

Cystinuria and Factor VII Deficiency in Scottish Deerhounds: Studies Underway for Talk at National Specialty in May 2010

There are very few Deerhound breeders who have not produced at least one dog affected by either cystinuria or Factor VII deficiency—two disease-predisposing conditions that are found in the breed. Unfortunately, there are still many unanswered questions about what having these conditions means for our dogs and for us as their breeders and owners. At the 2010 National Specialty we are going to try to tease through all of the available information by having Dr. Paula S. Henthorn, of the University of Pennsylvania (where many of us send our Factor VII and cystinuria tests), present the health seminar on these two subjects.

To be most beneficial to the breed, Dr. Henthorn's colleague, Dr. Urs Giger has offered to conduct two little studies between now and the show so Dr. Henthorn can report on them at the specialty. However, to do this, he needs people with dogs that have tested as affecteds for these conditions to help.

Cystinuria

Cystinuria is a defect of the kidneys where dogs don't reabsorb the amino acids cystine, ornithine, lysine, and arginine (collectively known as COLA) normally. Amino acids are the building blocks of proteins. The cystine in the urine can precipitate and when concentrated forms crystals and stones, which can cause life-threatening urinary blockages. For more information on cystinuria, see the "Health and Husbandry" column in the September/October 2003 *Claymore* or go to the "Health Issues" page at www.deerhound.org.

There is still a lot we don't know about cystinuria. There is a screening test for detecting cystine in urine, but some dogs don't appear to produce cystine consistently, so they might test negative but still have the condition. And some dogs test positive for cystine, yet never seem to have a problem, while others block repeatedly and need major surgery to prevent further blockages or expensive medication to reduce the amount of cystine produced. We still don't know why some dogs block and others don't. It appears that only males seem to be affected, but the mode of inheritance of cystinuria in Deerhounds is still unknown.

In people, who also get cystinuria, many individuals are able to avert forming stones by changing their diet. However, this has not proven to be as easy to do in dogs, so Dr. Giger would like to find out more about how a dog's diet can influence stone formation. **Thus, if you have a dog that has tested positive for cystine, please participate in the study to give us an idea on how diet affects urinary cystine excretion.**

The study is simple: all you have to do is change your dog's diet for a short period of time according to the study guidelines, collect and store a couple of urine samples both before and after the change, and send the samples and a completed form to PennGen.

So if you have a dog that has tested positive for cystine on the PennGen test, please consider enrolling your dog in this study. Your dog only has had to test positive, not form stones or have been blocked.

For more information, contact Dr. Giger at penngen@vet.upenn.edu.

Factor VII Deficiency

Factor VII deficiency is a bleeding disorder where one of the clotting factors, called Factor VII, is absent in affected Deerhounds. While this is a relatively mild hereditary bleeding disorder and treatments to stop bleeding are available, these dogs can be at risk of bleeding to death during surgery or if severely injured.

Unlike cystinuria, we have a genetic test for Factor VII deficiency that definitely tells us which dogs are affected or are asymptomatic carriers of this condition, which is caused by a simple recessive trait. However, we still are not entirely sure what having this disease actually means. Some dogs are affected, yet test normal on a clotting profile and undergo major surgery without a problem. Others bleed to death. So the purpose of this study is to dig deeper into the clotting profiles of affected dogs to try to determine the course of this disorder. If a dog tests normal on a clotting profile, is the normal clotting range accurate? Will the dog always test normal? Is there a correlation of the clotting profile results to the bleeding tendency?

This is important, because we have to determine just how much of a problem Factor VII deficiency is in our breed so we can best learn how to manage affected individuals, and depending on how severe the bleeding tendency turns out to be, it might influence breeding decisions.

To learn more, Dr. Giger would like to get results of clotting profiles (PT and PTT) from Factor VII-deficient dogs and relatives. The test can be done by your veterinarian, using their regular lab, or by PennGen. So if you have a Factor VII-deficient dog, please consider having a clotting profile done and sending the results to PennGen or sending the blood directly to PennGen, where they will do the clotting profile. If you've already done a clotting profile on your Factor VII-deficient dog, please send the results to PennGen.

For more information, contact Dr. Giger at penngen@vet.upenn.edu.

This is our opportunity to ask questions about cystinuria and Factor VII. While you can always ask a question at the seminar, you can also send questions to Betty Stephenson at the email or phone number below. These questions will be given to Dr. Henthorn before the specialty to facilitate coverage of these topics in his talk. So whether your question is about breeding strategies, managing the diseases, or whatever, please let Betty know.

This seminar is a joint project of the 2010 Specialty Committee and the newly formed SDCA Breeder's Education Committee, which has been charged by the Board to be a resource on breeding issues. For more information on the BEC, contact Dr. Betty Stephenson at kyleakindh@aol.com or at (270) 782-9224.

Thanks to Dr. Henthorn for speaking and to Dr. Giger for offering to do the studies, as well as for information provided and review of this article.

Miranda Levin