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HEREDO-DEGENERATIVE DISEASES

TUBEROUS SCLEROSIS

A report of a joint meeting of the Forum on Mental Retardation of the Royal Society of Medicine and The Tuberous Sclerosis Association of Great Britain states that the prevalence of tuberous sclerosis (TS) for children under 15 years may be 1 in 10,300. Evidence for the assignment of the TS gene to chromosome 9 was reviewed, and high levels of carbohydrate specific to a glycoprotein-fibronectin in the affected skin of TS patients noted. Infantile spasms associated with TS may have a better prognosis than in non-TS patients. Cognitive deterioration in TS is usually related to underlying pathology rather than seizure frequency or severity. Of 88 children with TS followed until the age of 5 years, 81% of those who were walking showed severe psychiatric disorder, 59% were autistic, and 70% had hyperkinetic behavior disorders. Infantile spasms were associated with autism in 68%, 79% were severely mentally handicapped, and 47% were in special care units of schools. The detection of rhabdomyomata by echocardiography was a valuable diagnostic technique, positive in 47% of 60 children and adults with TS. A greater awareness and early diagnosis of TS is called for. (Corbett J, Hunt A. Recent research on tuberous sclerosis (TS), JRSM Aug 1988;81:481-482).

> COMMENT. Forehead plaques, smooth patches of slightly raised skin with a reddish or yellowish discoloration, can be one of the earliest skin manifestations of TS. Wood's light examination of the skin for hypopigmented maculae is important in all infants with myoclonic spasms and hypsarrhythmia. (see Ped Neur Briefs 1987;1:3). MRI may help in predicting the

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eventual clinical severity of younger children with newly diagnosed TS, whereas the number of CT brain abnormalities does not correlate with prognosis. High-signal MRI lesions involving the cerebral cortex are characteristic of TS and correspond to hamartomas and gliotic areas seen pathologically. Periventricular calcific lesions are better visualized with CT than with MRI. (Roach et al Arch Neurol 1987;44:301).

MULTIPLE SULFATASE DEFICIENCY

A 9-year-old girl with a phenotype similar to a mucopolysaccharidosis (MPS) and a clinical history characteristic of late infantile metachromatic leukodystrophy (MLD) is reported from the Department of Neurology, National Defense Medical Center, Taipei, Taiwan, Republic of China; the Developmental and Metabolic Neurology Branch, NIH, Bethesda, Maryland; and Department of Pediatrics (Dr. Horwitz), University of Chicago, Chicago, Illinois. The girl's early history and development were normal up to 18 months of age. Following a high fever with a flu-like illness, her gait became unsteady and broad- based. Gradually her speech became slurred and her vocabulary deteriorated. Examination at 7 1/2 years showed short stature and microcephaly. She was autistic and inattentive, with marked cognitive impairment. She had hyperreflexia, extensor plantar responses, dysmetria, and incoordination. Dysmorphic features suggested MPS but dysostosis multiplex and organomegaly were absent. Funduscopic examination revealed a cherry-red-like spot and yellowish-granular appearance of the retina. Deficient activities of arylsulfatase-A, arylsulfatase-B, iduronate sulfatase, and heparan N-sulfatase in the leukocytes established the diagnosis as MSD. The total urinary content of the glycosaminoglycans was normal, but the concentration of heparan sulfate was increased, stressing the need for qualitative estimations when MSD is suspected. (Soong B-W, Casamassima AC, Fink JK, Constantopoulos G, Horwitz AL. Neurology August 1988;38:1273-75).

COMMENT. Multiple sulfatase deficiency or mucosulfatidosis (MSD) is an autosomal recessive genetic disease affecting the expression of lysosomal sulfatases with consequent accumulation of sulfate-containing glycolipids, glycosaminoglycans, and steroid sulfates in tissues and body tissues. The clinical manifestations represent a combination of 2 diseases: late infantile MLD and MPS. The disorder is rare and the authors cite 20 previous reports of this phenotype.

FRIEDREICH'S ATAXIA AND GLUCOSE METABOLISM

Glucose metabolism was investigated in 21 patients with FA at the Instituto Neurogico, Cattedra di Clinica Medica, Milan, Italy. Abnormalities of glucose tolerance occurred in 5 (23.8%) and 4 were diabetic (19%). By oral glucose tolerance tests, the plasma glucose levels of 5 patients were 140-200 mg/ml 2 hours after glucose ingestion. Plasma insulin levels of glucose-intolerant patients were significantly higher than controls after 180 minutes following glucose ingestion. Plasma glucagon levels of FA patients were higher