

Late-onset (rcd-4) progressive retinal atrophy in Irish Setters: Where are we, and where do we go from here?

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We now know from Dr. Cathryn Mellersh at the Animal Health Trust in the UK that there are at least three different inherited progressive retinal atrophy disorders in the breed; and early onset rcd-1, a still undefined middle-age onset PRA, and late-onset rcd-4 PRA.

The AHT reports a 30-40% carrier rate worldwide for the defective gene in Irish Setters. The rcd-4 gene that causes Irish Setter PRA is one that similarly causes autosomal recessive late-onset progressive retinal atrophy in man. It is the same genetic mutation causing late-onset rcd-4 PRA in Gordon Setters. Irish Setter owners will receive affected test results for dogs who have no observable vision problems. This is because this is a late-onset disorder. It was originally reported that the average onset of this form of PRA was around 10 years of age. This is the average age of Irish Setters recognized with visual impairment that test affected with rcd-4 PRA. The actual age of onset of Irish Setter rcd-4 PRA is possibly much older; with many affected dogs never reaching the age of onset of visual impairment. In addition, owners of very old Irish Setters with visual impairment may believe that it is "normal" for old dogs to not see well, and do not pursue a diagnosis of PRA. The fact of the matter is that there is a range of age of onset for the clinical signs of Irish Setter rcd-4 PRA where some may slowly lose their vision at younger than 10 years of age, and some many never show clinical signs of a vision problem.

Dr. Cathryn Mellersh at the AHT (cathryn.mellersh@aht.org.uk) is currently searching for the defective gene causing the middle-age onset form of PRA in the breed, and

is interested in cheek swab samples from affected dogs and their close relatives.

Because there is more than one form of PRA in the breed, and because Irish Setters can also have other disorders of the eyelids, cornea, lens, and retina, the rcd-4 genetic test does not replace the need for annual CERF examinations of the eyes.

The most important thing that we need to do about rcd-4 PRA is to not devastate the Irish Setter gene pool with widespread spaying/neutering, and the removing of quality dogs from breeding. Aside from the loss of quality dogs, the breed cannot withstand the removal of 30% to 40% of breeding dogs from the gene pool and maintain breed genetic diversity. This is not the only direct gene test that is available for the breed. We must all recognize that the proper use of genetic tests for recessive disease is to breed quality carrier dogs to quality clear dogs, and replace the carrier parent with a clear-testing offspring that is of equal or better quality.

If a quality dog that you determine deserves to be bred tests as a carrier, you certainly can and should breed the dog. You must make a decision counter to the emotional reaction when you received the carrier test result. Making a decision to not breed a quality dog based on a single testable gene is not appropriate. As long as carriers are not bred to carriers, no affected dogs will be produced. This is a testable and controllable gene. By dealing with rcd-4 PRA in an objective and informed manner, we can continue to produce quality Irish Setters and work away from this single gene hereditary disorder. The goal is to slowly decrease the carrier frequency in the population and slowly replace carrier breeding stock with normal offspring. This will take many generations. **A genetic test should not alter WHO gets bred, only WHO the dog gets BRED TO.**

Lastly, it is important to remember that this is about the dogs. You belong to a community that loves Irish Setters. No one wants to produce carrier or affected dogs. The stigmatizing of breeders and quality dogs due to carrier status is an old, outdated and an unacceptable practice. We need to be able to raise the level of conversation to constructive communication. Rcd-4 test results should be reported to the OFA open health database. This includes clear, carrier, and affected dogs. An application form is available in the DNA tests section of the OFA website (www.offa.org/pdf/dnaapp_bw.pdf) The application should be mailed with a copy of the official test results from the Animal Health Trust. The fee for entering rcd-4 results into the OFA database is \$15 for clear and carrier dogs, and is free for dogs testing affected. With several genetic tests available and more on the way, we know that there are no “perfect” dogs. By working together you can improve your breeding attitudes, your breeding programs, and the overall health of the Irish Setter breed.

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