

## FACTOR VII DEFICIENCY—OUR FIRST GENETIC TEST

Printed in The Claymore

We've hit a milestone for our breed—the development of the first genetic test for a life-threatening disease which occurs in the Scottish Deerhound. Bleeding excessively after major surgery (like spaying) has long been known to be a problem in Deerhounds. As of March 2006, we know the reason for this: **Factor VII deficiency**. Factor VII is a blood clotting factor, and it's been known to affect Beagles since 1996. The team of researchers headed by Dr. Mary Beth Callan at University of Pennsylvania has recently identified the reason for the deficiency—a genetic mutation which is an autosomal recessive trait. We couldn't have been luckier to have been in the right place at the right time—a Deerhounder whose imported bitch was spayed and bled from the incision, requiring seven plasma transfusions to survive had both a veterinary team with the expertise to save the bitch and to send the blood to Cornell to try to identify the deficiency and, the wherewithal and desire to follow this through yielded a bonanza—the newly identified mutation in Beagles is the same in, at least, *this* Deerhound. Dr. Debbie Cutter, another Deerhounder, member of the Health and Genetics Committee and a geneticist herself read about the bleeding Deerhound on the list and did an internet search for a Factor VII deficiency test which might be available. One was listed, Dr. Mary Beth Callan's Beagle project. Dr. Callan, when contacted by Debbie, wanted more blood samples from possibly affected Deerhounds to determine if this was a widespread phenomenon. Debbie remembered that one of my bitches had nearly bled to death after spaying. Luckily, I had a 4.5cc aliquot of blood left from my Cailie, (Ch Kyleakin Marcail o'Jubalhil, CD) who bled excessively after I spayed her in 1999, responded to a blood transfusion and second surgery, and later died of osteosarcoma. Dr. Callan was able to run the test on 1cc of my stored blood so I could keep the rest for future research efforts, and she, too, was homozygous for the recessive mutation (i.e. she had two copies of the gene). That means she had to have acquired the genes from both parents, and so Factor VII deficiency in Scottish Deerhounds must also occur in this country with fair regularity (Cailie's parents came from two entirely different lines). I sent blood on one of her offspring, and he turned out, as expected, to be an unaffected carrier. A third dog, related to Cailie by sharing her sire in addition to a common pair of great grandparents on her dam's side, tested clear.

### What does this mean to breeders?

First, let me reiterate what I've said before about autosomal recessive disorders: they're the best thing we could have. **Genetic testing isn't about eliminating animals from your breeding program. It's about selecting mates for them so affected offspring are not created.** With autosomal recessive traits, carrier animals can be bred to clear animals, and the next generation's individuals can be selected from to avoid the gene entirely if possible, and if not, at least they aren't affected, and the following generation can be selected clear. Even affected individuals can be bred if they are bred to non-carrier animals and no affected puppies will be produced.

The genetic test for Factor VII deficiency is available. It can be used before breeding any bitch to any stud to avoid the possibility of inadvertently producing another bleeder. It can be used before spaying any bitch so an intelligent decision can be made about whether to spay her or to spay her with clotting factors on hand to use if needed. This article also includes the information disseminated by the research team working on this disorder. It describes the disorder, its variable presentation, and a little additional information about the research and researchers. I believe, from anecdotal evidence spanning several decades, this disorder is fairly common and fairly severe in our breed. I have heard numerous stories from breeders across the board about bitches bleeding to death after major surgery, often being diagnosed as "DIC" (disseminated intravascular coagulation) when no causative factor could be identified. Blood samples from individuals (or their close relatives) which appear to have bled excessively would be welcomed by Dr. Callan.

[Test Submission Form](#)  
[Description](#)

Or go to the following website:

[http://w3.vet.upenn.edu/research/centers/pennngen/forms/factorVIIdeficiency\\_ScottishDeerhounds.htm](http://w3.vet.upenn.edu/research/centers/pennngen/forms/factorVIIdeficiency_ScottishDeerhounds.htm)