

Genomics England's Newborn Genomes Programme

Researching the potential of whole genome sequencing to help newborns with a rare genetic condition live their best possible lives.

Every day, nine babies in the UK are born with a rare genetic condition that could be treated, prevented, or even cured if only it had been diagnosed when those babies were newborns. The Newborn Genomes Programme is aiming to find out if this situation can be changed through a 3-part research study which will begin in a selection of NHS Trusts in 2023.

The 3 research questions we want to answer are:

- 01 Can we better diagnose, and therefore care for, children with rare diseases?
- 02 Can we give researchers opportunities to improve their understanding of rare disease, to develop new treatments, and diagnoses, and better understand how our genes affect our health?
- 03 Should we, and if so *how* should we, use a baby's genome throughout their lifetime as a resource they and their doctors can use if, for example, they become ill as they get older?

Our study in numbers...

Research study beginning **in 2023**

2022

2023

2024

2025



Aiming to find the **9** children born each day in the UK with a rare, treatable genetic disease – where early intervention is crucial



1000

Expecting 1000 **positive results** during the study

To find these children, we'll analyse

100,000+

newborn genomes for a specific set of childhood-onset, actionable conditions

6 linked design elements for the Programme – all underpinned by ethics:

1. Establishing principles to guide which conditions newborns should be screened for
2. Understanding the best way to recruit newborns and families to our research
3. Developing an appropriate consent process across the 3 parts of the Programme
4. Identifying the best way to a) take samples from newborns; b) sequence those samples
5. Establishing a process to return results to parents as quickly as possible
6. Ensuring care and treatment pathways / support are in place where positive results returned