Case reports

Neurovisceral sphingomyelinosis in a Siamese cat

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Summary. This report presents the clinical, morphological and biochemical findings on an 11-month-old female Siamese cat with neurovisceral sphingomyelinosis. Gross pathological features and histochemical findings are compared with the human disease counterpart and the previously described animal models. Hepatomegaly was observed while splenomegaly was not. Although sphingomyelin in liver and spleen was biochemically elevated, histochemical results in this case were slightly different from those previously recorded in human and feline Niemann-Pick disease. These results suggest that this feline case might be a different type of animal Niemann-Pick disease to that reported previously.

Key words: Sphingomyelinosis — Neuropathology — Histochemistry — Cat

Case report

A 6-month-old female Siamese cat, one of a litter of three, was admitted to our laboratory because of neurological signs such as ataxic gait, head tremor and loss of equilibrium. According to the owner, one of her littermates died at birth with the same neurological signs, but the other and her parents are normal. Routine hematological investigations and electroencephalogram disclosed no abnormality. Her neurological signs gradually progressed and she died at 11 months of age.

Materials and methods

The CNS and several visceral organs were removed and fixed in 10% buffered neutral formalin. For light microscopy, paraffin sections were stained with hematoxylin-eosin (H&E), periodic acid-Schiff (PAS), Luxol fast blue (LFB) and alcian blue (pH 2.5). Frozen sections were stained with PAS, LFB, Oil red O, Sudan black B and Baker’s acid hematin [7].

For electron microscopy, small pieces of formalin-fixed tissues were osmicated and embedded in epoxy resin. Ultrathin sections were stained with uranyl acetate and lead citrate and examined with a JEM 100CX-II electron microscope.

For biochemical analysis, samples from formalin-fixed liver and spleen from the affected cat and a normal female cat of similar age were used. Lipids were extracted with Folch’s method and separated by thin-layer chromatography using silica gel 60 plates (Merck, West Germany) [3, 11].

Pathological findings

At necropsy, the liver was enlarged and yellowish. No other gross lesions were observed.

By light microscopy, most neurons in the brain, spinal cord and enteric plexuses were swollen (Fig. 1). Their cytoplasm was pale, foamy or granulated; so-called “soap-bubble” in appearance. Loss of Nissl substance and margination of nuclei were occasionally observed. Cerebral and spinal endothelial storage was not found (Fig. 1). In addition, many glial cells were swollen (Fig. 2) and axonal swellings (spheroids) were found in CNS. The storage materials stained only slightly with PAS, LFB and alcian blue in paraffin sections, whereas they stained intensely.

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Fig. 1. Swelling neurons in Nucl. n. oculomotorii. H&E, ×125

Fig. 2. Many swelling glial cells in cerebral medulla. Luxol fast blue H&E, ×100

Fig. 3. Inclusion bodies in a myelinated and an unmyelinated axons. Spinal cord, ×3500

Fig. 4. Membrane-bound inclusions in neuronal perikarya. Spinal cord. ×15700. Insert Typical membranous cytoplasmic bodies. ×26200

Fig. 5. Many foamy cells in spleen. H&E, ×75

Fig. 6. Hepatocytes and Kupffer cells were swollen. H&E, ×75

Fig. 7. Irregularly membranous inclusions in splenocyte. ×7000

Fig. 8. Thin layer chromatography of lipids extracted from tissues of affected and normal cat. Samples were homogenized in ten volume of a mixture of chloroform: methanol (2:1, vol/vol) and the lower phase was washed again with the Folch upper phase. The lower phase was dried and the residue dissolved in chloroform methanol (2:1, vol/vol). Equal size samples per wet weight of each organ were spotted on silica gel 60 plate and then developed in a mixture of chloroform: methanol: water (15:6:1, vol/vol/vol). The plates were dried and spots located by spraying resorcinol and heating at 100°C for 20 min. 1, Normal spleen; 2, affected spleen; 3, normal liver; 4, affected liver; 5, sphingomyelin standard (Sigma, USA). A comparison of specimens 1 and 2, and 3 and 4, shows increased amounts of sphingomyelin in the affected cat.