

NCL Description for Dalmatians

Age of onset of clinical signs: 6 months to 1 year

Age of death or euthanasia: 1.5 to 8 years

Abnormalities often observed by the owner:

Mental changes: Affected dogs become aggressive and affected females tend to cannibalize pups. There is a tendency toward self-mutilation and grinding of teeth. Affected dogs may run erratically.

Changes in gait and posture: Tremor and ataxia appeared starting at 15 to 20 months of age. At 22 months of age pronounced staggering and bumping into objects were noted.

Visual abnormalities: Visual impairment is one of the earliest signs with an onset at about six months of age. This is manifested as delayed visual reactions that caused affected dogs to bump into objects. This progresses to complete blindness.

Seizures/convulsions: Seizures occur starting at 15 to 20 months of age.

Other changes: Affected dogs consume considerably less food than unaffected peers.

Abnormalities observed upon clinical examinations:

Clinical neurologic changes: None reported other than above.

Clinical ophthalmic changes: No abnormalities reported on funduscopic examination.

Visual abnormalities: Affected dogs are functionally blind late in the disease.

Retinal changes: Gross morphology of the retina is relatively normal with preservation of all cell layers. However lipopigment accumulation was reported in cells of every retinal layer.

Electroretinography (ERG): None reported

Other clinical findings: None reported

Histopathology

Brain: Brain weights were reduced by about 35% relative to normal control Dalmatians. Autofluorescent granules were present in neuronal perikarya of the cerebral cortex, hippocampus, basal ganglia, and brainstem nuclei. The autofluorescence was yellow-green. The granular material appeared blue with the Luxol-Fast-blue stain, red with the PAS stain, black with the Sudan-black-B stain, and red with the Oil-red-O stain. At the ultrastructural level the disease-related intracellular inclusions had granular and membraneous-lamellar components and varied in fine structural detail. No particular ultrastructural type of pigment correlated with a particular type of cell.

Eyes: Cells in all layers of the retina contained membrane-bound inclusions that had varying ultrastructural appearances. The contents of the inclusions were described as being of the following types: lamellar, fingerprint, and curvilinear. Usually a cell contained only one of these types, but occasionally more than one type of inclusion was present in the same cell.

Other organs and structures: Autofluorescent storage bodies accumulated in most visceral tissues including the skin. These had similar ultrastructural appearances to those present in brain and retina. Skin biopsies showed increasing numbers of storage bodies with increasing age.

Mode of inheritance: Not reported. Likely autosomal recessive.

Gene containing mutation: Unknown.

References:

Goebel HH and Dahme E: Ultrastructure of retinal pigment epithelial and neural cells in the neuronal ceroid-lipofuscinosis affected Dalmatian dog (1986) *Retina* 6:179-187.

Jolly RD, Palmer DN, Studdert VP Sutton RH, Kelly WR, Koppang N, Dahme G, Hartley WJ, Patterson JS, and Riis RC: Canine ceroid-lipofuscinoses: A Review and classification. (1994) *J. Small Animal Prac.* 35:299-306.

Goebel HH. Bilzer T. Dahme E. Malkusch F: Morphological studies in canine (Dalmatian) neuronal ceroid-lipofuscinosis (1988) *American Journal of Medical Genetics* - Supplement. 5:127-39.