ENCEPHALOPATHIES

PORPHYRIA PRESENTING AS DIFFUSE ENCEPHALOPATHY

An 18-year-old female presenting with seizures, myalgias, abdominal pain, headache and vomiting had multiple large contrast-enhancing white matter lesions on MRI and was diagnosed with acute intermittent porphyria (AIP), in a report from the Department of Neurology, Mayo Clinic, Rochester, MN. She was febrile, confused, complained of visual hallucinations, and had increased blood pressure and tachycardia. Liver enzymes were elevated and electrolytes lowered. AIP was considered because of seizures, dysautonomia, and abdominal pain. Uroporphyrin levels were 2,186 mcg/24 hrs (normal, 3-25), and porphobilinogen, 312.8 mg/24 hrs (normal, 0-0.5). Urine was dark, resembling ale or tea. Fecal porphyrin profile excluded hereditary coproporphyria and variegate porphyria. One week after treatment with IV hematin and dextrose, MRI lesions resolved. (Maramattom BV, Zaldivar RA, Glynn SM et al. Acute intermittent porphyria presenting as a diffuse encephalopathy. **Ann Neurol** April 2005;57:581-584). (Respond: Dr Wijdicks, Mayo Clinic College of Medicine, Department of Neurology, W8B, 200 First Street SW, Rochester, MN 55905).

COMMENT. AIP is an autosomal dominant disorder caused by a genetic deficiency of PBG deaminase enzyme involved in heme synthesis. Most patients are asymptomatic, but under stress, during menstruation, surgery, fasting or exposure to drugs, an acute attack may occur, with heme precursors, aminolevulinic acid and PBG, excreted in the urine. Treatment with barbiturates will aggravate or precipitate an attack. Episodes present with abdominal pain, vomiting, seizures, neuropathy, dysautonomia, and psychiatric symptoms. Encephalopathy, a presenting symptom in the above case, is unusual, but combined with abdominal pain, should arouse suspicion of AIP. A more common variety of porphyria in childhood is congenital erythropoietic porphyria, presenting with cutaneous photosensitivity and hemolytic anemia, but no neurologic symptoms.

CYSTIC LEUKOENCEPHALOPATHY WITH NORMOCEPHALY

A new syndrome of nonprogressive encephalopathy with normo- or microcephaly and early onset of psychomotor impairment is described in 15 children, in a report from the University of Gottingen and other centers in Europe and the US. Clinical findings include patients of Turkish origin; consanguinity of parents in 5; 2 patients siblings and 2 first cousins; age at onset birth to 18 months; microcephaly in 9 and normocephaly in 6; impaired motor and mental development; epilepsy in 8; spasticity in 12; hearing impairment in 5; normal peripheral nerves; course stable or slowly progressive. Tests for metabolic and infectious disease were negative. MRI findings included supratentorial white matter lesions with a high signal on T2 weighted images involving mainly periventricular regions, sparing of central white matter and cerebellar white matter, cystic lesions in anterotemporal lobes not connected to the ventricles, pericystic abnormal myelination on FLAIR-weighted images, enlarged temporal horn, and no involvement of gray matter. The cause is unknown, but an autosomal recessive inheritance is suspected and is under investigation. (Henneke M, Preuss

N, Engelbrecht V et al. Cystic leukoencephalopathy without megalencephaly: a distinct disease entity in 15 children. **Neurology** April (2 of 2) 2005;64:1411-1416). (Reprints: Dr Jutta Gartner, Department of Pediatrics and Pediatric Neurology, Georg August University, Faculty of Medicine, Robert-Koch-Strasse 40, 37075 Gottingen, Germany).

COMMENT. The authors distinguish this autosomal recessive disease from a previously described cystic leukoencephalopathy with megalencephaly (MLC), caused by a mutation of MLC1 gene. (Van der Knapp et al, 1995; **Progress in Pediatric Neurology III,** 1997;p557). In MLC, the MRI showed supratentorial white matter swelling and subcortical cysts, which contrasted with a mild clinical course.

CEREBRAL PALSY AND NEONATAL ENCEPHALOPATHY

The type and severity of cerebral palsy (CP) and pattern of associated disability in children with or without preceding neonatal encephalopathy (NE) were compared in a population-based case-control study of patients followed for 6 years at the Children's Hospital, Westmead, Sydney, Australia. Of 276 infants with NE, 25 (9.1%) died in the neonatal period, and of the 251 neonatal NE survivors, 32 (13%) developed CP by 5 years of age. Of term infants with CP, 24% followed NE. CP following NE was more likely in males, more severe, spastic quadriplegic in type, and more commonly complicated by cognitive and speech impairment, epilepsy, severe disability, and death by 6 years. (Badawi N, Felix JF, Kurinczuk JJ et al. Cerebral palsy following newborn encephalopathy: a population-based study. **Dev Med Child Neurol** May 2005;47:293-298). (Respond: Nadia Badawi PhD FRACP, Department of Neonatology, Children's Hospital at Westmead, Locked Bag 4001, Westmead, NSW 2145, Australia).

COMMENT. Term infants with CP and NE have a poorer prognosis compared to those without NE. One out of every five will die in the first 5 years of life. In a commentary by Dr Karin B Nelson, National Institutes of Health, USA (**Dev Med Child Neurol** 2005;47:292), the importance of causal factors in NE was stressed, a subject addressed by the authors in a previous study (Badawi N et al. **BMJ** 1998;317:1549-1553 and 1554-1558).

SPASTIC DIPLEGIC AND TETRAPLEGIC CEREBRAL PALSY COMPARED

Risk factors of cerebral palsy (CP), seizures, CP severity, EEG, and MRI findings were compared in 38 children with spastic diplegic (DCP) and 48 with spastic tetraplegic (TCP), in a report from Medical University of Bialystok, Poland. The Apgar score was lower in TCP cases than DCP, the gross motor function was more limited, mental retardation more frequent, cerebral atrophy on MRI more frequent (31% cf 5%), epilepsy more common (50% cf 16%) and more often intractable. Periventricular leukomalacia on MRI was more frequent in DCP (76%) than in TCP (44%). Gestational history was not related to increased risk of DCP or TCP; the frequencies of cesarean section, low birth weight, and perinatal pathology were the same in both groups. (Kulak W, Sobaniec W, Smigielska-Kuzia J et al. **Pediatr Neurol** May 2005;32:311-317). (Respond: Dr Kulak, Department of Pediatric Neurology an