



Generation
Study

Introducing the Genomics England Generation Study



Introducing the Generation Study

<https://www.generationstudy.co.uk/>

- The Generation Study is a long-term research study being run by Genomics England in partnership with NHS England.
- The study involves collecting a blood sample after birth and aims to understand if we can improve how we diagnose and treat genetic conditions by looking at the DNA of newborn babies.
- The Study will sequence and analyse the genomes of 100,000 newborn babies in England to look for certain treatable genetic conditions, with treatment options available through the NHS in England.
- The Study also aims to understand how babies' genomic data could be stored over their lifetime and used for research.

V1.0 IRAS number: 324562 Dat e: 31/03/2023

Genomics England | Generation Study



Free and optional

Place your local sticker here

Generation Study

Test your baby for 200+ genetic conditions

Contribute to research that could lead to new treatments. Sign up when you're pregnant, and the test will happen shortly after birth. Any suspected conditions can be treated in the NHS.



Scan to visit our website or talk to your study team

Current NHS Newborn Blood Spot (NBS) Screening Programme

Newborns can currently be screened for these conditions via a blood spot test.



There is a 97% uptake of newborns screening in the UK

- Sickle cell disease
- Cystic fibrosis
- Congenital hypothyroidism
- Phenylketonuria
- Medium-chain acyl-CoA dehydrogenase deficiency
- Maple syrup urine disease
- Isovaleric acidaemia (IVA)
- Glutaric aciduria type 1
- Homocystinuria
- Tyrosinaemia type 1 (to be added from September 2025)
- In service evaluation for SCID (in two thirds of England)

What is screening?

Screening

A way of identifying apparently healthy people who may have an increased risk of a particular condition.

Diagnosis

The process of identifying a condition from its signs, symptoms and test results.

Why is the generation study being done?

A

NHS screening currently **only looks for 10 conditions**, rather than screening the baby's genome. **We are testing a broader approach.**

B

“There is a clear potential for genomics in the testing for many of the conditions currently included in the blood spot test.” Generation Genome 2020

C

Support for genome sequencing in newborn screening from Genetic Alliance UK in the **Patient Charter on Newborn Screening 2018** and in the **Genomic Analysis in Children Task and Finish Group 2019**

D

Screens 200+ diseases in a single test, enabling a new screening platform expandable to even more conditions.

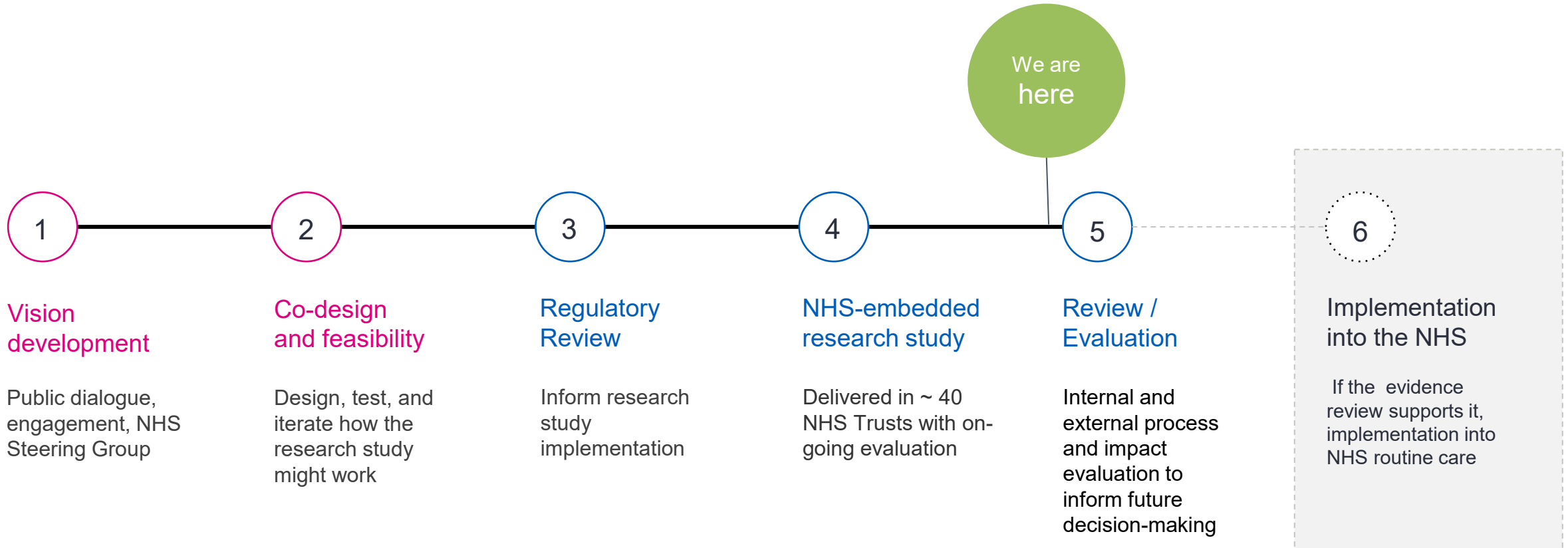
A genome and genome sequencing

- A genome is a person's entire genetic sequence — their body's instruction manual.
- Genomes are made up of a chemical called DNA and contain thousands of genes.
- Genes tell our bodies how to grow and develop.
- Looking into someone's genome can give us information. That's because changes in a genome can lead to health problems, including rare conditions.

- To study someone's genome, we take a DNA sample. It usually comes from a few drops of blood.
- From that, we create a digital file of their genome. This process is called genome sequencing.
- It's like taking a full body scan, but the radiologist only focusses on looking at certain images/areas of the body.
- Important to avoid the potential misconception that if no conditions are suspected, this is a 'clean bill of health' for the baby

Study Timeline

Where are we?



68 hospitals across 43 NHS Trusts are now live and recruiting.
With >36,500 parents recruited, and 28,000 samples from newborns collected.
>25,000 No-condition suspected results.
>93 condition suspected results (0.4%).

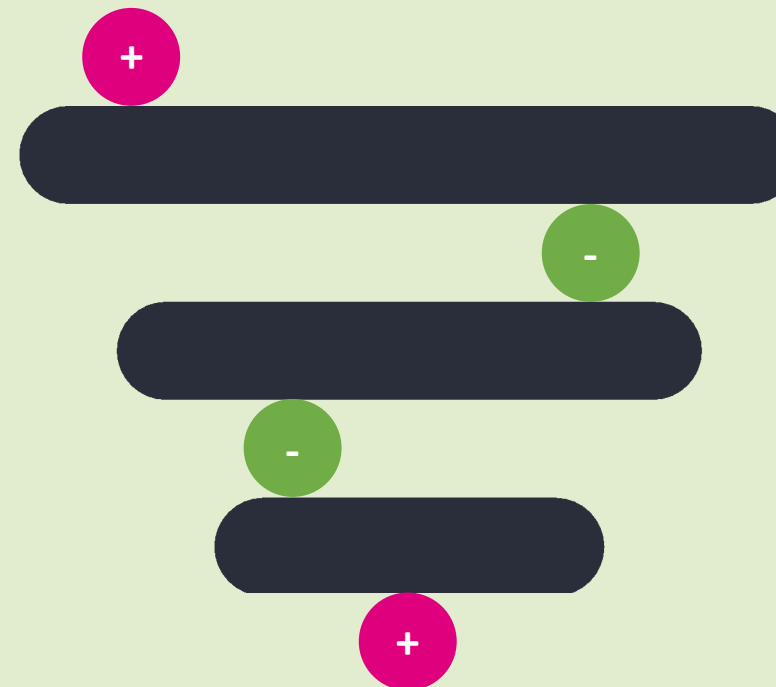
Choosing which conditions to screen for

The challenge: there are thousands of conditions that *could* be detected through whole genome sequencing – but we may not want to look for all of them

The programme will only screen for a **specific set of conditions, genes and variants**

Principles and criteria for screening already exist – we are taking a **bespoke approach** in the context of a **UK-based research programme**

Four principles...



Guiding Principles for Condition Selection

- **Principle A:** there is strong evidence that the genetic variant or variants cause the condition and can be reliably detected
- **Principle B:** a high proportion of individuals who have the genetic variant or variant would be expected to have symptoms that would have a debilitating impact on quality of life if left undiagnosed
- **Principle C:** early or pre-symptomatic intervention for the condition has been shown to lead to substantially improved outcomes in children, compared to intervention after the onset of symptoms
- **Principle D:** Conditions screened for are only those for which the interventions are equitably accessible for all

Learn more:

<https://www.genomicsengland.co.uk/initiatives/newborns/choosing-conditions>

Establishing conditions, genes and variants in six stages...



Co-developed



Evidence-based



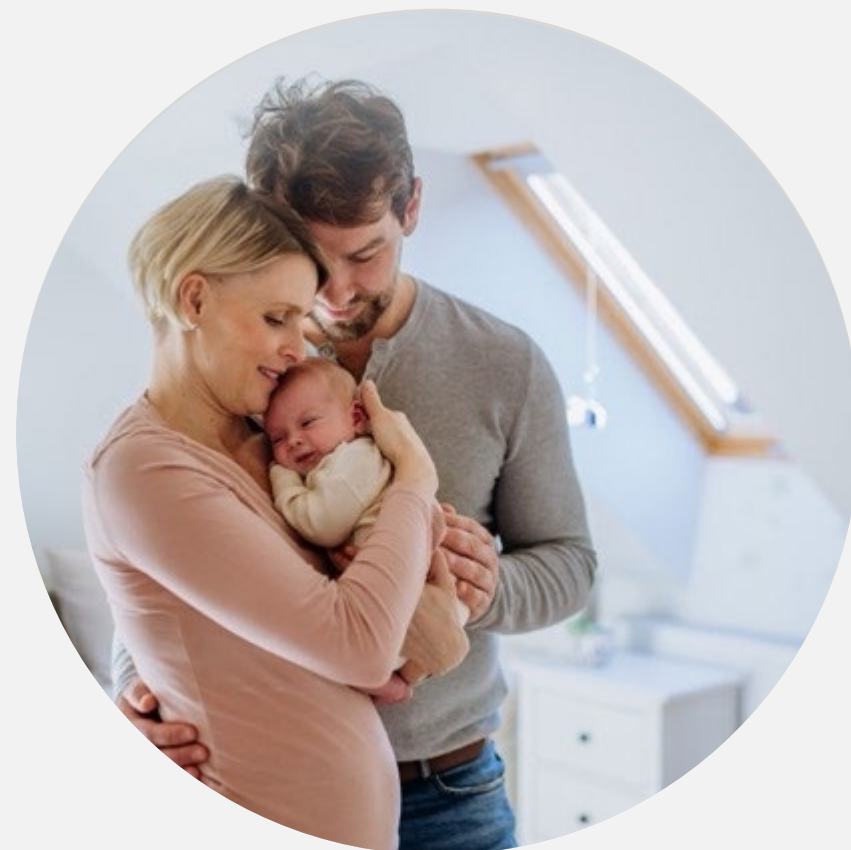
Transparent



Flexible

Conditions Screened For

- Since initial publication of the provisional conditions list in October 2023, 12 conditions (including 21 genes) have been removed from the list.
- Removed due to conflicting evidence or a poor ability to detect genetic variants in those genes.
- The current list of conditions includes 208 conditions (462 genes)
- The list can be found here <https://www.genomicsengland.co.uk/initiatives/newborns/choosing-conditions/conditions-list-generation-study>



Adding New Conditions

- Review and implementation of changes followed the same sign-off processes used to finalise the initial list of conditions in the study, including engagement with patient organisations, NHS England and relevant Clinical Reference Groups.
- January 2026 - plan to add approximately 50 new conditions (around 100 genes) subject to review by NHS England to ensure availability of treatments in the NHS and that there is capacity and capability in NHS services to accommodate these new conditions.
- The finalised list will be published on the Generation Study and Genomics England websites in early 2026.
- Babies in the Generation Study will be tested for the conditions listed at the time they joined the study, and will not be re-analysed if this list changes.
- The process for choosing conditions can be found <https://www.genomicsengland.co.uk/initiatives/newborns/choosing-conditions>

Participant journey



Participant Information



Participant information sheet



Leaflet



Site pull-up banner



Leaflet

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Participant information:

<https://www.generationstudy.co.uk/overview-of-the-study/join-the-study>

List of participating hospitals:

<https://www.generationstudy.co.uk/register-your-interest/participating-hospitals>

Limitations of Genomic Screening

- No screening test is perfect and there will always be some incorrect results.
- There can be 'false negative' and 'false positive' results

False positive: where the screening identifies a condition that turns out not to be there.

False negative: where the screening misses a problem and suggests there are no suspected conditions.

- **These can be harmful**, as someone may either be falsely reassured or be unnecessarily worried and perhaps have invasive tests or treatments which they do not need.

- There are only a specific range of genetic variants being screened for.

Communicating results

Communicating 'condition suspected':

- **~1% of babies (~1000 expected)**
- How: by phone from an NHS specialist, a few weeks after birth
- Regional result coordinators to liaise with Genomics England and the relevant NHS specialist team
- Aim to arrange follow-up for baby and family to be for confirmatory testing and appropriate further care
- Avenues of support through the specialist team, study-funded genetic counselling and patient organisations
- Regional result coordinator would contact NHS screening laboratory if the suspected condition can also be detected by standard newborn screening

Communicating 'no conditions suspected':

- **~99% of babies**
- How: by email/letter from Genomics England, a few months after birth
- Copy also sent to the baby's GP for their record
- Clarifies difference from NHS blood spot test and that this is not a 'clean bill of health'
- Includes information about what to expect in the future as a study participant

Communicating 'sample failure / no results returned'

- How: by email/letter from Genomics England
- May occur if samples are not taken or the test could not be completed

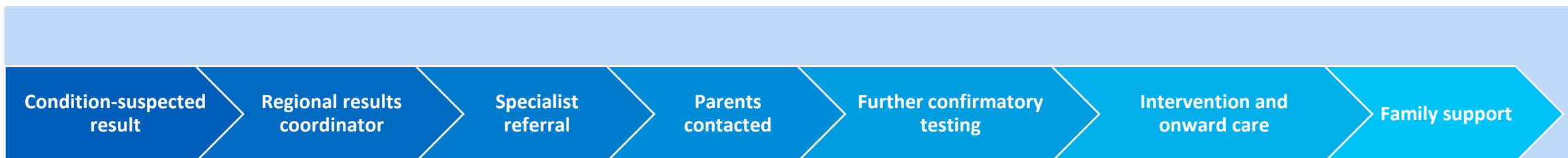
Condition Suspected Result Communication

The Regional Result Coordinator will contact the relevant specialist clinical team in the NHS and send them the following:

- Genomic Research Report
- Patient Contact Information
- Result Call Guideline
- Result follow-up email to parent
- Confirmatory testing pathway
- Condition information sheet
- Link to diagnostic outcomes form for evaluation








Onward support for Families



Avenues of support:

- NHS Specialist team (consultants, nurse specialists, psychologists, allied health professionals)
- Additional study-funded genetic counselling
- Patient organisations
- Genomics England Service Desk

Take Home Messages

-  The Generation Study does NOT screen for all genetic conditions – it only screens for a specific set of conditions, genes, and variants.
-  When no condition is suspected this is not a ‘clean bill of health’ for the baby.
-  It is important that babies participating in the Study continue to be offered the standard newborn blood spot test as well
-  When a condition suspected result is given, further testing to confirm the diagnosis is needed.
-  A baby’s genome data will be stored in the National Genomic Research Library (NGRL) for ongoing research