

## NCL Description for Chihuahuas

**Age of onset of clinical signs:** 13 - 21 months

**Age of death or euthanasia:** 2 years

**Abnormalities often observed by the owner:**

***Mental changes:*** aggression, decreased sense of smell

***Changes in gait and posture:*** circling

***Visual abnormalities:*** impaired vision

***Seizures/convulsions:*** none observed in the case reported

***Other changes:*** none reported

**Abnormalities observed upon clinical examinations:**

***Clinical neurologic changes:*** specific exam findings were not described

***Clinical ophthalmic changes:*** loss of pupillary light reflexes

***Visual abnormalities:*** visual dysfunction

***Retinal changes:*** normal retinal examination

***Electroretinography (ERG):*** not performed

***Other clinical findings:*** none reported

**Histopathology**

***Brain:*** Neuronal swelling throughout the CNS, with cytoplasmic accumulation of yellowish pigment granules (a storage product). This storage product demonstrates yellow-green autofluorescence, and stains in a pattern consistent with ceroid and lipofuscin. This storage product is thought to be a lipid-binding protein. Pigment deposition was most abundant in the neurons of the hippocampus and thalamus, and in the Purkinje cells of the cerebellum. Immunohistochemistry (GFAP) revealed astrocytosis; gemistocytes were present throughout the CNS, and laminar gliosis was present in the cerebral cortex. The cerebellum had loss of Purkinje cells, Bergmann glial proliferation, and a decreased molecular layer. Concurrent congenital hydrocephalus was also present in the case reported (dilated lateral and third ventricles, with open fontanelles); and no obstructive lesions were present.

***Eyes:*** Retinal neurons were also swollen and contained pigment granules. These pigments stained positive with periodic acid Schiff, Schmorl method for lipofuscin, and oil red O for lipid.

***Other organs and structures:*** no abnormalities were identified in other organs; no storage products were found in any tissues outside the CNS.

**Mode of inheritance:** Autosomal recessive inheritance is suspected.

**Gene containing mutation:** Unknown

**References:**

Kuwamura M, et al. Neuronal ceroid-lipofuscinosis and hydrocephalus in a Chihuahua. Journal of Small Animal Practice (2003); 44:227-230.