

Genomic Newborn Screening in Belgium: Opening the Debate of Public Health Evaluation

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Abstract

Accumulating evidence from international and Belgian pilot studies suggests that genomic newborn screening (gNBS) has reached a level of maturity that now justifies structured evaluation within a public health framework. Recent international pilot studies, including large Belgian data, now provide evidence that first-tier or adjunct genomic screening is technically feasible, clinically informative, and highly acceptable to parents. Belgium has played a leading role in this field. The BabyDetect program has shown that targeted next-generation sequencing can be implemented in a maternity setting with high parental uptake and a meaningful added diagnostic yield beyond conventional screening. At the same time, the rapid development of international programs has exposed major unresolved questions regarding gene selection, variant interpretation, health-economic evaluation, data governance, and equitable access. In this Insight, we

argue that the debate should now move beyond whether gNBS is technically possible and instead focus on how it can be evaluated and governed responsibly within the Belgian healthcare system. We review the current evidence, highlight the specific Belgian contribution, and outline the principal challenges that must be addressed before broader public implementation can be responsibly considered.

Introduction

Newborn screening (NBS) is one of the most successful public health interventions of the past 50 years. Since its introduction for phenylketonuria screening in the 1960s, NBS has progressively expanded to include an increasing number of conditions, primarily metabolic and endocrine disorders, and has substantially improved outcomes through presymptomatic diagnosis and early treatment. Today, NBS programs are implemented in all high-income countries, although the number and type of screened diseases vary considerably between jurisdictions. In Belgium, this variability is also observed between the Flemish and French-speaking communities, each operating its own screening program with distinct panels of conditions. Despite these differences, both systems share a common objective: early identification of severe, treatable diseases to enable timely intervention

and improve long-term outcomes. However, as in all other countries, conventional NBS in Belgium still relies predominantly on biochemical assays performed on dried blood spots. This model, though highly effective for many metabolic and endocrine disorders, limits the detection of conditions for which no reliable biomarker exists. As a result, many severe, early-onset diseases that would be relevant targets for NBS cannot currently be identified through standard biochemical screening, thereby limiting opportunities for timely diagnosis and early intervention. In this context, genomic newborn screening (gNBS) has emerged as a promising solution to these limitations. By analysing a predefined set of genes causing actionable childhood-onset disorders rather than targeting diseases individually, genomic screening offers a more comprehensive and flexible approach. It enables the inclusion of conditions that cannot be detected using conventional biochemical methods and allows rapid

adaptation of screening panels as new therapies become available. The purpose of this Insight is not to present gNBS as ready for immediate universal implementation, but to examine whether the scientific evidence has become sufficiently mature to justify a structured Belgian public health strategy. We review the current evidence, highlight the specific Belgian contribution, and outline the conditions required for scientifically robust, ethically responsible, and socially acceptable implementation.

What do current pilot studies actually show?

Over the past decade, gNBS has transitioned from conceptual exploration to real-world pilot implementation. Although pilot programs differ substantially in sequencing strategy, target population, gene content, and reporting criteria, the available evidence now allows several conclusions to be drawn regarding feasibility, parental acceptability, diagnostic yield, and the major unresolved challenges for public-health implementation.

Belgian Experience: The BabyDetect Program

Belgium has been at the forefront of genomic NBS implementation with the BabyDetect pilot program, which provides particularly valuable real-world data from our own healthcare context. This study used targeted next-generation sequencing (tNGS) to screen for 165 treatable paediatric conditions across 405 genes. In the initial feasibility and acceptability study conducted in Liège, 90.5% of parents consented to genomic screening, demonstrating remarkably high acceptance; a key prerequisite for any population-based screening program (1). This study also showed that structured pre-test information could be embedded in implementation through dedicated materials and trained consent procedures. The subsequent population-based BabyDetect study further confirmed the feasibility and clinical utility of this approach. Among 3,847 screened newborns, 71 positive cases were identified, corresponding to a screen-positive rate of 1.8% (2). Of these, 41 were G6PD deficiency, a condition also included in conventional biochemical newborn screening in Liège. The remaining 30 positive cases corresponded to conditions not detected by conventional screening, illustrating the added yield that a first-tier genomic strategy may provide. All positive cases were managed through a dedicated clinical pathway for confirmatory assessment, specialist follow-up, counselling, and access to care. Importantly, false-positive and false-negative rates were very low; with one false-negative case related to a previously unreported variant not included in the interpretation pipeline. BabyDetect also illustrates the importance of iterative panel governance, as reporting criteria for selected cardiomyopathy-associated genes were refined during the study in light of variable age of onset and phenotypic heterogeneity. This underscores both the robustness of the approach and the need for

continuous refinement of variant databases and interpretation pipelines. Together, these elements illustrate that gNBS implementation requires not only analytical validity, but also clear communication on the scope and limits of screening, predefined reporting rules, and the capacity to ensure rapid and appropriate follow-up for newborns and families.

International Experience: The GUARDIAN and BabyScreen+

Findings from international pilot programs broadly reinforce the signals observed in Belgium. In the United States, the GUARDIAN study evaluated genome sequencing as an adjunct to traditional NBS in a diverse population. Among 4,000 newborns, 72% of families consented to participate, and sequencing was successfully completed in 99.6% of cases (3). The reported screen-positive rate was 3.7%, reflecting the inclusion of a broader set of conditions, including those without established biochemical markers, as well as differences in variant-reporting policy. In particular, the study reported selected variants of uncertain significance (VUS) in specific situations, including autosomal recessive conditions when a VUS co-occurred with a pathogenic or likely pathogenic variant. This broader reporting framework partly explains the higher positivity rate compared with more conservative targeted-panel approaches. This higher rate compared to targeted panels underscores the impact of gene selection and reporting criteria on screening outcomes. In Australia, the BabyScreen+ study evaluated whole-genome sequencing in 1,000 newborns, targeting 605 genes associated with early-onset, treatable conditions. Sixteen infants (1.6%) were identified with actionable findings, most of which were not detected by standard screening (4). Parental acceptability was extremely high, with more than 99% of participants supporting the availability of genomic screening for all parents and their newborns, and psychosocial impact was minimal, with low levels of decisional regret reported.

Cross-study signals: consistency in performance, divergence in panel design

Despite important differences in populations, gene panels, sequencing technologies, and baseline screening programs, a remarkable convergence emerges across studies. In BabyDetect, the overall screen-positive rate was 1.8%. When excluding G6PD deficiency, the screen-positive rate was 0.7%. Similarly, the GUARDIAN authors reported that, when excluding G6PD deficiency, the frequency of positive genomic screening was 0.6%, compared with 0.3% for traditional newborn screening in New York State. This convergence is noteworthy given the differences in panel composition, study design, and reporting policies. It suggests that targeted genomic newborn screening may yield relatively consistent positivity rates across programs when comparable inclusion

criteria are applied. These empirical yields can also be interpreted in light of epidemiological estimates for early-onset rare single-gene disorders. In BabyDetect, the screen-positive rate after exclusion of G6PD deficiency and cardiomyopathy-associated findings subject to refined reporting criteria is approximately 0.5%. This closely aligns with Blencowe et al.'s modelled baseline birth prevalence of early-onset rare single-gene disorders, approximately 4.5 per 1,000 births, or 0.45% (5). Parental acceptance of genomic newborn screening is also consistently high across major pilot studies, including the Belgian BabyDetect study, in which acceptance exceeded 90%. In addition to acceptability, reliability appears robust. Rates of false positives and false negatives remain low when conservative variant filtering strategies are applied. However, this approach may lead to underreporting of uncertain or borderline variants, reflecting a deliberate trade-off between sensitivity and specificity. Together, these data indicate that the central scientific question is no longer feasibility alone, but rather how to define the appropriate scope, governance, and evaluation framework of gNBS. At the same time, one major source of uncertainty remains the marked heterogeneity in condition and gene selection across existing programs. A recent comparative analysis identified more than 25 gNBS initiatives worldwide, highlighting substantial variability in design, sequencing strategy, target population, and genes screened (6). One of the most striking findings from this international comparison is the heterogeneity in gene selection. Across 27 programs, the number of genes analysed ranges from 134 to more than 4,000, with only 1.7% of genes shared by more than 80% of programs. This heterogeneity is informative: it reflects both the momentum of the field and the absence of consensus, underscoring the need for harmonization.

Discussion

Belgium as a Pioneer in Genomic Screening

Belgium is not entering this discussion from the sidelines. The country has already played a pioneering role in the implementation of genetic newborn screening, particularly through spinal muscular atrophy (SMA) screening, and has made major contributions to demonstrating both its clinical and medico-economic value. Early Belgian initiatives, notably the 2018 pilot program in Southern Belgium, were among the first large-scale European efforts to systematically screen newborns for SMA (7). This program rapidly expanded and transitioned into an official screening program, illustrating Belgium's leadership in translating screening innovation from research into public health practice. Importantly, this program has demonstrated huge health economic efficacy. The Belgian experience provided robust real-world evidence supporting the clinical effectiveness of early detection. SMA screening enabled identification of affected infants before symptom onset and to initiate treatment

within the first weeks of life, which is a critical determinant of therapeutic success (8). This led to markedly improved outcomes compared with symptomatic diagnosis, including survival, motor development, and reduced need for ventilatory or nutritional support. These findings aligned with broader international evidence that presymptomatic treatment dramatically modifies disease trajectory. Beyond clinical effectiveness, Belgium has also been instrumental in evaluating the economic impact of SMA screening. Studies based on Belgian cohorts demonstrated that early identification through gNBS is associated with improved quality of life and reduced healthcare burden compared with later diagnosis (9). Importantly, cost-effectiveness analyses using real-world Belgian data showed that SMA screening combined with early treatment yields substantial gains in quality-adjusted life years, is cost-effective from a healthcare perspective, and even cost-saving from a societal perspective (10). These analyses incorporate not only medical costs but also caregiver burden, further strengthening the case for screening. This experience is directly relevant to the debate on genomic newborn screening. It shows that moving from pilot implementation to public policy requires more than technical feasibility: it requires demonstration of clinical benefit, operational integration, and long-term economic value. Belgium's early adoption and comprehensive evaluation of SMA gNBS have influenced global implementation. It was among the first countries to screen a significant proportion of newborns before nationwide adoption became more widespread, thereby contributing to international momentum toward broader screening programs. Overall, Belgium has been central in establishing both the clinical benefits and economic justification of SMA newborn screening, supporting its integration into public health systems worldwide. In that respect, the BabyDetect program extends Belgian leadership from single-disease molecular screening to panel-based genomic screening. By generating Belgian data on feasibility, acceptability, workflow integration, and the yield of actionable findings, BabyDetect shows that the value of gNBS lies not only in the sequencing panel itself, but also in demonstrating how screening, confirmation, counselling, and early specialist referral can be integrated within a real healthcare system. This places Belgium in a particularly strong position to contribute to the next phase of this debate.

Key Challenges and Open Questions

The scientific promise of gNBS should not obscure the fact that several important challenges remain before broader public implementation. Technical feasibility alone is not sufficient: condition selection, interpretation policy, economic evaluation, governance, and equity must all be addressed within a coherent public-health framework.

1. Define transparent criteria for condition selection. Condition and gene selection remain insufficiently harmonized. As demonstrated by Minten et al., there is striking variability in the genes included across programs (6). This reflects the lack of an agreed framework for determining which disorders are sufficiently severe, sufficiently actionable, and sufficiently childhood-onset to justify population screening. However, this is not unique to genomic screening as even traditional metabolic screening panels differ between countries or regions. Given that genomic NBS is a relatively recent development (less than 5 years), such variability is expected. Ongoing international collaboration will be essential to support harmonization while acknowledging that some differences will necessarily remain across settings considering treatment availability, health-system capacity and care pathways.
2. Define clinical pathways before implementation. Beyond panel composition itself, genomic newborn screening requires predefined pathways for confirmatory testing, specialist referral, counselling, and access to treatment. This is particularly important when screening identifies conditions requiring rapid intervention or family-based assessment. Without such pathways, genomic screening risks generating results that cannot be translated into timely clinical benefit.
3. Risk of False Positives and False Negatives. Test performance depends not only on sequencing quality but also on interpretation policy. False positives are an inherent risk in any screening program. In genomic screening, this risk is influenced by variant interpretation and reporting thresholds. Conversely, false-negative results may result from technical limitations, incomplete coverage of selected genomic regions, limited detection of specific variant classes, or the exclusion of genes and variants from the reporting framework. Conservative pipelines that report only pathogenic or likely pathogenic variants reduce false-positive findings and downstream anxiety, but they may also miss some affected children. This sensitivity-specificity trade-off should be made explicit and evaluated prospectively rather than handled implicitly within bioinformatic workflows.
4. Medico-Economic Impact. The medico-economic case is not yet established at population level. While traditional NBS has demonstrated clear economic benefits, genomic screening involves higher upfront costs and a much broader range of conditions. The overall value proposition of a broad genomic panel depends on long-term outcomes, downstream investigations, treatment access, healthcare utilization, and family impact. Medico-economic evaluation should also consider the position of gNBS within the broader landscape of genomic testing across the life course. Prenatal carrier screening, diagnostic sequencing in symptomatic children or adults, conventional and genomic newborn screening, and family cascade testing address related but distinct clinical questions and should be considered as complementary components of genomic medicine. However, coordinated pathways, shared infrastructure, and clear rules for data use and reinterpretation may reduce duplication and support efficient use of healthcare resources. In this context, gNBS may also reduce healthcare burden by identifying actionable conditions before symptom onset, shortening diagnostic odysseys, and avoiding sequential investigations after clinical presentation. Future evaluations will therefore require large populations and long-term follow-up to capture not only sequencing costs, but also confirmatory testing, avoided diagnostic investigations, follow-up, family-based assessment, data storage, reinterpretation, and care-pathway organization (11–15).
5. Optimal Sequencing Strategies. The choice of technology remains debated. Targeted next-generation sequencing currently offers advantages in cost control and interpretability, but with limited flexibility regarding potential additional genes/diseases, whereas whole-exome and whole-genome sequencing offer broader coverage and future reusability of data but generate additional challenges related to storage, interpretation, and governance. Though sequencing costs are rapidly decreasing, data storage, security, and long-term governance remain critical concerns.
6. Societal Impact, Education, and Counselling. The implementation of gNBS raises societal questions that extend beyond parental acceptability. Its public-health objective remains aligned with conventional newborn screening: the early identification of serious conditions before clinical presentation. As genomic technologies are integrated into this framework, information and counselling procedures will need to address consent, variant interpretation, residual risk, familial implications, data

governance, and the definition of reportable results (16,17). The BabyDetect feasibility study illustrates that structured information can be embedded in implementation through dedicated materials and trained consent procedures(1). At population level, this will require adequate training of healthcare professionals involved in prenatal, maternity, neonatal, paediatric, genetic, and primary care, and should be evaluated prospectively together with psychosocial impact, equity of access, and clinical utility (16,18).

7. Transition from Pilot Evaluation to Public Health Policy. Transitioning from pilot studies to publicly funded programs will require robust evidence, including clinical effectiveness, cost-effectiveness, ethical and societal acceptability. Given the scale of data required, this transition is likely to take time. At the same time, this transition is no longer purely theoretical: some jurisdictions have already begun to implement publicly supported or region-wide genomic newborn screening initiatives, including Puglia in Italy and Abu Dhabi in the United Arab Emirates, while in the United States, some state-level programs such as Florida have introduced publicly financed genomic sequencing in neonatal or infant care pathways (19–21). In Belgium, genomic screening is also beginning to enter clinical practice through BabyDetect, a prescription-based test costing approximately €650 and partially reimbursed by some insurance organizations. This type of trajectory is not unprecedented, as similar patterns have been observed with non-invasive prenatal testing (NIPT), carrier screening (which remains largely self-funded), and some vaccines such as meningococcal conjugated vaccines, where individual access preceded broader reimbursement. This development nonetheless raises important questions about equity and access, as preventive medicine introduced through individual pathways may increase disparities before broader public implementation becomes possible. These disparities are not only financial: unequal access to information, awareness, and counselling may also restrict genuine freedom of choice, since some families may not even know that such screening exists or may not be equally supported in understanding its benefits and limitations. In that sense, access prior to public funding may represent a transitional phase in the integration of new preventive strategies, but cannot replace the need for an equitable population-based strategy.

Conclusion

Genomic newborn screening represents a major evolution in neonatal healthcare. Emerging evidence from international and Belgian studies suggests that it can complement conventional newborn screening by identifying severe, actionable childhood-onset conditions that are not accessible to biochemical screening alone. The field has therefore reached a stage at which the central question is no longer technical feasibility in isolation, but how genomic screening should be defined, evaluated, and governed within a public health framework. Several uncertainties remain before broad implementation can be justified, including the selection of screened conditions, the balance between sensitivity and specificity in variant interpretation, the long-term clinical and psychosocial consequences of screening, the medico-economic impact at population level, and the preservation of equitable access. Addressing these issues will require large-scale prospective evaluation and sustained follow-up. Belgium is well positioned to contribute to the next phase of gNBS evaluation. Through its experience in neonatal molecular screening, its work in SMA screening and the BabyDetect pilot program, it has already generated important real-world evidence within its own healthcare context. The next phase should focus on consolidating and extending this evidence base, necessary to inform policy decisions. In practical terms, future efforts should focus on two priorities. First, ensuring that healthcare professionals involved in prenatal, maternity, neonatal, paediatric, genetic, and primary care are adequately informed about the benefits, limitations, and clinical pathways of gNBS is essential, so that unequal access to information does not itself become a source of inequity. Second, the establishment of a large-scale, publicly funded pilot program at the federal level based on multidisciplinary collaboration and integrated medico-economic assessment appears essential to generate the evidence needed to guide the future integration of genomic screening into public health systems.

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