VETERINARY TECHNICAL DATASHEET

Alexander Disease; mutation originally found in Labrador Retriever

Mutation Found In :Labrador Retriever

Disorder Type

- Nervous system

Disease Severity

- Severe

Background

Alexander disease is a rare, fatal, hereditary neurological disease. The disorder causes progressively worsening weakness of voluntary movement in all limbs. The disorder is caused by the dysfunction of astrocyte cells in the central nervous system.

Key Signs

- Progressive tetraparesis
- Regurgitation
- Myoclonic jerks

Clinical Description

Clinical signs of this disorder emerge around three months of age and begin with weakness of movement in the limbs, causing a spastic swimming-puppy-like position of the front legs. The dog can also develop other neurological signs, such as mild vestibular signs and myoclonic jerks. The chest of affected puppies can be flat and regurgitation can occur. The disorder is progressive and in a few weeks, the limb weakness during voluntary movement progresses to where the dog is unable to stand. The disease should be a differential diagnosis based on breed history and clinical presentation when other possible causes have been ruled out. MRI and electrodiagnostic tests can be used for further diagnostics.

Mode of Inheritance

- autosomal recessive

Gene Name

- GFAP

Next Steps

The disorder is progressive and there is no cure. Treatment is supportive and the prognosis is poor.

References

Van Poucke M, Martlé V, Van Brantegem L, Ducatelle R, Van Ham L, Bhatti S, Peelman LJ. A canine orthologue of the human GFAP c.716G>A (p.Arg239His) variant causes Alexander disease in a Labrador retriever. Eur J Hum Genet. 2016 Jun;24(6):852-6. doi: 10.1038/ejhg.2015.223. Epub 2015 Oct 21.