

spasticity and weakness, without associated sensory, cerebellar or cranial nerve deficits. The age of onset ranges from 1-55 years and varies within specific kindreds. Heredity may be autosomal dominant, autosomal recessive or sex-linked. Involvement of the spinal cord is unique for a mitochondrial disorder.

SEIZURE DISORDERS

EPILEPSY IN A MITOCHONDRIAL DISORDER

Epilepsy in 9 (22%) of 37 members of a family with maternally inherited mitochondrial disease is reported from the Department of Neurology, University Hospital, Tromsø, Norway. The epilepsy began in infancy or childhood in 3 patients and during adult life in the remainder. The different clinical expressions of epilepsy in the family were myoclonus, partial epilepsy and generalized tonic-clonic seizures. The neuropsychiatric disorders included intellectual deterioration, muscle weakness, pigmental retinal degeneration, progressive hearing loss, cerebellar ataxia and in 1 patient a hemiplegia and paranoid psychosis. The CT scan showed cerebral atrophy, cerebellar atrophy and calcification in the basal ganglia. The EEG abnormalities included slowing and spikes, sometimes focal, and a bilateral paroxysmal response to photic stimulation. A muscle biopsy from the proband revealed a mild defect in the NADH-ubiquinone oxidoreductase step (complex 1) in the respiratory chain (Torbergson T et al. Epilepsy in a mitochondrial disorder. J Neur Neurosurg & Psych Dec 1991; 54:1073-1076). (Correspondence: Dr. Torbergson, Department of Neurology, 9038 University Hospital, Tromsø, Norway.)

COMMENT. The various clinical manifestations of epilepsy including partial motor and/or complex partial epilepsy in different branches of this family were remarkable and worthy of publication. The authors postulate that mitochondrial dysfunctions may be the cause of some epileptic syndromes of undetermined etiology.

CSF TRYPTOPHAN IN INFANTILE SPASMS

The levels of tryptophan (TRP) metabolites in the CSF of 8 patients with infantile spasms are reported from the Department of Pediatrics, St. Marianna University School of Medicine, Kawasaki 213, Japan. Concentrations in patients were compared to TRP metabolites in CSF from 20 age matched controls (mean age: 5.8 months). The levels of CSF serotonin, 5-hydroxyindoleacetic acid and kynurenine were significantly lower in infantile spasm patients compared to controls, whereas the levels of CSF 3-hydroxykynurenine was significantly higher in patients. Tryptophan and 5-hydroxytryptophan were not significantly different in patients compared to controls. The CSF was collected before treatment. The findings suggest that infantile spasms are associated with a decrease in serotonergic metabolites (Yamamoto H, Studies on CSF tryptophan metabolism in infantile spasms. Pediatr Neurol Nov/Dec 1991; 7:411-414). (Correspondence: Dr. Yamamoto,

COMMENT. Yamamoto H has also found a significant inverse correlation with aging in the CSF neurotransmitters during the neonatal period (Pediatr Neurol 1991; 7:406-10). Langlais PJ, Wardlow ML, and Yamamoto H report marked increases in 5-hydroxyindoleacetic acid and decreases in kynurenine levels in the CSF of 5 infants whose infantile spasms were eliminated or reduced following treatment with ACTH. The elimination of seizures by ACTH was accompanied by increased serotonin turnover (Pediatr Neurol 1991; 7:440-5).

Treatment of infantile spasms with IV gamma-globulins (1g/kg/day, the first 2 days, every 3 weeks, for 6 months) is reported in a prospective study of 23 children from the Unite de Neuropediatrie, Centre Gui de Chauillac, 34059, Montpellier, France (Echenne B et al. Brain Dev; 1991; 13:313-9). Five patients responded satisfactorily and 15 received no benefit. Deficiencies of IgA in one patient, IgG2 in 3, and IgG1 in one showed no correlation with therapeutic results. IV gamma-globulin is recommended only as an auxiliary therapy and not the treatment of choice for infantile spasms.

OTHER PAROXYSMAL DISORDERS

PAROXYSMAL OCULAR DOWNWARD DEVIATION

Paroxysmal ocular downward deviation in 13 children with spastic quadriplegia, mental retardation and cortical visual impairment is reported from the Department of Pediatric Neurology, Seirei-Mikatabara General Hospital, Hamamatsu, Shizuoka, Japan. The ocular abnormality was observed in infancy, predominantly in pre-term infants and it resolves spontaneously in many by 2 years of age. The movement differed from the setting sun sign or tonic downward deviation and was not accompanied by retraction of the eyelids. No patient had hydrocephalus or kernicterus (Yokochi K. Paroxysmal ocular downward deviation in neurologically impaired infants. Pediatr Neurol Nov/Dec 1991; 7:426-428). (Correspondence: Dr. Yokochi, Department of Pediatric Neurology, Seirei-Mikatabara General Hospital, Mikatabara 3453, Hamamatsu, Shizuoka 433, Japan.)

COMMENT. The CT and MRI in these patients showed periventricular leukomalacia or multicystic encephalomalacia with involvement of optic radiations. Paroxysmal ocular downward deviation is proposed as a new sign of brain damage with central visual impairment, particularly in preterm infants.