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NUTRITIONAL AND METABOLIC DISORDERS

CHLORIDE-DEFICIENT DIET AND COGNITIVE DEFICITS

Distinctive cognitive impairments and a specific neurobehavioral syndrome are reported in 11 of 13 children who had failed to thrive on chloride-deficient formula during infancy. They were examined four to nine years later at the Division of Pediatric Neurology, UCLA School of Medicine, Los Angeles, CA and the Department of Neurology, Boston University School of Medicine, Boston, MA. All patients during infancy had developed symptoms of hypokalemic, hypochloremic metabolic alkalosis and/or growth deceleration, concurrently with their exposure to chloride-deficient Neo-Mull-Soy formula. Head circumference at follow-up was within normal limits, three children had hypotonia, two diffuse hyperreflexia, one had choreiform movements and one bilateral intention tremor. The EEG was abnormal in six, with paroxysmal discharges seen in five children. CT and MRI studies were normal. Intelligence was within the average range for 11 of the 13 children tested. All had a neurobehavioral syndrome consisting of a language disorder primarily involving articulation, word finding and naming; visual-motor and fine motor difficulties; and attention deficit disorder often featuring repetitive behaviors, withdrawal and "over-focus". (Kaleita TA, Kinsbourne M, Menkes JH. A neurobehavioral syndrome after failure to thrive on chloride-deficient formula. Dev Med Child Neurol July 1991; 33:626-635).

COMMENT. A syndrome characterized by loss of appetite, failure to gain weight, muscular weakness, lethargy, vomiting and severe hypochloremic, hypokalemic, metabolic alkalosis has been described in infants fed chloride-deficient soy-based infant formulas. Clinical features resemble Bartter Syndrome, but the renal biopsy is normal and

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infants recover when dietary chloride supplements are provided. Previous follow-up reports have drawn attention to developmental delay (Chutorian et al. Pediatr Neurol 1985; 1:334-341) and the present study demonstrates specific cognitive, language and attentional deficits in later childhood.

MAPLE SYRUP URINE DISEASE AND CEREBRAL EDEMA

Cerebral edema causing death in four children with maple syrup urine disease (MSUD) is reported from St. Christopher's Hospital for Children, Temple University School of Medicine, Philadelphia, PA. An intercurrent infection that caused severe dehydration and acidosis precipitated the cerebral edema when the children were three to five years of age. All four had been adequately managed with few problems before the intercurrent illness. The diagnosis of cerebral edema was established by autopsy in one patient and demonstrated by CT in two, one also having a subarachnoid hemorrhage. The authors recommend early hospitalization and cautious rehydration in all children with MSUD in whom intercurrent infections develop in association with decreased nutrient intake or vomiting, or both. Early treatment of dehydration and acidosis may prevent the catastrophic consequences observed (Riviello JJ Jr et al. Cerebral edema causing death in children with maple syrup urine disease. J Pediatr July 1991; 119:42-45).

COMMENT. Acute metabolic decompensation in MSUD during otherwise minor illnesses has generally been presumed to result from massive release of leucine from protein catabolism. The dynamics of protein metabolism and implications for management are reported from the Murdoch Institute and the Department of Dietetics, Royal Children's Hospital, Melbourne, Australia, and the Nutrition Research Group, Clinical Research Centre, Harrow, UK (Thompson GN et al. J Pediatr July 1991; 119:35-41). Fasting appeared to be a more important cause of increased leucine levels than the catabolic effect of infection. Branched-chain amino acid restriction should be commenced at the start of minor illness in children with MSUD and the intake of other nutrients should be increased. Dietary supplementation reduces the risk of metabolic decompensation during acute illnesses and early and meticulous treatment of MSUD results in intellectually normal children according to a study from the Division of Biochemical Development and Molecular Diseases, Children's Hospital of Philadelphia, University School of Medicine, Philadelphia (Kaplan P et al. J Pediatr July 1991; 119:46-50). Affected children treated presymptomatically had higher IQ scores than their siblings treated when their disease was symptomatic.

NON-MENKES TYPE COPPER DEFICIENCY

A two year old girl with copper deficiency progressive neuronal disorder and granulocytopenia is reported from the Department of Pediatrics, Kyoto University, Japan. A familial granulocytopenia was noted at one month