

## **Progressive Retinal Atrophy (PRA)**

Progressive Retinal Atrophy (PRA) is a disease which causes dogs to become blind. This project, which is now complete, was the focus of research for over 25 years. Due to the support and diligence of Irish Setter owners and breeders, scientists were able to discover the mutation on the gene which causes PRA in Irish Setters. A DNA blood test was developed which reveals normal dogs, carriers and affected dogs. Dogs can be tested as soon after birth as it is safe to draw blood.

Before the discovery of the DNA blood test, breeders test-mated their dogs, an arduous and heartbreaking process, in order to eliminate carriers from the breeding pool. To keep track of those test-mated dogs, the Irish Setter Genetic Registry was formed, independently of ISCA, but closely affiliated with it. ISGR monitored all the paperwork and procedures that each individual followed. TM (test mated) numbers were given to all those dogs passing a test mating. If two TM dogs were mated, then their offspring were issued TMA (Test Mated Ancestry) numbers. Once the blood test was developed it was thought that ISGR would no longer be needed, but breeders found there still exists a need for a registry that would enable them to keep track of the status of their blood tested offspring.

The blood test is administered by a veterinarian and sent to Optigen, the only laboratory currently approved by ISCA to analyze and record the data on individual dogs. Once the dog has been DNA tested results are sent by Optigen to OFA. The owners of the dog receive a certificate from OFA, who is now administering ISGR. The owner of the dog must fill out an appropriate form and send it along to OFA with a fee of \$7.50 in order to be registered with ISGR.

If the sire and dam of a litter have both been DNA blood tested by Optigen, their offspring can receive a Gene Tested Ancestry (GTA) certificate. In order for this to be issued, the CERF certificate must be submitted along with an application obtained from ISGR. The combination of the DNA Optigen blood test, which enables the dog to get a permanent CERF number and the ISGR registry, offers breeders the proof that their puppies have been cleared of PRA.

### **A Message from Eddie Dziuk, CEO of OFA, regarding registering genetically clear PRA offspring at OFA**

There are essentially 2 ways of listing PRA clearances for Irish Setters, through the OFA and through the ISGR which the OFA administers for the ISCA.

OFA wise, owners can register their Optigen Test Results with the OFA, and receive an OFA number. Or for first generation offspring of clear parents where all three, sire/dam/offspring have been DNA profiled to verify parentage, the OFA will issue a clearance with a CBP (Clear By Parentage) suffix at the end of the assigned OFA number. The OFA's CBP policy can be found on the OFA website at: <http://www.offa.org/cbp.html>

This policy is applied consistently across all DNA based databases.

The 2<sup>nd</sup> way of listing clearances is through the ISGR. The OFA began administering the ISGR for the ISCA in early 2008. All historical data was imported, we maintain the database, and have been issuing ISGR numbers since we began the administration process. For ISGR, owners can submit their Optigen reports and they are assigned ISGR numbers in the format: ISGR-DNA-####-GT. The numbering format indicates that the dog was Gene Tested. For descendants of clear dogs, the only requirement to be issued a number is that the sire and dam both have existing ISGR clearance numbers. There is no restrictive limit on the number of generations that will be cleared, and there is no requirement that parentage verification be documented. These were the existing ISGR guidelines when we took over the program, and we continue to run ISGR under the same criteria. Clear descendants are issued numbers in the format: ISGR-GTA-###, GTA indicating "Gene Tested Ancestry".

BOTH OFA and ISGR PRA clearance numbers are displayed on the OFA website.

BOTH OFA and ISGR PRA clearance numbers meet the PRA requirement for CHIC.

Hopefully this clears up any misconceptions, more importantly misinformation.

To download the form for registering blood tested dogs with ISGR [click here](#).

To download the form for registering offspring of tested sires and dams [click here](#)

## Late Onset PRA

There have been recent breaking developments in the UK about a new form of PRA in the Irish setter, a late onset form of PRA rcd4, also referred to as LOPRA. This research has been done by Dr. Cathryn Mellersh and her team at the Animal Health Trust in the UK.

This is truly emerging information. The actual research has NOT been published or peer reviewed yet.

It is my goal as Health Chair to provide up to the minute factual scientific information and testing information regarding this in the Memo, as well as links here, on the ISCA website, to this information.

As you read through this information please keep in mind the advice received from Dr. Aguirre, who did the original PRA DNA work for ISCA.

In my correspondence with Gustavo Aguirre DVM PhD he provided this timely and important insight as we begin to test dogs in the US and get back results on our dogs:

" There is absolutely no need to panic and the breeding recommendations that we have in place for rcd1 apply to LOPRA. Affected dogs CAN and SHOULD be bred if of exceptional quality, but only to genetically normal dogs. Please keep reminding the club members that you are breeding dogs and not test results, and it is essential to maintain genetic diversity in the breed. "

Thanks to Dr. Gustavo Aguirre, who did ISCA's initial research on PRA, for his guidance, to Dr. Christine Haakenson at AKC Canine Health Foundation for her contacts at AHT, to Dr. Mellersh for her insight, to Dr. Jerold Bell for the article about how to use the info once you have test results, to Dr. Nigel Holmes, the Genetics Service Manager at AHT for his help getting testing services info to me as well as permission to publish a direct link to the AHT info on rcd4 on the ISCA website and to Kim Poile, Lab Tech at AHT for answering many questions as well!

Finally thanks to Connie Vanacore and Jan Ziech for helping me every step of the way and to ISCA member Terry Moberg for giving me an early heads up about it and ISCA Member from the UK David Bell for his assistance.

### **Information about rcd4 in the Irish setter and how to use test results:**

Please refer here to the ISCA Website, which will have all the most current links to info about what PRA is and what rcd4 LOPRA is.

On the website you will find also a wonderful article from Dr. Jerold Bell. The article from Dr. Bell provides insight as to how to use the test results. ([Dr. Bell's article](#))

Rather than publish text from the Animal Health Trust website, a link is provided below.

It is VERY IMPORTANT that members realize that the AHT website is constantly updated as information becomes available.

Please refer to the link for the most current emerging information, which is updated frequently:

[http://www.aht.org.uk/genetics\\_prarc4\\_irish.html](http://www.aht.org.uk/genetics_prarc4_irish.html)

Be sure to refresh your browser often to assure you are viewing the most current info.

### **New: rcd4 PRA DNA Test for Gordon and Irish Setters now available from the OFA!**

Researchers at the Animal Health Trust (AHT) in the UK have discovered the gene mutation for the rcd4 form of PRA which affects both Gordon and Irish Setters. The DNA test became commercially available through the AHT in March of 2011. The AHT has graciously shared the science with the OFA and the University of Missouri to facilitate the availability of the test to North American owners. Tests can be ordered online through the OFA website. DNA samples will be collected via FTA cards which are non-invasive and can easily be done at home by the owner. Testing will be done at the University of Missouri College of Veterinary Medicine Small Animal

Molecular Genetics Laboratory. The OFA will handle all order processing and reporting. Inclusion of the results in the OFA database is included with the order. The test is offered at \$65.

To order the test online through the OFA: <http://www.offa.org/dnatesting/rcd4.html>

Information from the AHT regarding rcd4 PRA in Irish Setters:

<http://www.ahtdnatesting.co.uk/news/10>

**Where to Record rcd4 Test Results:** Confidential test results will be sent to the OWNER ONLY. Once owners receive test results from AHT, they may choose to make the results public. To submit the results to OFA complete the form in the following link, attach the official test results received from AHT and send them to OFA. The OFA form to use is found at:

[http://www.offa.org/pdf/dnaapp\\_bw.pdf](http://www.offa.org/pdf/dnaapp_bw.pdf)

### **How Do I Decide Which of My Dogs to Test?**

As Health Chair, I have been asked by many of you how to decide which dogs to test. Here is my suggestion:

Consider testing any stud dog that has been used more than once or a young dog that you believe may be used at stud. This will help you and all the breeders who have used your dog once the research is published and peer reviewed about the heritability of rcd4.

Consider testing any bitch that you plan to breed over the next 12-18 months.

Consider testing any dog or bitch that you own, to help support this ground breaking research.

The greater the variety of bloodlines and ages of dogs from puppies to veterans that we have tested and the greater the number of samples we get, the more significant the data becomes.

Our path is so much simpler than years ago when to participate in PRA research, breeders had to test breed stock! DNA research has made it possible to every Irish setter owner and breeder to play a role in determining the incidence and significance of this emerging issue.

At the clinics held in fall 2011, and with samples sent by Irish setter owners to AHT, there were 106 samples analyzed. All 106 samples were CLEAR.

Does this mean that rcd4 is not an issue? NO! We must be diligent in continuing to test for this, and be certain that we are not complacent. It is always better to be ahead of a disease than chasing it!

Anne Marie Kubacz  
ISCA Health Chair