FATTY LIVER AND MCT DIET IN INTRACTABLE EPILEPSY

Fatty infiltration of the liver in 4 children treated with the medium chain triglyceride (MCT) diet is reported from the Depts of Paediatrics and Radiology, Leeds General Infirmary, Belmont Grove, Leeds, England. This was not associated with hepatic dysfunction and resolved after discontinuing the diet. The patient's ages ranged from 4 to 12 years. The seizures were astatic myoclonic in pattern and resistant to medications, including sodium valproate, clobazam, or carbamazepine. The duration of the diet was 2 to 3 years at time of diagnosis by ultrasound scan and, in one case, by liver biopsy. Triglyceride and cholesterol levels were normal in 2 and liver function tests normal in 3 patients tested. Seizure control was improved during treatment with the MCT diet. (Beverley D, Arthur R. Fatty liver and medium chain triglyceride (MCT) diet. Arch Dis Child July 1988;63:840-842).

> **COMMENT**. The mechanism of the fatty liver infiltration was undetermined. The standard ketogenic diet is not always associated with a significant increase in serum lipids but it is accompanied by a fall in blood pH, standard bicarbonate, and blood sugar. (Millichap JG et al. Amer J Dis Child 1964;107:593) The accumulation of excess hydrogen ion within the liver cell is cited as one possible mechanism of fatty change in the liver. The concomitant therapy with anticonvulsant drugs, particularly valproate, might also be contributory.

DEGENERATIVE DISORDERS

RETT SYNDROME

The case-histories of 7 girls and 1 woman, 2 to 25 years of age, are reported from the Clinical Genetics and Child Developmental Center, Department of Maternal and Child Health, Dartmouth Medical School, Hanover, NH. Diagnosis was made after 5 years of age in 4 of the patients when the characteristic hand wringing, hand washing movements were first noted. In addition to these stereotypic hand behaviors, other clinical features included loss of hand function, hyperventilation, bruxism, irritability or self-injury, sleep disturbance, strabismus, seizures, scoliosis, ataxia, and hypotonia in infancy. Head growth deceleration, short stature and retarded rate of growth occurred in almost all cases. Three were small for gestational age at birth and experienced neonatal feeding problems. Previous diagnoses included Angelman syndrome, encephalopathy, and encephalitis, and in 1 case the syndrome appeared to develop as a reaction to pertussis vaccine. Treatment was symptomatic and supportive. Music and motion such as rocking or riding in a car had a calming effect, particularly when screaming attacks and sleep disturbances were troublesome. The absence of a "biological marker" for Rett syndrome makes diagnosis difficult and may lead to confusion with other defined neurodegenerative disorders, such as leukodystrophies, spinocerebellar heredoataxias, neuronal ceroid lipofuscinosis, and ornithine transcarbamylase deficiency. (Moeschler JB et al. Rett Syndrome: natural history and management. Pediatrics July 1988;82:1-10).

COMMENT. The diagnostic criteria for Rett syndrome proposed by the International Rett Syndrome Association (see Ped Neur Briefs 1988;2:29) in April 1988 were separated into 1) necessary, 2) supportive, and 3) exclusion categories. Normal development through the first 6 or 18 months was regarded as a necessary criterion and intrauterine growth retardation and microcephaly at birth were thought to exclude the diagnosis. By these criteria, 4 of the 8 cases described here would not be accepted as examples of Rett syndrome or, alternatively, a "forme fruste" atypical variety of the syndrome might be recognized. Even the female sex is no longer considered a necessary diagnostic criterion and a less restrictive symptom complex is proposed by some. The occurrence of a similar history and syndrome is boys is not uncommon. A plethora of publications on Rett syndrome has appeared in the last 12 months but none has uncovered a specific cause. The present authors note that pertussis vaccine was considered causative in 7 of 19 girls with Rett syndrome reported from Scotland (Br Med J 1985; 219:579), and the onset of regression heralded by inconsolable screaming attacks had followed recent pertussis immunization in 1 patient in their series of 8.

SPINOCEREBELLAR ATAXIA

The onset below 15 years of age of autosomal dominant spinocerebellar ataxia (SCA) in 6 of 41 affected patients is reported from the Dept of Pediatrics, Baylor College of Medicine, Houston, TX. Linkage analysis was performed on 93 individuals in a seven-generation kindred, and strong evidence for linkage of the SCA to the human leukocyte antigen loci on the short arm of chromosome 6 was documented. Age at onset was 6 to 15 years, and clinical findings included ataxia, dysmetria, dysdiadochokinesia, intellectual deficit, ophthalmoparesis, dysarthria, dysphagia, amyotrophy, and cerebellar atrophy on CT scan. Progression was rapid, 1 patient dying 3 years after the onset of symptoms at age 12 years, and 2 patients aged 15 to 26 years are terminally ill. None had seizures, retinal degeneration, or optic atrophy. Poor intellectual performance preceded other neurological abnormalities in 5 children. Of the 6 patients with juvenile onset, 5 were offspring of affected males. This was the first report of childhood onset in the HLA-linked form of SCA. (Zoghbi HY et al. Spinocerebellar ataxia: variable age of onset and linkage to human leukocyte antigen in a large kindred. Ann Neurol June 1988;23:580-584).

COMMENT. The authors comment that all families with dominantly inherited SCA should undergo genetic studies to determine linkage to HLA. Families with the HLA-linked form of SCA may be advised to have HLA typing for presymptomatic or prenatal diagnosis.

The spinocerebellar ataxias are a heterogeneous group of diseases characterized by a slowly progressive loss of neurons in the cerebellum. Friedrich's ataxia, an autosomal recessive trait, presents in childhood, whereas the autosomal dominant varieties of SCA, described by Marie, are usually distinguished by an onset in adult life.