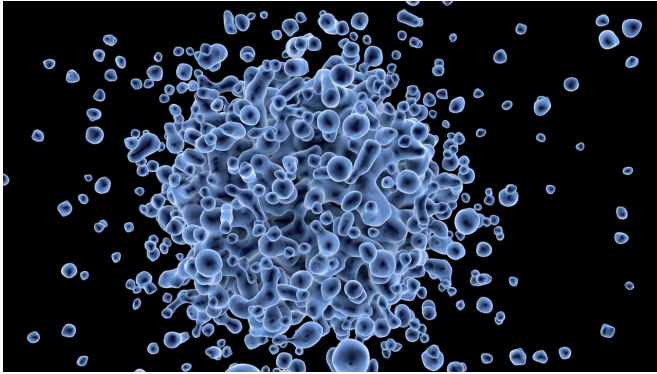


Gene associated with severe canine lung disease linked to human pulmonary issues

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In 2007, Finnish Airedale terrier breeders sent puppies that had died only a few days after being born for pathological examinations to the Finnish Food Safety Authority (Evira). Some litters had lost several puppies. A research project initiated at Evira under the direction of Professor Marjukka Anttila uncovered a problem in the puppies' lungs.

"There are vacuoles, or specialized membrane-bound organelles, in the epithelial, or superficial cells of the pulmonary alveoli, which are responsible for gas exchange. They are tasked with producing a substance that reduces [surface tension](#) on the surface of these cells, essential to breathing. The vacuoles in the lungs of the dead puppies hadn't matured normally, nor were they able to produce this surfactant," explains doctoral candidate Kati Dillard, LVM, from the Faculty of Veterinary Medicine, University of Helsinki, and the Finnish Food Authority.

A look at the dead puppies' pedigrees soon revealed that the cause was an inherited disease. The gene defect underlying the [disease](#) was located in the LAMP3 gene in Professor Hannes Lohi's laboratory. Gene discovery fits well with the

clinical picture. The gene produces a protein that functions precisely in the membranes of the vacuoles forming the surfactant.

Nearly 7,000 individual dogs representing nearly 300 different breeds were screened for the [gene defect](#), which was only found in the Airedale terrier breed. One-fifth of Airedale terriers were found to be carriers of the defect. In future, carriers can be identified with the help of a gene test, thus making it possible to avoid breeding combinations that give birth to sick dogs.

Potential link with certain breathing problems in human newborns

Neonatal respiratory diseases are typical not only to dogs. Similar defects in the formation of surfactant produced by the epithelial cells of pulmonary alveoli also occur in newborn babies.

"As the LAMP3 gene has not previously been associated with diseases, in the future its role in the breathing difficulties afflicting newborn babies should be investigated. This is another example of canine research that identifies a candidate gene for a [human disease](#)," says Professor Hannes Lohi, who headed the study at the Faculty of Veterinary Medicine and the Faculty of Medicine, University of Helsinki, and the Folkhälsan Research Center.

More information: Kati J. Dillard et al. Recessive missense LAMP3 variant associated with defect in lamellar body biogenesis and fatal neonatal interstitial lung disease in dogs, *PLOS Genetics* (2020). [DOI: 10.1371/journal.pgen.1008651](https://doi.org/10.1371/journal.pgen.1008651)

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