Study aims for DNA test to reduce glaucoma in Entlebucher Mountain Dogs

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Watching Finn and Luka, 2 beautiful and energetic Entlebucher Mountain Dogs, play fetch in their back-yard in Texas, you might not notice one of them relies on auditory and tactile cues to claim his prized tennis ball. Finn does this because, at 4 years of age, he has already lost both eyes to glaucoma.

Across the country at the University of Wisconsin School of Veterinary Medicine, where Finn's eyes were shipped after removal, Dr. Gillian McLellan prepares slides for analysis as part of an ongoing clinical study, “Genetic Basis of Glaucoma in Entlebucher Mountain Dogs.” Over the past 5 years, Dr. McLellan, along with Dr. Peter Muir, has led a team of researchers working to detect genetic associations and causal variants that predict the development of glaucoma in this breed. The end goal is to develop a genetic test that will lessen the occurrence of this very painful and rapidly blinding disease in thousands of dogs—not just Entlebucher Mountain Dogs.

The Entlebucher Mountain Dog is the smallest of the 4 Swiss Mountain Dog breeds. Known historically in Europe as an excellent herding dog, the breed is thought to have become nearly extinct around the time of World War I, after which renewed interest led to a small resurgence. Still, the breed’s low numbers and lack of genetic diversity were predictably problematic. Today’s Entlebucher Mountain Dog, considered a versatile, loving family pet, has a higher likelihood of developing certain health conditions, including glaucoma.

Dr. McLellan, who previously identified a gene that causes glaucoma in cats, is now looking to isolate and identify the same in Entlebucher Mountain Dogs with the hope of deepening our understanding of genetic determinants that cause glaucoma in animals and humans. “At this point, we are optimistic we’ll soon have a DNA test that will help breeders identify which dogs have 2 copies of the gene variant that causes glaucoma, which are ‘clear,’ and which are carriers, since it is likely a recessive trait in this breed,” she says. “Ideally, over time, that will be a factor in helping reduce the significant occurrence of glaucoma in Entlebuchers.”

Finn’s owners, Lilla and Chris Caho, fell in love with the breed and worked diligently to find and secure Finn—no easy task considering Entlebucher Mountain Dogs are still significantly fewer in number than more common breeds such as Retrievers and Labradors. They persisted and brought Finn home in the spring of 2020, and by all measures, he seemed healthy and happy. Two years later, they added Luka, a female Entlebucher, to the family.

Finn’s situation changed suddenly around Christmas 2023. One morning, they noticed Finn’s eye was cloudy and he lacked energy and was moving clumsily. Even though they immediately got Finn to his primary care veterinarian and a veterinary ophthalmologist, it was too late to save Finn’s eye. The veterinary ophthalmologist knew of Dr. McLellan’s work and suggested they could help spare dogs from Finn’s fate in the future if they submitted Finn’s eye to the University of Wisconsin.

Finn recovered from his first surgery and adapted well, with help from Luka (who is monitored closely and, so far, shows no signs of glaucoma). Unfortunately, the Caho family knew the signs when Finn’s remaining eye started showing symptoms about 4 months after the first. They went through the same process, including donating his eye for research. Thankfully, Finn continues to show resilience and adaptability in his recovery.

Dr. McLellan cites the support of dog owners who are willing to donate eyes as well as funding from the American Kennel Club’s Canine Health Foundation as critical to advancing the research.

“Clinical studies such as this one demonstrate why veterinary medical schools are often at the forefront of research that benefits both animals and humans,” McLellan says. “Every advancement we make as far as understanding the genetics and presentation of glaucoma in dogs is helping develop better treatment options and solutions for humans, too.”