Recessive Dystrophic Epidermolysis Bullosa: Rare Bilateral External Auditory Canal Stenosis and Surgical Treatment

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ABSTRACT: Epidermolysis bullosa (EB) is a family of rare genetic disorders affecting the skin and mucous membranes, causing blisters and lesions. Its treatment is based on the prevention of traumatic events that could favor the onset of blisters as well as careful wound care. New therapies, including gene therapy, are under investigation. In the case described here, a rare localization of EB at the level of the ear canal is described. To our knowledge, no surgical option for treatment has previously been described in the literature. The clinical features observed and the therapeutic modalities adopted are presented and discussed. A 56-year-old female patient came to our attention for bilateral progressive hearing loss. The patient was suffering from Dystrophic EB. Surgical correction of the stenosis through a retroauricular approach was planned, with the simultaneous reconstruction of the right external auditory canal using the canaloplasty technique combined with Thiersch skin grafting. The case we report here is, to our knowledge, the second describing the surgical treatment of ear canal stenosis secondary to EB. As a result of surgical correction of the stenosis, the ear regained its physiological function and there was an improvement in hearing. In the subsequent post-operative controls, there was no recurrence of the disease, from which the patient is still free 36 months after surgery. Although conservative treatment is a solid choice, our experience seems to indicate that the surgical option allows better management of the Dystrophic EB in the external auditory canal.

KEYWORDS: Epidermolysis bullosa, ear canal, otologic surgical procedures

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Introduction

Epidermolysis bullosa (EB) is a rare genetic disorder affecting the skin and mucous membranes, causing blisters and lesions. It can be transmitted in an autosomal dominant or recessive mode and manifests itself in different forms depending on its severity and location: simple, dystrophic, junctional, or mixed. The diagnosis is based on history and objective examination of the lesions, followed by histological and genetic confirmation. Currently, treatment is based on the prevention of traumatic events, favoring the onset of blisters and careful wound care. New therapies, such as gene ones, are under investigation.

Fantauzzi et al[1] described auricular localization of this disorder as rare since they reported only 2 cases out of 60 patients.

In this paper, we describe a patient with Recessive Dystrophic EB (RDEB) who presents total bilateral stenosis of the external auditory canals. The clinical features observed and the therapeutic modalities adopted are presented and discussed.

Case Presentation

A 56-year-old female reported a bilateral progressive hearing loss for 3 years. The patient had dystrophic bullous epidermolysis linked to skin alterations and symptomatic esophageal stenosis, treated by surgical dilation in 2007. Itch-like nodules and linear lesions on the lower legs were the onset of the disease and, involving also arms, abdomen, breast, and back a second time. Recently, some lesions also appeared in the occipital region.

The diagnosis of recessive DEB (RDEB) was made elsewhere based on a skin biopsy and was confirmed using molecular analysis that revealed a homozygous mutation in the G166A area of the gene COL7A1 (collagen type VII).

Otoscopy showed total stenosis of the external auditory canal (EAC) on both the right and the left. Pure tone audiometry showed a bilateral conductive hearing loss, more severe in the right ear (Figure 1) due to the previous multiple episodes of external otitis in the acute phase of EB. CT showed no significant alterations, except for stenosis of the ear canals (Figure 2). The surgical correction of the stenosis of the right ear was planned using a retroauricular approach, with a simultaneous reconstruction of the EAC using the canaloplasty technique combined with Thiersch skin grafting (Figure 3). This approach was preferred to the endoauricolar one because it allowed a better view of the EAC and complete removal of

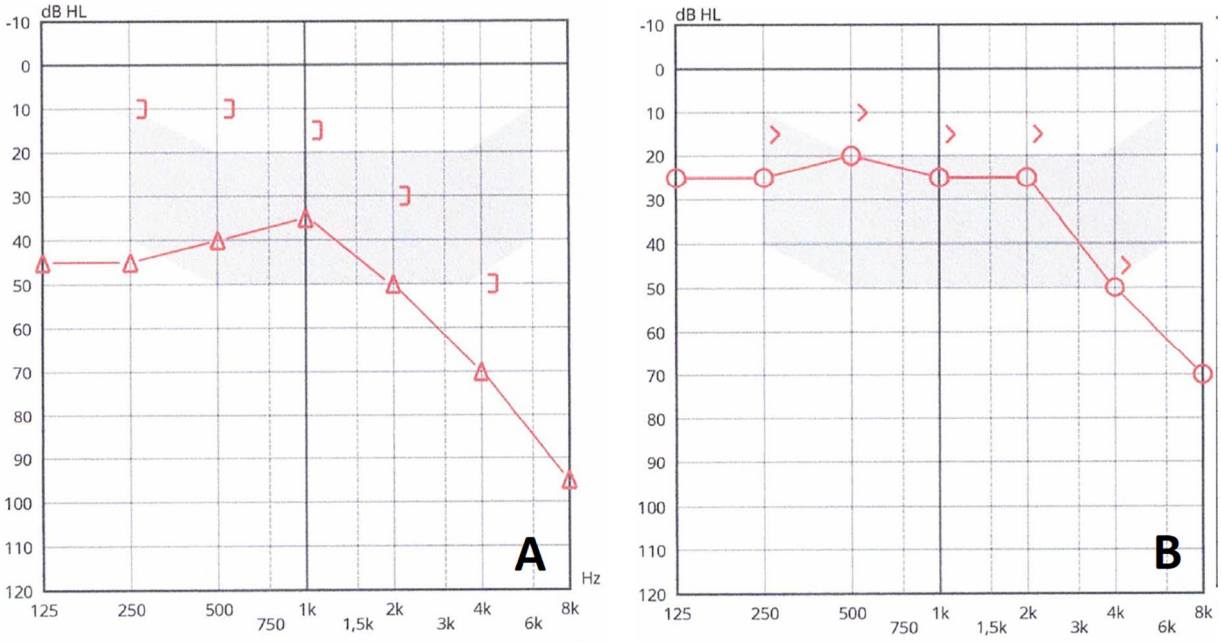

Figure 1. Pure tone audiometry. (A) Preoperative audiogram: moderate-severe conductive hearing loss. (B) Post-surgical audiogram: closure of the air-bone gap.

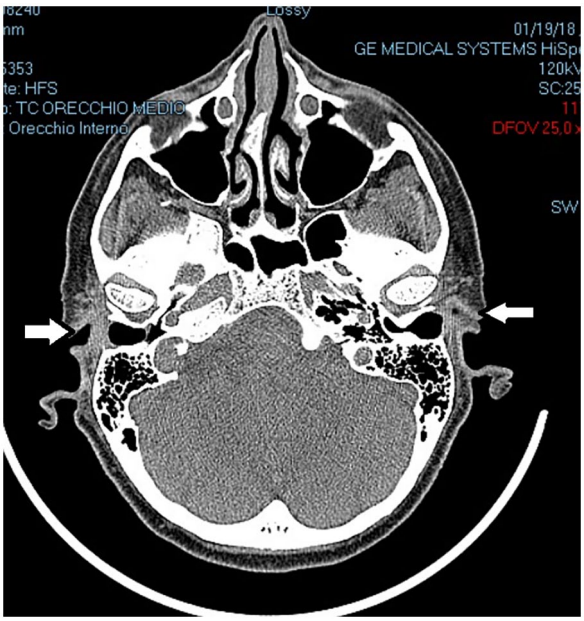

Figure 2. Preoperative Computed tomography. Arrows point at external auditory canal stenosis.

external stenotic tissue and since plastic surgery for ear canal enlargement was a preventive measure for recurrence of ear canal stenosis. Moreover, the Thiersch graft was performed on the retroauricular skin area for its low incidence of EB. A sample of one ear was excised for histological analysis, but the microscopic findings were not specific to the known disease. Indeed, the sample consisted of chronically inflamed dense connective tissue containing focally dilated glandular ducts and a small epidermoid cyst associated with a giant cell granuloma (Figure 4).

Postoperatively, she was treated for 1 month with antibiotic ear drops. A 1-year follow-up with otoscopy was performed every month and after 36 months she is disease-free.

Discussion

EB is a group of diseases that generally affected the skin of the limbs and the oral mucosa, less frequently the mucosa of the gastrointestinal and genitourinary tracts. Deficits in the skin and mucosal proteins develop vesicles and scarring, therefore, blisters have spontaneous onset or may be due to minimal trauma to the skin or mucosa.

The diagnosis is genetic on 16 distinct genes mutation, all of them codifying for proteins and influencing cellular integrity and adhesion, which have been correlated to EB. Moreover, it is advised to carry out also a clinical, electron microscopic, and immunohistological analysis.[2]

Four clinical subgroups[3] are described: EB simplex, junctional EB, Dystrophic EB, and Kindler disease.

EB simplex is an autosomal dominant expression of the disease that presents blisters on the epidermis but without any resulting scar.

Junctional EB, instead, is an autosomal recessive expression with blisters placed in the lamina lucida.

Finally, dystrophic EB is an autosomal dominant (DDEB) or recessive (RDEB) disease characterized by ungueal dystrophy, atrophy, mucosal lesions, and chromatic alterations of the skin.[4]

Kindler disease is determined by a mixed involvement of the level of skin cleavage.[3]

Clinical manifestations of DEB are usually less severe in DDEB than in RDBE but are both strongly involved in worsening patients' quality of life.[5,6]

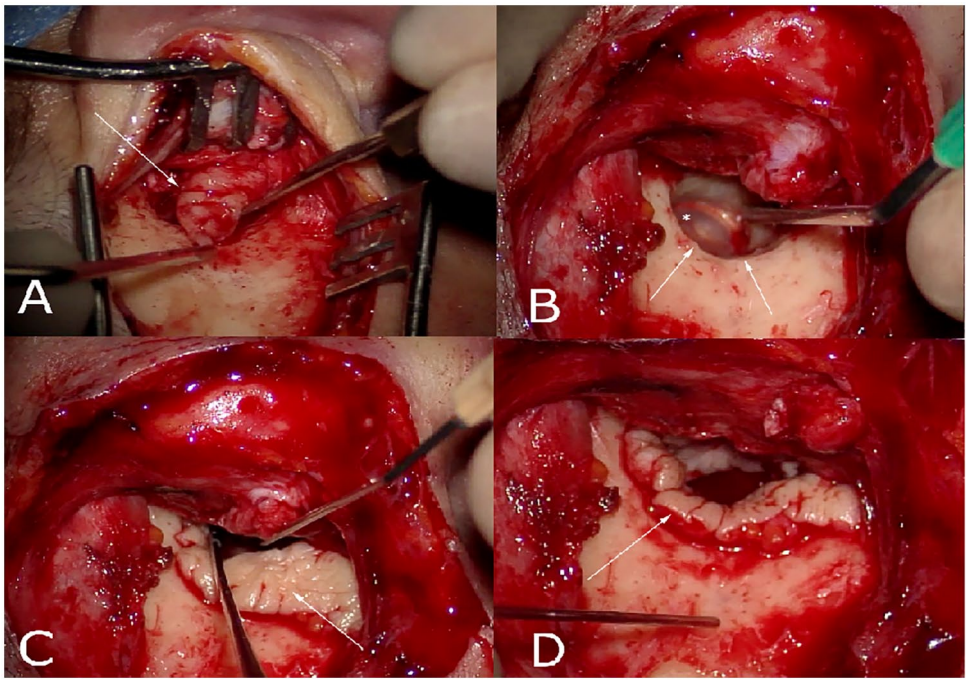

Figure 3. Intraoperative view. (A) Excision of the stenotic tissue (white arrow) from the external auditory canal. (B) Canaloplasty of the external auditory canal (white arrows) that allows a better exposition of the tympanic membrane (asterisk). (C) Positioning of the Thiersch graft (white arrow) in the external auditory canal. (D) Correctly positioned Thiersch graft (white arrow).

EB lesions can cause stenosis at the levels of the laryngeal and cervical esophagus levels[7] generally related to dysphagia. The treatment of choice in these cases is esophageal dilatation. In our case, the patient had already undergone such treatment, approximately 10 years before the presentation of the auricular location of the disease. Laryngeal or tracheal localization is possible, which usually presents with dyspnea, but the patients did not show it. Therefore, a tracheotomy was not necessaire.

However, despite the ENT district is affected primarily at the level of the oral mucosa, DDEB might as well produce auricular lesions with hearing loss.[5] An auricular location of EB has been described in the literature as a rare cutaneous manifestation affecting the pinna and occasionally the EAC. Only a few cases of ear canal involvement with hypoacusis and recurrent otitis are described in the literature, by Kastanioudakis et al[8] and Thawley et al.[9] In their case, no histological pattern was reported and a conservative approach was chosen, consisting of treatment of recurrent otitis as well as systemic infusion of phenytoin, vitamin E, minocycline, tetracycline, cyclosporine, and retinoid acid. Secondary prevention was chosen to avoid possible worsening. Surgical treatment was not considered due to the patient's pediatric age. Brown et al[10] described the case of a patient affected by RDEB with bilateral stenosis of EACs and consequent bilateral mixed hearing. One-side surgical implantation of BAHA allowed hearing recovery. The implant improved the patient's hearing, which had deteriorated due to malformation and recurrent infections linked to EB.

To our knowledge, this is the second paper describing the surgical treatment of ear canal stenosis secondary to RDEB.[9] All surgical options may increase the risk of inducing non-specific skin trauma developing skin changes and recurrence.[11] Therefore, the retroauricolar approach was preferred to the endoauricolar one because it allowed a better view of the EAC, while plastic surgery for ear canal enlargement was a preventive measure for recurrence of ear canal stenosis. Moreover, the Thiersch graft was performed on the retroauricular skin area for its low incidence of EB, decreasing the possibility of auricular recurrence. In subsequent postoperative controls, there was no recurrence of the disease, from which the patient is still free 36 months after surgery. Local therapy, as well as systemic steroid therapy, decreased the possibility of redevelopment of EB lesions. As a result of surgical correction of the stenosis, the ear regained its physiological function and there was an improvement in hearing. The closure of the air-bone gap was found after 3 months (Figure 1).

Conclusions

Due to the rarity of auricular involvement in the context of EB, identification of a method of choice for the treatment of ear canal stenosis has not yet been possible. Although conservative treatment is a solid option, our experience seems to indicate that the surgical option could represent favorable management for the patient with EB also in terms of recovery from hearing loss and improvement of the quality of life.

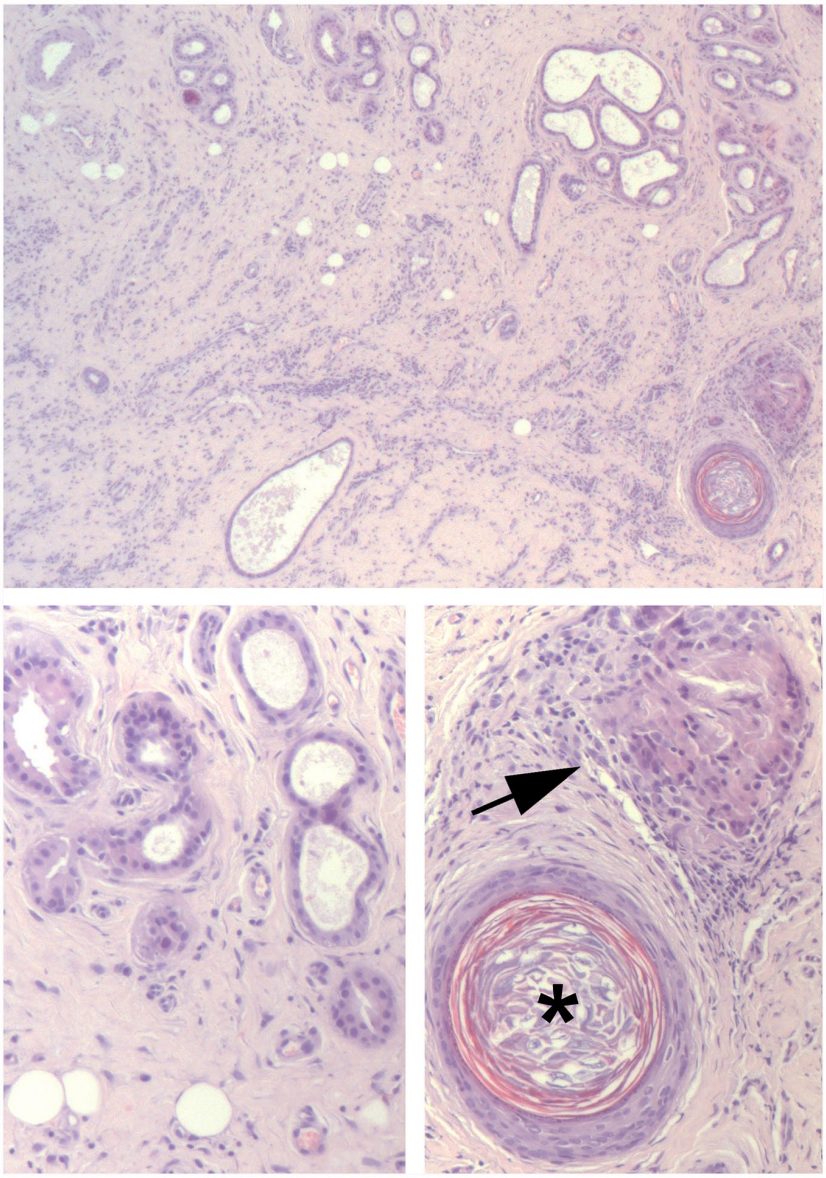

Figure 4. Low-power magnification of the excised sample is shown in the top panel. Some glandular ducts and the small epidermoid cyst (asterisk) associated with a giant cell granuloma (arrow) are shown in the left and right bottom panels, respectively.

Author Contributions

AM, AC, and GM: Made a substantial contribution to the concept or design of the work; acquisition, analysis, and interpretation of data. AM and AP: Drafted the article and revised it critically for important intellectual content. GI, SC, and AG: Approved the version to be published.

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REFERENCES

1. Fantauzzi SR, Maia OM, Cunha FC, Simões RV, Gonçalves DU, Maia AF. Otorhinolaryngological and esophageal manifestations of epidermolysis bullosa. *Braz J Otorhinolaryngol*. 2008;74:657-661.
2. Bardhan A, Bruckner-Tuderman L, Chapple ILC, et al. Epidermolysis bullosa. *Nat Rev Dis Primers*. 2020;6:78.
3. Has C, Bauer JW, Bodemer C, et al. Consensus reclassification of inherited epidermolysis bullosa and other disorders with skin fragility. *Br J Dermatol*. 2020;183:614-627.
4. Schaffer SR. Head and neck manifestations of epidermolysis bullosa. *Clin Pediatr (Phila)*. 1992;31:81-88.
5. Fulchand S, Harris N, Li S, et al. Patient-reported outcomes and quality of life in dominant dystrophic epidermolysis bullosa: a global cross-sectional survey. *Pediatr Dermatol*. 2021;38:1198-1201.
6. Eng VA, Solis DC, Gorell ES, et al. Patient-reported outcomes and quality of life in recessive dystrophic epidermolysis bullosa: a global cross-sectional survey. *J Am Acad Dermatol*. 2021;85:1161-1167.
7. Fine J, Johnson LB, Weiner M, Suchindran C. Tracheolaryngeal complications of inherited epidermolysis bullosa: cumulative experience of the national epidermolysis bullosa registry. *Laryngoscope*. 2007;117:1652-1660.
8. Kastanioudakis I, Bassioukas K, Ziavra N, Skevas A. External ear involvement in epidermolysis bullosa. *Otolaryngol Head Neck Surg*. 2000;122:618.
9. Thawley SE, Black MJ, Dudek SE, Spector GJ. External auditory canal stricture secondary to epidermolysis bullosa. *Arch Otolaryngol*. 1977;103:55-57.
10. Brown JR, Micah Milgraum D, Riyaz FR, Jahnke MN. Successful placement of a BAHA implant in a patient with epidermolysis bullosa: a case report and review of the literature. *Ann Otol Rhinol Laryngol*. 2017;126:778-780.
11. Košec A, Kostic M, Ajduk J, Ries M. Hypertrophic recurring lichen planus of the external auditory canal. *Eur Ann Otorhinolaryngol Head Neck Dis*. 2019;136:123-126.