

Liver Shunt in Scotties

Periodically, members of the Health Trust are approached by Scottie owners for information and recommendations on how to manage various health issues in our breed. We always try to answer these questions to the best of our current knowledge, and in many cases, we pledge to find out more. One such health issue that continues to be of concern for breeders is the risk of producing a puppy with a liver shunt. Some breeders are still not sure what this disease is, what it means for them, and how can they avoid it.

In her article entitled “Portosystemic Shunt (Liver Shunt) in the Scottish Terrier” published in *Bagpiper* #3, 1999, former STCA Health Trust Chairman and Health Committee member Linda Orsborn provided a comprehensive and excellent article for us on the topic. Linda researched thoroughly, took the time to interview scientists and owners of affected puppies, and shared her own personal experience with a liver shunt puppy named “Holly”. This article has stood the test of time, and today, 19 years later, it still is an excellent source of information on this devastating genetic disorder.

Linda wrote: *“This is a rare condition and not a widespread problem in the Scottish Terrier at this time. Unfortunately, genetic disorders in dogs can spread rapidly. Two factors, in particular, influence the spread of a genetic disease: 1. a lack of understanding about the disease and its symptoms; and 2. the widespread use of carrier dogs or bitches, well respected in the area of conformation, for breeding purposes.”*

These are critical points. They deserve our attention again in 2018, perhaps even more now than in 1999, when the words were written.

What is a liver shunt, and what are the symptoms?

The *portal vein* is a large vein that collects blood from the systemic circulation and carries it into the liver where toxins and other byproducts are removed. A liver shunt occurs when an abnormal connection exists between the portal vein and another vein allowing blood to bypass or *shunt* around the liver, thereby preventing the liver from metabolizing nutrients and removing wastes. In the majority of cases, a liver shunt is caused by a birth defect called a ***congenital portosystemic shunt***. In some cases, multiple small shunts form in an older dog because of severe liver disease such as ***cirrhosis***. These are referred to as ***acquired portosystemic shunts***.

Puppies born with a liver shunt are typically small and unthrifty. They often exhibit bizarre, neurological behavior, especially after eating, due to the build-up of wastes in the blood. The first step in diagnosis is a Bile Acid test which assesses the function of the liver. Follow-up ultrasound examination will reveal the abnormal shunting of blood flow and confirm the location and type of shunt. Surgery may correct the condition depending on the location of the shunt or shunts, but this procedure requires great skill and is very expensive.

What are the genetics behind liver shunt?

In separate research done on Cairns, Yorkshire Terriers, Tibetan Spaniels, Miniature Schnauzers and other high risk breeds, Dr. Karen Tobias at the University of Tennessee and Dr. Sharon Center at Cornell University concluded that the inheritance of liver shunt traits (Portosystemic Vascular Anomaly and the related Hepatic Microvascular Dysplasia) cannot be explained by simple autosomal recessive inheritance. In fact, it is now accepted that the expression of this complex disorder is “polygenic” or controlled by multiple genes. Some of these genes directly

interfere with the normal process of blood vessel formation in and around the liver, while others act to modify the expression of the trait thus causing 'incomplete penetrance' of the defect. Furthermore, the effect of environmental factors on the severity of the defect is not known, nor is the role of management of the dam during early pregnancy, possibly leading to changes in early developmental genes of the fetus.

In other words, with liver shunt, we most likely are not dealing with a single genetic or point mutation that can be easily identified, tracked phenotypically, predicted genetically, and tested for in our breeding stock like the simple test we do for vWD. Instead, there could be many DNA (gene) point mutations, deletions or inversions that all have a role in the occurrence of a shunt. The sire and the dam both contribute DNA segments and gene mutations to produce a puppy like Linda's puppy, Holly. Liver shunt is complex, difficult to predict, and at the present time, research has been inconclusive and essentially non-existent for Scottish Terriers.

What does it mean for a Scottie breeder?

For the individual breeding program, a liver shunt puppy represents emotional heartbreak, enormous monetary expense, and at the very least, an uncertain breeding program. For Scottish Terriers as a whole, liver shunt, if it becomes widespread genetically, can have far-reaching significance because of the increase in frequency of the causative genetic factors within the breed or population.

You might ask, "If it is rare and takes so many genetic variables to fall into place, why worry about it?" We need to worry about liver shunt because those genetic factors responsible for shunt are carried forward from both parents to the next generation. Generation after generation, the potential for producing shunt is preserved and spread. Genetic mutations do not fade away. They stay in the genetic make-up of each Scottie and are passed along to the next generations. Like a time-bomb waiting to go off, the right combination of genes from a breeding pair can produce unexpected results in the puppies.

Adding to the concern is the overuse of a 'Popular Sire' within a population, thus shrinking genetic diversity. We are all together in the small genetic pool of the Scottish Terrier, and what each one of us does individually can have a ripple effect realized by all. The repeated use of a tiny fraction of popular breeding males results in an over-representation of that one dog's genetic influence, good and bad, for generations to come. Furthermore, if that Popular Sire is a known producer of liver shunt, and if he and his offspring are used repeatedly in a population, the incidence of liver shunt can rise dramatically. Yes, it takes two to tango, and the contribution of the bitch is also important. But Popular Sires by definition are used extensively and bred into a wide diversity of bitch lines. In the future, liver shunts could appear suddenly in larger numbers than ever thought possible as the genetic factors required to produce a shunt are spread widely, deeply and silently through the population. This is exactly what happened in some breeds like the Yorkshire Terrier, where the incidence of liver shunt is unacceptably high.

Isn't a normal Bile Acid (BA) test result enough to clear a Scottie of liver shunt?

An **abnormal** bile acid test result is indicative of a liver function disorder, like a shunt, and it serves as a good screening test for more diagnostic work to pinpoint the problem. On the other hand, a **normal** bile acid test result tells you that the liver of the dog is functioning normally at that time. However, it does not tell you if the dog is carrying any of the causative genetic factors for producing a shunt, nor does it guarantee that the test result will remain normal for the life of the dog.

What can we do to avoid liver shunt?

- **Education is key.** The more we all understand about liver shunts, the better. We have available to us a wealth of resources to learn about what a shunt is, how we can recognize it in a litter, and what it means for the life of that puppy. We all need to educate ourselves by starting with Linda's article in the ScottiePhile collection on the www.STCA.biz website. For more detailed information on symptoms, diagnosis, and treatment, go to the University of Tennessee School of Veterinary Medicine website at <https://vetmed.tennessee.edu/vmc/SmallAnimalHospital/Documents/Shunts/MVD-Brochure.pdf> . Here you can download the excellent brochure entitled "A Basic Overview of Portosystemic Shunts" by Dr. Karen Tobias.
- **Get a diagnosis.** If you have the misfortune of producing a liver shunt puppy, be sure to get a definitive diagnosis using a Bile Acid test, ultrasound with Doppler, and other imaging modalities recommended by your veterinarian.
- **Necropsy:** If your puppy does not survive, be sure to have a necropsy done by a competent veterinary diagnostic lab. An excellent place to send a puppy for a necropsy is Cornell University College of Veterinary Medicine, Animal Health Diagnostic center. Your veterinarian has to make the contact for you and obtain instructions for shipping a refrigerated (not frozen) puppy for this postmortem exam. These exams typically can run up to \$200 or more plus the overnight shipping, but each situation varies depending on locality.
- **Contact the HTF.** A vital mission of the STCA's Health Trust is surveillance of health issues and tracking the health history of our breed. ***Please contact the HTF regarding your experience and provide confirmation of the diagnosis.*** Your information will be handled with confidentiality and respect and used to further the identification of the causal agents behind what appears to be a genetic defect.
- **Preserve genetic material.** Current technology allows us to easily preserve our breed's genetic heritage. We understand how devastating it is to lose a puppy, but we still ask that you consider contributing samples from affected puppies and family members to the genetic database of our breed. The HTF is currently finalizing plans for a DNA bank, and we hope soon to provide assistance and instructions to Scottie owners for the collection of samples, both tissue and blood, for DNA collection and preservation. This will be the case for any genetic disorder in our breed, such as CA or Cramp, and your contribution will allow this precious, irreplaceable material to be available for future Scottish Terrier research. **Please contact the HTF for more information.**
- **Stop the blame game.** No breeder ever wants to produce a problem. Genetic diseases and faults pop up for all breeders, at one time or another. Rather than blame and accuse, we need to be kind to one another and work together to educate, inform and avoid genetic diseases to the best of our ability. Remember, we are all in this together.
- **If you decide to continue with family lines known to carry the causative genetic factors for liver shunt:** Understand what you are doing and what it can mean for the litters on the way and the future of breed. Exercise full disclosure with the dog or bitch owners who come to you for breeding. Proceed with caution and be prepared to monitor offspring and pay for BA tests. Dr. Jerold Bell states that for polygenic diseases like liver shunt, the **breadth** of the pedigree (siblings, cousins, aunts and uncles), is more important than the **depth** of the pedigree (sire and dam, grandsires and granddams). Dr.

Bell suggests that if there are multiple generations of normalcy in the **breadth** of the pedigree, then there is less risk for the causative genetic factors to be carried forward.

Breeding is a gamble in any case. We all must make our breeding decisions with care and knowledge. Some breeders might decide to use the BA normal offspring of known shunt producers, or the siblings of affected Scotties, or even the siblings of known shunt producers. As in so many cases with breeding dogs, we must ask ourselves, is it worth the risk? The breeders who have experienced this disease and seen the suffering that a puppy must endure, would more than likely answer no.

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