Pediatric Neurology 100 (2019) 104

Contents lists available at ScienceDirect

Pediatric Neurology

journal homepage: www.elsevier.com/locate/pnu

Visual Diagnosis

"No-No" Head Tremor—A Nod to the Diagnosis

Ciara Terry, MB, BCh, BAO [a, *], Eva B. Forman, MRCPCH [a], Kathleen M. Gorman, MD [a], Mary D. King, MD [a, b]

[a] Department of Neurology and Neurophysiology, Temple Street Children's University Hospital, Dublin, Ireland
[b] Academic Centre on Rare Diseases, School of Medicine and Medical Science, University College Dublin, Ireland

ARTICLE INFO

Article history:
Received 11 January 2019
Accepted 7 March 2019
Available online 19 March 2019

Keywords:
Head tremor
Vitamin E deficiency
Friedreich ataxia
Cystic fibrosis
Abetaliproteinemia

This 16-year-old girl, the third child of nonconsanguineous parents, presented with a four-year history of head tremor, aggravated by stress and anxiety. She denied additional symptoms, although she was always described as clumsy. There was no relevant family history. Examination revealed a side-to-side "no-no" head tremor (see Video), hypermobile joints, areflexia, ataxia on tandem gait, absent vibration sensation below the knees, and a positive Romberg sign. Speech, eye movements, optic fundi, and tendon reflexes were normal. Brain magnetic resonance imaging, echocardiogram, and motor and sensory nerve conduction studies were normal. Plasma vitamin E level was 3.2 μmol/L (normal range: 9.8 to 30 μmol/L). Further evaluation including fecal elastase, celiac screen, and lipoproteins was normal. Treatment with high-dose oral vitamin E (17 mg/kg/day) was commenced. Plasma vitamin E levels are now normal following oral supplementation, and there is an improvement of symptoms, although they are not completely resolved.

Genetic studies revealed a homozygous missense mutation (c.458G>T, p.Gly153Val) of alpha-tocopherol transfer protein (*TTPA*), consistent with a diagnosis of autosomal recessive ataxia with vitamin E deficiency (AVED). *TTPA* encodes for a protein involved in the incorporation of alpha-tocopherol in very-low-density lipoproteins leading to low plasma vitamin E levels.

The differential diagnosis of vitamin E deficiency includes acquired deficiency secondary to malabsorption due to pancreatic enzyme deficiency in cystic fibrosis or to abetalipoproteinemia. AVED shares many clinical features with Friedreich ataxia, notably age of onset before age 20 years; cerebellar, pyramidal, and sensory signs; absent tendon reflexes and pes cavus. However, side-to-side head tremor, reported as a presenting feature in up to 40% of patients with AVED,[1] is rare in Friedreich ataxia, and this finding should alert clinicians to the possibility of this potentially treatable disorder. Lifelong high-dose oral vitamin E supplementation may arrest disease progression and reverse symptoms if commenced early in disease course.[2]

Supplementary data

Supplementary data to this article can be found online at https://doi.org/10.1016/j.pediatrneurol.2019.03.007

References

1. El Euch-Fayache G, Bouhlal Y, Amouri R, Feki M, Hentati F. Molecular, clinical and peripheral neuropathy study of Tunisian patients with ataxia with vitamin E deficiency. Brain. 2014 Feb;137:402–410.
2. Schuelke M, Mayatepek E, Inter M, et al. Treatment of ataxia in isolated vitamin E deficiency caused by alpha-tocopherol transfer protein deficiency. J Pediatr. 1999;134:240–244.

Financial disclosure: The authors have indicated that they have no financial relationships relevant to this article to disclose.
Potential conflict of interest: The authors have indicated that they have no potential conflicts of interest to disclose.
Funding source: No external funding.
* Communications should be addressed to: Dr. Terry; Department of Neurology and Neurophysiology; Temple Street Children's University Hospital; Temple Street; Dublin 1, Ireland.
E-mail address: Ciara.terry@ucdconnect.ie (C. Terry).

https://doi.org/10.1016/j.pediatrneurol.2019.03.007