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METABOLIC-DEGENERATIVE DISEASES

HARP SYNDROME

The clinical and laboratory studies of an 11 year old girl with hypoprebetalipoproteinemia, acanthocytosis, retinitis pigmentosa and pallidal degeneration are reported from the Developmental and Metabolic Neurology Branch, National Institute of Neurological Disorders and Stroke and Clinical Pathology Department, National Institutes of Health, Bethesda, MD. Development was normal until age 3 years, when spasticity began in the lower extremities and this was followed by language deterioration, generalized dystonia, orofacial dyskinesia, progressive dementia, and deterioration of visual acuity. At age 11 the fundi showed atypical retinitis pigmentosa and MRI showed a marked signal decrease in the pallidal nuclei on T_2 weighted images and the "eye-of-the-tiger" sign. Thirty to 40% acanthocytes were present in the peripheral blood and high resolution lipoprotein electrophoresis revealed an absence of the pre-beta fraction. Phase-contrast microscopy of whole blood showed 80-90% acanthocytes, confirmed by electron microscopy. Previous reports in 6 patients linking Hallervorden-Spatz disease with acanthocytosis have not included the orofacial dyskinesia and abnormal lipoproteins seen in this patient (Higgins JJ et al. Hypoprebetalipoproteinemia, acanthocytosis, retinitis pigmentosa, and pallidal degeneration (HARP syndrome). Neurology Jan 1992; 42:194-198). (Reprints: Dr. Joseph J. Higgins, Developmental and Metabolic Neurology Branch, NIND&S, National Institutes of Health, Bldg. 10, Rm. 3D03, Bethesda, MD 20892.)

COMMENT. This group of signs described by the acronym HARP syndrome has features in common with the rare association of Hallervorden-Spatz disease and acanthocytosis (Swisher CN, Menkes JH, Cancilla PA, Dodge PR. Trans Am Neurol Assoc 1972; 97:212) and with the syndrome of choreoacanthocytosis (Feinberg TE et al. Neurology 1991; 41:1000). A defect of vertical gaze noted in the present case report was also recorded in 3 sisters reported by Swisher et al.

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