Press Release

Major survey reveals rare disease community's overwhelming support for screening at birth

***7 May 2024, Paris* – EURORDIS-Rare Diseases Europe unveils new insights from a comprehensive survey on newborn screening for rare diseases, conducted by the organisation's *Rare Barometer* survey initiative and as part of the Screen4Care research project in May-July 2023.**

Developed in collaboration with a wide array of experts including policymakers, patient advocacy groups, and geneticists, this survey gathered feedback from 6,179 participants – 5,569 of whom are based in Europe – from over 50 countries, representing a diverse international community impacted by more than 1,300 distinct rare diseases.

Speaking about the survey's findings, **Jessie Dubief**, Social Research Director at EURORDIS, said:

> "Our latest survey results reflect our rare disease community's views on the potential benefits and challenges of newborn screening. Respondents overwhelmingly regard it as a means to access early diagnosis, shortening lengthy diagnostic journeys, securing appropriate healthcare, and ensuring newborns can live life to the fullest."

The key findings

1. **Our survey found that a wide majority would have liked the rare disease to be diagnosed at birth.**

Of those survey participants, **73%** either strongly agreed or agreed that they would have liked to have been diagnosed at birth or for the family member living with a rare disease to have been diagnosed at birth.

Conversely, only **11%** opposed early diagnosis, mainly due to concerns about potential anxiety, stigma, and discrimination, as well as the negative effects on insurance and banking policies. Despite being a minority of those surveyed, these participants' concerns, and reasoning highlight a pressing need for public policy interventions to address the mental health needs and social inequities that exist across Europe's rare disease community.

2. **The rare disease community strongly supports newborn screening for all rare conditions**

Most respondents stated that they would support newborn screening for all rare diseases, even when they would not have wanted their own rare disease to be diagnosed at birth.

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

- It would allow a quicker diagnosis, to the benefit of the individual person and their family carers.
- It would enable the person living with a rare disease to have their disabilities better recognised, more adequate social support and independent living.
- The rare disease could be followed-up and harm could be avoided through prevention practices.

Responding to the findings, **Virginie Bros-Facer**, Chief Executive Officer of EURORDIS, said:

"Our community's overwhelming support for expanded newborn screening, as revealed by our latest survey, highlights the need to further increase the capability of current newborn screening programmes across Europe. The identification of rare diseases during the newborn period can enable the provision of early targeted treatment and intervention plans, thus facilitating the delivery of appropriate social support, increasing independence, and ensuring improved healthcare follow-up and prevention of further complications.

"It is crucial that European policymakers, at both the national and international levels, act now to diminish disparities in newborn screening programmes across different countries, enhancing the quality of life for all children, irrespective of their birthplace.

"The technology is there to end the unacceptably long journeys to diagnosis many families impacted by a rare disease face. It is high time that advancements in public policy catch up with the scientific and technological advancements that we have seen in diagnostics."

The next steps

These findings highlight the community's strong support for newborn screening as an essential early healthcare intervention. Even when treatment options are limited, newborn screening is crucial for reducing health complications and improving quality of life.

In 2021, EURORDIS established *11 Key Principles for Newborn Screening* to foster a unified European approach to expanding birth screening programs. Adopting these principles would show the European Union and national governments' commitment to the highest health standards for all newborns.

The Rare Barometer team will further discuss these results at the upcoming *12th European Conference on Rare Diseases and Orphan Products (ECRD 2024)* on **15-16 May 2024**

Register for the conference now and benefit from a special discount using promo code: ***ECRD2024%15_Rbar***. The conference will be conducted in English with multilingual captioning available.

The survey report and its findings will be accessible in English, with additional factsheets available in 15 languages and detailed results for each question of the survey accessible in 24 languages ensuring broad access and engagement. Please visit the Rare Barometer website for more information.

- **Read the full paper & download the factsheet**

About EURORDIS-Rare Diseases Europe

EURORDIS-Rare Diseases Europe is a unique, non-profit alliance of over 1,000 rare disease organisations from 74 countries that work together to improve the lives of the 30 million people living with a rare disease in Europe. By connecting people, families, and rare disease groups, as well as by bringing together all stakeholders and mobilising the rare disease community, EURORDIS strengthens the patient voice and shapes research, policies, and services.

About Rare Barometer

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.

About Screen4Care

Screen4Care (2021-2026) is a research project funded by Innovative Medicines Initiative (IMI) offering an innovative research approach to accelerate rare disease diagnosis, which is based on two central pillars: genetic newborn screening and digital technologies. People living with rare diseases often find themselves on a burdensome diagnosis journey, enduring on average eight years of inconclusive consultations and possible misdiagnoses, leading to ineffective treatments and inefficient healthcare resource utilisation. Diagnosis odysseys and uncertainty around experienced symptoms continue to place a heavy burden on undiagnosed patients, their families, caretakers, doctors and society as a whole.

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