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).

COMMENT: This syndrome was first described by Segawa M et al in Japan (Therapy 1971;24:667) and should correctly be referred to as "Segawa Syndrome". Diurnal fluctuation of the dystonia is not invariably present and a trial of levodopa is worthwhile in possible variants of this dystonic syndrome. Emotional disturbance is a feature in some cases and may lead to a diagnosis of psychogenic etiology. In fact, in all cases of dystonia musculorum deformans (torsion dystonia) that I have treated, a diagnosis of conversion hysteria had previously been entertained and pyschotherapy prescribed.

CONGENITAL CNS DEFECTS

HYDROCEPHALUS AND SHUNT INFECTIONS

In the 10-year period, 1973-82, 431 children underwent cerebrospinal fluid shunt insertion for hydrocephalus at Children's Memorial Hospital, Chicago. The authors, now in Verona, Italy (Casella Postale 401.1-37100), have studied the relationship between the etiology of hydrocephalus, age at the time of shunt placement, and infection ate. Meningomyelocele was present in 40%, congenital communicating or obstructive hydrocephalus in 34%, and tumors in 18%. Intraventicular hemorrhage and meningitis were the causes in 5% and 3%, respectively. The age at surgery was less than 1 year in 83% and 1 week or younger in 18%. Each patient had an average of 3 procedures. Infections occurred as a complication of the shunt in 96 patients at rates of 22% per patient and 6% per procedure. Younger patients and those with meningomyelocele were most susceptible to infection. In the meningomyelocele group, infection occurred less often when shunted at 2 weeks of age or later, compared to 1 week or earlier, when the rate was 48%. (Ammirati M, Raimondi AJ. Cerebrospinal fluid shunt infections in children. Child's Nerv Syst 1987;4:106-109).

COMMENT: The rate of operative shunt infection reported in this study is high, and the authors are able to cite similar statistics from two other centers. Attempts to reduce the incidence of infection by perioperative antibiotics or a surgical isolator had not been successful. If a rate of infection of 20% or more per patient is the rule with the operative treatment of hydrocephalus, a reappraisal of techniques and indications for surgery would seem to be a necessity.

Recent experience at Children's Memorial Hospital indicates a rate of infection lower than that reported here, and Dr. Luis Yarsagaray at Loyola Stritch Medical Center, Chicago, recalls only 3 cases of shunt infection in a total of 2000 patients of all ages, both children and adults, that he has himself treated by surgery over a 17 year period (personal communication).

ARNOLD-CHIARI WITH MYELOMENINGOCELE

The outcome of 19 infants with complications of Arnold-Chiari malformation and meningomyelocele was reviewed at the Depts. of Pediatrics, Pathology, and Neurosurgery, University of Pennsylvania School of Medicine and the Children's Hospital of Philadelphia. Vocal cord paralysis and inspiratory stridor alone occurred in 10 (grade I), apnea was an additional symptom in 4 (grade II), and cyanotic spells and dysphagia were associated in 5 (grade III).