Glanzmann’s Thrombasthenia in Otterhounds

A bleeding disorder called Thrombasthenic thrombopathia was first described in Otterhounds in 1967.\(^1\) Affected dogs had mucosal bleeding and prolonged bleeding times. Platelet aggregation responses were minimal or lacking and clot retraction was severely impaired. Abnormal clot retraction tests and platelet aggregation studies were used as screening tests in an effort to eliminate Thrombasthenic thrombopathia from the Otterhound breed in the 70’s and 80’s. As a result of this testing, it was thought that the platelet disorder had been largely eliminated from the breed, however, in the early 1990’s, descendents of the dogs originally described with Thrombasthenic thrombopathia were identified with platelet dysfunction. Because Thrombasthenic thrombopathia closely resembled the Glanzmann’s thrombasthenia platelet disorder that had been described in Great Pyrenees dogs in 1996 and 2000,\(^2,3\) molecular studies were begun in Dr. Boudreaux’s laboratory to determine whether a mutation could be found that caused Thrombasthenic thrombopathia in Otterhounds. Blood samples were collected from the affected Otterhounds identified by Dr. James Catalfamo at Cornell University in the early 1990’s. As a result of this work, it was determined that Otterhound Thrombasthenic thrombopathia was identical to Glanzmann’s thrombasthenia (GT),\(^4\) based on the finding that affected Otterhounds had a mutation in the gene encoding for platelet glycoprotein I\(\text{IIb}\) (mutations in either of the genes encoding for glycoproteins I\(\text{IIb}\) or III\(\text{a}\) have been documented to cause GT in human beings). Although Otterhounds and Great Pyrenees both have mutations in the gene encoding for platelet glycoprotein I\(\text{IIb}\), and therefore both breeds are affected with GT, the mutations that cause the disease are different for the two breeds. The mutation causing GT in Otterhounds is in Exon 12 while the mutation causing GT in Great Pyrenees is in Exon 13.

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.

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, carriers can be accurately identified before breeding to avoid spreading the mutation and to avoid producing affected puppies. Carrier detection is vital in controlling spread of inherited defects and DNA testing is the only reliable method of detecting these animals.

The sample required for testing for GT in Otterhounds 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 single base pair change in Exon 12 causing Type I Glanzmann’s thrombasthenia in Otterhounds. 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.

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**Owner’s Signature**  
**Date**

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

**Address Results should be sent to:**

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Send samples to:  
Mary K. Boudreaux, DVM, PhD  
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College of Veterinary Medicine  
Auburn University, Alabama  36849-5519  
(334) 844-2692