Researchers identify a gene that causes canine hereditary deafness in puppies

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Finnish researchers have been the first to determine the cause for the nonsyndromic early-onset hereditary canine hearing loss in Rottweilers. The gene defect was identified in a gene relevant to the sense of hearing.

The study can also promote the understanding of mechanisms of hearing loss in human.

Hearing loss is the most common sensory impairment and a complex problem in humans, with varying causes, severity and age of onset. Deafness and hearing loss are fairly common also in dogs, but gene variants underlying the hereditary form of the disorder are so far poorly known.

Researchers from the University of Helsinki and the Folkhalsan Research Center focused on a rare type of hearing loss observed in Rottweilers. It begins early in puppyhood and progresses to deafness at the age of few months. A similar type of hearing loss was also seen in a small number of mixed-breed dogs, of which the majority had Rottweiler ancestry.

"We identified the variant in the LOXHD1 gene, which plays a key role in the function of the cilia of the cochlear sensory cells. While the exact mechanism of deafness is not known, variants of the same gene cause hereditary hearing loss in humans and mice as well," says Docent Marjo Hytonen from the University of Helsinki and the Folkhalsan Research Center.

Hearing impairment caused by the LOXHD1 gene defect is a recessively inherited trait, which means that to develop the disorder, the dog must have two copies of the defective gene, one from the father and one from the dam.

"Through our collaboration partner, we had the chance to investigate the prevalence and breed specificity of the gene variant in a unique global dataset of some 800,000 dogs. No surveys of similar scope have previously been published," says Professor Hannes Lohi from the University of Helsinki and the Folkhalsan Research Center.

New individual dogs that had inherited the gene defect and were also found to be deaf were identified in the screening.

"This enhances the significance of our finding. Thanks to our gene discovery, dogs used for breeding can now be tested for the defect. This makes it possible to avoid combinations that could result in puppies who will lose their hearing."

The recent study is part of a research program led by Professor Lohi and investigating the genetic background of hereditary diseases. Currently ongoing are several projects whose goals include the determination of genetic causes for hearing loss.

According to Marjo Hytonen, the preliminary results are promising.

"We have observed that both previously unknown hereditary congenital hearing loss and adult-onset hearing loss occur in several dog breeds. In addition to dogs, the preliminary findings open new avenues for investigating human hereditary hearing defects."

More information: Marjo K. Hytönen et al, Missense variant in LOXHD1 is associated with canine nonsyndromic hearing loss, *Human Genetics* (2021). DOI: 10.1007/s00439-021-02286-z

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