Medical Xpress

Millions of uncommon genetic variants found in Swedish study

August 24 2017

An extensive exercise to map genetic variation in Sweden has found 33 million genetic variants, 10 million of which were previously unknown. Large-scale DNA sequencing methods were used to analyse the whole genome of 1,000 individuals from different parts of the country. The study was led by researchers at Uppsala University, who have published their findings in the *European Journal of Human Genetics*.

"This resource will benefit many national research projects investigating the association between genetic variants and diseases," says Professor Ulf Gyllensten, Uppsala University and SciLifeLab, who has led the project.

The data will also be of immediate use in clinical diagnostics to determine whether a genetic variation in a patient is a cause of disease, or if it is also present among healthy individuals in the population.

"Our study shows the presence of millions of previously unidentified genetic variants in Sweden, the majority of which occur at low frequency in the population. It is crucial to identify these low frequency variants to facilitate the diagnosis of genetic diseases," says Adam Ameur, bioinformatician at Uppsala University and SciLifeLab, who has been responsible for the data analyses.

Several groups at SciLifeLab have been involved in the sequencing of the 1,000 DNA samples and in the development of data analysis methods. Very large amounts of data have been generated, over 100

terabytes for the entire project. Integrity and data security have been a high priority since the DNA sequences contain sensitive and personal information about the individuals.

"The resource is freely available, which enables researchers to quickly investigate genetic variant frequencies among the 1,000 Swedish individuals. However, a special request must be approved for access to data on individuals, and all processing must be performed within a custom-built computer system with extra high security," says Gyllensten.

More information: The variant frequency data is available from swefreq.nbis.se/

Adam Ameur et al. SweGen: a whole-genome data resource of genetic variability in a cross-section of the Swedish population, *European Journal of Human Genetics* (2017). DOI: 10.1038/ejhg.2017.130

Provided by Uppsala University