CASE # 1990-10

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Clinical History: The patient was a 12 year old boy with an incapacitating choreoathetoid movement disorder which began at 32 weeks of age during a brief, systemic illness characterized by diarrhea, vomiting, and otitis media. Laboratory studies including CSF examination, CBC, urinalysis, serum electrolytes, serum enzymes, uric acid, SMA_{12}, and ceruloplasmin disclosed only a slightly elevated LDH. The EEG was normal. His gestation, birth, and development had been previously unremarkable except that he was delivered without complications following a three hour induced labor. He weighed 10 lbs, 5 oz at birth. A maternal cousin had a seizure disorder but there was no other family history of neurological disease. His parents (who are unrelated) and older sibling were healthy.

During the ensuing twelve years the patient's cognitive and educational attainment reached second grade level. Because of his movement disorder, however, he never developed intelligible speech, had no voluntary use of his upper extremities, and was never able to sit, stand, walk, or keep his head erect without support. In the last four years of his life he had occasional, brief generalized seizures.

A CT of the head at 2 years of age was considered normal. Urine and plasma amino and organic acids were done at 12 years of age and were normal.

While in a spica cast following surgery for subluxation of the hip he developed decubiti and malnutrition. He died at home awaiting repair of the decubiti.

Necropsy findings: The formalin fixed brain was sent by the medical examiner to Children's Hospital of New Orleans for examination. The brain weighed 1390 grams. Grossly, the caudate and lentiform nuclei were symmetrically atrophic and firm.

Materials submitted: H&E section of corpus striatum

Points for discussion: Diagnosis