Case Report (Open access)

Keratosis pilaris spinulosa decalvans in Siblings: A Rare Cause of Scarring Alopecia

Pallavi Goyal[1*], Isha Gupta[2]

[1]Junior resident, Department of Dermatology, Mahatma Gandhi medical college & hospital, Jaipur, India

[2]Senior resident, Department of Dermatology, Mahatma Gandhi medical college & hospital, Jaipur, India

[*]**Address for Correspondence:** Dr. Pallavi Goyal, Junior resident, Department of Dermatology, Mahatma Gandhi Medical College & Hospital, Jaipur, India

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ABSTRACT- Keratosis follicularis spinulosa decalvans (KFSD) is a hereditary disorder of the hair follicle, which presents with scarring alopecia and follicular papules affecting the scalp and other areas of the body. Being X-linked it is more common in males but rarely, can be seen in females. We reported that this rare disorder in siblings affecting both male and female children.

Key-words- Keratosis follicularis spinulosa decalvans, Siblings, scarring alopecia, Child, Keratotic papules

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INTRODUCTION

Keratosis pilaris atrophicans is a broad term encompassing three rare and distinct clinical entities, including keratosis pilaris atrophicans faciei (KPAF), atrophoderma vermiculata and keratosis follicularis spinulosa decalvans. Common to these conditions are keratotic follicular papules, non-purulent inflammation of variable degree and atrophic end stages characterized by irreversible hair loss and/or atrophic depressions similar to pitted scars. It has X- linked inheritance; however cases have also been seen in families, suggesting an autosomal dominant pattern of inheritance [1]. Very few cases of KFSD have been reported in Indian literature. We hereby present a case report of KFSD, a rare cause of scarring alopecia in siblings.

CASE REPORT

Three year old male child born prematurely at 7 months of gestation period presented with history of absence of scalp and eyebrow hair since birth and rough skin over the scalp for 2 years. At birth, parents noticed the absence of scalp and eyebrow hair. After 1 year of age fine, brittle and scanty hairs appeared.

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On physical examination multiple follicular, skin coloured keratotic papules were present on the scalp (Fig. 1) and extensor aspect of bilateral arms. The hairs on the scalp were sparse, short and brittle with areas of scarring alopecia (Fig. 2).

Eyebrows and eyelashes were also sparse. Teeth, nails, palms and soles were found to be normal. There was no history of photophobia and ophthamological examination revealed no abnormal findings. His younger sister 2 years old of age had similar complaints. She also had similar lesions over the scalp with loss of scalp and eyebrow hairs (Fig. 4) and the parents were unaffected.

Hair microscopy and dermoscopy of scalp hair was done to rule out monilethrix and found to be normal. Punch biopsy was taken from male child but not from his younger sibling. Histopathological examination revealed dilation of follicular infundibulum and plugging with orthokeratotic corneocytes with some others showing mild spongiosis. Few follicles in deep dermis showed dense perifollicular fibroplasia and sparse superficial perivascular and periappendageal lymphocytic infiltrate (Fig. 5 & 6). Based on above clinical feature, family history and histopathology diagnosis of keratosis follicularis spinulosa decalvans were made.

Our patient was started on 0.5 mg/kg of isotretinoin and topical retinoic acid 0.025% at night time. Lesions on the body were treated with retinoic acid at night and urea containing moisturizers at day time. After 2 months, the patient had improvement in terms of increased hair growth and absence of disease progression (Fig. 3). Follicular lesions softened but persisted. He was asked to continue same treatment and come back for review in a month time.

Fig. 1: Multiple skin coloured keratotic follicular papules with sparse hair

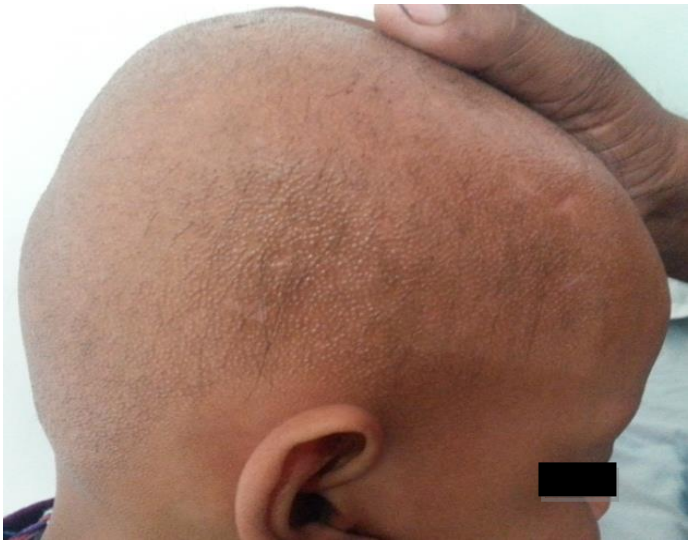

Fig. 2: Follicular papules with areas of Scarring alopecia before starting treatment

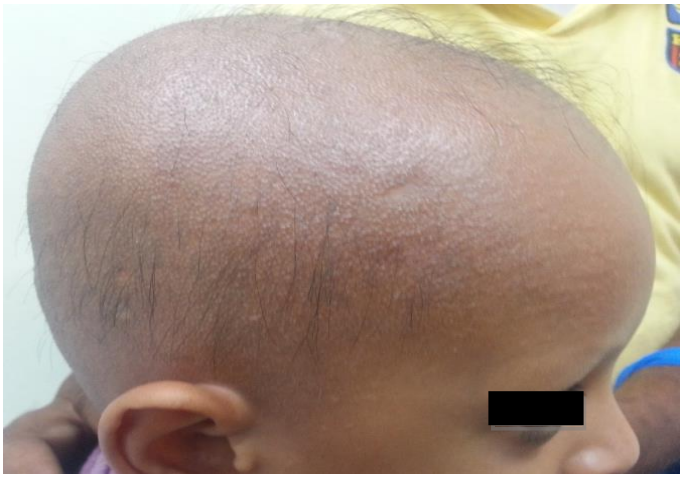

Fig. 3: After 1 month of giving isotretinoin therapy

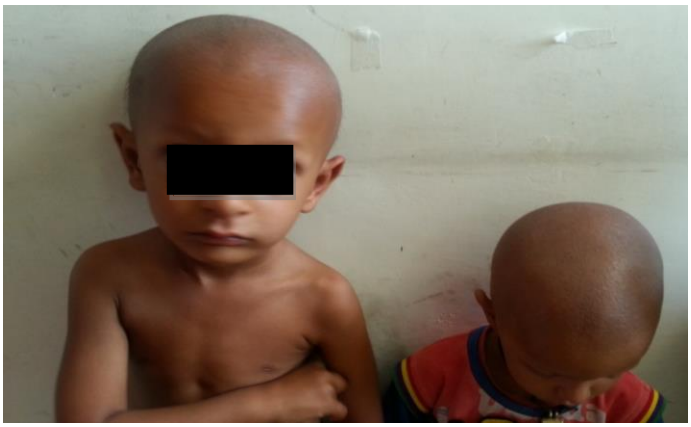

Fig. 4: Siblings with Sparse hair and Follicula keratotic papules

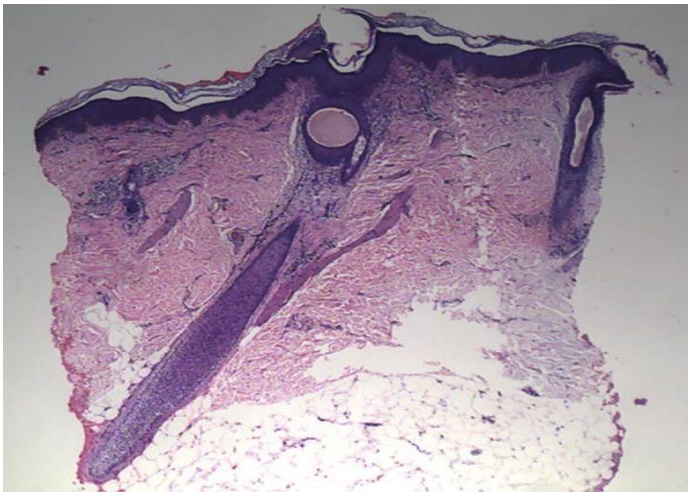

Fig. 5

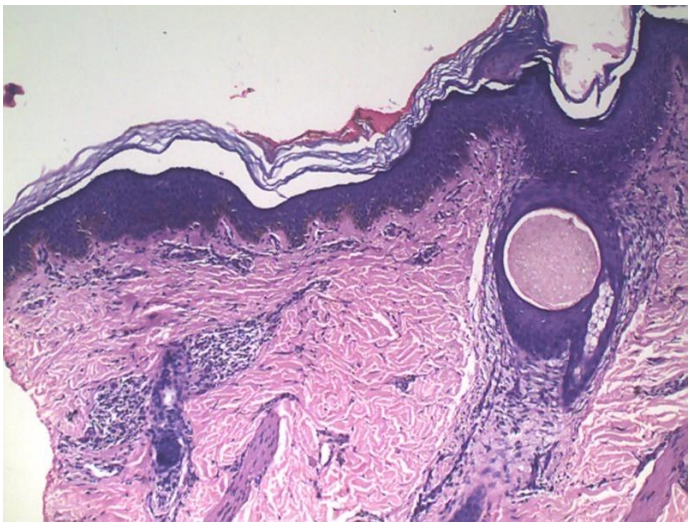

Fig. 6

Fig. 5 & 6: Dilation of follicular infundibulum and plugging with orthokeratotic corneocytes. Few follicles in deep dermis showed dense perifollicular fibroplasia and sparse superficial perivascular and periappendageal lymphocytic infiltrate

DISCUSSION

Keratosis follicularis spinulosa decalvans is a rare genetic disorder first described by Macleod however the descriptive term was first used by Siemens in 1926 [2]. It has X-linked dominant mode of inheritance mapped to chromosome Xp 22.13–p22.20. The candidate gene suggested is the membrane-bound transcription factor protease site 2 (MBTPS2) genes, which are required for cleavage of sterol regulatory element-binding proteins (SREBPs). Altered SREBP cleavage impairs cholesterol and lipid homeostasis in the skin, causing defective epidermal differentiation [3]. This condition affects predominantly males with females being carriers showing no disease or only mild forms. The process of lyonization (non–random X inactivation) may explain the expression of KFSD in women. However, cases have also been seen in families, suggesting autosomal dominant inheritance [1]. In the present case report, both brother and sister are affected without any other family members being involved. They may either have x-linked dominant inheritance with the process of lyonization explaining the manifestations in sister or autosomal dominant pattern of inheritance with variable penetrance.

The condition begins in infancy with follicular keratosis on the nose and cheeks and then on scalp, eyebrows, neck and body. Scarring alopecia of eyebrows and scalp begins in early childhood and progresses till puberty. Associated features include palmoplantar keratoderma, with predilection for calcaneal region, high periungual cuticles [4]. The ocular abnormalities include photophobia, keratitis, conjunctivitis, congenital glaucoma & lenticular cataract [5].

The differential diagnosis of KFSD includes ichthyosis follicularis alopecia, photophobia syndrome, lichen planopilaris and lichen spinulosa. IFAP syndrome is characterized by non-scarring alopecia in contrast to KFSD. The other two conditions can be differentiated by thorough clinic-pathological examination [2].

Treatment of KFSD is usually unsatisfactory. Topical treatment comprises mainly of keratolytics and emollients [1]. Systemic retinoids like isotretinoin and etretinate are beneficial in the early stages of the disease as they down regulate follicular hyperkeratosis and inflammation [6]. Dapsone was found to be useful due to its inhibition of leukocyte chemotaxis and stabilization of lysosomal enzymes [7]. Other treatments tried include antibiotics, intralesional and topical steroids. Hair reduction lasers have also been tried [8].

CONCLUSIONS

It is a rare cause of scarring alopecia affecting both brother and sister. Very few cases of Keratosis follicularis spinulosa decalvans have been reported in Indian literature especially in females.

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