

# [NM-310] Critical Airway in a Patient with Recessive Dystrophic Epidermolysis Bullosa Displaying Mild Cutaneous Manifestations

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## Introduction:

Recessive dystrophic epidermolysis bullosa (RDEB) is the most severe form of dystrophic epidermolysis bullosa (DEB). Anesthetizing patients with RDEB often involves difficult airway management. We present the case of a RDEB patient with minimal cutaneous manifestations but severe extracutaneous involvement and a critical airway.

## Case Report:

A female patient with RDEB presented frequently over 8 years for dilatation of esophageal strictures. Initially intubatable by direct laryngoscopy, she later required fiberoptic intubation (FOI) which became progressively more difficult due to severe pharyngeal scarring. FOI and esophageal dilatation eventually became impossible and the patient presented for lysis of pharyngeal adhesions by ENT. The patient displayed truncal skin lesions, no facial involvement, oral opening of 2 cm, and intact digits without fingernails. Stridor was noted at baseline with muffled and reduced vocal quality. During microdirect laryngoscopy epiglottic scarring to tongue and pharynx was noted. Supraglottic scars obliterated the upper esophagus and arytenoids. Adhesions were treated with balloon dilatation, sharp dissection, topical mitomycin C and steroid injection. Two years after her last surgery the pharynx permits tracheal and esophageal intubation.

## Discussion:

RDEB is a genetic disorder caused by mutations of the collagen VII gene. Faulty collagen VII production results in disorganized anchoring fibrils between the dermis and epidermis causing characteristic skin fragility and bulla formation (1). In addition to cutaneous manifestations, 90% of RDEB patients experience oral blistering with scarring and 50% have gastrointestinal involvement (2). Oral manifestations include tongue blisters and ankyloglossia. Loss of vestibles between teeth, lips and cheeks, buccal and oral cicatricial scarring results in severe limitation in oral opening. While differing levels of severity of RDEB is noted among affected siblings, it is unusual to find great disparity between cutaneous and extracutaneous manifestations in a single proband.

## Conclusions:

Airway management of patients with RDEB is temporally dynamic and complex often involving fiberoptic intubation. EB patients with mild cutaneous involvement and minor reduction of oral opening might be assumed to have a straightforward airway or be labeled as "mild EB". This case serves to demonstrate that cutaneous manifestations may demonstrate extreme disparity with airway and esophageal findings.

## References

- 1) Bruckner-Tuderman L, et al. Dystrophic epidermolysis bullosa: pathogenesis and clinical features. *Dermatol Clin* 28 (2010):107-114.
- 2) Fine JD, et al. *Epidermolysis bullosa: clinical, epidemiological, and laboratory advances and the findings of the national epidermolysis bullosa registry*. Baltimore: The Johns Hopkins University Press, 1999.

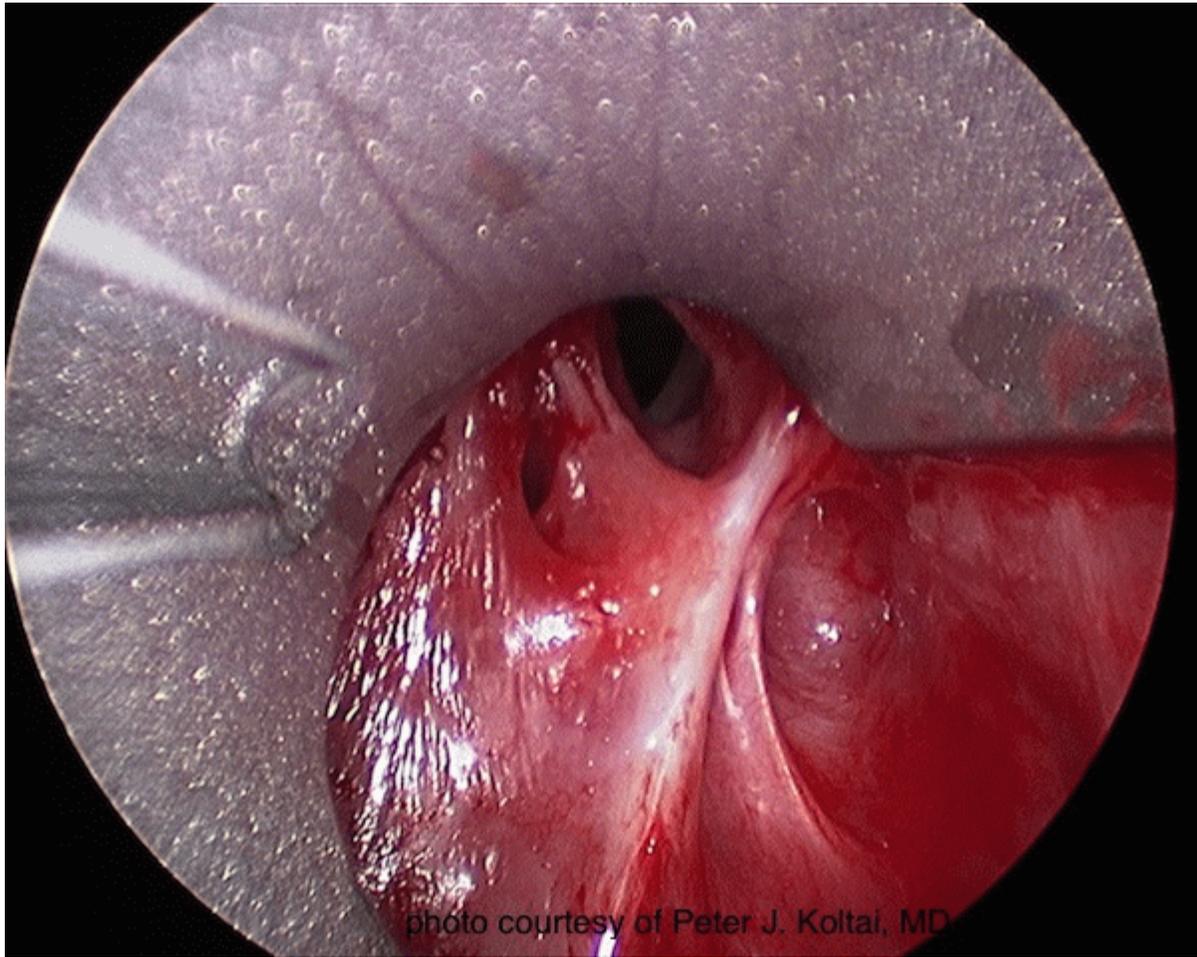


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