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Editorial Note

New Horizons in Newborn Screening: Is Sequencing the Next Frontier?

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The *Journal of Pediatric and Neonatal Sciences* continues to look ahead, not only reflecting on where our specialty stands today, but also where it is heading. In this issue, we present a carefully curated selection of original research, reviews, and clinical insights that address both the enduring challenges and the exciting advances shaping paediatrics and neonatology. This Editorial Note focuses on one such rapidly evolving frontier: the expanding role of newborn sequencing (NS) within a unified neonatal screening programme (NSP).

The early success of neonatal screening was built on the timely identification of a small number of conditions, notably congenital hypothyroidism and phenylketonuria, first introduced into screening programmes in 1963 [1]. From these foundational beginnings, the field has progressed remarkably. The journey from Sanger sequencing in the early 1980s to next-generation sequencing technologies in the 2000s has transformed what is technically feasible. These advances have reduced costs, improved accuracy, and shortened turnaround times, bringing the prospect of incorporating genomic sequencing into routine NSP closer to reality [2]. In doing so, they open new possibilities for truly personalised preventive care.

An NS-based screening approach offers clear advantages. Most notably, it enables earlier identification and avoids protracted diagnostic pathways for a broader range of preventable conditions. Unlike traditional biomarker-based programmes, genomic screening also supports the detection of monogenic disorders from birth. As whole genome sequencing (WGS) has rapidly expanded, the American College of Medical Genetics (ACMG) classification has been instrumental in standardising variant interpretation, fostering consistency and comparability across global variant databases.

Large-scale initiatives exemplify this momentum. *The Generation Study*, led by Genomics England in partnership with the National Health Service (NHS, England), is analysing a single blood sample from over 100,000 newborns to explore genes associated with more than 200 rare but preventable conditions. Importantly, the scale of this study has been matched by a strong emphasis on ethics, transparency, and public trust—recognised as essential foundations for any future widespread implementation [3]. In many ways, this work seeks to bridge the longstanding gap between technological capability and real-world adoption of WGS in newborn screening.

Across the Atlantic, the GUARDIAN study in the United States provides further insight into the scope and complexity of NS implementation. This programme examines 255 conditions, including 63 neurodevelopmental disorders for which confirmatory tests are currently unavailable [4]. When viewed alongside other international NS initiatives, it becomes clear that the spectrum of conditions amenable to early genomic diagnosis will continue to expand, with opportunities for early intervention, surveillance, and preventive strategies.

Despite this promise, significant challenges remain. Limited understanding of genomic medicine among healthcare professionals, lack of wider public awareness, and unequal access to genetic testing infrastructure continue to pose barriers in many regions. Encouragingly, the global extension of NS research and the enrichment of variant databases are beginning to address the historical overrepresentation of White populations in genetic datasets [5]. This imbalance has long hindered accurate interpretation of variants in underrepresented groups and must be corrected to ensure equity in genomic medicine.

There are also clear clinical advantages to sequencing-based approaches. Conventional biochemical screening is known to be less reliable in preterm neonates, often necessitating repeat sampling and causing delays in diagnosis. Newborn sequencing circumvents many of these limitations, offering a more stable and definitive diagnostic platform from the outset [6].

As lessons emerge from these large-scale studies, there is growing optimism that consensus will develop around core gene lists and conditions best suited to NS. However, acceptability remains a critical consideration. Current NSPs report acceptability rates approaching 99% [7], a benchmark that may be difficult to sustain with the introduction of genomic sequencing initially.

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Data from the GUARDIAN study indicate an acceptance rate of approximately 72% within its first year, with parental concerns centred on genetic testing itself and the privacy of genetic data [4]. Scalability—both at the point of testing and during follow-up of positive screens—also presents practical challenges [8]. Moreover, the interpretation of genomic findings, particularly variants of uncertain significance (VUS), requires careful initial evaluation and ongoing reappraisal as international databases evolve.

Nevertheless, the direction of travel is clear. As next-generation sequencing technologies accelerate and genomic databases become increasingly comprehensive, the foundations are being laid for newborn sequencing to become an integral part of future screening programmes. This evolution will be supported by a skilled and motivated workforce and driven by continued technological innovation. What lies ahead is an opportunity to deliver preventive care earlier in life than ever before.

This topic will resonate with clinicians, researchers, and policymakers alike as we continue to explore and define the next chapter in newborn screening. We thank you for your continued engagement and support of the *Journal of Pediatric and Neonatal Sciences.*

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