Mandel H, Reiss J, et al. Localization of a gene for molybdenum cofactor deficiency, on the short arm of chromosome 6, by homozygosity mapping. Am J Hum Genet July 1998;63:148-154). (Reprints: Dr Nadine Cohen, Department of Genetics, Tamkin Human Molecular Genetics Research Facility, Technion-Israel Institute of Technology, Bruce Rappaport Faculty of Medicine, POB 9649, Haifa 31096, Israel).

COMMENT. Molybdenum cofactor deficiency (MoCoD) is a fatal inherited, autosomal recessive, disorder manifesting with neonatal seizures unresponsive to therapy, opisthotonus, retardation, craniofacial dysmorphic features, ectopia lentis, and progressive neurologic deterioration. MoCoD is caused by abnormal biosynthesis of the Mo-complexed pterin cofactor for the enzymes: sulfite oxidase, xanthine dehydrogenase, and aldehyde oxidase. Postnatal diagnosis is suggested by hypouricemia and elevated urinary sulfite, and confirmed by sulfite oxidase deficiency in fibroblasts. Microsatellite markers may now be used for prenatal diagnosis, in addition to chorionic villus sampling and sulfite oxidase assay. Dietary therapy with methionine restriction and cysteine supplements has provided short-term clinical improvements (see Progress in Pediatric Neurology II, PNB Publ, 1994;p475).

SEIZURE DISORDERS

INFANTILE EPILEPTIC ENCEPHALOPATHY OF OHTAHARA

A case of infantile epileptic encephalopathy (Ohtahara syndrome) with EEG suppression-bursts at 2 days and associated with diffuse cerebral migrational and maturation disorder, diagnosed at autopsy at 19 months, is reported from Montreal Children's Hospital, McGill University, Montreal, Canada. Seizures of multiple types, usually generalized tonic clonic, began on the first day and persisted until the child died, despite various AEDs, ACTH, and a failed trial of the ketogenic diet. CT at 2 days showed a parietal skull fracture and extradural hemorrhage. MRIs at 1 and 5 months showed delayed myelination, a thin corpus callosum, and increased CSF spaces. EEGs after 5 months of age recorded generalized epileptiform slow spike-and-wave and multifocal abnormalities. CSF exam revealed absence of GABA. Diagnosis of cerebral migration disorder was made only at autopsy by microscopic exam, showing microdysgenesis of cerebral cortex, hippocampal cell loss and astrogliosis, immature neurons, and enlarged dentate and olivary nuclei. (Miller SP, Dilenge M-E, Meagher-Villemure K, O'Gorman AM, Shevell MI. Infantile epileptic encephalopathy (Ohtahara syndrome) and migrational disorder. Pediatr Neurol July 1998;19:50-54). (Respond: Michael I Shevell MD, Department of Neurology, Montreal Children's Hospital, 2300 Tupper Street, Montreal, Quebec H3H 1P3, Canada).

COMMENT. Congenital or acquired structural malformations of cortical development are usually associated with Ohtahara syndrome. Evidence of cortical migration defect may be overlooked on MRI and may be uncovered only at autopsy by microscopic examination.

HIPPOCAMPAL SCLEROSIS IN IDENTICAL TWINS

The genetic and acquired hypotheses of etiology of hippocampal sclerosis (HS) were studied at the University of Melbourne, Australia, using quantitative MRI in three monozygous (MZ) twin pairs, only the index twin having temporal lobe epilepsy and HS. The MZ twin pairs (mean age, 29 years) were compared with 30 age-matched control subjects having no neurologic disorder. All affected twins had HS and a history of prolonged seizures with fever in early childhood.