Case 1994-5

Submitted by: P.V. Gould and D.G. Munoz, University Hospital, University of Western Ontario, London, Ontario N6A 5A5

Clinical History:
The patient was a 40 year old male who developed bilateral sensorineural deafness at age 11. By age 20 he had an ataxic gait and progressive loss of vision. His brother underwent an orthotopic heart transplant for cardiomyopathy. The mother reportedly also suffered from cardiomyopathy and ataxia.

By age 36 our patient had marked external ophthalmoplegia with slight residual lateral gaze only — more so on the left. He had bilateral ptosis with facial weakness and prominent dysarthria. His motor strength was rated as 4/5 in all extremities. Sensation was intact except for the left hand which showed decreased pin prick, light touch and vibration. His gait was wide based and his coordination was impaired. An MRI scan revealed moderate cerebellar atrophy but no other lesions. He had no evidence of cardiomegaly. At age 39 his condition had deteriorated; he required a wheeled walker despite coenzyme Q therapy.

3 months later the patient developed pneumonia and went into acute respiratory failure. He remained dependent on mechanical ventilation thereafter due to significant respiratory muscle weakness. After 2 months he suffered a cardio-respiratory arrest but was successfully resuscitated. Following this episode he regained consciousness but had marked myoclonus and bouts of seizure activity which responded to standard anticonvulsant therapy. He was transferred back to his community hospital 3 months later, still respirator-dependent following tracheostomy. He suffered a terminal cardiac arrest there 3 months later.

Necropsy findings:
General autopsy revealed cerebellar atrophy and hypertrophic cardiomyopathy. The fresh brain weighed 1260 g. The brain was referred to our institution for further evaluation. The external examination confirmed symmetric atrophy of the cerebellum, but was otherwise unremarkable. Upon sectioning the brain we noticed multiple small areas of rarefaction in the tectum of the midbrain.

Material submitted:
- H&E stained section of cerebral cortex (a)
- H&E stained section of midbrain (b)

Points for discussion:
- Diagnosis
- Molecular defect