


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

 **13307** respondents worldwide and
10486 in Europe

 **27** languages

 **107** countries

 **1900+** diseases represented

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

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Chapter 14: In touch with other people living with rare diseases

Chapter 15: Consequences of diagnosis

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Chapter 1.

Sample description

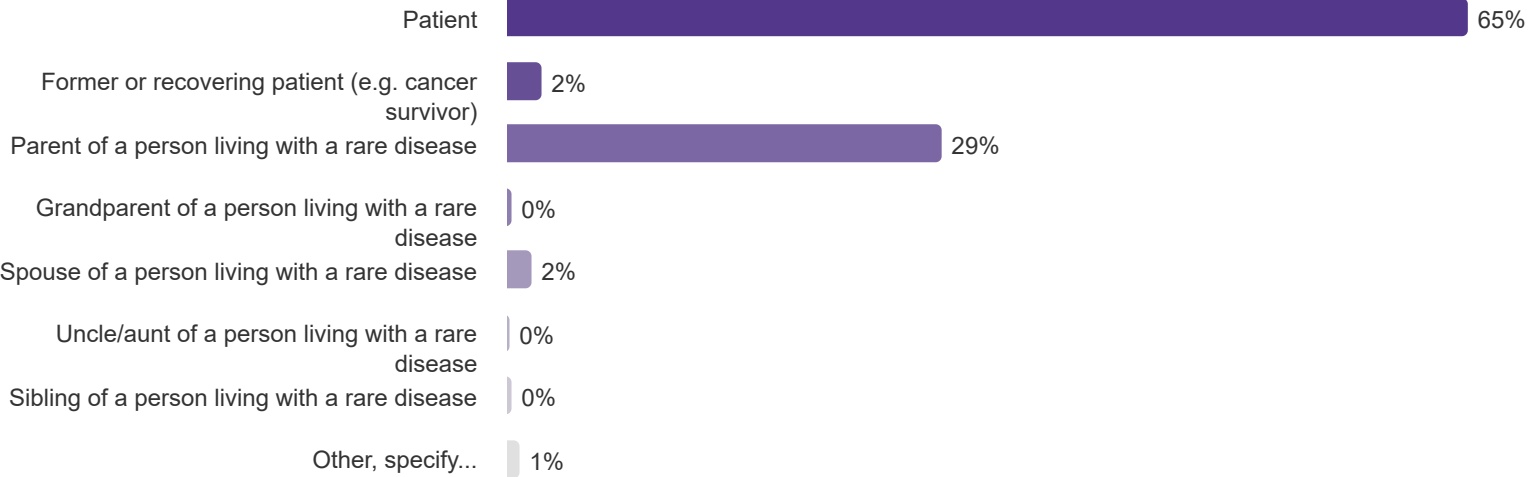
Number of respondents

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...



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).

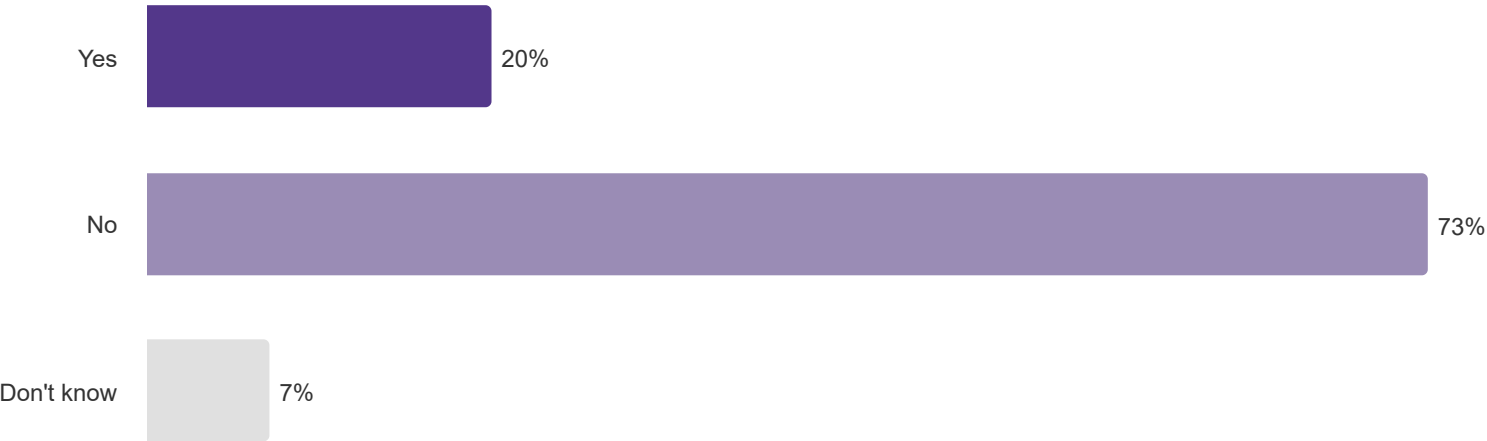
Are you a...



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?



Age of the respondent when the questionnaire was filled in

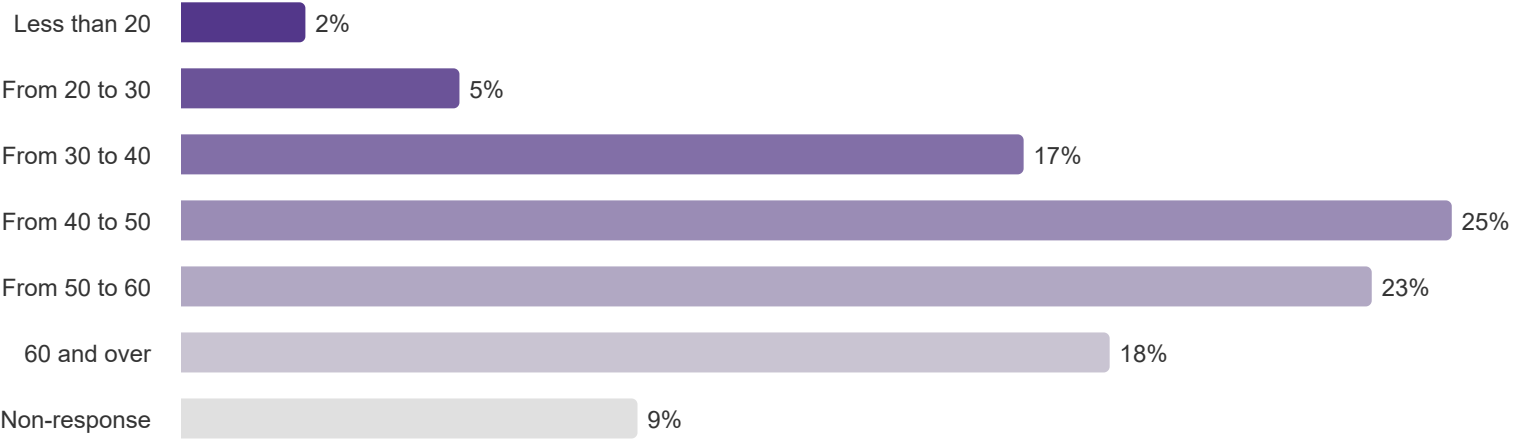
Calculated based on:

- 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



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

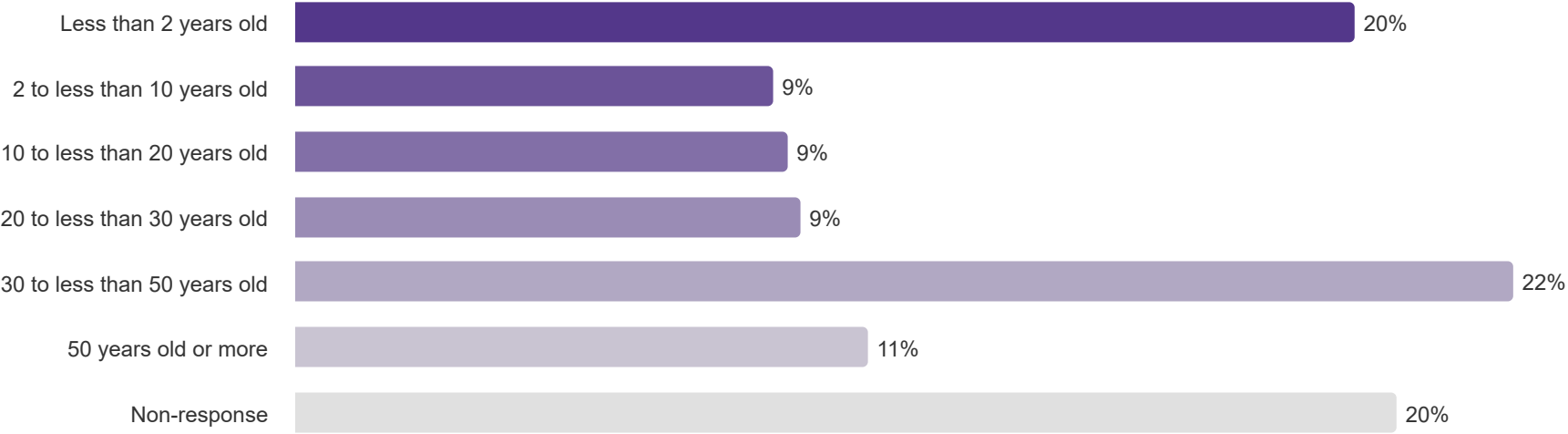
Calculated based on:

- 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

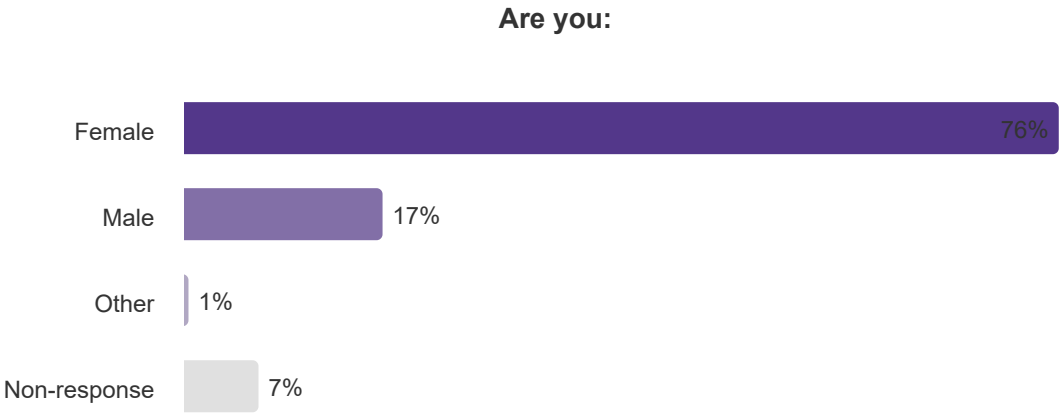
	N
Less than 2 years old	2,045
2 to less than 10 years old	925
10 to less than 20 years old	952
20 to less than 30 years old	978
30 to less than 50 years old	2,353
50 years old or more	1,107
Non-response	2,126
TOTAL	10,486

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



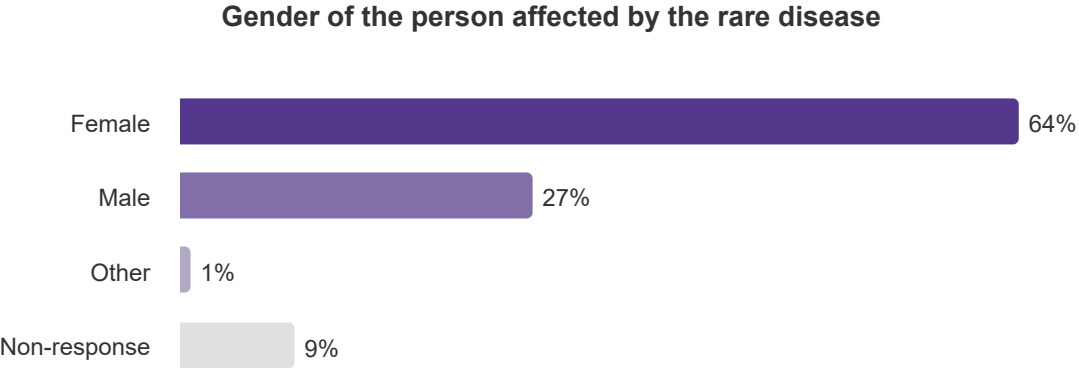
Gender of the respondent

Are you:	
	N
Female	7,930
Male	1,807
Other	56
Non-response	693
TOTAL	10,486



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

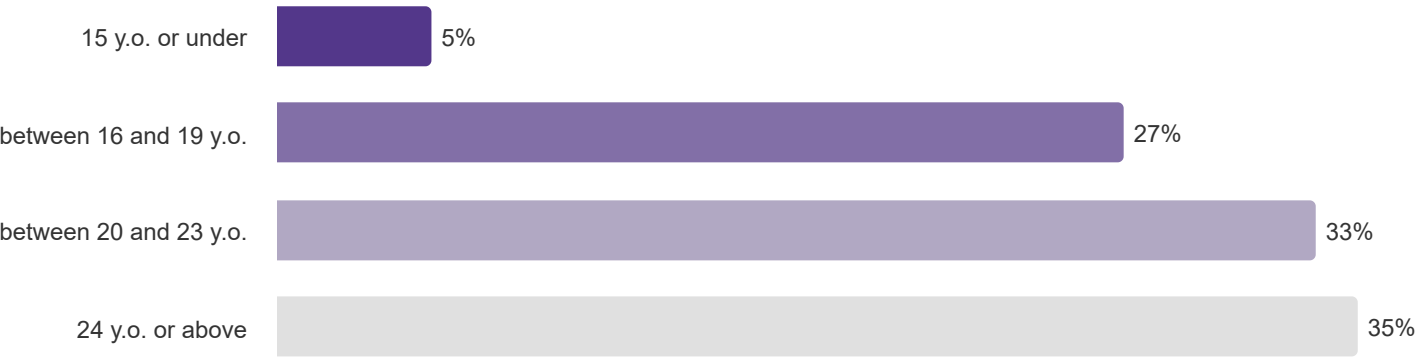


Education of the respondent

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

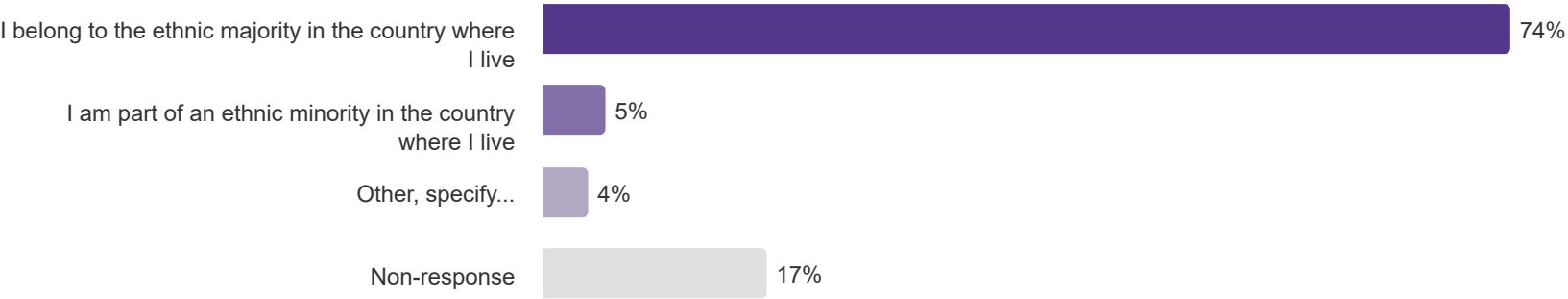
How old were you when you stopped full-time education?



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

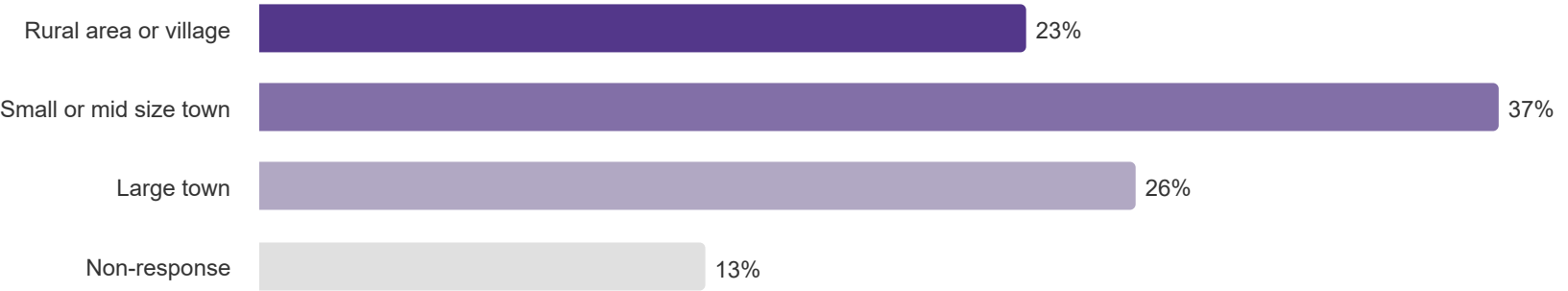
How would you best describe yourself?



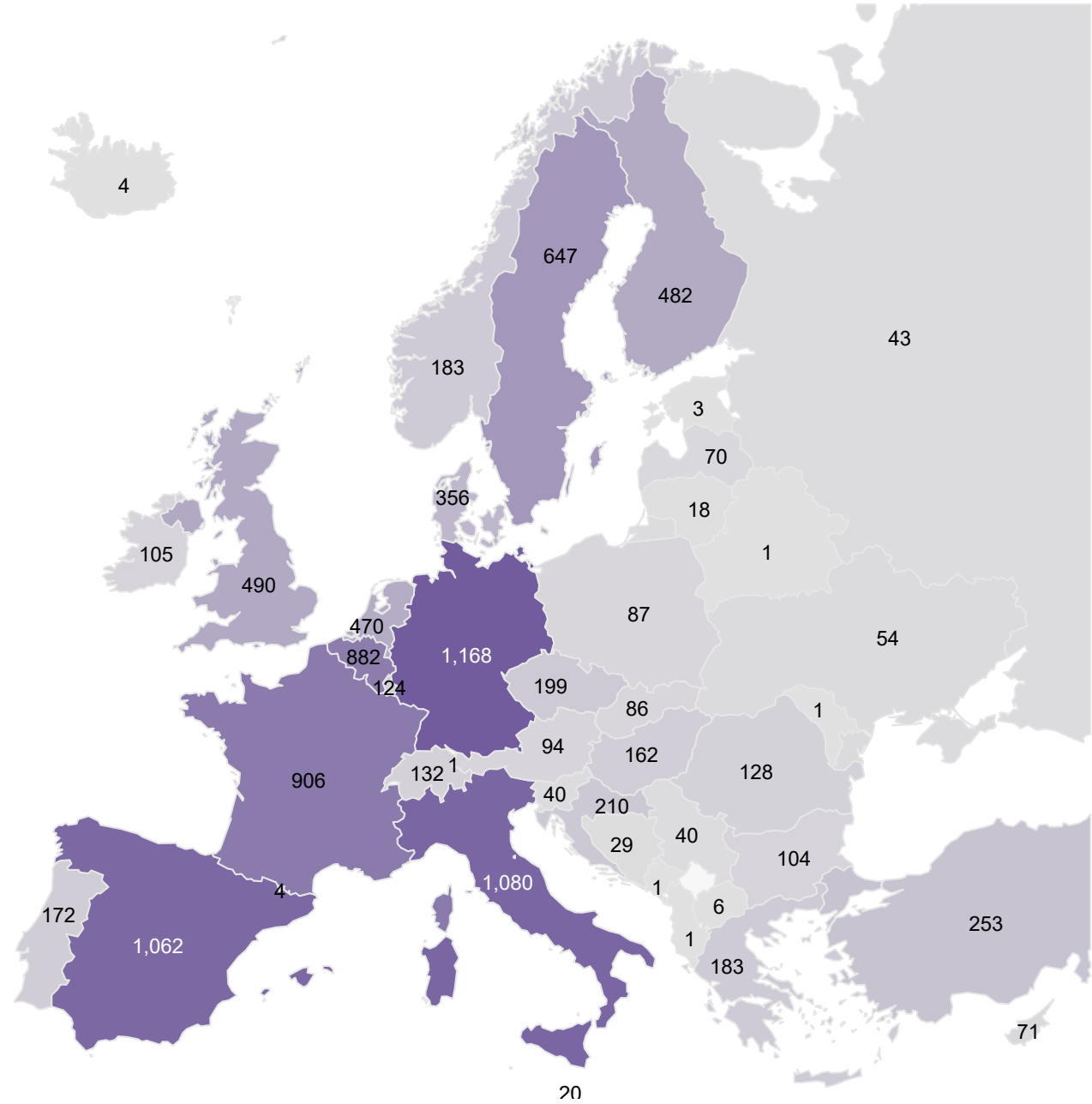
Would you say that you, or the person you care for, live in a:

	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

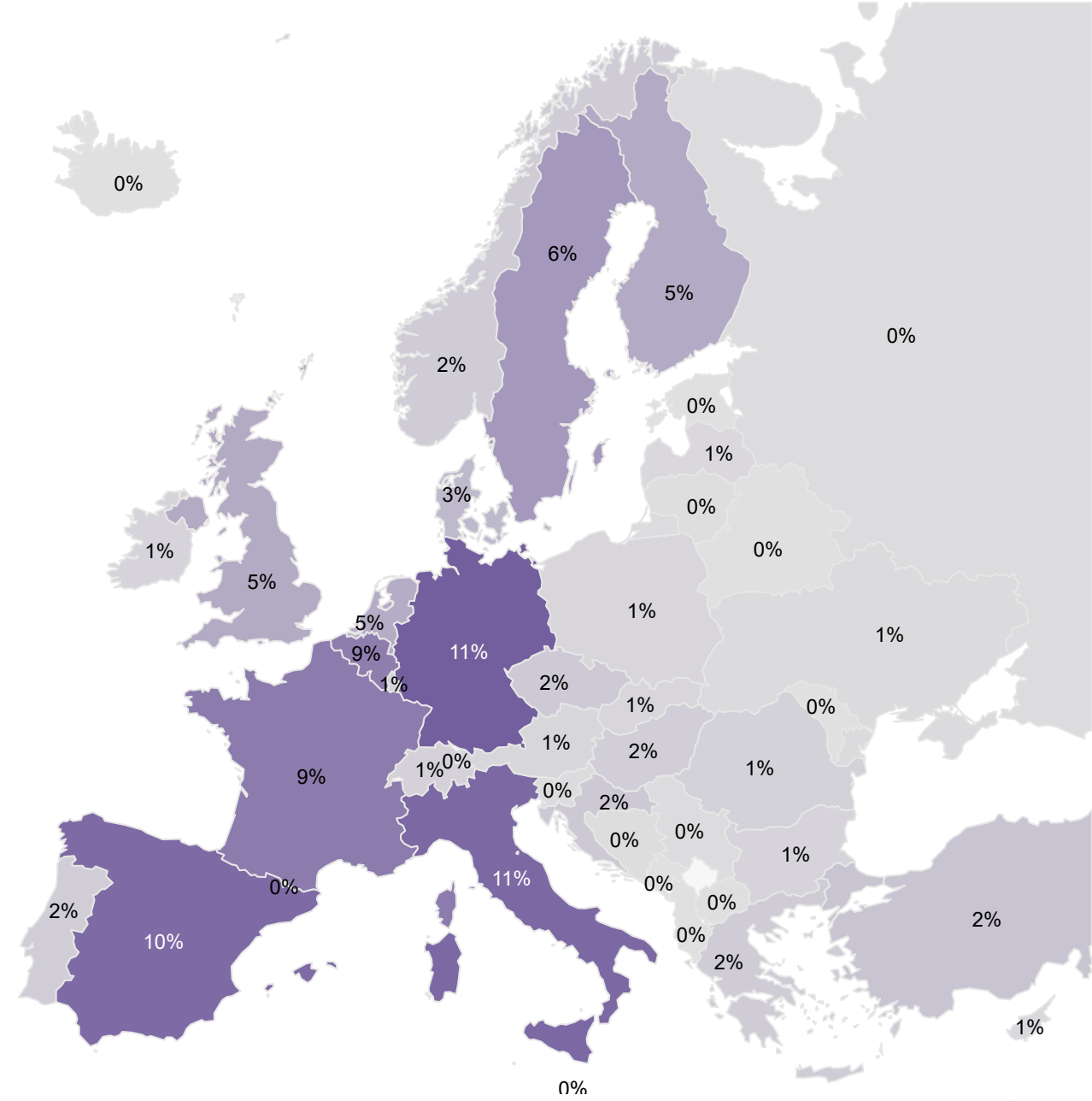
Would you say that you, or the person you care for, live in a:



In which country do you live?



In which country do you live?



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

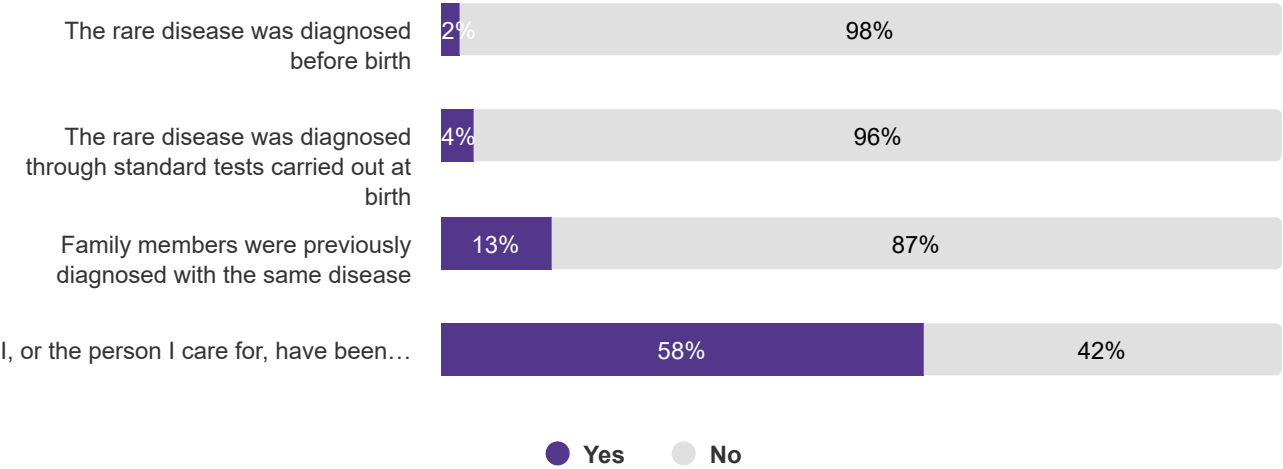
Please select the sentence that best describes your situation or the situation of the
person you care for:



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?



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=@ion=2&country=&province=&pp=200>

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

64

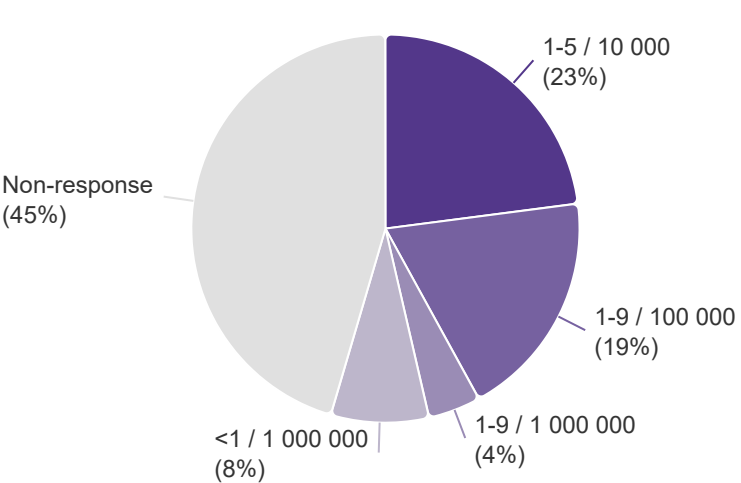
▼ Sample information : *NBS_recod among "Yes"*

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

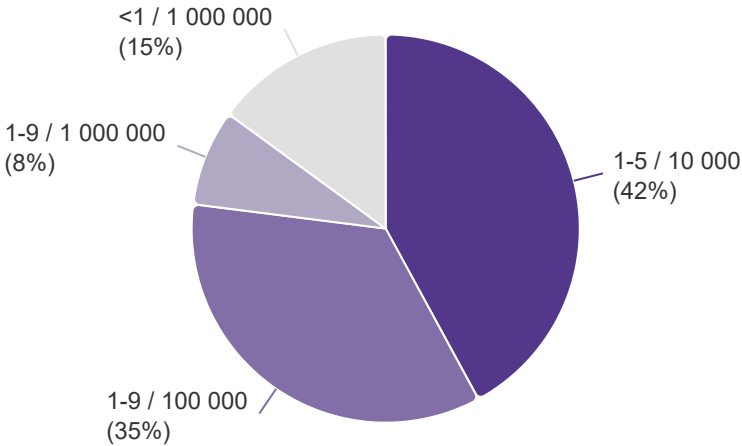
Point prevalence

	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%

Disease prevalence



Disease prevalence



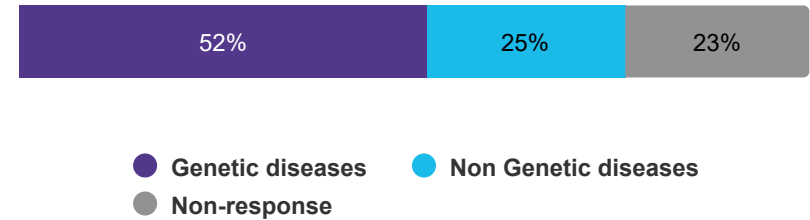
Variables calculated based on the name of respondents' disease and Orphanet data

orphanet.org

Genetic diseases

	N
Genetic diseases	5,447
Non Genetic diseases	2,627
Non-response	2,412
TOTAL	10,486

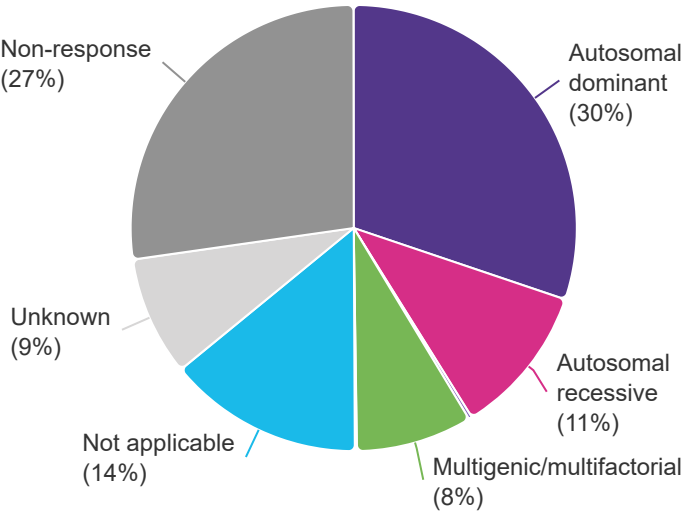
Genetic diseases



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



Number of diseases
(number of different orphacodes)

1,679

Diseases represented		
	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%

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

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

orphadata.org

Classification of rare diseases by Orphanet (one disease can appear in several groups)

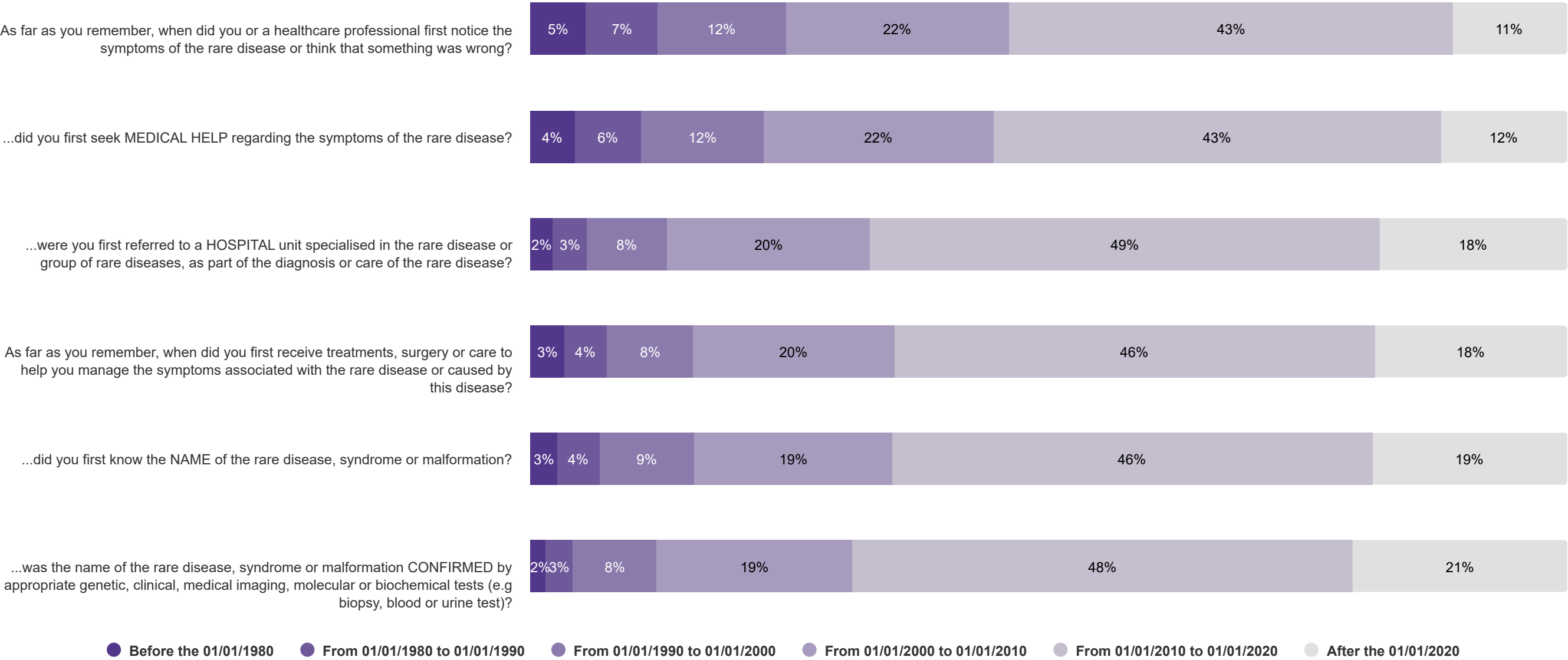
	N	%
Abdominal surgical diseases	239	3%
Allergic diseases	3	0%
Bone diseases	799	9%
Cardiac diseases	660	8%
Cardiac malformations	295	3%
Circulatory system diseases	1,351	16%
Developmental anomalies during embryogenesis	3,347	40%
Diseases due to toxic effects	3	0%
Endocrine diseases	995	12%
Gastroenterological 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%



Chapter 2.

Diagnosis journey

Dates of the different steps of the diagnosis journey





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.

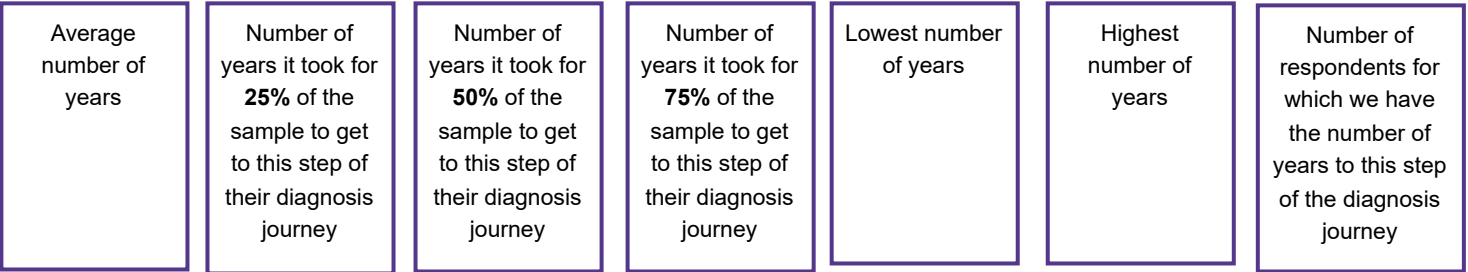
First medical contact = when they first sought medical help regarding the symptoms of the rare disease

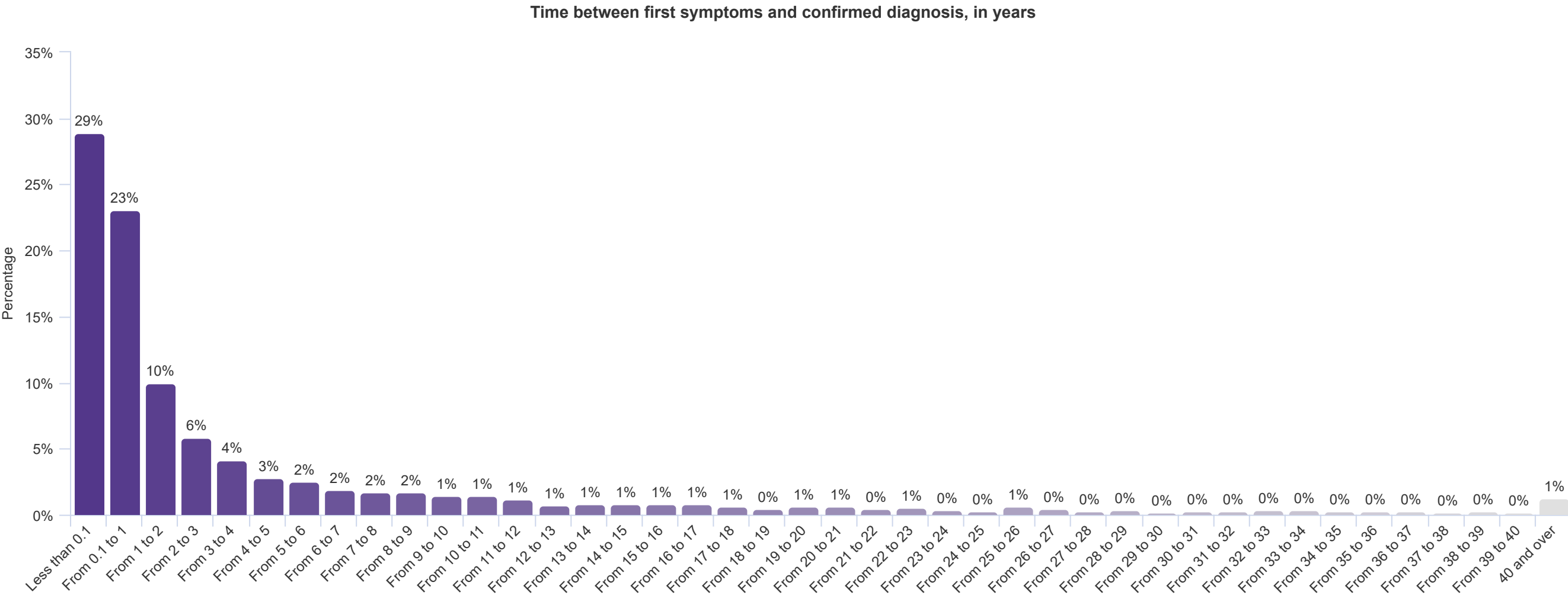
First symptomatic treatment = when they first receive treatments, surgery or care to help them manage the symptoms associated with the rare disease or caused by this disease.

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).





Time from first symptoms to the different steps of the diagnosis journey depending on the gender of the person affected by the rare disease, in years.

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

Time from first symptoms to the different steps of the diagnosis journey depending on the age of respondents when they stopped full-time education

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

Time from first symptoms to the different steps of the diagnosis journey depending if respondents are part of the ethnic majority in their country

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

Time from first symptoms to the different steps of the diagnosis journey depending on the type of town respondents live in

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

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.

Time from first symptoms to the different steps of the diagnosis journey depending if the person affected has been referred to a centre of expertise

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

Time from first symptoms to the different steps of the diagnosis journey depending if family members were previously diagnosed with the same disease

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

Time from first symptoms to the different steps of the diagnosis journey depending on the point prevalence of the disease

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](#)

Time from first symptoms to the different steps of the diagnosis journey depending if the rare disease is genetic or not (source: [orpha.data](#))

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	<u>4.1</u>	3,700	<u>4.6</u>	2,368	<u>4.2</u>	4,276	<u>5.9</u>	3,632
Non Genetic diseases	<u>0.1</u>	2,154	<u>2.4</u>	2,161	<u>2.3</u>	1,142	<u>2.2</u>	2,247	<u>2.6</u>	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



Chapter 3.

Age of the person
affected when first
symptoms were noticed

Time from first symptoms to the different steps of the diagnosis journey
depending on the age of the person affected by the rare disease when first symptoms were noticed

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	<u>1.1</u>	1,699	3.2	1,576	3.4	989	<u>4.4</u>	1,762	5.0	1,529
2 to less than 10 years old	<u>1.8</u>	758	<u>6.5</u>	705	<u>7.7</u>	417	<u>7.0</u>	789	<u>8.8</u>	666
10 to less than 20 years old	<u>3.1</u>	819	<u>8.3</u>	767	<u>9.7</u>	438	<u>8.0</u>	815	<u>10.4</u>	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	<u>-0.7</u>	2,062	<u>2.3</u>	1,904	<u>2.2</u>	1,120	<u>1.6</u>	2,005	<u>2.7</u>	1,671
50 years old or more	<u>-1.5</u>	941	<u>0.3</u>	943	<u>0.6</u>	549	<u>0.0</u>	972	<u>0.6</u>	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

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)													
	LESS THAN 2 YEARS OLD		2 TO LESS THAN 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	%
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)													
	LESS THAN 2 YEARS OLD		2 TO LESS THAN 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	%
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.

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:	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 THAN 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	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 THAN 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)

HOW WOULD YOU BEST DESCRIBE YOURSELF?	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 THAN 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	%
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)

GENETIC DISEASES	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 THAN 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	
	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.*

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	<u>1,838</u>	<u>90%</u>	<u>95</u>	<u>5%</u>	48	2%	64	3%	2,045	100%
2 to less than 10 years old	814	88%	54	6%	27	3%	30	3%	925	100%
10 to less than 20 years old	<u>789</u>	<u>83%</u>	<u>98</u>	<u>10%</u>	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	<u>2,009</u>	<u>85%</u>	<u>188</u>	<u>8%</u>	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: Point prevalence / Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable)

POINT PREVALENCE	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 THAN 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 (point prevalence between 5/10 000 and 1/100 000)	<u>772</u>	<u>20%</u>	<u>418</u>	<u>11%</u>	<u>481</u>	<u>13%</u>	<u>473</u>	<u>12%</u>	<u>1,127</u>	<u>29%</u>	<u>572</u>	<u>15%</u>	3,843	100%
Respondents with ultra-rare diseases (point prevalence <1/100 000)	<u>365</u>	<u>33%</u>	<u>170</u>	<u>16%</u>	<u>102</u>	<u>9%</u>	<u>107</u>	<u>10%</u>	<u>239</u>	<u>22%</u>	<u>111</u>	<u>10%</u>	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: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable)

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	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 THAN 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%

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 THAN 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)													
	LESS THAN 2 YEARS OLD		2 TO LESS THAN 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	<u>900</u>	<u>38%</u>	<u>314</u>	<u>13%</u>	<u>190</u>	<u>8%</u>	<u>210</u>	<u>9%</u>	<u>563</u>	<u>24%</u>	<u>189</u>	<u>8%</u>	2,366	100%
No	<u>1,099</u>	<u>19%</u>	<u>586</u>	<u>10%</u>	<u>744</u>	<u>13%</u>	<u>733</u>	<u>13%</u>	<u>1,723</u>	<u>30%</u>	<u>874</u>	<u>15%</u>	5,759	100%
Don't know	46	20%	25	11%	18	8%	35	15%	67	29%	<u>44</u>	<u>19%</u>	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)													
	LESS THAN 2 YEARS OLD		2 TO LESS THAN 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	<u>963</u>	<u>20%</u>	506	10%	<u>600</u>	<u>12%</u>	<u>651</u>	<u>14%</u>	<u>1,488</u>	<u>31%</u>	612	13%	4,820	100%
No	<u>908</u>	<u>30%</u>	<u>359</u>	<u>12%</u>	<u>295</u>	<u>10%</u>	<u>282</u>	<u>9%</u>	<u>723</u>	<u>24%</u>	413	14%	2,980	100%
Don't know	<u>174</u>	<u>31%</u>	60	11%	57	10%	<u>45</u>	<u>8%</u>	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)													
	LESS THAN 2 YEARS OLD		2 TO LESS THAN 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	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)													
	LESS THAN 2 YEARS OLD		2 TO LESS THAN 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	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.

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 THAN 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 than 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	<u>46</u>	<u>2%</u>	<u>270</u>	<u>13%</u>	<u>848</u>	<u>41%</u>	379	19%	138	7%	364	18%	2,045	100%
2 to less than 10 years old	11	1%	<u>70</u>	<u>8%</u>	381	41%	194	21%	74	8%	<u>195</u>	<u>21%</u>	925	100%
10 to less than 20 years old	9	1%	<u>70</u>	<u>7%</u>	<u>346</u>	<u>36%</u>	182	19%	86	9%	<u>259</u>	<u>27%</u>	952	100%
20 to less than 30 years old	6	1%	93	10%	424	43%	185	19%	79	8%	<u>191</u>	<u>20%</u>	978	100%
30 to less than 50 years old	<u>17</u>	<u>1%</u>	<u>217</u>	<u>9%</u>	1,062	45%	<u>530</u>	<u>23%</u>	197	8%	<u>330</u>	<u>14%</u>	2,353	100%
50 years old or more	12	1%	<u>138</u>	<u>12%</u>	<u>605</u>	<u>55%</u>	<u>193</u>	<u>17%</u>	<u>68</u>	<u>6%</u>	<u>91</u>	<u>8%</u>	1,107	100%
TOTAL	104	1%	1,048	10%	3,655	35%	1,048	10%	1,048	10%	1,048	10%	8,360	80%

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	<u>338</u>	<u>17%</u>	<u>679</u>	<u>33%</u>	<u>1,028</u>	<u>50%</u>	2,045	100%
2 to less than 10 years old	176	19%	377	41%	<u>372</u>	<u>40%</u>	925	100%
10 to less than 20 years old	176	18%	<u>489</u>	<u>51%</u>	<u>287</u>	<u>30%</u>	952	100%
20 to less than 30 years old	185	19%	<u>507</u>	<u>52%</u>	<u>286</u>	<u>29%</u>	978	100%
30 to less than 50 years old	479	20%	<u>1,183</u>	<u>50%</u>	<u>691</u>	<u>29%</u>	2,353	100%
50 years old or more	<u>242</u>	<u>22%</u>	<u>420</u>	<u>38%</u>	<u>445</u>	<u>40%</u>	1,107	100%
TOTAL	1,596	19%	3,655	44%	3,109	37%	8,360	80%

Under-represented elements

Over-represented elements

The relationship is very significant. p-value= < 0,01 ; Chi2= 293.4 ; dof= 10.

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 than 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	

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 than 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	2,488	28%	4,644	48%	2,484	28%	9,616	

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	<u>1,648</u>	<u>81%</u>	<u>332</u>	<u>16%</u>	<u>65</u>	<u>3%</u>	2,045	100%
2 to less than 10 years old	<u>657</u>	<u>71%</u>	<u>218</u>	<u>24%</u>	<u>50</u>	<u>5%</u>	925	100%
10 to less than 20 years old	484	51%	388	41%	80	8%	952	100%
20 to less than 30 years old	<u>425</u>	<u>43%</u>	<u>461</u>	<u>47%</u>	<u>92</u>	<u>9%</u>	978	100%
30 to less than 50 years old	<u>863</u>	<u>37%</u>	<u>1,304</u>	<u>55%</u>	186	8%	2,353	100%
50 years old or more	<u>300</u>	<u>27%</u>	<u>681</u>	<u>62%</u>	<u>126</u>	<u>11%</u>	1,107	100%
TOTAL	4,377	52%	3,384	40%	599	7%	8,360	

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	<u>1,788</u>	<u>87%</u>	<u>190</u>	<u>9%</u>	<u>67</u>	<u>3%</u>	2,045	100%
2 to less than 10 years old	<u>830</u>	<u>90%</u>	<u>73</u>	<u>8%</u>	22	2%	925	100%
10 to less than 20 years old	<u>894</u>	<u>94%</u>	<u>37</u>	<u>4%</u>	21	2%	952	100%
20 to less than 30 years old	<u>919</u>	<u>94%</u>	49	5%	<u>10</u>	<u>1%</u>	978	100%
30 to less than 50 years old	<u>2,192</u>	<u>93%</u>	<u>123</u>	<u>5%</u>	<u>38</u>	<u>2%</u>	2,353	100%
50 years old or more	<u>1,038</u>	<u>94%</u>	<u>49</u>	<u>4%</u>	20	2%	1,107	100%
TOTAL	5,004	60%	3,356	40%	400	5%	8,760	

Under-represented elements Over-represented elements

The relationship is very significant. p-value= < 0,01 ; Chi2= 83.7 ; dof= 10.

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

AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE)	Have you ever needed a genetic test but could not access it because... ...YOU COULD NOT AFFORD IT?							
	YES		NO		NOT RELEVANT		TOTAL	
	N	%	N	%	N	%	N	%
Less than 2 years old	199	10%	<u>1,565</u>	<u>77%</u>	<u>281</u>	<u>14%</u>	2,045	100%
2 to less than 10 years old	<u>111</u>	<u>12%</u>	<u>679</u>	<u>73%</u>	<u>135</u>	<u>15%</u>	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%	<u>638</u>	<u>65%</u>	228	23%	978	100%
30 to less than 50 years old	249	11%	<u>1,509</u>	<u>64%</u>	<u>595</u>	<u>25%</u>	2,353	100%
50 years old or more	<u>56</u>	<u>5%</u>	<u>681</u>	<u>62%</u>	<u>370</u>	<u>33%</u>	1,107	100%
TOTAL	837	10%	5,711	68%	1,812	22%	8,360	

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?

AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE)	Have you ever needed a genetic test but could not access it because... ...IT WAS NOT AVAILABLE IN YOUR COUNTRY?							
	YES		NO		NOT RELEVANT		TOTAL	
	N	%	N	%	N	%	N	%
Less than 2 years old	<u>279</u>	<u>14%</u>	<u>1,482</u>	<u>72%</u>	<u>284</u>	<u>14%</u>	2,045	100%
2 to less than 10 years old	<u>124</u>	<u>13%</u>	<u>667</u>	<u>72%</u>	<u>134</u>	<u>14%</u>	925	100%
10 to less than 20 years old	<u>131</u>	<u>14%</u>	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	<u>213</u>	<u>9%</u>	<u>1,467</u>	<u>62%</u>	<u>673</u>	<u>29%</u>	2,353	100%
50 years old or more	<u>74</u>	<u>7%</u>	<u>628</u>	<u>57%</u>	<u>405</u>	<u>37%</u>	1,107	100%
TOTAL	919	11%	5,467	65%	1,974	24%	8,360	

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?

AGE OF THE PERSON AFFECTED BY THE RARE DISEASE WHEN THE FIRST SYMPTOMS WERE NOTICED (CALCULATED VARIABLE)	Have you ever needed a genetic test but could not access it because... ...HEALTHCARE PROFESSIONALS WERE RELUCTANT OR NOT SUFFICIENTLY INFORMED?							
	YES		NO		NOT RELEVANT		TOTAL	
	N	%	N	%	N	%	N	%
Less than 2 years old	543	27%	<u>1,254</u>	<u>61%</u>	<u>248</u>	<u>12%</u>	2,045	100%
2 to less than 10 years old	<u>272</u>	<u>29%</u>	<u>535</u>	<u>58%</u>	<u>118</u>	<u>13%</u>	925	100%
10 to less than 20 years old	<u>321</u>	<u>34%</u>	<u>453</u>	<u>48%</u>	178	19%	952	100%
20 to less than 30 years old	<u>295</u>	<u>30%</u>	<u>478</u>	<u>49%</u>	205	21%	978	100%
30 to less than 50 years old	608	26%	<u>1,141</u>	<u>48%</u>	<u>604</u>	<u>26%</u>	2,353	100%
50 years old or more	<u>183</u>	<u>17%</u>	556	50%	<u>368</u>	<u>33%</u>	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...

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...															
	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%	<u>341</u>	<u>21%</u>	<u>269</u>	<u>16%</u>	<u>17</u>	<u>1%</u>	41	2%	<u>369</u>	<u>22%</u>	1,648	
2 to less than 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%	<u>48</u>	<u>10%</u>	<u>33</u>	<u>7%</u>	13	3%	8	2%	<u>154</u>	<u>32%</u>	484	
20 to less than 30 years old	122	29%	144	34%	54	13%	<u>22</u>	<u>5%</u>	16	4%	12	3%	118	28%	425	
30 to less than 50 years old	233	27%	277	32%	<u>98</u>	<u>11%</u>	<u>56</u>	<u>6%</u>	<u>43</u>	<u>5%</u>	15	2%	<u>260</u>	<u>30%</u>	863	
50 years old or more	88	22%	88	22%	<u>85</u>	<u>10%</u>	<u>44</u>	<u>5%</u>	9	2%	9	1%	<u>104</u>	<u>25%</u>	388	

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 than 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	100%	210	100%	6,700	100%	7,368	100%

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 than 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%
TOTAL	458	100%	572	100%	736	100%	1,132	100%	572	100%	161	100%	7,368	100%

Under-represented elements

Over-represented elements

The relationship is very significant. p-value= < 0,01 ; Chi2= 60.5 ; dof= 25.

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 than 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 than 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	

Under-represented elements

Over-represented elements

The relationship is very significant. p-value= < 0,01 ; Chi2= 46.6 ; dof= 10.

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%	<u>1,470</u>	<u>82%</u>	37	2%	1,787	100%
2 to less than 10 years old	<u>143</u>	<u>17%</u>	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%	<u>896</u>	<u>86%</u>	<u>9</u>	<u>1%</u>	1,038	100%
TOTAL	1,101	15%	8,412	89%	100	1%	9,613	100%

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	<u>515</u>	<u>25%</u>	<u>1,485</u>	<u>73%</u>	<u>45</u>	<u>2%</u>	2,045	100%
2 to less than 10 years old	<u>230</u>	<u>25%</u>	<u>676</u>	<u>73%</u>	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	<u>414</u>	<u>18%</u>	<u>1,908</u>	<u>81%</u>	31	1%	2,353	100%
50 years old or more	<u>145</u>	<u>13%</u>	<u>956</u>	<u>86%</u>	<u>6</u>	<u>1%</u>	1,107	100%
TOTAL	1,053	20%	8,555	78%	100	1%	9,708	100%

Under-represented elements

Over-represented elements

The relationship is very significant. p-value= < 0,01 ; Chi2= 110.7 ; dof= 10.

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 than 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	

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 than 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.

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 than 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 than 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.

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	134	7%	908	47%	717	37%	96	5%	72	4%	1,927	100%
2 to less than 10 years old	91	11%	432	50%	281	33%	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%	410	39%	403	39%	65	6%	50	5%	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	114	6%	1,156	60%	506	26%	97	5%	54	3%	1,927	100%
2 to less than 10 years old	64	7%	523	61%	216	25%	46	5%	13	2%	862	100%
10 to less than 20 years old	78	9%	516	59%	237	27%	31	4%	14	2%	876	100%
20 to less than 30 years old	72	8%	510	56%	290	32%	39	4%	6	1%	917	100%
30 to less than 50 years old	172	8%	1,161	54%	676	31%	129	6%	25	1%	2,163	100%
50 years old or more	84	8%	533	51%	353	34%	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.

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 than 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 than 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.

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 than 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 than 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 than 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 than 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...											
	...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 than 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.

Chapter 4.

Family members were
previously diagnosed

Question 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	<u>1.9</u>	785	<u>5.7</u>	757	<u>5.9</u>	527	<u>1.3</u>	834	<u>7.1</u>	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.

Question 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 THAN 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.*

Question 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	

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	

Under-represented elements Over-represented elements

The relationship is very significant. *p-value= < 0,01 ; Chi2= 13.5 ; dof= 3.*

Question asked only to respondents who are diagnosed

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

HOW WOULD YOU BEST DESCRIBE YOURSELF?	FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE					
	YES		NO		TOTAL	
	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

TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE	FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME DISEASE					
	YES		NO		TOTAL	
	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	

Under-represented elements Over-represented elements

The relationship is very significant. *p-value= < 0,01 ; Chi2= 39.3 ; dof= 2.*

Question 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	

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	<u>494</u>	<u>21%</u>	<u>1,887</u>	<u>79%</u>	2,381	100%
1-9 / 100 000	<u>222</u>	<u>11%</u>	<u>1,744</u>	<u>89%</u>	1,966	100%
1-9 / 1 000 000	57	13%	397	87%	454	100%
<1 / 1 000 000	<u>95</u>	<u>12%</u>	<u>727</u>	<u>88%</u>	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.*

Question 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	<u>294</u>	<u>65%</u>	<u>160</u>	<u>35%</u>	454	100%
Hypermobile Ehlers-Danlos syndrome	43	14%	268	86%	311	100%
Sarcoidosis	<u>7</u>	<u>4%</u>	<u>163</u>	<u>96%</u>	170	100%
Classical Ehlers-Danlos syndrome	13	10%	122	90%	135	100%
Williams syndrome	<u>0</u>	<u>0%</u>	<u>134</u>	<u>100%</u>	134	100%
Cystic fibrosis	14	11%	113	89%	127	100%
Myasthenia gravis	<u>3</u>	<u>3%</u>	<u>115</u>	<u>97%</u>	118	100%
Systemic sclerosis	<u>5</u>	<u>5%</u>	<u>100</u>	<u>95%</u>	105	100%
Tuberous sclerosis complex	8	8%	90	92%	98	100%
Neurofibromatosis type 1	<u>19</u>	<u>21%</u>	<u>73</u>	<u>79%</u>	92	100%
Interstitial cystitis	<u>2</u>	<u>3%</u>	<u>72</u>	<u>97%</u>	74	100%
Addison disease	5	7%	68	93%	73	100%
22q11.2 deletion syndrome	<u>3</u>	<u>4%</u>	<u>65</u>	<u>96%</u>	68	100%
Chronic inflammatory demyelinating polyneuropathy	<u>1</u>	<u>2%</u>	<u>62</u>	<u>98%</u>	63	100%
Perineural cyst	3	5%	58	95%	61	100%
Acute inflammatory demyelinating polyradiculoneuropathy	<u>1</u>	<u>2%</u>	<u>61</u>	<u>98%</u>	62	100%
Bett syndrome	<u>4</u>	<u>2%</u>	<u>56</u>	<u>98%</u>	60	100%

Under-represented elements

Over-represented elements

The relationship is very significant. p-value= < 0,01 ; Chi2= 3,382.7 ; dof= 1,629.

Question 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	88	12%	667	88%	755	100%

Under-represented elements

Over-represented elements

The relationship is very significant. p-value= < 0,01 ; Chi2= 2,168.5 ; dof= 34.

Question 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	<u>34</u>	<u>3%</u>	<u>275</u>	<u>21%</u>	604	46%	<u>184</u>	<u>14%</u>	<u>63</u>	<u>5%</u>	<u>149</u>	<u>11%</u>	1,309	100%
No	<u>112</u>	<u>1%</u>	<u>815</u>	<u>10%</u>	3,730	44%	<u>1,701</u>	<u>20%</u>	<u>665</u>	<u>8%</u>	<u>1,399</u>	<u>17%</u>	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	<u>858</u>	<u>66%</u>	<u>442</u>	<u>34%</u>	1,300	100%
No	<u>4,717</u>	<u>56%</u>	<u>3,647</u>	<u>44%</u>	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.*

Question 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.

Question 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%	<u>1,000</u>	<u>76%</u>	<u>190</u>	<u>15%</u>	1,309	100%
No	826	10%	<u>5,693</u>	<u>68%</u>	<u>1,903</u>	<u>23%</u>	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%	<u>956</u>	<u>73%</u>	<u>222</u>	<u>17%</u>	1,309	100%
No	924	11%	<u>5,448</u>	<u>65%</u>	<u>2,050</u>	<u>24%</u>	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.*

Question 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%	<u>795</u>	<u>61%</u>	<u>177</u>	<u>14%</u>	1,309	100%
No	2,121	25%	<u>4,485</u>	<u>53%</u>	<u>1,816</u>	<u>22%</u>	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	<u>311</u>	<u>34%</u>	<u>242</u>	<u>27%</u>	<u>106</u>	<u>12%</u>	<u>45</u>	<u>5%</u>	15	2%	14	2%	260	29%	906	
No	<u>1,079</u>	<u>26%</u>	<u>1,354</u>	<u>32%</u>	<u>685</u>	<u>16%</u>	<u>441</u>	<u>10%</u>	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.

Question 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.*

Question 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	

Under-represented elements Over-represented elements

The relationship is very significant. p-value= < 0,01 ; Chi2= 20.9 ; dof= 2.

Question 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.*

Question 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.

Question 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	

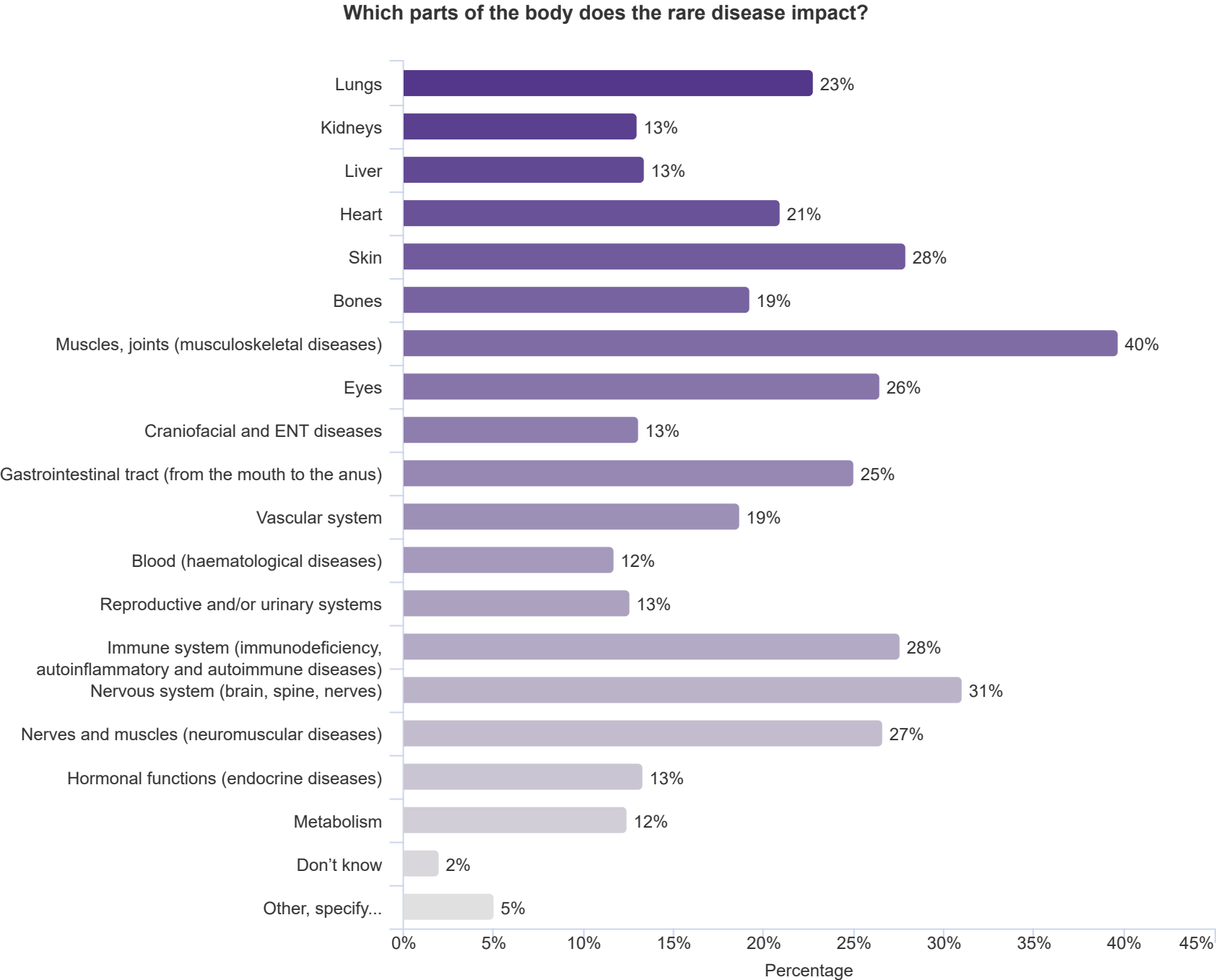
Under-represented elements Over-represented elements

The relationship is very significant. p-value= < 0,01 ; Chi2= 79.4 ; dof= 6.

Chapter 5.

Symptoms
characteristics:
body parts

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

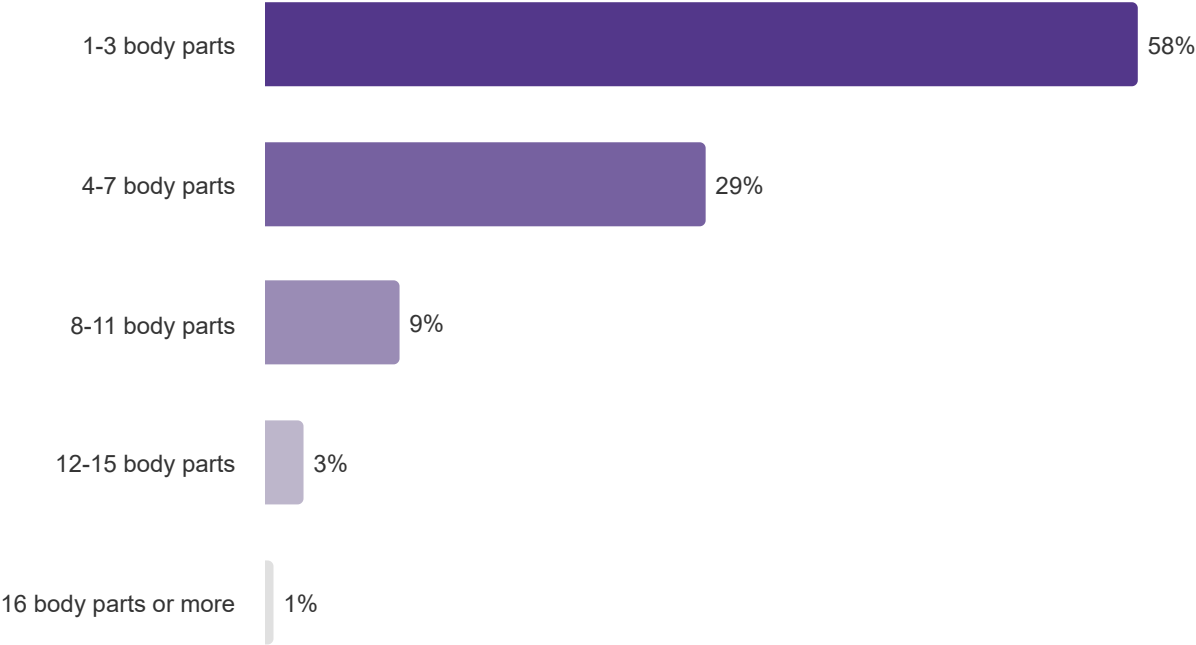


Complexity of the rare disease: number of body parts impacted by the rare disease

Number body parts impacted by the rare disease (or index of disease complexity)
Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?"

	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

Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable
computing answers to the question "Which parts of the body does the rare disease impact?"



Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?"	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

Cross: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / Are you a patient representative, i.e. involved in policy activities to support the cause of rare diseases?

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	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%

Under-represented elements Over-represented elements

The relationship is significant. p-value= 0.0 ; Chi2= 17.7 ; dof= 8.

Cross: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / Age of the person affected by the rare disease when the first symptoms were noticed (calculated variable)

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	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 THAN 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%

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 / Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?"

GENDER OF THE PERSON AFFECTED BY THE RARE DISEASE	NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"											
	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%

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 / Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?"

TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE	NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"											
	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.*

Cross: Would you say that you, or the person you care for, live in a: / Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?"

WOULD YOU SAY THAT YOU, OR THE PERSON YOU CARE FOR, LIVE IN A:	NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"					
	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: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / Please select the sentence that best describes your situation or the situation of the person you care for:

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	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

Cross: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / Genetic diseases

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	GENETIC DISEASES					
	GENETIC DISEASES		NON GENETIC DISEASES		TOTAL	
	N	%	N	%	N	%
1-3 body parts	<u>2,995</u>	<u>64%</u>	<u>1,665</u>	<u>36%</u>	4,660	100%
4-7 body parts	<u>1,726</u>	<u>72%</u>	<u>670</u>	<u>28%</u>	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	<u>41</u>	<u>82%</u>	<u>9</u>	<u>18%</u>	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: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / Point prevalence

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	POINT PREVALENCE					
	RESPONDENTS WITH LESS RARE DISEASES (POINT PREVALENCE BETWEEN 5/10 000 AND 1/100 000)		RESPONDENTS WITH ULTRA-RARE DISEASES (POINT PREVALENCE <1/100 000)		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: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / The rare disease was diagnosed before birth

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	THE RARE DISEASE WAS DIAGNOSED BEFORE BIRTH					
	YES		NO		TOTAL	
	N	%	N	%	N	%
1-3 body parts	<u>147</u>	<u>3%</u>	<u>5,539</u>	<u>97%</u>	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: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / The rare disease was diagnosed through standard tests carried out at birth

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	THE RARE DISEASE WAS DIAGNOSED THROUGH STANDARD TESTS CARRIED OUT AT BIRTH					
	YES		NO		TOTAL	
	N	%	N	%	N	%
1-3 body parts	<u>256</u>	<u>5%</u>	<u>5,307</u>	<u>95%</u>	5,563	100%
4-7 body parts	111	4%	2,678	96%	2,789	100%
8-11 body parts	<u>24</u>	<u>3%</u>	<u>851</u>	<u>97%</u>	875	100%
12-15 body parts	<u>2</u>	<u>1%</u>	<u>250</u>	<u>99%</u>	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.*

Cross: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	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: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	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.

Cross: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / ...wrongly attributed to another physical disease?

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	...WRONGLY ATTRIBUTED TO ANOTHER PHYSICAL DISEASE?							
	YES, ONE TIME		YES, SEVERAL TIMES		NO		TOTAL	
	N	%	N	%	N	%	N	%
1-3 body parts	<u>1,230</u>	<u>20%</u>	<u>2,152</u>	<u>35%</u>	<u>2,721</u>	<u>45%</u>	6,103	100%
4-7 body parts	552	18%	<u>1,513</u>	<u>49%</u>	<u>1,016</u>	<u>33%</u>	3,081	100%
8-11 body parts	<u>135</u>	<u>14%</u>	<u>587</u>	<u>62%</u>	<u>229</u>	<u>24%</u>	951	100%
12-15 body parts	<u>30</u>	<u>10%</u>	<u>213</u>	<u>74%</u>	<u>43</u>	<u>15%</u>	286	100%
16 body parts or more	<u>3</u>	<u>5%</u>	<u>55</u>	<u>85%</u>	<u>7</u>	<u>11%</u>	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: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / ...neglected, not taken seriously and/or considered as psychological?

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	...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	<u>759</u>	<u>12%</u>	<u>2,405</u>	<u>39%</u>	<u>2,939</u>	<u>48%</u>	6,103	100%
4-7 body parts	385	12%	<u>1,592</u>	<u>52%</u>	<u>1,104</u>	<u>36%</u>	3,081	100%
8-11 body parts	<u>79</u>	<u>8%</u>	<u>652</u>	<u>69%</u>	<u>220</u>	<u>23%</u>	951	100%
12-15 body parts	<u>17</u>	<u>6%</u>	<u>231</u>	<u>81%</u>	<u>38</u>	<u>13%</u>	286	100%
16 body parts or more	6	9%	<u>54</u>	<u>83%</u>	<u>5</u>	<u>8%</u>	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: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / 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.

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	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	<u>1,656</u>	<u>27%</u>	<u>2,467</u>	<u>40%</u>	<u>1,980</u>	<u>32%</u>	6,103	100%
4-7 body parts	774	25%	<u>1,661</u>	<u>54%</u>	<u>646</u>	<u>21%</u>	3,081	100%
8-11 body parts	<u>203</u>	<u>21%</u>	<u>614</u>	<u>65%</u>	<u>134</u>	<u>14%</u>	951	100%
12-15 body parts	<u>46</u>	<u>16%</u>	<u>219</u>	<u>77%</u>	<u>21</u>	<u>7%</u>	286	100%
16 body parts or more	<u>4</u>	<u>6%</u>	<u>57</u>	<u>88%</u>	<u>4</u>	<u>6%</u>	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: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / Genetic test(s) looking for genetic changes (also called mutations or variants)

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	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	<u>3,054</u>	<u>50%</u>	<u>2,561</u>	<u>42%</u>	488	8%	6,103	100%
4-7 body parts	<u>1,741</u>	<u>57%</u>	<u>1,098</u>	<u>36%</u>	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.

Cross: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / Other test(s) such as clinical examination(s), medical imaging (MRI, scans...), biopsy, biochemical test(s) (blood or urine tests...), etc

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	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	

Under-represented elements Over-represented elements

The relationship is very significant. *p-value= < 0,01 ; Chi2= 20.2 ; dof= 8.*

Cross: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / ...you could not afford it?

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

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	...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	

Under-represented elements Over-represented elements

The relationship is very significant. *p-value= < 0,01 ; Chi2= 255.4 ; dof= 8.*

Cross: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / ...it was not available in your country?

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	Have you ever needed a genetic test but could not access it because... ...IT WAS NOT AVAILABLE IN YOUR COUNTRY?							
	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	

Under-represented elements Over-represented elements

The relationship is very significant. p-value= < 0,01 ; Chi2= 148.7 ; dof= 8.

Cross: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / ...healthcare professionals were reluctant or not sufficiently informed?

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	Have you ever needed a genetic test but could not access it because... ...HEALTHCARE PROFESSIONALS WERE RELUCTANT OR NOT SUFFICIENTLY INFORMED?							
	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	

Under-represented elements Over-represented elements

The relationship is very significant. p-value= < 0,01 ; Chi2= 383.6 ; dof= 8.

Cross: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / To your knowledge, the genetic test(s) that were conducted targeted...

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	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	
<div><div></div> Under-represented elements<div></div> Over-represented elements</div>																

The relationship is very significant. p-value= < 0,01 ; Chi2= 70.0 ; dof= 24.

Cross: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / Did you ever request a private company or laboratory to conduct genetic testing to diagnose the disease?

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	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	
<div><div></div> Under-represented elements<div></div> Over-represented elements</div>								

The relationship is very significant. p-value= < 0,01 ; Chi2= 52.6 ; dof= 8

Cross: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / In general, how satisfied are you with how the results of the GENETIC TESTS were given to you?

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	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	%
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: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / 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)?

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	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	%
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.

Cross: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / Genetic tests

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	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: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / Other diagnostic tests such as clinical examination(s), medical imaging (MRI, scans...), biopsy, biochemical test(s) (blood or urine tests...), etc.

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	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	

Under-represented elements Over-represented elements

The relationship is significant. *p-value= 0.0 ; Chi2= 18.8 ; dof= 8.*

Cross: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / Additional advice from a healthcare professional specialised in the rare disease (in person or virtually)

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	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: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / ...psychological support

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	...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: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / ...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.

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	...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	<u>1,316</u>	<u>22%</u>	<u>266</u>	<u>4%</u>	<u>764</u>	<u>13%</u>	<u>1,113</u>	<u>18%</u>	<u>2,644</u>	<u>43%</u>	6,103	100%
4-7 body parts	597	19%	<u>97</u>	<u>3%</u>	<u>486</u>	<u>16%</u>	<u>387</u>	<u>13%</u>	<u>1,514</u>	<u>49%</u>	3,081	100%
8-11 body parts	<u>136</u>	<u>14%</u>	<u>20</u>	<u>2%</u>	<u>156</u>	<u>16%</u>	<u>115</u>	<u>12%</u>	<u>524</u>	<u>55%</u>	951	100%
12-15 body parts	<u>30</u>	<u>10%</u>	<u>4</u>	<u>1%</u>	41	14%	<u>12</u>	<u>4%</u>	<u>199</u>	<u>70%</u>	286	100%
16 body parts or more	<u>4</u>	<u>6%</u>	4	6%	<u>16</u>	<u>25%</u>	<u>0</u>	<u>0%</u>	<u>41</u>	<u>63%</u>	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: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / ...financial support including social security benefits

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	...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	<u>855</u>	<u>14%</u>	<u>169</u>	<u>3%</u>	<u>630</u>	<u>10%</u>	<u>2,368</u>	<u>39%</u>	<u>2,037</u>	<u>34%</u>	6,059	100%
4-7 body parts	428	14%	<u>55</u>	<u>2%</u>	<u>408</u>	<u>13%</u>	<u>931</u>	<u>30%</u>	<u>1,237</u>	<u>40%</u>	3,059	100%
8-11 body parts	<u>98</u>	<u>10%</u>	16	2%	<u>140</u>	<u>15%</u>	<u>201</u>	<u>21%</u>	<u>491</u>	<u>52%</u>	946	100%
12-15 body parts	<u>23</u>	<u>8%</u>	<u>1</u>	<u>0%</u>	40	14%	<u>40</u>	<u>14%</u>	<u>181</u>	<u>64%</u>	285	100%
16 body parts or more	<u>1</u>	<u>2%</u>	2	3%	<u>14</u>	<u>22%</u>	<u>4</u>	<u>6%</u>	<u>43</u>	<u>67%</u>	64	100%
TOTAL	1,405	13%	243	2%	1,232	12%	3,544	34%	3,989	38%	10,413	

Under-represented elements

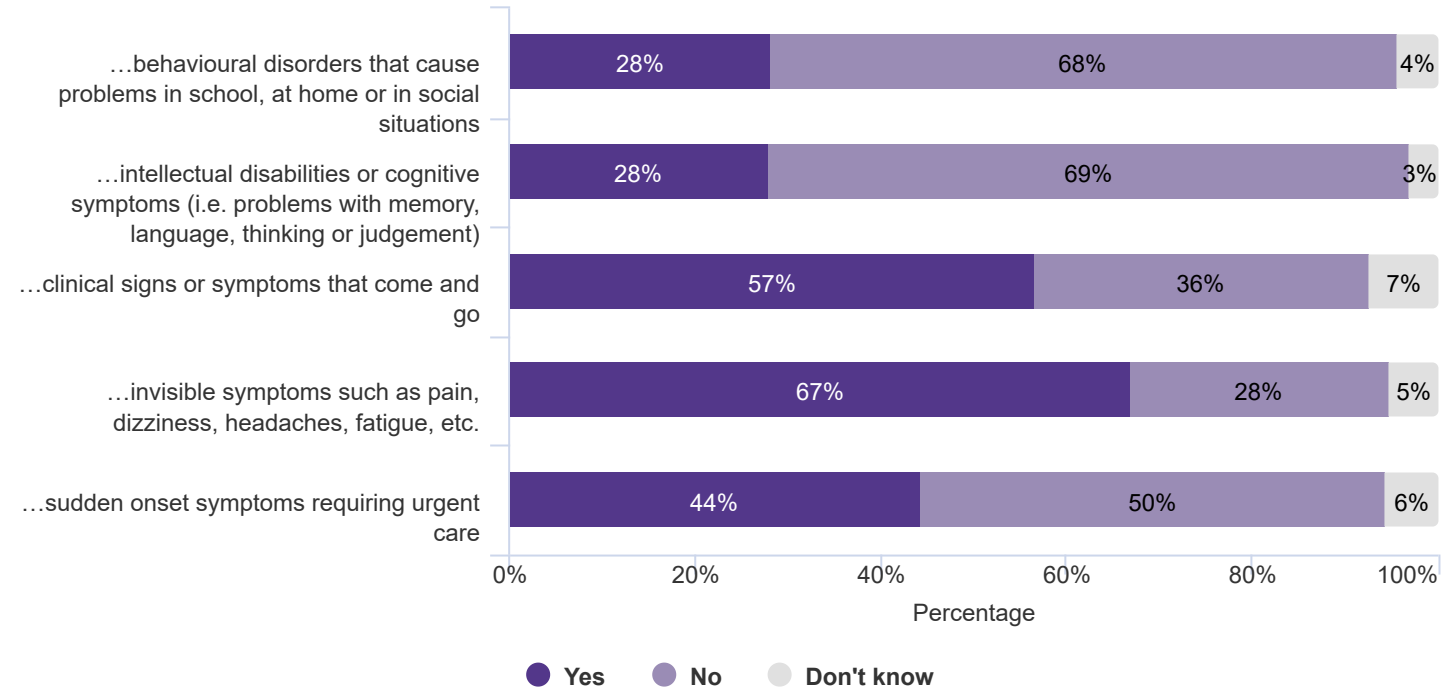
Over-represented elements

The relationship is very significant. p-value= < 0,01 ; Chi2= 363.7 ; dof= 16.

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

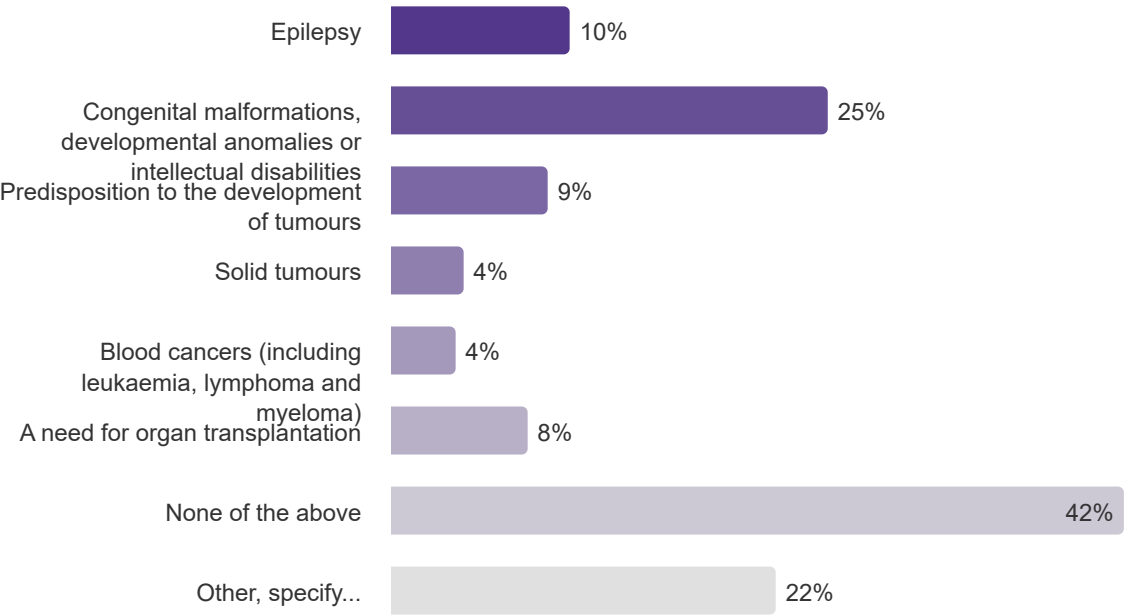
Did the first symptoms include...



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:



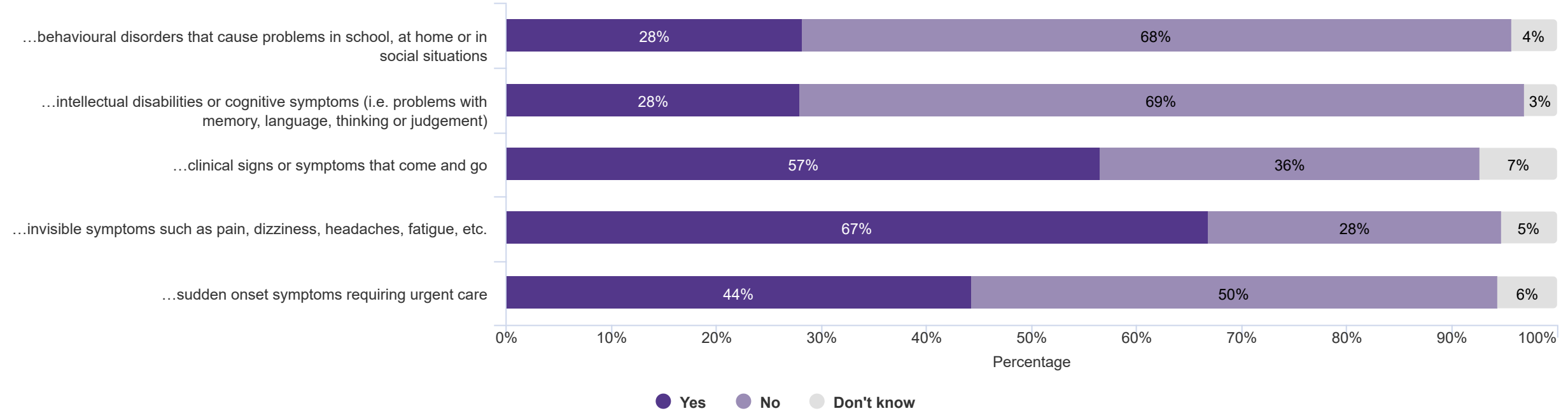
Chapter 6.

Symptoms
characteristics:
types of symptoms

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...



Did the first symptoms include...

Time from the first symptoms to the different steps of the diagnosis journey depending on the type of symptoms

...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

Did the first symptoms include...

Time from the first symptoms to the different steps of the diagnosis journey depending on the type of symptoms

...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

Did the first symptoms include...

Time from the first symptoms to the different steps of the diagnosis journey depending on the type of symptoms

...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	<u>4.0</u>	4,324	<u>4.4</u>	2,467	<u>4.1</u>	4,488	<u>5.4</u>	3,680
No	0.6	2,744	<u>2.8</u>	2,547	<u>3.0</u>	1,600	<u>2.8</u>	2,858	<u>3.8</u>	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

Did the first symptoms include...

Time from the first symptoms to the different steps of the diagnosis journey depending on the type of symptoms

...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	<u>3.9</u>	5,099	<u>4.5</u>	2,939	<u>4.2</u>	5,271	<u>5.1</u>	4,309
No	0.6	2,071	<u>2.6</u>	1,902	<u>2.6</u>	1,197	<u>2.4</u>	2,191	<u>3.9</u>	1,877
Don't know	0.5	377	<u>1.8</u>	321	2.6	199	<u>2.7</u>	381	<u>3.8</u>	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

Did the first symptoms include...

Time from the first symptoms to the different steps of the diagnosis journey depending on the type of symptoms

...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						
	YES		NO		DON'T KNOW		TO
	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 / ...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						
	YES		NO		DON'T KNOW		TO
	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 / ...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)						
	YES		NO		DON'T KNOW		TO
	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 / ...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						
	YES		NO		DON'T KNOW		TO
	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.

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	

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? / ...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	<u>224</u>	<u>49%</u>	<u>204</u>	<u>45%</u>	27	6%	455	100%
between 16 and 19 y.o.	<u>1,165</u>	<u>47%</u>	<u>1,141</u>	<u>46%</u>	<u>158</u>	<u>6%</u>	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	<u>1,337</u>	<u>43%</u>	<u>1,659</u>	<u>53%</u>	<u>149</u>	<u>5%</u>	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%	<u>248</u>	<u>8%</u>	<u>1,161</u>	<u>39%</u>	<u>642</u>	<u>22%</u>	<u>253</u>	<u>9%</u>	<u>608</u>	<u>21%</u>	2,957	100%
No	96	1%	<u>852</u>	<u>12%</u>	<u>3,218</u>	<u>45%</u>	<u>1,314</u>	<u>19%</u>	<u>507</u>	<u>7%</u>	<u>1,098</u>	<u>15%</u>	7,085	100%
Don't know	<u>14</u>	<u>3%</u>	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: ...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	

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	

Under-represented elements Over-represented elements

The relationship is very significant. p-value= < 0,01 ; Chi2= 27.0 ; dof= 2.

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.

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.*

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.

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%	<u>1,368</u>	<u>47%</u>	<u>1,054</u>	<u>36%</u>	2,936	100%
No	<u>1,383</u>	<u>19%</u>	<u>3,010</u>	<u>42%</u>	<u>2,843</u>	<u>39%</u>	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%	<u>1,602</u>	<u>55%</u>	<u>1,007</u>	<u>34%</u>	2,936	100%
No	876	12%	<u>3,161</u>	<u>44%</u>	<u>3,199</u>	<u>44%</u>	7,236	100%
Don't know	43	14%	<u>171</u>	<u>54%</u>	<u>100</u>	<u>32%</u>	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%	<u>1,493</u>	<u>51%</u>	<u>656</u>	<u>22%</u>	2,936	100%
No	1,817	25%	<u>3,366</u>	<u>47%</u>	<u>2,053</u>	<u>28%</u>	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

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	

Under-represented elements Over-represented elements

The relationship is very significant. p-value= < 0,01 ; Chi2= 826.2 ; dof= 4.

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.

Chapter 7.

Prevention

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.*

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 ; χ^2 = 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 ; χ^2 = 5.4 ; dof= 1.

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?													
	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. $p\text{-value}= 0.0$; $\text{Chi}^2= 14.3$; $\text{dof}= 5$.

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?													
	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. $p\text{-value}= < 0,01$; $\text{Chi}^2= 141.8$; $\text{dof}= 5$.

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.

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.

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.

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.*

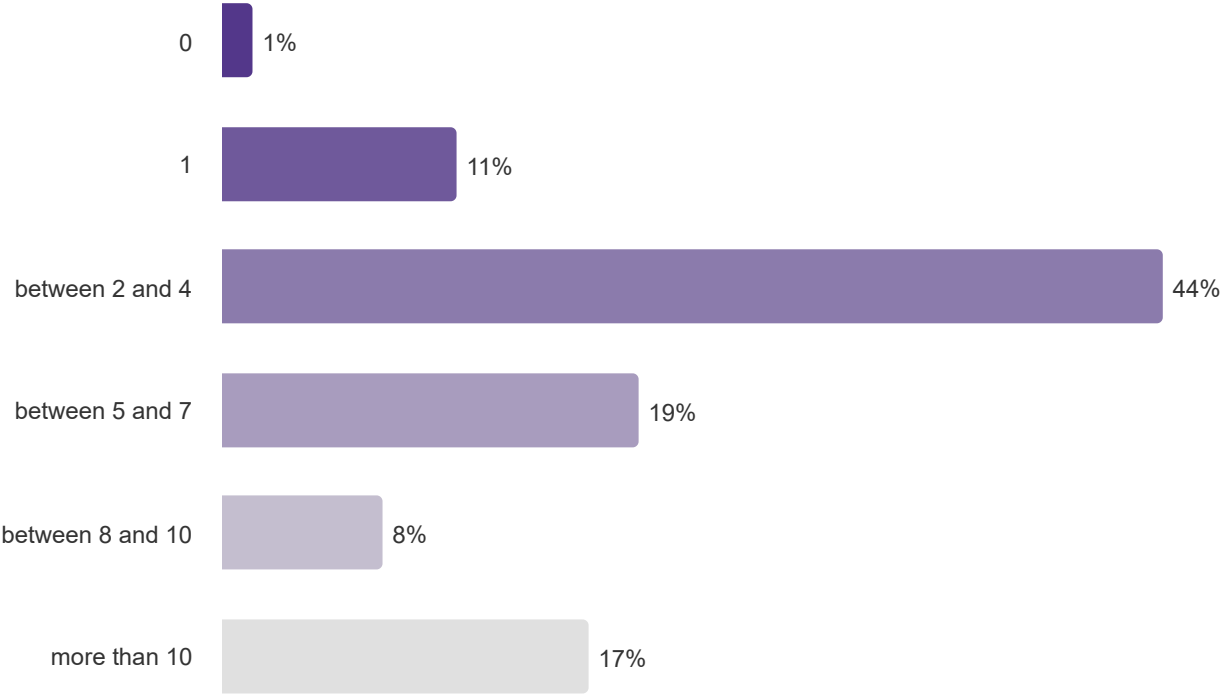
Chapter 8.

Number of healthcare professionals consulted

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?



Time from the first symptoms to the different steps of the diagnosis journey depending on the number of healthcare professional consulted while seeking a diagnosis

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	<u>1.3</u>	846	<u>2.4</u>	815	<u>3.1</u>	532	<u>1.0</u>	924	<u>2.5</u>	824
between 2 and 4	0.7	3,394	<u>2.5</u>	3,213	<u>2.5</u>	2,042	<u>1.8</u>	3,478	<u>3.1</u>	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	<u>4.4</u>	575	4.8	304	<u>5.5</u>	592	<u>6.9</u>	455
more than 10	<u>-0.3</u>	1,383	<u>6.9</u>	1,218	<u>8.6</u>	651	<u>9.7</u>	1,286	<u>10.9</u>	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.

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 than 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.

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?											
	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?											
	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?	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 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: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis?

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	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	%
1-3 body parts	<u>863</u>	<u>14%</u>	<u>3,067</u>	<u>50%</u>	<u>1,113</u>	<u>18%</u>	<u>406</u>	<u>7%</u>	<u>654</u>	<u>11%</u>	6,103	100%
4-7 body parts	<u>345</u>	<u>11%</u>	<u>1,171</u>	<u>38%</u>	<u>700</u>	<u>23%</u>	<u>258</u>	<u>8%</u>	<u>607</u>	<u>20%</u>	3,081	100%
8-11 body parts	<u>83</u>	<u>9%</u>	<u>269</u>	<u>28%</u>	174	18%	<u>93</u>	<u>10%</u>	<u>332</u>	<u>35%</u>	951	100%
12-15 body parts	<u>12</u>	<u>4%</u>	<u>54</u>	<u>19%</u>	<u>38</u>	<u>13%</u>	27	9%	<u>155</u>	<u>54%</u>	286	100%
16 body parts or more	<u>2</u>	<u>3%</u>	<u>8</u>	<u>12%</u>	8	12%	7	11%	<u>40</u>	<u>62%</u>	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.

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)	<u>1,182</u>	<u>13%</u>	<u>4,122</u>	<u>46%</u>	1,731	19%	<u>645</u>	<u>7%</u>	<u>1,368</u>	<u>15%</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>65</u>	<u>9%</u>	<u>236</u>	<u>31%</u>	<u>172</u>	<u>23%</u>	<u>85</u>	<u>11%</u>	<u>202</u>	<u>27%</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>25</u>	<u>8%</u>	<u>94</u>	<u>31%</u>	61	20%	28	9%	<u>98</u>	<u>32%</u>	306	100%
I know that the disease is rare but the name or the cause have NOT BEEN IDENTIFIED	<u>27</u>	<u>8%</u>	<u>109</u>	<u>31%</u>	68	20%	30	9%	<u>114</u>	<u>33%</u>	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	<u>118</u>	<u>26%</u>	<u>230</u>	<u>50%</u>	<u>53</u>	<u>12%</u>	<u>22</u>	<u>5%</u>	<u>35</u>	<u>8%</u>	458	100%
Hypermobile Ehlers-Danlos syndrome	<u>7</u>	<u>2%</u>	<u>44</u>	<u>14%</u>	55	17%	<u>39</u>	<u>12%</u>	<u>172</u>	<u>54%</u>	317	100%
Sarcoidosis	20	12%	86	51%	37	22%	10	6%	<u>17</u>	<u>10%</u>	170	100%
Classical Ehlers-Danlos syndrome	<u>5</u>	<u>4%</u>	<u>33</u>	<u>24%</u>	25	18%	11	8%	<u>63</u>	<u>46%</u>	137	100%
Williams syndrome	<u>35</u>	<u>26%</u>	57	42%	27	20%	5	4%	<u>12</u>	<u>9%</u>	136	100%
Cystic fibrosis	<u>34</u>	<u>27%</u>	62	48%	22	17%	<u>3</u>	<u>2%</u>	<u>7</u>	<u>5%</u>	128	100%
Myasthenia gravis	10	8%	<u>67</u>	<u>56%</u>	20	17%	8	7%	15	13%	120	100%
Systemic sclerosis	13	12%	<u>66</u>	<u>62%</u>	17	16%	6	6%	<u>5</u>	<u>5%</u>	107	100%
Tuberous sclerosis complex	<u>19</u>	<u>19%</u>	50	51%	19	19%	4	4%	<u>6</u>	<u>6%</u>	98	100%
Neurofibromatosis type 1	15	16%	49	53%	15	16%	6	7%	<u>7</u>	<u>8%</u>	92	100%
Interstitial cystitis	<u>3</u>	<u>4%</u>	<u>22</u>	<u>30%</u>	<u>25</u>	<u>34%</u>	<u>12</u>	<u>16%</u>	12	16%	74	100%
Addison disease	6	8%	35	48%	18	25%	5	7%	9	12%	73	100%
22q11.2 deletion syndrome	<u>17</u>	<u>25%</u>	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	<u>2</u>	<u>3%</u>	<u>36</u>	<u>58%</u>	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%	<u>16</u>	<u>31%</u>	10	19%	7	13%	11	21%	52	100%
Fragile X syndrome	4	8%	23	47%	12	24%	6	12%	4	8%	49	100%
Behçet disease	<u>1</u>	<u>2%</u>	<u>14</u>	<u>30%</u>	<u>15</u>	<u>32%</u>	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?											
	0-1		BETWEEN 2 AND 4		BETWEEN 5 AND 7		BETWEEN 8 AND 10		MORE THAN 10		TOTAL	
	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.

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	%
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?

...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	%
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.*

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	%
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?											
	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.

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. $p\text{-value} < 0,01$; $\text{Chi}^2 = 143.7$; $\text{dof} = 8$.

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. $p\text{-value} < 0,01$; $\text{Chi}^2 = 886.3$; $\text{dof} = 8$.

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.

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%	<u>1,102</u>	<u>46%</u>	476	20%	179	7%	<u>372</u>	<u>15%</u>	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%	<u>1,151</u>	<u>42%</u>	528	19%	219	8%	<u>509</u>	<u>18%</u>	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')	<u>184</u>	<u>10%</u>	813	45%	356	20%	134	7%	307	17%	1,794	100%
Group B ('Western Europe')	<u>682</u>	<u>13%</u>	<u>2,323</u>	<u>46%</u>	988	19%	377	7%	<u>735</u>	<u>14%</u>	5,105	100%
Group C ('Northern Europe')	394	12%	<u>1,297</u>	<u>40%</u>	631	19%	253	8%	<u>698</u>	<u>21%</u>	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.

Chapter 9.

Referred to a Centre of Expertise

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

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					
	YES		NO		TOTAL	
	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

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					
	YES		NO		TOTAL	
	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						
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					
	YES		NO		TOTAL	
	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						
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					
	YES		NO		TOTAL	
	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.*

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.*

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)	<u>5,298</u>	<u>59%</u>	<u>3,692</u>	<u>41%</u>	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	<u>322</u>	<u>43%</u>	<u>429</u>	<u>57%</u>	751	100%
I only have PARTIAL information on the name of the rare disease or the gene involved or the type of disease	<u>159</u>	<u>52%</u>	<u>146</u>	<u>48%</u>	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.*

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	

Under-represented elements Over-represented elements

The relationship is very significant. $p\text{-value} = < 0,01$; $\text{Chi}^2 = 9.8$; $\text{dof} = 1$.

Cross: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / I, or the person I care for, have been referred to a hospital unit specialised in the rare disease or group of rare diseases

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	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. $p\text{-value} = 0.0$; $\text{Chi}^2 = 12.1$; $\text{dof} = 4$.

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 / 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.

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?

I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES	Have you ever needed a genetic test but could not access it because... ...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.

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?

I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES	Have you ever needed a genetic test but could not access it because... ...IT WAS NOT AVAILABLE IN YOUR COUNTRY?							
	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?

I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES	Have you ever needed a genetic test but could not access it because... ...HEALTHCARE PROFESSIONALS WERE RELUCTANT OR NOT SUFFICIENTLY INFORMED?							
	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.

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.

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.

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

...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	<u>2.6</u>	1,448	<u>2.4</u>	838	<u>2.7</u>	1,506	<u>3.5</u>	1,274
YES, several times	0.4	3,471	<u>4.8</u>	3,203	<u>5.7</u>	1,753	<u>5.9</u>	3,389	<u>6.9</u>	2,720
NO	0.7	2,807	<u>2.4</u>	2,671	<u>2.8</u>	1,744	<u>1.4</u>	2,948	<u>3.0</u>	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.

...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	<u>2.4</u>	899	<u>2.3</u>	521	<u>2.4</u>	951	<u>3.0</u>	805
YES, several times	0.5	3,785	<u>5.1</u>	3,486	<u>6.0</u>	1,867	<u>5.7</u>	3,691	<u>6.9</u>	2,927
NO	0.5	3,077	<u>2.0</u>	2,937	<u>2.2</u>	1,947	<u>1.5</u>	3,201	<u>3.0</u>	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.

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.

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.

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

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?							
	NO		YES, ONE TIME		YES, SEVERAL TIMES		TOTAL	
	N	%	N	%	N	%	N	%
Hereditary hemorrhagic telangiectasia	<u>265</u>	<u>58%</u>	<u>61</u>	<u>13%</u>	<u>132</u>	<u>29%</u>	458	100%
Hypermobile Ehlers-Danlos syndrome	<u>35</u>	<u>11%</u>	<u>31</u>	<u>10%</u>	<u>251</u>	<u>79%</u>	317	100%
Sarcoidosis	<u>36</u>	<u>21%</u>	<u>51</u>	<u>30%</u>	83	49%	170	100%
Classical Ehlers-Danlos syndrome	<u>18</u>	<u>13%</u>	<u>16</u>	<u>12%</u>	<u>103</u>	<u>75%</u>	137	100%
Williams syndrome	<u>76</u>	<u>56%</u>	24	18%	<u>36</u>	<u>26%</u>	136	100%
Cystic fibrosis	<u>67</u>	<u>52%</u>	19	15%	<u>42</u>	<u>33%</u>	128	100%
Myasthenia gravis	38	32%	<u>37</u>	<u>31%</u>	45	38%	120	100%
Systemic sclerosis	44	41%	25	23%	38	36%	107	100%
Tuberous sclerosis complex	<u>63</u>	<u>64%</u>	16	16%	<u>19</u>	<u>19%</u>	98	100%
Neurofibromatosis type 1	<u>58</u>	<u>63%</u>	14	15%	<u>20</u>	<u>22%</u>	92	100%
Interstitial cystitis	<u>9</u>	<u>12%</u>	16	22%	<u>49</u>	<u>66%</u>	74	100%
Addison disease	25	34%	17	23%	31	42%	73	100%
22q11.2 deletion syndrome	<u>37</u>	<u>54%</u>	11	16%	<u>20</u>	<u>29%</u>	68	100%
Chronic inflammatory demyelinating polyneuropathy	24	37%	18	28%	23	35%	65	100%
Perineural cyst	<u>9</u>	<u>14%</u>	7	11%	<u>47</u>	<u>75%</u>	63	100%
Acute inflammatory demyelinating polyradiculoneuropathy	23	37%	<u>22</u>	<u>35%</u>	<u>17</u>	<u>27%</u>	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	<u>29</u>	<u>59%</u>	10	20%	<u>10</u>	<u>20%</u>	49	100%
Behçet disease	<u>3</u>	<u>6%</u>	10	21%	<u>34</u>	<u>72%</u>	47	100%
Primary sclerosing cholangitis	<u>25</u>	<u>54%</u>	10	22%	<u>11</u>	<u>24%</u>	46	100%

Under-represented elements Over-represented elements

The relationship is very significant. p-value= < 0,01 ; Chi2= 4,655.5 ; dof= 3,350.

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%
Polycystic disease	12%	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.

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	<u>1,008</u>	<u>42%</u>	418	17%	981	41%	2,407	100%
1-9 / 100 000	778	39%	<u>409</u>	<u>20%</u>	812	41%	1,999	100%
1-9 / 1 000 000	166	36%	95	21%	198	43%	459	100%
<1 / 1 000 000	335	39%	<u>124</u>	<u>14%</u>	<u>397</u>	<u>46%</u>	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	<u>256</u>	<u>11%</u>	<u>1,184</u>	<u>49%</u>	<u>967</u>	<u>40%</u>	2,407	100%
1-9 / 100 000	<u>260</u>	<u>13%</u>	<u>845</u>	<u>42%</u>	<u>894</u>	<u>45%</u>	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 / 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.

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%	<u>431</u>	<u>50%</u>	222	26%	856	100%
TOTAL	1,473	26%	2,665	47%	1,592	28%	5,721	

■ Under-represented elements ■ Over-represented elements

Cross: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / ...wrongly attributed to another physical disease?

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	...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	1
8-11 body parts	229	24%	135	14%	587	62%	951	1
12-15 body parts	43	15%	30	10%	213	74%	286	1

Under-represented elements Over-represented elements

The relationship is very significant. p-value= < 0,01 ; Chi2= 505.4 ; dof= 8.

Cross: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / ...neglected, not taken seriously and/or considered as psychological?

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	...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	1
4-7 body parts	385	12%	1,592	52%	1,104	36%	3,081	1
8-11 body parts	79	8%	652	69%	220	23%	951	1
12-15 body parts	17	6%	231	81%	38	13%	286	1

Under-represented elements Over-represented elements

The relationship is very significant. p-value= < 0,01 ; Chi2= 533.1 ; dof= 8.

Cross: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / 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.

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	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.

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.

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	

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	<u>2,772</u>	<u>46%</u>	705	12%	<u>2,521</u>	<u>42%</u>	5,998	100%
No	<u>1,509</u>	<u>34%</u>	527	12%	<u>2,379</u>	<u>54%</u>	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%	<u>2,613</u>	<u>44%</u>	<u>1,873</u>	<u>31%</u>	5,998	100%
No	1,157	26%	<u>2,359</u>	<u>53%</u>	<u>899</u>	<u>20%</u>	4,415	100%
TOTAL	2,669	26%	4,972	48%	2,772	27%	10,413	

Under-represented elements Over-represented elements

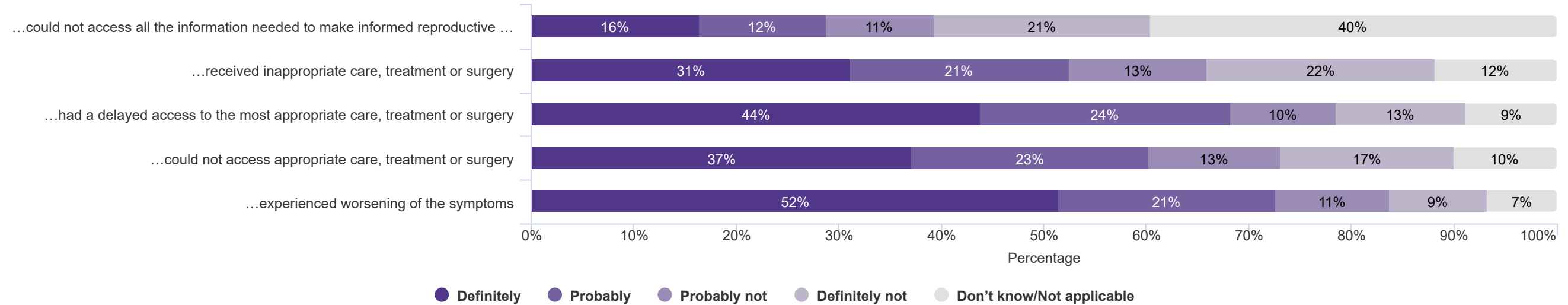
The relationship is very significant. *p*-value= < 0,01 ; Chi2= 165.6 ; dof= 2.

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...



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	

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	

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	

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	

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	

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											
	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											
	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											
	DEFINITELY		PROBABLY		PROBABLY NOT		DEFINITELY NOT		DON'T KNOW/NOT APPLICABLE		TOTAL	
	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											
	DEFINITELY		PROBABLY		PROBABLY NOT		DEFINITELY NOT		DON'T KNOW/NOT APPLICABLE		TOTAL	
	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.

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											
	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											
	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	

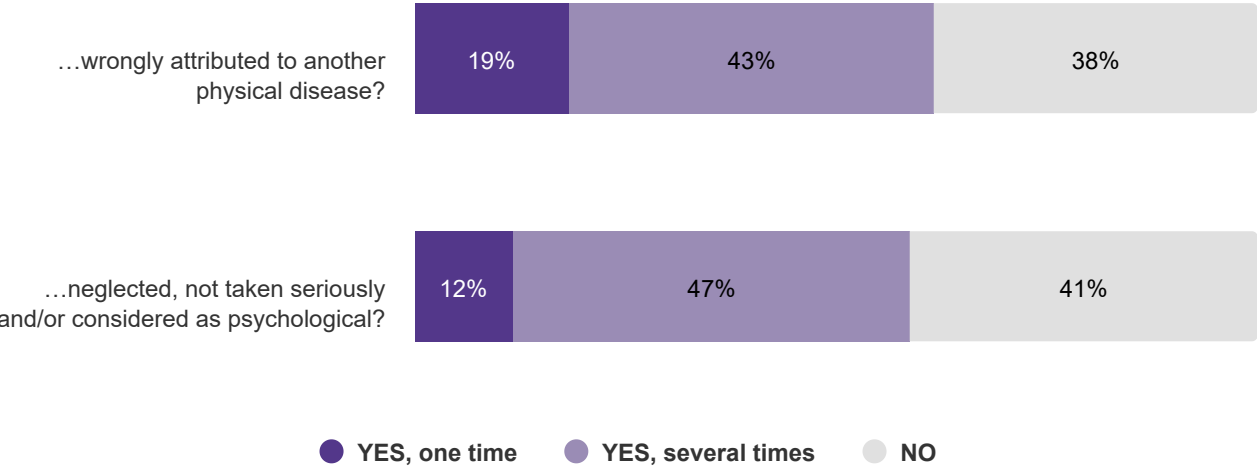
Under-represented elements Over-represented elements

The relationship is very significant. *p-value*= < 0,01 ; *Chi2*= 440.8 ; *dof*= 4.

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

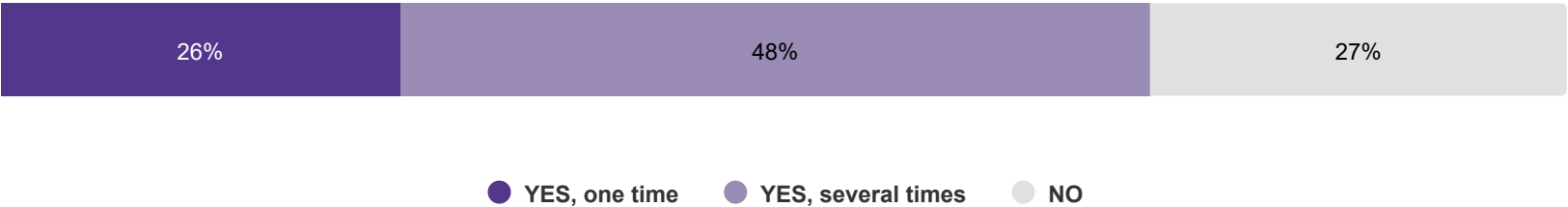
Did it ever happen that the symptoms of the rare disease were...



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.



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.

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

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

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?

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	
<div><div></div> Under-represented elements</div> <div><div></div> Over-represented elements</div>								
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 PROFESSIONALS 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	
<div><div></div> Under-represented elements</div> <div><div></div> Over-represented elements</div>								
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	
<div><div></div> Under-represented elements</div> <div><div></div> Over-represented elements</div>								
The relationship is very significant. p-value= < 0,01 ; Chi2= 797.1 ; dof= 4.								

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.

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	

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.

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.

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.

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.							
	YES, ONE TIME		YES, SEVERAL TIMES		NO		TOTAL	
	N	%	N	%	N	%	N	%
Yes	219	26%	<u>397</u>	<u>48%</u>	<u>215</u>	<u>26%</u>	831	100%
No	1,171	26%	1,967	44%	<u>1,377</u>	<u>30%</u>	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.

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.							
	YES, ONE TIME		YES, SEVERAL TIMES		NO		TOTAL	
	N	%	N	%	N	%	N	%
Yes	337	24%	<u>768</u>	<u>55%</u>	<u>298</u>	<u>21%</u>	1,403	100%
No	2,036	26%	<u>3,780</u>	<u>48%</u>	<u>2,092</u>	<u>26%</u>	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%	<u>1,086</u>	<u>52%</u>	<u>468</u>	<u>22%</u>	2,083	100%
No	2,095	26%	<u>3,856</u>	<u>47%</u>	<u>2,243</u>	<u>27%</u>	8,194	100%
Don't know	59	28%	<u>76</u>	<u>36%</u>	<u>74</u>	<u>35%</u>	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.

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.

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%	<u>2,465</u>	<u>46%</u>	<u>1,472</u>	<u>28%</u>	5,326	100%
YES, through online communities	1,264	25%	<u>2,602</u>	<u>52%</u>	<u>1,126</u>	<u>23%</u>	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%	<u>215</u>	<u>39%</u>	<u>190</u>	<u>35%</u>	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.

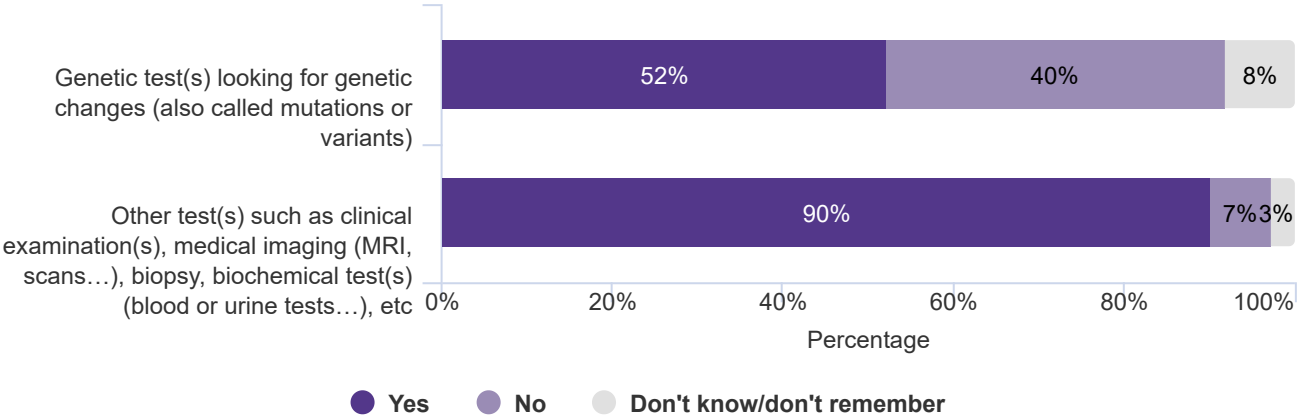
Chapter 11.

Diagnostic tests conducted (declarative)

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?



Cross: Genetic diseases / Genetic test(s) looking for genetic changes (also called mutations or variants)

GENETIC 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	%
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

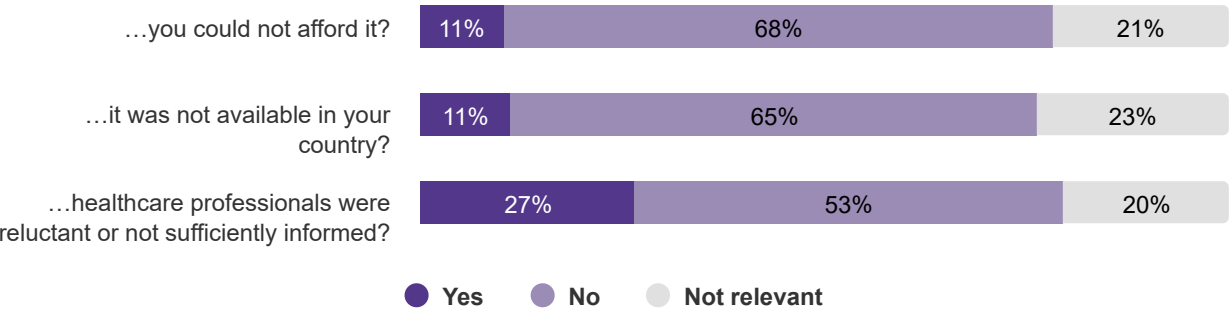
The relationship is very significant. $p\text{-value} = < 0,01$; $\text{Chi}^2 = 1,811.7$; $\text{dof} = 2$.

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

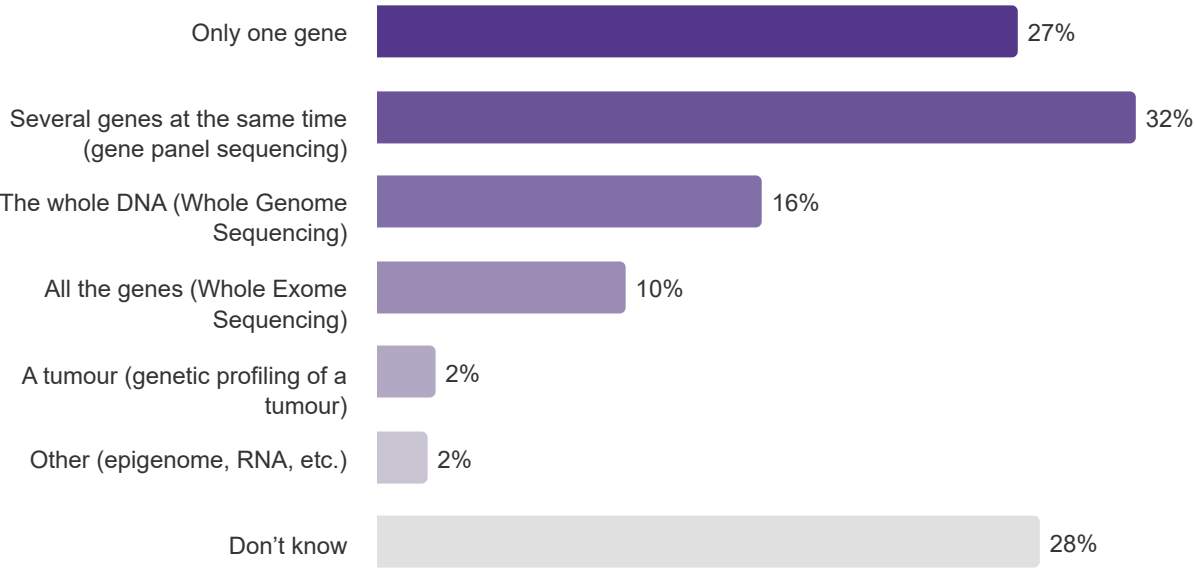
Have you ever needed a genetic test but could not access it because...



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 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

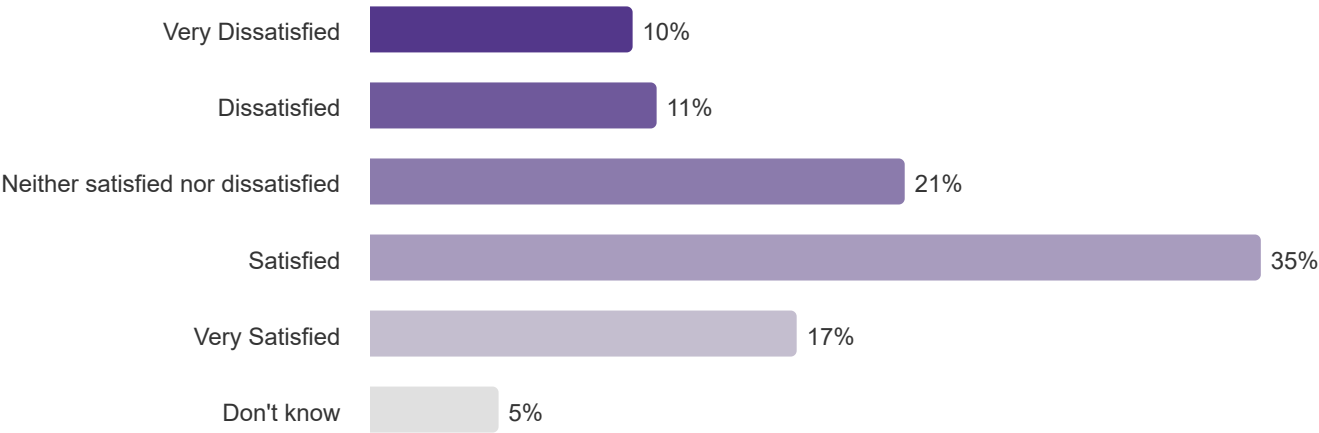
Did you ever request a private company or laboratory to conduct genetic testing to diagnose the disease?



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?

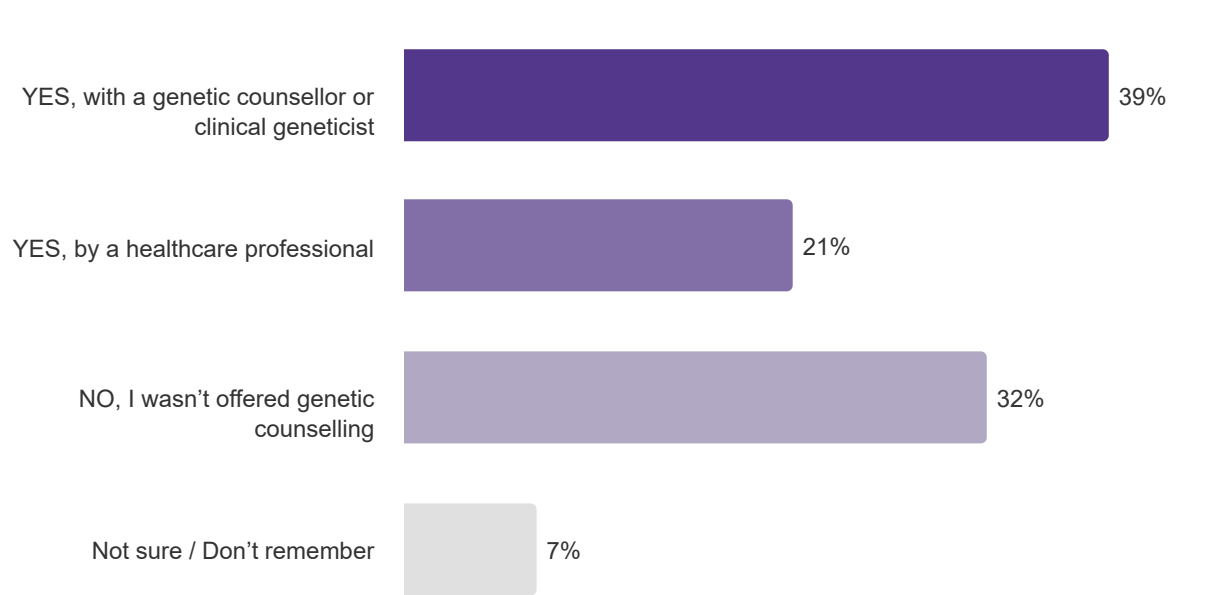


Only respondents who had genetic tests

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)?



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	<u>5.6</u>	3,565
No	0.2	3,263	3.4	3,138	3.7	1,564	3.6	3,216	<u>3.9</u>	2,506
Don't know/don't remember	0.5	534	<u>2.1</u>	529	<u>2.2</u>	320	<u>2.5</u>	531	<u>2.9</u>	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	<u>5.3</u>	705	<u>6.1</u>	334	<u>5.6</u>	742	<u>7.0</u>	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	<u>3.1</u>	1,589	3.6	853	3.6	1,659	<u>4.1</u>	1,355

Under-represented elements Over-represented elements

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

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.

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.

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.

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	

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	

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	

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	

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.

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?

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%	<u>183</u>	<u>6%</u>	3,113	100%
Male	182	10%	189	10%	375	21%	<u>658</u>	<u>37%</u>	<u>333</u>	<u>18%</u>	<u>64</u>	<u>4%</u>	1,801	100%
Other	6	10%	7	11%	18	29%	16	26%	8	13%	<u>7</u>	<u>11%</u>	62	100%
TOTAL	517	10%	576	12%	1,049	21%	1,730	35%	850	17%	254	5%	4,976	

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	

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	

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?

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	

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?							
	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?							
	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.

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	

Under-represented elements Over-represented elements

The relationship is not significant. p-value= 0.3 ; Chi2= 7.3 ; dof= 6.

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	<u>41</u>	<u>16%</u>	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%	<u>71</u>	<u>27%</u>	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	<u>711</u>	<u>41%</u>	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%	<u>2,899</u>	<u>41%</u>	<u>525</u>	<u>7%</u>	7,125	100%
I am part of an ethnic minority in the country where I live	<u>269</u>	<u>58%</u>	<u>147</u>	<u>32%</u>	<u>49</u>	<u>11%</u>	465	100%
Other, specify...	169	50%	131	39%	<u>37</u>	<u>11%</u>	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%	<u>15</u>	<u>4%</u>	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.

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...

HOW WOULD YOU BEST DESCRIBE YOURSELF?	TO YOUR KNOWLEDGE, THE GENETIC TEST(S) THAT WERE CONDUCTED TARGETED...															
			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.

Cross: How would you best describe yourself? / Did you ever request a private company or laboratory to conduct genetic testing to diagnose the disease?

HOW WOULD YOU BEST DESCRIBE YOURSELF?	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	%
I belong to the ethnic majority in the country where I live	401	11%	185	5%	3,115	84%	3,701	100%
I am part of an ethnic minority in the country where I live	43	16%	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?

HOW WOULD YOU BEST DESCRIBE YOURSELF?	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	%
I belong to the ethnic majority in the country where I live	374	10%	431	12%	776	21%	1,295	35%	657	18%	168	5%	3,701	100%
I am part of an ethnic minority in the country where I live	33	12%	29	11%	65	24%	92	34%	32	12%	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)?

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)?									
	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	%
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)

TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE	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	%
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.

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...				...YOU COULD NOT AFFORD IT?				
TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE	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.

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.*

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%	<u>279</u>	<u>24%</u>	432	37%	<u>161</u>	<u>14%</u>	53	5%	1,158	100%
Group B ('Western Europe')	<u>330</u>	<u>12%</u>	<u>332</u>	<u>12%</u>	577	21%	917	34%	<u>416</u>	<u>15%</u>	130	5%	2,702	100%
Group C ('Northern Europe')	<u>118</u>	<u>8%</u>	149	10%	<u>270</u>	<u>18%</u>	526	36%	<u>320</u>	<u>22%</u>	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')	<u>410</u>	<u>35%</u>	229	20%	<u>447</u>	<u>39%</u>	72	6%	1,158	100%
Group B ('Western Europe')	1,049	39%	<u>629</u>	<u>23%</u>	<u>817</u>	<u>30%</u>	207	8%	2,702	100%
Group C ('Northern Europe')	<u>617</u>	<u>42%</u>	<u>283</u>	<u>19%</u>	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.

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	

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	

Under-represented elements Over-represented elements

The relationship is not significant. *p-value= 0.3 ; Chi2= 4.7 ; dof= 4.*

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.

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	

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	

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)?									
	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	%
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	

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)	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	%
Hereditary hemorrhagic telangiectasia	115	31%	131	36%	45	12%	<u>7</u>	<u>2%</u>	<u>2</u>	<u>1%</u>	<u>1</u>	<u>0%</u>	92	25%	369	
Hypermobile Ehlers-Danlos syndrome	<u>15</u>	<u>16%</u>	<u>51</u>	<u>54%</u>	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	<u>6</u>	<u>9%</u>	<u>36</u>	<u>56%</u>	7	11%	3	5%	1	2%	2	3%	16	25%	64	
Williams syndrome	<u>49</u>	<u>37%</u>	39	30%	17	13%	<u>5</u>	<u>4%</u>	1	1%	0	0%	30	23%	131	
Cystic fibrosis	<u>47</u>	<u>39%</u>	42	35%	13	11%	6	5%	0	0%	0	0%	<u>22</u>	<u>18%</u>	121	
Myasthenia gravis	<u>1</u>	<u>6%</u>	8	47%	2	12%	0	0%	<u>2</u>	<u>12%</u>	<u>3</u>	<u>18%</u>	5	29%	17	
Systemic sclerosis	0	0%	0	0%	1	25%	0	0%	0	0%	0	0%	<u>3</u>	<u>75%</u>	4	
Tuberous sclerosis complex	17	19%	33	37%	12	13%	<u>3</u>	<u>3%</u>	1	1%	4	4%	26	29%	89	
Neurofibromatosis type 1	20	32%	14	22%	12	19%	<u>1</u>	<u>2%</u>	<u>6</u>	<u>10%</u>	2	3%	20	32%	63	
Interstitial cystitis	0	0%	0	0%	1	25%	0	0%	<u>1</u>	<u>25%</u>	<u>1</u>	<u>25%</u>	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%	<u>8</u>	<u>50%</u>	16	
Perineural cyst	1	17%	4	67%	2	33%	0	0%	<u>1</u>	<u>17%</u>	0	0%	2	33%	6	
Acute inflammatory demyelinating polyradiculoneuropathy	0	0%	0	0%	0	0%	0	0%	0	0%	0	0%	<u>3</u>	<u>100%</u>	3	
Rett syndrome	<u>27</u>	<u>48%</u>	19	34%	10	18%	<u>10</u>	<u>18%</u>	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...

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...															
	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)	<u>1,346</u>	<u>27%</u>	<u>1,510</u>	<u>31%</u>	<u>765</u>	<u>16%</u>	<u>458</u>	<u>9%</u>	121	2%	<u>97</u>	<u>2%</u>	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	<u>49</u>	<u>21%</u>	<u>93</u>	<u>39%</u>	31	13%	30	13%	5	2%	<u>10</u>	<u>4%</u>	75	31%	239	
I only have PARTIAL information on the name of the rare disease or the gene involved or the type of disease	<u>30</u>	<u>18%</u>	53	32%	<u>36</u>	<u>22%</u>	<u>31</u>	<u>19%</u>	6	4%	4	2%	48	29%	164	
I know that the disease is rare but the name or the cause have NOT BEEN IDENTIFIED	<u>33</u>	<u>18%</u>	<u>73</u>	<u>40%</u>	<u>47</u>	<u>26%</u>	<u>46</u>	<u>25%</u>	3	2%	5	3%	41	23%	181	
Other, specify...	2	40%	2	40%	1	20%	<u>2</u>	<u>40%</u>	0	0%	<u>1</u>	<u>20%</u>	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.*

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	<u>1,195</u>	<u>50%</u>	<u>1,026</u>	<u>43%</u>	186	8%	2,407	100%
1-9 / 100 000	1,111	56%	751	38%	137	7%	1,999	100%
1-9 / 1 000 000	<u>300</u>	<u>65%</u>	<u>132</u>	<u>29%</u>	27	6%	459	100%
<1 / 1 000 000	<u>547</u>	<u>64%</u>	<u>258</u>	<u>30%</u>	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	<u>3,458</u>	<u>58%</u>	<u>2,038</u>	<u>34%</u>	<u>502</u>	<u>8%</u>	5,998	100%
No	<u>1,998</u>	<u>45%</u>	<u>2,097</u>	<u>47%</u>	<u>320</u>	<u>7%</u>	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.*

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?

I, OR THE PERSON I CARE FOR, HAVE BEEN REFERRED TO A HOSPITAL UNIT SPECIALISED IN THE RARE DISEASE OR GROUP OF RARE DISEASES	Have you ever needed a genetic test but could not access it because... ...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.*

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.*

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.

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...	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...	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.

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	

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	

Under-represented elements Over-represented elements

The relationship is very significant. *p-value= < 0,01 ; Chi2= 288.0 ; dof= 18.*

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.

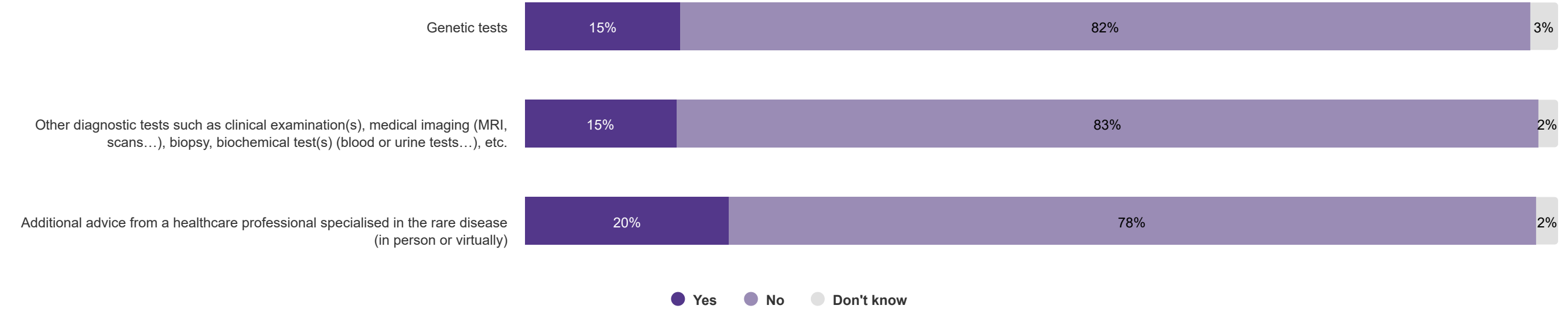
Chapter 12.

Cross-border healthcare

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	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.

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	<u>4.3</u>	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	<u>1.5</u>	106	<u>2.5</u>	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.

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	<u>0.1</u>	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.

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.*

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.

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 IMAGING (MRI, SCANS...), BIOPSY, BIOCHEMICAL TEST(S) (BLOOD OR URINE TESTS...), ETC.							
	YES		NO		DON'T KNOW		TOTAL	
	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	

[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	

[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 svndrome	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	<u>32</u>	<u>8%</u>	<u>368</u>	<u>90%</u>	8	2%	408	100%
Hypermobile Ehlers-Danlos syndrome	46	16%	235	83%	2	1%	283	100%
Sarcoidosis	<u>14</u>	<u>8%</u>	<u>154</u>	<u>91%</u>	1	1%	169	100%
Classical Ehlers-Danlos syndrome	14	11%	110	89%	0	0%	124	100%
Williams syndrome	<u>5</u>	<u>6%</u>	72	90%	3	4%	80	100%
Cystic fibrosis	19	17%	<u>89</u>	<u>77%</u>	<u>7</u>	<u>6%</u>	115	100%
Myasthenia gravis	<u>33</u>	<u>28%</u>	<u>83</u>	<u>70%</u>	2	2%	118	100%
Systemic sclerosis	<u>6</u>	<u>6%</u>	<u>100</u>	<u>94%</u>	0	0%	106	100%
Tuberous sclerosis complex	<u>5</u>	<u>5%</u>	<u>91</u>	<u>95%</u>	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%	<u>32</u>	<u>97%</u>	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.

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	<u>7</u>	<u>7%</u>	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	<u>19</u>	<u>7%</u>	<u>223</u>	<u>88%</u>	<u>12</u>	<u>5%</u>	254	100%
Circulatory system diseases	<u>82</u>	<u>9%</u>	<u>823</u>	<u>89%</u>	20	2%	925	100%
Clinical sign	0	0%	0	0%	0	0%	0	100%
Developmental anomalies during embryogenesis	<u>272</u>	<u>12%</u>	<u>1,942</u>	<u>86%</u>	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	<u>584</u>	<u>15%</u>	<u>3,190</u>	<u>83%</u>	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	<u>66</u>	<u>10%</u>	<u>585</u>	<u>87%</u>	21	3%	672	100%
Immunological diseases	<u>42</u>	<u>21%</u>	<u>158</u>	<u>77%</u>	4	2%	204	100%
Inborn errors of metabolism	<u>139</u>	<u>22%</u>	<u>469</u>	<u>75%</u>	18	3%	626	100%
Infectious diseases	0	0%	1	100%	0	0%	1	100%
Infertility	41	13%	250	82%	<u>13</u>	<u>4%</u>	304	100%

Under-represented elements Over-represented elements

The relationship is very significant. p-value= < 0,01 ; Chi2= 201.0 ; dof= 68.

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.

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.*

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:	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	%
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.

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.

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.*

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
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	
<div><div></div> Under-represented elements</div> <div><div></div> Over-represented elements</div>								
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
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	
<div><div></div> Under-represented elements</div> <div><div></div> Over-represented elements</div>								
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	
<div><div></div> Under-represented elements</div> <div><div></div> Over-represented elements</div>								
The relationship is very significant. p-value= < 0,01 ; Chi2= 58.3 ; dof= 4.								

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.

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.

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.*

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.

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.

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.

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.*

Chapter 13.

Support

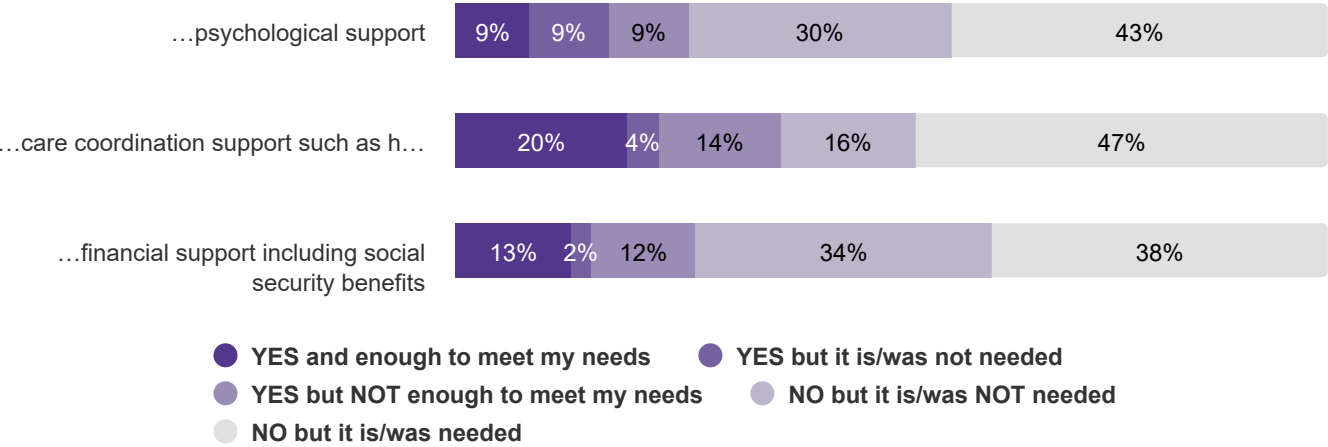
During your search for a diagnosis, were you offered...

	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

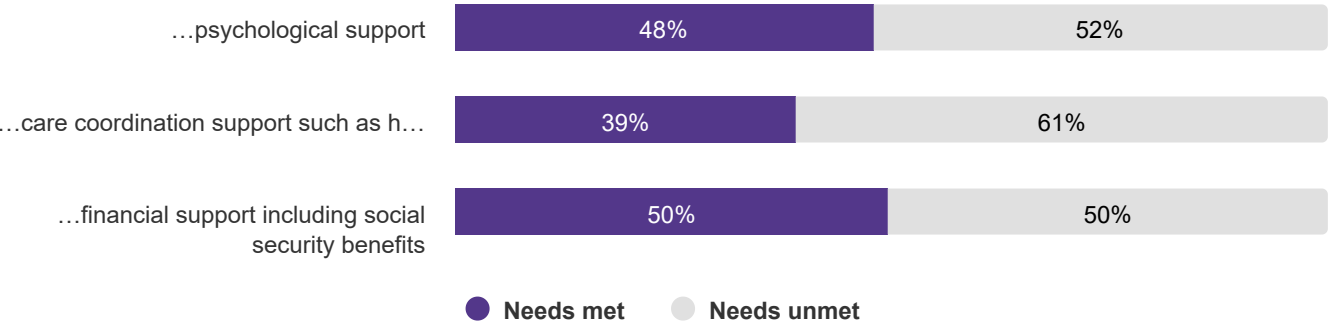
Needs met: YES and enough to meet my needs + YES but it is/was not needed + NO but it is/was NOT needed.

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...



During your search for a diagnosis, were you offered...



Time from first symptoms to the different steps of the diagnosis journey if psychological support needs were met during the search for a diagnosis

...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	<u>-0.3</u>	685	<u>2.1</u>	670	<u>2.0</u>	471	<u>2.0</u>	695	<u>2.5</u>	584
YES but it is/was not needed	0.3	688	3.1	664	3.6	461	3.4	717	<u>4.0</u>	615
YES but NOT enough to meet my needs	0.5	739	3.5	674	4.4	425	<u>4.5</u>	724	5.1	555
NO but it is/was NOT needed	0.8	2,255	3.9	2,166	3.9	1,280	<u>3.1</u>	2,330	4.9	1,965
NO but it is/was needed	0.5	3,453	3.6	3,148	4.3	1,698	<u>4.1</u>	3,377	<u>5.2</u>	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.

Time from first symptoms to the different steps of the diagnosis journey if care coordination needs were met during the search for 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.	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	<u>2.3</u>	1,535	<u>2.4</u>	1,117	<u>1.7</u>	1,619	<u>2.8</u>	1,425
YES but it is/was not needed	0.5	258	<u>2.2</u>	248	<u>2.0</u>	167	<u>2.2</u>	268	<u>3.2</u>	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	<u>2.7</u>	1,149	4.4	978
NO but it is/was needed	0.5	3,793	<u>4.2</u>	3,443	<u>5.2</u>	1,727	<u>4.8</u>	3,707	<u>5.9</u>	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.

Time from first symptoms to the different steps of the diagnosis journey if financial support needs were met during the search for a diagnosis

...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.

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.

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%	<u>89</u>	<u>20%</u>	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%	<u>497</u>	<u>16%</u>	1,400	46%	3,022	100%
24 y.o. or above	<u>574</u>	<u>18%</u>	104	3%	442	14%	474	15%	<u>1,551</u>	<u>49%</u>	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%	<u>81</u>	<u>18%</u>	<u>118</u>	<u>26%</u>	178	39%	451	100%
between 16 and 19 y.o.	299	12%	<u>43</u>	<u>2%</u>	284	12%	831	34%	<u>1,003</u>	<u>41%</u>	2,460	100%
between 20 and 23 y.o.	398	13%	75	2%	349	12%	<u>1,103</u>	<u>37%</u>	<u>1,079</u>	<u>36%</u>	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.

Cross: How would you best describe yourself? / ...psychological support

HOW WOULD YOU BEST DESCRIBE YOURSELF?	...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	%
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.

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.											
	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 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											
	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 belong to the ethnic majority in the country where I live	898	13%	155	2%	857	12%	<u>2,494</u>	<u>35%</u>	<u>2,685</u>	<u>38%</u>	7,089	100%
I am part of an ethnic minority in the country where I live	60	13%	14	3%	68	15%	<u>118</u>	<u>25%</u>	<u>204</u>	<u>44%</u>	464	100%
Other, specify...	45	13%	8	2%	42	12%	<u>96</u>	<u>28%</u>	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											
	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')	170	9%	180	10%	144	8%	<u>481</u>	<u>27%</u>	<u>819</u>	<u>46%</u>	1,794	100%
Group B ('Western Europe')	<u>399</u>	<u>8%</u>	<u>426</u>	<u>8%</u>	<u>429</u>	<u>8%</u>	1,501	29%	<u>2,350</u>	<u>46%</u>	5,105	100%
Group C ('Northern Europe')	<u>313</u>	<u>10%</u>	306	9%	<u>352</u>	<u>11%</u>	<u>1,097</u>	<u>34%</u>	<u>1,205</u>	<u>37%</u>	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.

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	<u>20</u>	<u>4%</u>	<u>28</u>	<u>6%</u>	<u>20</u>	<u>4%</u>	<u>250</u>	<u>55%</u>	<u>140</u>	<u>31%</u>	458	100%
Hypermobile Ehlers-Danlos syndrome	28	9%	<u>40</u>	<u>13%</u>	<u>52</u>	<u>16%</u>	<u>69</u>	<u>22%</u>	128	40%	317	100%
Sarcoidosis	11	6%	10	6%	<u>5</u>	<u>3%</u>	<u>71</u>	<u>42%</u>	73	43%	170	100%
Classical Ehlers-Danlos syndrome	7	5%	16	12%	<u>21</u>	<u>15%</u>	36	26%	57	42%	137	100%
Williams syndrome	15	11%	7	5%	13	10%	<u>28</u>	<u>21%</u>	<u>73</u>	<u>54%</u>	136	100%
Cystic fibrosis	16	13%	<u>21</u>	<u>16%</u>	14	11%	<u>25</u>	<u>20%</u>	52	41%	128	100%
Myasthenia gravis	10	8%	<u>3</u>	<u>3%</u>	6	5%	40	33%	61	51%	120	100%
Systemic sclerosis	6	6%	8	7%	7	7%	<u>43</u>	<u>40%</u>	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%	<u>45</u>	<u>61%</u>	74	100%
Addison disease	3	4%	11	15%	<u>1</u>	<u>1%</u>	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%	<u>18</u>	<u>28%</u>	65	100%
Perineural cyst	7	11%	9	14%	5	8%	16	25%	26	41%	63	100%
Acute inflammatory demyelinating polyradiculoneuropathy	6	10%	<u>11</u>	<u>18%</u>	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	32%	10	20%	40	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.

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.

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											
	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	25	10%	27	11%	<u>35</u>	<u>15%</u>	<u>55</u>	<u>23%</u>	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%	<u>229</u>	<u>35%</u>	270	41%	660	100%
Cardiac malformations	<u>36</u>	<u>12%</u>	<u>17</u>	<u>6%</u>	32	11%	<u>64</u>	<u>22%</u>	<u>146</u>	<u>49%</u>	295	100%
Circulatory system diseases	<u>87</u>	<u>6%</u>	<u>89</u>	<u>7%</u>	<u>92</u>	<u>7%</u>	<u>533</u>	<u>39%</u>	<u>550</u>	<u>41%</u>	1,351	100%
Clinical sign	0	0%	0	0%	0	0%	0	0%	0	0%	0	100%
Developmental anomalies during embryogenesis	<u>250</u>	<u>7%</u>	<u>269</u>	<u>8%</u>	<u>318</u>	<u>10%</u>	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%	<u>44</u>	<u>14%</u>	34	11%	<u>67</u>	<u>22%</u>	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%	<u>70</u>	<u>25%</u>	137	48%	284	100%
Hematological diseases	40	10%	36	9%	<u>48</u>	<u>12%</u>	110	27%	178	43%	412	100%
Hepatic diseases	<u>58</u>	<u>7%</u>	78	9%	<u>53</u>	<u>6%</u>	<u>408</u>	<u>46%</u>	<u>294</u>	<u>33%</u>	891	100%
Immunological diseases	28	10%	21	7%	27	9%	77	27%	133	47%	286	100%
Inborn errors of metabolism	<u>79</u>	<u>10%</u>	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%	<u>2</u>	<u>67%</u>	0	0%	3	100%
Bone diseases	165	21%	30	4%	<u>127</u>	<u>16%</u>	114	14%	363	45%	799	100%
Cardiac diseases	<u>168</u>	<u>25%</u>	31	5%	94	14%	103	16%	<u>264</u>	<u>40%</u>	660	100%
Cardiac malformations	71	24%	11	4%	51	17%	37	13%	125	42%	295	100%
Circulatory system diseases	<u>315</u>	<u>23%</u>	44	3%	194	14%	<u>256</u>	<u>19%</u>	<u>542</u>	<u>40%</u>	1,351	100%
Clinical sign	0	0%	0	0%	0	0%	0	0%	0	0%	0	100%
Developmental anomalies during embryogenesis	642	19%	<u>93</u>	<u>3%</u>	461	14%	495	15%	<u>1,656</u>	<u>49%</u>	3,347	100%
Diseases due to toxic effects	0	0%	<u>1</u>	<u>33%</u>	0	0%	1	33%	1	33%	3	100%
Endocrine diseases	204	21%	36	4%	147	15%	150	15%	458	46%	995	100%
Gastroenterological diseases	<u>95</u>	<u>31%</u>	14	5%	44	14%	37	12%	<u>115</u>	<u>38%</u>	305	100%
Genetic diseases	1,100	20%	192	4%	<u>773</u>	<u>14%</u>	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	<u>256</u>	<u>29%</u>	33	4%	<u>100</u>	<u>11%</u>	<u>204</u>	<u>23%</u>	<u>298</u>	<u>33%</u>	891	100%
Immunological diseases	55	19%	10	3%	<u>57</u>	<u>20%</u>	43	15%	121	42%	286	100%
Inborn errors of metabolism	<u>197</u>	<u>25%</u>	<u>38</u>	<u>5%</u>	110	14%	108	14%	<u>321</u>	<u>41%</u>	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.

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%	0	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											
	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%	<u>773</u>	<u>9%</u>	<u>2,827</u>	<u>31%</u>	<u>3,823</u>	<u>42%</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	61	8%	66	9%	<u>89</u>	<u>12%</u>	<u>185</u>	<u>24%</u>	<u>359</u>	<u>47%</u>	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%	<u>54</u>	<u>18%</u>	<u>69</u>	<u>23%</u>	136	44%	306	100%
I know that the disease is rare but the name or the cause have NOT BEEN IDENTIFIED	<u>44</u>	<u>13%</u>	28	8%	35	10%	<u>80</u>	<u>23%</u>	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)	1,935	21%	348	4%	1,262	14%	1,439	16%	4,064	45%	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	62	8%	19	3%	97	13%	102	13%	480	63%	760	100%
I only have PARTIAL information on the name of the rare disease or the gene involved or the type of disease	35	11%	15	5%	45	15%	31	10%	180	59%	306	100%
I know that the disease is rare but the name or the cause have NOT BEEN IDENTIFIED	49	14%	8	2%	56	16%	48	14%	187	54%	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.*

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	...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	%
1-5 / 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	...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	%
1-5 / 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.

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: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / ...psychological support

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	...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: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / ...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.

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	...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	<u>1,316</u>	<u>22%</u>	<u>266</u>	<u>4%</u>	<u>764</u>	<u>13%</u>	<u>1,113</u>	<u>18%</u>	<u>2,644</u>	<u>43%</u>	6,103	100%
4-7 body parts	597	19%	<u>97</u>	<u>3%</u>	<u>486</u>	<u>16%</u>	<u>387</u>	<u>13%</u>	<u>1,514</u>	<u>49%</u>	3,081	100%
8-11 body parts	<u>136</u>	<u>14%</u>	<u>20</u>	<u>2%</u>	<u>156</u>	<u>16%</u>	<u>115</u>	<u>12%</u>	<u>524</u>	<u>55%</u>	951	100%
12-15 body parts	<u>30</u>	<u>10%</u>	<u>4</u>	<u>1%</u>	41	14%	<u>12</u>	<u>4%</u>	<u>199</u>	<u>70%</u>	286	100%
16 body parts or more	<u>4</u>	<u>6%</u>	4	6%	<u>16</u>	<u>25%</u>	<u>0</u>	<u>0%</u>	<u>41</u>	<u>63%</u>	65	100%

Under-represented elements Over-represented elements

The relationship is very significant. p-value= < 0,01 ; Chi2= 245.7 ; dof= 16.

Cross: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / ...financial support including social security benefits

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	...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.

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											
	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	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	

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.											
	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											
	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											
	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	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	

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.											
	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	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	

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	<u>430</u>	<u>15%</u>	<u>50</u>	<u>2%</u>	<u>503</u>	<u>17%</u>	<u>626</u>	<u>21%</u>	<u>1,304</u>	<u>45%</u>	2,913	100%
No	943	13%	<u>182</u>	<u>3%</u>	<u>681</u>	<u>9%</u>	<u>2,822</u>	<u>39%</u>	<u>2,559</u>	<u>36%</u>	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	<u>494</u>	<u>8%</u>	<u>501</u>	<u>8%</u>	<u>588</u>	<u>10%</u>	<u>1,661</u>	<u>28%</u>	<u>2,696</u>	<u>45%</u>	5,940	100%
No	<u>372</u>	<u>10%</u>	<u>391</u>	<u>10%</u>	<u>293</u>	<u>8%</u>	<u>1,265</u>	<u>33%</u>	<u>1,467</u>	<u>39%</u>	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.

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											
	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	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	

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											
	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	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	

Under-represented elements Over-represented elements

The relationship is very significant. p-value= < 0,01 ; Chi2= 112.6 ; dof= 8.

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.

Cross: How many different healthcare professionals did you consult (in person or virtually) while seeking a diagnosis? / ...psychological support

HOW MANY DIFFERENT HEALTHCARE PROFESSIONALS DID YOU CONSULT (IN PERSON OR VIRTUALLY) WHILE SEEKING A DIAGNOSIS?	...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	%
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.

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.											
	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.*

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	

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	

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.

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.

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%	<u>699</u>	<u>36%</u>	<u>696</u>	<u>36%</u>	1,932	100%
YES, several times	<u>448</u>	<u>10%</u>	<u>63</u>	<u>1%</u>	<u>582</u>	<u>13%</u>	<u>1,259</u>	<u>28%</u>	<u>2,129</u>	<u>48%</u>	4,481	100%
NO	<u>688</u>	<u>17%</u>	<u>132</u>	<u>3%</u>	<u>430</u>	<u>11%</u>	<u>1,586</u>	<u>40%</u>	<u>1,164</u>	<u>29%</u>	4,000	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. $p\text{-value} = < 0,01$; $\text{Chi}^2 = 400.0$; $\text{dof} = 8$.

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	<u>338</u>	<u>7%</u>	<u>356</u>	<u>7%</u>	<u>565</u>	<u>11%</u>	<u>1,135</u>	<u>23%</u>	<u>2,540</u>	<u>51%</u>	4,934	100%
NO	<u>471</u>	<u>11%</u>	<u>485</u>	<u>11%</u>	<u>259</u>	<u>6%</u>	<u>1,646</u>	<u>38%</u>	<u>1,445</u>	<u>34%</u>	4,306	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. $p\text{-value} = < 0,01$; $\text{Chi}^2 = 512.9$; $\text{dof} = 8$.

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.

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.

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.*

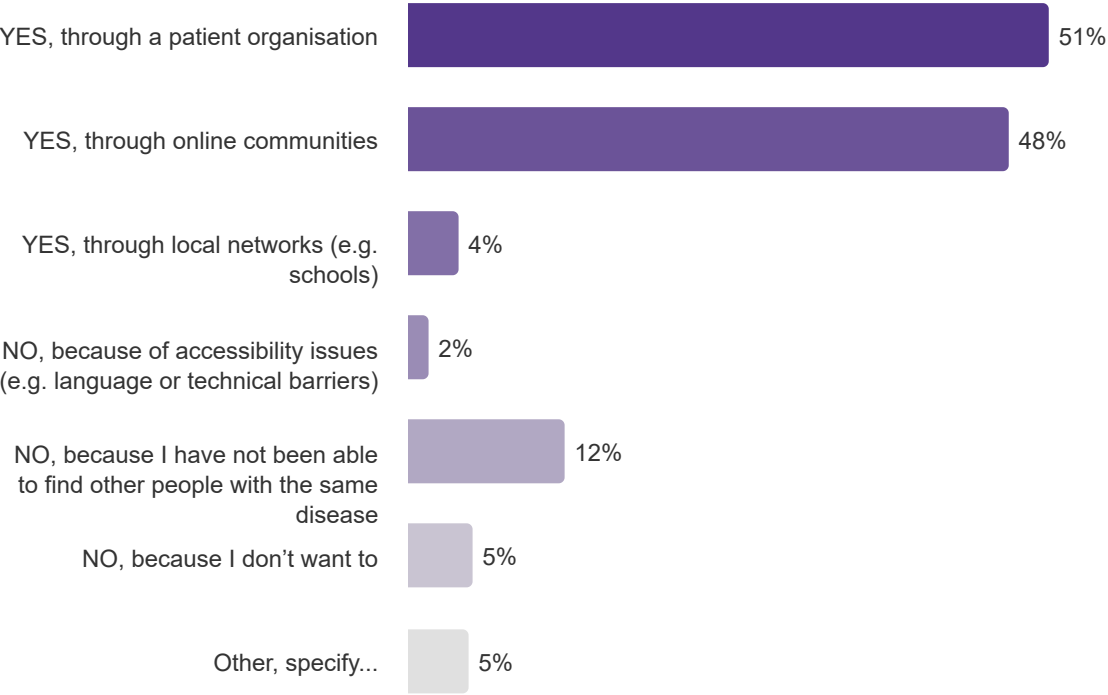
Chapter 14.

In touch with other
people living with the
same 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?

	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?



Respondents could choose several items

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%	<u>3,358</u>	<u>50%</u>	<u>240</u>	<u>4%</u>	<u>96</u>	<u>1%</u>	<u>791</u>	<u>12%</u>	356	5%	338	5%	6,772	
Former or recovering patient (e.g. cancer survivor)	<u>108</u>	<u>44%</u>	<u>94</u>	<u>38%</u>	13	5%	5	2%	39	16%	<u>35</u>	<u>14%</u>	11	4%	247	
Parent of a person living with a rare disease	1,560	51%	<u>1,416</u>	<u>46%</u>	<u>160</u>	<u>5%</u>	<u>74</u>	<u>2%</u>	410	13%	<u>121</u>	<u>4%</u>	136	4%	3,078	
Grandparent of a person living with a rare disease	22	55%	18	45%	<u>5</u>	<u>13%</u>	1	3%	5	13%	3	8%	0	0%	40	
Spouse of a person living with a rare disease	<u>81</u>	<u>44%</u>	<u>56</u>	<u>30%</u>	9	5%	<u>8</u>	<u>4%</u>	<u>37</u>	<u>20%</u>	<u>16</u>	<u>9%</u>	11	6%	186	
Uncle/aunt of a person living with a rare disease	15	65%	7	30%	<u>3</u>	<u>13%</u>	0	0%	3	13%	1	4%	0	0%	23	
Sibling of a person living with a rare disease	25	52%	<u>16</u>	<u>33%</u>	1	2%	2	4%	2	4%	<u>6</u>	<u>13%</u>	5	10%	48	
Other, specify...	<u>34</u>	<u>37%</u>	<u>27</u>	<u>29%</u>	5	5%	4	4%	<u>23</u>	<u>25%</u>	<u>9</u>	<u>10%</u>	<u>13</u>	<u>14%</u>	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	<u>1,135</u>	<u>56%</u>	998	49%	<u>113</u>	<u>6%</u>	41	2%	235	11%	<u>65</u>	<u>3%</u>	<u>84</u>	<u>4%</u>	2,045	
2 to less than 10 years old	478	52%	<u>419</u>	<u>45%</u>	45	5%	<u>30</u>	<u>3%</u>	121	13%	45	5%	50	5%	925	
10 to less than 20 years old	477	50%	<u>515</u>	<u>54%</u>	<u>52</u>	<u>5%</u>	11	1%	104	11%	49	5%	48	5%	952	
20 to less than 30 years old	<u>471</u>	<u>48%</u>	505	52%	36	4%	<u>7</u>	<u>1%</u>	116	12%	<u>66</u>	<u>7%</u>	48	5%	978	
30 to less than 50 years old	1,228	52%	1,167	50%	<u>76</u>	<u>3%</u>	30	1%	270	11%	101	4%	115	5%	2,353	
50 years old or more	547	49%	<u>487</u>	<u>44%</u>	<u>31</u>	<u>3%</u>	15	1%	149	13%	<u>66</u>	<u>6%</u>	<u>68</u>	<u>6%</u>	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.*

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.*

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.

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?																
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?															
	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.

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?														
	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...		TOT
	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.

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.

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	<u>1,376</u>	<u>57%</u>	1,196	50%	108	4%	27	1%	186	8%	124	5%	<u>150</u>	<u>6%</u>	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: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / 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?

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	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.

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.*

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%	<u>149</u>	<u>5%</u>	<u>82</u>	<u>3%</u>	379	13%	<u>108</u>	<u>4%</u>	141	5%	2,936	
No	3,694	51%	3,431	47%	<u>277</u>	<u>4%</u>	<u>93</u>	<u>1%</u>	874	12%	<u>428</u>	<u>6%</u>	358	5%	7,236	
Don't know	143	46%	143	46%	10	3%	<u>15</u>	<u>5%</u>	<u>57</u>	<u>18%</u>	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.

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%	<u>3,046</u>	<u>51%</u>	255	4%	115	2%	<u>667</u>	<u>11%</u>	<u>270</u>	<u>5%</u>	272	5%	5,940	
No	1,919	51%	<u>1,620</u>	<u>43%</u>	154	4%	62	2%	<u>524</u>	<u>14%</u>	<u>240</u>	<u>6%</u>	193	5%	3,788	
Don't know	<u>344</u>	<u>45%</u>	<u>326</u>	<u>43%</u>	27	4%	13	2%	<u>119</u>	<u>16%</u>	37	5%	<u>49</u>	<u>6%</u>	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.

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.

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%	<u>2,272</u>	<u>49%</u>	199	4%	77	2%	600	13%	<u>207</u>	<u>4%</u>	207	4%	4,648	
No	2,673	51%	<u>2,442</u>	<u>47%</u>	210	4%	95	2%	<u>617</u>	<u>12%</u>	<u>313</u>	<u>6%</u>	278	5%	5,251	
Don't know	<u>262</u>	<u>45%</u>	278	47%	27	5%	<u>18</u>	<u>3%</u>	<u>93</u>	<u>16%</u>	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.

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 ; *Chi*2= 8.1 ; dof= 6.

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.

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.

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.

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.*

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.*

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.

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	<u>2,898</u>	<u>53%</u>	2,576	47%	<u>254</u>	<u>5%</u>	<u>122</u>	<u>2%</u>	704	13%	<u>235</u>	<u>4%</u>	278	5%	5,490	
No	<u>2,063</u>	<u>49%</u>	<u>2,076</u>	<u>50%</u>	155	4%	<u>54</u>	<u>1%</u>	<u>475</u>	<u>11%</u>	<u>250</u>	<u>6%</u>	187	4%	4,171	
Don't know/don't remember	<u>365</u>	<u>44%</u>	<u>340</u>	<u>41%</u>	27	3%	14	2%	<u>131</u>	<u>16%</u>	<u>62</u>	<u>8%</u>	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...

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.

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	<u>697</u>	<u>59%</u>	<u>490</u>	<u>42%</u>	61	5%	23	2%	<u>116</u>	<u>10%</u>	57	5%	52	4%	1,179	
NO, I wasn't offered genetic counselling	<u>836</u>	<u>47%</u>	<u>891</u>	<u>50%</u>	78	4%	48	3%	<u>265</u>	<u>15%</u>	<u>60</u>	<u>3%</u>	89	5%	1,770	
Not sure / Don't remember	204	50%	181	45%	15	4%	7	2%	52	13%	<u>29</u>	<u>7%</u>	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.

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.

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.

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	<u>1,215</u>	<u>58%</u>	<u>870</u>	<u>42%</u>	99	5%	29	1%	<u>216</u>	<u>10%</u>	<u>135</u>	<u>6%</u>	97	5%	2,083	
YES but it is/was not needed	200	51%	171	44%	16	4%	5	1%	45	12%	<u>32</u>	<u>8%</u>	16	4%	391	
YES but NOT enough to meet my needs	<u>788</u>	<u>54%</u>	680	46%	63	4%	32	2%	183	13%	62	4%	73	5%	1,463	
NO but it is/was NOT needed	<u>770</u>	<u>47%</u>	<u>714</u>	<u>44%</u>	<u>51</u>	<u>3%</u>	<u>18</u>	<u>1%</u>	204	13%	<u>148</u>	<u>9%</u>	<u>100</u>	<u>6%</u>	1,627	
NO but it is/was needed	<u>2,353</u>	<u>48%</u>	<u>2,557</u>	<u>52%</u>	207	4%	<u>106</u>	<u>2%</u>	<u>662</u>	<u>13%</u>	<u>170</u>	<u>3%</u>	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.

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?							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...	
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.



Chapter 15.

Consequence of being diagnosed



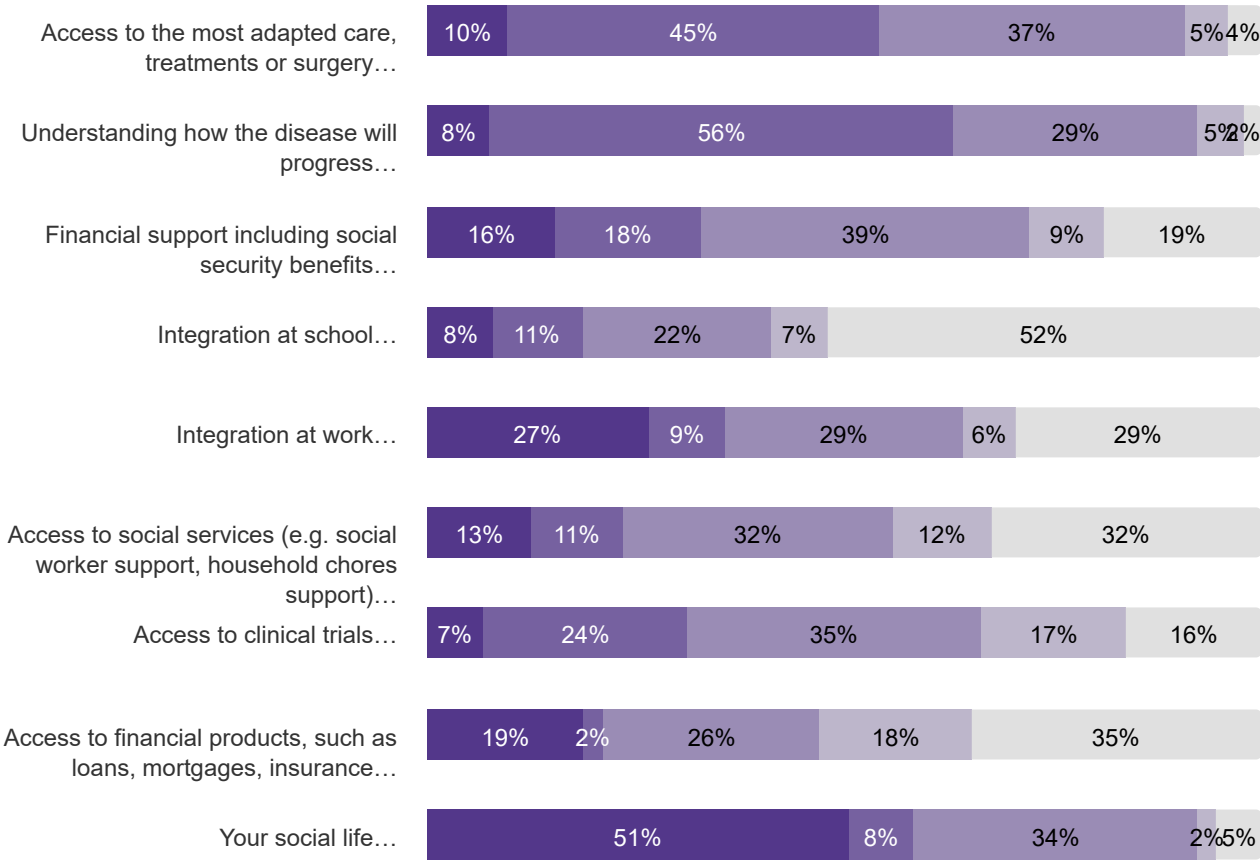
9. Consequences of diagnosis

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

Question 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	<u>3.9</u>	3,210	<u>4.3</u>	1,897	3.8	3,347	<u>5.2</u>	2,918
...has remained the same	0.5	2,526	<u>3.1</u>	2,293	3.4	1,353	3.3	2,659	4.4	2,285
Don't know	1.2	299	<u>2.4</u>	257	<u>2.3</u>	143	2.7	315	<u>3.0</u>	244
Not relevant	0.3	239	2.6	184	<u>2.5</u>	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.

Question 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.

Question 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.

Question 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.

Question 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	<u>4.5</u>	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	<u>2.7</u>	338	3.2	204	<u>2.1</u>	398	<u>3.4</u>	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.

Question 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.

Question 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	<u>0.1</u>	1,727	4.0	1,692	4.2	1,047	3.8	1,834	<u>5.6</u>	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	<u>4.0</u>	1,007
Not relevant	<u>1.1</u>	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.

Question 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	<u>4.7</u>	725	4.1	1,377	<u>5.7</u>	1,179
...has improved	0.1	157	<u>2.3</u>	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	<u>5.3</u>	1,625
Don't know	0.5	1,239	3.2	1,152	3.2	723	<u>3.0</u>	1,316	<u>4.0</u>	1,102
Not relevant	0.4	2,322	3.3	2,248	3.3	1,327	3.3	2,486	<u>4.2</u>	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.

Question 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.

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.

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.

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	

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	

Under-represented elements Over-represented elements

The relationship is very significant. p-value= < 0,01 ; Chi2= 45.4 ; dof= 12.

The relationship is very significant. $p\text{-value} = < 0,01$; $\text{Chi}^2 = 52.6$; $\text{dof} = 12$.

The relationship is significant. $p\text{-value} = 0.0$; $\text{Chi}^2 = 23.6$; $\text{dof} = 12$.

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	<u>54</u>	<u>13%</u>	<u>62</u>	<u>15%</u>	91	22%	<u>40</u>	<u>10%</u>	<u>166</u>	<u>40%</u>	413	100%
between 16 and 19 y.o.	174	8%	<u>200</u>	<u>9%</u>	<u>435</u>	<u>19%</u>	<u>197</u>	<u>9%</u>	<u>1,277</u>	<u>56%</u>	2,283	100%
between 20 and 23 y.o.	226	8%	283	10%	628	22%	<u>161</u>	<u>6%</u>	<u>1,526</u>	<u>54%</u>	2,824	100%
24 y.o. or above	250	9%	<u>339</u>	<u>12%</u>	<u>735</u>	<u>25%</u>	<u>159</u>	<u>5%</u>	<u>1,420</u>	<u>49%</u>	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%	<u>22</u>	<u>5%</u>	<u>98</u>	<u>24%</u>	<u>37</u>	<u>9%</u>	<u>139</u>	<u>34%</u>	413	100%
between 16 and 19 y.o.	<u>673</u>	<u>29%</u>	<u>185</u>	<u>8%</u>	<u>598</u>	<u>26%</u>	153	7%	674	30%	2,283	100%
between 20 and 23 y.o.	<u>696</u>	<u>25%</u>	262	9%	833	29%	<u>141</u>	<u>5%</u>	<u>892</u>	<u>32%</u>	2,824	100%
24 y.o. or above	763	26%	<u>300</u>	<u>10%</u>	<u>905</u>	<u>31%</u>	169	6%	<u>766</u>	<u>26%</u>	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.

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.

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.

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.

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.

Cross: How would you best describe yourself? / Your social life...

HOW WOULD YOU BEST DESCRIBE YOURSELF?	YOUR SOCIAL LIFE...											
	...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...

TYPOLOGY OF COUNTRIES BASED ON SIZE AND WELFARE	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	%
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.

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.*

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')	<u>243</u>	<u>17%</u>	145	10%	<u>590</u>	<u>40%</u>	175	12%	<u>308</u>	<u>21%</u>	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')	<u>315</u>	<u>11%</u>	<u>349</u>	<u>12%</u>	<u>777</u>	<u>27%</u>	<u>367</u>	<u>13%</u>	<u>1,040</u>	<u>37%</u>	2,848	100%
TOTAL	1,124	13%	999	11%	2,876	32%	1,061	12%	2,862	32%	8,922	

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')	<u>146</u>	<u>10%</u>	<u>315</u>	<u>21%</u>	<u>643</u>	<u>44%</u>	<u>206</u>	<u>14%</u>	<u>157</u>	<u>11%</u>	1,467	100%
Group B ('Western Europe')	<u>299</u>	<u>6%</u>	1,165	25%	<u>1,511</u>	<u>33%</u>	775	17%	<u>865</u>	<u>19%</u>	4,615	100%
Group C ('Northern Europe')	187	7%	701	25%	988	35%	<u>571</u>	<u>20%</u>	<u>405</u>	<u>14%</u>	2,852	100%
TOTAL	632	7%	2,181	24%	3,142	35%	1,552	17%	1,427	16%	8,934	

Under-represented elements Over-represented elements

The relationship is very significant. *p-value= < 0,01 ; Chi2= 141.4 ; dof= 8.*

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...											
	...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')	295	20%	<u>46</u>	<u>3%</u>	<u>521</u>	<u>36%</u>	271	18%	<u>334</u>	<u>23%</u>	1,467	100%
Group B ('Western Europe')	<u>816</u>	<u>18%</u>	92	2%	1,221	26%	835	18%	<u>1,651</u>	<u>36%</u>	4,615	100%
Group C ('Northern Europe')	<u>576</u>	<u>20%</u>	58	2%	<u>587</u>	<u>21%</u>	531	19%	<u>1,099</u>	<u>39%</u>	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...											
	...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')	<u>796</u>	<u>54%</u>	109	7%	<u>464</u>	<u>32%</u>	28	2%	64	4%	1,461	100%
Group B ('Western Europe')	2,309	50%	<u>394</u>	<u>9%</u>	1,615	35%	82	2%	<u>213</u>	<u>5%</u>	4,613	100%
Group C ('Northern Europe')	1,420	50%	<u>197</u>	<u>7%</u>	961	34%	<u>72</u>	<u>3%</u>	<u>198</u>	<u>7%</u>	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.*

Cross: Genetic diseases / Access to the most adapted care, treatments or surgery...

GENETIC DISEASES	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	%
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...

GENETIC DISEASES	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	%
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.*

Cross: Genetic diseases / Financial support including social security benefits...

GENETIC DISEASES	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	%
Genetic diseases	458	16%	<u>560</u>	<u>19%</u>	1,139	39%	243	8%	<u>501</u>	<u>17%</u>	2,901	100%
Non Genetic diseases	315	15%	<u>351</u>	<u>16%</u>	823	38%	202	9%	<u>469</u>	<u>22%</u>	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...

GENETIC DISEASES	INTEGRATION AT SCHOOL...											
	...HAS GOTTEN WORSE		...HAS IMPROVED		...HAS REMAINED THE SAME		DON'T KNOW		NOT RELEVANT		TOTAL	
	N	%	N	%	N	%	N	%	N	%	N	%
Genetic diseases	<u>474</u>	<u>9%</u>	<u>722</u>	<u>14%</u>	<u>1,434</u>	<u>27%</u>	342	7%	<u>2,260</u>	<u>43%</u>	5,232	100%
Non Genetic diseases	<u>175</u>	<u>7%</u>	<u>127</u>	<u>5%</u>	<u>327</u>	<u>13%</u>	156	6%	<u>1,749</u>	<u>69%</u>	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.

Cross: Genetic diseases / Access to clinical trials...

GENETIC DISEASES	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	%
Genetic diseases	360	7%	<u>1,367</u>	<u>26%</u>	1,863	36%	853	16%	<u>789</u>	<u>15%</u>	5,232	100%
Non Genetic diseases	173	7%	<u>576</u>	<u>23%</u>	889	35%	453	18%	<u>442</u>	<u>17%</u>	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...

GENETIC DISEASES	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	%
Genetic diseases	989	19%	<u>138</u>	<u>3%</u>	<u>1,486</u>	<u>28%</u>	942	18%	<u>1,676</u>	<u>32%</u>	5,231	100%
Non Genetic diseases	485	19%	<u>38</u>	<u>2%</u>	<u>538</u>	<u>21%</u>	442	17%	<u>1,030</u>	<u>41%</u>	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.*

Cross: Genetic diseases / Your social life...

GENETIC DISEASES	YOUR SOCIAL LIFE...											
	...HAS GOTTEN WORSE		...HAS IMPROVED		...HAS REMAINED THE SAME		DON'T KNOW		NOT RELEVANT		TOTAL	
	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...

POINT PREVALENCE OF 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	%
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.

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.*

Cross: Point prevalence of the rare disease / Integration at school...

POINT PREVALENCE OF 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	%
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...

POINT PREVALENCE OF 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	%
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.

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)...											
	...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	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...											
	...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	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: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / Access to the most adapted care, treatments or surgery...

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	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	888	48%	4,020	45%	3,346	37%	457	5%	342	4%	8,024	100%

Under-represented elements

Over-represented elements

The relationship is very significant. p-value= < 0,01 ; Chi2= 146.8 ; dof= 16.

Cross: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / Understanding how the disease will progress...

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	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	693	28%	4,994	55%	2,649	29%	524	6%	175	2%	8,024	100%

Under-represented elements

Over-represented elements

The relationship is very significant. p-value= < 0,01 ; Chi2= 39.6 ; dof= 16.

Cross: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / Integration at work...

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	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%

Under-represented elements Over-represented elements

The relationship is very significant. p-value= < 0,01 ; Chi2= 115.0 ; dof= 16.

Cross: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / Access to social services (e.g. social worker support, household chores support)...

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	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%

Under-represented elements Over-represented elements

The relationship is very significant. p-value= < 0,01 ; Chi2= 213.4 ; dof= 16.

Cross: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / Access to clinical trials...

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	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%

Under-represented elements Over-represented elements

The relationship is very significant. p-value= < 0,01 ; Chi2= 137.3 ; dof= 16.

Cross: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / Access to financial products, such as loans, mortgages, insurance...

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	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%

Under-represented elements Over-represented elements

The relationship is very significant. p-value= < 0,01 ; Chi2= 171.4 ; dof= 16.

Cross: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / Your social life...

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	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	<u>2,396</u>	<u>46%</u>	432	8%	<u>2,003</u>	<u>38%</u>	112	2%	<u>304</u>	<u>6%</u>	5,247	100%
4-7 body parts	<u>1,479</u>	<u>56%</u>	<u>185</u>	<u>7%</u>	<u>791</u>	<u>30%</u>	53	2%	141	5%	2,649	100%
8-11 body parts	<u>504</u>	<u>62%</u>	61	7%	<u>211</u>	<u>26%</u>	14	2%	<u>29</u>	<u>4%</u>	819	100%
12-15 body parts	<u>155</u>	<u>66%</u>	26	11%	<u>47</u>	<u>20%</u>	4	2%	<u>3</u>	<u>1%</u>	235	100%
16 body parts or more	<u>37</u>	<u>69%</u>	4	7%	12	22%	0	0%	1	2%	54	100%

Under-represented elements Over-represented elements

The relationship is very significant. p-value= < 0,01 ; Chi2= 169.6 ; dof= 16.

Cross: Number body parts impacted by the rare disease (or index of disease complexity) Calculated variable computing answers to the question "Which parts of the body does the rare disease impact?" / Access to financial products, such as loans, mortgages, insurance...

NUMBER BODY PARTS IMPACTED BY THE RARE DISEASE (OR INDEX OF DISEASE COMPLEXITY) CALCULATED VARIABLE COMPUTING ANSWERS TO THE QUESTION "WHICH PARTS OF THE BODY DOES THE RARE DISEASE IMPACT?"	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	<u>863</u>	<u>16%</u>	116	2%	1,337	25%	938	18%	<u>2,004</u>	<u>38%</u>	5,258	100%
4-7 body parts	528	20%	54	2%	<u>749</u>	<u>28%</u>	499	19%	<u>825</u>	<u>31%</u>	2,655	100%
8-11 body parts	<u>214</u>	<u>26%</u>	21	3%	193	24%	162	20%	<u>229</u>	<u>28%</u>	819	100%
12-15 body parts	<u>86</u>	<u>37%</u>	6	3%	56	24%	39	17%	<u>48</u>	<u>20%</u>	235	100%
16 body parts or more	<u>24</u>	<u>44%</u>	3	5%	10	18%	10	18%	<u>8</u>	<u>15%</u>	55	100%

Under-represented elements Over-represented elements

The relationship is very significant. p-value= < 0,01 ; Chi2= 171.4 ; dof= 16.

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	

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	

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.

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.

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.*

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	<u>1,448</u>	<u>57%</u>	<u>241</u>	<u>10%</u>	<u>702</u>	<u>28%</u>	50	2%	<u>87</u>	<u>3%</u>	2,528	100%
No	<u>2,915</u>	<u>48%</u>	<u>454</u>	<u>7%</u>	<u>2,256</u>	<u>37%</u>	<u>110</u>	<u>2%</u>	<u>370</u>	<u>6%</u>	6,105	100%
Don't know	<u>208</u>	<u>56%</u>	<u>13</u>	<u>4%</u>	<u>106</u>	<u>29%</u>	<u>23</u>	<u>6%</u>	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	<u>323</u>	<u>13%</u>	<u>1,053</u>	<u>42%</u>	935	37%	120	5%	87	3%	2,518	100%
No	<u>535</u>	<u>9%</u>	<u>2,879</u>	<u>46%</u>	2,289	37%	312	5%	243	4%	6,258	100%
Don't know	31	13%	<u>88</u>	<u>35%</u>	92	37%	<u>25</u>	<u>10%</u>	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	

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	

Under-represented elements Over-represented elements

The relationship is very significant. *p-value= < 0,01 ; Chi2= 141.6 ; 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	

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	

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	

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	

Under-represented elements Over-represented elements

The relationship is very significant. p-value= < 0,01 ; Chi2= 141.1 ; dof= 8.

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.

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	

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	

Under-represented elements Over-represented elements

The relationship is very significant. p-value= < 0,01 ; Chi2= 105.0 ; dof= 8.

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	

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	

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	

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	

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	<u>2,794</u>	<u>55%</u>	392	8%	<u>1,633</u>	<u>32%</u>	<u>85</u>	<u>2%</u>	<u>189</u>	<u>4%</u>	5,093	100%
No	<u>1,468</u>	<u>45%</u>	269	8%	<u>1,229</u>	<u>37%</u>	69	2%	<u>249</u>	<u>8%</u>	3,284	100%
Don't know	309	49%	47	7%	202	32%	<u>29</u>	<u>5%</u>	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	<u>648</u>	<u>11%</u>	<u>2,719</u>	<u>45%</u>	<u>2,144</u>	<u>36%</u>	<u>277</u>	<u>5%</u>	<u>193</u>	<u>3%</u>	5,981	100%
No	<u>205</u>	<u>8%</u>	1,111	43%	<u>993</u>	<u>39%</u>	137	5%	<u>125</u>	<u>5%</u>	2,571	100%
Don't know	36	8%	190	40%	179	38%	<u>43</u>	<u>9%</u>	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...

...INVISIBLE SYMPTOMS SUCH AS PAIN, DIZZINESS, HEADACHES, FATIGUE, 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	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...

...INVISIBLE SYMPTOMS SUCH AS PAIN, DIZZINESS, HEADACHES, FATIGUE, 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	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.

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	

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	

Under-represented elements Over-represented elements

The relationship is very significant. p-value= < 0,01 ; Chi2= 187.7 ; dof= 8.

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.*

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	<u>1,316</u>	<u>22%</u>	<u>114</u>	<u>2%</u>	<u>1,468</u>	<u>25%</u>	1,079	18%	<u>2,004</u>	<u>34%</u>	5,981	100%
No	<u>341</u>	<u>13%</u>	<u>74</u>	<u>3%</u>	<u>744</u>	<u>29%</u>	461	18%	<u>949</u>	<u>37%</u>	2,569	100%
Don't know	<u>58</u>	<u>12%</u>	12	3%	133	28%	<u>108</u>	<u>23%</u>	161	34%	472	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= 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	<u>3,337</u>	<u>56%</u>	468	8%	<u>1,835</u>	<u>31%</u>	<u>105</u>	<u>2%</u>	<u>228</u>	<u>4%</u>	5,973	100%
No	<u>1,007</u>	<u>39%</u>	219	9%	<u>1,065</u>	<u>42%</u>	57	2%	<u>213</u>	<u>8%</u>	2,561	100%
Don't know	227	48%	<u>21</u>	<u>4%</u>	164	35%	<u>21</u>	<u>4%</u>	<u>37</u>	<u>8%</u>	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.*

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	

■ Under-represented elements ■ Over-represented elements

The relationship is very significant. $p\text{-value} = < 0.01$: $\text{Chi}^2 = 106.2$: $\text{dof} = 8$.

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	

■ Under-represented elements ■ Over-represented elements

The relationship is very significant. $p\text{-value} = < 0.01$; $\text{Chi}^2 = 75.6$; $\text{dof} = 8$.

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...											
	...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.*

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	<u>1,282</u>	<u>32%</u>	379	9%	<u>1,056</u>	<u>26%</u>	228	6%	<u>1,073</u>	<u>27%</u>	4,018	100%
No	<u>990</u>	<u>22%</u>	397	9%	<u>1,394</u>	<u>31%</u>	280	6%	<u>1,444</u>	<u>32%</u>	4,505	100%
Don't know	139	29%	42	9%	137	28%	<u>43</u>	<u>9%</u>	<u>121</u>	<u>25%</u>	482	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= 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	<u>621</u>	<u>15%</u>	467	12%	1,322	33%	486	12%	<u>1,121</u>	<u>28%</u>	4,017	100%
No	<u>431</u>	<u>10%</u>	499	11%	1,429	32%	<u>501</u>	<u>11%</u>	<u>1,645</u>	<u>37%</u>	4,505	100%
Don't know	<u>82</u>	<u>17%</u>	45	9%	155	32%	<u>79</u>	<u>16%</u>	<u>121</u>	<u>25%</u>	482	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= 137.0 ; dof= 8.

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.

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	<u>2,240</u>	<u>56%</u>	<u>343</u>	<u>9%</u>	<u>1,194</u>	<u>30%</u>	69	2%	<u>171</u>	<u>4%</u>	4,017	100%
No	<u>2,074</u>	<u>46%</u>	344	8%	<u>1,715</u>	<u>38%</u>	91	2%	<u>281</u>	<u>6%</u>	4,505	100%
Don't know	257	53%	<u>21</u>	<u>4%</u>	155	32%	<u>23</u>	<u>5%</u>	26	5%	482	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= 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%	<u>16</u>	<u>8%</u>	206	100%
No	865	10%	3,933	45%	3,241	37%	447	5%	<u>326</u>	<u>4%</u>	8,812	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= 9.8 ; dof= 4.

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	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	16	8%	26	13%	<u>68</u>	<u>33%</u>	16	8%	<u>80</u>	<u>39%</u>	206	100%
No	730	8%	933	11%	<u>1,961</u>	<u>22%</u>	597	7%	<u>4,591</u>	<u>52%</u>	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	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	<u>42</u>	<u>21%</u>	22	11%	71	35%	17	8%	52	25%	204	100%
No	<u>2,367</u>	<u>27%</u>	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.

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)...											
	...HAS GOTTEN WORSE		...HAS IMPROVED		...HAS REMAINED THE SAME		DON'T KNOW		NOT RELEVANT		TOTAL	
	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...											
	...HAS GOTTEN WORSE		...HAS IMPROVED		...HAS REMAINED THE SAME		DON'T KNOW		NOT RELEVANT		TOTAL	
	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...											
	...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...											
	...HAS GOTTEN WORSE		...HAS IMPROVED		...HAS REMAINED THE SAME		DON'T KNOW		NOT RELEVANT		TOTAL	
	N	%	N	%	N	%	N	%	N	%	N	%
Yes	80	39%	20	10%	84	41%	5	2%	15	7%	204	100%
No	4,487	51%	688	8%	2,979	34%	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.

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%	<u>34</u>	<u>6%</u>	550	100%
No	832	10%	3,786	45%	3,122	37%	420	5%	<u>308</u>	<u>4%</u>	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%	<u>19</u>	<u>5%</u>	362	100%
No	653	8%	4,708	56%	2,483	29%	457	5%	<u>152</u>	<u>2%</u>	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.

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...											
	...HAS GOTTEN WORSE		...HAS IMPROVED		...HAS REMAINED THE SAME		DON'T KNOW		NOT RELEVANT		TOTAL	
	N	%	N	%	N	%	N	%	N	%	N	%
Yes	<u>42</u>	<u>24%</u>	28	16%	<u>55</u>	<u>31%</u>	21	12%	31	18%	177	100%
No	<u>884</u>	<u>15%</u>	1,027	18%	<u>2,288</u>	<u>39%</u>	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...											
	...HAS GOTTEN WORSE		...HAS IMPROVED		...HAS REMAINED THE SAME		DON'T KNOW		NOT RELEVANT		TOTAL	
	N	%	N	%	N	%	N	%	N	%	N	%
Yes	33	9%	<u>70</u>	<u>19%</u>	<u>127</u>	<u>35%</u>	<u>43</u>	<u>12%</u>	<u>90</u>	<u>25%</u>	363	100%
No	696	8%	<u>860</u>	<u>10%</u>	<u>1,837</u>	<u>22%</u>	<u>552</u>	<u>7%</u>	<u>4,525</u>	<u>53%</u>	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.*

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.

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.*

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.*

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...

FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME 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	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. $p\text{-value}= 0.1$; $\text{Chi}^2= 7.8$; $\text{dof}= 4$.

Cross: Family members were previously diagnosed with the same disease / Your social life...

FAMILY MEMBERS WERE PREVIOUSLY DIAGNOSED WITH THE SAME 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	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. $p\text{-value}= < 0,01$; $\text{Chi}^2= 32.7$; $\text{dof}= 4$.

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	

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	

Under-represented elements Over-represented elements

The relationship is very significant. p-value= < 0,01 ; Chi2= 77.2 ; dof= 16.

Cross: ...psychological support / Financial support including social security benefits...

FINANCIAL SUPPORT INCLUDING SOCIAL SECURITY BENEFITS...

...PSYCHOLOGICAL 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	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	

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...

...PSYCHOLOGICAL 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	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	

Under-represented elements Over-represented elements

The relationship is very significant. p-value= < 0,01 ; Chi2= 198.3 ; dof= 16.

Cross: ...psychological support / Integration at work...

...PSYCHOLOGICAL SUPPORT	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	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	

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)...

...PSYCHOLOGICAL 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	
	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	

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.

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)...											
	...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	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...											
	...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	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.

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	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	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	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	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	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	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	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.

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.*

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.*

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...											
	...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.*

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.

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.

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. $p\text{-value} = < 0,01$; $\chi^2 = 117.7$; $\text{dof} = 24$.

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? / 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	<u>287</u>	<u>6%</u>	<u>1,458</u>	<u>31%</u>	<u>1,621</u>	<u>34%</u>	<u>689</u>	<u>15%</u>	<u>690</u>	<u>15%</u>	4,745	100%
YES, through online communities	<u>342</u>	<u>8%</u>	1,057	24%	1,598	36%	800	18%	<u>629</u>	<u>14%</u>	4,426	100%
YES, through local networks (e.g. schools)	19	5%	<u>119</u>	<u>31%</u>	133	35%	59	15%	54	14%	384	100%
NO, because of accessibility issues (e.g. language or technical barriers)	<u>23</u>	<u>16%</u>	<u>19</u>	<u>13%</u>	48	34%	32	23%	19	13%	141	100%
NO, because I have not been able to find other people with the same disease	<u>89</u>	<u>9%</u>	<u>151</u>	<u>15%</u>	353	36%	<u>200</u>	<u>20%</u>	<u>183</u>	<u>19%</u>	976	100%
NO, because I don't want to	24	5%	<u>78</u>	<u>17%</u>	144	32%	<u>105</u>	<u>23%</u>	<u>97</u>	<u>22%</u>	448	100%
Other, specify...	31	7%	107	24%	<u>133</u>	<u>30%</u>	68	15%	<u>101</u>	<u>23%</u>	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.

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.

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.

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

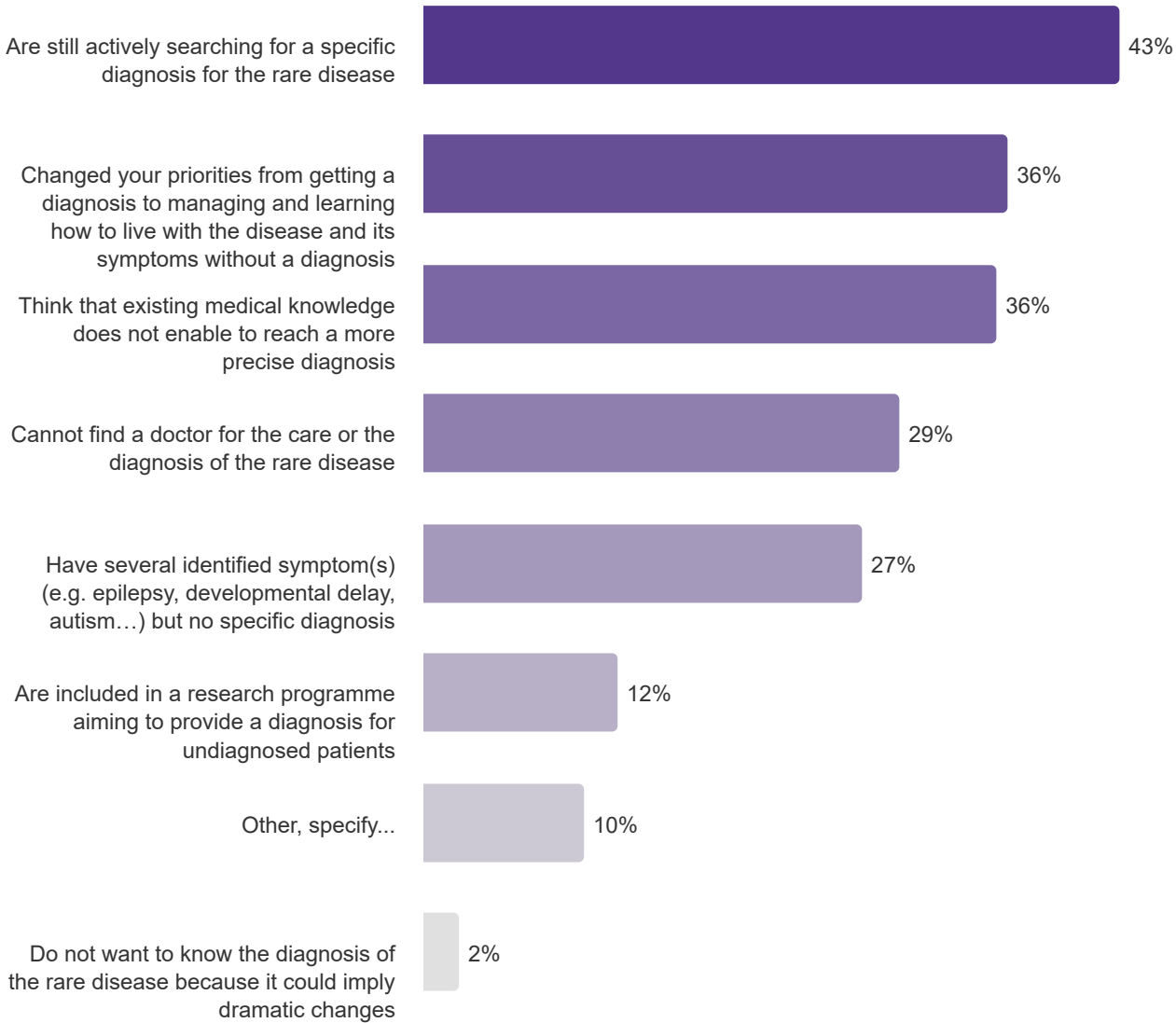
The relationship is very significant. p-value= < 0,01 ; Chi2= 146.4 ; dof= 16.

Chapter 16.

Questions for
undiagnosed /
unsolved cases

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:



Please select all the statements that describe your situation. You or the person you care for:

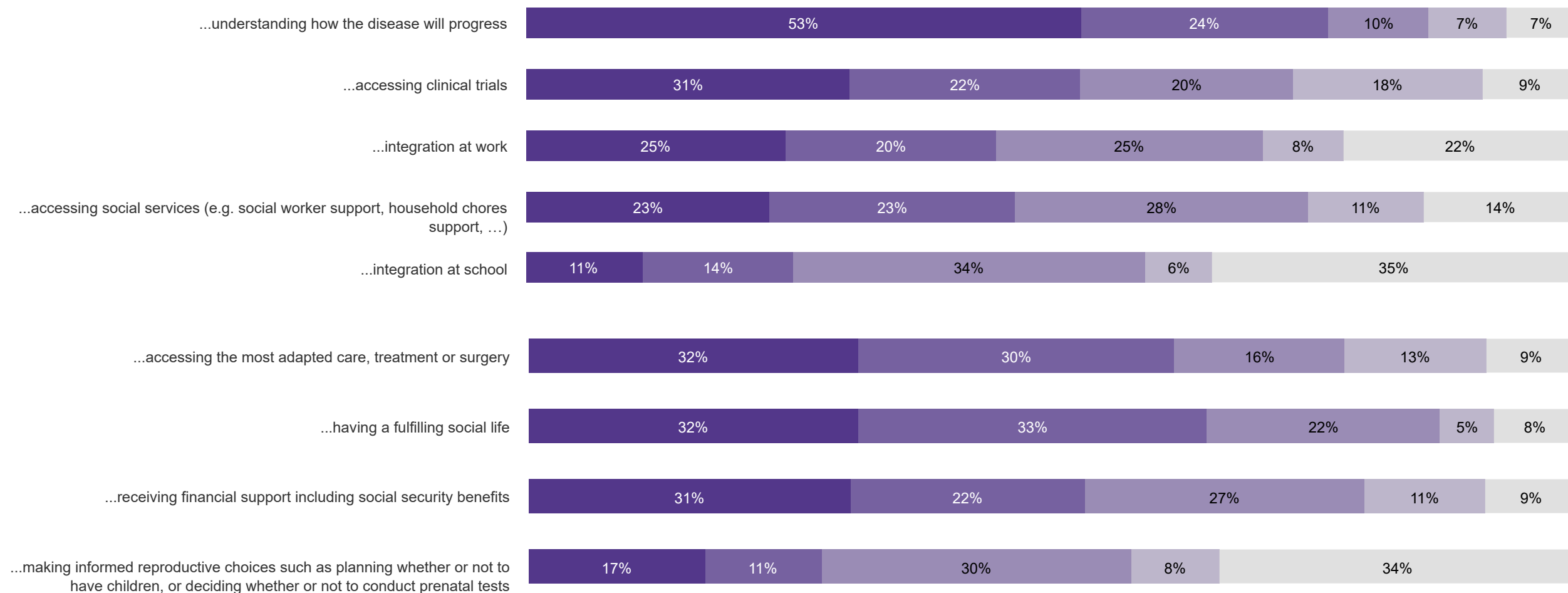
	N
Are still actively searching for a specific diagnosis for the rare disease	278
Changed your priorities from getting a diagnosis to managing and learning how to live with the disease and its symptoms without a diagnosis	233
Think that existing medical knowledge does not enable to reach a more precise 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



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...



● YES, completely ● YES, partially ● NO ● Don't know ● Not relevant

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

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!

