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# Laryngeal stenosis in epidermolysis bullosa dystrophica

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**Abstract**

Epidermolysis bullosa dystrophica is a rare hereditary skin disease of infancy in which minor trauma causes blister formation. We report a rare case of epidermolysis bullosa dystrophica (recessive) with a stenosis of the larynx due to epiglottic deformity. We performed a tracheotomy, and we detained a trachea aperture in the long term this time. In a characteristic of a disease though stimulation to the trachea causes the erosion on a trachea mucous membrane, postoperative course was uneventful and we are going to observe it in future.


*Keywords:* Epidermolysis bullosa dystrophica; Laryngeal stenosis; Tracheostomy

## 1. Introduction

Epidermolysis bullosa hereditaria is a disorder characterized by repeated occurrence of bullae, vesicles and erosion on the skin and mucous membranes induced by mechanical stimuli, leading to scar formation, atrophy, and deformities of the limbs. This clinical entity is classified according to the mode of inheritance and clinical manifestations into the following four categories: simple type, junctional type, dominant dystrophic type, and recessive dystrophic type. We encountered a male patient aged 32 years with congenital, recessive epidermolysis bullosa dystrophica suffering airway stenosis due to deformation of the epiglottis. A tracheostomy was performed, and the patient was followed up with regard to the condition of tracheal stoma and airway mucosa. The surgical intervention we performed may be associated with the risk of postoperative complications resulting from restenosis of the tracheal stoma or vesiculation of the tracheal mucosa because of the characteristics of the disease. However, our patient's postoperative course has been satisfactory and uncomplicated under airway management with a button cannula, and the patient is currently being followed on an outpatient basis.

## 2. Case report

A 32-year-old man began suffering from dyspnea around May 1, 2005, and on May 6, 2005, he was sent to the Emergency Department of this hospital due to aggravation of dyspnea. The patient had no family history of any similar disorders. He had experienced repeated blistering all over body with erosions. He had not only dental changes, nail changes, alopecia, esophageal stricture but also mitten hand deformity (Fig. 1). The diagnosis of epidermolysis bullosa dystrophica (recessive) was confirmed by skin biopsy, clinical findings and genetic screening. He had been receiving treatment for recessive epidermolysis bullosa dystrophica at the Department of Dermatology of this hospital since his birth.

Laryngofiberscopic examination revealed a marked deformation of the epiglottis, and the patient was admitted to the hospital.

On admission, the epiglottis was noted to be covered with a white coating, eroded, extremely deformed and swollen on laryngoscopy. The vocal cord was not endoscopically identifiable (Fig. 2).

A serum CRP level of 5.1 mg/dl and various other signs of inflammation were noted, and the patient was diagnosed with acute epiglottitis and was begun on intravenous corticosteroid and antibiotic drip infusion. The pulse rate and arterial oxygen

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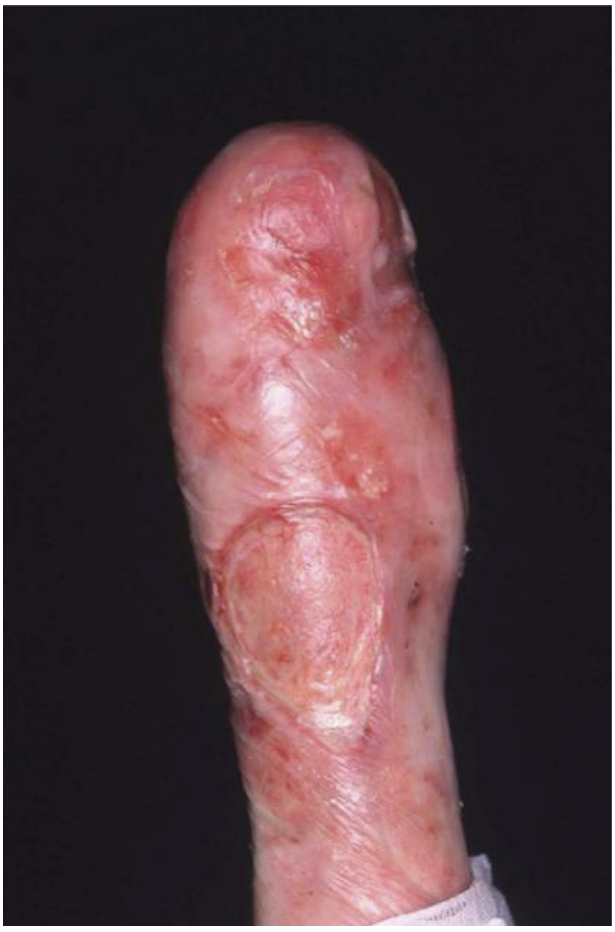

Fig. 1. This is the patient's hand. Hand deformities are as a mitten like (June 7, 2003).

saturation were monitored using an $SPO_2$ monitor (pulse-oximeter) attached to the patient during hospitalization. The symptoms gradually improved, and the patient was started on a diet on the third hospital day. The dyspnea resolved, and the

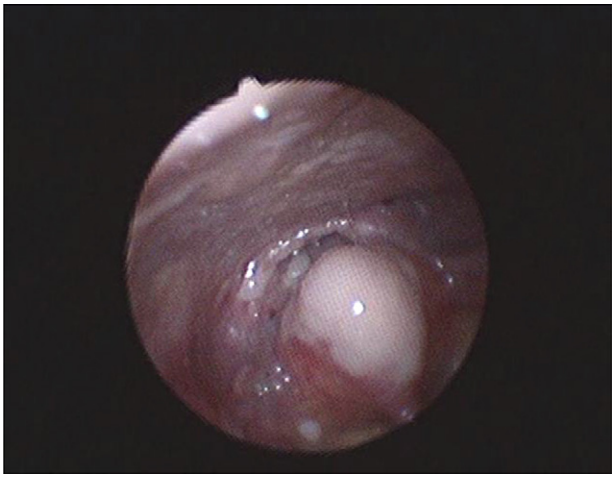

Fig. 2. Endoscopic view showing swelling and transformation of an epiglottis and white moss adhere to epiglottis. We could not confirm the vocal cord (May 6, 2005).

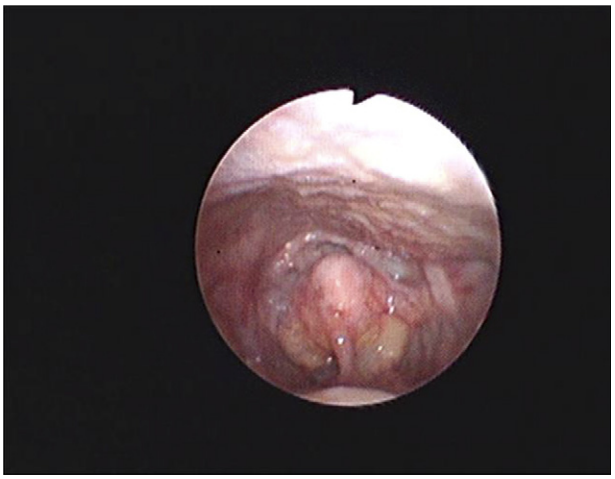

Fig. 3. Endoscopic view at the time of a discharge, white moss of an epiglottis disappeared, and the swelling was reduced (May 13, 2005).

patient was discharged on May 13. At discharge, no epiglottic white coating was noted with alleviation of the swelling (Fig. 3). The patient was followed thereafter by regular checkups of the pharynx on an outpatient basis. However, the symptom of dyspnea developed again, and the patient was admitted to the Emergency Department of this hospital on August 22, 2005. Laryngofiberscopy disclosed a severe deformation and swelling of the epiglottis, making it difficult to identify the vocal cord. At the second admission we monitored him using pulse-oximeter ($SPO_2$). $SPO_2$ was 98% but he repeated suffering from breathing symptom. Laboratory tests revealed a hemoglobin 8.5 g/dl, white blood cell count of 9500/$mm^3$, CRP of 7.5 mg/dl. Tracheostomy was considered to be indicated for this condition on account of the findings of marked epiglottic deformation with repeated dyspneic manifestations, and an inferior tracheostomy was performed on August 22. During operation, an air mattress was placed on the operating table to prevent pressure to and friction against the skin that would cause blister formation. In particular, the weight-bearing regions were protected using an

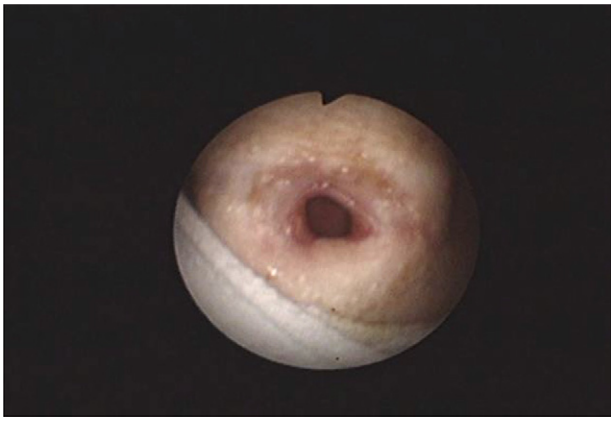

Fig. 4. The scar was no bullae, vesicles and erosion after tracheostomy (April 4, 2006).