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).