Spitz Thrombopathia

Spitz Thrombopathia was first described in 1994. The disorder closely resembles the disorder described in Basset hounds called Basset hound Thrombopathia and 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 Spitz Thrombopathia 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. The affected Spitz dogs described in 1994 experienced nose bleeds, gastrointestinal hemorrhage, gingival bleeding, shifting leg lameness, and hematomas. Other clinical signs to be aware of include skin bruising and excessive bleeding of the gingiva during permanent tooth eruption. 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.

Until recently, 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 Spitz Thrombopathia 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 Spitz dogs can now be identified by simply submitting a blood sample through the mail.

Boudreaux MK, Crager C, Dillon AR, Stanz K, Toivio-Kinnucan M: Identification of an intrinsic platelet function disorder in Spitz dogs. J Vet Int Med 8(2):93-98, 1994.

The sample required for testing for Spitz Thrombopathia 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 (use ice packs; do not freeze the samples) 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 mutation described as causing Spitz Thrombopathia in Eskimo Spitz 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:

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