

**Canadian Association of Neuropathologists**  
**L'Association Canadienne des Neuropathologistes**

**CASE No 10**

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This female was the second child born to third cousins of Anglo-Irish ancestry. Family history was negative. A 32 week ultrasound showed IUGR with oligohydramnios necessitating an emergency caesarean section. Birth weight was 0.812kg (<0.4th centile); physical exam was otherwise normal. A cranial ultrasound on day 2 of life revealed bilateral, small echogenic calcified foci in the frontal regions and the thalami. An infection screen was negative. She was slow to feed and was discharged home on day 54 of life tolerating breast feeds. At this time her height and weight were < 0.4th centile.

Ophthalmological examination on day 54 revealed abnormal retinal vascularisation and pre-retinal hemorrhages peripherally and over the macula of the left eye, and incomplete vascularisation of the right retina. A diagnosis of retinopathy of prematurity was made. Despite bilateral laser treatment, vitrectomies and photocoagulation she experienced bilateral retinal detachment with scarring and was left with light perception only.

In view of central hypotonia and peripheral hypertonia, a diagnosis of cerebral palsy was made at age 7 months. At 13 months of age she developed short-lasting, jerky movements of the arms and legs. An EEG demonstrated focal epileptiform activity over the left superior frontal, central and posterior temporal regions. Cranial MRI revealed features of a non-progressive hydrocephalus with a narrowed aqueduct and small fourth ventricle as well widespread abnormalities of the white matter in the frontal lobes and periventricularly. Brain CT showed extensive calcification of the periventricular, thalamic, basal ganglia and dentate regions. At 19 months of age she experienced two generalized seizures and was treated with phenobarbital.

She failed to thrive and at 21 months her weight and length were both below the 3rd centile. At 3 years she was noted to have generalized osteopenia and compression fractures of thoraco-lumbar vertebrae with loss of height and anterior wedging.

At 3 years 2 months she presented with recurrent meleana and haematemesis. Endoscopic biopsy demonstrated duodenitis and dilated veins. Abdominal ultrasound showed para-umbilical and gastrohepatic varices. A liver biopsy revealed portal fibrosis with abnormally large vascular channels. She had a normal ALP with a minimal elevation of transaminases and normal bleeding indices. Metabolic, biochemical, immunological, infective and chromosomal studies were non-contributory. Stomach and small bowel biopsies were suggestive of ischemic injury and underlying telangiectasia. A Whipple's procedure was performed in an attempt to control the gastro-intestinal hemorrhage; however, her liver function continued to deteriorate and she died aged 3 years 9 months.

Material Submitted: H&E section of the thalamus

Question: Differential Diagnoses?