

Case 11

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Clinical Abstract:

The patient was a 30 month old male admitted to Long Island Jewish Medical Center one week antemortem for dehydration and vomiting.

He was the 3020 gm. product of a 37 week gestation to a 32 year old, G1 P0 woman who had been given human chorionic gonadotropin and clomiphene during and before pregnancy. The pregnancy was otherwise uneventful, and the patient was born via normal spontaneous vaginal delivery with Apgars of 8 and 9. At birth, a large cephalohematoma was noted, and at 8 hours of age skull x-rays revealed multiple depressed fractures involving the occipital and parietal bones. The infant moved all extremities and displayed no neurologic deficits. At 5 days, vertical nystagmus and staring episodes were noted. EEG was suggestive of a paroxysmal disorder. No seizure activity was identified, and the patient was discharged at 3 weeks to home care.

At 2 months of age the infant developed generalized seizures and was placed on Phenobarbital. At this time he was appropriate size and weight for age, could smile, turn his head, and rolled from side to side. However, over the ensuing months his seizure activity continued, and by 6 months of age the infant had lost developmental milestones. He was hypotonic, had little visual following, did not smile, and had poor head control. By 8 months of age the infant was in the 3rd percentile for height, weight and head circumference. At that time, an extensive biochemical workup was performed. From that time until his death, the infant was unable to do more than lie supine and verbalize sounds. He was unable to speak, follow visual stimuli, and did not smile.

Over the two ensuing years the patient had multiple admissions for vomiting, respiratory infection, weight loss and dehydration. He was treated with antibiotics and supportive therapy, and each time discharged to home care. During his last admission for vomiting and dehydration, the patient succumbed to bilateral bronchopneumonia.

Material submitted: One H & E slide from the cerebellum
One 2 x 2 kodachrome of the base of the brain

Points for discussion: Diagnosis and pathogenesis