VETERINARY TECHNICAL DATASHEET

Neuronal Ceroid Lipofuscinosis 8, (NCL8); mutation originally found in Australian Shepherd

Mutation Found In :Australian Shepherd

Disorder Type

- Nervous system

Disease Severity

- Severe

Background

Neuronal ceroid lipofuscinoses (NCLs) are a group of inherited progressive neurodegenerative lysosomal storage disorders. Neuronal ceroid lipofuscinoses are characterized by excessive accumulation of lipofuscin and ceroid lipopigments in the central nervous system and other tissues. Different forms of NCLs differ by age of onset and pattern of progression. Usually progressive loss of vision is the first observable sign. In addition, the clinical signs of NCLs include ataxia (uncoordinated movements), seizures, and behavioral changes, such as aggression. NCL type 8 is encountered in English Setters, though this particular variant has been described in the Australian Shepherd.

Key Signs

- Vision impairment
- Ataxia
- Behavioral changes
- Seizures

Clinical Description

The first signs of NCL8 are usually observed at the age of 1 to 2 years. The clinical signs include ataxia (uncoordinated movements), behavioral changes, vision loss, and epileptic seizures. NCL8 is a progressive condition. The lifespan of affected dogs is rarely over two years of age due to severity of the clinical signs.

Mode of Inheritance

- autosomal recessive

Gene Name

- CLN8

Next Steps

Treatment is supportive care, however, due to the progressive nature of the condition, clinical signs typically lead to euthanasia on welfare grounds.

References

Guo J, Johnson GS, Brown HA, Provencher ML, da Costa RC, Mhlanga-Mutangadura T, Taylor JF, Schnabel RD, O'Brien DP, Katz ML. A CLN8 nonsense mutation in the whole genome sequence of a mixed breed dog with neuronal ceroid lipofuscinosis and Australian Shepherd ancestry. Mol Genet Metab 112:302-9, 2014.