

Case #9

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This white female infant was born at 39 weeks gestation, the first child of a 17 year old mother. The pregnancy was unremarkable apart from a brief febrile illness during the 4th month and the onset of albuminuria a few days prior to delivery. The child was born as a double footling breech after a 12 hour labor and was noted to be cyanotic. Convulsions developed soon after birth, which improved after suctioning and administration of oxygen. Blood sugar levels of 3 mg% and 9 mg% were recorded, and the infant was treated with intravenous dextrose and intramuscular Glucagon.

On physical examination, the patient was found to be markedly floppy, and the striking hypertonia permitted easy palpation of the abdominal viscera. Multiple congenital anomalies were found; notably, a prominently rotund face, low set ears, bilateral epicanthic folds, corneal clouding, bilateral simian creases, clubbed feet, and a protruding upper lip. There was moderate hepatosplenomegaly and the kidneys were apparently of normal size.

The clinical course over the next 5 days was marked by transient hypoglycemia, and an inability to feed, requiring gavage feeding. On the 4th day of life, the patient developed an episode of gastrointestinal bleeding and a platelet count of 87,000 with a normal prothrombin time was found. This failed to respond to therapeutic measures and the patient died after five days of life.

General Pathology: The foramen ovale was patent, and the pulmonary artery gave rise to the descending aorta. The ascending aorta continued as the innominate artery and terminated as the left subclavian artery. The liver showed some patchy fibrosis, and subcapsular and medullary cysts were seen in the kidney.

CNS Pathology: The brain weighed 370 grams and was externally unremarkable except for some slight thickening of some of the temporal gyri. Coronal sections showed focal blurring of the normal cortico-medullary junction. A small sub-ependymal cyst measuring 4 mm. in diameter was present in the left caudate nucleus.

Microscopic Pathology: Two slides, stained with Hematoxylin-eosin and Oil Red O.

Points for Discussion:

1. Diagnosis.
2. Pathological features of the disease.
3. Etiology and pathogenesis.