Discovery of genetic defect that triggers epilepsy

April 17 2013

Researchers at the University Department of Neurology at the MedUni Vienna have identified a gene behind an epilepsy syndrome, which could also play an important role in other idiopathic (genetically caused) epilepsies. With the so-called "next generation sequencing", with which genetic changes can be identified within a few days, it was ascertained that the CNTN2 gene is defective in this type of epilepsy.

This was investigated by a team led by Elisabeth Stögmann in collaboration with Cairo's Ain Shams University and the Helmholtz Centre Munich with reference to a particular Egyptian family, in which

five sick children have resulted from the marriage of one healthy cousin to his, likewise healthy, second cousin. The children affected suffer from a specific epilepsy syndrome, in which different types of epileptic attacks occur. This constellation has the "advantage", according to Stögmann, that both alleles of the gene, which is how one designates different forms of the gene, demonstrate this defect: "As a result the defect becomes symptomatic and identifiable.

"20,000 to 25,000 genes, including all the "protein coding" ones, were sequenced for this. When this was done a mutation was found in the CNTN2 gene. CNTN2 undertakes an important function in the anchoring of potassium channels to the synapses. The mutation makes it no longer possible to generate this protein and, as a consequence, the potassium channels no longer remain affixed to the synapses. The researchers suspect that the epilepsy in this family is triggered by the altered function of the potassium channels.

This discovery, which has now been published in the top journal *Brain*, is providing the stimulus for further research to investigate this particular gene in other epilepsy patients as well. Approximately one percent of the population suffers from active epilepsy in which regular epileptic fits occur. The danger of suffering from an epileptic fit once in your life lies at approximately four to five percent. Genetic factors play a major part in the occurrence of epilepsies.

More information: Stogmann, E. et al. Autosomal recessive cortical myoclonic tremor and epilepsy: association with a mutation in the potassium channel associated gene CNTN2, *Brain*, 2013 Apr; 136(Pt 4):1155-60. doi: 10.1093/brain/awt068. Epub 2013 Mar 21.

Provided by Medical University of Vienna