

# VETERINARY TECHNICAL DATASHEET

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



Mutation Found In :Cairn Terrier, Norfolk Terrier

## Disorder Type

- Blood

## Disease Severity

- Mild

## Background

Congenital macrothrombocytopenia is a blood disorder characterized by oversized platelets and a low platelet count. Platelets, also called thrombocytes, are produced in the bone marrow and play an important role in the blood clotting system when a blood vessel is injured. The original mutation was first identified in the Cavalier King Charles Spaniel and is known to cause macrothrombocytopenia in several other breeds as well. This particular mutation variant however has been identified in Norfolk Terriers and Cairn Terriers.

## Key Signs

- Low platelet count
- Macroplatelets

## Clinical Description

Congenital macrothrombocytopenia is characterized 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 lower platelet count. Macrothrombocytopenia is not associated with spontaneous bleeding.

## Mode of Inheritance

- autosomal recessive

## Gene Name

- TUBB1

## Next Steps

While this disorder is relatively mild, it is important not to mistake macrothrombocytopenia for other more severe conditions characterized by low platelet counts.

## References

Gelain ME, Tutino GF, Pogliani E, Bertazzolo W. Macrothrombocytopenia in a group of related Norfolk terriers. Vet Rec 167:493-4, 2010.