Case Report

Keratosis follicularis spinulosa decalvans in a female

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ABSTRACT

Keratosis follicularis spinulosa decalvans (KFSD), is a rare follicular syndrome associated with widespread keratosis pilaris and progressive scarring alopecia. This genodermatoses often starts at infancy or early childhood with an X-linked mode of inheritance. Males are predominantly affected and females frequently show no disease or only a mild form. We describe this not so common entity of KFSD in a nine year old female child.

Key words: Keratosis follicularis spinulosa decalvans, female, child

INTRODUCTION

Keratosis pilaris atrophicans (KPA) is the umbrella term for a group of three rare and distinct clinical entities representing the scarring types of keratosis pilaris. Three categories of KPA include: Keratosis pilaris atrophicans faciei (KPAF), Atrophoderma vermiculatum (AV) and Keratosis follicularis spinulosa decalvans (KFSD). They have the following features in common: keratotic follicular papules, nonpurulent inflammation of variable degree, and atrophic end stages characterized by irreversible hair loss and/or atrophic depressions similar to pitted scars. All of these disorders appear to be hereditary. Although in its characteristic form it is considered as a harmless condition, in many instances the lesion can cause cosmetic problems. No effective therapy is available.

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CASE REPORT

A nine year old girl born from a first-degree consanguinous marriage, visited our outpatient department with complaints of rough skin over the scalp since five years and over the body since eight years of age in association with partial loss of scalp and eyebrow hair. At birth, her parents noted the absence of scalp and eyebrow hair, which gradually, over the next three to four years grew to some measure and eventually became scanty. Family history was noncontributory. Physical examination disclosed multiple follicular flesh-colored horny papules over the scalp, eyebrows, cheeks and both upper and lower limbs. A closer view of the scalp, cheek and eyebrow revealed fine scaling and areas of scarring alopecia, punctate atrophy and hair loss of the lateral half of eyebrows [Figures 1-3]. The teeth, nails, palms and soles were found to be normal. She gave no history of photophobia. Ophthalmic examination revealed no abnormal findings.

Hair microscopy was done to rule out monilethrix and in this case was found to be normal. A punch biopsy specimen from the scalp showed follicular plugging in the epidermis with mild acanthosis, early perifollicular fibrosis with mild lymphocytic infiltrate. The dermis

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was decreased in thickness with inconspicuous ill-formed sebaceous units. The hair shafts appeared to be normal [Figure 4]. With all the above findings in hand, a clinical diagnosis of keratosis follicularis spinulosa decalvans was made. Our patient was started on oral azithromycin for five days in view of the infection she had secondary to the excoriation, topical retinoic acid 0.025% on alternate days for the scalp at night and salicylic-steroid lotion during the day. The body was treated with urea and lecithin containing moisturizers. A month later, patient had marked improvement in terms of decreased roughness, increased hair growth and absence of disease progression. She was asked to continue the same topicals and come back for review in a month's time.

DISCUSSION

Keratosis follicularis spinulosa decalvans, a relatively rare disorder, was first described by Macleod,[1] however, it was Siemens who first used this descriptive term in 1926.[2] It has a X-linked mode of inheritance mapped to a locus at Xp 22.13 - p 22.2.[3] However, the condition has also been seen in families in which the pattern of transmission suggests an autosomal dominant inheritance.[4] Some authors used the appellation of folliculitis spinulosa decalvans (FSD) to define the autosomal dominant form, which could be differentiated from the commoner X-linked one by more pronounced follicular inflammation, particularly evident on the scalp.[5] In general, men are more severely affected while female heterozygotes may exhibit a milder phenotype in families with an X-linked dominant inheritance.[6] The process of lyonization (non-random X inactivation) may explain expression of KFSD in women. A study done by Aten *et al.*, showed that KFSD patients carry mutations in the MBTPS2 gene, and that this affects the normal

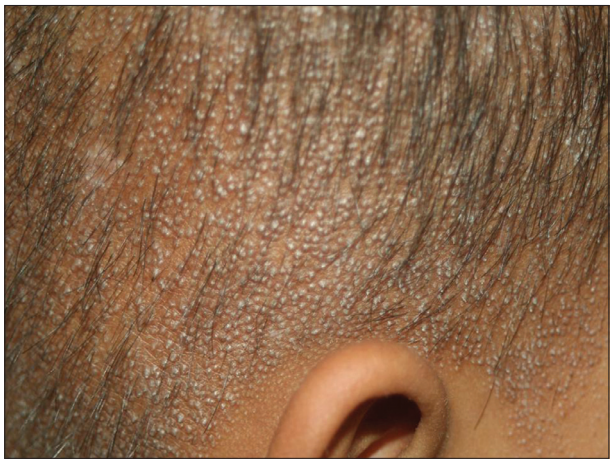

Figure 1: Scalp showing areas of scarring alopecia and KP lesions

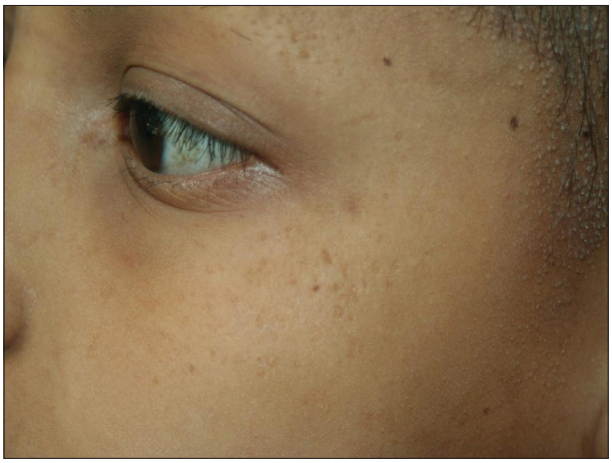

Figure 2: Cheeks showing punctate atrophy

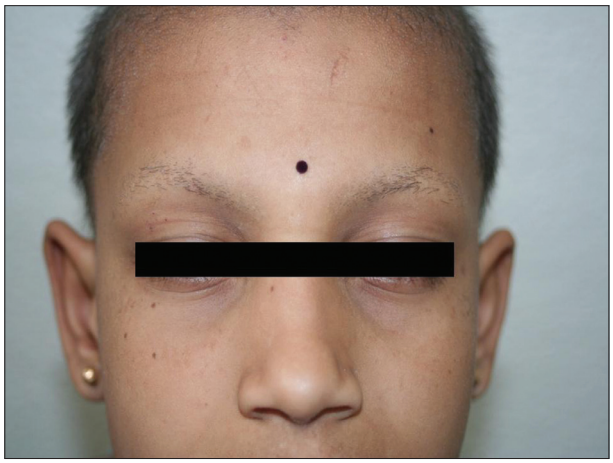

Figure 3: Bilateral loss of lateral one-third of eyebrows

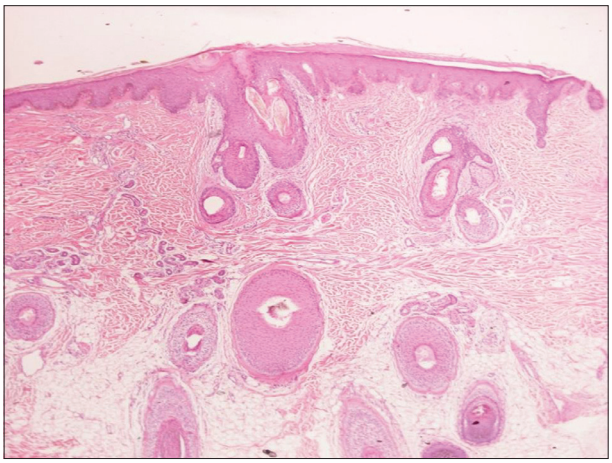

Figure 4: Microscopy-follicular plugging in the epidermis with mild acanthosis, early perifollicular fibrosis with mild lymphocytic infiltrate (H and E, ×200)

function of the protein by lowering MBTPS2 activity. MBTPS2 is required for cleavage of sterol regulatory element-binding proteins.[7]

The condition begins in infancy with numerous horny follicular plugs and milia on the nose and cheeks and later on the eyebrows, scalp, neck, and body. Scarring alopecia of the scalp, eyebrows, and eyelashes becomes apparent in childhood and progresses until puberty. Associated features include palmoplantar keratoderma, with predilection for the calcaneal region, and the unusual sign of high cuticles (or long cuticles).[6] The ocular abnormalities include photophobia, keratitis, conjunctivitis, congenital glaucoma and lenticular cataract.[2] Less commonly reported features are atopy, deafness, mental retardation, acne keloidalis nuchae and tufted hair folliculitis , aminoaciduria and woolly hair.[8-11]

KFSD simulates the ichthyosis follicularis alopecia photophobia (IFAP) syndrome. The latter is characterized by non-scarring alopecia, extensive KP, severe photophobia and corneal dystrophy.[12,13] The presence of scarring alopecia in our patients favors the diagnosis of KFSD over the IFAP syndrome. The other follicular conditions that it needs to be differentiated from are lichen planopilaris and lichen spinulosus. The former is characterized by atrophic or scarring patches on the scalp with complete loss of follicular orifices. The surrounding marginal hair follicles and residual small hairy islands within the patch reveal perifollicular erythematous macules and scale. Acuminate keratotic plugs can be frequently observed in the margins of the expanding area of alopecia.[14] The latter, a rare and benign disorder is characterized by follicular keratotic papules involving the neck, buttocks, abdomen, trochanteric regions, knees and extensor surfaces of the arms. Lichen spinulosus has a predilection for acral areas unlike keratosis pilaris, which is frequently limited to the upper aspects of the arms and legs. The horny spine that is characteristic of lichen spinulosus can be removed, leaving behind a tiny funnel-like orifice in the papule, whereas an entire individual lesion can be removed with the plug in keratosis pilaris.[15]

So far no effective therapy is known to work for KFSD. Frequent application of topical keratolytic agents and emollients improve skin texture.[4] Antibiotics such as tetracyclines, sulfonamides (dapsone), macrolides, penicillins and rifampin have been used at therapeutic doses and found to be ineffective.[4,16] Topical and intralesional corticosteroids were tried but caused transient improvement. Etretinate and isotretinoin have also been used but with variable results.[4,17] It is likely that retinoids, which are useful in disorders of keratinization, act by downregulating the process of follicular hyperkeratosis and inflammation. Laser-assisted hair removal with the long-pulse non-Q-switched ruby laser has been found to be useful in progressive or recalcitrant KFSD.[18]

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