

CASE 4

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This ten year old male with familial dysautonomia was hospitalized repeatedly because of aspiration pneumonia, apneic episodes, feeding and swallowing difficulties. He also showed poor temperature control with hyperpyrexia, labile blood pressure with occasional hypertension, absent tearing, bilateral corneal ulcers, and slow motor development. He was the third child of a mother whose first baby, a male, died at 3 weeks of age with bilateral aspiration pneumonia, under-nutrition and "congestion of the brain." A second boy is living and well. At two years neurological examination elicited hypotonia, areflexia, good sensory response to pain, incoordination of head, arms and legs, with poor motor development. He was unsteady on his feet but was beginning to walk. Since the age of two years improvement was noted in swallowing, temperature control, intellectual and motor development; however, he did not talk until four years of age. When he was four and a half years old osteochondrosis of the left femoral capital epiphysis was diagnosed and a brace was used until he was eight. There was no scoliosis. During this time his gait was wide based, somewhat shuffling and staggering, with the head thrust forward. He could climb trees. Intellectual function remained adequate. At eight, he showed gross ataxia with dysmetria of the arms. Speech was dysarthric, reflexes were hypoactive, muscle tone increased and pain response was still present. Slow improvement continued in swallowing control over the next two years. He remained unsteady on his feet and staggered slightly. Difficulty in walking did not increase in the dark. His vision was poor. There were no known fundoscopic abnormalities. Serology was never tested. He died suddenly at ten years of age.

Visceral pathology: Acute and chronic pneumonia. Sympathetic chains not examined.

Summary of the CNS lesions: Demyelination of fasciculus cuneatus, most prominent in sacral cord. Process extends through to cervical levels but becomes less obvious. Fasciculus gracilis only minimally involved. Dorsal roots at lower cord levels also show myelin loss. Myelin in brain stem, reticular formation pale and fragmented. Patchy demyelination of the optic nerves, tracts and radiations. Ancient degenerative lesion calcarine cortex (unilateral).

Sections: LFB-CEV stain

Diagnosis: Familial dysautonomia and ataxia (clinical)
Focal demyelination of CNS as indicated above.

Points for discussion:

- 1) Relationship between dysautonomia and ataxia
- 2) Pathological data from other cases of dysautonomia
- 3) Basis for demyelination in optic system
- 4) Primary localization of cord lesion to fasciculus cuneatus