

Sequencing for a Lifetime: Value, Feasibility, and the Governance Gap in Lifelong Genomic Medicine

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Abstract

Background A vision of lifelong genomic medicine, in which stored genomic data can inform a lifetime of care has long animated the field of genomic medicine. Component pieces of this vision are being researched or are already in clinical practice, including dozens of projects around the world sequencing healthy newborns, along with reanalysis of stored genomic data. Whether lifelong genomic medicine is desirable, and, if so, whether it is feasible, has not been explored in the literature.

Methods and Findings We conducted and thematically analyzed interviews with over 50 US-based healthcare professionals, including clinical geneticists, genetic counselors, primary care clinicians, laboratory personnel, and those who have implemented genomic screening in health systems. We found broad endorsement of the value of lifelong genomic medicine across groups. Perceived clinical value stemmed from the existence of genomic information relevant at multiple stages of life, the ability to query the genome if an individual's medical circumstances change, and the ability to inform patients about relevant evolving scientific advances. Participants also articulated an efficiency argument for reanalyzing stored genomic data rather than retesting. The clinical value was contested by a few participants, who argued for more targeted testing for the clinical situation and disputed the efficiency argument. Many participants viewed the model as inevitable, with operational precedent already established for many component activities. The feasibility of lifelong genomic medicine was limited not by scientific barriers but by governance gaps spanning delivery models, consent, data stewardship, recontact, and the pediatric-to-adult transition. These gaps have equity implications that are cumulative and mutually reinforcing.

Conclusions The concept of lifelong genomic medicine was widely viewed as acceptable and desired. However, until the governance infrastructure is established, including accountability, funding, data stewardship, and recontact mechanisms, population-scale genomic sequencing risks proceeding faster than the frameworks needed to make it responsible.

Introduction

A proposed vision for the future of population-wide genomic medicine is to sequence babies and to keep the genome “on file” to inform a lifetime of care [1–4]. For example, Biesecker et al., advocate for the genome to be sequenced at birth, and genetic results to be sequentially disclosed over time “at a life stage when interventions are known to be beneficial” [1]. They give examples of newborn screening conditions at birth, Wilms’s tumour and retinoblastoma to children, aortopathy and cardiomyopathy to teenagers, and carrier risks and colon and breast cancer risk to adults. Storing an individual’s genome would also enable recontact when knowledge changes, either because new information changes the classification of variants [5], new genes are linked to conditions [6], or because new interventions — and in particular an increasing array of gene and cell therapies — suggest the benefit of reporting more results. It could also enable querying for information that might shed light on a newly developed clinical phenotype. We refer to this vision of referring back to a sequenced genome to inform a lifetime of care as “lifelong genomic medicine,” and it has long animated the field of genomic medicine [7]. With improvements in technology, data management and AI, there is genuine excitement about this finally being possible.

Many research projects around the world are sequencing newborns to study the integration of genomic sequencing into newborn screening or the newborn period [8]. The prospect of lifelong genomic medicine is hence becoming less abstract. For example, the UK government is currently sequencing 100,000 newborns as part of Genomics England’s Generation Study. In addition, they aim to “explore the risks and benefits of storing an individual’s genome over their lifetime” [9]. Outside the context of screening newborns, there are several implementation studies that maintain genomic data on file as a resource, for example from the Geno4ME Study [10,11]. Several companies are also offering a sequencing and storage model that becomes a “lifetime digital genomic resource” [12].

It may, however, be that the benefits of keeping a genome on file are outweighed by costs of referring back to legacy data, the risks of discriminatory misuse, and the challenges of data governance [13,14]. Alternative models include not storing the genomic data and instead resequencing the genome if there is a clinical indication [15], or only sequencing a subset of the genome (i.e., panels) [14].

Because many of the component pieces of lifelong genomic medicine are already in place — storage of data sequenced in the context of a clinical indication, reanalysis of genomic data, recontacting patients if variants are reclassified, construction of age-based panels based on actionability — the views of professionals on the feasibility and value of lifelong genomic medicine

could refine this vision, including potentially suggesting that the vision should be abandoned. As those who would be responsible for delivering, interpreting, and communicating genomic information across a patient's lifetime, healthcare professionals are uniquely positioned to assess both the clinical value and the feasibility of lifelong genomic medicine, and their views may shape whether such programs are adopted.

Two qualitative interview studies with Australian healthcare professionals on genomic newborn screening touch on the potential for genomic data to be retained and reused across the life course, with participants in both studies seeing value in long-term storage for future reanalysis [16,17]. To our knowledge, no studies have examined professional perspectives on lifelong genomic medicine as a central question.

We conducted semi-structured interviews with over 50 US-based healthcare professionals to gain their perspectives on retaining and reusing genomic data over a lifetime, including clinical geneticists, genetic counselors, lab personnel, primary care clinicians (PCCs), and those with experience implementing population screening in healthcare systems. Here we report their perspectives on the value and feasibility of lifelong genomic medicine. Results related to the staging of genomic information by age will be reported separately.

Methods

Participants

Multiple types of healthcare professionals were recruited into this qualitative interview study to enable triangulation of perspectives. This included clinical geneticists, genetic counselors (pediatric, adult, and maternal fetal medicine) and primary care clinicians. While the genetics providers have the most experience with genomic medicine, primary care clinicians would likely be closely or even primarily involved if lifelong genomic medicine were to be implemented. Laboratory personnel, mostly laboratory directors, were also included, as they have experience managing genomic data. Finally, we included those who have led operational roles integrating genomic screening (i.e. genomic testing in the absence of an indication) into health systems. Some of these individuals were genetic counselors or clinical geneticists; others were primarily administrators with leadership roles (including some who had MDs). We refer to this group as “genomic screening implementers”.

We used purposive sampling to recruit individuals from all groups. An invitation to participate was distributed via email, some individualized and some via listservs. We used snowball sampling to recruit additional participants, asking interviewees for recommendations of others we should interview within their own institutions, particularly those who might have different perspectives or different subject area expertise on the themes covered.

We recruited healthcare providers from some of the BabySeq Project sites [18]. The BabySeq project studies the integration of genomic sequencing into newborn care and has recruited hundreds of babies across the US. For our study, the BabySeq sites we recruited from were Boston Children's Hospital, the University of Alabama Medical Center in Birmingham, Corewell Health in Detroit, and Children's Hospital of Philadelphia, including those working in academic hospitals and community clinics. We also invited genetic counselors at the University of Pennsylvania Health System, a sister institution to the Children's Hospital of Philadelphia. The study was advertised to individuals at these institutions via email listservs. Response rates could not be calculated for participants recruited, as the total number of recipients was not ascertained.

We identified the most active laboratories that conduct genomic sequencing in the US from the list of top submitters of variants to ClinVar and contacted their laboratory directors via email with an invitation to participate [19]. Of 14 emailed, 7 accepted the invitation. For genomic screening implementers (those with experience implementing programs that have launched genomic screening programs), a narrative review identified ten clinical and research programs that use genomic sequencing for population-based screening in a healthcare system in the US [20]. The names and email addresses of individuals associated with these programs were ascertained from publicly available information and the authors knowledge. The list was supplemented with others known to the research team to be involved in projects not covered by the review. Email invitations were sent to 18 individuals from this list, 12 of whom agreed to participate.

All prospective participants received a fact sheet with basic information about the study, including its purpose, before agreeing to participate (see Supplementary Information).

Data Collection

Semi-structured interviews were conducted using an interview guide (see Supplementary Information) that was designed based on review of the literature and knowledge of the research team. Some components of the interview guide were common across all types of providers; some were specific to clinical versus operational roles. Topics covered included desirability of lifelong genomic medicine, and facets of current day processes to understand feasibility, including, for example, clinician experiences with longitudinal relationships, current treatment of sequence information after a report is generated, privacy considerations.

All interviews were conducted by AL between August 2024 and February 2025. Interviews were approximately 60 minutes long, conducted over Zoom videoconferencing and recorded. Only the interviewer and interviewee were present. Field notes were taken immediately after each interview. Auto-generated transcripts were manually cleaned and de-identified to protect privacy and confidentiality. Transcripts were not returned to participants. A short demographic survey was emailed to participants after the interview. Interviewees were mailed a \$100 check as a thank you for their time. No repeat interviews were conducted.

Analysis

De-identified transcripts were coded by AL. Analysis drew on principles of framework analysis [21,22], combining deductive and inductive elements. Domains were defined *a priori* based on the research questions. The study adopted a pragmatist orientation, prioritizing the generation of findings with direct relevance to policy and practice. Within each domain, transcripts were inductively coded using Atlas.ti software inductively using *in vivo* and descriptive codes [23]. Codes were iteratively grouped into themes and subthemes through repeated comparison across codes, transcripts, and stakeholder groups; the coding framework underwent multiple revisions as groupings were refined. Four transcripts were coded by a second author (IH) who helped identify themes and refine the codebook. Code frequency was tracked by stakeholder group throughout, enabling systematic comparison of where professional perspectives converged and diverged. Throughout analysis, AL maintained analytical memos documenting evolving interpretations as the coding framework developed. Emerging themes were additionally sense-checked through presentation to and discussion with research colleagues. No substantively new themes emerged in the final interviews within each participant type. Participant checking of findings was not conducted; given the multi-stakeholder design and the analytical focus on cross-group comparison, we prioritized peer debriefing and co-coding over individual participant validation. The complete coding framework, including code frequencies by stakeholder groups, is provided in the Supplementary Information.

During this process, coded materials pertaining to population genomic screening broadly — rather than lifelong genomic medicine — were separated for analysis elsewhere. This paper focuses specifically on lifelong genomic medicine. Codes related to the staging of information by age are also analyzed elsewhere.

As a researcher working at the intersection of genomics and ethics, AL (DPhil, she/her) was a partial insider to the professional community studied. This may have facilitated candor but may also have led some participants to foreground ethical considerations. A small number of participants were known to AL prior to the study; all participants were informed of the confidentiality of their responses. IH (MD) is a clinical geneticist and researcher, and her expertise provided an additional analytical perspective and helped surface assumptions that might otherwise have gone unexamined. Both AL and IH are experienced qualitative researchers.

This study was approved by the Mass General Brigham IRB.

Results

Participant characteristics

Fifty-two healthcare professionals participated, including genetic counselors (n=16), clinical geneticists (n=6), laboratory personnel (n=8), primary care clinicians (n=10), and genomic screening implementers (n=12). Participants were predominantly female (n=28) and White (n=36),

with a mean age of 45 years (range 27–69) and a range of career experience from early-career to highly senior professionals ([Table 1](#)).

Table 1. Participant Characteristics.

Demographic data were missing for 4 participants (3 Primary Care Clinicians and 1 Genetic Counselor (adult)) and are excluded from the counts presented.

Perceived Value of lifelong genomic medicine

Many participants felt that the lifelong genomic medicine model has clinical value, could help advance future research, and may be more cost effective than other models of genomic medicine. Although they recognized the complexity of this approach, and some questioned whether the investment needed was justified in light of alternative models that are available, many felt that the lifelong genomic medicine model is already being developed and its implementation is inevitable.

Clinical Value across the lifespan

There was broad agreement across participant groups that genomic information becomes relevant at different life stages, making the lifelong genomic medicine model clinically and intuitively attractive, see [Figure 1](#). Examples cited included adult-onset actionable conditions, carrier screening at reproductive age, and polygenic risk for common disease, including predispositions relevant to adolescence such as mental health and substance use.

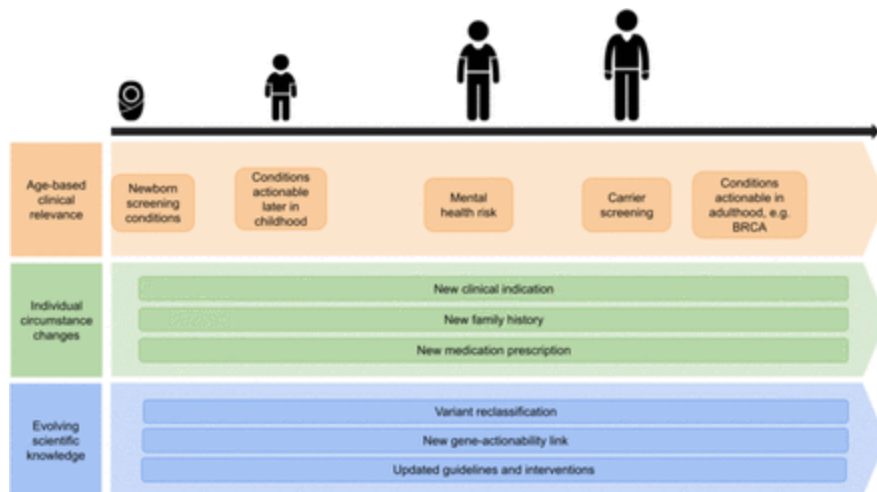


Figure 1: Clinical value of the lifelong genomic medicine model

Participants also saw value in the lifelong genomic medicine model as individual circumstances change. This included when medications are prescribed (pharmacogenomic results), when family history changes, and when phenotypes emerge or change. Scientific progress was also identified as a source of ongoing value. This included the evolution of knowledge linking genotype to phenotype, both for updates to variant-level pathogenicity classifications and new associations

between genes and diseases. It also included updates to guidelines, which could include new interventions associated with particular genetic variation, or new data on optimal screening regimens.

Beyond individual clinical benefit, participants, in particular genomic screening implementers, identified the potential for broader population-level learning to advance research, improve variant interpretation, and enable a learning health system. Moreover, this value could aid institutional buy-in.

Participants saw value in storing and reanalyzing genomic data over retesting. Genetic counselors and lab personnel noted that compared to retesting, reanalysis was cheaper, faster, and does not require a new sample. Others stressed that current practice of not reusing data is wasteful. As one genomic screening implementer put it, *“if you think about how we use genetics traditionally, it’s a waste... You order these multiple tests, they land in the medical record, they get lost, people get multiple tests ordered of the same thing”* (P24, genomic screening implementers).

The lifelong genomic medicine model depends on having broad genomic data available for future queries, and participants saw this as justified because broader testing has repeatedly proven its value. They also noted that the increasing norm of running virtual panels on exome or genome backbones meant the distinction between “panel” and “genome” was becoming artificial. Genomic screening implementers noted that patients themselves sometimes expressed frustration that previously collected genomic data could not be re-analyzed.

Perceived inevitability and its limits

Across many participant groups there was a pervasive sense that some form of lifelong genomic medicine was inevitable. For example one pediatric genetic counselor noted *“I have yet to see any major barriers that would stop this train from going down the tracks”* (P3, GC pediatrics). Genomic screening implementers were particularly emphatic, for example *“every other system that we talk to, with very few exceptions, are bullish. They’re like, ‘Yeah, this isn’t an if, it’s a when’”* (P24, health systems).

Participants across most groups articulated the complexity involved in implementing the lifelong genomic medicine vision. As one adult genetic counselor observed, *“I mean, it’s not impossible. I just — there’s so many moving parts”* (P43, GC adult). Another participant articulated concern about reputational risk if the complexity were not adequately handled.

Genomic screening implementers counterbalanced these concerns with pragmatism: it is acceptable not to get everything right the first time, and that the scale of the challenge should not be allowed to freeze the process.

A smaller number of participants questioned whether investment needed for lifelong genomic medicine was justified. The argument that finite resources might be better directed toward expanding access to interventions with a more established evidence base — what several

described as “low-hanging fruit” — was made most pointedly by laboratory personnel, but also by some genetic counselors and genomic screening implementers. One lab participant captured the counter narrative to data accumulation: *“I feel like my opinion has changed dramatically over the last decade. I very much began as a ‘data is awesome, more data is more awesome’ person, and now I’m more in the camp of ‘data is a burden’. There are important questions that you can answer effectively now, and there’s ways of making that harder for yourself.’ In many cases, more data just makes it harder to do the thing that you’re trying to do now”* (P10, lab personnel).

Some participants raised retesting rather than reanalysis as an alternative to maintaining stored data. One clinical geneticist noted that it was currently faster to order a new test than to get reanalysis results. And two participants described the “one and done” philosophy as naive. *“I’m wary of a public endeavour to say just have this one test, and you’re set up for life, because that’s not the truth... There’s no one single diagnostic test that’s going to cover you for everything. I talked to so many parents about this, and they say, ‘So I never have to have any other genetic test, because I had all my genetics looked at?’”* (P3, GC pediatrics).

Some participants, particularly laboratory personnel, questioned the need for whole genome sequencing. They stressed that genome-wide data is very complicated, and that there is no other area of medicine where we collect information because it might be useful in the future, and that they believe the focus on collecting genome-wide data drives payer anxiety about downstream costs. An advantage of gene panels raised by one PCC is that panel testing could remove the burden of choice from parents about what to learn when.

Perceived feasibility of lifelong genomic medicine

Participants identified a series of structural prerequisites for lifelong genomic medicine, spanning the choice of delivery model, data governance, patient-facing processes, the longevity and reusability of stored data, and the mechanisms by which stored data would generate clinical action over time. For many individual components, operational precedent exists; the challenges lie in connecting them into a coherent system, and in governance gaps that cut across nearly every area. Delivery model options are compared in [Table 2](#). [Figure 2](#) maps the component infrastructure required; [Table 3](#) details the operational precedent, unresolved challenges, and equity implications for each.

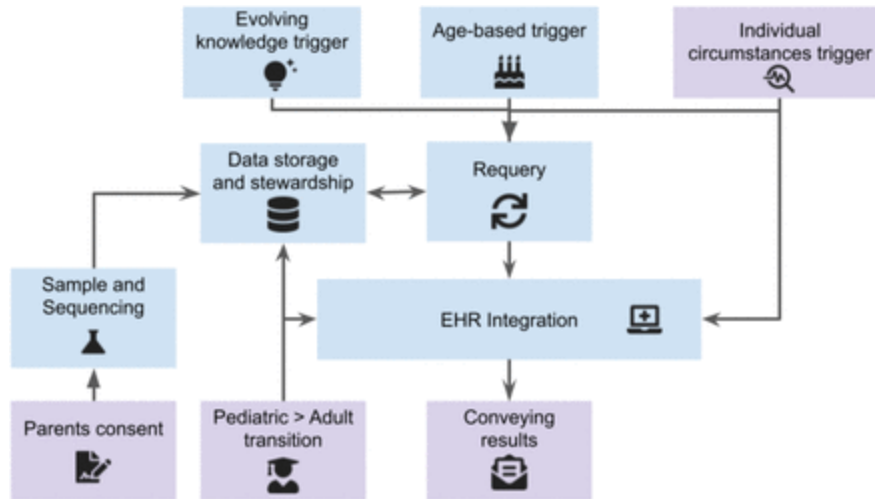


Figure 2: Schematic of the component infrastructure required for lifelong genomic medicine. Boxes represent functional components, arrows indicate dependencies and data flow between components. Purple boxes represent patient facing processes. Operational precedent, unresolved challenges, and equity implications for each component are detailed in [Table 3](#).

Table 2. Delivery model options for lifelong genomic medicine.

Advantages, challenges, and open questions are compared across three institutional pathways identified by participants: newborn screening programs, health system–based clinical offerings, and industry or direct-to-consumer models. Options are not mutually exclusive; a staged approach beginning in clinical settings was favored across multiple groups.

Table 3: Feasibility landscape for lifelong genomic medicine.

For each component of the envisioned model, operational precedent, unresolved challenges, and equity implications are identified from participant interviews. Components are ordered to reflect the flow from initial sequencing through longitudinal management.

No clear delivery pathway

Participants were divided on which institutional setting was best positioned to deliver lifelong genomic medicine, see [Table 2](#). Questions about revenue models and long-term financial sustainability arose repeatedly. Who would pay for the initial sequencing, subsequent reanalyses, and data storage is unresolved, and it isn't clear if insurance would cover those costs. Several participants acknowledged that there is no obvious access pathway for lifelong genomic medicine in the US, given the disseminated nature of healthcare in the US.

Health systems were identified by several as the most natural home for such a program, given their ability to coordinate longitudinal care, integrate data into electronic health records, and maintain patient relationships over time. Some genomic screening implementers noted that healthy whole-genome offerings were already beginning to emerge as clinical products. However, participants also recognized that health systems do not serve all patients equally, raising equity concerns.

Some participants advocated for the integration of lifelong genomic medicine into NBS, primarily on the basis of equity arguments, as NBS reaches almost every child. Other advantages noted included the ability to cross-correlate with biochemical NBS, the uniformity that would be possible within a jurisdiction, and the existence of a pre-existing model.

Arguments for separating lifelong genomic medicine from NBS included: the fundamentally different consent regimes that would be appropriate for lifelong genomic medicine compared to NBS, which currently operates mostly under implied consent; the lack of staffing or funding within state NBS programs; the fact that NBS is not designed for non-infant-relevant information; and the need to protect existing NBS programs from destabilization. Many participants suggested lifelong genomic medicine could start in the clinical context, and migrate to NBS as infrastructure improves, evidence accumulates, and acceptance matures.

Some participants identified roles for industry partners, including supporting PCCs and providing data persistence outside health systems through DTC channels, though clinical geneticists and lab personnel questioned the durability and business viability of commercial partners.

Patient-facing processes

With lifelong genomic medicine participants would be consenting not to a single test but to an open-ended ongoing relationship, concerning data whose clinical significance will change. Participants would need to understand that not everything will be examined, that knowledge evolves, and that they may be recontacted.

Privacy concerns were present across participant groups but were not dominant; most described the proportion of patients who actively objected to genomic data storage as small. However, the indefinite timeframe of the lifelong genomic medicine model compounds these concerns in ways that are distinctive to this model. Participants described commercial data sharing practices as already widespread but poorly transparent, and high-profile breaches such as 23andMe had shaped patient expectations about data security. More prominently, fear of insurance discrimination persisted regardless of existing legal protections and was raised across participant groups as a primary driver of whether patients would consent. Participants also identified mistrust of institutions, particularly among minority communities, as shaping who would engage with a program that asks for open-ended data retention.

Managing the transition from pediatric to adult care was identified as a structural vulnerability of the lifelong genomic medicine model, discussed across participant groups and with particular depth by genetic counselors and clinical geneticists (who also pointed out that this transition is challenging across all of medicine). There is some precedent to rely on, but concerns about scalability were common. The challenge is not only logistical but legal, with parents losing access to a child's medical record when the child turns 18: *"The kid turns 18, we're not allowed to contact the parents anymore now, but we only have the parents' contact information. I guess the logical thing would be to contact the parents at 17 years, 11 months, or something. None of this is very*

well structured, actually not structured at all yet" (P2, clinical genetics). Within genetics, the field is in the early days of working out how to manage this handoff, with current practices varying, for example regarding whether reanalysis continues after the child turns 18 years. Some participants framed the transition not only as a vulnerability but as an opportunity to reengage participants.

Sustaining contact with patients over the lifelong genomic medicine was described as one of the most intractable practical challenges of the lifelong genomic medicine model, with broad uncertainty about how this would work in practice. People move, change contact details, and frequently do not update their healthcare providers. People may forget testing even happened. As one clinical geneticist noted: *"Healthcare [is] so screwed up, we can barely take care of the patients we have in front of us, let alone trying to shepherd these patients all throughout their lifetime, and having the ability to give them information back and forth... and none of that coordination generates any revenue for my health system"* (P48, clinical genetics). One pediatric genetic counselor noted that genetics professionals tell patients they are a lifetime resource, while a PCC observed that genetics does not in practice maintain longitudinal relationships with patients.

Data longevity and reuse

Whether stored genomic data would remain technically viable for reanalysis over the multi-decade timeframe envisioned for lifelong genomic medicine prompted substantive discussion, particularly among laboratory personnel and clinical geneticists. The general view was that whole-genome short-read data was robust and would retain analytical value for at least ten to fifteen years (longer than exome data, which was seen as less future-proof). Laboratory personnel described using data ten or more years old in current reanalysis workflows, and several genomic screening implementers cited examples of disclosed findings derived from sequencing performed over a decade earlier. The transition to long-read sequencing was anticipated as a likely inflection point that would require some reassessment of whether reanalysis or retesting was more appropriate for older samples. But while newer technology might be preferred for individuals with unsolved phenotypes, for healthy individuals enrolled in population screening, existing data was likely to remain sufficient for much longer. Several participants expressed the view that the reanalysis-versus-retest decision should not be treated as a barrier to establishing the lifelong genomic medicine model now, noting that the field could address this question as the technology evolved. One lab participant also raised the possibility of retaining the biological sample itself as a complement to or hedge against data obsolescence.

Lifelong genomic medicine requires not just storing data but maintaining a live, queryable infrastructure over decades to inform clinical care. A clear picture emerged from laboratory personnel that whereas data storage costs were not prohibitive, and indeed many labs keep data indefinitely, the significant costs are maintaining integrations (particularly with EHRs), security audits, regulatory requirements, and the infrastructure required to enable authorized access over time.

Data stewardship was identified as an open question by clinical geneticists, lab personnel, pediatric genetic counselors, and genomic screening implementers, with proposed data stewards ranging from health systems, to labs, to parents. Several noted that everyone wants to control data, partly to enable AI applications. *“[E]verybody’s angling for data storage because they want the data, they want to be able to use it, sell it, research. ... [it’s] a business marketplace more than being very patient centric”* (P48, clinical genetics).

From stored data to clinical action

While variant reclassification and evolving guidelines affect all genetic testing, lifelong genomic medicine transforms these from incidental possibilities into structural commitments. The value proposition of maintaining genomic data over a lifetime rests on the promise that the data will be revisited as knowledge changes, yet responsibility for acting on that evolving knowledge is fragmented. Participants across several groups described encouraging patients to check back periodically — telling patients that contact is “a two-way street,” or advising parents to return in ten to twenty years to see if knowledge had changed — while acknowledging that patients told to follow up with genetics “never do.” This was recognized as inadequate, but with no clear alternative established. One primary care clinician observed that genetics is “pretty unique in the changing knowledgebase and revisiting data,” reinforcing that this is not a routine challenge with established precedent from other areas of medicine. Several noted that liability concerns drove clinical practice in ways not always aligned with patient-centered goals, and several expressed distress at the prospect of holding potentially actionable information without a clear mechanism to act: *“if we have this information, you wanna make sure you can always get it to those people... we never want to be losing sleep because we have this information, and we can’t get it to the person who needs it”* (P28, GC MFM).

Participants described substantial operational precedent for the component activities that would underlie this chain. Laboratories had established variant reclassification pipelines and proactively sent updated reports to ordering providers. Clinics recontacted patients when genes were added to guidelines or panels grew, and some had gone back to research participants when the ACMG secondary findings list was updated. Some laboratories had developed streamlined processes for variant reinterpretation, and some genomic screening implementers described existing “sequence once, query often” workflows in which reanalysis of screening data for new indications was already occurring. However, the current reclassification landscape was described as “in flux,” with practices unstandardized: some laboratories reported only upgrades to pathogenic, others reported all changes, some did not issue downgrades, and reclassified variants did not always result in a new report. Not all updated results were wanted by physicians. At least one major commercial laboratory had ceased providing variant status updates. Current processes were not scalable to population level, with staffing insufficient for recontact when classifications change. Participants recognized that for lifelong genomic medicine to be sustainable at scale, the

processes of reanalysis and recontact would need to be substantially more automated, and that there was no funding model for periodic reanalysis, with current reliance on philanthropy creating unfunded mandates that become unmet obligations.

The aspiration across participant groups was that any treating provider would have access to the genome, with relevant findings surfaced through clinical decision support. The lifelong genomic medicine model places specific demands on EHR integration beyond what one-time testing requires: a new phenotype or updated family history would need to trigger a relook at the genome, evolving guidelines would need to surface updated recommendations, and critically, when patients move between health systems their genomic record would need to follow. Participants across multiple groups noted that EHRs in the US do not talk to each other, and that patients who move are disconnected from their data. Working examples of EHR integration for genomic data existed, and some genomic screening implementers described efforts to route reinterpretations through Epic. But the current state was described as minimal with integrations fragile, expensive to maintain, and dependent on EHR vendor priorities. Clinical decision support for genomics was described as non-obvious because of diverse workflows. Laboratory personnel preferred their own portals and resisted interfacing with EHRs, creating an unclear boundary between the testing laboratory and the health system.

Discussion

Summary of findings

This is the first study to examine professional perspectives on lifelong genomic medicine as a primary research question, drawing on interviews with 52 participants across multiple stakeholder groups. Participants broadly endorsed the clinical value of maintaining genomic data over a lifetime, and many described lifelong genomic medicine as inevitable — yet this perception rested on conviction about the field's trajectory rather than on demonstrated readiness. The central finding of this study is that the main constraints on realizing the vision of lifelong genomic medicine are not primarily scientific or clinical, but the absence of governance structures, accountability frameworks, and institutional infrastructure needed to sustain genomic information as a clinical resource across a lifetime. Much of what lifelong genomic medicine envisions is already happening in fragmented form in different settings, including recontact post variant reclassification, recontact when new genes become actionable, reanalysis when phenotype changes, and clinical decision support with EHR integration. What is missing is the connective tissue that would make it coherent.

Lifelong genomic medicine as a governance problem

Our data suggests that while the technical challenges associated with lifelong genomic medicine are either solved or well on their way to being solved, there are huge governance gaps with unclear resolution: who is responsible for acting on evolving knowledge, who pays for reanalysis,

who maintains contact, who holds the data, who bears liability. Previous work from the Australian genomic NBS context identified the need for data governance and surfaced the need for consideration of managing the genomic data generated [16,17]. Our study identifies in detail the specific accountability, recontact, consent, and data stewardship challenges that emerge when genomic information is maintained as an evolving resource over decades. There is a danger that the perception that lifelong genomic medicine is inevitable may be treated as a downstream concern rather than a prerequisite for implementation, with technological momentum outpacing institutional readiness.

Governance feasibility challenges are compounded by the unsettled science: if disease is far from guaranteed to manifest (i.e., the penetrance of a condition given a genetic variant is low) [24] and the downstream burdens of screening remain unknown, the clinical significance of the information returned to patients will change over time. The “data is a burden” counternarrative from laboratory personnel illustrates how the unsettled science and the governance problem interact: participants were not just articulating that lifelong genomic medicine faces feasibility challenges, but that we don’t yet understand the clinical significance of the underlying data, which compounds the feasibility challenges.

We report a lack of a clear delivery model in the US context. NBS offers the strongest equity case as it reaches nearly every child, but this rests on its current position as part of public health rather than part of a traditional medical health system. Lifelong genomic medicine would need to operate within the medical health system. Moreover, parents already view genomic sequencing as requiring a higher standard of consent than standard NBS [25].

Lifelong genomic medicine adds further complexity, as consent is not only to a test but to an open-ended relationship with data whose significance will change unpredictably over decades, something categorically different from what NBS currently asks of families. The staged model many participants favored, beginning in clinical settings and migrating toward NBS as infrastructure and evidence matures, may be a pragmatic path forward.

Existing frameworks for responsible genomic data governance have been developed primarily for research data sharing contexts [26,27], yet our participants describe a governance vacuum for *clinical* genomic data held over decades. Proposed stewards ranged from health systems to laboratories to patients, with participants noting that competing institutional interests, including data’s commercial and AI value, risk creating a marketplace dynamic rather than patient-centered governance. The concern about commercial data interests is consistent with public attitudes research [28,29].

The emotional and professional burdens our participants described — liability anxiety, the distress of holding potentially actionable information without a clear mechanism to act — echoes findings by Carrieri et al. in the context of targeted genetic testing in the UK [30], but are amplified here by the volume, timescale, and absence of an ongoing clinical relationship that lifelong genomic

medicine entails. The emotional weight of these unresolved obligations is notable: participants were not describing occasional discomfort but instead a structural feature of practice that would intensify as the volume of stored data grew.

The pediatric-to-adult transition introduces a structural vulnerability with no evidence base in genomics specifically. Maintaining contact as someone passes the legal cutoff at 18 is not a solved logistical challenge. Yet it is when an individual becomes an adult that they could consent for much relevant information, including carrier status and adult actionability conditions. Moreover, in keeping with evolving assent models, the child should be involved with decisions about the return of genomic information, for example during adolescence [31].

The equity implications of these findings are not confined to any single domain but are cumulative and mutually reinforcing, reflecting familiar patterns of compounding disadvantage [32]: those least connected to health systems are most likely to face digital consent barriers at enrollment, most likely to be lost to follow-up when they move, and most likely to miss the pediatric-to-adult transition handoff. These issues are on top of known patterns in which those least well represented in genomic databases are least well served by genomic medicine [33,34]. This suggests a clear focus on the creation of equitable systems for lifelong genomic medicine will be needed.

Several research priorities emerge from our findings. This study captures professional perspectives; engagement with diverse patient and advocate stakeholders is now needed, including populations most likely to be excluded or to decline participation in lifelong genomic medicine. A scoping review of the reclassification and recontact literature argues specifically for deliberative methods — in which participants discuss, learn from others, and refine their views — as more productive than further surveys within homogeneous samples [35]. Pilot programs specifically designed around the pediatric-to-adult transition in genomic medicine, which our participants identified as a structural vulnerability with essentially no existing evidence base, are a priority. Comparative evaluation of who is reached and who is missed under different delivery pathways would inform more inclusive program design. Empirical work on recontact and engagement over time is needed to move beyond speculation about how longitudinal genomic relationships would function in practice.

Limitations

This study has several limitations. All interviews were conducted and primarily coded by a single researcher (AL), which may have introduced interpretive consistency at the cost of analytical breadth. This was mitigated through co-coding by a second researcher (IH), iterative discussion of the coding framework, and sense-checking of emerging themes with research colleagues. Our sample was US-based, and many of the governance and infrastructure challenges we identify — particularly EHR fragmentation, the absence of a centralized screening infrastructure, and uncertainty about institutional home — reflect structural features of the US health system. Findings may not generalize to contexts with more centralized delivery, though the core questions

of accountability, consent, and data stewardship are likely to arise in any setting. Participants were recruited in part through BabySeq Project sites and those leading genomic screening programs, resulting in our sample being weighted toward professionals with prior exposure to population-scale genomic sequencing through this pilot program. This may have produced both more informed and more favorable views of lifelong genomic medicine than would be found among the broader professional community. While we recruited across multiple professional groups to enable triangulation, we did not include patient or public perspectives; the extent to which professional assessments of patient concerns — around consent, trust, masking, and privacy — reflect patients' own views remains an open question (we are exploring parents' views in another study). Finally, lifelong genomic medicine does not yet exist as an implemented program: participants were reasoning about a largely hypothetical model, and the challenges they anticipate may differ from those that emerge in practice.

Conclusions

The component pieces of lifelong genomic medicine are no longer hypothetical. Genomic newborn screening is being researched across multiple countries, “sequence once, query often” workflows are already implemented in some health systems, and variant reclassification and ad hoc recontact are routine in clinical genetics. Yet the governance structures needed to sustain these practices as a coherent, lifelong intervention do not exist. Our findings show that responsibility is fragmented, unfunded, or unassigned across nearly every dimension required, from accountability for acting on reclassified variants, to stewardship of data whose significance will change over decades, to maintaining contact with individuals who may not know they were sequenced. Until that scope is defined and the structural prerequisites are in place, lifelong genomic medicine risks proceeding faster than the frameworks needed to make it responsible.

Data Availability

The qualitative nature of the data and the small, specialized professional community from which participants were drawn mean that interview transcripts cannot be shared publicly without risk of participant re-identification, even after de-identification. Participants were assured of confidentiality, and the study was approved under these conditions by the Mass General Brigham IRB. The interview guide, participant fact sheet, and complete coding framework with code frequencies by stakeholder group are provided as Supporting Information. Requests for additional information about the analytical process may be directed to the corresponding author.

Supporting Information Captions

S1 Text. Participant fact sheet. Information sheet provided to prospective participants prior to enrollment, describing the study purpose, procedures, and confidentiality protections.

S2 Text. Interview guide. Semi-structured interview guide used across participant groups. Most components were common across all provider types; sections specific to particular participant groups are indicated.

S3 Table. Coding framework with code frequencies by stakeholder group. Complete coding framework showing all codes, their grouping into subthemes and themes, and frequencies by participant group (clinical geneticists, genetic counselors, laboratory personnel, primary care clinicians, healthcare systems representatives).

S4 Checklist. COREQ checklist. Consolidated Criteria for Reporting Qualitative Research (COREQ) 32-item checklist for interviews and focus groups.

Study Title: An interview study to help assess the clinical value and feasibility of revealing genetic information at multiple points of the lifespan

Principal Investigator: Robert Green, MD, MPH Fact Sheet

- This is a Mass General Brigham research study. The purpose of this research is to help assess the value and feasibility of revealing genetic results over the lifespan. This research study is funded by the NIH.
- We are asking you to participate because you are a professional with relevant expertise to help the research team address their research questions. About 64 people will participate in total.
- If you are being emailed directly by the study team, your email was obtained from publicly available information, given your professional role in either a genomic testing lab or a program integrating genomic screening into health systems.
- In this study we will use the platform Zoom to conduct a one-on-one virtual interview. This virtual interview will take 60 minutes and will be recorded. We will ask you questions that draw on your professional expertise; there is no preparation needed on your part.
- You will receive a \$100 check for your time. We will need to collect your social security number and address to issue the check. This information will be kept secure and will not be used as study data.
- This interview will be transcribed and analyzed by the study team. We may use direct quotes from participants when presenting our findings.
- The video recordings will only be available to the study team. Your de-identified information may be used or shared with other researchers without your additional informed consent.

- The main risk is a potential loss of privacy and/or confidentiality. We have multiple procedures in place to protect privacy and confidentiality. While we cannot guarantee that the identity of the participants will not be revealed in the event of a breach or other unexpected event, we have precautions in place to keep your information secure. Study data will only be accessible to study staff; your identifiers will be separated from your responses and all data will be coded. We will use the secure systems at Mass General Brigham, endorsed by our Information security team, to secure both your study data and the information provided for issuing the check.
- Participation is voluntary and you can stop at any time.

Anna C F Lewis, DPhil, is the main point of contact for this research study. You may contact her through email at aclewis@bwh.harvard.edu. The PI's contact information is rcgreen@bwh.harvard.edu. If you'd like to speak to someone not involved in this research about your rights as a research subject, or any concerns or complaints you may have about the research, contact the MGB Human Research Committee at (857) 282-1900.

Interview guide for "An interview study to help assess the clinical value and feasibility of revealing genetic information at multiple points of the lifespan"

This Interview guide should be tailored to the specific respondent I am interviewing.

For everyone

Preamble

Hello, []. Thank you for agreeing to take part in this interview study. My name is

Anna Lewis and I'm a research scientist at Brigham and Women's Hospital division of genetics. My mentor is Dr Robert Green and he is the PI of this study.

As a reminder, we are conducting a research study. The purpose of this interview is to help assess the value and feasibility of revealing genetic results over the lifespan. There are no right or wrong answers to any questions.

I sent you a fact sheet about this study. In brief, this interview will be recorded and then transcribed and analyzed. All identifying information will be removed and all of the analyzed data will remain anonymous. This interview will last about 60 minutes. For participating in this interview, we will send you a \$75 gift card as a thank you for your time. Your participation in this interview is completely voluntary and if at any point you wish to stop the interview, you are welcome to do so. Additionally, if there is a question you would rather not answer, just let me know.

As a thank you for your time, we will send you a \$100 check. Per hospital policy, I need to ask you for your SSN in order to issue the check. I'll do so at the end of the interview; it will be used solely for this purpose, kept secure, and will not be connected to your responses.

Before we get started, do you have any questions about what I have presented, or any questions about the Fact sheet?

Do you consent to be part of this study? Is it okay if I begin recording now?

Introduction to the Project: Frame some of the key pieces of background, as well as the motivations and concerns. Explain why they were selected to be interviewed.

1. Background Questions: This basic set of open ended questions are designed to get the respondent comfortable sharing and telling us about their work.

- What is the focus of your professional practice?
- What led you to this area?

For health care providers (Aim 1a)

2a. Reactions to receiving screening information.

First ask for examples of where they receive both genetic and (where appropriate) non-genetic examples. For these examples, ask:

- What makes it useful?
- What makes it a burden?
- How does uncertainty factor in?
- Examples of receiving information before they would initiate a risk mitigation strategy?

Idea of “patients in waiting”

3a. How they view their obligations over time to patients and their parents

- Example of a variant of uncertain significance for a child with a phenotype
- Example of a child with a pathogenic variant but not yet showing signs of having the condition (low penetrance?)

4a. Their experiences of longitudinal relationships with patients and their parents

- Any experiences of patient-parental divergence of opinion with regards to genetic knowledge?
- Any observations of the resources families need access to in order for the value of genetic information to be realized?

5a. Attitudes toward when information should be shared.

- Are there example genetic conditions where you think it would be optimal to do population screening later in childhood?
- In other words, do you see any clinical value in withholding any later childhood onset information?

6a. Their expectations of what patients and parents would want

- What would you anticipate parents would want when it comes to just-in-time information, versus receiving it all at once?
- What about patients themselves, as they reach the age of assent?

For those with operational roles in generating genomic reports (Aim 1b): not all questions will be relevant to all roles

2b. [Lab personnel] Treatment of sequence information after an initial report is generated

- Current treatment -- does anything happen? For example, reinterpretation of variants?
- Do you envision this changing in the nearterm, and why?

3b. [Those embedded in health system] Vision for integration of genomic information into health system

- What is the near term vision for the integration of genomics into the health system?
- [If not in the near term vision] Do you share a medium term vision of using the genome as a resource over the lifespan? Why or why not?

4b. How they view their obligations to the individuals who they provide testing for.

- Does the lab have any legal or regulatory responsibility to the individuals you provide reports for, after the report is signed out?

What about reporting variants that have taken on a new significance?

- Do you think the lab has any *ethical* responsibility in this area?
- Do you think anyone else, perhaps the ordering physician, has any ethical responsibility in this area?

5b. [Lab personnel] Whether, and how, their lab currently distinguishes in theory and in practice genetic information from the querying of that information.

When a lab generates a variant file, in some sense there is information that is both “there and not there” -- a simple look up could reveal a pathogenic variant, but it won't necessarily be looked up.

Does your lab distinguish in theory or in practice genetic information from the querying of that information?

6b. Privacy, security and longevity of information.

- What do you consider necessary, currently to maintain the privacy, security and longevity of sequence information?
- Do you see any of this changing in the near or mid term?

For everyone

7. Their views on using the genome as a resource over the lifespan

Do you think that this vision, of sequencing once and querying many times, is one that the field should be pursuing?

- Why or why not? Any considerations we haven't covered? (anticipate a mixture of practical and ethical concerns)
- Any additional practical considerations that we haven't touched upon?
- If ethical concerns haven't come up explicitly: Do you have any ethical concerns?
- If equity hasn't come up: Do you foresee any equity barriers?

8. Wrap up.

- Is there anyone else you recommend I should talk to about these kinds of questions? Particularly someone who might have a contrasting view to your own?

If so, can I send you a recruitment email, for you to forward to this/these individuals, copying me?

- Is there anything else you would like to add?
- Do you have any questions for me?

I am going to stop recording now. Please can I ask you for your SSN? I will store this securely and we will only use it to issue your check. Can I also please confirm what address you would like the check sent to?

Thank you!

S4 Checklist. COREQ (COnsolidated criteria for REporting Qualitative research) Checklist

Developed from: Tong A, Sainsbury P, Craig J. Consolidated criteria for reporting qualitative research (COREQ): a 32-item checklist for interviews and focus groups. *International Journal for Quality in Health Care*. 2007. Volume 19, Number 6: pp. 349–357.

Manuscript: *Sequencing for a Lifetime: Value, Feasibility, and the Governance Gap in Lifelong Genomic Medicine*

Acknowledgements

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References

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