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Tremor and Other Hyperkinetic Movements

Video Abstracts

Under-Recognition of Cervical Dystonia: An Essential Tremor Patient with Numerous Textbook Features of Cervical Dystonia

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Abstract

Background: Physical examination findings of dystonia are often underrecognized, especially in the setting of other movement disorders such as essential tremor (ET).

Phenomenology Shown: A patient with ET exhibited numerous textbook features of cervical dystonia, which were misattributed to ET by a primary care physician and two neurologists.

Educational Value: To provide a clear and unmistakable visual example of the clinically significant characteristics of cervical dystonia in the setting of concomitant ET.

Keywords: Essential tremor, cervical dystonia, clinical, torticollis, neurological examination

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Dystonia, a movement disorder characterized by excessive muscle contractions that can be sustained or intermittent, is often misdiagnosed by physicians as essential tremor (ET), particularly when tremor is present. In one study, 8% of patients with an intake diagnosis of "ET" were found to have isolated dystonia rather than ET.[1] Here, we present an educational video of a 77-year-old patient enrolled in an ET study at Yale University. Our aim is to aid physicians in the recognition of cervical dystonia. The intention of the ET study was to enroll ET patients who had no additional neurological diagnoses such as dystonia. As part of our patient's screening questionnaire, she reported having been diagnosed with ET and denied ever being diagnosed with dystonia. She had developed upper limb tremor as a young adult, which had gradually worsened. In addition, she noted onset of head tremor in her mid-60s. She had an extensive family history of ET, with multiple affected first- and second-degree relatives. There was no family history of dystonia. Over the years, she had been under the care of a primary care physician and two neurologists who had attributed her upper limb and head tremors to ET. On our neurological examination, there was moderate postural tremor, moderate-to-severe kinetic tremor, and an occasional intention tremor of her upper extremities (Videos 1–5), confirming her ET diagnosis. In addition, she had each of the following significant features of cervical dystonia: (1) head tremor that was somewhat irregular (Videos 5 and 6), (2) head tremor that had a directional quality (Videos 5 and 6), (3) neck turning

Video 1. **On Arm Extension, There Is Moderate Tremor on the Right That Involves Wrist Flexion–Extension Movements.** Tremor on the left is far milder during arm extension.

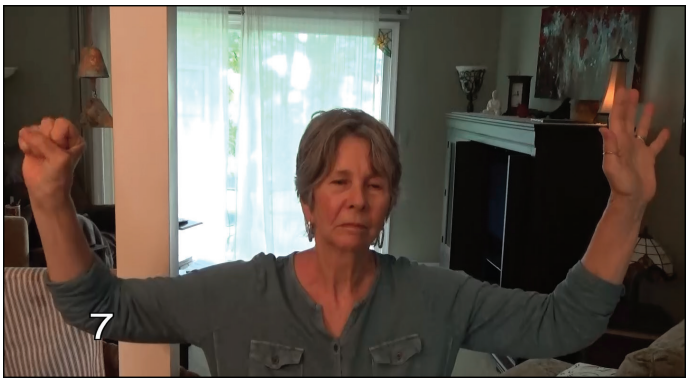
Video 2. **During the Wing-Beat Position, Some Dystonic Flexion and Abduction of the Right Pinky Emerges.** The postural tremor is also apparent on the right more than the left.

Video 3. **The Patient Pouring.** There is moderate kinetic tremor on the right and mild kinetic tremor on the left.

Video 4. **The Patient Using a Spoon.** There is moderate to severe tremor on the right with spilling.

Video 5. **During the Finger–Nose–Finger Maneuver, There Is Mild Kinetic Tremor on the Right; an Intentional Component Is Sometimes Observed.** There is also a horizontal neck tremor that is somewhat irregular and slightly directional to the left.

Video 6. **While Performing Finger Taps, the Irregular, Directional Neck Tremor Is Observed.** The head has a tendency to turn to the left. There is mild to moderate hypertrophy of the right sternocleidomastoid muscle.



Video **7. The Patient with Both Hands in the Air.** While opening and closing the fist, the right thumb is slightly flexed in a dystonic posture.

Video **9. The Patient Standing.** She also notes the presence of neck discomfort.

Video **8. Close-up Shot of Patient's Head while Sitting.** The patient notes that she always turns her head to the left and that turning to the left lessens her head tremor.

Video **10. Patient Lying on Back.** While supine, the neck tremor persists rather than resolving.

(left laterocollis) (Video 6), which was even acknowledged by the patient (Videos 8 and 9), (4) a null point – lessening of the head tremor when she turned her head to the left (Videos 8 and 9), (5) hypertrophy of the right sternocleidomastoid muscle (Video 6), (6) cervical discomfort (Videos 8 and 9), and (7) the persistence of neck tremor while supine (Video 10).[2] There were also a few signs of subtle limb dystonia (Videos 2 and 7). Our patient displayed long-standing, typical, familial ET, with co-occurring dystonia on examination. The concomitant presence of dystonic movements/postures in some ET patients is increasingly being recognized,[3] although there is debate as to when the presence of dystonia on examination necessitates an additional diagnosis (i.e., a second diagnosis of dystonia). Regardless, physical examination findings of dystonia are often underrecognized or are attributed to ET, which can lead to errors in clinical research and treatment. Recognition of these physical examination findings, along with the use of other methods to identify dystonia (e.g., electromyography to test for the presence of abnormal patterns of neck muscle activation), may reduce such errors.

References

1. Jain S, Lo SE, Louis ED. Common misdiagnosis of a common neurological disorder: how are we misdiagnosing essential tremor? *Arch Neurol* 2006;63:1100–1104. doi: 10.1001/archneur.63.8.1100

2. Louis ED. Essential tremor: a nuanced approach to the clinical features. *Pract Neurol* 2019;19(5):389–398. doi: 10.1136/practneurol-2018-002183

3. Bhatia KP, Bain P, Bajaj N, Elble RJ, Hallett M, Louis ED, et al. Consensus statement on the classification of tremors from the task force on tremor of the International Parkinson and Movement Disorder Society. *Mov Disord* 2018;33:75–87. doi: 10.1002/mds.27121