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VIDEO | ENDOSCOPY

Esophageal Striae as a Possible Gastrointestinal Manifestation of Hypermobile Ehlers-Danlos Syndrome

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CASE REPORT

Ehlers-Danlos syndrome (EDS) is a heterogeneous group of connective tissue disorders that affects multiple organ systems, including the skin, joints, cardiovascular system, and gastrointestinal tract. There are 13 different subtypes of EDSs.[1] For most, specific genes have been identified.[2] However, hypermobile EDS (h-EDS) lacks a specific genetic locus, and the diagnosis is based on clinical criteria. Joint hypermobility and skin features are common. Motility-related gastrointestinal and cardiovascular symptoms can occur.[3,4]

A 27-year old woman presented with slowly progressive dysphagia. The patient described recurrent episodes of dysphagia for solids and liquids. Symptoms started with myalgias, joint aches, headaches, and brain fog. A neurologic workup, including a brain magnetic resonance imaging, a lumbar puncture, neurophysiologic testing, and tilt test, was performed, and a diagnosis of postural tachycardia syndrome and migraine was made. A rheumatologic evaluation showed joint hypermobility with a Beighton scale of 7/9. Genetic testing for EDS subtypes was negative. A clinical diagnosis of h-EDS was made. Gastrointestinal symptoms included heartburn, globus sensation, and chronic constipation.

Physical examination showed atrophic scars, joint hypermobility and slightly hyperextensible skin, stretch marks of the thigh, and piezogenic papules (Figure 1 and Video 1). Esophageal high-resolution manometry was consistent with ineffective esophageal motility as defined by the Chicago classification 4.0 (Figure 2). Endoscopy showed linear stretch marks (striae) in the esophagus (Figure 3 and Video 1), which distended and collapsed with inspiration and expiration.

This is the first report of an endoscopic finding of esophageal striae as a manifestation of h-EDS. Stretch marks are generally caused by ruptured elastic fibers and incomplete repair by local fibroblasts.[5] We postulate that repetitive stretching of the esophageal wall leads to small ruptures in collagen bundles of the mucosa and the submucosa of patients with h-EDS. Esophageal striae should raise the suspicion of EDS in patients with typical clinical features.

Figure 1. Physical examination findings suggestive of EDS: from left to right: atrophic scar, hypermobile thumb touching the forearm, and piezogenic papules. EDS, Ehlers-Danlos syndrome.

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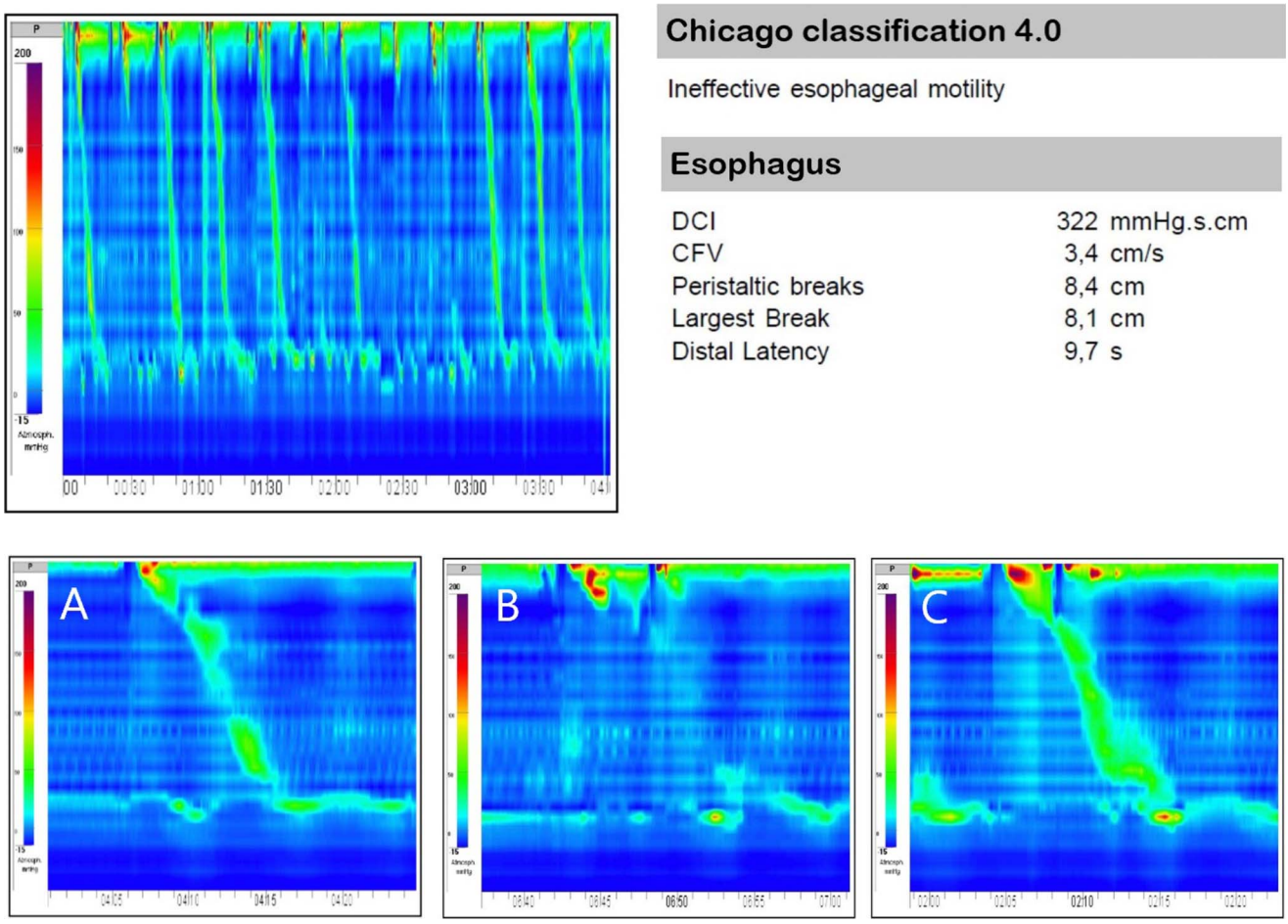

Figure 2. High-resolution manometry findings: ineffective esophageal motility as defined by the Chicago classification 4.0 with more than 70% ineffective swallows. Examples are (A) fragmented swallow, (B) failed peristalsis, and (C) weak contraction.

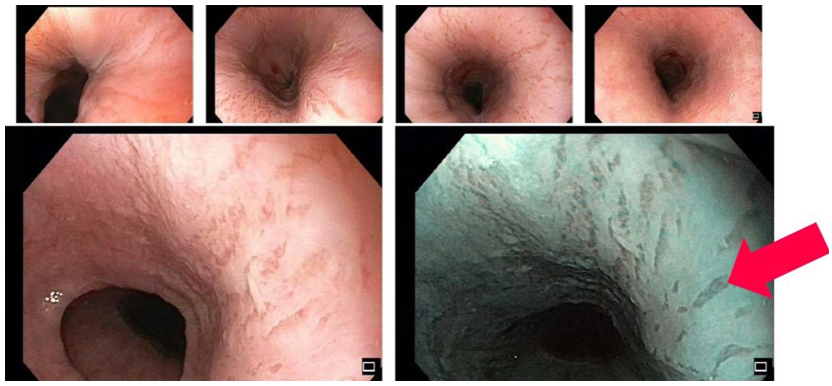

Figure 3. Endoscopic appearance of the esophagus. Esophageal stretch marks are seen, which widened with and collapsed with inspiration and expiration, respectively (arrow).

DISCLOSURES

Author contributions: All authors contributed significantly to the case, analysis, editorial input and reviewing the manuscript. The video was taken by AJ Eckardt who also drafted the manuscript. AJ Eckardt is the article guarantor.

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Previous presentation: The case was presented at the German meeting "Viszeralmedizin", Leipzig, Germany, May 14, 2022 as a video session ("Eine schwer zu schluckende Diagnose").

Informed consent was obtained for this case report.

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