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New study yields important clues to the genetics of epilepsy

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An international team of researchers has discovered a significant genetic component of Idiopathic Generalized Epilepsy (IGE), the most common form of epilepsy. Epilepsy is a neurological disorder characterized by sudden, uncontrolled electrical discharges in the brain expressed as a seizure. The new research, published in this week's issue of *EMBO Reports*, implicates a mutation in the gene for a protein, known as cotransporter KCC2.

KCC2 maintains the correct levels of chloride ions in neurons, playing a major part in regulating excitation and inhibition of neurons. The results indicate that a genetic mutation of KCC2 might be a risk factor for developing IGE.

"We found a clear statistical association between two variants of KCC2 and severe IGE in a large French-Canadian patient sample," said Dr. Guy Rouleau, Director of the Montreal Neurological Institute and Hospital-The Neuro, at McGill University and the McGill University Health Centre, and senior author of the study. "Our data not only corroborate recent findings by other groups but vastly extend them from genetic, physiological and biochemical standpoints." The first authors on the paper are Dr. Kristopher Kahle, chief neurosurgery resident at Massachusetts General Hospital and post-doctoral fellow at Harvard University, and Dr. Nancy Merner, a former post-doctoral fellow in Dr. Rouleau's laboratory and now a professor at Auburn University.

The study examined 380 French Canadians with IGE living in Montreal

and Quebec City. Results were compared to data from a control group of more than 1,200 people. "KCC2 is a hot topic in neuroscience given its important role in neuronal signaling and in its potential role in neurological diseases such as epilepsy, neuropathic pain, and other diseases," said Dr. Rouleau.

Each day in Canada, an average of 42 people learn that they have epilepsy. In 50 - 60% of cases, the cause of epilepsy is unknown. The major form of treatment is long-term drug therapy. Drugs are not a cure and can have numerous, sometimes severe, side effects. Brain surgery is recommended only when medication fails and when the seizures are confined to one area of the brain where brain tissue can be safely removed without damaging personality or function.

The Neuro has been at the forefront of epilepsy treatment and research for over half a century. The "Montreal Procedure" developed at The Neuro revolutionized the surgical treatment for epilepsy. The use of EEG and MRI for the study and treatment of epilepsy was also pioneered at The Neuro. The Neuro's Epilepsy Program has a multi-disciplinary team of epileptologists, neurosurgeons, nurses, neuropsychologists, neuropsychiatrists, social workers, EEG technologists, nurse clinicians and case managers. The Program provides both in-patient and out-patient evaluation and treatments for more than 3000 patients a year and the EEG Clinic conducts almost 4,000 electroencephalograms each year.

More information: Paper: embor.embopress.org/content/ea … 06/10/embr.201438840

Provided by McGill University

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