Esophageal Epidermoid Metaplasia in an Obese Patient with Ehlers-Danlos Disease

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Received: November 05, 2019; Published: November 12, 2019

Keywords: Epidermoid Metaplasia; Esophagus; Ehlers-Danlos Disease; Pathology; Endoscopy

Epidermoid metaplasia in the esophagus, also known as leukoplakia, is an extremely rare disease with risk for squamous dysplasia and squamous cell carcinoma in the vicinity. A 51-year-old obese female with history of Ehlers-Danlos disease and cervical fusion of C4-6 presented with cervical vague dysphagia for more than a year, felt as a tightness, for solids and liquids. Flexible laryngoscopy was normal. Dysphagia and food impaction resolved without endoscopic intervention. Her vital signs and physical exam were normal. Her body mass index (BMI) was 38.28 kg/m². Endoscopic examination showed a large hiatal hernia in the lower third of the esophagus. At the gastro-esophageal junction, a sharply demarcated, localized moderate mucosal change characterized by white, altered texture with cross-hatching was found and biopsies were taken with a cold forceps for histology (Figure 1).

![Figure 1](image-url)

H&E stain showed squamous esophageal mucosa with reactive changes including basal layer hyperplasia and mild spongiosis but was negative for intestinal metaplasia or dysplasia (Figure 2).

Citation: Jialing Huang, et al. “Esophageal Epidermoid Metaplasia in an Obese Patient with Ehlers-Danlos Disease”. EC Gastroenterology and Digestive System 6.12 (2019): 01-03.
Focal leukoplakia (epidermoid metaplasia) was identified with abrupt transition from adjacent reactive squamous epithelium. The lesional squamous epithelium had a prominent granular layer overlayed by a thick, compact cornified layer (hyperorthokeratosis) in a basket-wave pattern. The granular layer was maximally 4-cells in thickness and contained basophilic keratohyalin granules (Figure 3).

Esophageal epidermoid metaplasia with unknown etiology often involves the upper-to-middle third of the esophagus in middle-aged women. It is associated with increased risk for squamous cell dysplasia and carcinoma. The neoplastic nature of the lesional cells is supported by their clonality as evidenced by the gene alterations of esophageal squamous cell carcinoma seen in 67% of the cases. Some authors believe this condition represents a precursor status. Therefore, it warrants close follow up histopathologically.

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Contribution of Authors

Dr. Jialing Huang put all data together and wrote the manuscript. Dr. Bart Kummer performed the endoscopic examination and provided endoscopic pictures and explanations. Dr. Wenqing Cao made the pathologic interpretation.

Volume 6 Issue 12 December 2019
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