

CASE 9

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This 3 1/2-year-old boy was noted to be jaundiced in the neonatal period. A liver biopsy at age 2 months revealed intrahepatic biliary atresia. The extra-hepatic biliary tree was demonstrated to be patent. The child's psychomotor development was somewhat retarded. At age 2 1/2 years he was noted to be small for his age and very jaundiced. Xanthomas were noted on the gums and numerous xanthomatous deposits were present on the fingers, palms, elbows, legs and feet.

At age 3 years, he was hospitalized because of increasing irritability, lethargy, bleeding and a seizure involving the right upper extremity. He was febrile and had a large head. Liver and spleen were enlarged. Jerking of the right arm and occasionally of the left arm and fingers were noted. His eyes tended to deviate to the right. Hyperlipemic vessels and blurring of the temporal disc margin were noted in the right fundus. There were two diopters of papilledema in the left fundus. He had bilateral Babinski signs. A ventriculogram was done but no abnormalities were noted. Following ventriculography he developed a left hemiplegia and bloody discharge from site of the right parietal needle puncture. The patient apparently improved somewhat although he remained hemiparetic and blind. A liver transplant was performed 5 months later and the patient died the following day.

Autopsy findings: Hemoperitoneum, **atelectasis** of the lungs and pulmonary and aortic atherosclerosis.

Neuropathological findings: Extensive old encephalomalacia involving the parietal and occipital lobes, bilaterally, and old hemorrhage in the right corpus striatum and external and extreme capsules; swollen axons in the sensory nuclei of the medulla and in Clarke's column. Numerous xanthomas were found on the inner surface of the ventral aspect of the spinal dura, chiefly in the cervical and lumbar segments. Similar lipid deposits were found in the cerebral dura and choroid plexus.

Submitted are: 1 slide and 1 Kodachrome of spinal dura.

Points for discussion:

1. Pathogenesis of lipid deposits in dura.
2. Distribution of these deposits.
3. Chemical nature of lipid.