reported in 10 children studied at the University of Arkansas, Little Rock. (Davis P, Landers A, Gentry B et al. Perceptual and Motor Skills Dec 1997;85:809-810).

MOYAMOYA SYNDROME AND CONGENITAL HEART DISEASE

The association of moyamoya syndrome and congenital heart disease is described in 5 patients at Children's Hospital, Boston, MA. Stroke was the presenting sign in 3 and seizures in 2. Coarctation of the aorta with septal or valvular defects was diagnosed in 3, and tetralogy of Fallot in 2. Moyamoya, diagnosed after surgery for congenital heart disease, was treated by cerebral revascularization. (Lutterman J, Scott M, Nass R, Geva T. Moyamoya syndrome associated with congenital heart disease. Pediatrics Jan 1998;101:57-60). (Respond: Tal Geva MD, Department of Cardiology, Children's Hospital, 300 Longwood Ave, Boston, MA 02115).

COMMENT. Moyamoya syndrome should be considered in the differential diagnosis of stroke or seizures associated with congenital heart disease, both before or after surgery.

METABOLIC DISORDERS

BIOTINIDASE DEFICIENCY: EARLY PRESENTATION

Two infants with manifestations of biotinidase deficiency presenting at age 3 weeks and 2 weeks are reported from the University of Aarhus, Roskilde County Hospital, and Herning Central Hospital, Denmark. Patient 1, born to related Kurdic parents, developed a generalized skin rash at 3 weeks, generalized tonic-clonic seizures up to 20 times daily at 6 weeks, and visual inattention, hypertonia and hyperreflexia on admission at 8 weeks. EEG showed epileptiform activity. Valproic acid was ineffective. Metabolic screening showed urinary B-hydroxyisovalerate and B-methylcrotonylglycine, and very low serum biotinidase activity. After oral biotin (5mg x 3 daily) the seizures stopped within a few days, and at 2 year follow up psychomotor development was normal except for hearing loss. MRI showing cerebral atrophy initially was normal at 12 months. Patient 2, the second child of related Kurdic parents, presented at 1 hour after birth with respiratory distress and septicemia. She had dry and squamous skin at 2 weeks, loss of hair at 4 weeks, and on readmission at 6 weeks she was lethargic, hypotonic, and hypothermic. Visual inattention, trembling, tense fontanelle, tonic clonic seizures, conjunctivitis, and alopecia were noted. EEG showed diffuse slowing and a right occipital spike focus. CT was suggestive of periventricular leukodystrophy. Serum lactate and pyruvate were elevated. Organic aciduria and absent serum biotinidase confirmed the diagnosis of biotinidase deficiency. Oral biotin (10mg daily) was begun at 7 weeks, and seizures were controlled and other manifestations improved within 2 weeks. At 18 month follow-up, development and CT were normal. The authors advocate routine neonatal screening for biotinidase deficiency in Denmark. (Haagerup A, Andersen JB, Blichfeldt S, Christensen MF. Biotinidase deficiency: two cases of very early presentation. Dev Med Child Neurol Dec 1997;39:832-835). (Respond: Dr Annette Haagerup, Institute of Human Genetics, University of Aaarhus, DK-8000 Aarhus C, Denmark).

COMMENT. Biotinidase deficiency is an autosomal recessive disorder causing multiple carboxylase deficiency and usually manifested at 3 to 6 months of age with intractable seizures, hypotonia, skin rash, alopecia, and developmental delay. Lactic acidosis leads to coma and death in untreated cases.

Late presentation of biotinidase deficiency is described in a previously healthy 5-year-old girl who developed acute visual loss and optic atrophy, and an ataxic gait. Classical signs of biotinidase deficiency were absent. (Rahman S, Standing S, Dalton RN, Pike MG. Dev Med Child Neurol Dec 1997;39:830-831).

Biotin deficiency and chronic anticonvulsant therapy. Nine adults treated with various anticonvulsants, including phenytoin and carbamazepine, compared to 17 controls showed a twofold increase in the 24-hour urinary excretion of bisnorbiotin, biotin sulfoxide, and 3-hydroxyisovaleric acid, metabolites of biotin, whereas urinary and serum biotin concentrations were unchanged. Long-term treatment with anticonvulsants may be associated with an increased biotin catabolism. (Mock DM, Dyken ME. Neurology Nov 1997;49:1444-1447).

DEGENERATIVE DISEASES

MITOCHONDRIAL DNA MUTATION IN RETT SYNDROME

Analysis of mitochondrial DNA from 15 children with Rett syndrome (RS) and 14 of their mothers is reported from the Department of Pediatrics, Beijing Medical University, China. Polymerase chain reaction amplification and single strand conformation polymorphism analysis showed mutations in region 2650-3000 encoding 16S rRNA of mtDNA in 13 patients with RS and 11 mothers. DNA sequence analysis and mismatch PCR results confirmed a point mutation (C --> T) at position 2835 in 7 patients with RS and in 6 of their mothers, that was absent in controls. (Tang J, Qi Y, Bao X-H, Wu X-R. Mutational analysis of mitochondrial DNA of children with Rett syndrome. Pediatr Neurol Nov 1997;17:327-330). (Respond: Dr Xi-Ru Wu, Department of Pediatrics, The First Teaching Hospital, Beijing Medical University, Beijing 100034, PR China).

COMMENT. Most cases of Rett syndrome are sporadic, but a few familial examples are reported. A maternal inheritance pattern suggests that mitochondrial DNA may be involved. The mutations observed in the mtDNA of patients with Rett syndrome and their mothers lends support to the hypothesis of a genetic basis for the disorder in some cases.

Japanese monozygotic female twins with Rett syndrome are reported from Fukuoka University, Japan. (Ogawa A, Mitsudome A, Yasumoto S, Matsumoto T. Brain Dev Dec 1997;19:568-570). The two 28-year-old patients had discordant characteristics regarding seizures, scoliosis, and stereotypic hand movements in adolescence. The authors cite 7 pairs of monozygotic twins with RS reported in the literature, and 11 pairs of dizygotic twins, only one twin affected, always the female.

CSF SUBSTANCE P LEVELS IN RETT SYNDROME

The cerebrospinal fluid (CSF) levels of neuropeptide substance P were measured in 20 patients with Rett syndrome and controls at Kurume University, Japan, and other centers. CSF substance P levels are constant in control children between 2 and 12 years, and show a gradual decrease through adolescence, reaching a plateau at 20 years. Significant reductions in substance P in patients with RS compared to controls were present at the early phases of the disease, at age 2 to 3 years, and were not age dependent. Childhood RS levels were 50% of controls in the same age group, and adults with RS had 37% of control adult