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Case Report

Case Report on a Mitten Hand Contracture in Epidermolysis Bullosa

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Abstract Epidermolysis Bullosa is a disorder characterized by skin fragility. Repetitive injury followed by healing can result in severe deformity due to scarring. The available evidence on whether surgery enhances hand function is somewhat inconclusive because there is a shortage of large, well-controlled studies. Nevertheless, the majority of the data suggest that surgery has the potential to enhance hand function. We present a case of Epidermolysis Bullosa in a 6-year-old patient who presented with bilateral mitten hand deformity. Right-hand contracture, pseudosyndactyly release, and defect resurfacing with a full-thickness skin graft (FTSG) were done. The patient had a smooth recovery and regained the ability to grasp objects.

Keywords: Epidermolysis bullosa, Hand deformities, Skin transplantation

Introduction

Epidermolysis Bullosa is characterized by skin fragility, which causes blisters to form even in response to minimal mechanical stress and results in a breakdown of the dermo-epidermal junction. It covers a broad spectrum of clinical presentations affecting the skin and other body systems due to the absence of essential adhesion proteins. This is caused by mutations leading to the loss of function in the genes encoding laminin 332 or type VII collagen to milder forms of skin fragility brought on by minute molecular anomalies, such as single amino acid substitutions.[1,2] The shearing force from daily activities makes the hand more susceptible to injury. Blisters can be superficial and result in erosions or more substantial, resulting in ulcerations. Contractures, pseudosyndactyly, mitten deformity, nail loss, and atrophy of the finger and thumb tips are brought on by progressive scarring.[3-5] Surgery can be used to correct hand deformity caused by Epidermolysis Bullosa because they generally result in considerable physical disabilities and decreased quality of life.[6] However, there is currently no solid data foundation on which to design surgical therapy of the hand in Epidermolysis Bullosa.[4] We describe the operation performed on a 6-year-old Epidermolysis Bullosa patient.

Case Presentation

A 6-year-old girl with Epidermolysis bullosa was referred to our team following the development of mitten hand deformity in both her hands (Figure 1). She had this deformity for 2 years and has been losing the function of her hands. She had previously been able to write and play with toys due to her intact pinch and grip functions. The disease's progressive nature resulted in severe flexion contractures, hence losing these abilities. She has not been able to seek medical attention since 2019 due to the COVID-19 pandemic, resulting in the rapid progression of the disease.

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Figure 1 Bilateral mitten hand deformity



Figure 2 Contracture release on the right hand at the level of the MCP and PIP joint with pseudosyndactyly release



Figure 3 90° flexion of PIP joint and full release of the pseudosyndactyly



Figure 4 Child regains the ability to grasp objects 3 months after surgery.

Detailed multidisciplinary discussions with the pediatric, pediatric intensivist, and dermatology team concluded that surgery could be performed to prevent further loss of function and aid in the child's developmental milestones. The surgery of choice was a contracture release on the right hand at the level of the metacarpal phalangeal joint (MCPJ) with pseudosyndactyly release. A full-thickness skin graft (FTSG) harvested from the right infraclavicular region was used to cover the MCPJ due to limited donor sites and the high risk of donor site morbidity if a split skin graft is taken.

Release was initiated, freeing the fingertips from the palm, and continued towards the metacarpophalangeal joints. Gentle and steady traction was applied without shearing forces (Figure 2). We were able to obtain a 90° flexion of the proximal interphalangeal joint (PIP) and full release of the pseudosyndactyly (Figure 3). A full release of the scar was limited by joint contracture, limited skin graft donor site, and difficult dissection due to skin fragility.

A full-thickness skin graft was applied over the MCPJ, and the other areas were dressed with modern dressings, as dermal elements were preserved throughout. Subsequent dressing changes were performed weekly under general anesthesia for a month. A custom-made padded splint was applied after the wounds were healed to maintain the hand in a functional position. The patient had a smooth recovery and regained the ability to grasp objects after the operation (Figure 4).

Discussion

Epidermolysis Bullosa is a group of rare genetic disorders. According to the National Epidermolysis Bullosa Registry, the incidence of epidermolysis bullosa is 1 in every 51,000 live births in the United States, with a frequency of 11.1 per million live births.[7]

The morphological study of a skin sample by immunohistological approaches, as well as the mutational investigation of the candidate gene, are used to diagnose and classify EB.[2] Epidermolysis Bullosa is divided into four basic types: simplex (70%), dystrophic (25%), junctional (5%), and Kindler syndrome; nonetheless, the first three subtypes account for 99% of the patient population.[2,8]

Epidermolysis bullosa negatively affects quality of life and places a financial burden on patients and their families, according to a survey by Anna et al. This includes frequent hospital visits, not working or working fewer hours, pain and itching, wound care, divorce or separation, limited daily activities, socialization, and emotional stress. Patients and carers indicated that the average amount (in USD) spent on epidermolysis bullosa care per month was $262.34 and $682.16, respectively.[6]

A comprehensive multidisciplinary team strategy that prioritizes the patient's needs is critical.[3,4,9] Our patient was referred early to the Pediatric Dermatology and Pediatric Intensive Care teams for planning. Early admission was vital to optimizing chronic normochromic normocytic anemia and electrolyte imbalances. A minimal touch guideline was established to guide the process of transfer, intubation, surgery, extubation, and post-surgical care to decrease further skin damage.

The available evidence on whether surgery enhances hand function is somewhat inconclusive because there is a shortage of large, well-controlled studies. Nevertheless, the majority of the data suggest that surgery has the potential to enhance hand function.[3] There are numerous methods described for managing and covering wounds after contracture release, but as of now, there is no consensus on the preferred approach.[10]

FTSG, split-thickness skin graft (SSG), dermal graft, skin substitutes, and non-adhesive dressings are among the approaches documented.[4-6,11,12] We performed an FTSG and contemporary dressing because full-thickness grafts are generally recommended due to superior functional outcomes and lesser risk for secondary contracture formation, and split-thickness skin grafts were not advantageous for graft take.[13,14] There was also the fear of a nonhealing donor site following a split-thickness skin graft harvest, and harvesting an FTSG allowed us to close the wound primarily. Skin substitutes were not available for our patient.

Following surgery, the hand therapist created a custom-made padded splint and performed controlled active mobilization of the metacarpophalangeal joints and interphalangeal joints after 1 month. These splints were maintained for 3 months and reduced to night splinting for a total of 1 year. Long-term use of splints has been linked to the avoidance of recurring hand contractures.[4,15]

Conclusion

Surgical correction of mitten hand contracture in epidermolysis bullosa can improve a patient's quality of life. The surgery has allowed the child to regain the ability to grasp objects, a critical factor in cognitive and physical development. However, extended follow-up is required to confirm the long-term maintenance of hand function. Additionally, a more significant number of cases and studies are needed to provide further support for the observed outcome.

Conflict of interest

The authors do not have any conflict of interest.

Consent

Written consent has been taken from the parents.

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REFERENCES

1. 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(4):614-27. doi: 10.1111/bjd.18921.
2. Has C, Fischer J. Inherited epidermolysis bullosa: New diagnostics and new clinical phenotypes. Exp Dermatol. 2019;28(10):1146-52. doi: 10.1111/exd.13668.
3. Has C, Liu L, Bolling MC, et al. Clinical practice guidelines for laboratory diagnosis of epidermolysis bullosa. Br J Dermatol. 2020;182(3):574-92. doi: 10.1111/bjd.18128.
4. Bernardis C, Box R. Surgery of the hand in recessive dystrophic epidermolysis bullosa. Dermatol Clin. 2010;28(2):335-41. doi: 10.1016/j.det.2010.01.013.

5. Terrill PJ, Mayou BJ, Pemberton J. Experience in the surgical management of the hand in dystrophic epidermolysis bullosa. Br J Plast Surg. 1992;45(6):435-42. doi: 10.1016/0007-1226(92)90207-e.
6. Bruckner AL, Losow M, Wisk J, et al. The challenges of living with and managing epidermolysis bullosa: insights from patients and caregivers. Orphanet J Rare Dis. 2020;15(1):1. doi: 10.1186/s13023-019-1279-y.
7. Fine JD. Epidemiology of Inherited Epidermolysis Bullosa Based on Incidence and Prevalence Estimates From the National Epidermolysis Bullosa Registry. JAMA Dermatol. 2016;152(11):1231-8. doi: 10.1001/jamadermatol.2016.2473.
8. DEBRA International. What is EB? DEBRA International; 2023 [cited 2023 December 3]. Available from: https://www.debra-international.org/what-is-eb.
9. Tuncer S, Sezgin B, Kaya B, et al. An algorithmic approach for the management of hand deformities in dystrophic epidermolysis bullosa. J Plast Surg Hand Surg. 2018;52(2):80-6. doi: 10.1080/2000656X.2017.1338183.
10. Anstadt EE, Turer DM, Spiess AM, et al. Surgical Management of Hand Deformity in Epidermolysis Bullosa: Initial Experience and Technique. Plast Reconstr Surg Glob Open. 2020;8(3):e2666. doi: 10.1097/GOX.0000000000002666.
11. Tian F, Li B, Tian LJ. Treatment of severe hand deformities caused by epidermolysis bullosa. Orthopedics. 2011;34(11):e780-3. doi: 10.3928/01477447-20110922-37.
12. Sibbald RG, Zuker R, Coutts P, et al. Using a dermal skin substitute in the treatment of chronic wounds secondary to recessive dystrophic epidermolysis bullosa: a case series. Ostomy Wound Manage. 2005;51(11):22-46.
13. Prasetyono TO, Sadikin PM, Saputra DK. The use of split-thickness versus full-thickness skin graft to resurface volar aspect of pediatric burned hands: A systematic review. Burns. 2015;41(5):890-906. doi: 10.1016/j.burns.2015.01.011.
14. Nakamura S, Saito S, Ishida Y, et al. Full-thickness Skin Grafts for Hand Contractures in an Adult Patient with Junctional Epidermolysis Bullosa: A Case Report. J Plast Recontr Surg. 2023;2(4):156-62. doi: 10.53045/jprs.2022-0049.
15. Formsma SA, Maathuis CB, Robinson PH, et al. Postoperative hand treatment in children with recessive dystrophic epidermolysis bullosa. J Hand Ther. 2008;21(1):80-4. doi: 10.1197/j.jht.2007.10.001.