Finding a genetic cause for severe childhood epilepsies

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(Medical Xpress)—A large scientific study has discovered new genes causing severe seizure disorders that begin in babies and early childhood. The finding will lead to new tests to diagnose these conditions and promises to lead to improved outcomes.

Epileptic encephalopathies are severe seizure disorders occurring in infants and children. The seizures are accompanied by slow development and intellectual problems.

Paediatric neurologist and researcher Professor Ingrid Scheffer from the University of Melbourne and the Florey Institute of Neuroscience and Mental Health, and the clinical leader of the study said "these children have devastating disorders. Finding the cause is the first step in developing targeted treatments."

"Overall, our findings have important implications for making a diagnosis in patients, optimizing therapy and genetic counseling for families," she said.

The study published in *Nature Genetics* today revealed two new genes associated with these severe epilepsies.

In the study, researchers analysed the genes of 500 children who have epileptic encephalopathies.

Using recent advances in genetic testing, next generation sequencing of a

gene panel was performed. Researchers analysed 65 genes of which 19 had previously been associated with epileptic encephalopathies and 46 were hypothesized to potentially cause these devastating disorders.

Results revealed that mutations that cause epileptic encephalopathy were found in 52 out of the 500 patients (more than 10% of the study population).

Mutations were found in 15 of the 65 genes, including two new genes, CHD2 and SYNGAP1, which have not previously been found to cause epileptic encephalopathies.

"This is a very exciting breakthrough which could lead to dramatic benefits in the lives of the children who suffer this condition," Professor Scheffer said.

These genes will now become a diagnostic test for children with these severe epilepsies and enable genetic counseling in their families.

Collaborators on the study included geneticists from the University of Washington, US and paediatric neurologists from around Australia, New Zealand, Denmark and Israel. The study was funded by the National Health and Medical Research Council, Australia and the National Institutes of Health, US.

More information: Targeted resequencing in epileptic encephalopathies identifies de novo mutations in CHD2 and SYNGAP1, DOI: 10.1038/ng.2646

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