

Esophageal Bullae in Epidermolysis Bullosa

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Background

- Epidermolysis bullosa (EB) is a heterogeneous group of genetic disorders of the stratified squamous epithelium, characterized by skin blistering and scarring.
- Dystrophic EB involves genetic mutations in *COL7A1*, which encodes type VII collagen, and can result in fragility of both dermal and mucosal epithelium.
- Patients frequently develop symptoms of dysphagia.

Patient presentation

- A 59-year-old woman with dystrophic EB presented to the outpatient endoscopy center with odynophagia and dysphagia.
- On exam, the patient had multiple dermal sequelae of EB including anonychia and fragile blistering wounds (Figure 1).
- The patient reported an extensive history of difficulty swallowing solid food, but also acute limitations in oral intake of soft solids and liquids due to pain with any swallowing.

References

1. Pope et al, J Pediatr Gastroenterol Nutr 2020.
2. Anderson et al, Gastrointest Endosc 2004.



Figure 1. Skin manifestations of epidermolysis bullosa. Anonychia, epidermal bullae, blistering wounds, and generalized thin fragile skin were apparent on visual inspection.

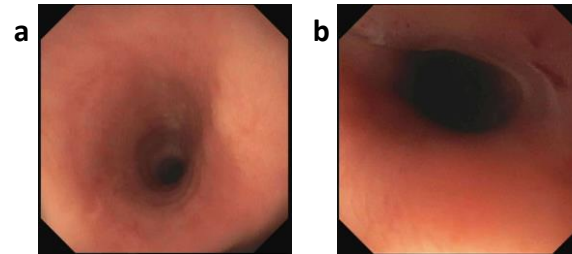


Figure 2. Upper esophageal stenosis was apparent (a) and treated with Savary dilation (b).

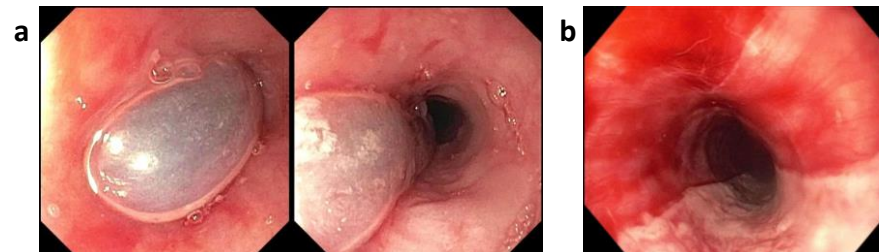


Figure 3. Mucosal bulla (a) and sloughing (b) observed in the esophagus.

Clinical course

- Upper endoscopy found an upper esophageal stricture that was treated with Savary dilation to 8 mm (Figure 2). Mucosal biopsies were normal on H&E staining. The patient had mild symptomatic improvement.
- Repeat upper endoscopy for dysphagia three weeks later showed an upper esophageal stricture (8 mm in diameter), a large mucosal bulla (Figure 3a), and denuded sloughed mucosa (Figure 3b). Dilation with passage of a standard gastroscop, resulted in moderate mucosal tear.
- After endoscopy, the patient advanced her diet to a soft diet. Persisting odynophagia symptoms were treated with viscous lidocaine, magic mouthwash and proton pump inhibition.

Discussion

- Mechanical irritation from food intake can result in cycles of esophageal bulla formation, wound healing, and scarring in patients with EB.
- Esophageal strictures are common and most frequently at the proximal esophagus in EB^{1,2}.
- Retrospective assessment has shown success of endoscopic dilation in treating symptoms of dysphagia^{1,2}.