

# Newborn Genomes Programme



Continuing the conversation about offering whole genome sequencing (WGS) to all newborns





**1 Vision development**  
Public engagement, expert interviews, and NHS steering committee set up to gauge support and interest, and develop the vision

September 2021

**2 Co-design and feasibility**  
Design, test, and iterate how the pilot might work and feel

**3 Review**  
Review to inform pilot implementation

**4 NHS research pilot**  
Proof-of-concept in several Trusts and through the NHS Genomic Medicine Service to evaluate benefits and implementation

**5 Review**  
NHS Genomic Medicine Service Research Collaborative to coordinate the evaluation of the evidence from the pilot to inform decisions on health services

**6 Implementation into the NHS**  
If the pilot is successful, implementation into NHS routine care

This document shares the joint NHS England and NHS Improvement (NHSE/I) and Genomics England vision for the Newborn Genomes Programme, developed following our public and expert dialogues. It outlines initial design themes, and the iterative, collaborative approach we will take over the coming years.

The Newborn Genomes Programme will co-design and run an ethics-approved research pilot embedded in the NHS to explore the benefits, challenges, and practicalities of offering whole genome sequencing (WGS) to all newborns to accelerate diagnosis and access to treatments for rare genetic conditions. The programme will aim to:

- 1** Evaluate the utility, feasibility, and impact on the NHS of screening for a larger number of childhood-onset rare genetic conditions in newborns, including what support they will need
- 2** Understand how, with consent, the genomic and health data could be used for research to enable new diagnostic discoveries and treatments to be developed
- 3** Explore the potential risks, benefits, and broader implications of storing an individual's genome over their lifetime

The NHS research pilot will align clinical care with research and will feed back insights to parents through the NHS within a research protocol. It's crucial that these research aims are carried out by maintaining public trust while considering the impact on NHS clinical care and workforce. We'll ensure our work is human-centred, ethical, and translatable into a clinical service.



**Sarah-Jane Marsh**  
**Chair**, Newborn Genomes Programme  
NHS Steering Group  
**CEO**, Birmingham Women's and Children's NHS Foundation Trust



Genomics is transforming our healthcare system, enhancing our ability to provide personalised care, with so much more to discover.

By establishing the clinical pathways for WGS in newborn babies, this groundbreaking programme will evaluate such an approach for its benefit and impact, with a view to incorporating it into the standard NHS offer in a safe and effective manner, providing a supportive experience for families.

Having worked with children with rare diseases for over a decade, this really is a unique opportunity to add years to life, and life to years, whilst facilitating research to continually enhance care across all disease groups.

I am proud to work in a universal health service free at the point of need, that operates at the limits of science, and I truly believe we are in a fantastic place to lead the way globally on a programme of such huge scale and significance.



**Rebecca Middleton**  
**Vice-Chair**,  
**Participant Panel**,  
Genomics England



I am heartened to hear that the programme will have the necessary time to consider potential patient journeys, and consider equity of access and family support.

The public dialogue demonstrated that people see the potential power of genome sequencing but want us to get this right, from the very start. Co-designing the pilot with patients, families, and clinicians will ensure that we understand their needs and concerns, and act on experience. This is a really exciting and powerful opportunity.

Our work in offering WGS to all newborns will focus on understanding and evaluating if and how the following potential benefits can be achieved:



### 1 Early diagnosis and care for childhood-onset rare genetic conditions

- Postnatal screening only**
- Actionable, childhood-onset conditions only**

Building on the principles of the NHS newborn screening programme, up to 200,000 babies' genomes will be sequenced and analysed for a set of actionable genetic conditions which may affect their health in early years. This aims to ensure timely diagnosis, access to treatment pathways, and enable better outcomes and quality of life for babies and their families.

We will work with parents, individuals with a rare genetic condition, multi-professional NHS workforce and experts to co-design future care and support pathways, the feasibility of delivery, and how they will be implemented and resourced, and to develop the principles to define actionability and which conditions to include.



### 2 Enabling research and new treatments for NHS patients

With parents' consent, babies' genomes will be de-identified and added, alongside their health data, to Genomics England's Research Environment. This will enable vetted academic, clinical, and commercial healthcare researchers to improve understanding of health and disease, develop new diagnostics and treatments, and understand how current therapies can be improved or repurposed.

Building on our existing governance practices for data protection and access, we will collaborate with experts, individuals with a rare genetic condition, and the public to ensure this data is accessed and used appropriately, now and in the future.



### 3 Exploring the potential of a lifetime genomic record

Storing babies' genomes securely, regardless of their newborn screening outcome, could allow them to be reanalysed as needed, potentially enabling access to new developments in genomics throughout their lifetime.

We will explore the benefits as well as the ethical and practical implications of storing participants' genomes, how and when it's appropriate for participants' data to be reanalysed in accordance with their choice, and the impact on them and their future clinical care.



# Today

Working in partnership to deliver the 100,000 Genomes Project and the NHS Genomic Medicine Service, NHSE/I and Genomics England have established the infrastructure to support WGS diagnostic and treatment pathways for children and adults with symptoms, and developed the sequencing and analytical pipelines to ensure a safe and effective service.

For many individuals with rare conditions and their families, WGS has brought an end to the diagnostic odysseys that they had been on – providing more certainty and choice. It has demonstrated the life-changing potential of using genomic medicine in healthcare.



# Tomorrow

We want to build on these strong foundations. Considering our increased understanding of the links between the genome and health, as well as the analytical and healthcare capabilities we have already developed in WGS and the NHS, we believe now is the time to explore extending this life-changing potential to newborns.

Providing WGS for newborns could help transform diagnostic odysseys like Jessica's, ensuring babies get access to appropriate treatments and interventions much earlier. It could also enable researchers to discover and develop new ways to use genomic medicine to help treat and save lives – and it could usher in a future of personalised, preventative healthcare.



## Every year 3,000+ babies

could benefit from **life-saving** or **life-changing** interventions thanks to WGS\*

\*conservative number based on an assessment of the potential of WGS as a screening test in newborns, by the Genomics Analysis in Children Task and Finish Group



## Jessica's story

Jessica was born with GLUT1 deficiency syndrome, which affects her movement, general development, and causes epilepsy.

It took years to find a diagnosis for Jessica - a diagnostic odyssey which included MRI scans under general anaesthesia, lumbar punctures, and EEGs.

Taking part in the 100,000 Genomes Project enabled her diagnostic odyssey to end, allowing her parents and doctors to focus on helping her live a healthier life.



**If we had this done when Jessica was born and found out the results straight away, we would have been on the right track immediately."**

**Jessica's Mum**

**While earlier diagnosis can feel life-changing, tailored intervention may have the potential to limit or prevent conditions from progressing altogether.**

By aligning clinical care with research, we hope to accelerate the discovery, development, trial, and approval of treatments for clinical use in the UK. Examples of rare genetic conditions for which therapies are already being trialled or used include:

- **Duchenne muscular dystrophy:** Translarna helps some children produce a protein essential to muscle development, when they would otherwise develop progressive weakness
- **RPE65-mediated retinal dystrophy:** Luxturna improves vision in children who would otherwise experience progressive, irreversible vision loss
- **Haemophilia A:** Roctavian increases the production of a protein needed for the blood to clot, and reduces the need for regular replacement therapy



# Our engagement efforts so far

We have been working with a diverse range of communities and groups to understand the ambitions, benefits, and challenges for a pilot. We will continue to work with them to explore these and to define the key themes and questions we need to look at when designing the pilot.



# A collaborative and iterative way forward

We will keep talking and listening to the public and experts in a dialogue regarding the most appropriate uses of WGS in newborns.

At the same time, we'll collaborate with parents, individuals with a rare genetic condition, and professional experts to co-design key elements of the process and ensure equitable access for all communities.



## Public dialogue

Together with the UK National Screening Committee, we commissioned an in-depth national dialogue involving over 130 members of the public from a range of backgrounds and places within the UK, including individuals with genetic conditions, new/expectant parents, ethnic minorities, and young adults.

This initial piece of work found high levels of support and excitement for a pilot, and uncovered some concerns which were fed into our vision. The report and findings were launched in July 2021 at an event attended by over 1000 members of the public.



## NHS Steering Group

The NHS Steering Group was established to support and develop the programme and its design, and understand the implementation considerations necessary to support the potential rollout of a NHS Newborn WGS service.

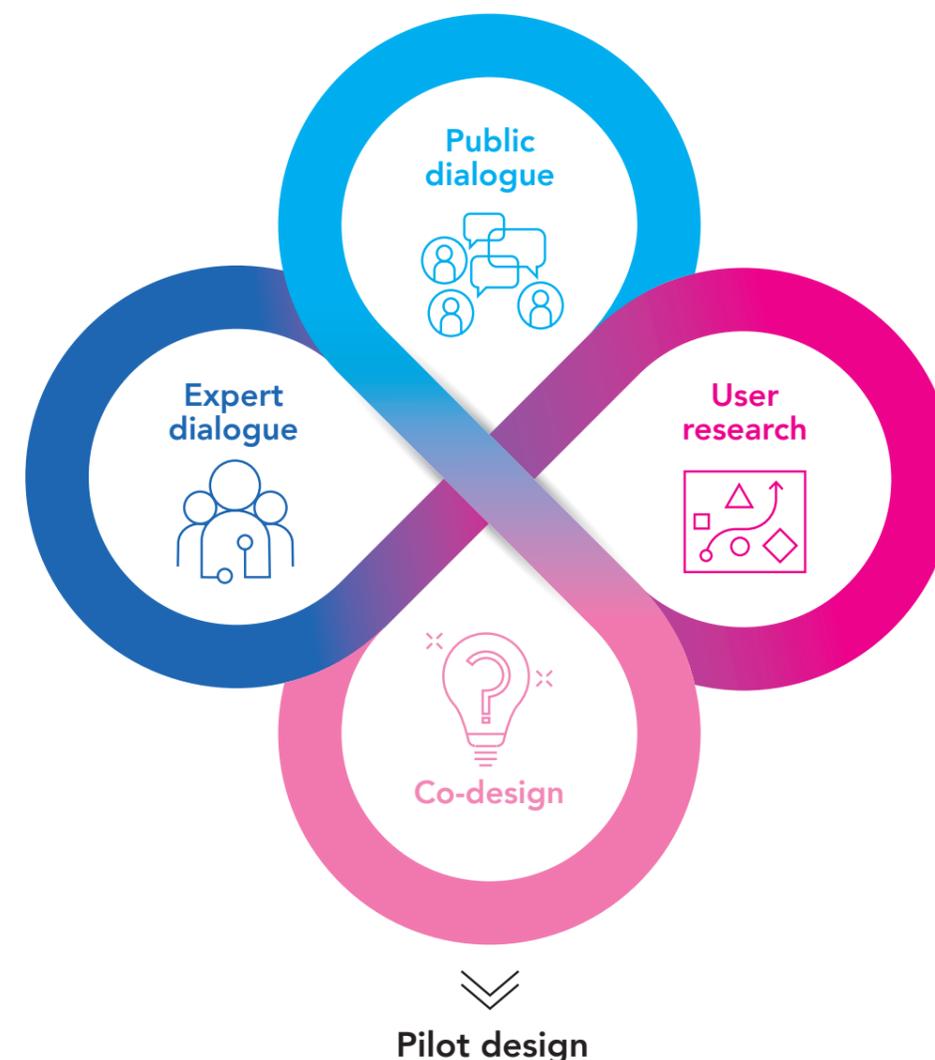
The group shares ideas, concerns, and expectations as key themes are developed for the design and potential rollout of the pilot.

The list of NHS Steering Group members is on page 10.



## Expert interviews

We have interviewed a number of communities with expertise across maternity healthcare, genomic healthcare, ethics, and people with lived experience in order to road-test the vision and define initial design themes to prioritise.



Our dialogues with the public and experts have highlighted some initial themes to prioritise in the co-design and feasibility phase:



### The benefits, limitations, and unknowns of WGS as a screening tool

The potential of WGS for screening is enormous, but there are questions remaining:

- How it will integrate with the NHS newborn screening blood spot test
- How conclusively it can predict disease in pre-symptomatic babies, and how it impacts their diagnostic experience
- How it should be paired with other screening and diagnostic tools
- How to best guide parents through that uncertainty to avoid unnecessary 'medicalisation'

“Up until recently we have started with signs/symptoms and used genomics to confirm a diagnosis. In newborn screening we start with a genome and try and interpret what symptoms a baby might develop. Given the 4 million or so variants per genome that need to be filtered, this inverted approach is likely to be more difficult than it sound”

Clinical Geneticist

“I used to be quite scared that people would be uncomfortable with the notion [of uncertainty], but in my experience, if you are upfront and honest with them, they understand.”

Genetic Counsellor

### Co-developed principles for including conditions in the screening panel

The logic behind which conditions are included in the initial panel of conditions we will screen for must be developed in a transparent and equitable way, with the active involvement of rare disease communities as well as experts.

“By only looking at clinical markers of severity, you don't get the lived experience, which is ultimately what the parents are interested in. What is my child's life going to be like? The expert knowledge [patients] have is really important to include.”

Professor of Social Science in Medicine & Genomics

“The most important word is 'improved outcome'. What counts as an appropriate treatment and what we mean by actionability are going to be central themes in the programme.”

Clinical Geneticist

### Person-centred consent across screening, research, and reanalysis

We need to set the bar to ensure all parents are empowered to make informed choices in terms of opting into the programme and understanding what it entails - and how their children will be able to make their own choices too.

“Pregnant women already have a tsunami of information coming towards them. Genomics is difficult for many people to understand, it will need more than just giving them written information.”

Obstetrician

“At the beginning [of the 100,000 Genomes Project], lots of people signed up for additional findings - but then life happens. It's important that reanalysis is not just based on clinical care but takes into account the patient's appetite for knowledge at that time. We must ensure the patient is in the driver's seat at all times.”

100,000 Genomes Project Participant, Adult Rare Disease Patient

### A supportive and inclusive experience for all families

It's crucial that we design the end-to-end experience thoughtfully and inclusively, so that parents and children aren't overwhelmed and always know where to find support, no matter their background or screening outcome.

“Very much part of getting to know that family is understanding their culture, ensuring that we are led by them in terms of what their needs are, but also advocate for the child by linking them with the experts or specialists that they need.”

Health Visitor

“I am a firm believer that emotional support is most effective from someone who has experienced the emotional turmoil that a patient's family is feeling. On completion of genetic counselling, ongoing support and follow-up could be carried out by that particular group. e.g. cystic fibrosis group.”

Public Dialogue Participant, Genetic Conditions Group

### Trusted and future-proofed genomic data storage and usage

People see the potential in the re-analysis of genomic data for healthcare purposes throughout an individual's lifetime. However, it's crucial we ensure the public trusts that their data is appropriately used and safe-guarded, and define together what it means to future-proof it.

“Genomics England's current method of having deliberative discussion on whether or not to provide access to the [research] environment, and how that access is provided - both of those are jewels in the crown of the process.”

Genetic Alliance UK

“In the future, you might build in a regular review: if the child is a certain age, you're looking for a spectrum of conditions likely to arise at that age. And obviously, once the child is mature enough to make their own decision about taking part, you've got the issue around re-consenting.”

Health Policy Researcher

### A sustainable and scalable programme for the NHS

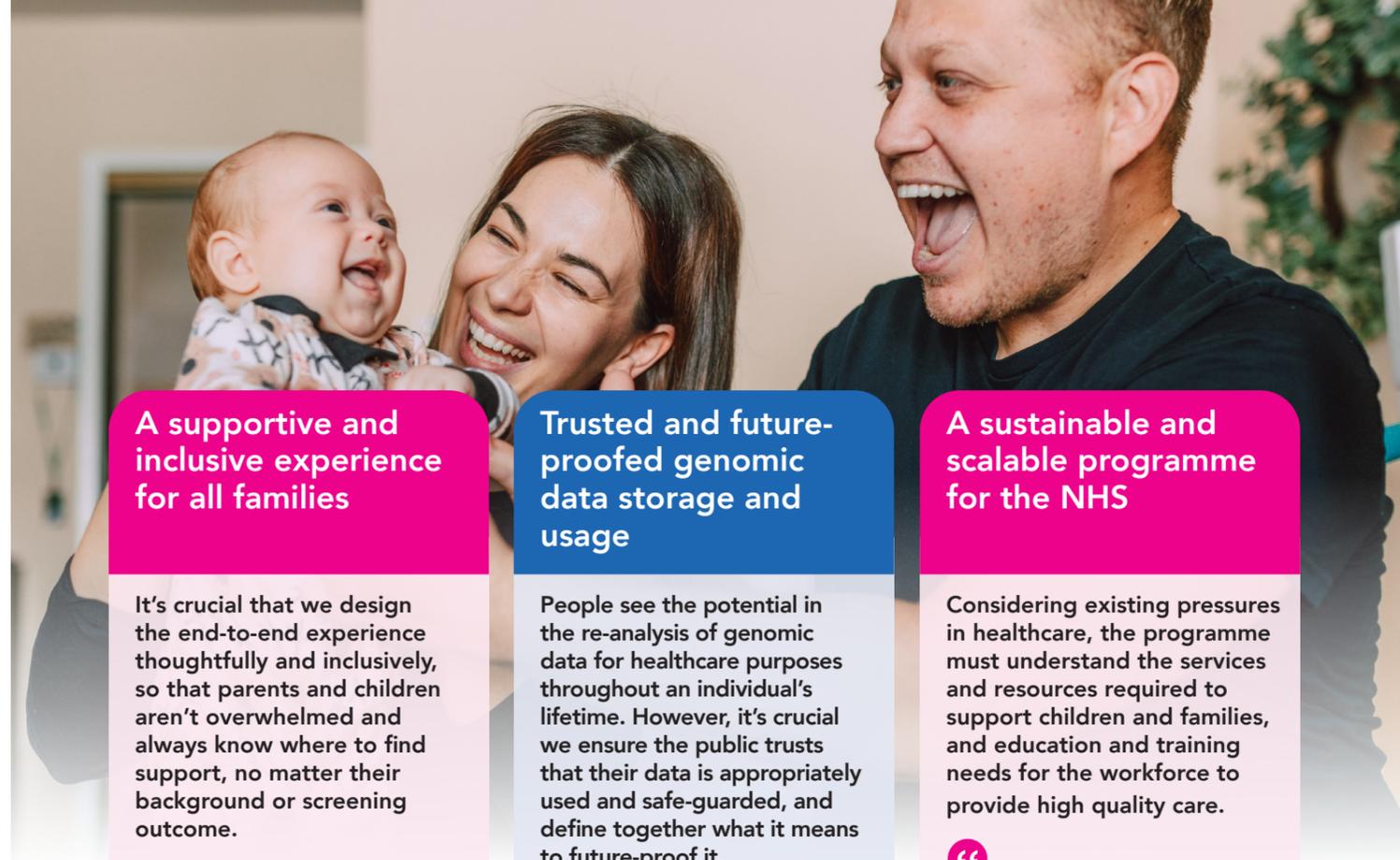
Considering existing pressures in healthcare, the programme must understand the services and resources required to support children and families, and education and training needs for the workforce to provide high quality care.

“As a midwife I would want to have clear guidance on what I need to discuss with a woman. What resources can we give them, where can I signpost them to find answers?”

Midwife

“What will provide much better quality of care is making sure that the clinical pathways and the guidance are very robust. If a condition won't appear until a child is 8 or 10, we need to know exactly what symptoms might come with that condition, what referrals might be needed and when.”

General Practitioner



**Who we'll involve:**

**Public and rare disease interest groups**

A representative sample of new parents, patients with rare genetic conditions, 100,000 Genomes Project Participant panel members

**Royal Colleges**

Midwives, GPs, Paediatrics and Child Health, Nursing, Physicians, Obstetricians and Gynaecologists

**Healthcare professionals**

Health visitors, midwives, neonatal nurses, GPs, paediatricians, genetic counsellors, clinical geneticists, clinical scientists

**NHSE/I, Maternity and Child Services, Diagnostic services**

Various teams

**Ethics experts**

**UK National Screening Committee**

**Genomic Laboratory Hubs & Genomic Medicine Service Alliances**

**Healthcare researchers**

Academic, clinical, and healthcare industry



**NHS Steering Group**

**Carmel Bagness**

Professional Lead, Midwifery and Women's Health, Royal College of Nursing [RCN]

**Dr Emma Baple**

Medical Lead for Rare Disease, South West Genomic Laboratory Hub

**Prof Lucy Chappell**

Chief Scientific Adviser, Department of Health and Social Care

**Dr Ronny Cheung**

Consultant General Paediatrician, Evelina London Children's Hospital

**Prof Lyn Chitty**

Professor of Genetics and Fetal Medicine, Great Ormond Street Hospital NHS Foundation Trust

**Dr Chris Dewhurst**

Clinical Director for Family Health (Maternity and Neonatal Services), Liverpool Women's NHS Foundation Trust

**Prof Jacqueline Dunkley-Bent**

Chief Midwifery Officer for England, NHS England and NHS Improvement [NHSE/I]

**Dr Ngozi Edi-Osagie**

Chair, Clinical Reference Group for Neonatal Critical Care, NHSE

**Dr David Elliman**

Clinical Lead for NHS Newborn Blood Spot Screening Programme, Public Health England [PHE]

**Dr Frances Elmslie**

Clinical Director, NHS South East Genomic Medicine Service Alliance

**Prof Dame Sue Hill**

Senior Responsible Owner, Genomics Programme NHSE/I

**Dr Larissa Kerecuk**

Rare Disease Lead, Birmingham Women's and Children's NHS Foundation Trust

**Prof Mark Kilby**

Incoming Chair, Genomics Taskforce, Royal College of Obstetricians and Gynaecologists

**Donna Kirwan**

Genomics Midwifery Lead, Genomics Unit, NHSE/I

**Michelle Lyne**

Education Adviser, Royal College of Midwives

**Prof Anne Mackie**

Director of Screening, PHE

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CEO, Birmingham Women's and Children's NHS Foundation Trust

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**Prof Anne-Marie Slowther**

Professor of Clinical Ethics, University of Warwick

**Prof Helen Stokes-Lampard**

Chair, Academy of Medical Royal Colleges

**Alice Tuff-Lacey**

Programme Lead, Newborns, Genomics England

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CEO, Genomics England



# Stay in touch

Find out more at:  
[www.genomicsengland.co.uk/newborns](http://www.genomicsengland.co.uk/newborns)

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programme in detail, please contact  
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