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 2006 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 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.

Boudreaux MK, Catalfamo JL, Klok M: Calcium-diacylglycerol guanine nucleotide exchange factor I gene mutations associated with loss of function in canine platelets. Translational Res 150(2):81-92, 2007.

Specimen requirements: At least 1ml EDTA whole blood (purple top tube). Do not cross contaminate samples during collection particularly if more than one dog is collected at the same time. Label all specimens clearly. Protect the tubes to prevent breakage during shipping. All methods of shipping are acceptable. **Blood samples do not require ice**.

Ship to: Hemostasis Laboratory, Dr. Peter W. Christopherson 166 Greene Hall Auburn University, AL 36849-5519

Fee for testing: \$130.00 (payment options listed below)

Invoice payments are due within 30 days from the invoice date and can be made securely online: https://www.aub.ie/payinvoice, by mailed check payable to: Pathobiology Diagnostic Services, or through wire transfer (email weldolm@auburn.edu for wire transfer instructions)

Questions regarding invoicing and/or payments: weldolm@auburn.edu



Hemostasis Laboratory Department of Pathobiology Dr. Peter W. Christopherson, DVM, PhD, DACVP 166 Greene Hall Auburn University, AL 36849-5519 PH: 334-844-2797 / 334-844-2697, Fax: 334-844-2652 Email: <u>chrispw@auburn.edu</u>

OFFICE USE ONLY

ACCESSION

DATE

HEMOSTASIS LABORATORY

Spitz Thrombopathia

SAMPLE DATE:

AGE AT TIME OF SAMPLING OR DATE OF BIRTH:

ANIMAL NAME:

BREED:

SEX: MALE F

FEMALE

ANIMAL REGISTRATION NUMBER (if applicable):

NAME OF SIRE (if applicable):

REGISTRATION NUMBER OF SIRE (if applicable):

NAME OF DAM (if applicable):

REGISTRATION OF DAM (if applicable):

PERTINENT HISTORY:

OWNER INFORMATION	VETEI	VETERINARIAN'S INFORMATION (BILLING INFORMATION)	
	(В		
	REFERRING VETERINAR	REFERRING VETERINARIAN	
NAME	CLINIC		
ADDRESS	ADDRESS		
CITY/TOWN	CITY/TOWN		
PROVINCE	PROVINCE		
POSTAL CODE	POSTAL CODE	COUNTRY	
COUNTRY	PHONE	FAX	
PHONE	EMAIL		
	FAX RESULTS	EMAIL RESULTS	

RESULTS (if you would like the results sent to additional emails and/or faxes please list below)

EMAIL 1:	FAX 1:
EMAIL 2:	FAX 2:

SPECIMEN REQUIREMENTS: EDTA WHOLE BLOOD (1ML)

TURNAROUND TIME FOR RESULTS: TYPICALLY 8 TO 10 WORKING DAYS UPON

ARRIVAL HARD COPIES OF REPORTS AVAILABLE UPON REQUEST