MOVEMENT DISORDERS

IDIOPATHIC PAROXYSMAL TONIC UPWARD GAZE

A case of paroxysmal tonic upward gaze with neck flexion in a 14-month-old boy is reported from a Department of Neurology in Izmir, Turkey. The paroxysms began at age 11 months, they recurred several times each day, and episodes lasted 1 or 2 hours. Downbeat nystagmus was elicited on attempted downward gaze. Horizontal eye movements were normal during an attack. Episodes were associated with a wide-based ataxic gait. Video EEG during an episode was normal, and EEG recorded between episodes was also normal. Tests including brain MRI, CSF, amino acids, organic acids, serum lactate, and thyroid were unremarkable. Levodopa treatment was without benefit. Six months later, the episodes ceased spontaneously and showed no recurrence after 1-year-follow-up. The ocular and neurologic examinations were normal. (Ozbay OE. Idiopathic paroxysmal tonic upward gaze. **Pediatr Neurol** 2012 Oct;47(4):306-8) (Respond: Dr Ozbay, Department of Neurology, School of Medicine, Ege University, Bornova, Izmir, Turkey, 35040. E-mail: ozgul.ekemekci@ege.edu.tr).

COMMENT. First described in 1988 (Ouvrier EA, Billson F. Benign paroxysmal tonic upgaze of childhood. **J Child Neurol** 1988 Jul;3(3):177-80), the authors reviewed reported cases in the literature in 2005 and suggested the deletion of "benign" since some cases are more persistent and are exacerbated by fatigue, immunization, and febrile illness. The disorder is idiopathic or symptomatic of underlying disease. The pathophysiology is unclear but the tectum in upper dorsal brainstem, the anatomic site of vertical eye movements, is probably involved. Lesions affecting the brainstem, epilepsy, oculogyric crises, and retinal disease are included in the differential diagnosis. Association with genetic disorders such as Beckwith-Wiedemann syndrome (macrosomia, macroglossia) has been reported. The present case report is an example of an idiopathic, benign form of paroxysmal tonic upward gaze.

PAROXYSMAL KINESIGENIC DYSKINESIA AND INFANTILE CONVULSIONS

A team of twelve geneticists and neurologists from centers in the Netherlands studied the phenotypes and penetrance of paroxysmal kinesigenic dyskinesia (PKD), infantile convulsion and choreoathetosis (ICCA) syndrome, and benign familial infantile convulsions (BFIC), caused by PRRT2 mutations. Three different PRRT2 heterozygous mutations were detected in 2 families with ICCA, 2 families with PKD, and one individual with sporadic PKD. PRRT2 mutations were not detected with febrile convulsions or with migraine. The estimated penetrance of PRRT2 mutations in cases involving only PKD was 61%; it was nearly complete if infantile convulsions (BFIC) were also included. The identification of PRRT2 as a major gene for the PKD-ICCA-BFIC spectrum allows better disease classification, molecular confirmation of the clinical diagnosis, and genetic testing and counseling. (van Vliet R, van Andel J, Brilstra E, et al. PRRT2 phenotypes and penetrance of paroxysmal kinesigenic dyskinesia and infantile

convulsions. **Neurology** 2012 Aug 21;79(8):777-84). (Response and reprints: Dr Matt-Kievit. E-mail: j.a.maat@erasmusmc.nl).

COMMENT. PRRT2 mutations are the major cause of PKD or ICCA, but they are not involved in the etiology of febrile convulsions and migraine. Paroxysmal kinesigenic dyskinesia/choreoathetosis is characterized by brief attacks of involuntary movements (dystonia, chorea, athetosis, and ballism), precipitated by a kinesigenic trigger such as sudden movement or startle. Attacks last <1 minute, without loss of consciousness; they begin during childhood, and are controlled by anticonvulsant medication. Neurologic examination and MRI are normal, and ictal EEG shows nonspecific abnormalities. PKD is sporadic or familial. Familial cases have an autosomal dominant transmission with incomplete penetrance. PKD may be associated with BFIC or ICCA and with migraine.

UTILITY OF THE ELECTROENCEPHALOGRAM

EEG IN PREDICTION OF EARLY NEURODEVELOPMENT OF PRETERM INFANTS

Researchers at Anjo Kosei Hospital and other centers in Japan and at Washington University, St Louis, MO, USA studied the prognostic value of conventional EEG for the identification of preterm infants admitted to the Anjo neonatal ICU and at risk for adverse neurodevelopment. Serial EEG recordings were conducted during 3 time periods, at least once each within days 6 (first period), during days 7 to 19 (second period), and days 20 to 36 (third period). Neurodevelopment outcomes were assessed at a corrected age of 12 to 18 months. Of 333 preterm infants (<34 weeks' gestation), 33 (10%) had developmental delay and 34 (10%) had cerebral palsy. In the infant's 780 EEG records studied between 2002 and 2008, abnormalities were significantly predictive of developmental delay and cerebral palsy at all 3 time periods. Acute stage background EEG abnormalities were graded as mild, moderate, and severe, and were characterized as suppressed with decreased continuity, lower amplitude, and/or attenuated fast-wave. Chronic stage background was characterized by a disorganized pattern with sharp waves or a dysmature pattern. The grade of EEG abnormalities correlated with the incidence of developmental delay or cerebral palsy in all periods ($p<0.001$). EEG abnormality in the second period was an independent predictor of developmental delay and cerebral palsy. (Hayashi-Kurahashi N, Kidokoro H, Kubota T, et al. EEG for predicting early neurodevelopment in preterm infants: an observational cohort study. **Pediatrics** 2012 Oct;130(4):e891-7). (Respond: Hiroyuki Kidokoro MD, Department of Pediatrics, Washington University in St Louis, 660 South Euclid Ave, St Louis, MO 63110. E-mail: kidokoro_h@kids.wustl.edu).

COMMENT. EEG abnormalities within the first month of life of preterm infants significantly predict adverse neurodevelopment at age 12 to 18 months. The EEG has prognostic value independent of neuroimaging and clinical risk factors. A disorganized EEG pattern precedes abnormalities on ultrasound and is a better prognostic marker in the neonatal care setting (Kidokoro H et al. **Neuropediatrics** 2010 Dec;41(6):241-5).