

VETERINARY TECHNICAL DATASHEET

Spongy Degeneration with Cerebellar Ataxia, (SDCA); mutation originally found in Belgian Shepherd



Mutation Found In :Belgian Sheepdog, Belgian Malinois

Disorder Type

- Nervous system

Disease Severity

- Moderate/severe

Background

Spongy Degeneration with Cerebellar Ataxia is similar to a rare neurological disorder in humans, SeSAME (Seizures, sensorineural deafness, ataxia, mental retardation, and electrolyte imbalance) syndrome. The disease occurs in Belgium Shepherds, especially the Malinois type.

Key Signs

- Ataxia
- Myokymia
- Neuromyotonia seizures

Clinical Description

The age of onset of SDCA is usually 6-8 weeks. The first observable sign is poor coordination of movements (ataxia). Affected dogs may also suffer from episodic myokymia and/or seizures. Myokymia is characterized by uncontrollable twitching of the muscles that tends to run through a muscle in waves. Myokymia episodes can be precipitated by exercise or excitement. The condition can lead to neuromyotonia (generalized muscle stiffness). Neuromyotonia episodes are incapacitating attacks during which the dog becomes rigid and falls down. Affected dogs remain aware of their surroundings during neuromyotonia attacks and are at risk of overheating. Long-term prognosis of this disorder is poor.

Mode of Inheritance

- autosomal recessive

Gene Name

- SDCA1

Next Steps

There is no curative treatment for the disease. Antiepileptic drugs can be used to decrease seizure activity but long-term prognosis is still poor.

References

Mauri N, Kleiter M, Leschnik M, Högler S, Dietschi E, Wiedmer M, Dietrich J, Henke D, Steffen F, Schuller S, Gurtner C, Stokar-Regenscheit N, O'Toole D, Bilzer T, Herden C, Oevermann A, Jagannathan V, Leeb T. A Missense Variant in KCNJ10 in Belgian Shepherd Dogs Affected by Spongy Degeneration with Cerebellar Ataxia (SDCA1). *G3* (Bethesda). 2017 Feb; 7(2): 663–669. Published online 2016 Dec 21. doi: 10.1534/g3.116.038455 PMID: PMC5295610