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Surgical Management of Psudosyndactyly in Recessive Dystrophic Epidermolysis Bullosa: About Three Case Reports

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Abstract **Case Report**

Recessive dystrophic epidermolysis bullosa (RDEB) is defined as a congenital disorder resulting from mutations in the COL7A1 gene, often resulting in hand contractures and pseudosyndactyly. While multiple treatments exist to improve hand malformations, there are currently still no radical cures for this disease because of its high recurrence rate. This paper reports our experience of management of hand deformities in three cases of Recessive dystrophic epidermolysis bullosa patients with surgical management and postoperative skin dressings. Hand function was significantly improved after the complete release of pseudosyndactyly and the achievement of satisfying digital web spaces. Patients were followed up for one year and showed good results after functional hand training activities.

Keywords: Recessive dystrophic epidermolysis bullosa - pseudosyndactyly - cocoon hands.

INTRODUCTION

Epidermolysis bullosa (EB) comprises a heterogeneous group of rare genetic diseases associated with skin blistering caused by minimal trauma [1]. A major and common EB subtype, recessive dystrophic EB (RDEB), is characterized by altered wound healing, inflammatory dysbalance, and fibrotic changes associated with reduced to absent collagen VII [2]. Because of its exposed position and its continued use in daily activities, the hand is constantly at risk of microtrauma and is therefore one of the organs most affected by the disease with highly disabling deformities that represent a challenging field in hand surgery practice.

Although various gene and protein therapies have been investigated, nowadays there is no cure for EB. The treatment consists of skin care, prevention of skin trauma, adequate nutrition, and medical and surgical treatment of complications [3]. Surgical treatment of these patients is a challenge for the hand surgeon; obtaining successful results and avoiding recurrences are still major problems.

Here, we report three cases of hand deformities in RDEB patients with surgical management and postoperative skin covering. To our knowledge, the patients we were reporting represent uncommon cases from the Morrocan population reported in the literature so far.

CASE PRESENTATION 1

We report the first patient, a 9-year-old, who was referred to our department for management of pseudo syndactyly in February 2016. The history of the disease dates back to the age of one month when he was diagnosed with Recessive dystrophic epidermolysis bullosa, with no parental consanguinity reported. At the age of 8 years old, he presented erythematous-vesicular lesions on the knees, inguinal region, and both hands (Figure 1). He also developed pseudo-syndactyly of both hands, predominantly on the left hand, leading to difficulty with hand mobility.

Laboratory Findings showed hypochromic microcytic anemia (6g/dL), hypoproteinemia, elevated erythrocyte sedimentation rate (ESR), C-reactive protein (CRP), and fibrinogen.

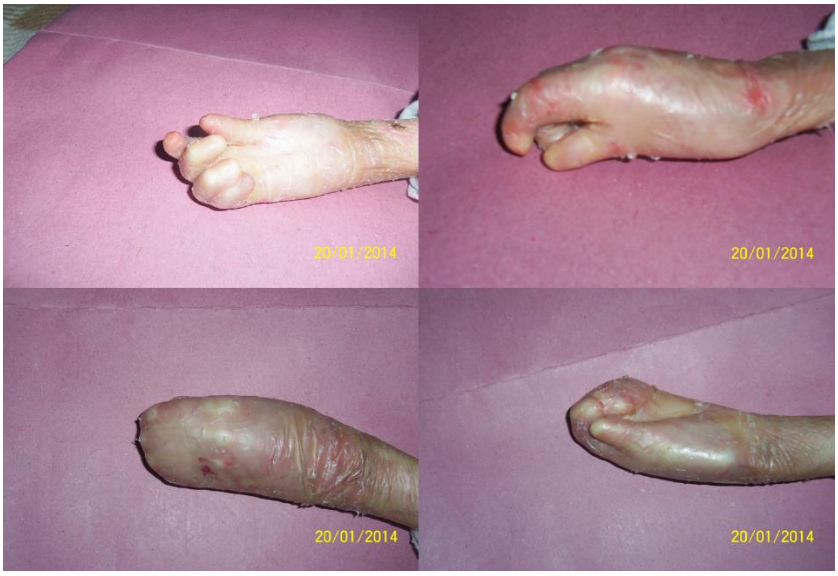

Figure 1: Pictures of the first patient showing a pseudosyndactyly referring to a mitten hands deformity

CASE PRESENTATION 2

Here we report the second patient, a 3-year-old girl, an only child, with a history of parental consanguinity, diagnosed with Recessive dystrophic epidermolysis bullosa since birth. She was referred to our department for management of pseudo-syndactyly of the hands in September 2017. He presented, at the age of 2 years, generalized erythematous-vesicular lesions, predominantly on the knees, ankles, both hands, and the neck. She also developed pseudo-syndactyly of both hands (Figure 2), leading to difficulty with hand mobility, especially the left hand.

The first patient initially underwent a first release of the thumb and index finger, as well as the second interdigital space. Subsequently, he underwent a secondary procedure for the release of the fourth and fifth interdigital spaces.

The second patient only underwent a release of the thumb and index finger, as well as the second interdigital space.

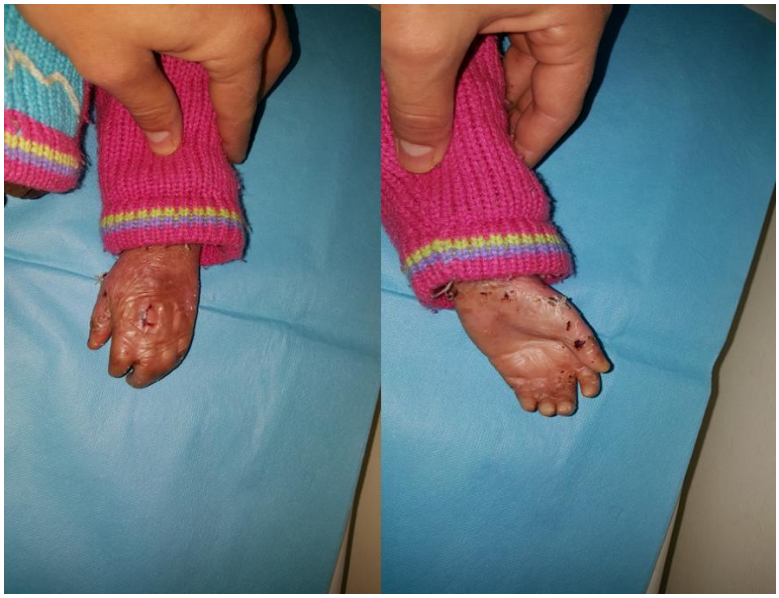

Figure 2: Pictures of the second patient showing a pseudosyndactyly referring to a mitten hands deformity

CASE PRESENTATION 3

The third patient is a 7-year-old boy. He was referred to our department for the management of flexion contracture and mitten deformity. The history of the disease dates back to birth when he was diagnosed with Recessive dystrophic epidermolysis bullosa, with no parental consanguinity reported. At the age of 7 years old, he presented erythematous-vesicular lesions on the inguinal region, the shoulders, and both hands (Figure 3).

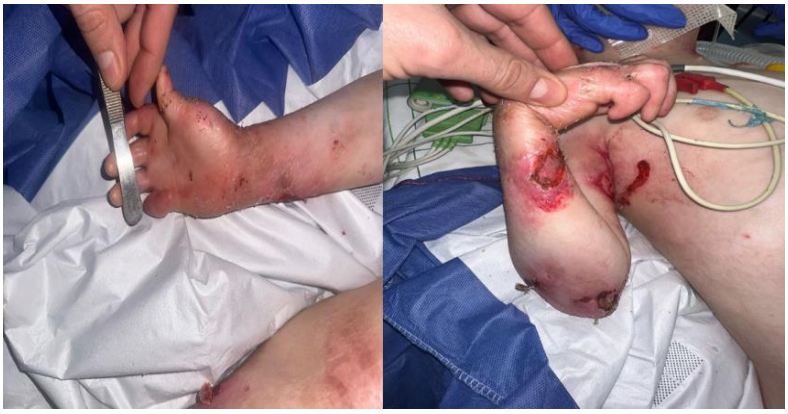

Figure 3: Pictures of the third patient showing erythematous-vesicular lesions, flexion contracture, and mitten deformity

SURGICAL APPROACH

All patients were carefully placed on an operation table which was prepared to prevent pressure ulcers. Large padding was used at the pressure zones (Figure 4), and repositioning of the patients during the procedure should be avoided. We proceed with an incision at the dorsal side of the first interdigital space extending from the base of the metacarpal and continuing into the palm up to the proximal palmar crease. This was coupled with a fasciectomy. Flexion contractures were then released using a palmar transverse incision (scarification) at the interphalangeal and metacarpophalangeal joint. Then We placed axial wires in the distal phalanx, which we pushed further to the level of the proximal phalanx, to maintain finger extension. To cover the soft tissue defect we chose the full-thickness skin graft, preloved from the inguinal region (Figure 5 & 6).

Dressing in Vaseline gauze was applied, and changed every 48 hours, with good evolution.

All patients were discharged after two weeks, and functional hand training activities were started with good results after a one-year follow-up (Figure 7).

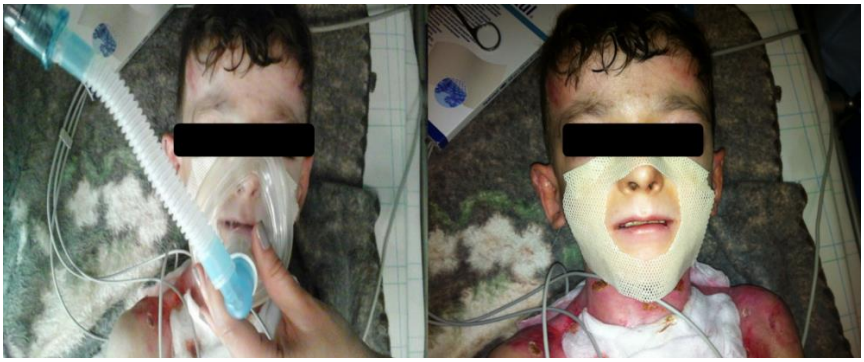

Figure 4: Pictures of the first patient showing facial protection in pressure zones

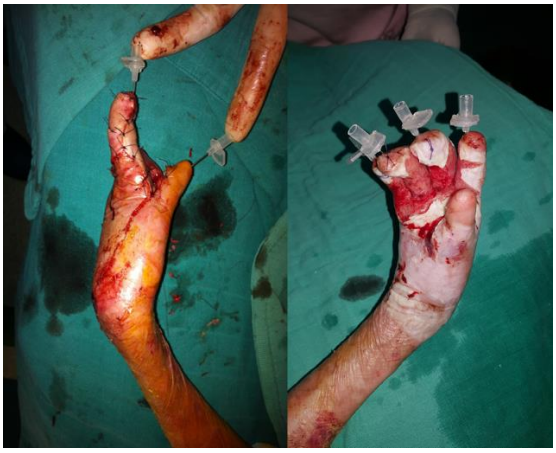

Figure 5: First patient preoperative images after a skin graft

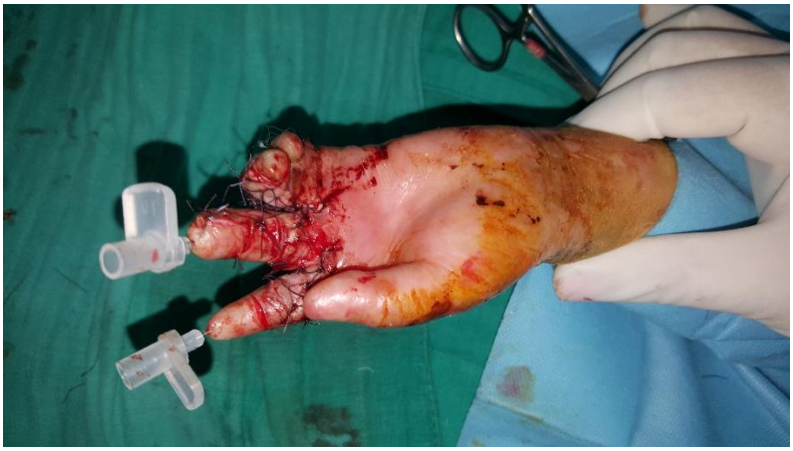

Figure 6: Second patient preoperative image after a skin graft

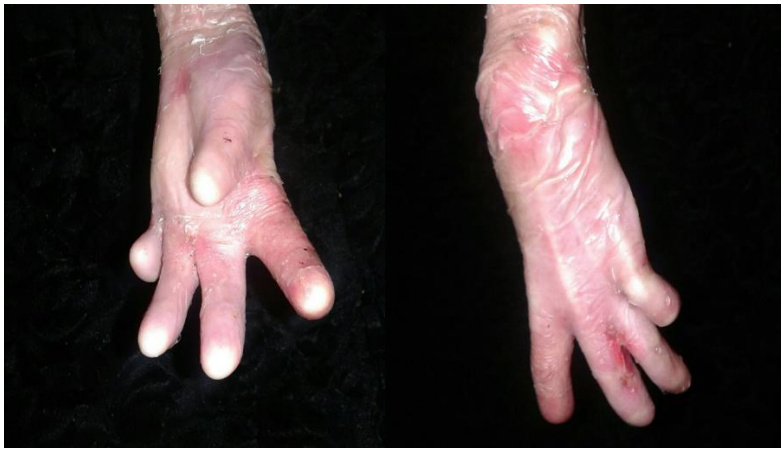

Figure 7: Image showing the First Patient results after a one-year follow-up

DISCUSSION

Epidermolysis bullosa (EB) is the name given to a heterogeneous group of rare, inherited skin diseases, It was first described by Von Hebra in 1870 [1]. characterized by fragility of the epidermis. The underlying genetic abnormalities cause destabilization at the dermo-epidermal junction. Although 30 subtypes are described, the main clinical types of EB are simplex (if the blisters are within the basal keratinocytes), junctional (within the lamina lucida), dystrophic (in the superficial papillary dermis), and Kindler syndrome (a mixed type) [1, 4]. Dystrophic EB (DEB) may be transmitted as an autosomal dominant or recessive subtype. Recessive dystrophic EB (RDEB) is the more frequent DEB subtype with a prevalence of about 2 in 1,000,000 population [1, 5]. In addition, RDEB is related to the loss of collagen VII expression that causes clinical symptoms at birth, including generalized blisters, ulceration of the mucous membranes, and atrophic scars. These symptoms gradually deteriorate with disease progress and can cause chronic blood loss, recurrent infections, and malnutrition. This can result in anemia, delayed puberty, and osteoporosis [6, 7]. RDEB-generalized patients who survive past childhood frequently develop metastatic squamous cell carcinoma (SCC), which often leads to death [8, 9].

Deformities of the hand in RDEB include both the cutaneous and musculoskeletal systems [10]. These progressive deformations lead to an inevitable decrease in hand function. With each relatively minor traumatic episode, ulceration produces fibrinous adhesions and scars. The result is an obliteration of the inter-digital spaces, progressing to the formation of pseudosyndactylies. The same process occurs in the palm, initially causing an adduction contracture of the thumb. In some cases, severe mitten-like deformities can cause significant dysfunction that leads to the loss of ability to perform even the most basic daily tasks [10]. These hand deformities are known as ‘cocoon hands’ [11].

RDEB is burdened with a high psychological impact on patients and their families. A multidisciplinary approach to managing the disease is fundamental and surgery plays a critical role. “Mitten hands” compromise the functional development of the hands in children with RDEB. So, an early surgical approach is of paramount importance. Fine *et al*., showed that about 61% of RDEB patients required hand surgery, typically needing 5 or more operations [5]. Other studies revealed that pseudosyndactylies in patients with this disease are characterized by high rates of recurrence [13].

Surgery is still crucial to correct severe hand deformities, improving patient's quality of life. In 1995, Ciccarelli *et al*., proposed many indications for surgery, such as palmar contracture, contracture of the proximal interphalangeal (IP) joint > 30°, severe small finger deformity, pseudosyndactyly extending to the proximal IP joint, and severe impairment of daily activities [14].

It is important to know that conventional surgical techniques for "mitten hand" are limited to the release of pseudosyndactylies and wound coverage, achieving optimal epithelialization, beginning early mobilization, and providing long-term stability with minimal recurrence [13]. Various methods are described, but no consensus exists. The simplest procedure to remove layers of tissue to allow finger separation was first described by Kitlowski and Banfield in 1948 [15]. This technique led to a rapid recurrence of deformities within a few months of surgery. A "decocooning" (removal of the superficial epidermal layer) of the hand to identify interdigital spaces has been advised by Greider, Flatt, and Campiglio *et al*., [16, 17]. Meanwhile, other authors have not found this necessary [18]. According to Cavallo and Smith, the identification of these spaces can be done simply by palpation [19]. On the one hand, the release of pseudosyndactylies has neither been recommended by Horner *et al*., nor by Cuono and Finseth because of the rapid recurrence of digital contractures [20, 21]. On the other hand, Terrill et al. demonstrated a separation of the digits maintained beyond 3 years [22]. Moreover, Abboud *et al*., showed that results are rather in favor of the release of digital pseudosyndactylies, as 77 percent of operated patients were able to maintain digital function beyond 3 years [23].

The type of wound coverage that ensures high effectiveness of treatment and low rates of recurrence continues to be debated. Some authors recommended using skin grafts for secondary surgical wounds. Luria *et al*., [5], Terrill *et al*., [22], and Horner *et al*., [20] used a variety of techniques for split-thickness skin grafts. In addition, Rees *et al*., [24] and Ladd *et al*., [25] chose to graft full-thickness skin. They believed that grafts could protect the exposed neurovascular bundles and tendons, facilitate wound healing, and perhaps reduce the recurrence of hand contracture. However, Vozdvizhensky *et al*., [26] and Ciccarelli *et al*., [14] held the opposite view, suggesting that skin grafting not be used to avoid damage in donor-site skin, which may result in extra trauma and bullae. Ciccarelli *et al*., [14] showed that skin grafting is unnecessary as it does not affect the average recurrence rate. Bioengineered skin was also reported in the treatment of RDEB but is very costly and difficult to obtain, which curtails its application [12].

CONCLUSION

Hand deformities occur in most patients with RDEB, and include adduction contractures of the first web space, pseudosyndactyly, and flexion contractures of the IP, MCP, and wrist joints. All structures in the hand may be involved. The severity of the deformity worsens with age, and surgical correction becomes increasingly difficult. Recurrent deformity occurs within 2 to 5 years. Meticulous skin care and the use of well-fitted splints supervised within a multidisciplinary team setting are essential. To date, there is no strong evidence base on which to plan surgical treatment of the hand in RDEB.

Declaration of patient consent: The authors certify that they have obtained all appropriate patient consent forms.

Conflicts of interest: The authors declare no conflicts of interest.

Author contributions

All authors participated in this work. All have read and approved the final version of the manuscript.

REFERENCES

1. Hsu, C. K., Wang, S. P., Lee, J. Y. Y., & McGrath, J. A. (2014). Treatment of hereditary epidermolysis bullosa: updates and future prospects. *American journal of clinical dermatology*, *15*, 1-6.
2. Laimer, M., Prodinger, C., & Bauer, J. W. (2015). Hereditary epidermolysis bullosa. *JDDG: Journal der Deutschen Dermatologischen Gesellschaft*, *13*(11), 1125-1133.
3. El Hachem, M., Zambruno, G., Bourdon-Lanoy, E., Ciasulli, A., Buisson, C., Hadj-Rabia, S., ... & Bodemer, C. (2014). Multicentre consensus recommendations for skin care in inherited epidermolysis bullosa. *Orphanet Journal of Rare Diseases*, *9*, 1-20.
4. Has, C., Bauer, J. W., Bodemer, C., Bolling, M. C., Bruckner-Tuderman, L., Diem, A., ... & Mellerio, J. E. (2020). Consensus reclassification of inherited epidermolysis bullosa and other disorders with skin fragility. *British Journal of Dermatology*, *183*(4), 614-627.
5. Fine, J. D. (2016). Epidemiology of inherited epidermolysis bullosa based on incidence and prevalence estimates from the National Epidermolysis Bullosa Registry. *JAMA dermatology*, *152*(11), 1231-1238.
6. Luria, S., Radwan, S., Zinger, G., & Eylon, S. (2014). Hand surgery for dystrophic epidermolysis bullosa. *Journal of Pediatric Orthopaedics*, *34*(7), 710-714.
7. Pfendner, E. G., & Lucky, A. W. (2006). Dystrophic epidermolysis bullosa. NCBI bookshelf. A service of the National Library of Medicine [updated 2018 Sep 13]. National Institutes of Health; Seattle (WA), p. 21.
8. Montaudié, H., Chiaverini, C., Sbidian, E., Charlesworth, A., & Lacour, J. P. (2016). Inherited epidermolysis bullosa and squamous cell carcinoma: a systematic review of 117

cases. *Orphanet journal of rare diseases*, *11*(1), 117–129.
9. Hon, K. L. E., Li, J. J., Cheng, B. L., Luk, D. C., Murrell, D. F., Choi, P. C., & Leung, A. K. (2015). Age and etiology of childhood epidermolysis bullosa mortality. *Journal of Dermatological Treatment*, *26*(2), 178-182.
10. McGrath, J. A., O'Grady, A., Mayou, B. J., & Eady, R. A. J. (1992). Mitten deformity in severe generalized recessive dystrophic epidermolysis bullosa: histological, immunofluorescence, and ultrastructural study. *Journal of cutaneous pathology*, *19*(5), 385-389.
11. Eisenberg, M., & Llewelyn, D. (1998). Surgical management of hands in children with recessive dystrophic epidermolysis bullosa: use of allogeneic composite cultured skin grafts. *British journal of plastic surgery*, *51*(8), 608-613.
12. Fivenson, D. P., Scherschun, L., & Cohen, L. V. (2003). Apligraf in the treatment of severe mitten deformity associated with recessive dystrophic epidermolysis bullosa. *Plast Reconstr Surg*, *112*(2), 584–588.
13. Tuncer, S., Sezgin, B., Kaya, B., Ayhan, S., & Latifoglu, O. (2018). An algorithmic approach for the management of hand deformities in dystrophic epidermolysis bullosa. *J Plast Surg Hand Surg*, 52, 80-86.
14. Ciccarelli, A. O., Rothaus, K. O., Carter, D. M., & Lin, A. N. (1995). Plastic and reconstructive surgery in epidermolysis bullosa: clinical experience with 110 procedures in 25 patients. *Annals of plastic surgery*, *35*(3), 254-261.
15. Kitlowski, E. A., & Banfield, E. E. (1948). Epidermolysis bullosa hereditaria; report of a case with unusual associated deformities. *Plastic and reconstructive surgery*, *3*(4), 481–488.
16. Greider, J. L., & Flatt, A. E. (1983). Care of the hand in recessive epidermolysis bullosa. Plast Reconstr Surg, 72, 222–225.
17. Campiglio, G. L., Pajardi, G., & Rafanelli, G. (1997). A new protocol for the treatment of hand deformities in recessive dystrophic epidermolysis bullosa (13 cases). *Ann Hand Surg*, *16*(2), 91-101.
18. Bernardis, C., & Box, R. (2010). Surgery of the hand in recessive dystrophic epidermolysis bullosa. *Dermatol Clin*, 28, 335–341.
19. Cavallo, A. V., & Smith, P. J. (1998). Surgical treatment of dystrophic epidermolysis bullosa of the hand. *Tech Hand Upper Extrem Surg*, *2*(3), 184–195.
20. Horner, R. L., Wiedel, J. D., & Bralliar, F. (1971). Involvement of the hand in epidermolysis bullosa. *J Bone Joint Surg*, 53, 1347.
21. Cuono, C., & Finseth, F. (1978). Epidermolysis bullosa: current concepts and management of the advanced hand deformity. *Plast Re- constr Surg*, 62, 280–285.
22. Terrill PJ, Mayou BJ, Pemberton J. Experience in the surgical management of the hand in dystrophic epidermolysis bullosa. Br J Plast Surg. 1992;45:435–42.
23. Abboud, L., Leclerc-Mercier, S., Bodemer, C., & Guéro, S. (2022). Hand surgery in recessive dystrophic epidermolysis bullosa: Our experience with dermal substitutes. *Journal of plastic, reconstructive & aesthetic surgery: JPRAS*, *75*(1), 314–324.
24. Rees, T. D., & Swinyard, C. A. (1967). Rehabilitative digital surgery in epidermolysis bullosa. *Plast Reconstr Surg*, *40*(2), 169–174.
25. Ladd, A. L., Kibele, A., & Gibbons, S. (1996). Surgical treatment and postoperative splinting of recessive dystrophic epidermolysis bullosa. *J Hand Surg Am*, *21*(5), 888–897.
26. Vozdvizhensky, S. I., & Albanova, V. I. (1993). Surgical treatment of contracture and syndactyly of children epidermolysis bullosa. *Br J Plast Surg*, *46*(4), 314–316.