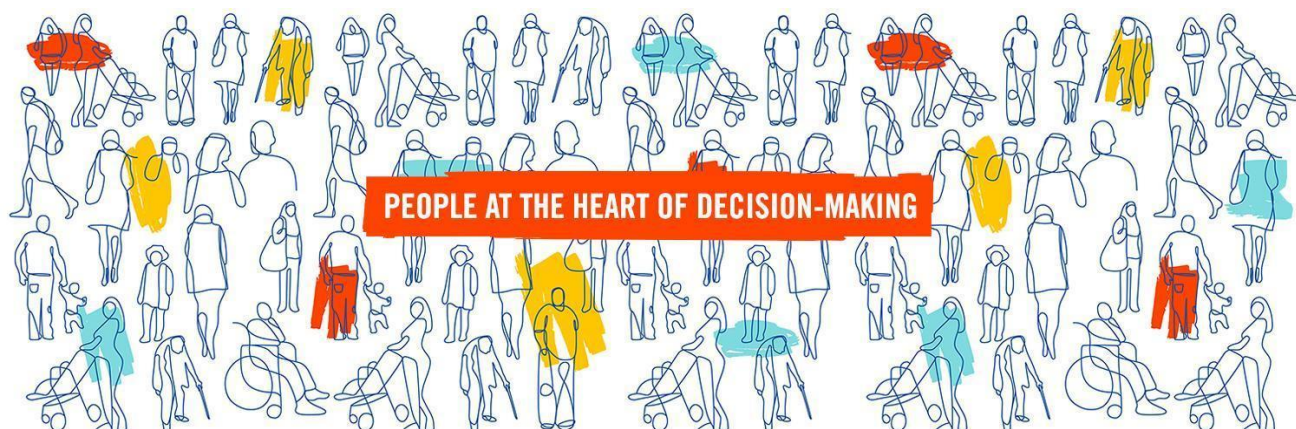




Newborn Genomes Programme: Developing draft principles for feeding back results to families

Working group consensus building report: phase 1

March 2022



About Involve

We're the UK's leading public participation charity, on a mission to put people at the heart of decision-making.

We believe that decision-making in the UK needs to be more:

- **Open** - so that people can understand, influence and hold decision-makers to account for the actions and inactions of their governments;
- **Participatory** - so that people have the freedom, support and opportunity to shape their communities and influence the decisions that affect their lives; and,
- **Deliberative** - so that people can exchange and acknowledge different perspectives, understand conflict and find common ground, and build a shared vision for society.

Our work seeks to create:

1. **New innovations** - to demonstrate better ways of doing democracy;
2. **New institutions** - to put people at the heart of decision-making;
3. **New norms** - to make democracy more open, participatory and deliberative.

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Our values

- **Collaboration** – because change comes when broad coalitions of people work towards a common vision.
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- **Purpose** – because participation must have an impact. We reject tokenistic or ineffectual engagement.

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1. Background and overview

Genomics England's Newborn Genomes Programme is embarking on an NHS-embedded research study to explore the benefits, challenges, and practicalities of sequencing and analysing the genomes of newborns. The study aims to generate evidence regarding whether whole genome sequencing (WGS) could complement (rather than replace) the existing NHS newborn screening service. While principles and criteria for screening already exist,¹ it was felt that a unique approach needed to be taken for this Programme, as it is a research study and looking at the overall effectiveness of a specific technology (WGS).

To do this, Genomics England has convened a working group with expertise from 22 individuals with clinical, scientific, ethical, policy, patient and public backgrounds to inform the **conditions (and the genes and variants which cause them) that will be included in the initial testing panel for newborn genome sequencing**. The Working group's objectives are:

- Inform the development of a set of principles to assess which conditions should be included in the newborn testing panel
- Advise on a consultation process to ensure the development of the principles and the testing panel can be appropriately assessed by relevant groups
- Consider how to disseminate the principles and outputs from this group (e.g., with healthcare professionals, patient and policy groups)

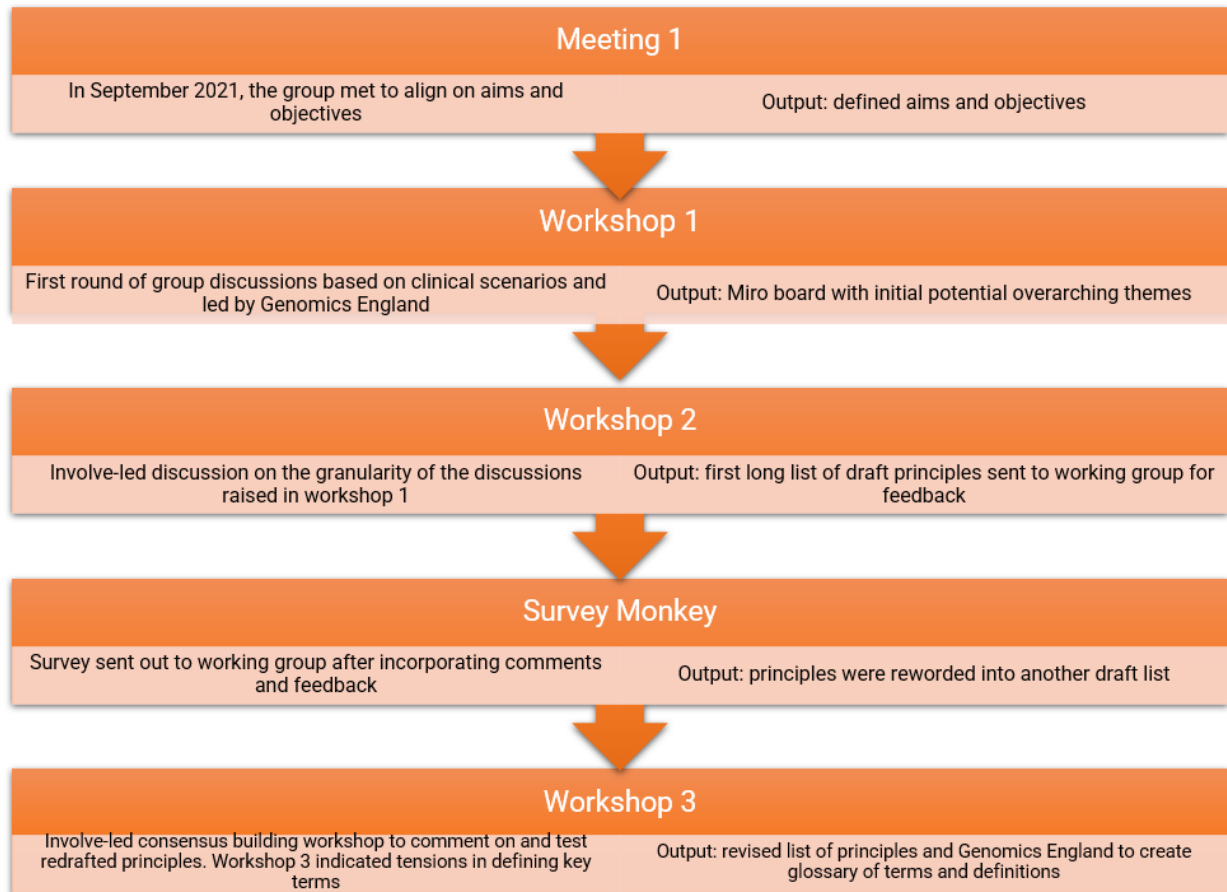
These principles will apply to the conditions, genes, and variants that would be looked for in the Newborn Genomes Programme's research study, with the opportunity to identify a larger number of conditions than is currently screened for by the NHS newborn blood spot test.

A [public dialogue](#) conducted in July 2021 for Genomics England stated that the larger number of conditions that should be screened for should include "those that impact the infant in early childhood *and* [where] there are treatments and interventions to cure, prevent, or slow progression of disease". The working group was convened to provide more granularity and thought on what this criterion means for different choices of conditions, genes, and variants.

The working group consisted of around 20 individuals from different backgrounds (see Appendix for full membership), as well as individuals from NHS England and NHS Improvement Genomics Unit and Genomics England, met four times between September 2021 and February 2022 to develop and deliberate on a draft set of principles (see chart below). Involve supported the consensus building and decision-

¹ See, for example, <https://www.gov.uk/government/publications/evidence-review-criteria-national-screening-programmes>; and <https://apps.who.int/iris/handle/10665/37650>.

making process of the working group. During the sessions, case studies and relevant issues were discussed to surface important considerations. In between meetings, the group had the opportunity to share their comments and thoughts on the draft principles via additional conversations and email. The draft principles were continuously refined based on this feedback.



Following this process, the group agreed on five draft principles (discussed in further detail below).

Include the genetic variant in the screening programme if:

- A: There is strong evidence that the genetic variant(s) causes the condition
- B: A high proportion of individuals who have the genetic variant(s) would be expected to have symptoms that would significantly impact their quality of life if left untreated
- C: Pre-symptomatic or early treatment(s) for the condition has been shown to lead to improved health outcomes for the child, compared to treatment after onset of symptoms.
- D: There is a minimally invasive confirmatory test that can establish whether or not the child has the condition.
- E: Conditions screened for are only those for which the socially acceptable interventions are equitably accessible for all as standard of care within the NHS.

The next steps following the establishment of these draft principles will be:

- An open feedback process, including engagement with different professional, patient, and public groups
- An assessment of the outcomes of this engagement process, and analysis of how the draft principles might need to be revised
- The working group will deliberate on revisions and agree on a finalised set of principles
- Application of the principles to define a list of conditions, genes, and variants that will be looked for in the Programme.

The outcomes of this process will be shared openly by Genomics England.

This report summarises:

- The areas discussed by the working group, the key themes emerging, and how discussions evolved (**Chapter 2**)
- The Group's current recommendations for wording the principles (**Chapter 3**)
- Recommendations for what should be covered in the next stage of wider engagement with patients, public, and other stakeholders (**Chapter 4**)

2. Thematic analysis

Overview

Over the course of working group meetings, decisions to be made on the conditions to be included in the Programme were iterated and discussed. The group talked about how genetic conditions can present variably between individuals and the unique experience each person with a condition might have; and the challenge of identifying variants before an individual develops symptoms of a condition.

We note comments made by participants in the sessions throughout this section. We also indicate *when* in the process the comments were made (i.e., Workshop 2, post-Workshop 2 in written feedback, Workshop 3) to illustrate the group's journey. Comments from Workshop 1 are not collated as it took place at the point where the working group was convened.

Group members had different levels of technical knowledge about genomics and screening processes, scientific and clinical expertise, and experiences of working with the public and patients. This meant that working group members brought distinct views on the various principles. Despite this, by the end of the third session, broad consensus was achieved on the thematic areas which would be important.

The group affirmed that the principles should guide the Programme's decision-making, and should **reflect any key trade-offs between looking for a broader number of conditions versus being more conservative in their approach**. While the working group overall had good understanding of the technical issues that lay behind some of the trade-offs, it highlighted the need for Genomics England to add definitions and technical information to the final principles to enable their accessibility for different audiences (including clinical and scientific experts, and the public).

The following themes emerged as the working group discussed the core issues and trade-offs.

Penetrance (how likely a person is to have the condition if they have the genetic variation associated with that condition) and expressivity (the range of signs and symptoms associated with a condition, and their severity)

The working group questioned how best to balance the need for research on variants where less is known about penetrance, with the need to gain the best value from the process by looking for variants known to be associated with high penetrance.

As part of this, several questions emerged in Workshop 2:

- Should the choice be made to include only those variants with the highest known penetrance?

- Considering that in many conditions penetrance increases with age, should sequencing partially depend on penetrance by the time a person reaches a certain age?
- Should variants that are only known to be associated with a non-penetrant form of the condition be excluded? If so, does this limit the ability to learn more about the clinical validity of a variant (how well the variant relates to presence, absence, or risk of disease)?
- If there is uncertainty with respect to the variant's penetrance, what should be done?

“What we would need to know is if we find a gene what are the chances that it will give rise to symptoms and to what degree this would impair well-being” (feedback after Workshop 2)

In Workshop 3, the discussion around penetrance focussed on how being screened for a variant with low penetrance might affect families, as there would be uncertainty about what the implications of the test might be. This led to discussion about the test's sensitivity and specificity, and recommendations for only screening for conditions where confirmatory tests would be available (see more in next section). In the Workshop 3, group members tended to agree that, based on current knowledge, looking only for pathogenic variants might be too narrow for the Programme. As a research study, the Programme could help generate more information about a condition, including understanding variant classification and penetrance.

“We do not have the evidence to apply this in rare conditions - we may just not be able to determine this beyond frequent variants/mutations” (Workshop 3)

The group recommended that Genomics England consider the extent of its research programme's aims, including whether it has a remit to look for variants currently known to have lower penetrance.

Testing: at what level to set sensitivity and specificity of the screening

In the earlier meetings, the group considered how sensitive or specific testing should be, but it took until the Workshop 3 for the group to articulate the core trade-off at the heart of the question around sensitivity and specificity: if the sensitivity of WGS is set too high, it will not pick up enough cases for the Programme to be economically viable (when balancing against the money saved in treatment and the benefits of early diagnosis). However, if sensitivity is set too low, more cases may be identified where the likelihood of having the condition is uncertain (false positives). The uncertainty of this diagnosis risks harming babies and families. While this is a factor to consider in any screening programme, this was felt to be particularly important when using genomic sequencing as a technology for identifying conditions.

- “It can create long periods of unknowns for families who may constantly worry at every cough or cold” (Workshop 3)

It was also acknowledged that maximising sensitivity could come at the expense of specificity, meaning that more cases would be missed in order to avoid more false positives. The group felt it was likely that other stakeholders would have the same challenge in navigating this trade-off.

The group highlighted that sensitivity and specificity could vary significantly by condition. One example mentioned was spinal muscular atrophy, where there is a high-cost gene therapy treatments available; specificity and sensitivity would be expected to be relatively high based on the variant known to cause the condition. However, the knowledge and variability of variants associated with other conditions may be much less certain.

There was also uncertainty around the unique selling point of this study over the testing that is already available, and a need to explore what “socially acceptable” levels of specificity and sensitivity might be. This will need to be clearly addressed in the next stages of the process.

By the end of the deliberations, there was general consensus around the importance of both sensitivity and specificity, with some members questioning whether both analytical and clinical validity need to be maximised. The group felt unable to recommend set quantitative definitions for these concepts and felt that parameters should be set by the Genomics England team.

- “It’s difficult to see where the balance should lie, it’s a gene by gene problem” (Workshop 3)

Frequency: how rare is the condition in the population as a whole?

The group initially focused their discussion on the frequency of the variant in certain population groups as opposed to others, and were concerned about the overall equity of the sequencing programme.

There was discussion about screening for conditions which disproportionately affect minoritised groups or specific subgroups; or to setting out decision-making transparently.

- “Should we think about a proportionate impact within sub-populations?” (Feedback after Workshop 2)

It was also acknowledged that the frequency of a condition is often a consideration for inclusion in screening programmes. The group concluded that the frequency of the disease or condition in the population was not the most important issue for this

programme, especially given the technology enables many conditions to be examined at once, regardless of frequency.

Treatment/intervention: how to define what constitutes a treatment, and what is the threshold for any required outcome in terms of the patient's quality of life?

At the beginning of its deliberations, the group was concerned with defining 'treatable' and 'actionable' in the context of any treatment or intervention. Different considerations were brought up, including effectiveness of early treatment; trade-offs of treating early versus the harms of unnecessary treatments (for false positive cases); and the impact on quality of life. As the discussion progressed, the group also considered whether the Programme should also find conditions *without* a curative treatment available. The group indicated that the important consideration would be whether something could be done to improve the child's quality of life, *balanced* against the certainty of the treatment working and the invasiveness of the treatment.

- "Some conditions just need a vaccine while others need a bone marrow transplant" (Feedback after Workshop 2)
- "This treatment [gene therapy for spinal muscular atrophy] has been justified on the grounds that the treatment costs are offset by the savings on long term care for untreated individuals as well as the significant improvements in quality of life for the individual." (Feedback after Workshop 2)

Ultimately, the group argued that the Programme should not be too conservative and focus only on cures, but that it should only screen for things where actionability can lead to significant positive health outcomes. To assess the treatability or actionability of a condition, the group suggested using different dimensions of evidence, which should be explored with publics and patients.

Timing of intervention

In Workshop 2, the discussion focused predominantly on screening for conditions according to when symptoms appear. There was consideration of timing of treatment versus timing of onset of symptoms - for example, if a disease which would cause symptoms in adult life could be screened for and treated in early childhood, then it would be legitimate to consider screening. The group considered the benefits as well as risks and harms of any intervention and stressed the importance of questioning who the beneficiary would be.

- "The best interest of the child is the key question and this relates back to quality of life" (Workshop 3)

Role of patient experience in designing criteria around quality of life

While it was considered important for Genomics England to set the technical thresholds within the criteria for screening, there were some areas where the role of patient experience was seen as critical. While well-understood concepts like the

Quality Adjusted Life Year (QALY) were considered important, it was also argued that the voice of the families affected should also be built into the principles development process, given that it is essential to understand what people themselves value in their quality of life.

- “A violinist losing a finger might affect their quality of life more than another person having a more serious illness” (Workshop 3)

Regular review of criteria and principles

From the beginning of its deliberations, the group agreed that the criteria and principles should be regularly reviewed to change as science and society moves on. The group indicated that patients’ experiences should continue to add perspective to these reviews.

Technical definitions

The group generally struggled to set and define technical criteria like percentages and proportions because of their subjective nature, and due to limited knowledge about many rare genetic conditions.

Distinguishing between conditions with different levels of evidence for treatment - should there be more than one list?

As the Newborn Genomes Programme will be a research study, the working group considered whether parents should have the option to receive findings in distinct groups: for example, a group with conditions where the evidence for treatment or impact on quality of life is much clearer, versus conditions where there is relatively little evidence, or available treatments are in an experimental phase.

3. Principles in detail

Involve's recommendation on the principles

Our recommendation is that Genomics England move to **five core principles which should underlie the choice of any condition (and gene variants causing them) to be analysed and fed back to families as part of the initial screen.**

Deliberation from the working group revealed further queries and recommendations for further refinement. There were also some differences of opinion on how best to prioritise the principles and how to present them as a set. Therefore, we have recommended a further compression of the principles and some reordering, plus a way of presenting them which can be tested in the next stage. Of note, some working group members commented that this compression went too far the other way – i.e., “lost too much detail about analytical and clinical validity overall” – which will need to be further explored via engagement in the next stage and in further iterations.

Based on questions raised by the working group, we added technical details provided by Genomics England to define terms and provide clarity. At the next stage, there needs to be further exploration of terms such as ‘strong’, ‘most’, ‘how sensitive’, and ‘how specific’.

When reviewing the principles below, please note the following points / the following should be considered:

- Anything included in the programme has to adhere to **all** five principles. (principle A AND principle B AND principle C AND principle D AND principle E).
- Three of the five principles include detailed criteria and issues to consider. The group wanted all these definitions and issues to be considered as part of deciding whether the genetic variant fulfils each principle. However, not all the criteria have to be met for the principle to stand, and the programme team should be able to use some discretion (e.g., applying criterion one AND/OR criterion two).
- Where there was disagreement or uncertainty in the group about how to word a principle, alternative wording has been suggested that may be further discussed in the next engagement stage
- Some genetic conditions will be rare, such that there are only a few reported cases in the literature. As such, data about penetrance (the proportion of individuals with the variant that will develop the condition) and the natural history of the disease will be limited, particularly in the context of identifying a variant in a presymptomatic individual. Therefore, all of the principles should be considered in the context of existing knowledge, which will change over time in light of new information. There was discussion in Workshop Two about this limitation, and the opportunity for the Newborn Genomes Programme to gather this information in the context of a research study.
- Whole genome sequencing in newborns will overall aim to have a high specificity in order to minimise the potential of false positives. As a result, the sensitivity of individual genes will vary. The individual and sum of all genetic disease sensitivities and specificities will need to be assessed.

- It is essential that these principles should be reviewed over time. Additionally, conditions, genes, and variants should be reviewed and adjusted in relation to the principles during the course of the study.

The principles

Include the genetic variant in the screening programme if there is strong evidence...

A: That the *genetic variant(s)* causes the condition

Technical notes:

This refers to pathogenic and likely pathogenic variants, in line with UK Best Practice Guidelines for Variant Classification in Rare Disease

(<https://www.acgs.uk.com/media/11631/uk-practice-guidelines-for-variant-classification-v4-01-2020.pdf>)

B: That a high proportion of individuals who have the genetic variant(s) would be expected to have symptoms that would significantly impact their quality of life if left untreated

Alternative...

That individuals who have the genetic variant(s) would be expected to have symptoms that would significantly impact their quality of life if left untreated

Criteria: *To satisfy this principle, the following must be considered:*

i) symptoms have a significant impact on quality of life, measured in QALYs where available or taking into consideration factors such as how serious the condition is and the testimony of people affected by the condition

ii) if left untreated, symptoms would typically start in childhood (less than 16 years).

Alternatives...

if left untreated, symptoms would typically start in early childhood

if left untreated, symptoms would typically start by age 5

Technical notes:

This principle refers to penetrance, and knowledge of penetrance in individuals with a known pathogenic variant may be limited. Due to ascertainment bias and/or lack of an evidence base this information may not be available for all the disorders in the test.

Based on the workshop on some of the case examples (e.g. complement component 5 deficiency), there was discussion regarding whether a condition with lower penetrance (Principle B) may still be considered if the available treatment is low risk and with significant benefit (Principle C).

C: Pre-symptomatic or early treatment(s) for the condition has been shown to lead to improved health outcomes for the child, compared to treatment after onset of symptoms.

Criteria: *To satisfy this principle, the following must be considered:*

i) the treatment available has to be initiated in early childhood

ii) the treatment available for the condition could either cure, delay, or modify the course of the condition

iii) the treatment available provides a measurable improvement in quality of life, measured in QALYs where available or taking into consideration factors such as available clinical evidence and the testimony of people affected by the condition

Technical information:

Treatment may be defined as an intervention that can either cure, delay, or modify the course of the condition

D: There is a minimally invasive confirmatory test that can establish whether or not the child has the condition.

E: Conditions screened for are only those for which the socially acceptable interventions are equitably accessible for all as standard of care within the NHS.

4. Conclusions and recommendations

Conclusions

In Involve's view, the process of developing the principles has been robust, and the principles developed reflect the key concerns of the working group. They can now be taken forward into greater exploration with the public and patient groups, with further iteration from the working group.

The general public, patient groups, and a range of healthcare professionals (those who are not genomics specialists) will have a view on the principles, including from a lay perspective and in terms of the principles which will have most impact on the lives of patients and people living with rare diseases.

The issues around the Programme's screening choices are complex; this has been apparent as a group of experts from various disciplines have taken a number of sessions to come to the level of consensus we have reached.

To get actionable feedback from wider groups, the issues need to be framed properly and questions crafted carefully so that the public, patients, healthcare professionals, and other stakeholders can see what they are being asked to do and what decisions they are being asked to make.

Raising awareness and promoting engagement with the principles will also be important. This will depend on appropriate framing, crafting questions, and providing digestible information which gets to the heart of the issues.

Recommendations for next stages of engagement

To share the principles with a wider audience, the following will be crucial:

Education about genomics and the particular challenges of the screening programme.

There are a number of useful presentations created by Genomics England and members of the working group for this stage, which should act as starting points for a body of information to share with other stakeholders.

Covering the key areas that working group stakeholders recommended for future discussion with publics and patients.

Key areas to explore with families and patients (arising from working group discussions):

- Experience and beliefs about the *testing* process for both newborn screening and confirmatory tests. Plus any trade-offs between perceived *invasiveness* of tests, risks of doing (or not doing) *confirmatory* tests, and *timeliness* of getting results back. What are the principles and red lines for patients and members of the public here?
- Experience of the *diagnostic odyssey* and discussion of how decisions made around the screening programme could help or hinder this.

- A deeper sense of how patients and frontline clinicians think about how *patient experience of their condition* should be defined. Terminology like ‘severity’, ‘quality of life’, or ‘experience of symptoms’, should be discussed, to inform the way that assessment of quality of life is made in the principles. While the public and, to some extent, healthcare professionals cannot define the precise assessment criteria, it is important to know the best ways of framing the impacts of conditions on quality of life, and the impacts of uncertainty around testing on quality of life.
- Issues of *consent*: what should be the balance between early screening and leaving the (newborn) child until an age where they are able to decide for themselves what to test for?
- Issues of *equity*: how should the wider outcomes of different groups in society be considered with relation to the programme?

Exploration of principles in detail. Wider engagement should explore the principles as they stand in the light of the **key trade-offs** for the programme. Using case studies and testing the draft principles against them will be important: including the ‘edge cases’ which different principles might lead to including or excluding.

Focusing on lived experience of patients. An exploration of the principles should be set in context of people’s lived experience with the conditions they have, their diagnostic odyssey, and their experience of having a genetic condition within their families. It will be important to ask sensitively about the inflection points on their journeys, and the decisions that were, and could have been, made early in their children’s lives.

Recommendations for the presentation of the principles

We recommend that the rubric to go along with the principles should explain:

- The nature of the programme and its aims
- The fact that the principles’ criteria have been selected to balance the patient’s experience of the process, and familial and social elements, with the potential certainty of diagnosis, likelihood of different prognoses, and the nature of possible treatment.

At the next stage of engagement, different ways of showing the principles **visually** should be trialled, for example as a decision tree or a series of weighted criteria.

For any condition screened for, where the aim is to diagnose or help patients, the programme should produce guidance – e.g., a timeframe for intervention for each condition, ways of communicating the uncertainty of the screening process, any additional tests that are required to confirm the results of the screening test, age of onset, and prognosis to families and to society as a whole; with consideration of

second order impacts, and ethnic and other biases – as they relate to that specific condition.

Recommendations on glossary

For future stakeholder engagement, we recommend a glossary should be provided to include recommended definitions and descriptions of the issues. The definitions should be ready when materials for any future engagement are developed.

Recommendations on usage of other screening criteria

At different points in the discussion and feedback, working group members of recommended there should be consideration of, and learning from, other past and current screening criteria.

Wilson and Jungner (1968) 'Principles and Practice of Screening for Disease' outlined 10 principles that should be considered for diagnostic genetic testing. These are still the nominal foundation for screening, but they have evolved and been scrutinised in the light of advances in genomics and other research areas over the years. The following are, in summary, the dimensions in which the current draft principles deviate from Wilson and Jungner's:

- Issues of equity and the possibility of perpetuating inequalities
- Including the child or young person in decision making process
- Testing closer to age of onset
- Treatment with focus on benefits, including improvements in quality of life
- Issues of consent

These changes reflect the important differences between Wilson and Jungner's 1968 socio-political context and that in 2022 England as well as accounting for the agency of, and consequences for, both patients and their families.

Stakeholders also mentioned taking guidance from the UK National Screening Committee - [criteria](#)

There are some key differences in the use of these in the national screening programme and in the new study:

- The new study will focus on severity more than frequency of the condition because the programme is about rare disease, thus other factors are prioritised
- Planned review of selection methods.

Concluding note

This report has explored the working group's rich discussions and deliberations on the principles which will provide the foundation for the Newborn Genomes Programme's approach to choosing the conditions, genes, and variants which will be initially screened for. This is a first step in what we hope will be a wider programme of

engagement, which should reflect a range of stakeholders' views, concerns, and expectations of the principles which will underpin this important research study.

Appendix

Working group members

** Indicates the individual is also a member of the NHS Steering Group*

- **Chair:** Dr Emma Baple, Medical Lead for Rare Disease, SW GLH (CHAIR)*
- Professor Elijah Behr, Professor of Cardiovascular Medicine, St George's University of London
- Dr Felicity Boardman, Professor in Medicine, Ethics and Society, Warwick Medical School
- Dr Mike Champion, Consultant in Children's Inherited Metabolic Diseases And Clinical Lead, Evelina London Children's Hospital
- Dr Ngozi Edi-Osagie, Consultant Neonatologist, Manchester University NHS Foundation Trust*
- Dr David Elliman, Clinical Lead for NHS Newborn Infant Physical Examination Programme and NHS Newborn Blood Spot Screening Programme, Public Health England*
- Dr Francis Elmslie, Consultant Clinical Geneticist, St George's University Hospitals NHS Foundation Trust*
- Brad Gudger, Founder and Director, Alike
- Alison Hall, Senior Advisor (Humanities), PHG Foundation
- Georgia Hayes, Institute of Health Visiting
- Michelle Lyne, Royal College of Midwives*
- Dr Emma McCann, Medical Director, North West Genomics Laboratory Hub
- Dominic McMullan, Consultant Clinical Scientist, Central and South Genomics Laboratory Hub
- Simon Ramsden, Consultant Clinical Scientist, North West Genomics Laboratory Hub
- Jo Revill, CEO, Royal College of Paediatrics and Child Health*
- Professor Rob Taylor, Head of Highly Specialised Mitochondrial Laboratory, Newcastle upon Tyne Hospitals NHS Foundation Trust
- Sally Shillaker, Institute of Health Visiting
- Professor Caroline Wright, Professor in Genomic Medicine, University of Exeter
- Sarah Wynn, CEO, Unique

NHS England & NHS Improvement team

- Sarah Jevons, Head of Policy and Strategy
- Donna Kirwan, Genomics Midwifery Lead
- Alexandra Pickard, Deputy Director for Genomics*

- Laurence Russell, Senior Policy and Strategy Manager

Genomics England team

- Arzoo Ahmed, Ethics Lead
- Dr David Bick, Principal Clinician
- Dasha Deen, Genome Data Scientist
- Kate Harvey, Engagement Manager
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- Alice Tuff-Lacey, Programme Lead*
- Amanda Pichini, Clinical Lead for Genetic Counselling*
- Dr Richard Scott, Chief Medical Officer*
- Simon Wilde, Engagement Director