

Genetic Mutation Associated With Hereditary Cataract In The Australian Shepherd

Background

During 2006 researchers at the Animal Health Trust, UK identified a genetic mutation in a small number of Australian Shepherds affected with Hereditary Cataract (HC). To further investigate the relationship between the mutation and HC in the breed the AHT, in collaboration with researchers at the University of Helsinki, Finland, recruited a large number of additional Australian Shepherd samples from the UK, Europe and USA. All the samples were genotyped for the mutation and the results were analysed along with each dog's cataract status.

The results show the mutation is a risk factor for the development of cataract in the Australian Shepherd. There are currently no reports in the scientific literature describing the clinical aspects of hereditary cataracts in the Australian Shepherd but anecdotal evidence indicates bilateral posterior polar cataracts are the most prevalent form. Our study implies that the likelihood of developing bilateral posterior cataracts is approximately 11 times higher for Australian Shepherds that carry the mutation (either one or two copies) than it is for dogs that don't carry the mutation. The mutation appears to be dominant, which means that dogs only need to inherit a single copy of the mutation to be at increased risk of developing cataracts. This also means that dogs/bitches that have produced offspring with cataracts are not necessarily carriers of the mutation as the affected offspring may have inherited a single copy of the mutation from their other parent.

What The Results Mean

If your dog carries one or two copies of the mutation it has an increased chance of developing bilateral posterior cataracts. We currently estimate it is about 11 times more likely to develop this type of cataract than a dog that doesn't carry the mutation. Our estimate of the increased likelihood may change slightly as we analyse more data but is unlikely to change significantly. If your dog does not carry the mutation it has a lower chance of developing cataracts, during its lifetime. It is possible that additional mutations exist in the Australian Shepherd population that are also risk factors for the development of cataracts; our DNA test does not currently detect these mutations.

If your dog carries a single copy of the mutation it will pass the mutation (and the increased risk of developing cataracts) onto to approximately 50% of its offspring. If it carries two copies of the mutation it will pass the mutation onto all of its offspring.