DEGENERATIVE AND METABOLIC DISEASES

DIAGNOSIS OF HALLERVORDEN-SPATZ DISEASE

The *in vivo* diagnosis of Hallervorden-Spatz disease is discussed in relation to the clinical manifestations and MRI findings in two children examined at the Department of Paediatrics, University Hospital of Aarhus, Denmark. Characteristic clinical findings of the late infantile type are a gradual onset with gait disturbance, corticospinal tract signs, rigidity and dystonia, especially oromandibular involvement, and mental deterioration. MRI may be normal at first and will later show hypo-intensity in the globus pallidus in T2-weighted images and an area of hyperintensity in the anteromedial portion, corresponding to the 'eye-of-the-tiger' sign. In some cases, both globus pallidus and substantia nigra are involved, showing hypo-intensities consistent with iron deposition. The diagnostic findings in five additional cases reported in the literature are also tabulated. Age at onset ranged from 1 to 4 years. MRIs were positive when examined at 7 to 12 years. (Ostergaard JR et al. *In vivo* diagnosis of Hallervorden-Spatz disease. Dev Med Child Neurol Sept 1995;37:827-833). (Respond: JR Ostergaard MD PhD, Department of Paediatrics, Aarhus Kommunehospital, University Hospital of Aarhus, DK-8000 Aarhus, Denmark).

> COMMENT. Hallervorden-Spatz disease occurs as 1) a classic post-infantile type, with onset between 7 and 15 years; 2) late infantile type, with onset before 6 years of age and leading to death within 10 years; and 3) an adult form, onset between 20 and 60 years, and fatal within 10 years. In addition to the clinical and MRI findings, the diagnosis of Hallervorden-Spatz disease is made by exclusion of other neurodegenerative disorders, some having identical MRI changes, including the 'eye-of-the-tiger' sign. The rare association of Hallervorden-Spatz disease and acanthocytosis has been described by Swisher CN et al (Trans Am Neurol Assoc 1972;97:212), and HARP syndrome, characterized by hypoprebetalipoproteinemia, acanthocytosis, retinitis pigmentosa, and pallidal degeneration, may include the 'eye-of-the-tiger' sign in the MRI. Reports of HARP syndrome and commentaries are included in Progress in Pediatric Neurology II, PNB Publishers, 1994, p477; and Ped Neur Briefs April 1995;9:26-27).

TESTS FOR SUSPECTED INBORN ERRORS OF METABOLISM

The initial laboratory assessment of infants and children with suspected inborn errors of metabolism (IEM) is reviewed by the Department of Medical Genetics, Mayo Clinic, Rochester, MN. Classes of IEM include organic acidemias, aminoacidopathies, urea cycle defects, glycogen storage diseases, lysosomal storage diseases, B-oxidation defects, and peroxisomal disorders. Signs and symptoms of IEM include failure to thrive, loss of milestones, vomiting, seizures, coma, hepatosplenomegaly, dysmorphic features, sparse or abnormal textured hair, cataract and other eye findings, and urine or body odor. Initial tests suggested include blood gases, glucose, urinary ketones, ammonia, electrolytes, uric acid, liver function, lactate and pyruvate, carnitine, free fatty acids, B-hydoxybutyrate, and acetoacetate. (Lindor NM, Karnes PS. Initial assessment of infants and children with suspected inborn errors of metabolism. Mayo Clin Proc October 1995;70:987-988). (Reprints: Dr NM Lindor,

Department of Medical Genetics, Mayo Clinic, 200 First Street SW, Rochester, MN 55905).

COMMENT. Examples of IEM requiring additional preliminary tests include Menkes' kinky-hair disease (serum copper and ceruloplasmin), and molybdenum cofactor deficiency (urine sulfite dipstick).

MOVEMENT DISORDERS

CLASSIFICATION OF PAROXYSMAL DYSKINESIAS

Forty six patients, ages ranging 1 to 77 years, with paroxysmal dyskinesias and classified according to precipitating factors and duration of attacks were reported from the Movement Disorder Clinic, Department of Neurology, Baylor College of Medicine, Houston, TX. Paroxysmal kinesigenic dyskinesia (PKD), occurring abruptly after a sudden movement, affected 13 patients; paroxysmal nonkinesigenic dyskinesia (PNKD) occurred spontaneously in 26; exertion-induced attacks (PED) affected 5; and episodes were precipitated only by sleep (PHD) in 1. The etiology was idiopathic in 22 and secondary to psychogenic illness in 9, to stroke in 4, trauma (3), encephalitis (2), multiple sclerosis (2), kernicterus (1), and migraine (1). Short duration (<5 min) and long-lasting attacks (>5 min) were about equal in incidence. None had loss of consciousness or other evidence of seizures and EEGs were normal in 34 tested. MRI was normal in 25 tested. Nine of 10 (90%) patients with PKD improved with medications, mainly carbamazepine, phenytoin, or clonazepam, compared to only 7 of 19 (37%) with PNKD. (Demirkiran M, Jankovic J. Paroxysmal dyskinesias: clinical features and classification. Ann Neurol October 1995;38:571-579). (Respond: Dr Jankovic, Department of Neurology, Baylor College of Medicine, 6550 Fannin, Suite 1801, Houston, TX 77030).

COMMENT. In contrast to the original (Mount and Reback, 1940) and other previous reports which empasized genetic and familial factors in etiology, the majority of the patients in the above series had sporadic and secondary paroxysmal dyskinesias. Menkes JH, in his Textbook of Child Neurology (Lea & Febiger, 1985), provides an excellent account of the various types of paroxysmal dyskinesia, classified according to 1) movement pattern - choreiform, athetoid, dystonic, or tonic; 2) familial or acquired; 3) kinesigenic or nonkinesigenic; 4) acquired etiology - perinatal asphyxia, reflex epilepsy, metabolic disorders (eg. idiopathic hyperparathyroidism), and multiple sclerosis. The specificity of response of the kinesigenic dyskinesias to anticonvulsant drugs, especially phenytoin, has been documented previously. Rare cases of hypnogenic dyskinesia, responding to lorazepam, and one patient with paroxysmal diplopia due to superior oblique myokymia following head injury, responding to carbamazepine, are described in the present series. An overlap between paroxysmal dyskinesia and epilepsy, migraine, and paroxysmal ataxia is discussed. The possible relation between migraine and paroxysmal dyskinesia mentioned in this report has not previously been noted.

HEREDITARY MYOKYMIA AND PAROXYSMAL ATAXIA

A family with autosomal dominant hereditary myokymia and paroxysmal ataxia, linked to chromosome 12p, is described from University