

Case Report

Paroxysmal Kinesigenic Dyskinesia: An Uncommon Familial Movement Disorder

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Abstract: Paroxysmal kinesigenic dyskinesia is a rare hyperkinetic movement disorder, characterized by short duration dystonic or choreoathetoid movements precipitated by sudden movement, a change in position, or hyperventilation. It can be difficult to distinguish this syndrome from seizures. We report a case of 15 years old boy and his 53 years old father, who presented with abnormal involuntary unilateral dystonic movements triggered by sudden movement and would last few seconds. All symptoms showed excellent response to carbamazepine. Although an uncommon movement disorder, it is important to recognize clinical presentation of paroxysmal kinesigenic dyskinesia as most patients respond very well to medical treatment.

Keywords: Familial, Paroxysmal kinesigenic dyskinesia (PKD), Carbamazepine.

INTRODUCTION

Paroxysmal dyskinesias (PDs) represent a rare heterogeneous group of movement disorders with typical childhood onset [1, 2]. PDs include paroxysmal kinesigenic dyskinesia (PKD), induced by sudden involuntary movements; paroxysmal non kinesigenic dyskinesia (PKD), which occur at rest; paroxysmal exertion-induced dyskinesia (PED), triggered by prolonged exercise; and paroxysmal hypnogenic dyskinesia (PHD), which occur in sleep [3]. Lack of familiarity with these conditions and a normal neurological examination between attacks often cause diagnostic delay or results in an incorrect diagnosis. Since most patients with paroxysmal dyskinesias respond very well to medical treatment, it is important to recognize their clinical presentation and differentiation from epilepsy [4, 5].

Herein, we report an Indian family (son and his father) suffered from PKD.

CASE REPORT

A 15 year old boy presented to us with history of episodic abnormal involuntary twisting and curling of upper and lower limbs of either side with occasional involvement of face, lasted for few (10-30) seconds and precipitated by sudden movement or sudden change in velocity during a movement, at a frequency of 8-10 attacks per month. Movements started after 12 year of age. He denied any aura, changes in level of consciousness, visual or speech problems, tongue

biting, bladder and bowel incontinence during attacks or postictal confusion. His birth history was uneventful and development milestones were normal. His neurological and other systemic examinations were normal. His 53 year old father also had history of similar abnormal movements, started when he was about 15 year old with a frequency of 15-20 episodes per month. His movements were also unilateral and triggered by sudden movements. His father's physical and neurological examinations were normal and he was not on any treatment. A workup included blood counts, electrolytes, blood sugar, serum calcium, thyroid functions, vasculitic workup, magnetic resonance imaging (MRI) of brain and electroencephalogram (EEG). All investigations were within normal limits. We made clinical diagnosis of PKD as per Bruno's criteria [4]. Genetic study was done after getting informed consent, which revealed mutation in PRRT2 gene on chromosome 16, which confirmed the diagnosis. Both, patient and his father were put on carbamazepine and symptoms showed marked remission.

DISCUSSION

Paroxysmal kinesigenic dyskinesia (PKD) is a rare neurologic condition that has an estimated prevalence of 1 in 150,000 [5]. It is characterized by unilateral or bilateral involuntary movements, precipitated by sudden movements such as standing up from sitting position, being startled or change in velocity. Attacks include combinations of dystonia,

choreoathetosis or ballismus, sometimes preceded by an 'aura' like sensation and without loss of consciousness [6]. Limbs are commonly involved and generally manifest unilaterally but may alternate or even bilateral. Neck, trunk and face may also be affected [7]. Onset of PKD is typically between 6 months to 40 years of age, with male predominance [1, 2].

There are two types of PKD, primary and secondary. Primary PKD is classified as familial and sporadic. Familial is most commonly inherited as autosomal dominant with variable penetrance. The gene associated with PKD has been mapped to chromosome 16, and mutations in PRRT2 gene was identified to cause PKD. This gene consists of four exons encoding the proline-rich transmembrane protein 2, which encompasses 340 amino acids and contains two predicted transmembrane domains [8]. PRRT2 is highly expressed during the development of the nervous system, and a truncating mutation alters the subcellular localization of the PRRT2 protein. PRRT2 has been found to be mainly expressed in the basal ganglia, a brain area possibly involved in the PKD pathogenesis. Secondary PKD is caused by multiple sclerosis, stroke, hypocalcemia, hypoglycemia, pseudo-hypoparathyroidism, hyperglycemia or head injury [8].

We reported a father and son, who were diagnosed using strict set of guidelines as per Bruno *et al.* [4]. Diagnostic criteria include: age at onset between 1-20 yrs, the attacks of involuntary movements last less than one minute and have a known kinesigenic trigger, no pain or loss of consciousness during attacks, exclusion of other organic diseases, normal neurologic examination between attacks, frequency of attacks can range from fewer than 1 per month to as many as 100 per day, symptoms are usually unilateral and control of attacks with anticonvulsants like carbamazepine. We ruled out all possible secondary causes of PKD by relevant investigations. Diagnosis is supported by clinical criteria, positive family history and good response to carbamazepine.

As PKD patients respond well to pharmacotherapy, it is important that an early diagnosis is made and treatment initiated with carbamazepine, which is drug of choice, but other anticonvulsants such as phenytoin, oxcarbazepine and barbiturates has also been found beneficial [3].

CONCLUSION

A careful differentiation from other movement disorders can help to 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(3): 206-211.

2. Bhatia KP; The paroxysmal dyskinesias. *J Neurol.*, 1999; 246(3): 149-155.
3. Mehta SH, Morgan JC, Sethi KD; Paroxysmal dyskinesias. *Curr Treat Options Neurol.* 2009; 11(3): 170-178.
4. Bruno MK, Hallett M, Gwinn-Hardy K, Sorensen B, Considine E, Tucker S *et al.*; Clinical evaluation of idiopathic paroxysmal kinesigenic dyskinesia: New diagnostic criteria. *Neurology*, 2004; 63(12): 2280–2287.
5. Van Rootstelaar AF, Van Westrum SS, Velis D N, Tijssen MA; The Paroxysmal dyskinesias. *Pract Neurol.*, 2009; 9(2): 102-109.
6. Spacey S, Adams P; Familial Paroxysmal Kinesigenic Dyskinesia. In Pagon RA, Adam MP, Ardinger HH, Wallace SE, Amemiya A, Bean LJH editors; *GeneReviews*® [Internet]. Seattle (WA): University of Washington, Seattle, 1993-2015.
7. Unterberger I, Trinka E; Diagnosis and treatment of paroxysmal dyskinesias revisited. *Ther Adv Neurol Disord.*, 2008; 1(2): 4–11.
8. Prabakara S, Kolandaswamy A; Molecular analysis of PRRT2 gene in case of paroxysmal kinesigenic dyskinesia patient. *Ann Indian Acad Neurol.*, 2014; 17(4): 459-462.