VETERINARY TECHNICAL DATASHEET

Intestinal Cobalamin Malabsorption or Imerslund-Gräsbeck Syndrome, (IGS); mutation originally found in Border Collie

Mutation Found In :Border Collie

Disorder Type

- Metabolic

Disease Severity

- Moderate

Background

Intestinal cobalamin malabsorption or Imerslund-Gräsbeck syndrome (IGS) is a metabolic disorder encountered in several breeds. Cobalamin or vitamin B12 is required for the normal function of many enzymes. The underlying cause for intestinal cobalamin malabsorption is a defect in cobalamin receptors in the ileum. Cobalamin is stored in the body before birth, but once those stores are consumed early in life, cobalamin must to be acquired from food.

Key Signs

- Growth retardation
- Anemia
- Neutropenia
- Loss of appetite

Clinical Description

Initial signs of intestinal cobalamin malabsorption can be seen in puppies 6 to 12 weeks of age, when cobalamin store become depleted. Puppies with IGS suffer from weakness and loss of appetite and fail to grow normally Bloodwork shows anemia, neutropenia, and low cobalamin concentrations. High levels of homocysteine and methylmalonic acid can also be observed in the blood. Proteinuria is typically present.

Mode of Inheritance

- autosomal recessive

Gene Name

- CUBN

Next Steps

Treatment consists of regular vitamin B12 supplementation; injection provides rapid absorption but oral vitamin B12 may also be used.

References

Owczarek-Lipska M, Jagannathan V, Drögemüller C, Lutz S, Glanemann B, Leeb T, Kook PH. A frameshift mutation in the cubilin gene (CUBN) in Border Collies with Imerslund-Gräsbeck syndrome (selective cobalamin malabsorption). PLoS One 8:e61144, 2013.