CASE 1993 - 2

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CLINICAL HISTORY:

The patient was a 41 year old male with a long history of mental impairment and a severe progressive motor disability, who expired in a chronic care facility.

A childhood history reveals that the patient was slow to develop and required special education classes upto 16 years of age. Progressive deficits in motor strength and gait difficulties were noted in early to mid childhood. Subsequently slurred speech and dysphagia developed. In childhood he had measles and carried a diagnosis of "Galactosemia" He had bilateral cataract extractions at the age of 18 years. He also had history of extraction of multiple carious teeth and bilateral ankle swelling.

The patient's mother died of cancer at age 51 years and father died of a heart attack at age 62 years. Both parents had "diabetes". Out of the 5 siblings, a sister (2 years younger than the patient), and a brother (2 years older than the patient) had similar afflictions. Three other siblings were unaffected.

Physical examination revealed severe intellectual disability, an ataxic-spastic gait, dysarthria, and evidence of polyneuropathy.

LABORATORY STUDIES:

CBC - normal, serum cholesterol - normal, Galactose tolerance test and Galactose transferase levels were normal; 17 Ketosteroid secretions = 7.9 mg/24 hour (normal 3-10 mg); 24 hour urine for amino acids revealed "uniformly low levels". Chromosomal analysis was normal. Chest x-ray and EKG were normal at age of 27 years. CT scan (brain) showed diffuse, cortical atrophy at age 35 years.

NECROPSY FINDINGS:

At autopsy the brain showed cerebellar atrophy and cut sections revealed multiple ill defined gray-yellow lesions in the basal ganglia and cerebellum.

MATERIAL SUBMITTED:

One H&E stained slide, cerebellum

POINTS OF DISCUSSION: 1) Diagnosis
2) Pathogenesis