HGM2013-ICG-1478

Rare Genetic Variants in Health and Disease: 10,000 Genomes - The UK10K Project

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UK 10K
RARE GENETIC VARIANTS IN HEALTH AND DISEASE

4,000 whole genome samples sequenced
Average 6x depth

- 2,000 ALSPAC
- 2,000 TwinsUK

SNP, indel and larger structural variants associated with phenotypic traits to determine which were linked to disease and which were not

Candidate variants identified in subjects with extreme disease phenotypes

Low frequency variants (down to 0.1% allele frequency) imputed into studies of non-sequenced individuals with GWAS data to increase study power

>5,500 whole exome samples sequenced
Average 72x depth

- 3,000 Neuro disorders
  - 2,113 Schizophrenia
  - 887 Autism
- >1,500 Obesity
  - 1,000 SCOOP study
  - 430 Generation Scotland
  - 68 TwinsUK
- 1,000 Rare disorders
  - 125 Ciliopathies
  - 125 Coloboma
  - 125 Severe insulin resistance
  - 125 Congenital heart disease
  - 125 Familial hypercholesterolaemia
  - 125 Neuromuscular disease
  - 125 Learning disability (FIND study)
  - 125 Thyroid disorders

BAM and VCF files deposited in the European Genome-phenome Archive to create a managed-access data resource

The data generated by UK10K is a valuable resource for research into human genetics, and it is hoped the discovery of rare and low frequency disease-causing variants will lead to further insight into the diagnosis and treatment of disease