

The Missing Link in Newborn Screening:

Why Europe Needs Shared
Implementation Knowledge

EUROPE • SCREENING • IMPLEMENTATION • KNOWLEDGE

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Executive Summary

“Countries are largely tackling these challenges in isolation.”

Newborn screening (NBS) gives babies the earliest possible chance at a healthy life, allowing serious conditions to be identified and treated before irreversible harm occurs. Yet access to this opportunity varies sharply across the European Union: national panels range from over 40 screened conditions to as few as 2, creating a "postcode lottery" in which a child's chance of early diagnosis depends on where they are born.

This variation stems not only from differing national decisions on which conditions to screen for, but from roadblocks at two distinct stages: the decision to expand a screening panel, and the practical implementation of that decision once approved. Gaps in evidence, limited budgets, laboratory capacity, workforce shortages, and slow-moving governance structures can each delay expansion by years.

Critically, countries are largely tackling these challenges in isolation. Despite having introduced conditions such as spinal muscular atrophy (SMA), severe combined immunodeficiency (SCID), and cystic fibrosis (CF) through very different governance, funding, and clinical models, much of this real-world experience remains undocumented or inaccessible to policymakers elsewhere.

This paper argues that closing this knowledge gap through structured, cross-country exchange of implementation experience is essential to ensuring every newborn in Europe receives timely access to screening, diagnosis, and care.

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1. What is newborn screening

SECTION 01

This white paper focuses primarily on the expansion and implementation of NBS programmes across the 27 EU Member States, while drawing on wider European experience where relevant. Newborn screening (NBS) is a universal, population-based public health programme whose aim is to identify babies right after birth who are asymptomatic but at risk of a serious, often life-threatening or life-altering condition, allowing action to be taken before irreversible damage occurs [1], [2]. Screening is typically performed under an opt-out or presumed-consent model, though a minority of countries treat participation as mandatory [3], [4]. Most of the conditions targeted through NBS are rare genetic diseases for which early diagnosis and treatment can substantially improve outcomes. The best-known NBS test is newborn bloodspot screening (often referred to as the Guthrie test or heel-prick test), in which a small blood sample is collected through a heel prick and analysed [5]. Results are typically available within a few days, and in most European countries parents are contacted only when an abnormal result is detected (“No news is good news”). Other commonly used NBS approaches are physiological and include a hearing screening to check for hearing loss and screening for critical congenital heart disease (CCHD). A positive (abnormal) screening result does not necessarily mean the baby has a condition, but that complementary testing is needed to confirm or rule out the diagnosis.

Many advances continue to be made in NBS techniques. For example, the range of biomarkers measurable from dried blood spots has expanded. Additionally, second-tier testing, in which additional analyses are performed on the same initial sample following an abnormal first tier screening result, is increasingly being implemented to reduce false-positive screening results. Genetic and genomic testing are also emerging as additional approaches to NBS. While traditional bloodspot screening primarily detects biochemical or other markers associated with disease, DNA-based testing can directly identify disease-causing genetic variants and may enable screening for actionable genetic conditions for which suitable biochemical markers are not available. Importantly, genomic screening should currently be viewed as complementary to, rather than a replacement for, established newborn bloodspot screening [6], [7], [8], [9], [10].

2. Why newborn screening matters

SECTION 02

NBS turns the first days of life into an opportunity to prevent avoidable harm. NBS programmes can identify infants who appear healthy but may have a serious, often life-threatening or life-altering condition. For these children, early detection is not simply a diagnostic advantage: it is the difference between treatment starting before irreversible damage occurs and diagnosis arriving only after symptoms have already appeared.

For several conditions, the period between birth and the onset of symptoms represents a critical window. If a diagnosis is made and care begins within that window, outcomes that might otherwise include developmental regression, permanent disability, or death can be substantially improved. In many cases, a child’s health trajectory can come to resemble that of an unaffected child. If the window is missed, the same condition may become a lifelong burden for the child and family, and a continuing challenge for health systems. NBS is not limited to conditions for which a curative or disease-modifying treatment is available. Screening programmes may also include conditions where an early diagnosis enables timely monitoring, symptom management, supportive care, informed reproductive decision-making, or access to specialised services. Even when the disease course cannot be fundamentally altered, early identification can help avoid unnecessary examinations, reduce uncertainty, and improve care planning for affected children and their families.

Early detection can also shorten the diagnostic odyssey faced by many families affected by rare diseases. Without a screening pathway, children may move through years of clinical appointments, inconclusive tests, misdiagnoses, and ineffective treatments before receiving an accurate diagnosis. Recent evidence suggests that the average diagnostic delay for rare diseases remains close to five years in Europe [11]. By identifying eligible conditions before symptoms emerge, NBS can replace such uncertainty and delay with timely diagnosis and care.

The value of NBS extends beyond clinical outcomes. Early detection can also reduce healthcare use and related costs as well as the burden placed on families and society [12], [13]. Dutch and Belgian modelling on spinal muscular atrophy (SMA) found that healthcare costs decreased when patients were identified through NBS rather than after clinical diagnosis [12], [13].

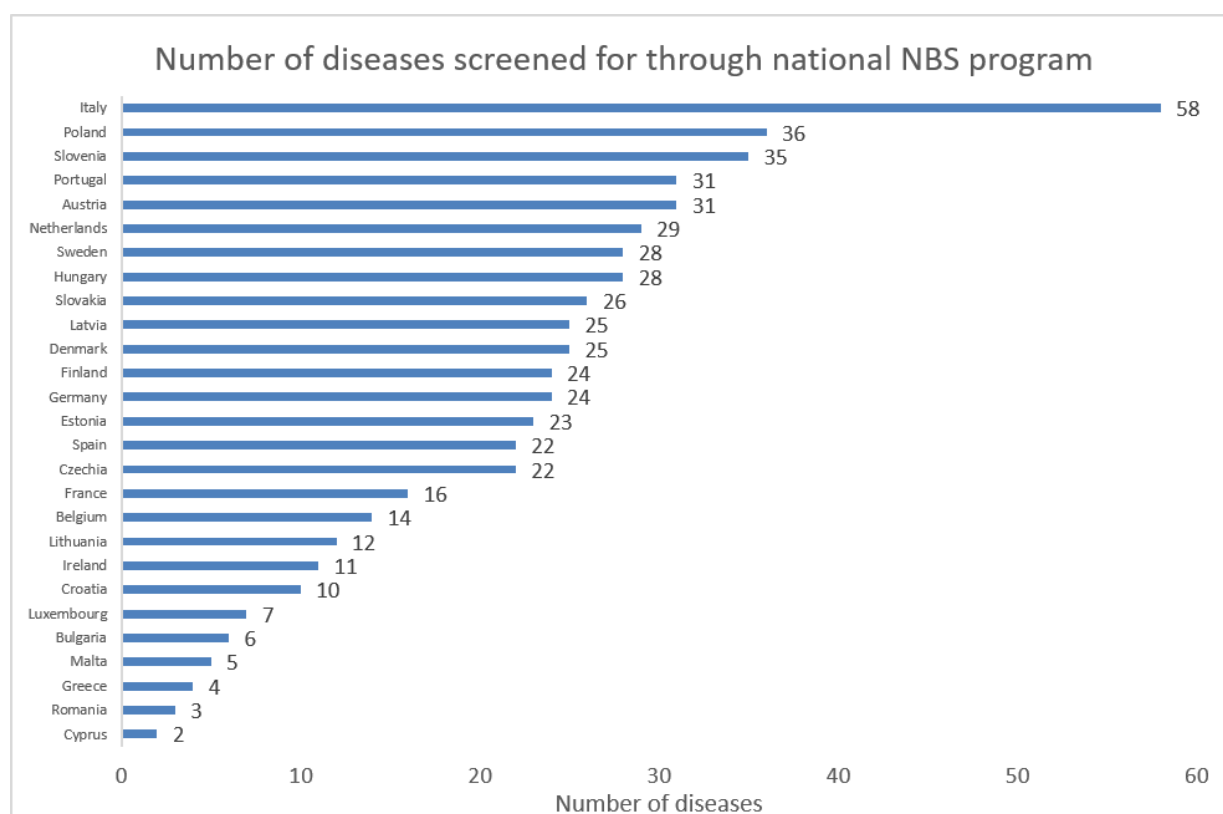
NBS therefore represents much more than a screening test: it is a mechanism for ensuring that children with rare conditions are identified as early as possible and given the best available opportunity for optimal outcomes and quality of life. However, there is substantial variation across EU member states in both the number and type of conditions included in NBS programmes [5]. This results in a “postcode lottery” for early diagnosis, where the country or region where a child is born determines whether they are screened for a specific condition at birth and can access early diagnosis and treatment, potentially influencing not only their future health and quality of life but, in some cases, their survival.

3. Variation in NBS expansion and implementation in practice

SECTION 03

Although NBS is well established across the EU, there is no single EU-wide NBS programme or harmonized EU screening panel, as the decision to include a new indication to the NBS programme is a national or regional responsibility. This results in a large variation in the number of screened conditions across countries. Italy is at the top of the range, screening for more than 50 conditions, followed by Slovenia and Poland. At the other end of the range, Cyprus screens for 2 conditions (Figure 1). Regional differences may also exist within individual countries.

Figure 1. Number of conditions included in national newborn screening programmes across EU Member, May 2026.
Source: unpublished European landscape assessment of newborn screening programmes.



This variation is not only limited to how many conditions are screened, but also which conditions are included. There is limited consensus across countries on which diseases should be included in NBS programmes. Only congenital hypothyroidism and phenylketonuria appear in every Member State's panel, meaning two countries with similarly sized panels may nonetheless screen for substantially different conditions (Figure) [1]. For example, as of the early 2020s, SMA was included in NBS programmes in some countries (e.g. Wallonia (Belgium), Germany) while only piloted or regionally approved in others (France, Czech Republic, and parts of Italy and Spain) [14]. For severe combined immunodeficiency (SCID), Catalonia became the first region in Spain and one of the first in Europe to introduce universal SCID screening in 2017, years before (or in the continued absence of) equivalent programmes elsewhere [15].

The variation observed across European NBS panels reflects differences in how countries evaluate, approve and implement new conditions for NBS. To add an indication to a NBS panel, it must go through a pathway of being selected as a candidate, followed by the actual implementation pathway to add it to the panel. This is a multi-stage process that moves from evidence generation to policy evaluation, political approval, operational preparation, implementation, and post-implementation monitoring, which can in general be summarized in five important stages [1], [16], [17], [18] (Table 1):

Table 1. The five-stage pathway for adding a condition to a NBS panel.

1	Identification of a candidate condition	Triggered by new evidence, availability of treatment, technological advances, pilot studies, or advocacy from clinicians, researchers, patients, or public authorities.
2	Evidence assessment	Review of disease burden, natural history, screening test performance, treatment effectiveness, ethical considerations, and economic impact.
3	Policy recommendation and decision-making	Evaluation by national screening committees, health technology assessment bodies, ministries of health, or advisory groups.
4	Operational implementation	Adaptation of laboratory workflows, workforce training, procurement of equipment, development of confirmatory testing pathways, clinical referral networks, communication materials, and quality assurance systems.
5	Monitoring and evaluation	Measurement of programme performance, outcomes, false-positive rates, diagnostic yield, timeliness, and patient impact.

Experience across Europe demonstrates that countries with similar evidence bases frequently arrive at different decisions or reach the same decision at very different speeds. This suggests that implementation is influenced by factors extending beyond scientific evidence alone [16], [17].

- Governance and decision-making structures
- Some countries operate highly formalised review systems with predefined assessment procedures and predictable review cycles. Others rely on more ad hoc processes, making it difficult for stakeholders to anticipate when new conditions will be considered. Transparent governance structures generally provide greater predictability and facilitate stakeholder engagement [1], [15].
- Availability of implementation resources
- The addition of even a single condition may require new laboratory equipment, validation studies, IT adaptations, workforce training, quality management procedures, and long-term funding commitments. Countries with established implementation infrastructure can often move more quickly than those starting from limited capacity [18], [20].
- Evidence requirements
- Countries differ in the type and quantity of evidence required before introducing a condition. Some place considerable emphasis on randomised clinical evidence and health-economic evaluations, while others accept observational evidence, real-world outcomes, or pilot programme results. These differences can significantly affect implementation timelines [16], [17].
- Stakeholder engagement
- Implementation increasingly depends on collaboration between laboratories, clinicians, patient organisations, policymakers, public health agencies, and industry. The EURORDIS principles emphasise that all stakeholders should contribute to the design and governance of NBS programmes, yet approaches to stakeholder involvement remain highly variable across Europe [1], [21].
- These structural differences in governance, resources, evidence requirements, and stakeholder engagement do not merely explain why panels vary but translate into concrete roadblocks at both the decision-making and implementation stages of NBS expansion.

4. Roadblocks to NBS expansion in practice

SECTION 04

The roadblocks to NBS expansion can vary substantially between countries and can arise both during the decision-making process to expand an NBS panel and during implementation after an expansion has been approved or legislated.

One of the major barriers during the decision-making process is the lack of evidence needed to support decisions on NBS expansion [2], [18]. Even when evidence is available, many countries do not systematically consider key categories of evidence, such as test accuracy, whether early treatment improves outcomes, and the potential harms of overdiagnosis [22]. Furthermore, expanding NBS programmes may require financial resources and investments in laboratory infrastructure, technology and workforce capacity, which need to be considered when introducing new conditions [5], [18].

Importantly, approval of an NBS expansion does not automatically translate into implementation in practice. Several roadblocks can delay or hinder implementation in practice, including limitations in laboratory infrastructure, clinical pathways, workforce capacity, or funding arrangements [1], [16]:

- The laboratory infrastructure might not have the required technology to perform new screening tests or the required second-tier testing capacity to manage false positives [3]. The emergence of genetic NBS introduces additional implementation requirements, including sequencing and bioinformatics capacity, variant interpretation, data governance and access to appropriate genetic expertise and counselling [23], [24].
- A positive screen can only lead to improved outcomes if appropriate reference centres are available to confirm the diagnosis and initiate treatment [25].
- The right healthcare providers need to be available and trained to provide associated follow-up testing and (genetic) counselling [23].

These barriers result in substantial delays in implementing new indications after a positive decision has been made, making time one of the major barriers to timely expansion of screening programmes [2], [18]. As implementation speed differs markedly between countries, ranging from rapid roll-out to multi-year delays, children may miss the opportunity for presymptomatic diagnosis and treatment initiation at the time when treatment is expected to provide the greatest benefit [1]. As screening technologies evolve, Member States will increasingly face common implementation questions, creating further opportunities for EU-level knowledge sharing and collaboration.

5. Recommendations

SECTION 05

Accelerating the expansion and effective implementation of NBS programmes is essential to ensure that every newborn in the EU has an equal opportunity for a healthy start in life and to reduce the burden on healthcare systems. As effective treatments and management options become available for a growing number of rare diseases, differences in access to NBS risk translating into even greater inequalities in access to timely diagnosis, treatment and health outcomes.

Several recommendations have been made to advance both the decision to expand and the practical implementation of NBS in the European Union:

- Adopt a standardized, evidence-based prioritisation approaches for candidate conditions [26], [27].
- Establish dedicated NBS decision-making bodies that regularly consider candidate conditions for inclusion [28]
- Pursue EU-level coordination and funding mechanisms [1]
- Invest in laboratory infrastructure, including capacity for quality assurance and second-tier testing capacity [3]
- Train specialists and counsellors in parallel with panel expansion [23]

Beyond these measures, structured cross-country knowledge exchange is essential to ensure that governance, funding, laboratory, and clinical models already tested in one country can inform decisions elsewhere.

6. The Missing Piece: Cross-Country Knowledge Sharing

SECTION 06

Despite increasing attention to collaboration on NBS, systematic exchange of implementation experience and best practices between EU Member States remains limited. Countries have introduced conditions such as SMA, SCID, cystic fibrosis, and numerous metabolic disorders using different governance, funding, laboratory, and clinical models. Yet much of this experience remains undocumented, difficult to access, or insufficiently compared across countries, thereby creating an incomplete understanding of why implementation succeeds in some settings while progressing slowly in others. This knowledge gap creates a significant obstacle to improving access to screening across Europe.

Policymakers seeking to expand programmes often have limited visibility of how other countries addressed similar challenges, what barriers they encountered, and which implementation strategies proved effective [16]. As a result, countries often face implementation challenges in isolation, despite similar issues having already been addressed elsewhere. Better operational intelligence could therefore support Europe in several ways [1], [29].

All stakeholders, including patients, physicians, policymakers and industry, would benefit from a clearer understanding of how NBS programmes operate in practice:

- Better knowledge of the operational steps would support stakeholders aiming at expanding the NBS programme in their country or region.
- Understanding the roadblocks encountered by individual countries, and how these have been overcome, can provide practical insights into how the processes for assessing and implementing new conditions within NBS programmes can be improved.
- Comparing why some countries have rapidly added conditions such as SMA or SCID while others still lack them entirely could help countries learn from each other's experiences and identify which approaches might be adapted to their own healthcare system rather than expecting a single pathway to fit every country given how differently national systems are structured.

As a first step, we propose the development of an EU-wide comparative study of NBS implementation, documenting how different Member States have addressed the governance, funding, laboratory, clinical and operational challenges associated with adding conditions such as SMA and SCID to their NBS programmes. The objective would not be to impose a single implementation model across the EU, but to create a shared evidence base from which Member States can identify approaches relevant to their own healthcare systems. This would provide a concrete foundation for narrowing the variation in NBS access across Europe. Shared understanding of this kind is essential to improve NBS and for babies to receive timely access to screening, diagnosis, and care, regardless of where they are born.

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