Esophagitis and almost complete esophageal occlusion in a girl with epidermolysis bullosa

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Epidermolysis bullosa is a genetically transmitted skin disorder that typically manifests with trauma-induced skin blistering, scarring and in some cases mucosal involvement. Esophageal webs, strictures or stenosis can be found in about a third of the patients with the recessive dystrophic type of this disease. We report a six-year-old girl with recessive dystrophic epidermolysis bullosa and progressive dysphagia. Almost complete esophageal occlusion was found on barium swallow and esophagoscopy (endoscopic signs of esophagitis were also seen). She was treated successfully with fluoroscopically guided balloon dilatation and esomeprazole. Fluoroscopically guided balloon dilatation and postdilatation use of esomeprazole is a safe and effective therapy for recessive dystrophic epidermolysis bullosa patients with almost complete esophageal occlusion and esophagitis.

Key words: epidermolysis, esophageal occlusion, esophagitis.

Epidermolysis bullosa (EB) is a heterogeneous group of genetically transmitted disorders characterized by skin blistering and scarring after minor trauma. EB is divided into three major types (simplex, junctional, dystrophic) depending on the dermal layer of blister formation[1,2] Each of the major types has several subtypes. Dystrophic epidermolysis bullosa (DEB) is caused by the collagen VII gene (COL7A1 and HS-RDEB) mutation and abnormalities of anchoring fibrils connecting the epidermis to the dermis[3,4]. Apart from skin, in patients with DEB, blistering can affect different parts of the gastrointestinal tract, such as the oropharynx, esophagus, small and large intestine, rectum, and anus. Recurrent oropharyngeal blisters can limit the range of motion in temporomandibular joints and cause shrinkage of the oral cavity and mouth (microstomia) that can seriously compromise the patient's ability to eat. Occlusive esophageal lesions (webs, stenosis and strictures) may be found in about 35% of patients with recessive DEB (RDEB) in any part of the esophagus[5]. Half of the strictures occur in the proximal third of the esophagus, 25% in the distal third, and the rest in multiple sites[6]. In patients with RDEB, the esophagus can also be affected by: prestenotic dilatation, esophagitis, shortening of the esophagus and hiatal hernia, decreased peristalsis, and esophageal atony, etc.[7-9]. Up-to-date experience has shown that the treatment of choice for esophageal strictures in children with RDEB is balloon dilatation[10,11].

Case Report

A six-year-old girl was admitted to our hospital due to progressive dysphagia. She complained of poor appetite and the sensation of "food getting stuck". She had had skin blisters since infancy when she was diagnosed as RDEB. On admission, she was noted to have signs of malnutrition and skin blisters on her lips, ears and hands. She had small oral erosions and could not fully open her mouth. Except for elevated C-reactive protein (40), another laboratory analysis (complete blood count, blood urea nitrogen [BUN], creatinine, alanine aminotransferase [ALT], aspartate aminotransferase [AST]) showed no abnormalities. Barium esophagogram revealed a 4 cm long stricture of the mid-esophagus (Fig. 1). On endoscopy, due to microstomia and the

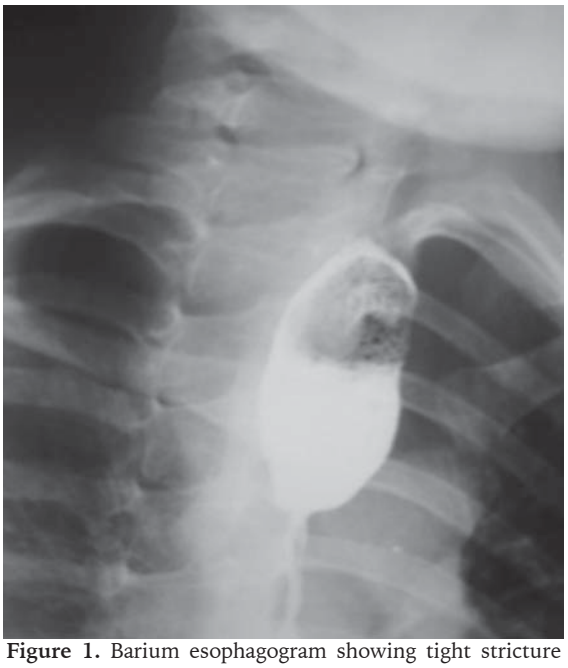

Figure 1. Barium esophagogram showing tight stricture of the mid-esophagus.

presence of oral erosions and blisters on her lips, we did not use classical endoscopic bite guard but instead gently placed two airways on both sides of her jaw, thus opening her mouth sufficiently for passing the endoscope (Olympus GIF Q180). Esophagoscopy showed almost complete esophageal occlusion and endoscopic signs of esophagitis in the form of non-confluent white exudates (Fig. 2). Three sessions of fluoroscopically guided esophageal balloon dilatation using a balloon catheter (Medi/Tech/Boston Scientific) were done. Under fluoroscopic guidance, a guidewire with soft tip was introduced through the nostrils and positioned in the stomach. A balloon catheter (6 mm) was then placed across the stricture and slowly inflated with contrast for 30 seconds. Balloon catheter size was increased by 2 mm for subsequent sessions. The interventions were performed without any complications. Both endoscopic and radiologic procedures were done in deep sedation achieved with midazolam. The patient took esomeprazole for four weeks after balloon dilatation and was symptom-free during that period. Due to the possibility of esophageal stenosis recurrence, her regular follow-up visits to our outpatient department have been continued.

Discussion

One of the most debilitating features in patients with RDEB is the development of esophageal strictures, which can produce profound dysphagia with nutritional failure, food bolus obstruction, esophagitis, and aspiration pneumonia. Different strategies have been reported for the management of esophageal strictures in RDEB, including medical therapy, fluoroscopically or endoscopically assisted balloon dilatation, and surgical esophageal reconstruction by colon interposition.

Medical therapy with corticosteroids, phenytoin, verapamil, and immunoglobulins was not proven effective in a large series of RDEB patients with esophageal stricture[12-15]. Bougienage has been used previously for treating esophageal stenosis in RDEB patients, but it is now considered to be contraindicated because tangential shearing forces cause detachment of the esophageal mucus membrane, which leads to scar formation and finally esophageal stricture. Today, the initial therapy of choice for esophageal stenosis in patients with DEB is fluoroscopically or endoscopically guided balloon dilatation. It is less harmful than bougienage because it produces longitudinal forces to the esophageal wall, and can be done safely and repeatedly since children with RDEB require repeated dilatations for recurrent strictures. Azizkhan et al.[10] performed 92 balloon dilatations under fluoroscopic guidance in 25 RDEB pediatric patients without

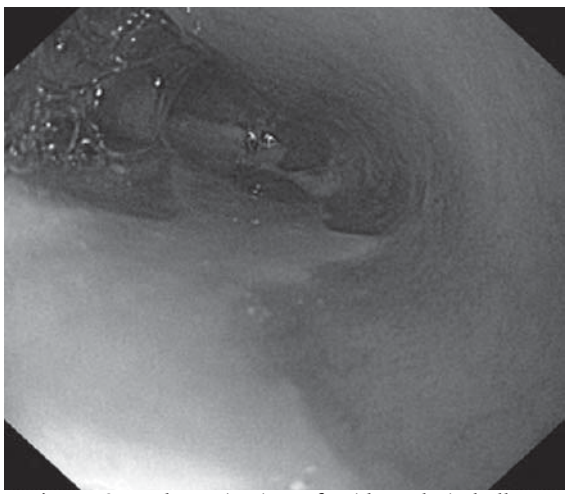

Figure 2. Endoscopic view of epidermolysis bullosa esophagitis and esophageal stricture.

any complications. Most patients reported immediate relief of symptoms, rapid recovery and resumption of adequate food intake (on average 4 procedures per patient were performed). With the advent of small caliber endoscopes, endoscopy has become much safer for RDEB children. A group from Stanford University developed an outpatient technique using fluoroscopically guided balloon dilatation and limited upper endoscopy with small caliber endoscopes[16]. With this technique, they have performed 109 dilatations in 22 children with immediate clinical improvement and negligible complications.

In both series, recurrent esophageal strictures requiring repeated dilatations were often noted. To date, no effective preventive measure against stricture recurrence has been found. Authors from San Diego recently reported two children with EB in whom topical steroid therapy (budesonide) reduced the rate of esophageal stricture recurrence[17]. Further studies with a greater number of patients would be useful to better understand the efficacy and safety of this therapeutic option. One of the preventive treatments may be post-dilatation topical application of mitomycin-n[18]. However, large experience with this therapy in patients with DEB is lacking.

Nutritional support is very important for the prevention of esophageal strictures because poor nutrition can worsen not only skin changes, but also esophageal lesions. Nutritional support can be provided by percutaneous endoscopic gastrostomy placement or partial parenteral nutrition. A group from Japan reported successful treatment with balloon dilatation and extensive nutritional support (provided by partial parenteral nutrition) in nine children with esophageal strictures complicating RDEB[19].

Surgical intervention (colonic interposition) is not the first therapy of choice for the vast majority of patients with RDEB[20]. It is reserved for patients with severe esophageal strictures that cannot be managed by esophageal dilatation.

Esophageal strictures represent one of disabilities that can be found frequently in RDEB patients. The healthcare team must be familiar with the complexity of these patients and should be experienced in diagnostic and therapeutic procedures. Fluoroscopically guided balloon dilatation and post-dilatation use of esomeprazole is a safe and effective therapy for RDEB patients with almost complete esophageal occlusion and esophagitis.

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