Macrothrombocytopenia; disease-linked variant originally found in Norfolk and Cairn Terrier

In brief
Congenital macrothrombocytopenia is a blood disorder characterised by oversized platelets and a low platelet count. The causative mutation was originally identified in the Cavalier King Charles Spaniel. The same mutation is known to cause macrothrombocytopenia in several other breeds as well. A similar mutation has also been identified in Norfolk Terriers and Cairn Terriers. Congenital macrothrombocytopenia is inherited as an autosomal recessive trait.

Clinical overview
Platelets, also called thrombocytes, are produced in the bone marrow. Platelets play an important role in the blood clotting system when a blood vessel is injured. Congenital macrothrombocytopenia is characterised by oversized platelets, also called macroplatelets, and an abnormally low number of platelets. Macroplatelets function normally and the affected dogs do not exhibit any health problems due to either the size or the fewer numbers of its platelets. Macrothrombocytopenia is not associated with spontaneous bleeding; however, it is important not to mistake macrothrombocytopenia for other more severe conditions characterised by low platelet counts.

References

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

Scientific articles