

# Gene discovery leads to new epilepsy treatment

6 July 2015

The discovery of the gene responsible for a form of epilepsy in girls has led to a new treatment for the disorder.

An international team led by Professor Jozef Gecz, a genetics expert from the University of Adelaide in South Australia, first identified the responsible gene and mutations in this female-only epileptic syndrome in 2008.

Now, in breakthrough research that will help thousands of young girls who suffer from the rare yet debilitating condition, Professor Gecz has found a treatment for this disorder and Marinus Pharmaceuticals is recruiting affected girls for a world-first clinical trial.

Professor Gecz, who works at the University of Adelaide's Robinson Research Institute, says this condition is unique as it presents almost exclusively in girls while boys with mutations in the gene are not affected.

"We discovered that this condition is caused by an inherited mutation of the protocadherin 19 (PCDH19) gene, located on the X-chromosome," says Professor Gecz, Head of Neurogenetics at the University of Adelaide. "And interestingly, both males and females can be born with this mutation but only females suffer from the symptoms of the condition."

Girls are affected because they have two X-chromosomes, one healthy and one with the PCDH19 mutation, which would usually protect them from a X-chromosome borne disease, but in this case it drives the disorder.

Professor Gecz has worked with the families of girls with this female-only epilepsy from all over the world and says while the condition affects everyone differently, in most cases it is highly incapacitating.

According to Professor Gecz, this form of epilepsy affects up to 30,000 girls in the United States and approximately 1000 in Australia.

"Girls born with this [gene mutation](#) appear perfectly normal in the first few months of their lives but when they reach about eight months of age, they start suffering from debilitating and frequent seizures. The girls also commonly suffer from [intellectual disability](#) and autism – it's a truly terrible disease which impacts the whole family.

"Through our current research we found that sufferers are deficient in a hormone called allopregnanolone.

"We know that hormones play a critical role in this condition because the seizures often stop once the [girls](#) reach puberty – however the autism and intellectual disability remain. We expect that the longer we can delay the onset of seizures, the less the sufferer might be affected by the autism and intellectual disability."

His findings are so promising that Marinus Pharmaceuticals has commenced a clinical trial to test the effect of a synthetic form of the neurosteroid allopregnanolone, called ganaxolone.

The research is published in Oxford Journals, *Human Molecular Genetics*.

**More information:** "Mutations of protocadherin 19 in female epilepsy (PCDH19-FE) lead to allopregnanolone deficiency." *Hum. Mol. Genet.* first published online June 29, 2015 [DOI: 10.1093/hmg/ddv245](#)

Provided by The Lead Australia

APA citation: Gene discovery leads to new epilepsy treatment (2015, July 6) retrieved 28 April 2021 from <https://medicalxpress.com/news/2015-07-gene-discovery-epilepsy-treatment.html>

*This document is subject to copyright. Apart from any fair dealing for the purpose of private study or research, no part may be reproduced without the written permission. The content is provided for information purposes only.*