

A gene discovery in truffle dogs sheds new light on the mechanisms of childhood epilepsy

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A new epilepsy gene, LGI2, has been found in the Lagotto Romagnolo dogs, known from their gift for truffle hunting. The gene discovery made by Professor Hannes Lohi and his research group at the University of Helsinki and the Folkhälsan Research Center offers a new candidate gene for human benign childhood epilepsies characterized by seizure remission. The research will be published in the prestigious scientific journal *PLoS Genetics*.

Epilepsy is the most common neurological disease in children. It occurs in 0.5 percent of all 2-10 year-old children – during the ages when the development of the nerves in the brain is at its strongest. Childhood epilepsies are characterized by remission: the seizures set in and last for a while before they disappear completely. The mechanisms related to the remission have remained unknown. The new [gene discovery](#) made by the research group of Professor Lohi and colleagues in the Lagotto Romagnolo breed gives us new perspectives into the development of a child's brain and the remission mechanisms in childhood epilepsies. In addition, the identified gene has enabled the development of a DNA test for the Lagotto Romagnolo breed.

- This gene discovery is significant for both dogs and humans. Every third Lagotto Romagnolo carries the gene mutation in its genome and we have now developed a gene test to be used by breeders to eliminate the disease from the breed. Furthermore, the gene has not previously been

linked to human epilepsies, which makes it a new candidate gene for especially childhood epilepsies, explains Hannes Lohi.

An epileptic seizure is caused by an electronic disturbance in brain function. Epilepsies form a heterogeneous group of syndromes of the nervous system in which the causes, the age of onset and the treatment vary significantly. [Epilepsy](#) is most common in the ends of the age spectrum – childhood and old age.

- With this study we gain crucial insight into the pathways and mechanisms that control the development of a child's brain, optimizing its structure for electrical stability and seizure-freedom in the rest of adult life. This study will open vast avenues of research in uncovering the molecular bases of the transformation of the brain from its immature state in infancy to its maximal abilities in adolescence and early adulthood, says Dr. Berge Minassian, senior co-author of the study and a senior scientist and pediatric epileptologist at The Hospital for Sick Children in Toronto, Canada.

Epilepsy is the most common disease of the nervous system in dogs, and different types of hereditary epilepsy exist in many breeds. Lohi has previously identified the first canine epilepsy gene, EPM2B, in the Miniature Dachshund. The newly identified mutation in the LGI2 gene is the first idiopathic epilepsy gene in dogs.

Previous clinical studies have demonstrated the focal remitting epilepsy in Lagotto Romagnolo puppies. Seizures causing tremor, trembling, shaking and wheezing set in at around four weeks of age and last for one to two months before a complete cease. The seizure frequency varies significantly even within the same litter and severity can vary from very mild to attacks of unconsciousness. The mildest seizures may even go unnoticed. Between seizures some dogs may experience ataxic episodes with difficulties in movement coordination and motor function.

Lagotto's epilepsy resembles human benign childhood epilepsies with remission.

- We tested the mutation in about 40 different breeds and in dogs with a very early age of onset of epilepsy, but it was present only in Lagottos. On the other hand, the study revealed another form of epilepsy in the breed, unconnected with this mutation and with an age of onset in adulthood. In addition, the breed has a progressive juvenile ataxia (lack of motor coordination) with similar onset and symptoms to juvenile epilepsy except that it does not remit - ataxic puppies have to be euthanized usually by the first year of life. More samples are needed for both adult-onset epilepsy and ataxia to enable us to investigate their genetics further, says a primary author of the study, Eija Seppälä, PhD.

Together with his research group Lohi has built a large canine DNA bank in Finland with over 35 000 samples from 250 breeds. The DNA bank has played an important role in the present and ongoing studies.

- We also study the epilepsies in other breeds, and several new epilepsy loci have been discovered recently. I believe that there will be more similar success stories such as the case of the truffle dogs in future. Canine epilepsies are natural, spontaneous and resemble human epilepsies, offering us a great opportunity to advance the epilepsy research for the benefit of both humans and dogs, Lohi asserts.

Lohi's research group operates in two campuses at the Faculties of Veterinary Medicine and Medicine at the University of Helsinki and also at the Folkhälsan Research Center.

More information: Jokinen T, Metsahonkala L, Bergamasco L-A, Viitmaa R, Syrja P, Lohi H, Snellman M, Jeserevics J, and Cizinauskas S. Benign Familial Juvenile Epilepsy in Lagotto Romagnolo Dogs, *J Vet Int Med*, 21(3):464-71, 2007.

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