Pediatric Neurology: Chapter 88. Paroxysmal movement disorders and episodic ataxias (Handbook of Clinical Neurology)

Emilio Fernández-Alvarez, Belén Perez-Dueñas

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This chapter summarizes clinical symptoms of some paroxysmal dyskinesias (PDs) of infancy and childhood, as well as episodic ataxias. PDs refer to a complex group of disorders whose main feature is the occurrence of sudden, intermittent attacks of abnormal postures and involuntary movements. PDs can sometimes be symptomatic (secondary PDs), but usually an underlying cerebral lesion is not present (primary PDs). Some of the primary PDs are transient, such as benign paroxysmal torticollis of infancy. Chronic PDs are subdivided into nonkinesigenic (Mount and Reback type), kinesigenic (Kertesz type), and exercise-induced (Lance type) but cases that overlap with these types are on record. They are autosomal dominant inherited conditions. The myofibrillogenesis regulator-1 gene is responsible for nonkinesigenic PDs. To date, the genetic basis of kinesigenic PDs remains unknown. Several clinical entities associated epilepsy with PDs, such as infantile convulsions and choreoathetosis (ICCA). Exercise-induced PD type can be produced by mutations in the SLC2A1 gene that encodes Glut1 (glucose transporter type1). Episodic ataxias are inherited disorders of intermittent ataxia. The attacks are brief and triggered by abrupt exercise and emotional stimulus. Between attacks, palpebral and hand muscle myokymia is often seen in episodic ataxia type 1 (EA1). In episodic ataxia type 2 (EA2) interictal nystagmus is usually present. Some of these latter patients develop progressive ataxia with vermian atrophy. This disorder is associated with mutations in the human Ca channel alfa 1 subunit CACN1A4 gene.

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