Hereditary Elliptocytosis

In brief

Hereditary elliptocytosis is characterised 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.

Clinical overview

Most typically, canine hereditary elliptocytosis is encountered as an asymptomatic condition that is detected during examination of the dog for an unrelated disease. Only one genetic mutation associated with elliptocytosis was previously described. In a case study on one dog, a Labrador x 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 β-spectrin. The studied dog was found to be heterozygous for a mutation in the β-spectrin encoding gene. Further information on the mutation is needed to examine whether homozygosity for the mutation leads to a more severe hemolytic, elliptocytic anaemia.

References

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

Scientific articles


Disease severity
Mild

Clinical signs
- Abnormal oval shaped red blood cells
- Possible hemolytic anemia

Mode of Inheritance

Results of the genetic test are reported as follows:
- Clear
- At risk

Mutation(s) found in:
Chow Chow
Golden Labrador Retriever
Labrador Retriever