VETERINARY TECHNICAL DATASHEET

Neuronal Ceroid Lipofuscinosis 1, (NCL1); mutation originally found in Dachshund

Mutation Found In :Dachshund (Shorthaired), Dachshund (Miniature Shorthaired), Dachshund (Wirehaired), Dachshund (Miniature Wirehaired), Dachshund (Longhaired), Dachshund (Miniature Longhaired)

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. Type 1 form of NCL is encountered in Dachshunds.

Key Signs

- Vision impairment
- Ataxia
- Behavioral changes
- Seizures

Clinical Description

The onset of clinical signs is usually by 9 months of age. NCL type 1 is a progressive condition characterized by rapidly progressing vision impairment, ataxia (uncontrolled movements), and behavioral changes, such as anxiety, sound sensitivity, and inability to recognize familiar individuals. Clinical signs of NCL1 also include general weakness and uncontrolled rhythmic head movements.

Mode of Inheritance

- autosomal recessive

Gene Name

- PPT1

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

Sanders DN, Farias FH, Johnson GS, Chiang V, Cook JR, O'Brien DP, Hofmann SL, Lu JY, Katz ML. A mutation in canine PPT1 causes early onset neuronal ceroid lipofuscinosis in a Dachshund. Mol Genet Metab 100(4):349-56, 2010.