

## A New Form of Ichthyosis in Golden Retrievers

For many months, a few GRCA members have openly discussed skin disease in litters of Golden Retrievers who had been DNA tested for the common form of Golden Retriever ichthyosis (GR ICH1, a PNPLA1 mutation) and were not affected by GR ICH1. Recently, an established group of canine skin disease researchers confirmed to the GRCA Health and Genetics committee (H&G) that a different gene can cause a recessive form of ichthyosis in Golden Retrievers and that this new ichthyosis causes more clinical signs than GR ICH1. Clinical signs in affected dogs (those inheriting the gene from both sire and dam) may include more severe skin disease, chronic skin infections, and stunted growth. Some affected Golden Retrievers are euthanized.

A DNA test is now available from PennGen ([www.vet.upenn.edu/research/academic-departments/clinical-sciences-advanced-medicine/research-labs-centers/penngen/penngen-tests](http://www.vet.upenn.edu/research/academic-departments/clinical-sciences-advanced-medicine/research-labs-centers/penngen/penngen-tests)). As described, breeders can avoid producing affected puppies as long as at least one of the parents is normal/clear for this new ichthyosis. However, these findings are not yet reviewed by the veterinary scientific community (peer-reviewed). It is, nevertheless, important that members know the experience of breeders who have produced affected puppies, sought the advice of experts, and have helped gather samples needed to identify the underlying cause. For this reason, we requested the attached article by Sharon MacDermott. Breeders like Sharon, who recognize a problem in their litters and help in identifying the underlying cause, are heroes who can make future puppies healthier.

For those unfamiliar with ichthyosis, the DNA test for GR ICH1 is available from several laboratories and is usually simply called an ichthyosis test (Golden Retriever form) — but only tests for a specific recessive mutation in the PNPLA1 gene (1). Most Golden Retrievers with GR ICH1 have dandruff as puppies with little or no evidence of skin disease as adults, although clinical signs vary a lot (1). However, the term “ichthyosis” is actually the gen-

eral name for skin disease characterized by excess scaling. Many different types of ichthyosis can affect dogs and humans (2; 3). Ichthyosis occurs when the skin has a decreased ability to act as a barrier. This can be because of a problem in environment (such as fleas or a bacterial infection) and/or a genetic defect (2). Ichthyosis caused by

a genetic defect is known as primary ichthyosis. In both people and dogs, mutations in many different genes can cause ichthyosis (2; 3). As noted above, this new type of Golden Retriever ichthyosis is reported by the research team and breeders to cause more significant skin disease than GR ICH1. This new form of Golden Retriever ichthyosis is caused by a different genetic mutation than one causing GR ICH1 and is cur-



*A special needs Golden with this condition who is living a good life with care from her devoted owner.*

rently known as GR ICH2.

Breeders may want to consider this new information when deciding whether or not to use the DNA test for the new type of ichthyosis now available from PennGen. Carriers for this condition can be bred but should only be bred to dogs who are normal/clear for GR ICH2. As with other recessive conditions, **automatic exclusion of carriers from breeding based on carrier status for recessive genes is not recommended; genetic diversity is important for breed preservation.**

Additional recommendations from the GRCA Health & Genetics Committee will be based upon peer-reviewed data when available. ♦

### References

1. Grall A, Guaguere E, Planchais S, Grond S, Bourrat E, et al. 2012. PNPLA1 mutations cause autosomal recessive congenital ichthyosis in golden retriever dogs and humans. *Nat Genet* 44:140-7
2. Mauldin EA. 2013. Canine ichthyosis and related disorders of cornification. *Vet Clin North Am Small Anim Pract* 43:89-97
3. Takeichi T, Akiyama M. 2016. Inherited ichthyosis: Non-syndromic forms. *J Dermatol* 43:242-51

# Yet Another Ichthyosis...

by Sharon MacDermott

My name is Sharon MacDermott and I've been breeding Golden Retrievers for a very long time with the kennel name Sunkyst. Some of you know me, many do not. I would like to tell my story of producing four litters that were affected with ichthyosis ...and not the GR ICH1, the form of ichthyosis we test for using the current ichthyosis test. I do DNA testing for this and other Golden Retriever diseases.

It all began close to seven years ago when I produced two puppies that were somewhat reddened in color and had a crackly appearance to their skin. They were more dense in consistency compared to their littermates. This denseness appeared to be fluid, but nothing compared to what is known as anasarca. They were normal in every way, except for this tightness of skin and slight swelling caused by this fluid. The face appeared puffy, making the eyes look deeper with orbital swelling. The crackle appearance of the skin was just that. It looked somewhat like mosaic or cracked glass. Unfortunately, I lost these two shortly after a planned c-section. At the time of this breeding and whelp, we were not doing DNA testing with our dogs for GR ICH1 or other diseases.

It wasn't until four years later, that the same skin problem appeared again. This time, out of 10 puppies in the litter, only one was affected. This puppy had crackled skin and felt dense. She also had the minimal swelling like the two from my first litter. Both the sire and dam of this litter were clear of GR ICH1, since that testing was now available. This particular puppy developed dandruff at approximately 4 weeks of age and then a few weeks later, seborrhea on her neck, chest, under her front legs and on the back of her rear legs. No other medical issues were found on a veterinary exam. Since I was totally baffled by all this, I contacted the genetic lab that tested the sire and dam for ichthyosis. They immediately retested the sire, dam and the puppy. They all came back as clear. I let it go, believing it was some sort of freakish occurrence. However, I posted my concern on producing this in two separate litters in one of the Golden Retriever discussion groups, but no one claimed to have experienced this.

Another year went by and I had a litter of 11. The sire was a carrier of GR ICH1 and the dam was clear. If you know anything about genetics, this is perfectly acceptable when breeding and you will not produce affected offspring, only a percentage of clear and percentage of carrier. Out of the 11 puppies, two were affected and had the very same symptoms as the previous litters. Now I was very concerned and even more frustrated, for I consider myself a very ethical breeder, following all the guidelines set forth by the GRCA and also doing DNA testing. Why am I producing such puppies? I then presented this to the Golden Retriever discussion group on Facebook yet again, reaching out to anyone for answers from those who had ever encountered this with their puppies. I received a private message from another Golden breeder who was experiencing the same thing. She too was very concerned and wanted answers. It is the most disheartening feeling to place affected puppies into the arms of their new families at time of pickup, knowing there will be extra time, care and expense required for these puppies. The one amazing aspect of these affected puppies is that they don't scratch as if itchy and never develop hot spots and have the most

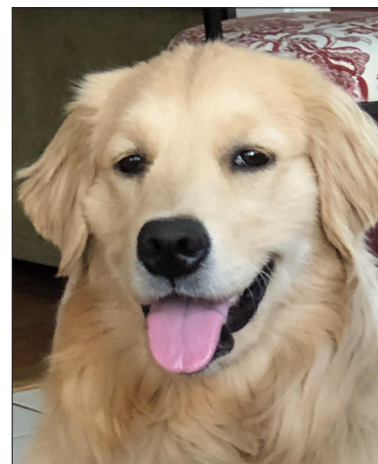
amazing coats. Go figure! It was at this time that out of pure frustration, I contacted the genetic lab I was using and begged for help. They had no answers but directed me to someone who could help.

This is when my journey into new discovery began.

Blood was sent from all affected puppies, littermates, parents and grandparents (if available). This required lots of hard work to convince families to send blood samples from their puppies/dogs, so the Golden community could find answers to why affected puppies were being produced — but I knew there was a problem and we needed answers. DNA was then extracted and testing began. Five other Golden breeders with similarly affected puppies graciously jumped on board and submitted DNA on their affected Golden Retrievers. I was so grateful that others helped with this research. The more samples we had, the better the hope we would find answers. Thank you all so much — I am forever grateful. But there were no answers at the time my fourth litter of affected puppies was produced. This litter had yet the most affected puppies produced in a litter. Half the litter was affected. Out of 10 puppies, five were affected with this dreadful skin disease.

Research now has provided answers. There *is* in fact another mutation which affects our wonderful breed and causes ichthyosis. Please understand that this new type of ichthyosis isn't just simple dandruff. This new form of ichthyosis is far more complex than GR ICH1. This form of ichthyosis causes not only dandruff, but seborrhea; and the care for affected Golden Retrievers is more involved, with extra time being spent on weekly baths, use of mousse every other day and omega fatty acids. There is no special food out there, so they have continued what I have recommended from day one. These families are by far the true heroes in my book. To dedicate so much time and effort to the care of these special needs puppies/dogs is nothing short of amazing. I have been told by the families of these affected puppies/dogs that they are the best dog and/or Golden they have ever had, and the extra care is minor compared to the love they receive.

Thank you for allowing me to tell my story. I hope you all better understand the nature of this new disease. Now that the recessive mutation that causes this disease has been discovered, testing of our dogs, when it becomes available, will allow this disease to be eradicated simply by avoiding breeding two healthy carriers to each other. The oldest affected puppy from my litters is now six years of age, and he and the others, to this day, don't suffer from any other medical issues but occasional ear and eye infections. Their owners worked hard to be sure they have wonderful lives, while dedicated breeders worked with talented researchers so that a simple test can prevent future puppies from facing the challenges of this previously unknown disease. ❖



***A much loved special needs Golden Retriever who has faced the challenge of this new condition.***