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Tremor and Other Hyperkinetic Movements

Video Abstracts

Pseudodystonic Posture Secondary to Klippel–Feil Syndrome and Diastematomyelia

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Abstract

Background: Dystonic postures possess a great number of differential diagnoses.

Phenomenology Shown: We describe a pseudodystonic posture in a 61-year-old woman with skeletal and extra-skeletal abnormalities.

Educational Value: Klippel–Feil syndrome represents an unusual cause of pseudodystonic posture to be considered in the differential diagnosis of dystonia.

Keywords: Pseudodystonia, Klipel-Feil Syndrome, diastematomyelia

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A 61-year-old female born of full-term normal delivery after a non-consanguineous marriage, reported congenital short neck with abnormal posture, unilateral sensorineural hearing loss, and migraine. She sought consultation at our center for chronic headaches and reported that when she was a teenager she was misdiagnosed with cervical dystonia. Clinical examination showed head tilt, limitation of movement at the cervical level, and low posterior hair implantation (Video 1). Spinal magnetic resonance imaging revealed vertebral fusion between the medulla and C4 as well as partial splitting between C2 and C3.

Klippel–Feil syndrome (KFS) is a rare congenital disorder (1 in 50,000 people), characterized by a defect in the formation or segmentation of the cervical vertebrae with short neck, low posterior hairline, and limited neck movement.[1] Extra-skeletal manifestations and spinal abnormalities including posterior fossa cysts or diastematomyelia may occur in KFS.[2]

In this case, KFS represents an unusual mimicking dystonia or pseudodystonic syndrome where sustained muscle contractions, unusual twisting movements, and/or abnormal postures occur secondary to underlying abnormalities.

Video 1. Clinical Examination. Head tilt, posture-mimicking laterocollis (pseudodystonia) with low posterior hairline in Klippel–Feil syndrome.

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