Glanzmann's Thrombasthenia in Great Pyrenees dogs

A bleeding disorder called Glanzmann's thrombasthenia (GT) was recognized and described in a Great Pyrenees dog in 1996.[1] GT has been recognized for many years in humans and is due to a congenital/inherited membrane defect in platelets. Platelets are small, circulating cytoplasmic fragments that are the first line of defense in stopping the flow of blood from injured blood vessels. An important aspect of platelet function is their ability to stick to each other and plug holes in damaged vessels until blood clotting and tissue repair can occur. The platelets of people and dogs with GT are defective in their ability to stick to each other. Therefore, these individuals are at increased risk for spontaneous hemorrhage and they are also at high risk for excessive hemorrhage as a result of injury or surgery. The type of spontaneous bleeding that occurs with GT includes excessive gingival bleeding during tooth eruption, nose bleeds, and superficial skin bleeds. Young dogs less than 18 months of age are especially prone to excessive, spontaneous bleeding. The Great Pyrenees dog described in 1996 bled excessively during tooth eruption as a puppy and also had nose bleeds until the age of 23 months. She continued to have minor bruising and skin hemorrhages throughout her life. A major obstacle to identifying other dogs with GT was the necessity of performing highly specialized functional and biochemical diagnostic tests and also the necessity of patients being on the premises during these studies. Until a few years ago, the disease could not be diagnosed without bringing the dog to the testing facility. In addition, carriers of the disease could not be readily identified by these methods. In early 1999, the gene that encodes for one of the proteins defective on the platelet surface in GT was sequenced and the molecular basis for the disease was determined.[2] By using DNA testing, affected and carrier animals can now be identified by simply submitting a blood sample through the mail. By using DNA testing, families of Great Pyrenees dogs carrying the mutation for GT have been identified in Illinois, Indiana, and Florida and individual dogs have been identified in Oklahoma, Minnesota, Missouri, and Mississippi. Carrier detection is vital in controlling spread of inherited defects and DNA testing is the only reliable method of detecting these animals.

1. Boudreaux MK, Kvam K, Dillon AR, Bourne C, Scott M, Schwartz KA, Toivio-Kinnucan M. Type I Glanzmann's Thrombasthenia in a Great Pyrenees Dog. Veterinary Pathology 33:503-511, 1996.
2. Lipscomb DL, Bourne C, Boudreaux MK: Two genetic defects in alpha IIb are associated with Type I GT in a Great Pyrenees dog: a 14-base insertion in exon 13 and a splicing defect of intron 13. Veterinary Pathology 37:581-588, 2000.

The sample required for testing for GT in Great Pyrenees dogs is a 2 ml EDTA tube (purple top) containing at least 1 ml of whole blood. Care should be taken to not cross contaminate samples during collection, particularly if more than one dog is collected at the same time. Samples should be labeled clearly so that there is no confusion regarding sample identification. Samples should be kept cold (ice packs) and shipped overnight to the address below. Take care to make sure tubes are protected well to prevent breakage during shipping. Please do not ship on Friday or the day before a holiday. The fee for testing is $100 per sample. **Make checks payable to: Auburn University, Department of Pathobiology.**

Please provide the following information on each dog being tested:

Name and AKC Registration Number _____

Male or Female (Circle one)

Age at time of sampling or Date of Birth _____

AKC Registration Number of Sire _____

AKC Registration Number of Dam _____

I am hereby requesting this sample be tested for the 14-base pair insertion mutation causing Type I Glanzmann's thrombasthenia in Great Pyrenees dogs. I understand that my individual test results will only be released to me. I certify that I am the owner of this dog. I understand and agree that the results of this test may be confidentially combined with those of other owners and used in aggregate result form for research purposes including publication. I understand in aggregate result form my individual results will not be identifiable specifically to my dog. I release Dr. Boudreaux and any associates working with her and Auburn University from all liability regarding this sample.

_____ _____

Owner's Signature **Date**

_____ _____

Owner's Name (print clearly or type) **Telephone number**

Address Results should be sent to: _____

_____

_____

Send samples to:

Mary K. Boudreaux, DVM, PhD
Department of Pathobiology
166 Greene Hall
College of Veterinary Medicine
Auburn University, Alabama 36849-5519
(334) 844-2692