MOYAMOYA DISEASE

Sixteen children with moyamoya disease and involvement of the posterior cerebral artery are reported from the Departments of Neurophysiology, Neurosurgery, and Radiology, Faculty of Medicine, Kyushu University, Fukuoka, Japan. Eight patients had complete occlusion of posterior cerebral arteries and the other eight had nonocclusive disease. All patients showed patent ophthalmic arteries bilaterally. Pattern-reversal visual-evoked potentials showed abnormalities in 75% of the posterior cerebral artery occlusive group and no abnormalities in the nonocclusive group. Abnormalities were also found in positron emission tomography, computed tomography, and the clinical examination of the visual fields. The authors concluded that the pattern-reversal visual-evoked potentials was the most practical means to explore posterior cerebral artery occlusion in the course of moyamoya disease. (Tashima-Kurita S et al. Moyamoya disease. Posterior cerebral artery occlusion and pattern-reversal visual-evoked potential. Arch Neurol May 1989; 46:550-553).

COMMENT. Moyamoya disease is characterized by progressive occlusion of cerebral arteries and predominantly the anterior and middle cerebral arteries. The incidence of posterior cerebral artery involvement and visual disturbances is approximately 25% and the clinical manifestations include decreased visual acuity, homonymous hemianopsia, constriction of the visual fields, and scintillating scotoma.

METABOLIC DISORDERS

CNS COMPLICATIONS OF CYSTINOSIS

Fourteen patients with cystinosis, eight males and six females ranging in age from 13 to 24 years (mean 18.1 years) were examined for neurological involvement at the National Institute of Neurologic and Communicative Disorders and Stroke, the National Institutes of Health Clinical Center, Bethesda, Maryland. Two patients had neurological symptoms, including bradykinesia, dementia and spasticity, and behavioral and cognitive disturbances; 12 patients had CT evidence of generalized cerebral atrophy; two had multifocal intracerebral mineralization on CT scan; two had abnormal electroencephalograms and only one patient was entirely normal. Patients with neurologic symptoms or markedly abnormal CT scans were older and had a longer interval between their initial renal transplantation and the examination at follow-up than those patients who were normal or who had only mild cerebral atrophy. The neurologic and neuropsychometric abnormalities correlated with the degree of roentgenographic abnormality. The patients with nervous system abnormalities were not distinguished by patterns of medication use or the relative severity of cystinosis. The differential diagnosis included other complications from renal failure, dialysis and immunosuppression. (Fink JK et al. Neurologic complications in long-standing nephropathic cystinosis. Arch Neurol May 1989; 46:543-548).

COMMENT. The central nervous system involvement in nephropathic cystinosis has not been implicated until recently. Survival into adulthood following renal dialysis and transplantation has drawn attention to the sequelae of long-standing cystinosis. Cystinosis is a rare autosomal recessive disorder of children and adults characterized biochemically by the intracellular accumulation of cystine crystals in the kidneys, bone marrow, and cornea as well as the reticuloendothelial system. With increasing longevity, involvement of other organs such as the thyroid, pancreas, and central nervous system has become apparent. Three different patterns of the disease are recognized depending on the degree of cystine accumulation: Infantile nephropathic form, intermediate or late onset adolescent form, and a benign adult form. The clinical manifestations of infantile cystinosis include recurrent episodes of dehydration, the Fanconi renal tubular syndrome, retarded growth, anemia, photophobia, retinopathy, and vitamin D resistant rickets. Long-term treatment with Cysteamine may delay or prevent the accumulation of cystine crystals in various organs and may alter the prognosis. (daSilva VA et al. N Engl J Med 1985; 313:1460). Biopsy specimens of cerebral cortex and meninges have revealed cystine crystals in the walls of arachnoidal blood vessels and in the cytoplasm of cortical neurons, basal ganglia, thalamus, cerebellum and posterior pituitary. The relationship of these crystalline deposits to neurological abnormalities has not been determined.

MANNOSIDOSIS AND COGNITIVE FUNCTIONING

Longitudinal assessments of the biochemistry and cognitive functioning in three brothers with mannosidosis are reported from the Department of Pediatrics in Human Development; Michigan State University, East Lansing, Michigan. The patients were followed from three or four years of age. The biochemical findings demonstrated profound deficits of leukocyte alpha mannosidase that remained stable over time and were very similar to levels of the same enzyme activity in fibroblasts. Cognitive tests including general intelligence, language, visual spatial skills, and overall adaptive abilities, were generally uniform with no signs of progressive deterioration except for receptive language abilities. When examined initially the patients were mildly retarded. Loss of receptive vocabulary abilities seen above the age of six years may have been related to a conductive hearing loss of 30-40 dB and frequent otitis media. There was a lack of correspondence between the level of enzyme deficiency and the degree of mental dysfunction. Sequential data were obtained for six years for the oldest brother, five years for the middle brother, and four years for the youngest brother. The authors suggest that hearing should be constantly examined to address potential sensory deprivation as it affects cognitive functioning in children with mannosidosis. (Noll RB et al. Long-term follow-up of biochemical and cognitive functioning in patients with mannosidosis. Arch Neurol May 1989; 46:507-509).

COMMENT. Children with mannosidosis have coarse features, slight hepatosplenomegaly and psychomotor retardation. After two