MUSCLE DISEASES

MRI IN CONGENITAL MYOTONIC DYSTROPHY

The results of CT and MRI of the brain were analyzed and neurological development was assessed from the neonatal period in 7 children with congenital myotonic dystrophy aged 2-8 years at the Division of Neurology, Chiba Children's Hospital and Department of Pediatrics, Chiba University School of Medicine, Chiba, Japan. Ventricular dilatation seen on the first day of life in 2 of 3 infants had not progressed on follow-up CTs at intervals of 1-6 years. Areas of periventricular hyperintensity on T2 weighted MRIs were seen in all children, and an asphyxial episode at birth was responsible for the MRI abnormalities. Low developmental quotients ranging from 12 to 72 were not correlated with the extent of the periventricular hyperintensity or the ventriculomegaly (Tanabe Y et al. Neuroradiological findings in children with congenital myotonic dystrophy. Acta Paediatr Aug 1992; 81:613-617). (Correspondence: Dr. Y. Tanabe, Division of Neurology, Chiba Children's Hospital, 579-1 Heta-cho, Chiba, Japan 280-02.)

COMMENT. The mental retardation commonly found in children with congenital myotonic dystrophy was not attributable to brain damage due to perinatal asphyxia. There was no significant correlation between the severity of mental impairment and the degree of structural change in the brain assessed by periventricular hyperintensity on the MRI.

The frequency of cerebral atrophy, large ventricles, and dysmyelination in patients with Fukuyama's congenital muscular dystrophy and occidental congenital muscular dystrophy are compared in a study from Riyadh, Saudi Arabia (Cook J, Gascon G et al. Ann Neurol Sept 1992; 32:439 (abstr)). In 37 children with Fukuyama's congenital muscular dystrophy, 75% were mentally retarded, 62% had cerebral atrophy, 81% large ventricles and 53% dysmyelination. The mental retardation in this disorder is attributed to a brain dysgenesis.

WERDNIG-HOFFMANN: DOMINANT INHERITANCE

A family in which both Werdnig-Hoffmann disease and chronic distal spinal muscular atrophy occurred, with apparent autosomal dominant inheritance, is reported from the Department of Neurology, Mayo Clinic Jacksonville, FL and Johns Hopkins University School of Medicine, Baltimore, MD. The female proband developed symptoms of Werdnig-Hoffmann disease at 2 months of age and died at 10 months. The proband's father and his 2 brothers developed bilateral progressive atrophy and weakness of the hands and legs in their second decade of life. The mother had no symptoms or signs of motor neuron disease, but EMG revealed distal denervation of the limbs. Family studies suggested autosomal dominant inheritance, although the Werdnig-Hoffmann disease may have been influenced by a maternally derived trait. (Boylan KB, Cornblath DR. Werdnig-Hoffmann Disease and chronic distal spinal muscular atrophy with apparent

autosomal dominant inheritance. Ann Neurol Sept 1992; 32:404-407.) (Correspond: Dr. Boylan, Mayo Clinic, 4500 San Pablo Road, Jacksonville, FL 32224.)

COMMENT. Werdnig-Hoffmann disease is generally regarded as an autosomal recessive disorder with linkage to chromosome 5q11.2-13.3. Other studies have suggested genetic heterogeneity.

The prognosis of patients with Werdnig-Hoffmann disease and a clinical scoring system are evaluated by Russman BS et al. (Ann Neurol Sept 1992; 32:439 (abstr)). The 7 criteria for a poor prognosis were poor movement in utero, presence of tongue fasciculation, a poor cry, inability to hold the head at 3 months, inability to roll over by 6 months, loss of function, and diaphragmatic breathing. Five patients with scores of 4-7 died prior to 15 months of age, while 1 patient with a score of 1 died at 31 years. Patients whose scores during the first 6 months of life were 0 or 1 are living and range in age from 3-27 years. Death prior to age 2-4 years is not invariable.

A unique presentation of neonatal adrenoleukodystrophy as a progressive spinal muscular atrophy is reported in 2 siblings who later developed the symptoms and signs of encephalopathy (Miles DK et al. Ann Neurol Sept 1992; 32:466 (abstr)). Peroxisomal diseases must be considered in the differential diagnosis of Werdnig-Hoffmann disease.

HYPEREKPLEXIA REVISITED

The August 1992 issue of Ped Neur Briefs included 3 articles on hyperekplexia. An additional report from the Hammersmith Hospital, London illustrates the difficulties of clinical diagnosis of startle disease (hyperekplexia). At 1 hour after birth the infant had generalized jerks diagnosed as convulsions and treated with phenobarbitone. She had pronounced truncal hypotonia, but on day 3 she developed hypertonia in the limbs, exaggerated tendon reflexes, and jitteriness. The hypertonia diminished during sleep but increased when she was touched. Jerks and stiffening were precipitated by handling, tapping, and especially by turning to the prone position. Pyridoxine and phenobarbitone reduced the frequency and severity of the attacks but did not relieve the touch-induced myoclonus. Clonazepam introduced on day 28 controlled the symptoms. Forcible flexion of the infant also stopped the jerks and stiffening. The measurement of CSF GABA concentration may be helpful in the diagnosis. (Dubowitz LMS et al. Low cerebrospinal fluid concentration of free gamma-aminobutyric acid in startle disease. Lancet July 11, 1992; 340:80-81.) (Correspondence: Dr. L.M.S. Dubowitz, Department of Paediatrics and Neonatal Medicine, Hammersmith Hospital, Du Cane Road, London W12 OHS, UK.)

COMMENT. I am startled by the unexplained rash of recent articles on hyperekplexia! Ryan SG et al. have studied 4 large clonazepam-responsive families with a hereditary hyperekplexia, using a panel of chromosome 5q polymorphic DNA markers. "Multilocus linkage analysis in each family confirms the previous chromosomal assignment