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

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Abstract

Vohwinkel Syndrome, also known as Keratoderma Hereditarium Mutulans, is an extremely rare dominant autosomal keratosis. It typically presents with “starfish” keratoses on the knuckles, palmarplant 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 of the GJB2 gene, which codes for the connexin 26 protein.2 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.3 Medical treatment includes systemic synthetic retinoids derived from vitamin A, which improve the differentiation and proliferation of the epidermal tissues.4,5 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.

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 interphalangeal (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 thickening 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 5th 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 gauges, 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. Our patient showed a typical clinical pattern with honeycomb, diffuse palmar plantar keratoderma, starfish keratosis of the dorsal surfaces of the digits, fibrous constrictions 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, palmar plantar keratoderma of Sybert, and palmar plantar 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. There are no sufficiently distinctive histopathological pictures to differentiate palmar plantar keratoderma in one type or another. In our case histological examination showed hyperkeratosis, acanthosis, and hypergranulosis which are suggestive for pseudoainhum. Unlike other Authors, Luk et al. 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. 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. 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
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 et al. (1986)11   | 1              | 54  | M   | No             | Yes       | Z-plasties                                      | No                | 12 months |
| Pisco et al. (1995)17 | 1              | 33  | F   | Yes            | Yes       | Z-plasty + skin graft                           | Vitamin A ointments | 2 months |
| Solis et al. (2001)18 | 1              | 17  | F   | Yes            | Yes       | Cross finger-flap coverage + split thickness skin graft | Ammonium lactate 12% lotion | No        |
| Atabay et al. (2001)19| 1              | 24  | F   | No             | Yes       | Z-plasties                                      | No                | 12 months |
| Sinha et al. (2009)2   | 2              | 46,30| M, F| Yes            | No         | 1) Z and Y-V plasties; 2) thickness skin graft | 1) Acitretin; 2) no | 5 years, 8 years |
| Bassett et al. (2010)10| 1              | 48  | F   | Not available  | No         | Cross finger flap                               | Systemic retinoids | 18 months |
| Liebman et al. (2013)14| 1              | 23  | F   | Not available  | No         | Full-thickness skin graft                       | No                | 16 months |
| Zhang et al. (2016)15  | 1              | 24  | F   | Yes            | Yes       | Abdominal skin flap                             | No                | 18 months |
| Zamiri et al. (2018)16 | 1              | 62  | M   | Not available  | No         | Full thickness skin grafts                      | Acitretin         | 36 months |

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