

At a Crossroads: The Impact of Genetic Testing on Breed Diversity



“Big Red,” a popular Disney movie from 1962, made the Irish Setter a Top 10 breed in American Kennel Club registrations, a lofty ranking that came with a price, says Anne Marie Kubacz, of Jackson, New Jersey.

“By the early ’70s, there was an explosion of progressive retinal atrophy (PRA), which began as night blindness in the breed. Breeders unknowingly produced puppies that showed signs around 6 weeks of age and became blind by 1 to 2 years old,” says Kubacz, health chair of the Irish Setter Club of America (ISCA) Foundation. “It was an emotionally charged disease, and everyone wanted to do the right thing.”

Seeking answers, the parent club began working with Gustavo Aguirre, VMD, PhD, professor of medical genetics and ophthalmology at the University of Pennsylvania School of Veterinary Medicine. Dr. Aguirre found that rod-cone dysplasia type 1-PRA in young Irish Setters was caused by a nonsense mutation in the cGMP phosphodiesterase β -subunit gene (PDE6B). Identifying that cGMP was critically involved in the disease was based on a study published in September 1978 in *Science*. Once the gene and mutation were identified in 1994, a direct DNA test empowered breeders to selectively breed carriers, the dogs most responsible for disseminating the unwanted gene, to genetically normal dogs, and select genetically clear offspring for use in future breeding.

“Because of *red-1*, our club has always been a proponent of supporting research to help advance the health of our breed as well as that of all dogs,” says Kubacz, one of eight ISCA representatives — more than any other club — who attended the 2019 AKC Canine Health Foundation National Parent Club Canine Health Conference sponsored by Purina.

“Although our club usually sends two or three people, our board members felt it was important for more of us to attend this year,” Kubacz says. “They felt it was important to learn about the outcomes of the Canine Health Foundation work we support and to help sort out the currently available genetic testing for *red-1* and other diseases. We really are at a crossroads.”

Genetic testing for canine diseases, including new panel DNA testing and how these tests impact breed diversity and the health of dog breeds, was a featured topic at the AKC Canine Health Foundation National Parent Club Canine Health Conference. In the wake of concerns about preserving rare dog breeds and an overall decline in purebred dog breeding — the once Top 10-ranked Irish Setter is now 77th in AKC breed registrations — these topics are becoming increasingly important to parent clubs and breeders.

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<https://www.purinaproclub.com/resources/dog-articles/todays-breeder/genetic-diversity-testing-irish-setters>

The ABC Canine Health Foundation is committed to advancing the health of dogs through sound scientific research while also providing educational outreach for breeders, veterinarians and other dog professionals,” says Dr. Diane Brown, CEO of the Foundation. “Thanks to our partnership with Purina, we are able to hold the canine health conference every other year to bring these groups together with canine health researchers, including geneticists, to share and discuss complex matters such as DNA testing and breed diversity.”
