NIHR | BioResource

Rare Diseases BioResource

Over 60 disease areas including immunology, neuroscience, haematology, rheumatology, cardiovascular disease, and more

24,536 participants*

100+ publications supported

***as of July 2026**

Data types available

- ThermoFisher UKBiobank v2.1 Genotype Array (1,600 participants)
- Whole Genome Sequencing (subset of ~8,500 participants, Illumina 150bp)
- Metabolome data (subset of 344 participants Metabolon)
- Clinical Report Form

Long Read Sequencing (LRS) Project

- Currently in production, target is 4,000 Rare Disease participants using Oxford Nanopore Technology with 30x coverage genomes
- Potential to provide additional detail when analysing genomes, particularly for complex structural variants, repeat regions and methylation
- Aim to sequence thousands of genomes over next year, in the following areas:
  - **Rare Diseases**
  - Eating Disorders (Mental Health)
  - Population cohort

RNA Omics Project

- ~1,000 participants across 17 rare diseases were recruited into the project - **full list**
- Short and long read WGS
- RNA Sequencing of 4 blood cell types: Neutrophils, CD4, Monocytes, Platelets
- Proteomics (mass spec) of the 4 cell types and plasma
- Blood count data

For more information, visit our website, or get in touch with us via email

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