Neuronal Ceroid Lipofuscinoses Type 6, (NCL6)

In brief

Neuronal ceroid lipofuscinoses (NCLs) are a group of inherited progressive neurodegenerative lysosomal storage disorders. Neuronal ceroid lipofuscinoses are characterised 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 behavioural changes, such as aggression. NCL type 6 is encountered in the Australian Shepherd and is inherited in an autosomal recessive manner.

Clinical overview

The clinical signs of NCL6 can be observed at 1 1/2 years of age. The first clinical sign is usually vision impairment progressing to complete blindness in one month’s time. The clinical signs include also anxiety, sensitivity to touch and sound and ataxia (uncoordinated movements). NCL6 is a progressive condition. Affected dogs will not survive and they are usually euthanised after the onset of the clinical signs.

References

Online database
Online Mendelian Inheritance in Animals, OMIA (http://omia.angis.org.au/). Faculty of Veterinary Science, University of Sydney.

Scientific articles