Press release

EURORDIS reiterates its call for harmonised newborn screening programmes across Europe at the Czech EU Presidency technical meeting

22 July, Paris **- EURORDIS-Rare Diseases Europe will join a technical meeting, organised under the auspices of the Czech Presidency of the Council of the EU on 23 July, to call for harmonised newborn screening programmes across Europe. This would ensure universal and equal access to newborn screening, help attain the highest standard of health for children born with a rare disease, and promote good practice among the Member States.**

EURORDIS alongside its National Alliance for Rare Diseases in the Czech Republic (ČAVO) will join Vlastimil Válek, the Czech Minister of Health, national and European policy makers, researchers and civil society organisations at the technical meeting "Early Diagnosis of Patients with Rare Disorders in the EU: Crucial Role of the Newborn Screening", to discuss EU action in the field of early detection and improve the newborn screening (NBS) systems in EU.

Building on the conclusions of the 2021 Slovenian EU Presidency meeting [1], this gathering will also see a discussion on the roadmap for the establishment of an NBS expert group, with the goal to help inform the European Commission and to act as a source for unbiased, evidence-based advice to support decision-making at a national level.

> *"With 72% of rare diseases being of genetic origin, screening for rare conditions in newborns holds the potential to lengthen, improve, and even save many lives. We want to see Europe take action, to reduce existing inequalities in newborn screening between countries and increase the quality of life of all children, no matter where they are born."*
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> – Gulcin Gumus, Research and Policy Project Manager at EURORDIS-Rare Diseases Europe

The rare disease patient community recognises the positive impact of NBS and its significant contribution to disease prevention, treatment and care. Building on the 11 Key principles of newborn screening [2], our involvement in Screen4Care [3] and the Rare Barometer survey [4], EURORDIS and its members demand that all newborns across the EU benefit from it where possible and, in particular, call for:

- Transparent and equal access to treatment and testing resources.
- Broad and fair implementation of NBS programmes.
- The inclusion of all stakeholders in the NBS process.
- Clear roles, responsibilities, and accountability frameworks in the NBS system.
- The establishment of an NBS expert group, to support efforts at the national level through coordinated work at the EU level, in areas where it can bring added value and optimise countries' efforts.

This technical meeting is an official event of the Czech Presidency of the EU Council, which will be continued with an expert meeting on rare diseases, where the possibility of establishing a European action plan for rare diseases is likely to be discussed after explicit support from Jakub Dvořáček and Vlastimil Válek for strengthened EU action on rare diseases.

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[1] Under its EU presidency, Slovenia spearheaded an initiative designed to enhance cooperation and equity in the provision of newborn and FH paediatric screening within the EU. The accompanying event of the Slovenian Presidency, "Achieving equity and innovation in newborn screening and in familial hypercholesterolemia paediatric screening across Europe", had among its objectives to identify good practice in the field of NBS in the EU, define the main gaps and discuss outlines to improve the equity of provision in the field of NBS, and explore and agree on next

steps, to ensure a systematic and comprehensive approach to screening in Europe that leaves no-one behind. The meeting saw the participation of medical experts, policymakers at EU and national levels, patient organisations, representatives of the main professional organisations, regulators and HTA agencies.

[2] EURORDIS published the position paper Key Principles for Newborn Screening in January 2021. A multi-stakeholder EURORDIS Working Group on Newborn Screening developed the 11 Key Principles, establishing the scope of newborn screening, but also the steps that should be taken at the national and European levels to make it a success.

[3] Screen4Care is a new EU Research Project focussed on accelerating diagnosis for Rare Disease patients through genetic newborn screening and advanced analysis methods such as machine learning and Artificial Intelligence.

[4] In 2021, EURORDIS Rare Barometer conducted the Survey on patients' opinion on the future of rare diseases, which showed that 95% of people living with rare diseases supported newborn screening for rare conditions.

About newborn screening

Newborn screening (NBS) is the process of systematically testing newborns for certain treatable diseases. With a simple blood test by heel prick, health professionals can check for certain treatable rare disorders and conditions that are not clinically evident in the newborn period. Since its introduction in the early 1960s, continued advancements in screening tools and treatment options have expanded the range of NBS programmes to include potentially detectable rare diseases (RDs).

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

EURORDIS-Rare Diseases Europe

EURORDIS-Rare Diseases Europe is a unique, non-profit alliance of 1000 rare disease patient 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 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. Follow @eurordis or see the EURORDIS Facebook page. For more information, visit eurordis.org.

Press contact

Stanislav Ostapenko
Communications Senior Manager
EURORDIS-Rare Diseases Europe
stanislav.ostapenko@eurordis.org
+33 1 56 53 52 61