Paroxysmal Kinesigenic Dyskinesia

A 14-year-old boy presented with history of multiple daily episodes of abnormal movements of the body for the last 2 years. The event consisted of sudden onset of uncontrollable, bizarre, twisting movement of both arms and legs lasting for 10-30 seconds with no impairment of consciousness. These events were often precipitated by sudden movement. There was no family history of similar events. His neurological examination, magnetic resonance imaging and electroencephalography were unremarkable.

He was on six antiepileptic drugs (valproate, levitiracetam, phenytoin, topiramate, lacosamide, clobazam) with no reduction in frequency of these events. Home video was reviewed and clinical possibility of paroxysmal kinesigenic dyskinesia (PKD) was considered (***Fig.1*** and ***Web Video* 1**). A homozygous single base pair deletion (p.Arg217GlufsTer12) in exon 2 of the *PRRT2* gene was detected on Next generation sequencing. Subsequently, he was treated with oral carbamazepine, and showed a dramatic reduction in frequency of these events. Subsequently, all other antiepileptic drugs were tapered; he is event-free for last 1 year on carbamazepine (10 mg/kg/day).

Primary paroxysmal kinesigenic dyskinesia (PKD) is a hyperkinetic movement disorder characterized by recurrent, brief (usually less than a minute) episodes of choreathetoid or dystonic movement without alteration of consciousness. These events can be precipitated by sudden movement; hence, the term kinesigenic. In contrast, paroxysmal non kinesigenic dyskinesia (PNKD) is trigerred by emotional stress, fatigue and not by sudden motor movement. PKD has favourable prognosis with good response to treatment. *Proline-rich transmembrane protein (PRRT2)* gene mutation has been implicated in benign familial infantile epilepsy, infantile convulsions and choreathetosis and PKD. The present case is a sporadic *PRRT2* mutation-related PKD that needs wider, recognition among pediatricians.

Fig. 1 *A still image from the video of paroxysmal kinesigenic dyskinesia depicting abnormal twisting movement of upper limb during the clinical event (See video at website).*

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