Surgical management of pseudoainhum in loricrin keratoderma

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INTRODUCTION

In Vohwinkel syndrome (of which loricrin keratoderma is a variant), surgical management of pseudoainhum often results in recurrence. To the authors' knowledge, only 2 cases of disease-free intervals greater than 24 months have been reported in scientific literature. In this report, the authors report one of the longest disease-free intervals in the literature.

CASE REPORT

A 5-year-old boy with a known diagnosis of loricrin keratoderma presented to the clinic for evaluation of a painful right fifth toe, with associated color changes. His mother had noted generalized dry skin prone to fissuring despite the use of several topical therapies.

Physical examination showed fine scale on all areas of the body. A faint hourglass constriction of approximately 5 mm was present over the proximal aspect of the phalanx of the right fifth toe (Fig 1). The toe was noted to have slight congestion distal to the site of constriction, with capillary refill of 1 second. Additional constriction bands were present at the proximal aspect of the interphalangeal joint of the fourth and fifth digits of the hands bilaterally (Fig 2). Constrictions on the left hand were asymptomatic. Radiographs of the foot revealed no bony abnormalities.

Initially, conservative management with topical emollients (petrolatum, 12% ammonium lactate lotion, and pumice bar) was pursued. However, the constriction progressed. At the site of the constriction band on the right fifth toe, the patient developed a local infection, resolved with oral antibiotics. Surgical intervention was pursued to prevent further progression, with initial release of the constriction bands on the right fifth toe and finger. The right finger constriction band was corrected via excision followed by Z-plasty flaps overlying the radial proximal aspect of the interphalangeal joint. The right toe constriction band was excised along the medial border of the proximal aspect of the interphalangeal joint, followed by full-thickness skin graft

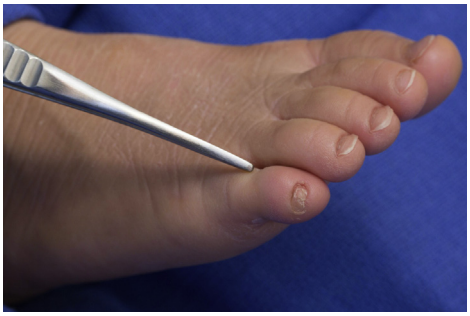

Fig 1. Loricrin keratoderma. Faint hourglass constriction of approximately 5 mm over the proximal aspect of the phalanx of the right fifth toe.

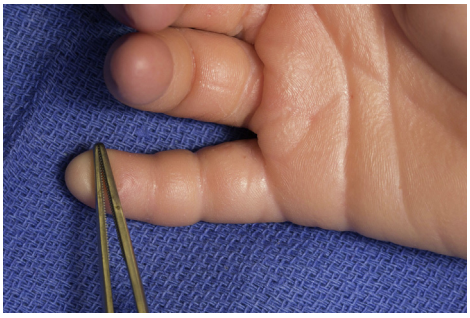

Fig 2. Loricrin keratoderma. Constriction band at the proximal aspect of the interphalangeal joint of the right little finger.

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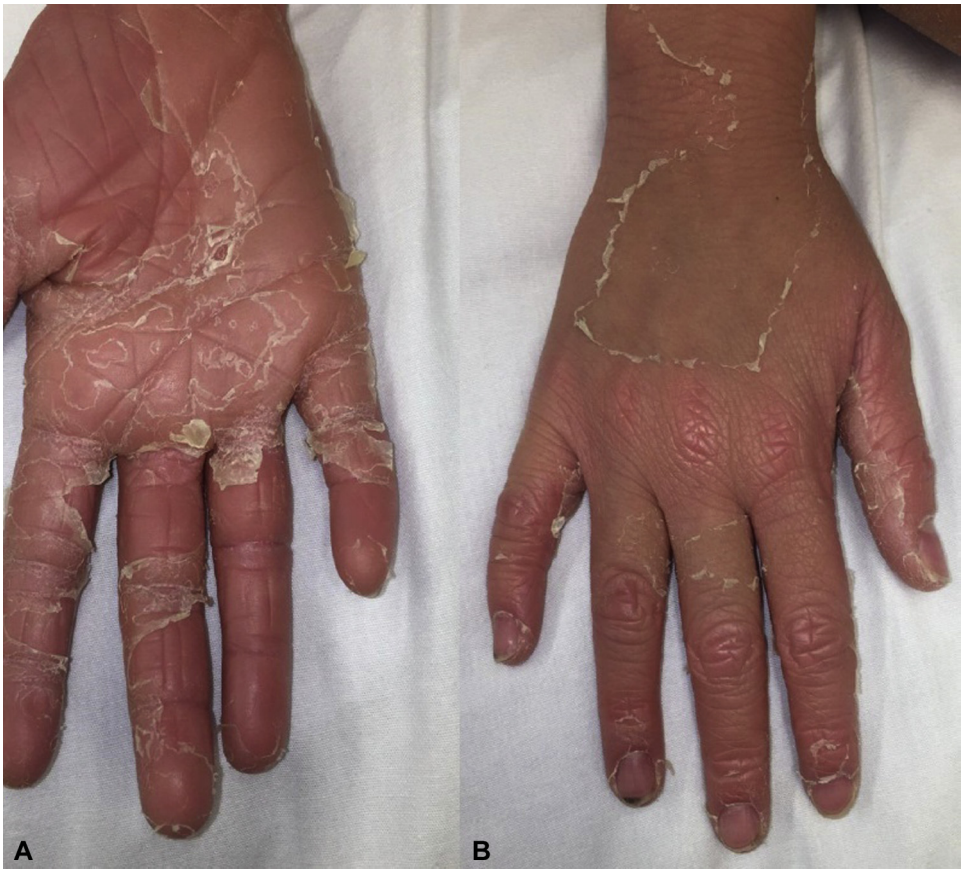

Fig 3. Loricrin keratoderma. Twenty-four months postexcision followed by Z-plasty flaps overlying the radial proximal aspect of the interphalangeal joint of the right little finger. **A**, Palmar aspect. **B**, Dorsal aspect.

from the right side of the groin. Constriction release of the left fifth finger was pursued later.

At 24-month follow-up, the incisions were appropriately healed, without evidence of recurrence or ischemia (Fig 3). Unfortunately, no immediate postoperative photograph of the toe was available. Final pathology showed markedly hyperkeratotic skin.

DISCUSSION

Constriction bands fall into 2 categories: ainhum and pseudoainhum. *Ainhum* refers to a primary process of autoamputation, almost exclusively found on the fifth toe of male individuals accustomed to going barefoot in underdeveloped countries of Africa. *Pseudoainhum* refers to the secondary process of autoamputation, that which mimics ainhum.

The pathophysiology of pseudoainhum is classified into congenital constricting bands caused by the umbilical cord; constricting bands from external forces, such as hair or thread; and constricting bands secondary to another disease process. These diseases may be hereditary or nonhereditary. Known hereditary causes include psoriasis, lamellar ichthyosis, pachyonychia congenita, and Vohwinkel syndrome.

Classic Vohwinkel syndrome, also known as keratoderma hereditarium mutilans, is an inherited palmoplantar keratoderma characterized by honeycomblike palmoplantar keratoderma, starfish-shaped keratotic plaques, and sensorineural deafness. Vohwinkel syndrome is rare; approximately 50 cases have been recorded in the literature. It occurs as the result of a heterozygous mutation in the gap junction beta 2 *(GJB2)* gene encoding protein connexin 26. A variant of Vohwinkel syndrome, loricrin keratoderma, is an inherited palmoplantar keratoderma featuring a similar honeycomblike keratoderma, but with the addition of prominent generalized ichthyosis and without sensorineural deafness. Loricrin keratoderma is a result of heterozygous frameshift insertion or deletion mutations in *LOR* encoding the loricrin protein integral to the cornified cell envelope. Both conditions commonly result in the gradual development of pseudoainhum.[1]

Treatment of loricrin keratoderma, particularly the constriction bands, has included both medical

Table I. Literature review: surgical treatment of pseudoainhum

Treatment	Clinical diagnosis	Duration of success	Comment
Full-thickness skin graft (Simkin)[2]	Epidermolytic ichthyosis	2 y	Subsequent to failed Z-plasty
Full-thickness skin graft (Zamiri)[3]	Loricrin palmoplantar keratoderma	36 mo	Donor site was left forearm
Z-plasty (Christopher)[4]	Erythropoietic protoporphyria	Not specified	
Z-plasty (Atabay)[5]	Vohwinkel syndrome	3 y	
Z-plasty (Raque)[6]	Unspecified congenital collagen dysplasia	6 mo	Excision of band, staged Z-plasties; constriction bands both legs
Z-plasty (Shetty)[7]	Amniotic band syndrome	Not specified	Staged Z-plasty
Z-plasty (Pickus)[8]	Vohwinkel syndrome	1 y	
Flap (Kim)[9]	Epidermolysis bullosa	5 mo	Circumferential excision, followed by cross-finger flap and full-thickness skin graft
Flap (Bassetto)[10]	Vohwinkel syndrome	18 mo	Cross-finger flap
Flap (Zhang)[11]	Vohwinkel syndrome	18 mo	Distant abdominal skin flap
Flap (Pisoh)[12]	Vohwinkel syndrome	2 mo	Lateral digital flap, transposed into the palmar defect, and a split-thickness graft, harvested from the forearm, to cover the secondary defect
Flap (Solis)[13]	Vohwinkel syndrome	Unspecified	Multiple surgical releases, including cross-finger flap
Surgical release (Kumar)[14]	Psoriasis	2 wk	
Surgical release (McLaurin)[15]	Psoriasis	4 mo	
Surgical extirpation of foreign body (Sebaratnam)[16]	Hair-thread tourniquet syndrome	Unspecified	
Excision (Pirozzi)[17]	Presumed amniotic band	6 mo	Elliptical incision and resection of bone
Excision (Besonhe)[18]	Collodion baby	4 mo	"The membrane was opened like a book and completely removed"

and surgical therapies. Medical management is frequently disappointing, often leading to surgical intervention as tissue constriction progresses.

Surgical options described for management of digital constriction bands include excision alone, excision followed by local tissue rearrangement, skin grafting, local flaps, or distant flaps. Given the rarity of the disease, it is difficult to suggest a preferred approach. Split-thickness skin grafts can have more contraction than full-thickness ones. Therefore, full-thickness skin grafts are always used on the digits. However, regardless of the approach, recurrence after surgery is common, especially after surgery involving local flaps. Table I includes a summary of surgical treatments for pseudoainhum reported previously. Because underlying etiology influences response to treatment, caution is needed in comparing outcomes across etiologies.

Pisoh et al[12] reported recurrence as quickly as 2 months. This was after a lateral digital flap, transposed into the palmar defect, and a split-thickness graft, harvested from the forearm, to cover the secondary defect. The longest known disease-free follow-up is 36 months, as reported by 2 studies: Atabay et al,[5] after treating with Z-plasty, and Zamiri and Watson,[3] after treating with full-thickness skin graft harvested from the forearm. It is theorized that the incorporation of unaffected, healthy tissue from a distant site avoids the use of local pathologic tissue that, on a cellular level, may retain the potential to reestablish constriction. In the future, definitive treatment may be achieved via gene therapy.

CONCLUSION

In current literature, surgical treatment of pseudoainhum in Vohwinkel syndrome often results in recurrence of the constriction band by 12-month follow-up. With no recurrence at 24-month follow-up, we report one of the longest disease-free intervals after surgical intervention of constriction bands via Z-plasty and full-thickness skin grafting. Z-plasty and skin grafting are relatively quick procedures, with minimal morbidity, offering an excellent treatment option when constriction bands threaten digital loss.

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