

Differentiating Keratosis Follicularis Spinulosa Decalvans From Monilithrix : Two Rare Causes Of Scarring Alopecia In Young Male Siblings.

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Abstract: Keratosis follicularis spinulosa decalvans (KFSD) is a rare disorder affecting the hair follicles characterized by progressive cicatricial alopecia of the scalp and eyebrows, with usually X-linked recessive mode of transmission hence predominantly affecting Males.[1]

Monilethrix is a rare structural Hair shaft disorder with autosomal dominant mode of transmission, characterized by short, fragile, brittle hair that breaks spontaneously resulting in patchy dystrophic alopecia.[2]

Key words: Keratosis Follicularis Spinulosa Decalvans, Monilithrix, Hereditary Scarring Alopecias

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I. Introduction

KFSD is a rare type of primary scarring alopecia with lymphocytic predominance. It is an X-linked disorder of the hair follicle which was first described by Macleod, but the term KFSD was used by Siemen in 1926 when he reported the disorder in a Bavarian family.^[3]

The differential diagnosis of KFSD include ichthyosis follicularis alopecia (non -scarring) photophobia (IFAP) syndrome , Lichen planopilaris, Lichen spinulosus , Graham Little Piccardi Lasseur syndrome (GLPLS) and Monilethrix^[4,5]

Where the Lichen planopilaris, Lichen spinulosus can be differentiated clinically and histopathologically which shows vacuolar degeneration and interface dermatitis, IFAP can be excluded by non scarring pattern of hair loss and Graham Little Piccardi Lasseur syndrome (GLPLS) differentiated on the basis of clinical features - scarring alopecia of the scalp whereas nonscarring alopecia of axillae and pubic region.^[6]

First described by Walter Smith in 1897, Monilethrix is a rare structural hair shaft disorder with autosomal dominant inheritance characterized by a periodic thinning of the hair-shaft leading to a characteristic beaded or 'necklace' like appearance of the hair^[7]

Monilithrix can be differentiated from KFSD on the basis of clinical features, trichogram studies, genetic studies, histopathology, and microscopic examination of hair shaft.

II. Case Report

We present case history of two brothers aged 2.5 years and 5 years, born of a non-consanguineous marriage, who presented to Dermatology OPD accompanied by their Mother, with complaints of progressive hair loss over the scalp since birth with sparse eye brows.

Mother states that symptoms started 15 days after birth characterized by recurrent, raised, scaly, itchy papules over some areas of scalp, which were insidious in onset and gradually progressed to involve entire scalp, There was sparse growth of hairs over these papules which eventually shed off, leading to scarring hair loss, later on eyebrows were also involved. Similar complaints were present in younger brother suggesting X-linked inheritance. However she gave no history of similar complaints in her family.

On examination- multiple patches of cicatricial alopecia with sparse hair growing out of the follicular hyperkeratotic papules with perifollicular erythema of size <0.5cm, in the frontal, parietal, and temporal areas of the scalp (fig. 1). Presence of sparse hair over the eyebrows, eyelashes was noted (fig.2). No skin abnormality was observed. The oral cavity, nails, palms, and soles were found to be normal. Rest of the systemic evaluation revealed no abnormal findings.

Biopsy from the scalp was taken from younger brother and was sent for Histopathology which revealed presence of orthokeratosis with follicular plugging, mild perivascular lymphocytic infiltrate and presence of hair shaft within the keratinous material.

Considering the mode of inheritance of disease among two brothers, clinical features, and histopathological examination a diagnosis of KFSD was made.

III. Discussion

Monilithrix and KSFD both are responsible for hereditary form of scarring alopecia of childhood. Monilithrix being a disorder of Hair shaft whereas KSFD being disorder of Hair Follicles presenting with scarring alopecia and follicular papules affecting the scalp and other areas of the body^[1,2]

Monilethrix has predominant AD inheritance, genetic studies found human hair basic keratin hHb1 and hHb6 mutation as the chief cause. Also mutations in the hair cortex-specific keratin genes KRT81, KRT83, and KRT86 lead to AD monilethrix whereas mutations in the desmoglein 4 gene cause an AR form^[8] KFSD has X-linked inheritance where males are more commonly and severely affected than female heterozygotes. Gene studies discovered mutation in membrane-bound transcription factor protease site 2 (MBTPS2) gene at loci Xp22.13–22.2^[9] which eventually impairs cholesterol and lipid homeostasis in the skin causing defective epidermal differentiation^[10]

Clinically, in Monilithrix patients have normal hair at birth, but within the first months of life, they present with short, dull and fragile hairs that breaks easily especially in the sites of friction such as the top of the head, the nape and occipital areas. In severe forms, eyelashes, eyebrows, and pubic hairs may get involved^[11]

In KFSD the symptoms usually appear during the 1st weeks or months of life and are characterized by scarring alopecia associated with follicular hyperkeratosis of the skin, especially in the region of the face, and absence of the follicles of the hair, eyelashes, and eyebrows^[12] Ocular symptoms like photophobia, keratitis, conjunctivitis, glaucoma, cataract, and corneal dystrophy may be seen. In some cases palmoplantar keratoderma is also present^[13]

Histopathologically KFSD is characterized by follicular hyper and parakeratosis, complete degeneration of the inner and outer root sheath and fibrosis of hair follicle. A perifollicular inflammatory cell infiltrate composed of lymphocytes, plasma cells and neutrophils is present along with a mild perivascular lymphocytic infiltrate^[14]

Treatment comprises mainly of keratolytics, emollients and Systemic retinoids such as isotretinoin and etretinate^[15]. Others include tetracyclines, sulfonamides (dapson), macrolides, penicillins, and rifampin. For Hair growth- slight improvement with Minoxidil is reported^[16] For recalcitrant KFSD Hair removal with the long-pulse non-Q-switched ruby laser has been found to be useful^[17]

Our cases presented with multiple follicular dark brown to black, monomorphic, keratotic papules over the scalp, along with scarring alopecia over the scalp and eyebrows with no other cutaneous symptom. The oral cavity, nails, palms, and soles, eyes and laboratory parameters were found to be normal. Maternal Family history was negative. Similar complaints present in both of the male children of the family. With all the above finding, a diagnosis of KFSD was made. Our patients were started on topical Minoxidil 2% solution and are being followed up.

IV. Declaration Of Patient Consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Nil.

CONFLICT OF INTEREST

Nil

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