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CASE REPORT

A Case of Paroxysmal Kinesigenic Dystonia : Epilepsy Mimic

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Abstract:

Background: *We report a rare case of movement disorder Paroxysmal kinesigenic dystonia who was misdiagnosed as Epilepsy and Conversion disorder.* ***Case report:*** *A 35 -year female presented with episodic painful twisting of her Right hand and arm only when she is awake. These events were triggered by sudden movements and would last several seconds to minutes. Her symptoms were unilateral and her physical and neurological examinations were normal. Treatment with anticonvulsants Oxcarbazepine improved her symptoms.* ***Conclusion:*** *Although an uncommon movement disorder, it is important to recognize the clinical presentation of Paroxysmal kinesigenic dystonia as most patients respond very well to medical treatment.*

Key Words: *Anticonvulsants; Oxcarbazepine; Movement disorder; Paroxysmal Nonkinesigenic dystonia; Medical treatment.*

Introduction:

Paroxysmal dyskinesias (PDs) are a rare heterogeneous group of disorders characterized by sudden attacks of involuntary movements like chorea, athetosis, dystonia, hemiballismus or combination of them. Paroxysmal kinesigenic dyskinesia (PKD) involves sudden attack of dyskinesias induced by voluntary movement. PKD most commonly occurs sporadically or as an autosomal dominant trait with variable penetrance. The exact pathophysiology of PKD is unknown although basal ganglia dysfunction appears to play a major role. Although the precise gene remains unknown, genetic linkage studies have isolated loci on chromosome 16. The episodic nature of PKD and its relationship with other episodic disease, such as Migraine, epilepsy & episodic ataxia, suggests channelopathy as a possible underlying pathology. PKD may remit spontaneously, but it also responds well to anti-convulsants as well as other agents[1, 2,3].

PD subtypes include

1) Paroxysmal kinesigenic dyskinesia (PKD) which induced by sudden voluntary movements;
2) Paroxysmal nonkinesigenic dyskinesia (PNKD) which occurs at rest: PKND is characterized by spontaneous attack that tends to be more dystonic in nature. Attacks tend to be longer & less frequent than PKD lasting 10 minutes to 6 hours. Attacks are precipitated by alcohol, caffeine, stress & less responsive to anti consultants.
3) Paroxysmal exertion-induced dyskinesia (PED) triggered by prolonged exercise: PED involves attacks provoked by ongoing exertion & lasting for 5-30 minutes, attacks occurs 10-15 minutes after exercise whereas attack of PKD occurs immediately on movements. In PED attacks are dystonic & involve the exercised body parts, they stop within 10-15 minutes of rest. Anticonvulsants have not proved useful in PED.
4) Paroxysmal hypnogenic dyskinesia (PHD) which occur in sleep. It is characterized by the patient suddenly awakening with cry alongwith involuntary dystonic & ballistic movements lasting upto 45 seconds[1, 2, 3].

Most cases of PKD are primary, categorized as either familial (usually autosomal dominant) or

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idiopathic. Primary PKD usually has its onset in childhood between 6-16 years with a range of 4 months to 57 yrs. Males are more affected than females having ratio 2:1 to 4:1. The condition may be sporadic. It is inherited as autosomal dominant trait with variable penetrance. The clinical features is quiet distinctive of sudden onset involuntary movement precipitated by sudden movement. Attacks are usually unilateral followed bilateral attack. Extremities are more affected than face, neck & trunk. There is no alteration or loss of consciousness. However, in some cases a specific cause for the PKD has been identified, such as multiple sclerosis (MS), vascular lesions, trauma, or acquired metabolic abnormalities. Primary PKD is short lasting < 5 min, occurs in childhood whereas secondary PKD are tends to be longer in duration > 5 min & frequent in adult. Both children and adults are affected by PDs, which often cause diagnostic delays or an incorrect diagnosis due to a normal neurological examination between attacks and lack of familiarity with these movement disorders. Since most patients with PDs respond very well to medical treatment, it is important to recognize their clinical presentation [4, 6, 7].

Here in we report a case of PKD in a patient which is diagnosed as a focal epilepsy and Conversion disorder.

Case Report:

A 35-year-old female was admitted in our department with history of episodes of painful twisting of her right hand and arm for last 6 months. These abnormal events would last anywhere between several seconds to minutes and were triggered by sudden movements and also by when she was abruptly asked to do something. Her symptoms were unilateral and she denied any aura, post ictal confusion, changes in her level of consciousness, visual or speech problems, tongue biting, or bowel and bladder incontinence. It is for first time & no family history. Both her general physical and neurological examinations were normal in between attack. And during attack there was painful dystonic limb posturing with intact consciousness. Magnetic resonance imaging of the brain was also unremarkable. She was started on phenytoin but this was changed to Oxcarbazepine when she admitted in Department of neurology, Bangabandhu Sheikh Mujib Medical University (BSMMU), Dhaka, Bangladesh. Her symptoms significantly improved with Oxcarbazepine treatment. Secondary causes of PNKD were ruled out.

Discussion:

PKD is a rare neurologic condition that has an estimated prevalence of 1 in 150,000 [4]. The onset of PKD is typically between 6-months to 40-years of age, with males more commonly affected than females [1, 2, 5]. Although the Pathophysiology of PKD remains unknown, several familial cases have been linked to a peri-centromeric chromosome 16 locus [8].

Diagnostic criteria include[6,7]

1. An identified kinesigenic trigger for the attacks, short duration of attacks (< 1 minute),
2. No pain or loss of consciousness during attacks,
3. Exclusion of other organic diseases,
4. Normal neurologic examination between attacks,
5. Age at onset between 1-20 years if no family history of PKD, and
6. Control of attacks with phenytoin or carbamazepine

A startle, sudden movement, hyperventilation, or continuous exercise can precipitate attacks, which typically last between a few seconds to 5 minutes [2]. The frequency of attacks can range from as many as 100 per day to fewer than 1 per month, with the extremities more often affected than the face, neck, or trunk. Symptoms are often unilateral, but can become bilateral, and there is usually a short refractory period before another attack can be triggered[9]. With PKD patients responding well to pharmacotherapy, it is important that an early diagnosis is made. Carbamazepine is the drug of choice, but a beneficial response has also been reported for other anticonvulsants such as phenytoin, oxcarbazepine, and barbiturates[10]. A careful differentiation from other movement disorders can also help avoid years of anguish and uncertainty for both patients and their families.

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