VETERINARY TECHNICAL DATASHEET

Glycogen Storage Disease Type IIIa, (GSD IIIa); mutation originally found in Curly Coated Retriever

Mutation Found In :Curly Coated Retriever

Disorder Type

- Metabolic

Disease Severity

- Moderate

Background

Glycogen storage disease type IIIa (GSD IIIa) is a disorder of glycogen metabolism first identified in Curly Coated Retrievers. GSD IIIa affected dogs have a congenital deficiency in glycogen debranching enzyme activity, which is 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

- Fatigue
- Exercise intolerance
- and Episodic hypoglycemic collapse

Clinical Description

The clinical signs of GSD IIIa include fatigue, exercise intolerance, and hypoglycemic collapses caused by low blood sugar. Characteristic signs can typically be observed at around 14 months of age.

Mode of Inheritance

- autosomal recessive

Gene Name

- AGL

Next Steps

Management of affected puppies consists of a high-starch diet to prevent hypoglycemia at young ages and high-protein diets which may have a positive effect on cardiomyopathy associated with GSD IIIa. The use of medium-chain triglycerides has also shown positive therapeutic effects. Dietary therapies, however, do not prevent long-term complications of GSD IIIa.

References

Gregory BL, Shelton GD, Bali DS, Chen YT, Fyfe JC. Glycogen storage disease type IIIa in curly-coated retrievers. J Vet Intern Med 21:40-6, 2007.