Atypical Symptom of Ehlers-Danlos Syndrome Impeding Diagnosis: Feeling of Spinal Instability

ROBERT GASIK and TADEUSZ STYCZYNSKI

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To the Editor:

Ehlers-Danlos syndrome (EDS) has a genetic background and is caused by defects in collagen biosynthesis. The main features of EDS are hyperelasticity, fragility of skin, hyperlaxity of the joints, and bleeding diathesis. Several types of EDS are recognized, depending on the specific gene mutations and presented phenotypes. In type VI the primary symptom is kyphoscoliosis.

We describe a patient with EDS with underreported disease related findings that impeded diagnosis: a feeling of spinal instability and balance control disturbances.

A 20-year-old man was admitted to the clinic with spinal instability (Figure 1) and balance control disturbances. The symptoms made normal walking impossible and compromised other daily activities. Because of his symptoms he had been hospitalized several times on pediatric wards with a diagnosis of kyphoscoliosis and Scheuerman syndrome. During the hospitalizations he received psychological care, as no organic causes of the symptoms could be identified.

Physical examination revealed no symptoms of nervous system or locomotor system disturbance. Manipulation, by a patient with EDS, of the costal arch, exceeded the physiologic range of movement and changing the shape of the chest; this is possible due to hypermobility of the costal cartilage connecting ribs 8, 9, and 10 with the sternum, and sternocostal and costotransverse joints.

Balance disturbances reported by the patient resulted from gravity-induced changes in spinal curvature caused by low muscle tone and hypermobility in spinal joints. Insufficient muscular stabilization of the spine was indirectly responsible for development of hyperkyphosis. This caused hypermobility of the gravity center, interfering with patient’s posture disturbances. The described etiology of posture disturbances is supported by the evolution of the disease. Loading to the spine induced by position change from lying to standing or by standing revealed insufficiency of spinal structures responsible for an upright position. In several dozen seconds the position evolved into thoracic hyperkyphosis, which forced the patient to seek support to avoid a fall. A psychogenic etiology of the described disorders can be excluded based on his repeated cycles of changes in spine curvatures during standing, visibly wide range of thoracic spine curvature during standing and sitting, attempts by the patient to maintain correct posture, and psychological examination. Neurological examination helped to exclude another etiology of posture disturbance, e.g., Parkinson’s disease or dystonic disorder.

Apart from the described case, the biomechanics of poor balance control is often seen in various other motor organ pathologies, e.g., in older people in whom thoracic hyperkyphosis and compromised muscular stabilization increase the risk of falls.

ROBERT GASIK, MD, PHD; TADEUSZ STYCZYNSKI, MD, PHD, Spondylo-Neurosurgery and Neurology Department; and the Pain Clinic, Eleonora Reicher’s Rheumatology Institute, Warsaw, Poland.

E-mail: robertg@ir.ids.pl

REFERENCES