

Background

Epidermolysis bullosa (EB) is a rare genetic condition resulting in extremely fragile skin that easily blisters with shearing or friction. There are three major forms of EB which all present with sloughing of the skin due to mutations in genes involving the mechanical stability and adhesion of skin. The simplex form of EB results from mutations in keratin genes and causes blistering within the epidermis. This case reviews a patient with EB simplex and bilateral cleft lip and palate. To our knowledge, there are only two documented cases of a cleft lip or palate repair being performed on a patient with EB, the most recent being reported in 2005 (Cooper et al., 2003; Ozgur et al, 2005). The objective of this report is to demonstrate how extensive pre-, peri-, and intra-operative planning can improve outcomes in patients who need additional care for EB and other coexisting medical conditions.

Patient Presentation

- Bilateral cleft lip and palate detected prenatally and confirmed at birth
- Bullae were discovered during routine pediatrics exam 24 hours after birth (Figure 1)
- Dermatologist suspected EB simplex from genetic testing results and referred patient to a national EB center, where diagnosis was confirmed

Pre-Surgical Preparation

- Nasoalveolar molding (NAM) with Craniofacial Orthodontics with limited success due to bullae formation of the intraoral mucosa

Cleft Lip Adhesion

- Bilateral cleft lip adhesion performed at 4-months of age (Figure 7)
- No surgical complications and no new bullae during admission

Cleft Lip Repair

- Definitive cleft lip repair performed at 7-months of age using the Mulliken technique
 - Tip rhinoplasty with placement of nostril retainer stents
 - Bilateral myringotomy and tympanostomy tube placement by Otolaryngology
- No surgical complications and no new bullae during admission
- Sutures were removed a week later with no complications (Figure 8)

Introduction

Our patient is a now 10-month-old female with EB simplex, who received care for bilateral cleft lip and palate. A treatment plan for cleft lip and palate repair was created through communication with members of the local craniofacial team, dermatologist, anesthesia, and a national EB center, in cooperation with the patient's parents.



Figure 1: Bullae discovered by pediatrics during exam at 24h of life



Figure 2: At 2 months of age, prior to surgical intervention



Figure 3: Before lip adhesion with sutured endotracheal tube and silicon eye tape



Figure 4: Lesions on the right foot



Figure 7: After cleft lip adhesion



Figure 8: After definitive cleft lip repair and suture removal at 10mo

Peri- and Intra-operative Precautions

- Education of all members of the nursing staff in pre-op, peri-op, PACU, and pediatrics floor prior to her arrival.
- Alcohol free skin prep prior to any adhesive application
- Vaseline gauze wrapped under cast padding prior to blood pressure cuff placement (Figure 5)
- Ear Pulse Oximetry probe
- Silicon tape for eye protection, which was provided by the parents (Figure 3)
- Securing the endotracheal tube using a circum-mandibular suture with a xeroform pledget to avoid using tape (Figure 3)
- EKG pads were trimmed to minimize adhesive contact (Figure 6)
- Use of a Bovie grounding mat instead of an adhesive grounding pad
- Silicone tape used to secure IV



Figure 5: Blood pressure cuff over Vaseline gauze and cast padding



Figure 6: Trimmed EKG pads

Conclusion

Due to extensive planning and multidisciplinary teamwork our patient has had successful cleft lip adhesion and repair with no complications and no new skin lesions upon discharge home or on post-operative follow-up. A palatoplasty is scheduled for the child and the same precautions will be taken. Treating patients with cleft lip and palate along with EB requires careful planning and communication to prevent complications. Educating all team members and following established guidelines for patients with EB while modifying traditional cleft treatment plans is the best way to maximize outcomes.

References

1. Cooper J, Seung-Jun O, Thaller SR. Case report: Cleft palate closure in 18-month-old female with epidermolysis bullosa. *The Cleft Palate-Craniofacial Journal*. 2003;40:88-90.
2. Ozgur F, Sonmez E, Tuncbilek G. Cleft lip and cleft palate closure in 13 month-old female with epidermolysis bullosa. *The Journal of Craniofacial Surgery*. 2005;16:843-847.