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Paroxysmal kinesigenic dyskinesia – case report and brief literature review

Wanninayake L[1,2], Somarathne A[2], Nair N[2], Ranawaka UK[2,3]

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Abstract

Paroxysmal dyskinesias are a group of rare movement disorders with several distinct varieties. Clinical history is the key to the diagnosis of the paroxysmal dyskinesias. This case report describes a patient with paroxysmal kinesigenic dyskinesia, the commonest form of paroxysmal dyskinesia. Awareness of paroxysmal dyskinesias would help physicians to recognise these rare disorders and initiate appropriate treatment without delay.

Key words: paroxysmal kinesigenic dyskinesia, paroxysmal dyskinesia

Introduction

Paroxysmal dyskinesias are a group of rare movement disorders which are underdiagnosed and often misdiagnosed.[1,2] The first case was documented by Mount et al in 1940 and over the years several different types of paroxysmal dyskinesias have been described.[3] Classification of paroxysmal dyskinesias into different types is based on the trigger for the dyskinesia. The three main types are: paroxysmal kinesigenic dyskinesia (PKD; triggered by sudden activity), paroxysmal non-kinesigenic dyskinesia (PNKD; dyskinesia without a trigger), and paroxysmal exercise-induced dyskinesia (PED; triggered by exercise).[4] Each type is further subclassified into short lasting (duration less than 5 minutes) or long lasting (over 5 minutes) subtypes, based on the duration of the attacks.[5] Each subtype can be idiopathic or secondary to an underlying cause, and several secondary causes have been described for each type.[6,7] Idiopathic cases are sporadic or familial, usually with autosomal dominant inheritance. Familial PKD, PNKD, PED are linked to three separate genetic mutations: proline-rich transmembrane protein 2 (PRRT2), myofibrillogenesis regulator 1 (MR-1), and glucose transporter 1 (SLC2A1) genes, respectively.[8] A paroxysmal movement disorder that occurs in sleep (paroxysmal hypnogenic dyskinesia) was previously categorised as a type of paroxysmal dyskinesia, but is now considered a form of frontal lobe epilepsy.[6,9]

The first report of a paroxysmal dyskinesia from Sri Lanka was in 2003 by Ranawaka et al who described paroxysmal exercise-induced dyskinesia in a 27-year-old man.[10] Alibhoy et al subsequently reported 6 cases of paroxysmal kinesigenic dyskinesia.[11] Paroxysmal dyskinesias remain a group of movement disorders that are frequently misdiagnosed due to their rarity and lack of awareness among the clinicians.

Case report

We report a 14-year-old schoolboy presenting with episodic involuntary movements in the left upper limb, lower limb and left side of face. He had presented to the outpatient department (OPD) of the Colombo North Teaching Hospital (CNTH) with increasing frequency of the attacks and was admitted by a concerned OPD medical officer. The frequency had increased from 1-2

[1]*Department of Pharmacology, Faculty of Medicine, University of Colombo,Sri Lanka,* [2]*University Medical Unit, North Colombo Teaching Hospital, Ragama, Sri Lanka,* [3]*Department of Medicine, Faculty of Medicine, University of Kelaniya, Sri Lanka.*

Correspondence: LW, e-mail: leonardwanni@gmail.com

https://orcid.org/0000-0002-6794-2778

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attacks a day to over 10 attacks per day over the course of 2 months. Observation of the attacks revealed that they were brief, lasting from a few seconds to a maximum duration of 2 minutes. They were triggered by standing from a seated position and starting to walk or run. During the attack, there was dystonia of the left upper limb and left lower limb, with left sided oro-facial dyskinesias without associated dysarthria. There was no loss of consciousness during the attacks and no drowsiness following the attacks. The attacks were not triggered by caffeine. The increasing frequency of attacks was distressing for the boy and the parents, however school performance or day to day activities were not adversely affected. There was no history of birth insults, childhood brain injury, seizures, cognitive impairment, or developmental delay. No history of head trauma or substance abuse was noted. There was no family history of similar movement disorders or seizure disorders.

The boy was appropriately built for his age. He was alert and oriented. Speech content, fluency and articulation were normal. No skin rashes were noted. Cardiac, respiratory, and abdominal examinations were normal. Neurological examination of the cranial nerves, upper and lower limbs, coordination, and gait did not reveal any abnormality between attacks. Fundi were normal on ophthalmoscopy. There were no Kayser-Fleischer rings on slit lamp examination.

The full blood count, serum electrolytes, serum creatinine, capillary blood glucose, liver profile, thyroid function tests and creatinine phosphokinase levels were within normal limits. Brain imaging with non-contrast CT scan and contrast enhanced magnetic resonance imaging [MRI] were normal. EEG was within normal limits.

A clinical diagnosis of paroxysmal kinesigenic dyskinesia was made and the patient was started on carbamezapine 100 mg twice daily. Following two doses of carbamazepine, the dyskinetic movements completely disappeared and there was no recurrence of attacks. He is being followed up in the outpatient neurology clinic and remains asymptomatic.

Discussion

Paroxysmal dyskinesias can present with a variety of abnormal movements such as dystonic, choreic, athetoid, ballistic or a combination of these.[2,12] A careful clinical history with description of the attacks and recognition of the triggers is of paramount importance for diagnosis.

Paroxysmal kinesigenic dyskinesia (PKD), which is the commonest of the paroxysmal dyskinesias, is typically triggered by sudden change in movement, such as standing from a seated position, or beginning to walk or run. The attacks can also be triggered by hyperventilation, startle, and sudden light or noise stimulation.[6] About 70% of patients notice an aura of pins and needles sensation prior to the attacks.[13] The attacks are usually short lasting (<1 minute), typically affecting one side of the body. Dystonia is the commonest type of abnormal movement seen. There is no loss of consciousness during the attacks. Attacks are frequent, with most patients experiencing about 20 attacks per day, and some may have over 100 attacks per day.[1,4,13] Longer attacks are usually due to a secondary cause.[12] A set of diagnostic criteria with 6 elements was recently proposed for PKD (see Table 1).[13] EEG and neuroimaging findings are usually normal.[13]

Table 1. Diagnostic criteria for PKD

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

Although most PKD cases are idiopathic, its association with a variety of secondary causes such as multiple sclerosis, metabolic abnormalities, stroke, and trauma has been well documented.[4,13] PKD has shown a clear association with the syndrome of infantile convulsion and choreoathetosis (ICCA) which shares the PRRT2 mutation in chromosome 16.[14] A different locus, termed EKD2, on chromosome 16 was also reported to be associated with PKD.[15] PKD responds well to treatment with phenytoin and carbamazepine, at much smaller doses than used in the treatment of epilepsy.[16,17] Several other antiepileptics have been successfully used in patients.[18] The target of treatment is to achieve complete remission of the attacks.[18] The prognosis of PKD is usually very good with the attack frequency reducing with age and complete cessation often seen in adulthood.[13]

Our patient fulfilled all the clinical criteria for the diagnosis of PKD.[13] He reported the typical triggers of standing from a seated position, and initiation of walking and running. He complained of an aura with

numbness in the upper and lower limbs prior to the attacks. The attacks lasted less than 2 minutes and he did not lose consciousness during the attacks. There was no family history of a similar disorder, and no obvious secondary cause was identified. He responded dramatically to carbamazpepine. We were unable to perform genetic testing due to resource limitations.

PKD needs to be distinguished from other types of paroxysmal dyskinesias as treatment would differ. Paroxysmal non-kinesigenic dyskinesia (PNKD) is triggered by alcohol or caffeine. Similar to PKD, associated aura in the form of numbness and dysarthria can be seen.[13] Attacks usually last from 10 minutes to 1 hour, sometimes several hours. The attack frequency is less compared to PKD. The predominant movement is dystonic; however, other hyperkinetic movements have also been described. The patient is able to function normally in between the attacks.[13,19] Trigger avoidance is one of the mainstays of management of PNKD.[18] Benzodiazepines such as clonazepam and diazepam have been effective in treatment of PNKD, with clonazepam being the drug of choice.[5,19] PNKD does not respond well to carbamazepine. Similar to PKD, the attack frequency reduces with age.[9]

Paroxysmal exercise-induced dyskinesia (PED) is characterized by dyskinesias following a prolonged duration of exertion. It may rarely be triggered by exposure to cold.[9] Attacks last from 2-30 minutes (maximum 2 hours), and usually cease about 10 minutes after stopping exercise.[9] The attacks mainly affect the lower limbs, and the predominant movements are chorea and dystonia.[20] Associated auras are not seen. PED may respond to ketogenic diet or modified Atkins diet. Exercise limitation is effective but avoidance of exercise is not advised.[18] Response to medication is generally unsatisfactory.[18]

Paroxysmal dyskinesias are often misdiagnosed as simple or complex partial seizures. The key to establishing the diagnosis is careful history taking with pattern recognition. Video footage of the attacks would be helpful for diagnosis. The pattern of identified trigger, short duration of attacks, preservation of consciousness during attacks, lack of post-ictal symptoms, normal neurological examination and normal EEG would favour a diagnosis of paroxysmal dyskinesia over a seizure or epilesy.[21] Functional movement disorders should also be considered in the differential diagnosis of paroxysmal dyskinesias. Functional movements disorders would have variability, inconsistency, suggestibility, distractibility and suppressibility of the movements during the examination whereas paroxysmal dyskinesias would be less variable, more consistent and not distractable or suppressible during examination. Changes in the movement phenotype and duration in the same individual is likely to occur in functional movement disorders. Functional movement disorders are often seen following physical or emotional life events in contrast to paroxysmal dyskinesias which do not usually follow such life events.[22,23] The response of functional movement disorders to anticonvulsants would be less compared to paroxysmal dyskinesias.[23]

Conclusion

Patients with various movement disorders are regularly encountered by physicians, especially in outpatient settings. This case report of paroxysmal kinesigenic dyskinesia highlights the importance of a careful history in the evaluation of patients with abnormal movements, and the need for awareness regarding this rare and interesting group of movement disorders. Careful clinical evaluation is the key to avoiding delayed diagnosis and misdiagnosis.

Conflicts of interests

None.

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