Case #2

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Reference: (NP 4362, University of Washington Medical School)

This 14 year old boy was said to have Batten's disease since about age 10. He was first evaluated at age 11 in November, 1971 for intellectual deterioration and incoordination. A rectal biopsy at that time revealed storage material in the ganglion cells. In April, 1972, examination showed an obese, dysarthric and sluggish boy who walked stooped over, bent at the knees and hips. He had limited upward gaze, questionable non-macular vision in the left, abnormal ocular fundi with pigmentary degenerated macula, hyperactive DTR's, sustained ankle clonus, upgoing toes, moderate spasticity (worse in the legs than in the arms), ataxia and Romberg's sign. There was a small right subcapsular cataract. EEG was abnormally slow and mildly epileptiform. Urine screen for mucopolysaccharides was negative. The diagnosis was Batten-Spielmeyer-Vogt disease (or juvenile cerebromacular degeneration). The patient also had skin lesions suggesting epidermolysis bulbosa in the extremities and trunk for 3 years, but a skin biopsy in May, 1972 showed only subacute dermatitis.

The patient was reevaluated in January, 1974 for a decubitus ulcer of the right hip. He had not walked for 6 months, his extremities were weak and wasted. He responded to pain and the simplest commands but was unable to speak, only uttering sounds. He had difficulty swallowing and choked frequently. Seizures, which had been only occasional in May 1972, had increased to one per day and included jerking of the mouth and upper limbs. Neurological examination in March, 1974 showed frequent yawning and chewing movements, positive snout reflex and glabellar rugae. He was able to seen and follow an object but did not respond to hand clapping. Gag reflex was absent. Muscle tone was increased with contractures of both elbows and knees. DTR's were 4+ and Babinski's sign was present bilaterally. He was on Dilantin, 200 mgs. per day, which seemed to control the myoclonic jerks. He was seen again in September 1974, essentially unchanged.

The patient died in October, 1974 of bronchopneumonia following a cold for several days.

General autopsy: Bronchopneumonia

Gross neuropathology: Mild to moderate degree of ventricular dilatation, granular appearance of cerebellar cut surface. Whole brain weight: 1030 grams.

Microscopic slides: Frontal cortex: One stained with LFB-PAS-H, one unstained.

Points for Discussion:

1. Diagnosis.