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NEUROMUSCULAR DISORDERS

PERIPHERAL NEUROPATHY IN KRABBE'S DISEASE

A 13-year-old female child presenting with scoliosis, pes cavus, distal lower extremity weakness, sensory loss, and delayed nerve conduction velocities, initially diagnosed as Charcot-Marie-Tooth disease, later developed lower extremity spasticity and laboratory evaluations were consistent with Krabbe's disease, in a report from the Alfred I duPont Institute, Wilmington, DE. An MRI showed diffuse white matter demyelination. Galactocerebroside beta-galactosidase assay levels were abnormally low. Nerve biopsy showed Schwann cell inclusions consisting of globoid clusters and signs of demyelination. DNA analysis identified two mutations, one allele derived from the mother that abolished expression of the enzyme GCBGase, and the other associated with some enzyme activity and accounting for the late onset and slow progression of the disease. (Marks HG, Scavina MT, Kolodny EH, Palmieri M, Childs J. Krabbe's disease presenting as a peripheral neuropathy. Muscle & Nerve August 1997:1024-1028). (Respond: Dr Harold Marks, Division of Neurology, Alfred I dePont Institute, PO Box 269, Wilmington, DE 19899).

COMMENT. Globoid cell leukodystrophy (Krabbe's disease) is an autosomal recessive disorder that usually presents acutely at or before 6 months of age with irritability, stiffness, tonic spasms, optic atrophy, hyperacusis, and terminally, flaccidity and absent reflexes. CSF protein is elevated and motor nerve conduction velocities are reduced. Near absence of leukocyte or skin fibroblast GCBG or galactosyl ceramidase is a definitive diagnostic test. Late-onset cases may account for 10-15%, and present between 2 and 3 years, 3 and 10 years, or after 10 years. Clinical presentations are variable, and the juvenile or adult-onset forms are often misdiagnosed. A Babinski sign in a patient with an otherwise characteristic peripheral neuropathy should prompt an evaluation for Krabbe's disease.

MOTOR NEUROPATHY IN VARICELLA

An acute motor axonal neuropathy developed within 4 days of the

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