DNA Test for Primary Lens Luxation Now Available

From University of Missouri, College of Veterinary Medicine, Oct. 15, 2009. Submitted by the PBGVCA Health Committee.

A mutation that causes development of primary lens luxation (PLL) in many breeds of dogs has been identified by a team of researchers led by Dr. Gary Johnson and Dr. Elizabeth Giuliano at the University of Missouri College of Veterinary Medicine. A DNA test for this mutation became available in mid-September of this year through a partnership with OFA (Orthopedic Foundation for Animals).

Shortly after the announcement by the University of Missouri, researchers at the Animal Health Trust (AHT) in England also announced they had found a mutation for PLL. Dr. Catherine Mellersh and Dr. David Sargen from the AHT contacted Dr. Johnson, and both research teams have agreed to share data and co-publish this discovery. PLL testing will also be available through the AHT in England at a price comparable to the fee at OFA.

Primary lens luxation is an eye problem well-known in many terrier breeds as well as Tibetan terriers, Chinese cresteds, Australian cattle dogs and other breeds. The lens is held in place in the eye by fibers known as zonules. If these zonules stretch or break, the lens can fall out of place, or luxate. This often requires immediate veterinary attention to remove the displaced lens and prevent painful secondary glaucoma, and sometimes loss of vision.

Research at the University of Missouri led to identification of a DNA mutation that predicts which dogs are at risk for developing PLL as they age. A few months later, working independently and using other breeds, researchers at AHT found the same mutation. This independent confirmation of the finding makes both labs confident the correct mutation has been identified, and that the test is valid for many breeds. A simple DNA test will reveal if a dog is NORMAL (has two normal copies of the gene), a CARRIER (has one normal copy and one mutated copy of the gene) or AFFECTED (has two mutated copies of the gene). Wise use of this test will allow breeders to avoid producing individuals destined to develop PLL while still retaining many other desirable traits in their dogs.

Testing and Inheritance of PLL. From previous pedigree studies, there has been general agreement that PLL is inherited as a simple recessive trait. This means a dog needs two mutated, or "bad," copies of the gene to show the disease. With the PLL mutation identified and the research groups able to compare notes on dogs used in the study, it has become apparent that there are some exceptions. While the vast majority of dogs with PLL have tested AFFECTED, a small percentage of the dogs that test CARRIER are also at risk of developing PLL. Owners and breeders should be aware of this and understand the implications of the test results so they can make well-informed decisions for the future of individual dogs and the breed as a whole.

Dogs that test AFFECTED have two mutated copies of the gene. The vast majority of these dogs will luxate at four to eight years of age, the typical age of onset for PLL. There were a few dogs in the study group that tested as AFFECTED but did not luxate until after eight years of age, and some dogs testing AFFECTED have died from other causes without luxating. A search of published veterinary literature revealed about 10 percent of dogs reported to be clinically affected with PLL had onset of symptoms after age eight. Because of this, the test results will say "AFFECTED/HIGH RISK."

As stated earlier, dogs testing CARRIER are at a slight risk of developing PLL. Carriers have one normal and one mutated copy of the gene. They could pass either the normal copy or the mutated copy on to their offspring. Because there were a very few cases of dogs in the research groups testing CARRIER who did appear to have PLL, the test results will say "CARRIER/LOW RISK."

A dog testing NORMAL has two normal copies of the gene, is not at risk for developing PLL and can only pass a normal copy of the gene to any offspring.

Breeders and individual owners are now able to test any dog using the testing kit that can be ordered online through the OFA Web site, www.OFFA.org. DNA is collected using a cheek swab, and the barcoded sample will be tested by the Animal Molecular Genetics Lab at the University of Missouri with results reported directly to the owner by OFA.

Owners who had submitted samples for research prior to Sept. 1, 2009, may request test results for their dogs using the Test Request Form for Existing Samples at www.OFFA.org. These requests are now being accepted.

Owners of dogs diagnosed as AFFECTED with PLL by an ACVO- or ECVO-boarded ophthalmologist are eligible to receive a free DNA test if they send a blood sample, pedigree copy and a copy of the ophthalmologist's report. Go to www.OFFA.org for the instructions and form to submit samples from affected dogs. Samples from affected dogs may be sent now as well.

Thanks go to the clubs and many individual owners who have supported this research and participated in the project by supplying samples and information on their dogs, as well as monetary support.

Questions? Contact Project Coordinator Liz Hansen at HansenL@missouri.edu. For additional information, visit the Glaucoma and Lens Luxation section at www.CanineGeneticDiseases.net. ■

This study was supported in part by the PBGV Health and Rescue Foundation because PLL is one of the eye diseases that occurs in PBGVs. We thank the PBGV owners who submitted DNA samples from affected dogs to be included in this study.