European Reference Network
for rare or low prevalence complex diseases

Network Neuromuscular Diseases (ERN EURO-NMD)

Newborn Screening in Neuromuscular Diseases

Satellite Scientific Symposium organized by ERN EURO-NMD
March, 6th 2025

Newborn Screening in Rare Diseases: The role of patient organizations in driving NBS research and policies

Gulcin Gumus
EURORDIS

Funded by the European Union

Our Strategy

EMPOWER

To empower organisations and advocates representing people living with rare diseases across all rare diseases and all European countries.

PARTNER

To establish and facilitate networks with strategic partners and key stakeholders.

ADVOCATE

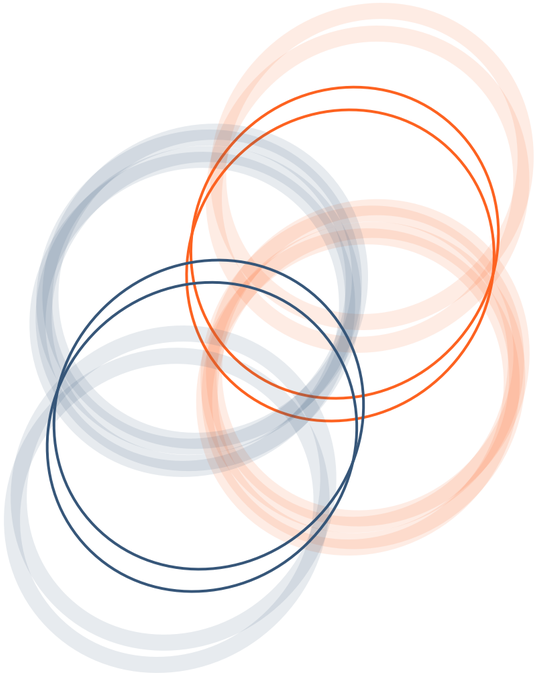
To advocate for policies and innovative solutions driven by the needs of people living with rare diseases.

PEOPLE FIRST

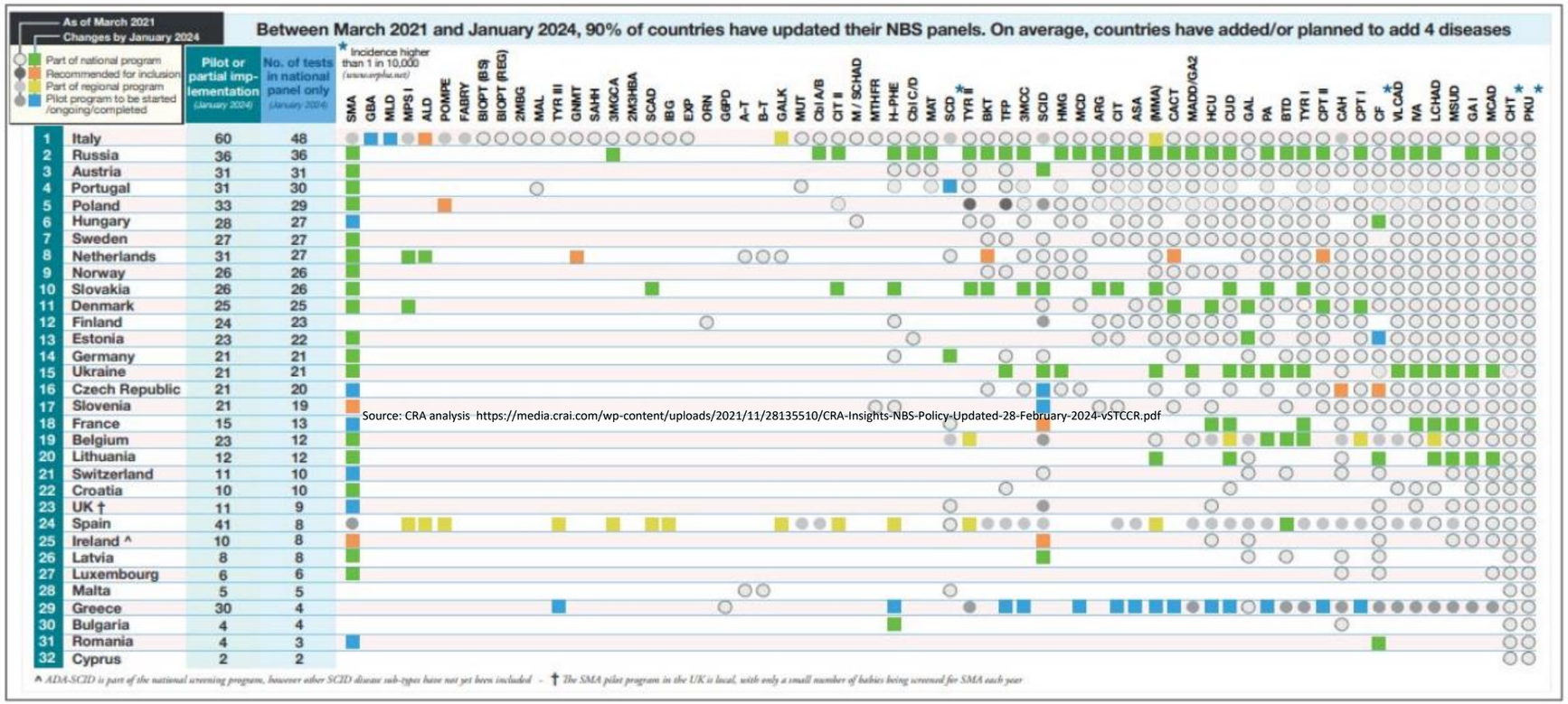
based on evidence and experience of unmet needs and preferences

Outline

- NBS **Advocacy for Joint Action**
  - NBS-Working Group & Policy actions
- NBS **Research**
  - EURORDIS' role in Screen4Care
- NBS **Networks and Dissemination**
  - Consortia, Scientific Societies, Networks



The NBS Landscape in Europe



Source: CRA analysis https://media.crai.com/wp-content/uploads/2021/11/28135510/CRA-Insights-NBS-Policy-Updated-28-February-2024-vSTCCR.pdf

Current Status of Newborn Screening in Southeastern Europe

Vanesa Koracin[1], Matej Mlinaric[2], Ivo Baric[3], Ian Brincat[4], Maja Djordjevic[5], Ana Drole Torkar[2,6], Ksenija Fumic[7], Mirjana Kocova[8], Tatjana Milenkovic[9], Florentina Moldovanu[10], Vjosa Mulliqi Kotori[11], Michaela Iuliana Nanu[10], Ziga Iztok Remec[12], Barbka Repic Lampret[6,12], Dimitrios Platis[13], Alexey Savov[14], Mira Samardzic[15], Biljana Suzic[16], Ildiko Szatmari[17], Alma Toromanovic[18], Mojca Zerjav Tansek[2,6], Tadej Battelino[2,6] and Urh Groselj[2,6*]

International Journal of *Neonatal Screening* MDPI

Article

Neonatal Screening in Europe Revisited: An ISNS Perspective on the Current State and Developments Since 2010

J. Gerard Loeber [1,*], Dimitris Platis [2], Rolf H. Zetterström [3], Shlomo Almashanu [4], François Boemer [5], James R. Bonham [6], Patricia Borde [7], Ian Brincat [8], David Cheillan [9], Eugenie Dekkers [10], Dobry Dimitrov [11], Ralph Fingerhut [12], Leifur Franzson [13], Urh Groselj [14], David Hougaard [15], Maria Knapkova [16], Mirjana Kocova [17], Vjosa Kotori [18], Viktor Kozich [19], Anastasiia Kremezna [20], Riikka Kurkijärvi [21], Giancarlo La Marca [22], Ruth Mikelsaar [23], Tatjana Milenkovic [24], Vyacheslav Mitkin [25], Florentina Moldovanu [26], Uta Ceglarek [27], Loretta O'Grady [28], Mariusz Oltarzewski [29], Rolf D. Pettersen [30], Danijela Ramadza [31], Damilya Salimbayeva [32], Mira Samardzic [33], Markhabo Shamsiddinova [34], Jurgita Songailienė [35], Ildiko Szatmari [36], Nazi Tabatadze [37], Basak Tezel [38], Alma Toromanovic [39], Irina Tovmasyan [40], Natalia Usurelu [41], Parsla Vevere [42], Laura Vilarinho [43], Marios Vogazianos [44], Raquel Yahyaoui [45], Maximilian Zeyda [46] and Peter C. J. I. Schielen [1]

Between March 2021 and January 2024, 90% of countries have updated their NBS Panels

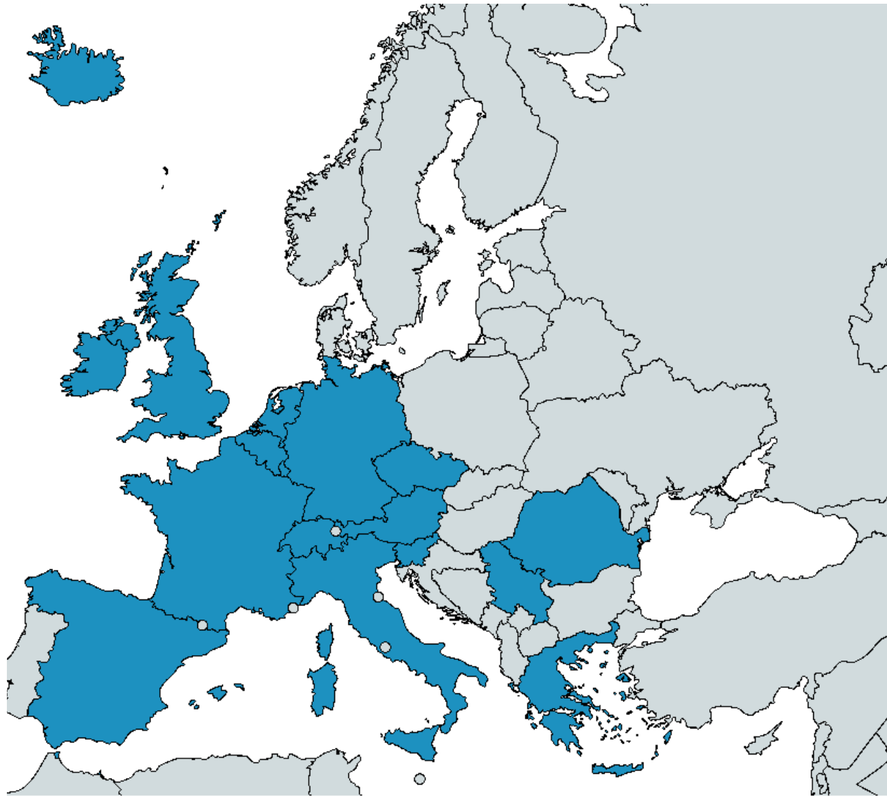
EURORDIS Newborn Screening Working Group

35+ Members

17 countries

Multidisciplinary WG

NBS Policy in Europe



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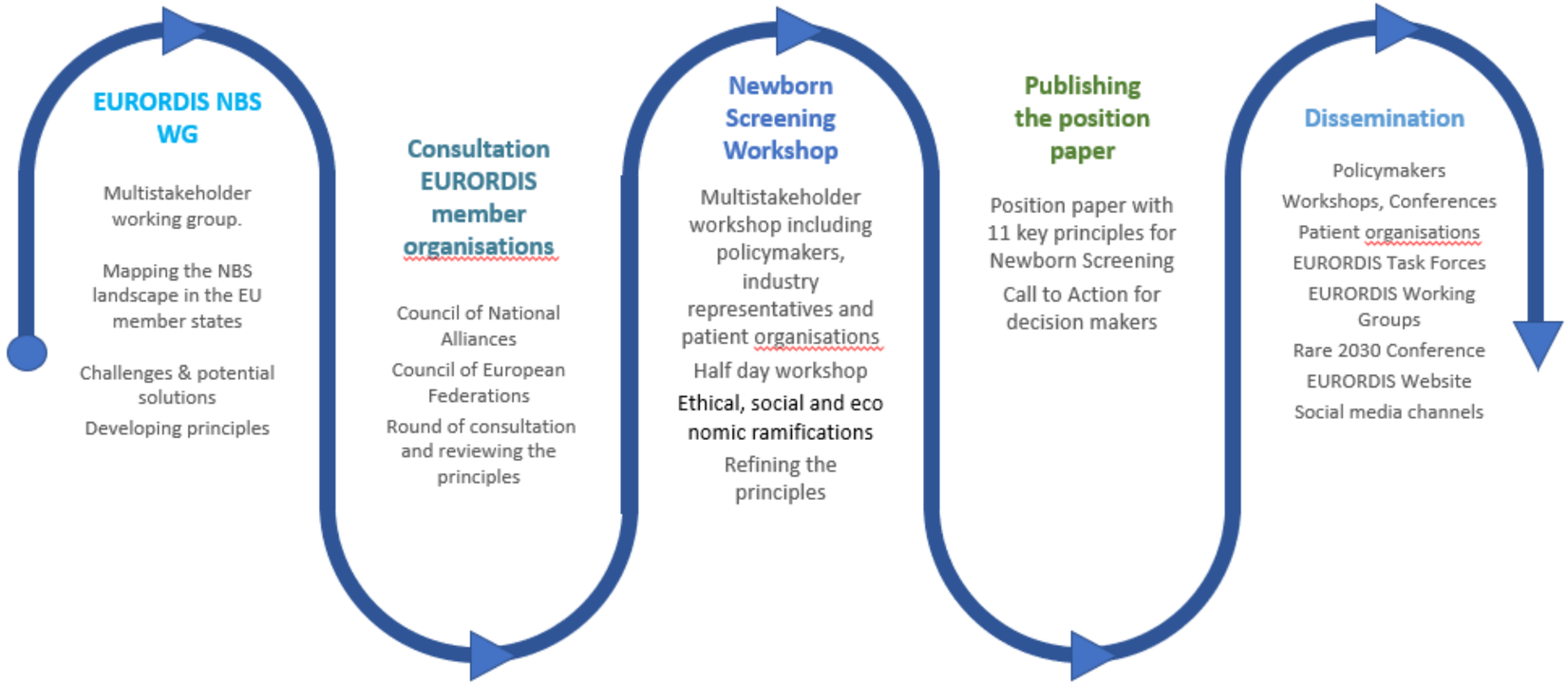


EURORDIS.ORG

EURORDIS Newborn Screening Working Group

Position Paper available in 13 languages

Czech English
German
Georgian Greek
Italian
Macedonian Portuguese
Serbian
Slovenian Spanish
Turkish

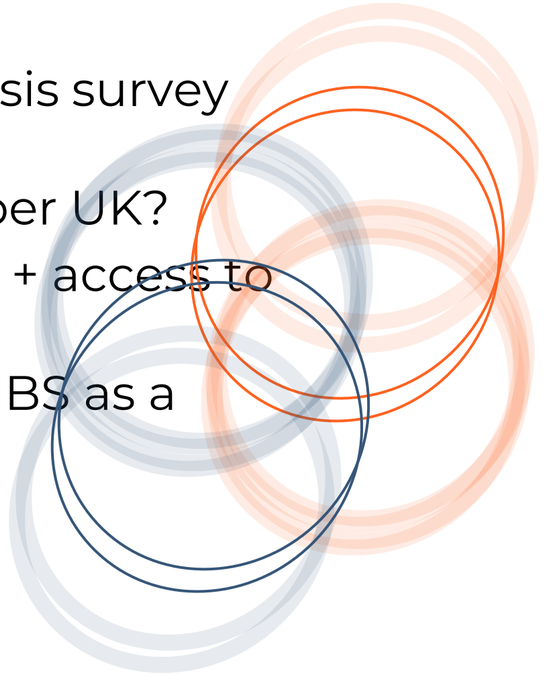
In progress: Ukranian, Icelandic, French

Next steps

Updates to Priorities

We currently **updating our advocacy messages** and have started exploring with the NBS WG how to do this for NBS in the context of the political context and the Draghi Report (setting the strategic guidelines for the new EU mandate):

- Link early screening and the development of new therapies - earlier you screen the more likely the therapy is to be effective
- Make sure all countries profit equally from advances > linked to diagnosis survey results
- Push for EU level analysis of Key Principles to engage stakeholders as per UK?
- Importance of empowering families to access NBS as future outcomes + access to social services usually needs concrete diagnosis
- Not just about innovation, but next steps to implement innovations - NBS as a beacon/example of how to take forward and deliver



From PRINCIPLES to ACTION

NBS Technical Meetings

EU2022.CZ

- Technical Meetings on NBS
- Achieving Equity and Innovation in Newborn Screening (11 October 2021, Slovenia)
- NBS Expert Meeting (23 July 2022, Czech Republic)
- Expert conference on rare diseases(25-26 October, 2022 Czech Republic)

EURORDIS' role

- One of the main stakeholders&organisers
- The views of people living with rare diseases and their families: 11 key principles & Panel discussion

Promotion of best practices

Promoting Italian model as best practice

- The most extensive newborn screening programme in Europe
- Organised as a "system"
- Embedded into the national health care system
- Aligned with 11 Key Principles!

EURORDIS' role

- Supporting and promoting the campaign to sign the petition

Polish Presidency Event

POLAND25.EU

Polish Presidency event: Conference on Rare Diseases

10-11 April 2025

Warsaw, Poland

More information to follow

39th Workshop of the EURORDIS Round Table of Companies (ERTC): Addressing systemic inequalities through patient-driven innovation & research

Aims:

•Demonstrate the added value of the rare disease patient community in shaping research, development and access pathways;

•Review our community's advocacy asks by addressing barriers to needs-led innovation;

•Explore opportunities to embed these priorities into future European research policies and funding programmes.

Advancing equity in newborn screening session

Maria Martinez - Illumina
Tamara Danguloff- BabyDetect
Zhana Chokeli - SCN2A Association

European Parliament Event on Rare Disease Day

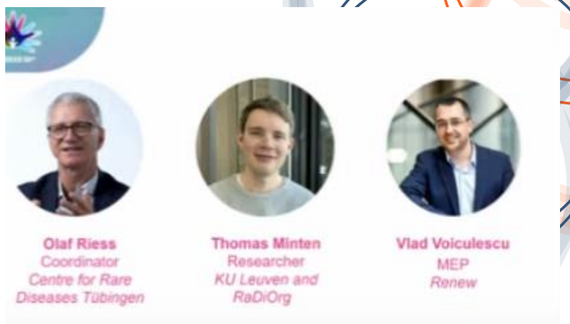
5 March 2025, Brussels

Easier, faster and more accurate diagnosis of rare diseases

Strategies to improve diagnosis rates
reduce waiting times address inequities
access to evolving technologies

To ensure equal diagnostic opportunities, EU-level initiatives should enhance cross-country collaboration**, particularly in newborn screening**. Strengthening Centres of Expertise and European Reference Networks is essential to streamline diagnoses and improve outcomes for rare disease patients

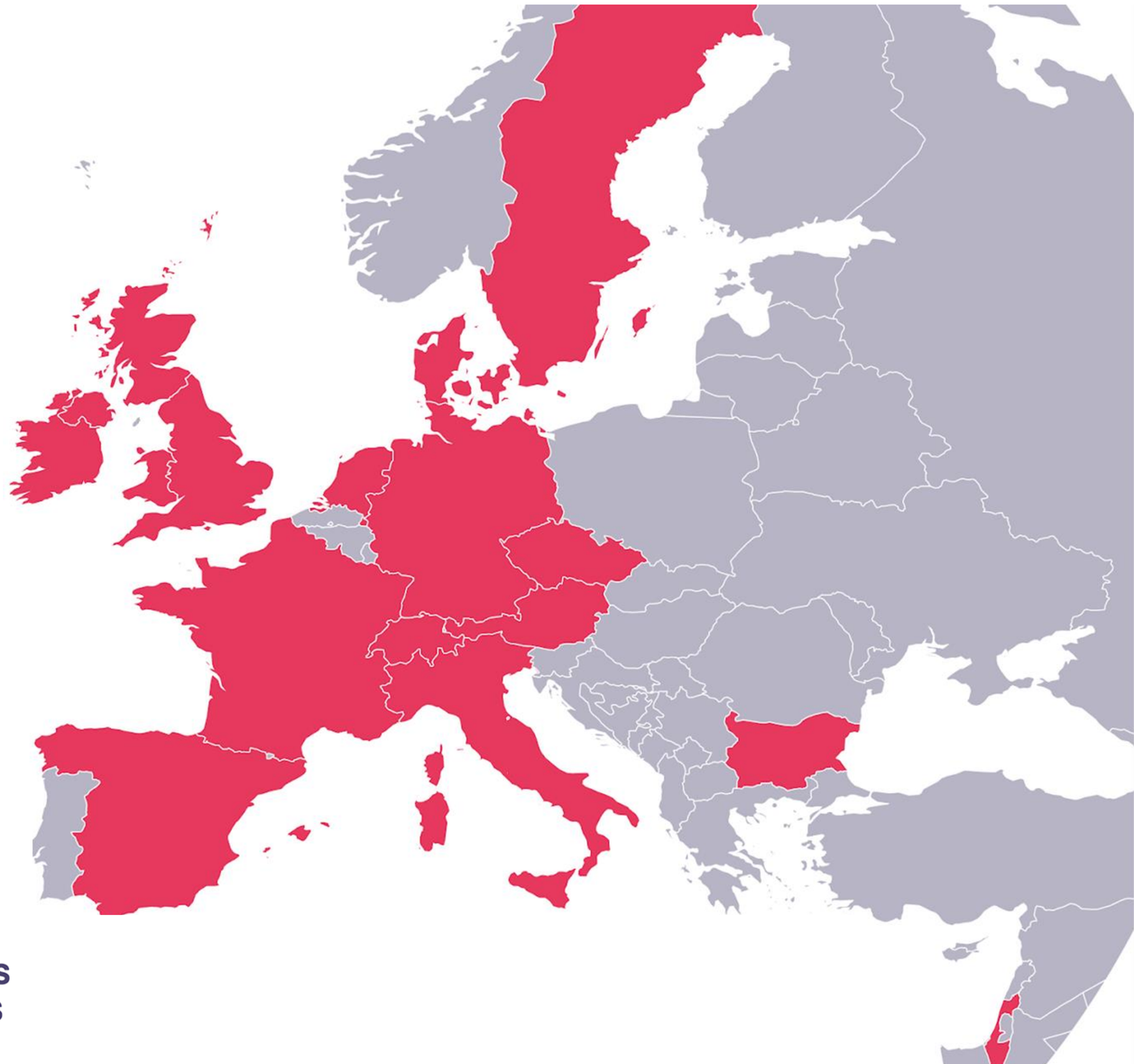
– ***Olaf Riess****, Institute of Medical Genetics and Applied Genomics and Centre for Rare Diseases, Tübingen*
– ***Thomas Minten****, KU Leuven*
– *MEP* ***Vlad Voiculescu*** *(Renew)*



EURORDIS & NBS Research

SCREEN 4CARE

Accelerating Diagnosis for Rare Disease Patients Through Genetic Newborn Screening and Artificial Intelligence



START DATE
1 OCTOBER 2021

DURATION
5 YEARS

BUDGET
25 MIO €

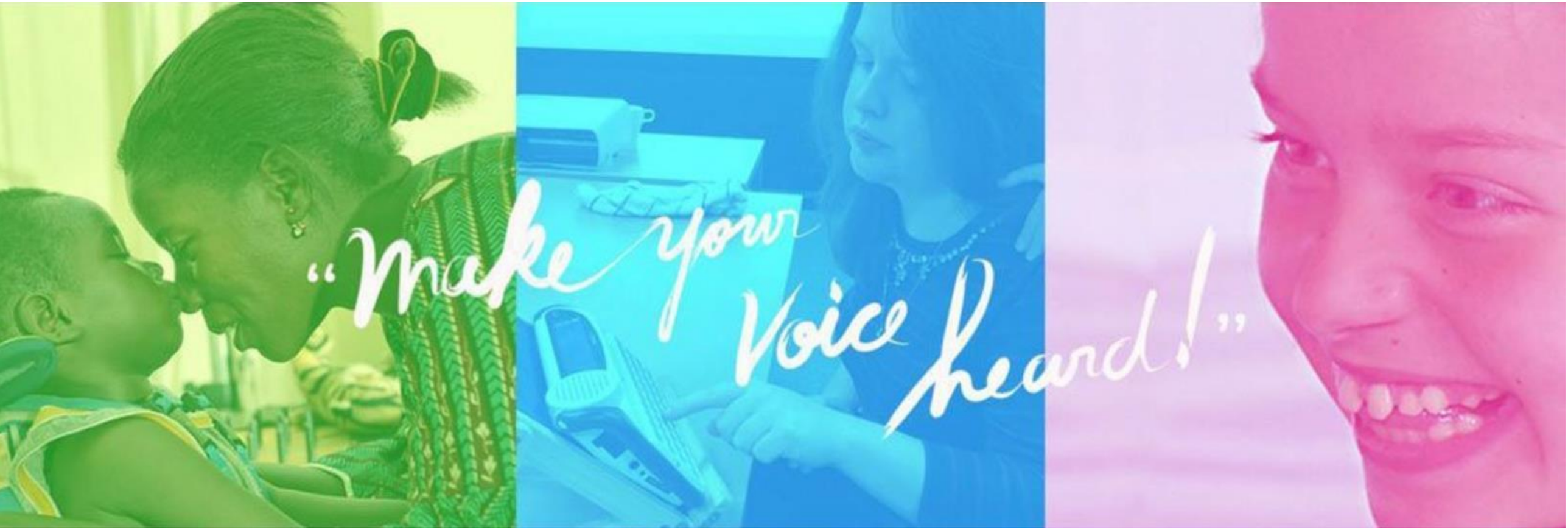
14 COUNTRIES
35 PARTNERS

Rare Barometer Programme

Rare Barometer

A EURORDIS INITIATIVE

EURORDIS' survey initiative



Rare Barometer Programme

EURORDIS' survey initiative

Rare Barometer
A EURORDIS INITIATIVE

Goal

Transforming opinions and experiences
about topics that directly affect people living with rare diseases
into figures and facts
to feed the rare disease community's
advocacy work

Surveys

Patients, families and patient representatives

1-3 surveys per year

23 languages

Worldwide

Results sent to participants

Panel

20 000+
people living with rare diseases who participate in EURORDIS' surveys and studies

People DO NOT have to register to participate in surveys

eurordis.org/rare-barometer

Rare Barometer Survey on Newborn Screening

SHAPING THE ONLINE QUESTIONNAIRE

Literature review

Identify main issues and criteria to define treatable and actionable conditions for newborn screening

Expert consultations

Input into priorities and considerations regarding issues and criteria for newborn screening

Topic Expert Committee

Contribute to clarifying topics and criteria to include in the questionnaire

EURORDIS' Council of National Alliances Members

Input on topics and criteria to be included in the questionnaire

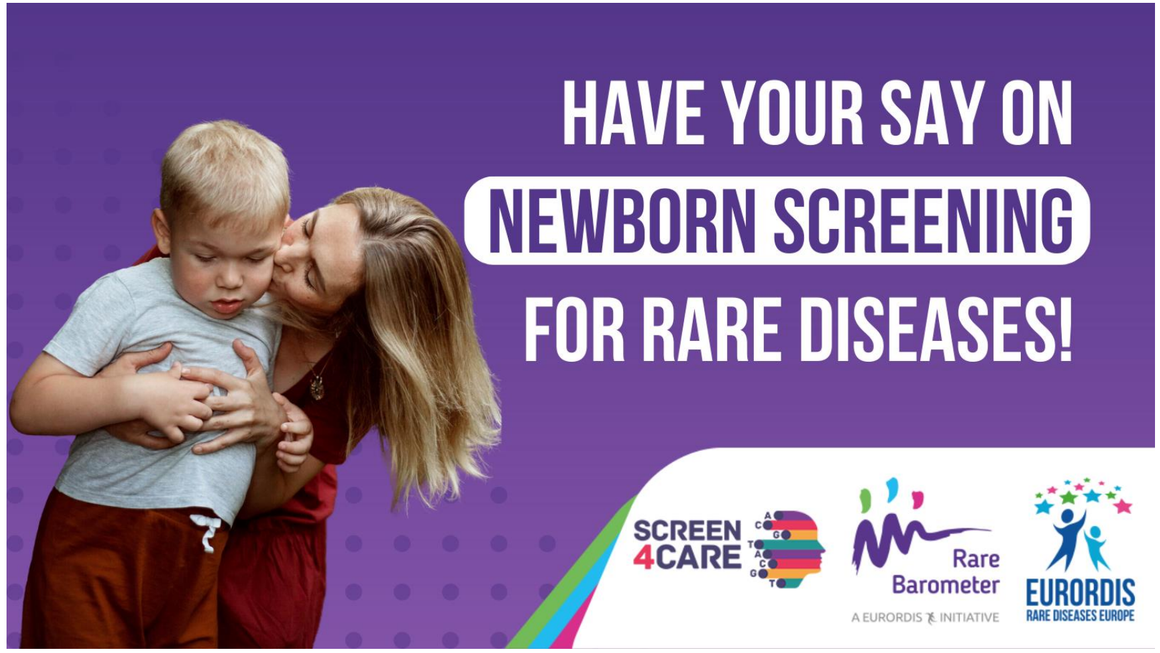
Feedback on the questionnaire

Pilot test with patients and carers

15 participants

Translations checked **in 15 languages** by native speakers

Rare Barometer Survey on Newborn Screening



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

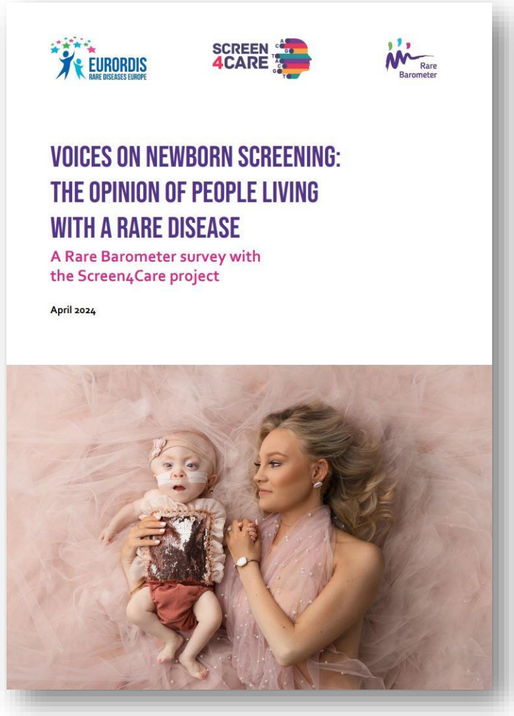
NEWBORN SCREENING RESULTS

rare.barometer@eurordis.org

REPORT

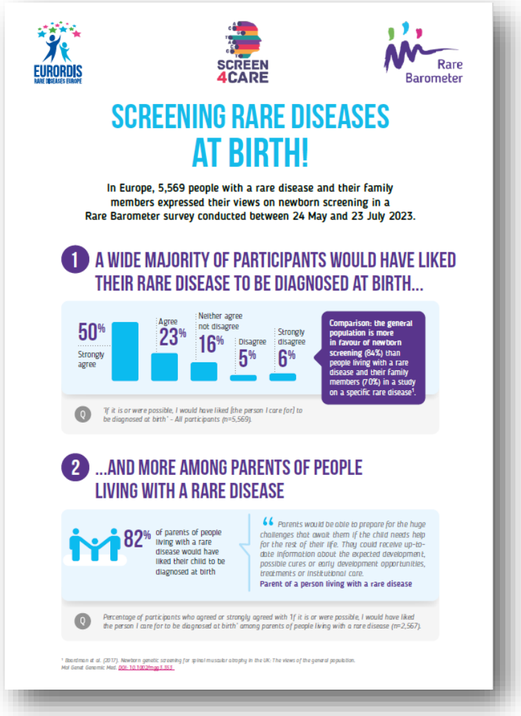
30 pages – English

DOI upcoming



tiny.cc/RB_NBS_report

FACTSHEETS & DASHBOARDS



tiny.cc/RB_NBS

WEBINAR ON SURVEY RESULTS

tiny.cc/RB_NBS_webinar

RARE ON AIR PODCAST

eurordis.org/rare-on-air

Screen4Care NBS Forum

- 40 Members
  - Newborn Screening Working Group
  - Patient Advisory Board
  - Digital and Data Advisory Board
  - Screen4Care members

- Results produced in Screen4Care analysed, consolidated, distilled to key messages for important stakeholder groups
- Breakout sessions

- Meets 2 times a year

1st NBS Forum F2F meeting (October 2023)

- The landscape of gNBS
- Define the criteria on actionability

List of actionable conditions for gNBS

2nd NBS Forum F2F meeting(22 October 2024)

- Implications of genetic NBS and what will matter to drive adoption
- How can we lower barriers for genetic NBS from an ethical / implementation /economic and social perspective.

Considerations for Patient focused advocacy on gNBS

Screen4Care NBS Forum

Challenges for Implementation

- Capacity planning
- Importance of the **communication of results.**
- Importance **of joint action and involvement of all stakeholders**
- It has to be country-based as it has to be aligned with approved treatments.
- The NBS approach needs to be flexible enough to add/remove genes in a reasonable time
- Genetic newborn screening programmes can gain valuable insights from **discussions on respecting patient autonomy and the importance of incorporating equality and diversity** into gNBS initiatives.

Ideas for the future

- Best practices
- Care pathways for infants identified after gNBS
- Thinking about consent, costs, reliability, problems of data storage
- Experiences of participants' feedback/ Impact of identification of positive cases to families

Networks and Dissemination

- **IRDIRC Newborn Screening Working Group**

Title: Patient Organizations: Advocating for Timely Newborn Screening & Improved Quality of Life

(Published)

- **International Consortium of Newborn Sequencing** (posters) 9-10 October
  - Title: Considerations for the expansion of newborn screening programmes: insights from the Rare Barometer survey on the opinion of people living with a rare disease (Jessie Dubief)
  - Title: Patient-Driven Partnership: Developing a Gene Panel for Actionable Conditions for Newborn Screening Using a Collaborative Approach (Gulcin Gumus)

- **European Society of Human Genetics**

ESHG Newborn Screening Workshop (June 2025)

Key Messages

- NBS research is continuously developing
- Advancing research in newborn screening requires **a collective effort** from all stakeholders
- **Dialogue** between patients, parents, policymakers and treatment developers, together with clinicians with academic experts on newborn screening.

- There are many ways to actively integrate RD community in newborn screening Research.
- Keep in mind that the impact of early diagnosis can be life changing for patients.