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Novel gene may provide clues on how we resist disease

As part of an international collaboration involving scientists from Australia, the United Kingdom and the United States, Assoc Prof Jozef Gecz from the Women's and Children's Hospital, Adelaide has discovered a novel gene, which when mutated, causes intellectual disability and may be a contributing factor in Autism Spectrum Disorder. The gene is also part of a widespread disease-preventing control system in all human cells.

This international Wellcome Trust-funded project involved 600 families world-wide of which 150 came from Australia. To date, in excess of \$9 million has been channelled into the project.

Neurogenetics Laboratory Head, Assoc Professor Jozef Gecz believes this gene is particularly important as it tells us a great deal about the basic biology of cells.

"This gene is part of an important multiprotein quality control system found in nearly every cell of our body which was discovered a number of years ago," Prof Gecz said.

"These genes are intimately involved in policing our cells to keep down the background noise, to make sure that our genetic material turns over at the correct level and no incorrect proteins are produced. Sometimes making incorrect proteins is more harmful than not making any proteins at all.

"We were very surprised to find that this gene was involved in intellectual disability, and that the four affected families we studied all had quite different clinical presentations.

"We would have expected such an important gene, when mutated, to have much more of a deleterious effect on the body. This tells us that human cells are so robust that even when a very important gene is mutated there are alternative routes that can be used to keep checks on the cells. But, in this case, as these are not quite as good as the original gene, then intellectual disability occurs.

"This has major implications for our understanding of how our bodies resist genetic diseases, and may be very important in the development of therapy to treat intellectual disability."

Approximately 3,000 babies are born with intellectual disability in Australia annually. Of these, each year about two babies will be born with a mutation in this gene. One of these children will also have Autism Spectrum Disorder.

"Because each of these families studied, as part of our large international project, has on average over six or more members, the 150 Australian families studied really represent over 1000 individuals who directly benefit from this research," Prof Gecz said.

"Having the knowledge as to whether you do or don't carry the gene defect, allows families to make important decisions about having children. It helps families feel safe to have children. They also feel less isolated as they become aware of others with the same problems."

The research is an international collaboration between organisations which include the Wellcome Trust Sanger Institute and the Cambridge Institute of Medical Research in the UK, Greenwood Genetic Centre in the USA, Hunter Genetics in New South Wales and the University of Adelaide and Women's and Children's Hospital in Adelaide, South Australia.

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