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Tremor and Other Hyperkinetic Movements

Video Abstracts

Axial Sensory Tricks in Chorea–Acanthocytosis: Insights into Phenomenology

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Abstract

Background: Trunk flexion and axial extension are characteristic symptoms of chorea–acanthocytosis (ChAc).

Phenomenology Shown: A 41-year-old male with ChAc (confirmed by *VPS13A* mutations) reported that his involuntary axial movements were significantly ameliorated by either folding his arms over his chest or putting his hands behind his head.

Educational Value: These apparent "sensory tricks" suggest a dystonic pathophysiology, and also merit further study to analyze their potential for symptom control in ChAc.

Keywords: Chorea–acanthocytosis, trunk flexion, axial extension

Citation: Bhidayasiri R, Jitkritsadakul O, Walker RH. Axial sensory tricks in chorea-acanthocytosis: insights into phenomenology. Tremor Other Hyperkinet Mov. 2017; 7. doi: 10.7916/D8PV6RWW

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Editor: Elan D. Louis, Yale University, USA

Received: April 26, 2017 **Accepted:** May 22, 2017 **Published:** June 29, 2017

Funding: This study was supported by the International Research Network grant from the Thailand Research Fund (IRN59W0005), and the Chulalongkorn Academic Advancement Fund into its 2nd Century Project of Chulalongkorn University, Bangkok, Thailand.

Financial Disclosures: Dr. Bhidayasiri serves as an editorial board member of *Parkinsonism and Related Disorders* and the *Journal of the Neurological Sciences*, and the scientific advisory board for Britannia pharmaceuticals, receives royalties from Wiley and Humana Press.

Conflicts of Interest: The authors have no conflict of interest.

Ethics Statement: All patients that appear on video have provided written informed consent; authorization for the videotaping and for publication of the videotape was provided.

Various types of axial movements are described in chorea–acanthocytosis (ChAc), including tongue protrusion, feeding dystonia, head drops, trunk flexion, and axial extension, which significantly affect the patient's daily activities.[1,2] As these axial hyperkinesias are generally unresponsive to oral medications, some patients may develop their own coping strategies, including our patient, who found a novel method to improve trunk flexion and axial extension.

This 41-year-old male with ChAc (confirmed by identification of *VPS13A* mutations; disease duration of 5 years) suffers from alternating trunk flexion and axial extension while sitting upright that are so severe that he is unable to perform daily activities while sitting. In order to eat, he has to lie at 45 degrees and be fed by caregivers. He also suffers tongue protrusion dystonia, postural sway when he attempts to walk, and has a mild degree of parkinsonism. He discovered that both his trunk flexion and his axial extension considerably reduced when he folded his arms over his chest (Video segment 1) or put both hands behind his neck (Video segment 2). Although the improvement only lasts between 10 and 30 seconds, he can alternate between the tricks to continue to ameliorate his symptoms. This strategy enables him to sit upright while eating, although caregivers still feed him.

The precise phenomenology of alternating trunk flexion and axial extension is debated.[1] When these movements are analyzed by serial photographs taken every 5 seconds, they begin as a sudden loss of axial muscle tone (cervical and trunk muscles), leading to a large-amplitude sway of low frequency in either the mediolateral or the anteroposterior direction (Supplementary figures 1 and 2). These movements are variously described as myoclonic-like, ballistic, tic, choreic, or dystonic, and are reported as a debilitating feature of advanced disease, leading to injuries to the back of the head and forehead, or can even be misinterpreted as self-harm.[1]

In our case, these movements were associated with numerous falls from chairs, restricting our patient's ability to perform daily tasks in an upright position. The beneficial nature of his apparent "sensory tricks" suggests an underlying dystonic pathophysiology.

Video 1. Segment 1. Axial sensory tricks in Chorea-Acanthocytosis. The patient was asked to sit and relax while he exhibited trunk flexion and axial extension movements. When asked to fold his arms over his chest, these movements subsided although neck flexion movements were still observed. He claimed that he utilized this method when he needed to sit still. When his arms dropped to his sides, the trunk flexion and axial extension movements returned. **Segment 2.** The patient exhibited trunk flexion and axial extension movements together with continuous movements of his neck. When he pressed both his hands on the sides of his neck, all these movements subsided, but recurred as soon as his hands were returned to his sides.

This suggestion is also supported by a previous report of a trick observed in ChAc patients that reduces feeding and orolingual dystonia.[2] Indeed, the benefits of sensory trick-like maneuvers have been reported in patients with idiopathic jaw-opening dystonia where the application of a small stick between cheek and teeth or biting on a stick significantly lessen dystonic jaw activities as well as clinical severity.[3] Although the debate on the phenomenology of alternating trunk flexion and axial extension will continue, we believe that axial sensory tricks should be further developed into a device that provides a rehabilitation program for axial symptoms in ChAc.

Supplementary Material

All supplementary figures referenced in this article are available here: https://doi.org/10.7916/D8WH3118.

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