SCREENING RARE DISEASES AT BIRTH!

Key findings from a Rare Barometer survey on the opinion of people living with a rare disease on newborn screening

April 2024

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

70%
of rare diseases are genetic

70%
of rare diseases appear in childhood

Early diagnosis is key for people living with a rare disease as it allows families to plan for their child's care and treatment, to prevent severe disabilities from developing, and even to save lives. **Access to early diagnosis can be improved through newborn screening**, which is the process of systematically testing newborns just after birth and making sure that they can receive appropriate care and follow-up.

While there are still discrepancies in newborn screening programmes across Europe, the concept of newborn screening is widely accepted among the rare disease community: a previous Rare Barometer survey showed that **95% of people living with a rare disease were in favour of performing tests to diagnose rare diseases at birth**[1].

The survey presented here, conducted by Rare Barometer with the Screen4Care project, goes further and explores the point of view of people living with a rare disease and their close family members on the possibility to screen their condition at birth, thus considering their direct experience. **These insights valuably contribute to the development of newborn screening programmes across Europe.**

EUROPEAN RESULTS

A large scale quantitative survey conducted by Rare Barometer with the Screen4Care Research Project

24 MAY → 23 JULY 2023

5,569 respondents in Europe

24 languages

38 countries

TARGET POPULATION:
people living with a rare disease or family members (parents and close relatives)

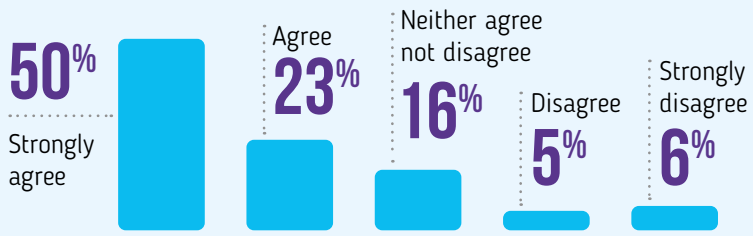
1,331 diseases represented

[1] *EURORDIS-Rare Diseases Europe, Rare disease patients' opinion on the future of rare diseases. A Rare Barometer survey for the Rare 2030 Foresight Study. June 2021.*

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

People living with a rare disease were asked if they would have liked to be diagnosed at birth, while family members were asked if they would have liked the person living with a rare disease to be diagnosed at birth.

73% of the participants either strongly agreed or agreed that they would have liked to be diagnosed at birth, or the person living with a rare disease to be diagnosed at birth.



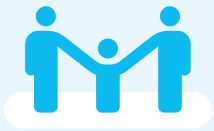
Comparison: the general population is more in favour of newborn screening (84%) than people living with a rare disease and their family members (70%) in a study on a specific rare disease[2].

Q *'If it is or were possible, I would have liked [the person I care for] to be diagnosed at birth' - All participants (n=5,569).*

Only 11% of the participants either disagreed or strongly disagreed with this proposition. Those participants were mostly concerned by the anxiety associated with the diagnosis, and by the stigma and discrimination that the child and the family could face at work or school, or by policies that insurance companies or banks would apply after the diagnosis.

2 PARENTS OF PEOPLE LIVING WITH A RARE DISEASE STRONGLY SUPPORT THE DIAGNOSIS OF THEIR CHILD AT BIRTH

82% of parents of people living with a rare disease would have liked their child to be diagnosed at birth



Comparison: parents of people living with a rare disease were also more in favour of newborn screening than adults living with a rare disease in a study on a specific condition[3].

"Parents would be able to prepare for the huge challenges that await them if the child needs help for the rest of their life. They could receive up-to-date information about the expected development, possible cures or early development opportunities, treatments or institutional care. I would definitely support this because it would have been a great help to me in the last 24 years."

Parent of a person living with a rare disease

Q *Percentage of participants who agreed or strongly agreed with 'If it is or were possible, I would have liked the person I care for to be diagnosed at birth' among parents of people living with a rare disease (n=2,567).*

[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. DOI: 10.1002/mgg3.353

[3] Boardman et al. (2019). Newborn screening for haemophilia: The views of families and adults living with haemophilia in the UK. Haemophilia. DOI: 10.1111/hae.13706

3 MOST PEOPLE LIVING WITH A RARE DISEASE WOULD HAVE LIKED TO BE DIAGNOSED AT BIRTH...

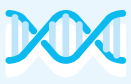
63% of people living with a rare disease would have liked to be diagnosed at birth

Percentage of participants who agreed or strongly agreed with 'If it is or were possible, I would have liked to be diagnosed at birth' among people living with a rare disease (n=2,701).

4 ...AND MORE FOR PAEDIATRIC AND GENETIC DISEASES

More people living with a rare disease would have liked to be diagnosed at birth when:

73% the first symptoms of their rare disease usually appear before or during infancy[4]



70% their rare disease is genetic[4]

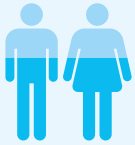
69% their rare disease was diagnosed more than 5 years after the first medical encounter

68% the symptoms of the rare disease are improving

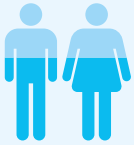
> *"Knowing about the genetic predisposition to a particular disease, you can notice the first symptoms in a timely manner and adjust your lifestyle or treatment to avoid transition to more severe forms. My diagnosis was made late, after 10 years of incorrect and ineffective treatment. Due to late diagnosis, I lost a lot in my quality of life, which I never managed to restore. With timely diagnosis, I could be living a full life".*
>
> **Person living with a rare disease**

Percentage of participants who would have liked to be diagnosed at birth among people living with a rare disease with an onset before or during infancy[4] (n=497), of genetic origin[4] (n=1,518), diagnosed more than 5 years after medical contact (n=808) or with improving symptoms (n=313).

People living with a rare disease were equally willing to be diagnosed at birth regardless of whether they received treatments or not.



63% Treatments or interventions



62% No treatment or intervention

> *"Regardless of available treatments, a diagnosis is often necessary to better understand what is happening to us. It is often indispensable to receive reasonable medical care or help in everyday life.*
>
> **Person living with a rare disease**

Percentage of people living with a rare disease who would have liked to be diagnosed at birth among those who answered either 'Yes, even partially (e.g. for one of the symptoms)' or 'No' to the question 'Did you receive or are you receiving treatment(s) or intervention(s) to lessen or control the effects of the rare disease, including medication, surgery, diet or other medical means?'.

[4] *Natural history and classification of rare diseases based on Orphanet data: orphadata.com*

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

Most participants support newborn screening for all rare diseases, 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).*

More information:
eurordis.org/voices or rare.barometer@eurordis.org

Full report in English: tiny.cc/survey_NBS_RD

Rare on Air podcast: eurordis.org/rare-on-air

Screen4Care research project: screen4care.eu

[5] *Etchegary et al. (2012) Interest in newborn genetic testing: a survey of prospective parents and the general public. Genet Test Mol Biomarkers. DOI: 10.1089/gtmb.2011.0221*

THANK YOU **to all people living with a rare disease who participated in the survey, and to Rare Barometer and Screen4Care partners!**

Rare Barometer
A EURORDIS INITIATIVE

Rare Barometer is the survey programme run independently by EURORDIS-Rare Diseases Europe and is a not-for-profit initiative. It conducts regular studies to identify the perspectives and needs of the rare disease community in order to be their voice within European and International initiatives and policy developments. Rare Barometer brings together more than 20,000 people living with a rare disease or family members to make the voice of the rare disease community stronger. For more information please visit eurordis.org/voices