Gene involved brain development and intellectual disability identified

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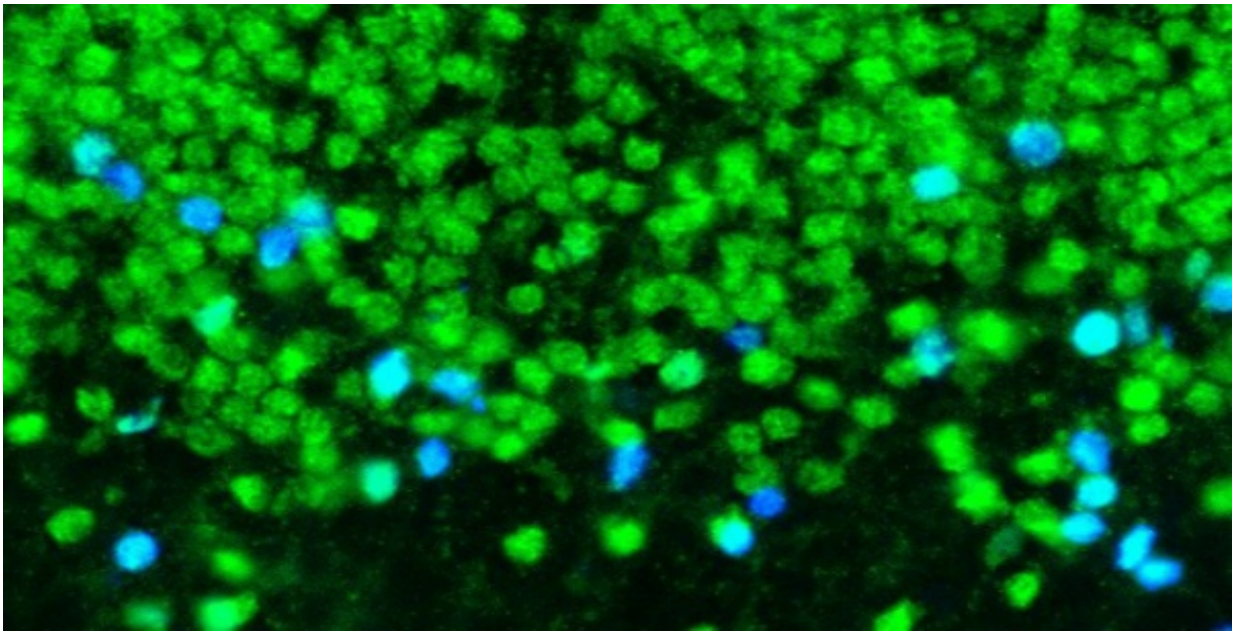

Image of fluorescence staining to identify EURL in nerve cells of the brain (cyan). Credit: Julian Heng

INVESTIGATING the genetics of Down Syndrome has led WA researchers to find a new gene responsible for brain development and intellectual disability.

Down Syndrome is a genetic disorder caused by the presence of an extra copy of all or parts of chromosome 21. People with Down Syndrome

have some characteristic physical features, some health and development challenges and some level of intellectual disability.

Researchers were puzzled, however, by rare cases of children with abnormalities in chromosome 21 who were intellectually disabled, but did not display the typical clinical features normally present in Down Syndrome.

"This led us to predict that genetic factors specifically for intellectual disability might be present on chromosome 21," said Associate Professor Julian Heng, Head of the Brain Growth and Disease Laboratory at the Harry Perkins Institute of Medical Research.

The research team focused on a gene known as 'EURL' or C21ORF91—originally detected as a gene in Early Undifferentiated Retina and Lens.

"Prior to our discovery, there were only inferences made as to the possible function for EURL in cells," A/Prof Heng says.

Their work found EURL to control the formation of neural circuits in the brain, inferring that this gene is likely responsible for causing intellectual disability in Down Syndrome.

A/Prof Heng believes that the functions for EURL within brain cells can be manipulated by blocking its activity as a signalling molecule or by preventing the gene from being switched on.

"In both cases, such manipulations might be useful to consider when there are extra copies of the EURL gene in a child, such that we might be able to balance the quantity of functional EURL which, in turn, could lead to improvements in the brain's neural circuitry."

A/Prof Heng says the next step in the research is to develop a cell-based assay to determine how EURL controls fetal brain growth.

"Such an assay is critical to the evaluation of therapeutics which can correct EURL gene dosage imbalances and restore normal function in affected neural tissues," he says.

He adds, "the promise of Genomic Medicine is only limited by our imagination and, clearly, the community must be involved in any conversation as to the responsible use of such technologies which could improve the lives of humans."

This means future genomic medicine to correct the imbalance of gene products in brain cells, could possibly improve mental health and mean a better quality of life for those with certain forms of intellectual disability .

More information: Shan Shan Li et al. The HSA21 gene EURL/C21ORF91 controls neurogenesis within the cerebral cortex and is implicated in the pathogenesis of Down Syndrome, *Scientific Reports* (2016). DOI: 10.1038/srep29514

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