Paroxysmal Kinesigenic Dyskinesia

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Abstract
We present a case of paroxysmal kinesigenic dyskinesia (PKD) in a 21 year old girl, with no family history of similar episodes. The episodes were short (lasting less than a minute), frequent, occurring 5 to 10 times a day, self-limiting dystonia of her right upper limb precipitated by sudden movement. She also had a past history of partial seizures with secondary generalization in her childhood. She responded to phenytoin, with cessation of events after 1 month of treatment. This case impresses upon the hypothesis stating the association between seizure activity and PKD probably due to a common foci of origin. Awareness of this condition is required as it is easily treatable but frequently misdiagnosed.

Introduction
Paroxysmal dyskinesia (PD) encompasses a rare group of movement disorders. It consists of sudden attacks of either chorea, athetosis, dystonia, hemiballismus or a combination of these.¹ They are further divided into subtypes based on the aggravating factors. The most common among these is Paroxysmal Kinesigenic Dyskinesia (PKD)¹ which is aggravated by sudden movement. It is etiologically classified as primary (familial or sporadic) and secondary. A lot of studies have been done to identify the underlying genetics. Diagnosis is a clinical one, with investigations targeted at ruling out secondary causes or other possible causes. Treatment success depends on the type of PD. Patients with PKD have shown to benefit the most with anticonvulsant therapy.²

Case Report
A 21 year old female visited the Neurology Clinic on 13/8/12 with attacks of abnormal posturing of her right upper limb for the past 3 months. The episode was characterized by outward twisting of her right upper limb. Each episode lasted for 10 to 20 seconds, with spontaneous resolution. She had at least one episode daily with a maximum of 5 to 10 episodes per day. Aggravating factors included sudden movements such as on initiation of walking or change of position like rising from a sitting position. These episodes were sudden in onset, without any tingling sensation, or prior warning. She had no history of any loss of consciousness.

She has a past history of partial seizures with secondary generalization, first noticed at the age of 1 year 10 months. She was started on Tab Carbamazepine. A few months after her last seizure attack in 2000, she was switched over to Tab Phenytin, gradually, tapered to 50 mg a day and was to be stopped after 2 months. However, she continued it till Jan, 2012. She had been off Phenytoin for 5 months, when she noticed her first incidence of abnormal posturing. On presentation, she was not taking any medication.

There was no history of any such similar episode in her family members and no family history of epilepsy. She had no history of any developmental delay. There were no other comorbidities.

On examination, patient was of average build with a PR- 80/min, regular, BP-120/80 mm Hg. General examination and systemic examination were unremarkable. On investigation: Hb- 11.89 g/dl, WBC- 5800/mm³, N62, L32 E5 M1, Platelet: 1,86,400/mm³, RBS: 98 mg/dl, LFT- normal, S. Ca – 8.96, S. PO₄ -3.01, ANA (HEP-2) – 1:80 (negative), ASO titre - 116.52 (N<200), serum ceruloplasmin - 56.36 mg/dL (Normal). CT brain- normal, awake EEG: unremarkable except for occasional non-specific fronto-central slowing.

Final diagnosis was paroxysmal kinesigenic dyskinesia (dystonia) with a history of partial seizures with secondary generalization.

She was restarted on Tab Phenytoin 50 mg OD. Gradually increased to 100 mg OD and finally to her current dose of 100 mg BD. The episodes decreased in frequency and at after 1 month the attacks completely stopped.

Discussion
Familial Paroxysmal Choreoathetosis coined by Mount and Reback in 1940 was the first mention of a similar movement disorder, precipitated by coffee and alcohol. In 1967, Kertesz³ identified sudden movement as the precipitating factor of such attacks and termed it paroxysmal kinesigenic choreoathetosis. The current term paroxysmal kinesigenic dyskinesia was coined in 1995 by Demirkiran and Jankovic¹ acknowledging that these attacks can manifest as any form of dyskinesia – chorea, athetosis, dystonia, hemiballismus or a combination of these. Paroxysmal dyskinesia is an uncommon group of movement disorders. It is characterized by attacks of sudden involuntary movements with intact consciousness. Based on the duration of the attack it can be short lasting (<5 mins) or long lasting (>5 mins). Based on the aggravating factors Demirkiran and Jankovic suggested a classification:¹ (1) paroxysmal kinesigenic dyskinesias (PKD) aggravated by sudden movement, (2) paroxysmal non-

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kinesigenic dyskinesias (PNKD) not triggered by sudden movement, but by hyperventilation, stress, caffeine, alcohol, (3) paroxysmal exercise-induced (exertion-induced) dyskinesias (PED) as a result of prolonged periods of physical exercise and (4) paroxysmal hypogenic dyskinesias (PHD) or nocturnal paroxysmal dystonia (NPD) refers to attacks of dystonia during non-REM sleep. Various genes have been implicated for the different types and Bruno et al. reported genotype phenotype correlations.

Paroxysmal kinesigenic dyskinesias (PKD) are the commonest of the types. It is characterized by episodes (paroxysmal) aggravated by sudden voluntary movement (kinesigenic) resulting in involuntary movements (dyskinesia). Etiologically they are divided into primary (familial-autosomal dominant or sporadic) and secondary. Secondary causes include hypocalcemia, hypoglycemia, hyperglycemia, central nervous system trauma, multiple sclerosis (MS), stroke, hyperthyroidism or hypoparathyroidism. The onset is usually before the age of 20 years, but may be variable especially in secondary cases. It has a higher predilection in males. Due to proximity of the loci on chromosome 16, PKD has also been linked with infantile convulsions and choreoathetosis (ICCA) syndrome, in which patients develop paroxysmal choreoathetosis with a history of afebrile seizures during infancy. Characteristically, the precipitating factor is standing up quickly or getting startled. Consciousness is intact and it may be preceded by an aura comprising of paresthesia or abnormal sensation of the affected area. Limbs are most commonly involved, but the muscles of the face, neck and trunk may be involved too. The frequency of episodes varies from 1 per month to up to 100 per day, with majority having daily episodes. Most attacks are brief, lasting less than 1 minute. After the attack, neurological examination is normal. If severe, the episodes may interfere with walking, standing and affect activities of daily living.

The pathophysiology remains unknown. Dysfunction of the basal ganglia may play a role as evidenced by SPECT studies revealing hyperactivity in the basal ganglia. Our patient, also had a history of partial seizure prior to the PKD, thus supporting this hypothesis of the origin of seizure-like discharges from the basal ganglia. Another hypothesis suggested is the origin of discharges from the basal ganglia with spread to the cerebral cortex. A locus for PKD has been identified on 16p11.2-q11.2. Mutations of this PRRT2 gene on chromosome 16 have been attributed to this disorder. There is no definitive diagnostic test for PKD. Routine blood tests, other tests, or neuroimaging (CT, MRI) or neurophysiologic studies (EEG) may be conducted to confirm or rule out secondary causes, or rule out other possible causes, especially epilepsy. PKD has an excellent response to anticonvulsant medications. Phenytoin was one of the first effective medication used. Carbamazepine is which is found to be effective in a majority is also commonly used. Other effective medications include phenobarbitone and benzodiazepines. Precipitating factors should be avoided. Some cases are seen to spontaneously resolve. Awareness of this condition is required as it is frequently misdiagnosed.

References