **%5** | **514** | **%5** | **10.486** | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 71,7 ; dof= 12.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: …intellectual disabilities or cognitive symptoms (i.e. problems with memory, language, thinking or judgement) / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?**

| …INTELLECTUAL DISABILITIES OR COGNITIVE SYMPTOMS (I.E. PROBLEMS WITH MEMORY, LANGUAGE, THINKING OR JUDGEMENT) | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY… | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 1.489 | %51 | 1.418 | %48 | 149 | %5 | 82 | %3 | 379 | %13 | 108 | %4 | 141 | %5 | 2.936 | |
| No | 3.694 | %51 | 3.431 | %47 | 277 | %4 | 93 | %1 | 874 | %12 | 428 | %6 | 358 | %5 | 7.236 | |
| Don't know | 143 | %46 | 143 | %46 | 10 | %3 | 15 | %5 | 57 | %18 | 11 | %4 | 15 | %5 | 314 | |
| **TOTAL** | **5.326** | **%51** | **4.992** | **%48** | **436** | **%4** | **190** | **%2** | **1.310** | **%12** | **547** | **%5** | **514** | **%5** | **10.486** | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 84,3 ; dof= 12.*

Rare Barometer
A EURORDIS INITIATIVE

EURORDIS
RARE DISEASES EUROPE

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: …clinical signs or symptoms that come and go / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?**

| …CLINICAL SIGNS OR SYMPTOMS THAT COME AND GO | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | | | | | | | | | | | | |
| | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY... | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 3.063 | %52 | 3.046 | %51 | 255 | %4 | 115 | %2 | 667 | %11 | 270 | %5 | 272 | %5 | 5.940 | |
| No | 1.919 | %51 | 1.620 | %43 | 154 | %4 | 62 | %2 | 524 | %14 | 240 | %6 | 193 | %5 | 3.788 | |
| Don't know | 344 | %45 | 326 | %43 | 27 | %4 | 13 | %2 | 119 | %16 | 37 | %5 | 49 | %6 | 758 | |
| **TOTAL** | **5.326** | **%51** | **4.992** | **%48** | **436** | **%4** | **190** | **%2** | **1.310** | **%12** | **547** | **%5** | **514** | **%5** | **10.486** | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 85,1 ; dof= 12.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

**Cross: ...invisible symptoms such as pain, dizziness, headaches, fatigue, etc. / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?**

| ...INVISIBLE SYMPTOMS SUCH AS PAIN, DIZZINESS, HEADACHES, FATIGUE, ETC. | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY... | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 3.518 | %50 | 3.489 | %50 | 286 | %4 | 124 | %2 | 869 | %12 | 355 | %5 | 338 | %5 | 7.020 | |
| No | 1.558 | %53 | 1.238 | %42 | 119 | %4 | 46 | %2 | 353 | %12 | 167 | %6 | 148 | %5 | 2.916 | |
| Don't know | 250 | %45 | 265 | %48 | 31 | %6 | 20 | %4 | 88 | %16 | 25 | %5 | 28 | %5 | 550 | |
| **TOTAL** | 5.326 | %51 | 4.992 | %48 | 436 | %4 | 190 | %2 | 1.310 | %12 | 547 | %5 | 514 | %5 | 10.486 | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 53,0 ; dof= 12.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: ...sudden onset symptoms requiring urgent care / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?**

| ...SUDDEN ONSET SYMPTOMS REQUIRING URGENT CARE | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY... | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 2.391 | %51 | 2.272 | %49 | 199 | %4 | 77 | %2 | 600 | %13 | 207 | %4 | 207 | %4 | 4.648 | |
| No | 2.673 | %51 | 2.442 | %47 | 210 | %4 | 95 | %2 | 617 | %12 | 313 | %6 | 278 | %5 | 5.251 | |
| Don't know | 262 | %45 | 278 | %47 | 27 | %5 | 18 | %3 | 93 | %16 | 27 | %5 | 29 | %5 | 587 | |
| **TOTAL** | **5.326** | **%51** | **4.992** | **%48** | **436** | **%4** | **190** | **%2** | **1.310** | **%12** | **547** | **%5** | **514** | **%5** | **10.486** | |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 37,2 ; dof= 12.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

**Only respondents living with a diagnosed rare disease**

## Cross: The rare disease was diagnosed before birth / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?

| THE RARE DISEASE WAS DIAGNOSED BEFORE BIRTH | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY... | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 131 | %59 | 95 | %43 | 6 | %3 | 2 | %1 | 23 | %10 | 15 | %7 | 7 | %3 | 222 | |
| No | 4.953 | %52 | 4.623 | %49 | 406 | %4 | 155 | %2 | 1.052 | %11 | 485 | %5 | 464 | %5 | 9.509 | |
| TOTAL | 5.084 | %52 | 4.718 | %48 | 412 | %4 | 157 | %2 | 1.075 | %11 | 500 | %5 | 471 | %5 | 9.731 | |

🟣 *Under-represented elements*   🔵 *Over-represented elements*

*The relationship is not significant. p-value= 0,2 ; Chi2= 8,1 ; dof= 6.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

**Only respondents living with a diagnosed rare disease**

**Cross: The rare disease was diagnosed through standard tests carried out at birth / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?**

| THE RARE DISEASE WAS DIAGNOSED THROUGH STANDARD TESTS CARRIED OUT AT BIRTH | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY... | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 243 | %61 | 166 | %42 | 18 | %5 | 5 | %1 | 33 | %8 | 19 | %5 | 16 | %4 | 396 | |
| No | 4.736 | %52 | 4.473 | %49 | 383 | %4 | 148 | %2 | 1.013 | %11 | 469 | %5 | 445 | %5 | 9.139 | |
| **TOTAL** | **4.979** | **%52** | **4.639** | **%49** | **401** | **%4** | **153** | **%2** | **1.046** | **%11** | **488** | **%5** | **461** | **%5** | **9.535** | |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is significant. p-value= 0,0 ; Chi2= 14,1 ; dof= 6.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Only respondents living with a diagnosed rare disease**

**Cross: Family members were previously diagnosed with the same disease / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?**

| FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY... | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 751 | %57 | 576 | %44 | 64 | %5 | 14 | %1 | 112 | %9 | 71 | %5 | 117 | %9 | 1.309 | |
| No | 4.333 | %51 | 4.142 | %49 | 348 | %4 | 143 | %2 | 963 | %11 | 429 | %5 | 354 | %4 | 8.422 | |
| **TOTAL** | **5.084** | **%52** | **4.718** | **%48** | **412** | **%4** | **157** | **%2** | **1.075** | **%11** | **500** | **%5** | **471** | **%5** | **9.731** | |

■ *Under-represented elements*　　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 79,4 ; dof= 6.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

**Cross: I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?**

| I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | | | | | | | | | | | | | | | |
| | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY... | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 3.221 | %54 | 2.810 | %47 | 261 | %4 | 103 | %2 | 689 | %11 | 348 | %6 | 296 | %5 | 5.998 | |
| No | 2.071 | %47 | 2.150 | %49 | 170 | %4 | 86 | %2 | 615 | %14 | 197 | %4 | 215 | %5 | 4.415 | |
| TOTAL | 5.292 | %51 | 4.960 | %48 | 431 | %4 | 189 | %2 | 1.304 | %13 | 545 | %5 | 511 | %5 | 10.413 | |

■ *Under-represented elements*　　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 48,1 ; dof= 6.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

**Cross: …wrongly attributed to another physical disease? / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?**

| …WRONGLY ATTRIBUTED TO ANOTHER PHYSICAL DISEASE? | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY... | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, one time | 1.019 | %52 | 885 | %45 | 74 | %4 | 27 | %1 | 248 | %13 | 97 | %5 | 80 | %4 | 1.950 | |
| YES, several times | 2.209 | %49 | 2.385 | %53 | 201 | %4 | 96 | %2 | 567 | %13 | 184 | %4 | 209 | %5 | 4.520 | |
| NO | 2.098 | %52 | 1.722 | %43 | 161 | %4 | 67 | %2 | 495 | %12 | 266 | %7 | 225 | %6 | 4.016 | |
| TOTAL | 5.326 | %51 | 4.992 | %48 | 436 | %4 | 190 | %2 | 1.310 | %12 | 547 | %5 | 514 | %5 | 10.486 | |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 92,8 ; dof= 12.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: …neglected, not taken seriously and/or considered as psychological? / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?**

| …NEGLECTED, NOT TAKEN SERIOUSLY AND/OR CONSIDERED AS PSYCHOLOGICAL? | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY... | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, one time | 635 | %51 | 582 | %47 | 54 | %4 | 18 | %1 | 166 | %13 | 69 | %6 | 59 | %5 | 1.246 | |
| YES, several times | 2.419 | %49 | 2.608 | %53 | 223 | %5 | 100 | %2 | 599 | %12 | 202 | %4 | 230 | %5 | 4.934 | |
| NO | 2.272 | %53 | 1.802 | %42 | 159 | %4 | 72 | %2 | 545 | %13 | 276 | %6 | 225 | %5 | 4.306 | |
| **TOTAL** | **5.326** | **%51** | **4.992** | **%48** | **436** | **%4** | **190** | **%2** | **1.310** | **%12** | **547** | **%5** | **514** | **%5** | **10.486** | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 98,4 ; dof= 12.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Has the person affected by the rare disease already been misdiagnosed? Calculated variable that computes the number of times the person affected by the rare disease was misdiagnosed. / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?**

| HAS THE PERSON AFFECTED BY THE RARE DISEASE ALREADY BEEN MISDIAGNOSED? CALCULATED VARIABLE THAT COMPUTES THE NUMBER OF TIMES THE PERSON AFFECTED BY THE RARE DISEASE WAS MISDIAGNOSED. | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY... | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, one time | 1.389 | %52 | 1.264 | %47 | 103 | %4 | 42 | %2 | 325 | %12 | 142 | %5 | 128 | %5 | 2.683 | |
| YES, several times | **2.465** | **%49** | **2.602** | **%52** | 224 | %4 | 100 | %2 | 634 | %13 | **215** | **%4** | 233 | %5 | 5.018 | |
| NO | **1.472** | **%53** | **1.126** | **%40** | 109 | %4 | 48 | %2 | 351 | %13 | **190** | **%7** | 153 | %5 | 2.785 | |
| **TOTAL** | **5.326** | **%51** | **4.992** | **%48** | **436** | **%4** | **190** | **%2** | **1.310** | **%12** | **547** | **%5** | **514** | **%5** | **10.486** | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 84,3 ; dof= 12.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Genetic test(s) looking for genetic changes (also called mutations or variants) / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?**

| GENETIC TEST(S) LOOKING FOR GENETIC CHANGES (ALSO CALLED MUTATIONS OR VARIANTS) | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY... | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 2.898 | %53 | 2.576 | %47 | 254 | %5 | 122 | %2 | 704 | %13 | 235 | %4 | 278 | %5 | 5.490 | |
| No | 2.063 | %49 | 2.076 | %50 | 155 | %4 | 54 | %1 | 475 | %11 | 250 | %6 | 187 | %4 | 4.171 | |
| Don't know/don't remember | 365 | %44 | 340 | %41 | 27 | %3 | 14 | %2 | 131 | %16 | 62 | %8 | 49 | %6 | 825 | |
| **TOTAL** | 5.326 | %51 | 4.992 | %48 | 436 | %4 | 190 | %2 | 1.310 | %12 | 547 | %5 | 514 | %5 | 10.486 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 80,3 ; dof= 12.*

**Cross: …healthcare professionals were reluctant or not sufficiently informed? / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?**

| Have you ever needed a genetic test but could not access it because… ...HEALTHCARE PROFESSIONALS WERE RELUCTANT OR NOT SUFFICIENTLY INFORMED? | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY… | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 1.325 | %47 | 1.494 | %53 | 132 | %5 | 70 | %2 | 399 | %14 | 87 | %3 | 119 | %4 | 2.805 | |
| No | 2.930 | %53 | 2.502 | %45 | 237 | %4 | 81 | %1 | 661 | %12 | 322 | %6 | 274 | %5 | 5.556 | |
| Not relevant | 1.071 | %50 | 996 | %47 | 67 | %3 | 39 | %2 | 250 | %12 | 138 | %6 | 121 | %6 | 2.125 | |
| TOTAL | 5.326 | %51 | 4.992 | %48 | 436 | %4 | 190 | %2 | 1.310 | %12 | 547 | %5 | 514 | %5 | 10.486 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

The relationship is very significant. p-value= < 0,01 ; Chi2= 105,1 ; dof= 12.

Have you ever needed a genetic test but could not access it because…

**Cross: To your knowledge, the genetic test(s) that were conducted targeted… / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?**

| TO YOUR KNOWLEDGE, THE GENETIC TEST(S) THAT WERE CONDUCTED TARGETED… | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY… | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Only one gene | 860 | %59 | 681 | %47 | 67 | %5 | 20 | %1 | 154 | %11 | 55 | %4 | 75 | %5 | 1.460 | |
| Several genes at the same time (gene panel sequencing) | 942 | %54 | 858 | %50 | 91 | %5 | 39 | %2 | 235 | %14 | 44 | %3 | 82 | %5 | 1.731 | |
| The whole DNA (Whole Genome Sequencing) | 418 | %48 | 411 | %47 | 47 | %5 | 20 | %2 | 159 | %18 | 30 | %3 | 35 | %4 | 880 | |
| All the genes (Whole Exome Sequencing) | 247 | %44 | 293 | %52 | 27 | %5 | 12 | %2 | 111 | %20 | 16 | %3 | 24 | %4 | 567 | |
| A tumour (genetic profiling of a tumour) | 67 | %50 | 63 | %47 | 5 | %4 | 4 | %3 | 20 | %15 | 8 | %6 | 10 | %7 | 135 | |
| Other (epigenome, RNA, etc.) | 59 | %50 | 61 | %52 | 8 | %7 | 4 | %3 | 22 | %19 | 6 | %5 | 3 | %3 | 117 | |
| Don't know | 730 | %48 | 670 | %44 | 56 | %4 | 47 | %3 | 184 | %12 | 103 | %7 | 96 | %6 | 1.511 | |
| **TOTAL** | **2.898** | **%53** | **2.576** | **%47** | **254** | **%5** | **122** | **%2** | **704** | **%13** | **235** | **%4** | **278** | **%5** | **5.490** | |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 159,5 ; dof= 36.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: After the tests were performed, were you offered genetic counselling (e.g. given information about how your genetic condition might affect you and your family)? / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?**

| AFTER THE TESTS WERE PERFORMED, WERE YOU OFFERED GENETIC COUNSELLING (E.G. GIVEN INFORMATION ABOUT HOW YOUR GENETIC CONDITION MIGHT AFFECT YOU AND YOUR FAMILY)? | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY... | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, with a genetic counsellor or clinical geneticist | 1.161 | %54 | 1.014 | %47 | 100 | %5 | 44 | %2 | 271 | %13 | 89 | %4 | 109 | %5 | 2.137 | |
| YES, by a healthcare professional | 697 | %59 | 490 | %42 | 61 | %5 | 23 | %2 | 116 | %10 | 57 | %5 | 52 | %4 | 1.179 | |
| NO, I wasn't offered genetic counselling | 836 | %47 | 891 | %50 | 78 | %4 | 48 | %3 | 265 | %15 | 60 | %3 | 89 | %5 | 1.770 | |
| Not sure / Don't remember | 204 | %50 | 181 | %45 | 15 | %4 | 7 | %2 | 52 | %13 | 29 | %7 | 28 | %7 | 404 | |
| TOTAL | 2.898 | %53 | 2.576 | %47 | 254 | %5 | 122 | %2 | 704 | %13 | 235 | %4 | 278 | %5 | 5.490 | |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 68,0 ; dof= 18.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Genetic tests / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?

| GENETIC TESTS | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY... | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 399 | %48 | 414 | %50 | 48 | %6 | 27 | %3 | 125 | %15 | 32 | %4 | 33 | %4 | 831 | |
| No | 2.434 | %54 | 2.094 | %46 | 202 | %4 | 93 | %2 | 555 | %12 | 195 | %4 | 240 | %5 | 4.515 | |
| Don't know | 65 | %45 | 68 | %48 | 4 | %3 | 2 | %1 | 24 | %17 | 8 | %6 | 4 | %3 | 143 | |
| TOTAL | 2.898 | %53 | 2.576 | %47 | 254 | %5 | 122 | %2 | 704 | %13 | 235 | %4 | 277 | %5 | 5.489 | |

■ *Under-represented elements*　　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 27,3 ; dof= 12.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

**Cross: Other diagnostic tests such as clinical examination(s), medical imaging (MRI, scans…), biopsy, biochemical test(s) (blood or urine tests…), etc. / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?**

| OTHER DIAGNOSTIC TESTS SUCH AS CLINICAL EXAMINATION(S), MEDICAL IMAGING (MRI, SCANS…), BIOPSY, BIOCHEMICAL TEST(S) (BLOOD OR URINE TESTS…), ETC. | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY… | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 698 | %50 | 696 | %50 | 80 | %6 | 32 | %2 | 215 | %15 | 63 | %4 | 62 | %4 | 1.403 | |
| No | 4.029 | %51 | 3.789 | %48 | 320 | %4 | 127 | %2 | 962 | %12 | 414 | %5 | 390 | %5 | 7.908 | |
| Don't know | 77 | %45 | 77 | %45 | 2 | %1 | 5 | %3 | 23 | %14 | 13 | %8 | 5 | %3 | 170 | |
| **TOTAL** | **4.804** | **%51** | **4.562** | **%48** | **402** | **%4** | **164** | **%2** | **1.200** | **%13** | **490** | **%5** | **457** | **%5** | **9.481** | |

🟣 *Under-represented elements*  🔵 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 33,2 ; dof= 12.*

**Cross: Additional advice from a healthcare professional specialised in the rare disease (in person or virtually) / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?**

| ADDITIONAL ADVICE FROM A HEALTHCARE PROFESSIONAL SPECIALISED IN THE RARE DISEASE (IN PERSON OR VIRTUALLY) | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY... | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 1.136 | %55 | 1.099 | %53 | 105 | %5 | 51 | %2 | 245 | %12 | 76 | %4 | 76 | %4 | 2.083 | |
| No | 4.096 | %50 | 3.799 | %46 | 326 | %4 | 130 | %2 | 1.029 | %13 | 453 | %6 | 428 | %5 | 8.194 | |
| Don't know | 94 | %45 | 94 | %45 | 5 | %2 | 9 | %4 | 36 | %17 | 18 | %9 | 10 | %5 | 209 | |
| **TOTAL** | **5.326** | **%51** | **4.992** | **%48** | **436** | **%4** | **190** | **%2** | **1.310** | **%12** | **547** | **%5** | **514** | **%5** | **10.486** | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 72,0 ; dof= 12.*

**Cross: …psychological support / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?**

| …PSYCHOLOGICAL SUPPORT | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY... | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| YES and enough to meet my needs | 512 | %56 | 435 | %47 | 49 | %5 | 15 | %2 | 104 | %11 | 47 | %5 | 52 | %6 | 922 | |
| YES but it is/was not needed | 511 | %54 | 428 | %45 | 45 | %5 | 14 | %1 | 108 | %11 | 65 | %7 | 48 | %5 | 955 | |
| YES but NOT enough to meet my needs | 471 | %49 | 471 | %49 | 41 | %4 | 25 | %3 | 123 | %13 | 40 | %4 | 45 | %5 | 952 | |
| NO but it is/was NOT needed | 1.625 | %51 | 1.427 | %45 | 109 | %3 | 49 | %2 | 377 | %12 | 218 | %7 | 165 | %5 | 3.165 | |
| NO but it is/was needed | 2.207 | %49 | 2.231 | %50 | 192 | %4 | 87 | %2 | 598 | %13 | 177 | %4 | 204 | %5 | 4.492 | |
| TOTAL | 5.326 | %51 | 4.992 | %48 | 436 | %4 | 190 | %2 | 1.310 | %12 | 547 | %5 | 514 | %5 | 10.486 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 78,6 ; dof= 24.*

Rare Barometer
A EURORDIS INITIATIVE

EURORDIS
RARE DISEASES EUROPE

**Cross: …care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc. / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?**

| …CARE COORDINATION SUPPORT SUCH AS HELP TO FIND THE NECESSARY INFORMATION ON THE DISEASE AND THE RIGHT PROFESSIONALS, ARRANGING APPOINTMENTS WITH DIFFERENT HEALTH PROVIDERS, ETC. | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | YES, THROUGH A PATIENT ORGANISATION | | YES, THROUGH ONLINE COMMUNITIES | | YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS) | | NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS) | | NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE | | NO, BECAUSE I DON'T WANT TO | | OTHER, SPECIFY... | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % | N | % | N | % |
| YES and enough to meet my needs | 1.215 | %58 | 870 | %42 | 99 | %5 | 29 | %1 | 216 | %10 | 135 | %6 | 97 | %5 | 2.083 | |
| YES but it is/was not needed | 200 | %51 | 171 | %44 | 16 | %4 | 5 | %1 | 45 | %12 | 32 | %8 | 16 | %4 | 391 | |
| YES but NOT enough to meet my needs | 788 | %54 | 680 | %46 | 63 | %4 | 32 | %2 | 183 | %13 | 62 | %4 | 73 | %5 | 1.463 | |
| NO but it is/was NOT needed | 770 | %47 | 714 | %44 | 51 | %3 | 18 | %1 | 204 | %13 | 148 | %9 | 100 | %6 | 1.627 | |
| NO but it is/was needed | 2.353 | %48 | 2.557 | %52 | 207 | %4 | 106 | %2 | 662 | %13 | 170 | %3 | 228 | %5 | 4.922 | |
| TOTAL | 5.326 | %51 | 4.992 | %48 | 436 | %4 | 190 | %2 | 1.310 | %12 | 547 | %5 | 514 | %5 | 10.486 | |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 206,7 ; dof= 24.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: …financial support including social security benefits / Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease?**

| …FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS | ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | **YES, THROUGH A PATIENT ORGANISATION** | **YES, THROUGH ONLINE COMMUNITIES** | **YES, THROUGH LOCAL NETWORKS (E.G. SCHOOLS)** | **NO, BECAUSE OF ACCESSIBILITY ISSUES (E.G. LANGUAGE OR TECHNICAL BARRIERS)** | **NO, BECAUSE I HAVE NOT BEEN ABLE TO FIND OTHER PEOPLE WITH THE SAME DISEASE** | **NO, BECAUSE I DON'T WANT TO** | **OTHER, SPECIFY…** | **TOTAL** |
| YES and enough to meet my needs | %56 | %44 | %4 | %2 | %13 | %6 | %4 | |
| YES but it is/was not needed | %58 | %40 | %5 | %1 | %12 | %7 | %5 | |
| YES but NOT enough to meet my needs | %49 | %48 | %5 | %4 | %14 | %4 | %5 | |
| NO but it is/was NOT needed | %53 | %45 | %3 | %1 | %11 | %6 | %5 | |
| NO but it is/was needed | %47 | %51 | %5 | %2 | %13 | %4 | %5 | |
| **TOTAL** | **%51** | **%48** | **%4** | **%2** | **%13** | **%5** | **%5** | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 148,2 ; dof= 24.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

# Chapter 15.

## Consequence of being diagnosed

Rare
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EURORDIS
RARE DISEASES EUROPE

# 9. Consequences of diagnosis

Questions asked only to respondents who are diagnosed

**Since receiving a diagnosis for the rare disease, how have the following aspects changed for you?**

| | …HAS GOTTEN WORSE | …HAS IMPROVED | …HAS REMAINED THE SAME | DON'T KNOW | NOT RELEVANT | TOTAL |
|---|---|---|---|---|---|---|
| Access to the most adapted care, treatments or surgery… | 889 | 4.020 | 3.316 | 457 | 342 | 9.024 |
| Understanding how the disease will progress… | 694 | 4.999 | 2.644 | 494 | 175 | 9.006 |
| Financial support including social security benefits… | 928 | 1.056 | 2.345 | 532 | 1.126 | 5.987 |
| Integration at school… | 746 | 960 | 2.030 | 613 | 4.675 | 9.024 |
| Integration at work… | 2.411 | 818 | 2.587 | 551 | 2.638 | 9.005 |
| Access to social services (e.g. social worker support, household chores support)… | 1.134 | 1.011 | 2.906 | 1.066 | 2.887 | 9.004 |
| Access to clinical trials… | 637 | 2.197 | 3.173 | 1.564 | 1.452 | 9.023 |
| Access to financial products, such as loans, mortgages, insurance… | 1.715 | 200 | 2.345 | 1.648 | 3.114 | 9.022 |
| Your social life… | 4.571 | 708 | 3.064 | 183 | 478 | 9.004 |

**Since receiving a diagnosis for the rare disease, how have the following aspects changed for you?**

| | …has gotten worse | …has improved | …has remained the same | Don't know | Not relevant |
|---|---|---|---|---|---|
| Access to the most adapted care, treatments or surgery… | %10 | %45 | %37 | %5 | %4 |
| Understanding how the disease will progress… | %8 | %56 | %29 | %5 | %2 |
| Financial support including social security benefits… | %16 | %18 | %39 | %9 | %19 |
| Integration at school… | %8 | %11 | %22 | %7 | %52 |
| Integration at work… | %27 | %9 | %29 | %6 | %29 |
| Access to social services (e.g. social worker support, household chores support)… | %13 | %11 | %32 | %12 | %32 |
| Access to clinical trials… | %7 | %24 | %35 | %17 | %16 |
| Access to financial products, such as loans, mortgages, insurance… | %19 | %2 | %26 | %18 | %35 |
| Your social life… | %51 | %8 | %34 | %2 | %5 |

● …has gotten worse  ● …has improved  ● …has remained the same  ● Don't know  ● Not relevant

Rare Barometer
A EURORDIS INITIATIVE

EURORDIS
RARE DISEASES EUROPE

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

**Questions asked only to respondents who are diagnosed**

**Since receiving a diagnosis for the rare disease, how have the following aspects changed for you?**

| Access to the most adapted care, treatments or surgery… | | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|---|
| | | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| …has gotten worse | | 0,4 | 647 | 3,8 | 569 | 3,3 | 328 | 3,6 | 679 | 4,8 | 535 |
| …has improved | | 0,4 | 3.169 | 3,9 | 3.210 | 4,3 | 1.897 | 3,8 | 3.347 | 5,2 | 2.918 |
| …has remained the same | | 0,5 | 2.526 | 3,1 | 2.293 | 3,4 | 1.353 | 3,3 | 2.659 | 4,4 | 2.285 |
| Don't know | | 1,2 | 299 | 2,4 | 257 | 2,3 | 143 | 2,7 | 315 | 3,0 | 244 |
| Not relevant | | 0,3 | 239 | 2,6 | 184 | 2,5 | 110 | 2,8 | 251 | 4,4 | 195 |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is not significant. p-value= 0,4 ; Fisher= 0,9.*

*Inter variance= 41,0. Intra variance= 44,2.*

Rare Barometer
A EURORDIS INITIATIVE

EURORDIS
RARE DISEASES EUROPE

Questions asked only to respondents who are diagnosed

**Since receiving a diagnosis for the rare disease, how have the following aspects changed for you?**

| Understanding how the disease will progress… | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| …has gotten worse | 0,3 | 494 | 3,0 | 447 | 3,0 | 238 | 3,0 | 533 | 4,2 | 415 |
| …has improved | 0,4 | 3.948 | 3,7 | 3.847 | 4,0 | 2.305 | 3,7 | 4.177 | 4,9 | 3.616 |
| …has remained the same | 0,7 | 1.979 | 3,4 | 1.825 | 3,6 | 1.063 | 3,3 | 2.052 | 4,8 | 1.764 |
| Don't know | 0,6 | 331 | 3,4 | 286 | 3,0 | 158 | 3,5 | 356 | 4,6 | 283 |
| Not relevant | -0,1 | 118 | **1,0** | 97 | **1,7** | 61 | 2,8 | 121 | 2,8 | 91 |

🟥 *Under-represented elements*　　🟦 *Over-represented elements*

*The relationship is not significant. p-value= 0,4 ; Fisher= 1,0.*
*Inter variance= 44,6. Intra variance= 44,3.*

**Questions asked only to respondents who are diagnosed**

**Since receiving a diagnosis for the rare disease, how have the following aspects changed for you?**

| Financial support including social security benefits… | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| …has gotten worse | 0,4 | 683 | 4,2 | 644 | 5,2 | 347 | 4,6 | 707 | 5,8 | 551 |
| …has improved | 0,8 | 845 | 5,2 | 827 | 5,6 | 488 | 5,2 | 862 | 6,8 | 761 |
| …has remained the same | 0,7 | 1.817 | 4,6 | 1.731 | 5,1 | 999 | 4,7 | 1.886 | 6,3 | 1.610 |
| Don't know | 0,7 | 381 | 3,8 | 364 | 4,3 | 215 | 3,1 | 398 | 4,4 | 310 |
| Not relevant | 0,4 | 816 | 3,7 | 800 | 3,9 | 448 | 3,2 | 878 | 4,8 | 746 |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is not significant. p-value= 0,8 ; Fisher= 0,4.*
*Inter variance= 27,2. Intra variance= 60,8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Questions asked only to respondents who are diagnosed

## Since receiving a diagnosis for the rare disease, how have the following aspects changed for you?

| Integration at school… | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| …has gotten worse | 0,1 | 579 | 2,8 | 534 | 3,1 | 302 | 2,8 | 602 | 3,9 | 508 |
| …has improved | 0,1 | 728 | 2,8 | 702 | 2,6 | 418 | 2,8 | 795 | 3,7 | 671 |
| …has remained the same | 0,5 | 1.558 | 3,1 | 1.421 | 3,1 | 871 | 3,5 | 1.624 | 4,8 | 1.410 |
| Don't know | 0,8 | 428 | 2,7 | 394 | 2,6 | 247 | 2,0 | 460 | 3,4 | 376 |
| Not relevant | 0,5 | 3.587 | 4,0 | 3.462 | 4,5 | 1.993 | 4,1 | 3.770 | 5,2 | 3.212 |

■ *Under-represented elements* ■ *Over-represented elements*

*The relationship is not significant. p-value= 0,3 ; Fisher= 1,3.*

*Inter variance= 57,0. Intra variance= 44,2.*

Rare Barometer
A EURORDIS INITIATIVE

EURORDIS
RARE DISEASES EUROPE

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

Questions asked only to respondents who are diagnosed

**Since receiving a diagnosis for the rare disease, how have the following aspects changed for you?**

| Integration at work… | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| …has gotten worse | 0,2 | 1.901 | 3,5 | 1.793 | 3,6 | 1.004 | 3,4 | 1.989 | 4,5 | 1.642 |
| …has improved | 0,5 | 663 | 3,9 | 639 | 4,2 | 370 | **4,5** | 683 | 5,5 | 593 |
| …has remained the same | 0,6 | 2.006 | 3,5 | 1.858 | 3,4 | 1.122 | 3,6 | 2.091 | 4,9 | 1.840 |
| Don't know | 0,5 | 355 | **2,7** | 338 | 3,2 | 204 | **2,1** | 398 | **3,4** | 323 |
| Not relevant | 0,5 | 1.945 | 3,5 | 1.874 | 4,3 | 1.125 | 3,6 | 2.078 | 4,7 | 1.771 |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is not significant. p-value= 0,5 ; Fisher= 0,9.*

*Inter variance= 37,7. Intra variance= 44,3.*

**Questions asked only to respondents who are diagnosed**

**Since receiving a diagnosis for the rare disease, how have the following aspects changed for you?**

| Access to social services (e.g. social worker support, household chores support)… | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| …has gotten worse | 0,2 | 846 | 3,1 | 772 | 3,7 | 426 | 3,5 | 867 | 4,5 | 716 |
| …has improved | 0,1 | 795 | 3,7 | 781 | 3,6 | 483 | 3,7 | 856 | 4,6 | 755 |
| …has remained the same | 0,6 | 2.281 | 3,6 | 2.105 | 3,9 | 1.211 | 4,2 | 2.392 | 5,4 | 2.051 |
| Don't know | 0,6 | 785 | 3,4 | 743 | 4,1 | 444 | 2,7 | 828 | 4,2 | 667 |
| Not relevant | 0,5 | 2.162 | 3,6 | 2.100 | 3,6 | 1.261 | 3,1 | 2.295 | 4,4 | 1.979 |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is not significant. p-value= 0,2 ; Fisher= 1,4.*
*Inter variance= 61,4. Intra variance= 44,3.*

**Questions asked only to respondents who are diagnosed**

**Since receiving a diagnosis for the rare disease, how have the following aspects changed for you?**

| Access to clinical trials… | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | **MEAN** | **N** | **MEAN** | **N** | **MEAN** | **N** | **MEAN** | **N** | **MEAN** | **N** |
| …has gotten worse | 0,0 | 456 | 3,3 | 398 | 2,9 | 212 | 2,9 | 487 | 4,7 | 384 |
| …has improved | 0,1 | 1.727 | 4,0 | 1.692 | 4,2 | 1.047 | 3,8 | 1.834 | 5,6 | 1.599 |
| …has remained the same | 0,6 | 2.474 | 3,5 | 2.327 | 3,7 | 1.315 | 3,7 | 2.577 | 4,7 | 2.234 |
| Don't know | 0,4 | 1.157 | 3,2 | 1.077 | 3,2 | 619 | 3,4 | 1.214 | 4,0 | 1.007 |
| Not relevant | 1,1 | 1.065 | 3,1 | 1.018 | 3,9 | 638 | 3,3 | 1.138 | 4,3 | 952 |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Fisher= 4,4.*
*Inter variance= 193,2. Intra variance= 44,1.*

Rare Barometer
A EURORDIS INITIATIVE

EURORDIS
RARE DISEASES EUROPE

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

Questions asked only to respondents who are diagnosed

Since receiving a diagnosis for the rare disease, how have the following aspects changed for you?

| Access to financial products, such as loans, mortgages, insurance… | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| …has gotten worse | 0,4 | 1.317 | 4,0 | 1.261 | 4,7 | 725 | 4,1 | 1.377 | 5,7 | 1.179 |
| …has improved | 0,1 | 157 | 2,3 | 150 | 3,6 | 89 | 2,7 | 161 | 3,6 | 144 |
| …has remained the same | 0,6 | 1.844 | 3,7 | 1.701 | 4,1 | 967 | 4,0 | 1.910 | 5,3 | 1.625 |
| Don't know | 0,5 | 1.239 | 3,2 | 1.152 | 3,2 | 723 | 3,0 | 1.316 | 4,0 | 1.102 |
| Not relevant | 0,4 | 2.322 | 3,3 | 2.248 | 3,3 | 1.327 | 3,3 | 2.486 | 4,2 | 2.126 |

■ *Under-represented elements*　　■ *Over-represented elements*

The relationship is not significant. p-value= 0,8 ; Fisher= 0,4.

Inter variance= 15,9. Intra variance= 44,3.

Rare Barometer
A EURORDIS 'X' INITIATIVE

EURORDIS
RARE DISEASES EUROPE

Questions asked only to respondents who are diagnosed

**Since receiving a diagnosis for the rare disease, how have the following aspects changed for you?**

| Your social life… | TIME BETWEEN FIRST SYMPTOMS AND FIRST MEDICAL CONTACT, IN YEARS | | TIME BETWEEN FIRST SYMPTOM AND FIRST SYMPTOMATIC TREATMENT, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND FIRST REFERRAL TO A CENTRE OF EXPERTISE, IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND INITIAL DIAGNOSIS (FIRST HEARING THE NAME OF THE DISEASE), IN YEARS | | TIME BETWEEN FIRST SYMPTOMS AND CONFIRMED DIAGNOSIS, IN YEARS | |
|---|---|---|---|---|---|---|---|---|---|---|
| | MEAN | N | MEAN | N | MEAN | N | MEAN | N | MEAN | N |
| …has gotten worse | 0,4 | 3.556 | 3,4 | 3.342 | 3,4 | 1.898 | 3,2 | 3.728 | **4,1** | 3.126 |
| …has improved | 0,2 | 555 | **4,7** | 543 | **5,2** | 325 | **5,0** | 575 | **6,6** | 490 |
| …has remained the same | 0,7 | 2.320 | 3,7 | 2.209 | 4,2 | 1.350 | 4,0 | 2.454 | **5,5** | 2.156 |
| Don't know | 0,7 | 117 | 2,1 | 110 | 3,8 | 66 | 3,2 | 127 | 4,0 | 108 |
| Not relevant | **-0,4** | 321 | **1,7** | 297 | **1,9** | 186 | **1,9** | 354 | **3,0** | 288 |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is weakly significant. p-value= 0,1 ; Fisher= 2,3.*
*Inter variance= 101,9. Intra variance= 44,3.*

Rare Barometer
A EURORDIS INITIATIVE

EURORDIS
RARE DISEASES EUROPE

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Gender of the person affected by the rare disease / Access to the most adapted care, treatments or surgery…**

| GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE | ACCESS TO THE MOST ADAPTED CARE, TREATMENTS OR SURGERY… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Female | 603 | %10 | 2.638 | %44 | 2.267 | %38 | 299 | %5 | 216 | %4 | 6.023 | %100 |
| Male | 250 | %10 | 1.220 | %46 | 917 | %35 | 130 | %5 | 107 | %4 | 2.624 | %100 |
| Other | 11 | %13 | 29 | %35 | 30 | %37 | 6 | %7 | 6 | %7 | 82 | %100 |
| **TOTAL** | **864** | **%10** | **3.887** | **%45** | **3.214** | **%37** | **435** | **%5** | **329** | **%4** | **8.729** | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is weakly significant. p-value= 0,1 ; Chi2= 14,5 ; dof= 8.*

**Cross: Gender of the person affected by the rare disease / Understanding how the disease will progress…**

| GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE | UNDERSTANDING HOW THE DISEASE WILL PROGRESS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Female | 474 | %8 | 3.250 | %54 | 1.850 | %31 | 346 | %6 | 103 | %2 | 6.023 | %100 |
| Male | 189 | %7 | 1.572 | %60 | 682 | %26 | 124 | %5 | 57 | %2 | 2.624 | %100 |
| Other | 7 | %9 | 35 | %43 | 32 | %39 | 4 | %5 | 4 | %5 | 82 | %100 |
| **TOTAL** | **670** | **%8** | **4.857** | **%56** | **2.564** | **%29** | **474** | **%5** | **164** | **%2** | **8.729** | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 41,4 ; dof= 8.*

**Cross: Gender of the person affected by the rare disease / Financial support including social security benefits…**

| GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE | FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Female | 748 | %16 | 828 | %18 | 1.846 | %39 | 438 | %9 | 839 | %18 | 4.699 | %100 |
| Male | 165 | %14 | 213 | %18 | 461 | %38 | 85 | %7 | 275 | %23 | 1.199 | %100 |
| Other | 7 | %21 | 4 | %12 | 13 | %38 | 4 | %12 | 6 | %18 | 34 | %100 |
| **TOTAL** | **920** | **%16** | **1.045** | **%18** | **2.320** | **%39** | **527** | **%9** | **1.120** | **%19** | **5.932** | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 23,1 ; dof= 8.*

**Cross: Gender of the person affected by the rare disease / Integration at school…**

| GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE | INTEGRATION AT SCHOOL… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Female | 475 | %8 | 497 | %8 | 1.241 | %21 | 416 | %7 | 3.394 | %56 | 6.023 | %100 |
| Male | 229 | %9 | 387 | %15 | 677 | %26 | 159 | %6 | 1.172 | %45 | 2.624 | %100 |
| Other | 14 | %17 | 13 | %16 | 24 | %29 | 6 | %7 | 25 | %30 | 82 | %100 |
| **TOTAL** | **718** | **%8** | **897** | **%10** | **1.942** | **%22** | **581** | **%7** | **4.591** | **%53** | **8.729** | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 168,2 ; dof= 8.*

## Cross: Gender of the person affected by the rare disease / Integration at work…

| GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE | INTEGRATION AT WORK… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Female | 1.711 | %28 | 552 | %9 | 1.691 | %28 | 332 | %6 | 1.737 | %29 | 6.023 | %100 |
| Male | 611 | %23 | 239 | %9 | 804 | %31 | 170 | %6 | 799 | %30 | 2.623 | %100 |
| Other | 20 | %24 | 4 | %5 | 20 | %24 | 12 | %15 | 26 | %32 | 82 | %100 |
| TOTAL | 2.342 | %27 | 795 | %9 | 2.515 | %29 | 514 | %6 | 2.562 | %29 | 8.728 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 39,9 ; dof= 8.*

## Cross: Gender of the person affected by the rare disease / Access to social services (e.g. social worker support, household chores support)…

| GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE | ACCESS TO SOCIAL SERVICES (E.G. SOCIAL WORKER SUPPORT, HOUSEHOLD CHORES SUPPORT)… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Female | 777 | %13 | 619 | %10 | 1.946 | %32 | 749 | %12 | 1.932 | %32 | 6.023 | %100 |
| Male | 299 | %11 | 350 | %13 | 837 | %32 | 260 | %10 | 877 | %33 | 2.623 | %100 |
| Other | 15 | %18 | 8 | %10 | 29 | %35 | 10 | %12 | 20 | %24 | 82 | %100 |
| TOTAL | 1.091 | %13 | 977 | %11 | 2.812 | %32 | 1.019 | %12 | 2.829 | %32 | 8.728 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 34,0 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Gender of the person affected by the rare disease / Access to clinical trials…

| GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE | ACCESS TO CLINICAL TRIALS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Female | 449 | %7 | 1.397 | %23 | 2.106 | %35 | 1.108 | %18 | 963 | %16 | 6.023 | %100 |
| Male | 167 | %6 | 707 | %27 | 930 | %35 | 389 | %15 | 431 | %16 | 2.624 | %100 |
| Other | 4 | %5 | 18 | %22 | 30 | %37 | 17 | %21 | 13 | %16 | 82 | %100 |
| TOTAL | 620 | %7 | 2.122 | %24 | 3.066 | %35 | 1.514 | %17 | 1.407 | %16 | 8.729 | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 28,8 ; dof= 8.*

## Cross: Gender of the person affected by the rare disease / Access to financial products, such as loans, mortgages, insurance…

| GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE | ACCESS TO FINANCIAL PRODUCTS, SUCH AS LOANS, MORTGAGES, INSURANCE… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Female | 1.207 | %20 | 108 | %2 | 1.541 | %26 | 1.113 | %18 | 2.054 | %34 | 6.023 | %100 |
| Male | 438 | %17 | 81 | %3 | 722 | %28 | 435 | %17 | 947 | %36 | 2.623 | %100 |
| Other | 22 | %27 | 0 | %0 | 12 | %15 | 27 | %33 | 21 | %26 | 82 | %100 |
| TOTAL | 1.667 | %19 | 189 | %2 | 2.275 | %26 | 1.575 | %18 | 3.022 | %35 | 8.728 | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 53,9 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Gender of the person affected by the rare disease / Your social life…

| GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE | YOUR SOCIAL LIFE… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Female | 3.194 | %53 | 444 | %7 | 2.004 | %33 | 110 | %2 | 271 | %4 | 6.023 | %100 |
| Male | 1.200 | %46 | 238 | %9 | 942 | %36 | 59 | %2 | 184 | %7 | 2.623 | %100 |
| Other | 43 | %52 | 3 | %4 | 24 | %29 | 3 | %4 | 9 | %11 | 82 | %100 |
| TOTAL | 4.437 | %51 | 685 | %8 | 2.970 | %34 | 172 | %2 | 464 | %5 | 8.728 | %100 |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 61,6 ; dof= 8.*

## Cross: How old were you when you stopped full-time education? / Access to the most adapted care, treatments or surgery…

| HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION? | ACCESS TO THE MOST ADAPTED CARE, TREATMENTS OR SURGERY… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 15 y.o. or under | 59 | %14 | 183 | %44 | 130 | %31 | 25 | %6 | 16 | %4 | 413 | %100 |
| between 16 and 19 y.o. | 260 | %11 | 961 | %42 | 840 | %37 | 147 | %6 | 75 | %3 | 2.283 | %100 |
| between 20 and 23 y.o. | 257 | %9 | 1.271 | %45 | 1.070 | %38 | 109 | %4 | 117 | %4 | 2.824 | %100 |
| 24 y.o. or above | 261 | %9 | 1.314 | %45 | 1.072 | %37 | 140 | %5 | 116 | %4 | 2.903 | %100 |
| TOTAL | 837 | %10 | 3.729 | %44 | 3.112 | %37 | 421 | %5 | 324 | %4 | 8.423 | %100 |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 45,4 ; dof= 12.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: How old were you when you stopped full-time education? / Understanding how the disease will progress…

| HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION? | UNDERSTANDING HOW THE DISEASE WILL PROGRESS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 15 y.o. or under | 40 | %10 | **205** | **%50** | 130 | %31 | 24 | %6 | **14** | **%3** | 413 | %100 |
| between 16 and 19 y.o. | **216** | **%9** | **1.179** | **%52** | **710** | **%31** | 140 | %6 | 38 | %2 | 2.283 | %100 |
| between 20 and 23 y.o. | 203 | %7 | 1.570 | %56 | 844 | %30 | 152 | %5 | 55 | %2 | 2.824 | %100 |
| 24 y.o. or above | **189** | **%7** | **1.731** | **%60** | **780** | **%27** | 149 | %5 | 54 | %2 | 2.903 | %100 |
| **TOTAL** | **648** | **%8** | **4.685** | **%56** | **2.464** | **%29** | **465** | **%6** | **161** | **%2** | **8.423** | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 52,6 ; dof= 12.*

## Cross: How old were you when you stopped full-time education? / Financial support including social security benefits…

| HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION? | FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 15 y.o. or under | 44 | %20 | 31 | %14 | 87 | %40 | 22 | %10 | 35 | %16 | 219 | %100 |
| between 16 and 19 y.o. | 275 | %17 | 269 | %16 | **600** | **%37** | **170** | **%10** | 321 | %20 | 1.635 | %100 |
| between 20 and 23 y.o. | 274 | %15 | 326 | %17 | 762 | %40 | 158 | %8 | 367 | %19 | 1.887 | %100 |
| 24 y.o. or above | 278 | %15 | **364** | **%19** | 743 | %39 | 155 | %8 | 349 | %18 | 1.889 | %100 |
| **TOTAL** | **871** | **%15** | **990** | **%18** | **2.192** | **%39** | **505** | **%9** | **1.072** | **%19** | **5.630** | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is significant. p-value= 0,0 ; Chi2= 23,6 ; dof= 12.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: How old were you when you stopped full-time education? / Integration at school…

| HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION? | INTEGRATION AT SCHOOL… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 15 y.o. or under | 54 | %13 | 62 | %15 | 91 | %22 | 40 | %10 | 166 | %40 | 413 | %100 |
| between 16 and 19 y.o. | 174 | %8 | 200 | %9 | 435 | %19 | 197 | %9 | 1.277 | %56 | 2.283 | %100 |
| between 20 and 23 y.o. | 226 | %8 | 283 | %10 | 628 | %22 | 161 | %6 | 1.526 | %54 | 2.824 | %100 |
| 24 y.o. or above | 250 | %9 | 339 | %12 | 735 | %25 | 159 | %5 | 1.420 | %49 | 2.903 | %100 |
| **TOTAL** | **704** | **%8** | **884** | **%10** | **1.889** | **%22** | **557** | **%7** | **4.389** | **%52** | **8.423** | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 109,2 ; dof= 12.*

## Cross: How old were you when you stopped full-time education? / Integration at work…

| HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION? | INTEGRATION AT WORK… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 15 y.o. or under | 117 | %28 | 22 | %5 | 98 | %24 | 37 | %9 | 139 | %34 | 413 | %100 |
| between 16 and 19 y.o. | 673 | %29 | 185 | %8 | 598 | %26 | 153 | %7 | 674 | %30 | 2.283 | %100 |
| between 20 and 23 y.o. | 696 | %25 | 262 | %9 | 833 | %29 | 141 | %5 | 892 | %32 | 2.824 | %100 |
| 24 y.o. or above | 763 | %26 | 300 | %10 | 905 | %31 | 169 | %6 | 766 | %26 | 2.903 | %100 |
| **TOTAL** | **2.249** | **%27** | **769** | **%9** | **2.434** | **%29** | **500** | **%6** | **2.471** | **%29** | **8.423** | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 69,7 ; dof= 12.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: How old were you when you stopped full-time education? / Access to social services (e.g. social worker support, household chores support)…**

| HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION? | ACCESS TO SOCIAL SERVICES (E.G. SOCIAL WORKER SUPPORT, HOUSEHOLD CHORES SUPPORT)… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 15 y.o. or under | 65 | %16 | 53 | %13 | 133 | %32 | 46 | %11 | 116 | %28 | 413 | %100 |
| between 16 and 19 y.o. | 341 | %15 | 221 | %10 | 690 | %30 | 315 | %14 | 716 | %31 | 2.283 | %100 |
| between 20 and 23 y.o. | 316 | %11 | 299 | %11 | 929 | %33 | 289 | %10 | 991 | %35 | 2.824 | %100 |
| 24 y.o. or above | 331 | %11 | 370 | %13 | 967 | %33 | 331 | %11 | 904 | %31 | 2.903 | %100 |
| **TOTAL** | **1.053** | **%13** | **943** | **%11** | **2.719** | **%32** | **981** | **%12** | **2.727** | **%32** | **8.423** | |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 63,1 ; dof= 12.*

**Cross: How old were you when you stopped full-time education? / Access to clinical trials…**

| HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION? | ACCESS TO CLINICAL TRIALS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 15 y.o. or under | 32 | %8 | 112 | %27 | 132 | %32 | 73 | %18 | 64 | %15 | 413 | %100 |
| between 16 and 19 y.o. | 196 | %9 | 520 | %23 | 768 | %34 | 449 | %20 | 350 | %15 | 2.283 | %100 |
| between 20 and 23 y.o. | 158 | %6 | 653 | %23 | 1.029 | %36 | 454 | %16 | 530 | %19 | 2.824 | %100 |
| 24 y.o. or above | 210 | %7 | 759 | %26 | 1.023 | %35 | 495 | %17 | 416 | %14 | 2.903 | %100 |
| **TOTAL** | **596** | **%7** | **2.044** | **%24** | **2.952** | **%35** | **1.471** | **%17** | **1.360** | **%16** | **8.423** | |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 58,4 ; dof= 12.*

## Cross: How old were you when you stopped full-time education? / Access to financial products, such as loans, mortgages, insurance…

| HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION? | ACCESS TO FINANCIAL PRODUCTS, SUCH AS LOANS, MORTGAGES, INSURANCE… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | ...HAS GOTTEN WORSE | | ...HAS IMPROVED | | ...HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 15 y.o. or under | 79 | %19 | 11 | %3 | 104 | %25 | 80 | %19 | 139 | %34 | 413 | %100 |
| between 16 and 19 y.o. | 411 | %18 | 41 | %2 | **549** | **%24** | **472** | **%21** | 810 | %35 | 2.283 | %100 |
| between 20 and 23 y.o. | 556 | %20 | 56 | %2 | 742 | %26 | **454** | **%16** | 1.016 | %36 | 2.824 | %100 |
| 24 y.o. or above | 554 | %19 | 73 | %3 | **807** | **%28** | 517 | %18 | **952** | **%33** | 2.903 | %100 |
| **TOTAL** | **1.600** | **%19** | **181** | **%2** | **2.202** | **%26** | **1.523** | **%18** | **2.917** | **%35** | **8.423** | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 33,1 ; dof= 12.*

## Cross: How old were you when you stopped full-time education? / Your social life…

| HOW OLD WERE YOU WHEN YOU STOPPED FULL-TIME EDUCATION? | YOUR SOCIAL LIFE… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | ...HAS GOTTEN WORSE | | ...HAS IMPROVED | | ...HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 15 y.o. or under | 228 | %55 | 29 | %7 | **122** | **%30** | 7 | %2 | 27 | %7 | 413 | %100 |
| between 16 and 19 y.o. | **1.235** | **%54** | 164 | %7 | **736** | **%32** | 46 | %2 | **102** | **%4** | 2.283 | %100 |
| between 20 and 23 y.o. | **1.369** | **%48** | 210 | %7 | **1.024** | **%36** | **44** | **%2** | **177** | **%6** | 2.824 | %100 |
| 24 y.o. or above | 1.431 | %49 | **254** | **%9** | 995 | %34 | **72** | **%2** | 151 | %5 | 2.903 | %100 |
| **TOTAL** | **4.263** | **%51** | **657** | **%8** | **2.877** | **%34** | **169** | **%2** | **457** | **%5** | **8.423** | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 39,8 ; dof= 12.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

## Cross: How would you best describe yourself? / Access to the most adapted care, treatments or surgery…

| HOW WOULD YOU BEST DESCRIBE YOURSELF? | ACCESS TO THE MOST ADAPTED CARE, TREATMENTS OR SURGERY… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| I belong to the ethnic majority in the country where I live | 649 | %10 | 2.955 | %45 | 2.456 | %37 | 325 | %5 | 249 | %4 | 6.634 | %100 |
| I am part of an ethnic minority in the country where I live | 56 | %13 | 154 | %37 | 159 | %38 | 28 | %7 | 23 | %5 | 420 | %100 |
| Other, specify… | 40 | %14 | 121 | %41 | 93 | %32 | 22 | %7 | 19 | %6 | 295 | %100 |
| TOTAL | 745 | %10 | 3.230 | %44 | 2.708 | %37 | 375 | %5 | 291 | %4 | 7.349 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 30,6 ; dof= 8.*

## Cross: How would you best describe yourself? / Understanding how the disease will progress…

| HOW WOULD YOU BEST DESCRIBE YOURSELF? | UNDERSTANDING HOW THE DISEASE WILL PROGRESS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| I belong to the ethnic majority in the country where I live | 482 | %7 | 3.757 | %57 | 1.921 | %29 | 352 | %5 | 122 | %2 | 6.634 | %100 |
| I am part of an ethnic minority in the country where I live | 52 | %12 | 200 | %48 | 123 | %29 | 34 | %8 | 11 | %3 | 420 | %100 |
| Other, specify… | 40 | %14 | 142 | %48 | 76 | %26 | 31 | %11 | 6 | %2 | 295 | %100 |
| TOTAL | 574 | %8 | 4.099 | %56 | 2.120 | %29 | 417 | %6 | 139 | %2 | 7.349 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 55,6 ; dof= 8.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

## Cross: How would you best describe yourself? / Financial support including social security benefits…

| HOW WOULD YOU BEST DESCRIBE YOURSELF? | FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| I belong to the ethnic majority in the country where I live | 664 | %15 | 775 | %18 | 1.718 | %39 | 392 | %9 | 839 | %19 | 4.388 | %100 |
| I am part of an ethnic minority in the country where I live | 68 | %25 | 45 | %16 | 89 | %32 | 31 | %11 | 44 | %16 | 277 | %100 |
| Other, specify... | 36 | %17 | 24 | %11 | 77 | %36 | 30 | %14 | 47 | %22 | 214 | %100 |
| TOTAL | 768 | %16 | 844 | %17 | 1.884 | %39 | 453 | %9 | 930 | %19 | 4.879 | |

🟪 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 32,9 ; dof= 8.*

## Cross: How would you best describe yourself? / Integration at school…

| HOW WOULD YOU BEST DESCRIBE YOURSELF? | INTEGRATION AT SCHOOL… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| I belong to the ethnic majority in the country where I live | 548 | %8 | 701 | %11 | 1.543 | %23 | 436 | %7 | 3.406 | %51 | 6.634 | %100 |
| I am part of an ethnic minority in the country where I live | 57 | %14 | 54 | %13 | 89 | %21 | 45 | %11 | 175 | %42 | 420 | %100 |
| Other, specify... | 23 | %8 | 29 | %10 | 47 | %16 | 30 | %10 | 166 | %56 | 295 | %100 |
| TOTAL | 628 | %9 | 784 | %11 | 1.679 | %23 | 511 | %7 | 3.747 | %51 | 7.349 | |

🟪 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 45,8 ; dof= 8.*

## Cross: How would you best describe yourself? / Integration at work…

| HOW WOULD YOU BEST DESCRIBE YOURSELF? | INTEGRATION AT WORK… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| I belong to the ethnic majority in the country where I live | 1.749 | %26 | 630 | %9 | 1.963 | %30 | 395 | %6 | 1.897 | %29 | 6.634 | %100 |
| I am part of an ethnic minority in the country where I live | 124 | %30 | 33 | %8 | 113 | %27 | 35 | %8 | 115 | %27 | 420 | %100 |
| Other, specify… | 95 | %32 | 25 | %8 | 53 | %18 | 27 | %9 | 95 | %32 | 295 | %100 |
| TOTAL | 1.968 | %27 | 688 | %9 | 2.129 | %29 | 457 | %6 | 2.107 | %29 | 7.349 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 29,5 ; dof= 8.*

## Cross: How would you best describe yourself? / Access to social services (e.g. social worker support, household chores support)…

| HOW WOULD YOU BEST DESCRIBE YOURSELF? | ACCESS TO SOCIAL SERVICES (E.G. SOCIAL WORKER SUPPORT, HOUSEHOLD CHORES SUPPORT)… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| I belong to the ethnic majority in the country where I live | 819 | %12 | 758 | %11 | 2.225 | %34 | 773 | %12 | 2.059 | %31 | 6.634 | %100 |
| I am part of an ethnic minority in the country where I live | 72 | %17 | 52 | %12 | 136 | %32 | 55 | %13 | 105 | %25 | 420 | %100 |
| Other, specify… | 47 | %16 | 34 | %12 | 78 | %26 | 40 | %14 | 96 | %33 | 295 | %100 |
| TOTAL | 938 | %13 | 844 | %11 | 2.439 | %33 | 868 | %12 | 2.260 | %31 | 7.349 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 20,7 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: How would you best describe yourself? / Access to clinical trials…

| HOW WOULD YOU BEST DESCRIBE YOURSELF? | ACCESS TO CLINICAL TRIALS… | | | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| I belong to the ethnic majority in the country where I live | 482 | %7 | 1.727 | %26 | 2.437 | %37 | 1.154 | %17 | 834 | %13 | 6.634 | %100 |
| I am part of an ethnic minority in the country where I live | 43 | %10 | 85 | %20 | 144 | %34 | 83 | %20 | 65 | %15 | 420 | %100 |
| Other, specify… | 19 | %6 | 59 | %20 | 81 | %27 | 70 | %24 | 66 | %22 | 295 | %100 |
| **TOTAL** | **544** | **%7** | **1.871** | **%25** | **2.662** | **%36** | **1.307** | **%18** | **965** | **%13** | **7.349** | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 51,0 ; dof= 8.*

## Cross: How would you best describe yourself? / Access to financial products, such as loans, mortgages, insurance…

| HOW WOULD YOU BEST DESCRIBE YOURSELF? | ACCESS TO FINANCIAL PRODUCTS, SUCH AS LOANS, MORTGAGES, INSURANCE… | | | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| I belong to the ethnic majority in the country where I live | 1.195 | %18 | 152 | %2 | 1.838 | %28 | 1.237 | %19 | 2.212 | %33 | 6.634 | %100 |
| I am part of an ethnic minority in the country where I live | 100 | %24 | 13 | %3 | 115 | %27 | 76 | %18 | 116 | %28 | 420 | %100 |
| Other, specify… | 52 | %18 | 1 | %0 | 56 | %19 | 68 | %23 | 118 | %40 | 295 | %100 |
| **TOTAL** | **1.347** | **%18** | **166** | **%2** | **2.009** | **%27** | **1.381** | **%19** | **2.446** | **%33** | **7.349** | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 32,4 ; dof= 8.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

## Cross: How would you best describe yourself? / Your social life…

| | YOUR SOCIAL LIFE… | | | | | | | | | | | |
| HOW WOULD YOU BEST DESCRIBE YOURSELF? | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| I belong to the ethnic majority in the country where I live | 3.337 | %50 | 528 | %8 | 2.278 | %34 | 134 | %2 | 357 | %5 | 6.634 | %100 |
| I am part of an ethnic minority in the country where I live | 250 | %60 | 28 | %7 | 115 | %27 | 11 | %3 | 16 | %4 | 420 | %100 |
| Other, specify... | 163 | %55 | 17 | %6 | 86 | %29 | 6 | %2 | 23 | %8 | 295 | %100 |
| TOTAL | 3.750 | %51 | 573 | %8 | 2.479 | %34 | 151 | %2 | 396 | %5 | 7.349 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 23,5 ; dof= 8.*

## Cross: Typology of countries based on size and welfare / Access to the most adapted care, treatments or surgery…

| | ACCESS TO THE MOST ADAPTED CARE, TREATMENTS OR SURGERY… | | | | | | | | | | | |
| TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Group A ('Eastern Europe') | 173 | %12 | 618 | %42 | 547 | %37 | 79 | %5 | 51 | %3 | 1.468 | %100 |
| Group B ('Western Europe') | 454 | %10 | 2.100 | %46 | 1.701 | %37 | 195 | %4 | 165 | %4 | 4.615 | %100 |
| Group C ('Northern Europe') | 259 | %9 | 1.247 | %44 | 1.044 | %37 | 179 | %6 | 123 | %4 | 2.852 | %100 |
| TOTAL | 886 | %10 | 3.965 | %44 | 3.292 | %37 | 453 | %5 | 339 | %4 | 8.935 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 28,6 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Typology of countries based on size and welfare / Understanding how the disease will progress…

| TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE | UNDERSTANDING HOW THE DISEASE WILL PROGRESS… | | | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Group A ('Eastern Europe') | 207 | %14 | 666 | %46 | 435 | %30 | 120 | %8 | 34 | %2 | 1.462 | %100 |
| Group B ('Western Europe') | 302 | %7 | 2.732 | %59 | 1.307 | %28 | 206 | %4 | 66 | %1 | 4.613 | %100 |
| Group C ('Northern Europe') | 179 | %6 | 1.557 | %55 | 879 | %31 | 163 | %6 | 71 | %2 | 2.849 | %100 |
| TOTAL | 688 | %8 | 4.955 | %56 | 2.621 | %29 | 489 | %5 | 171 | %2 | 8.924 | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 177,0 ; dof= 8.*

## Cross: Typology of countries based on size and welfare / Financial support including social security benefits…

| TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE | FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS… | | | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Group A ('Eastern Europe') | 146 | %22 | 110 | %16 | 307 | %46 | 49 | %7 | 62 | %9 | 674 | %100 |
| Group B ('Western Europe') | 464 | %15 | 611 | %19 | 1.227 | %39 | 254 | %8 | 617 | %19 | 3.173 | %100 |
| Group C ('Northern Europe') | 314 | %15 | 322 | %15 | 783 | %38 | 227 | %11 | 439 | %21 | 2.085 | %100 |
| TOTAL | 924 | %16 | 1.043 | %18 | 2.317 | %39 | 530 | %9 | 1.118 | %19 | 5.932 | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 91,3 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Typology of countries based on size and welfare / Integration at school…

| TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE | INTEGRATION AT SCHOOL… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Group A ('Eastern Europe') | 175 | %12 | 224 | %15 | 422 | %29 | 137 | %9 | 510 | %35 | 1.468 | %100 |
| Group B ('Western Europe') | 389 | %8 | 470 | %10 | 1.072 | %23 | 279 | %6 | 2.405 | %52 | 4.615 | %100 |
| Group C ('Northern Europe') | 176 | %6 | 254 | %9 | 522 | %18 | 190 | %7 | 1.710 | %60 | 2.852 | %100 |
| TOTAL | 740 | %8 | 948 | %11 | 2.016 | %23 | 606 | %7 | 4.625 | %52 | 8.935 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 263,3 ; dof= 8.*

## Cross: Typology of countries based on size and welfare / Integration at work…

| TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE | INTEGRATION AT WORK… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Group A ('Eastern Europe') | 433 | %30 | 118 | %8 | 456 | %31 | 151 | %10 | 304 | %21 | 1.462 | %100 |
| Group B ('Western Europe') | 1.386 | %30 | 440 | %10 | 1.423 | %31 | 216 | %5 | 1.148 | %25 | 4.613 | %100 |
| Group C ('Northern Europe') | 569 | %20 | 256 | %9 | 690 | %24 | 180 | %6 | 1.153 | %40 | 2.848 | %100 |
| TOTAL | 2.388 | %27 | 814 | %9 | 2.569 | %29 | 547 | %6 | 2.605 | %29 | 8.923 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 352,0 ; dof= 8.*

## Cross: Typology of countries based on size and welfare / Access to social services (e.g. social worker support, household chores support)…

| TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE | ACCESS TO SOCIAL SERVICES (E.G. SOCIAL WORKER SUPPORT, HOUSEHOLD CHORES SUPPORT)… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Group A ('Eastern Europe') | 243 | %17 | 145 | %10 | 590 | %40 | 175 | %12 | 308 | %21 | 1.461 | %100 |
| Group B ('Western Europe') | 566 | %12 | 505 | %11 | 1.509 | %33 | 519 | %11 | 1.514 | %33 | 4.613 | %100 |
| Group C ('Northern Europe') | 315 | %11 | 349 | %12 | 777 | %27 | 367 | %13 | 1.040 | %37 | 2.848 | %100 |
| TOTAL | 1.124 | %13 | 999 | %11 | 2.876 | %32 | 1.061 | %12 | 2.862 | %32 | 8.922 | %100 |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 159,2 ; dof= 8.*

## Cross: Typology of countries based on size and welfare / Access to clinical trials…

| TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE | ACCESS TO CLINICAL TRIALS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Group A ('Eastern Europe') | 146 | %10 | 315 | %21 | 643 | %44 | 206 | %14 | 157 | %11 | 1.467 | %100 |
| Group B ('Western Europe') | 299 | %6 | 1.165 | %25 | 1.511 | %33 | 775 | %17 | 865 | %19 | 4.615 | %100 |
| Group C ('Northern Europe') | 187 | %7 | 701 | %25 | 988 | %35 | 571 | %20 | 405 | %14 | 2.852 | %100 |
| TOTAL | 632 | %7 | 2.181 | %24 | 3.142 | %35 | 1.552 | %17 | 1.427 | %16 | 8.934 | %100 |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 141,4 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Typology of countries based on size and welfare / Access to financial products, such as loans, mortgages, insurance…

| TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE | ACCESS TO FINANCIAL PRODUCTS, SUCH AS LOANS, MORTGAGES, INSURANCE… | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Group A ('Eastern Europe') | 295 | %20 | 46 | %3 | 521 | %36 | 271 | %18 | 334 | %23 | 1.467 | %100 |
| Group B ('Western Europe') | 816 | %18 | 92 | %2 | 1.221 | %26 | 835 | %18 | 1.651 | %36 | 4.615 | %100 |
| Group C ('Northern Europe') | 576 | %20 | 58 | %2 | 587 | %21 | 531 | %19 | 1.099 | %39 | 2.851 | %100 |
| TOTAL | 1.687 | %19 | 196 | %2 | 2.329 | %26 | 1.637 | %18 | 3.084 | %35 | 8.933 | |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 172,2 ; dof= 8.*

## Cross: Typology of countries based on size and welfare / Your social life…

| TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE | YOUR SOCIAL LIFE… | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Group A ('Eastern Europe') | 796 | %54 | 109 | %7 | 464 | %32 | 28 | %2 | 64 | %4 | 1.461 | %100 |
| Group B ('Western Europe') | 2.309 | %50 | 394 | %9 | 1.615 | %35 | 82 | %2 | 213 | %5 | 4.613 | %100 |
| Group C ('Northern Europe') | 1.420 | %50 | 197 | %7 | 961 | %34 | 72 | %3 | 198 | %7 | 2.848 | %100 |
| TOTAL | 4.525 | %51 | 700 | %8 | 3.040 | %34 | 182 | %2 | 475 | %5 | 8.922 | |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 40,7 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Genetic diseases / Access to the most adapted care, treatments or surgery…

| | ACCESS TO THE MOST ADAPTED CARE, TREATMENTS OR SURGERY… | | | | | | | | | | | |
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| GENETIC DISEASES | N | % | N | % | N | % | N | % | N | % | N | % |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| Genetic diseases | 517 | %10 | 2.337 | %45 | 1.918 | %37 | 250 | %5 | 210 | %4 | 5.232 | %100 |
| Non Genetic diseases | 232 | %9 | 1.188 | %47 | 901 | %36 | 126 | %5 | 87 | %3 | 2.534 | %100 |
| TOTAL | 749 | %10 | 3.525 | %45 | 2.819 | %36 | 376 | %5 | 297 | %4 | 7.766 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is not significant. p-value= 0,3 ; Chi2= 5,0 ; dof= 4.*

## Cross: Genetic diseases / Understanding how the disease will progress…

| | UNDERSTANDING HOW THE DISEASE WILL PROGRESS… | | | | | | | | | | | |
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| GENETIC DISEASES | N | % | N | % | N | % | N | % | N | % | N | % |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| Genetic diseases | 387 | %7 | 3.015 | %58 | 1.453 | %28 | 248 | %5 | 116 | %2 | 5.219 | %100 |
| Non Genetic diseases | 193 | %8 | 1.374 | %54 | 783 | %31 | 145 | %6 | 37 | %1 | 2.532 | %100 |
| TOTAL | 580 | %7 | 4.389 | %57 | 2.236 | %29 | 393 | %5 | 153 | %2 | 7.751 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 17,6 ; dof= 4.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Genetic diseases / Financial support including social security benefits…

| | FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS… | | | | | | | | | | | |
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| GENETIC DISEASES | N | % | N | % | N | % | N | % | N | % | N | % |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| Genetic diseases | 458 | %16 | 560 | %19 | 1.139 | %39 | 243 | %8 | 501 | %17 | 2.901 | %100 |
| Non Genetic diseases | 315 | %15 | 351 | %16 | 823 | %38 | 202 | %9 | 469 | %22 | 2.160 | %100 |
| **TOTAL** | **773** | **%15** | **911** | **%18** | **1.962** | **%39** | **445** | **%9** | **970** | **%19** | **5.061** | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 22,1 ; dof= 4.*

## Cross: Genetic diseases / Integration at school…

| | INTEGRATION AT SCHOOL… | | | | | | | | | | | |
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| GENETIC DISEASES | N | % | N | % | N | % | N | % | N | % | N | % |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| Genetic diseases | 474 | %9 | 722 | %14 | 1.434 | %27 | 342 | %7 | 2.260 | %43 | 5.232 | %100 |
| Non Genetic diseases | 175 | %7 | 127 | %5 | 327 | %13 | 156 | %6 | 1.749 | %69 | 2.534 | %100 |
| **TOTAL** | **649** | **%8** | **849** | **%11** | **1.761** | **%23** | **498** | **%6** | **4.009** | **%52** | **7.766** | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 509,4 ; dof= 4.*

## Cross: Genetic diseases / Integration at work…

| GENETIC DISEASES | INTEGRATION AT WORK… | | | | | | | | | | | |
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| Genetic diseases | 1.282 | %25 | 484 | %9 | 1.606 | %31 | 328 | %6 | 1.518 | %29 | 5.218 | %100 |
| Non Genetic diseases | 788 | %31 | 225 | %9 | 619 | %24 | 127 | %5 | 773 | %31 | 2.532 | %100 |
| TOTAL | 2.070 | %27 | 709 | %9 | 2.225 | %29 | 455 | %6 | 2.291 | %30 | 7.750 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 57,4 ; dof= 4.*

## Cross: Genetic diseases / Access to social services (e.g. social worker support, household chores support)…

| GENETIC DISEASES | ACCESS TO SOCIAL SERVICES (E.G. SOCIAL WORKER SUPPORT, HOUSEHOLD CHORES SUPPORT)… | | | | | | | | | | | |
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| Genetic diseases | 628 | %12 | 691 | %13 | 1.795 | %34 | 577 | %11 | 1.527 | %29 | 5.218 | %100 |
| Non Genetic diseases | 334 | %13 | 190 | %8 | 705 | %28 | 318 | %13 | 984 | %39 | 2.531 | %100 |
| TOTAL | 962 | %12 | 881 | %11 | 2.500 | %32 | 895 | %12 | 2.511 | %32 | 7.749 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 125,8 ; dof= 4.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Genetic diseases / Access to clinical trials…

| | ACCESS TO CLINICAL TRIALS… | | | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| GENETIC DISEASES | N | % | N | % | N | % | N | % | N | % | N | % |
| Genetic diseases | 360 | %7 | 1.367 | %26 | 1.863 | %36 | 853 | %16 | 789 | %15 | 5.232 | %100 |
| Non Genetic diseases | 173 | %7 | 576 | %23 | 889 | %35 | 453 | %18 | 442 | %17 | 2.533 | %100 |
| TOTAL | 533 | %7 | 1.943 | %25 | 2.752 | %35 | 1.306 | %17 | 1.231 | %16 | 7.765 | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 16,5 ; dof= 4.*

## Cross: Genetic diseases / Access to financial products, such as loans, mortgages, insurance…

| | ACCESS TO FINANCIAL PRODUCTS, SUCH AS LOANS, MORTGAGES, INSURANCE… | | | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| GENETIC DISEASES | N | % | N | % | N | % | N | % | N | % | N | % |
| Genetic diseases | 989 | %19 | 138 | %3 | 1.486 | %28 | 942 | %18 | 1.676 | %32 | 5.231 | %100 |
| Non Genetic diseases | 485 | %19 | 38 | %2 | 538 | %21 | 442 | %17 | 1.030 | %41 | 2.533 | %100 |
| TOTAL | 1.474 | %19 | 176 | %2 | 2.024 | %26 | 1.384 | %18 | 2.706 | %35 | 7.764 | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 80,1 ; dof= 4.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Genetic diseases / Your social life…

| | YOUR SOCIAL LIFE… | | | | | | | | | | | |
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| **GENETIC DISEASES** | N | % | N | % | N | % | N | % | N | % | N | % |
| Genetic diseases | 2.429 | %47 | 452 | %9 | 1.915 | %37 | 109 | %2 | 313 | %6 | 5.218 | %100 |
| Non Genetic diseases | 1.481 | %59 | 159 | %6 | 747 | %30 | 46 | %2 | 98 | %4 | 2.531 | %100 |
| **TOTAL** | 3.910 | %50 | 611 | %8 | 2.662 | %34 | 155 | %2 | 411 | %5 | 7.749 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 101,4 ; dof= 4.*

## Cross: Point prevalence of the rare disease / Access to the most adapted care, treatments or surgery…

| | ACCESS TO THE MOST ADAPTED CARE, TREATMENTS OR SURGERY… | | | | | | | | | | | |
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| **POINT PREVALENCE OF THE RARE DISEASE** | N | % | N | % | N | % | N | % | N | % | N | % |
| 1-5 / 10 000 | 228 | %10 | 1.125 | %48 | 801 | %34 | 124 | %5 | 75 | %3 | 2.353 | %100 |
| 1-9 / 100 000 | 196 | %10 | 825 | %42 | 757 | %39 | 86 | %4 | 86 | %4 | 1.950 | %100 |
| 1-9 / 1 000 000 | 50 | %11 | 207 | %46 | 156 | %35 | 17 | %4 | 19 | %4 | 449 | %100 |
| <1 / 1 000 000 | 85 | %11 | 311 | %39 | 328 | %41 | 34 | %4 | 39 | %5 | 797 | %100 |
| **TOTAL** | 559 | %10 | 2.468 | %44 | 2.042 | %37 | 261 | %5 | 219 | %4 | 5.549 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 35,8 ; dof= 12.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Point prevalence of the rare disease / Understanding how the disease will progress…

| POINT PREVALENCE OF THE RARE DISEASE | UNDERSTANDING HOW THE DISEASE WILL PROGRESS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 1-5 / 10 000 | 154 | %7 | 1.400 | %60 | 645 | %27 | 115 | %5 | 36 | %2 | 2.350 | %100 |
| 1-9 / 100 000 | 147 | %8 | 1.081 | %55 | 570 | %29 | 102 | %5 | 49 | %3 | 1.949 | %100 |
| 1-9 / 1 000 000 | 30 | %7 | 247 | %55 | 140 | %31 | 23 | %5 | 9 | %2 | 449 | %100 |
| <1 / 1 000 000 | 80 | %10 | 395 | %50 | 256 | %32 | 43 | %5 | 21 | %3 | 795 | %100 |
| TOTAL | 411 | %7 | 3.123 | %56 | 1.611 | %29 | 283 | %5 | 115 | %2 | 5.543 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 33,6 ; dof= 12.*

## Cross: Point prevalence of the rare disease / Financial support including social security benefits…

| POINT PREVALENCE OF THE RARE DISEASE | FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 1-5 / 10 000 | 244 | %14 | 323 | %18 | 680 | %39 | 169 | %10 | 332 | %19 | 1.748 | %100 |
| 1-9 / 100 000 | 205 | %16 | 245 | %19 | 496 | %38 | 110 | %8 | 250 | %19 | 1.306 | %100 |
| 1-9 / 1 000 000 | 47 | %18 | 48 | %19 | 95 | %37 | 25 | %10 | 42 | %16 | 257 | %100 |
| <1 / 1 000 000 | 76 | %16 | 83 | %18 | 185 | %40 | 32 | %7 | 86 | %19 | 462 | %100 |
| TOTAL | 572 | %15 | 699 | %19 | 1.456 | %39 | 336 | %9 | 710 | %19 | 3.773 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is not significant. p-value= 0,7 ; Chi2= 9,5 ; dof= 12.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Point prevalence of the rare disease / Integration at school…

| | INTEGRATION AT SCHOOL… | | | | | | | | | | | |
| POINT PREVALENCE OF THE RARE DISEASE | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 1-5 / 10 000 | 190 | %8 | 227 | %10 | 472 | %20 | 166 | %7 | 1.298 | %55 | 2.353 | %100 |
| 1-9 / 100 000 | 141 | %7 | 184 | %9 | 443 | %23 | 112 | %6 | 1.070 | %55 | 1.950 | %100 |
| 1-9 / 1 000 000 | 47 | %10 | 65 | %14 | 106 | %24 | 26 | %6 | 205 | %46 | 449 | %100 |
| <1 / 1 000 000 | 68 | %9 | 89 | %11 | 218 | %27 | 51 | %6 | 371 | %47 | 797 | %100 |
| TOTAL | 446 | %8 | 565 | %10 | 1.239 | %22 | 355 | %6 | 2.944 | %53 | 5.549 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 48,0 ; dof= 12.*

## Cross: Point prevalence of the rare disease / Integration at work…

| | INTEGRATION AT WORK… | | | | | | | | | | | |
| POINT PREVALENCE OF THE RARE DISEASE | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 1-5 / 10 000 | 602 | %26 | 216 | %9 | 685 | %29 | 135 | %6 | 712 | %30 | 2.350 | %100 |
| 1-9 / 100 000 | 503 | %26 | 178 | %9 | 559 | %29 | 118 | %6 | 590 | %30 | 1.948 | %100 |
| 1-9 / 1 000 000 | 114 | %25 | 42 | %9 | 139 | %31 | 29 | %6 | 125 | %28 | 449 | %100 |
| <1 / 1 000 000 | 204 | %26 | 58 | %7 | 211 | %27 | 42 | %5 | 280 | %35 | 795 | %100 |
| TOTAL | 1.423 | %26 | 494 | %9 | 1.594 | %29 | 324 | %6 | 1.707 | %31 | 5.542 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is not significant. p-value= 0,4 ; Chi2= 12,6 ; dof= 12.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Point prevalence of the rare disease / Access to social services (e.g. social worker support, household chores support)…**

| POINT PREVALENCE OF THE RARE DISEASE | ACCESS TO SOCIAL SERVICES (E.G. SOCIAL WORKER SUPPORT, HOUSEHOLD CHORES SUPPORT)… | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 1-5 / 10 000 | 260 | %11 | 214 | %9 | 709 | %30 | 302 | %13 | 865 | %37 | 2.350 | %100 |
| 1-9 / 100 000 | 269 | %14 | 235 | %12 | 625 | %32 | 198 | %10 | 621 | %32 | 1.948 | %100 |
| 1-9 / 1 000 000 | 49 | %11 | 63 | %14 | 132 | %29 | 48 | %11 | 157 | %35 | 449 | %100 |
| <1 / 1 000 000 | 108 | %14 | 99 | %12 | 269 | %34 | 90 | %11 | 229 | %29 | 795 | %100 |
| TOTAL | 686 | %12 | 611 | %11 | 1.735 | %31 | 638 | %12 | 1.872 | %34 | 5.542 | |

■ *Under-represented elements*　　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 48,1 ; dof= 12.*

**Cross: Point prevalence of the rare disease / Access to clinical trials…**

| POINT PREVALENCE OF THE RARE DISEASE | ACCESS TO CLINICAL TRIALS… | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 1-5 / 10 000 | 142 | %6 | 596 | %25 | 822 | %35 | 425 | %18 | 368 | %16 | 2.353 | %100 |
| 1-9 / 100 000 | 141 | %7 | 466 | %24 | 716 | %37 | 318 | %16 | 309 | %16 | 1.950 | %100 |
| 1-9 / 1 000 000 | 29 | %6 | 128 | %29 | 148 | %33 | 81 | %18 | 63 | %14 | 449 | %100 |
| <1 / 1 000 000 | 59 | %7 | 195 | %24 | 298 | %37 | 116 | %15 | 129 | %16 | 797 | %100 |
| TOTAL | 371 | %7 | 1.385 | %25 | 1.984 | %36 | 940 | %17 | 869 | %16 | 5.549 | |

■ *Under-represented elements*　　■ *Over-represented elements*

*The relationship is not significant. p-value= 0,2 ; Chi2= 15,0 ; dof= 12.*

## Cross: Point prevalence of the rare disease / Access to financial products, such as loans, mortgages, insurance…

| POINT PREVALENCE OF THE RARE DISEASE | ACCESS TO FINANCIAL PRODUCTS, SUCH AS LOANS, MORTGAGES, INSURANCE… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 1-5 / 10 000 | 437 | %19 | 39 | %2 | 608 | %26 | 441 | %19 | 828 | %35 | 2.353 | %100 |
| 1-9 / 100 000 | 398 | %20 | 35 | %2 | 510 | %26 | 313 | %16 | 693 | %36 | 1.949 | %100 |
| 1-9 / 1 000 000 | 86 | %19 | 15 | %3 | 110 | %24 | 86 | %19 | 152 | %34 | 449 | %100 |
| <1 / 1 000 000 | 139 | %17 | 25 | %3 | 225 | %28 | 133 | %17 | 275 | %35 | 797 | %100 |
| TOTAL | 1.060 | %19 | 114 | %2 | 1.453 | %26 | 973 | %18 | 1.948 | %35 | 5.548 | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is significant. p-value= 0,0 ; Chi2= 21,5 ; dof= 12.*

## Cross: Point prevalence of the rare disease / Your social life…

| POINT PREVALENCE OF THE RARE DISEASE | YOUR SOCIAL LIFE… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 1-5 / 10 000 | 1.142 | %49 | 206 | %9 | 839 | %36 | 50 | %2 | 113 | %5 | 2.350 | %100 |
| 1-9 / 100 000 | 1.017 | %52 | 140 | %7 | 653 | %34 | 35 | %2 | 103 | %5 | 1.948 | %100 |
| 1-9 / 1 000 000 | 228 | %51 | 38 | %8 | 136 | %30 | 13 | %3 | 34 | %8 | 449 | %100 |
| <1 / 1 000 000 | 392 | %49 | 60 | %8 | 281 | %35 | 20 | %3 | 42 | %5 | 795 | %100 |
| TOTAL | 2.779 | %50 | 444 | %8 | 1.909 | %34 | 118 | %2 | 292 | %5 | 5.542 | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is weakly significant. p-value= 0,1 ; Chi2= 18,8 ; dof= 12.*

## Cross: Disease complexity classified into five groups, based on the number of affected body parts. / Access to the most adapted care, treatments or surgery…

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | ACCESS TO THE MOST ADAPTED CARE, TREATMENTS OR SURGERY… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 1-3 body parts | 433 | %8 | 2.340 | %44 | 1.962 | %37 | 291 | %6 | 234 | %4 | 5.260 | %100 |
| 4-7 body parts | 269 | %10 | 1.199 | %45 | 990 | %37 | 115 | %4 | 82 | %3 | 2.655 | %100 |
| 8-11 body parts | 113 | %14 | 376 | %46 | 269 | %33 | 40 | %5 | 21 | %3 | 819 | %100 |
| 12-15 body parts | 53 | %23 | 90 | %38 | 80 | %34 | 7 | %3 | 5 | %2 | 235 | %100 |
| 16 body parts or more | 21 | %38 | 15 | %27 | 15 | %27 | 4 | %7 | 0 | %0 | 55 | %100 |
| TOTAL | 889 | %10 | 4.020 | %45 | 3.316 | %37 | 457 | %5 | 342 | %4 | 9.024 | |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 146,8 ; dof= 16.*

## Cross: Disease complexity classified into five groups, based on the number of affected body parts. / Understanding how the disease will progress…

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | UNDERSTANDING HOW THE DISEASE WILL PROGRESS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 1-3 body parts | 359 | %7 | 2.899 | %55 | 1.575 | %30 | 310 | %6 | 106 | %2 | 5.249 | %100 |
| 4-7 body parts | 213 | %8 | 1.503 | %57 | 752 | %28 | 129 | %5 | 52 | %2 | 2.649 | %100 |
| 8-11 body parts | 87 | %11 | 450 | %55 | 235 | %29 | 36 | %4 | 11 | %1 | 819 | %100 |
| 12-15 body parts | 24 | %10 | 122 | %52 | 68 | %29 | 16 | %7 | 5 | %2 | 235 | %100 |
| 16 body parts or more | 11 | %20 | 25 | %46 | 14 | %26 | 3 | %6 | 1 | %2 | 54 | %100 |
| TOTAL | 694 | %8 | 4.999 | %56 | 2.644 | %29 | 494 | %5 | 175 | %2 | 9.006 | |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 39,6 ; dof= 16.*

Rare Barometer — A EURORDIS ❋ INITIATIVE

EURORDIS RARE DISEASES EUROPE

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Disease complexity classified into five groups, based on the number of affected body parts. / Financial support including social security benefits…

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 1-3 body parts | 438 | %13 | 583 | %17 | 1.353 | %39 | 314 | %9 | 785 | %23 | 3.473 | %100 |
| 4-7 body parts | 284 | %17 | 305 | %18 | 681 | %40 | 157 | %9 | 270 | %16 | 1.697 | %100 |
| 8-11 body parts | 142 | %24 | 121 | %20 | 224 | %38 | 52 | %9 | 58 | %10 | 597 | %100 |
| 12-15 body parts | 51 | %28 | 39 | %22 | 69 | %39 | 9 | %5 | 11 | %6 | 179 | %100 |
| 16 body parts or more | 13 | %32 | 8 | %20 | 18 | %44 | 0 | %0 | 2 | %5 | 41 | %100 |
| TOTAL | 928 | %16 | 1.056 | %18 | 2.345 | %39 | 532 | %9 | 1.126 | %19 | 5.987 | %100 |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 167,0 ; dof= 16.*

## Cross: Disease complexity classified into five groups, based on the number of affected body parts. / Integration at school…

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | INTEGRATION AT SCHOOL… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 1-3 body parts | 379 | %7 | 549 | %10 | 1.188 | %23 | 352 | %7 | 2.792 | %53 | 5.260 | %100 |
| 4-7 body parts | 243 | %9 | 293 | %11 | 618 | %23 | 186 | %7 | 1.315 | %50 | 2.655 | %100 |
| 8-11 body parts | 83 | %10 | 88 | %11 | 171 | %21 | 59 | %7 | 418 | %51 | 819 | %100 |
| 12-15 body parts | 30 | %13 | 25 | %11 | 42 | %18 | 14 | %6 | 124 | %53 | 235 | %100 |
| 16 body parts or more | 11 | %20 | 5 | %9 | 11 | %20 | 2 | %4 | 26 | %47 | 55 | %100 |
| TOTAL | 746 | %8 | 960 | %11 | 2.030 | %22 | 613 | %7 | 4.675 | %52 | 9.024 | %100 |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 39,0 ; dof= 16.*

## Cross: Disease complexity classified into five groups, based on the number of affected body parts. / Integration at work…

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | INTEGRATION AT WORK… | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 1-3 body parts | **1.224** | **%23** | 466 | %9 | **1.612** | **%31** | **346** | **%7** | **1.600** | **%30** | **5.248** | %100 |
| 4-7 body parts | **795** | **%30** | 254 | %10 | **710** | **%27** | 151 | %6 | 739 | %28 | **2.649** | %100 |
| 8-11 body parts | **269** | **%33** | 82 | %10 | **209** | **%26** | 41 | %5 | 218 | %27 | 819 | %100 |
| 12-15 body parts | **101** | **%43** | **10** | **%4** | **48** | **%20** | 10 | %4 | 66 | %28 | 235 | %100 |
| 16 body parts or more | **22** | **%41** | 6 | %11 | **8** | **%15** | 3 | %6 | 15 | %28 | 54 | %100 |
| **TOTAL** | **2.411** | **%27** | **818** | **%9** | **2.587** | **%29** | **551** | **%6** | **2.638** | **%29** | **9.005** | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 115,0 ; dof= 16.*

## Cross: Disease complexity classified into five groups, based on the number of affected body parts. / Access to social services (e.g. social worker support, household chores support)…

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | ACCESS TO SOCIAL SERVICES (E.G. SOCIAL WORKER SUPPORT, HOUSEHOLD CHORES SUPPORT)… | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 1-3 body parts | **572** | **%11** | **503** | **%10** | **1.614** | **%31** | 638 | %12 | **1.920** | **%37** | **5.247** | %100 |
| 4-7 body parts | 342 | %13 | **347** | **%13** | **909** | **%34** | 307 | %12 | **744** | **%28** | **2.649** | %100 |
| 8-11 body parts | **154** | **%19** | **109** | **%13** | 276 | %34 | 96 | %12 | **184** | **%22** | 819 | %100 |
| 12-15 body parts | **52** | **%22** | **40** | **%17** | **90** | **%38** | 20 | %9 | **33** | **%14** | 235 | %100 |
| 16 body parts or more | **14** | **%26** | **12** | **%22** | 17 | %31 | 5 | %9 | **6** | **%11** | 54 | %100 |
| **TOTAL** | **1.134** | **%13** | **1.011** | **%11** | **2.906** | **%32** | **1.066** | **%12** | **2.887** | **%32** | **9.004** | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 213,4 ; dof= 16.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Disease complexity classified into five groups, based on the number of affected body parts. / Access to clinical trials…

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | ACCESS TO CLINICAL TRIALS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 1-3 body parts | 310 | %6 | 1.266 | %24 | 1.815 | %35 | 921 | %18 | 947 | %18 | 5.259 | %100 |
| 4-7 body parts | 181 | %7 | 660 | %25 | 977 | %37 | 436 | %16 | 401 | %15 | 2.655 | %100 |
| 8-11 body parts | 96 | %12 | 213 | %26 | 284 | %35 | 147 | %18 | 79 | %10 | 819 | %100 |
| 12-15 body parts | 41 | %17 | 47 | %20 | 84 | %36 | 45 | %19 | 18 | %8 | 235 | %100 |
| 16 body parts or more | 9 | %16 | 11 | %20 | 13 | %24 | 15 | %27 | 7 | %13 | 55 | %100 |
| TOTAL | 637 | %7 | 2.197 | %24 | 3.173 | %35 | 1.564 | %17 | 1.452 | %16 | 9.023 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 137,3 ; dof= 16.*

## Cross: Disease complexity classified into five groups, based on the number of affected body parts. / Access to financial products, such as loans, mortgages, insurance…

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | ACCESS TO FINANCIAL PRODUCTS, SUCH AS LOANS, MORTGAGES, INSURANCE… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 1-3 body parts | 863 | %16 | 116 | %2 | 1.337 | %25 | 938 | %18 | 2.004 | %38 | 5.258 | %100 |
| 4-7 body parts | 528 | %20 | 54 | %2 | 749 | %28 | 499 | %19 | 825 | %31 | 2.655 | %100 |
| 8-11 body parts | 214 | %26 | 21 | %3 | 193 | %24 | 162 | %20 | 229 | %28 | 819 | %100 |
| 12-15 body parts | 86 | %37 | 6 | %3 | 56 | %24 | 39 | %17 | 48 | %20 | 235 | %100 |
| 16 body parts or more | 24 | %44 | 3 | %5 | 10 | %18 | 10 | %18 | 8 | %15 | 55 | %100 |
| TOTAL | 1.715 | %19 | 200 | %2 | 2.345 | %26 | 1.648 | %18 | 3.114 | %35 | 9.022 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 171,4 ; dof= 16.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: Disease complexity classified into five groups, based on the number of affected body parts. / Your social life…

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | YOUR SOCIAL LIFE… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | ...HAS GOTTEN WORSE | | ...HAS IMPROVED | | ...HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 1-3 body parts | 2.396 | %46 | 432 | %8 | 2.003 | %38 | 112 | %2 | 304 | %6 | 5.247 | %100 |
| 4-7 body parts | 1.479 | %56 | 185 | %7 | 791 | %30 | 53 | %2 | 141 | %5 | 2.649 | %100 |
| 8-11 body parts | 504 | %62 | 61 | %7 | 211 | %26 | 14 | %2 | 29 | %4 | 819 | %100 |
| 12-15 body parts | 155 | %66 | 26 | %11 | 47 | %20 | 4 | %2 | 3 | %1 | 235 | %100 |
| 16 body parts or more | 37 | %69 | 4 | %7 | 12 | %22 | 0 | %0 | 1 | %2 | 54 | %100 |
| TOTAL | 4.571 | %51 | 708 | %8 | 3.064 | %34 | 183 | %2 | 478 | %5 | 9.004 | %100 |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 169,6 ; dof= 16.*

## Cross: Disease complexity classified into five groups, based on the number of affected body parts. / Access to financial products, such as loans, mortgages, insurance…

| DISEASE COMPLEXITY CLASSIFIED INTO FIVE GROUPS, BASED ON THE NUMBER OF AFFECTED BODY PARTS. | ACCESS TO FINANCIAL PRODUCTS, SUCH AS LOANS, MORTGAGES, INSURANCE… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | ...HAS GOTTEN WORSE | | ...HAS IMPROVED | | ...HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| 1-3 body parts | 863 | %16 | 116 | %2 | 1.337 | %25 | 938 | %18 | 2.004 | %38 | 5.258 | %100 |
| 4-7 body parts | 528 | %20 | 54 | %2 | 749 | %28 | 499 | %19 | 825 | %31 | 2.655 | %100 |
| 8-11 body parts | 214 | %26 | 21 | %3 | 193 | %24 | 162 | %20 | 229 | %28 | 819 | %100 |
| 12-15 body parts | 86 | %37 | 6 | %3 | 56 | %24 | 39 | %17 | 48 | %20 | 235 | %100 |
| 16 body parts or more | 24 | %44 | 3 | %5 | 10 | %18 | 10 | %18 | 8 | %15 | 55 | %100 |
| TOTAL | 1.715 | %19 | 200 | %2 | 2.345 | %26 | 1.648 | %18 | 3.114 | %35 | 9.022 | %100 |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 171,4 ; dof= 16.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: …behavioural disorders that cause problems in school, at home or in social situations / Access to the most adapted care, treatments or surgery…

| …BEHAVIOURAL DISORDERS THAT CAUSE PROBLEMS IN SCHOOL, AT HOME OR IN SOCIAL SITUATIONS | ACCESS TO THE MOST ADAPTED CARE, TREATMENTS OR SURGERY… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 317 | %13 | 1.163 | %46 | 854 | %34 | 127 | %5 | 73 | %3 | 2.534 | %100 |
| No | 516 | %8 | 2.733 | %45 | 2.333 | %38 | 286 | %5 | 249 | %4 | 6.117 | %100 |
| Don't know | 56 | %15 | 124 | %33 | 129 | %35 | 44 | %12 | 20 | %5 | 373 | %100 |
| TOTAL | 889 | %10 | 4.020 | %45 | 3.316 | %37 | 457 | %5 | 342 | %4 | 9.024 | %100 |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 107,0 ; dof= 8.*

## Cross: …behavioural disorders that cause problems in school, at home or in social situations / Understanding how the disease will progress…

| …BEHAVIOURAL DISORDERS THAT CAUSE PROBLEMS IN SCHOOL, AT HOME OR IN SOCIAL SITUATIONS | UNDERSTANDING HOW THE DISEASE WILL PROGRESS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 259 | %10 | 1.386 | %55 | 690 | %27 | 153 | %6 | 40 | %2 | 2.528 | %100 |
| No | 398 | %7 | 3.447 | %56 | 1.832 | %30 | 303 | %5 | 127 | %2 | 6.107 | %100 |
| Don't know | 37 | %10 | 166 | %45 | 122 | %33 | 38 | %10 | 8 | %2 | 371 | %100 |
| TOTAL | 694 | %8 | 4.999 | %56 | 2.644 | %29 | 494 | %5 | 175 | %2 | 9.006 | %100 |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 72,0 ; dof= 8.*

## Cross: …behavioural disorders that cause problems in school, at home or in social situations / Financial support including social security benefits…

| …BEHAVIOURAL DISORDERS THAT CAUSE PROBLEMS IN SCHOOL, AT HOME OR IN SOCIAL SITUATIONS | FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 305 | %23 | 249 | %19 | 496 | %38 | 96 | %7 | 166 | %13 | 1.312 | %100 |
| No | 565 | %13 | 766 | %17 | 1.741 | %40 | 401 | %9 | 921 | %21 | 4.394 | %100 |
| Don't know | 58 | %21 | 41 | %15 | 108 | %38 | 35 | %12 | 39 | %14 | 281 | %100 |
| TOTAL | 928 | %16 | 1.056 | %18 | 2.345 | %39 | 532 | %9 | 1.126 | %19 | 5.987 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 128,0 ; dof= 8.*

## Cross: …behavioural disorders that cause problems in school, at home or in social situations / Integration at school…

| …BEHAVIOURAL DISORDERS THAT CAUSE PROBLEMS IN SCHOOL, AT HOME OR IN SOCIAL SITUATIONS | INTEGRATION AT SCHOOL… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 327 | %13 | 422 | %17 | 653 | %26 | 183 | %7 | 949 | %37 | 2.534 | %100 |
| No | 389 | %6 | 514 | %8 | 1.309 | %21 | 384 | %6 | 3.521 | %58 | 6.117 | %100 |
| Don't know | 30 | %8 | 24 | %6 | 68 | %18 | 46 | %12 | 205 | %55 | 373 | %100 |
| TOTAL | 746 | %8 | 960 | %11 | 2.030 | %22 | 613 | %7 | 4.675 | %52 | 9.024 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 392,9 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: …behavioural disorders that cause problems in school, at home or in social situations / Integration at work…

| …BEHAVIOURAL DISORDERS THAT CAUSE PROBLEMS IN SCHOOL, AT HOME OR IN SOCIAL SITUATIONS | INTEGRATION AT WORK… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 788 | %31 | 244 | %10 | 721 | %29 | 171 | %7 | 604 | %24 | 2.528 | %100 |
| No | 1.515 | %25 | 545 | %9 | 1.774 | %29 | 341 | %6 | 1.931 | %32 | 6.106 | %100 |
| Don't know | 108 | %29 | 29 | %8 | 92 | %25 | 39 | %11 | 103 | %28 | 371 | %100 |
| TOTAL | 2.411 | %27 | 818 | %9 | 2.587 | %29 | 551 | %6 | 2.638 | %29 | 9.005 | |

🔴 *Under-represented elements*    🔵 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 84,8 ; dof= 8.*

## Cross: …behavioural disorders that cause problems in school, at home or in social situations / Access to social services (e.g. social worker support, household chores support)…

| …BEHAVIOURAL DISORDERS THAT CAUSE PROBLEMS IN SCHOOL, AT HOME OR IN SOCIAL SITUATIONS | ACCESS TO SOCIAL SERVICES (E.G. SOCIAL WORKER SUPPORT, HOUSEHOLD CHORES SUPPORT)… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 431 | %17 | 364 | %14 | 924 | %37 | 268 | %11 | 541 | %21 | 2.528 | %100 |
| No | 644 | %11 | 611 | %10 | 1.866 | %31 | 738 | %12 | 2.246 | %37 | 6.105 | %100 |
| Don't know | 59 | %16 | 36 | %10 | 116 | %31 | 60 | %16 | 100 | %27 | 371 | %100 |
| TOTAL | 1.134 | %13 | 1.011 | %11 | 2.906 | %32 | 1.066 | %12 | 2.887 | %32 | 9.004 | |

🔴 *Under-represented elements*    🔵 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 259,5 ; dof= 8.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

## Cross: …behavioural disorders that cause problems in school, at home or in social situations / Access to clinical trials…

| …BEHAVIOURAL DISORDERS THAT CAUSE PROBLEMS IN SCHOOL, AT HOME OR IN SOCIAL SITUATIONS | ACCESS TO CLINICAL TRIALS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 230 | %9 | 623 | %25 | 919 | %36 | 413 | %16 | 349 | %14 | 2.534 | %100 |
| No | 375 | %6 | 1.509 | %25 | 2.138 | %35 | 1.054 | %17 | 1.040 | %17 | 6.116 | %100 |
| Don't know | 32 | %9 | 65 | %17 | 116 | %31 | 97 | %26 | 63 | %17 | 373 | %100 |
| TOTAL | 637 | %7 | 2.197 | %24 | 3.173 | %35 | 1.564 | %17 | 1.452 | %16 | 9.023 | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 63,2 ; dof= 8.*

## Cross: …behavioural disorders that cause problems in school, at home or in social situations / Access to financial products, such as loans, mortgages, insurance…

| …BEHAVIOURAL DISORDERS THAT CAUSE PROBLEMS IN SCHOOL, AT HOME OR IN SOCIAL SITUATIONS | ACCESS TO FINANCIAL PRODUCTS, SUCH AS LOANS, MORTGAGES, INSURANCE… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 514 | %20 | 74 | %3 | 731 | %29 | 507 | %20 | 708 | %28 | 2.534 | %100 |
| No | 1.126 | %18 | 125 | %2 | 1.531 | %25 | 1.044 | %17 | 2.289 | %37 | 6.115 | %100 |
| Don't know | 75 | %20 | 1 | %0 | 83 | %22 | 97 | %26 | 117 | %31 | 373 | %100 |
| TOTAL | 1.715 | %19 | 200 | %2 | 2.345 | %26 | 1.648 | %18 | 3.114 | %35 | 9.022 | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 97,6 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: …behavioural disorders that cause problems in school, at home or in social situations / Your social life…

| …BEHAVIOURAL DISORDERS THAT CAUSE PROBLEMS IN SCHOOL, AT HOME OR IN SOCIAL SITUATIONS | YOUR SOCIAL LIFE… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 1.448 | %57 | 241 | %10 | 702 | %28 | 50 | %2 | 87 | %3 | 2.528 | %100 |
| No | 2.915 | %48 | 454 | %7 | 2.256 | %37 | 110 | %2 | 370 | %6 | 6.105 | %100 |
| Don't know | 208 | %56 | 13 | %4 | 106 | %29 | 23 | %6 | 21 | %6 | 371 | %100 |
| TOTAL | 4.571 | %51 | 708 | %8 | 3.064 | %34 | 183 | %2 | 478 | %5 | 9.004 | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 157,7 ; dof= 8.*

## Cross: …intellectual disabilities or cognitive symptoms (i.e. problems with memory, language, thinking or judgement) / Access to the most adapted care, treatments or surgery…

| …INTELLECTUAL DISABILITIES OR COGNITIVE SYMPTOMS (I.E. PROBLEMS WITH MEMORY, LANGUAGE, THINKING OR JUDGEMENT) | ACCESS TO THE MOST ADAPTED CARE, TREATMENTS OR SURGERY… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 323 | %13 | 1.053 | %42 | 935 | %37 | 120 | %5 | 87 | %3 | 2.518 | %100 |
| No | 535 | %9 | 2.879 | %46 | 2.289 | %37 | 312 | %5 | 243 | %4 | 6.258 | %100 |
| Don't know | 31 | %13 | 88 | %35 | 92 | %37 | 25 | %10 | 12 | %5 | 248 | %100 |
| TOTAL | 889 | %10 | 4.020 | %45 | 3.316 | %37 | 457 | %5 | 342 | %4 | 9.024 | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 61,6 ; dof= 8.*

## Cross: …intellectual disabilities or cognitive symptoms (i.e. problems with memory, language, thinking or judgement) / Understanding how the disease will progress…

| …INTELLECTUAL DISABILITIES OR COGNITIVE SYMPTOMS (I.E. PROBLEMS WITH MEMORY, LANGUAGE, THINKING OR JUDGEMENT) | UNDERSTANDING HOW THE DISEASE WILL PROGRESS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 235 | %9 | 1.368 | %55 | 714 | %28 | 151 | %6 | 41 | %2 | 2.509 | %100 |
| No | 427 | %7 | 3.530 | %56 | 1.851 | %30 | 312 | %5 | 130 | %2 | 6.250 | %100 |
| Don't know | 32 | %13 | 101 | %41 | 79 | %32 | 31 | %13 | 4 | %2 | 247 | %100 |
| TOTAL | 694 | %8 | 4.999 | %56 | 2.644 | %29 | 494 | %5 | 175 | %2 | 9.006 | %100 |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 64,9 ; dof= 8.*

## Cross: …intellectual disabilities or cognitive symptoms (i.e. problems with memory, language, thinking or judgement) / Financial support including social security benefits…

| …INTELLECTUAL DISABILITIES OR COGNITIVE SYMPTOMS (I.E. PROBLEMS WITH MEMORY, LANGUAGE, THINKING OR JUDGEMENT) | FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 281 | %25 | 217 | %19 | 430 | %38 | 83 | %7 | 130 | %11 | 1.141 | %100 |
| No | 604 | %13 | 814 | %17 | 1.848 | %40 | 425 | %9 | 965 | %21 | 4.656 | %100 |
| Don't know | 43 | %23 | 25 | %13 | 67 | %35 | 24 | %13 | 31 | %16 | 190 | %100 |
| TOTAL | 928 | %16 | 1.056 | %18 | 2.345 | %39 | 532 | %9 | 1.126 | %19 | 5.987 | %100 |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 141,6 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: …clinical signs or symptoms that come and go / Access to social services (e.g. social worker support, household chores support)…**

| …CLINICAL SIGNS OR SYMPTOMS THAT COME AND GO | ACCESS TO SOCIAL SERVICES (E.G. SOCIAL WORKER SUPPORT, HOUSEHOLD CHORES SUPPORT)… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 707 | %14 | 553 | %11 | 1.653 | %32 | 601 | %12 | 1.579 | %31 | 5.093 | %100 |
| No | 348 | %11 | 391 | %12 | 1.041 | %32 | 367 | %11 | 1.137 | %35 | 3.284 | %100 |
| Don't know | 79 | %13 | 67 | %11 | 212 | %34 | 98 | %16 | 171 | %27 | 627 | %100 |
| TOTAL | 1.134 | %13 | 1.011 | %11 | 2.906 | %32 | 1.066 | %12 | 2.887 | %32 | 9.004 | %100 |

🟥 *Under-represented elements*　　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 41,9 ; dof= 8.*

**Cross: …clinical signs or symptoms that come and go / Access to clinical trials…**

| …CLINICAL SIGNS OR SYMPTOMS THAT COME AND GO | ACCESS TO CLINICAL TRIALS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 404 | %8 | 1.314 | %26 | 1.818 | %36 | 865 | %17 | 697 | %14 | 5.098 | %100 |
| No | 188 | %6 | 753 | %23 | 1.136 | %34 | 541 | %16 | 675 | %20 | 3.293 | %100 |
| Don't know | 45 | %7 | 130 | %21 | 219 | %35 | 158 | %25 | 80 | %13 | 632 | %100 |
| TOTAL | 637 | %7 | 2.197 | %24 | 3.173 | %35 | 1.564 | %17 | 1.452 | %16 | 9.023 | %100 |

🟥 *Under-represented elements*　　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 112,0 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: …clinical signs or symptoms that come and go / Access to financial products, such as loans, mortgages, insurance…

| …CLINICAL SIGNS OR SYMPTOMS THAT COME AND GO | ACCESS TO FINANCIAL PRODUCTS, SUCH AS LOANS, MORTGAGES, INSURANCE… | | | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 1.080 | %21 | 98 | %2 | 1.345 | %26 | 902 | %18 | 1.673 | %33 | 5.098 | %100 |
| No | 529 | %16 | 87 | %3 | 854 | %26 | 573 | %17 | 1.249 | %38 | 3.292 | %100 |
| Don't know | 106 | %17 | 15 | %2 | 146 | %23 | 173 | %27 | 192 | %30 | 632 | %100 |
| TOTAL | 1.715 | %19 | 200 | %2 | 2.345 | %26 | 1.648 | %18 | 3.114 | %35 | 9.022 | %100 |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 85,9 ; dof= 8.*

## Cross: …clinical signs or symptoms that come and go / Your social life…

| …CLINICAL SIGNS OR SYMPTOMS THAT COME AND GO | YOUR SOCIAL LIFE… | | | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 2.794 | %55 | 392 | %8 | 1.633 | %32 | 85 | %2 | 189 | %4 | 5.093 | %100 |
| No | 1.468 | %45 | 269 | %8 | 1.229 | %37 | 69 | %2 | 249 | %8 | 3.284 | %100 |
| Don't know | 309 | %49 | 47 | %7 | 202 | %32 | 29 | %5 | 40 | %6 | 627 | %100 |
| TOTAL | 4.571 | %51 | 708 | %8 | 3.064 | %34 | 183 | %2 | 478 | %5 | 9.004 | %100 |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 141,1 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: …intellectual disabilities or cognitive symptoms (i.e. problems with memory, language, thinking or judgement) / Access to financial products, such as loans, mortgages, insurance…

| …INTELLECTUAL DISABILITIES OR COGNITIVE SYMPTOMS (I.E. PROBLEMS WITH MEMORY, LANGUAGE, THINKING OR JUDGEMENT) | ACCESS TO FINANCIAL PRODUCTS, SUCH AS LOANS, MORTGAGES, INSURANCE… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 527 | %21 | 84 | %3 | 687 | %27 | 494 | %20 | 726 | %29 | 2.518 | %100 |
| No | 1.138 | %18 | 114 | %2 | 1.601 | %26 | 1.089 | %17 | 2.314 | %37 | 6.256 | %100 |
| Don't know | 50 | %20 | 2 | %1 | 57 | %23 | 65 | %26 | 74 | %30 | 248 | %100 |
| TOTAL | 1.715 | %19 | 200 | %2 | 2.345 | %26 | 1.648 | %18 | 3.114 | %35 | 9.022 | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 80,8 ; dof= 8.*

## Cross: …intellectual disabilities or cognitive symptoms (i.e. problems with memory, language, thinking or judgement) / Your social life…

| …INTELLECTUAL DISABILITIES OR COGNITIVE SYMPTOMS (I.E. PROBLEMS WITH MEMORY, LANGUAGE, THINKING OR JUDGEMENT) | YOUR SOCIAL LIFE… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 1.506 | %60 | 190 | %8 | 676 | %27 | 49 | %2 | 88 | %4 | 2.509 | %100 |
| No | 2.914 | %47 | 506 | %8 | 2.333 | %37 | 121 | %2 | 374 | %6 | 6.248 | %100 |
| Don't know | 151 | %61 | 12 | %5 | 55 | %22 | 13 | %5 | 16 | %6 | 247 | %100 |
| TOTAL | 4.571 | %51 | 708 | %8 | 3.064 | %34 | 183 | %2 | 478 | %5 | 9.004 | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 173,7 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: …clinical signs or symptoms that come and go / Access to the most adapted care, treatments or surgery…

| …CLINICAL SIGNS OR SYMPTOMS THAT COME AND GO | ACCESS TO THE MOST ADAPTED CARE, TREATMENTS OR SURGERY… | | | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 528 | %10 | 2.365 | %46 | 1.830 | %36 | 219 | %4 | 157 | %3 | 5.099 | %100 |
| No | 302 | %9 | 1.411 | %43 | 1.265 | %38 | 166 | %5 | 149 | %5 | 3.293 | %100 |
| Don't know | 59 | %9 | 244 | %39 | 221 | %35 | 72 | %11 | 36 | %6 | 632 | %100 |
| TOTAL | 889 | %10 | 4.020 | %45 | 3.316 | %37 | 457 | %5 | 342 | %4 | 9.024 | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 91,6 ; dof= 8.*

## Cross: …clinical signs or symptoms that come and go / Understanding how the disease will progress…

| …CLINICAL SIGNS OR SYMPTOMS THAT COME AND GO | UNDERSTANDING HOW THE DISEASE WILL PROGRESS… | | | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 427 | %8 | 2.871 | %56 | 1.474 | %29 | 248 | %5 | 74 | %1 | 5.094 | %100 |
| No | 217 | %7 | 1.810 | %55 | 992 | %30 | 175 | %5 | 91 | %3 | 3.285 | %100 |
| Don't know | 50 | %8 | 318 | %51 | 178 | %28 | 71 | %11 | 10 | %2 | 627 | %100 |
| TOTAL | 694 | %8 | 4.999 | %56 | 2.644 | %29 | 494 | %5 | 175 | %2 | 9.006 | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 73,8 ; dof= 8.*

## Cross: …clinical signs or symptoms that come and go / Financial support including social security benefits…

| …CLINICAL SIGNS OR SYMPTOMS THAT COME AND GO | FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 619 | %17 | 655 | %18 | 1.461 | %40 | 307 | %8 | 627 | %17 | 3.669 | %100 |
| No | 248 | %13 | 352 | %18 | 733 | %38 | 159 | %8 | 435 | %23 | 1.927 | %100 |
| Don't know | 61 | %16 | 49 | %13 | 151 | %39 | 66 | %17 | 64 | %16 | 391 | %100 |
| TOTAL | 928 | %16 | 1.056 | %18 | 2.345 | %39 | 532 | %9 | 1.126 | %19 | 5.987 | %100 |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 72,1 ; dof= 8.*

## Cross: …clinical signs or symptoms that come and go / Integration at school…

| …CLINICAL SIGNS OR SYMPTOMS THAT COME AND GO | INTEGRATION AT SCHOOL… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 470 | %9 | 521 | %10 | 1.036 | %20 | 308 | %6 | 2.764 | %54 | 5.099 | %100 |
| No | 220 | %7 | 380 | %12 | 839 | %25 | 221 | %7 | 1.633 | %50 | 3.293 | %100 |
| Don't know | 56 | %9 | 59 | %9 | 155 | %25 | 84 | %13 | 278 | %44 | 632 | %100 |
| TOTAL | 746 | %8 | 960 | %11 | 2.030 | %22 | 613 | %7 | 4.675 | %52 | 9.024 | %100 |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 105,0 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: …clinical signs or symptoms that come and go / Integration at work…

| …CLINICAL SIGNS OR SYMPTOMS THAT COME AND GO | INTEGRATION AT WORK… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 1.513 | %30 | 506 | %10 | 1.415 | %28 | 259 | %5 | 1.401 | %28 | 5.094 | %100 |
| No | 750 | %23 | 271 | %8 | 985 | %30 | 218 | %7 | 1.060 | %32 | 3.284 | %100 |
| Don't know | 148 | %24 | 41 | %7 | 187 | %30 | 74 | %12 | 177 | %28 | 627 | %100 |
| TOTAL | 2.411 | %27 | 818 | %9 | 2.587 | %29 | 551 | %6 | 2.638 | %29 | 9.005 | %100 |

🟥 *Under-represented elements* 🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 111,6 ; dof= 8.*

## Cross: …clinical signs or symptoms that come and go / Access to social services (e.g. social worker support, household chores support)…

| …CLINICAL SIGNS OR SYMPTOMS THAT COME AND GO | ACCESS TO SOCIAL SERVICES (E.G. SOCIAL WORKER SUPPORT, HOUSEHOLD CHORES SUPPORT)… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 707 | %14 | 553 | %11 | 1.653 | %32 | 601 | %12 | 1.579 | %31 | 5.093 | %100 |
| No | 348 | %11 | 391 | %12 | 1.041 | %32 | 367 | %11 | 1.137 | %35 | 3.284 | %100 |
| Don't know | 79 | %13 | 67 | %11 | 212 | %34 | 98 | %16 | 171 | %27 | 627 | %100 |
| TOTAL | 1.134 | %13 | 1.011 | %11 | 2.906 | %32 | 1.066 | %12 | 2.887 | %32 | 9.004 | %100 |

🟥 *Under-represented elements* 🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 41,9 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: …clinical signs or symptoms that come and go / Access to clinical trials…

| …CLINICAL SIGNS OR SYMPTOMS THAT COME AND GO | ACCESS TO CLINICAL TRIALS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 404 | %8 | 1.314 | %26 | 1.818 | %36 | 865 | %17 | 697 | %14 | 5.098 | %100 |
| No | 188 | %6 | 753 | %23 | 1.136 | %34 | 541 | %16 | 675 | %20 | 3.293 | %100 |
| Don't know | 45 | %7 | 130 | %21 | 219 | %35 | 158 | %25 | 80 | %13 | 632 | %100 |
| TOTAL | 637 | %7 | 2.197 | %24 | 3.173 | %35 | 1.564 | %17 | 1.452 | %16 | 9.023 | %100 |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 112,0 ; dof= 8.*

## Cross: …clinical signs or symptoms that come and go / Access to financial products, such as loans, mortgages, insurance…

| …CLINICAL SIGNS OR SYMPTOMS THAT COME AND GO | ACCESS TO FINANCIAL PRODUCTS, SUCH AS LOANS, MORTGAGES, INSURANCE… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 1.080 | %21 | 98 | %2 | 1.345 | %26 | 902 | %18 | 1.673 | %33 | 5.098 | %100 |
| No | 529 | %16 | 87 | %3 | 854 | %26 | 573 | %17 | 1.249 | %38 | 3.292 | %100 |
| Don't know | 106 | %17 | 15 | %2 | 146 | %23 | 173 | %27 | 192 | %30 | 632 | %100 |
| TOTAL | 1.715 | %19 | 200 | %2 | 2.345 | %26 | 1.648 | %18 | 3.114 | %35 | 9.022 | %100 |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 85,9 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: …clinical signs or symptoms that come and go / Your social life…

| …CLINICAL SIGNS OR SYMPTOMS THAT COME AND GO | YOUR SOCIAL LIFE… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 2.794 | %55 | 392 | %8 | 1.633 | %32 | 85 | %2 | 189 | %4 | 5.093 | %100 |
| No | 1.468 | %45 | 269 | %8 | 1.229 | %37 | 69 | %2 | 249 | %8 | 3.284 | %100 |
| Don't know | 309 | %49 | 47 | %7 | 202 | %32 | 29 | %5 | 40 | %6 | 627 | %100 |
| TOTAL | 4.571 | %51 | 708 | %8 | 3.064 | %34 | 183 | %2 | 478 | %5 | 9.004 | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 141,1 ; dof= 8.*

## Cross: …invisible symptoms such as pain, dizziness, headaches, fatigue, etc. / Access to the most adapted care, treatments or surgery…

| …INVISIBLE SYMPTOMS SUCH AS PAIN, DIZZINESS, HEADACHES, FATIGUE, ETC. | ACCESS TO THE MOST ADAPTED CARE, TREATMENTS OR SURGERY… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 648 | %11 | 2.719 | %45 | 2.144 | %36 | 277 | %5 | 193 | %3 | 5.981 | %100 |
| No | 205 | %8 | 1.111 | %43 | 993 | %39 | 137 | %5 | 125 | %5 | 2.571 | %100 |
| Don't know | 36 | %8 | 190 | %40 | 179 | %38 | 43 | %9 | 24 | %5 | 472 | %100 |
| TOTAL | 889 | %10 | 4.020 | %45 | 3.316 | %37 | 457 | %5 | 342 | %4 | 9.024 | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 58,2 ; dof= 8.*

## Cross: …invisible symptoms such as pain, dizziness, headaches, fatigue, etc. / Understanding how the disease will progress…

| | UNDERSTANDING HOW THE DISEASE WILL PROGRESS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| …INVISIBLE SYMPTOMS SUCH AS PAIN, DIZZINESS, HEADACHES, FATIGUE, ETC. | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 515 | %9 | 3.265 | %55 | 1.793 | %30 | 315 | %5 | 85 | %1 | 5.973 | %100 |
| No | 142 | %6 | 1.481 | %58 | 727 | %28 | 136 | %5 | 77 | %3 | 2.563 | %100 |
| Don't know | 37 | %8 | 253 | %54 | 124 | %26 | 43 | %9 | 13 | %3 | 470 | %100 |
| **TOTAL** | **694** | **%8** | **4.999** | **%56** | **2.644** | **%29** | **494** | **%5** | **175** | **%2** | **9.006** | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 65,6 ; dof= 8.*

## Cross: …invisible symptoms such as pain, dizziness, headaches, fatigue, etc. / Financial support including social security benefits…

| | FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| …INVISIBLE SYMPTOMS SUCH AS PAIN, DIZZINESS, HEADACHES, FATIGUE, ETC. | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 788 | %17 | 823 | %18 | 1.830 | %40 | 400 | %9 | 780 | %17 | 4.621 | %100 |
| No | 121 | %10 | 219 | %18 | 470 | %38 | 107 | %9 | 311 | %25 | 1.228 | %100 |
| Don't know | 19 | %14 | 14 | %10 | 45 | %33 | 25 | %18 | 35 | %25 | 138 | %100 |
| **TOTAL** | **928** | **%16** | **1.056** | **%18** | **2.345** | **%39** | **532** | **%9** | **1.126** | **%19** | **5.987** | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 92,8 ; dof= 8.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

## Cross: …invisible symptoms such as pain, dizziness, headaches, fatigue, etc. / Integration at school…

| …INVISIBLE SYMPTOMS SUCH AS PAIN, DIZZINESS, HEADACHES, FATIGUE, ETC. | INTEGRATION AT SCHOOL… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 514 | %9 | 520 | %9 | 1.146 | %19 | 395 | %7 | 3.406 | %57 | 5.981 | %100 |
| No | 191 | %7 | 366 | %14 | 755 | %29 | 174 | %7 | 1.085 | %42 | 2.571 | %100 |
| Don't know | 41 | %9 | 74 | %16 | 129 | %27 | 44 | %9 | 184 | %39 | 472 | %100 |
| TOTAL | 746 | %8 | 960 | %11 | 2.030 | %22 | 613 | %7 | 4.675 | %52 | 9.024 | %100 |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 251,3 ; dof= 8.*

## Cross: …invisible symptoms such as pain, dizziness, headaches, fatigue, etc. / Integration at work…

| …INVISIBLE SYMPTOMS SUCH AS PAIN, DIZZINESS, HEADACHES, FATIGUE, ETC. | INTEGRATION AT WORK… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 1.841 | %31 | 556 | %9 | 1.607 | %27 | 302 | %5 | 1.667 | %28 | 5.973 | %100 |
| No | 485 | %19 | 219 | %9 | 851 | %33 | 197 | %8 | 810 | %32 | 2.562 | %100 |
| Don't know | 85 | %18 | 43 | %9 | 129 | %27 | 52 | %11 | 161 | %34 | 470 | %100 |
| TOTAL | 2.411 | %27 | 818 | %9 | 2.587 | %29 | 551 | %6 | 2.638 | %29 | 9.005 | %100 |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 187,7 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: …invisible symptoms such as pain, dizziness, headaches, fatigue, etc. / Access to social services (e.g. social worker support, household chores support)…

| …INVISIBLE SYMPTOMS SUCH AS PAIN, DIZZINESS, HEADACHES, FATIGUE, ETC. | ACCESS TO SOCIAL SERVICES (E.G. SOCIAL WORKER SUPPORT, HOUSEHOLD CHORES SUPPORT)… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 846 | %14 | 614 | %10 | 1.873 | %31 | 718 | %12 | 1.922 | %32 | 5.973 | %100 |
| No | 235 | %9 | 333 | %13 | 859 | %34 | 286 | %11 | 848 | %33 | 2.561 | %100 |
| Don't know | 53 | %11 | 64 | %14 | 174 | %37 | 62 | %13 | 117 | %25 | 470 | %100 |
| TOTAL | 1.134 | %13 | 1.011 | %11 | 2.906 | %32 | 1.066 | %12 | 2.887 | %32 | 9.004 | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 66,9 ; dof= 8.*

## Cross: …invisible symptoms such as pain, dizziness, headaches, fatigue, etc. / Access to clinical trials…

| …INVISIBLE SYMPTOMS SUCH AS PAIN, DIZZINESS, HEADACHES, FATIGUE, ETC. | ACCESS TO CLINICAL TRIALS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 475 | %8 | 1.408 | %24 | 2.105 | %35 | 1.046 | %17 | 947 | %16 | 5.981 | %100 |
| No | 142 | %6 | 687 | %27 | 895 | %35 | 405 | %16 | 441 | %17 | 2.570 | %100 |
| Don't know | 20 | %4 | 102 | %22 | 173 | %37 | 113 | %24 | 64 | %14 | 472 | %100 |
| TOTAL | 637 | %7 | 2.197 | %24 | 3.173 | %35 | 1.564 | %17 | 1.452 | %16 | 9.023 | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 49,5 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: …invisible symptoms such as pain, dizziness, headaches, fatigue, etc. / Access to financial products, such as loans, mortgages, insurance…

| …INVISIBLE SYMPTOMS SUCH AS PAIN, DIZZINESS, HEADACHES, FATIGUE, ETC. | ACCESS TO FINANCIAL PRODUCTS, SUCH AS LOANS, MORTGAGES, INSURANCE… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 1.316 | %22 | 114 | %2 | 1.468 | %25 | 1.079 | %18 | 2.004 | %34 | 5.981 | %100 |
| No | 341 | %13 | 74 | %3 | 744 | %29 | 461 | %18 | 949 | %37 | 2.569 | %100 |
| Don't know | 58 | %12 | 12 | %3 | 133 | %28 | 108 | %23 | 161 | %34 | 472 | %100 |
| TOTAL | 1.715 | %19 | 200 | %2 | 2.345 | %26 | 1.648 | %18 | 3.114 | %35 | 9.022 | %100 |

🟪 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 118,2 ; dof= 8.*

## Cross: …invisible symptoms such as pain, dizziness, headaches, fatigue, etc. / Your social life…

| …INVISIBLE SYMPTOMS SUCH AS PAIN, DIZZINESS, HEADACHES, FATIGUE, ETC. | YOUR SOCIAL LIFE… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 3.337 | %56 | 468 | %8 | 1.835 | %31 | 105 | %2 | 228 | %4 | 5.973 | %100 |
| No | 1.007 | %39 | 219 | %9 | 1.065 | %42 | 57 | %2 | 213 | %8 | 2.561 | %100 |
| Don't know | 227 | %48 | 21 | %4 | 164 | %35 | 21 | %4 | 37 | %8 | 470 | %100 |
| TOTAL | 4.571 | %51 | 708 | %8 | 3.064 | %34 | 183 | %2 | 478 | %5 | 9.004 | |

🟪 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 258,9 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: …sudden onset symptoms requiring urgent care / Access to the most adapted care, treatments or surgery…

| …SUDDEN ONSET SYMPTOMS REQUIRING URGENT CARE | ACCESS TO THE MOST ADAPTED CARE, TREATMENTS OR SURGERY… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 438 | %11 | 1.930 | %48 | 1.369 | %34 | 169 | %4 | 120 | %3 | 4.026 | %100 |
| No | 395 | %9 | 1.915 | %42 | 1.769 | %39 | 233 | %5 | 202 | %4 | 4.514 | %100 |
| Don't know | 56 | %12 | 175 | %36 | 178 | %37 | 55 | %11 | 20 | %4 | 484 | %100 |
| TOTAL | 889 | %10 | 4.020 | %45 | 3.316 | %37 | 457 | %5 | 342 | %4 | 9.024 | %100 |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 106,2 ; dof= 8.*

## Cross: …sudden onset symptoms requiring urgent care / Understanding how the disease will progress…

| …SUDDEN ONSET SYMPTOMS REQUIRING URGENT CARE | UNDERSTANDING HOW THE DISEASE WILL PROGRESS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 359 | %9 | 2.268 | %56 | 1.133 | %28 | 197 | %5 | 61 | %2 | 4.018 | %100 |
| No | 284 | %6 | 2.512 | %56 | 1.363 | %30 | 243 | %5 | 104 | %2 | 4.506 | %100 |
| Don't know | 51 | %11 | 219 | %45 | 148 | %31 | 54 | %11 | 10 | %2 | 482 | %100 |
| TOTAL | 694 | %8 | 4.999 | %56 | 2.644 | %29 | 494 | %5 | 175 | %2 | 9.006 | %100 |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 75,6 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: …sudden onset symptoms requiring urgent care / Financial support including social security benefits…

| …SUDDEN ONSET SYMPTOMS REQUIRING URGENT CARE | FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 524 | %19 | 488 | %18 | 1.042 | %39 | 204 | %8 | 435 | %16 | 2.693 | %100 |
| No | 351 | %12 | 523 | %18 | 1.173 | %40 | 281 | %9 | 630 | %21 | 2.958 | %100 |
| Don't know | 53 | %16 | 45 | %13 | 130 | %39 | 47 | %14 | 61 | %18 | 336 | %100 |
| **TOTAL** | **928** | **%16** | **1.056** | **%18** | **2.345** | **%39** | **532** | **%9** | **1.126** | **%19** | **5.987** | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 92,8 ; dof= 8.*

## Cross: …sudden onset symptoms requiring urgent care / Integration at school…

| …SUDDEN ONSET SYMPTOMS REQUIRING URGENT CARE | INTEGRATION AT SCHOOL… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 391 | %10 | 433 | %11 | 879 | %22 | 275 | %7 | 2.048 | %51 | 4.026 | %100 |
| No | 311 | %7 | 482 | %11 | 1.043 | %23 | 282 | %6 | 2.396 | %53 | 4.514 | %100 |
| Don't know | 44 | %9 | 45 | %9 | 108 | %22 | 56 | %12 | 231 | %48 | 484 | %100 |
| **TOTAL** | **746** | **%8** | **960** | **%11** | **2.030** | **%22** | **613** | **%7** | **4.675** | **%52** | **9.024** | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 45,2 ; dof= 8.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

## Cross: …sudden onset symptoms requiring urgent care / Integration at work…

| …SUDDEN ONSET SYMPTOMS REQUIRING URGENT CARE | INTEGRATION AT WORK… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 1.282 | %32 | 379 | %9 | 1.056 | %26 | 228 | %6 | 1.073 | %27 | 4.018 | %100 |
| No | 990 | %22 | 397 | %9 | 1.394 | %31 | 280 | %6 | 1.444 | %32 | 4.505 | %100 |
| Don't know | 139 | %29 | 42 | %9 | 137 | %28 | 43 | %9 | 121 | %25 | 482 | %100 |
| TOTAL | 2.411 | %27 | 818 | %9 | 2.587 | %29 | 551 | %6 | 2.638 | %29 | 9.005 | %100 |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 127,4 ; dof= 8.*

## Cross: …sudden onset symptoms requiring urgent care / Access to social services (e.g. social worker support, household chores support)…

| …SUDDEN ONSET SYMPTOMS REQUIRING URGENT CARE | ACCESS TO SOCIAL SERVICES (E.G. SOCIAL WORKER SUPPORT, HOUSEHOLD CHORES SUPPORT)… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 621 | %15 | 467 | %12 | 1.322 | %33 | 486 | %12 | 1.121 | %28 | 4.017 | %100 |
| No | 431 | %10 | 499 | %11 | 1.429 | %32 | 501 | %11 | 1.645 | %37 | 4.505 | %100 |
| Don't know | 82 | %17 | 45 | %9 | 155 | %32 | 79 | %16 | 121 | %25 | 482 | %100 |
| TOTAL | 1.134 | %13 | 1.011 | %11 | 2.906 | %32 | 1.066 | %12 | 2.887 | %32 | 9.004 | %100 |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 137,0 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: …sudden onset symptoms requiring urgent care / Access to clinical trials…

| …SUDDEN ONSET SYMPTOMS REQUIRING URGENT CARE | ACCESS TO CLINICAL TRIALS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 328 | %8 | 1.085 | %27 | 1.390 | %35 | 627 | %16 | 595 | %15 | 4.025 | %100 |
| No | 261 | %6 | 1.015 | %22 | 1.623 | %36 | 813 | %18 | 802 | %18 | 4.514 | %100 |
| Don't know | 48 | %10 | 97 | %20 | 160 | %33 | 124 | %26 | 55 | %11 | 484 | %100 |
| TOTAL | 637 | %7 | 2.197 | %24 | 3.173 | %35 | 1.564 | %17 | 1.452 | %16 | 9.023 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 92,4 ; dof= 8.*

## Cross: …sudden onset symptoms requiring urgent care / Access to financial products, such as loans, mortgages, insurance…

| …SUDDEN ONSET SYMPTOMS REQUIRING URGENT CARE | ACCESS TO FINANCIAL PRODUCTS, SUCH AS LOANS, MORTGAGES, INSURANCE… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 937 | %23 | 86 | %2 | 1.003 | %25 | 709 | %18 | 1.290 | %32 | 4.025 | %100 |
| No | 690 | %15 | 109 | %2 | 1.226 | %27 | 810 | %18 | 1.678 | %37 | 4.513 | %100 |
| Don't know | 88 | %18 | 5 | %1 | 116 | %24 | 129 | %27 | 146 | %30 | 484 | %100 |
| TOTAL | 1.715 | %19 | 200 | %2 | 2.345 | %26 | 1.648 | %18 | 3.114 | %35 | 9.022 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 119,4 ; dof= 8.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: …sudden onset symptoms requiring urgent care / Your social life…

| …SUDDEN ONSET SYMPTOMS REQUIRING URGENT CARE | YOUR SOCIAL LIFE… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | **2.240** | **%56** | **343** | **%9** | **1.194** | **%30** | 69 | %2 | **171** | **%4** | 4.017 | %100 |
| No | **2.074** | **%46** | 344 | %8 | **1.715** | **%38** | 91 | %2 | **281** | **%6** | 4.505 | %100 |
| Don't know | 257 | %53 | **21** | **%4** | 155 | %32 | **23** | **%5** | 26 | %5 | 482 | %100 |
| **TOTAL** | **4.571** | **%51** | **708** | **%8** | **3.064** | **%34** | **183** | **%2** | **478** | **%5** | **9.004** | **%100** |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 129,8 ; dof= 8.*

**Only respondents living with a diagnosed rare disease**

## Cross: The rare disease was diagnosed before birth / Access to the most adapted care, treatments or surgery…

| THE RARE DISEASE WAS DIAGNOSED BEFORE BIRTH | ACCESS TO THE MOST ADAPTED CARE, TREATMENTS OR SURGERY… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 22 | %11 | 84 | %41 | 75 | %36 | 9 | %4 | **16** | **%8** | 206 | %100 |
| No | 865 | %10 | 3.933 | %45 | 3.241 | %37 | 447 | %5 | **326** | **%4** | 8.812 | %100 |
| **TOTAL** | **887** | **%10** | **4.017** | **%45** | **3.316** | **%37** | **456** | **%5** | **342** | **%4** | **9.018** | **%100** |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is significant. p-value= 0,0 ; Chi2= 9,8 ; dof= 4.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

**Only respondents living with a diagnosed rare disease**

### Cross: The rare disease was diagnosed before birth / Understanding how the disease will progress…

| THE RARE DISEASE WAS DIAGNOSED BEFORE BIRTH | UNDERSTANDING HOW THE DISEASE WILL PROGRESS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 18 | %9 | 100 | %49 | 57 | %28 | 20 | %10 | 9 | %4 | 204 | %100 |
| No | 676 | %8 | 4.896 | %56 | 2.586 | %29 | 472 | %5 | 166 | %2 | 8.796 | %100 |
| TOTAL | 694 | %8 | 4.996 | %56 | 2.643 | %29 | 492 | %5 | 175 | %2 | 9.000 | |

🟣 *Under-represented elements*   🔵 *Over-represented elements*

The relationship is very significant. p-value= < 0,01 ; Chi2= 15,8 ; dof= 4.

### Cross: The rare disease was diagnosed before birth / Financial support including social security benefits…

| THE RARE DISEASE WAS DIAGNOSED BEFORE BIRTH | FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 27 | %25 | 17 | %16 | 44 | %41 | 5 | %5 | 14 | %13 | 107 | %100 |
| No | 899 | %15 | 1.038 | %18 | 2.299 | %39 | 527 | %9 | 1.112 | %19 | 5.875 | %100 |
| TOTAL | 926 | %15 | 1.055 | %18 | 2.343 | %39 | 532 | %9 | 1.126 | %19 | 5.982 | |

🟣 *Under-represented elements*   🔵 *Over-represented elements*

The relationship is significant. p-value= 0,0 ; Chi2= 11,1 ; dof= 4.

**Only respondents living with a diagnosed rare disease**

## Cross: The rare disease was diagnosed before birth / Integration at school…

| THE RARE DISEASE WAS DIAGNOSED BEFORE BIRTH | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 16 | %8 | 26 | %13 | 68 | %33 | 16 | %8 | 80 | %39 | 206 | %100 |
| No | 730 | %8 | 933 | %11 | 1.961 | %22 | 597 | %7 | 4.591 | %52 | 8.812 | %100 |
| TOTAL | 746 | %8 | 959 | %11 | 2.029 | %22 | 613 | %7 | 4.671 | %52 | 9.018 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 18,3 ; dof= 4.*

## Cross: The rare disease was diagnosed before birth / Integration at work…

| THE RARE DISEASE WAS DIAGNOSED BEFORE BIRTH | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 42 | %21 | 22 | %11 | 71 | %35 | 17 | %8 | 52 | %25 | 204 | %100 |
| No | 2.367 | %27 | 796 | %9 | 2.515 | %29 | 534 | %6 | 2.583 | %29 | 8.795 | %100 |
| TOTAL | 2.409 | %27 | 818 | %9 | 2.586 | %29 | 551 | %6 | 2.635 | %29 | 8.999 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is weakly significant. p-value= 0,1 ; Chi2= 9,0 ; dof= 4.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Only respondents living with a diagnosed rare disease**

## Cross: The rare disease was diagnosed before birth / Access to social services (e.g. social worker support, household chores support)…

| THE RARE DISEASE WAS DIAGNOSED BEFORE BIRTH | ACCESS TO SOCIAL SERVICES (E.G. SOCIAL WORKER SUPPORT, HOUSEHOLD CHORES SUPPORT)… | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 32 | %16 | 24 | %12 | 66 | %32 | 26 | %13 | 56 | %27 | **204** | **%100** |
| No | 1.101 | %13 | 987 | %11 | 2.838 | %32 | 1.040 | %12 | 2.828 | %32 | **8.794** | **%100** |
| **TOTAL** | **1.133** | **%13** | **1.011** | **%11** | **2.904** | **%32** | **1.066** | **%12** | **2.884** | **%32** | **8.998** | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is not significant. p-value= 0,5 ; Chi2= 3,2 ; dof= 4.*

## Cross: The rare disease was diagnosed before birth / Access to clinical trials…

| THE RARE DISEASE WAS DIAGNOSED BEFORE BIRTH | ACCESS TO CLINICAL TRIALS… | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 21 | %10 | 50 | %24 | 66 | %32 | 31 | %15 | 38 | %18 | **206** | **%100** |
| No | 614 | %7 | 2.146 | %24 | 3.105 | %35 | 1.532 | %17 | 1.414 | %16 | **8.811** | **%100** |
| **TOTAL** | **635** | **%7** | **2.196** | **%24** | **3.171** | **%35** | **1.563** | **%17** | **1.452** | **%16** | **9.017** | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is not significant. p-value= 0,3 ; Chi2= 4,9 ; dof= 4.*

**Only respondents living with a diagnosed rare disease**

## Cross: The rare disease was diagnosed before birth / Access to financial products, such as loans, mortgages, insurance…

| THE RARE DISEASE WAS DIAGNOSED BEFORE BIRTH | ACCESS TO FINANCIAL PRODUCTS, SUCH AS LOANS, MORTGAGES, INSURANCE… | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 37 | %18 | 2 | %1 | 65 | %32 | 35 | %17 | 67 | %33 | 206 | %100 |
| No | 1.677 | %19 | 198 | %2 | 2.279 | %26 | 1.611 | %18 | 3.045 | %35 | 8.810 | %100 |
| TOTAL | 1.714 | %19 | 200 | %2 | 2.344 | %26 | 1.646 | %18 | 3.112 | %35 | 9.016 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is not significant. p-value= 0,3 ; Chi2= 4,5 ; dof= 4.*

## Cross: The rare disease was diagnosed before birth / Your social life…

| THE RARE DISEASE WAS DIAGNOSED BEFORE BIRTH | YOUR SOCIAL LIFE… | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | <u>80</u> | <u>%39</u> | 20 | %10 | <u>84</u> | <u>%41</u> | 5 | %2 | 15 | %7 | 204 | %100 |
| No | <u>4.487</u> | <u>%51</u> | 688 | %8 | <u>2.979</u> | <u>%34</u> | 178 | %2 | 462 | %5 | 8.794 | %100 |
| TOTAL | 4.567 | %51 | 708 | %8 | 3.063 | %34 | 183 | %2 | 477 | %5 | 8.998 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is significant. p-value= 0,0 ; Chi2= 11,4 ; dof= 4.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

**Only respondents living with a diagnosed rare disease**

## Cross: The rare disease was diagnosed through standard tests carried out at birth / Access to the most adapted care, treatments or surgery…

| THE RARE DISEASE WAS DIAGNOSED THROUGH STANDARD TESTS CARRIED OUT AT BIRTH | ACCESS TO THE MOST ADAPTED CARE, TREATMENTS OR SURGERY… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 55 | %10 | 231 | %42 | 194 | %35 | 36 | %7 | 34 | %6 | 550 | %100 |
| No | 832 | %10 | 3.786 | %45 | 3.122 | %37 | 420 | %5 | 308 | %4 | 8.468 | %100 |
| TOTAL | 887 | %10 | 4.017 | %45 | 3.316 | %37 | 456 | %5 | 342 | %4 | 9.018 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is significant. p-value= 0,0 ; Chi2= 12,6 ; dof= 4.*

## Cross: The rare disease was diagnosed through standard tests carried out at birth / Understanding how the disease will progress…

| THE RARE DISEASE WAS DIAGNOSED THROUGH STANDARD TESTS CARRIED OUT AT BIRTH | UNDERSTANDING HOW THE DISEASE WILL PROGRESS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 24 | %7 | 202 | %56 | 95 | %26 | 22 | %6 | 19 | %5 | 362 | %100 |
| No | 653 | %8 | 4.708 | %56 | 2.483 | %29 | 457 | %5 | 152 | %2 | 8.453 | %100 |
| TOTAL | 677 | %8 | 4.910 | %56 | 2.578 | %29 | 479 | %5 | 171 | %2 | 8.815 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 23,3 ; dof= 4.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Only respondents living with a diagnosed rare disease**

## Cross: The rare disease was diagnosed through standard tests carried out at birth / Financial support including social security benefits…

| THE RARE DISEASE WAS DIAGNOSED THROUGH STANDARD TESTS CARRIED OUT AT BIRTH | FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS… | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 42 | %24 | 28 | %16 | 55 | %31 | 21 | %12 | 31 | %18 | 177 | %100 |
| No | 884 | %15 | 1.027 | %18 | 2.288 | %39 | 511 | %9 | 1.095 | %19 | 5.805 | %100 |
| TOTAL | 926 | %15 | 1.055 | %18 | 2.343 | %39 | 532 | %9 | 1.126 | %19 | 5.982 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 13,4 ; dof= 4.*

## Cross: The rare disease was diagnosed through standard tests carried out at birth / Integration at school…

| THE RARE DISEASE WAS DIAGNOSED THROUGH STANDARD TESTS CARRIED OUT AT BIRTH | INTEGRATION AT SCHOOL… | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 33 | %9 | 70 | %19 | 127 | %35 | 43 | %12 | 90 | %25 | 363 | %100 |
| No | 696 | %8 | 860 | %10 | 1.837 | %22 | 552 | %7 | 4.525 | %53 | 8.470 | %100 |
| TOTAL | 729 | %8 | 930 | %11 | 1.964 | %22 | 595 | %7 | 4.615 | %52 | 8.833 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 124,8 ; dof= 4.*

**Only respondents living with a diagnosed rare disease**

## Cross: The rare disease was diagnosed through standard tests carried out at birth / Integration at work…

| THE RARE DISEASE WAS DIAGNOSED THROUGH STANDARD TESTS CARRIED OUT AT BIRTH | INTEGRATION AT WORK… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 109 | %20 | 66 | %12 | 165 | %30 | 68 | %12 | 141 | %26 | 549 | %100 |
| No | 2.300 | %27 | 752 | %9 | 2.421 | %29 | 483 | %6 | 2.494 | %30 | 8.450 | %100 |
| TOTAL | 2.409 | %27 | 818 | %9 | 2.586 | %29 | 551 | %6 | 2.635 | %29 | 8.999 | |

■ *Under-represented elements* ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 56,4 ; dof= 4.*

## Cross: The rare disease was diagnosed through standard tests carried out at birth / Access to social services (e.g. social worker support, household chores support)…

| THE RARE DISEASE WAS DIAGNOSED THROUGH STANDARD TESTS CARRIED OUT AT BIRTH | ACCESS TO SOCIAL SERVICES (E.G. SOCIAL WORKER SUPPORT, HOUSEHOLD CHORES SUPPORT)… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 54 | %15 | 61 | %17 | 106 | %29 | 56 | %16 | 84 | %23 | 361 | %100 |
| No | 1.051 | %12 | 921 | %11 | 2.740 | %32 | 977 | %12 | 2.763 | %33 | 8.452 | %100 |
| TOTAL | 1.105 | %13 | 982 | %11 | 2.846 | %32 | 1.033 | %12 | 2.847 | %32 | 8.813 | |

■ *Under-represented elements* ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 28,1 ; dof= 4.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

**Only respondents living with a diagnosed rare disease**

## Cross: The rare disease was diagnosed through standard tests carried out at birth / Access to clinical trials…

| THE RARE DISEASE WAS DIAGNOSED THROUGH STANDARD TESTS CARRIED OUT AT BIRTH | ACCESS TO CLINICAL TRIALS… | | | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 37 | %7 | 135 | %25 | 188 | %34 | 105 | %19 | 84 | %15 | 549 | %100 |
| No | 598 | %7 | 2.061 | %24 | 2.983 | %35 | 1.458 | %17 | 1.368 | %16 | 8.468 | %100 |
| TOTAL | 635 | %7 | 2.196 | %24 | 3.171 | %35 | 1.563 | %17 | 1.452 | %16 | 9.017 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is not significant. p-value= 0,8 ; Chi2= 1,5 ; dof= 4.*

## Cross: The rare disease was diagnosed through standard tests carried out at birth / Access to financial products, such as loans, mortgages, insurance…

| THE RARE DISEASE WAS DIAGNOSED THROUGH STANDARD TESTS CARRIED OUT AT BIRTH | ACCESS TO FINANCIAL PRODUCTS, SUCH AS LOANS, MORTGAGES, INSURANCE… | | | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 37 | %10 | 14 | %4 | 88 | %24 | 106 | %29 | 117 | %32 | 362 | %100 |
| No | 1.643 | %19 | 179 | %2 | 2.201 | %26 | 1.503 | %18 | 2.943 | %35 | 8.469 | %100 |
| TOTAL | 1.680 | %19 | 193 | %2 | 2.289 | %26 | 1.609 | %18 | 3.060 | %35 | 8.831 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 46,6 ; dof= 4.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

**Only respondents living with a diagnosed rare disease**

## Cross: The rare disease was diagnosed through standard tests carried out at birth / Your social life…

| THE RARE DISEASE WAS DIAGNOSED THROUGH STANDARD TESTS CARRIED OUT AT BIRTH | YOUR SOCIAL LIFE… | | | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 141 | %39 | 54 | %15 | 124 | %34 | 16 | %4 | 26 | %7 | 361 | %100 |
| No | 4.352 | %51 | 634 | %8 | 2.864 | %34 | 162 | %2 | 440 | %5 | 8.452 | %100 |
| TOTAL | 4.493 | %51 | 688 | %8 | 2.988 | %34 | 178 | %2 | 466 | %5 | 8.813 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 48,6 ; dof= 4.*

## Cross: Family members were previously diagnosed with the same disease / Access to the most adapted care, treatments or surgery…

| FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | ACCESS TO THE MOST ADAPTED CARE, TREATMENTS OR SURGERY… | | | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 131 | %11 | 520 | %43 | 420 | %35 | 74 | %6 | 52 | %4 | 1.197 | %100 |
| No | 756 | %10 | 3.497 | %45 | 2.896 | %37 | 382 | %5 | 290 | %4 | 7.821 | %100 |
| TOTAL | 887 | %10 | 4.017 | %45 | 3.316 | %37 | 456 | %5 | 342 | %4 | 9.018 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is weakly significant. p-value= 0,1 ; Chi2= 7,7 ; dof= 4.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Only respondents living with a diagnosed rare disease**

### Cross: Family members were previously diagnosed with the same disease / Understanding how the disease will progress…

| FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | UNDERSTANDING HOW THE DISEASE WILL PROGRESS… | | | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | ...HAS GOTTEN WORSE | | ...HAS IMPROVED | | ...HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 101 | %8 | 654 | %55 | 384 | %32 | 39 | %3 | 17 | %1 | 1.195 | %100 |
| No | 593 | %8 | 4.342 | %56 | 2.259 | %29 | 453 | %6 | 158 | %2 | 7.805 | %100 |
| **TOTAL** | **694** | **%8** | **4.996** | **%56** | **2.643** | **%29** | **492** | **%5** | **175** | **%2** | **9.000** | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 18,9 ; dof= 4.*

### Cross: Family members were previously diagnosed with the same disease / Financial support including social security benefits…

| FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS… | | | | | | | | | | | |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| | ...HAS GOTTEN WORSE | | ...HAS IMPROVED | | ...HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 127 | %13 | 141 | %14 | 400 | %41 | 109 | %11 | 200 | %20 | 977 | %100 |
| No | 799 | %16 | 914 | %18 | 1.943 | %39 | 423 | %8 | 926 | %19 | 5.005 | %100 |
| **TOTAL** | **926** | **%15** | **1.055** | **%18** | **2.343** | **%39** | **532** | **%9** | **1.126** | **%19** | **5.982** | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 20,8 ; dof= 4.*

**Only respondents living with a diagnosed rare disease**

## Cross: Family members were previously diagnosed with the same disease / Integration at school…

| FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | INTEGRATION AT SCHOOL… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 68 | %6 | 81 | %7 | 291 | %24 | 87 | %7 | 670 | %56 | 1.197 | %100 |
| No | 678 | %9 | 878 | %11 | 1.738 | %22 | 526 | %7 | 4.001 | %51 | 7.821 | %100 |
| TOTAL | 746 | %8 | 959 | %11 | 2.029 | %22 | 613 | %7 | 4.671 | %52 | 9.018 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 37,7 ; dof= 4.*

## Cross: Family members were previously diagnosed with the same disease / Integration at work…

| FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | INTEGRATION AT WORK… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 277 | %23 | 95 | %8 | 369 | %31 | 86 | %7 | 368 | %31 | 1.195 | %100 |
| No | 2.132 | %27 | 723 | %9 | 2.217 | %28 | 465 | %6 | 2.267 | %29 | 7.804 | %100 |
| TOTAL | 2.409 | %27 | 818 | %9 | 2.586 | %29 | 551 | %6 | 2.635 | %29 | 8.999 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 14,5 ; dof= 4.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

**Only respondents living with a diagnosed rare disease**

## Cross: Family members were previously diagnosed with the same disease / Access to social services (e.g. social worker support, household chores support)…

| FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | ACCESS TO SOCIAL SERVICES (E.G. SOCIAL WORKER SUPPORT, HOUSEHOLD CHORES SUPPORT)… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | ...HAS GOTTEN WORSE | | ...HAS IMPROVED | | ...HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 134 | %11 | 113 | %9 | 340 | %28 | 147 | %12 | 461 | %39 | 1.195 | %100 |
| No | 999 | %13 | 898 | %12 | 2.564 | %33 | 919 | %12 | 2.423 | %31 | 7.803 | %100 |
| TOTAL | 1.133 | %13 | 1.011 | %11 | 2.904 | %32 | 1.066 | %12 | 2.884 | %32 | 8.998 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 30,8 ; dof= 4.*

## Cross: Family members were previously diagnosed with the same disease / Access to clinical trials…

| FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | ACCESS TO CLINICAL TRIALS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | ...HAS GOTTEN WORSE | | ...HAS IMPROVED | | ...HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| Yes | 85 | %7 | 345 | %29 | 393 | %33 | 192 | %16 | 182 | %15 | 1.197 | %100 |
| No | 550 | %7 | 1.851 | %24 | 2.778 | %36 | 1.371 | %18 | 1.270 | %16 | 7.820 | %100 |
| TOTAL | 635 | %7 | 2.196 | %24 | 3.171 | %35 | 1.563 | %17 | 1.452 | %16 | 9.017 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 15,5 ; dof= 4.*

**Only respondents living with a diagnosed rare disease**

### Cross: Family members were previously diagnosed with the same disease / Access to financial products, such as loans, mortgages, insurance…

| | ACCESS TO FINANCIAL PRODUCTS, SUCH AS LOANS, MORTGAGES, INSURANCE… | | | | | | | | | | | |
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | N | % | N | % | N | % | N | % | N | % | N | % |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| Yes | 262 | %22 | 26 | %2 | 306 | %26 | 213 | %18 | 390 | %33 | 1.197 | %100 |
| No | 1.452 | %19 | 174 | %2 | 2.038 | %26 | 1.433 | %18 | 2.722 | %35 | 7.819 | %100 |
| TOTAL | 1.714 | %19 | 200 | %2 | 2.344 | %26 | 1.646 | %18 | 3.112 | %35 | 9.016 | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is weakly significant. p-value= 0,1 ; Chi2= 7,8 ; dof= 4.*

### Cross: Family members were previously diagnosed with the same disease / Your social life…

| | YOUR SOCIAL LIFE… | | | | | | | | | | | |
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE | N | % | N | % | N | % | N | % | N | % | N | % |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| Yes | 521 | %44 | 95 | %8 | 465 | %39 | 31 | %3 | 83 | %7 | 1.195 | %100 |
| No | 4.046 | %52 | 613 | %8 | 2.598 | %33 | 152 | %2 | 394 | %5 | 7.803 | %100 |
| TOTAL | 4.567 | %51 | 708 | %8 | 3.063 | %34 | 183 | %2 | 477 | %5 | 8.998 | |

🟥 *Under-represented elements*　🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 32,7 ; dof= 4.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: …psychological support / Access to the most adapted care, treatments or surgery…

| …PSYCHOLOGICAL SUPPORT | ACCESS TO THE MOST ADAPTED CARE, TREATMENTS OR SURGERY… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES and enough to meet my needs | 66 | %9 | 384 | %50 | 261 | %34 | 41 | %5 | 20 | %3 | 772 | %100 |
| YES but it is/was not needed | 62 | %8 | 381 | %46 | 306 | %37 | 30 | %4 | 43 | %5 | 822 | %100 |
| YES but NOT enough to meet my needs | 104 | %13 | 348 | %44 | 278 | %35 | 44 | %6 | 23 | %3 | 797 | %100 |
| NO but it is/was NOT needed | 217 | %8 | 1.232 | %45 | 1.035 | %37 | 142 | %5 | 136 | %5 | 2.762 | %100 |
| NO but it is/was needed | 440 | %11 | 1.675 | %43 | 1.436 | %37 | 200 | %5 | 120 | %3 | 3.871 | %100 |
| TOTAL | 889 | %10 | 4.020 | %45 | 3.316 | %37 | 457 | %5 | 342 | %4 | 9.024 | %100 |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 71,4 ; dof= 16.*

## Cross: …psychological support / Understanding how the disease will progress…

| …PSYCHOLOGICAL SUPPORT | UNDERSTANDING HOW THE DISEASE WILL PROGRESS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES and enough to meet my needs | 63 | %8 | 466 | %61 | 204 | %26 | 24 | %3 | 13 | %2 | 770 | %100 |
| YES but it is/was not needed | 48 | %6 | 466 | %57 | 247 | %30 | 42 | %5 | 17 | %2 | 820 | %100 |
| YES but NOT enough to meet my needs | 75 | %9 | 409 | %51 | 241 | %30 | 52 | %7 | 19 | %2 | 796 | %100 |
| NO but it is/was NOT needed | 146 | %5 | 1.545 | %56 | 850 | %31 | 147 | %5 | 71 | %3 | 2.759 | %100 |
| NO but it is/was needed | 362 | %9 | 2.113 | %55 | 1.102 | %29 | 229 | %6 | 55 | %1 | 3.861 | %100 |
| TOTAL | 694 | %8 | 4.999 | %56 | 2.644 | %29 | 494 | %5 | 175 | %2 | 9.006 | %100 |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 77,2 ; dof= 16.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

## Cross: …psychological support / Financial support including social security benefits…

| | FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS… | | | | | | | | | | | |
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| …PSYCHOLOGICAL SUPPORT | N | % | N | % | N | % | N | % | N | % | N | % |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| YES and enough to meet my needs | 63 | %14 | 95 | %22 | 176 | %40 | 22 | %5 | 84 | %19 | 440 | %100 |
| YES but it is/was not needed | 72 | %13 | 99 | %18 | 218 | %40 | 51 | %9 | 107 | %20 | 547 | %100 |
| YES but NOT enough to meet my needs | 113 | %25 | 78 | %17 | 185 | %40 | 26 | %6 | 57 | %12 | 459 | %100 |
| NO but it is/was NOT needed | 215 | %10 | 325 | %16 | 795 | %38 | 217 | %10 | 539 | %26 | 2.091 | %100 |
| NO but it is/was needed | 465 | %19 | 459 | %19 | 971 | %40 | 216 | %9 | 339 | %14 | 2.450 | %100 |
| TOTAL | 928 | %16 | 1.056 | %18 | 2.345 | %39 | 532 | %9 | 1.126 | %19 | 5.987 | %100 |

🟥 *Under-represented elements*     🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 209,4 ; dof= 16.*

## Cross: …psychological support / Integration at school…

| | INTEGRATION AT SCHOOL… | | | | | | | | | | | |
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| …PSYCHOLOGICAL SUPPORT | N | % | N | % | N | % | N | % | N | % | N | % |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| YES and enough to meet my needs | 51 | %7 | 130 | %17 | 156 | %20 | 45 | %6 | 390 | %51 | 772 | %100 |
| YES but it is/was not needed | 52 | %6 | 85 | %10 | 171 | %21 | 56 | %7 | 458 | %56 | 822 | %100 |
| YES but NOT enough to meet my needs | 110 | %14 | 106 | %13 | 161 | %20 | 51 | %6 | 369 | %46 | 797 | %100 |
| NO but it is/was NOT needed | 133 | %5 | 216 | %8 | 614 | %22 | 200 | %7 | 1.599 | %58 | 2.762 | %100 |
| NO but it is/was needed | 400 | %10 | 423 | %11 | 928 | %24 | 261 | %7 | 1.859 | %48 | 3.871 | %100 |
| TOTAL | 746 | %8 | 960 | %11 | 2.030 | %22 | 613 | %7 | 4.675 | %52 | 9.024 | %100 |

🟥 *Under-represented elements*     🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 198,3 ; dof= 16.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: …psychological support / Integration at work…

| | INTEGRATION AT WORK… | | | | | | | | | | | |
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| …PSYCHOLOGICAL SUPPORT | N | % | N | % | N | % | N | % | N | % | N | % |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| YES and enough to meet my needs | 199 | %26 | 80 | %10 | 207 | %27 | 33 | %4 | 251 | %33 | 770 | %100 |
| YES but it is/was not needed | 190 | %23 | 89 | %11 | 223 | %27 | 49 | %6 | 269 | %33 | 820 | %100 |
| YES but NOT enough to meet my needs | 257 | %32 | 81 | %10 | 178 | %22 | 56 | %7 | 224 | %28 | 796 | %100 |
| NO but it is/was NOT needed | 538 | %20 | 223 | %8 | 845 | %31 | 181 | %7 | 971 | %35 | 2.758 | %100 |
| NO but it is/was needed | 1.227 | %32 | 345 | %9 | 1.134 | %29 | 232 | %6 | 923 | %24 | 3.861 | %100 |
| TOTAL | 2.411 | %27 | 818 | %9 | 2.587 | %29 | 551 | %6 | 2.638 | %29 | 9.005 | %100 |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 213,3 ; dof= 16.*

## Cross: …psychological support / Access to social services (e.g. social worker support, household chores support)…

| | ACCESS TO SOCIAL SERVICES (E.G. SOCIAL WORKER SUPPORT, HOUSEHOLD CHORES SUPPORT)… | | | | | | | | | | | |
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| …PSYCHOLOGICAL SUPPORT | N | % | N | % | N | % | N | % | N | % | N | % |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| YES and enough to meet my needs | 78 | %10 | 160 | %21 | 236 | %31 | 76 | %10 | 220 | %29 | 770 | %100 |
| YES but it is/was not needed | 59 | %7 | 115 | %14 | 260 | %32 | 95 | %12 | 291 | %35 | 820 | %100 |
| YES but NOT enough to meet my needs | 149 | %19 | 124 | %16 | 264 | %33 | 91 | %11 | 168 | %21 | 796 | %100 |
| NO but it is/was NOT needed | 203 | %7 | 228 | %8 | 752 | %27 | 327 | %12 | 1.247 | %45 | 2.757 | %100 |
| NO but it is/was needed | 645 | %17 | 384 | %10 | 1.394 | %36 | 477 | %12 | 961 | %25 | 3.861 | %100 |
| TOTAL | 1.134 | %13 | 1.011 | %11 | 2.906 | %32 | 1.066 | %12 | 2.887 | %32 | 9.004 | %100 |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 557,1 ; dof= 16.*

## Cross: …psychological support / Access to clinical trials…

| …PSYCHOLOGICAL SUPPORT | ACCESS TO CLINICAL TRIALS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES and enough to meet my needs | 38 | %5 | 228 | %30 | 243 | %31 | 131 | %17 | 132 | %17 | 772 | %100 |
| YES but it is/was not needed | 40 | %5 | 200 | %24 | 301 | %37 | 126 | %15 | 155 | %19 | 822 | %100 |
| YES but NOT enough to meet my needs | 88 | %11 | 187 | %23 | 269 | %34 | 140 | %18 | 113 | %14 | 797 | %100 |
| NO but it is/was NOT needed | 140 | %5 | 693 | %25 | 945 | %34 | 467 | %17 | 516 | %19 | 2.761 | %100 |
| NO but it is/was needed | 331 | %9 | 889 | %23 | 1.415 | %37 | 700 | %18 | 536 | %14 | 3.871 | %100 |
| TOTAL | 637 | %7 | 2.197 | %24 | 3.173 | %35 | 1.564 | %17 | 1.452 | %16 | 9.023 | |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 108,8 ; dof= 16.*

## Cross: …psychological support / Access to financial products, such as loans, mortgages, insurance…

| …PSYCHOLOGICAL SUPPORT | ACCESS TO FINANCIAL PRODUCTS, SUCH AS LOANS, MORTGAGES, INSURANCE… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES and enough to meet my needs | 140 | %18 | 24 | %3 | 182 | %24 | 132 | %17 | 294 | %38 | 772 | %100 |
| YES but it is/was not needed | 160 | %19 | 23 | %3 | 200 | %24 | 142 | %17 | 297 | %36 | 822 | %100 |
| YES but NOT enough to meet my needs | 195 | %24 | 23 | %3 | 197 | %25 | 155 | %19 | 227 | %28 | 797 | %100 |
| NO but it is/was NOT needed | 361 | %13 | 47 | %2 | 699 | %25 | 474 | %17 | 1.179 | %43 | 2.760 | %100 |
| NO but it is/was needed | 859 | %22 | 83 | %2 | 1.067 | %28 | 745 | %19 | 1.117 | %29 | 3.871 | %100 |
| TOTAL | 1.715 | %19 | 200 | %2 | 2.345 | %26 | 1.648 | %18 | 3.114 | %35 | 9.022 | |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 207,9 ; dof= 16.*

## Cross: …psychological support / Your social life…

| …PSYCHOLOGICAL SUPPORT | YOUR SOCIAL LIFE… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES and enough to meet my needs | 364 | %47 | 83 | %11 | 269 | %35 | 17 | %2 | 37 | %5 | 770 | %100 |
| YES but it is/was not needed | 369 | %45 | 71 | %9 | 312 | %38 | 12 | %1 | 56 | %7 | 820 | %100 |
| YES but NOT enough to meet my needs | 523 | %66 | 71 | %9 | 172 | %22 | 11 | %1 | 19 | %2 | 796 | %100 |
| NO but it is/was NOT needed | 1.041 | %38 | 188 | %7 | 1.226 | %44 | 63 | %2 | 239 | %9 | 2.757 | %100 |
| NO but it is/was needed | 2.274 | %59 | 295 | %8 | 1.085 | %28 | 80 | %2 | 127 | %3 | 3.861 | %100 |
| TOTAL | 4.571 | %51 | 708 | %8 | 3.064 | %34 | 183 | %2 | 478 | %5 | 9.004 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 475,9 ; dof= 16.*

## Cross: …care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc. / Access to the most adapted care, treatments or surgery…

| …CARE COORDINATION SUPPORT SUCH AS HELP TO FIND THE NECESSARY INFORMATION ON THE DISEASE AND THE RIGHT PROFESSIONALS, ARRANGING APPOINTMENTS WITH DIFFERENT HEALTH PROVIDERS, ETC. | ACCESS TO THE MOST ADAPTED CARE, TREATMENTS OR SURGERY… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES and enough to meet my needs | 101 | %6 | 960 | %53 | 622 | %34 | 73 | %4 | 61 | %3 | 1.817 | %100 |
| YES but it is/was not needed | 26 | %8 | 147 | %46 | 121 | %38 | 12 | %4 | 12 | %4 | 318 | %100 |
| YES but NOT enough to meet my needs | 135 | %11 | 558 | %44 | 451 | %36 | 67 | %5 | 46 | %4 | 1.257 | %100 |
| NO but it is/was NOT needed | 98 | %7 | 596 | %43 | 541 | %39 | 84 | %6 | 81 | %6 | 1.400 | %100 |
| NO but it is/was needed | 529 | %13 | 1.759 | %42 | 1.581 | %37 | 221 | %5 | 142 | %3 | 4.232 | %100 |
| TOTAL | 889 | %10 | 4.020 | %45 | 3.316 | %37 | 457 | %5 | 342 | %4 | 9.024 | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 146,4 ; dof= 16.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: …care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc. / Understanding how the disease will progress…**

| …CARE COORDINATION SUPPORT SUCH AS HELP TO FIND THE NECESSARY INFORMATION ON THE DISEASE AND THE RIGHT PROFESSIONALS, ARRANGING APPOINTMENTS WITH DIFFERENT HEALTH PROVIDERS, ETC. | UNDERSTANDING HOW THE DISEASE WILL PROGRESS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES and enough to meet my needs | **87** | **%5** | **1.145** | **%63** | **478** | **%26** | **68** | **%4** | 35 | %2 | **1.813** | **%100** |
| YES but it is/was not needed | 19 | %6 | 171 | %54 | 106 | %33 | 18 | %6 | 4 | %1 | **318** | **%100** |
| YES but NOT enough to meet my needs | 107 | %9 | 685 | %55 | 360 | %29 | 76 | %6 | 26 | %2 | **1.254** | **%100** |
| NO but it is/was NOT needed | **73** | **%5** | 759 | %54 | **449** | **%32** | 76 | %5 | **41** | **%3** | **1.398** | **%100** |
| NO but it is/was needed | **408** | **%10** | **2.239** | **%53** | 1.251 | %30 | **256** | **%6** | **69** | **%2** | **4.223** | **%100** |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 113,4 ; dof= 16.*

**Cross: …care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc. / Financial support including social security benefits…**

| …CARE COORDINATION SUPPORT SUCH AS HELP TO FIND THE NECESSARY INFORMATION ON THE DISEASE AND THE RIGHT PROFESSIONALS, ARRANGING APPOINTMENTS WITH DIFFERENT HEALTH PROVIDERS, ETC. | FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES and enough to meet my needs | **111** | **%10** | 210 | %19 | 441 | %40 | 104 | %9 | **243** | **%22** | **1.109** | **%100** |
| YES but it is/was not needed | 23 | %11 | 40 | %19 | 70 | %33 | 24 | %11 | **53** | **%25** | **210** | **%100** |
| YES but NOT enough to meet my needs | **150** | **%21** | 135 | %19 | 276 | %38 | 56 | %8 | **107** | **%15** | **724** | **%100** |
| NO but it is/was NOT needed | **91** | **%8** | **158** | **%15** | 418 | %39 | 106 | %10 | **309** | **%29** | **1.082** | **%100** |
| NO but it is/was needed | **553** | **%19** | 513 | %18 | 1.140 | %40 | 242 | %8 | **414** | **%14** | **2.862** | **%100** |
| **TOTAL** | **928** | **%16** | **1.056** | **%18** | **2.345** | **%39** | **532** | **%9** | **1.126** | **%19** | **5.987** | |

🟥 *Under-represented elements*    🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 213,6 ; dof= 16.*

**Cross: …care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc. / Integration at school…**

| …CARE COORDINATION SUPPORT SUCH AS HELP TO FIND THE NECESSARY INFORMATION ON THE DISEASE AND THE RIGHT PROFESSIONALS, ARRANGING APPOINTMENTS WITH DIFFERENT HEALTH PROVIDERS, ETC. | INTEGRATION AT SCHOOL… | | | | | | | | | | | |
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES and enough to meet my needs | 107 | %6 | 266 | %15 | 402 | %22 | 124 | %7 | 918 | %51 | 1.817 | %100 |
| YES but it is/was not needed | 20 | %6 | 25 | %8 | 70 | %22 | 34 | %11 | 169 | %53 | 318 | %100 |
| YES but NOT enough to meet my needs | 111 | %9 | 171 | %14 | 317 | %25 | 90 | %7 | 568 | %45 | 1.257 | %100 |
| NO but it is/was NOT needed | 65 | %5 | 104 | %7 | 297 | %21 | 89 | %6 | 845 | %60 | 1.400 | %100 |
| NO but it is/was needed | 443 | %10 | 394 | %9 | 944 | %22 | 276 | %7 | 2.175 | %51 | 4.232 | %100 |
| TOTAL | 746 | %8 | 960 | %11 | 2.030 | %22 | 613 | %7 | 4.675 | %52 | 9.024 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 166,9 ; dof= 16.*

**Cross: …care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc. / Integration at work…**

| …CARE COORDINATION SUPPORT SUCH AS HELP TO FIND THE NECESSARY INFORMATION ON THE DISEASE AND THE RIGHT PROFESSIONALS, ARRANGING APPOINTMENTS WITH DIFFERENT HEALTH PROVIDERS, ETC. | INTEGRATION AT WORK… | | | | | | | | | | | |
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES and enough to meet my needs | 365 | %20 | 213 | %12 | 526 | %29 | 102 | %6 | 607 | %33 | 1.813 | %100 |
| YES but it is/was not needed | 71 | %22 | 18 | %6 | 95 | %30 | 34 | %11 | 100 | %31 | 318 | %100 |
| YES but NOT enough to meet my needs | 384 | %31 | 95 | %8 | 355 | %28 | 91 | %7 | 329 | %26 | 1.254 | %100 |
| NO but it is/was NOT needed | 256 | %18 | 118 | %8 | 427 | %31 | 92 | %7 | 504 | %36 | 1.397 | %100 |
| NO but it is/was needed | 1.335 | %32 | 374 | %9 | 1.184 | %28 | 232 | %5 | 1.098 | %26 | 4.223 | %100 |
| TOTAL | 2.411 | %27 | 818 | %9 | 2.587 | %29 | 551 | %6 | 2.638 | %29 | 9.005 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 208,6 ; dof= 16.*

**Cross: …care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc. / Access to social services (e.g. social worker support, household chores support)…**

| …CARE COORDINATION SUPPORT SUCH AS HELP TO FIND THE NECESSARY INFORMATION ON THE DISEASE AND THE RIGHT PROFESSIONALS, ARRANGING APPOINTMENTS WITH DIFFERENT HEALTH PROVIDERS, ETC. | ACCESS TO SOCIAL SERVICES (E.G. SOCIAL WORKER SUPPORT, HOUSEHOLD CHORES SUPPORT)… | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES and enough to meet my needs | 144 | %8 | 294 | %16 | 506 | %28 | 191 | %11 | 677 | %37 | 1.812 | %100 |
| YES but it is/was not needed | 25 | %8 | 31 | %10 | 107 | %34 | 42 | %13 | 113 | %36 | 318 | %100 |
| YES but NOT enough to meet my needs | 190 | %15 | 155 | %12 | 423 | %34 | 169 | %13 | 317 | %25 | 1.254 | %100 |
| NO but it is/was NOT needed | 100 | %7 | 100 | %7 | 367 | %26 | 162 | %12 | 668 | %48 | 1.397 | %100 |
| NO but it is/was needed | 675 | %16 | 431 | %10 | 1.503 | %36 | 502 | %12 | 1.112 | %26 | 4.223 | %100 |
| TOTAL | 1.134 | %13 | 1.011 | %11 | 2.906 | %32 | 1.066 | %12 | 2.887 | %32 | 9.004 | |

🟥 *Under-represented elements* 🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 415,5 ; dof= 16.*

**Cross: …care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc. / Access to clinical trials…**

| …CARE COORDINATION SUPPORT SUCH AS HELP TO FIND THE NECESSARY INFORMATION ON THE DISEASE AND THE RIGHT PROFESSIONALS, ARRANGING APPOINTMENTS WITH DIFFERENT HEALTH PROVIDERS, ETC. | ACCESS TO CLINICAL TRIALS… | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES and enough to meet my needs | 61 | %3 | 604 | %33 | 577 | %32 | 267 | %15 | 307 | %17 | 1.816 | %100 |
| YES but it is/was not needed | 20 | %6 | 77 | %24 | 123 | %39 | 47 | %15 | 51 | %16 | 318 | %100 |
| YES but NOT enough to meet my needs | 99 | %8 | 302 | %24 | 463 | %37 | 238 | %19 | 155 | %12 | 1.257 | %100 |
| NO but it is/was NOT needed | 56 | %4 | 340 | %24 | 442 | %32 | 230 | %16 | 332 | %24 | 1.400 | %100 |
| NO but it is/was needed | 401 | %9 | 874 | %21 | 1.568 | %37 | 782 | %18 | 607 | %14 | 4.232 | %100 |
| TOTAL | 637 | %7 | 2.197 | %24 | 3.173 | %35 | 1.564 | %17 | 1.452 | %16 | 9.023 | |

🟥 *Under-represented elements* 🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 275,3 ; dof= 16.*

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RARE DISEASES EUROPE

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: …care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc. / Access to financial products, such as loans, mortgages, insurance…**

| …CARE COORDINATION SUPPORT SUCH AS HELP TO FIND THE NECESSARY INFORMATION ON THE DISEASE AND THE RIGHT PROFESSIONALS, ARRANGING APPOINTMENTS WITH DIFFERENT HEALTH PROVIDERS, ETC. | ACCESS TO FINANCIAL PRODUCTS, SUCH AS LOANS, MORTGAGES, INSURANCE… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES and enough to meet my needs | **260** | **%14** | **68** | **%4** | 449 | %25 | 306 | %17 | **733** | **%40** | 1.816 | %100 |
| YES but it is/was not needed | 49 | %15 | 12 | %4 | 84 | %26 | 56 | %18 | 117 | %37 | 318 | %100 |
| YES but NOT enough to meet my needs | 239 | %19 | 24 | %2 | 349 | %28 | **285** | **%23** | **360** | **%29** | 1.257 | %100 |
| NO but it is/was NOT needed | **207** | **%15** | **17** | **%1** | 337 | %24 | 234 | %17 | **604** | **%43** | 1.399 | %100 |
| NO but it is/was needed | **960** | **%23** | **79** | **%2** | 1.126 | %27 | 767 | %18 | **1.300** | **%31** | 4.232 | %100 |
| **TOTAL** | **1.715** | **%19** | **200** | **%2** | **2.345** | **%26** | **1.648** | **%18** | **3.114** | **%35** | **9.022** | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 199,8 ; dof= 16.*

**Cross: …care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc. / Your social life…**

| …CARE COORDINATION SUPPORT SUCH AS HELP TO FIND THE NECESSARY INFORMATION ON THE DISEASE AND THE RIGHT PROFESSIONALS, ARRANGING APPOINTMENTS WITH DIFFERENT HEALTH PROVIDERS, ETC. | YOUR SOCIAL LIFE… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES and enough to meet my needs | **747** | **%41** | **175** | **%10** | **725** | **%40** | 35 | %2 | **130** | **%7** | 1.812 | %100 |
| YES but it is/was not needed | **135** | **%42** | 21 | %7 | **134** | **%42** | 9 | %3 | 19 | %6 | 318 | %100 |
| YES but NOT enough to meet my needs | **683** | **%54** | 104 | %8 | 397 | %32 | 26 | %2 | **44** | **%4** | 1.254 | %100 |
| NO but it is/was NOT needed | **538** | **%39** | 95 | %7 | **615** | **%44** | 26 | %2 | **123** | **%9** | 1.397 | %100 |
| NO but it is/was needed | **2.468** | **%58** | 313 | %7 | **1.193** | **%28** | 87 | %2 | **162** | **%4** | 4.223 | %100 |
| **TOTAL** | **4.571** | **%51** | **708** | **%8** | **3.064** | **%34** | **183** | **%2** | **478** | **%5** | **9.004** | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 322,2 ; dof= 16.*

## Cross: …financial support including social security benefits / Access to the most adapted care, treatments or surgery…

| …FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES and enough to meet my needs | 83 | %7 | 626 | %52 | 406 | %34 | 44 | %4 | 43 | %4 | 1.202 | %100 |
| YES but it is/was not needed | 15 | %7 | 106 | %52 | 71 | %35 | 4 | %2 | 8 | %4 | 204 | %100 |
| YES but NOT enough to meet my needs | 117 | %11 | 448 | %44 | 373 | %36 | 61 | %6 | 27 | %3 | 1.026 | %100 |
| NO but it is/was NOT needed | 195 | %6 | 1.455 | %46 | 1.167 | %37 | 156 | %5 | 165 | %5 | 3.138 | %100 |
| NO but it is/was needed | 475 | %14 | 1.364 | %40 | 1.283 | %38 | 190 | %6 | 98 | %3 | 3.410 | %100 |
| TOTAL | 885 | %10 | 3.999 | %45 | 3.300 | %37 | 455 | %5 | 341 | %4 | 8.980 | |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 195,3 ; dof= 16.*

## Cross: …financial support including social security benefits / Understanding how the disease will progress…

| …FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES and enough to meet my needs | 75 | %6 | 741 | %62 | 297 | %25 | 56 | %5 | 29 | %2 | 1.198 | %100 |
| YES but it is/was not needed | 14 | %7 | 116 | %57 | 61 | %30 | 8 | %4 | 5 | %2 | 204 | %100 |
| YES but NOT enough to meet my needs | 113 | %11 | 551 | %54 | 285 | %28 | 58 | %6 | 15 | %1 | 1.022 | %100 |
| NO but it is/was NOT needed | 132 | %4 | 1.825 | %58 | 968 | %31 | 140 | %4 | 72 | %2 | 3.137 | %100 |
| NO but it is/was needed | 358 | %11 | 1.741 | %51 | 1.023 | %30 | 228 | %7 | 52 | %2 | 3.402 | %100 |
| TOTAL | 692 | %8 | 4.974 | %55 | 2.634 | %29 | 490 | %5 | 173 | %2 | 8.963 | |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 165,5 ; dof= 16.*

## Cross: …financial support including social security benefits / Financial support including social security benefits…

| …FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES and enough to meet my needs | 56 | %9 | 234 | %36 | 300 | %46 | 29 | %4 | 29 | %4 | 648 | %100 |
| YES but it is/was not needed | 9 | %8 | 15 | %13 | 64 | %53 | 7 | %6 | 25 | %21 | 120 | %100 |
| YES but NOT enough to meet my needs | 127 | %24 | 127 | %24 | 220 | %42 | 22 | %4 | 27 | %5 | 523 | %100 |
| NO but it is/was NOT needed | 137 | %6 | 296 | %12 | 854 | %35 | 281 | %12 | 859 | %35 | 2.427 | %100 |
| NO but it is/was needed | 599 | %26 | 384 | %17 | 907 | %40 | 193 | %9 | 186 | %8 | 2.269 | %100 |
| TOTAL | 928 | %16 | 1.056 | %18 | 2.345 | %39 | 532 | %9 | 1.126 | %19 | 5.987 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 1.246,3 ; dof= 16.*

## Cross: …financial support including social security benefits / Integration at school…

| …FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES and enough to meet my needs | 67 | %6 | 197 | %16 | 288 | %24 | 80 | %7 | 570 | %47 | 1.202 | %100 |
| YES but it is/was not needed | 11 | %5 | 26 | %13 | 58 | %28 | 15 | %7 | 94 | %46 | 204 | %100 |
| YES but NOT enough to meet my needs | 121 | %12 | 144 | %14 | 258 | %25 | 75 | %7 | 428 | %42 | 1.026 | %100 |
| NO but it is/was NOT needed | 154 | %5 | 225 | %7 | 632 | %20 | 182 | %6 | 1.945 | %62 | 3.138 | %100 |
| NO but it is/was needed | 390 | %11 | 361 | %11 | 783 | %23 | 257 | %8 | 1.619 | %47 | 3.410 | %100 |
| TOTAL | 743 | %8 | 953 | %11 | 2.019 | %22 | 609 | %7 | 4.656 | %52 | 8.980 | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 321,6 ; dof= 16.*

## Cross: …financial support including social security benefits / Integration at work…

| …FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS | INTEGRATION AT WORK… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES and enough to meet my needs | 215 | %18 | 140 | %12 | 322 | %27 | 75 | %6 | 446 | %37 | 1.198 | %100 |
| YES but it is/was not needed | 29 | %14 | 24 | %12 | 71 | %35 | 19 | %9 | 61 | %30 | 204 | %100 |
| YES but NOT enough to meet my needs | 308 | %30 | 77 | %8 | 285 | %28 | 76 | %7 | 276 | %27 | 1.022 | %100 |
| NO but it is/was NOT needed | 664 | %21 | 259 | %8 | 991 | %32 | 161 | %5 | 1.062 | %34 | 3.137 | %100 |
| NO but it is/was needed | 1.185 | %35 | 314 | %9 | 903 | %27 | 218 | %6 | 781 | %23 | 3.401 | %100 |
| TOTAL | 2.401 | %27 | 814 | %9 | 2.572 | %29 | 549 | %6 | 2.626 | %29 | 8.962 | |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 313,4 ; dof= 16.*

## Cross: …financial support including social security benefits / Access to social services (e.g. social worker support, household chores support)…

| …FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS | ACCESS TO SOCIAL SERVICES (E.G. SOCIAL WORKER SUPPORT, HOUSEHOLD CHORES SUPPORT)… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES and enough to meet my needs | 90 | %8 | 245 | %20 | 381 | %32 | 111 | %9 | 371 | %31 | 1.198 | %100 |
| YES but it is/was not needed | 11 | %5 | 29 | %14 | 73 | %36 | 24 | %12 | 67 | %33 | 204 | %100 |
| YES but NOT enough to meet my needs | 187 | %18 | 137 | %13 | 402 | %39 | 122 | %12 | 174 | %17 | 1.022 | %100 |
| NO but it is/was NOT needed | 146 | %5 | 236 | %8 | 823 | %26 | 384 | %12 | 1.548 | %49 | 3.137 | %100 |
| NO but it is/was needed | 693 | %20 | 358 | %11 | 1.209 | %36 | 421 | %12 | 719 | %21 | 3.400 | %100 |
| TOTAL | 1.127 | %13 | 1.005 | %11 | 2.888 | %32 | 1.062 | %12 | 2.879 | %32 | 8.961 | |

■ *Under-represented elements*　■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 1.079,0 ; dof= 16.*

## Cross: …financial support including social security benefits / Access to clinical trials…

| …FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS | ACCESS TO CLINICAL TRIALS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES and enough to meet my needs | 45 | %4 | 374 | %31 | 379 | %32 | 202 | %17 | 202 | %17 | 1.202 | %100 |
| YES but it is/was not needed | 8 | %4 | 61 | %30 | 74 | %36 | 37 | %18 | 24 | %12 | 204 | %100 |
| YES but NOT enough to meet my needs | 105 | %10 | 235 | %23 | 380 | %37 | 159 | %15 | 147 | %14 | 1.026 | %100 |
| NO but it is/was NOT needed | 109 | %3 | 826 | %26 | 1.054 | %34 | 546 | %17 | 603 | %19 | 3.138 | %100 |
| NO but it is/was needed | 367 | %11 | 693 | %20 | 1.265 | %37 | 614 | %18 | 470 | %14 | 3.409 | %100 |
| TOTAL | 634 | %7 | 2.189 | %24 | 3.152 | %35 | 1.558 | %17 | 1.446 | %16 | 8.979 | |

🟪 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 263,0 ; dof= 16.*

## Cross: …financial support including social security benefits / Access to financial products, such as loans, mortgages, insurance…

| …FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS | ACCESS TO FINANCIAL PRODUCTS, SUCH AS LOANS, MORTGAGES, INSURANCE… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES and enough to meet my needs | 147 | %12 | 60 | %5 | 314 | %26 | 215 | %18 | 466 | %39 | 1.202 | %100 |
| YES but it is/was not needed | 25 | %12 | 6 | %3 | 58 | %28 | 40 | %20 | 75 | %37 | 204 | %100 |
| YES but NOT enough to meet my needs | 275 | %27 | 26 | %3 | 278 | %27 | 193 | %19 | 254 | %25 | 1.026 | %100 |
| NO but it is/was NOT needed | 384 | %12 | 41 | %1 | 722 | %23 | 545 | %17 | 1.446 | %46 | 3.138 | %100 |
| NO but it is/was needed | 874 | %26 | 66 | %2 | 956 | %28 | 645 | %19 | 867 | %25 | 3.408 | %100 |
| TOTAL | 1.705 | %19 | 199 | %2 | 2.328 | %26 | 1.638 | %18 | 3.108 | %35 | 8.978 | |

🟪 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 534,3 ; dof= 16.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

## Cross: …financial support including social security benefits / Your social life…

| …FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS | YOUR SOCIAL LIFE… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES and enough to meet my needs | 516 | %43 | 117 | %10 | 463 | %39 | 21 | %2 | 81 | %7 | 1.198 | %100 |
| YES but it is/was not needed | 75 | %37 | 15 | %7 | 98 | %48 | 5 | %2 | 11 | %5 | 204 | %100 |
| YES but NOT enough to meet my needs | 610 | %60 | 81 | %8 | 282 | %28 | 25 | %2 | 24 | %2 | 1.022 | %100 |
| NO but it is/was NOT needed | 1.289 | %41 | 248 | %8 | 1.303 | %42 | 62 | %2 | 235 | %7 | 3.137 | %100 |
| NO but it is/was needed | 2.058 | %61 | 242 | %7 | 908 | %27 | 68 | %2 | 124 | %4 | 3.400 | %100 |
| **TOTAL** | 4.548 | %51 | 703 | %8 | 3.054 | %34 | 181 | %2 | 475 | %5 | 8.961 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 373,8 ; dof= 16.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

**Cross: Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease? / Access to the most adapted care, treatments or surgery…**

| ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | ACCESS TO THE MOST ADAPTED CARE, TREATMENTS OR SURGERY… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, through a patient organisation | 435 | %9 | 2.407 | %51 | 1.596 | %34 | 160 | %3 | 148 | %3 | 4.746 | %100 |
| YES, through online communities | 439 | %10 | 1.937 | %44 | 1.690 | %38 | 221 | %5 | 139 | %3 | 4.426 | %100 |
| YES, through local networks (e.g. schools) | 34 | %9 | 201 | %52 | 124 | %32 | 10 | %3 | 15 | %4 | 384 | %100 |
| NO, because of accessibility issues (e.g. language or technical barriers) | 16 | %11 | 38 | %27 | 67 | %48 | 15 | %11 | 5 | %4 | 141 | %100 |
| NO, because I have not been able to find other people with the same disease | 115 | %12 | 332 | %34 | 404 | %41 | 80 | %8 | 45 | %5 | 976 | %100 |
| NO, because I don't want to | 41 | %9 | 172 | %38 | 163 | %36 | 37 | %8 | 35 | %8 | 448 | %100 |
| Other, specify… | 40 | %9 | 194 | %44 | 146 | %33 | 30 | %7 | 30 | %7 | 440 | %100 |
| **TOTAL** | **889** | **%10** | **4.020** | **%45** | **3.316** | **%37** | **457** | **%5** | **342** | **%4** | **9.024** | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 233,2 ; dof= 24.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease? / Understanding how the disease will progress…**

| ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | UNDERSTANDING HOW THE DISEASE WILL PROGRESS… | | | | | | | | | | TOTAL | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, through a patient organisation | **273** | **%6** | **3.033** | **%64** | **1.206** | **%25** | **147** | **%3** | 80 | %2 | 4.739 | %100 |
| YES, through online communities | 333 | %8 | **2.508** | **%57** | 1.273 | %29 | 227 | %5 | **71** | **%2** | 4.412 | %100 |
| YES, through local networks (e.g. schools) | 30 | %8 | **235** | **%61** | 102 | %27 | **12** | **%3** | 5 | %1 | 384 | %100 |
| NO, because of accessibility issues (e.g. language or technical barriers) | **24** | **%17** | **45** | **%32** | 49 | %35 | **21** | **%15** | 2 | %1 | 141 | %100 |
| NO, because I have not been able to find other people with the same disease | **127** | **%13** | **341** | **%35** | **369** | **%38** | **116** | **%12** | 22 | %2 | 975 | %100 |
| NO, because I don't want to | 28 | %6 | **206** | **%46** | **166** | **%37** | **36** | **%8** | 12 | %3 | 448 | %100 |
| Other, specify... | 39 | %9 | 238 | %54 | 124 | %28 | 19 | %4 | **19** | **%4** | 439 | %100 |
| **TOTAL** | **694** | **%8** | **4.999** | **%56** | **2.644** | **%29** | **494** | **%5** | **175** | **%2** | **9.006** | |

■ *Under-represented elements*   ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 474,5 ; dof= 24.*

**Cross: Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease? / Financial support including social security benefits…**

| ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, through a patient organisation | _442_ | _%14_ | _618_ | _%20_ | 1.262 | %40 | _249_ | _%8_ | 571 | %18 | **3.142** | %100 |
| YES, through online communities | _514_ | _%17_ | 545 | %18 | 1.184 | %39 | 264 | %9 | _521_ | _%17_ | **3.028** | %100 |
| YES, through local networks (e.g. schools) | 34 | %16 | 42 | %19 | 84 | %38 | 22 | %10 | 37 | %17 | **219** | %100 |
| NO, because of accessibility issues (e.g. language or technical barriers) | _19_ | _%27_ | 8 | %11 | 27 | %39 | 8 | %11 | 8 | %11 | **70** | %100 |
| NO, because I have not been able to find other people with the same disease | 111 | %18 | _74_ | _%12_ | 261 | %41 | 64 | %10 | 121 | %19 | **631** | %100 |
| NO, because I don't want to | 46 | %14 | 56 | %18 | 109 | %34 | 26 | %8 | _83_ | _%26_ | **320** | %100 |
| Other, specify... | 42 | %14 | 57 | %19 | 104 | %34 | 30 | %10 | _70_ | _%23_ | **303** | %100 |
| **TOTAL** | **928** | **%16** | **1.056** | **%18** | **2.345** | **%39** | **532** | **%9** | **1.126** | **%19** | **5.987** | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 69,6 ; dof= 24.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease? / Integration at school…**

| ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | INTEGRATION AT SCHOOL… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, through a patient organisation | 373 | %8 | 594 | %13 | 1.084 | %23 | 303 | %6 | 2.392 | %50 | 4.746 | %100 |
| YES, through online communities | 368 | %8 | 437 | %10 | 955 | %22 | 305 | %7 | 2.361 | %53 | 4.426 | %100 |
| YES, through local networks (e.g. schools) | 38 | %10 | 82 | %21 | 90 | %23 | 18 | %5 | 156 | %41 | 384 | %100 |
| NO, because of accessibility issues (e.g. language or technical barriers) | 23 | %16 | 16 | %11 | 39 | %28 | 7 | %5 | 56 | %40 | 141 | %100 |
| NO, because I have not been able to find other people with the same disease | 92 | %9 | 70 | %7 | 242 | %25 | 63 | %6 | 509 | %52 | 976 | %100 |
| NO, because I don't want to | 30 | %7 | 36 | %8 | 93 | %21 | 36 | %8 | 253 | %56 | 448 | %100 |
| Other, specify... | 36 | %8 | 42 | %10 | 82 | %19 | 27 | %6 | 253 | %58 | 440 | %100 |
| **TOTAL** | **746** | **%8** | **960** | **%11** | **2.030** | **%22** | **613** | **%7** | **4.675** | **%52** | **9.024** | |

🟥 *Under-represented elements*   🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 127,4 ; dof= 24.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease? / Integration at work…**

| ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | INTEGRATION AT WORK… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, through a patient organisation | 1.245 | %26 | 489 | %10 | 1.405 | %30 | 253 | %5 | 1.347 | %28 | 4.739 | %100 |
| YES, through online communities | 1.261 | %29 | 406 | %9 | 1.205 | %27 | 267 | %6 | 1.273 | %29 | 4.412 | %100 |
| YES, through local networks (e.g. schools) | 105 | %27 | 39 | %10 | 120 | %31 | 21 | %5 | 99 | %26 | 384 | %100 |
| NO, because of accessibility issues (e.g. language or technical barriers) | 45 | %32 | 9 | %6 | 42 | %30 | 5 | %4 | 40 | %28 | 141 | %100 |
| NO, because I have not been able to find other people with the same disease | 259 | %27 | 56 | %6 | 286 | %29 | 66 | %7 | 307 | %32 | 974 | %100 |
| NO, because I don't want to | 90 | %20 | 44 | %10 | 132 | %29 | 36 | %8 | 146 | %33 | 448 | %100 |
| Other, specify... | 99 | %23 | 32 | %7 | 114 | %26 | 29 | %7 | 165 | %38 | 439 | %100 |
| **TOTAL** | **2.411** | **%27** | **818** | **%9** | **2.587** | **%29** | **551** | **%6** | **2.638** | **%29** | **9.005** | |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 74,7 ; dof= 24.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease? / Access to social services (e.g. social worker support, household chores support)…**

| ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | ACCESS TO SOCIAL SERVICES (E.G. SOCIAL WORKER SUPPORT, HOUSEHOLD CHORES SUPPORT)… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, through a patient organisation | **556** | **%12** | **622** | **%13** | 1.559 | %33 | **515** | **%11** | 1.486 | %31 | **4.738** | **%100** |
| YES, through online communities | 566 | %13 | 521 | %12 | 1.443 | %33 | 534 | %12 | **1.348** | **%31** | **4.412** | **%100** |
| YES, through local networks (e.g. schools) | 53 | %14 | **68** | **%18** | 133 | %35 | 43 | %11 | **87** | **%23** | **384** | **%100** |
| NO, because of accessibility issues (e.g. language or technical barriers) | **28** | **%20** | **8** | **%6** | **58** | **%41** | 14 | %10 | **33** | **%23** | **141** | **%100** |
| NO, because I have not been able to find other people with the same disease | **145** | **%15** | **79** | **%8** | 306 | %31 | 123 | %13 | 321 | %33 | **974** | **%100** |
| NO, because I don't want to | 47 | %10 | 41 | %9 | **116** | **%26** | 59 | %13 | **185** | **%41** | **448** | **%100** |
| Other, specify… | 59 | %13 | 58 | %13 | **113** | **%26** | 47 | %11 | **162** | **%37** | **439** | **%100** |
| **TOTAL** | **1.134** | **%13** | **1.011** | **%11** | **2.906** | **%32** | **1.066** | **%12** | **2.887** | **%32** | **9.004** | |

■ *Under-represented elements* ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 117,7 ; dof= 24.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease? / Access to clinical trials…**

| ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | ACCESS TO CLINICAL TRIALS… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, through a patient organisation | 287 | %6 | 1.458 | %31 | 1.621 | %34 | 689 | %15 | 690 | %15 | 4.745 | %100 |
| YES, through online communities | 342 | %8 | 1.057 | %24 | 1.598 | %36 | 800 | %18 | 629 | %14 | 4.426 | %100 |
| YES, through local networks (e.g. schools) | 19 | %5 | 119 | %31 | 133 | %35 | 59 | %15 | 54 | %14 | 384 | %100 |
| NO, because of accessibility issues (e.g. language or technical barriers) | 23 | %16 | 19 | %13 | 48 | %34 | 32 | %23 | 19 | %13 | 141 | %100 |
| NO, because I have not been able to find other people with the same disease | 89 | %9 | 151 | %15 | 353 | %36 | 200 | %20 | 183 | %19 | 976 | %100 |
| NO, because I don't want to | 24 | %5 | 78 | %17 | 144 | %32 | 105 | %23 | 97 | %22 | 448 | %100 |
| Other, specify... | 31 | %7 | 107 | %24 | 133 | %30 | 68 | %15 | 101 | %23 | 440 | %100 |
| **TOTAL** | 637 | %7 | 2.197 | %24 | 3.173 | %35 | 1.564 | %17 | 1.452 | %16 | 9.023 | |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 264,4 ; dof= 24.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease? / Access to financial products, such as loans, mortgages, insurance…**

| ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | ACCESS TO FINANCIAL PRODUCTS, SUCH AS LOANS, MORTGAGES, INSURANCE… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, through a patient organisation | 892 | %19 | 119 | %3 | 1.276 | %27 | 825 | %17 | 1.633 | %34 | 4.745 | %100 |
| YES, through online communities | 915 | %21 | 86 | %2 | 1.123 | %25 | 831 | %19 | 1.471 | %33 | 4.426 | %100 |
| YES, through local networks (e.g. schools) | 74 | %19 | 12 | %3 | 107 | %28 | 63 | %16 | 128 | %33 | 384 | %100 |
| NO, because of accessibility issues (e.g. language or technical barriers) | 37 | %26 | 3 | %2 | 40 | %28 | 22 | %16 | 39 | %28 | 141 | %100 |
| NO, because I have not been able to find other people with the same disease | 175 | %18 | 21 | %2 | 246 | %25 | 183 | %19 | 350 | %36 | 975 | %100 |
| NO, because I don't want to | 71 | %16 | 9 | %2 | 98 | %22 | 88 | %20 | 182 | %41 | 448 | %100 |
| Other, specify... | 94 | %21 | 10 | %2 | 93 | %21 | 78 | %18 | 165 | %38 | 440 | %100 |
| **TOTAL** | 1.715 | %19 | 200 | %2 | 2.345 | %26 | 1.648 | %18 | 3.114 | %35 | 9.022 | |

🟥 *Under-represented elements*  🟦 *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 44,9 ; dof= 24.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: Are you, or the person you care for, in touch with other people living with the same rare disease or with an undiagnosed rare disease? / Your social life…**

| ARE YOU, OR THE PERSON YOU CARE FOR, IN TOUCH WITH OTHER PEOPLE LIVING WITH THE SAME RARE DISEASE OR WITH AN UNDIAGNOSED RARE DISEASE? | YOUR SOCIAL LIFE… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES, through a patient organisation | 2.290 | %48 | 452 | %10 | 1.694 | %36 | 79 | %2 | 223 | %5 | 4.738 | %100 |
| YES, through online communities | 2.375 | %54 | 342 | %8 | 1.407 | %32 | 83 | %2 | 205 | %5 | 4.412 | %100 |
| YES, through local networks (e.g. schools) | 189 | %49 | 39 | %10 | 138 | %36 | 2 | %1 | 16 | %4 | 384 | %100 |
| NO, because of accessibility issues (e.g. language or technical barriers) | 75 | %53 | 9 | %6 | 50 | %35 | 0 | %0 | 7 | %5 | 141 | %100 |
| NO, because I have not been able to find other people with the same disease | 486 | %50 | 55 | %6 | 338 | %35 | 31 | %3 | 64 | %7 | 974 | %100 |
| NO, because I don't want to | 206 | %46 | 24 | %5 | 170 | %38 | 12 | %3 | 36 | %8 | 448 | %100 |
| Other, specify... | 199 | %45 | 33 | %8 | 156 | %36 | 11 | %3 | 40 | %9 | 439 | %100 |
| TOTAL | 4.571 | %51 | 708 | %8 | 3.064 | %34 | 183 | %2 | 478 | %5 | 9.004 | |

■ *Under-represented elements*  ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 109,0 ; dof= 24.*

*Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"*

*Sample size 10486 responses*

**Cross: …care coordination support such as help to find the necessary information on the disease and the right professionals, arranging appointments with different health providers, etc. / Access to the most adapted care, treatments or surgery…**

| …CARE COORDINATION SUPPORT SUCH AS HELP TO FIND THE NECESSARY INFORMATION ON THE DISEASE AND THE RIGHT PROFESSIONALS, ARRANGING APPOINTMENTS WITH DIFFERENT HEALTH PROVIDERS, ETC. | ACCESS TO THE MOST ADAPTED CARE, TREATMENTS OR SURGERY… | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | …HAS GOTTEN WORSE | | …HAS IMPROVED | | …HAS REMAINED THE SAME | | DON'T KNOW | | NOT RELEVANT | | TOTAL | |
| | N | % | N | % | N | % | N | % | N | % | N | % |
| YES and enough to meet my needs | 101 | %6 | 960 | %53 | 622 | %34 | 73 | %4 | 61 | %3 | 1.817 | %100 |
| YES but it is/was not needed | 26 | %8 | 147 | %46 | 121 | %38 | 12 | %4 | 12 | %4 | 318 | %100 |
| YES but NOT enough to meet my needs | 135 | %11 | 558 | %44 | 451 | %36 | 67 | %5 | 46 | %4 | 1.257 | %100 |
| NO but it is/was NOT needed | 98 | %7 | 596 | %43 | 541 | %39 | 84 | %6 | 81 | %6 | 1.400 | %100 |
| NO but it is/was needed | 529 | %13 | 1.759 | %42 | 1.581 | %37 | 221 | %5 | 142 | %3 | 4.232 | %100 |
| **TOTAL** | **889** | **%10** | **4.020** | **%45** | **3.316** | **%37** | **457** | **%5** | **342** | **%4** | **9.024** | |

■ *Under-represented elements*    ■ *Over-represented elements*

*The relationship is very significant. p-value= < 0,01 ; Chi2= 146,4 ; dof= 16.*

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

# Chapter 16.

## Questions for undiagnosed respondents

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Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

**Question asked only to respondents who are undiagnosed (partial diagnosis or unsolved cases)**

## Please select all the statements that describe your situation. You or the person you care for:

Are still actively searching for a specific diagnosis for the rare disease — %43

Changed your priorities from getting a diagnosis to managing and learning how to live with the disease and its symptoms without a diagnosis — %36

Think that existing medical knowledge does not enable to reach a more precise diagnosis — %36

Cannot find a doctor for the care or the diagnosis of the rare disease — %29

Have several identified symptom(s) (e.g. epilepsy, developmental delay, autism…) but no specific diagnosis — %27

Are included in a research programme aiming to provide a diagnosis for undiagnosed patients — %12

Other, specify... — %10

Do not want to know the diagnosis of the rare disease because it could imply dramatic changes — %2

## Please select all the statements that describe your situation. You or the person you care for:

| | N |
|---|---|
| Are still actively searching for a specific diagnosis for the rare disease | 278 |
| Changed your priorities from getting a diagnosis to managing and learning how to live with the disease and its symptoms without a diagnosis | 233 |
| Think that existing medical knowledge does not enable to reach a more precise diagnosis | 229 |
| Cannot find a doctor for the care or the diagnosis of the rare disease | 190 |
| Have several identified symptom(s) (e.g. epilepsy, developmental delay, autism…) but no specific diagnosis | 175 |
| Are included in a research programme aiming to provide a diagnosis for undiagnosed patients | 78 |
| Other, specify... | 65 |
| Do not want to know the diagnosis of the rare disease because it could imply dramatic changes | 15 |
| **TOTAL** | **645** |

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# 10. Consequences of being undiagnosed

Question asked only to respondents who are undiagnosed (partial diagnosis or unsolved cases)

## Does the lack of a precise diagnosis for the rare disease prevent you from...

| Category | YES, completely | YES, partially | NO | Don't know | Not relevant |
|---|---|---|---|---|---|
| ...understanding how the disease will progress | %53 | %24 | %10 | %7 | %7 |
| ...accessing clinical trials | %31 | %22 | %20 | %18 | %9 |
| ...integration at work | %25 | %20 | %25 | %8 | %22 |
| ...accessing social services (e.g. social worker support, household chores support, …) | %23 | %23 | %28 | %11 | %14 |
| ...integration at school | %11 | %14 | %34 | %6 | %35 |
| ...accessing the most adapted care, treatment or surgery | %32 | %30 | %16 | %13 | %9 |
| ...having a fulfilling social life | %32 | %33 | %22 | %5 | %8 |
| ...receiving financial support including social security benefits | %31 | %22 | %27 | %11 | %9 |
| ...making informed reproductive choices such as planning whether or not to have children, or deciding whether or not to conduct prenatal tests | %17 | %11 | %30 | %8 | %34 |

● YES, completely   ● YES, partially   ● NO   ● Don't know   ● Not relevant

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RARE DISEASES EUROPE

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

**Question asked only to respondents who are undiagnosed (partial diagnosis or unsolved cases)**

## Does the lack of a precise diagnosis for the rare disease prevent you from...

| | YES, COMPLETELY | YES, PARTIALLY | NO | DON'T KNOW | NOT RELEVANT | TOTAL |
|---|---|---|---|---|---|---|
| ...accessing clinical trials | 218 | 154 | 143 | 127 | 62 | 704 |
| ...understanding how the disease will progress | 369 | 164 | 67 | 51 | 46 | 697 |
| ...integration at school | 79 | 101 | 236 | 44 | 244 | 704 |
| ...integration at work | 173 | 140 | 177 | 53 | 154 | 697 |
| ...accessing social services (e.g. social worker support, household chores support, …) | 162 | 163 | 195 | 76 | 101 | 697 |

| | YES, PARTIALLY | YES, COMPLETELY | NO | DON'T KNOW | NOT RELEVANT | TOTAL |
|---|---|---|---|---|---|---|
| ...accessing the most adapted care, treatment or surgery | 212 | 223 | 115 | 95 | 60 | 705 |
| ...making informed reproductive choices such as planning whether or not to have children, or deciding whether or not to conduct prenatal tests | 78 | 118 | 206 | 58 | 237 | 697 |
| ...receiving financial support including social security benefits | 155 | 214 | 185 | 79 | 60 | 693 |
| ...having a fulfilling social life | 232 | 220 | 155 | 36 | 54 | 697 |

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"

Sample size 10486 responses

# THANK YOU!

Thank you to all the people living with rare diseases who participated in the survey, and to the Rare Barometer partners and corporate donors in 2021.

A special thank you to our National Alliances and European Federations who helped us spread the word about the survey and contributed to the great number of respondents.

Together we can make the voice of the rare disease community stronger.