Congenital Macrothrombocytopenia

Cavalier King Charles Spaniels (CKCS) have a high prevalence (30 to 50% of dogs in the United States) of a macrothrombocytopenia that is inherited as an autosomal trait. The disorder is characterized by platelet numbers ranging between 50,000 and 100,000/µl with many of the circulating platelets being larger than normal. Platelet counts performed using "in house" instrumentation by veterinarians can potentially be even lower than 50,000/uL depending on the methodology of the particular counting instrument. Affected dogs do not have a bleeding diathesis; however, the existence of low platelet numbers can be confused with and must be distinguished from acquired causes of thrombocytopenia including thrombocytopenias secondary to infectious agents, consumption, medications, immune-mediated causes, and others. Unfortunately, CKCS have received inappropriate treatment with antibiotics, steroids, or other medications because of confusion or lack of awareness of this disorder by veterinarians. A molecular assay developed at Auburn University can confirm an inherited cause for macrothrombocytopenia to help veterinarians distinguish inherited from acquired macrothrombocytopenia in canine patients with low platelet counts.

Two mutations have been identified in the gene encoding beta1-tubulin, a protein involved in platelet production by megakaryocytes, that are responsible for congenital macrothrombocytopenia observed in CKCS [1] and in Norfolk and Cairn Terriers [2]. Since the identification of the mutation in CKCS, the identical mutation has also been documented in other breeds of dogs with congenital macrothrombocytopenia including Chihuahua, Labrador retriever, Poodle, English Toy Spaniel, Labradoodle, Shih Tzu, Maltese, Jack Russell, Havanese, Boxer, Cocker Spaniel, and Bichon. Other breeds are likely to be identified with this mutation, and congenital macrothrombocytopenia should be suspected in any dog that has a persistently low platelet count in the absence of history or evidence of abnormal bleeding and is non-responsive to treatment with antibiotics or steroids. DNA assays are available at Auburn University to determine the presence or absence of these mutations.

1. Davis B, Toivio-Kinnucan M, Schuller S, Boudreaux MK. Mutation in Beta1-tubulin correlates with macrothrombocytopenia in Cavalier King Charles Spaniels. J Vet Int Med 22:540-545, 2008.

2. Gelain ME, Bertazzolo W, Tutino G, Pogliani E, Cian F, Boudreaux MK. A novel point mutation in the β1-tubulin gene in asymptomatic macrothrombocytopenic Norfolk and Cairn Terriers. Vet Clin Pathol 43(3):317-21, 2014.

Specimen requirements: At least 1ml EDTA whole blood (purple top tube). Please also include 3 to 5 blood smears fixed in methanol (Diff-Quik #1). 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, 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

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Dr. Peter W. Christopherson, DVM, PhD, DACVP
166 Greene Hall
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OFFICE USE ONLY

ACCESSION _____

DATE _____

HEMOSTASIS LABORATORY

Congenital Macrothrombocytopenia (Beta1-tubulin)

SAMPLE DATE: _____ AGE AT TIME OF SAMPLING OR DATE OF BIRTH: _____

ANIMAL NAME: _____ BREED: _____ SEX: ___ MALE ___ FEMALE

ANIMAL REGISTRATION NUMBER (if applicable): _____

PERTINENT HISTORY:

OWNER INFORMATION	VETERINARIAN'S INFORMATION (BILLING INFORMATION)
	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

SAVE AS A PDF RESET FORM

06/27/2023