COMMENT. Early diagnosis by newborn screening is recommended by the authors. Biotinidase enzyme deficiency and a trial of biotin should be considered in infants or young children with poorly controlled seizures, especially in those with hypotonia, ataxia, skin rash, alopecia, metabolic ketoacidosis, or organic aciduria. Symptoms resolve rapidly after biotin therapy, but neurologic damage may be irreversible if diagnosis is delayed. (see Ped Neur Briefs, Jan 1990).

## MOLYBDENUM-COFACTOR DEFICIENCY AND SEIZURES

The clinical, biochemical, and neuropathological findings in two neonates with molybdenum-cofactor deficiency presenting with convulsions are reported from the Academic Medical Center, Amsterdam, The Netherlands. Patient 1 was admitted at day two with feeding problems, jitteriness, an abnormal cry, apneic spells, and partial and generalized seizures. Head circumference, weight and length were above the 97th percentile. EEGs showed a burst suppression pattern. CT revealed diffuse hypodense ischemic changes. Plasma and urinary cysteine were decreased, and urine sulphite, S-sulphocysteine, taurine, and thiosulphate were increased. Purine analysis showed elevated xanthine and hypoxanthine in the urine, while uric acid was very low. Seizures were refractory, and the infant died on the 10th day. Cultured fibroblasts from a skin biopsy showed absent sulfite oxidase activity. Autopsy findings were meningeal fibrosis, loss of cortical neurons, gliosis and cystic lysis of white matter. Patient 2 was admitted at age 4 days with feeding difficulties, hypertonia, jitteriness, opisthotonus, and high-pitched cry. Generalized and partial tonic-clonic seizures were resistant to treatment. EEG was a multifocal epileptic pattern. An initial diagnosis of postanoxic encephalopathy was changed to molybdenum-cofactor deficiency at 3 years, on reexamination prompted by the revelation of parental consanguinity. Clinically deteriorated, he had spastic tetraplegia, intractable epilepsy, and characteristic metabolic changes. He died 8 months later. Diagnosis was confirmed by liver biopsy and sulfite oxidase and xanthine oxidase analyses. Lens dislocation, a frequent feature of the syndrome, was absent. (Slot HMJ et al. Molybdenum-cofactor deficiency: an easily missed cause of neonatal convulsions. Neuropediatrics June 1993; 24: 139-142). (Respond: Mrs HMJ Slot MD, Dept of Neonatology, Academic Medical Center, Meibergdreef 9, 1105 AZ Amsterdam, The Netherlands).

COMMENT. Molybdenum-cofactor deficiency is an inborn error of metabolism that results in characteristic biochemical and clinical symptoms of sulphite oxidase and xanthine dehydrogenase deficiences. Diagnosis can be made simply with a sulphite strip test in fresh urine and by measuring uric acid excretion. Antenatal diagnosis is possible by chorionic villus sampling and sulphite oxidase assay.