

## VETERINARY TECHNICAL DATASHEET

Neuronal Ceroid Lipofuscinosis 7, (NCL7); mutation originally found in Chinese Crested Dog and Chihuahua



Mutation Found In :Chinese Crested, Chihuahua

### 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. Clinical signs may start with behavioral changes and develop into impairment of vision and degeneration of cognitive and motor skills. A causative mutation for NCL7 has been found in Chinese Crested dogs and the Chihuahua.

### Key Signs

- Progressive neurological signs
- Blindness
- Cognitive impairment
- Motor impairment

### Clinical Description

Affected dogs seem normal as puppies but develop progressive neurological signs around one year of age. First signs of the disease include exhibiting forms of compulsory behavior, such as excessive licking of a body part. As the disease progresses, affected dogs show further changes in behavior and develop motor and vision impairment. Affected dogs may be progressively more fearful and hyper-responsive to stimuli. Affected dogs may also develop epileptic seizures in later stages.

### Mode of Inheritance

- autosomal recessive

### Gene Name

- MFSD8

### 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, O'Brien D, Mhlanga-Mutangadura T, Olby N, Taylor J, Schnabel R, Katz M, Johnson G. A rare homozygous MFSD8 single-base-pair deletion and frameshift in the whole genome sequence of a Chinese Crested dog with neuronal ceroid lipofuscinosis. BMC Vet Res;10:960, 2015.