



FREE Eye exam by board certified ophthalmologist at ISCA National in KY

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Time is approaching for the Jane Gavin Health Clinic at ISCA National specialty 2023.

In the April 2021 Memo there is an article about test breeding for PRA. Ironically and sadly, just after that article was published, I was contacted by a member whose dog seemed to be having vision issues. There was a local eye clinic with a board-certified ophthalmologist and much to their shock, the dog was diagnosed with having PRA. Since this dog is genetically clear of both rcd-1 and rcd-4, the hope was that with a more complete eye exam, another cause for the dog's progressive blindness could be found. Unfortunately, a second board-certified ophthalmologist confirmed the PRA diagnosis.

I was grateful to the dog's owner for being willing to have the dog examined for a **third** time by Dr Aguirre, who did all the original PRA research for Irish setters. Unfortunately, he too confirmed that the dog has PRA, that it is likely a late onset PRA which would not have been seen on exam much before the dog turned 5. Dr Aguirre could find no inflammatory reason why this dog had developed PRA.

Meanwhile thanks to the ISCA Foundation, tests were repeated with PawPrint Genetics (original testing was done via Optigen for this dog) as were tests on sibling and dam and all are genetically clear of PRA rcd-1 and rcd-4 with the current tests we have. Dr Aguirre also suggested that a genetic panel for ALL inherited eye disease in every breed with genetic eye diseases be run to see if there would be a genetic "hit" of any inherited eye disease in another breed. A multitude of tests were run and there was no positive genetic hit found.

So of course, my question to Dr Aguirre was: How should ISCA proceed with this development? His response was that it has always been his advice that dogs should have an **eye EXAM by a board-certified ophthalmologist as a young dog and then again as an older dog so that we can be vigilant about the possibility of new eye disease emerging.**

Beginning in 2022 ISCA will offer a free ophthalmology exam at our National specialty to all dogs who sign up in advance for the exam. It is very important that we see as many dogs as possible at this clinic of all ages from puppy to veteran so we can screen and see if there is evidence that a new form of PRA is emerging.

Is it possible that this incidence of PRA is a de novo mutation (something unique to this affected dog and only this affected dog)? That is a possibility. But the only way we will know if it is truly something aberrant vs something emerging in our breed is to ramp up making sure breeders know how important it is to have their dog's eyes certified via EXAM at least twice in their lives: once as a young dog and then again when the dog is over 5.

I am asking that EVERY Irish setter that is attending that National take advantage of this and contribute to the health of our breed by having this exam done. It is a simple matter of having drops placed in the dog's eyes to dilate the pupils and then about 15-30 minutes later having the eye exam performed in a dark room by the Ophthalmologist.

Below I have reprinted the article from April 2021 explaining the history of PRA in the Irish setter. **Now more than ever, as Health Chair I am imploring every Irish setter owner to PLEASE help us screen as many Irish setters as possible at the National Specialty. Please do not use the excuse that you are readying dogs for the show or want to socialize with your friends. There will be plenty of time to do that at the National outside of taking time out of your day to sign up in advance for the eye exam and to show up for the eye exam when scheduled.**

For many of you, test breeding is not something you've had to do. But back in those days, the only option we had to know if our dogs were going to produce PRA was to test breed.

We truly hope that the dog involved in the new PRA diagnosis is a de novo exception and not the beginning of a new form of PRA that we need to be vigilant about

Here is the reprint from April 2021 Memo:

I was truly fortunate to live near University of Pennsylvania and work with Dr Aguirre as he closed in on the genetic link for PRA rcd-1 (early onset PRA) in the Irish setter. He had already discovered that PRA had variants in each breed, so that a genetic test had to be specific for each breed. We were fortunate that in the Irish setter PRA was a simple recessive, so he targeted our breed in his initial genetic study.

Thanks to Dr Aguirre and the work and support of many Irish setter breeders and the ISCA Foundation, a genetic test was discovered. This was truly groundbreaking back in the day when this research was not assisted by computer matching of genes! Optigen became the lab where all ISCA genetic tests for PRA were performed. The cost of the testing was not inexpensive, but it was far less expensive and much less emotional than breeding a litter of puppies, not knowing if any would be blind and knowing all would be genetic carriers of the disease.

Sometimes the question arises as to why ISCA only recognized Optigen and the answer is simple: we needed to be sure that the integrity of the lab was beyond question, as generations of Irish setters could be affected if the test was not accurate. There were other testing agencies that sprung up, hoping to take advantage of genetic testing, and most were less expensive than Optigen.

While ISCA did not recognize those labs, some breeders chose to use them. Sadly, there was at least one group of Irish setters detonated when a supposedly genetically clear litter tested by a lab other than Optigen had a blind puppy in it. Testing of the sire and dam via Optigen revealed that both were carriers, not clear, and had produced a blind offspring. This sad event pretty much ended breeders going outside of approved labs for testing.

When rcd-4 (late onset PRA) was discovered in some Irish setters, ISCA worked quickly with the Animal Health Trust in England to determine the incidence in Irish setters in the US. The ISCA Foundation sponsored clinics and I flew to specialties all over the US collecting cheek swab samples to send to the Animal Health Trust for testing. Fortunately, the incidence is unlikely in most bloodlines, but testing is still important!

When Optigen notified us that they were being bought out by another lab, ISCA began our search for a new partner. Thanks to Jerold Bell DVM for his assistance and Dr Aguirre for his guidance, PawPrint Genetics is now approved by ISCA for testing for rcd-1 PRA and rcd-4 PRA ; their Irish setter panel included tests for both of these plus Degenerative Myelopathy, Leukocyte Adhesion Deficiency, Globoid Cell Leukodystrophy, and Von Willebrand disease 1. There is a year-round code that breeders can use for 50% off this test cost: ISCA2021. At our National specialty and the two weeks surrounding the National specialty dates, the discount is 55%.

Another question that arises is what do you do if in fact one of your dogs is discovered to be a carrier of PRA? Fortunately, genetic testing allows you to go forward with that dog in your breeding program. You would breed that Irish setter to a genetically clear Irish setter and then test each puppy in the litter to determine who is a carrier and who is genetically clear. Ideally the genetically clear puppies are the ones to go forward with in a breeding program. It is true that in some countries, breeding a genetic carrier of PRA is forbidden, but that approach is foolish and a misuse of the basis for genetic testing! As we have heard from many veterinary experts, genetic diversity is especially important in our dogs, and eliminating an otherwise healthy dog from a breeding program based on its carrier status of PRA makes no sense and potentially can reduce a gene pool.

This test is now a cheek swab test that owners can do at home. The instructions are clear and easy to follow. To process dogs most efficiently at our health clinic, we will still draw blood for the test at our health clinic. Once results are received from PawPrint Genetics, you can send those results to OFA to be recorded in their CHIC database. For multiple tests, it is \$7.50 per test. This is the link to the form: <https://www.ofa.org/application-forms>

It is still possible to have genetically clear dogs by parentage via OFA, which as taken over the application for genetically clear dogs from the Irish Setter Club Genetic Registry. Here is that link: https://irishsetterclub.org/wp-content/uploads/2019/11/ISGR_DNA_App.pdf

However, Dr Aguirre warned us back in the 80s we needed to test at least every third generation. Frankly at the small cost associated with this test now, most breeders chose to simply test every dog they breed. Certainly, all frequently used stud dogs should be tested directly.

One final question to address: why is this test available via cheek swab when for many years the only way to get the test via Optigen was to have blood drawn by a vet or at our health clinic? The simple answer is that genetic testing and the use of cheek swabs for DNA has become more reliable in terms of the amount of DNA that can be extracted from a cheek swab! Science makes progress and ISCA embraces that progress once it is proven successful.