

Molecular Genetic Study of Inherited Cataracts in Miniature and Toy Poodles – We Need Your Help!

What is the problem? Cataracts consist as any opacity/cloudiness of the lens, which may impair vision or cause blindness. They are a common cause of blindness in humans and are common in animals. The opacity may vary in size, shape, localization, age of onset, progression rate and cause. Cataracts are found in many dog breeds – and Poodles are no exception. Albeit the causes are multiple, an estimated 6% of all Poodles are diagnosed with inherited cataracts!

What are cataracts? How do they form? Cataracts usually arise due to abnormal lens proteins that misfold, lose solubility or are altered in any other way. In other words, the constituents of the crystalline lens lose their structure and integrity, collapse on each other and lead to a mass of cluttered material a few millimeters in size or, in the worst and more progressed cases, involving the whole lens with a proportional impairment of vision. Cataracts have various causes: trauma, radiation, chronic disease, or as a collateral effect of drugs or medications; nutrition plays a role in cataract development as well. Of course, a very important triggering factor of cataracts is aging; the majority of cataract cases manifest themselves in the later stages of life.

Nonetheless, a number of cataracts have an onset in the earlier years of the dog's life. Although some individuals use the term "Juvenile Cataracts," a genetically inherited defect is implicated (or at the very least, strongly suspected), and the appropriate term is Inherited Cataracts. Dogs with inherited cataracts are born with normal lenses, which then proceed to degenerate over time, leading to visual impairment and then blindness later in life. Inherited cataracts affect 6 percent of American Poodles, with disease beginning sometime between 2-5 years of age.

How can I help? The Poodle Club of America Foundation has funded a continuation of a three year research study to be carried out at the University of Pennsylvania and OptiGen, LLC to identify the molecular genetic basis of inherited cataracts, and develop a DNA-based diagnostic test that can be used to identify dogs that are genetically normal, carriers or affected. By judiciously using the DNA test information, breeders could minimize the risk of producing affected dogs while maintaining genetic diversity of the breed.

We are asking for assistance from all the breeders and owners of dogs (Standard, Miniature and Toy Poodles) that are normal (> 5 years of age) or affected with cataracts presumed to be inherited based on a clinical examination by a board certified veterinary ophthalmologist (ACVO or ECVO). We need samples, preferably in the form of a small 2-3 ml blood sample stored in an EDTA (purple top) tube. The sample, along with a pedigree and a completed cataract research form designed specifically for this study

will be of critical help for the ongoing studies. Please make sure that only one form is used for each study dog. For more information and to download a sample form, go to http://www.optigen.com/opt9_poodlecataractresearch.html. Complete the first section of owner and dog information. In the second section of the form, the examining ophthalmologist provides brief clinical descriptions and, if possible, clinical photographs. Send the completed form, any current/previous eye exam records, and 2-3 ml of whole unclotted blood in EDTA to OptiGen 767 Warren Rd. Ithaca NY 14850. A copy of the dog's pedigree (5-6 generations) also will be essential.

Once we enroll your dog in the study, we ask that you provide us with follow up information in the form of examination records to confirm that the dog remains normal, or, in the case of dogs with cataracts, that the cataracts are progressing in the expected manner for the inherited defect.

OK, but why is my contribution important? Your contribution is very important, and for several reasons!

- You can contribute to the health of the breed and canine health in general.
- Because of the similarities between dogs and humans, discoveries have a potential for giving scientists better insights on human cataracts.
- Many genetic studies can be carried out only when a “critical mass” of well diagnosed samples is reached, to give the scientists involved the appropriate amount of information for analyses and comparisons. Each and every sample could make the difference!

###