

DNA Test for Primary Lens Luxation (PLL)

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Instead of the promised third column on the upper arm assembly, a proven new health test for Norwich has prompted me to invite Blair Kelly to write this month's column. Mr. Kelly obtained his first Norwich from Phyllis K. Pullen, MD, in 1997. He breeds under the Shaksper prefix and has been an AKC agility judge since 2001.

FIRST DNA TEST FOR A NORWICH HEALTH PROBLEM IDENTIFIED

In 2004 Dr. Phyllis Pullen noticed that her not-quite-6-year-old Norwich, Ch. Jerusalem Chutney (Chute) had developed obvious pain in one eye. She remembers, "I saw a canine ophthalmologist that day. He told me that this was due to a luxated lens that had been floating around for some time and suddenly blocked the duct that carries the vitreous fluid out of the eye, causing glaucoma." The vet told Dr. Pullen that primary lens luxation (PLL) had caused the glaucoma.

Glaucoma is a descriptive term that basically means an abnormal fluid buildup inside the eye. The eye is filled with a liquid called aqueous humor. Normally the amount of this transparent fluid is carefully balanced to keep the eyeball in its proper shape and continuously bathed.

This balance can be disturbed if for some reason the fluid cannot be removed as quickly as it is produced. This causes pressure to build up. Without treatment the elevated pressure will destroy retinal cells and cause damage to the optic nerve as well. Glaucoma is a leading cause of blindness in dogs (and people).

PLL is a hereditary eye disease where the supporting ligaments holding the lens of the eye are weak. These weak ligaments can allow the lens of the eye to detach (usually between the ages of 4 and 8 years), which can lead to painful glaucoma and blindness. PLL is common in many terrier breeds.

In Chute's case, it was too late to save the eye. Dr. Pullen relates, "The opposite eye had a loose lens also, so this lens was reattached during the surgery to remove the blind eye." Thus while one eye was lost, the other was saved.

In my readings I came across information about the PLL DNA test, which was discovered in late 2009 and developed at the University of Missouri and made available by the Orthopedic Foundation for Animals (OFA). The test had not been proven to be applicable to Norwich Terriers, however.

I remembered that Chute had lost his eye because of a lens-luxation problem, so I asked Dr. Pullen about it. She verified that Chute had been seen by an ACVO (American College of Veterinary Ophthalmologists) specialist who had diagnosed Chute's glaucoma as being caused by PLL.

At my behest, Dr. Pullen was kind enough to have Chute tested with the PLL DNA test. The test came back positive. This verified that the mutation causing PLL in Norwich terriers and the mutation found by the University of Missouri DNA test are one and the same. Dr. Pullen says she never would have noticed that Chute had PLL if he had not developed glaucoma.

The PLL mutation is a simple recessive, meaning that a dog is either clear (has no copies of the bad mutation), a carrier (has one copy of the bad mutation), or affected (has two copies of the bad

mutation). Two copies of the bad mutation are necessary for the disease to express itself—which normally happens after an age when many Norwich are bred.

Now that there is a verified DNA test for PLL in Norwich, responsible breeders can use it to avoid producing PLL-affected individuals of the breed. There is no longer a need to exclude any Norwich with PLL (nor closely related dogs) from a breeding program and from our already small Norwich gene pool. As long as affected and carrier animals are only bred to clear animals, the disease should not develop in the progeny. Thus genetic diversity need not be lost while the bad mutation is eliminated from the gene pool. And if two Norwich are bred who are both PLL-clear, then the progeny are PLL-clear “by descent” and do not need to be tested.

The OFA website allows one to see which Norwich have been tested for PLL and provides their PLL status. If you do an “Advanced Search,” you can specify Norwich Terrier as the breed and “Primary Lens Luxation” as the report type. This will show all the Norwich who have been tested and their PLL status.

Many health tests are interpretive (such as hip X-rays), requiring several experts to agree before giving a ruling. (This is similar to the requirement where several conformation judges must evaluate a dog and agree on its superior quality before the dog can be called a champion.) However, a DNA test is the gold standard in health testing, as it reads the DNA and tells definitively if a dog is affected, a carrier, or a clear for certain trait or defect.

The PLL DNA test is a simple cheek-swab test and can be ordered from the OFA website for a cost of \$65 — (www.offa.org/dnatesting/pll.html)

It would be terrible if another Norwich is bred who develops PLL and loses sight when a tool — the PLL DNA test — exists to prevent it. —B.K.

Thank you, Blair, for helping to get this crucial information out to the fancy. The Norwich community would also like to thank Dr. Phyllis Pullen for testing Chute and for agreeing to make Chute’s PLL status public. Chute passed away earlier this year from unrelated causes at age 13½. Dr. Pullen notes, “Chute had no further trouble with the repaired eye for the rest of his life and went on to compete in obedience, rally, and agility.”

Please have your Norwich tested! A CERF exam will not tell you if your Norwich is a carrier.

— Leandra Little, AKC Gazette Breed Columnist · Weehawken NJ · llittle9@earthlink.net

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