

Announcing!
The completion of a genetic test for
IMPROPER COAT (IC)
in Portuguese Water Dogs

For more than a decade, you have helped us analyze the complex genetics that regulate conformation, behavior and most importantly those complicated diseases that afflict both our dogs and ourselves during later life e.g. cancer, IBD or Addison's disease. We are pleased to announce, as a thank-you gift for your help, the completion of a genetic test for Improper Coat (IC).

The test, described further below, is a "gene" test, not a linkage test. We do not know if this is the only gene that can cause IC, but it is responsible for the majority of cases. The test readily determines if a Portuguese Water Dog is free of the causative gene or is a carrier of that gene.

The Georgie Project

At this time, it seems appropriate to review a bit of the history of the road we have traveled together: It is 24 years since my daughter presented us with a six-week-old puppy that she rescued. It was two more years before we learned that "Georgie" was a Portuguese Water Dog. Fourteen years ago, Georgie died of autoimmune hemolytic anemia. Her death was not an end, but rather a beginning: the search for another PWD led to the creation of the Georgie Project, in which Portuguese Water Dog owners and breeders collaborate with researchers in my laboratory at the University of Utah to study the genetics of the breed with the ultimate goal of



"Dutch" photo courtesy Connie Millard

*Thank
You*

By Dr. Gordon Lark

improving the health of the breed and of all dogs everywhere. This was possible because all of you were dedicated to this common goal. We first encountered this positive attitude when we sent tape measures to 500 of you and got measurements back from ~300 a 60% response, unbelievable!!! Those measures

allowed us to determine that it would be possible to analyze the genetic basis for PWD conformation based on skeletal radiographs. Subsequent research established that the quantitative genetics of dogs was within reach and with your collaboration we helped convince the scientific community to sequence the dog genome and the molecular basis for dog genetics was established.

Autopsy

Recently, when writing about the Georgie Project, NIH author Alison Davis noted that "the benefits of autopsy to medical research are enormous. Modern medicine got its start, for the most part, from making connections between a patient's symptoms and diseased organs found after death. Autopsy has several purposes and benefits. Examining a body after death can confirm or disprove a clinical diagnosis." See: <http://www.georgieproject.com/new/autopsy/summary.html>

Today, we work, through autopsy, to determine the genetic components that regulate Portuguese Water Dog health. We have autopsied more than 200 dogs that you sent to us during the past three to four years and will need to autopsy about 300 more during the next four to five years in order to complete our genetic analysis of "health at time of death." We will also need to find new sources of funding for these 300 autopsies.

The initial results are truly astounding. The presence of multiple sub-clinical pathologies, not diagnosed prior to death, is typical of many (perhaps most) of the autopsies that we perform. We have found significant frequencies of thyroid atrophy, pancreatitis, IBD, and, of course different cancers. All of these appear to be heritable. That is, they have a strong genetic component and we should be able to identify the genetic loci that are involved, once we have enough autopsies. Go to <http://www.georgieproject.com/new/autopsy/summary.html> For more information about the Georgie Autopsy Project.

The test for improper coat (IC)

In the fall of 2007 we were contacted by Joan Bendure who inquired whether it was possible to develop a test for Improper Coat (IC). In December of that year, Bev Rafferty provided us with compelling evidence for the recessive nature of IC based on breedings that she had done. Bev's cooperation and help convinced us to try and map the IC genetic locus. Despite preliminary attempts to map a gene for IC we were unsuccessful in doing so during the following year.

Recently we participated in a multi-breed study by the Ostrander laboratory at NIH that identified various genes regulating dog coats, including a gene responsible for furnishings (specific patches of hair that grow out faster than the rest of the dog coat). As part of that study one IC-affected PWD was genotyped and lacked a part of the DNA sequence ("indel") associated with the presence of furnishings. This suggested that the presence or absence of this mutation might be responsible for IC. The presence of this "indel" makes the sequence longer than DNA from dogs that lack the "indel." We therefore tested for the presence of this mutation in affected, carrier and clear dogs. The region containing the mutation produced a clear distinction between affected, carrier and clear dogs. This was true of 5 affected, 19 carrier and 10 clear dogs tested. Based on the marker tests for 287 dogs the mutated allele was probably introduced into the PWD population in the 1970s and the current allele frequency in the PWD population is about 15%. A follow up study that analyzed a litter from a carrier to affected cross (provided by Mimi Johnson and Kari Lavalli), confirmed these results.

As a consequence, together with Heidi Parker of the Ostrander lab we have developed a DNA test that determines whether a dog is a carrier of IC.

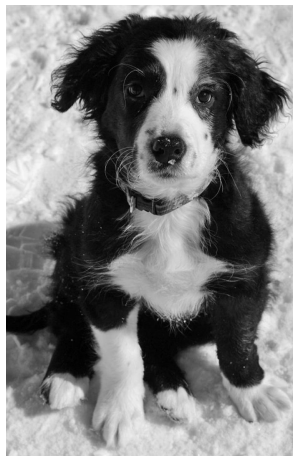
It is important to note that other, infrequent genes also may determine IC and that this test is only reliable as a test for the presence or absence of this particular mutation. As more dogs are tested we can more accurately estimate the probability that one or more other genes are involved. Currently we believe that this gene is responsible for more than 95% of the cases of improper coat.

This test will be offered by OptiGen. OptiGen has retained DNA from many PWDs previously tested for *prcd*-PRA and you will be able to request an IC test on stored samples or to provide cheek swabs. See OptiGen's home page for further details at www.optigen.com.

Our profound thanks to all who have supported The Georgie Project over the years.



Improperly coated Portuguese Water Dogs



OptiGen is pleased to offer the new test for Improper Coat described by Dr. Lark in this issue of The Courier:

The IC13 Test.

**Please check
www.optigen.com
for details.**



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Improperly Coated Portuguese Water Dogs

by Karen Miller

Twenty five years ago I went to see a six-week-old litter by a well known American sire out of an imported dam. There were eight black puppies, all wavy. Five romped in a wonderful playground with things to climb and jump on, a pool to wade in, and toys, lots of toys. On the far side of the yard was a small pen with the other three littermates.

They had no toys nor was anyone paying them any attention.

They were waiting to go to the Humane Society.

They were unwanted, unsaleable, improperly coated Portuguese Water Dogs.



What is “improper coat?” Why are breeders so concerned about it? What does it look like? Why does it happen? What can be done?

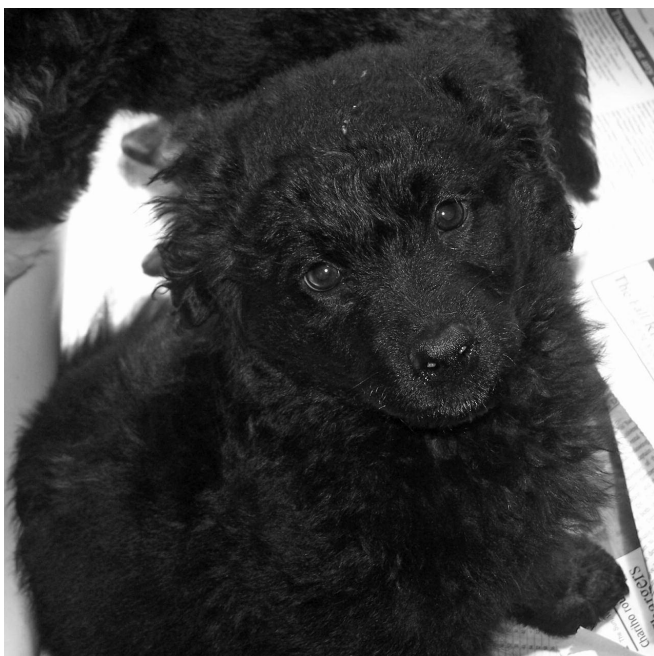
Improper coat (IC) is a genetic mutation found in pure bred Portuguese Water Dogs, probably originating with the inclusion of some other breed at some point in our history. It is not a health issue. An IC Portuguese Water Dog is healthy and in every other way can be as correct to the Breed Standard, structurally and mentally, as any of its properly coated littermates.

That being said, the Portuguese Water Dog is a “coated” breed. To the extent that “hair” makes the dog, it does so in our breed. That lush, non-shedding, single coat, either wavy or curly is an adored as well as required characteristic of our Breed Standard:

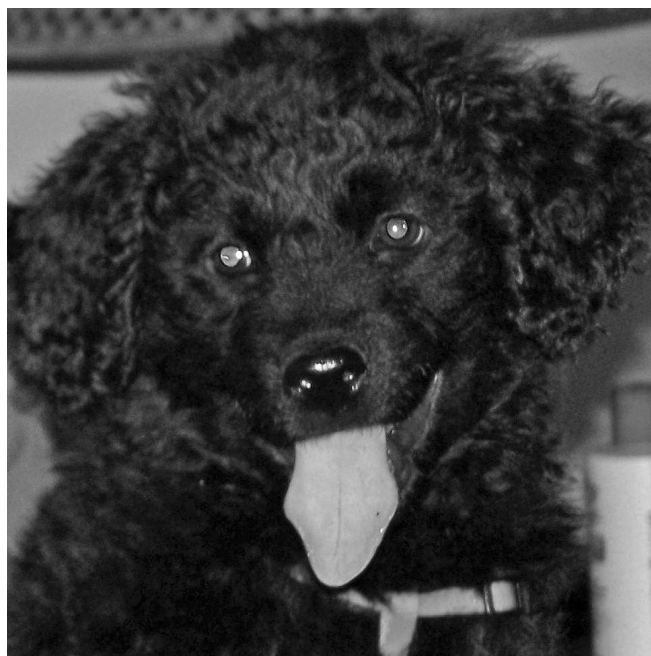
“A profuse, thickly planted coat of strong, healthy hair, covering the whole body evenly, except where the forearm meets the brisket and in the groin area, where it is thinner.”

The hair of the improperly coated dog may be discernibly curly or wavy but bears little resemblance to a correct Portuguese Water Dog coat. The hair of the IC dog’s muzzle, in fact, the face and head, doesn’t grow out long, it remains short and flat to the skin, looking more like a Border Collie coat. The hair on the legs is short and sometimes sparse. The coat along the neck, withers and the loin is frequently longer and thicker. The coat is also likely to shed.

Rescue Chair Mary Harkins: “Improperly coated dogs are very hard to place. They just don’t look like Portuguese Water Dogs and most people want dogs that do. Plus there’s the shedding problem. One of the reasons people are attracted to our breed is because they don’t shed; IC dogs do. I can place a blind dog more easily.”



Darwin at 8 weeks



Darwin at 9 weeks

The telltale signs that identify an improperly coated puppy may be clear at two weeks of age or not until four weeks. Many breeders don't recognize them until much closer to placement time.

Breeder Jane Harding: "I have had improperly coated dogs in my 23 years of breeding. I thought I could always tell IC at two weeks of age – those pups with more visible toe nails, the more pointy noses, the coat that was too straight and thick. Recently I saw a litter at two weeks and saw nothing unusual when I trimmed their toe nails, but by four weeks the IC was clear. Pat Volz has told me that if you brush, with your finger, against the lay of the coat, up the muzzle and over the eyes, and it's sparse, that is also a good indicator of an IC coat when compared to what you are positive is a proper coat. Pat thinks you can do this at two weeks."



Breeder Lisa Wiley: "I have never had any trouble placing IC puppies. Many times a visiting family falls in love with an IC puppy. Many owners like the ease of coat care. I do not sell them to families with allergies. My daughter Lauren is very allergic to shedding dogs although she does not react to IC Portuguese Water Dogs. Families with allergic relatives tell me the same thing. There are people who are allergic to

them just as there are people who are allergic to normally coated PWDs. My IC puppies are wonderful family dogs. One is a hearing ear dog for a mother of twins. Another was recently on the Animal Planet show *Go Fetch*. One went to a family that got their first normally coated PWD from Deyanne Miller; they went home with a wonderful little IC

girl. The IC puppies seem to have the best structure. I don't know why but it always seems that bitch I was thinking about keeping turns out IC."

A properly placed IC Portuguese Water Dog can and will be all a Portuguese Water Dog can be. Being sure they get to the right homes is frequently of more concern to breeders than with normally coated puppies. Growing up in the correct environment can produce spectacular results.



Breeder Karen Kirby Ash: "My Dolly is a beautiful dog and her IC coat shows off her head and structure. Her coat is shiny and dries quickly, a big plus for a working water dog. Occasionally competing in agility and obedience with Dolly has been a challenge as she is black and white and is repeatedly mistaken for a Border Collie. Once in obedience, I had a

judge who decided I was trying to pass off Dolly as a pure bred dog. This judge talked to the show chair about 'an exhibitor having the nerve to bring a mixed breed dog into her ring!' The show chair, who knew Dolly, confirmed that she was a pure bred Portuguese Water Dog with an improper coat. The obedience judge responded that I should have euthanized her!"

Euthanize Dolly? Unconscionable! What was that stupid person thinking? Improperly coated Portuguese Water Dogs are shown in every venue except conformation and are respected in every venue, a true exemplar being Presidio Dutch of Alto Mare, "Dutch."

Courier Editor Terry Cardillino: "Dutch's career as a Search and Rescue (SAR) dog encompassed dozens of searches in

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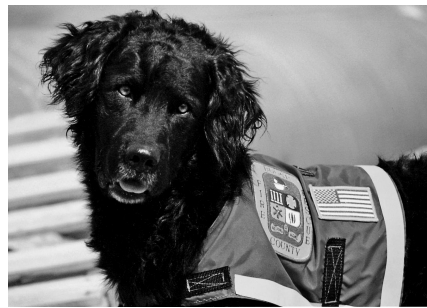


Darwin at 21 weeks



Darwin at 21 weeks

Missouri and beyond. He was the first Portuguese Water Dog ever to earn a Search and Rescue Wilderness certification, a Cadaver certification, and FEMA Urban Search and Rescue Advanced certification. He competed in AKC events simultaneously with his SAR career. His exceptional talent earned



Dutch

him an Agility High in Trial his very first time in the agility ring, a Tracking Dog Excellent title, a Courier Water Dog Excellent title, and he was the winner of the 1999 PWDC Super Dog competition.”

We can easily see the coat differences between IC and properly coated dogs. What we have not been able to see is the gene or genes that produce the IC coat type. And that’s been of interest to many breeders for a very long time.

Breeder-Judge Linda Fowler: “My first stud dog sired 14 litters and produced no IC until the eighth breeding and thereafter only two litters had IC, a total of seven IC pups. His son, Adam, produced 16 litters, three of which included IC, for a total of eight IC pups. If indeed this is a simple recessive, as people continue to tell me, these percentages just don’t make sense.”

Breeder Bev Rafferty: “For as long as I’ve been breeding Portuguese Water Dogs, there had been two opposing beliefs about the mode of inheritance for improper coats. One group was convinced that it was caused by a simple recessive gene, the other equally convinced that it was not. Finally, an informal group of PWD fanciers decided to try and answer the question once and for all. It is very easy to conduct a test-breeding for a simple recessive trait. You breed two ‘affected’ parents together, and all puppies in the litter will exhibit that trait. If even a single puppy does not, then that mode of inheritance (a simple recessive trait) does not apply. Test-breeding for improper coats is also easy because you’re not purposely breeding for a health problem.



Photo: Joseph Casey

“In 2006, after the appropriate health testing on the parents, we brought two improperly coated Portuguese Water Dogs together, resulting in six puppies. All puppies were improperly coated, which gave us a 98.5% statistical probability that we were dealing with a simple recessive gene – certainly enough to convince even the doubters among us. I contacted Dr. Gordon Lark to ask if there was interest in DNA from the parents and puppies. We’re happy that we had the opportunity to contribute to the Georgie Project’s ongoing good work. A few people deserve acknowledgement for their participation, including Jean Combs, Debbie Norris, Lynette Appelhans, Martha Thomas, Karen Arends, and Cathy Cates.”

The Georgie Project’s commitment to creating a test for improper coat has been an important service to our breed. Now we can identify which breeding dogs carry the improper coat gene and react accordingly.

Breeder Joan Bendure: “Let’s face it, without good research and gene tests there is no way for breeders to know what genes our dogs are carrying and what we might produce in a litter. I know breeders will say that IC is not a health problem, and I know it does not make a dog sick, but what about improperly coated PWDs if they end up in a shelter? Not being recognized as a PWD can be a death sentence! I am thankful that Georgie was willing to take on this research and even more elated that a DNA test has been developed. I know from published papers that Georgie has had a significant role in dog research worldwide and that Georgie has been able to have the Portuguese Water Dog breed included in dog research in general.”

Breeder Linda Fowler: “I am thrilled to know that we will have a test available. I just hope that this does not throw out a number of worthy stud dogs.”

That’s a wise thought.

Be aware that along with the good news, and of critical importance, is a warning to breeders: Identifying the IC gene in breeding stock is all well and good. Breeding a carrier (or even an IC dog) to a normal is fine and sensible to avoid producing improperly coated dogs. But, and it’s a big “but,” not using IC carriers in breeding programs is NOT fine. There is lots more research that needs to be done to fully understand how this gene works. We’ve thrown the baby out with the bath water in many previous breeding decisions, let’s learn from those mistakes, not repeat them.

Research scientist Kevin Chase: “This is an important regulatory gene. There are also some 30 other genes in close proximity to it. Removing this variant of the gene from the breeding population would remove a tremendous amount of genetic diversity in the breed. This would be very dangerous.”