CASE 7

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This 11 months' old male infant was the product of a normal pregnancy and delivery. The family history was not remarkable; there were two normal older siblings. The neonatal course was normal until bilateral hernia operations in the third month. At that time he began to regurgitate feeding, failed to gain weight and developmental progress halted. He developed generalized and myoclonic seizures and became unresponsive to stimuli. When examined at eight months of age, he showed myoclonic seizures and a decerebrate posture with opisthotonus. EEG showed random spikes with irregular background. He was maintained on tube feeding and had several bouts of pneumonitis and atelectasis, with one episode of cardiac arrest. He was found dead in his crib.

General autopsy findings showed only tracheobronchitis and pneumonitis.

The fresh brain weighed 720 grams. It appeared atrophic, particularly in the frontal lobes and cerebellum. The fixed brain showed increased resistance to cutting. The white matter of the hemispheres appeared markedly shrunken, brownish-gray discolored, and diffusely spongy. The gray matter did not appear affected. The white matter of brain stem and cerebellum were yellowed and remarkably firm and rubbery. There was obscuration of nuclear outlines. The pyramidal tracts in the medulla were cavitated. The spinal cord was not examined.

Microscopically there was a mild diffuse cortical gliosis, with mild to moderate nerve cell loss most marked in the hippocampus. Nuclei of basal ganglia and brain stem are distorted by the adjacent degenerated white matter. The white matter shows an absence of myelin and extensive infiltration with hypertrophic astrocytes, pleomorphic microglia, and many multinucleated cells with strongly PAS-positive cytoplasm. The medullary cavities are punched out and encapsulated. Below this level, in the upper spinal cord, the white matter shows the previously described degeneration, with marked loss of anterior horn cells.

The submitted slide is a section at the level of the inferior olivary nuclei, stained with Luxol Fast Blue-Periodic Acid Schiff.

Diagnosis: Globoid Body Leukodystrophy

For discussion: Nature of the bilateral medullary cavitation, its etiology, and the relation of infantile cavitary encephalomalacia to the leukodystrophies.