VETERINARY TECHNICAL DATASHEET

Glycogen Storage Disease Type Ia, (GSD Ia)

Mutation Found In :Maltese

Disorder Type

- Metabolic

Disease Severity

- Severe

Background

Glycogen storage disease type Ia (GSD Ia) is a severe metabolic disorder encountered in the Maltese. GSD Ia-affected dogs have a congenital deficiency in the enzyme glucose-6-phosphatase required for breakdown of glycogen. Glycogen is a large polysaccharide that serves as energy storage in the body and an energy source for cells. Insufficient enzyme function causes cellular accumulation of glycogen especially in the liver and impairment of endogenous glucose production.

Key Signs

- Hepatomegaly
- Hypoglycemia
- Lactic acidosis
- Hyperuricemia
- Coma
- Death

Clinical Description

GSD Ia is characterized by hypoglycemia (low blood sugar). Failure of glucose metabolism causes glycogen accumulation leading to hypoglycemia, lactic acidosis causing low blood pH, coma, and death. Affected puppies are weak and they rarely survive more than a few weeks.

Mode of Inheritance

- autosomal recessive

Gene Name

- G6PC

Next Steps

Management of affected puppies consists of a high-starch diet fed frequently throughout the day to control hypoglycemia with only small amounts of protein and fat in the diet to provide required nutrients. Experimental gene therapy has successfully treated this disorder in dogs.

References

Kishnani PS, Bao Y, Wu JY, Brix AE, Lin JL, Chen YT. Isolation and nucleotide sequence of canine glucose-6-phosphatase mRNA: identification of mutation in puppies with glycogen storage disease type Ia. Biochem Mol Med 61(2):168-177, 1997.