The gene for day blindness in the dachshund has been found

May 27 2009

Wire-haired dachshund suffering from the disease has a bad eyesight in daylignt and becomes gradually blind. Photo: Frode Lingaas

A PhD project by Anne Caroline Wiik has discovered the genetic cause of day blindness or "cone-rod dystrophy" in the wire-haired dachshund. The disease was discovered in two litter mates in 1999 and has since been studied in both clinical and genetic trials in offspring of these.

In her thesis, Anne Caroline Wiik concentrated on finding the genetic mutation that causes this disease. Day blindness is a recessive, heritable disease in which both parents need to be carriers in order for the disease to develop.

Inherited photoreceptor diseases, or diseases in the sensory cells of the retina (rods and cones), occur naturally in both animals and man. They

comprise the most common form of inherited retinal disease in people, with an occurrence of approximately 1 in 4,000.

The project began with a candidate gene study, in which genes known to cause similar diseases in people were investigated to see if they had any connection to the canine disease. Ten genes were studied, without a connection being found between the genes and eye disease.

A new method for finding genetic mutations

Wiik was then among the first researchers to be given an opportunity to try a new method of finding genes causing the disease in dogs. By comparing all genes in a sample of sick and healthy dogs, an area that might contain the diseased gene was isolated. Some seventy separate genes occurred within this particular region, and the most likely were studied more closely in the hunt for damaging mutations.

Part of the gene NPHP4 (Nephronophthisis 4) was shown to be lacking in the sick dogs. This genetic mutation leads to the dog losing important functions, which especially affects the cones of the eye, and eventually also the rods.

A series of dachshunds were subsequently tested for the mutation. Gene-testing of wire-haired dachshunds showed that some 9.8% of the dogs carried the mutation, which seems to have appeared around 1970. With such a high percentage of the dachshund population affected, the risk that two randomly chosen dogs will both be carriers is quite high, and a genetic test is now available to reduce the possibility of the disease appearing in more litters.

Cand. Scient. Anne Caroline Wiik defended her PhD thesis, entitled "Genetic studies of cone-rod dystrophy in the standard wire-haired dachshund", at the Norwegian School of Veterinary Science, on May 13,

2009.

Provided by Norwegian School of Veterinary Science (news : web)