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Scientists discover childhood epilepsy genes

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Singapore: National Institute of Health (NIH)-funded genetic study of childhood epilepsies has linked two new genes to severe forms of disease and provides a novel strategy for identifying therapy targets. This study used a cutting-edge genetic technique, called exome sequencing, to search for new mutations that are not inherited. The results suggest this may be a highly effective way to find and confirm many disease-causing gene mutations.

"It appears that the time for using this approach to understand complex neurological disorders has arrived," said Dr David Goldstein, director, Center for Human Genome Variation, Duke University Medical Center, and a leader of the study. "This moderately-sized study identified an unusually large number of disease-causing mutations and provides a wealth of new information for the epilepsy research community to explore."

Severe childhood epilepsy gene mutations researchers from the Epi4K project used advanced genetic techniques to search for mutations that are likely to be involved with severe childhood epilespies.

The study is part of a worldwide, $25 million project called Epilepsy 4000 (Epi4K External Web Site Policy). Epi4K's mission is to use the latest genetic techniques to sequence and analyze DNA from 4000 epilepsy patients and their relatives. To do this, the researchers and NIH staff involved organized a team of international research institutions devoted to the mission, called the Epilepsy Centers without Walls. This approach facilitates the sharing and analysis of DNA sequences and patient information among the dozens of institutions participating in the project.

"These promising results highlight the strength of supporting large international research teams devoted to studying the genetics behind highly complex neurological disorders," said Dr Story Landis, director, NIH's National Institute of Neurological Disorders and Stroke (NINDS). The project is also led by Dr Daniel Lowenstein, vice chair, Department of Neurology, University of California, San Francisco (UCSF) and Dr Sam Berkovic, director of the Epilepsy Research Center at the University of Melbourne, Australia, on behalf of an international team of investigators.

Epilepsy is a group of neurological disorders caused by abnormal firing of nerve cells in the brain which often produces debilitating seizures and a range of other symptoms. Although some studies have found genes associated with rare inherited forms of epilepsy, finding genes associated with the majority of epilepsies has been difficult.

"Unlike some diseases many of the genetic mutations associated with severe childhood epilepsies appear to be new mutations that are not inherited," said Dr Randall Stewart, a program director at NINDS. "This Epi4K-EPGP project was established to find such mutations."