Case 7

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Ref #: 

Clinical Abstract:

The patient, weighed 3.5 kilograms at birth. His mother, a G2 P1 Abl 26 year
old caucasian had developed a febrile flu-like illness during the 8th month of
pregnancy. Delivery, at 40 weeks gestation, was uncomplicated. There was no family
history of consanguinity or neurological illness except for a maternal cousin who
had mental retardation of unknown etiology. The patient had Apgar scores of 6 and 7
at 1 and 5 minutes respectively. He was microcephalic, had bilateral spastic hemi-
paresis and experienced infantile spasms. CT scan at 6 weeks of age showed that the
lateral ventricles were enlarged and the right cerebral hemisphere was smaller than
the left.

During the first year of life the patient's psychomotor development was
severely delayed. On examination at 18 months of age he weighed 9 kilograms, had
a height of 84.5 cms. and had severe scoliosis of the lower spine. His head
circumference was 45.5 cms. Deep tendon reflexes, which had previously been brisk,
were diminished or absent. The patient withdrew from pain but was otherwise
unresponsive. The optic disks were thought to be normal. EMG showed neurogenic
abnormalities and peroneal nerve conduction velocities were 13 meters/sec (normal
39-51 meters/sec). Gastrocnemius muscle and sural nerve biopsies were performed.
The muscle showed groups of round atrophic fibers surrounded by adipose and connective
tissue without inflammation or necrosis. Myofibers were small (7.5 to 15 microns in
diameter) and showed type II C fiber staining characteristics except in one fascicle
which included a group of hypertrophic type I fibers. The sural nerve biopsy showed
loss of myelinated fibers of all sizes and some thinly myelinated fibers, but no
evidence of active degeneration, demyelination or storage abnormality. The patient
was diagnosed as having Werdnig-Hoffmann's disease. At 3 years of age, since the
parents were considering having another child and in view of the patient's atypical
presentation, biopsies were taken from the other sural nerve and the frontal cortex
for further neuropathological evaluation. The sural nerve showed further loss of
nerve fibers, most remaining fibers appearing atrophic. The frontal cortex and
superficial white matter showed only mild gliosis. The patient experienced increasingly
frequent bouts of aspiration that led to a terminal pneumonia at 4 years of age.
The brain weighed 1,020 gms. and was mildly asymmetric (the left cerebral hemisphere
being larger than the right). The optic nerves were thin and gray. In coronal sections
the lateral ventricles were dilated, the left more than the right. The corpus callosum
was markedly thinned. The cerebellum, brain stem and spinal cord were grossly normal.

Material submitted: 1 H&E slide and 1 unstained slide of the spinal cord.

Points for discussion:
1) Is this a progressive illness?
2) What relation does the pathologic change in the neurons have to other degenerative
disorders?