VETERINARY TECHNICAL DATASHEET

GM2 Gangliosidosis; mutation originally found in Japanese Chin

Mutation Found In :Japanese Chin

Disorder Type

- Neuromuscular

Disease Severity

- Severe

Background

GM2 gangliosidosis, is a lysosomal storage disease causing progressive degeneration of nervous tissue. The disorder is known to affect Japanese Chins and is also known as Tay-Sachs disease. A related condition is also seen in Toy Poodles and Golden Retrievers but the underlying mutation is still unknown in Golden Retrievers.

Key Signs

- Ataxia
- Intention tremors

Clinical Description

First signs of GM2 gangliosidosis are usually observed at one year of age. Characteristic signs include ataxia (uncoordinated movements), intention tremors of the head, vision impairment, altered mental status, and feeding difficulties. GM2 gangliosidosis is a progressive condition, so affected dogs will have increasing difficultly with everyday tasks such as walking and going up stairs, and may need assistance. Affected dogs are usually euthanized on welfare grounds by two years of age.

Mode of Inheritance

- autosomal recessive

Gene Name

- HEXA

Next Steps

Upon initial observation of clinical signs, affected dogs should be closely monitored to assess welfare, and devise a supportive care treatment plan. As clinically signs are progressive, affected dogs are usually euthanized on welfare grounds by 2 years of age.

References

GM2 gangliosidosis associated with a HEXA missense mutation in Japanese Chin dogs: a potential model for Tay Sachs disease.