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CORRESPONDENCE

Pyruvate Dehydrogenase Complex Deficiency Due to *PDHA1* Mutation—A Rare Treatable Cause for Episodic Ataxia in Children

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To the Editor: Episodic or recurrent cerebellar ataxia in children is rare, and neurometabolic conditions are an important cause [1]. Here, we present a young boy with episodic ataxia due to pyruvate dehydrogenase complex (PDC) deficiency with good response to thiamine supplementation.

A 5-y-old-boy, born of nonconsanguineous parentage, presented to us with three episodes of fever-triggered ataxia since 3 y of age. Each episode lasted for 2–7 d. On examination during the third event, he had impaired tandem gait, left upper limb incoordination and bilateral optic disc pallor. The child was completely normal interictally. He had unremarkable birth and developmental histories and family history was noncontributory.

His serum and cerebrospinal fluid lactate were elevated, while other metabolic parameters were normal. MRI brain revealed symmetric T2-weighted and fluid-attenuated inversion recovery hyperintensity in the bilateral dentate nuclei without any white matter or corpus callosum involvement. Clinical exome sequencing revealed a pathogenic hemizygous 4-base-pair duplication in exon 12 (c.1273_1276dup) of the *PDHA1* gene (frameshift mutation). He was initiated on high-dose thiamine, levocarnitine, and alpha lipoic acid following which he had only one minor episode in the next 3 y.

Involvement of corpus callosum, grey matter nuclei in brainstem, basal ganglia, thalamus, dentate nucleus, and ventriculomegaly are the neuroimaging findings reported in PDC deficiency [2, 3]. The diagnostic clues in this patient were elevated lactate and dentate nucleus involvement [2, 4]. However, isolated dentate nucleus involvement has not been reported yet. Usually, frameshift mutations are reported to cause more severe and fatal manifestations of PDC deficiency as compared to missense mutations, but our patient had a mild phenotype. Treatment options include high-dose thiamine, alpha lipoic acid, ketogenic diet, and dichloroacetate [2, 3]. PDC deficiency merits consideration in a child with episodic ataxia, since it is eminently treatable.

Declarations

Conflict of Interest None.

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