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Position Statement:

An Urgent Call for EU-level Collaboration on Newborn Screening

A joint statement from rare disease community members and organisations

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Newborn screening (NBS) saves lives by detecting serious, often rare conditions, before symptoms appear. Yet, in Europe, lifesaving NBS programmes strongly vary across countries.

The signatories, representing groups and individuals within the rare disease community, call upon the European Union to establish a multi-stakeholder Newborn Screening Group to support Member States in strengthening NBS programmes. This initiative would help promote expanding newborn screening programmes and ensure that every newborn child in Europe has an equal chance for a healthy start in life.

Where are we today: Persistent Inequalities Across Europe

Health inequalities are embedded in EU health systems from birth, as newborn screening policies differ significantly across Member States. European countries screen newborns for anywhere between 2 and 49 conditions, and on average, for fewer disorders than other high-income countries such as Australia and the USA. Implementation speed also differs markedly: some countries roll out nationwide screening for newly added conditions rapidly once a decision is taken, whereas others face multi-year delays before implementation is achieved. This means that for a newborn within the European Union, timely access to rare disease treatment may be lacking and therapy options depend on place of birth rather than on medical need.

These substandard newborn screening policies in several European countries have already had devastating consequences. For life-threatening but treatable conditions such as MCAD deficiency, spinal muscular atrophy (SMA), metachromatic leukodystrophy (MLD) and severe combined immunodeficiency (SCID), unequal and delayed adoption of screening has resulted in many preventable child deaths and cases of irreversible disability in Europe. Beyond the human toll, these delays also undermine national and EU competitiveness through preventable
healthcare expenditure, caregiver burden, and long-term productivity losses over the past decade.

In many cases, establishing a comprehensive NBS system, aligned with regulatory decision-making and timely patient access to innovative therapies, acts as the "last-mile" step that completes the medicines development lifecycle, ensuring that investments made along the pathway translate into real-world benefits for the final recipient: every newborn in Europe.

These disparities are avoidable, and their human cost is indefensible. Recent global developments reinforce the urgency of action. The 2023 UN General Assembly Resolution on persons living with rare diseases and the 2025 WHO Resolution on rare diseases both recognise newborn screening as an essential preventive measure.

Yet implementation across Europe remains fragmented and lags behind this global momentum. Persistent national and regional differences mean that not all newborns benefit equally from scientific and clinical advances in screening and early treatment.

Why Newborn Screening and Early Detection Matter

Newborn screening, a system of tests and follow-up for several rare diseases conducted on newborn babies shortly after birth, is a proven public health programme that enables early detection and timely treatment shortly after birth. Newborn screening can prevent irreversible disability, especially for conditions where prompt intervention is essential. While the vast majority of screened newborns are healthy, for those diagnosed with treatable conditions, early detection transforms lives.

Early detection through newborn screening not only can improve clinical outcomes but also spares children and families a long and often distressing diagnostic odyssey. For many conditions, screening at birth has also proven to be cost-efficient or even cost-saving by a timely intervention and minimising long-term disability. By shortening or preventing the diagnostic odyssey for many families, newborn screening contributes to better use of specialist services and more efficient care pathways across the health system.

Up to 70% of rare diseases have a paediatric onset, and about 72% are genetic. This underscores the potential of newborn screening to identify a large proportion of rare conditions early in life and to improve the quality of life for children with these conditions.

Moreover, health care costs for rare diseases are significant. A 2024 study estimated the cost of just 43 rare diseases at €249.3 billion per year across nine European countries— six times the cost borne by reference populations. Newborn screening and early detection, followed by appropriate care pathways, can therefore have a meaningful positive impact both on individuals' lives and health system sustainability.

For European countries, improving newborn screening policies is therefore an important tool to strengthen health systems as well as to improve outcomes for children with rare diseases. The current lack of universal newborn screening and genetic testing represents a missed opportunity to reduce the high economic burden associated with the "diagnostic odyssey". The 2024 study demonstrates that every misdiagnosis and delay in starting treatment incurs additional economic costs, particularly through increased direct medical and non-medical expenses. For example, total costs were significantly higher for those experiencing longer delays between diagnosis and treatment. By enabling immediate intervention, newborn screening avoids the expensive, inappropriate medical exams and late-stage hospitalisations that occur when a disease has already progressed, thereby protecting both the clinical condition of the patient and the long-term sustainability of the healthcare system.

Why the EU Must Act

Despite its commitment to high-quality healthcare, the European Union lacks a cohesive strategy on newborn screening. Several European countries face significant challenges with newborn screening,

including opaque internal processes, duplicated evidence-gathering efforts, limited budget allocations, regional differences, slow processes and unnecessary delays in implementation.

Moreover, the lack of coordination and coherence across Europe further increases fragmentation and amplifies the complexity of regulatory and access pathways for innovative therapies. This, in turn, weakens timely patient access, reduces efficiency in the use of public resources, and dilutes the impact of policies and initiatives designed to strengthen EU competitiveness. Without a unified approach, the full potential of newborn screening remains unrealised across Europe.

EU-level cooperation through a structured European NBS Group, would drastically support and improve national newborn screening programmes. Collaboration is essential for NBS evidence-gathering since the rarity of conditions makes it difficult for individual countries to assess evidence alone. Moreover, European shared guidelines and evidence reviews would reduce duplication, decrease screening gaps and promote equity for European newborns.

Calls for European coordination are longstanding. These reasons have led patients' organisations, clinicians, and experts to repeatedly urge for European coordination on newborn screening. An expert opinion document commissioned by the European Commission in 2014 recommended a similar EU-level NBS committee. Yet, more than a decade later, these recommendations remain unimplemented.

A Multi-stakeholder European NBS Group

The multi-stakeholder European NBS Group we call for should foster structured cooperation while respecting national competences. Key areas of work for the group should include:

- Providing scientific review of evidence for screening targets;
- Developing EU-level guidelines for target newborn screening disorders;
- Establishing EU-level recommendations for education, training, and family communication;
- Sharing data on programme outputs and HTA evaluations;
- Expected Outcomes of Increased Cooperation and EU Support

Conclusion: A Call to Shared Action

Europe prides itself on equity, solidarity, and excellence in healthcare. Yet, for too many children with rare conditions, these values remain unattained. The disparities in newborn screening are avoidable; their human cost is indefensible and undermine those values Europe stands for.

Now is the time to act. In a rapidly evolving newborn screening landscape, it is more urgent than ever to start with systematic European collaboration. Let us not lose another decade. A coordinated European effort can ensure that every child, regardless of birthplace, receives an equal chance for a healthy start in life.

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