

What's the resolutive surgery for pseudo-ainhum in Vohwinkel syndrome? A case report and review of the literature

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of the GJB2 gene, which codes for the connexin 26 protein.⁴ The classic variant of this disease is characterized by fibrous bands (pseudoainhum) on the fifth finger that can lead to a progressive self-amputation, starfish-shaped keratosis on the elbows, knees, wrists and knuckles of the hand, and deafness. Nail dystrophy, alopecia, onychogriphosis and various neurological anomalies may also occur. Another variant of this disease is the Camisa variant. This is caused by a mutation of the loricrina protein - synthesized in the grainy layer of the epidermis - and leads to ichthyosis.⁵ Medical treatment includes systemic synthetic retinoids derived from vitamin A, which improve the differentiation and proliferation of the epidermal tissues.^{6,7} Different experimental surgical techniques, such as local tissue rearrangements, local flaps and skin grafts, have been described for the treatment of pseudoainhum. In the present study we report the case of a fifty-year-old woman affected by Vohwinkel syndrome showing constriction bands (pseudoainhum) in the right fifth digit.

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Abstract

Vohwinkel Syndrome, also known as Keratoderma Hereditarium Mutilans, is an extremely rare dominant autosomal keratosis. It typically presents with “starfish” keratoses on the knuckles, palmoplantar keratoderma (PPK), hearing impairment and mutilating digital constriction bands (pseudoainhum) that cause strangulation, often leading to autoamputation of the affected digit. Both medical and surgical treatment haven't shown to date consistent results, in the treatment of pseudoainhum. In this study we present the case of a woman with Vohwinkel syndrome who showed constriction bands causing ischemic changes of the 5th digit of the right hand for which she was treated with surgery. We also present a review of the literature for the management of this disease.

Introduction

Vohwinkel syndrome, or hereditary mutilating keratoderma, is a rare connective tissue disease, and only a few cases have been described in the literature. It most commonly affects caucasian women, and it occurs during childhood with worsening in adolescence. It can be sporadic, or inherited with an autosomal dominant pattern and a variable phenotype.¹⁻³

It was described by Vohwinkel in 1929, and it is caused by a heterozygous mutation

Case Report

A 55-year-old caucasian woman, with bilateral sensorineural hearing loss, came to our attention complaining of severe pain in the fifth digit of the right hand. The pain had been mild during the last year and became acute in the last two months. Clinical examination showed “honeycomb” palmar keratosis, starfish keratosis on the dorsal side of both hands and feet (which the patient had been suffering since the age of five), digital constriction bands in the proximal interphalangea (PIP) joint of the right fifth digit and skin dyschromia (Figure 1a). Neurovascular deficit with pain, paresthesia and hypoesthesia of the fifth finger were also present. Her father, sister and grandmother had the same syndromic pattern. Based on these clinical findings a diagnosis of Vohwinkel syndrome was made. Plain radiographs of the right hand showed periosteal thinning of the proximal phalanx of the fifth digit and soft tissue alteration (Figure 1b).

MR confirmed the periosteal thinning and showed a lack of visualization of a segment (8 mm) of the extensor tendon at the proximal interphalangeal joint of the fifth digit, compatible with a complete lesion (Figure 1c). Marked thinning of the fifth digit's flexor tendon, thickening of the flexor tendons of the other fingers, and thinning of their extensor counterparts, were also evident.

Conservative surgical treatment was

proposed to the patient, but owing to the high recurrence rate described in the literature she refused this type of approach and preferred amputation of the proximal phalanx of the fifth digit of the right hand.

In anesthesia, a skin incision was made through the proximal interphalangeal joint and a capsulotomy was performed. We then proceeded with the amputation and disarticulation of the head of the proximal phalanx, followed by proper hemostasis, tendon plasty procedure and suturing (Figure 2). The incision was medicated with sterile gauzes, and an elastic dressing was applied.

Post-surgical follow-up consisted in anterior-posterior and latero-lateral radiographs of the right hand, and an in-hospital observation period of 24 hours during which neither vascular nor nervous deficits emerged. Optimal articular range of motion was maintained and pain was greatly reduced. Histological examination showed (severe hyperkeratosis, hypergranulosis, and mild acanthosis, of the cutaneous component consistent with the diagnosis of Vohwinkel Syndrome (Figure 3).

At 15 days follow-up, the surgical wound was clean, no sign of infection was evident, and the patient didn't complain any pain. At 12-month follow-up, the patient did not report pain, and plain radiographs showed no abnormalities at the site of amputation.

Discussion and Conclusions

In our case the diagnosis of Vohwinkel syndrome was made on the basis of clinical findings and then confirmed by histological examination, as previously reported in the literature.⁸ Our patient showed a typical clinical pattern with honeycomb, diffuse palmoplantar keratoderma, starfish keratosis of the dorsal surfaces of the digits, fibrous constriction bands (pseudoainhum) of the 5th digit, hearing loss, deaf mutism and scarring alopecia. Differential diagnosis includes other keratodermas presenting with digital autoamputation, such as: Mal de Meleda, Olmsted syndrome, acral keratoderma, pachyonychia congenita, palmoplantar keratoderma of Sybert, and palmoplantar keratoderma of Gamborg-Nielsen, as well as acquired dermatoses that can lead to the appearance of constricting bands, such as leprosy, tertiary syphilis, ainhum, scleroderma, amniotic bands, Raynaud syndrome, and syringomyelia.⁹ There are no sufficiently distinctive histopathological pictures to differentiate palmoplantar keratoderma in one type or another.^{10,11} In our case histological examination showed hyperkeratosis, acanthosis, and hypergranulosis which are suggestive for pseudoainhum.^{12,13} Unlike other Authors, Luk *et al.*¹ considered hypogranulosis an atypical histological finding for pseudoainhum. Pseudo-ainhum is a congenital disorder characterized by fibrous bands development on fingers and toes, while ainhum is an idiopathic disease involving the fifth toe of black people. In both cases, if not treated, fibrosis can progressively compromise the mobility and the neurovascular function leading to self-amputation.¹⁴ In the few cases of Vohwinkel syndrome reported in the literature the results of medical and surgical treatments were often inconsistent owing to the lack of histologic examination and uncertain diagnosis.^{2,6,15-17} Systemic retinoids and derivatives of vitamin A have been used to treat these patients, since they may facilitate the differentiation and proliferation of the epidermal skin layer, which could help to treat pseudoainhum. However, this therapy has given only modest results, and its use is very limited due to its toxicity and teratogenicity. To date no standard surgical treatment or guidelines exist for the treatment of pseudoainhum in patients with Vohwinkel disease, however four surgical techniques have been described for the treatment pseudoainhum (Table 1). Z-plasty has been used to treat early stages of ainhum (phase I-II) to release the constrictive base. For phases III and IV, amputation is



Figure 1. a) Palmar keratosis, starfish keratosis and digital constriction bands on the PIP joint; b) radiographs show periosteal thinning of the proximal phalanx; c) MRI shows periosteal thinning and a lack of visualization of the extensor tendon at the proximal PIP.

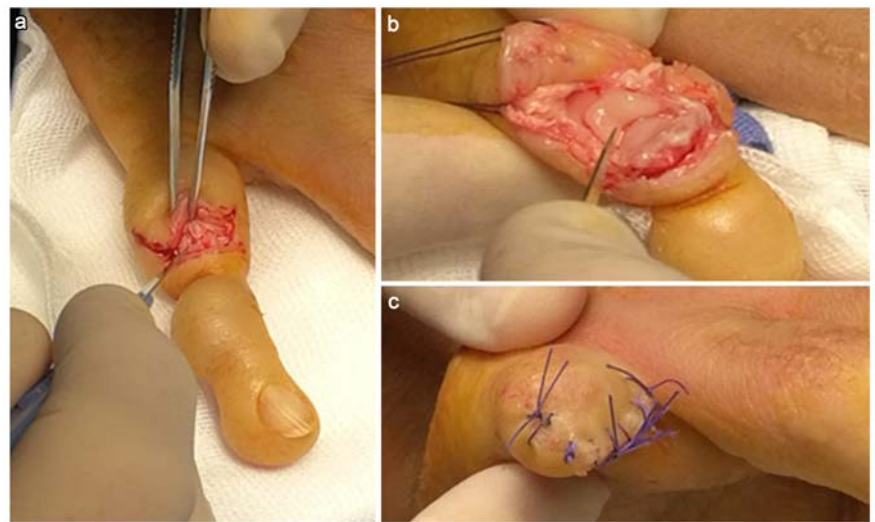


Figure 2. a) Incision; b) disarticulation of the head of the proximal phalanx, followed by proper hemostasis, tendon plasty procedure, and c) suturing.

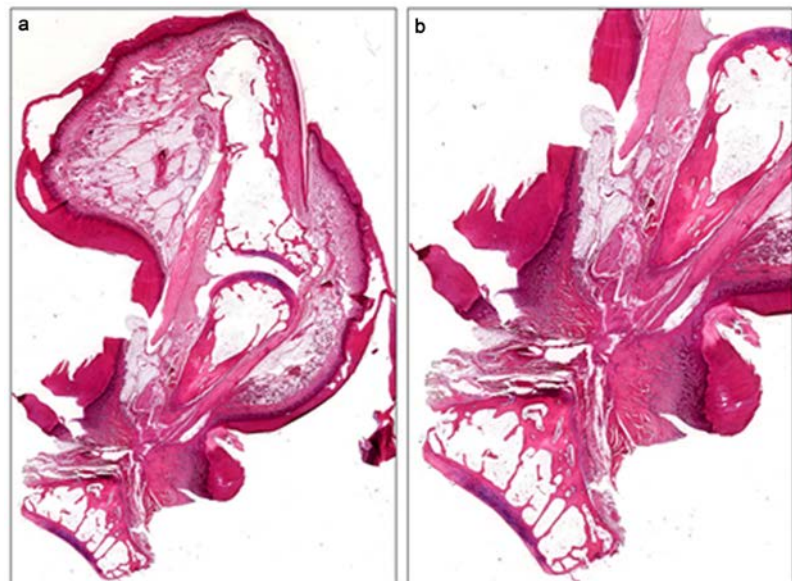


Figure 3. a) Orthokeratotic hyperkeratosis, acanthosis and hypergranulosis of the finger and constriction of the proximal PIP; b) Higher magnification of circumferential hyperkeratotic constriction band in the proximal interphalangeal joint. a, b) ematoxylin and eosin stain. Original magnifications: a) 10x; b) 20x.

Table 1. Different surgical techniques for the treatment of pseudo ainhum.

Author (year)	N. of patients	Age	Sex	Family history	Hystology	Surgery	Medical treatment	Follow-up
Luk <i>et al.</i> (1986) ¹	1	54	M	No	Yes	Z-plasties	No	12 months
Pisoh <i>et al.</i> (1995) ¹⁷	1	33	F	Yes	Yes	Z-plasty + skin graft	Vitamin A ointments	2 months
Solis <i>et al.</i> (2001) ²⁰	1	17	F	Yes	Yes	Cross finger-flap coverage + split thickness skin grafts	Ammonium lactate 12% lotion	No
Atabay <i>et al.</i> (2001) ¹²	1	24	F	No	Yes	Z-plasties	No	12 months
Sinha <i>et al.</i> (2009) ²	2	46, 30	M, F	Yes	No	1) Z and Y-V plasties; 2) thickness skin graft	1) Acetretin; 2) no	5 years, 8 years
Bassetto <i>et al.</i> (2010) ¹⁹	1	48	F	Not available	No	Cross finger flap	Systemic retinoids	18 months
Liebman <i>et al.</i> (2013) ¹⁶	1	23	F	Not available	No	Full-thickness skin graft	No	16 months
Zhang <i>et al.</i> (2016) ¹³	1	24	F	Yes	Yes	Abdominal skin flap	No	18 months
Zamiri <i>et al.</i> (2018) ¹⁵	1	62	M	Not available	No	Full thickness skin grafts	Acitretin	36 months

generally necessary, unless self-amputation has not already taken place.¹⁸ Z-plasty, consisting in the surgical excision of the fiber band and lengthening by Z-plasty, was the first technique to be used in 1986 by Luk *et al.*¹ and then by Pisoh *et al.* in 1995.¹⁷ However, the lack of follow-up data represents an important limit of these two studies. Two other cases reported in the literature, treated with Z-plasty describe the recurrence of the fiber band 36 and 6 months after surgery, respectively.^{2,12} Crossing finger flap used by Bassetto *et al.*¹⁹ and Solis RR²⁰ is a technique to cover the palmar area of the PIP - in which the fibrous band was removed - and had satisfactory results at 18-month follow-up. This procedure was used for the first time by Atasoy *et al.*²¹ in 1979 to treat digital injuries. The limit of this technique is the thinness and the lack of adequate subcutaneous tissue at the level of the proximal interphalangeal joints.

The full-thickness skin graft consists of full-thickness excision of the constriction band with identification and preservation of the neurovascular bundles, and application of a skin graft harvested from the groin crease or from the forearm.^{16,17} Lieban *et al.* used this technique successfully, with amelioration of the pseudo-ainhum, without recurrence of the treated area after 16 months of follow-up. Zamiri *et al.* also reported a successful treatment of pseudoainhum using the full-thickness excision techniques, with no recurrence after 36 months. In 2016, Zhang *et al.*¹³ designed a surgical approach for digital constriction bands using a distant abdominal skin flap to repair the wound after excising the constriction band from the digits. No recurrence occurred after 18 months of follow-up. Using a distant abdominal skin flap can ensure blood supply, and flap survival as well as improve the appearance.

This technique may permanently solve the constriction band recurrence problem; however, it requires a long treatment period, with 3 operations under local anesthesia, and a relatively long local immobilization time.

In our case we had to perform an amputation due to the patient's poor compliance, and to the clinical severity (neurovascular impairment of the finger). According to the literature, with special reference to the work of Liebamann *et al.*,¹⁶ the excision of the constriction bands may not be sufficient. Indeed, the condition could recur over time considering the characteristics of the disease, and its tendency towards specific areas of the fingers. The skin graft tends to maintain the characteristics of the donor site, therefore it could definitively solve the problem, if taken from not affected areas.

In our review of the literature we report many surgical techniques for the treatment of pseudoainhum, however in all cases the authors report a short term follow up which doesn't allow to define a resolute treatment.

We believe that a multidisciplinary approach for the management of these patients could help to prevent long-term self-amputation.

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