Department of Medical Genetics, Mayo Clinic, 200 First Street SW, Rochester, MN 55905).

COMMENT. Examples of IEM requiring additional preliminary tests include Menkes' kinky-hair disease (serum copper and ceruloplasmin), and molybdenum cofactor deficiency (urine sulfite dipstick).

MOVEMENT DISORDERS

CLASSIFICATION OF PAROXYSMAL DYSKINESIAS

Forty six patients, ages ranging 1 to 77 years, with paroxysmal dyskinesias and classified according to precipitating factors and duration of attacks were reported from the Movement Disorder Clinic, Department of Neurology, Baylor College of Medicine, Houston, TX. Paroxysmal kinesigenic dyskinesia (PKD), occurring abruptly after a sudden movement, affected 13 patients; paroxysmal nonkinesigenic dyskinesia (PNKD) occurred spontaneously in 26; exertion-induced attacks (PED) affected 5; and episodes were precipitated only by sleep (PHD) in 1. The etiology was idiopathic in 22 and secondary to psychogenic illness in 9, to stroke in 4, trauma (3), encephalitis (2), multiple sclerosis (2), kernicterus (1), and migraine (1). Short duration (<5 min) and long-lasting attacks (>5 min) were about equal in incidence. None had loss of consciousness or other evidence of seizures and EEGs were normal in 34 tested. MRI was normal in 25 tested. Nine of 10 (90%) patients with PKD improved with medications, mainly carbamazepine, phenytoin, or clonazepam, compared to only 7 of 19 (37%) with PNKD. (Demirkiran M, Jankovic J. Paroxysmal dyskinesias: clinical features and classification. Ann Neurol October 1995;38:571-579). (Respond: Dr Jankovic, Department of Neurology, Baylor College of Medicine, 6550 Fannin, Suite 1801, Houston, TX 77030).

COMMENT. In contrast to the original (Mount and Reback, 1940) and other previous reports which empasized genetic and familial factors in etiology, the majority of the patients in the above series had sporadic and secondary paroxysmal dyskinesias. Menkes JH, in his Textbook of Child Neurology (Lea & Febiger, 1985), provides an excellent account of the various types of paroxysmal dyskinesia, classified according to 1) movement pattern - choreiform, athetoid, dystonic, or tonic; 2) familial or acquired; 3) kinesigenic or nonkinesigenic; 4) acquired etiology - perinatal asphyxia, reflex epilepsy, metabolic disorders (eg. idiopathic hyperparathyroidism), and multiple sclerosis. The specificity of response of the kinesigenic dyskinesias to anticonvulsant drugs, especially phenytoin, has been documented previously. Rare cases of hypnogenic dyskinesia, responding to lorazepam, and one patient with paroxysmal diplopia due to superior oblique myokymia following head injury, responding to carbamazepine, are described in the present series. An overlap between paroxysmal dyskinesia and epilepsy, migraine, and paroxysmal ataxia is discussed. The possible relation between migraine and paroxysmal dyskinesia mentioned in this report has not previously been noted.

HEREDITARY MYOKYMIA AND PAROXYSMAL ATAXIA

A family with autosomal dominant hereditary myokymia and paroxysmal ataxia, linked to chromosome 12p, is described from University

Hospital Groningen, The Netherlands. The proband, a 20 year old woman, had an onset of attacks of 'swinging legs', dizziness, involuntary jerky limb movements, dysarthria, and gait ataxia beginning at 6 years of age. Attacks up to four times daily and lasting 10 seconds to 5 min occurred at rest, during exercise, or when startled, standing up, or running. Myokymia of the hands and semirhythmical movements of fingers were noted on examination, and EMG showed myokymic discharges. An attack provoked by knee bends consisted of rhythmic involuntary shaking of limbs, and tremor and dysmetria on finger-to-nose test. Acetazolamide prevented attacks, but treatment was limited by paraesthesiae and development of tolerance. A 22 year old brother was also affected from 6 years of age, and the myokymia and ataxia were complicated by paroxysmal kinesigenic dystonia at 15 years, after a mild head injury. Another family member also had attacks of paroxysmal choreoathetosis. Carbamazepine controlled the attacks of dystonia and choreoathetosis but not the ataxia. Data on 6 affected family members are tabulated. (Lubbers WJ, Brunt ERP, et al. Hereditary myokymia and paroxysmal ataxia linked to chromosome 12 is responsive to acetazolamide. J Neurol Neurosurg Psychiatry October 1995;59:400-405). (Respond: Dr ERP Brunt, Department of Neurology, University Hospital Groningen, PO Box 30.001, 9700 RB Groningen, The Netherlands).

COMMENT. Provisional diagnoses of basilar migraine and epilepsy had been made initially in two of three children with familial paroxysmal ataxia reported in the UK (Hawkes CH, 1992). The EEG was normal and the MRI showed atrophy of the superior cerebellar vermis. All responded to acetazolamide. (see Progress in Pediatric Neurology II, PNB Publ, 1994, p149-150). MRI findings were not reported in the above series. An overlap and relation between paroxysmal types of ataxia, myokymia, choreoathetosis and dystonia is strengthened by this report, although different responses to acetazolamide and carbamazepine may suggest separate etiologies.

SEIZURE DISORDERS

HEREDITARY ROLANDIC EPILEPSY AND SPEECH DYSPRAXIA

A syndrome of nocturnal oro-facio-brachial partial seizures, secondarily generalized partial seizures, centro-temporal epileptiform discharges, associated with oral and speech dyspraxia and cognitive impairment, is described in a family of 9 affected members in three generations reported from Austin Hospital, Heidelberg (Melbourne), the University of Melbourne, and the Royal Children's Hospital, Melbourne, Australia. All affected individuals had nocturnal rolandic seizures limited to midchildhood. Inheritance of epilepsy and speech dyspraxia was autosomal dominant, with 100% penetrance for speech dyspraxia. Clinical anticipation was noted across the three generations of affected individuals, with increasingly severe speech dyspraxia, epilepsy as well as cognitive impairment. The syndrome has features resembling benign rolandic epilepsy (BRE) and may help in identifying the gene for BRE which is unassociated with clinical anticipation. It differs from the syndromes of Landau-Kleffner and epilepsy with continuous spike and wave during slow-wave sleep. (Scheffer IE et al. Autosomal dominant rolandic epilepsy and speech dyspraxia: a new syndrome with anticipation. Ann Neurol October 1995;38:633-642).