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Case Reference no: PM83-89

Clinical History: Female 19 years

She fed poorly as a neonate and was hypotonic; she sat independently at 1 year of age and did not walk until 2 years and 3 months. Speech development was normal. Her gait was always ataxic.

12 years: External ophthalmoplegia, retinopathy and cerebellar syndrome.

14 years: Insulin-dependent diabetes.
16 years: Mild bulba weakness, brisk reflexes and exterior plantars.
19 years: Fit after a fall. Admitted unconscious to hospital.

Biochemical and haematological screens normal, including blood lactate concentrations (apart from mild hypoglycaemia) - ECG: bifascicular block, CSF: xanthochromic with 3g/l proteins. EEG: widespread theta and slow activity with superimposed episodes of high amplitude irregular slow waves - CT scan: extensive white matter low attenuation.

The patient did not regain consciousness and died 3 weeks later.

Necropsy findings: General post-mortem examination showed bronchopneumonia and oedema, pyelonephritis and cystitis. The brain weighed 1150g and showed the appearance of a diffuse white matter disorder.

Material submitted: H&E sections

Points for discussion: 1. Diagnosis
  2. Pathogenesis