Case 7

Submitted by: William W. Pendlebury, M.D. and Daniel P. Perl, M.D.
Laboratory of Neuropathology
Department of Pathology
University of Vermont College of Medicine
Medical Alumni Building
Burlington, Vermont 05405-0068

Reference No: XED 82-9

Clinical Abstract:

The patient was the product of a G_6 P_{1041} 26 year old white woman, born at 35 weeks gestation. There was no history of consanquity. Of the four abortions, the first three had been spontaneous and the last therapeutic. The fifth pregnancy had resulted in delivery of a 35 week gestational age, 5 pound 13 ounce male infant, presently alive and well. Family history revealed Down's syndrome in a maternal sibling and paternal second cousin. A maternal aunt was said to have had polydactyly. Chromosomal studies performed on both parents were reported as normal. Details of the present pregnancy are not known, but it was said to have been uncomplicated. Delivery was preceeded by meconium-stained fluid, and the baby was delivered vaginally in a frank breech presentation. Apgar scores were 3 and 6, and the baby was intubated immediately. Birth weight was 2520 grams, and multiple congenital anomalies were noted. There was dolichocephaly and bathrocephy with premature closure of fontanelles. There was micrognathia, microglossia, posterior cleft palate, bifid epiglottis, and bilateral postaxial polydactyly of hands and feet. X-ray examination revealed no osseous formation in any of the 6th digits. There was also 2-3 syndactyly of both feet. There was a sacral dimple, and the anus was patent. The patient died at 20 hours from cardiopulmonary arrest.

Autopsy confirmed the previously noted anomalies. Additionally, the patient was found to have a right cervico-thoracic scoliosis, bilateral renal ectopia and hypoplasia, and bilateral pulmonary hypoplasia. Gross examination of the brain revealed a weight of 210 grams after formatin fixation and a 1.3 cm smooth, firm, spherical mass arising from the area of the right mammillary body and extending across the midline.

Material Submitted: 2 2x2 kodachromes of gross lesion
1 H & E stained section from autopsy

Points for discussion:
1. Diagnosis
2. Pathogenesis