

mutations. Onset is with visual failure due to retinal degeneration, noted between the ages of 5 and 8 years. This is followed by epilepsy, psychomotor deterioration, sleep disturbance, and extrapyramidal symptoms. Affected females have acne, hirsutism, and obesity. Death follows in the early twenties.

EARLY DIAGNOSIS OF EPILEPSY SYNDROMES

The classification of epilepsy syndromes made initially on the basis of information at time of diagnosis was compared to that made 2 years later in a cohort of 613 children, followed by participating physicians in Connecticut, between 1993 and 1997. After 2 years, syndrome classifications were the same in 86% of the cohort. The diagnosis was changed in 10% (mainly incomplete syndromes), and syndrome evolution, mainly West to Lennox-Gastaut, occurred in 4%. Significant changes were rare. (Berg AT, Shinnar S, Levy SR et al. How well can epilepsy syndromes be identified at diagnosis? A reassessment 2 years after initial diagnosis. *Epilepsia* October 2000;41:1269-1275). (Reprints: Dr Anne T Berg, Department of Biological Sciences, Northern Illinois University, DeKalb, IL 60115).

COMMENT. The identification of epileptic syndromes, for the most part, may be made accurately at the time of the initial presentation and diagnosis. Changes in diagnosis at follow-up, necessary in only 14%, are explained by difficulties in classification of incomplete syndromes and the evolution of West to Lennox-Gastaut syndromes with age and maturation.

Epileptic syndromes posing problems in diagnosis. Hirsch E et al (Strasbourg, France) review the heterogeneous nature and clinical management of partial epilepsies and incomplete syndromes. BECTS are the most common idiopathic localization-related epilepsy, and may be triggered by carbamazepine in some cases. Primary reading epilepsy and idiopathic occipital lobe epilepsies with photosensitivity are an overlap of idiopathic localization-related and generalized epilepsies, and respond to sodium valproate. Other variants of idiopathic localization-related epilepsies include autosomal dominant nocturnal frontal lobe epilepsy and benign familial infantile convulsions. AED resistance can be due to errors in diagnostic classification of these epilepsy syndromes. EEG-video evaluation may be necessary in refractory seizures. (Hirsch E et al. New insights into the clinical management of partial epilepsies. *Epilepsia* Oct 2000;41(suppl 5):S13-S17).

Post-ictal paralysis in BECTS. Dai A et al (State University of New York, Buffalo, NY) found a 9% association of post-ictal paresis among 68 children with benign rolandic epilepsy, and 50% had brief post-ictal aphasia. Todd's paresis and aphasia do not exclude the diagnosis of BREC, and these transient complications are clinically benign. (Abstracts from the Annual Meeting of the AES, Los Angeles, CA, Dec 1-6, 2000. *Epilepsia* Oct 2000;4, suppl 7:88).

ANOXIC AND VASCULAR DISORDERS

HYPOTHERMIA TREATMENT FOR NEONATAL ENCEPHALOPATHY

Treatment with mild whole body hypothermia after birth asphyxia was evaluated in 10 of 16 newborns with EEG burst suppression evidence of a bad prognosis, followed at the Imperial College School of Medicine, London, UK. All infants selected for treatment had convulsions and a severe encephalopathy. Hypothermia was instituted within 6 hours of birth and continued for 48 hours.

Changes included prolonged metabolic acidosis, decreased blood potassium, lower heart rate, and higher mean blood pressure, but were not associated with clinical complication. Unusual MRI findings in 3 infants were sinus thromboses and cerebral infarction. Six infants had normal follow-up neurologic exams or only minor abnormalities. Three infants died and 1 had major abnormalities. EEG activity returned to a more continuous pattern by 13 hours in 5 infants, although seizures recurred. EEG suppression was greater in infants with a poor outcome than in the 6 who recovered. Of 6 untreated infants with a normal initial EEG, none developed severe encephalopathy or neurologic sequelae. (Azzopardi D, Robertson NJ, Cowan FM et al. Pilot study of treatment with whole body hypothermia in neonatal encephalopathy. Pediatrics October 2000;106:684-694). (Reprints: Denis Azzopardi MD, FRCP, Department of Pediatrics, Imperial College School of Medicine, London, UK).

COMMENT. Prolonged mild hypothermia has been evaluated in the treatment of asphyxiated neonates at high risk of developing severe neonatal encephalopathy. A burst suppression pattern in the EEG is indicative of increased risk of severe encephalopathy and possible trial of hypothermia.

OUTCOME OF NEONATAL CEREBRAL INFARCTION

The long-term neurodevelopmental outcome of CT-documented cerebral infarction was evaluated in 46 children followed for a mean of 42 months (range, 18-164 months) at the Glenrose Rehabilitation Hospital, and University of Alberta, Edmonton, Canada. Outcome was normal in 15 and abnormal in 31, with multiple disabilities in 23, cerebral palsy in 22, and cognitive impairment in 19. Risk factors for long-term disability were neonatal seizures and abnormal neurologic exam at discharge. (Sreenan C, Bhargava R, Robertson CMT. Cerebral infarction in the term newborn: clinical presentation and long-term outcome. J Pediatr September 2000;137:351-355). (Reprints: Charlene Robertson MD, FRCPC, Neonatal and Infant Follow-up Clinic, Glenrose Rehabilitation Hospital, 10230-111 Ave, Edmonton, Alberta, Canada T5G 0B7).

COMMENT. The long-term outcome in term neonates with cerebral infarction is normal in one third and abnormal in two thirds. Risk factors for disability are seizures and an abnormal neurologic examination at the time of discharge.

MENTAL RETARDATION SYNDROMES

FRAGILE X SYNDROME WORKSHOP

The proceedings of the 9th International Workshop on Fragile X Syndrome and X-Linked Mental Retardation, held in Strasbourg, France, are reported from the University Hospital of Leuven, Belgium.

Several examples of X-Linked MR syndromes were presented. *Mohr-Tranebjaerg syndrome* (MTS), characterized by deafness, dystonia and mental retardation (MR), was discussed as a mitochondrial disorder with a gene named DDP for deafness/dystonia peptide. Among a set of 8 patients with MTS, only one of 4 studied showed ragged-red fibers on muscle biopsy whereas 4/4 had increased numbers of mitochondria.

In two papers on *fragile X syndrome* (FXS), a total of 27 children (24 boys and 3 girls), the diagnosis had been missed by the referring physicians. Clinical manifestations were nonspecific, a positive family history was not considered, or