VETERINARY TECHNICAL DATASHEET

Neuronal Ceroid Lipofuscinosis 10, (NCL10); mutation originally found in American Bulldog

Mutation Found In :Bulldog (American)

Disorder Type

- Nervous system

Disease Severity

- Moderate/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. Though progressive loss of vision is usually the first observable sign, and is followed by other brain-related signs, NCL type 10 significantly differs from the other forms of NCL as the clinical signs are all related to locomotion. NCL10 is encountered in the American Bulldog.

Key Signs

- Progressive ataxia
- Hypermetria
- Muscle weakness
- Paraparesis

Clinical Description

Type 10 neuronal ceroid lipofuscinosis differs from the other forms of NCL because it doesn't impair the vision of affected dogs. In addition, affected dogs don't suffer from clinical signs associated with brain function, such as seizures or behavioral changes. The first signs of NCL10 can be observed at around 1.5 years of age. The clinical signs include progressive ataxia (uncoordinated movements) and hypermetria (overreaching movements). Affected dogs suffer from pelvic limb weakness progressing to weakness of all limbs. NCL10 is a slowly progressing disorder leading to euthanasia or premature death before 7 years of age.

Mode of Inheritance

- autosomal recessive

Gene Name

- CTSD

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

Awano T, Katz ML, O'Brien DP, Taylor JF, Evans J, Khan S, Sohar I, Lobel P, Johnson GS. A mutation in the cathepsin D gene (CTSD) in American Bulldogs with neuronal ceroid lipofuscinosis. Mol Genet Metab 87:341-348, 2006.