postural abnormalities in infancy, peripheral weakness without ataxia, and atypical cases may last 5 to 12 hours. The EA1 locus is mapped to chromosome 12q, and 19 missense mutations in KCNA1 have been reported.

Type 2 (EA2), the most common EA, is characterized by longer episodes of ataxia (hours) with spontaneous nystagmus (usually vertical and downbeat) and mildly progressive baseline ataxia. A gaze-evoked nystagmus is elicited between attacks. Attacks begin in early childhood, and are associated with vertigo, nausea, vomiting, and migraine headaches. They are responsive to acetazolamide. EA2 is allelic with familial hemiplegic migraine type 1 (FHM1). The EA2 locus is mapped to chromosome 19p, similar to that of FHM1. EA 3 – 6 are described in only one or two families, but with distinctive genetic features and mapped to different chromosomes. Two North Carolina kindreds with EA4 had late-onset vestibulocerebellar ataxia, vertigo and interictal nystagmus. Linkage analysis ruled out EA1 and EA2 loci.

Differential diagnosis of EA syndromes includes epilepsy, paroxysmal dyskinesias and migraine. Myokymia, an irregular undulation of the surface of muscles, distinguishes cases of EA1, and baseline nystagmus, ataxia and headaches are typical of EA2. Genetic testing is available for EA1 and EA2. In treatment, carbamazepine, valproic acid and acetazolamide are effective for EA1, and acetazolamide, flunarizine and 4-aminopyridine in EA2. (Jen JC, Graves TD, Hess EJ et al. Primary episodic ataxias: diagnosis, pathogenesis and treatment. Brain October 2007;130:2484-2493). (Respond: Joanna C Jen, UCLA Neurology, 710 Westwood Plaza, Los Angeles, CA 90095).

C O M M E N T. Episodic ataxias are characterized by attacks of incoordination and imbalance, with onset in early childhood, and associated with myokymia, nystagmus and sometimes migraine headache or seizures. Attacks may be controlled by acetazolamide and/or carbamazepine. EAs are inherited as autosomal dominant channelopathies with mutations commonly in two genes.

METABOLIC DISORDERS

PHENOTYPE OF MITOCHONDRIAL DNA 3243A>G MUTATION

The prevalence and common clinical manifestations of the mitochondrial DNA 3243A>G mutation in children in a defined population in Finland were studied at the Universities of Oulu and Turku and other centers. Three probands were detected with encephalopathy, diabetes mellitus, or sensorineural hearing impairment, and 27 children as potential mutation carriers, with a prevalence of 18.4 in 100,000. Clinical features in 24 children in 5 families with 3243A>G mutation included migraine and learning disabilities, short stature, sensorineural hearing loss, exercise intolerance, delayed motor and speech development, and progressive encephalopathy. The prevalence was relatively high in the pediatric population, but morbidity in children is low. (Uusimaa J, Moilanen JS, Vainionpaa L, et al. Prevalence, segregation, and phenotype of the mitochondrial DNA 3243A>G mutation in children. Ann Neurol Sept 2007;62:278-287). (Respond: Dr Majamaa, Department of Neurology, University of Turku, FIN-20014 Turku, Finland. E-mail: kari.majamaa@utu.fi).
COMMENT. The 3243A>G mutation is the most common cause of the MELAS syndrome. Infants may present with the classic mitochondrial encephalomyopathy, lactic acidosis, and strokelike episodes while others show failure to thrive, hypotonia, seizures, cardiomyopathy, and lactic acidosis, dependent on the amount of mutant gene in different tissues. The clinical phenotype varies widely in symptoms and their severity. Many children with the 3243A>G mutation are asymptomatic, and those with symptoms usually present with sensorineural hearing loss, short stature, migraine, exercise intolerance, and learning difficulties. Encephalomyopathy is uncommon and morbidity relatively low.

VASCULAR DISORDERS

RISK OF EPILEPSY AFTER PERINATAL STROKE

The prevalence and severity of epilepsy after 6 months of age in 64 children with a history of perinatal stroke were studied by a retrospective review of patients at Riley Hospital for Children, Indianapolis, IN. Forty eight (75%) presented in the NICU with seizures and were treated with phenobarbital. Comorbidities included infection in 11 (17%), cardiac abnormalities in 11 (17%), ECMO in 4 (6%), and renal failure in 3 (5%). Seven (11%) had a family history of seizures. Prenatal ultrasound was positive for stroke in 4 (6%) patients. Infarction was confirmed by CT or MRI, and an abnormal initial EEG was recorded in 40 (91%). Follow-up data were available on 61 (95%) patients, and 41 (67%) had developed epilepsy between 6 months and last follow-up (mean 43 mos; range 9-178 mos). Five (8%) had infantile spasms. Seizures resolved in 13 (32% of 41 with epilepsy), and medications had been discontinued. Infarct on prenatal ultrasound (p=.0065) and family history of seizures (p=.0093) were significantly associated with an earlier development of seizures after 6 months of age: median time 3.8 months with, vs 53.9 without, positive ultrasound; and 1.1 months with, vs 53.9 without, positive family history. No variables were correlated with time to resolution of seizures, or with epilepsy occurrence after 6 months of age. (Golomb MR, Garg BP, Carvalho KS, Johnson CS, Williams LS. Perinatal stroke and the risk of developing childhood epilepsy. J Pediatr October 2007;151:409-413). (Reprints: Dr Meredith Golomb, Indiana University School of Medicine, Blg XE, Room 040 (Pediatric Neurology), 575 West Dr, Indianapolis, IN 46202).

COMMENT. Perinatal stroke is frequently followed by the development of epilepsy in childhood. Evidence of infarction on prenatal ultrasonography and a family history of epilepsy are predictive of an earlier onset of seizures, but no risk factors used in this study predicted time of seizure resolution or whether the patients would develop epilepsy.

Frequency of electrographic seizures and PEDS after intracerebral hemorrhage was studied in 102 consecutive adult patients with ICH who underwent continuous electroencephalographic monitoring (cEEG) at Columbia University, NY. (Claassen J et al. Neurology Sept 2007;69:1356-1365). Seizures occurred in one third of patients with ICH and more than one half were purely electrographic seizures. Electrographic seizures were associated with expanding hemorrhages, and PEDs were independently associated with cortical ICH and poor outcome. Continuous EEG monitoring is essential for detection of subtle seizures.

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