Submitted by: Edward A. Klein, M.D., Department of Pathology, Staten Island University Hospital, Staten Island, New York 10305

This 11 month old female was the product of an uncomplicated full term pregnancy, born via vaginal delivery with good Apgar scores. She was discharged within one week after an unremarkable postnatal course. At age 4 months the patient suffered a generalized motor seizure and was diagnosed to have internal hydrocephalus. Treatment included anticonvulsant medication and a ventriculo-peritoneal shunt. Successive CAT scans revealed progressive, slight increase of ventricular size with no increase of head circumference, in addition to rarefaction of the cerebral white matter. Clinically, the patient showed progressive psychomotor retardation with failure to achieve milestones, progressive decortication with spasticity, and episodic seizures. The patient remained at home during the final 3 months and was brought to the ER in status epilepticus two weeks before death. The patient remained comatose subsequently and died at home.

Clinical workup for the following gave negative or normal results:
Antibodies for TORCH, Borrelia, E. Barr virus,
Long chain fatty acids, Lactate, Pyruvate,
Amino and organic acids, Phytoic acid
Metachromatic leukodystrophy and Krabbe's disease
Karyotype

Family history was negative for any neurologic illness.

Autopsy Findings:

Cachectic white female with body weight and height below 3rd percentile and normocephalic (head circumference 45cm). General autopsy unremarkable. Brain weight 884 grams (normal 850), The cerebrum had a grossly normal external appearance with normal gyral pattern. However, the cerebral cortical mantle was easily compressible and the frontal and parietal lobes could be collapsed with slight pressure. The cerebellum was globally small but otherwise unremarkable externally. Coronal sections of the cerebrum revealed diffuse softening and loss of cerebral white matter with cavitation in the centrum semiovale. The cavitation was present in all lobes except temporal, worst in the frontal to the point of subtotal destruction. The cavities had poorly defined margins and did not spare "U" fibers. The loss of white matter was nearly symmetric with no recognizable geographic or "tigroid" pattern. Infratentorially, cavitation was present in the dentate hila and pontine tegmentum. The cerebral cortical ribbon was grossly normal except for a shrunken appearance of the hippocampi. The lateral ventricles were slightly enlarged without obstruction, and the shunt tube was patent.

Material Submitted:
(1) H&E slide of frontal lobe
(2) Electron micrograph of astrocyte in cerebral cortex

Points for Discussion:
(1) Diagnosis
(2) Pathogenesis