

Paroxysmal Kinesigenic Dyskinesia in a Child with Positive Family History: A Case Report

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ABSTRACT:

Paroxysmal kinesigenic dyskinesia (PKD) is a rare movement disorder characterized by brief, recurrent episodes of involuntary movements triggered by sudden voluntary actions [1]. It typically presents in childhood and shows an excellent response to sodium channel blocking antiepileptic drugs [3,7]. We report an 11-year-old boy with classical PKD features, a positive maternal family history, normal neuroimaging, and a marked therapeutic response to low-dose oxcarbazepine after failure of sodium valproate and brivaracetam. Genetic analysis revealed a heterozygous pathogenic PRRT2 frameshift mutation, confirming autosomal dominant episodic kinesigenic dyskinesia (OMIM #128200) [2,4]. This case highlights the importance of early recognition and targeted therapy in this highly treatable pediatric movement disorder [6].

Keywords: *Paroxysmal kinesigenic dyskinesia; PRRT2; childhood movement disorders; familial dyskinesia; oxcarbazepine*

INTRODUCTION:

Paroxysmal kinesigenic dyskinesia (PKD) is the most common form of paroxysmal dyskinesia, typically presenting in childhood or early adolescence [1,3]. It is characterized by brief attacks of dystonia, chorea, or mixed hyperkinetic movements precipitated by sudden voluntary motion, without loss of consciousness [1]. Most cases follow an autosomal dominant inheritance pattern and are commonly associated with pathogenic variants in the PRRT2 gene [2,4,5]. Early recognition is crucial, as PKD responds dramatically to low-dose sodium channel-blocking antiepileptic drugs, preventing misdiagnosis and unnecessary treatment [6,7].

Case Presentation:

An 11-year-old boy, born of a non-consanguineous marriage, presented with recurrent paroxysmal involuntary movements since the age of four years. Episodes were triggered by sudden initiation of voluntary movements such as standing or walking. Attacks were brief, stereotyped, and self-limiting. There was no loss of consciousness, post-ictal confusion, or autonomic features. Neurological examination between attacks was

normal. A positive family history was present in the mother. Brain magnetic resonance imaging was normal. The child had previously been treated with sodium valproate and brivaracetam without clinical benefit. He was subsequently initiated on oxcarbazepine, following which there was a marked reduction in the frequency and severity of attacks, with excellent symptomatic control. Genetic testing revealed a heterozygous pathogenic frameshift mutation in exon 2 of the PRRT2 gene (c.649dup; p.Arg217ProfsTer8), confirming autosomal dominant episodic kinesigenic dyskinesia.

DISCUSSION:

This case represents a classical presentation of PRRT2-related paroxysmal kinesigenic dyskinesia. Hallmark features include childhood onset, kinesigenic trigger, preserved consciousness, positive family history, and excellent response to sodium channel-blocking agents [1,6]. Frameshift mutations involving exon 2 of PRRT2 are among the most frequently reported pathogenic variants in familial PKD [2,4,5].

Failure to respond to broad-spectrum antiepileptic drugs such as sodium valproate and brivaracetam, contrasted

with the dramatic response to oxcarbazepine, further supports the diagnosis, as PKD is particularly sensitive to sodium channel blockade [6,7]. Early recognition is essential to avoid misdiagnosis as focal epilepsy, which often leads to unnecessary investigations and prolonged ineffective treatment [3,8,10].

The prognosis of PKD is generally favorable, with many patients experiencing a reduction in attack frequency over time and the possibility of medication tapering under supervision [6,9].

CONCLUSION:

This genetically confirmed case highlights the importance of recognizing paroxysmal kinesigenic dyskinesia as a distinct, highly treatable pediatric movement disorder. Awareness of its characteristic clinical features and therapeutic response can facilitate early diagnosis and significantly improve patient outcomes [6].

Informed Consent:

Written informed consent was obtained from the patient's parent for publication of this case report.

Conflict of Interest:

The authors declare no conflict of interest.

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