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