

Rare Diseases BioResource

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



24,536 participants



100+ publications supported

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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