INTRODUCTION

Vohwinkel syndrome (VS), also known as keratoderma hereditaria mutilans, is a rare, autosomal dominant, and syndromic form of diffuse palmoplantar keratoderma (PPK) which manifests as hyperkeratosis of the palms and soles with a honeycomb appearance.1

Vohwinkel's syndrome is classified into two variants: (1) a deafness-associated variant (Classical) and (2) an ichthyosis-associated variant (Camisa syndrome).2 Camisa syndrome, also called as variant Vohwinkel's syndrome or loricrin keratoderma, is a rare variant that is associated with ichthyosis most commonly ichthyosis vulgaris and lamellar ichthyosis.3 Pseudoainhum (constricting circumferential band around a digit or limb) is one of the classical features of this syndrome while starfish-shaped keratotic papules and deafness are not observed.4,5

Recently, it has been shown that gain-of-function mutations in LOR on 1q21.3. underlies the ichthyotic variant while that in connexin 26, genes causes VS with deafness.6,7 The histological features of Camisa syndrome include hyperkeratosis with orthokeratosis and focal parakeratosis, acanthosis, elongation of rete ridges, and sparse dermal lymphocytic infiltrate with normal appendages.8,9 Retinoids such as acitretin and isotretinoin have been proven to be effective in hereditary palmoplantar keratoderma and preventing pseudoainhum.10,11

Here, we report a case of Camisa syndrome with pseudoainhum in the fifth toe leading to amputation as timely treatment was not sought.

KEYWORDS
acitretin, camisa syndrome, pseudoainhum

CASE REPORT

We present a case of 38-year-old woman who presented to our Dermatology OPD with complaints of palmoplantar thickening and scaling. During her childhood, she had noticed slight scaling localized to bilateral palms and soles which then progressed to become brownish honeycomb transgradient hyperkeratosis over a period of around ten years (Figure 1A, B; Figure 2A, B). It was associated with pain, tightening, and winter aggravation. She also had generalized dryness of the skin. However, she did not receive any treatment, and at the age of twenty-five years, she gradually developed a constriction band around the fifth toe which gradually tightened leading to pseudoainhum with amputation at the level of the proximal interphalangeal joint (Figure 2A, B). Her hearing, vision, hair, and nails were normal. Among her two children, the elder son is eleven years old and is also affected and the younger daughter is unaffected till this date. Considering the
presence of transgradient honeycomb keratoderma with generalized ichthyosis and pseudoainhum in the absence of deafness, a diagnosis of Camisa syndrome was made.

3 | TREATMENT AND FOLLOW-UP

Initially, she was treated with oral acitretin, and currently, she is under maintenance therapy with topical urea and salicylic acid. She is also being closely followed up to assess for further progression of disease and formation of pseudoainhum.

4 | DISCUSSION

Vohwinkel syndrome was first described in 1929 by Vohwinkel. VS with ichthyosis (Camisa's syndrome) is a rare group of inherited genodermatoses, with very few cases reported in the literature. It has an autosomal dominant inheritance due to mutations in LOR, starting in childhood, and occurring predominantly in females and Caucasians. Our case and her son also had childhood onset. Furthermore, it can be made out that the mother had sporadic condition and the son got it from her in an autosomal dominant fashion.

Clinical features of Camisa's syndrome include generalized ichthyosis and honeycomb-like palmoplantar keratoderma, with or without varying degrees of constricted digits, erythematous plaques, and/or erythroderma. Honey comb-like keratoderma, pseudoainhum, and ichthyosis were present in our patient, and her hearing was normal. Pseudoainhum and amputation are most common in the fifth toe, which was the case in our patient as well. (Table 1).

Here, we list the case reports and series of Camisa syndrome and associated pseudoainhum. (Table 1).

Acitretin is a feasible, conservative, and durable therapeutic modality for the treatment of pseudoainhum-like mutilations associated with ichthyoses and mutilating keratodermas. The use of isotretinoin in a non-continuous regimen is a reasonable approach in women of childbearing age, especially in cases at risk of amputation due to severe pseudoainhum. For pseudoainhum threatening, the viability of the digits excision of the constricting band followed by Z-plasty has been reported to be effective. Successful full-thickness skin grafting for pseudoainhum
TABLE 1  Case reports of Camisa disease

| SN  | Authors                      | Country          | No. of cases | Cases with Pseudoainhum | Digits                        | Age of pseudoainhum | Amputation | Treatment                                |
|-----|------------------------------|------------------|--------------|--------------------------|-------------------------------|---------------------|------------|------------------------------------------|
| 1   | Kura, Parsewar               | India            | 1            |                          | Left 5th toe                  | 14 years            | Absent     | Acitretin 25 mg PO BD x 6 months        |
| 2   | Reinehret al                 | Brazil           | 19 (1 family)|                          | 2nd and 5th finger of hand    | Not mentioned       | Absent     | Not mentioned                           |
| 3   | O’Driscoll et al             | UK               | 14 (1 family)|                          | 5th toes of bilateral feet    | Not mentioned       | Left 5th toe at 26 years and right 5th toe at 44 years | Surgical amputation done |
| 4   | Korge et al                  | Scotland         | 8 (1 family) | Present but number not specified | Not mentioned               | Not mentioned       | One had amputation | Not mentioned                           |
| 5   | Armstrong et al              | UK               | 8 (1 family) |                          | Not mentioned                | Not mentioned       | Absent     | Not mentioned                           |
| 6   | Maestrini et al, Schmuth et al | USA             | 16 (1 family)| Nearly all (number not specified) | 4th or 5th digit of hands or feet | Not mentioned       | Some had amputation of bilateral 5th toe | Not mentioned                           |
| 7   | Rajashekar et al             | India            | 1            |                          | Right 1st and 5th toes       | Not mentioned       | Absent     | Not mentioned                           |
| 8   | Corte et al                  | Brazil           | 2 (1 family) |                          | Left 5th toe                 | Not mentioned       | Present in 1 patient | Not mentioned                           |
| 9   | Camisa, Rossano              | Not available    | Not available | Absent                   | Not applicable               | Not applicable      | Not applicable | Isotretinoin                            |
| 10  | Takahashi et al              | Japan            | 1            |                          | All fingers and 5th toes     | Not mentioned       | Absent     | Not mentioned                           |
| 11  | Zamiri et al                 | UK               | 1            |                          | All fingers                  | Not mentioned       | Absent     | Acitretin, keratolytic, full-thickness skin graft |                                |
| 12  | Suzuki et al                 | Japan            | 8 (2 families)|                          | Not mentioned                | Not mentioned       | Not mentioned | Not mentioned                           |
| 13  | Nico, Fernandes              | Brazil           | 1            |                          | Right 5th toe                | Not mentioned       | Right 5th toe at 25 years | Isotretinoin 0.5 mg/kg x 8 months |
| 14  | Hotz et al                   | Brazil, Brazilian origin, France | 4 (3 families) |                          | 1st—mild constriction 2nd—Fingers 3rd – 5th toe | Not mentioned       | Absent     | Not mentioned                           |
| 15  | Kinsler et al                | Not specified    | 5 (1 family) | Not mentioned            | Not applicable               | Not applicable      | Not applicable | Not mentioned                           |
| 16  | Mu–noz-Aceituno et al        | Spain            | 2 (1 family) | Absent                   | Not applicable               | Not applicable      | Not applicable | Emollients                              |
| 17  | Matsumoto et al              | Japan            | 3 (1 family) |                          | PIP of all fingers           | 1st—3 years         | Absent     | Not mentioned                           |
| 18  | Gedicke et al                | Germany          | 5 (1 family) | Absent                   | Not applicable               | Not applicable      | Not applicable | Not mentioned                           |

(Continues)
has also been reported. Our patient was treated with oral acitretin and topical urea and salicylic acid. This has halted the further progression of disease and loss of other digits.

This case highlights the importance of recognition and prompt treatment of Camisa syndrome to prevent the pseudoainhum formation and improve the quality of life.

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CONFLICTS OF INTEREST
The authors declare no conflict of interest.

AUTHOR CONTRIBUTIONS
Bibisha Baaniya involved in preparation of manuscript and editing. Sudha Agrawal involved in Idea and literature review.

CONSENT
Patient provided written consent for publication of this case report.

DATA AVAILABILITY STATEMENT
Data will be made available upon request.

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