CASE 3

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Reference No.:

Clinical Abstract:

This 3 1/2 year old boy, with severe psychomotor retardation, was born to non-consanguineous parents after a normal pregnancy and delivery. His birth weight was 4054 grams, length 53.5 cm, head circumference 37 cm and he appeared normal. At one month of age the parents voiced concern about "nodding movements" of the head. On physical examination his visual status was questioned, he exhibited head lag and the lower limbs were felt to be "stiff" with occasional scissoring. Two months later, examination revealed roving eye movements, poor head control, cortical blindness, developing spasticity and hepatomegaly. Seizures developed shortly afterwards. The initial EEG was normal, as was the CT scan.

Initially a poor feeder, his only weight gain was associated with a switch in formula (he was found to be lactose intolerant). However, as time progressed his parameters (weight, length and head circumference) began to decline. Seizures proved impossible to control for any prolonged period of time. Later EEG's showed generalized abnormalities with/without evidence of seizure activity. An extensive investigation and numerous consultations (including other centres) failed to give any diagnostic insights.

INVESTIGATIONS

CSF - (protein, glucose, cells), normal
  - measles antibody, negative
  - immunoglobulins, negative
Skin biopsy - normal
Liver biopsy - changes consistent with toxic injury
Toxoplasma antibodies - negative
Lactic acid - Normal (CSF not done)
Amino acid screen (serum and urine) - normal
Hexaminidase A, B glucosidase, B galactosidase, sphingomyelinase, oligosaccharides, mucopolysaccharides - all normal.

At autopsy, the child weighed approximately 13 kg, had a body length of 97 cm and a head circumference of 49 cm. The brain weighed 1140 gms (cerebellum and brainstem 35 gms) and had a normal gyral pattern.

Material submitted: One 2x2 kodachrome (cerebral hemisphere)
  : One H&E slide (cerebral cortex, white matter)
  One unstained slide

Points for discussion: 1. Diagnosis
  2. Pathogenesis