Progressive Retinal Atrophy in Standard Wirehaired Dachshunds of European Descent.

By Jolanta Jeanneney, April 10, 2011.

We have some important news to report about PRA (Progressive Retinal Atrophy) in the gene pool of standard wirehaired dachshunds descended from European lines and now residing and being bred in North America. The PRA gene mutation was found in one of our wirehaired dachshunds. He has been genetically tested to be a carrier, but he will not develop PRA himself. This report is long, so please bear with me. First some background information is in order.

PRA is an inherited disease of the retina in dogs, and the end result is blindness. PRA occurs in both eyes simultaneously, and it does not cause pain. It occurs in most breeds of dogs and can occur in mixed breeds as well. There are many different types of PRA, and this term is used as an umbrella term for a number of inherited retinal degenerative diseases.

In spring 1996 one of the older wirehaired dachshunds in the Deer Search family was diagnosed with PRA. The Deer Search dogs were descendants from German imports. On May 5, 1996 Dr. Gregory Acland and Julie Ann Alling of Cornell University conducted the PRA Clinic for dachshunds of Deer Search breeding.

The results were very encouraging for dachshund owners and breeders. Thirty-nine (39) wirehaired dachshunds were examined and symptoms of PRA were not found in any of them. Since Dr. Acland had examined twelve other dachshunds earlier that spring, a total of fifty-one (51) dogs were tested. Of the fifty-one dogs only one had PRA symptoms.

During the testing of Deer Search wirehaired dachshunds in spring 1996 Dr. Acland collected blood from 12 dogs for genetic testing, and 10 of them were related to the individual diagnosed with PRA. We also included two of our imports, Gerte vom Dornenfeld and Fausto de la Grande Futaie, in the test.

Thirteen years passed.

At the end of January 2009 I was contacted by Dr. Acland. He wrote:

"All of the 12 dogs examined and collected in 1996 were tested for two types of PRA mutations:
1. the RPGRIP1 mutation reportedly associated with PRA in MLHDs (mini longs) and other breeds -- all dogs tested normal/clear (i.e homozygous wild type) for this mutation;

2. the deletion mutation reported to be associated with PRA in SWHDs (standard wirehaired dachshunds) in Europe reported in http://genome.cshlp.org/content/18/9/1415 *A deletion in nephronophthisis 4 (NPHP4) is associated with recessive cone-rod dystrophy in standard wire-haired dachshund* by Anne Caroline Wiik, Claire Wade, Tara Biagi, Ernst-Otto Ropstad, Ellen Bjerkås, Kerstin Lindblad-Toh, and Frode Lingaas.

The individual diagnosed with PRA in spring 1996 tested homozygous for the mutation reported in the paper. Four related dachshunds have tested as

carriers and the rest tested clear." (By the way, our Gerte and Fausto tested clear).

The NPHP4 mutation is recessive and two copies of the mutation have to be present for a dog to develop disease. The DNA test can determine which dogs have no mutation at all, which ones have one copy (and these dogs are called carriers of the disease), and which ones have two copies and most likely will develop the disease.

I asked Dr. Acland whether other breeds tested positive for this specific mutation and the answer was "no". So far only wirehaired dachshunds have tested positive.

Now the foundation dogs of Deer Search are gone, and the bloodlines tested in 1996 are extinct. No descendants from these bloodlines are being used for breeding these days, and only one dachshund back in 1996 was diagnosed with PRA. However, we know that the mutation is present in the European gene pool of standard wires.

> The DTK (German Teckel Club) requires that every dog used for breeding has to be checked by an ophthalmologist for the presence of PRA and cataracts every other year, and only dogs that are PRA and cataracts-free are approved for breeding. If a dog shows symptoms, his name is published in the DTK Zuchtbuch and this information becomes public. Breeders, based on this info, can adjust their breeding strategies. We have been following these guidelines, and our dogs have their eyes checked on a regular basis.
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> There is a very long document available online, which includes names of standard wires who produced offspring diagnosed with PRA. If a disease-free dog produces even one offspring with symptoms of PRA, it means that he is a PRA carrier. The document can be located at http://www.teckelzucht.info/pra-und-katarakt-beim-teckel.pdf. The names of wires are under subheading 6.3.6. When we import dachshunds from Germany we always request breeders for dogs with pedigrees that don't contain PRA carriers in the last 3 generations.
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> However, until now there was no DNA test available and only a small percentage of carriers was identified by the fact that they produced offspring with PRA when bred to another carrier.

Since in the last fifteen years John and I imported a number of wires from Europe, we asked Dr. Ackland to test them for the mutation and he agreed. On March 26, 2009 we took all our eleven dogs to Cornell. Their eyes were examined, and DNA samples were collected.

Two more years passed, and finally a month ago we got a long overdue e-mail stating that of the eleven dogs tested, ten were normal (mutation-free) and one was a carrier (has one copy of the mutation). This particular dog was imported from Germany.

What do the results mean?

Actually the results are very encouraging as almost all our dogs used for breeding in the last decade tested free of mutation. Only one dog has one copy of the mutation. We are lucky! This dog has sired five litters in the United States, and his offspring that will be used for further breeding will be DNA tested, and carriers will be allowed to be bred to clear, mutation-free dogs only.

What can we do?

I have not found any lab in the States offering the test for the NPHP4 mutation on a commercial basis, but hopefully things will change in the future. Luckily, the Animal Health Trust in UK offers the DNA test for the NPHP4 mutation. To read more about it go to http://www.aht.org.uk/genetics_pracrd.html. One can request a test kit from http://www.ahtdnatesting.co.uk/. The test fee can be paid by a credit card. Collecting DNA is very easy (cheek swab) and anyone can do it. The fee is 54 British pounds (around $88), and the shipment is free.

Which dogs should be tested?

For us in the United States the highest priority is to test dogs that were imported from Europe and used for breeding. If a dog tests positive for the mutation, then his offspring used for breeding should be tested too. There is no need to test offspring from two parents who already tested clear.

What are breeding recommendations?

When a DNA test is available the best professional recommendation regarding handling disease caused by a recessive mutation is to use carriers (who have one copy of mutation) only for breeding to clear dogs (without mutation). Removing all carriers from the gene pool would reduce genetic diversity too much. The goal is to produce healthy PRA-free dogs. The dogs showing symptoms of PRA could come only from breeding a carrier to another carrier or affected dog.

Final thoughts

The availability of DNA test for NPHP4 mutation will help breeders a great deal with controlling PRA in standard wirehaired dachshunds out of European bloodlines. However, it is necessary to have a large picture in mind when dealing with genetic-based diseases. PRA is just one of many factors that should be considered whether the dog should be bred or not. One of the best articles on this topic is at http://www.lgd.org/library/PadgettDefects.htm.

This report has been produced by Jolanta Jeanneney, Ph.D. based on her experience and education, and it is not associated or endorsed by any club such as DTK, NATC, DCA, UBT or Deer Search. You can reach her at jola@born-to-track.com.