

NCL Description for Cocker Spaniels

Age of onset of clinical signs: 1.5 - 6 years

Age of death or euthanasia: 1.5 - 6 years

Abnormalities often observed by the owner:

Mental changes: Aggression, irritability, dementia

Changes in gait and posture: progressive difficulty walking; weakness and uncoordinated movement.

Visual abnormalities: blindness may be present in some cases

Seizures/convulsions: reported in some cases

Other changes: Jaw champing, head tremors, emaciation

Abnormalities observed upon clinical examinations:

Clinical neurologic changes: Gait abnormalities, weakness, uncoordinated movement (hypermetric ataxia, proprioceptive deficits, exaggerated spinal reflexes)

Clinical ophthalmic changes: ophthalmic changes have been reported

Visual abnormalities: blindness has been reported

Retinal changes: retinal atrophy has been reported

Electroretinography (ERG): not described in reported cases

Other clinical findings: none reported

Histopathology

Brain: Yellow-brown granules were present in neuronal cytoplasm of some neurons in the brain and spinal cord. Storage granules were most abundant in the spinal cord and cerebellum. Cells in the cerebral cortex, brain stem, and Purkinje cells were less affected. Affected neurons were swollen, and often had an eccentric nucleus due to displacement by accumulation of granules. These granules exhibited yellow-green autofluorescence and staining patterns consistent with ceroid and lipofuscin. Degenerative changes within the CNS were also described (neuronal necrosis, Wallerian degeneration, and axonal dystrophy).

Eyes: mild irregular loss of photoreceptor cells; massive accumulation of fluorescent material in the retinal pigmented epithelium.

Other organs and structures: Storage product also accumulated within the smooth muscle cells of the intestines, pancreas, urinary bladder, and walls of small arteries.

Mode of inheritance: Autosomal recessive inheritance is suspected.

Gene containing mutation: Unknown

References:

Minatel L, et al. Ceroid-lipofuscinosis in a Cocker Spaniel dog. *Veterinary Pathology* (2000); 37:488-490.

Jolly RD, et al. Generalised ceroid-lipofuscinosis and brown bowel syndrome in Cocker spaniel dogs. *New Zealand Veterinary Journal* (1994); 42:236-239.

Nimmo JS, et al. Neuronal and Generalized Ceroid-Lipofuscinosis in a Cocker Spaniel. *Veterinary Pathology* (1982); 19:623-628.