Case Report

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Keratosis Follicularis Spinulosa Decalvans: A Series of Three Cases in a Family

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Abstract

Keratosis follicularis spinulosa decalvans (KFSD) is a rare genodermatoses characterized by hyperkeratotic follicular papules on the scalp, with progressive cicatricial alopecia of the scalp, eyelashes, and eyebrows. Ocular involvement with photophobia and keratitis, and dental anomalies may also be seen. Due to the genetic and clinical heterogeneity of similar disorders, a definitive diagnosis of KFSD is often daunting. We report a case series from India of three siblings, including a young girl and her two elder brothers, all of whom had features consistent with KFSD. There morphological heterogeneity and differential diagnosis of the condition is also discussed.

Keywords: Keratosis Pilaris; Follicularis; Keratosis; Alopecia; Cicatricial.

Case Details

Case 1

A 14-year-old-girl (Proband) born out of first degree consanguineous marriage presented with complaints of loss of hair and spiny lesions on the scalp and eye brows since childhood. She recalled complete absence of hairs at birth and gradual appearance of sparsely distributed fine hairs over scalp with age. The spiny lesions on scalp and eyebrows were itchy, red and scaly. History of atopy and photophobia were absent. There was no history of similar complaints in the parents, although her brothers were detected to have similar skin lesions (*vide infra*). The child was stunted for her age. On cutaneous examination there was presence of erythematous hyperkeratotic papules associated with hypotrichosis, with fine, short, brittle hair, affecting the scalp, eyelashes and eyebrows with evidence of cicatricial alopecia at places, and presence of ichthyosis over the limbs [Figure 1]. Keratosis pilaris was noted on the extensor surfaces of the upper and lower extremities. There was no palmoplantar hyperkeratosis. Nails, teeth and genitalia were unremarkable. Trichoscopy revealed follicular keratosis with evidence of cicatricial alopecia, perifollicular scaling, and irregular eyelashes with scaling and erythema at the follicular base [Figure 2]. Two of the four brothers of the proband were also affected.

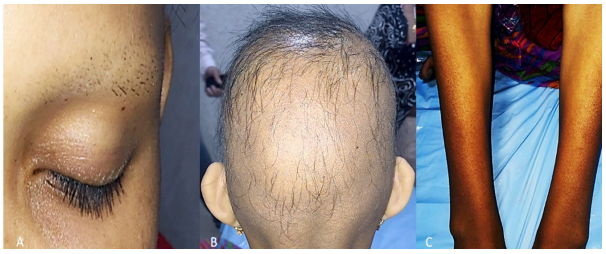

Fig. 1: Clinical features of the 14-year-old girl showing: **(A)** follicular papules with scaling and hypotrichosis of the eyebrows, **(B)** erythematous follicular hyperkeratosis over a background of sparse scalp hair, and **(C)** ichthyotic affection of the forearms

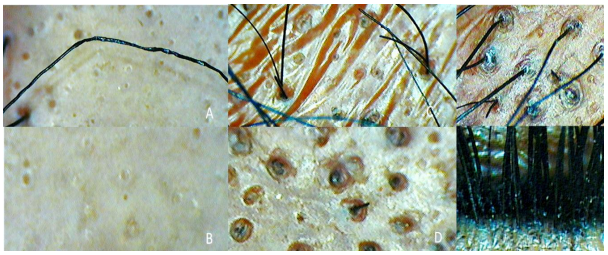

Fig. 2: Trichoscopic findings of the 14-year-old girl: **(A)** pigmented hair with wavy shaft suggestive of woolly hair, **(B)** cicatricial alopecia with loss of follicular orifices, **(C)** tufts of hair on the background of cicatricial alopecia, **(D)** follicular keratosis, **(E)** perifollicular scales, and **(F)** Irregular eyelash hairs with scaling at the base. (20 ×, non-polarized mode).

Case 2

The index patient's 22-year-old brother had similar complaints with sparse scalp, facial and body

hair, cicatricial alopecia of the scalp with hyperkeratosis, woolly hair and perifollicular scales, and ichthyosis [Figure 3]. Few follicular papules and minimal scarring alopecia were seen over the neck. Trichoscopy revealed wavy hair shafts suggestive of woolly hairs with loss of follicular orifices suggestive of scarring alopecia.

Case 3

The index patient's another 18-year-old brother had a similar history of absence of hair at birth with spontaneously regrowth at the age of 5 years. However, currently he has few patches of cicatricial alopecia over the scalp and sparse facial hairs, but with normal eyebrows, eyelashes, and body hairs.

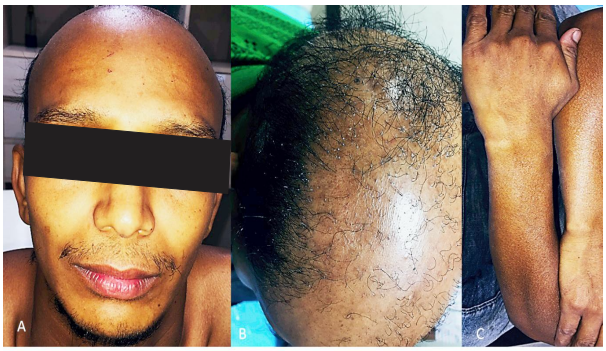

Fig. 3: Clinical features of the 22-year-old brother of the proband showing: **(A)** sparse scalp, facial hairs with irregular eyelashes, **(B)** perifollicular scaling and woolly hairs on background of cicatricial alopecia, and **(C)** ichthyosis on forearms

The constellation of clinical and trichoscopic findings in the patient, and her two brothers was consistent with a diagnosis of familial keratosis follicularis spinulosa decalvans (KFSD). Patients were offered scalp biopsy for histopathological confirmation but consent for the same was not obtained.

KFSD is one of several related disorders that are distinguished by the presence of keratosis pilaris with inflammation and subsequent atrophy. These disorders have been grouped under the category of keratosis pilaris atrophicans (KPA), which comprises keratosis pilaris atrophicans faciei, atrophoderma vermiculatum, and KFSD [1,2].

There are many sporadic cases, but the most intensive manifestations are found in men suggesting a pattern of X-linked pattern of inheritance. Rare instances of male-male transmission suggested the existence of an autosomal dominant transmission [3-6]. The suspected genetic locus has been traced to a mutation in MBTPS2 gene [6]. Phenotypic manifestations of KFSD are usually limited to skin, teeth, and eye. However, rare findings, including developmental delay, growth retardation, abnormal genitalia, and dysmorphic facies have been reported [5].

The overlap in clinical features between subtypes of scarring follicular keratoses can lead to misdiagnoses. While cicatricial alopecia of the scalp and eyebrows is a hallmark of the KFSD, other distinguished features of this disorder include. photophobia, widespread hyperkeratosis pilaris-like lesions, and teeth abnormalities.

The ichthyosis follicularis alopecia photophobia (IFAP) syndrome constitutes a close clinical differential of KFSD. IFAP is characterized by non-scarring alopecia, extensive keratosis pilaris, severe photophobia and corneal dystrophy [7]. The presence of scarring alopecia in our patients favoured the diagnosis of KFSD over the IFAP syndrome.

Although there is insufficient data and documentation with respect to the histopathological findings of scalp biopsy specimens of KFSD, a skin biopsy specimen may be sent for histological examination, including special stains for collagen and elastic fibers.

Histopathology helps to rule out other diseases in the differential diagnosis. The histological hallmark of KFSD appears to be compact hyperkeratosis and hypergranulosis of the upper follicular epithelium, indicating abnormal keratinization. There is follicular plugging and absence of hair shafts.

Treatment of KFSD is difficult and unsatisfactory. The aim of treatment is to stop the progression of the alopecia and clinical improvement of the areas that present with erythema and scaling.

Numerous treatments have been utilized, including topical and intralesional corticosteroids, topical and systemic antibiotics, dapsone, and systemic retinoids [8]. Despite follicular hyperkeratosis being an important component of the disorder, outcome of systemic retinoid therapy has been unsatisfactory.

Systemic isotretinoin therapy at a dosage of 1 mg/kg for 4 months caused slight to no improvement in 3 patients with KFSD and was associated with flaring-up of inflammation in one patient [9,10]. The relief induced by any treatment is at most temporary, followed by an almost imminent relapse upon discontinuation of the therapy. The current case series of the kindred of three siblings with KFSD has been reported to highlight the clinical and dermoscopic features of this rare genodermatosis.

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