Researchers publish far-reaching genetic study of 1,000 UK people

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A unique population-based study of all the genes in 1,000 people born in the UK in 1958 is being made available online by researchers at the Institute of Cancer Research, London, to further understanding of the role gene variation plays in disease.The paper, which will be invaluable for clinical genomics and translational research, makes large-scale gene variation from exome sequence data from the UK general population, openly available for the first-time.

Titled 'The ICR1000 UK exome series: a resource of gene variation in an outbred population', the study has been led by Professor Nazneen Rahman and colleagues from the Division of Genetics & Epidemiology at the Institute of Cancer Research and has been published on open science platform F1000Research. It includes 1,000 people in the UK, who are part of the 1958 Birth Cohort - a study which follows the lives of 17,000 people born in England, Scotland and Wales in a single week in 1958.

The study by Professor Nazneen Rahman and colleagues provides a baseline of gene variation in a general population and, unlike most published exome series, the individual exome sequences are available for use by other researchers, on application to the 1958 Birth Cohort committee. Other clinical data and samples are also available regarding these individuals which increases the potential usefulness of the exome data to other researchers whilst ensuring anonymity for the individuals.

A high quality analysis pipeline was applied to the data including

accurate detection of indels, a very important class of gene variation that can cause disease. This sets the study apart from many other datasets which exclude all or some indels because they have proved tricky to identify, or state that indel calling is not of a high quality.

Professor Nazneen Rahman said: "We have analysed all the genes in 1,000 members of the general UK population. It has proved invaluable to us in both our disease gene discovery research and our work to translate gene testing into the clinic.

"We have found this data to be of tremendous value and it continues to be useful for us every day. Hundreds of research and clinical laboratories are doing similar work and we thought they would also find it valuable, so we decided to make our data available to them via F1000Research."

Michael Markie, Associate Publisher of F1000Research, said: "Vast quantities of genomic data currently reside in disparate silos making it difficult for scientists to access it and utilise it for their own research.

"F1000Research provides a simple and effective way to make genomic data publicly available to further disease research and we hope that more members of the genomic community will embrace it."

More information: f1000research.com/articles/4-883/v1

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