

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/ μ L 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 β 1-tubulin gene in asymptomatic macrothrombocytopenic Norfolk and Cairn Terriers. Vet Clin Pathol 43(3):317-21, 2014.

The sample required for testing for congenital macrothrombocytopenia is a 2 ml EDTA tube (purple top) containing at least 1 ml of whole blood. Please also include 3 to 5 unstained blood smears. 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. Take care to make sure tubes and slides 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.**
Congenital Macrothrombocytopenia Test Form

This document should be used when submitting samples for testing.

Please provide the following information on each dog being tested:

Name and AKC Registration Number _____
(if available)

Breed _____

Male or Female (Circle one)

Age at time of sampling or Date of Birth _____

Owner's Name (print clearly) _____

Date _____

Veterinarian/Requester Telephone number _____

Veterinarian/Requester Email address _____

**Name and Address Results
should be sent to:
(print clearly or type)** _____

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
FAX: (334) 844-2652

The fee for testing is \$100 per sample.

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

Sample is EDTA whole blood (1 ml) and 3 to 5 unstained blood smears.

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