

WELCOME!

THIS WEBINAR WILL START AT 14:30 CET



Please make sure that your microphone is muted



Use the “raise hand” symbol if you want to raise a question in public



Feel free to leave your camera on or off



Use the chat to introduce yourself and for asking questions

RESULTS OF THE RARE BAROMETER SURVEY: THE DIAGNOSIS ODYSSEY OF PEOPLE LIVING WITH A RARE DISEASE

How to use them in your advocacy work and your actions!

OVERVIEW

1

RESULTS OF THE DIAGNOSIS SURVEY

Welcome and introduction

The Diagnostic odyssey: a Rare Barometer study

Results presented in the peer-reviewed article:

- Measuring the time to diagnosis
- The diagnosis odyssey: a long journey
- The diagnosis odyssey: a difficult journey
- Post-diagnosis changes
- Main determinants of diagnostic delays
- Key results

2

USING THE RESULTS

Factsheet & dashboard

Accessing tailored results (EURORDIS members)

Example of uses of the results:

- Myasthenia Gravis – Liselotte Chirakow, Anna Kole
- HHT – Claudia Crocione
- French public campaign
- Global Commission – Toon Digneffe, Danielle Dong

Upcoming Rare Barometer projects

CONTEXT

eurordis.org/our-priorities/diagnosis

Many people with rare diseases remain undiagnosed or misdiagnosed for years.

Timely and accurate diagnosis is essential to access to the right treatments and support, and ultimately improve quality of life.

Diagnosis of rare diseases is often difficult and can take years, leading to significant delays in treatment and considerable challenges for patients, families, and healthcare systems.

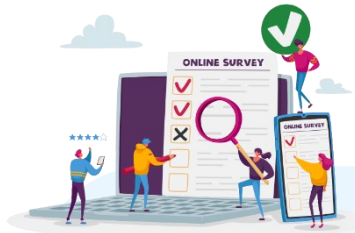
Policy goals set for the diagnosis of rare diseases range **from six months of coming to medical attention** (Rare 2030 recommendation 2) **to one year of coming to medical attention** (IRDiRC Goal 1)



THE RARE BAROMETER PROGRAMME

eurordis.org/voices

EURORDIS' survey initiative to support evidence-based advocacy



Surveys

People living with a rare disease & family members

1-3 surveys per year

25 languages

Worldwide

Up to 13,000
respondents to our surveys



Panel

20,000+
people living with a rare
disease registered

2,300+ rare diseases

120+ countries

*People DO NOT have to register
to participate in surveys*



Make your voice heard!

Collective results shared
with participants, patient
organisations, decision makers
and the wider public

*Information is only accessible to the Rare
Barometer team, saved on a secured server
in Europe*

GOALS OF THE RARE BAROMETER DIAGNOSIS STUDY

- Measuring the **time necessary to obtain a diagnosis** as a rare disease patient
- Understanding the **different steps** of the diagnosis odyssey
- Defining the **obstacles limiting the access** to diagnosis
- Identifying **best practices, tools, support and services** contributing to a faster diagnosis
- Identifying the **role** of new technologies

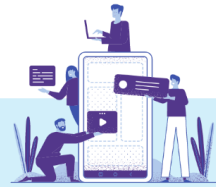


SHAPING THE ONLINE QUESTIONNAIRE



Literature review

Identify **indicators** already existing or still missing



Online panel

Refine the **diagnosis concept** and identify what is **new in the field**

61 participants



Topic Expert Committee

Contribute to identifying **issues and indicators** to include in the questionnaire



EURORDIS' Council of National Alliances Members

Input on **topics and indicators** to be included

Feedback on the questionnaire



Pilot test with patients and family members

9 participants

Translations checked **in 15 languages** by native speakers

“opinionway
healthcare

ONLINE QUESTIONNAIRE

17 MARCH> **15 JUNE** 2022

13,304 respondents
worldwide and

10,486 in
Europe

27 languages



TARGET POPULATION

All patients living with a rare disease and their family members, including unsolved cases (undiagnosed)

104 countries

1900+ diseases represented



RARE
DISEASES
INTERNATIONAL



GLOBAL COMMISSION

to End the Diagnostic Odyssey for Children with a Rare Disease

PUBLISHED PAPER

European Journal of Human Genetics

[nature.com/articles/s41431-024-01604-z](https://www.nature.com/articles/s41431-024-01604-z)

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- 204
- 3048
- 6394
- 9216

PUBLISHED PAPER

European Journal of Human Genetics

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Time to diagnosis and determinants of diagnostic delays of people living with a rare disease: results of a Rare Barometer retrospective patient survey

[Fatoumata Faye](#), [Claudia Crocione](#), [Roberta Anido de Peña](#), [Simona Bellagambi](#), [Luciana Escati Peñaloza](#), [Amy Hunter](#), [Lene Jensen](#), [Cor Oosterwijk](#), [Eva Schoeters](#), [Daniel de Vicente](#), [Laurence Faivre](#), [Michael Wilbur](#), [Yann Le Cam](#) & [Jessie Dubief](#) 

[European Journal of Human Genetics](#) **32**, 1116–1126 (2024) | [Cite this article](#)

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ARTICLE OPEN

 Check for updates

Time to diagnosis and determinants of diagnostic delays of people living with a rare disease: results of a Rare Barometer retrospective patient survey

Fatoumata Faye¹, Claudia Crocione², Roberta Anido de Peña³, Simona Bellagambi⁴, Luciana Escati Peñaloza², Amy Hunter⁵, Lene Jensen⁶, Cor Oosterwijk⁷, Eva Schoeters⁸, Daniel de Vicente⁹, Laurence Faivre¹⁰, Michael Wilbur¹¹, Yann Le Cam¹ and Jessie Dubief¹² 

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Timely diagnosis is one of the most serious challenges faced by people living with a rare disease (PLWRD), and this study estimates that in Europe, the average total diagnosis time (TDT) is close to 5 years. We investigated the duration of the TDT for PLWRD in Europe, the difficulties associated with their diagnosis odyssey and the main determinants of diagnosis delays for all rare diseases (RD). We conducted a survey of PLWRD and their families using Rare Barometer, the survey initiative of EURORDIS-Rare Diseases Europe. In geographical Europe, we surveyed 6507 people living with 1675 RD in 41 countries. We then performed a descriptive analysis and ordinal logistic regressions to identify the main determinants of diagnosis delays. Average TDT is 4.7 years. 56% of respondents were diagnosed more than 6 months after a first medical contact. The main determinants of diagnosis delays are symptom onset before 30 years of age, especially during childhood (OR = 3.11; 95% CI: 2.4–4.0) and adolescence (OR = 4.79; 95% CI: 3.7–6.2), being a woman (OR = 1.22; 95% CI: 1.1–1.4), living in Northern Europe (OR = 2.15; 95% CI: 1.8–2.6) or Western Europe (OR = 1.96; 95% CI: 1.6–2.3), the number of healthcare professionals consulted (OR = 5.15; 95% CI: 4.1–6.4), misdiagnosis (OR = 2.48; 95% CI: 2.1–2.9), referral to a centre of expertise (OR = 1.17; 95% CI: 1.0–1.3), unmet needs for psychological support (OR = 1.34; 95% CI: 1.2–1.5) and financial support (OR = 1.16; 95% CI: 1.0–1.3), having a genetic disease (OR = 1.33; 95% CI: 1.1–1.5) and a family history of an RD (OR = 1.36; 95% CI: 1.1–1.6). These determinants can inform policies and actions to improve access to diagnosis for all PLWRD.

European Journal of Human Genetics (2024) 32:1116–1126; <https://doi.org/10.1038/s41431-024-01604-z>

INTRODUCTION

In Europe, rare diseases (RD) have a point prevalence below 5 cases per 10,000 inhabitants. Over 6000 distinct RD have been identified to date, and together they affect approximately 30 million people in Europe and 300 million worldwide [1]. An estimated 72% of RD are genetic and 70% have a paediatric onset [1].

Accurate and timely diagnosis is critical to end the diagnostic odyssey experienced by PLWRD and ensure their access to appropriate health and social care. Early diagnosis allows to provide supportive holistic care as early as possible, can delay or reduce the apparition of symptoms and impairments, and defer the apparition of co-morbidities. Goals set for the diagnosis of RD range from six months (European Rare 2030 foresight study [2]) to one year (International Rare Diseases Research Consortium – IRDiRC [3]) of coming to medical attention. The available evidence [4–12] is primarily condition-specific or country-specific, but shows that the diagnostic search of PLWRD often involves visits with several healthcare professionals, numerous tests, misdiagnoses or inappropriate treatments, including surgeries, which is why it is often referred to as a ‘diagnostic odyssey’. The introduction of

next-generation sequencing should improve the diagnosis of people living with genetic RD who have been undiagnosed for years. Still, these technologies are being implemented at different rates in Europe, varying from countries with long-withstanding programmes granting automatic access, to others where they are still being deployed.

The first aim of this study is to estimate the Total Diagnostic Time (TDT) for all PLWRD in Europe i.e., the number of years between symptom onset and the confirmed diagnosis, and to describe the difficulties associated with the diagnostic odyssey. The second aim of this study is to identify the main determinants of diagnostic delays (TDT > 1 year) for PLWRD in Europe while distinguishing patient delays (PD), i.e., delays between symptom onset and the first medical contact, and health system delays (HSD), i.e., delays between the first medical contact and the confirmed diagnosis.

METHODS

Survey development

This study was conducted within the Rare Barometer programme, the survey initiative of EURORDIS-Rare Diseases Europe [13]. It relies on an

¹EURORDIS-Rare Diseases Europe, Paris, France. ²IRiE Europe, Roma, Italy. ³FADEPOF - Federación Argentina de Enfermedades Poco Frecuentes, Caba, Argentina. ⁴UNIAMO - Federazione Italiana Malattie Rare, Roma, Italy. ⁵Genetic Alliance UK, London, UK. ⁶Genetic Alliance - Rare Disease Denmark, Taastrup, Denmark. ⁷VOP - Vereniging Samenwerkende Ouder En Patiëntenorganisaties, Soest, Netherlands. ⁸RadiOrg - Rare Diseases Belgium asbl/Vzw, Brussels, Belgium. ⁹FEDER - Federación Española De Enfermedades Raras, Madrid, Spain. ¹⁰Centre Hospitalier Universitaire Dijon-Bourgogne, Dijon, France. ¹¹Email: jessie.dubief@eurordis.org

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SPRINGER NATURE

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European Journal of Human Genetics

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Thank you!

to all the participants in the quantitative survey and the online discussion forum, and to all those who made this survey possible

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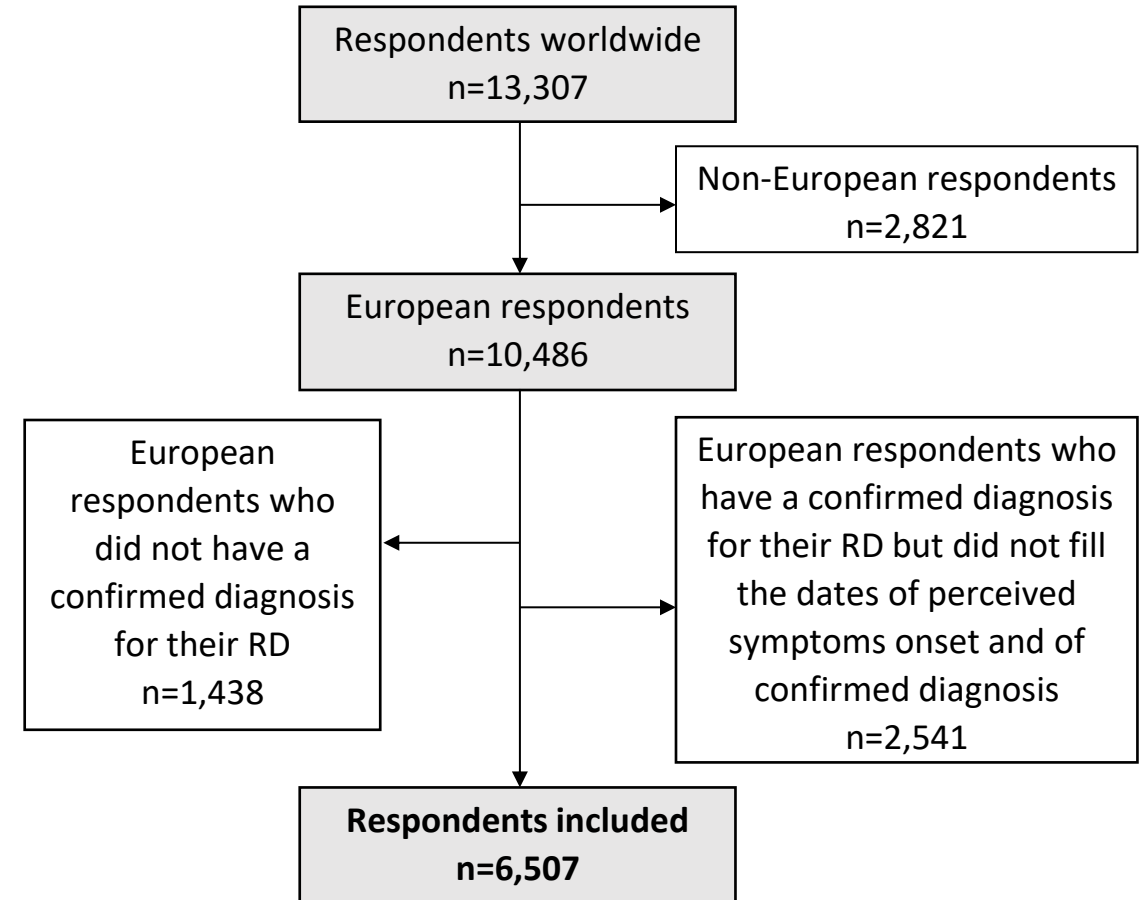
The authors are grateful to all the people living with a rare disease who participated in the quantitative survey and online discussion forum. The authors would also like to thank (i) Sandra Courbier (EURORDIS) for making this survey possible and directing its first stages; (ii) Erwan Berjonneau (EURORDIS), Andrea Osvoll (EURORDIS) and Victoria Chiseliov (EURORDIS) for their participation in the study; Rita Francisco (EURORDIS) for her review of the manuscript; (iii) the Topic Expert Committee members for their fruitful contribution: Shirlene Badger (Illumina), Simona Bellagambi (UNIAMO), Virginie-Bros Facer (EURORDIS), Valentina Botarelli (EURORDIS), Marie-Hélène Boucand (physician and PLWRD), Hélène Cederroth (Wilhelm Foundation), Anne-Sophie Chalandon (Sanofi), Alexandra le Chaffotec (Paris School of Business), Milda Galkute (FEDER), Edith Gross (EURORDIS), Sigrid Hendriks (VSOP), Amy Hunter (Genetic Alliance UK), Anna Kole (EURORDIS), Maryam Matar (United Arab Emirates genetic disease association), Flaminia Macchia (Rare Diseases International), Alexandre Mejat (AFM-Téléthon), Hlawulani Mkhabela (Rare Diseases International), Linn Parrish (Takeda), Lauren Roberts (Genetic Alliance UK), Amy Simpson (Genetic Alliance UK) and Ariane Weinman (EURORDIS); (iv) the Global Commission to End the Diagnosis Odyssey of Children Living with a Rare Disease and Rare Diseases International for their help reaching out to patient communities outside Europe; (v) all the patient organisations that helped disseminate the survey in their communities, and especially FEBER and FEBRARAS (Brazil), Rare Diseases Croatia, Rare Diseases Czech Republic, Harso-Rare Disease Alliance Finland, Alliance Maladies Rares (France), ACSE (Germany), Rare Diseases Greece, Rare Diseases Hungary-HUFERDIS, Rare Diseases Ireland, Alan-Maladies Rares Luxembourg, Malaysian Rare Disorders Society, Rare Disorders NZ (New Zealand), FFO (Norway), RD-Portugal, Romanian National Alliance for Rare Diseases, Rare Diseases South Africa NPC, Rare Diseases Sweden, Proraris (Switzerland), Rare Diseases Turkey, NORD (USA), HHT Europe, Sarcoidosis Europe, European Myasthenia Gravis Association, European Federation of Williams Syndrome, CF Europe (cystic fibrosis), Federation of European Scleroderma Associations, NF Patients United (Neurofibromatosis) and European Tuberous Sclerosis Complex Association.

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[nature.com/articles/s41431-024-01604-z](https://www.nature.com/articles/s41431-024-01604-z)

- 1** Estimate the time necessary to obtain a **diagnosis** for all people living with a rare disease in Europe, **and describe the difficulties** associated with the diagnostic odyssey.
- 2** Identify the main **determinants of the diagnostic delays** (1+ year after symptom onset) for people living with a rare disease in Europe, while differentiating the impact of **patient delays and of healthcare system delays**.



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European Journal of Human Genetics

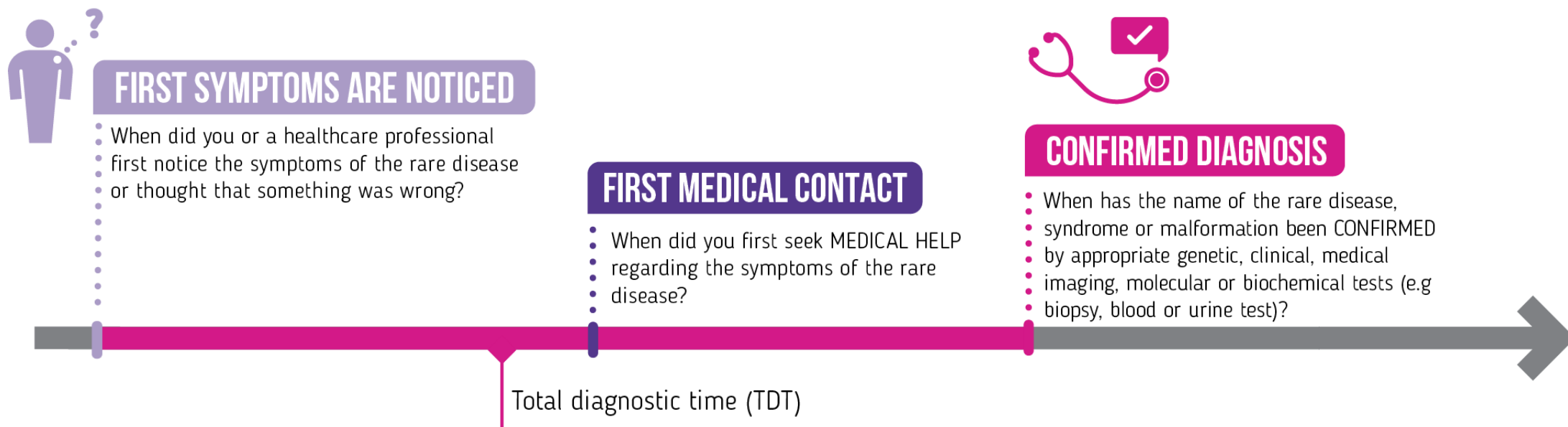
First study to investigate the time to diagnosis for people living with such a broad scope of rare diseases in 41 European countries.

Limitations inherent to any declarative retrospective survey on rare diseases:

- (i) by lack of epidemiological studies on rare diseases, recruitment bias cannot be estimated precisely;
- (ii) this study does not allow to monitor the evolution of time to diagnosis over time as this measure is subject to too many structural effects and does not fully capture the changing situation regarding genetic tests, which are increasingly realised at first intention and lower cost, at least in some European countries.

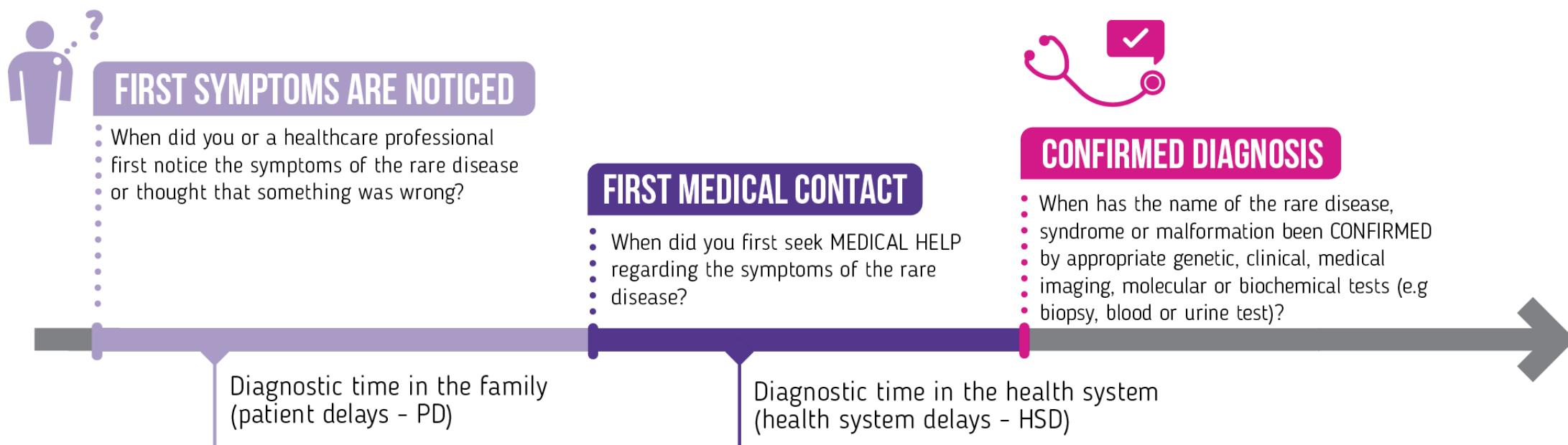
1. MEASURING THE TIME TO DIAGNOSIS

Questionnaire in English:
tiny.cc/questionnaire_diagRD



1. MEASURING THE TIME TO DIAGNOSIS

Questionnaire in English:
tiny.cc/questionnaire_diagRD



2. THE DIAGNOSIS ODYSSEY: A LONG JOURNEY

Time to diagnosis: distribution

Table 1. Characteristics of the respondents.

Variable	Category	Distribution % (n)	Total diagnosis time in years				
			Mean	Median (IQR)	% < 1	% 1–4	% ≥ 5
Total		100 (6507)	4.7	0.8 (0.1–5.0)	52	23	25

Distribution: percentage and frequencies of respondents for the categories of each variable (percentages in column-totals may not be equal between variables because of missing values); % < 1, % 1–4, % ≥ 5 = percentage of respondents who experienced a TDT of less than 1 year; 1–4 year; 5 years or more in each category (percentages in row).

% Percentage, N number of observations (totals are not equal between categories because of missing values), IQR Interquartile range.

Bold values refer to the corresponding figures for the whole sample (6507 people): average TDT is 4.7 years, median TDT is 0.8 years (IQR = 0.1-5.0), 52% respondents were diagnosed in less than 1 year, 23% within 1 to 4 years, and 25% in within 5 years or more.

Mean = 4.7 years = average value

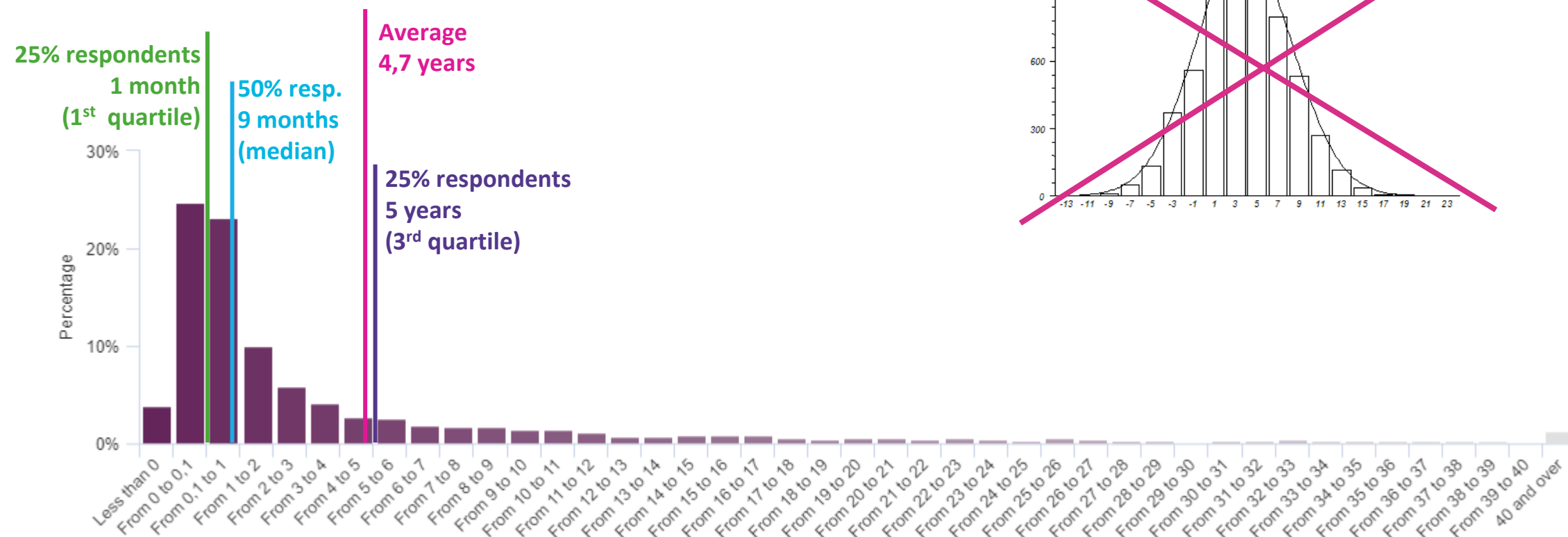
Median = 0.8 years (9 months) = 50% of responses below and 50% of responses above

First quartile (Q1) = 0.1 year (1 month) = 25% of responses below and 75% of responses above

Third quartile (Q3) = 5 years = 75% of responses below and 25% of responses above

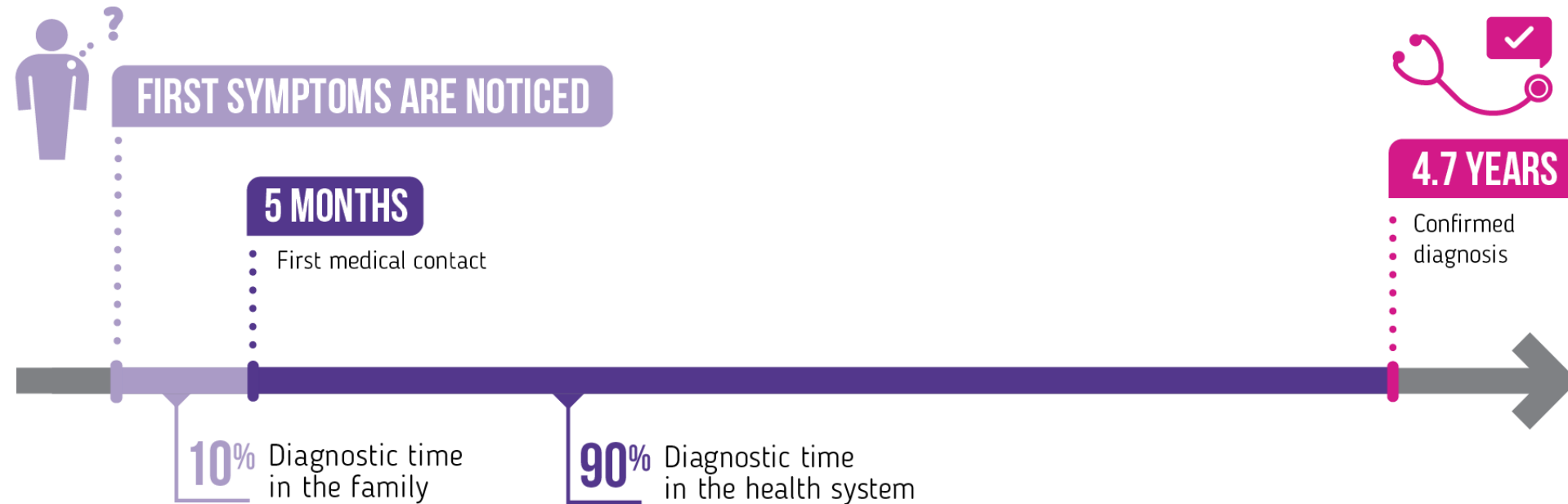
2. THE DIAGNOSIS ODYSSEY: A LONG JOURNEY

Time to diagnosis: distribution



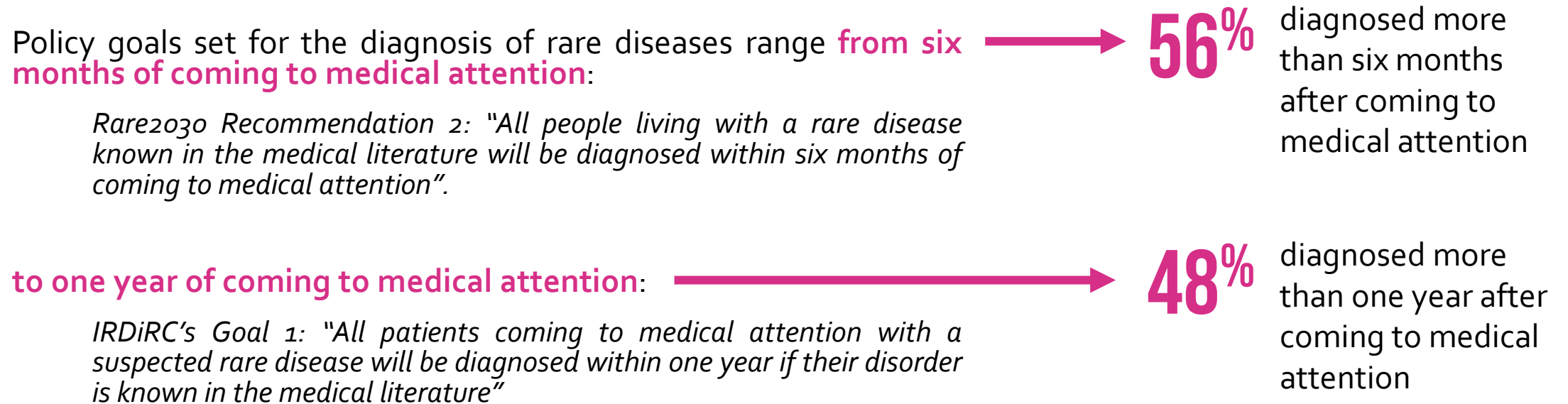
2. THE DIAGNOSIS ODYSSEY: A LONG JOURNEY

Most of the diagnostic time stems from health systems



2. THE DIAGNOSIS ODYSSEY: A LONG JOURNEY

Comparison with policy objectives



3. THE DIAGNOSIS ODYSSEY: A DIFFICULT JOURNEY

Associated difficulties



22% consulted at least 8 healthcare professionals
28%

of those diagnosed within one year consulted at least 5 healthcare professionals



73% were misdiagnosed at least one time
62%

of those diagnosed within one year were misdiagnosed at least one time



40% were NOT referred to a Centre of Expertise*
38%

of those diagnosed within one year were NOT referred to a Centre of Expertise

*A Centre of Expertise is a hospital unit specialised in a rare disease or in a group of rare diseases

3. THE DIAGNOSIS ODYSSEY: A DIFFICULT JOURNEY

Consequences of misdiagnosis

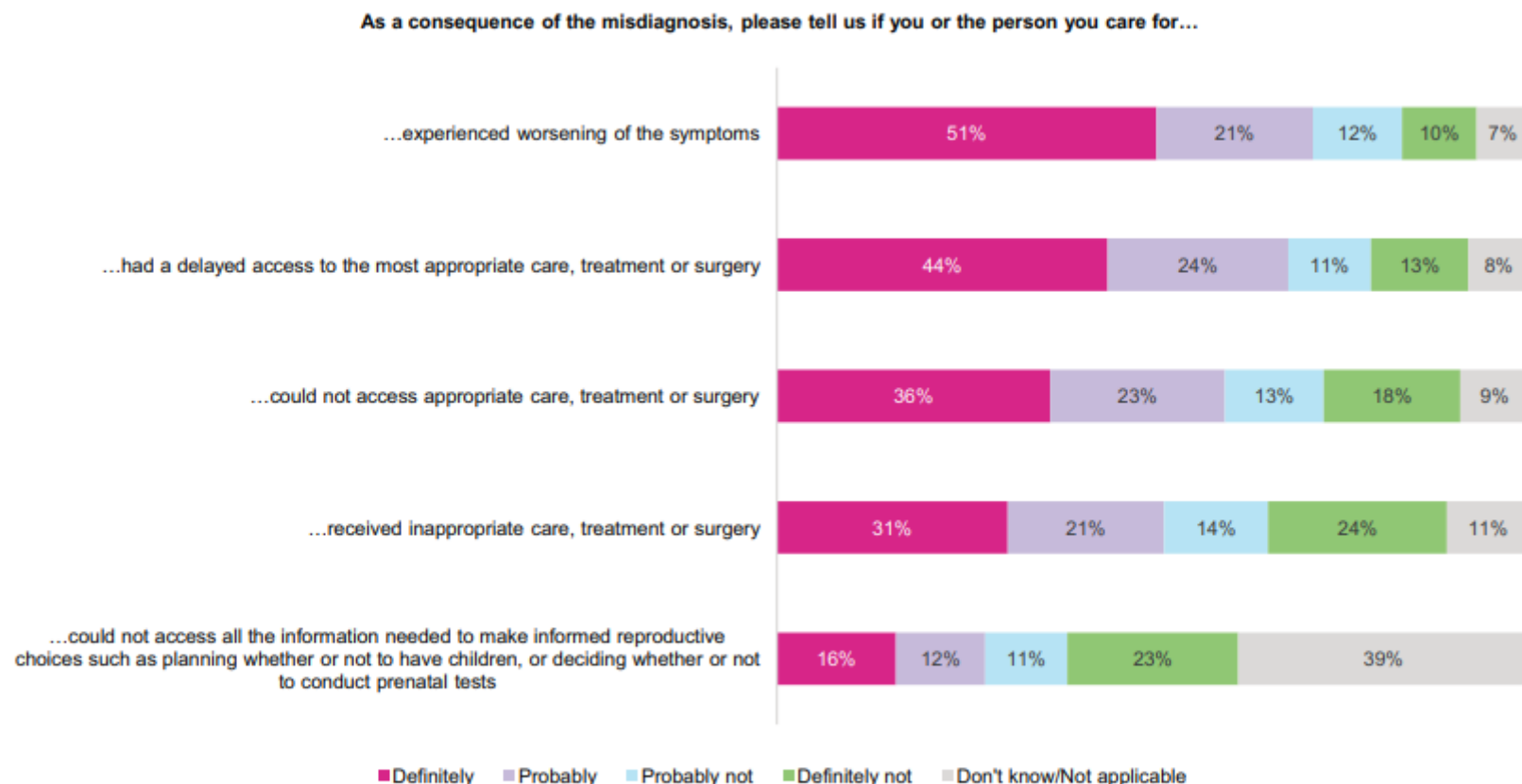


Fig. 2 Consequences of misdiagnosis. Respondents who said that their RD, or the RD of the person they care for, have been misdiagnosed at least once ($n = 4756$), i.e. that the symptoms of the RD have been attributed to another physical disease, or that symptoms were neglected, not taken seriously or considered as psychological.

3. THE DIAGNOSIS ODYSSEY: A DIFFICULT JOURNEY

Access to genetic tests



55%

Had a genetic test as part of the diagnosis of the rare disease

Q: Which tests were performed as part of the diagnosis of the rare disease? Genetic test(s) looking for genetic changes (also called mutations or variants).

Among those who had a genetic test and were diagnosed after 2010:

- 34% had gene-panel sequencing,
- 26% had single-gene sequencing,
- 24% had genome sequencing or exome sequencing,
- 25% didn't know which test was conducted.

3. THE DIAGNOSIS ODYSSEY: A DIFFICULT JOURNEY

Access to genetic tests



23%

Perceived hesitation from healthcare professionals to prescribe genetic tests

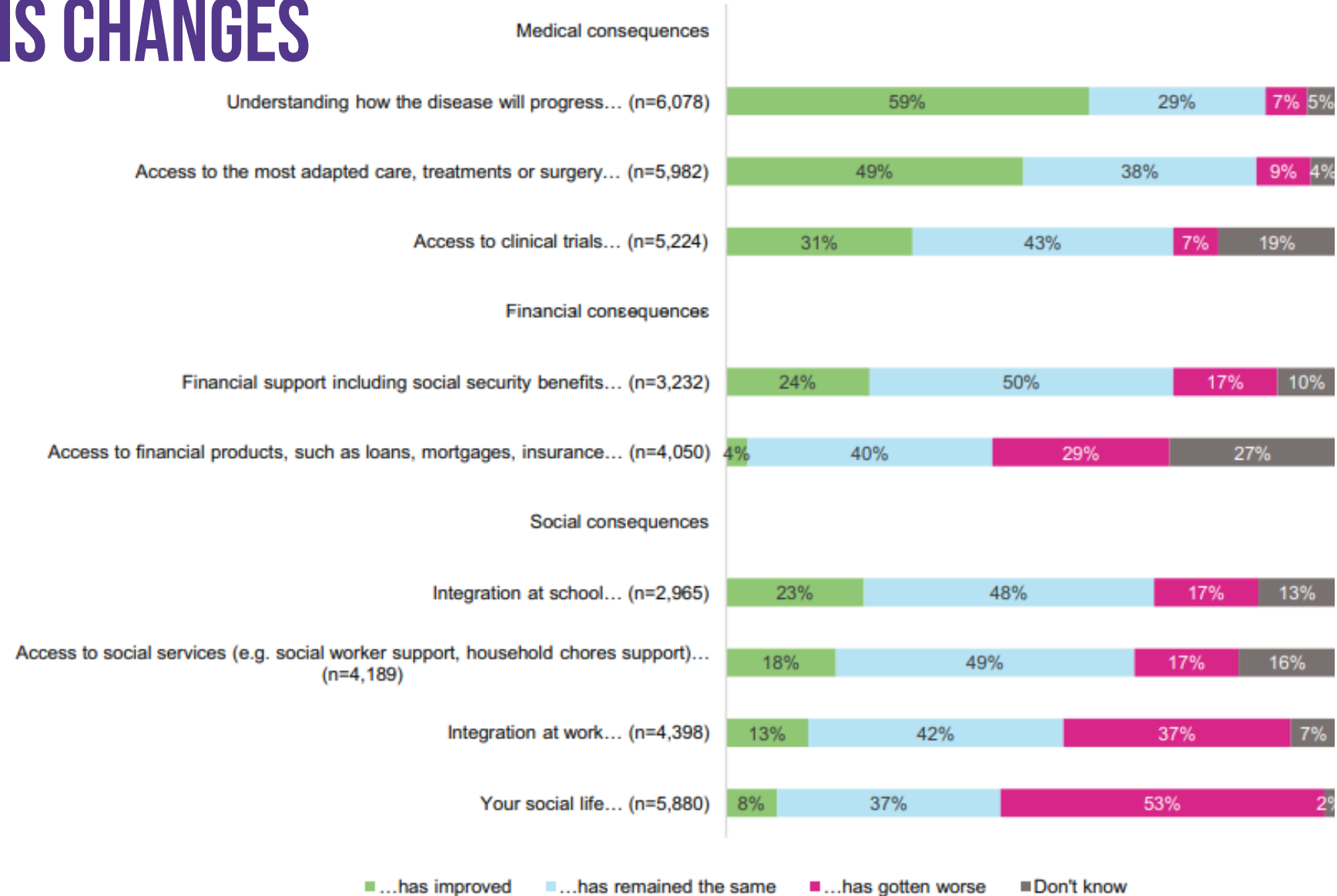
Q: Have you ever needed a genetic test but could not access it because healthcare professionals were reluctant or not sufficiently informed?

52% of those who perceived hesitation from healthcare professionals still had access to genetic tests, and 90% of them had a genetic condition

4. POST-DIAGNOSIS CHANGES

Since receiving a diagnosis for the rare disease, how have the following aspects changed for you or for the person you care for?

Responses 'not relevant' were removed: totals are not equal between categories because of varying number of responses 'not relevant' and missing values.



5. MAIN DETERMINANTS OF DIAGNOSTIC DELAYS

Variables

First, we crossed the median time to diagnosis with 70 variables (patients' sociodemographic and diagnosis-related characteristics; Kruskal-Wallis test) and we kept the 20 variables that had significant p-values.

> *Additional file 4 and Table 1 (pp. 1119-1120)*

Only one time-related variable included: see additional file 5 for methodological considerations.

Non-significant variables:

- City size, education: usual health determinants.
- Point prevalence: for diagnosis, 'rare is rare'.

Variables	chi2	p-value
SOCIODEMOGRAPHIC CHARACTERISTICS		
Age of the patient at perceived symptom onset	404.844	0.0001
Gender of the patient	39.098	0.0001
Country group	28.356	0.0001
CHARACTERISTICS OF THE DIAGNOSIS JOURNEY (HEALTHCARE SYSTEM)		
Number of healthcare professionals consulted	693.249	0.0001
Misdiagnosis	450.761	0.0001
Genetic tests	129.731	0.0001
Healthcare professionals were reluctant or not sufficiently informed to prescribe genetic tests (declarative)	230.671	0.0001
The patient was referred to a Centre of Expertise	31.301	0.0001
CHARACTERISTICS OF THE DIAGNOSIS JOURNEY (FAMILY AND SUPPORT)		
Family members already diagnosed	8.676	0.0032
Financial support	24.923	0.0001
Psychological support	37.601	0.0001
CHARACTERISTICS OF THE RARE DISEASE AND ASSOCIATED SYMPTOMS		
Number of body parts affected	84.727	0.0001
Genetic disease	123.975	0.0001
Skin diseases	121.883	0.0001
Gastroenterological diseases	32.464	0.0001
Gynaecological diseases	13.923	0.0002
Renal diseases	23.473	0.0001
Odontologic diseases	47.234	0.0001
Neurological diseases	0.045	0.8321
Sudden onset of symptoms	4.675	0.0966
Outbreaks	65.564	0.0001

Kruskal–Wallis equality-of-populations rank test

5. MAIN DETERMINANTS OF DIAGNOSTIC DELAYS

Results

Secondly, we included the 20 variables in ordinal logistic regressions with:

- **Patient delays**
(< 0 ; 0-0.25 years; ≥ 0.25 years)
- **Health system delays**
(< 1 year; 1-4 years; ≥ 5 years).
- **Diagnosis delays**
(< 1 year; 1-4 years; ≥ 5 years).

The model allows to estimate the specific influence of each of the 20 variables on the patient, health system and diagnosis delays (all things being equal).

Table 2 (pp. 1121-1122)

Table 2. Main variables associated with diagnosis delays, and their association with patient delays and health system delays.

Variables	Category	Patient delay		Health system delay		Diagnosis delay	
		OR (95% CI)	P	OR (95% CI)	P	OR (95% CI)	P
Sociodemographic characteristics							
Age of the patient at perceived symptom onset	<2	1.92	***	1.17	NS	2.26	***
		(1.5–2.4)		(0.9 – 1.5)		(1.8 – 2.8)	
	2–9	1.57	***	1.95	***	3.10	***
		(1.2–2.0)		(1.5–2.5)		(2.4–3.9)	
	10–19	2.70	***	1.71	***	4.74	***
		(2.1–3.5)		(1.3–2.2)		(3.7–6.2)	
	20–29	1.74	***	1.54	***	2.44	***
		(1.4–2.2)		(1.2–2.0)		(1.9–3.1)	
	30–49	1.33	***	1.36	***	1.70	***
	(1.1–1.6)		(1.1–1.7)		(1.4–2.0)		
	50 and more	ref (-)		ref (-)		ref (-)	
Gender of the patient	Male	ref (-)		ref (-)		ref (-)	
	Female	1.13	*	1.27	***	1.22	***
		(1.0–1.3)		(1.1–1.5)		(1.1–1.4)	
Country Group	Eastern, Central and Southern Europe	ref (-)		ref (-)		ref (-)	
	Western Europe	1.23	**	1.89	***	1.95	***
		(1.0–1.5)		(1.5–2.3)		(1.6–2.3)	
	Northern Europe	1.07	NS	2.30	***	2.11	***
		(0.9–1.3)		(1.9–2.8)		(1.7–2.5)	
Characteristics of the diagnosis journey (healthcare system)							

5. MAIN DETERMINANTS OF DIAGNOSTIC DELAYS

Results

Table 1 (pp. 1119-1120)

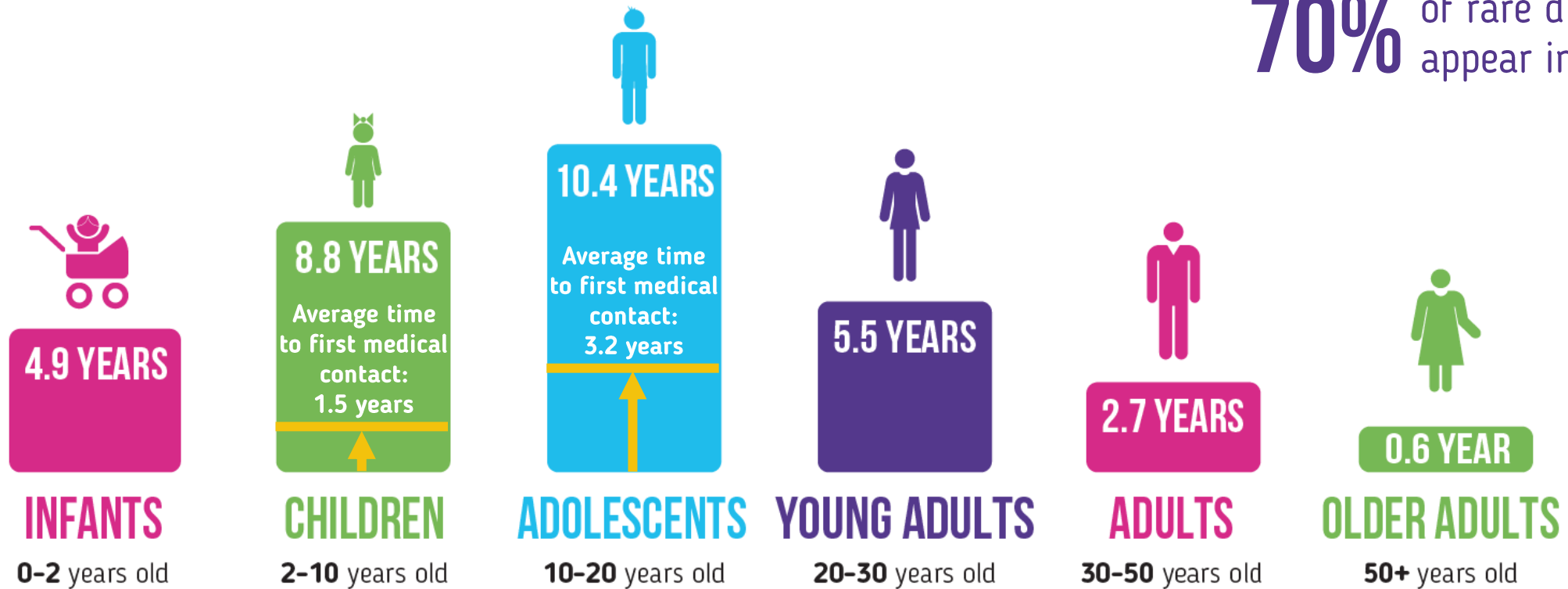
Table 1. Characteristics of the respondents.

Variable	Category	Distribution % (n)	Total diagnosis time in years				
			Mean	Median (IQR)	% < 1	% 1–4	% ≥ 5
Sociodemographic characteristics							
Age of the patient at perceived symptom onset	<2	25 (1502)	4.9	0.8 (0.1–4.5)	52	24	24
	2–9	11 (666)	8.8	2.0 (0.1–12.0)	42	19	39
	10–19	11 (629)	10.4	5.3 (0.3–18.3)	34	15	51
	20–29	12 (691)	5.5	1.3 (0.2–7.9)	46	22	33
	30–49	28 (1671)	2.7	0.7 (0.1–3.4)	55	26	19
	50 and more	14 (807)	0.6	0.3 (0.0–1.2)	72	22	6
Age of the patient at the time of the study	<2	2 (108)	0.5	0.2 (0.0–0.6)	83	15	2
	2–9	11 (648)	1.2	0.5 (0.1–1.6)	63	32	5
	10–19	12 (709)	2.7	0.9 (0.1–4.6)	50	26	24
	20–29	10 (570)	4.1	1.1 (0.1–6.5)	48	22	30
	30–49	29 (1752)	5.7	1.1 (0.1–7.3)	49	21	30
	50 and more	37 (2206)	6.2	0.8 (0.1–7.0)	51	20	29
Date of the confirmed diagnostic	<2000	12 (775)	2.5	0.3 (0.0–3.3)	61	19	20
	2000–2009	19 (1217)	4.5	0.6 (0.0–4.6)	55	20	25
	2010–2014	18 (1153)	4.8	0.8 (0.1–4.8)	53	22	25
	2015–2019	31 (1989)	5.5	1.0 (0.1–5.8)	49	24	27
	2020–2022	20 (1309)	5.2	1.1 (0.2–5.8)	47	26	27
Gender of the patient	Female	70 (4193)	5.4	1.0 (0.1–6.0)	50	22	28
	Male	30 (1839)	3.7	0.6 (0.1–3.6)	56	23	22

5. MAIN DETERMINANTS OF DIAGNOSTIC DELAYS

Age at first symptoms

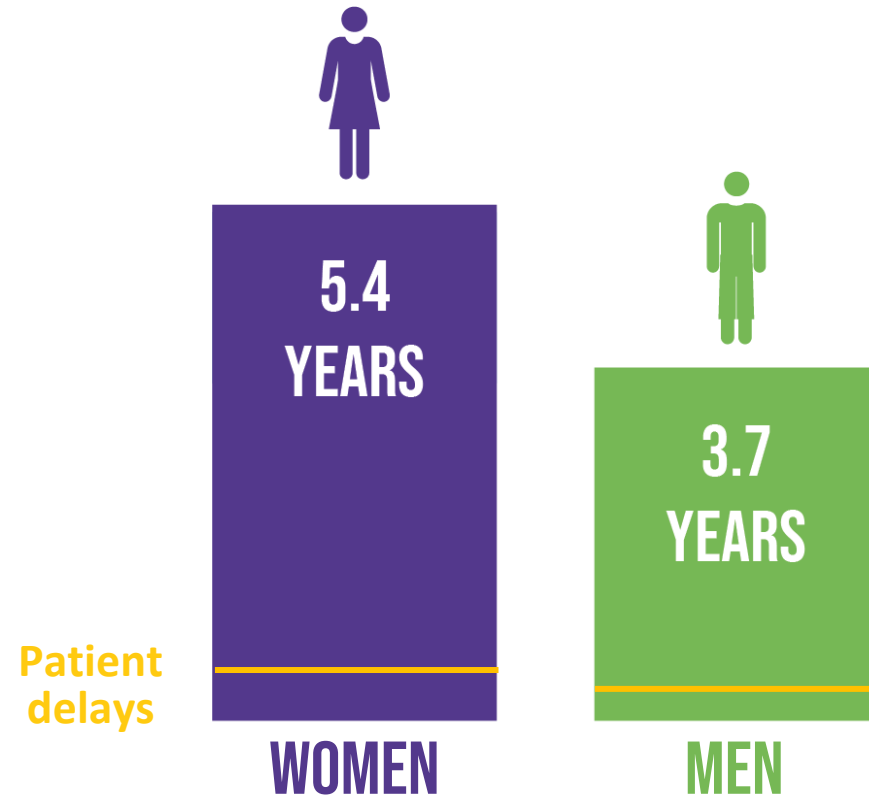
70% of rare diseases appear in childhood



Average number of years between first symptoms and confirmed diagnosis depending on the age of the patient at symptom onset
Logit: OR = 2.26 for infants; 3.10 for children; 4.74 for adolescents; 2.44 for young adults; 1,70 for adults; ref=older adults.

5. MAIN DETERMINANTS OF DIAGNOSTIC DELAYS

Sex of the patient



Average number of years between first symptoms and confirmed diagnosis depending on the sex of the patient.
Logit: OR = 1.22 for women.

5. MAIN DETERMINANTS OF DIAGNOSTIC DELAYS

Referral to a Centre of Expertise

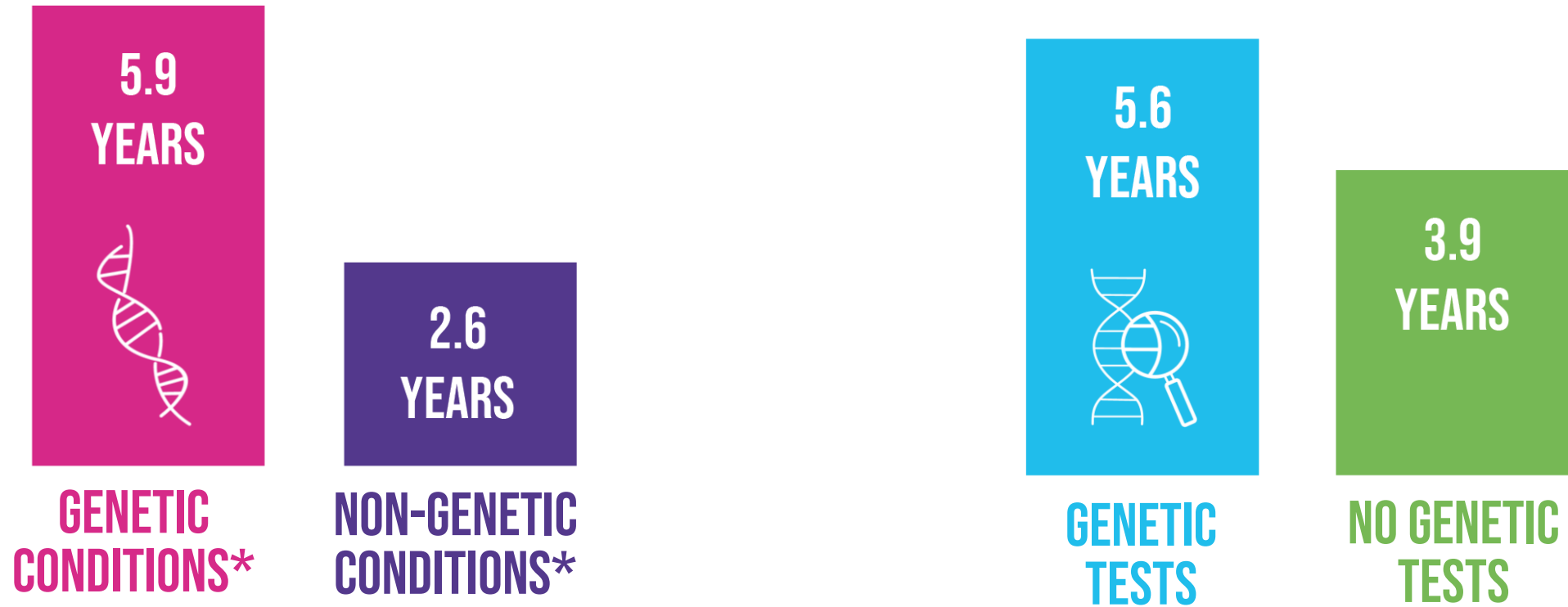


Average number of years between first symptoms and confirmed diagnosis depending if patients were referred to a Center of Expertise (a hospital unit specialised in a rare disease or in a group of rare diseases)

Logit: OR = 1.17 when not referred to a CoE

5. MAIN DETERMINANTS OF DIAGNOSTIC DELAYS

Genetic conditions and genetic tests



*Orphanet data

All respondents (genetic and non-genetic conditions)

Average number of years between first symptoms and confirmed diagnosis depending if the condition is genetic, and if the patient already had genetic tests. *Logit: OR = 1.34 for genetic conditions and 1.55 for genetic tests.*

5. MAIN DETERMINANTS OF DIAGNOSTIC DELAYS

Financial and psychological support needs

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

...financial support including social security benefits?

...psychological support?



YES and enough to cover my needs
YES but it is/was not needed
NO but it is/was NOT needed

Needs met

YES but NOT enough to cover my needs
NO but it is/was needed

Needs unmet

5. MAIN DETERMINANTS OF DIAGNOSTIC DELAYS

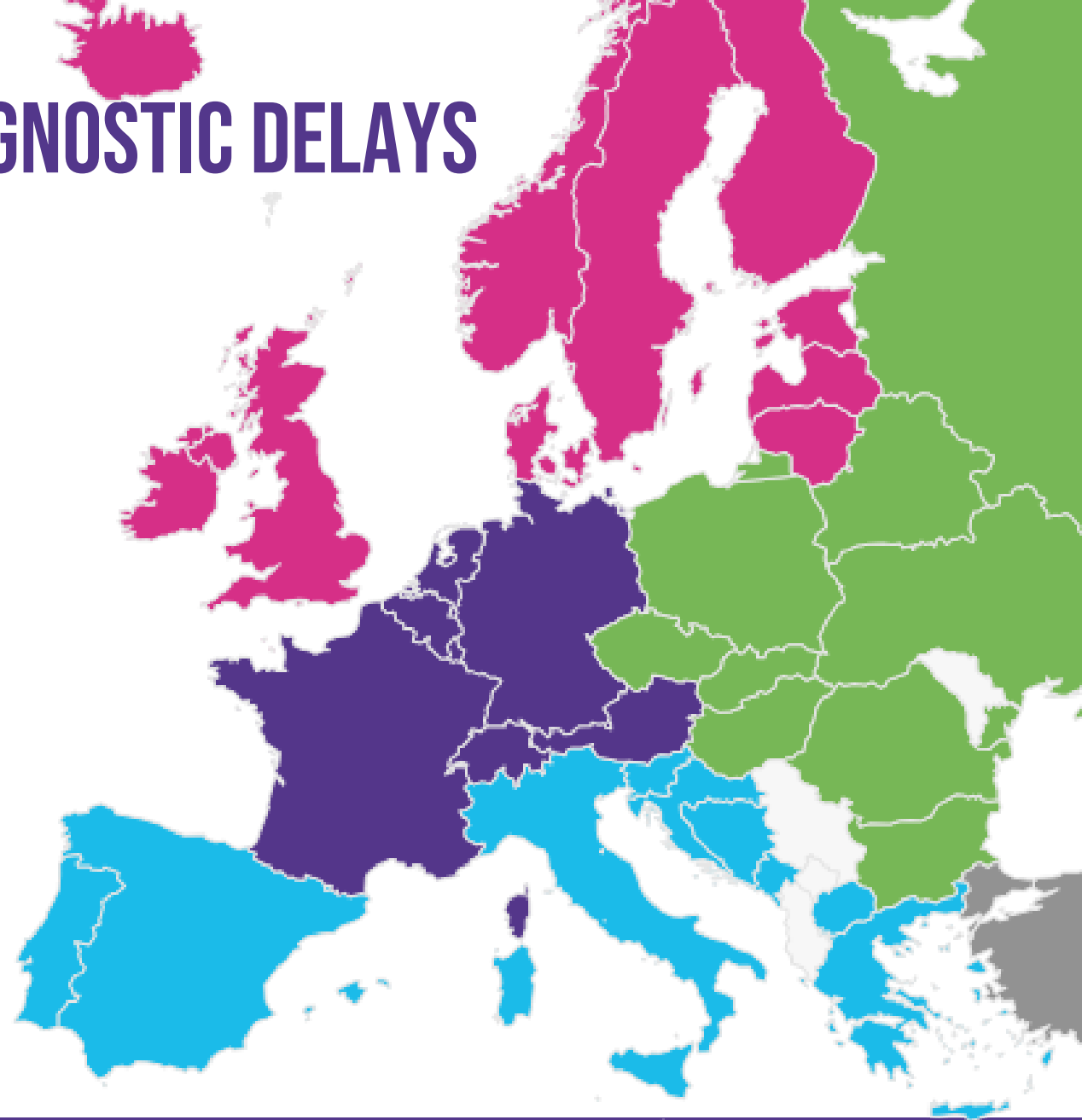
Financial and psychological support needs



Average number of years between first symptoms and confirmed diagnosis depending if patient's needs were met for financial and psychological support. Logit: OR = 1.17 for financial support and 1.33 for psychological support

5. MAIN DETERMINANTS OF DIAGNOSTIC DELAYS

European country groups



5. MAIN DETERMINANTS OF DIAGNOSTIC DELAYS

European country groups: consequences

Time to diagnosis for all people with rare diseases cannot be used as a stand-alone indicator to monitor rare disease diagnosis at a national and a European level (at least not across diseases: it is useful per disease):

- 1) Measured time to diagnosis may increase when rare diseases are better known and better managed in national healthcare systems, or when new diagnosis technologies allow to end more diagnostic odysseys.
- 2) This measure does not include rare disease patients who are still waiting for a confirmed diagnosis.

However, when coupled with a better understanding of the main determinants of diagnostic delays, total diagnostic time is a robust indicator to inform policies and actions that could be taken to improve access to diagnosis for people with rare diseases.

6. KEY RESULTS

The diagnostic odyssey is long...

Close to 5 years on average

90% of this time stems from the health system (time in the family is also key for children and adolescents)

...and difficult

Several consultations, misdiagnosis is very common (73%) and has negative consequences, 40% were not referred to a centre of expertise.

Access to diagnosis improved access to healthcare but not to psychosocial support

Prioritising actions in the health system:

- Improving awareness of all rare diseases among healthcare professionals, especially in primary care and among paediatricians
- Improving referral to centre of expertise

More holistic care

Better public awareness of rare diseases could help improve social acceptance

6. KEY RESULTS

Additional difficulties were encountered by some people with rare diseases:

Children and adolescents (9-10 years to diagnosis), both from patient delays (1.5-3 years) & health system delays (7 years)



Improve public and healthcare professionals' awareness of rare diseases among children and adolescents.

Women (+1.7 years), mostly from health systems



Considering symptoms of girls and women

People with rare genetic disorders (access to genetic tests)



Improve access to effective and available diagnostic testing technologies.

Psychological support, mostly impact on health system delays



Improve access to psychological support for undiagnosed patients.

Q&A

WE ARE ON A 10 MIN BREAK



Please make sure that your microphone is muted



Use the “raise hand” symbol if you want to raise a question in public

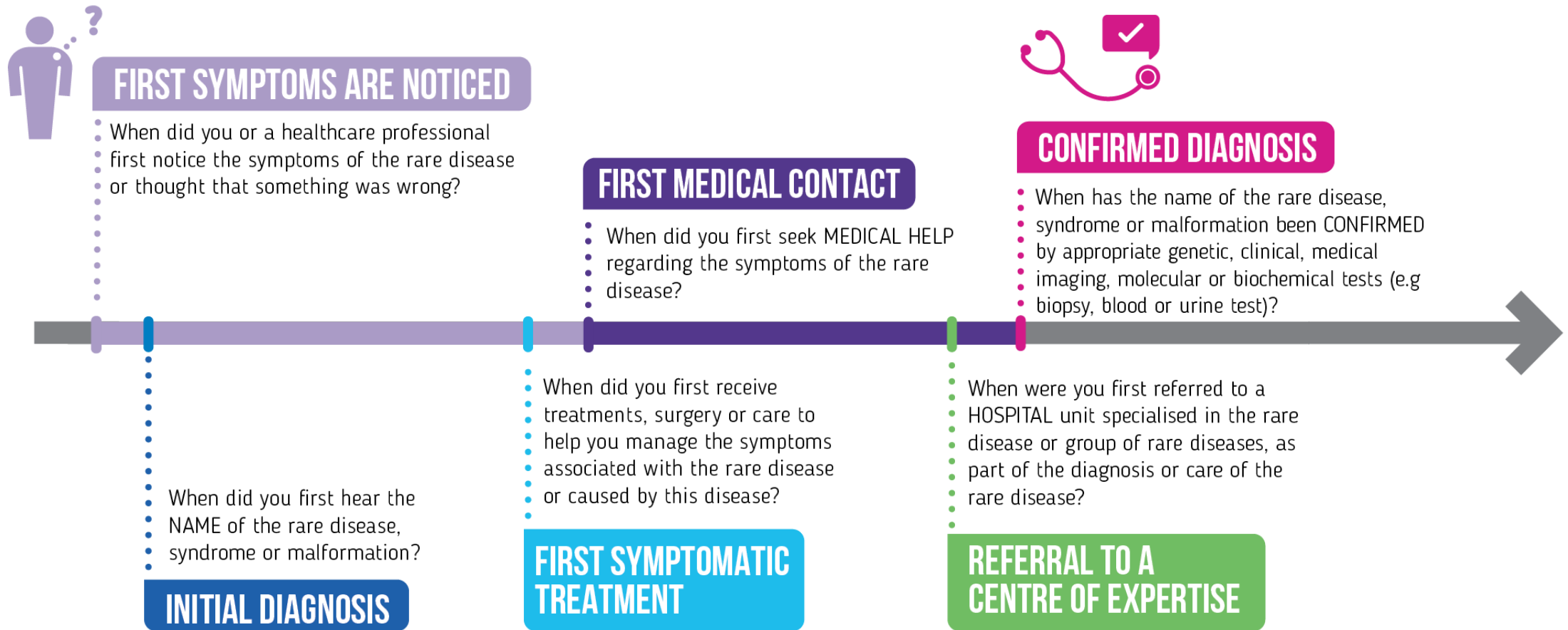


Feel free to leave your camera on or off



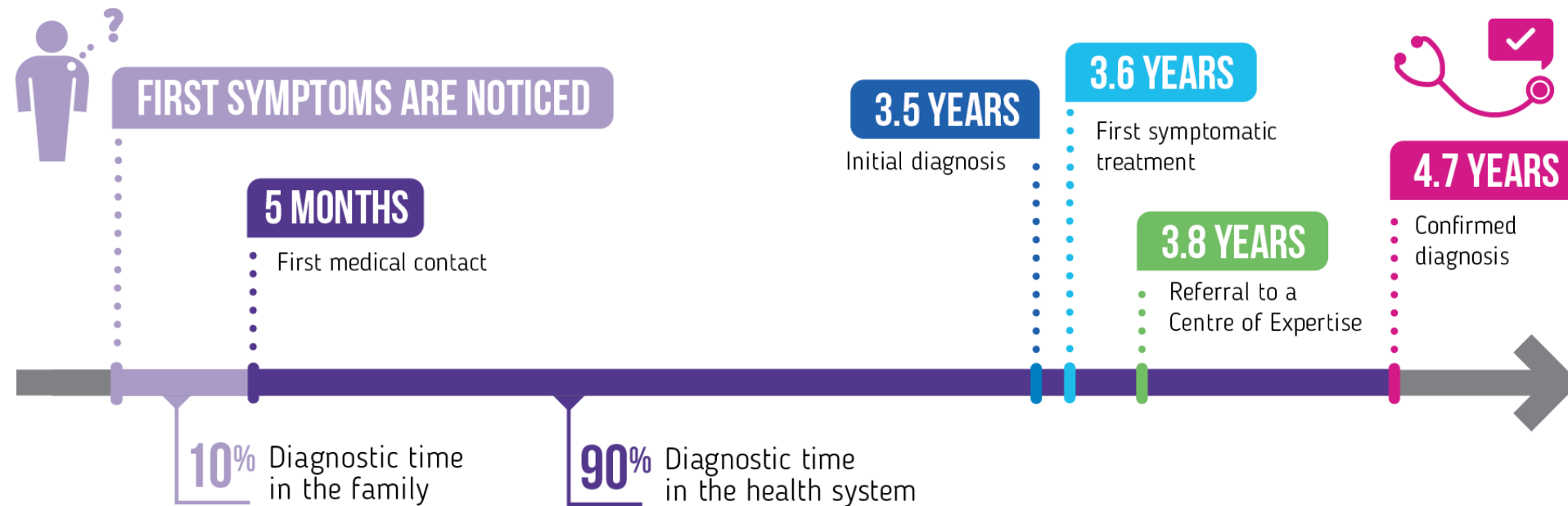
Use the chat to introduce yourself and for asking questions

THE 6 STEPS OF THE DIAGNOSIS ODYSSEY



THE FULL DIAGNOSIS ODYSSEY

Presented in factsheets



Average time for people living with a rare disease who reached each step.

A Centre of Expertise is a hospital unit specialised in the rare disease or group of rare diseases.

FACTSHEET

tiny.cc/RB_diag



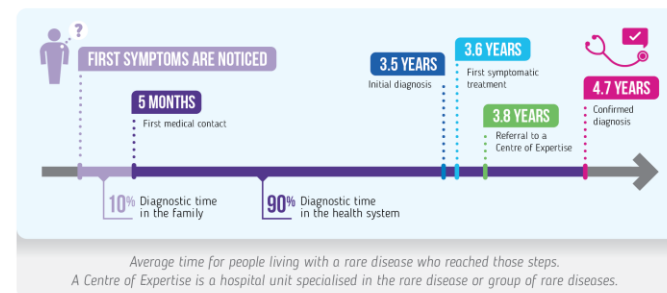
THE DIAGNOSTIC ODYSSEY OF PEOPLE LIVING WITH A RARE DISEASE



Key findings from a Rare Barometer survey
May 2024



Average time between first symptoms and further steps of the diagnostic odyssey



Difficulties associated with the diagnostic odyssey



30 MILLION
people are living with a rare disease
in Europe and 300 million worldwide



NO CURE
for the vast majority of diseases
and few treatments available



THANK YOU
to everyone who participated in the survey,
and to the Rare Barometer partners!

For more information visit eurordis.org/voices or email rare.barometer@eurordis.org
Peer-reviewed article on European results: <https://www.nature.com/articles/s41431-024-01604-z>



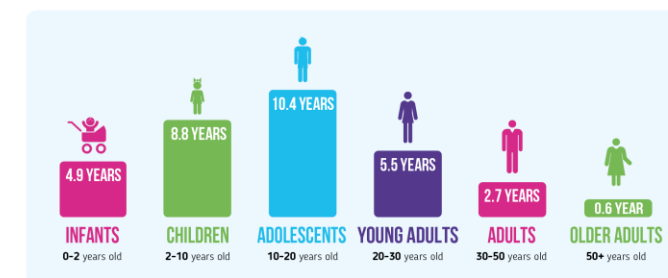
THE DIAGNOSTIC ODYSSEY OF PEOPLE LIVING WITH A RARE DISEASE



Key findings from a Rare Barometer survey
Average number of years between first symptoms and confirmed diagnosis



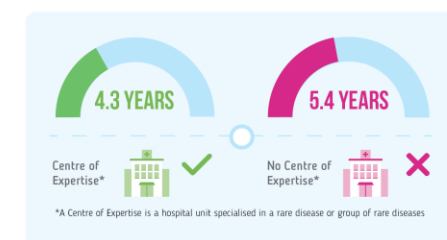
Children and adolescents have a **longer** diagnosis journey than adults



Women have a **longer** diagnosis journey than men



The diagnosis journey is **shorter** when people living with a rare disease are referred to a Centre of Expertise*



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For more information visit eurordis.org/voices or email rare.barometer@eurordis.org
Peer-reviewed article on European results: <https://www.nature.com/articles/s41431-024-01604-z>

DASHBOARD

450+ pages

tiny.cc/RB_diag

Sample information : IN_TOUCH is not among "Non-response" And CONSENT is not among "No" And EUROPE_VS_WORLD among "Europe"
Sample size 10486 responses



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).

Average number of years

Number of years it took for 25% of the sample to get to this step of their diagnosis journey

Number of years it took for 50% of the sample to get to this step of their diagnosis journey

Number of years it took for 75% of the sample to get to this step of their diagnosis journey

Lowest number of years

Highest number of years

Number of respondents for which we have the number of years to this step of the diagnosis journey

DASHBOARD

450+ pages

tiny.cc/RB_diag

1. Description of the sample
2. Average diagnosis journey (time from first symptoms to other steps of the diagnosis journey)
3. Age of the person affected when first symptoms were noticed.
4. Family members previously diagnosed with the same rare disease or not
5. Characteristics of first symptoms 1: body parts impacted, disease complexity, rare disease causes,
6. Characteristics of first symptoms (2): behavioural disorders, invisible symptoms...
7. Prevention: diagnosed before birth or at birth; family members previously diagnosed with the same rare disease.
7. Number of healthcare professionals consulted.
8. Referred to a centre of expertise.
9. Misdiagnosis (physical and psychological).
10. Diagnosis tests conducted, including questions on genetic tests.
11. Cross-border healthcare
12. Support: psychological, care coordination, financial.
13. Access to patient organisations and to other people living with the same rare disease.
14. Consequences of diagnosis.
15. Consequences of being undiagnosed.

TAILORED RESULTS

rare.barometer@eurordis.org

Collective results shared with EURORDIS members and ERNs if 30+ respondents

	European/ international results for one disease or group of diseases	Results for one country on all rare diseases	Specific results (one group of diseases in one country...)	European results for diseases of each ERN	European results
EURORDIS European & international Federations	X				X
EURORDIS National Alliances		X			X
Other EURORDIS members			X		X
European Reference Networks (ERNs)				X	X
Anyone (EURORDIS website)					X

TAILORED FACTSHEET

rare.barometer@eurordis.org



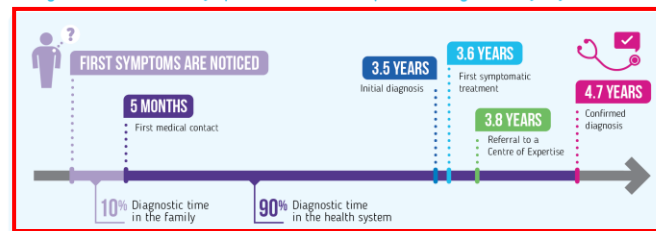
THE DIAGNOSTIC ODYSSEY OF PEOPLE LIVING WITH A RARE DISEASE



Key findings from a Rare Barometer survey
May 2024



Average time between first symptoms and further steps of the diagnostic odyssey



Average time for people living with a rare disease who reached those steps.
A Centre of Expertise is a hospital unit specialised in the rare disease or group of rare diseases.

Difficulties associated with the diagnostic odyssey



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NO CURE
for the vast majority of diseases
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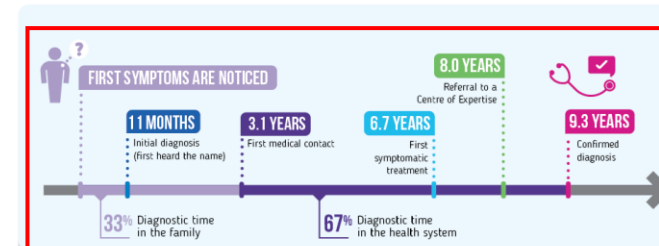
THE DIAGNOSTIC ODYSSEY OF PEOPLE LIVING WITH HHT



Key findings from a Rare Barometer survey
May 2024

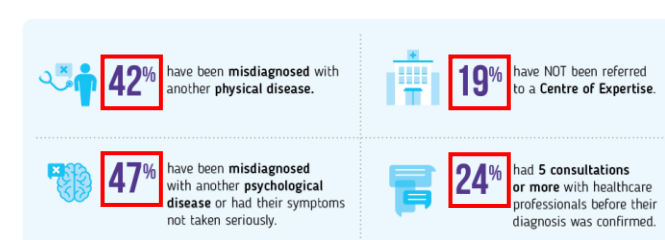


Average time between first symptoms and further steps of the diagnostic odyssey



Average time for people living with HHT who reached those steps.
*A Centre of Expertise is a hospital unit specialised in the rare disease or group of rare diseases.

Difficulties associated with the diagnostic odyssey



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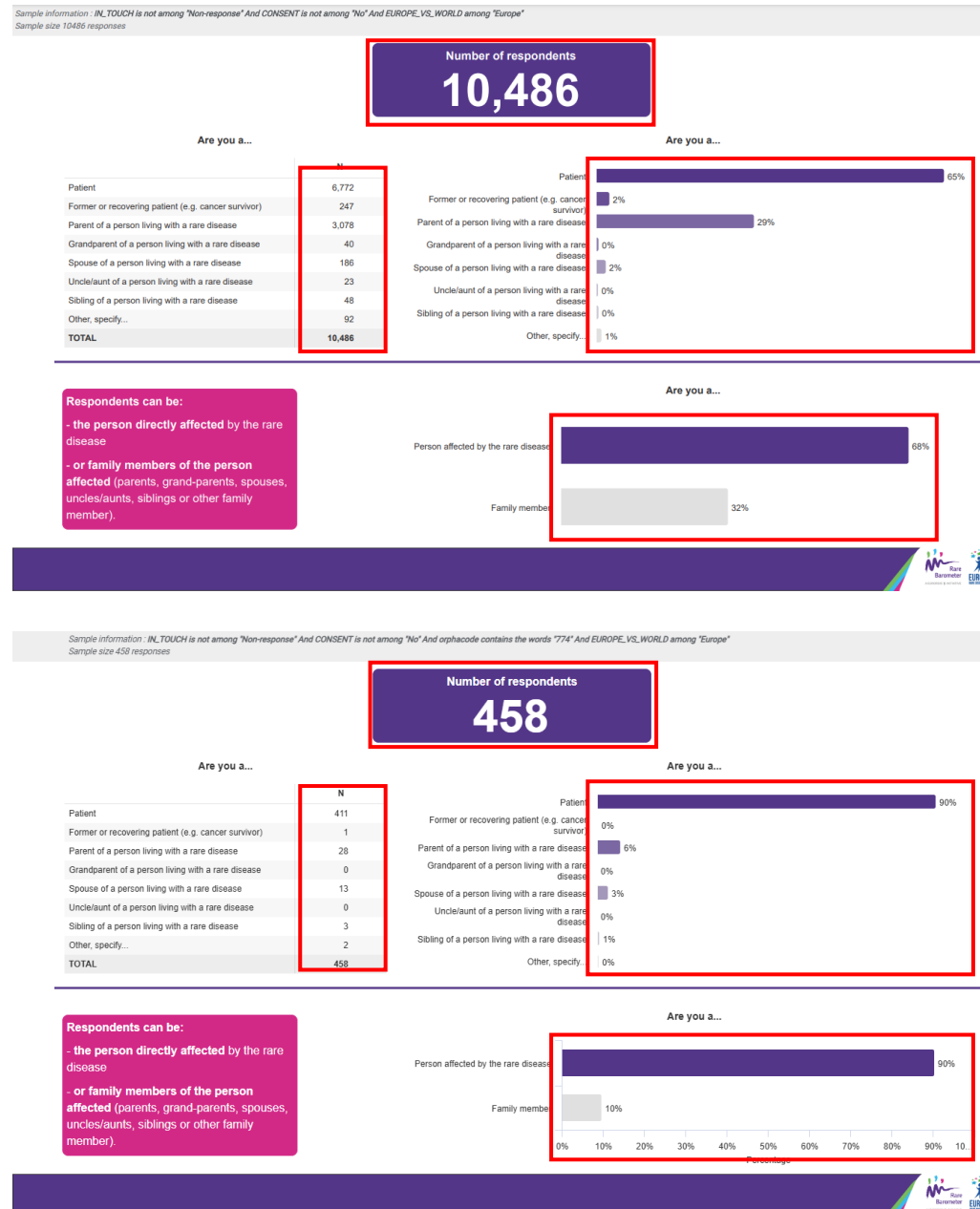


THANK YOU
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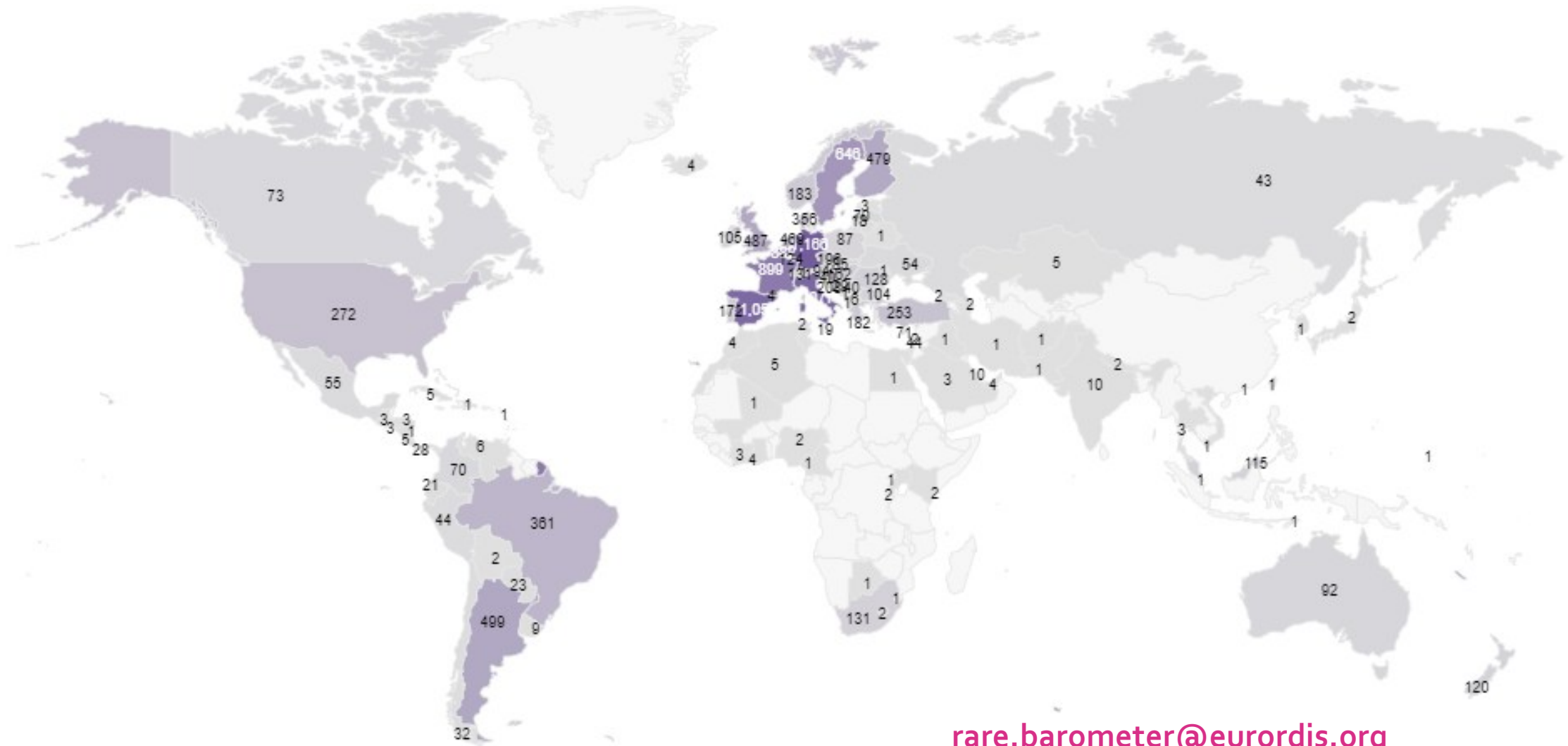
For more information visit eurordis.org/voices or email rare.barometer@eurordis.org
Peer-reviewed article on European results: <https://www.nature.com/articles/s41431-024-01604-z>

TAILORED DASHBOARD

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@eurordis.org](https://rare.barometer@eurordis.org)



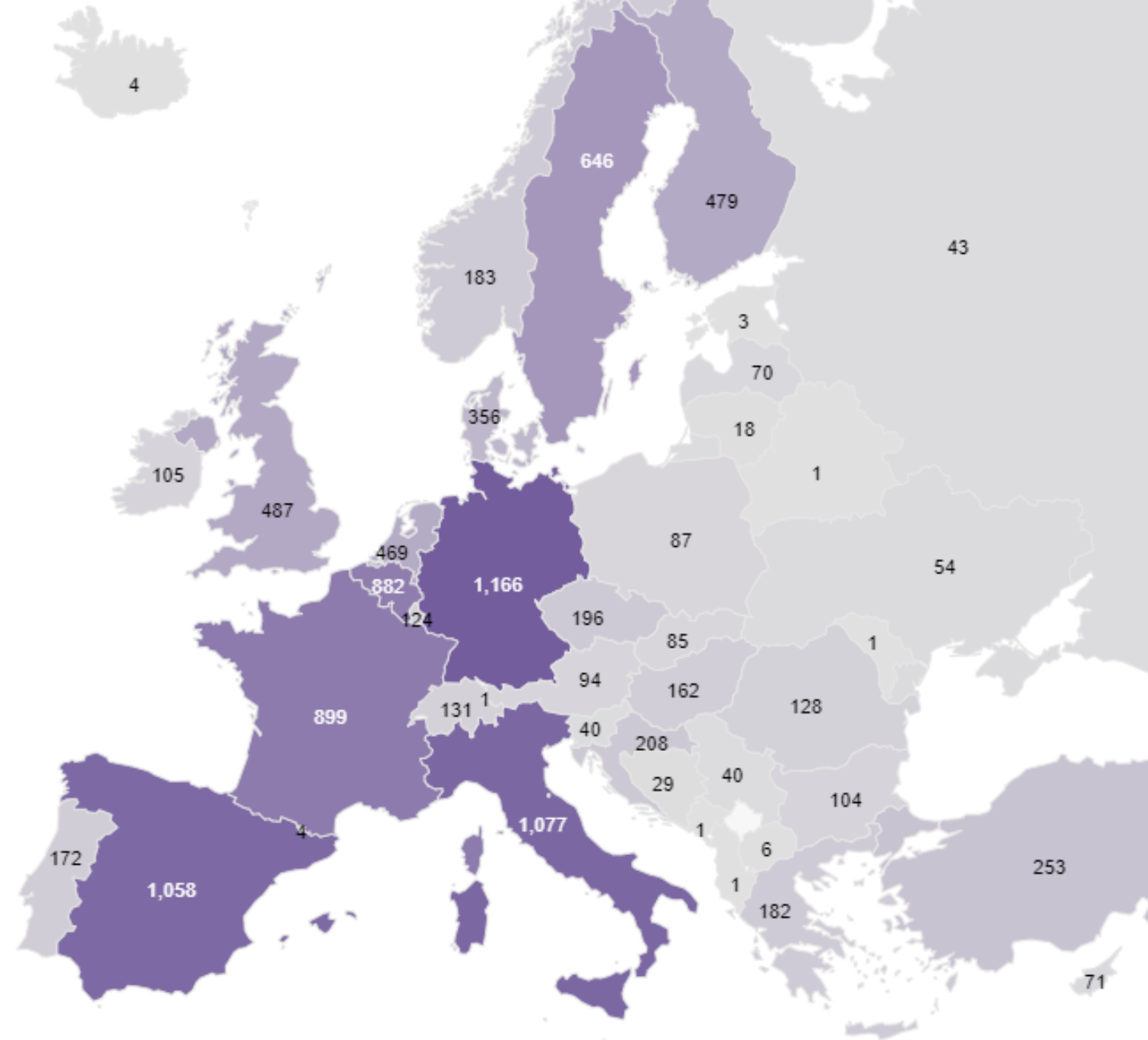
DIAGNOSIS ODYSSEY – RESPONDENTS IN THE WORLD



rare.barometer@eurordis.org

DIAGNOSIS ODYSSEY - RESPONDENTS IN EUROPE

rare.barometer@eurordis.org



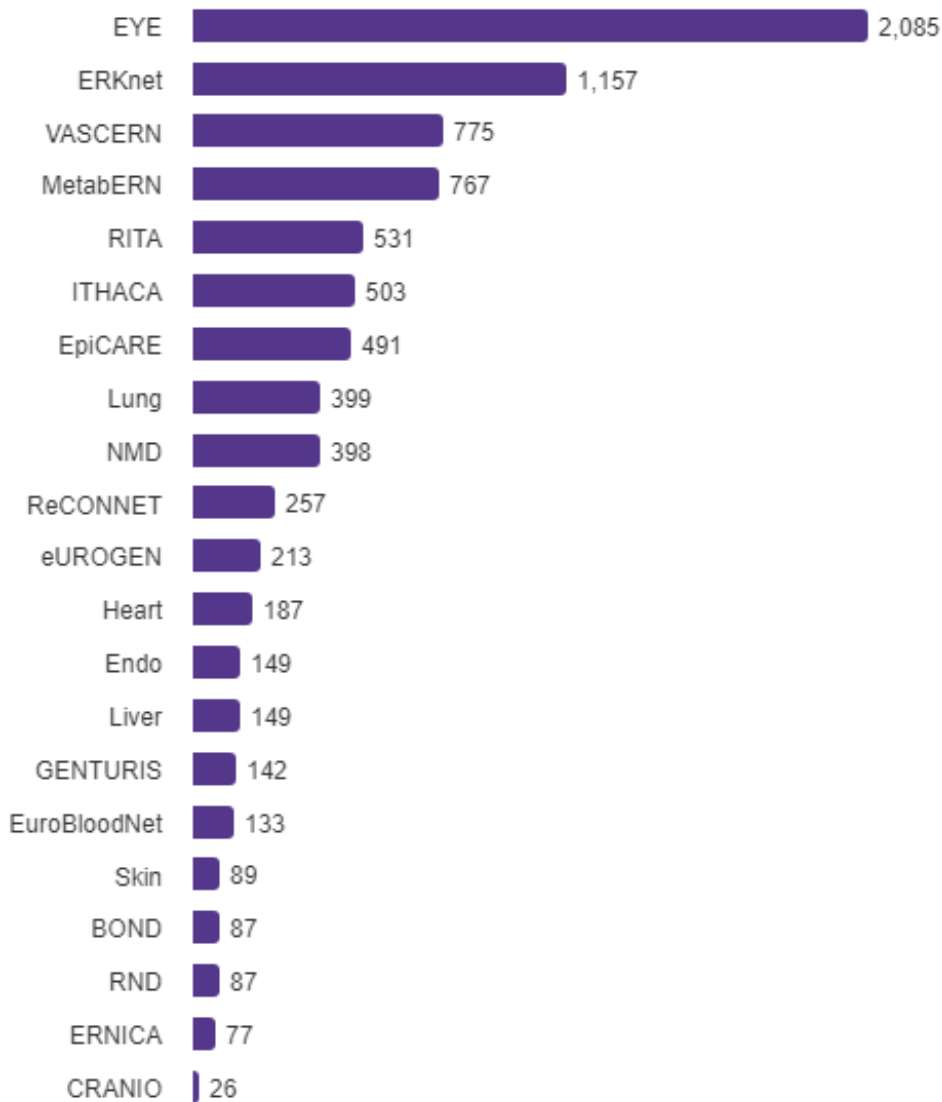
DIAGNOSIS ODYSSEY - EUROPEAN FEDERATIONS

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HHT Europe	458	European Federation for Hereditary Spastic Paraplegia	52
Federation of European Scleroderma Associations	200	European Fragile X Network	49
Sarcoidosis	178	Sclerosing Cholangitis	46
Lupus Europe	150	European Society for Phenylketonuria	45
European Myasthenia Gravis Association	139	OIFE - Osteogenesis Imperfecta Federation Europe	43
European Federation of Williams Syndrome	136	Albi France	41
CF Europe	128	Duchenne Muscular Dystrophy	41
NF Patients United	125	European Federation of Associations of Patients with Haemochromatosis	41
European Tuberous Sclerosis Complex Association	98	SMA Europe	35
PHA Europe (Pulmonary Arterial Hypertension)	86	MPS Europe	34
22Q11 Europe	80	European Idiopathic Pulmonary Fibrosis & Related Disorders Federation	32
Multinational Interstitial Cystitis Association	74	European Huntington Association	27
Marfan Europe Network	72		
Rett Syndrome Europe	65		
Perineural cyst	63		

DIAGNOSTIC ODYSSEY ERNS

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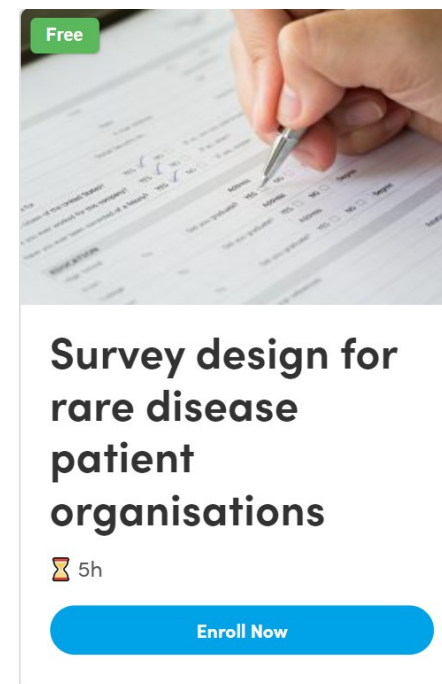
RARE BAROMETER RESOURCES TO GO FURTHER

Rare Barometer guide for EURORDIS members



tiny.cc/RBguide_members

Open Academy online course on survey design



Initiated by the ERNs
and healthcare team



Boston
Scientific
Foundation Europe



openacademy.eurordis.org

Examples of uses of the results



COMMUNICATING ON SPECIFIC DISEASE RESULTS

Myasthenia Gravis (MG)

Who: EUMGA & UCB.

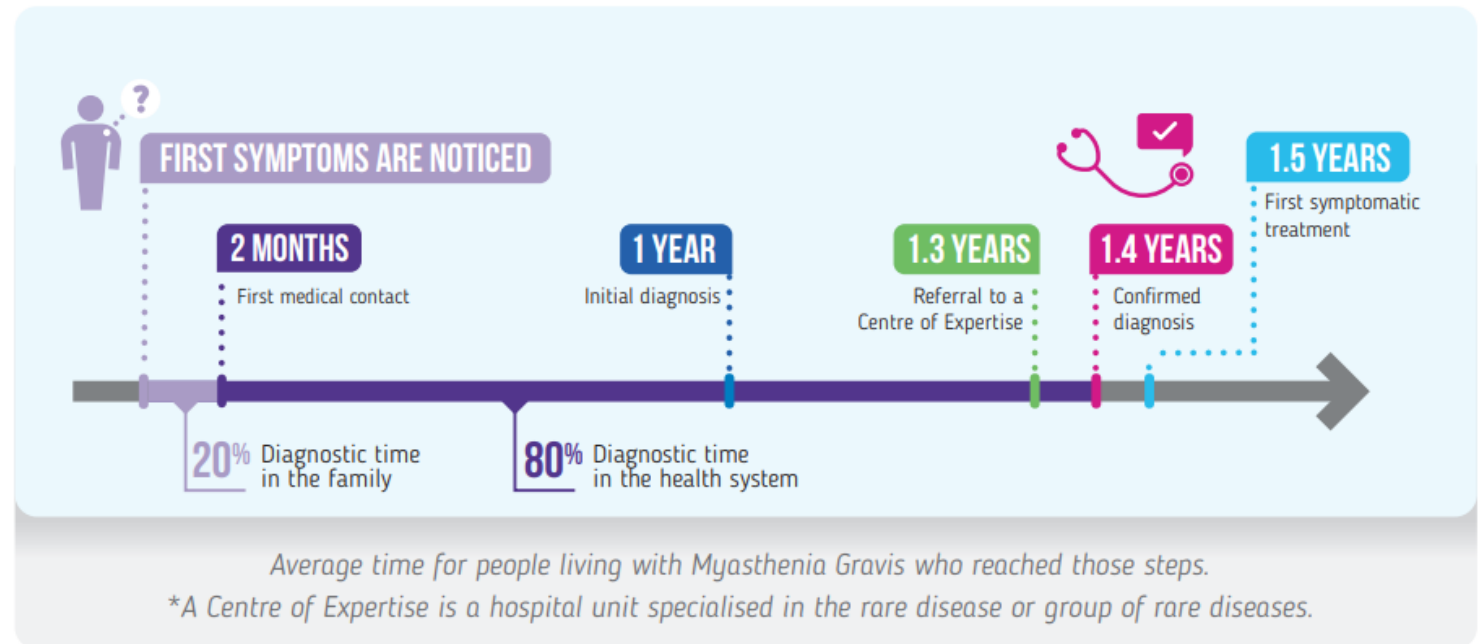
How: presenting MG results at the UCB Rare Disease Connect in Neurology conference (RDCN)

Main results: diagnostic odyssey for people living with MG

- Main results, dashboard and factsheet accessed by EUMGA
- Presented alongside scope of literature on delays in diagnosis for people living with MG by UCB



Average time between first symptoms and further steps of the diagnostic odyssey



Rare Disease Connect
in Neurology (RDCN)
2024 Annual Meeting

Welcome

14-15 November 2024

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Rare Disease Connect in Neurology



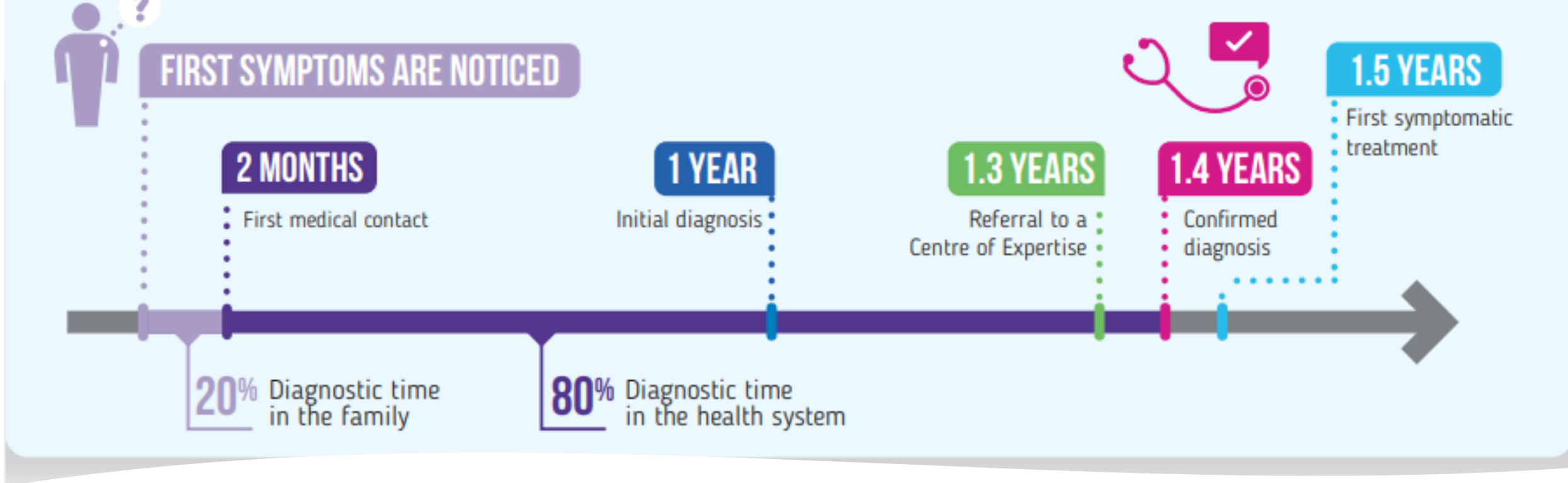
RDCN Patient Track

Over a four-day learning journey, sessions covered topics including consensus guidelines,

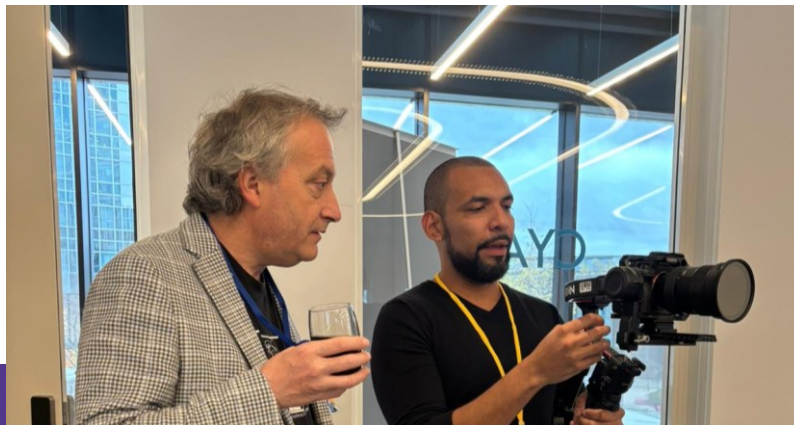
delays in

diagnosis, the importance of registries, quality of life and patient experience studies, the importance of multidisciplinary care and the role of nurses and patients in shared decision making.





- Differing perspectives between HCPs and patients?
- Gender differences
- Fluctuating symptoms resulting in difficulty to diagnose, misdiagnosis
- Delayed referrals
- Unnecessary exacerbations especially at beginning of disease
- More evidence-based awareness raising is needed for anyone working in the MG Community





SCIENTIFIC COMMUNICATION AND STRATEGIC PLANNING

HHT-Europe: Claudia Crocione

Presentation

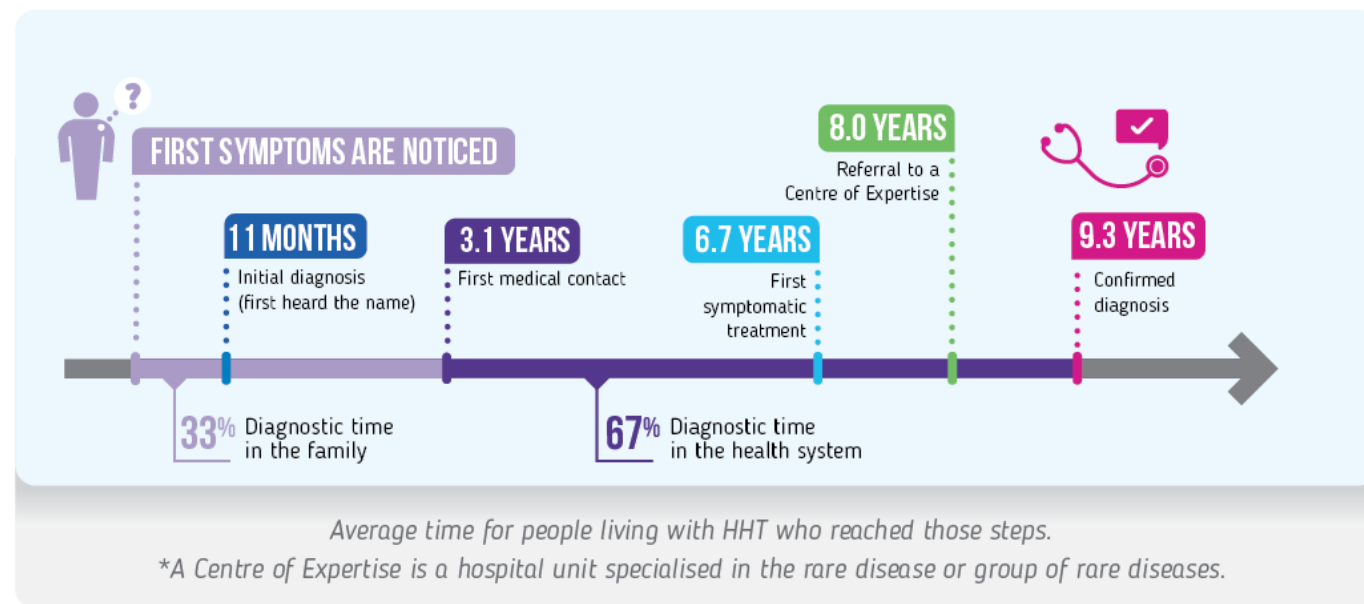
Participation: dissemination, **458** respondents in Europe & **145** in Italy.

Co-author of the EJHG article: more than 300 responses, helped analyse specific results.

Specific results for HHT: longer time to diagnosis when the condition is known in the family; those delays mostly stemmed from patient delays (diagnostic time in the family).



Average time between first symptoms and further steps of the diagnostic odyssey



RDD AWARENESS CAMPAIGN ON THE DIAGNOSIS ODYSSEY

Alliance maladies rares (France)

National campaign for Rare Disease Day:
“Let’s speed up diagnosis” (3 years)

Target: general public

Aspects addressed: several consultations and exams, ‘culture of doubt’, disease unknown or ‘it’s in your head’, 5 years, impact on daily life, isolation

Call to action:

- call the French hotline
- visit www.orpha.net

Communications material:

- Short films: 30 sec for TV; 1 min for cinema
30 sec: <https://youtu.be/ofOjF4Lk1EY>
1 min: <https://youtu.be/W3GQ9CCCBII>
- Ribbon: pharmacies, doctors, politicians... (public affairs)
- Posters (medical cabinets, pharmacies)
- Contact card of the RD hotline



Highest impact on calls to the hotline: talking about RDs and diagnostic odyssey on national TV (ex. morning shows with big audience).

RDD AWARENESS CAMPAIGN ON THE DIAGNOSIS ODYSSEY

Alliance maladies rares (France)

Film: “Let’s speed up diagnosis”

Brief: written internally

Agency: Strike

Budget:

- Film 70k€. Cheaper if reuse: contact Strike (pb@strike.paris)
- Rights to show the movie every year (France)
- Voices = employees and families

“It's always the same story. First, you sense that something is wrong. You go to the doctor. He examines us. He says ‘come back if it doesn't go away’. We come back. He prescribes tests. Nothing happens. A year passes, then two, then five. You lose your self-confidence, your social life, your job, and after years of odyssey, you identify a rare disease. It's always the same story. Let's change it together”.

1 French person in 20 lives with a rare disease. To help them, let's speed up diagnosis.

For advice, information and guidance: Call Maladies Rares Info Services. Visit www.orpha.net

<https://youtu.be/ofOjF4Lk1EY>



The Global Commission to End the Diagnostic Odyssey for Children with a Rare Disease



GLOBAL
COMMISSION

to End the Diagnostic Odyssey
for Children with a Rare Disease

Global Commission members



Yann Le Cam
 EURORDIS
RARE DISEASES EUROPE



Alaa Hamed
 sanofi



Neil Inhaber
 Takeda



Moeen AlSayed




Gareth Baynam
 Perth Children's Hospital
 IRDIRC



Kym Boycott
 CHEO
RESEARCH INSTITUTE
INSTITUT DE RECHERCHE



Pamela Gavin
 NORD
National Organization for Rare Disorders



Roberto Giugliani
 UFRGS
UNIVERSIDADE FEDERAL
DO RIO GRANDE DO SUL



Alexandra Heumber
Perry
 RARE
DISEASES
INTERNATIONAL



Kevin Huang




Derralynn Hughes
Royal Free London 
NHS Foundation Trust



Maryam Mohd.
Fatima Matar



Dau-Ming Niu



Anne O'Donnell-Luria



Carmencita Padilla



Mike Porath



Marshall Summar



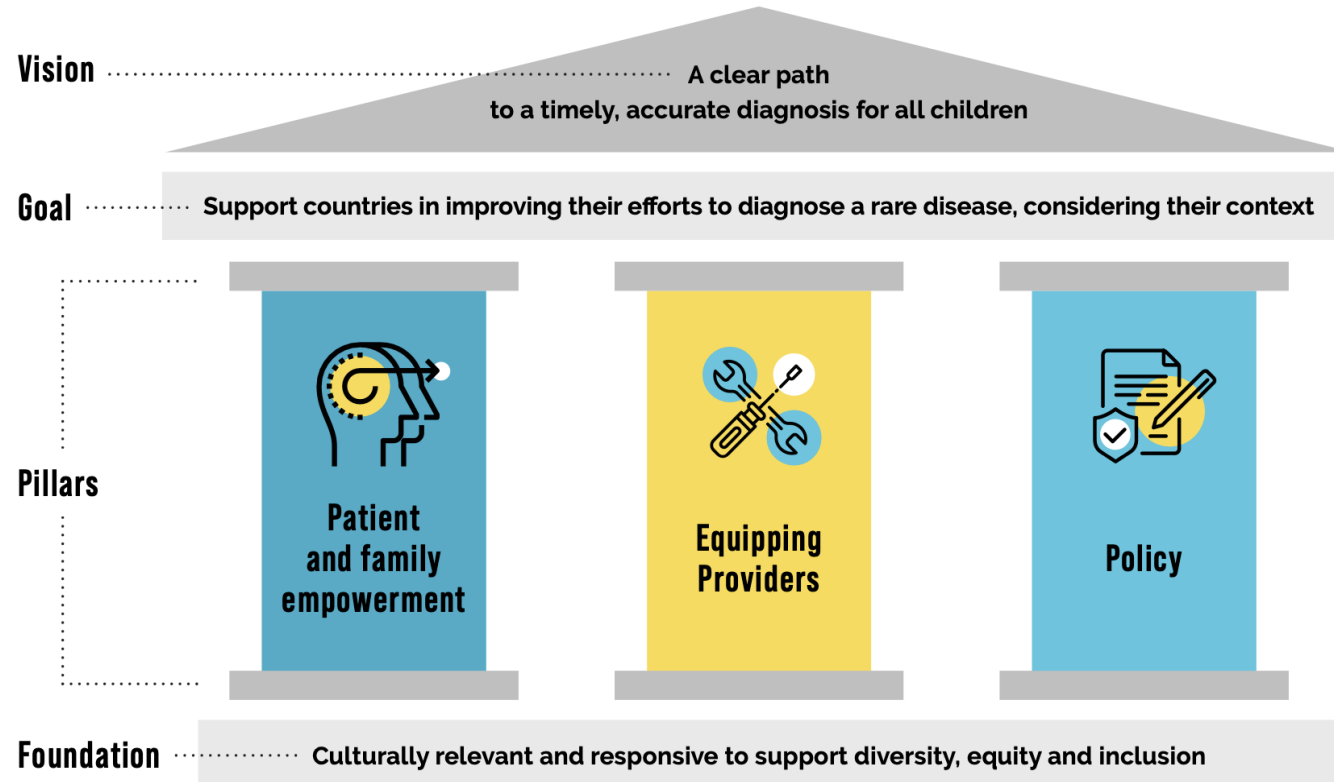
Durhane Wong-Rieger



Samuel Agyei Wiafe



Our vision, mission and framework



MISSION

Harness the power of innovation, advances in genetics and motivation of families to accelerate the time to diagnose a rare disease for children around the world



Amplifying innovations in diagnosis: 10 case studies

Exemplars from eight different countries, two global and one regional effort



Case Study	Pillar
Casa Dos Raros (Brazil)	Patient & Family Empowerment
The Global Nursing Network for Rare Diseases (Global)	Equipping Providers
New Zealand's First Rare Disease Strategy (New Zealand)	Policy
UTOPIA (Australia & Singapore)	Equipping Providers
DxGPT (Global)	Patient & Family Empowerment Equipping Providers
Newborn Screening Program and Rare Disease Act (Philippines)	Policy
Ghana Diagnostic Access Program (Ghana)	Patient & Family Empowerment
Nanbyo Act (Japan)	Policy
Africa Rare Disease Initiative (Democratic Republic of Congo)	Equipping Providers
Undiagnosed Hackathon (Europe)	Patient & Family Empowerment

Casa dos Raros: Brazil

Significantly **reduced the time to diagnose** a patient with a rare disease to an **average of 58 days from first contact** with the Center.

This timeline is considerably shorter than the **national average of 5.4 years for patients to receive a diagnosis** and much faster than the average two-year wait for a first consultation with rare disease specialists

FRAMEWORK PILLAR: Patient & Family Empowerment

CASE STUDY: *Casa dos Raros*, Brazil



A new integrated diagnostic and treatment center in Brazil that has significantly reduced the time to diagnose a rare disease.

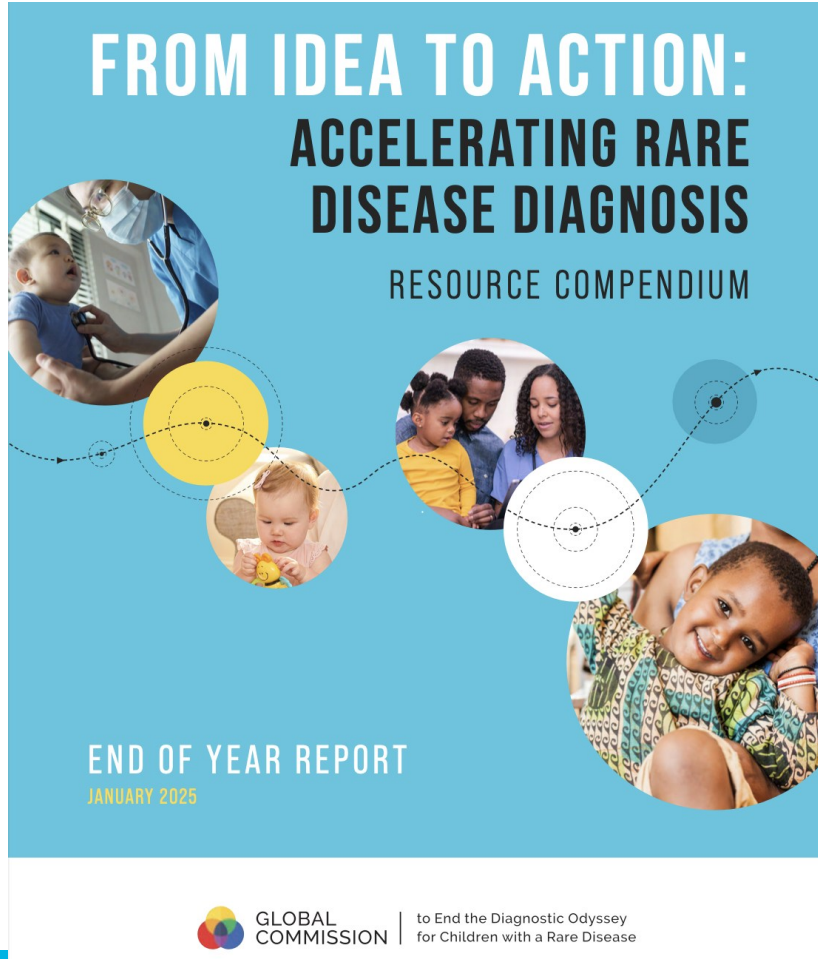
Context

It takes an average of 5.4 years for someone with a rare disease in Brazil to finally be diagnosed after first presenting symptoms.¹ *Casa dos Raros* ("House of Rares") is a new, comprehensive rare disease center in Porto Alegre that has succeeded in diagnosing patients in an average of 58 days from first contact while the average wait time for a first consultation with a rare disease reference center affiliated with the public health system is around two years.

Casa dos Raros was established to accelerate diagnosis, facilitate research, develop educational materials for healthcare teams, and improve the accessibility of treatment for people living with a rare disease. Specifically, it seeks to expand the reach of the rare disease reference centers affiliated with the public health system, which offer clinical care, lab testing, and counselling, but have long wait times and have been unable to fully meet the demand of individuals and families seeking a timely diagnosis.



Call to action



Please send us examples of compelling innovations in diagnosis at info@globalrarediseasecommission.com

Learn more about our work and read our case studies: <https://globalrarediseasecommission.com/>

Look out for our forthcoming report in early February 2025



[Follow us on LinkedIn](#)



[Check out our website](#)

Thank you!

Toon Digneffe, Head of Public Affairs & Public Policy,
Takeda, Toon.Digneffe@takeda.com

Danielle Dong, Scientific Advocacy Lead Global Medical
Affairs, Rare Disease, Sanofi, Danielle.Dong@sanofi.com

The Global Commission to End the Diagnostic Odyssey for Children with a Rare Disease is an association composed of a diverse group of organizations and individuals committed to shortening the time for an accurate diagnosis for all children with rare diseases. The views expressed herein are solely those of the Commission and do not represent any individual, co-chair, or member organization's views or opinions.

EU Policy Event to mark Rare Disease Day

5 March 2025, 13:30–15:30

European Parliament, Brussels

**"Impact of Rare Diseases:
More Than You Can Imagine"**

Hosted by MEP Stine Bosse & EURORDIS

Opportunity to:

1. Delve into the impact of rare diseases.
2. Explore the challenges faced by people living with rare diseases and their families.
3. Discuss concrete EU-level solutions.



EU Policy Event to mark Rare Disease Day

Opportunity to use results of several Rare Barometer Surveys, to:

1. Build a robust narrative of the journey of people with rare diseases.
2. Bringing policy maker's attention to the issues that EU policy and initiatives can help solve.

Using figures on:

1. Time to diagnosis.
2. Limited treatments.
3. Daily life with a rare disease e.g. disability, employment, mental health.



**IMPACT OF RARE DISEASES:
MORE THAN
YOU CAN IMAGINE**

RESULTS OF THE 'DAILY LIFE' SURVEY

tiny.cc/daily_life_RD_webinar

February 13th

10 JULY> 8 SEPTEMBER 2024

10,478 respondents worldwide and 9,591 in Europe

25 languages



TARGET POPULATION

All patients living with a rare disease and their family members, including unsolved cases (undiagnosed)

92 countries

1,754 rare diseases represented



THANK YOU!

To the Rare Barometer participants,
partners and corporate donors in 2024

rare.barometer@eurordis.org



Co-funded by
the Health Programme
of the European Union

