50th ANNUAL DIAGNOSTIC SLIDE SESSION 2009

CASE 2009-09

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Clinical History: This infant girl was born at 37 weeks gestation via C-section to a 28-year-old G1P0 woman. Maternal testing was positive for Group B Streptococcus but was otherwise unremarkable. At 22 4/7 weeks, ultrasound showed mildly echogenic and enlarged kidneys at the 95th percentile but normal volume of amniotic fluid. Amniocentesis was declined at this time. By 30 5/7 weeks, the estimated fetal weight was 8th percentile and there was severe oligohydramnios, bilateral large echogenic kidneys and an echogenic focus in the left cardiac ventricle. The baby was delivered by elective C -section on 01/12/08 with APGARs of 7 and 8. Weight was 1890 g (<10%), head circumference 33 cm (50%), and length 32cm (<10%). In the delivery room, she required blow-by O2 and continuous positive airway pressure.

She was admitted to the NICU, where she was intubated and received surfactant twice. She was extubated to an oxyhood at 8 hours of life but then was reintubated at 24 hours due to increased work of breathing. Chest X-ray showed left lung opacity and blood gas showed severe metabolic acidosis (pH 7.03) despite aggressive attempts to correct with a bicarbonate drip. Lactate was 7.5. Over the next 24 hours of life, the baby became hypotensive requiring saline boluses and a dopamine drip. An echo was done, which showed a patent ductus arteriosus and patent foramen ovale with bidirectional flow and normal low or mildly depressed left ventricular function. Labs showed that she was coagulopathic (INR 2/ PT 24/ PTT 51). She also had electrolyte disturbances including hyperkalemia (8.1) and hypoglycemia (down to 38). Initial ammonia was 223, rising to 496.

On exam the baby was noted to be dysmorphic with a prominent forehead and a strong odor “like sweaty feet”. The anterior and posterior fontanelles were large. There was a 1-2/6 systolic ejection murmur. There was a paucity of spontaneous movement but arousal to tactile stimulation. Pupils were 2-4 mm, eyes midposition, tone normal, movements of all extremities, reflexes 2/4. She had 2-3 episodes of left arm quivering and stiffening associated with lip quivering lasting 1-3 minutes, associated with bradycardia, hypertension, and desaturation.

Metabolic and genetic testing was pursued. Head ultrasound was negative for intraventricular hemorrhage.

The prognosis was poor and care was redirected. The baby died at 2 d of age.

Autopsy findings: Pulmonary hypoplasia and bilobed lungs; cardiomegaly with biventricular hypertrophy; cytoplasmic lipid deposition in myocardiocytes, hepatocytes, renal tubular epithelium, and skeletal myofibers; renal dysplasia with dilated tubules; and fetal osteosclerosis.

Material submitted:
1. Glass slide from parietal lobe
2. Gross image of brain

Points for discussion:
1. Diagnosis
2. Pathogenesis of brain abnormality