

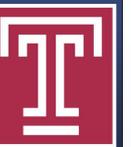
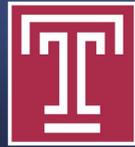
# A Case Report of Elective Amputation Following Pseudo-ainhum Secondary to Vohwinkel Syndrome

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## Statement of Purpose and Literature Review

Vohwinkel Syndrome (VS), also known as mutilating palmoplantar keratoderma, is a rare autosomal dominant genetic disorder. In fact, only about 50 cases have been reported in medical literature. There are two types of VS dependent on the mutation of specific genes. Mutation of the GJB2 gene is associated with hearing loss, hyperkeratosis of the palms and soles, constricting bands or pseudo-ainhum of the digits, and starfish-shaped hyperkeratosis. Mutation of the loricrin gene has similar clinical findings, but with the absence of hearing loss. Treatment is primarily symptom-based, with topical keratolytic and systemic retinoids to treat hyperkeratosis and with reconstruction or amputation for treatment of digital pseudo-ainhum [1].

Given the relative rarity of the condition in the medical literature, there are no clear guidelines when it comes to the management of patients with VS. Most treatment plans are individualized based on patient symptoms.

**In this report we review a case of a female patient who presented with a symptomatic pseudo-ainhum of the right 5th digit secondary to Vohwinkel Syndrome.**

## References

- [1] Bassetto, F., Tiengo, C., Sferrazza, R., Belloni-Fortina, A., Alaibac, M. Vohwinkel Syndrome: Treatment of Pseudo-ainhum. *International Journal of Dermatology* 2010, 49, 79-82.
- [2] Dumet Fernandes, J., Menta Simonsen Nico, M. Low Dose Isotretinoin Prevents Digital Amputation in Loricrin Keratoderma (Vohwinkel Syndrome with Ichthyosis). *Journal of the German Society of Dermatology*. 2017.665-666.
- [3] Prieto Herman Reinehr, C., Peruzzo, J., Cestari, T. Vohwinkel Syndrome: Ichthyosiform Variant in a Family. *Anais Brasileiros de Dermatologia*. 2018; 93 (5): 723-5.
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## Case Report

A 23-year-old female of Southeast Asian heritage previously diagnosed with VS and with a family history of VS presented with a painful right 5th digit secondary to pseudo-ainhum. Progressive worsening of deformity and pain was noted over a period of several months without alleviating factors. She had a prior history of left 5<sup>th</sup> toe amputation for the same issue 5 years prior. The band involved approximately the medial 2/3 of the digit at the level of the proximal phalanx (Figure 1). Hyperkeratosis of the palms and soles consistent with her diagnosis of VS were also apparent (Figure 2).

An elective 5th metatarsal-phalangeal joint disarticulation with primary closure was performed (Figure 3). This healed uneventfully with suture removal at two weeks (Figure 4). Complete resolution of symptoms was noted, although she remains at risk for development of future issues.



## Discussion

Vohwinkel Syndrome (VS) is a rare autosomal dominant genetic disorder with little supporting medical literature. This is particularly true within the foot and ankle surgical literature. Although this case did not require the most challenging surgical technique certainly, it does provide insight into the clinical pathology and pathogenesis of a rare condition with lower extremity manifestations.

A review of the contemporary literature demonstrates that most pseudo-ainhums of the digits are eventually treated with amputation, although some have advocated for the initial use of topical keratolytics and systemic retinoids [1,2,3]. This has been described with mixed results. Dumet Fernandes et al. showed that a daily dose of 0.35 mg/kg for a month followed by an eight month daily dose of 0.5mg/kg showed significant improvement in the annular constriction of a 5th finger [2]. This prevented the need for an amputation. However, in a case reported by Bassetto et al., the use of systemic retinoids did not show clinical improvement in the pseudo-ainhum of the left 5th digit, which eventually required amputation[3].

**It is our hope that this brief case report increases awareness and knowledge of a rare genetic disorder with lower extremity manifestations.**

