

Australian Scientists' global effort to improve diagnosis of human genetic disorders and genetic variations

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An Australian led global initiative to improve the diagnosis of genetic disorders and reduce errors in the reporting of genetic variations was published in the prestigious scientific journal Science on Friday 7 November.

Over 60 percent of people worldwide will be affected by a genetic change at some point in their lives that can result in a range of diseases such as cystic fibrosis, epilepsy and cancer.

In a world first, The Human Variome Project aims to collect information on every variation in every gene worldwide. However, out of the 20,000 human genes mapped, only 3,000 have any information available on their variations. "Currently there is no standardized way to capture this information and make it of use to clinicians," says Professor Richard Cotton, lead author of the paper, Convenor of the Human Variome Project and honorary researcher at the University of Melbourne. "There is a staggering error rate of 40 percent in some reporting of genetic variations. This means clinicians and specialists cannot solely rely on the research literature to inform the life and death decisions of diagnosis and prognosis of genetic disorders."

The project aims to produce standards for the storage, transmission and use of genetic variation information which for many will reduce the enormously time consuming task of seeking data to assist in providing patients with information.

The Science paper details the establishment of a range of pilot projects being organised around the world that will examine how to systematically collect genetic, clinical and biochemical information in either a country specific or gene specific manner.

"Once these pilot projects are complete, we will be able to roll out suitable systems around the globe and improve the health of billions of people," Professor Cotton said.

In order to raise funding for this enormous project, the HVP has created a charity called 'Adopt a Gene' whereby organizations and support groups can sponsor the curation of specific genes. "People in the past thought this task was too big, too impossible, but the news is it's happening," Professor Cotton said.

The Australian-led global project combines the talents of University of Melbourne researchers and colleagues within the Florey Neuroscience Institutes, the Department of Medicine at The Royal Melbourne Hospital and the Epilepsy Research Centre, as well as international colleagues from around the globe.

Neurologist Professor Sam Berkovic of the University of Melbourne and Austin Health, and co author on the paper explains the significance of gaining better access to genetic variations for diseases affecting the brain.

"There is a real challenge for neurologists to ascertain the genetic makeup of the many diseases affecting the brain such as epilepsy, Alzheimer's and degenerative disorders. Access to extremely varied genetic information is critical as patients develop these diseases over a period of time," Professor Berkovic said.

"This project opens the doors to earlier understanding and treatment of these complex conditions," he said.

The Human Variome Project has the support of WHO, UNESCO and OECD countries.

For more information visit www.humanvariomeproject.org

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The Human Variome Project is pursuing this mission by creating systems and standards for the storage, transmission and use of genetic variation information. Coordinated from Melbourne, Australia, the Human Variome Project's activities are distributed among 14 Key Action Areas, each of which is administered by an HVP Working Group. HVP Working Groups are responsible for producing recommendations for systems, standards and processes.

[You can find this press release here](#)