Fluoroscopically guided endoluminal balloon dilation of oesophageal stricture due to dystrophic epidermolysis bullosa in two sisters

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Abstract

Dystrophic epidermolysis bullosa is an inherited disorder with frequent oesophageal stricture formation. There is no satisfactory medical treatment of dysphagia however ; endoluminal balloon dilation is a novel method with satisfactory results. Intrafamilial cases of dystrophic epidermolysis bullosa manifest variable clinical presentations. We report two sisters with dystrophic epidermolysis bullosa simultaneously presenting with dysphagia. Fluoroscopically guided endoscopic balloon dilation revealed almost complete resolution of dysphagia in both patients. Our cases represented a striking similarity in their clinical picture and response to treatment. Balloon dilation in these cases is a safe and effective approach. (Acta gastroenterol. belg., **2006**, 69, **327-329**).

Key words : oesophagus, stricture, balloon dilation, epidermolysis bullosa, treatment.

Introduction

Epidermolysis Bullosa (EB) is a heterogeneous group of disorders of the stratified epithelium, characterized by skin blistering, scarring and in some cases, mucosal involvement after minor mechanical trauma. EB is divided into three categories depending on the dermal layer of involvement : EB simplex (intraepidermal), junctional EB (the lamina lucida of the epidermal basement membrane) and dystrophic EB (DEB) in which the undersurface of the dermo-epidermal junction, i.e., beneath the lamina densa, is affected.

DEB is a hereditary blistering disorder, which the oesophageal mucosa is frequently involved (1). The autosomal inherited form, recessive dystrophic epidermolysis bullosa (RDEB), is responsible for oesophageal lesions consisting of web or stenosis. Blister formation leads to ulceration, wound healing, scaring with fibrosis and subsequent stricture formation. Minor traumas, iatrogenic invasive procedures may worsen clinical picture (1). Blister formation is associated with reduction or complete absence of anchoring fibrils at the epidermal-dermal junction leading to loosening of adhesion to the basement membrane (1,2). Molecular defect in all forms of DEB is a mutation in the COL7A1 gene encoding type VII collagen, a protein of the epidermal attachment complex (3).

Oesophageal involvement is the most frequent extracutaneous gastrointestinal manifestation of the disease (1). However, junctional epidermolysis bullosa, a very rare form of the disease, is associated with pyloric atresia and death in the first months of life (3). Upper oesophageal segments are usually affected and stricture formation causes dysphagia.

Patients with oesophageal disease usually become symptomatic after the first 2 decades of life. Clinical manifestations occur earliest in RDEB (4). Almost all patients with RDEB experience dysphagia in the third decade of life (5). Fifty percent of strictures occur in the proximal third of the oesophagus, usually near the cricoid cartilage and cricopharyngeal muscle, where the oesophageal lumen is narrowest, 25% in the distal third and the rest in multiple sites (6). Gingival blistering leads to scarring, ankyloglossia, microstomia, and carious teeth, all common features of severe dystrophic EB, which cause food to be poorly masticated, thus exacerbate the oesophageal problem (7).

The management of these oesophageal strictures is controversial. Although bougienage has been used (6), the tangential force applied sloughs the fragile mucosa and produces an unacceptably high risk of perforation (8). Total colonic interposition has considerable improvement of symptoms however, with high mortality and morbidity rates (9,10).

In our report, we describe two female patients, who are sisters with similar cutaneous and oesophageal involvement. Successful balloon dilation and improvement of symptoms are discussed. Another interesting feature of these cases is the familial involvement and similar natural course of the disease.

Case reports

Case 1

41-year-old female, who had been suffering from RDEB since birth, developed progressive dysphagia for the last six months. The patient has severe scarring of her skin, especially in her face and extremities due to prior blistering lesions. She could only swallow liquid foods for the last month. Upper gastrointestinal endoscopy revealed a web-like stricture at 21 cms from

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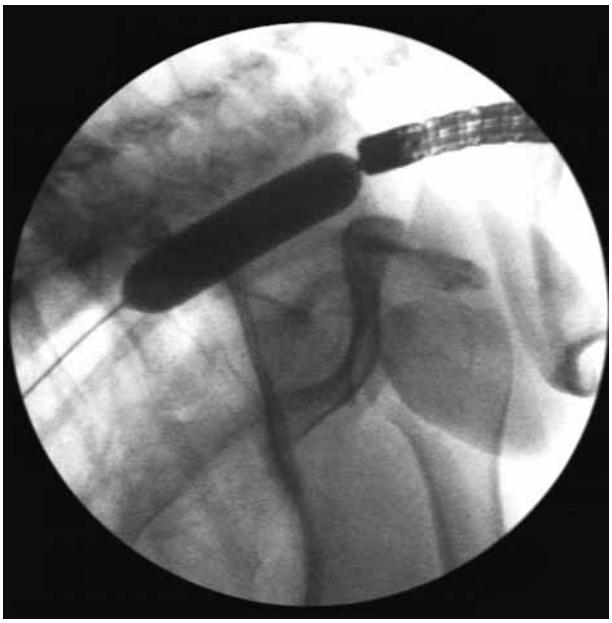

Fig. 1. — The balloon was inflated with a diluted water-soluble contrast agent up to 18 mm in diameter under endoscopic vision. Inflation was maintained for 2 minutes. The balloon was completely expanded and there was no indentation on the balloon (Fluoroscopic view).

the incisors, which could not be passed. She was hospitalized for balloon dilation to relieve dysphagia.

Premedication was done with a local anaesthetic (Xylocain, Astra, Södertalje, Sweden) applied to the oropharinx and light sedation with intravenous 5 mg midazolam (Dormicum, Roche, Switzerland). The procedure was performed endoscopically with the guidance of fluoroscopy. Under fluoroscopical control, the endoscope (Pentax IPM G300) was inserted above stricture and balloon catheter was advanced through the stricture (Boston-Scientific CRE Wire-guided balloon dilator, 15-16.5-18 mm diameter, USA). Under endoscopic vision, the balloon was inflated with a diluted water-soluble contrast agent up to 18 mm in diameter. Inflation was maintained for 2 minutes. The balloon was completely expanded and there was no indentation on the balloon (Fig. 1).

There were no complications related to the procedure. After 24 hours, an oesophagogram was performed before refeeding the patient. The control oesophagogram showed improvement of the oesophageal strictures. Then she had started liquids and a soft diet. She was discharged on the second day of the procedure with proton pump inhibitor (lansoprazol 30 mg a day) maintenance therapy and soft, blenderized diet. Dysphagia symptom resolved almost completely until now (12 months).

Case 2

38 years-old female (sister of the former patient) admitted with progressive dysphagia symptom, concurrent with her sister. Duration of RDEB was similar and the patient did not seek any help for her disorder until now. She had similar skin findings with her sister. Dysphagia lasted for 3 months of period and worsened in the last 15 days. She was unable to eat solid foods. The oesophagography showed narrowing of the oesophageal lumen, which was more prominent in the upper oesophageal segment. She was hospitalized for endoscopic balloon dilation procedure.

The same endoscopic procedure including premedication was performed. Successful oesophageal dilation was achieved with the above-mentioned protocol. There were no complications related to the procedure. The control oesophagography showed improvement of the strictures in the upper oesophagus. She was discharged with proton pump inhibitor (lansoprazol 30 mg a day) therapy and soft, blenderized diet. However, her symptoms partially resolved and another dilation procedure was required after a month. Her symptoms have improved after this treatment and she is able to eat soft diet without symptoms at the end of twelve months.

Discussion

DEB is a genodermatosis resulting from resulting from mutations in COL7A1, the gene encoding type VII collagen (1). The site and specific mutation determine the clinical phenotype, which ranges widely from a severe mutilating condition to a relatively mild disorder. Worldwide prevalence of DEB (per million) ranges from 1.2 in South Africa to 24.4 in Scotland (1). Oesophageal involvement usually manifests at paediatric ages. However, in the autosomal dominant form of DEB, adult patients predominate (4% vs. 23%) (1).

There is no currently available medical treatment for oesophageal lesions in RDEB. Dilation of oesophageal strictures with bougienage is reported to be successful in some case reports however ; it may result in exacerbations or complications such as perforation (2,11). Balloon dilation prevents longitudinal shear stress related trauma associated with bougienage.

Surgical treatments such as colonic interposition are considered as high risk procedures because of limited exposure of the airway, malnutrition, and postoperative secondary infections. For this reason, these operations should be performed in experienced centres and multidisciplinary approach is mandatory (9,10).

Percutaneous gastrostomy tube placement is an attractive option because it guarantees delivery of a definable level of nutrition (12,13). However, endoscopic balloon dilation yields comparable results, enabling maintenance of adequate nutrition with the added psychological benefit of allowing the patient to eat, albeit a specialized diet. A trial directly comparing the two methods would likely be unethical because of the possibility of irreversible occlusion of the oesophagus.

Fluoroscopical balloon dilation is reported as a successful method without any significant complications (2,14). Endoscopic dilation under fluoroscopy is a

similar method. Experienced endoscopist is able to reach the stenotic segment without further damage to the mucosa. In addition, the direct visual aid in this manner avoids unnecessary mucosal trauma during guide wire replacement.

Anderson *et al.* reported endoscopic balloon dilation of 53 EB patients with oesophageal strictures. For all but three patients, there was an improvement in the dysphagia score. There was no significant post-procedure morbidity (15).

Nutritional aspects of EB are of paramount importance. These patients should be kept on a soft and blenderized diet. Any further mechanical trauma aggravates the scar formation in the oesophagus. We also advise maintenance proton pump inhibitor therapy, in order to avoid acid-related chemical trauma.

In our cases, above-mentioned approach has proven to be successful. Mid-term results are comparable with other methods of therapy. However, the most striking aspect of our cases is the patients being sisters. Despite a recent report about the intrafamilial phenotypic variation according to skin metalloproteinase expression (16,17), our cases presented with almost identical clinical picture.

As far as we know, this is the first report of simultaneous oesophageal phenotypic expression of recessive dystrophic epidermolysis bullosa and similar treatment outcome in two sisters.

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