

A novel locus identified for glaucoma in Dandie Dinmont Terrier dog breed

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Professor Hannes Lohi's research group at the University of Helsinki and Folkhälsan Research Center, Finland, has identified a novel locus for glaucoma in Dandie Dinmont Terrier. The locus on canine chromosome 8 includes a 9.5 Mb region that is associated with glaucoma. The canine locus shares synteny to human chromosome 14, which has been previously associated with different types of human glaucomas. However, the actual glaucoma causing mutation in Dandies remains unknown. The study was published in the scientific journal *PLOS ONE* on August 14, 2013

Glaucoma is one the most common blindness causing disease both in human and in dog. Glaucoma is an optic neuropathy, which destroys the [retinal ganglion cells](#) and damages the optic nerve causing irreversible blindness. Possible elevation of the intraocular pressure may cause considerable pain. In humans glaucoma is broadly classified into three types, open-angle, closed-angle and [congenital glaucoma](#). Several loci have been mapped in humans, but only a few [causative genes](#) are known and the genetic basis remains poorly understood. In dogs several different glaucoma types are diagnosed, but only one causative gene, ADAMTS10, is known to cause open-angle glaucoma in the Beagle and in other breeds the genetic background of glaucoma is still unknown.

Collaboration between researchers benefit the glaucoma study

The glaucoma research in Dandie Dinmont Terriers has started in the United States by Doctor Gary Johnson at the University of Missouri. In the University of Helsinki samples from affected and healthy dogs have been collected since 2007. The researcher decided to collaborate to collect samples from all around the world. "Because Dandie Dinmont Terrier is globally a small breed, collecting enough samples was quite challenging", says the research leader, Professor Hannes Lohi.

Glaucoma is quite common disease in Dandie Dinmont Terrier resembling human closed-angle glaucoma. The affected dogs have very narrow or collapsed iridocorneal angles leading to obstruction of the normal outflow of the aqueous humor. This causes elevation of the intraocular pressure (IOP). Elevated IOP can be treated, but usually the most effective treatment is to remove the affected eye. Glaucoma is usually bilateral so both eyes may be removed. The average age of onset is about 7 years. As the disease is diagnosed in older dogs the affected dogs may have been used for breeding before the disease onset.

Abnormal iridocorneal angles are commonly diagnosed in the breed and many dogs are affected with pectinate ligament dysplasia (PLD). Pectinate ligament form the internal boundary of the canine iridocorneal angle. In the normal canine eye the pectinate ligament is presented as pillar of tissue, which provides support for the iris to the posterior cornea. As part of the research, 18 healthy Finnish Dandies were clinically studied by a veterinary ophthalmologist Elina Pietilä. "Based on the clinical study and ophthalmological reports collected from the affected dogs, PLD causes an elevated risk for glaucoma. 72.3 % of the clinically studied dogs had PLD, but no glaucoma was diagnosed. PLD does not always lead to glaucoma development but also other eg. genetic factors have an effect on glaucoma development", informs MSc Saija Ahonen. "It is possible that some of the studied dogs will develop glaucoma later in life, so clinically studied dogs will be followed", continues Ahonen.

The glaucoma locus was identified using genome wide association analysis

To identify the glaucoma causing gene a pedigree was constructed around the affected dogs and a genome wide association analysis was performed with 23 cases and 23 controls. A locus on canine [chromosome 8](#) was identified including 21 genes.

"The locus identification was a huge breakthrough in the project. In addition, the same chromosome has been associated with glaucoma in humans, so we can be fairly sure that we have mapped the glaucoma associated region", informs Professor Hannes Lohi. "The actual causative mutation has not been identified. Based on the results the genetic background of glaucoma may be complex meaning that multiple genes of mutation in the regulatory regions may affect [glaucoma](#) development", adds Professor Lohi.

Even though we know the where the associated region is, we cannot develop a gene test for the breed, which would be very helpful for the breeders. The locus identification gives us a lot of new information and we can now concentrate more detailed to the specific region", comment MSc Saija Ahonen.

This study is dedicated to Celia Danks, a devoted Dandie breeder and supporter, who sadly passed away 2011.

More information: Ahonen, S. et al. (2013) Genome-Wide Association Study Identifies a Novel Canine Glaucoma Locus, *PLoS ONE* 8(8): e70903. [DOI: 10.1371/journal.pone.0070903](https://doi.org/10.1371/journal.pone.0070903)

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