

of age and she developed truncal ataxia and progressive neurologic regression at age two years, shortly after a ten day fever. She was admitted with a diagnosis of acute cerebellar ataxia and later developed dysarthria, mild dysphagia and was then unable to speak two word sentences. She was referred to the Utano National Hospital, Kyoto, three months after the onset. She was well nourished, her hair was normal, and she had no hepatosplenomegaly. Truncal and limb ataxia, horizontal nystagmus, increased reflexes and extensor plantar reflexes were noted. CT showed cerebellar atrophy and symmetric low-density areas in the bilateral periventricular white matter. Sural nerve biopsy revealed axonal degeneration. Blood lactate levels were high (25.8/mg/dl) and serum levels of copper and ceruloplasmin and urinary excretion of copper were low. Cultured skin fibroblasts showed normal copper uptake. Copper given orally 0.1 mg/kg daily for three days resulted in increased serum copper and ceruloplasmin levels, normal granulocyte count, hemoglobin level, blood lactate and pyruvate levels, but neither clinical improvement nor normalization of copper, lactate and pyruvate levels in the CSF were noted. The patient deteriorated slowly, the MRI showed extension of high signal T₂-weighted images and newly appearing lesions in the basal ganglia. At age three years (11 months after onset) she had spastic tetraplegia with respiratory difficulty and was placed on a respirator. (Fujii T et al. Non-menkes-type copper deficiency with regression, lactic acidosis, and granulocytopenia. Neurology August 1991; 41:1263-1266).

COMMENT. The clinical and MRI features of this patient ruled out the diagnosis of Wilson's disease, and Menkes' disease was unlikely in the absence of pili torti and with a normal copper uptake in cultured fibroblasts. The authors speculated that copper transport across the intestinal wall and across the blood brain barrier was impaired in their patient, and that long-term copper deficiency in the brain might have caused irreversible damage to cytochrome *c* oxidase production. (Dr. Fujii is presently at The Burke Rehab Ctr., White Plains, NY).

FOLIC ACID AND NEURAL TUBE DEFECTS

The results of a randomized double-blind prevention trial conducted at 33 centers in seven countries to determine the effects of folic acid supplements around the time of conception in the prevention of neural tube defects is reported by the MRC Vitamin Study Research Group, Department of Environmental and Preventive Medicine, Medical College of St. Bartholomew's Hospital, London EC1, UK. A total of 1,817 women at high risk of having a pregnancy with a neural tube defect were allocated at random to one of four groups - folic acid, other vitamins, both, or neither. Of 27 mothers giving birth to a child with a neural tube defect, six were in the folic acid groups and 21 in the two other groups, a 72% protective effect. Other vitamins showed no significant protective effect. Capsules for those in the folic acid group contained 4 mg of folic acid, one a day until 12 weeks of pregnancy (Wald N et al. Prevention of neural tube defects: results of the Medical Research Council vitamin study. Lancet July 20, 1991; 338:131-137).

COMMENT. This study establishes the specific role of folic acid in the prevention of neural tube defects. Folic acid supplementation in a dose of 4/mg/day is now recommended for all women who have had a previously affected pregnancy, and the diet of all women who may bear children should contain an adequate amount of folic acid. An editorial in this issue of *The Lancet* comments that this supplement did not prevent all neural tube defects and six unsuccessful cases were not accounted for by unusually low serum folic acid concentrations. A heterogeneous aetiology for this group of disorders is suggested. The dosage of folic acid necessary to prevent neural tube defects is not entirely established, and the optimal duration of treatment before conception is uncertain. Can culturally appropriate dietary guidelines be prepared as a matter of urgency for all ethnic groups? Can the requisite folic acid be eaten in food instead of given as a supplement? Providing specific dietary advice about the prevention of malformations to women before pregnancy is a challenging public health problem.

EPILEPTIC SYNDROMES

EPILEPTIC PALATAL MYOCLONUS

A 14 year old girl with epilepsy partialis continua complicated by palatal myoclonus of focal cortical origin is reported from the Comprehensive Epilepsy Center, Graduate Hospital; and University of Pennsylvania School of Medicine, Philadelphia, PA. Her seizures consisted of staring, impairment of consciousness, and left facial twitching. Later she developed intermittent stuttering speech in association with palatal and lingual movements occurring up to 70 times a day. Several episodes of epilepsy partialis continua, lasting up to two weeks, with noted in the next three years. These were characterized by left facial and pharyngeal movements. They were refractory to medications. The EEG revealed theta and delta activity in the right central region with intermixed spikes, and a SPECT showed a focal area of increased perfusion in the right centrotemporal region. The epilepsy partialis continua remitted after several weeks. (Tatum WO et al. Epileptic palatal myoclonus. *Neurology* August 1991; 41:1305-1306).

COMMENT. The myoclonus epilepsies of childhood are divided into three major groups: (1) the progressive myoclonus epilepsies which include a number of degenerative and storage diseases; (2) the myoclonus epilepsies symptomatic of non-progressive brain damage usually of prenatal or perinatal origin; (3) the cryptogenic myoclonus epilepsies (Aicardi J. *Int Pediatr* 1991; 6:195-200). More than 15 specific disorders may cause the progressive myoclonus epilepsy syndrome; reviewed by Berkovic SF et al. (*J Clin Neurophysiol* July 1991; 8:261-274). The major causes of PME are the Unverricht-Lundborg type, Lafora disease, neuronal ceroid lipofuscinoses, MERRF and sialidoses. Myoclonus is best controlled by valproate and/or clonazepam.