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

years of age, growth slows down, the tongue enlarges and a lumbar kyphosis and prominent forehead develop. Based on the findings in this study, overall intellectual functioning was low but showed no evidence of progressive deterioration except for receptive language skills.

BRAINSTEM AUDITORY RESPONSE TEST

BAER IN HIGH RISK INFANTS

Brainstem auditory responses (BAER) performed on 667 high risk infants from an infant special care unit were evaluated at the Department of Otolaryngology, University of Texas Medical Branch, Galveston, Texas. Infants who failed the test were classified into two groups; those who failed at 30 dB hearing level and those who failed at 45 dB hearing level. At follow-up examination in one, three, or six months, 8 (1.2%) had severe sensorineural hearing impairments (since only 50% returned for follow-up, 2.4% was a more accurate incidence). Conductive hearing loss was found in 15.7% (17/108) of those who passed 30 dB level and in 34.3% (12/34) of those who failed. The use of BAER testing at levels less than 45 dB permitted detection of middle ear disorders. All of the infants who failed at 45 dB hearing level and had abnormal results at the 3-4 month follow-up examination had severe sensorineural or moderate to severe mixed hearing losses. For the group that failed at 30 dB hearing level and were abnormal at follow-up, 80% had conductive hearing disorders and 20% had mild sensorineural hearing impairments. Infants enrolled in a parent-infant program for hearing impairment by 6 months of age were referred from the BAER program. (Kramer S J et al. Auditory brainstem responses and clinical follow-up of high-risk infants. Pediatrics March 1989; 83:385-392).

COMMENT. The brainstem auditory evoked response test (BAER) is effective in the early detection of hearing impairments in high risk neonates, and the degree and type of hearing loss may be predicted. However, the children who were referred to the BAER program represented only 31% of the total number of parent-infant program children with congenital hearing impairment and only 50% of the children with multiple handicaps. Some of the hearing impaired children entering the parent-infant program at this center during the period of the study were referred from sources other than the BAER program and were much older when enrolled. An infant hearing assessment program for only high risk infants would fail to identify approximately one-half of hearing impaired children.

TAURINE AND BAER MATURATION

A blinded randomized trial of taurine supplementation of preterm infants was conducted at the Department of Pediatrics, University of Texas Southwestern Medical Center, Dallas, and Ross Laboratories, Columbus, Ohio. Infants who received taurine supplementation had more mature brainstem auditory evoked responses with a reduction in the interval between stimulus and response at two different stimulation