COMMENT. HARP syndrome is distinguished from Hallervorden-Spatz disease (HSD) by acanthocytosis and the abnormality of lipoprotein. The authors note that all cases of HARP syndrome have been sporadic and lack the autosomal recessive feature of HSD. For further reports of HARP syndrome, see Progress in Pediatric Neurology II, PNB Publ, 1994, p477.

## FRIEDREICH'S ATAXIA WITH RETAINED REFLEXES

Genetic linkage analyses in 11 patients from 6 families with Friedreich's ataxia (FA) phenotype, including cardiomyopathy, but retained reflexes (FARR), are reported from the University of Naples and C Besta Neurological Institute, Milan, Italy; and La Fe University Hospital, Spain. Mean age of onset was 13.5 years. Inheritance was autosomal recessive. All patients had progressive ataxia, dysarthria, dysmetria, scoliosis and pes cavus. FARR mapped to the FA locus on chromosome 9q13-21.1, suggesting that FARR is a variant phenotype of FA. (Palau F et al. Early-onset ataxia with cardiomyopathy and retained tendon reflexes maps to the Friedreich's ataxia locus on chromosome 9q. Ann Neurol March 1995;37:359-362). (Respond: Prof Filla, Clinica Neurologica, Universita Frederico II, via Pansini 5, 80131 Napoli, Italy).

COMMENT. The diagnosis of FARR, a variant of Friedreich's ataxia, should be considered in patients with early onset cerebellar ataxia, cardiomyopathy, and sensory neuropathy. Barbeau found absence of deep tendon reflexes to be a required criterion in the diagnosis of FA (Can J Neurol Sci 1978a;5:57-59), whereas Bell and Carmichael allowed hyperactive reflexes in some cases (Treas Hum Inherit 1939;4:141-281). (Bala V Manyam, personal communication).

## INFANTILE LEUKOENCEPHALOPATHY WITH MILD COURSE

Eight children, including 2 siblings, with infantile onset cerebral leukoencephalopathy and megalencephaly, and mild neurological signs and symptoms, are reported from Free University Hospital, and Academic Medical Center, Amsterdam, The Netherlands. Ataxia and spasticity were slowly progressive, while intellectual functioning was preserved for a few years. MRI showed swelling of supratentorial hemispheral white matter, subcortical cysts, and sparing of corpus callosum and internal capsule. Metabolic studies were negative. (van der Knaap MS, Barth PG et al. Leukoencephalopathy with swelling and a discrepantly mild clinical course in eight children. Ann Neurol March 1995;37:324-334). (Respond: Dr MS van der Knaap, Department of Child Neurology, Free University Hospital, PO Box 7057. 1007 MB Amsterdam, The Netherlands).

COMMENT. This type of infantile leukoencephalopathy is distinguished from Canavan and Alexander diseases by an MRI showing severe white matter abnormalities which contrasted with a slow clinical progressive course. Lysosomal and other metabolic white matter disorders characterized by megalencephaly were also ruled out biochemically and clinically.

**LATE ONSET KRABBE'S DISEASE WITH PRESERVED INTELLECT** in a 24-year-old Swedish male patient is reported from the County Hospital of Jonkoping, and the University of Goteborg, Sweden. (Arvidsson J,

Hagberg B et al. Late onset globoid cell leukoencephalopathy (Krabbe's disease) - Swedish case with 15 years of follow-up. Acta Paediatr Feb 1995;84:218-21). The disease presented with visual dysfunction at 4 years of age. At 8 years he developed a limp and ataxia and within 6 months he was wheel-chair dependent. Epilepsy began at 14 years. Speech became dysarthric on entering school, but he was able to stay in the mainstream educational system. Leukocyte galactosylceramidase activity was reduced.

## CONGENITAL LACTIC ACIDOSIS: PET AND MRS STUDIES

Positron emission tomography (PET) and proton magnetic resonance spectroscopy (MRS) identified an increase in rate of cerebral glycolysis (PET) and cerebral lactate (MRS) in 2 children with defective mitochondrial respiration and congenital lactic acidosis studied at the Universitat zu Koln, Germany, and the University Hospital, Nijmegen, The Netherlands. These changes were not apparent in a child with lactic acidosis and normal respiratory chain activity. Defects of oxidative phosphorylation may cause increases in glycolysis and accumulation of cerebral lactate. (Duncan DB et al. Positron emission tomography and magnetic resonance spectroscopy of cerebral glycolysis in children with congenital lactic acidosis. Ann Neurol March 1995;37:351-358). (Respond: Prof Dr Heiss, Max-Planck-Institut fur neurologische Forschung, Gleueler Str 50, 50931 Koln, Germany).

COMMENT. PET and MRS have been used to demonstrate the metabolic changes associated with defective mitochondrial respiration in the brain without resort to diagnostic muscle biopsy. For further discussion of MRS in mitochondrial disorders, see Progress in Pediatric Neurology II, 1994, p454.

# VASCULAR DISORDERS

## NEONATAL MIDDLE CEREBRAL ARTERY STROKE

The presentation, EEG, imaging studies, and outcome of six term neonates with middle cerebral artery infarcts are reported from Mannheim Hospital, University of Heidelberg, Germany. Birth was by cesarean section in 5 cases. Apgars were normal in 4. Seizures occurred in 4 within 1 to 3 days and in 2 at the 5th and 9th days. EEGs showed focal slowing followed by focal spike-wave activity, correlating with the structural lesion defined by neuroimaging. EEG abnormalities sometimes antedated a positive ultrasound. A late intra-uterine event was suggested. Only 2 had obvious hemiparesis at discharge, but all children showed developmental delay and spastic hemiparesis at 3 to 10 year follow up. One had infantile spasms and hemihypsarrhythmia, relieved only after drainage of a cyst and shunt procedure. Four developed epilepsy late. (Koelfen W, Freund M, Varnholt V. Neonatal stroke involving the middle cerebral artery in term infants: Clinical presentation, EEG and imaging studies, and outcome. Dev Med Child Neur March 1995;37:204-212). (Respond: Dr Wolfgang Koelfen, Kinderklinik Mannheim, Theodor Kutzer Ufer, 68127 Mannheim, Germany).

COMMENT. This study confirms previous reports of late onset of epilepsy and development of spastic hemiparesis and cognitive deficits following an apparent early favorable outcome in term neonates suffering middle