The Royal College of Pathologists of Australasia

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The largest genomic datasets provide researchers with an important resource for the future

Professor David Thomas, a speaker at the Royal College of Pathologists of Australasia's (RCPA) annual conference, 'Pathology Update 2018', will discuss preliminary findings from the Medical Genome Reference Bank (MGRB) program. The Australian study is already one of the largest genomic datasets ever produced in the country, and the largest dataset on the well elderly worldwide. The MGRB program is sequencing and analysing genomes from healthy, aged individuals to create a database that is depleted of damaging genetic variants. The study aims to provide a universal 'control' set for future disease-focused genomic research, whilst offering unique insights into the ageing process.

The results are providing valuable datasets for health and medical researchers in Australia and around the world and are providing an understanding of genetics in the healthy ageing population. The project, which was funded by NSW Health through the Office for Health and Medical Research, is being undertaken at the Kinghorn Centre for Clinical Genomics (Garvan Institute of Medical Research) in collaboration with two leading Australian research studies in older people: the ASPREE (ASPirin in Reducing Events in the Elderly) study (Monash University, Melbourne) and the 45 and Up study (Sax Institute, Sydney).

Whole genome sequencing of approximately 4000 Australians over the age of 70 years, who do not have any history of cancer, cardio-vascular disease or dementia, is being performed by Genome.One, a wholly owned subsidiary of Garvan, and home to the largest sequencing facility in the Southern Hemisphere.

Prof Thomas, Director of The Kinghorn Cancer Centre and Head of Garvan's Cancer Division and co-lead of MGRB says,

"We hope this dataset will help us understand what genetic variants cause disease, by filtering out the vast amount of genetic variation that we carry that is compatible with a long, healthy life. We will use the MGRB as a control for understanding genetic risk in young people with cancer. By contrasting the extremes, we get more power to understand the genetic basis for disease.

"For example, approximately 5% of the elderly cohort appears to have what we might currently think of as clinically significant genetic findings, but without any overt evidence of disease. This finding is important information in interpreting the same genetic variant when found in a young person, to help decide its clinical meaning.

"The study, which has already generated a vast amount of genomic information, assumes the relative depletion of those 'risk genes' (damaging genetic variants) in the population, which normally cause the disease to develop. In addition to studying rare genetic variations that cause clinical syndromes, the MGRB enables study of common variations which are widely present in the community, and which have disproportionate contribution to the risk of common diseases like dementia.

"Just as importantly, we can use the MGRB to understand the genetic basis of health in ageing: do our genes make a difference in how we age? We are beginning to have fascinating insights into what we can learn from the whole genome as we get older.

"We can also observe the ageing process reflected in these genomes, and we've made some very interesting observations about features which potentially reflect a person's biological age more effectively than chronological age."

As one of the largest whole genome studies of an elderly well population, it is hoped that this Australian study will enable clinicians and researchers to access results that will assist their own clinical interpretation and research.

"We are putting together a major research paper on the first results from the MGRB. A major goal is to create a resource for the clinical genetics and genomic research communities to help them with their own research. We have already received 27 applications from researchers, some as far afield as the U.S, to utilise the data set for their research, which is very promising," says Prof Thomas.

Prof Thomas is a speaker at the RCPA's Pathology Update 2018 conference, 'A Bridge to the Future', which takes place at the ICC Sydney from 2nd until 4th March 2018.

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About the Royal College of Pathologists of Australasia:

The RCPA is the leading organisation representing pathologists in Australasia. Its mission is to train and support pathologists and to improve the use of pathology testing to achieve better healthcare.

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