Case #5

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The patient was a 32 year old white female. The history of neurological symptoms began at age 5 with bilateral foot drop which progressed to leg weakness and inability to walk by age 8. Because of the suspicion of a spinal cord tumor, a spinal laminectomy was performed which revealed no cord lesion. She attended public school through the 8th grade and had a tutor through the 10th grade. Her intelligence was judged to be borderline normal. For a year or so before her death, she complained of gradually deteriorating vision (in both eyes) that was uncorrected by glasses. Two months before her death, she developed somnolence and dysarthria together with diplopia associated with a divergent strabismus. An ophthalmological examination one month before death, reported bilateral optic atrophy and a divergent strabismus with inability to adduct either eye past the midline. At that time, a diagnosis of demyelinating disease was made and, in the last 3 weeks of life, she became progressively more obtunded, expiring after one week of deepening coma.

A detailed family history indicated no neurological disorders. Additionally, the history states that she was not an alcoholic nor was she malnourished.

The general postmortem examination findings were limited to fibrosis and shrinkage of the thyroid gland and moderate coronary arteriosclerosis.

The brain weighed 1150 grams. Serial coronal sections through the cerebral hemispheres revealed multifocal lesions. The head of the caudate nucleus contained areas of cystic necrosis which were more extensive on the right than on the left but also extended into the body of the caudate on both sides. Similar lesions in various stages of necrosis were present in the left putamen, thalamus, and hypothalamus exclusive of the mamillary bodies. The optic tracts and chiasm appeared small but otherwise not definitely abnormal. Serial transverse sections through the brainstem disclosed extensive softening in the tegmentum of the brainstem, extending from the rostral midbrain to the medulla. A large necrotic lesion was also evident in the deep cerebellar white matter and dentate. Sections through the spinal cord showed pale, granular posterior columns.

Submitted are: 1 slide of the hypothalamus with mamillary bodies and 1 slide of the rostral pons; both stained with H & E.

Points for Discussion:

1. Incidence of Leigh's encephalopathy in adults. 2. Relationship to the infantile form. 3. Relationship to Wernicke's encephalopathy. 4. Familial incidence. 5. Possible biochemical defect(s).