

## Basset Hound Thrombopathia

Basset Hound Thrombopathia (BHT) was first described by Johnstone and Lotz in 1979. Since that time investigators at Cornell, Michigan State, and Auburn University have studied the disorder in affected and carrier Basset hounds in attempts to understand the functional, biochemical, and molecular bases for the disorder. The disorder is due to a congenital/inherited signal transduction 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 dogs with BHT are defective in their ability to stick to each other due to the inability of the platelets to transmit internal signals properly. 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. Affected Basset hounds experience spontaneous mucosal type bleeding (including gingival bleeding, particularly during permanent tooth eruption, gastrointestinal bleeding, urinary tract bleeding, and nose bleeds), and petechial and ecchymotic hemorrhages of the skin (bruising that can range from small, pinpoint lesions to lesions as large as a half-dollar or larger) and hemorrhage at the tips of the ears secondary to trauma. Gastrointestinal bleeding may or may not be apparent. If bleeding is severe, the stools will appear black and tarry. Gastrointestinal bleeds can also be slow and insidious (microscopic and not visibly apparent) resulting in iron deficiency anemia with time.

For many years the disease could not be diagnosed without bringing dogs to a testing facility that specialized in studying platelet function disorders in animals. Although these methods were accurate in diagnosing affected dogs, the methods could not readily identify carriers of the disease. Carrier detection is vital in controlling spread of inherited defects and DNA testing is the only reliable method of detecting these animals. During the summer of 2006, the molecular basis for BHT was determined at Auburn University. A mutation was found in a gene that encodes for a signal transduction protein vitally important in transmitting signals that result in normal platelet aggregation and granule release. By using DNA testing, affected and carrier Basset hounds can now be identified by submitting a blood sample through the mail.

- **Johnstone IB, Lotz F: An inherited platelet function defect in Basset hounds Can Vet J 20:211-215, 1979.**
- **Catalfamo JL, Raymond SL, White JG, Dodds WJ: Defective platelet-fibrinogen interaction in hereditary canine thrombopathia. Blood 67:1568-1577, 1986.**
- **Boudreaux MK, Dodds WJ, Slauson DO, Catalfamo JL: Evidence for regulatory control of canine platelet phosphodiesterase. Biochem Biophys Res Commun 140(2):580-594, 1986.**
- **Boudreaux MK, Dodds WJ, Slauson DO, Catalfamo JL: Impaired cAMP metabolism associated with abnormal function of thrombopathic canine platelets. Biochem Biophys Res Commun 140:595-601, 1986.**
- **Patterson WR, Estry DW, Schwartz KA, Borchert RD, Bell TG: Absent platelet aggregation with normal fibrinogen binding in Basset hound hereditary thrombopathy. Thromb Haemostas 62(3):1011-1015, 1989.**
- **Boudreaux MK, Catalfamo JL, Klok M: Calcium-diacylglycerol guanine nucleotide exchange factor I gene mutations associated with loss of function in canine platelets. Transl Res 150(2):81-92, 2007.**

The sample required for testing for BHT 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 shipped to the address below. Take care to make sure tubes are protected well to prevent breakage during shipping. The fee for testing is \$100 per sample.

**Make checks payable to: Auburn University, Department of Pathobiology.**

**Basset Hound Thrombopathia Test Form**

Please provide the following information on each dog being tested:

Name and Registration Number \_\_\_\_\_

Male or Female (Circle one)

Age at time of sampling or Date of Birth \_\_\_\_\_

Name and Registration Number of Sire \_\_\_\_\_

Name and Registration Number of Dam \_\_\_\_\_

I am hereby requesting this sample be tested for the mutation described as causing Basset hound Thrombopathia in Basset hounds. 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)

\_\_\_\_\_  
Email address/ 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

email: [boudrmk@auburn.edu](mailto:boudrmk@auburn.edu)

FAX: (334) 844-2652

The fee for testing is \$100 per sample. Sample is EDTA whole blood (1 ml).

**Make checks payable to: Auburn University, Department of Pathobiology.**

Turnaround time for results is typically 3 to 5 working days.