CASE 5

A66-556

Submitted by Dr. R. A. Clasen, Presbyterian - St. Luke's Hospital, Chicago, Illinois

The patient was born to a 36 year old gravida IX, para IX white female. At the time all siblings were living and well. The patient's birth weight was 8 pounds 3 ounces. His development was never considered normal. He sat up at 5 months; walked and talked at 3 years. A diagnosis of progeria was made at 1 year of age. The patient never attended school but was able to walk around his neighborhood and recognized and called his neighbors by name. He was first seen at our outpatient clinic at the age of 17 years because of transient paralysis and an increasingly clumsy gait. At that time he could smile and say "hello doctor." He presented the typical physical appearance of progeria. One month later the patient was admitted to hospital because of the sudden onset of right-sided paresis and loss of speech. His motor symptoms improved but he was never again able to make sounds intelligible to anyone but his mother. On the second hospital day, he developed a right-sided seizure. At the time of his discharge he was able to follow simple commands. The patient was admitted to our hospital for the last time at the age of 19 because of loss of voice, and passing black stools. Examination of the spinal fluid revealed a protein of 705 mg%. A cell count showed 1,000 RBCs and one mononuclear WBC. He expired on the 3rd hospital day.

At autopsy the body weighed 13 lbs. and measured 89 cm. in length. Except for a general reduction in size and absence of testes, no gross visceral anomalies were demonstrated. Atherosclerotic changes were absent. Microscopically, the viscera showed disseminated granulomatous lesions, morphologically, resembling Boeck's sarcoid. A pure culture of alpha streptococci was obtained from the blood. The brain weighed 500 grams. The meninges were markedly thickened. The gyral pattern of the cerebral hemispheres was normal. The cut section showed marked symmetrical dilatation of the ventricles. The white matter generally was atrophic and showed a grayish discoloration. There were calcific changes in both basal ganglia and dentate nuclei.

The slides submitted are from the cortex and subcortical white matter stained by the phloxine-fast green FCF-gallocyanin technique (Jour. Neuropath. Exp. Neurol. 23: 156-160) and from the basal ganglia stained by the hematoxylin-eosin technique.

The changes observed in the brain of this case are similar to those recently described by Dr. Malamud and his coworkers in a case diagnosed clinically as Pelizaeus-Merzbacker disease (Jour. Neurol. Neurosurg. & Psychiat. 28:540-547, Dec. 1965). It is suggested that our case represents a similar genetic defect.