

Undescended Testicles (Cryptorchidism) Research

November, 2008

There are 2 research groups studying the problem of cryptorchidism (retained testicles) in Shelties. Both research projects are sponsored by the Canine Health Foundation and the ASSA. Although both studies concern the same topic, the investigators are using different approaches to study the problem. **Breeders are urged to contribute to both studies if possible.**

Dr. Agoulnik, at Baylor Medical School, (Canine Health Foundation Grant No. 882-A) is using a “global” approach to find genes associated with cryptorchidism by looking at larger strands of DNA to find an association with the trait and further pinpointing the genes from there. Because he needs a large quantity of good quality DNA for his work, he must use blood samples or testicular tissue rather than DNA collected using cheek brushes.

Dr. Rothschild, a geneticist at Iowa State University, (Canine Health Foundation Grant No. 1018-A) has been studying cryptorchidism in species other than dogs. He is looking to see if the genes that cause the problem in man, pigs, and mice also are associated with the problem in dogs. The fact that DNA from cheek brush samples can be used for this study makes it easy for owners to collect the DNA samples themselves.

Both studies are described below:

Undescended Testicles (Cryptorchidism) Research

Canine Health Foundation Grant No. 882-A

Cryptorchidism or retained testicles is the most common birth defect in purebred dogs. Two major health consequences of cryptorchidism are infertility at adulthood and significantly increased risk of testicular cancer. Because of the cancer risk, the retained testicles should be removed. Currently, there is no genetic diagnostic test to predict the risk of this disease in a dog's progeny. The main objective of this study is to develop a DNA test that may predict the risk of a dog to produce cryptorchidism in its male offspring. Blood samples or tissues samples (testicles) removed during surgery from cryptorchid dogs and their normal littermates are needed for the current DNA research sponsored by AKC CHF. The information generated by this study can be used to identify animals with the mutant gene and thus will provide breeders with informative breeding recommendations.

Samples are needed from cryptorchid Shelties and their normal male littermates. (Samples from normal male littermates are not required, but would be helpful.)

Material needed for participation:

- 2-6 cc of whole blood in purple topped EDTA tubes **or**
- Frozen testicular tissue removed during castration
- AKC registration number and/or pedigree
- Consent form (obtained from Dr. AgoulNIK)

This should all be sent over night with a cold pack to:

Alexander I. AgoulNIK, Ph.D.
Associate Professor
Department of Obstetrics and Gynecology
Baylor College of Medicine
One Baylor Plaza,
Room BCMD-207D, MS BCM612
Houston, TX 77030

tel. 1-713-798-6087

fax. 1-713-798-5074

[e-mail: agoulNIK@bcm.edu](mailto:agoulNIK@bcm.edu)

Prior to collecting the samples, please notify Dr AgoulNIK to request the consent form and shipping instructions. If you explain the purpose of the blood draw, most veterinarians will draw the blood sample at no charge or for a reduced amount. The study will pay for shipping costs. We sincerely thank everybody who helps with our research aimed to benefit all dog breeders.

Comparative Gene Discovery for Canine Cryptorchidism

Canine Health Foundation Grant No. 1018-A

Dr. Rothschild, the primary investigator, is a geneticist at Iowa State University. He has been studying cryptorchidism in species other than canine. He is looking to see if the genes that cause the problem in man, pigs, and mice also are associated with the problem in dogs. He is currently working with Siberian Huskies but is banking DNA from other breeds.

After being contacted by the ASSA Research Advisory Committee, he agreed to include Shelties in his study. He would be greatly appreciative and could put the samples to good use. Because obtaining sufficient samples from the right sets of dogs (see below) has been a slow, time consuming process with the Siberian Huskies, he asked that one of the ASSA representatives

collect the samples and then send them to him once the needed numbers have been obtained. Mary Mahaffey, DVM is willing to take the lead on this. This is what is needed for the study:

- Cheek brush DNA samples from 20 groups of the following: sire, dam, at least one normal male pup and one cryptorchid pup. (Grandparents would be good also if obtainable).
- Pedigree of the puppies.

The fact that cheek brush samples rather than blood samples are used in this study makes it easier to obtain the samples as owners can collect the DNA themselves. Obtaining 20 combinations as described above may not be easy, but can be done. DNA collected on cheek brushes can be kept indefinitely as long as the brushes are dry and kept in paper envelopes. Once 20 of the above combinations are obtained, all will be sent to Dr. Rothschild at the same time.

If the owners wish to keep the information confidential, put all the materials for each litter in a large envelope and indicate on the outside of the envelope that DNA samples of the sire, dam, affected and unaffected male offspring are included inside (without listing the dog names). Then put that envelope inside a larger one and mail it to Mary Mahaffey, DVM at the address below.

To participate in the study, contact Mary Mahaffey and she will send you DNA collection kits and instructions.

Mary Mahaffey, DVM
1611 Cedar Road
Watkinsville, GA 30677

E-mail address: lastly3345@bellsouth.net
