MedicalXpress

Australia leads world first global effort to improve diagnosis of genetic disorders

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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 has been published today in the prestigious scientific journal *Science*.

"There is a staggering error rate of up to 40 percent in some reporting of genetic variations," said Professor Richard Cotton, lead author of the paper, Convenor of the Human Variome Project and honorary researcher at the University of Melbourne.

"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."

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, we aim to collect information on every fault in every gene worldwide."

"Ultimately the project will provide the first global standardization of the reporting of genetic mutations and their effect on human health so clinicians can reliably diagnose, treat and inform patients," he 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.

The project has the support of World Health Organization, UNESCO and OECD countries.

The completion of the Human Genome Project in early 2000 empowered researchers with the genomic mapping of the human body. But out of the 20,000 human genes mapped, only 3,000 have any information available on their variations.

"In the next few years it is expected that the number of genes in which disease-causing variations are recognized will increase dramatically," Professor Cotton said.

"Currently there is no standardized way to capture this information and make it of use to clinicians."

The Human Variome Project will 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.

Countries already signed on to these pilots include Australia, China, Japan and Kuwait.

"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," he said.

One of the areas the pilot will be tested is colon cancer. World leader in colon cancer Professor Finlay Macrae in the University's Department of Medicine at the Royal Melbourne Hospital is a co- author on the paper.

As Secretary of InSiGHT, the International Society for Gastrointestinal Hereditary Tumours, Professor Macrae has been instrumental in establishing some of the first gene specific pilots for four of the genes predisposing to colon cancer.

"Colon cancer is the commonest internal cancer affecting both men and women in the western world, "Professor Macrae said.

"Genetic predispositions to colon cancer are now well recognized. Testing for mutations in some of these genes is critical to establishing risk for bowel cancer in some families. However, the information needed to interpret mutations is widely scattered and not readily available. "

"Providing systems to comprehensively and readily access this information is the aim of the worldwide InSiGHT/Human Variome Project pilot," he said.

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 make up 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."

Source: University of Melbourne