

CASE 9

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This 15 year old male had developed normally until 3 years of age, when he was hit on the head with a hammer without any immediate sequelae. Later he began to stumble, became less coordinated and steadily regressed mentally. Neurological studies revealed evidence of mental deficiency, spasticity and incoordination; no definite diagnosis was made. He was subsequently institutionalized, becoming extremely stiff, spastic and immobile. He never had seizures, choreo-athetosis or myoclonus. He became emaciated and terminally developed episodes of cyanosis and respiratory difficulty. During one of these episodes he died.

Autopsy; Marked cerebral atrophy (660 grams), severe bilateral bronchopneumonia, generalized cachexia and atrophy of skeletal muscles. The slide distributed is a Nissl-stained section from the cerebral cortex and is typical of the laminar neuronal degeneration seen throughout the cerebral cortex, with relative sparing of the hippocampal formation. The brain stem and cerebellum (weight 147 grams) were relatively uninvolved.

Diagnosis; Poliodystrophy (Alper's disease)

Points for discussion: Relationship to Jacob-Creutzfeldt's disease, and "Walnut brain" (Courville)