Press release

Rare Barometer launches new survey on newborn screening

31 May 2023, Paris – Rare Barometer, a EURORDIS - Rare Diseases Europe global survey initiative, has launched a new survey on the opinion of people living with a rare disease on newborn screening. The survey is open to families with a rare disease from any country in the world, and is translated into 24 languages.

Newborn screening is the process of systematically testing newborns just after birth for certain diseases, allowing for earlier diagnoses, better informed families, and improved follow-up healthcare.

Launching the survey, **Jessie Dubief, EURORDIS Rare Barometer Senior Manager** said:

> "While newborn screening programmes traditionally aimed to identify infants for only treatable conditions, where early identification helps to avoid irreversible health damage, our new survey is also examining rare disease patients' views on newborn screening for actionable conditions which may not be treatable but could still be addressed through other health interventions. Screening for more conditions at birth may allow for accelerated diagnoses of rare conditions, earlier interventions, and parents' improved ability to make informed reproductive choices.
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> Our survey will gather insight into rare disease patients' views on the benefits and possible disadvantages of newborn screening, including its impact on access to treatment, psychological wellbeing, and family dynamics. The respondents will be asked to give their opinion on newborn screening for their own disease, but also in more general terms".

With only 6% of rare diseases currently having an approved treatment, it is particularly important that other means of improving health and wellbeing are explored for those who currently do not have access to treatments.

Approximately 70% of rare diseases occur during childhood, but many show no symptoms for days or months following birth.

The Key principles for newborn screening, set out by EURORDIS, alongside its Council of National Alliances and Council of European Federations, assert patients' perspective that screening for untreatable but actionable diseases may still enable the more appropriate and effective management of the condition. Even in the absence of a cure or a treatment, an early diagnosis can lead to improved health and social outcomes for the newborn, and strategies of integrated care that address the multiple impacts of a condition on one's life.

The questionnaire was designed with the help of a topic expert committee composed of relevant experts in the field of newborn screening, including policy experts, patient advocacy organisations, sociologists, ethicists, geneticists and corporate partners. The survey has been developed as part of the Screen4Care research project, of which EURORDIS is a partner of.

Edith Gross, EURORDIS Health and Social Science Senior Project Manager commented:

> "The Screen4Care project is aiming to shorten the pathways to diagnosis of rare diseases, through advanced technologies such as newborn screening and artificial intelligence. The survey will provide the project with a strong empirical basis for selecting conditions to be screened on some 20,000 newborns in Italy, Germany, France and Greece. Seeking the perspective of people living with a rare condition will be at the centre of this survey, which will serve to establish a robust approach to ethical, legal and societal elements of newborn screening.

The survey will help EURORDIS generate new knowledge about the perspective of people living with rare diseases on newborn screening worldwide, as part of its advocacy work and involvement in the Screen4Care research project.

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About EURORDIS-Rare Diseases Europe

EURORDIS-Rare Diseases Europe is a unique, non-profit alliance of over 930 rare disease patient organisations from 73 countries that work together to improve the lives of the 30 million people living with a rare disease in Europe. By connecting patients, families and patient 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 patient services.

About Rare Barometer

The Rare Barometer Programme is a EURORDIS-Rare Diseases Europe initiative created to systematically collect patients' and carers' opinions on transversal topics and introduce them into the policy and decision-making processes. The objective is to transform the opinions and experiences of people living with a rare disease and their close family members into facts and figures that can be shared with a wider public and with decision-makers.

About Screen4Care

Screen4Care is a 5-year European Project funded under the IMI2 (Innovative Medicine Initiative) aimed at shortening the pathway to diagnosis using advanced technologies, including newborn screening and artificial intelligence applied to the diagnosis of rare diseases.

About rare diseases

The European Union considers a disease as rare when it affects less than 1 in 2,000 citizens. Over 6,000 different rare diseases have been identified to date affecting an estimated 30 million people in Europe and 300 million worldwide. 72% of rare diseases are genetic whilst others are the result of infections (bacterial or viral), allergies and environmental causes, or are degenerative and proliferative. 70% of those genetic rare diseases start in childhood.

Due to the low prevalence of each disease, medical expertise is rare, knowledge is scarce, care offerings inadequate and research limited. Despite their great overall number, rare disease patients are the orphans of health systems, often denied diagnosis, treatment and the benefits of research.