NCL Description for Miniature Schnauzers

Age of on-set of clinical signs: 2 – 4 years

Age of euthanasia: 3 - 5 years

Abnormalities often observed by the owner:
***Visual abnormalities*:** The first signs of disease in this breed are visual problems. Initially there is a reluctance to climb stairs and inability to navigate in unknown surrounding; bumps into objects. The onset of blindness is rapid, within 5 months after initial visual problems.
***Mental changes*:** Confusion, unawareness of surroundings, loss of memory for tasks that are normally fulfilled, and aimless wandering.
***Changes in gait and posture*:** None reported.
Seizures/convulsions: Bouts of trembling, although these are not identified as seizures.
Other changes: None reported.

Abnormalities observed upon clinical examinations:.
***Clinical ophthalmic changes*:** Abnormal pupillary light reflexes; present but sluggish and do not constrict normally.
***Visual abnormalities*:** Moderate visual impairment to blindness. The visual problems progress rapidly.
***Retinal changes*:** Ophthalmoscopy shows a patchy granular appearance of the tapetal fundus, with changes in tapetal reflectivity (hypo- and hyperreflective areas), generalized slight vascular attenuation and optic disc pallor.
Electroretinography (ERG): Reduced scotopic responses, normal photopic responses.
Clinical neurologic changes: None found at examination.
***Other clinical findings*:** None reported.

Histopathology
***Brain*:** Extensive accumulation of autofluorescent storage material occurs in the cerebral cortex. Purkinje cells are swollen and vacuolated with a content of variable amounts of granular cytoplasmic material. There are clusters of glial cells with granular material around many of the blood vessels in the Purkinje cell layer and in other small vessels throughout the brain. Storage bodies label with antibodies to sphingolipid activator proteins (SAPs) A and D, but not with antibodies to mitochondrial ATP synthase subunit C. Ultrastructurally, the storage body contents are granular (not finger-print or observed in whorls, as in CL of many other breeds or species).
Eyes: Severe photoreceptor degeneration with accumulation of autofluorescent storage bodies in many layers of the retina. Granular material is mainly present in pigment epithelial cells, blood vessel endothelium and in ganglion cells.
***Other organs and structures*:** Extensive storage body accumulation in macrophages is observed in the liver.

Mode of inheritance: Autosomal recessive inheritance is suspected.

Gene containing mutation: Unknown

References:

Palmer DN, Tyynela J, van Mil HC, Westlake JV and Jolly RD. Accumulation of sphingolipid activator proteins (SAPs) A and D in granular osmiophilic deposits in Miniature Schnauzer dogs with ceroid lipofuscinosis. *J Inherit Metab Dis 20*: 74-84 (1997).

Smith RIE, Sutton RH, Jolly RD and Smith KR. A retinal degeneration associated with ceroid-lipofuscinosis in adult Miniature Schnauzers. Vet and Comp Ophthalmology, 6: 187-191 (1996).

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