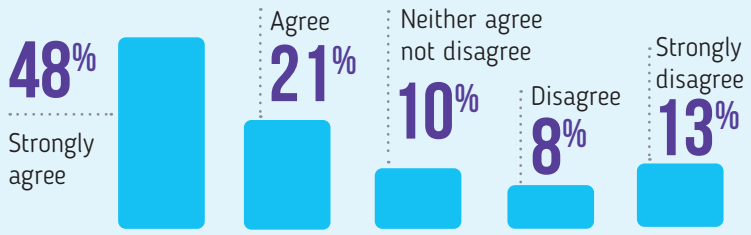
SCREENING RARE DISEASES AT BIRTH!

In Europe, 117 people with a rare urogenital disease and their family members expressed their views on newborn screening in a Rare Barometer survey conducted between 24 May and 23 July 2023.

1 A WIDE MAJORITY OF PARTICIPANTS WOULD HAVE LIKED THEIR RARE UROGENITAL 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[1].

Q *'If it is or were possible, I would have liked [the person I care for] to be diagnosed at birth' - All participants (n=117*

2 ...AND MORE AMONG PARENTS OF PEOPLE LIVING WITH A RARE UROGENITAL DISEASE

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

> *"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.*
> **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=65).*

[1] *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 MOST PEOPLE LIVING WITH A RARE UROGENITAL DISEASE WOULD HAVE LIKED TO BE DIAGNOSED AT BIRTH

51% of people living with a rare urogenital disease would have liked to be diagnosed at birth

Q *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=49).*

4 THE RARE UROGENITAL 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:**

- Knowing the child's mental or physical limitations in advance can allow more appropriate parenting.
- It can allow family members to know whether they carry the variant causing the disease.
- 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)[2].

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=117).*

[2] *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