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New Epilepsy Gene Located in Dogs

ScienceDaily (Mar. 23, 2012) — A new epilepsy gene for idiopathic epilepsy in Belgian Shepherds has been found in the canine chromosome 37. The research of Professor Hannes Lohi and his group conducted at the University of Helsinki and the Folkhälsan Research Center opens new avenues for the understanding of the genetic background of the most common canine epilepsies. The research also has an impact on the understanding of common epilepsies in humans.

Belgian shepherd dogs. (Credit: © Cerae / Fotolia)

The research is published in the scientific journal *PLoS ONE* on March 23, 2012.

Epilepsy affects about 1-5% of the human population at some stage of live, and it includes a host of syndromes the age of onset, causes and prognosis of which vary significantly. Based on their basic mechanisms epilepsy syndromes are divided into genetic (idiopathic) epilepsies, structural / metabolic (symptomatic) epilepsies and epilepsies of unknown cause. Symptomatic causes refer to discernible external or structural change, whereas with idiopathic epilepsy there is a strong genetic background. A common denominator between the different syndromes are reoccurring epileptic seizures, which are divided according to an international classification into two main groups -- focal and generalized seizures -- based on clinical symptoms and research findings. About two thirds of the seizures in adults are focal in nature and one third generalized. In children and teenagers the occurrence of generalized forms of epilepsy is greater (ca. 50%).

Identification of the epilepsy gene on process

Genetic factors are estimated to play a role in the development of epilepsy in as many as 40% of epilepsy patients. Several genes affecting the development of symptomatic epilepsies have already been identified, but the genetic background of multifactorial idiopathic epilepsies often remain unknown. Both focal and generalized idiopathic epilepsies occur in Belgian Shepherds. The research group of Professor Hannes Lohi, working in collaboration with Danish, Swedish and American researchers in an EU-funded project, has made a major breakthrough by identifying a chromosome region associated with the most common form of epilepsy in dogs. By comparing the genome of dogs with epilepsy and healthy control dogs a gene region in chromosome 37 was discovered, which if homozygous, increases the risk of epilepsy seven-fold. In addition the research findings indicate that other, still unknown, genetic risk factors may be present in the breed.

The identified region has excellent neurological candidate genes for epilepsy and ongoing follow-up research is aimed to identify the specific gene causing epilepsy. Epilepsy genes have not previously been identified in this chromosome region, so the discovery will reveal an entirely new epilepsy gene in dogs and possibly also in humans. The type of epilepsy occurring in Belgian Shepherds is extremely common in also other breeds and thus the discovery may have an impact on the understanding of the epilepsies in different dog breeds.

"There are only few genes in the identified region and I believe that the ongoing analyses will help us to discover the specific epilepsy gene," says Professor Hannes Lohi who led the research. "This would give us a better understanding of the disease mechanisms and provide us with new diagnostic tools for the disease."

The Research group of Hannes Lohi has begun an extensive gene-sequencing project in which the entire identified chromosome region will be 'read through' with a next-generation sequencing method. By identifying the specific gene mutation an individual's epilepsy risk could be assessed, although the gene mutation may also be common in dogs that never become symptomatic of epilepsy.

Epilepsy is common among Belgian Shepherds

"The identified genomic region is likely to be the strongest single risk factor for epilepsy in Belgian Shepherds, and we are studying an interesting gene variant causing an amino acid change in the protein level. However, this homozygous amino acid change is also present in one fifth of healthy Belgian Shepherds. The research continues in the breed and aims to identify the specific mutation for genetic testing in this loci and possible in other chromosomes. The need for the gene test is urgent since as much as 20% of the dogs in this breed are estimated to have epilepsy," comments the first author of the article Eija Seppälä, PhD.

The age of onset of idiopathic epilepsy in Belgian Shepherds is on average 3 years, although the range varies greatly. A seizure often begins as a focal seizure, and the owner may observe abnormal movement, usually on only one side of the dog's body. At this point the dog often seeks out the owner, drools or vomits. With most Belgian Shepherds the seizure becomes generalized and is accompanied with loss of consciousness and irregular cramping in the limbs. The dog may also urinate or defecate involuntarily.

Earlier gene discoveries of the research group

Epilepsy is the most common disorder of the nervous system in dogs and different types of genetic epilepsies occur in dozens of dog breeds. The research group has previously identified the first epilepsy gene for symptomatic epilepsy, EPM2B, in Miniature Wirehaired Dachshunds, as well as more recently a gene, LGI2, associated with transient idiopathic epilepsy in Lagotto Romagnolos. The group has also participated in the discovery of a gene for symptomatic epilepsy in Tibetan Terriers. Lohi and his research group have built a canine DNA bank in Finland, which currently holds almost 40 000 samples from more than 250 different breeds. The DNA bank has played a major role in the execution of among others the current research to be published in March, 2012.

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Journal Reference:

1. Seppälä EH, Koskinen LLE, Gulløv CH, Jokinen P, Karlskov-Mortensen P, Bergamasco L, Baranowska Körberg I, Cizinauskas S, Oberbauer AM, Berendt M, Fredholm M, Lohi H. **Identification of a novel idiopathic epilepsy locus in Belgian Shepherds**. *PloS ONE*, 2012 DOI: 10.1371/journal.pone.0033549

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Helsingin yliopisto (University of Helsinki) (2012, March 23). New epilepsy gene located in dogs. *ScienceDaily*. Retrieved March 24, 2012, from http://www.sciencedaily.com /releases/2012/03/120323205337.htm

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