

CASE 1993-3

Submitted by: Dr. Samuel K. Ludwin and Dr. David Hurlbut
Departments of Pathology, University of Western Ontario
and St. Joseph's Health Centre

Department of Pathology, Health Sciences Centre
University of Western Ontario
London, Ontario, N6A 5C1. Canada.

Clinical History: This 36 week gestation male infant was delivered by Caesarean section 17 h 20m following spontaneous rupture of membranes. Caesarean section was performed because of a nonreactive nonstress test. The pregnancy for the 30 year old mother (TPAL 1001) was uneventful apart from decreased fetal movements. APGAR scores were 2,3,6 at 1,5 and 10 minutes, respectively. The infant required resuscitation by the neonatal intensivists. He had multiple congenital anomalies including micrognathia, limb contractures and cleft palate. The infant was incubated and ventilated. Subsequent complete neurological examination revealed no spontaneous limb movements, and absent deep tendon reflexes. Echocardiography showed a large atrial septal defect. Following further investigations compassionate care was instituted. The baby died at 10 days after delivery. Consent for a complete autopsy was obtained from the mother.

Necropsy findings: Atrial Septal Defect

Material submitted: 1 glass slide of basal ganglia

Points for discussion: 1. Diagnosis
2. Nosology
3. Is this all one disease?