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Genomic newborn screening: data retention for research and clinical reuse

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The integration of genomic sequencing into public health newborn screening (NBS), gNBS, could identify far more children that would meet screening guidelines than existing biochemical NBS. The retention of genomic data from gNBS could have huge benefits for research and could also enable potential clinical reuse. Many different ethical frameworks can support not seeking parental permission for traditional NBS, and indeed, most programs around the world do not do so, and are either mandatory or allow for an opt-out. Many NBS programs retain the underlying sample for anonymized or pseudonymized/coded research. This is proving to be a controversial aspect of NBS. While the appropriate consent regime(s) for the screening aims for gNBS remain unclear, we put forward arguments for the appropriate consent regimes for the retention and use of genomic data in gNBS. We review the different ethical frameworks that justify screening on the one hand, and further storage and uses of the data on the other. We argue that parental permission via an informed choice should be sought for genomic data retention for research purposes, that individual genomic data may be retained by the program for QA/QI purposes (but only for long enough to permit these purposes), and that no parental permission is needed to update aggregated genomic databases (e.g., allele frequencies). For clinical recontact, the appropriate consent regime for retaining genomic data will depend on the jurisdiction, but parents should be very thoroughly educated on the prospect of re-contact if this is planned.

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INTRODUCTION

The goal of classical newborn screening (NBS) programs is to detect asymptomatic newborns at risk for disorders that are serious and actionable during childhood [1]. NBS has been a huge public health success story. Consent regimes for NBS vary by jurisdiction (Box 1). In the majority of NBS programs, screening is mandatory or some form of implied consent is used, whereby prospective parents are given a brochure after birth or during the prenatal period informing them about the heel prick blood test to be performed at birth and informing them of the opportunity to ask questions. They are told that screening to find asymptomatic but at-risk newborns is carried out on all newborns. Following screening, if an at-risk newborn is found, a confirmatory diagnostic test is undertaken with parental consent. Parents may also be notified that they can choose to object and opt out prior to screening. There are also some jurisdictions—including, for example, Italy, Greece, Austria, and Nebraska [2, 3]—that have a mandatory NBS program, with no opt-outs.

Many projects around the world are currently assessing the feasibility of introducing genomic sequencing into NBS programs (gNBS) generally within the context of a research study [4]. gNBS is motivated by sequencing's ability to screen for more conditions and benefit more newborns [5, 6].

However, there are features of gNBS that suggest the need to consider whether their current consent regimes are appropriate [7]. Some of these features relate to the screening itself, specifically the large increase in the number of conditions that could be screened, and the ability to screen for conditions that do not meet the standard NBS criteria. There are also features of gNBS that relate specifically to potential retention of genomic data generated during gNBS. Such retention would enable research, and, new in the context of gNBS, potential clinical reuse (we delineate these potential uses below) [8–10].

We believe that it is too early to settle the case of the appropriate consent regime for gNBS screening, but that more can be said about the appropriate consent regimes for retention and use of the genomic data generated. We first outline the benefits and risks of genomic data retention, and then review the legal and ethical frameworks for different activities. We use this to argue that parental permission should in general be required. Exceptions are for aggregated genomic databases, for QA/QI (quality assurance/quality improvement) only for as long as necessary to achieve QA/QI goals, and, depending on local context, for clinical reuse. We indicate what further work is needed to operationalize these requirements.

GENOMIC DATA RETENTION

In traditional NBS, both the underlying sample, in this case typically a dried bloodspot card (DBS), and any connected metadata and generated data, are retained for some duration of time in order to enable suitable confirmatory tests to be

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performed, and for the lab to perform QA/QI activities. Many NBS programs will store samples beyond this initial time frame, to enable suitably qualified researchers access for studies that pass ethical review. Retention policies, including the period of retention, can differ dramatically between jurisdictions, in the case of different US states ranging from a few months to indefinitely [11].

gNBS programs would generate large amounts of genomic data, which can be categorized as outlined in Table 1: aggregated genomic data; anonymized individual level genomic data; pseudonymized/coded individual level genomic data; genomic data linked to identifiers. These are subject to different regulatory regimes. NBS programs will need to define policies relevant to these different data types, distinguishing by the intended use of that data. An individual's genomic data is currently generally considered "anonymized", though this may change. We note that on December 17th, 2025, the NIH proposed making such data subject to controlled access requirements [12].

There are three broad sets of purposes that would prompt retaining genomic data: improving the screening program, research, and clinical reuse. And there are associated risks. This changing set of benefits and risks for data retention poses a qualitatively different ethical challenge than consent for the initial screen.

Benefits

Program improvements via QA/QI. QA/QI is the ongoing use of data to monitor and enhance how well the program identifies, follows up, and benefits the target population. Such activities are a legal or regulatory requirement for clinical laboratories in many jurisdictions, including under the US Clinical Laboratory Improvement Amendments [13] and ISO 15189 accreditation frameworks in Europe and beyond [14]. Using genomic data to update allele frequency databases relied on to accurately interpret whether variants are pathogenic or benign would be in scope. The complete underlying genomic data could also be helpful, for example, as part of adding new genes to those screened, or for improving the phasing of variants. Determining appropriate data retention policies is further complicated by the fact that the boundary between QA/QI and research is inherently blurry—a challenge that predates gNBS [15], but that is especially acute for genomic data given its richness and multi-purpose utility.

Research. There is much excitement that data from gNBS will reveal more underlying genetic diversity than current volunteer population biobanks. Currently NBS programs include a large proportion of the population and the inclusion of this data (in aggregated genomic databases and/or in anonymized individual level genomic databases) could hence help address the lack of diversity and underrepresentation in genomic datasets, widely viewed as holding back genomic science and its applications [16–18]. Population-scale sequencing data could dramatically accelerate variant interpretation efforts by providing unbiased allele frequency data.

gNBS programs would also have the opportunity to establish pediatric population biobanks, by linking to other data sources such as electronic health records or public records. There are not that many longitudinal data sources for pediatric populations, and many benefits could follow from the establishment of these biobanks, including research that could eventually enable new treatments, including via facilitation of rare disease clinical trials. This could include the creation of unbiased penetrance and expressivity estimates, and a better understanding of the natural history of rare genetic conditions identified presymptomatically [10].

Clinical reuse. NBS has long grappled with research use; clinical reuse is substantially novel for gNBS. Retaining genomic data could enable: reanalysis as knowledge evolves (e.g., variants become classifiable as pathogenic or genes are added to screening panels); reporting of results that become actionable later in childhood; and access to data if a relevant phenotype emerges. We are not aware of any gNBS programs currently employing clinical re-use later in the lifecourse, though at least one program is actively considering this [19].

Risks

Privacy risks related to storing genomic data. Even in the case of anonymized or pseudonymized/coded data, privacy concerns can persist, given the (slight) possibility of reidentification. Re-identification risks are not static but depend on the (growing) availability of other information about the individual in public or commercial databases, as well as the regulatory and technical protections in place. Breaches of privacy could have long-lasting implications for the screened individuals and their families,

Table 1. Categories of data and relevant regulatory domains, using the EU and the US as examples.

Category of data	Description	Relevant regulatory domains
Aggregated genomic data	Examples include allele frequency databases (such as GnomAD) and genome wide association summary statistics	Generally falls outside of regulatory scope as not individually identifiable: not personal data under GDPR; not human subjects data under Common Rule; not PHI under HIPAA Re-identification risks from some aggregated data may lead to regulatory oversight, e.g., requirement for controlled access
Anonymized individual level genomic data	Examples include genomic reference databases such as 1KG data	Currently such genomic data is not in itself considered identifiable, and hence is not considered personal data under the GDPR, not identifiable private information under the Common Rule, and not PHI under HIPAA
Pseudonymized/coded individual level genomic data	Whoever is granted access (recipient) to the data does not have any identifying information nor the ability to reasonably obtain the identity of the individuals	Under GDPR, a recent ECJ decision (EDPS v SRB, 2025[57]) indicates this would not constitute personal data for the recipient; under the Common Rule, this would not be human subjects research; under HIPAA, this would not be PHI.
Genomic data linked to identifiers		Full protections apply: personal data under GDPR requiring consent; human subjects research under Common Rule requiring IRB oversight; PHI under HIPAA requiring Authorization or applicable exception

Table 2. Appropriate gNBS consent regimes for retained genomic data.

Category of genomic data	Supports which types of activity	Appropriate consent regime
Aggregated genomic data	QA/QI; research	Same as for initial screening
Anonymized individual level genomic data	QA/QI for the program	Same as for initial screening; should be retained only as long as necessary to support QA/QI
	If shared outside of the program (either open access or controlled access): other genetic testing/screening; research, especially population genomic research	Parental permission needed, either opt in or opt out depending on local context
Pseudonymized/coded individual level genomic data linked to other data, e.g., EHR for medical phenotypes, public records for social determinants of health	Can be used by the program to create pediatric population biobanks for all babies screened, to enable discovery research, including eventual therapy development	Parental permission needed, either opt in or opt out depending on local context
Genomic data linked to identifiers and a means to contact the child's parents	Clinical reuse: contacting parents if variant classification changes, or as new genes become clinically actionable, or for information that is actionable at older ages	Parental permission may be needed, depending on local requirements
Genomic data linked to phenotype for screen positive children	Evaluation of screening performance, refinement of condition lists, and assessment of long-term clinical utility/harms	Some uses are QA/AI and others will be seen as research, particularly if they involve sharing data with outside researchers. The line will depend on local policies/ requirements.

Parental permission could include a nurse or other professional who says "We're doing this, unless you say No" (opt out), or an offer to the parents for the particular uses of their newborn's genomic data (opt in). Which is more appropriate would depend on local context.

particularly in jurisdictions where protections against genetic discrimination are incomplete.

Beyond these objective risks, parental concerns about privacy — even where current re-identification risk is low — are themselves ethically significant. Parents express persistent and well-documented privacy concerns about storage of genomic data [20, 21]. Trust in the data steward is also a key determinant of parental decisions [22], and such trust can be systematically lower in those from groups previously harmed by the biomedical enterprise [23]. These concerns are not just hypothetical; privacy was the most common reason for decline in BabySeq [24].

The retention of dried blood spots for research has been a sticking point for NBS programs around the world. For example, in the Netherlands public attention was called to the retention of dried blood spots in 2000, and—based on poor parent understanding of long term retention and the lack of parental consent for such retention—subsequent policy discussions led to the establishment of destruction of samples after five years [15]. Concerns about state storage and secondary use of residual blood spots have been central issues in lawsuits in the US. In Texas, a lawsuit brought by parents prompted policy change to require explicit consent for dried blood spot retention and use for research [11]. There have been subsequent lawsuits in several other US states.

Costs of storing genomic data. There are substantial costs associated with storing genomic data (particularly raw data) for long periods of time, to have the computational infrastructure to retrieve it when necessary, and to have the governance structures in place to enable appropriate access to that data. These demands will be particularly high if the gNBS program chooses to make the data available to suitable third parties. Responsible data stewardship requires, as a matter of ethical best practice, that any proposed research use demonstrate equitable benefit to the communities contributing data, proportionate risk mitigation, and transparent governance with community input and oversight — safeguards grounded in the recognized need to maintain public trust and protect against exploitation in the absence of individual consent [25–27]. These demands could detract from the sustainability of the screening program.

Risks specific to clinical re-use. Expansion of return of results beyond initial screening for clinical re-use would be resource intensive and infeasible in many contexts. NBS programs have no ongoing clinical role with the child after the initial screening encounter; it is the child's pediatrician who is positioned to act on evolving clinical information over time. Pediatrician-mediated reuse, in which retained genomic data is made available through primary care rather than via direct recontact by the NBS program, may therefore be the more operationally viable model, though it would require data-sharing infrastructure between NBS programs and primary care that has yet to be established.

Additionally, clinical reuse may set parental expectations of ongoing follow-up — a presumption that any genomically relevant finding will prompt recontact. This risks undermining the bounded nature of NBS as a one-time screen rather than a longitudinal surveillance program.

RELEVANT FRAMEWORKS AND CONSENT

It is necessary to be deliberate about which ethical and legal principles we are applying to consent, and when these may differ by context between initial screening, retention for research, and retention for clinical reuse. We summarize our conclusions for genomic data retention and use in Table 2.

Existing NBS

There are several models of consent operating for biochemical NBS, see Box 1. Many jurisdictions do not require parental permission for newborn screening, based on the ethical justification that the potential harms to newborns from missed diagnoses outweigh considerations of parental autonomy. Conversely, shifting concerns about parental control over their newborns' healthcare and increasing attention to privacy around health data have led some jurisdictions, such as the UK, to adopt models that require parental permission [28].

At least four ethical frameworks relevant to NBS support either implied consent or mandatory screening. First, in the context of medical treatment consent is implied when a person agrees to become a patient, thereby accepting the constraints of a hospital or of the clinical setting, including the tests necessary to reach a

Box 1. Consent in NBS

In the context of newborn screening, Potter et al. distinguish two dimensions along which models of parental choice about NBS vary: first, whether parents have the authority to decide whether or not to have their infants screened, and (b) the extent to which parents are expected to engage in decision-making or think through the issues involved with NBS [53]. They further delineate the four implementation models that are in operation internationally, while acknowledging continua along both dimensions:

- Mandatory: parents have no authority to refuse screening, no expectation of parental engagement e.g., Nebraska, Austria, Italy, Mexico, Greece, Argentina.
- Opt-out: no decision required, no expectation of parental engagement. Parents do, however, have the authority to refuse screening. E.g. Netherlands, most Canadian provinces.
- Informed compliance: Decision required, little expectation for parental engagement. Also sometimes called "mandatory choice". Parents do give written or verbal informed consent, but, for example, operationalized as a notification or sign-off process. Given very high levels of compliance, it may not be presented as a true choice or otherwise routinized. E.g. France
- Informed choice: An opt-in decision required, definite expectation for parental engagement. E.g., the United Kingdom. Though note that it is unclear whether in the UK NBS is viewed as a choice [28]

diagnosis, propose a treatment, or suggest a prevention pathway. In contrast, interventions such as anesthesia or surgery may require a specific signed authorization. Overall, following admission to be treated at a clinic or hospital, consent to medical treatment need not be express, but rather *is usually implied from the words or conduct* of the patient [29]. (In contrast, for medical research, an express written consent is usually required though, increasingly, verbal consent may be accepted [30]). Today, once an infant is admitted, routine pediatric procedures are performed without parental permission from the parents since these are justifiable on the basis of an implied consent (usually verbal) to receive the professional standard of care. NBS programs (biochemical), when administered either in a hospital before discharge or in other clinical settings as part of newborn pediatric care, is seen in some jurisdictions as falling under this routine medical care approach [31]. In other jurisdictions where NBS is mandatory, screening is part of state-mandated pediatric care wherever administered.

Second, the absence of parental permission for newborn screening can be ethically and legally grounded within a child welfare framework and harm principal considerations under *parens patriae*, the legal doctrine whereby the state and the courts have the power to act on behalf of children for their protection as vulnerable citizens [32]. Different frameworks within pediatric bioethics set the threshold for state intervention differently [33–35], but under any of them the absence of parental permission reflects the view that screening carries minimal risk and that irreparable harm could be done if screening is delayed or not performed [36, 37]. In the case of parental objection to mandatory vaccination, the European Court of Human Rights held that the duty to vaccinate children fell within the State's margin of appreciation and was supported by medical evidence and public health interests, as well as being in the best interests of the child [38]. *Parens patriae* is a proportionality-constrained doctrine: it authorizes only the minimum intervention necessary to prevent a serious harm to the child, and it demands that such intervention be warranted by the child's own interest alone.

Third, rights based approaches within pediatric ethics further emphasize children as rights holders, and not just objects of protection. This includes rights to healthcare, and to the highest attainable standard of health [39, 40]. Knoppers et al. suggest the existence of "the right of the asymptomatic, at-risk newborn to be found" [41], based on the best interests of the child being primary [40], the right to the highest attainable standard of health [39, 40] and the human right to benefit from scientific advances [39, 42].

Fourth, a public health ethics framework is appropriate given that NBS searches and screens across a whole population [41]. Such frameworks are designed to mediate between individual rights and collective benefits. In this case, a public health ethics framework prioritizes the ability of the jurisdiction to be able to deliver the benefits to all, and making sure that neither benefits nor harms accrue systematically to subgroups [43]. Such frameworks also specify that interventions must satisfy proportionality i.e., that any intrusion on individual rights be no greater than necessary to achieve the public health goal, and that governance arrangements maintain public legitimacy through transparency and accountability to affected communities [44]. The accepted 1968 WHO screening criteria emphasize the need for: severity of the condition; possibility for pre-symptomatic detection; the availability of an effective treatment/actionability; validity/reliability/safety of the test; and cost-effectiveness [1]. Since 1968, other criteria have been added into public health programs, such as equity and access [45]. Public health approaches need to be sustainable, equitable, accessible, and make appropriate use of finite resources [11]. Lack of parental permission is justified based on the view that protection of children is in the public interest as well as in the child's best interests. Watts et al. argue that a public health ethics framework should be primary for gNBS, but that insights from individual-rights based and child welfare frameworks are additionally needed [46].

All four frameworks could be used to support current NBS being mandatory. Respecting parents rights to opt out are deemed to not outweigh the rights of the child to benefit from screening: "there are few, if any, compelling reasons why a competent parent would refuse to provide a simple and life-saving intervention for their child, or that the individual right of the parent to decide based on an unusual religious or political belief would outweigh the right of the infant to have an accepted treatment to prevent intellectual disability or death" [47].

However, several features of gNBS suggest that gNBS may warrant a more explicit consent process than traditional NBS, even where the latter does not require one [10, 41]. These include: the breadth of conditions screened, many of which lack the established actionability of traditional NBS targets; the generation of genomic data with implications for biological relatives; and the capacity for indefinite re-interrogation.

gNBS screening

Several suggestions have been made for consent regimes for gNBS [48]. These include tiered options for different groups of conditions (e.g., as implemented in the GUARDIAN study [49]), or including parental permission just for the genomic sequencing part of NBS. Alternatively, adding gNBS may lead to calls for making all of NBS (genomic and traditional) subject to parental permission [48].

We believe it is too early to settle the question of the appropriate consent regime for gNBS, because the relevant stakeholder engagement and associated ethical and legal analysis has not yet been completed. It will be essential to deepen engagement with parents, clinicians, legal and policy experts, and the public to assess the implications of maintaining mandatory or implied consent versus transitioning to a consented model for some or all aspects of gNBS. Such engagement is necessary to carefully weigh the possible benefits and harms of each approach, including how a consented model could empower families with greater choice, but might also result in fewer newborns being screened if consent rates decline, which would undermine its population goals.

Regardless of the model of consent adopted (including mandatory programs), all parents must receive suitable education, including on the nature of the test, the program's overall goals and limitations, and test limitations [5].

The approach to consent will depend on local context, including whether NBS already uses a consented model. Irrespective of the approach taken by a given jurisdiction to consent, the initial "case finding" approach on which population screening is based could well be undermined by the complexity of consent to gNBS [41]. At stake in the decision about the form of consent appropriate for gNBS is hence not just the successful rollout and uptake of a new technology, but maintaining trust in existing NBS programs.

Genomic data retention for QA/QI

This use can be justified in the same way as screening in the first place is justified (see section above, which all rest on a compelling balance of benefits to harms). We noted above the blurry line between QA/QI and research; heightened privacy concerns with genomic data would motivate erring on the side of caution. They would also suggest that data be kept for the minimum necessary time to enable these use cases. Programs should clearly delineate exactly how long they deem this time to be.

Genomic data retention for research

For screen-positive newborns, for which parents are standardly consented for follow-up testing, use of their data for research—e.g., condition prevalence and disease course—should follow the standard research consent paradigms used for data collected in medical settings.

From a regulatory point of view, the use of genomic data of all screened newborns without parental permission for inclusion in aggregated genomic databases, in anonymized genomic reference databases, or in pediatric population biobanks (using pseudonymized/coded individual-level genomic data) may be justifiable. This justification would come from a combination of compelling public interest, minimal individual risk, and precedent from other public health activities (e.g., disease surveillance) [50]. Waivers of individual express consent may be considered to be "in the public interest if there is substantial expected advancement of the health-related needs of members of a group whose interests are, or should be, of particular concern to the society in question" [27]. A de-identified genome may become re-identifiable over the course of a child's life, depending on the evolving data and regulatory environment, and in particular, what other genetic information and linked clinical data about the individual becomes available.

In the case of aggregated genomic databases use of gNBS genomic data is compelling as it poses incredibly low risks of re-identification. Such use should not require parental permission.

But for anonymized genomic reference libraries and for pseudonymized/coded individual level genomic data, even if this is legally permissible, there may be ethical and social reasons to seek parental permission. Such uses of genomic data without parental permission could lead to a loss of trust sufficient to jeopardize the goals of the screening program. Public attitudes research suggest a sizable minority of parents in many jurisdictions are concerned about having no control over the genomic data of their children [20, 21]. Public concern leading to restrictions on data sharing in the NBS context in the Netherlands and the US suggest that these concerns are not hypothetical [11, 15]; there is a real risk that the privacy concerns already present in NBS will increase when related to genomic data, and could lead to decline in trust, and more opt outs, in NBS in general.

Some jurisdictions have decided to implement parental permission for research in the context of NBS. In Victoria, Australia, parental permission for screening is explicit, and there is a separate consent for "secondary use of the sample in de-identified health research" [51]. Some US states, such as Michigan and Minnesota, require parents to give explicit permission before residual newborn screening samples can be used for research, even when the samples are coded or otherwise de-identified.

Equity concerns cut both ways. The case for broad retention is linked to the well-documented underrepresentation of diverse populations in genomic datasets: because NBS captures nearly all newborns regardless of whether families would voluntarily participate in research, gNBS data could help address consent bias — the tendency of voluntary research cohorts to systematically underrepresent certain communities — and the clinical disparities in variant interpretation this produces [16, 17]. However, communities historically marginalized by the biomedical enterprise have well-founded reasons to be cautious about having their children's genomic data retained without permission [23]; for these communities, the absence of meaningful consent risks eroding trust in NBS programs and reducing their sense of agency over their children's biological information. The appropriate balance between these considerations will depend on community and context.

While public health ethics frameworks would recognize substantial population-level benefit from research on retained genomic data, the same frameworks also emphasize that such benefits must not override individual rights or reasonably held community interests in control over the use of their children's genomic data. Where meaningful public unease persists, retaining data for research without explicit parental permission may fail the proportionality and legitimacy tests built into public health ethics [44]. It may fail proportionality because permission can be achieved using an opt-out model without burdening parents or undermining population health benefits. It may fail legitimacy because, as the Netherlands and US cases illustrate, retention without any permission mechanism has demonstrably eroded public confidence and constrained programs in practice. We hence argue that parental permission should be sought for sharing a newborn's genome with anonymized genomic reference databases or pediatric population biobanks. Such permission should not be "implied", but could be opt in (an offer to parents) or opt out (which could include a nurse or other professional who says "We're doing this, unless you say No"). Which is more appropriate would depend on local context, including the model adopted for gNBS as a whole.

The ambiguity inherent in the blurry line between QA/QI and bona fide research strengthens, rather than weakens, the case for transparency and parental permission. It suggests that part of a NBS program's ensuring ongoing public legitimacy will be careful delineation of exactly what uses of data, and why, will be considered to be necessary for improving the quality and utility of screening program data.

Genomic data retention for clinical reuse

As outlined above, there are risks associated with gNBS that could jeopardize the sustainability of the screening program, and a public health ethics lens would require this sustainability. For jurisdictions that are confident that NBS sustainability would not be impacted, the prospect of direct benefit to children would be aligned with a child-rights based approach, drawing on children's right to "the highest attainable standard of health" [9]. There is already precedent in clinical genetic testing to support the re-interrogation of genetic data as knowledge and phenotype evolve. The prospect of such clinical benefit could counterveil privacy concerns.

The appropriate parental permission for clinical reuse will depend on how gNBS is set up, and will be very sensitive to the education the parents receive. Above we delineated three main use cases: if genetic variation gains new clinical significance, for genetic variation that becomes relevant later in childhood, and to be reused if a phenotype suggestive of a genetic condition develops. It could be that some, but not all, of these uses are allowable without parental permission. Clear time limits on data

retention should be set in this context, in recognition of privacy concerns, and such data should only be retained if the program has active plans in place to enable these use cases.

CONCLUSION

The introduction of genomic sequencing into newborn screening programs raises many ethical, legal and social issues, but perhaps none more pressing to address than selection of the appropriate consent regime. While we believe that it is premature to settle the question of the most appropriate consent regime for gNBS screening itself, we have argued that there are no ethically compelling reasons to retain genomic data longer than necessary to meet the needs of screening in the absence of parental permission outside of some narrow use cases (see Table 1), namely to support QA/QI, to update aggregated genomic databases, and, potentially, and dependent on local context, for clinical reuse. Work is needed to clarify the QA/QI–research boundary, and to determine reasonable timelines for retention for QA/QI purposes. Exploration of potential clinical reuse is in its very early days, and much work lies ahead on this front for any gNBS program.

While there will be collective benefits to research performed on this data, there are also associated risks, and the balance of these benefits to risks is not yet clear. Even if it were to become much clearer through research, it is in the eyes of the public that this balance must be compelling, as any concerns could contribute to a loss of trust in NBS programs.

Given the very different ethical justifications for retention for research and for retention for clinical reuse, separate consents for each should be implemented where appropriate. Programs will need to determine whether this is best offered as an opt-in or opt-out in their specific context. Additional tiers of consent, for example, for enabling commercial versus non-commercial uses, could also fruitfully be explored in gNBS research projects.

We emphasize that consent for genomic data and reuse is only meaningful if supported by adequate communication and education. The educational burden for gNBS will already be high, requiring education for both professionals and the public, and in particular the "prenatal" public, delivered in a multi-modal fashion [52]. Parents will need clear answers related to data management and security plans, and what will happen to the sample and generated data both in the scenario where they do not explicitly opt in for retention, and when they do. Without such clarity, parental permission cannot function as an ethically robust safeguard.

REFERENCES

1. Wilson JMG, Jungner G. Principles and practice of screening for disease. World Health Organization; 1968.
2. Loeber JG, Platis D, Zetterström RH, Almashanu S, Boemer F, Bonham JR, et al. Neonatal screening in Europe revisited: an ISNS perspective on the current state and developments since 2010. Int J Neonatal Screen. 2021;7:15.
3. Kelly N, Makarem DC, Wasserstein MP. Screening of newborns for disorders with high benefit-risk ratios should be mandatory. J Law Med Ethics J Am Soc Law Med Ethics. 2016;44:231–40.
4. Minten, T, Bick, S, Adelson, S, Gehlenborg, N, Amendola, LM, Boemer, F, et al. Data-driven consideration of genetic disorders for global genomic newborn screening programs. Genet Med. 2025;27:101443.
5. Mori M, Chaudhari BP, Ream MA, Kemper AR. Promises and challenges of genomic newborn screening (NBS) - lessons from public health NBS programs. Pediatr Res. 2025;97:1327–36.
6. Friedman JM. Implementing genomic newborn screening as an effective public health intervention: sidestepping the hype and criticism. npj Genom Med. 2024;9:70.
7. Tomlinson AN, Skinner D, Perry DL, Scollon SR, Roche MI, Bernhardt BA. 'Not tied up neatly with a bow': professionals' challenging cases in informed consent for genomic sequencing. J Genet Couns. 2016;25:62–72.
8. Biesecker LG, Green ED, Manolio T, Solomon BD, Curtis D. Should all babies have their genome sequenced at birth? BMJ. 2021;375:n2679.
9. Gyngell C, Lynch F, Vears D, Bowman-Smart H, Savulescu J, Christodoulou J. Storing paediatric genomic data for sequential interrogation across the lifespan. J Med Ethics. 2025;51:205–11.
10. Stark Z, Scott RH. Genomic newborn screening for rare diseases. Nat Rev Genet. 2023;24:755–66.
11. Lewis MH, Goldenberg A, Anderson R, Rothwell E, Botkin J. State laws regarding the retention and use of residual newborn screening blood samples. Pediatrics. 2011;127:703–12.
12. NOT-OD-26-023: Request for Information on Draft NIH Controlled-Access Data Policy and Proposed Revisions to NIH Genomic Data Sharing Policy. https://grants.nih.gov/grants/guide/notice-files/NOT-OD-26-023.html.
13. 42 CFR Part 493 -- Laboratory Requirements. https://www.ecfr.gov/current/title-42/part-493.
14. ISO 15189:2022. *ISO* https://www.iso.org/standard/76677.html.
15. Douglas CMW, van El CG, Faulkner A, Cornel MC. Governing biological material at the intersection of care and research: the use of dried blood spots for biobanking. Croat Med J. 2012;53:390–7.
16. Wojcik GL, Graff M, Nishimura KK, Tao R, Haessler J, Gignoux CR, et al. Genetic analyses of diverse populations improves discovery for complex traits. Nature. 2019;570:514–8.
17. Manrai AK, Funke BH, Rehm HL, Olesen MS, Maron BA, Szolovits P, et al. Genetic misdiagnoses and the potential for health disparities. N Engl J Med. 2016;375:655–65.
18. Martin AR, Kanai M, Kamatani Y, Okada Y, Neale BM, Daly MJ. Clinical use of current polygenic risk scores may exacerbate health disparities. Nat Genet. 2019;51:584.
19. Leblond M, Galati M, Roberts J, Etheredge H, Willacy N, Özkurt Ö, et al. Co-creating the experience of consent for newborn genome sequencing: the generation study. Public Health Genom. 2024;27:210–27.
20. Hui LL, Nelson EAS, Deng HB, Leung TY, Ho CH, Chong JSC, et al. The view of Hong Kong parents on secondary use of dried blood spots in newborn screening program. BMC Med Ethics. 2022;23:105.
21. Cunningham S, O'Doherty KC, Sénécal K, Secko D, Avard D. Public concerns regarding the storage and secondary uses of residual newborn bloodspots: an analysis of print media, legal cases, and public engagement activities. J Community Genet. 2015;6:117–28.
22. Bombard Y, Miller FA, Hayeems RZ, Carroll JC, Avard D, Wilson BJ, et al. Citizens' values regarding research with stored samples from newborn screening in Canada. Pediatrics. 2012;129:239–47.
23. Shavers VL, Lynch CF, Burmeister LF. Knowledge of the Tuskegee study and its impact on the willingness to participate in medical research studies. J Natl Med Assoc. 2000;92:563–72.
24. Genetti CA, Schwartz TS, Robinson JO, VanNoy GE, Petersen D, Pereira S, et al. Parental interest in genomic sequencing of newborns: enrollment experience from the BabySeq project. Genet Med J Am Coll Med Genet. 2019;21:622–30.
25. Buseh AG, Stevens PE, Millon-Underwood S, Townsend L, Kelber ST. Community leaders' perspectives on engaging African Americans in biobanks and other human genetics initiatives. J Community Genet. 2013;4:483–94.
26. Kraft SA, Cho MK, Gillespie K, Halley M, Varsava N, Ormond KE, et al. Beyond consent: building trusting relationships with diverse populations in precision medicine research. Am J Bioeth AJOB. 2018;18:3–20.
27. Ballantyne A, Schaefer GO. Public interest in health data research: laying out the conceptual groundwork. J Med Ethics. 2020;46:610–6.
28. Ulph F, Dharni N, Bennett R, Lavender T. Consent for newborn screening: screening professionals' and parents' views. Public Health. 2020;178:151–8.
29. Robertson, GB. Legal Liability of Doctors and Hospitals in Canada, 5th edn. Carswell: Toronto, Ontario; 2017.
30. Noë A, Vaillancourt E, Zawati MH. Verbal consent in biomedical research: moving toward a future standard practice?. Front Genet. 2025;16:1472655.
31. Bombard Y, Miller FA, Hayeems RZ, Barg C, Cressman C, Carroll JC, et al. Public views on participating in newborn screening using genome sequencing. Eur J Hum Genet. 2014;22:1248–54.
32. Faden RR, Holtzman NA, Chwalow AJ. Parental rights, child welfare, and public health: the case of PKU screening. Am J Public Health. 1982;72:1396–1400.
33. Kopelman LM. The best-interests standard as threshold, ideal, and standard of reasonableness. J Med Philos. 1997;22:271–89.
34. Diekema D. Parental refusals of medical treatment: the harm principle as threshold for state intervention. Theor Med Bioeth. 2004;25:243–64.
35. Gillam L. The zone of parental discretion: an ethical tool for dealing with disagreement between parents and doctors about medical treatment for a child. Clin Ethics. 2016;11:1–8.

36. Faden R, Chwalow AJ, Holtzman NA, Horn SD. A survey to evaluate parental consent as public policy for neonatal screening. Am J Public Health. 1982;72:1347–52.
37. Currier RJ. Newborn screening is on a collision course with public health ethics. Int J Neonatal Screen. 2022;8:51.
38. Vavřička and Others v. the Czech Republic. European Court of Human Rights, Grand Chamber. 2021. https://hudoc.echr.coe.int/fre?i=001-209039 (accessed 6 Dec 2025).
39. UN General Assembly. International Covenant on Economic, Social and Cultural Rights. 1966. https://www.ohchr.org/en/instruments-mechanisms/instruments/international-covenant-economic-social-and-cultural-rights (accessed 6 Dec 2025).
40. UN General Assembly. Convention on the Rights of the Child, 20 November 1989. https://ihl-databases.icrc.org/en/ihl-treaties/crc-1989/article-24 (accessed 6 Dec 2025).
41. Knoppers BM, Bonilha AE, Laberge A-M, Ahmed A, Newson AJ. Genomic sequencing in newborn screening: balancing consent with the right of the asymptomatic at-risk child to be found. Eur J Hum Genet EJHG. 2025;33:182–8.
42. Nations U. Universal declaration of human rights. United Nations; 1948.
43. Newson AJ. The promise of public health ethics for precision medicine: the case of newborn preventive genomic sequencing. Hum Genet. 2022;141:1035–43.
44. Childress JF, Faden RR, Gaare RD, Gostin LO, Kahn J, Bonnie RJ, et al. Public health ethics: mapping the terrain. J Law Med Ethics J Am Soc Law Med Ethics. 2002;30:170–8.
45. Andermann A, Blancquaert I, Beauchamp S, Déry V. Revisiting Wilson and Jungner in the genomic age: a review of screening criteria over the past 40 years. Bull World Health Organ. 2008;86:317–9.
46. Watts G, Otlowski M, Banks R, Newson AJ. Three ethical approaches to expanding newborn screening through genomics: a critical comparative analysis. BMC Med Ethics. 2025;26:167.
47. McCandless SE, Wright EJ. Mandatory newborn screening in the United States: History, current status, and existential challenges. Birth Defects Res. 2020;112:350–66.
48. Friedman JM, Cornel MC, Goldenberg AJ, Lister KJ, Sénécal K, Vears DF. Genomic newborn screening: public health policy considerations and recommendations. BMC Med Genom. 2017;10:9.
49. Ziegler A, Koval-Burt C, Kay DM, Suchy SF, Begtrup A, Langley KG, et al. Expanded newborn screening using genome sequencing for early actionable conditions. JAMA. 2025;333:232–40.
50. Attachment E: Recommendations Regarding Research Uses of Newborn | HHS.gov. https://www.hhs.gov/ohrp/sachrp-committee/recommendations/2015-april-24-attachment-e/index.html (2015).
51. Charles T, Pitt J, Halliday J, Amor DJ. Implementation of written consent for newborn screening in Victoria, Australia. J Paediatr Child Health. 2014;50:399–404.
52. Rothwell E, Wong B, Anderson RA, Botkin JR. The influence of education on public trust and consent preferences with residual newborn screening dried bloodspots. J Empir Res Hum Res Ethics JERHRE. 2016;11:231–6.
53. Potter BK, Etchegary H, Nicholls SG, Wilson BJ, Craigie SM, Araia MH. Education and parental involvement in decision-making about newborn screening: understanding goals to clarify content. J Genet Couns. 2015;24:400–8.

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All authors conceived of the study, ACFL and BMK drafted the initial draft, all authors revised the draft and approved of the final version.

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COMPETING INTERESTS

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