

VETERINARY TECHNICAL DATASHEET

Hereditary Elliptocytosis



Mutation Found In :Chow Chow, Labrador Retriever

Disorder Type

- Blood

Disease Severity

- Mild/moderate

Background

Hereditary elliptocytosis is characterized by abnormally shaped red blood cells or erythrocytes in the blood. Abnormal cells have an oval shape instead of the characteristic biconcave shape of canine erythrocytes. Abnormally shaped red blood cells are called elliptocytes or ovalocytes. The genetic background of canine congenital elliptocytosis remains poorly understood. Most typically, canine hereditary elliptocytosis is encountered as an asymptomatic condition that is detected during examination of the dog for an unrelated disease.

Key Signs

- Abnormal oval shaped red blood cells
- Possible hemolytic anemia

Clinical Description

Only one genetic mutation associated with elliptocytosis has been previously described. In a case study on one dog, a Labrador and Chow Chow mixed breed, the patient presented with persistent elliptocytosis, decreased mechanical deformability of erythrocytes and decreased erythrocyte membrane stability. Molecularly, elliptocytosis was found to be due to a defect in the erythrocyte membrane protein beta-spectrin. The studied dog was found to carry one copy of a mutation in the beta-spectrin encoding gene. Further information on the mutation is needed to examine whether dogs with two copies of the mutation have a more severe hemolytic, elliptocytic anemia.

Mode of Inheritance

- autosomal dominant

Gene Name

- SPTB

Next Steps

It is advised that dogs with this condition have regular veterinary check-ups for monitoring purposes and supportive treatment as needed.

References

Di Terlizzi R, Gallagher PG, Mohandas N, Steiner LA, Dolce KS, Guo X, Wilkerson MJ, Stockham SL. Canine elliptocytosis due to a mutant beta-spectrin. Vet Clin Pathol 38(1):52-8, 2009.