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Embedding ethics into Genomics England's Generation Study

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ABSTRACT

The Generation Study is a large-scale research initiative led by Genomics England in partnership with the National Health Service, aiming to evaluate the use of whole genome sequencing in newborn screening, as well as ongoing research use of these genomic data. The Generation Study will sequence 100 000 newborn genomes in England to potentially identify approximately 200 rare and treatable conditions. This paper outlines the study's approach to embedding ethics from inception through implementation. A model of 'ethical embeddedness' that emphasises transparency, trustworthiness and responsiveness to uncertainty is utilised. Drawing on the deliberations of a multidisciplinary Ethics Working Group, public dialogue findings and design research, the paper presents key decisions and our approach to complex ethical challenges including consent, potential impact of the study on clinical services and navigating uncertainty. The paper also reflects on the ethical tensions inherent in balancing research ambitions with operational realities, particularly in a context of evolving genomic science and sometimes limited regulatory clarity. By embedding ethics into the study's design and delivery, we hope to foster public trust and inform future policy and practice.

INTRODUCTION

Genome sequencing is increasingly used in diagnosis and is being explored in screening and population health globally.[1] Patient organisations in England have called for expanded screening of newborn babies using technologies like genomic newborn sequencing (gNBS). This may identify a larger number of treatable genetic conditions earlier and support affected families sooner.[2 3] Currently, gNBS is the topic of extensive international research.[1]

Genomics England is examining the benefits, risks and broader implications of gNBS in newborns using whole genome sequencing (WGS) through the Generation Study (IRAS #324562), a research project in partnership with the National Health Service in England (Hereafter referred to as the NHS). It aims to sequence the genomes of 100 000 newborns, to identify around 200 rare and treatable conditions as soon as possible after birth. The current lack of evidence about WGS in gNBS requires the Generation Study to run as a research project rather than an embedded pilot[1] to generate evidence on the use of WGS in a gNBS context. The challenges associated with this task are numerous, and some are highlighted in a recent publication from Genetic Alliance on the current status of newborn screening in the UK.[4]

The Generation Study aims to do the following:

1. Evaluate the utility and feasibility of using WGS to screen newborns for a large number of rare conditions that can be treated in early childhood.
2. Enable wider research in genomics and health.
3. Explore the potential of storing and reanalysing an individual's genome over their lifetime to support their healthcare. This is an emerging field of inquiry internationally.[5 6]

The Generation Study began recruitment in April 2024 and several factors make it uniquely complex:

- The study has a very large sample, with wide eligibility criteria in maternity settings, to measure utility as well as explore feasibility and impact of offering gNBS at scale.
- Confirmatory testing and treatment for babies with condition suspected (CS) results is provided by the NHS.
- The study plans to store genomic data long term. These data will be accessible to vetted researchers, aligned to Genomics England's National Genomic Research Library (NGRL) Governance Framework.[7]

Population-wide studies of this magnitude and complexity create ethical challenges in their conception, design and implementation. This paper details our efforts to embed ethics into the Generation Study and ongoing work as the study matures.[8] We have chosen several ethical issues to discuss in detail as these may be most relevant to others working in the

field. But this paper is not a comprehensive accounting of all ethical issues across the Generation Study.

Our approach to embedding ethics

The Generation Study utilises an 'ethical embeddedness' approach developed over time: anticipating, considering and responding to ethical issues from study conception, and ongoing today. The approach draws on compelling ethical theories to help frame and elucidate key issues, rather than adopting a singular ethical framework hoping to cover all eventualities. 'Ethical Preparedness'[9] has been important in embedding ethical thinking as a continuum—this work taking place long before REC approval and ongoing. 'Genomic Uncertainty'[10] is instrumental in framing our thinking, given its main premise that genomics is a plethora of inherent unknowns. Carefully considering how the study is grounded within such uncertainty allows us to openly acknowledge the unknown. Finally, our approach to embedding ethics is bolstered by continual iteration with the public, health professionals and more recently, Generation Study participant parents themselves.

Why ethical embeddedness?

In a research project as complex as the Generation Study, operational ethical questions do not often lend themselves to resolution by applying guiding principles, as the weighting of ethical principles is context-specific and subjective. Consequently, consensus can be elusive, which can lead to paralysis in decision-making and a lack of clarity over the normative force of principles in the applied context. Or, principles are applied differentially in practice and may eventually become relativistic,[11] or worse, tokenistic, reflecting an 'ethics-washing' approach. Meaningfully responding to ethical issues involves accommodating nuance and complexity as they emerge. This can be enabled by embedding ethics-informed thinking. For the Generation Study, this entailed encompassing a range of views and values to pragmatically inform active decision-making, starting with a public dialogue[12] to explore public views before the study was funded. Thereafter, these views and values were foregrounded as the project developed at pace. Recent scholarship in research subjects advancing at pace and scale, such as in Artificial Intelligence and Machine Learning, has also taken the approach of ethical embeddedness—or 'Ethics by Design'—as a crucial starting point for complex and multisystem interventions.[13] This is compelling in landscapes that are evolving, uncertain and often ahead of clear regulation. The Generation Study reflects and manifests some of this complexity.[14]

Taking an 'ethical embeddedness' approach challenges us to operationalise ethics in the service of the overall programme objectives.[13] Our approach strives to foreground:

- Transparency—galvanising insights from public, patient, participant and expert engagement, openly working to identify, weigh up and address complex ethical issues and clearly demonstrating our thinking.
- Trustworthiness—demonstrating competence, reliability and accountability, especially in managing the limitations of our approach, the extent of our current knowledge, the aims of the research, its risks and the unanswered questions at the heart of the Generation Study.

The generation study ethical embeddedness model

The Ethics Working Group (EWG), a working group of external experts, was established alongside several other working groups (communicating results to families, evaluation and recruitment). To facilitate coherence across the working groups, cross-cutting workstreams were formed (including ethics, engagement and design research). Insights from three public dialogues, each addressing a research aim of the Generation Study, were incorporated, complemented by substantive engagement work on informed consent in under-represented populations. For example, the principle that all conditions screened on the Generation Study must have a treatment available to all children through the NHS was a critical finding from our first public dialogue and is a cornerstone of our 'choosing conditions' framework. In our engagement work with ethnic minority communities, the imperative not to 'target' under-represented groups, but rather to enter into meaningful and collaborative partnerships with these communities, was emphasised. This finding has influenced our funding and reimbursement models, as well as our ongoing engagement work towards facilitating equity of access to the Generation Study for birthing parents in England.

In an ongoing and iterative process, the practical implementation of ethics (among other subject areas) was addressed in design research,[8 15] where over 150 interactions with a range of participants and 10 hospital sites have informed study delivery to date. Among the participants in our design research are maternity and research staff, clinical specialists and clinical scientists. Design research helps us to triangulate a range of unique perspectives to drive the design of service pathways and products such as information collateral for participants and research staff, promoting trustworthiness and transparency in a tangible way.

The Ethics Working Group

The EWG comprised multidisciplinary experts from genomics, newborn screening, midwifery, law, patient advocacy and social science disciplines, all with ethics expertise. The group's aim was to identify and critically consider ethical issues inherent to the Generation Study, inputting in terms of transparency and trustworthiness. As a forum, the EWG facilitated a sense-check and challenge to our proposals. Open discussion of concerns and issues was essential, as was acknowledging that the study needed to be designed and implemented within certain funding and time constraints and as such, pragmatism was key.

Over the course of 18 months, the EWG met 11 times. Discussions included:

- Principles for selecting conditions, principles of study evaluation.
- Research strategy and linked data sets.
- Lifetime genomes strategy and consent model.
- Baby and mother sample study (BaMSS). A precursor study to decide on the most appropriate sample type for the Generation Study
- Consent model and debating key tensions.
- Consent process, information sharing and key messages.
- Newborn research workshop and recontact.
- NHS workforce and study staffing.
- Duty of care, withdrawal and recontact.
- Evaluation and ethical preparedness.[9]

Some ethical issues identified in EWG meetings were resolved. Solutions, often suggested by the EWG themselves, were implemented into the project and user tested where possible. For instance, in refining principles by which conditions for the Generation Study are selected, one criterion was that all conditions had a minimally invasive confirmatory test available on the NHS. The EWG advised that some confirmatory tests are not, and cannot be, minimally invasive. This framing was changed. The EWG also proposed a compelling strategy for withdrawal that combined a focus on the best interests of participant children with mediating autonomous parental decisions when a CS result is found in a baby. Now, if a parent withdraws their baby while the genome is under analysis in Genomics England's bio-pipeline, the result would still be returned to the parent through our established pathways for results return. This was based on EWG advice that once a CS result was known, this should be returned as it protects a child's right to be found and fosters Genomics England's duty of care to our newborn participants. This is made clear in our information sharing materials.

The EWG also guided our position on remote consenting. Remote consent does not involve seeking a 'wet signature', but rather is the culmination of an information sharing process. Ultimately, a verbal expression of willingness to participate is sought during a phone call or online meeting, and then recorded by the research staff member on an electronic consent form. A copy of this is then sent to the participant for their records. Practising midwives advised that remote consenting would enable greater accessibility and reduce research staff workload. However, others felt more comfortable with requiring a signature on a consent form, as a clear indication that a healthcare professional had been involved in sharing complex information—rather than parents signing up entirely independently. Our design research pointed to the need for remote consent for the majority of parents. Guided by the EWG and these positions, a decision was made to facilitate remote consent. This is obtained by trained research staff, who benefit from intensive training and regular consent webinars.

The EWG also helped us work through concerns that the Generation Study may decrease the uptake of standard NBS (standard of care newborn screening programme in England; screens infants for 10 rare conditions about 5 days postpartum) if parents wrongly believed that it was a clinically validated substitute, or otherwise conflated the two. This would result in some babies not receiving the NBS, which is a commissioned, clinical intervention. While this may not be wholly avoidable, we considered it a significant risk, requiring mitigation and ongoing monitoring. In mitigation, careful liaison with the NBS Programme and the UK National Screening Committee has taken place, and we tested materials to check that parents understood the Generation Study to be research at first glance. Early feedback from sites suggested that while participant parents did not conflate the interventions, research staff may have done so. Consequently, we revised a tranche of wording on our participant materials relating to the NBS.[16]

Other ethical issues were much more complex, requiring careful deliberation over a longer time. Here, we discuss a selection of issues to illustrate our efforts towards ethical embeddedness from the outset:

- Facilitating appropriate consent
- Managing uncertainty in genomic screening results.
- Resource impact on clinical services.

Facilitating appropriate consent

Consent in genomic research is widely debated. Questions arise about how information on future data use can meaningfully be shared when advances in genomics may be unknown and open-ended. The Joint Committee on Genomics and Medicine posits that present and future implications of genomics render 'specific' and 'fully informed' consent unachievable. Broad consent models may be more appropriate,[17] including in the gNBS setting,[18] if supported by robust governance and safeguards. It is also acknowledged that our approach to consent needs to align with the nature of the research at hand and wider societal needs.[19]

To realise the Generation Study's objectives, parents are invited to consent to screening (including receipt of a research result), long-term data storage in the NGRL and ongoing research access to these data. Consent is thus 'all-in' rather than a tiered model that separates the screening research from longer term research. This decision was significant. It was informed by extensive discussions with the EWG who contributed important challenge and validation, which directed our efforts to embed ethics. It was also informed by design research.

Critical to these discussions was the need to clearly delineate the Generation Study as a research project, and not a clinical service. By inextricably linking screening (objective 1) and future research (objectives 2 and 3), the consent model comprises a complete research package, leaving less space to conflate it with clinical provision. Equally important were considerations of equity. It was discussed that an all-in model may be off-putting

to communities historically under-represented in research—who may be mistrusting of ongoing data use.[20] This tension was balanced against concerns that the alternative tiered consent model would inherently be more complex, making it more difficult for individuals with lower literacy levels to engage with. This could potentially disempower parents from feeling able to make informed choices about participation and risk creating inequities in recruitment. These views were carefully considered and mitigation work—key to which is easy and accessible withdrawal as well as translations of information sharing materials into 17 most frequently spoken languages at study sites—is ongoing to empower appropriate decision-making within a diversity of communities.

Another consideration in favour of the all-in consent model was anecdotal evidence from the 100K Genomes Project that when offered tiered options, people were often more concerned with understanding the tiers than they were in understanding the core offering. This was confirmed in design research for the Generation Study, which found that all-in consent better supported understanding of the screening as a research intervention, rather than a clinical intervention alongside an invitation to share data for research. It is essential that people understand the core offering of the Generation Study, especially because it involves sending research results to parents. In the case of CS results, these can be life-altering. An independent evaluation partner is undertaking research into people's experience of consent; a key aspect of this evaluation will be exploration of some people's reasons for declining and their perceptions of the 'all-in' model.

Ongoing data storage and reanalysis, facilitated through the all-in consent model, allows us to evaluate aspects like false negative results and participant outcomes over time. This is essential for answering our research questions. It also promotes objectives two and three of the Generation Study:

- Enabling ongoing research into genomics and health.
- Exploring the benefits, risks and uses of a person's genome over their lifetime.

The storage and ongoing use of newborn babies' genomes to this scale over the life course (unless withdrawn) is unique to the Generation Study. However, it is starting to gain traction in other countries initiating gNBS research studies.[21] Ongoing research and lifetime genome work is open-ended, which challenges us to carefully consider and describe associated genomic uncertainty. Concerns about storage and reanalysis of data as a compulsory component of the consent model hinge on these uncertainties. Challenges to data security and access are also well documented.[22 23]

Transparently imparting information of this complexity, and conveying the uncertainties, is challenging. Yet it is essential to facilitate parental consent, informed to an appropriate level.[24] Ethical embeddedness compels us to anticipate and mitigate this complexity at the outset, in our case through extensive iteration. Consent too is seen as ongoing, and information supporting it will be gradually tailored to participant children as they get older. Genomics England has a good track record of managing participants' data in a trustworthy manner and we have been audited by the NHS on a number of occasions.

Managing uncertainty in genomic screening results

A criticism often levelled at gNBS is the anxiety arising from false positive or inconclusive results.[25] The relative harms of these, in more numerous individual cases, need to be weighed against the relatively smaller number of potential larger-magnitude benefits to society.[26] It has been argued that aggregation of many small harms may or may not outweigh aggregate benefits, and thus a higher level of individual risk to a larger number of people may or may not be acceptable. The types of harms in question may include causing parental anxiety, disrupting parental bonding and overmedicalisation of 'well' babies.[26] These will be considered in the evaluation of the Generation Study.[27] Of course, parents receiving a CS result will naturally feel worried in these circumstances, and while awaiting confirmatory testing. Parents receiving such results from the NBS share this experience.[28] Through close collaboration with the UK National Screening Committee, a pathway to ensure that parents do not receive discordant results in the event of a CS in a child has been developed. Research staff also use our 'decision cards' to assist parents making decisions within this uncertainty framework, and to promote engagement with these risks. We are fortunate to have external ethics expertise to support the results return process.

To manage uncertainty, we have emphasised the potential for incorrect results in our study information materials (including as a piece of 'narrative storytelling'—which our user research demonstrated better brought these concepts to life). We invite parents to carefully consider their appetite for this before making a participation decision. It has been demonstrated that people's preferences regarding screening results may change through deeper interrogation,[29] and we enable timely withdrawal options and ongoing psychological support should it be required.

The uncertainty ramifications of inconclusive results also require monitoring. Recent research with health professionals managing Cystic Fibrosis Screen Positive Inconclusive Diagnosis (CFSPID) children[30] and parents[31] suggests that screening programmes should consider prioritising sensitivity to mitigate the risk of missing affected children, while ensuring adequate communication and management of inconclusive results. This does not necessarily align with public perceptions[29] and demonstrates divergence between lived experience and public attitudes. We have sought to mitigate the potential detriments of inconclusive results with thorough, transparent information sharing about these uncertainties as a core component of the consenting process.[31] However, we cannot eliminate this.

As the Generation Study progresses, monitoring our false positive, false negative and inconclusive results is critical and ongoing.[32]

Resource impacts on clinical services

A major area of ethics debate in the EWG involved the Generation Study creating additional burden for already overstretched specialist clinical services. It would be ethically unacceptable for research activities in these settings to detract from clinical care. Many patients and families are awaiting clinical genomic results or interventions across England. They are already presenting with symptoms and in need of clinical management. Creating delays in their care by processing research findings from newborn babies, the vast majority of whom would receive a no condition suspected (NCS) result, would be inappropriate. To this end, much of the Generation Study system is automated and NCS cases will never be referred into the health system. A small percentage of results need manual intervention, minimising the scale of impact on diagnostic services.

For CS cases, we have embedded ethics by carefully designing the results return pathway to minimise impact on clinical services. Modelling suggests that the number of babies needing confirmatory testing will be low, between 500 and 1000. A proportion of these babies would regardless be referred into clinical care based on NBS results, or because they were later diagnosed with the condition through standard pathways (a false negative GS result). However, mitigating administrative burden for clinicians in these circumstances is still a challenge. For instance, clinicians reiterated the value of the 100K Genomes Project, but emphasised the additional administrative work associated with returning results.[33] In early result return from the Generation Study, we are anecdotally observing a similar trend. To minimise potential harms to families and burden on clinical teams, we codeveloped a range of tools and processes to support the return of results, including the following:

- A confirmatory test pathway for each condition, with information about the confirmatory genetic and non-genetic tests that should be initiated following receipt of a CS result from the study, as well as an overview of next steps for management and onward support.
- A call guideline to be used as an aide-memoire for the first result conversation with parents; prewritten template emails and parent-facing information sheets for each condition written specifically for screening results.
- Regional Results Coordinator support for coordinating next steps, and preresults MDTs if necessary.

While the potential impacts on clinical services are a substantial ethical issue, they are being monitored and mitigated. More broadly, these impacts are weighed against potential benefits of the study, such as possible long-term cost savings through reducing diagnostic odysseys and potential for enhanced quality of life that may come with early intervention.

CONCLUSION

A year after the Generation Study began recruitment, we reflect on our ethical embeddedness approach. Has it been useful in promoting trustworthiness and transparency? This appraisal is subjective. Through consistent monitoring, it appears that ethical issues where we most focused our efforts are progressing as we expected, and we are mitigating as planned. For instance, the support we have put in place for the consent process—while not perfect—appears to be well received, with many queries and questions coming through to us. However, some of our efforts still need honing, such as reducing the administrative burden of managing CS results. Areas of focus for ethical embeddedness can be mapped to our 'edge cases'—those eventualities that were deemed less likely to occur and require case-by-case appraisal. These have been challenging as the study rolls out. They include our approach to providing results in the case where a participant baby has died, our management of Subject Access Requests and some of our study exclusion criteria that may be open to interpretation and situational differences. Embedding ethics across some of the 'big ticket' issues has freed us up to focus on these 'edge cases' and has also given us the opportunity to iterate. The transparency of our approach has been acknowledged. In terms of trustworthiness, our success in this regard is evidenced by the parents who choose to trust us to manage their newborns' information, something we do not take for granted. It is heartening that this sentiment has been consolidated in recent user research with Generation Study participant parents.

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