Case 9

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Reference No. Vancouver General Hospital A82-127

This 3½ year old North American Indian girl presented with a gait disorder. She the first child of healthy, nonconsanguineous young parents. Twin sisters were thriving. A brother died of prematurity. There was a family history of diabetes mellitus. Towards the end of pregnancy, the mother's blood pressure was elevated. The infant was born at 36 weeks gestation weighing 2.2 kg. Initially she was tube fed. When discharged at age 1 month she behaved normally. The early milestones were normal. Subsequently she crawled, stood supported and walked freely at 11, 15 and 18 months respectively. Her first word was at 12 months. Thereafter speech development was slow and articulation poor. She had mild chicken pox at the age of 18 months, and had a susceptibility to respiratory infections, poor appetite and slow weight gain.

The presenting complaint was frequent falls from the age of 18 months. She continued to learn only slowly. She had not lost any skills. At the age of 3½ years height, weight and head circumference were low normal. She had a mild scoliosis, unsteady gait, slightly decreased strength and tone in the legs, equivocal plantar responses and uniformly absent deep tendon reflexes. She was mentally alert. On the Gesell behavioural schedule she functioned between 18 and 24 months in all except the adaptive area which was at 24 month level.

By 4 years of age, her height and weight were below normal. The plantar reflexes were extensor and response to painful and tactile stimuli was impaired in the legs. Strength and tone were diminished in the arms.

At age of 5 years she had nystagmus on lateral gaze and a wide based gait. Temperature and vibration sense were impaired in the legs. She was less alert mentally and responded poorly to auditory stimuli. On the Gesell behavioural schedule she functioned at the 15 - 18 month level. Occasional choreo-athetoid movements were present, especially in the arms, and these movements became more pronounced.

At 6½ years she was irritable, drowsy and unable to walk. Response to painful stimuli was impaired in the arms. She had periods of inappropriate laughing and crying. Her overable behaviour was at less than a 4 month level. Lower cranial nerve palsies were present resulting in facial and bulbar muscle weakness and impaired sensation over the upper part of the face. Attacks of tachycardia and bradycardia were reported. Pupillary light reaction was sluggish.

At the age of 6 years 10 months she was non-responsive. The left limbs were spastic and extended; on the right leg was extended and the arm flexed. Attacks of decerebrate posturing were present. She had occasional generalized seizures. She succumbed a month later from bilateral bronchopneumonia.

Extensive laboratory investigations were noncontributory. Lateral popliteral nerve was studied at age 3½ years and 6½ years. Nerve conduction volocity decreased from 47 to 33 m/sec, amplitude decreased from 4 mV to 1.3 mV, distal latency increased from 2.9 m/sec to 5.4 m/sec, proximal latency from 6.1 m/sec to 9.2 m/sec and nerve action potential decreased from 3 uV to 0. The EEG was initially normal but by 6-½ years showed a slow dysrhythymia. Visual evoked potential and brain auditory evoked response were abnormal. The serum immunoglobulin was always slightly elevated 122-150 mg% with an upper light of normal of 110 mg%. A sural nerve biopsy on E.M. showed a suggestion of increased endoneural collagen and also some loss of unmyelinated axons. Myelinated axons appeared generally intact.

MATERIAL SUBMITTED: 1 H & E stained section

POINTS FOR DISCUSSION: Diagnosis