https://doi.org/10.1038/s41576-026-00936-4

Navigating ethical, legal and social implications in genomic newborn screening

Anna C. F. Lewis, Gemma Brown, The GA4GH Working Group on genomic newborn screening & Yvonne Bombard

The implementation of genomic newborn screening (gNBS) poses myriad ethical, legal and social implications (ELSI) that require a public health ethics framework. Policy tools are needed to aid gNBS implementers in navigating key strategic ELSI questions to optimize implementation and realize the benefits of gNBS.

Newborn screening (NBS) is considered a premier public health success. NBS programmes cover an increasing proportion of babies worldwide, identifying a growing list of conditions that are severe, early-onset and treatable. Although the vast majority of screened conditions are genetic, to date, biochemical methods have been used predominantly as a first-line screen. However, decreasing sequencing costs and the large and increasing number of actionable genetic conditions undetectable by biochemical methods have prompted research to integrate genomic sequencing into NBS programmes (gNBS)[1]. The first such studies are at least 10 years old; however, there are currently dozens of projects, billed mostly as feasibility research studies, either launched or being launched around the world, mostly in well-resourced countries[2]. In 2025, Puglia, Italy, became the first jurisdiction to deploy genomic sequencing as a first-line screen alongside biochemical screening for all babies.

Those in the gNBS field must navigate many ethical, legal and social implications (ELSI). For example, should explicit consent be required for gNBS? What uses of the genomic data should be permitted, if any, beyond initial screening? Should the scope of conditions screened be increased to include those that are actionable but not treatable[3]? What about conditions that become actionable only later in childhood, or even in adulthood? gNBS raises a tension between public health and clinical care paradigms. For example, in clinical genetics, testing aims to improve the care of individuals and typically involves pre-test counselling and always requires explicit consent. Public health programmes, such as NBS, must benefit populations and be accessible, equitable, sustainable and represent a good use of often limited resources. In public health, we seek to advance the public benefits of a whole population, and often do so without explicit consent from individuals; NBS most commonly operates under an implied consent framework[4]. The gNBS field must not undermine these values nor the trust of the population.

Public health ethics and children's rights

Several overarching ideas are relevant to the ethical rollout of gNBS, although many implementation choices will need to be tailored to specific jurisdictions. Foremost is the central relevance of a public health ethics framework[4]. Such frameworks emphasize values such as public accountability, community engagement and trust, and stewardship of both resources and authority[5]. Because public health programmes operate in the best interests of a population, they should be informed by the views of that population[6].

Extending the benefits of NBS to infants with currently undetected conditions through gNBS must not disrupt existing NBS programmes or undermine the high levels of acceptance and public trust they rely on. Any implementation of gNBS within a public health ethics framework must be sustainable, equitable and focused on population benefit; this applies not just to the sequencing component, but to the entire system by which families are informed and children are followed up through downstream healthcare.

A framework that centres the health rights of children is also appropriate. Knoppers et al.[4] argue for the existence of a right for the asymptomatic at-risk child to be found. This right would support the inclusion of as many conditions that meet standard criteria as possible; however, operating within a public health ethics framework, such a right reinforces the need for gNBS to not disrupt existing NBS. The precise scope and limits of this right need to be specified — for example, whether it extends to children for whom treatments are not accessible or prognosis is uncertain. It must also be balanced against the right to not be exposed to undue harm from unnecessary surveillance or interventions that do not alter disease manifestation[7], as well as against programme feasibility.

Questions that guide decision-making

Several high-level questions to guide ELSI-relevant decision-making should be considered at the outset of gNBS research projects, pilot studies or implementations. Answers will be constrained, but not determined, by legal requirements and the broader health system.

What is the overall vision for how the potential implementation of gNBS will integrate with biochemical NBS, including the overall ethical justification or framing? One vision is to deploy sequencing like any other testing technology, with parameters as similar as possible to existing NBS. Another is to offer sequencing as a standalone consented test, alongside conventional NBS.

What consent process, if any, is envisioned for the potential deployment of gNBS, and how does this relate to how conventional NBS is implemented? Whether gNBS should be explicitly consented is a critical open question, further complicated by the fact that gNBS research projects necessarily require research consent.

How will the reuse of genomic data differ from the reuse of conventional NBS data? Most NBS programmes already store samples

and associated data and permit some research uses of these. Privacy concerns have led some to restrict allowable uses and shorten retention times[8,9]; gNBS will probably intensify these concerns owing to the uniquely identifying, predictive and long-term nature of genomic data.

Will there be the opportunity to screen or retest the same children in the future? This could include screening for conditions that become actionable later in childhood at a later stage. Keeping a genome 'on file' to inform a lifetime of care has long been a vision in genomic medicine, and in some integrated health systems, this possibility may be enabled by gNBS[1].

If the implementation effort is a research project or pilot study, what evidence does it need to generate to inform deployment decisions? Research success depends on informing implementation, including gathering data comparing the strategy of presymptomatic screening versus offering sequencing once children become symptomatic, and data to guide strategies for ELSI-laden decision points.

With whom should data be shared to maximize the positive effect on future children screened? Because many screened conditions are extremely rare, sharing outcome data will probably be central to programme success. Planning for such sharing within regulatory and other constraints is necessary to promote overall programme goals.

A policy tool

Answers to these key questions help shape strategies for specific ELSI-relevant decisions that arise across nine areas: stakeholder engagement during planning; consent models; conditions to include; testing and laboratory processes; results disclosure and management; post-test clinical management; data management; data revisiting for clinical purposes; and evaluation framework, governance systems and implementation continuity.

> "a roadmap … that is ethically robust, publicly accountable and worthy of the trust placed in NBS programmes"

We, an international and interdisciplinary group of gNBS experts, have designed a tool to aid in the navigation of ELSI for gNBS, under the auspices of the Global Alliance for Genomics and Health (GA4GH), an international community dedicated to advancing human health through genomic data. The GA4GH policy tool presents considerations for key strategic questions, as well as a list of 59 decision points with ELSI that we have identified on the basis of a scoping review of the field, conducted as part of our process[10]. Although the length of this list reveals the complexities of gNBS, delineating it should make navigation more tractable. For each decision point, we provide a brief synopsis of the ELSI at stake, along with an extended synthesis of the literature, highlighting implemented approaches where available. In addition to supporting individual projects, our tool also enables the comparison of approaches across projects.

Conclusions

A large range of ELSI with great complexity are integral to the successful implementation of gNBS. As gNBS projects and deployments within existing NBS programmes expand globally, across different jurisdictions with varying societal, economical and technological considerations, there is a need for structure and support to navigate this complexity. As genomic technologies are woven into NBS, the GA4GH policy tool offers a roadmap for doing so in a way that is ethically robust, publicly accountable and worthy of the trust placed in NBS programmes.

Anna C. F. Lewis [1,2,3] ✉, Gemma Brown[4], The GA4GH Working Group on genomic newborn screening* & Yvonne Bombard [5,6]

[1]Mass General Brigham, Boston, MA, USA. [2]Harvard Medical School, Boston, MA, USA. [3]Broad Institute, Boston, MA, USA. [4]Wellcome Genome Campus, Hinxton, UK. [5]Genomics Health Services Research Program, Li Ka Shing Knowledge Institute, St. Michael's Hospital, Unity Health Toronto, Toronto, Ontario, Canada. [6]Institute of Health Policy, Management, and Evaluation, University of Toronto, Toronto, Ontario, Canada. *A list of authors and their affiliations appears online.

✉e-mail: aclewis@bwh.harvard.edu

Published online: 04 February 2026

References

1. Stark, Z. & Scott, R. H. Genomic newborn screening for rare diseases. *Nat. Rev. Genet.* **24**, 755–766 (2023).
2. Minten, T. et al. Data-driven consideration of genetic disorders for global genomic newborn screening programs. *Genet. Med.* **27**, 101443 (2025).
3. Phillips, A., Siermann, M., Claesen-Bengtson, Z., Jackson, L. & Wright, C. F. What do we mean by actionability? Examining a key criterion in genomic prenatal and newborn screening. *Genet. Med.* **28**, 101624 (2025).
4. Knoppers, B. M., Bonilha, A. E., Laberge, A.-M., Ahmed, A. & Newson, A. J. Genomic sequencing in newborn screening: balancing consent with the right of the asymptomatic at-risk child to be found. *Eur. J. Hum. Genet.* **33**, 182–188 (2025).
5. Abbasi, M., Majdzadeh, R., Zali, A., Karimi, A. & Akrami, F. The evolution of public health ethics frameworks: systematic review of moral values and norms in public health policy. *Med. Health Care Philos.* **21**, 387–402 (2018).
6. Watts, G., Otlowski, M., Banks, R. & Newson, A. J. Three ethical approaches to expanding newborn screening through genomics: a critical comparative analysis. *BMC Med. Ethics* **26**, 167 (2025).
7. Horton, R. et al. Challenges of using whole genome sequencing in population newborn screening. *BMJ* **384**, e077060 (2024).
8. Douglas, C. M. W., van El, C. G., Faulkner, A. & Cornel, M. C. Governing biological material at the intersection of care and research: the use of dried blood spots for biobanking. *Croat. Med. J.* **53**, 390–397 (2012).
9. Lewis, M. H., Goldenberg, A., Anderson, R., Rothwell, E. & Botkin, J. State laws regarding the retention and use of residual newborn screening blood samples. *Pediatrics* **127**, 703–712 (2011).
10. Brown, G. et al. Genomic newborn screening: a scoping review of the field's evolution and associated ethical, legal, and social implications. Preprint at *SSRN* https://doi.org/10.2139/ssrn.5934255 (2025).

Acknowledgements

The authors acknowledge other members of the GA4GH genomic newborn screening working group, in particular A. Pichini, for their input to the policy tool.

Competing interests

Y.B. is co-founder of Genetics Adviser. The other authors declare no competing interests.

Related links

Global Alliance for Genomics and Health: https://www.ga4gh.org/about-us/
GA4GH policy tool: https://www.ga4gh.org/document/genomic-sequencing-in-newborn-screening-policy-tool/