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Clinical Abstract:

The patient was the product of an uneventful term pregnancy, labor and delivery, birth weight was 7 lbs, 12 ozs. He had no problems in the immediate postnatal period and sat at the age of 8 months. He spoke two to three words when he was 9 months old and began to walk at age 12 months. The mother noticed that he stopped speaking and gradually developed difficulty in walking at about 14 months of age. He was evaluated by an Orthopedic Surgeon who found no significant orthopedic problem. The child's motor abilities slowly deteriorated and at the age of two and a half years he was noted to have high-pitched cry and many adventitious movements. At three years of age his weight was thirty-four pounds; head circumference was below the 50th percentile and height was below the third percentile. Funduscopic examination revealed no abnormalities. No abnormal physical findings were detected in the lungs or heart. There was no abdominal organomegaly or bony deformity. Muscle tone was considered normal throughout and deep tendon reflexes were brisk. Bilateral ankle clonus was present and cranial nerve examination was unremarkable. Skeletal survey and bone age were normal. CT scan revealed no abnormality.

Laboratory Studies: A complete blood count was within normal limits, including the differential and morphologic examination. Cerebrospinal fluid was clear and colorless containing five lymphocytes/cubic millimeter. CSF protein was 9 mg/deciliter, glucose 56 mgm/deciliter, and the chloride was normal. Numerous other blood tests were done which included a moderately elevated SGOT, LDG, SGPT, and alkaline phosphatase levels. Urinalysis was normal. EEG showed an abnormal, poorly-organized, slow electroencephalogram with no consistent focal lateralizing or paroxysmal features. This was interpreted as representing a diffuse encephalopathy of an unspecified type. Urine screening test for amino acids, proline, hydroxylproline indoles, imidazole, methylmalonic acid, orotic acid and mucopolysaccharides, were all normal.

The patient's condition continued to deteriorate until he became completely uncommunicative except for constant screaming. He expired as a result of cardiorespiratory arrest associated with pneumonia at the age of three and one-half years. The family history was negative for any evidence of central nervous system disease.

MATERIAL SUBMITTED: One H & E slide and one unstained section.

Points for Discussion:

Diagnosis