HALLERVORDEN-SPATZ DISEASE: CLINICOPATHOLOGY

Clinical and pathological features of familial late infantile Hallervorden-Spatz disease (HSD) are reported in two sisters, one of whom died at 11 years, from the Institute for Neurological Sciences, University of Siena, Italy. Clinical diagnosis was confirmed by the classical "eye of the tiger" sign in the MRI. The appearance of the globus pallidus on MRI correlated with the pathological findings, showing pallidal axonal spheroids and iron deposits without involvement of the substantia nigra. Clinically, retinitis pigmentosa, acanthocytosis, and neuromuscular involvement with increased serum creatine kinase were observed in both patients. HSD is classified as a form of neuroacanthocytosis, along with choreo-acanthocytosis, McLeod syndrome, and HARP syndrome. These diseases have the following clinical features in common but variable in frequency: 1) acanthocytosis, 2) extrapyramidal movements, 3) neuromuscular involvement, and 4) retinitis pigmentosa. (Malandrini A et al. Clinicopathological study of familial late infantile Hallervorden-Spatz disease: a particular form of neuroacanthocytosis. Child's Nerv Syst March 1996;12:155-160). (Respond: Dr A Malandrini, Institute for Neurological Sciences, University of Siena, Viale Bracci, 2, I-53100 Siena, Italy).

COMMENT. Hallervorden-Spatz disease is a rare, progressive, and fatal degenerative disorder, with onset in late infancy, childhood or adulthood, characterized by a bizarre gait and speech disturbance, dystonic postures and choreo-athetotic movements, mental deterioration, retinitis pigmentosa, and occasionally, acanthocytosis. The coexistence of HSD and acanthocytosis in 3 sisters was reported by Swisher CN, Menkes JH, Cancilla PA and Dodge PR. Trans Am Neurol Assoc 1972;97:212. An autosomal recessive inheritance is suggested by familial cases. Diagnosis may be confirmed by the MRI and the "eye of the tiger sign" affecting the globus pallidus. For further reviews of HSD and related disorders, see Ped Neur Briefs Nov 1995;9:85, and Progress in Pediatric Neurology II, PNB Publ, 1994, p 477.

MUSCLE DISORDERS

DIAGNOSIS OF CONGENITAL MUSCULAR DYSTROPHY

Patterns of alkaline and acid phosphatases were compared with the distribution of merosin and dystrophin staining in muscle biopsies from 20 children with congenital muscular dystrophy (CMD) examined at the Department of Neurology, Washington University School of Medicine, St Louis, MO. A ratio of AcP:AlkP staining was calculated for each biopsy. In 9 patients with CMD with normal dystrophin, the AcP:AlkP ratio was low, whereas in 3 patients with CMD and reduced dystrophin and in 7 with Duchenne muscular dystrophy, the ratio was up to 15 times higher. Low AcP:AlkP ratios were correlated with absence of AcP-positive cells. Merosin staining was absent in 5 of 17 CMD patients, none of whom could walk, whereas all 12 with merosin-positive stains walked. (Connolly AM, Pestronk A, et al. Congenital muscular dystrophy syndromes distinguished by alkaline and acid phosphatase, merosin, and dystrophin staining. Neurology March 1996;46:810-814). (Respond: Dr Alan Pestronk, Department of Neurology, Box 8111, Washington University School of Medicine, 660 S Euclid Ave, St Louis, MO 63110).

COMMENT. Biopsies showing few acid phosphatase-positive cells in association with numerous alkaline phosphatase staining muscle fibers are