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Ear, Nose and Throat Involvement in Epidermolysis Bullosa

Ismail Cem Temel[1], Asli Bilgic[2*] and Dedee Frances Murrell[2,3]

*[*1]Department of Head and Neck Surgery, Chris O'Brien Life House, Sydney, Australia*

[2]Department of Dermatology, St George Hospital, UNSW, Sydney, Australia

[3]Faculty of Medicine, University of New South Wales, Sydney, Australia.

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ABSTRACT

Epidermolysis Bullosa (EB) is a group of inherited blistering diseases in which patients are prone to increased mechanical fragility of skin and mucous membranes. The ear, nose and throat (ENT) area is one of the main affected mucosal sites and involvement of this area significantly increases patients' morbidity and mortality. ENT involvement of EB can result in blistering of all oropharyngeal and tracheolaryngeal structures, ulcerations, thickening and scarring of these tissues. The most common surgical procedure within the ENT in EB is tracheostomy placement. Although children with EB need to be managed by multidisciplinary teams in specialist units, some general recommendations and instructions which can be given by all practitioners are presented in this article.

Keywords: Epidermolysis bullosa, Ear, Nose, Throat, Mucosal involvement

Abbreviations: EB: Epidermolysis Bullosa; EBS: Epidermolysis Bullosa Simplex; JEB: Junctional Epidermolysis Bullosa; DEB: Dystrophic Epidermolysis Bullosa; ENT: Ear, Nose and Throat; EBDASI: Epidermolysis Bullosa Disease Activity and Scarring Index

INTRODUCTION

Epidermolysis Bullosa (EB) is a group of inherited blistering diseases. Patients in this group are prone to increased mechanical fragility of skin and mucous membranes which can result in easily repeated blisters, erosions, poor healing and/or scarring, dystrophy of nails and systemic problems due to the involvement of mucous membranes. EB is classified into 4 major subtypes according to the involved mutated protein; EB simplex (EBS), junctional EB (JEB), dystrophic EB (DEB) and Kindler Syndrome. Although some forms of EB are quite mild and just cause blistering with mechanical trauma on acral sites, others such as recessive DEB and JEB can result in scarring causing loss of normal architecture and complications in oropharyngeal, laryngeal, oesophageal-gastrointestinal and genitourinary systems [1].

DISCUSSION

Ear, nose and throat involvement

The ear, nose and throat (ENT) area is one of the main affected mucosal sites **(Figures 1-4)** and involvement of this area significantly increases patients' morbidity and mortality. ENT involvement of EB varies according to the subtype of EB [2-4]. The main involvement of oropharyngeal and laryngeal mucous membranes are shown in **Table 1**. Apart from these manifestations, all EB types can present with ear (otitis media, otitis externa, ear blockage and otalgia) and nose problems (epistaxis, nose obstruction, nasal stiffness, hyposmia).

Laryngeal problems are early as within the first year of life. Thus, it is important to monitor patients carefully for early clinical signs of airway disease activity. Furthermore, because of the cumulative risk of laryngeal stenosis or stricture especially for patients with JEB, there is a need for continued close surveillance after approximately ages 6 to 9

Corresponding author: Asli Bilgic, MD, Visiting Fellow, Department of Dermatology, St George Hospital, UNSW, Sydney, NSW, Australia, Tel: (+61) 02 9598 5800; E-mail: bilgi_asli@yahoo.com

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[2]. Mild stridor can be managed with dexamethasone and adrenalin nebulisers. When exuberant granulation tissue evolves in ENT areas, vascular laser is an effective choice to protect the airway. However, if symptoms worsen, elective tracheostomy is recommended as the most helpful approach to prevent the risk of sudden airway occlusion occurring later within a surgically uncontrolled environment [2,5].

Table 1. Various symptoms and signs of ENT involvement in patients with EB according to the subtype of EB.

Manifestation	EB simplex	JEB	DEB	
			Recessive DEB	Dominant DEB
Oral				
Oral pain and burning	+	++	++	+
Bleeding	+	++	++	+
Microstomia	-	++	++	-
Ankyloglossia	-	++	++	-
Abnormal tongue mobility	-	++	++	-
Soft tissue abnormalities	+	++++	++++	++
Dental problems	-	++++	+++	-
Enamel hypoplasia	-	++++	-	-
Pharynx				
Dysphagia	-	++	++	-
Odynophagia	-	++	++	-
Larynx				
Dysphonia	-	++	+	-
Scarring of the true and false vocal cords	-	++	+	-
False membranes	-	++	+	-
Inspiratory stridor	-	+++	+	-
Hoarse cry	-	+++	+	-
Exuberant granulation tissue	-	+++	+++	+
Submucosal cyst formation	-	+	+	-
Laryngeal stenosis and/or stricture	-	+++	+	-

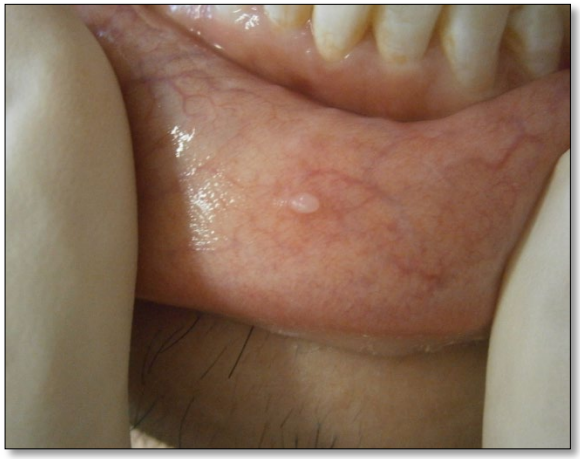

Figure 1. Labial mucosal blistering.

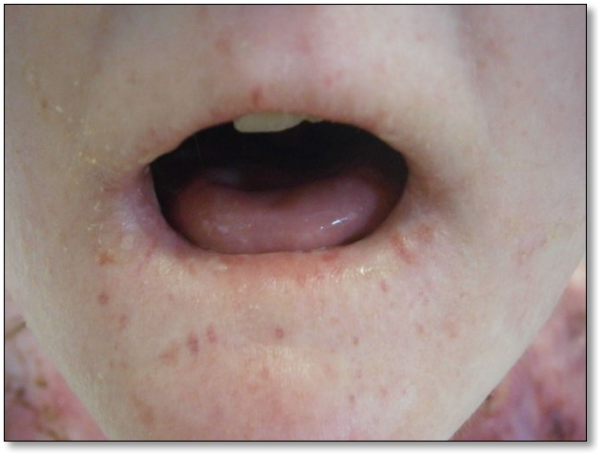

Figure 2. Microstomia and atrophy of the tongue due to the cycles of blistering and healing in and around the mouth.

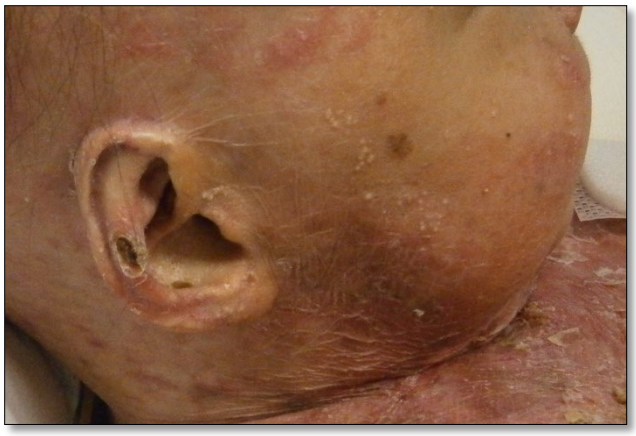

Figure 3. Residual wound and scarring due to bullae of the helix of the right ear.

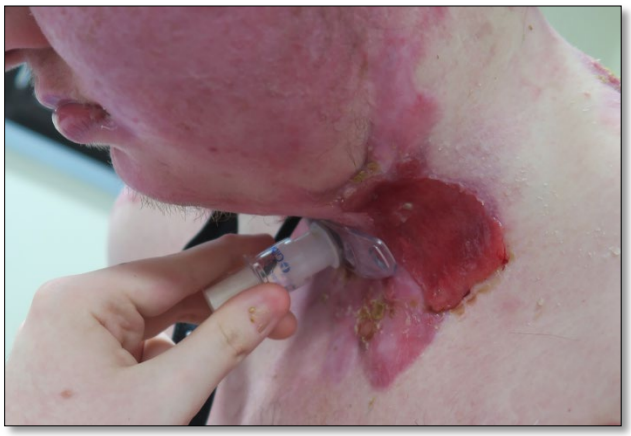

Figure 4. Tracheostomy is one of the most common ENT procedures in EB patients especially in JEB.

The most common surgical procedure within the ENT in EB is tracheostomy placement for JEB. Others include tonsillectomy, adenoidectomy and other surgical procedures within the ears or nasal cavity [2].

Moreover, patients with EB, especially more severe types (JEB) can have dental and enamel tissue problems. Soft tissues abnormalities, enamel hypoplasia, frequent dental pitting and caries are commonly seen with JEB and occasionally seen with recessive DEB [1]. Thus, all patients with EB should be evaluated by a dentist to identify any problem in need of attention.

Reliable and validated disease severity scoring systems which can consistently evaluate disease severity and demonstrate therapeutic response are increasingly important in the management of severe cutaneous diseases like EB. With this aim, Epidermolysis Bullosa Disease Activity and Scarring Index (EBDASI) was developed and validated by our group which separates disease activity from scarring [6]. It is important as damage scores may increase despite the resolution of activity when measuring treatment response, thus understating the measured benefit of medications. The EBDASI covers not only skin but also mucosal involvement in ENT. It quantifies the number and size of lesions (erosions, blisters, erythema, mucosal atrophy, fissures, stenosis), clinical findings (hoarseness) and damage of the involved areas [7,8].

MANAGEMENT

Children with EB usually need to be managed by multidisciplinary teams in specialist units. However, apart from referring patients to consultants specialized in EB; there are some general recommendations and instructions which can be given by all practitioners;

- Patients and their families should be informed about the role of traumatic physiological mechanisms to avoid the appearance of new mucosal lesions
- Gentle tooth brushing with small headed/soft-short brushes
- Soaking the tooth brush in warm water to soften
- Neutral, non-flavoured and alcohol-free formulations of toothpaste should be chosen
- Suggest consuming soft or pureed food in small amounts to prevent mucosal damage especially in severe forms
- Not to eat too much sugary food
- Mouth rinses with normal saline after meals to reduce prolonged oral clearance time
- Drainage or incision of oral bullae to avoid spreading
- Preventive dental care and frequent visits to a dental care provider are required to keep the teeth clean
- Fluoride therapy
- Basic therapeutic approaches suggested for patients with EB;
- Using sucralfate powder and/or topical lidocaine for oral mucosa lesions
- Try to eliminate factors such as including anaemia, malnutrition, infection which are affecting healing of EB lesions adversely [8,9].

CONCLUSION

ENT sites are often affected in EB. Thus, foreseeing these problems may prevent severe complications and provide better quality of life for EB patients.

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Dr. Ismail Cem Temel is a consultant ENT surgeon at Seydisehir State Hospital, Konya, Turkey. He is currently working as an observer at Head and Neck Surgery Unit, Chris O'Brien Lifehouse, Sydney, Australia.

Dr. Asli Bilgic is a consultant dermatologist and now working as a Visiting Fellow at St. George Hospital, UNSW. She is the recipient of Turkish Dermatology Society – Prof. Dr. Hulusi Behcet (Long-term Research) Scholarship in 2018.

CONFLICT OF INTEREST

None

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