MOVEMENT DISORDERS

PAROXYSMAL NONKINESIGENIC DYSKINESIA

A Canadian family of European descent with paroxysmal nonkinesigenic dyskinesia (PNKD), and a gene locus that links to a distinct region on chromosome 2q31, is reported by researchers at University of British Columbia, Canada. The age at onset in 10 affected members of the 14-member family was childhood in 2, teens in 2, and 30 to 50s in the remainder. They presented with episodes of dystonia, primarily affecting hands and feet symmetrically, and not triggered by movement, exercise, alcohol, caffeine or excitement. Attacks lasted 2-5 minutes and occurred daily. Treatment with various anticonvulsants, including acetazolamide, in one patient was ineffective. Mutation analysis of the MR-1 and GAD67 genes showed no linkage in affected family members. A genome-wide screen generated positive LOD scores at chromosome 2q31. (Spacey SD, Adams PJ, Lam PCP et al. Genetic heterogenicity in paroxysmal nonkinesigenic dyskinesia. **Neurology** May (2 of 2) 2006;66:1585-1590). (Reprints: Dr Sean Spacey, Neurology and Neurogenetics, Room S-127, 2211 Wesbrook Mall, UBC Hospital, Vancouver, BC V6T 2B5, Canada).

COMMENT. This family is distinguished from 10 previously reported PNKD families in not having a linkage to the myofibrillogenesis regulator 1 gene (MR-1), on chromosome 2q32-36, but instead, having a novel gene locus at chromosome 2q31. Also, caffeine and alcohol, which act as consistent triggers in other PNKD pedigrees with MR-1 gene mutations, failed to stimulate attacks. Two different genes appear to be responsible for PNKD, indicative of genetic heterogeneity.

ACTION DYSTONIA IN LESCH-NYHAN DISEASE

The motor disorder associated with Lesch-Nyhan disease (LND) was studied in a total of 44 patients (ages 2 to 38 years) seen at Johns Hopkins Hospital, Baltimore, MD, and other US and international centers. A characteristic motor syndrome begins with hypotonia and/or delayed motor development in the first 3-6 months of age, and is followed by involuntary movements between 6 and 24 months. The course is then relatively static with severe action dystonia, sometimes associated with choreoathetosis or ballismus, and less frequently, pyramidal signs affecting the lower extremities. The evolution of the motor syndrome of LND parallels the cognitive disability and behavioral syndrome that includes mental retardation and self-injurious behavior. Brain MRI in 25 patients showed cerebral atrophy or maldevelopment in 3, and 34% reduction in basal ganglia volume in 7 compared to normal controls. The results of this large multi-centre prospective study are compared with a review of 122 previous reports, including 254 patients. (Jinnah HA, Visser JE, Harris JC et al. Delineation of the motor disorder of Lesch-Nyhan disease. **Brain** May 2006;129:1201-1207). (Respond: HA Jinnah MD PhD, Meyer Room 6-181, Department of Neurology, Johns Hopkins Hospital, Baltimore, MD 21287).