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ATAXIA SYNDROMES

ATAXIA WITH IDIOPATHIC HYPOMYELINATION

A progressive ataxic diplegia syndrome of unknown etiology is reported in 4 unrelated girls evaluated at the National Institutes of Health, Bethesda, MD; Johns Hopkins University, Baltimore, MD; and Tufts, New England Medical Center, Boston, MA. Following normal early milestones, clumsiness and then progressive ataxia developed at 2 to 5 years of age. Seizures occurred in 3 of the 4 patients, some with fever. The ability to walk or sit independently was lost within one year of onset of ataxia. Other symptoms included progressive dysarthria and painful leg cramps. Two had optic atrophy. Deep tendon reflexes were markedly increased, plantar responses were extensor, and ankle clonus was elicited. Cognition was normal in two and mildly delayed in two. Early CTs and MRIs showed diffuse hypodensity of cerebral and cerebellar white matter, and later studies after clinical deterioration showed no progressive change or atrophy. Known metabolic and degenerative diseases were excluded. Open-brain biopsy specimens from two patients showed white matter hypomyelination, demyelination, and gliosis. Myelin-specific proteins and lipid analyses revealed decreased levels. Magnetic resonance spectroscopic imaging showed decrease of N-acetylaspartic acid, choline, and creatine in white matter, a diagnostic feature of the syndrome. (Schiffmann R et al. Childhood ataxia with diffuse central nervous system hypomyelination. Ann Neurol March 1994;35:331-340). (Respond: Dr Schiffmann, National Institutes of Health, Bldg 10, Rm 3D03, 9000 Rockville Pike, Bethesda, MD 20892).

> COMMENT. The MRSI findings appear to be unique to this childhood ataxic syndrome. The degree of white matter hypomyelination found early and before clinical deterioration and the absence of further white matter changes despite worsening of ataxia are remarkable findings.

SENSORY ATAXIA AND VITAMIN E DEFICIENCY

A progressive limb and gait ataxia, distal loss of proprioception and vibration sense, and areflexia, caused by a prolonged and severe vitamin E deficiency, are reported in four patients evaluated as adults at King's College,

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