MOLYBDENUM COFACTOR DEFICIENCY: DIETARY THERAPY

Moderate short-term clinical improvement was obtained with dietary methionine restriction and cysteine supplementation in a 5-month-old-girl with molybdenum cofactor deficiency (MoCD) treated at Yale University School of Medicine, New Haven, CT. Born to first-cousin parents, the infant's head circumference fell from the 25th percentile at birth to <5th at age 2 months. Developmental delay, spastic quadriplegia, opisthotonus, and irritability were remarkable at 3 months, and a CT showed multiple infarcts. Focal seizures began at 4 months. Laboratory abnormalities included an elevated plasma lactate level, low plasma cysteine, low serum uric acid, and urine positive for sulfites and *S*-sulfocysteine, consistent with sulfite oxidase deficiency. The diagnosis of MoCD was confirmed by absent urinary urothione. Within one month of dietary restriction of methionine to 30 mg/kg/d and supplementation of cysteine to 70 mg/kg/d the infant was more active and alert, irritability resolved, head growth resumed, and seizures were controlled. Urinary sulfites were undetectable. Dietary parental noncompliance resulted in recurrence of irritability, spasticity, opisthotonus, frequent infantile spasms and hypsarrythmia, lack of head growth, regression to a vegetative state by 16 months of age, and elevated sulfites in the urine, >400 mg/L. (Boles RG et al. Short-term response to dietary therapy in molybdenum cofactor deficiency. Ann Neurol Nov 1993;34:742-744). (Respond: Dr Boles, Dep of Genetics, Yale Univ School of Medicine, WWW 305, 333 Cedar Street, New Haven, CT 06510).

> **COMMENT.** MoCD is an autosomal recessive inborn error of metabolism involving 3 molybdenum-requiring enzymes: sulfite oxidase, xanthine dehydrogenase, and aldehyde oxidase. An infant with delayed development, microcephaly, refractory seizures, and lactic acidosis, should be tested for urinary sulfites and serum hypouricemia. Dietary therapy appears promising. The appearance of ectopia lentis, a characteristic finding, is often delayed. For MoCD as an overlooked cause of neonatal seizures, see Ped Neur Briefs July 1993;7:52.

SEIZURE DISORDERS

PROGNOSIS OF INFANTILE SPASMS

The developmental, neurologic, and seizure outcome and prognostic factors of 57 cases of infantile spasms were evaluated in relation to etiology (cryptogenic 17 cases, symptomatic 40 cases) at the Hospital for Sick Children, Toronto, Canada. Age of onset ranged from 4 weeks to 13 months (mean, 5 months), and duration of follow-up was 12 - 60 months. Cerebral dysgenesis was the most common cause (20%) in the symptomatic group. Neurologic deficits, especially hypotonia, were present in 23% of cryptogenic cases cf 75%