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Case report

Epidermolysis bullosa: an exceptional cause of dysphagia

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Abstract

Epidermolysis bullosa is a genetically transmitted disorder characterized by skin blistering and scarring after minor traumatism, involves also internal organs that are lined with squamous epithelium. Esophageal stricture is a rare but often serious complication of epidermolysis bullosa causing dysphagia. We report a case of epidermolysis bullosa complicating by esophageal stricture which was successfully managed with endoscopic balloon dilatation without complications. This case illustrate an example of unusual but serious aetiology of dysphagia in children.

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Introduction

Epidermolysis bullosa is a genetically transmitted disorder characterized by skin blistering and scarring after minor traumatism, involves also internal organs that are lined with squamous epithelium such as oesophagus, bronchus and anus. Esophageal stricture is a rare but often serious complication of epidermolysis bullosa causing dysphagia. We report a case of epidermolysis bullosa complicating by esophageal stricture which was successfully managed with endoscopic balloon dilation.

Patient and observation

A 15-year-old child born of consanguineous marriage with a history of spontaneous blistering on friction site since early infancy, diagnosed as having recessive dystrophic epidermolysis bullosa, presented with progressive odynophagia and dysphagia. General examination revealed that the patient was poorly built and nourished with multiple erythematous skin eruption with skin blisters on the trunk upper and low limbs, there was also a marked nail dystrophy (Figure 1) with severe hand deformities and distal phalangeal atrophy (Figure 2). Except for hypoproteinemia and calcium deficiency, other laboratory analysis included (complete blood count, urea nitrogen, creatinine) showed no abnormalities. Esophagoscopy showed tight stricture of the upper oesophagus, with friable erythematous mucosa and exudates. A barium swallow test revealed two short stricture in the proximal oesophagus (Figure 3). The patient underwent 2 sessions of endoscopic balloon dilatation at intervals of 3 months, under scopic guidance a balloon catheter was placed across the stricture and carefully inflated. The interventions were performed without incidents. We advised the patient the use of oral Omeprazole and prokinetic drugs, after three months the evolution was favourable with disappearance of dysphagia and satisfactory weight gain, two years later, the patient is still asymptomatic.

Discussion

Epidermolysis bullosa is a genetically transmitted disorder characterized by skin blistering and scarring after minor traumatism, the recessive subtype (RDEB: recessive dystrophic epidermolysis bullosa) also named Hallopeau-Siemens is the most severe. It is due to the lack of expression of type VII collagen which is essential for dermal anchoring and may be complicated with oesophagus involvement with estimated risk at 20% [1,2]. Esophageal involvement was described for the first time by Stout in 1929 [3]. It results from the trauma caused by the passage of solid or hot foods, resulting in a cleavage between esophageal epithelium and lamina propria when the lesions heal with scarring segmental stenosis result [4]. Dysphagia is often the tell-tale symptom of the esophageal stricture and usually starts insidiously during the first decade of life, but cases of late-onset are possible. Dysphasia is first reversible when caused by bullae or webs, becomes permanent when the inflammatory lesions are replaced by permanent scars and strictures [1]. Esophageal stenosis may be responsible for severe dysphagia with malnutrition, esophagitis and inhalation pneumonia, isolated cases of perforation, haemorrhage and esophageal cancer complicating epidermolysis bullosa have been reported in the literature [1].

Radiographic features of esophageal lesions may be different according to stages of the disease and barium oesophagograms revealed that half of strictures occur in the proximal third of the oesophagus, 25% in the distal third and they are multiple in 25% of cases [5], prestenotic dilatation, ulceration and spasm may be seen [1]. Endoscopy is not necessary for the diagnosis, if realized it can show ulceration or stenosis but it has especially a therapeutic interest [4]. The

therapeutic management of esophageal stenosis includes: mixed diet, medical treatment, endoscopic dilatation and rarely surgical management [2]. Medical treatments reported in the literature include phenytoin, verapamil, corticosteroids and immunoglobulins [6]. Endoscopic management includes bougienage with the inconvenience of a longitudinal friction likely to traumatize the esophageal mucosa and actually. The method of choice for esophageal stricture is the endoscopic balloon dilatation, it gives satisfactory results with less complications [4,7]. Because of its morbidity, surgical treatment is limited in case of failure of the endoscopic methods.

Conclusion

Epidermolysis bullosa is an exceptional cause of dysphagia, endoscopic balloon dilation associated with omeprazole therapy was safe and effective management of the esophageal stricture in this disease.

Competing interests

The authors declare no competing interest.

Authors' contributions

All the authors have read and agreed to the final manuscript.

Figures

Figure 1: skin reddening and loss of toenails

Figure 2: deformed fingers of the hand with distal phalangeal atrophy

Figure 3: barium swallow test revealed two short stricture (black and red narrow) in the proximal oesophagus

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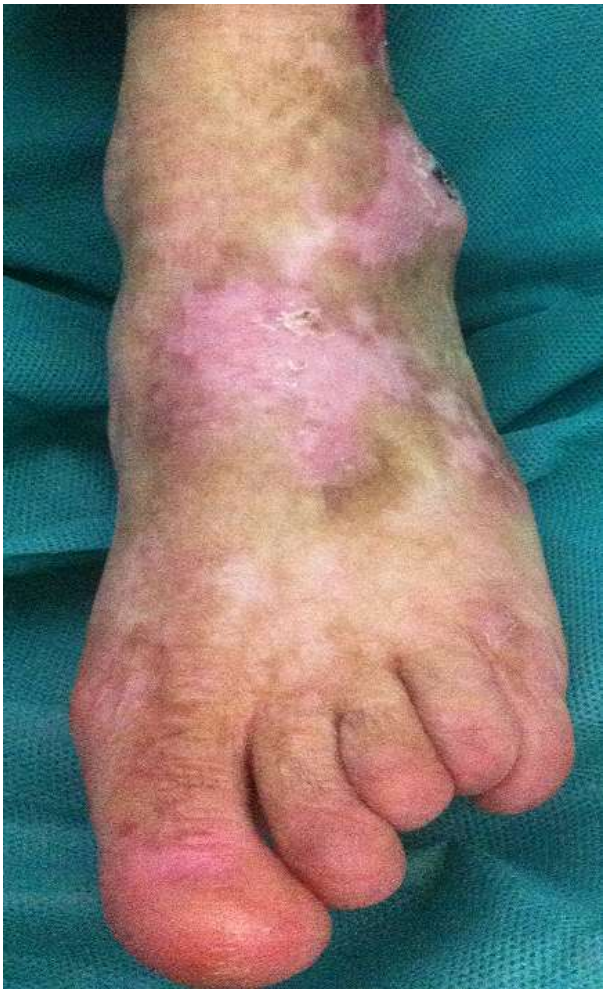

Figure 1: skin reddening and loss of toenails

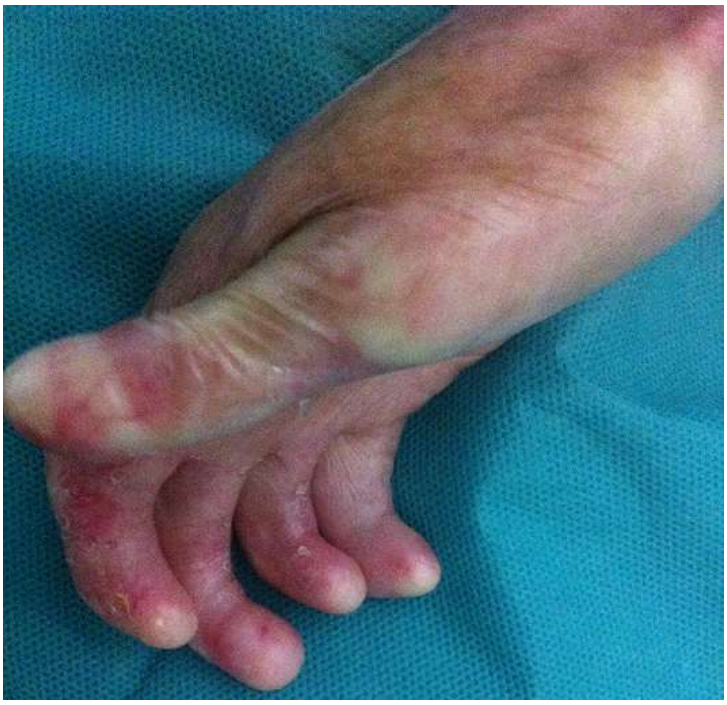

Figure 2: deformed fingers of the hand with distal phalangeal atrophy

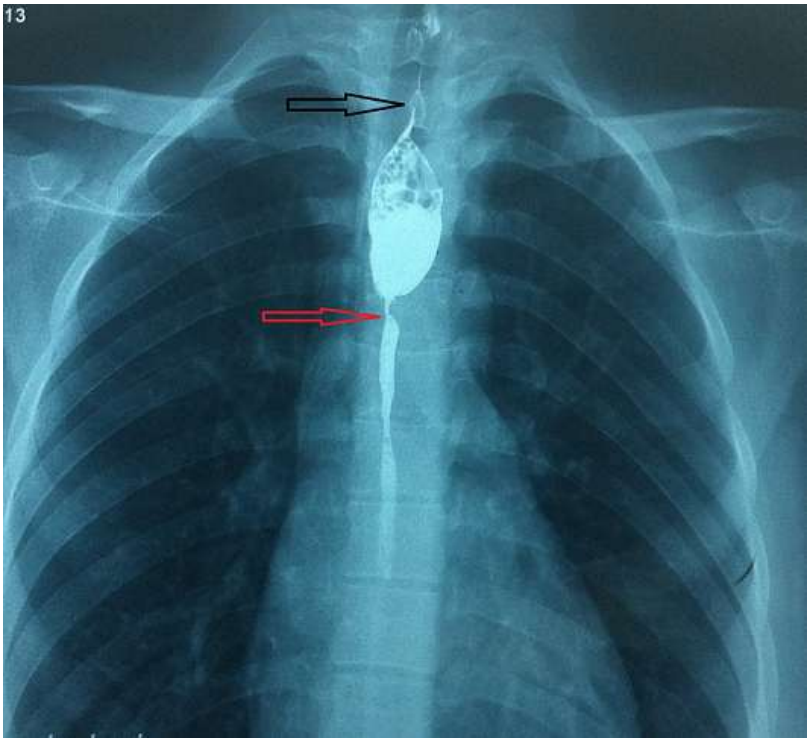

Figure 3: barium swallow test revealed two short stricture (black and red narrow) in the proximal oesophagus