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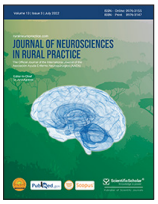

Letter to Editor

Associated movement disorder as a clue for the diagnosis of paroxysmal kinesigenic dyskinesia in a child with focal epilepsy

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Dear Editor,

A developmentally normal 10-year-old girl with an uneventful birth history presented with a history of the right focal tonic seizures with behavioral arrest for 6 months of age. She continued to have seizure episodes once in 6 months till 8 years of age while on levetiracetam and valproate. Parents also reported occasional abnormal movements (choreo-athetoid type), which were noticed during the initiation of movements. She was referred to us for no relief from symptoms despite two anti-seizure medications (ASMs). On examination, she had difficulty rising from the chair and initiating walking due to twisting and bending movements at the knees and ankles, which resolved with movements. The rest of her neurological assessment was normal. Her father had a similar history of only abnormal movements while rising from a chair and initiating walking since the age of 10, which resolved by 25 years without treatment. Based on the clinical presentation and significant family history, proline-rich transmembrane protein gene (PRRT2) mutation-related disorders and glucose transporter type-1 deficiency were considered. She was started on carbamazepine, and the choreoathetoid movements subsided over 6 months. Genetic testing was done at 6-month follow-up and revealed a heterozygous pathogenic variation c.47A>T (p.Glu16Val) in the exon 2 of the PRRT2 gene, suggestive of Paroxysmal kinesigenic dyskinesia (PKD).

PKD, a rare disorder with an autosomal dominant inheritance, is characterized by recurrent, transient episodes of chorea, dystonia with preserved consciousness and triggered by voluntary movement initiation due to pathogenic variations in the PRRT2 gene located on chromosome 16p11.2.[1] PRRT2 mutations are also associated with benign familial infantile seizures, hemiplegic migraine, episodic ataxia, and developmental delay.[2,3] Infantile convulsion and choreoathetosis syndrome is a variable form of PKD.[4] In patients with early-onset focal tonic seizures, a history of dyskinesias should be asked, along with a family history of kinesigenic dyskinesias, which serves as a diagnostic clue. These dyskinesias *per se* mimic epilepsy due to recurrent falls resulting in misdiagnosis. Like the index child, targeted therapy with ASMs, including carbamazepine, helps in complete remission in 80–90% of patients[5] and counseling is paramount.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

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Received: 02 March 2023 **Accepted:** 11 May 2023 **EPub Ahead of Print:** 31 May 2023 **Published:** 16 August 2023 **DOI**: 10.25259/JNRP_118_2023

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How to cite this article: Laxmi V, Gunasekaran P, Manjunathan S, Gupta R, Kumar A, Saini L. Associated movement disorder as a clue for the diagnosis of paroxysmal kinesigenic dyskinesia in a child with focal epilepsy. J Neurosci Rural Pract 2023;14:558-9.