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: