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. 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 is responsible for the inherited macrothrombocytopenia observed in CKCS[1]. Since the identification of this mutation in CKCS, the identical mutation has also been documented in other breeds of dogs with inherited macrothrombocytopenia including Chihuahua, Labrador retriever, Poodle, English Toy Spaniel, Labradoodle, Shih Tzu, Maltese, and Jack Russell. Other breeds are likely to be identified with this mutation and inherited macrothrombocytopenia should be suspected in any dog that has a persistently low platelet count that is non-responsive to treatment with antibiotics or steroids and has no history or evidence of abnormal bleeding. A DNA assay is available at Auburn University to determine the presence or absence of this mutation.

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.

The sample required for testing for inherited macrothrombocytopenia in 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.**

The following 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 or type) _____

Date _____

Veterinarian/Requester Telephone number _____

Veterinarian/Requester Email address _____

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

_____

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Send samples to:

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