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Tremor and Other Hyperkinetic Movements

Video Abstracts

Paroxysmal Kinesigenic Dyskinesia

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Abstract

Background: Paroxysmal kinesigenic dyskinesia (PKD) is a rare condition associated with heterozygous mutations in the proline-rich transmembrane protein 2 (*PRRT2*) gene.

Phenomenology Shown: In this article we illustrate the phenomenology of PKD in a male previously misdiagnosed with Tourette's syndrome.

Educational Value: Regardless of the underlying phenotype, PKD is highly responsive to some antiepileptic drugs.

Keywords: Paroxysmal kinesigenic dyskinesia, *PRRT2*, carbamazepine

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A 28-year-old male with paroxysmal and short-lasting dystonia triggered by sudden movement was evaluated at our center. Many short-lasting dystonia events occurred per day; these were preceded by aura-like symptoms (e.g., paresthesias in the affected limb). Consciousness was preserved during these episodes and physical examination between dystonia episodes was normal. Symptom onset in this case was at age 11. Epilepsy was suspected first; however, repeat investigations with electroencephalography and neuroimaging were normal. The patient's presentation was previously misinterpreted as Tourette's syndrome despite the absence of vocal tics. This presentation is rather compatible with paroxysmal kinesigenic dyskinesia (PKD), a rare movement disorder associated in most cases with heterozygous mutations in the proline-rich transmembrane protein 2 (*PRRT2*) gene (DYT10).[1] Targeted analysis of this gene (both sequencing and MLPA) was normal and whole-genome sequencing was not contributory in this case. Focal dystonia, in either the toes or the arms, was evident after standing up and running. In addition, running triggered generalized dystonia with falls and impaired speech, which are unusual in PKD (Video 1). Dystonia receded completely after treatment with carbamazepine was initiated. This good treatment response to antiepileptic drugs (carbamazepine or phenytoin) constitutes one of the criteria proposed for PKD.[2] Other hyperkinesias that can occur in PKD patients include chorea and/or ballism.[2,3] Aura-like symptoms (paresthesias and stiffness

Video 1. Phenomenology of Paroxysmal Kinesigenic Dyskinesia in a 28-year-old Male. **Segment 1.** Mild and short-lasting posturing in the left big toe after standing up. Running triggers generalized dystonia leading to a fall and impaired speech; consciousness is preserved. **Segment 2.** This segment shows the remarkable good response to carbamazepine.

in the affected limb) before the attacks, as described in this case, are very common.[3] Differential diagnosis of PKD includes several acquired conditions affecting the basal ganglia and familial conditions such as paroxysmal exercise-induced dyskinesia, paroxysmal hypnogenic dyskinesia, and glucose transporter type 1 deficiency syndrome (Glut1-DS).[3]

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