

CASE 8

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The patient was a _____ male, the product of a normal pregnancy and delivery, and had been apparently normal until 3-1/2 years of age, except that he was "slower" than his siblings. He walked alone at 13 months, was speaking words at 9 months and sentences by the 2nd year.

At 3-1/2 years he had a seizure and had 15 to 20 within the next year. At 4-1/2 years he was walking only with aid, his balance was poor and he was afraid of falling. He had difficulty speaking sentences, "blocked" on some words and was barely intelligible. He was taking Dilantin and phenobarbital for the seizures.

Physical examination showed a child with an intention tremor, "drooling", and "who had signs of cerebellar ataxia." No pyramidal tract signs were observed; cogwheeling was present. The right pupil was fixed but reacted consensually; the left reacted directly but not consensually. There "seemed to be some primary optic atrophy and a question of gaze palsy."

EEG showed "diffuse seizure activity with abnormal slow background activity and the absence of sleep spindles". A year _____ later it was more abnormal with multiple seizure foci mainly in the left hemisphere and maximal in the parietal area.

Pneumoencephalogram showed slight enlargement of the ventricles,

The child was tried on a number of drugs, continued his regression, developed pyramidal tract signs and myoclonic spasms, was institutionalized and died at the age of 8-1/2 years.

Subsequently a sibling has developed similar symptoms.