

Landseer Thrombopathia

Landseers of European Continental Type (ECT) have been described with a bleeding disorder secondary to platelet dysfunction. 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 Landseers with 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 dogs are at increased risk for spontaneous hemorrhage and they are also at high risk for excessive hemorrhage as a result of injury or surgery. Affected Landseers experience spontaneous mucosal type bleeding (including gingival bleeding, particularly during permanent tooth eruption, gastrointestinal bleeding, urinary tract bleeding, and nose bleeds), and petechial and ecchymotic hemorrhages of the skin (bruising that can range from small, pinpoint lesions to lesions as large as a half-dollar or larger). Skin lesions on the abdomen can best be observed where the hair tends to be thinner. 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 recently, 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 thrombopathia in Landseers was determined at Auburn University. A mutation was found in the gene that encodes for CalDAG-GEFI, 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 Landseers can now be identified by submitting a blood sample through the mail.

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.

The sample required for testing for thrombopathia in Landseers 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 shipped to the address below. Take care to make sure tubes are protected well to prevent breakage during shipping. The fee for testing is \$100 per sample. **Follow instructions for International shipping (see International Shipping Information link). Make checks payable to: Auburn University, Department of Pathobiology or contact Lori Carden (weldolm@vetmed.auburn.edu) for Bank Transfer information.**

Please provide the following information on each dog being tested (print clearly):

Name _____

Studbook _____

Number (nr) _____

Male or Female (Circle one)

Age at time of sampling or Date of Birth _____

Name, Studbook, and Number of Sire _____

Name, Studbook, and Number of Dam _____

I am hereby requesting this sample be tested for the mutation described as causing Thrombopathia in Landseers. 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) email address

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

email: boudrmk@auburn.edu FAX: (334) 844-2652
Sample is EDTA whole blood (1 ml). The fee for testing is \$100 per sample
Contact Lori Carden (weldolm@vetmed.auburn.edu) for Bank Transfer information.