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Paroxysmal Kinesigenic Dyskinesia in a Mother and Daughter

Waqas Ullah KHAN[1,2], G. STAIOS[1,2], Abdul Qayyum RANA[1]

[1]Parkinson's Clinic of Eastern Toronto & Movement Disorders Center, Rouge Valley Centenary Hospital;
[2]University of Toronto, Toronto, Ontario, Canada

Abstract

We report a rare case of familial paroxysmal kinesigenic dyskinesia. A 42-year-old woman and her 13-year-old daughter both presented with episodic curling of their hand and arm. These events were triggered by sudden movements and would last several seconds. Both patients' symptoms were unilateral and their physical and neurological examinations were normal. Treatment with carbamazepine improved their symptoms. Although an uncommon movement disorder, it is important to recognize the clinical presentation of paroxysmal kinesigenic dyskinesia as most patients respond very well to medical treatment.

Key Words: anticonvulsants; movement disorder; paroxysmal dyskinesia; paroxysmal kinesigenic dyskinesia; pharmacotherapy

Introduction

Paroxysmal dyskinesias (PDs) are a rare heterogeneous group of disorders characterized by sudden attacks of involuntary movements (1, 2). Both children and adults are affected by PDs, which include paroxysmal kinesigenic dyskinesia (PKD), induced by sudden voluntary movements; paroxysmal non-kinesigenic dyskinesia (PKND), which occur at rest; paroxysmal exertion-induced dyskinesia (PED), triggered by prolonged exercise; and paroxysmal hypnogenic dyskinesia (PHD), which occur in sleep (3, 4).

Lack of familiarity with these movement disorders and a normal neurological examination between attacks often cause diagnostic delays or an incorrect diagnosis. Since most patients with PDs respond very well to medical treatment, it is important to recognize their clinical presentation (4, 5). Herein, we report a case of PKD in a patient whose daughter developed a similar condition.

Case Report

A 42-year-old woman was referred to our clinic with respect to episodes of hand curling that started when she was 14-years of age. These abnormal events would last anywhere between 2 to 15 seconds and were triggered by sudden movements or when 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. During her childhood, activities like running or being called upon in school would provoke brief episodes of arm or hand curling. Both her general physical and neurological examinations were normal. Magnetic resonance imaging of the brain was also unremarkable. She was started on phenytoin while living in Guyana, but this was changed to carbamazepine when she arrived in Canada. Her symptoms significantly improved with carbamazepine treatment.

Interestingly when this woman's daughter reached the age of 13-years, she developed episodic curling of her hand and arm that would last several seconds. Similar to her mother, the daughter's symptoms were unilateral and triggered by sudden movements. Her general physical and neurological examinations were also normal, and she responded very well to carbamazepine. With the exception of these two cases, there is no family history of this or any other neurological disorders. Secondary causes of PKD 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, 5-8). Although the pathophysiology of PKD remains unknown, several familial cases have been linked to a pericentromeric chromosome 16 locus (9).

Diagnostic criteria include: an identified kinesigenic trigger for the attacks, short duration of attacks (< 1 minute), no pain or loss of consciousness during attacks, exclusion of other organic diseases, normal neurologic examination between attacks, age at onset between 1-20 years if no family history of PKD, and control of attacks with phenytoin or carbamazepine (4, 5). A startle, sudden movement, hyperventilation, or continuous exercise can precipitate attacks, which typically last between a few seconds to 5 minutes (2, 7, 8, 10). 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 (7, 8).

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 (11). A careful differentiation from other movement disorders can also help avoid years of anguish and uncertainty for both patients and their families.

REFERENCES

1. Weber YG, Lerche H. Genetics of paroxysmal dyskinesias. Curr Neurol Neurosci Rep. 2009;9:206-11.
2. Bhatia KP. The paroxysmal dyskinesias. J Neurol. 1999;246:149-55.
3. Mehta SH, Morgan JC, Sethi KD. Paroxysmal dyskinesias. Curr Treat Options Neurol. 2009;11:170-8.
4. Van Rootselaar AF, Van Westrum SS, Velis DN, Tijssen MA. The paroxysmal dyskinesias. Pract Neurol. 2009;9:102-9.
5. Bruno MK, Hallett M, Gwinn-Hardy K, Sorensen B, Considine E. *et al*. Clinical evaluation of idiopathic paroxysmal kinesigenic dyskinesia: new diagnostic criteria. Neurology. 2004;63:2280-7.
6. Blakeley J, Jankovic J. Secondary paroxysmal dyskinesias. Mov Disord. 2002;17:726-34.
7. Sethi KD. Paroxysmal dyskinesias. In: "Parkinson's Disease and Movement Disorders". Lippincott, Williams & Wilkins. 4eds., Philadelphia; 2002: 430-7.
8. Lotze T, Jankovic J. Paroxysmal kinesigenic dyskinesias. Semin. Pediatr Neruol. 2003;10:68-79.
9. Swoboda KJ, Soong B, McKenna C, Brunt ER, Litt M. *et al*. Paroxysmal kinesigenic dyskinesia and infantile convulsions: clinical and linkage studies. Neurology. 2000;55:224-30.
10. Demirkiran M, Jankovic J. Paroxysmal dyskinesias: clinical features and classification. Ann Neurol. 1995;38:571-9.
11. Mehta SH, Sethi KD. Therapeutics of paroxysmal dyskinesias. In: "Therapeutics of Parkinson's Disease and Other Movement Disorders. Wiley-Blackwell. 1 eds., West Sussex; 2008:91-120.

Abdul Qayyum Rana, M.D., FRCPC
Parkinson's Clinic of Eastern Toronto &
Movement Disorders Centre
Rouge Valley Centenary Hospital
2867 Ellesmere Road, Toronto, ON.,
Canada, M1E5E9
E-mail: ranaaq@yahoo.com