VETERINARY TECHNICAL DATASHEET

Cerebellar Hypoplasia; mutation originally found in Eurasier

Mutation Found In :Eurasier

Disorder Type

- Nervous system

Disease Severity

- Moderate/severe

Background

Cerebellar hypoplasia in Eurasier dogs resembles the human Dandy-Walker malformation. The disease results from the cerebellum, a part of the brain, not being formed properly and the condition is not curable. Several other breeds have been shown to exhibit a similar condition but it is not known if the causal mutation in the Eurasier is responsible for the condition in these other breeds.

Key Signs

- Ataxia
- Epileptic seizures
- Cerebellar hypoplasia

Clinical Description

Clinical signs of cerebellar hypoplasia include ataxia of varying severity, from mild truncal sway and subtly uncoordinated gait to severe cerebellar ataxia and falling or rolling. Some dogs may exhibit epileptic seizures, nystagmus, or tremors. Clinical signs are present from birth and are typically noticed when puppies start to walk. Affected dogs will lack a menace reflex. The most severely affected puppies are usually euthanized. Some dogs exhibit only minor ataxia in adulthood.

Mode of Inheritance

- autosomal recessive

Gene Name

- VLDLR

Next Steps

Treatment is supportive care and symptomatic depending on the severity of the dog's ataxia.

References

Gerber M, Fischer A, Jagannathan V, Drögemuller M, Drögemuller C, Schmidt M, Bernandino F, Manz E, Matiasek K, Rentmeister K, Leeb T. A deletion in the VLDLR gene in Eurasier dogs with cerebellar hypoplasia resembling a Dandy-Walker-like malformation (DWLM). PLoS ONE 10(2): e0108917, 2015.

Bernardino F, Rentmeister K, Schmidt M, Bruehschwein A, Matiasek K, Matiasek L, Lauda A, Schoon H, Fischer A. Inferior cerebellar hypoplasia resembling a Dandy-Walker like malformation in purebred Eurasier dogs with familial non-progressive ataxia: a retrospective and prospective clinical cohort study. PLoS ONE 10(2): e0117670, 2015.