CLINICAL HISTORY:

D.L. was a 41 year old white male with a long history of mental retardation and a severe progressive motor disability, who expired in a nursing home.

A childhood history reveals that the patient was slow to develop and required special education classes up to 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 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) have a similar affliction. Three other siblings are unaffected.

Physical examination revealed severe mental retardation, 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 reveal 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