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MOVEMENT DISORDERS

PAROXYSMAL DYSTONIC CHOREOATHETOSIS

The clinical characteristics of paroxysmal dystonic choreoathetosis (PDC) in 20 affected members of a large British family are reported from the Institute of Neurology, Queen Square, and the MR Unit, Hammersmith Hospital, London, UK. The findings are compared with those in 11 other PDC families cited in the literature. This hyperkinetic movement disorder consists of episodic attacks of chorea, dystonia, and ballism, beginning in infancy or childhood, and persisting with lesser frequency and severity into adulthood. Speech is frequently involved, diplopia may also occur, but consciousness is not disturbed. Frequency may be up to 3 times daily in childhood, mainly afternoon or evening, and yearly in adults. Attack duration in children is usually 30 min to 2 hours. Precipitants include stress, anger, and excitement, caffeine, alcohol, sleep deprivation, hunger, intercurrent illness, cold, prolonged exercise, and menstruation. Sudden movement or startle do not provoke attacks. Attacks are aborted by short periods of sleep, sometimes prevented by clonazepam, but are unaffected by various other antiepileptic drugs. Epilepsy and hysteria are common misdiagnoses. CSF monoamine metabolite levels may increase during attacks. MRI and PET scans are normal. Inheritance is autosomal dominant with high penetrance and linked to chromosome 2g. Male to femal ratio is 1.7:1. (Jarman PR. Bhatia KP. Davie C et al. Paroxysmal dystonic choreoathetosis: clinical features and investigation of pathophsiology in a large family. Mov Disord July 2000;15:648-657). (Reprints: Paul Jarman PhD, Department of Clinical Neurology, Institute of Neurology, Oueen Square, London, WC1N 3BG, UK).

COMMENT. In addition to paroxysmal dystonic choreoathetosis (PDC) that is non-kinesigenic, two further hyperkinetic movement disorders are described: 1) paroxysmal kinesigenic choreoathetosis (PKC), the most common, with attacks occurring frequently (100 or more daily), precipitated by sudden movement, and controlled by anticonvulsant drugs; and 2) paroxysmal exercise-induced dystonia (PED), of intermediate duration (5-30 min), the least common type. In addition, Nocturnal paroxysmal dystonia has been described (Lugaresi et al. 1986), usually

PEDIATRIC NEUROLOGY BRIEFS (ISSN 1043-3155) © 2000 covers selected articles from the world literature and is published monthly. Send subscription requests (S63 US; S65 Canada; \$73 airmail outside N America) to Pediatric Neurology Briefs - J. Gordon Millichap, M.D., F.R.C.P.-Editor, P.O. Box 11391, Chicago, Illinois, 60611, USA.

The editor is Pediatric Neurologist at Children's Memorial Hospital and Northwestern University Medical School, Chicago, Illinois, PNB is a continuing education service designed to expedite and facilitate review of current scientific information for physicians and other health professionals. Fax: 312-943-0123. Visit our web site: www.pnbpublishers.com misdiagnosed as nightmares, and generally treated as a complex partial seizure of frontal lobe origin. (see <u>Progress in Pediatric Neurology II</u>, PNB Publ, 1994, pp 193-4). Attacks of PDC, unlike PKC, were not benefited by anticonvulsants. PKC is sometimes referred to as PKD, and is often associated with infantile convulsions and epilepsy.

PAROXYSMAL KINESIGENIC DYSKINESIA AND EPILEPSY

Clinical and linkage data in 11 families with paroxysmal kynesigenic dyskinesia (PKD), with or without infantile convulsions (IC), were studied at the University of Utah, Salt Lake City, and multiple international centers. These families of diverse ethnic backgound had 95 affected individuals, the majority having chromosome 16 as the major locus, with heterogeneity. Kinesigenic dyskinesia (PKD) occurred in 42, infantile convulsions (IC) in 37, and a combined phenotype in 16. Infantile convulsions were usually afebrile and developed between 3 and 18 months; they remitted by age 3 years without treatment. Episodes of PKD began at 6 to 23 years (average 11 years), and were triggered by sudden movement, stress, and anxiety; they were controlled by anticonvulsant drugs, and were less frequent after age 25 years. EEGs were generally normal. The PKD/IC syndrome spans the phenotypic spectrum between epilepsy and movement disorder. (Swoboda KJ, Soong B W, McKenna C et al. Paroxysmal kinesigenic dyskinesia and infantile convulsions. Clinical and linkage studies. Neurology July (2 of 2) 2000:55:224-230), (Reprints: Dr Kathryn J Swoboda, University of Utah School of Medicine, Department of Neurology, Room 3R210, 50 North Medical Drive, Salt Lake City, UT 84132).

COMMENT. In an editorial, Berkovic SF classifies the Paroxysmal kinesigenic dyskinesia with infantile convulsions as a "channelopathy." (Paroxysmal movement disorders and epilepsy. Links across the channel. Neurology July 2000;55:169-170). Other paroxysmal disorders implicating channelopathies and manifested by seizures and dyskinesias include febrile seizures (sodium channel mutations), benign familial neonatal convulsions (potassium channel mutations), and episodic ataxia type 2 (calcium channel mutations). Different mutations are responsible for the diverse phenotypes. With PKD/IC syndrome, a single mutation may cause age-dependent symptoms: convulsions in infancy and dyskinesia in adolescence.

Risperidone and withdrawal dyskinesia is reported in a 13-year-old boy with ADHD, conduct disorder, and psychotic features, treated with lithium, valproic acid, and risperidone. (Lore C. <u>IAm Acad Child Adolese Psychiatry</u> August 2000;39:941). Two weeks after the resperidone was discontinued, a withdrawal dyskinesia manifested with mouth movements, neck twisting, and intermittent upward gaze. Although withdrawal dyskinesia may resolve within 6 weeks, the author raises concerns about the use of resperidone in ADHD children with ODD and CD, but without psychosis.

INTERNATIONAL PERSPECTIVE OF TOURETTE SYNDROME

The findings from a multisite, international database of 3500 individuals with Tourette syndrome (TS) from 22 countries are reported from the British Columbia Children's Hospital, Vancouver, Canada. Males outnumbered female patients, the average male:female ratio being 4.3:1. Mean age at onset of tics is 6.4 years, and mean age at diagnosis is 13 years. Comorbidities are reported in 88%, and the most common is ADHD with TS (60%). Other comorbid symptoms include OCD (27%), CD/ODD (15%), mood disorder (20%), ID (23%), and stuttering (8%).