

Scientists achieve major genetics breakthrough

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University of Adelaide geneticist Dr Jozef Gecz and a team of Belgium and UK scientists have achieved a major breakthrough in discovering the causes of intellectual disability.

Dr Gecz, a senior researcher who is based at the Women's and Children's Hospital in Adelaide, has collaborated with an international research team to reveal that various mutations of a small part of the X chromosome lead to mental retardation.

The breakthrough is reported in the February issue of the American Journal of Human Genetics and comes after an intensive collaboration with scientists from the Katholieke Universiteit Leuven in Belgium, the Wellcome Trust Sanger Institute in Cambridge, England and the Genetics of Learning Disability (GOLD) Service in NSW.

Dr Gecz says the duplicated genes – dubbed HSD17B10 and HUWE1 – produce excess protein, which appears to be the trigger leading to intellectual disability.

The researchers used specialised molecular technology to study the X chromosomes of more than 500 families diagnosed with various forms of X chromosome-linked mental retardation.

In six of the families they discovered that a certain part of the X chromosome had been duplicated. Four of these families were from Australia. Moreover, in one of these genes, HUWE1, they identified a

further three mutations in another three families, one of them from Australia.

“HUWE1 is a protein, which regulates TP53, an important tumour suppressor gene. One of TP53’s functions is to regulate the renewal of neuronal cells in the brain. That is where we see the connection with HUWE1 and intellectual disability,” Dr Gecz says.

“Through this research we hope to uncover the important role that these genes and their proteins play in the normal brain development and thus learning and memory.”

The collaboration between the University of Adelaide and the Belgium and UK researchers started back in 2001 at a scientific meeting in Italy and gathered pace at the World Congress of Human Genetics in Brisbane in 2006.

Dr Gecz’s research has been funded by the Australian National Health and Medical Research Council Program grant, which is administered through the University of Adelaide.

“Our next step is to see how frequent these mutations are among the isolated cases. Also, we will try to understand the molecular pathology of the associated disease and investigate the routes for possible future interventions and treatments.”

Intellectual disability affects 1-3% of the world’s population, with 30% more men affected than women.

Source: University of Adelaide

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