THE RARE BAROMETER PROGRAMME

EURORDIS' survey initiative to support evidence-based advocacy and actions

Jessie Dubief, Social Research Director, Rare Barometer lead

EURORDIS-RARE DISEASES EUROPE

eurordis.org

A unique alliance of rare disease patient organisations

1000+

MEMBER PATIENT ORGANISATIONS

OUTREACH TO OVER

2500

PATIENT GROUPS

57

NATIONAL ALLIANCES OF RARE DISEASE PATIENT ORGANISATIONS

77

EUROPEAN & INTERNATIONAL FEDERATIONS OF SPECIFIC RARE DISEASES

75

COUNTRIES (27 EU COUNTRIES)

FOUNDED IN

1997

35+

TEAM MEMBERS, WITH OFFICES IN PARIS, BRUSSELS, BARCELONA

EURORDIS-RARE DISEASES EUROPE

Our vision

EURORDIS' vision is a world where all people living with a rare disease can have longer and better lives and can achieve their full potential, in a society that values their well-being and leaves no one behind.

Eddison, xeroderma pigmentosum

THE RARE BAROMETER PROGRAMME

eurordis.org/voices

EURORDIS' survey initiative to support evidence-based advocacy and actions

Robustly collects experiences and opinions of people living with a rare disease and their close family members, on topics that directly affect them.

Transforms those experiences and opinions into facts and figures to feed the advocacy work of the rare disease community.

WHAT

Not-for-profit initiative

WHO

Run independently by EURORDIS-Rare Diseases Europe

WHEN

Created in 2016

WHY

Evidence-based advocacy

THE RARE BAROMETER PROGRAMME

eurordis.org/voices

EURORDIS' survey initiative to support evidence-based advocacy and actions

Surveys

People living with a rare disease & family members

1-3 studies per year

25 languages

Worldwide

Up to 13,000 respondents to our surveys

Panel

23,000+
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

GDPR compliant: information is only accessible to the Rare Barometer team, saved on secured servers in France

SURVEY TOPICS

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Topic	Publication date	World participants
Juggling care and daily life: the balancing act of the rare disease community	May 2017	3017
Access to treatment: unequal care for European rare disease patients	February 2017	1350
Rare disease patients' participation in research	February 2018	3213
Data sharing and data protection	July 2019	2013
Impact of COVID-19	October 2020	8552
The rare disease community's experience of medical care	January 2021	3905
Rare disease patients' opinion on the future of rare diseases	May 2021	3998
The diagnosis odyssey of people living with a rare disease	May 2024	13 304
Voices on newborn screening: the opinion of people living with a rare disease	April 2024	6179
The impact of living with a rare disease: barriers and enablers of independent living with social participation	February 2025	10 552
The mental wellbeing of people living with a rare disease and their family	*ongoing*	*ongoing*

RARE BAROMETER RESULTS

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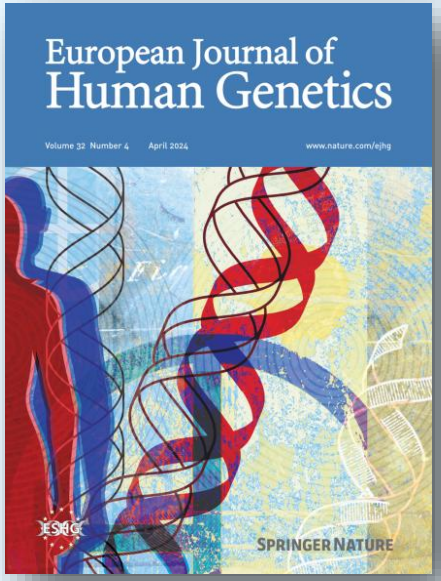
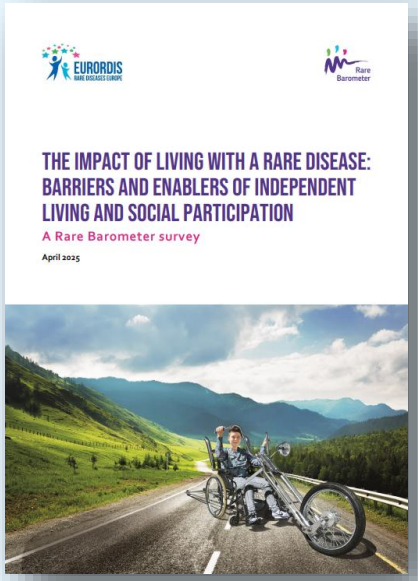
PEER-REVIEWED ARTICLES



REPORT

30-40 pages
Only available in English

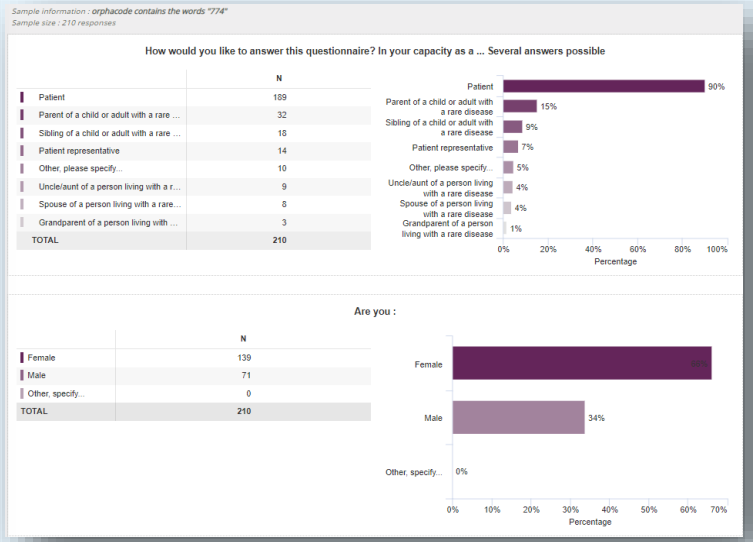


FACTSHEET

2 pages – 25 languages

DASHBOARD

Each question of the questionnaire
Frequency and percentages
25 languages



MORE DETAILED

MORE VISUAL

EURORDIS RARE DISEASES EUROPE

Rare Barometer
A EURORDIS INITIATIVE

TAILORED RESULTS FOR EURORDIS MEMBERS & ERNS

FACTSHEET
2 pager
25 languages

Tailored for EURORDIS members & ERNs with 30+ participants:
per country, geographical region, disease, group of diseases

Specific key results

EURORDIS RARE DISEASES EUROPE

RECONNAISSEZ LES HANDICAPS ET LES OBSTACLES !

Rare Barometer

Principaux résultats d'une enquête Rare Barometer sur l'impact de vivre avec une maladie rare

Février 2025

10 Juillet 8 Sept. 2024

465 répondants en Belgique

256 maladies rares représentées

1 LA GRANDE MAJORITÉ DES PERSONNES AYANT UNE MALADIE RARE VIVENT AVEC UN HANDICAP...

9/10 personnes ayant une maladie rare vivent avec un handicap

Q Washington Group Short Set on Functioning (WG-SS) : 94% des participants avaient « quelques difficultés », « beaucoup de difficultés » ou « ne pouvaient pas du tout » voir, entendre, marcher, se souvenir/se concentrer, prendre soin d'eux-mêmes (s'habiller ou se laver) ou communiquer ; Global Activity Limitation Index (GALI) : 90% des participants étaient « limités » ou « fortement limités » dans leurs activités habituelles en raison d'un problème de santé au cours des 6 derniers mois ou plus ; auto-identification : 91% des participants considéraient vivre avec un handicap visible, un handicap invisible ou les deux à la fois. Tous les participants (n=465).

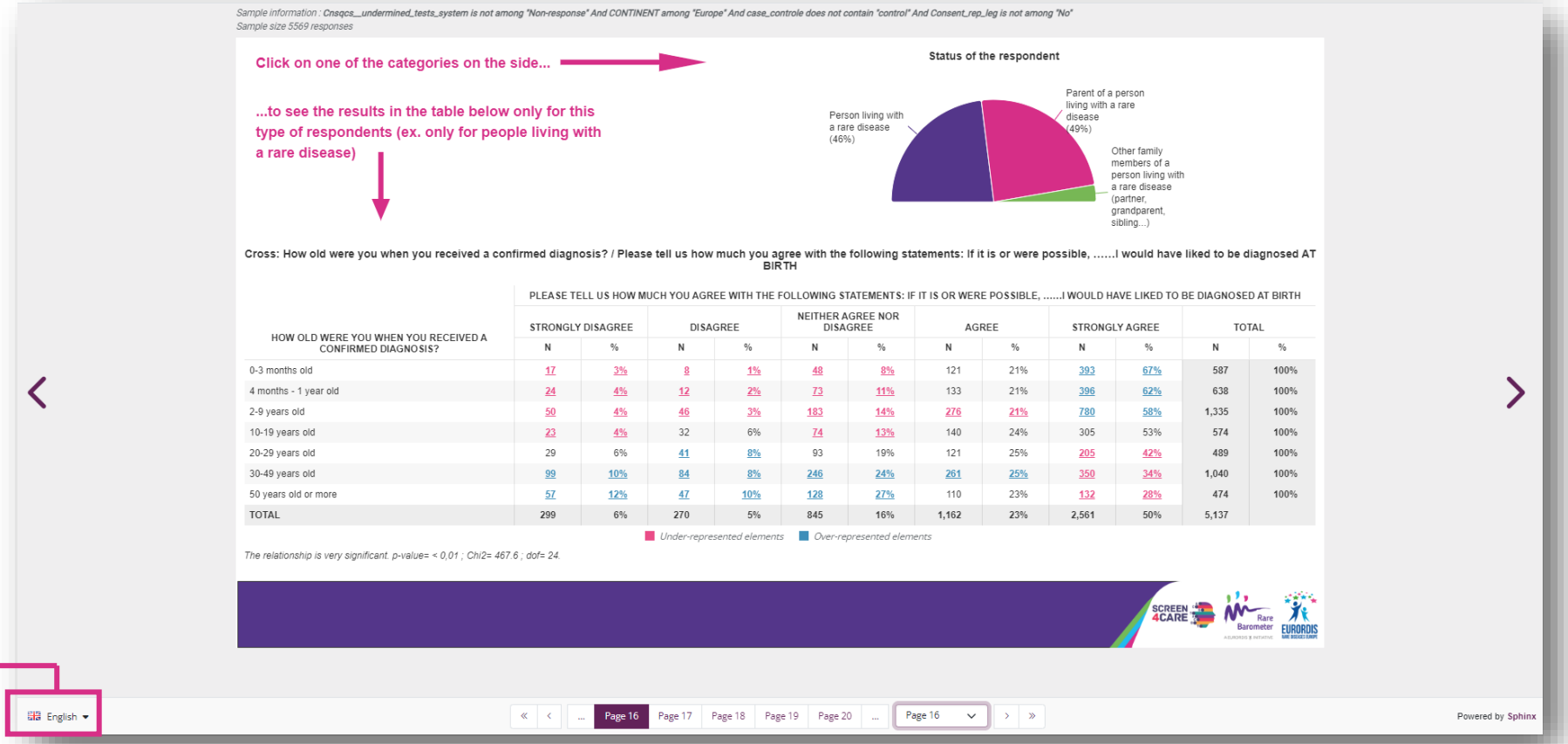
TAILORED RESULTS FOR EURORDIS MEMBERS & ERNS

ONLINE DASHBOARD

Each question of the questionnaire
Descriptive statistics

Tailored for EURORDIS members & ERNs with 30+ participants:
per country, geographical region, disease, group of diseases

25 languages



TAILORED RESULTS

eurordis.org/who-we-are/our-members

Collective results shared with EURORDIS members and ERNs with 30+ participants in their community

	European or international results for one disease or group of diseases	Results for all rare diseases in one country	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

INTERESTED IN SPECIFIC RESULTS?

Not a patient organisation:

- **Write to** rare.barometer@eurordis.org to know if the community you are interested in had 30+ participants in one of our surveys and get the contact of the patient organisation that received the results.
- **Reach out to the EURORDIS member representing the community you are interested in** (country, disease, group of diseases): if they have 30+ participants in their community, they have received their results and can decide to share them with you. eurordis.org/who-we-are/our-members

For patient organisations

If you are a **EURORDIS member** AND there are **30+ participants** in your community, you can access your tailored survey results!
EURORDIS membership: www.eurordis.org/get-involved/become-a-member/

1

STORY IDEA:
THE DIAGNOSTIC ODYSSEY

THE DIAGNOSTIC ODYSSEY OF PEOPLE WITH RARE DISEASES

17 MARCH ⇢ 15 JUNE 2022

13,304 respondents worldwide and **10,486** in Europe

27 languages

104 countries

1900+ diseases represented

TARGET POPULATION

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

RARE DISEASES INTERNATIONAL

GLOBAL COMMISSION
to End the Diagnostic Odyssey for Children with a Rare Disease

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THE DIAGNOSIS ODYSSEY

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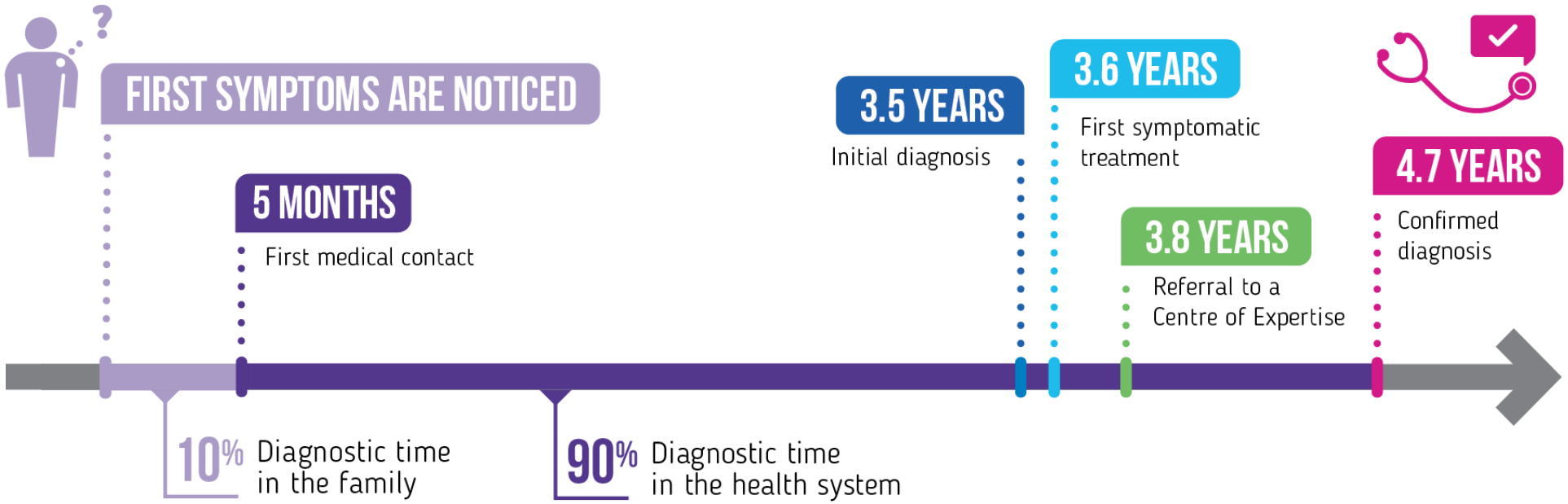
Most of the diagnostic time stems from health systems



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.

KEY RESULTS

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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 centres of expertise

More holistic care

Better public awareness of rare diseases could help improve social acceptance

KEY RESULTS

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Additional difficulties are encountered by some people with rare diseases:

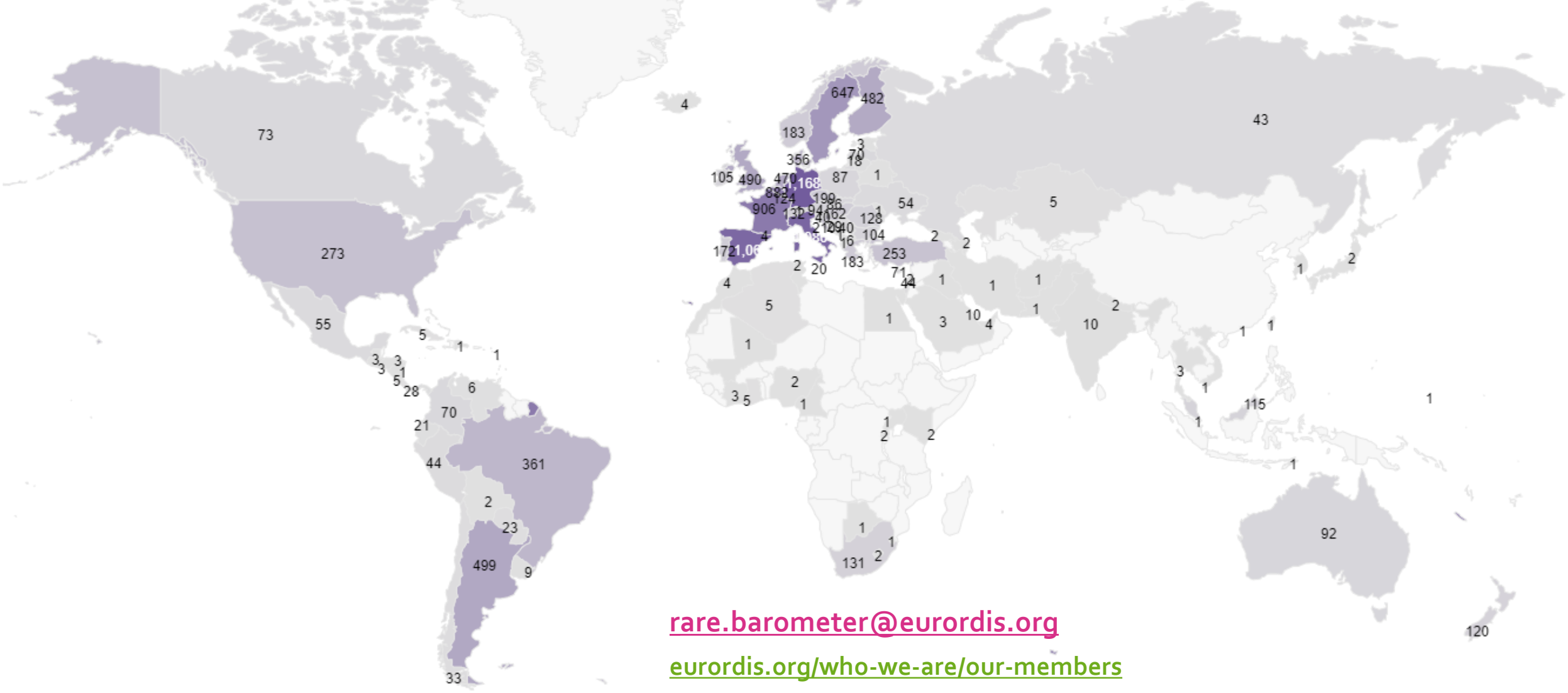
Children and adolescents (9-10 years to diagnosis), both from patient delays (1.5-3 years) & health system delays (7 years)	>	Improve public and healthcare professionals' awareness of rare diseases among children and adolescents.
Women (+1.7 year), mostly from health systems	>	Considering symptoms of girls and women
People with rare genetic disorders (access to genetic tests)	>	Improve access to effective and available diagnostic testing technologies.
Psychological support (+1.3 year), mostly impact on health system delays	>	Improve access to psychological support for undiagnosed patients.

DIAGNOSIS ODYSSEY - PARTICIPANTS IN EUROPE

rare.barometer@eurordis.org

eurordis.org/who-we-are/our-members

DIAGNOSIS ODYSSEY – PARTICIPANTS IN THE WORLD



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DIAGNOSIS ODYSSEY – PARTICIPANTS EUROPEAN FEDERATIONS

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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
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
European Huntington Association	27

DIAGNOSIS ODYSSEY – REFERENCES

rare.barometer@eurordis.org

Press release: Major survey reveals lengthy diagnostic delays for rare disease patients, 16 May 2024. eurordis.org/survey-reveals-lengthy-diagnostic-delays/

Press: EURACTIV, Study reveals long diagnostic delays for rare diseases in Europe, 14 May 2024. euractiv.com/news/study-reveals-long-diagnostic-delays-for-rare-diseases-in-europe/

Press: EURACTIV, Genetic diagnostic technology is a game changer for rare diseases but ethics concerns linger, 21 May 2024. euractiv.com/news/genetic-diagnostic-technology-a-game-changer-for-rare-diseases-but-ethics-concerns-linger/

Peer-reviewed paper: Faye, F., Crocione, C., Anido de Peña, R. et al. Time to diagnosis and determinants of diagnostic delays of people living with a rare disease: results of a Rare Barometer retrospective patient survey. Eur J Hum Genet 32, 1116–1126 (2024). doi.org/10.1038/s41431-024-01604-z

Factsheet key results: EURORDIS, The diagnosis odyssey of people living with a rare disease. Key findings of a Rare Barometer survey. 2024. tiny.cc/RB_diag

Webinar: Rare Barometer Diagnosis Survey Results – 30 January 2025. youtube.com/watch?v=DyO-l7B7LqM

Podcast: Rare on Air. Ayça Sahin's story and what our survey reveals about long diagnostic journeys. eurordis.org/rare-on-air/

More on rare disease diagnosis: EURORDIS. Earlier, faster and more accurate diagnosis. eurordis.org/our-priorities/diagnosis/

2

STORY IDEA: IMPACT OF RARE DISEASES ON DAILY LIFE

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THE IMPACT OF LIVING WITH A RARE DISEASE ON DAILY LIFE

10 JULY ⇢ 8 SEPTEMBER 2024

10,478 respondents worldwide and **9,591** in Europe

25 languages

92 countries

1,754 rare diseases represented

TARGET POPULATION

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

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MOST PEOPLE WITH RARE DISEASES LIVE WITH DISABILITIES

8/10 people with rare diseases live with disabilities

Q *WG-SS: 87% of the participants had 'some difficulties', 'a lot of difficulties' or 'could not at all' see, hear, walk, remember/concentrate, selfcare (dressing or washing over) or communicate; GALI: 83% of the participants were limited or severely limited in performing activities that people usually do because of a health problem during the last 6 or more months; Self-identification: 88% of the participants considered themselves as a person with a visible disability, an invisible disability or both. All participants (n=9591).*

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THEIR DISABILITIES ARE DIVERSE AND COMPLEX

Often time, they live with multiple disabilities

They can have difficulties in:

72% at least **2 activities**

53% at least **3 activities**

35% at least **4 activities**

Percentage of people with rare diseases who had 'some difficulties', 'a lot of difficulties' or 'could not do at all' in at least 2 domains, at least 3 domains or at least 4 domains of the WGSS - All participants (n=9591).

SOCIAL PARTICIPATION

Unemployment of people with rare diseases is higher than in the general population

23% of people with rare diseases are **unemployed**

Comparison: the unemployment rate in the general population of the European Union was **6.1%** in 2023[1].

Percentage of participants aged 16-64 who answered 'Unemployed' or 'Cannot work because of a disease' to 'What is your current situation?' (n=5332).

[1]Unemployment rates by sex, age and citizenship, Eurostat (https://ec.europa.eu/eurostat/databrowser/view/lfsa_urgan__custom_15225487/default/table?lang=en), consulted February 3rd 2025

DAILY LIFE - PARTICIPANTS IN EUROPE

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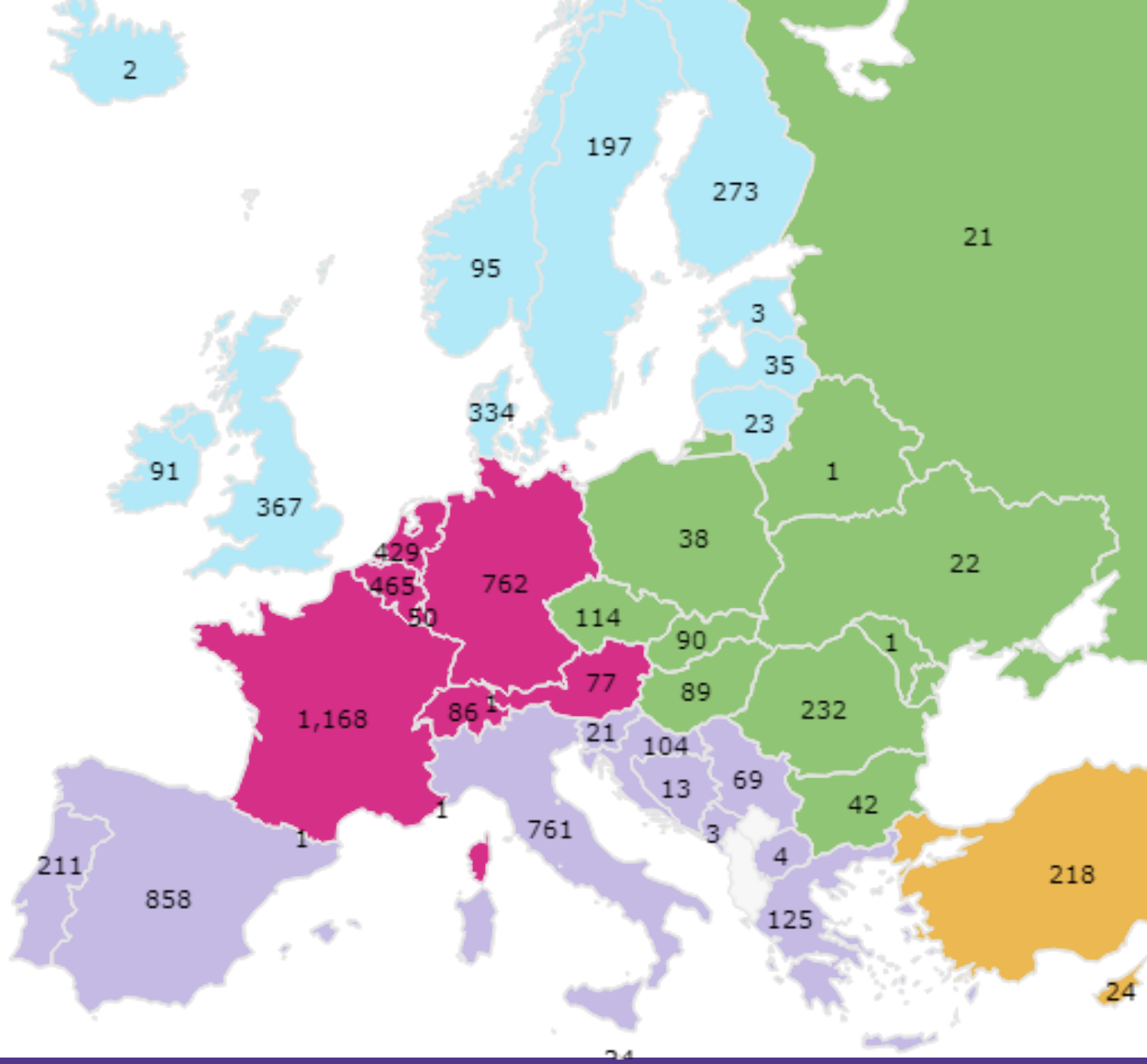
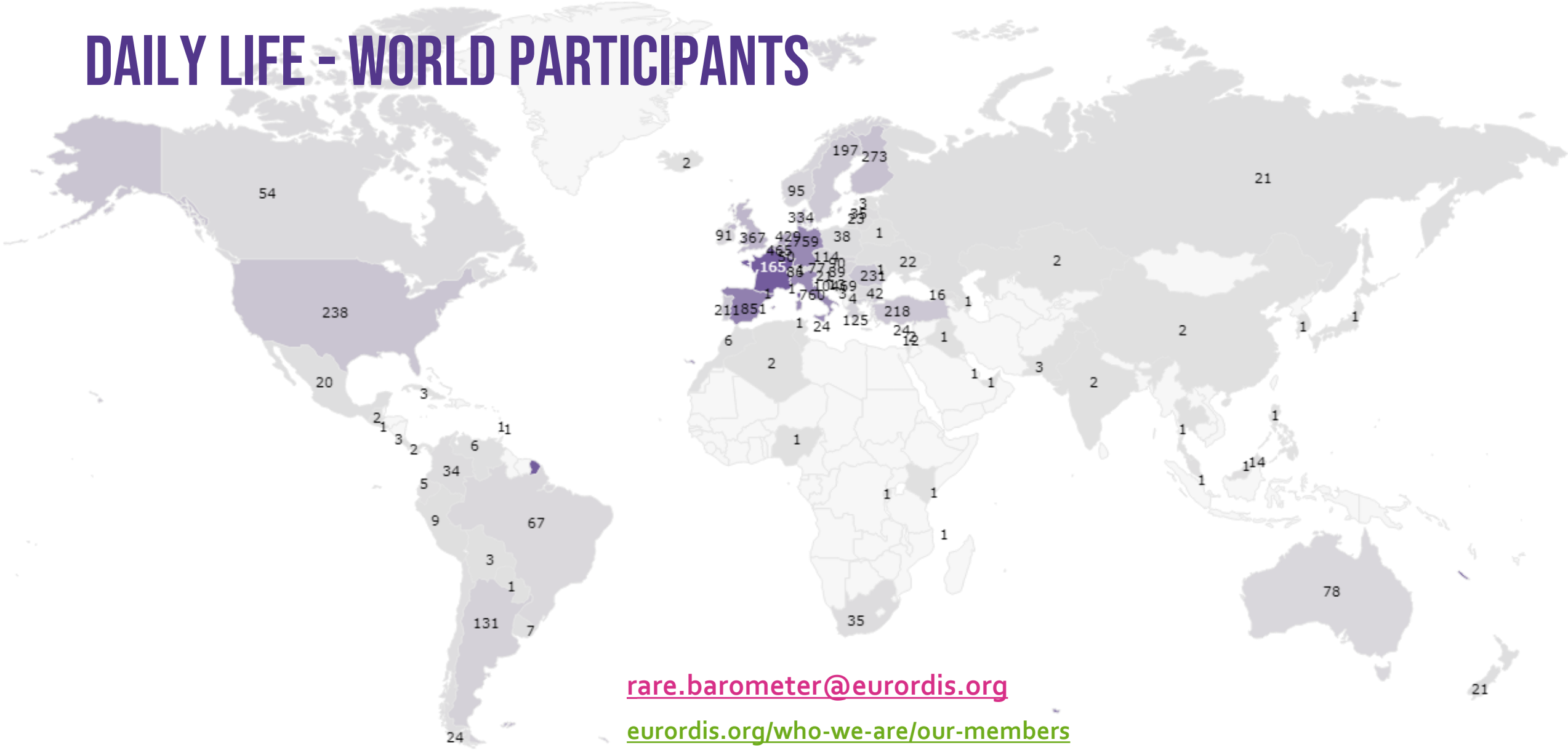
eurordis.org/who-we-are/our-members



DAILY LIFE - WORLD PARTICIPANTS



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DAILY LIFE – FEDERATIONS WITH 30+ RESPONDENTS

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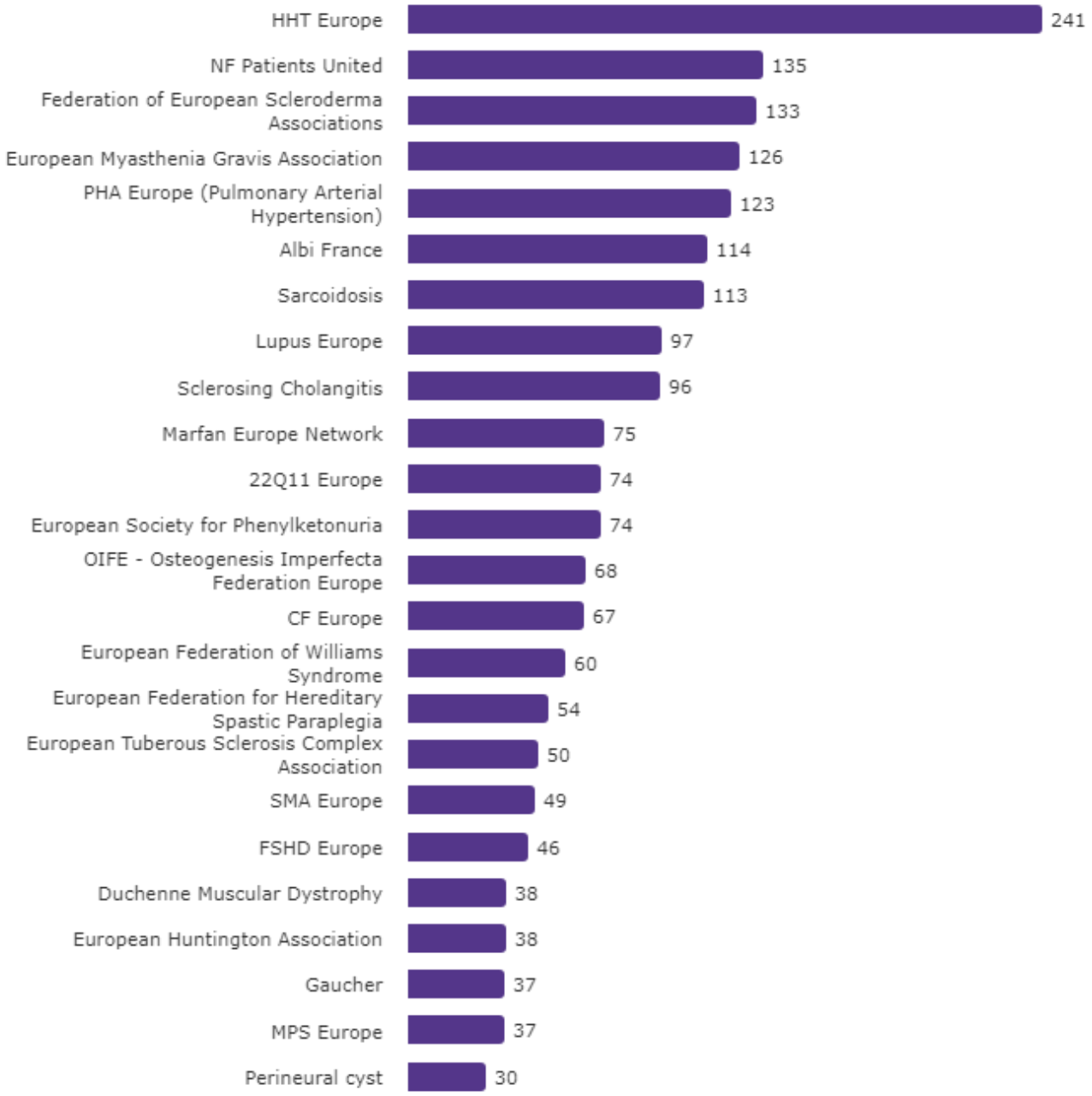

DAILY LIFE – REFERENCES

rare.barometer@eurordis.org

Press release: EURORDIS Rare Barometer finds major barriers to disability recognition and social participation. 13 February 2025. eurordis.org/rare-barometer-findings-daily-life-survey/

Report: Faye F., Castro R., Dubief J., The impact of living with a rare disease: barriers and enablers of independent living and social participation. A Rare Barometer survey. EURORDIS-Rare Diseases Europe. March 2025. doi.org/10.70790/PDIR1346

Factsheet key results: Faye F., Castro R., Dubief J., Recognising disabilities and barriers! Key findings from a Rare Barometer survey on the impact of living with a rare disease. EURORDIS-Rare Diseases Europe. February 2025. tiny.cc/RB_DailyLife_Results

Webinar: Rare Barometer Daily Life Survey Results – 13 February 2025. youtu.be/B3krthKaEf4

3

STORY IDEA: NEWBORN SCREENING

THE OPINION OF PEOPLE LIVING WITH A RARE DISEASE ON NEWBORN SCREENING

24 MAY → 23 JULY 2023

6,179 respondents worldwide and **5,569** in Europe

24 languages

TARGET POPULATION

All patients living with a rare disease and their family members

50 countries

1331 rare diseases represented

EURORDIS RARE DISEASES EUROPE

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RELYING ON THE LIVED EXPERIENCE OF PEOPLE LIVING WITH A RARE DISEASE

PEOPLE LIVING WITH A RARE DISEASE

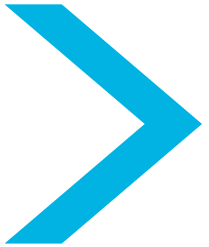
2,567 respondents (46%)

If it is or were possible, I would have liked to be diagnosed at birth

FAMILY MEMBERS

3,002 respondents (54%)
Mostly parents of people living with rare diseases (49% of the sample)

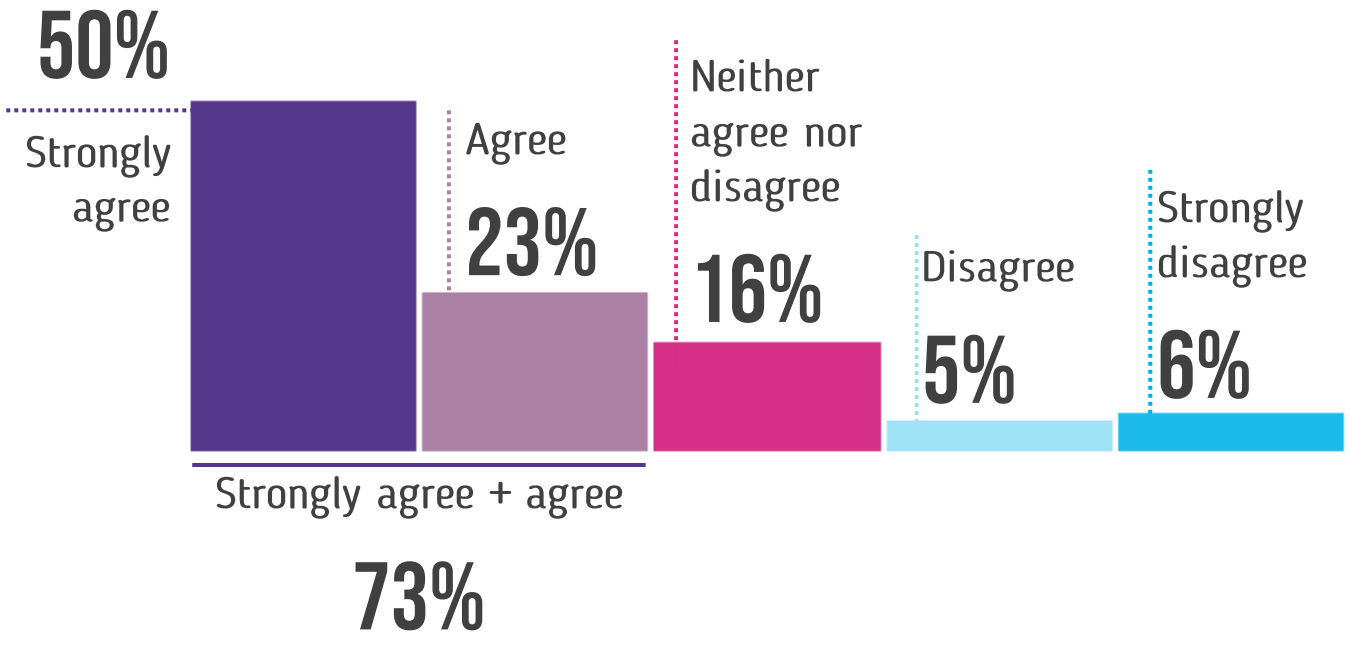
If it is or were possible, I would have liked the person I care for to be diagnosed at birth



Strongly agree
Agree
Neither agree nor disagree
Disagree
Strongly disagree

A WIDE MAJORITY OF PARTICIPANTS WOULD HAVE LIKED THEIR RARE DISEASE TO BE DIAGNOSED AT BIRTH

If it is or were possible, I would have liked [the person I care for] to be diagnosed at birth - *All respondents; n=5,569*



Comparison: the general population is more in favour of newborn screening (84%) than people living with a rare disease and their family members (70%) in a study on a specific rare disease[2].

[2]*Boardman et al. (2017). Newborn genetic screening for spinal muscular atrophy in the UK: The views of the general population. Mol Genet Genomic Med.*

THE RARE DISEASE COMMUNITY STRONGLY SUPPORTS NEWBORN SCREENING FOR ALL RARE CONDITIONS

A very large majority of participants support screening for all rare diseases when presented with specific consequences, even when they would not have liked their rare disease to be diagnosed at birth

90% **of the respondents think that any rare disease should be screened at birth if:**

It allows a quicker diagnosis, to the benefit of the individual person and their family carers.

It allows the person living with a rare disease to have their disabilities better recognised, more adequate social support and independent living.

The disease can be followed-up and harm can be avoided through prevention practices.

Comparison: 95% of the general population agreed that testing should be available for parents who wished it, even when respondents would decline it for their own newborns (around 85% said that they would probably or definitely have their newborn tested for a rare disease)[5].

Q *Percentage participants who agreed or strongly agreed with 'In your opinion, should any rare disease be screened at birth if no treatment exists and…' – All participants (n=5,569).*

[5]*Etchegary et al. (2012) Interest in newborn genetic testing: a survey of prospective parents and the general public. Genet Test Mol Biomarkers.*

NEWBORN SCREENING - PARTICIPANTS IN EUROPE

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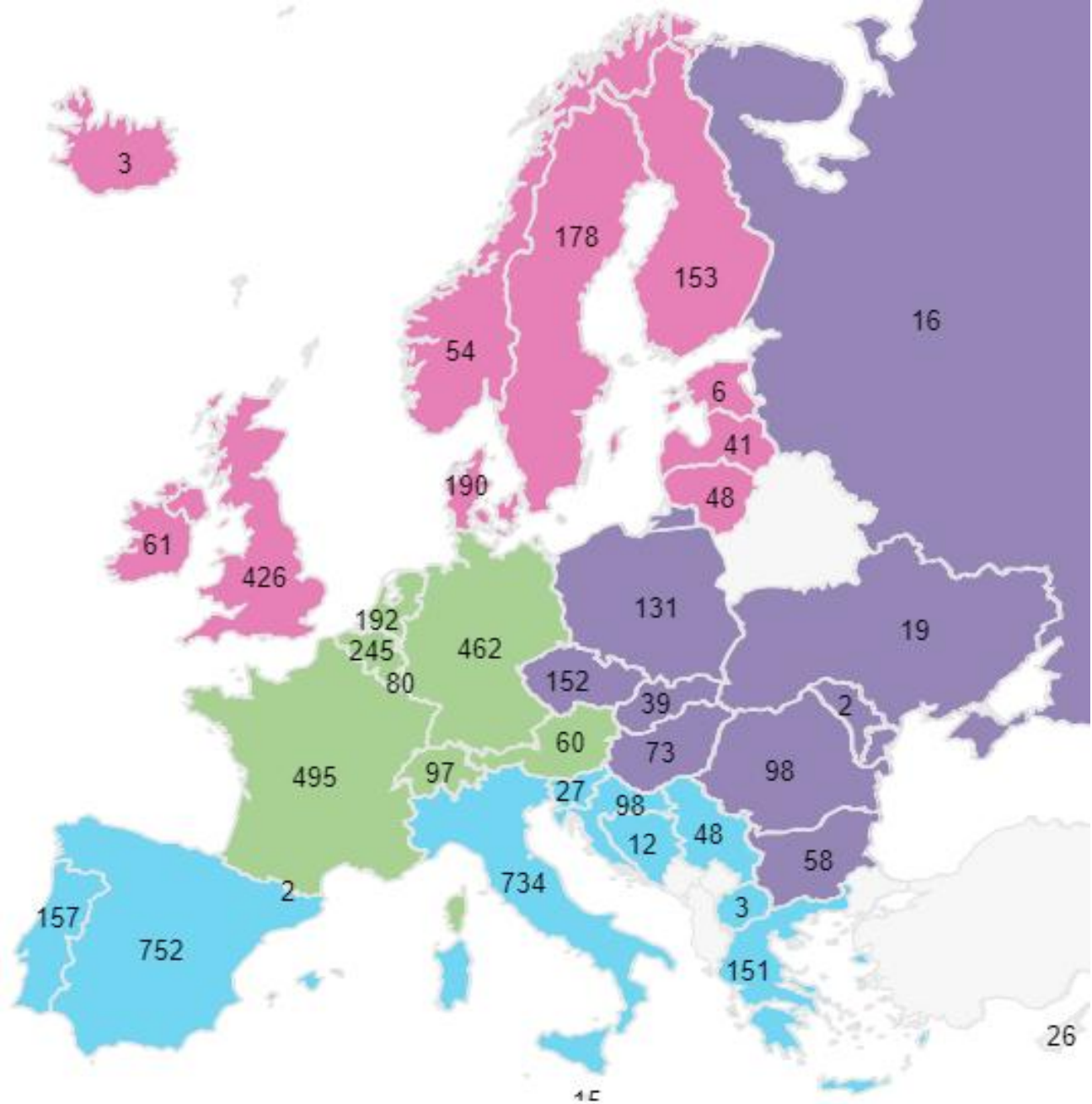
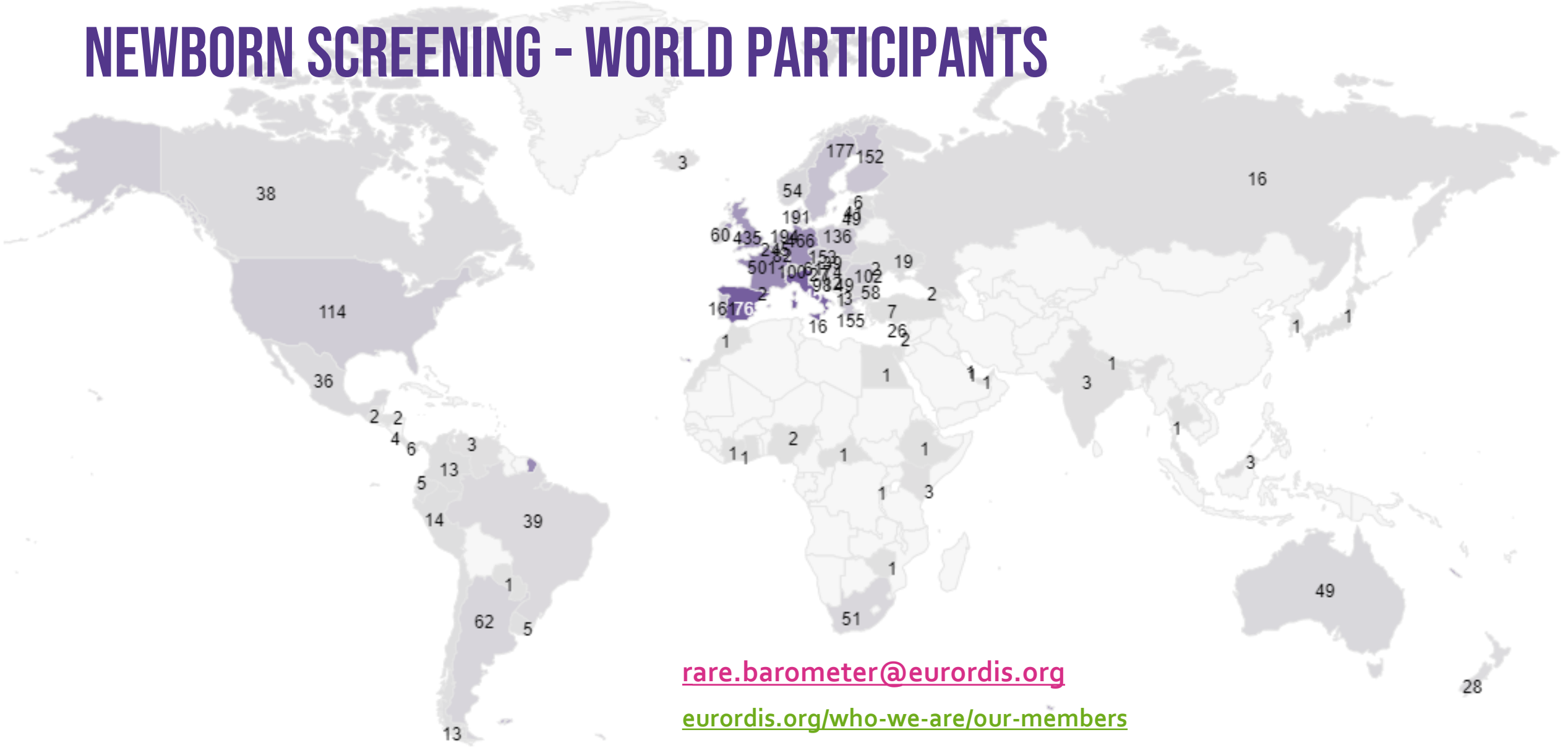
eurordis.org/who-we-are/our-members



NEWBORN SCREENING - WORLD PARTICIPANTS



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eurordis.org/who-we-are/our-members

NEWBORN SCREENING FEDERATIONS WITH 30+ PARTICIPANTS

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eurordis.org/who-we-are/our-members

Hereditary hemorrhagic telangiectasia	199
Cystic fibrosis	197
Hypermobile Ehlers-Danlos syndrome	120
Neurofibromatosis type 1	103
Fragile X syndrome	76
22q11.2 deletion syndrome	61
Tuberous sclerosis complex	49
Sarcoidosis	47
Duchenne muscular dystrophy	45
Phenylketonuria	40
Williams syndrome	39
Coffin-Lowry syndrome	37
Wilson disease	34
Fabry disease	33
Gaucher disease	32
Marfan syndrome	32
Classical Ehlers-Danlos syndrome	31
Osteogenesis imperfecta	31
Classic phenylketonuria	30
Rett syndrome	30

NEWBORN SCREENING – REFERENCES

rare.barometer@eurordis.org

Press release: Major survey reveals rare disease community's overwhelming support for screening at birth. 7 May 2024. download2.eurordis.org/pressreleases/2024-05-07-Press-release-on-survey-into-newborn-screening.pdf

Report: Dubief J., Gross E.S., Faye F., Voices on newborn screening: the opinion of people living with a rare disease. A Rare Barometer survey with the Screen4Care project. EURORDIS-Rare Diseases Europe. May 2024. doi.org/10.70790/NLMC2114

Factsheet key results: Dubief J. Screening Rare Diseases at Birth! Key findings from a Rare Barometer survey on the opinion of people living with rare diseases on newborn screening. April 2024. tiny.cc/RB_NBS

Webinar: Rare Barometer Webinar on the results of the newborn screening survey. youtube.com/watch?v=FeBjCEoYrNk

Position paper: Key principles for newborn screening. A EURORDIS Position Paper. January 2021. eurordis.org/publications/key-principles-for-newborn-screening/

Podcast: Rare on Air. Should more diseases be screened at birth? eurordis.org/rare-on-air/

THANK YOU!

to the Rare Barometer participants, partners and corporate donors in 2024-2025

rare.barometer@eurordis.org

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INNOVER POUR GUÉRIR

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CSL Behring
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Johnson & Johnson

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Orchard therapeutics

Roche

sanofi

Takeda

Zambon 1906

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