Von Willebrand's Disease (vWD) Type 2; German Pointer marker test

In brief
The clotting pathway is a complex process. The vWF glycoprotein complexes with factor VIII and both are required for platelet adhesion and preventing the rapid clearance of factor VIII. Von Willebrand’s disease (vWD) is a bleeding disorder affecting multiple breeds and several genetic variants have been characterised. Type 2 vWD affects several breeds, but the mutation tested here has been found to only be predictive of the risk of developing the condition in German Shorthaired Pointers and German Wirehaired Pointers. The disease causes moderate to severe bleeding tendency due to low level and abnormal structure of von Willebrand’s factor. The mode of inheritance is autosomal recessive.

Clinical overview
Type 2 von Willebrand’s disease is a moderate to severe clotting disorder. Typical symptoms include mucosal bleeding, such as epistaxis, bleeding from the gums, gastrointestinal bleeding, and blood in the urine. Exceptionally excessive and prolonged bleeding may be observed after a trauma or surgery, and spontaneous bleeding may also be encountered. This condition can lead to death if untreated.

References

Online database
Online Mendelian Inheritance in Animals, OMIA (http://omia.angis.org.au/). Faculty of Veterinary Science, University of Sydney; OMIA001339-9615.

Scientific articles
