

COMMENT. The authors comment that these MRI findings may be specific to infantile neuroaxonal dystrophy and permit early diagnosis. The clinical picture includes upper and lower motor neuron deficits, including progressive weakness, difficulty in walking, hypotonia, muscle atrophy, hyperactive reflexes, Babinski signs, and optic atrophy. Diagnosis is confirmed by peripheral nerve biopsy, which shows globular swellings on the axons. Axonal swelling may also occur in Hallervorden-Spatz disease, Friedreich's ataxia, and other neurodegenerative diseases, however.

MRI IN INFANTILE KRABBE DISEASE

Serial MRI findings paralleling the clinical deterioration of a year-old child with Krabbe disease are reported from the University of Rochester Medical Center, New York. At 13 months, an abnormal high signal on T2-weighted images in the frontoparietal white matter and cerebellar white matter was correlated with severe developmental delay, increased tone, and hyperextension (stage 2). At 32 months, MRI changes had progressed to include central and cortical atrophy, decreased white matter volume, thalamic and caudate atrophy, and abnormal high signal in all motor tracts except the anterior limb of the internal capsule. Clinically, the child was in stage 3, with spasticity of the upper extremities, peripheral neuropathy, and no response to auditory or visual stimuli. (Farley TJ et al. Serial MRI and CT findings in infantile Krabbe disease. Pediatr Neurol Nov/Dec 1992; **8**: 455-458). (Correspondence: Dr Ketonen, Dept Radiology, University of Rochester Medical Center, Box 648, 601 Elmwood Ave, Rochester, NY 14642).

COMMENT. The 3-stages of Krabbe disease described by Hagberg begin with hyperirritability, restlessness, and frequent crying. Convulsions may develop, induced by sensory stimuli. Development becomes delayed, tone is increased, and later, reflexes are difficult to elicit and are absent in the lower limbs. Terminally, the infant is flaccid and blind. CSF protein is elevated and nerve conduction is delayed. The absence of the enzyme galactosyl ceramide B-galactosidase in leukocytes and skin fibroblasts is diagnostic, a test result common to all variants and stages of Krabbe disease, including antenatal.

BIOCHEMICAL MARKER FOR MENKES DISEASE

Plasma and CSF levels of catechols in 10 patients with Menkes disease, ranging in age from 9 days to 27 months, were compared with control groups and patients with congenital absence of dopamine-B-hydroxylase (DBH) at the National Institutes of Health, Bethesda, MD. The neurochemical pattern in Menkes disease patients was characterized by high dihydroxyphenylalanine (DOPA), dopamine (DA), and dihydroxyphenylacetic acid (DOPAC) levels,

reduced dihydroxyphenylglycol(DHPG) levels, and high ratios of DOPA:DHPG and DOPAC:DHPG. In contrast to patients with absent DBH, norepinephrine (NE) levels were normal in 4 Menkes patients and in the CSF of all 10 patients. The pattern was consistent with partial deficiency of DBH activity and compensatory increases in catecholamine biosynthesis in sympathetic nerves and brain. The changes may provide a biochemical marker for Menkes disease. (Kaler SG et al. Plasma and cerebrospinal fluid neurochemical pattern in Menkes disease. Ann Neurol Feb 1993; **33**: 171-175). (Correspondence: Dr Kaler, National Inst of Health, Bldg 10, Rm 9S 242, 9000 Rockville Pike, Bethesda, MD 20892).

COMMENT. Menkes disease (Kinky-Hair disease) is a sex-linked recessive, neurodegenerative disorder of gray matter involving copper metabolism. Clinically, it is characterized by failure to thrive, cherubic facies, twisted and fractured hair, seizures, hypotonia, and progressive psychomotor deterioration. Biochemically, serum copper and ceruloplasmin levels are reduced or low-normal, and copper uptake by cultured fibroblasts is abnormally increased, permitting intrauterine diagnosis of the disease. Copper levels in liver and brain are low, but are high in the intestine and kidney. The incorporation of copper into enzymes such as DBH requiring this cofactor is impaired. DBH activity was assessed in the present study by measuring levels of DA, NE, and metabolites to provide a picture of catecholamine synthesis and turnover in infants with Menkes disease.

NEUROPATHIES

PROGRESSIVE MONONEUROPATHY

Six patients, aged 10 to 27 years, with insidiously progressive, painless weakness in the distribution of a single major lower extremity nerve are reported from the University of California, San Francisco. The duration at diagnosis varied from 3 months to 8 years. The sciatic nerve was involved in three patients, the common peroneal in two, and the femoral nerve in one. The appearance of the nerve at surgery was normal in two, atrophied in two, and fibrosed in one. EMG revealed a chronic axonal mononeuropathy without conduction block or focal conduction slowing. MRI, CT, and US imaging failed to reveal a nerve mass or compression. (Engstrom JW et al. Idiopathic, progressive mononeuropathy in young people. Arch Neurol Jan 1993; **50**: 20-23). (Reprints: Dr Engstrom, Box 0114, M794, Department of Neurology, University of California, San Francisco, CA 94143).

COMMENT. The authors ask the question: Could the patients have had a nerve tumor not uncovered by standard MRI or surgery and too proximal for localization by nerve conduction studies? The diagnosis of