VETERINARY TECHNICAL DATASHEET

Hereditary Ataxia or Cerebellar Ataxia; mutation originally found in Old English Sheepdog and Gordon Setter

WISDOM HEALTH™

Mutation Found In :Gordon Setter, Old English Sheepdog

Disorder Type

- Nervous system

Disease Severity

- Moderate

Background

Cerebellar ataxias are a widely varying group of disorders characterized by neuronal degeneration of the cerebellar cortex and are also known as cerebellar abiotrophy or cerebellar cortical degeneration in dogs. Degeneration of the cerebellar nerve cells leads to progressive ataxia (lack of normal coordination of movement). The onset of clinical signs and pattern of progression varies in different forms of the condition. The onset of clinical signs can be in puppyhood, at a young age, or in adulthood depending on the form of the cerebellar ataxia. Hereditary cerebellar ataxia in Old English Sheepdogs and Gordon Setters is a slowly progressing, early-onset form of hereditary ataxia caused by a mutation in RAB24 gene.

Key Signs

- Progressive ataxia
- Hypermetria
- Intention tremors
- Wide-based stance

Clinical Description

In Old English Sheepdogs, the first signs of cerebellar ataxia are usually observed at 6 months to 4 years of age. The onset of clinical signs in Gordon Setters is usually at the age of 6 to 10 months. Characteristic signs include ataxia (uncoordinated movements) and hypermetria (overreaching movements). Intention tremors, wide-based stance, and nystagmus (involuntary eye movements) are also observed on occasion. Cerebellar ataxia is a slowly progressing condition in both breeds.

Mode of Inheritance

- autosomal recessive

Gene Name

- RAB24

Next Steps

Upon initial observation of clinical signs, affected dogs should be monitored as the condition progresses and practical advice should be given to ensure quality of life is maintained. For instance as the disease progresses affected dogs may find going up and down stairs and moving on smooth surfaces challenging, so assistance may be needed.

References

Agler C, Nielsen DM, Urkasemsin G, Singleton A, Tonomura N, Sigurdsson S, Tang R, Linder K, Arepalli S, Hernandez D, Lindblad-Toh, K, van de Leemput J, Motsinger-Reif A, O'Brien, DP, Bell J, Harris T, Steinberg S, Olby N.J Canine Hereditary Ataxia in Old English Sheepdogs and Gordon Setters Is Associated with a Defect in the Autophagy Gene Encoding RAB24. PLoS Genet 10:e1003991, 2014.