

Macrothrombocytopenia of Cavalier King Charles Spaniels

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/ul with many of the circulating platelets being larger than normal. 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 that would confirm an inherited cause for macrothrombocytopenia would help veterinarians distinguish inherited from acquired macrothrombocytopenia in a particular animal.

During the summer of 2007, a mutation was identified in the gene that encodes beta1-tubulin, a protein involved in platelet production by megakaryocytes, that correlates with the macrothrombocytopenia observed in CKCS. A DNA assay is available at Auburn University to determine the presence or absence of this mutation. The following document should be used when submitting samples for testing:

The sample required for testing for inherited macrothrombocytopenia in CKCS 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 CKCS 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 macrothrombocytopenia in Cavalier King Charles Spaniels. 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/Email

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