

abnormal oligoclonal bands in 82%. (Duquette P, Murray TJ et al. Multiple sclerosis in childhood: Clinical profile in 125 patients. J. Pediatr 1987;111:359-63).

COMMENT: MS is probably more common in children than we suspect and the diagnosis should be considered especially in girls with initial sensory or visual symptoms that remit completely and later evolve in a relapsing-remitting manner. An onset at 2 years of age is the earliest case report (Bejar, Ziegler Arch Neurol 1984;41:881). The abrupt rise in incidence that coincides with puberty may be related to hormonal factors. Analysis of data from a Faroe Island epidemic of MS suggested a 2-stage process in the pathogenesis of MS: 1) acquisition of an exogenous factor such as infection and 2) the onset of host factors related to pubescence that allow the pathogenesis to proceed (Fischman HR. AM J Epidemiol 1981;114:244).

Oligoclonal bands in the CSF are the best single laboratory test for the presence of abnormal IgG in patients suspected of having MS. A combination of oligoclonal band and IgG synthesis tests is 97% sensitive for probable and definite MS. (Bloomer LC, Bray PF. Clin Chem 1981;27:2011). NMR imaging differentiates between gray and white matter and is superior to CT in the diagnosis of MS. (Young IR et al. Lancet 1981;2:1063).

DEGENERATIVE AND METABOLIC DISEASES

SPINOCEREBELLAR DEGENERATION

Two children, an 8-year old boy and a 6-year old girl, with progressive ataxia, dysmetria, hypoactive or absent deep tendon reflexes, equivocal plantar response, and sensory impairments, were investigated by pathologists and neurologists at the University of Vermont, Burlington, VT, and University of Saskatchewan, Saskatoon, Canada. The diagnosis of SCD was established by the clinical course and laboratory tests that were normal for arylsulfatase, amino acids, phytanic acid etc.

Rectal biopsy specimens were examined ultrastructurally by electron microscope and by a laser microprobe mass analyzer (LAMMA). Clusters of acicular osmophilic inclusions in the mitochondria of neuronal somata were consistent with crystals of calcium hydroxyapatite (CHA). The calcific nature of the deposits was confirmed by LAMMA. Similar mitochondrial inclusions were found in 10% of smooth muscle cells but not in skeletal muscle and nerve biopsy specimens. Tissue from control subjects had no mitochondrial acicular deposits. It has been suggested that the calcium overload may interfere with mitochondrial enzyme activity by disrupting oxidative phosphorylation. (Munoz DG, Emery ES III, Highland RA. Mitochondrial hydroxyapatite deposits in spinocerebellar degeneration. Ann Neurol 1987;22:258-263).

COMMENT: An abnormal oxidative phosphorylation in muscle mitochondria of patients with Freidreich's ataxia (FA) was previously demonstrated by Stumpf DA et al (Neurology 1982;32:221); mitochondrial malic enzyme activity was 10% of control level in FA fibroblasts. Also, glutamate dehydrogenase deficiency has been noted in cultured skin fibroblasts and leukocyte homogenates of patients with spinocerebellar syndrome (Plaitakis A et al. Ann Neurol 1980;7:297). These studies and the present report may eventually lead to carrier

detection and possible specific therapies for spinocerebellar degenerative disease.

MELAS SYNDROME

Melas syndrome consists of mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke. Three familial cases are described by members of the Departments of Neurology and Pediatrics, University of Texas Health Science Center, San Antonio, TX. In these 3 cases, the onset was in adulthood whereas the majority of previously described patients developed symptoms at 4 to 11 years of age. Early development is usually normal except for short stature. Other features include sensorineural hearing loss, headache, nausea and vomiting, seizures and basal ganglia calcifications by CT. The absence of ophthalmoplegia, heart block, retinal pigmentation, myoclonus, and cerebellar ataxia, seen in other mitochondrial myopathies, is noteworthy. The pathologic findings of MELAS are ragged red fibers, and lactic acidosis. Some have increased carnitine acetyl transferase activity in skeletal muscle.

The assessment of proposed treatments such as methylprednisolone and chlorpromazine is difficult because the course of MELAS is variable. The proband with the full syndrome in this report improved spontaneously and had remained stable for 16 months without therapy. (Driscoll PF, Larsen PD, Gruber AB. MELAS syndrome involving mother and two children. Arch Neurol 1987;44:971-973).

COMMENT: MELAS is familial and inheritance is almost exclusively by maternal transmission. Egger J and Wilson J at the Hospital for Sick Children, Great Ormond Street, London, report a high ratio of affected to unaffected siblings with mitochondrial cytopathy, making Mendelian inheritance unlikely (N Engl J Med 1983;309:142). Two other disorders associated with mitochondrial myopathy and cerebral disease are Kearns-Sayre syndrome and MERRF (myoclonus epilepsy and ragged red fibers). All 3 syndromes are characterized also by dementia, seizures, short stature, hearing loss and a positive family history. K-S syndrome includes ophthalmoplegia, retinal degeneration and cerebellar ataxia. MERRF includes myoclonus and ataxia. MELAS has cortical blindness and hemiparesis as distinctive features.

HEREDITARY PROGRESSIVE DYSTONIA

Four cases of hereditary progressive dystonia with diurnal fluctuation were treated at the Sackler School of Medicine, Tel-Aviv University and the Technion-Israel Institute of Technology, Haifa, Israel. All were sporadic, 3 presented as spastic diplegia or were misdiagnosed as spinocerebellar degeneration, two resembled torsion dystonia, and one had been diagnosed previously as Huntington's chorea and tics. The correct diagnosis was determined by the marked diurnal fluctuation of signs and symptoms, which worsened toward evening, and a prompt, pronounced, and sustained response to levodopa in moderate doses (100-375 mg). Treatment had been continued for 2 to 7 years. Polysomnographic studies were useful in diagnosis and showed increased body movements during REM sleep. Close relatives had increased leg movements in sleep. (Costeff H et al. Fluctuating dystonia responsive to levodopa. Arch Dis Childhood 1987;6:801-804).