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Keratosis Follicularis Spinulosa Decalvans with Associated Mental Retardation: Response to Isotretinoin

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Sir,

Keratosis follicularis spinulosa decalvans (KFSD), a rare disorder that was originally described by Siemens, often starts at infancy or early childhood with an X-linked mode of inheritance. Males are predominantly affected. It is characterized by photophobia, corneal dystrophy, widespread follicular hyperkeratosis, scarring alopecia of the scalp, eyebrows, and eyelashes. We report an 18-year-old boy, born of nonconsanguineous marriage, who presented with rough, keratotic papules over the scalp, neck, upper, and lower limbs, along with alopecia since birth. There was sparse growth of hairs over these papules which eventually shed off. There was no history of atopy, ichthyosis, photophobia, hypo or hyperhidrosis, skeletal abnormality, and eye or ear complaints. A family history was not contributory. Physical examination showed multiple follicular dark brown to black, monomorphic, keratotic papules over the scalp, neck, and other areas of the body (upper/lower limb and trunk) along with scarring alopecia over the scalp [Figure 1]. Pubic hairs were normal but hairs over axilla, eyebrows, eyelashes, and beard were sparse. The oral cavity, nails, palms, and soles were found to be normal. Rest of the systemic evaluation revealed no abnormal findings. Hair shaft microscopic examination revealed no abnormality. Neuropsychiatric evaluation revealed a borderline mental retardation (Intelligence quotient-60). Histopathology from a keratotic papule over the scalp showed follicular plugging, basket weave orthokeratosis, mild perivascular inflammatory infiltrate along with perifollicular fibrosis and a small vertical scar [Figure 2]. Magnetic resonance imaging brain did not reveal any abnormal finding. With all the above finding, a diagnosis of KFSD in association with mental retardation was made. Our patient was started on oral isotretinoin (0.5 mg/kg). Following 1 month of therapy, there was marked flattening of the keratotic papules, and significant hair growth was seen by the 3rd month [Figure 3]. He was followed up for the next 6 months.

The locus of mutation of KFSD has been localized to Xp22.[1] The candidate gene suggested is the membrane-bound transcription factor protease site 2 gene which is 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.[2]

The skin manifestations usually appear during the 1st weeks or months of life and are characterized by follicular hyperkeratosis of the skin, especially in the region of the face, associated with scarring alopecia and absence of the follicles of the hair, eyelashes, and eyebrows. Eye symptoms can be associated, the most typical of which are photophobia, keratitis, conjunctivitis, congenital glaucoma, lenticular cataract, and corneal dystrophy.[3] Some patient have associated features including palm plantar keratoderma and the unusual sign of long cuticles.[4] Our patient had associated mental retardation with a subnormal IQ. This is a rare finding associated with KFSD, with only 1 case report previously reported.[5] Other less commonly reported features are atopy, deafness, mental retardation, acne keloidalis nuchae, tufted hair folliculitis, aminoaciduria, and woolly hair.[6,7,8,9]

Treatment of KFSD is usually unsatisfactory. Topical treatment comprises mainly of keratolytics and emollients. Systemic retinoids such as isotretinoin and etretinate are beneficial in the early stages of the disease as they downregulate follicular hyperkeratosis and inflammation.[10] The condition is found to recur after stopping the treatment; however, our patient showed no recurrence in the 6 months of follow-up. Other treatment options such as tetracyclines, sulfonamides (dapsone), macrolides, penicillins, and rifampin have been found to be ineffective.[11] Topical and intralesional corticosteroids were tried but cause only transient improvement. Laser assisted hair removal with the long-pulse non-Q-switched ruby laser has been found to be useful in progressive or recalcitrant KFSD.[12]

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Conflicts of interest

There are no conflicts of interest.

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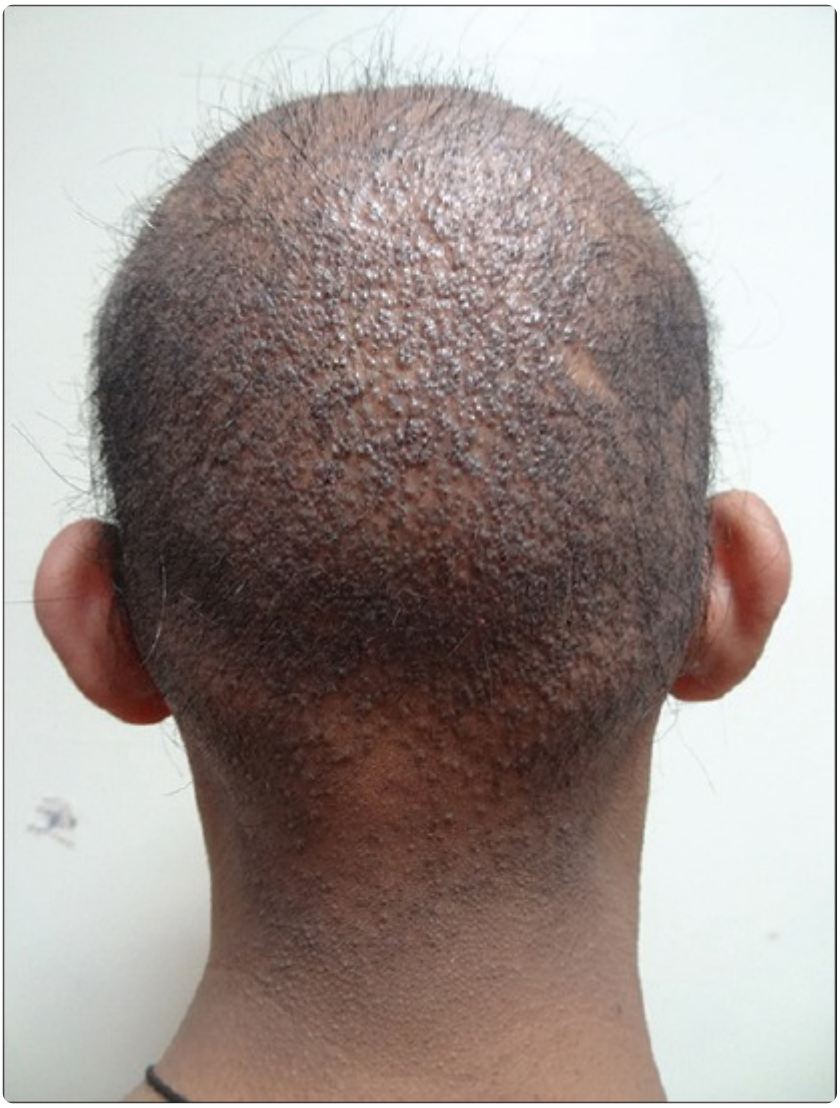
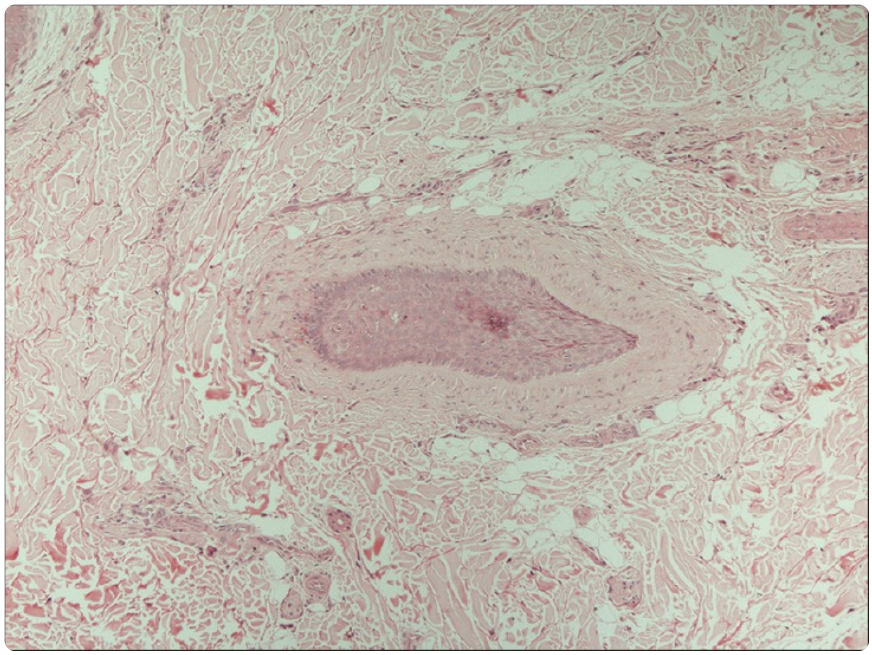
Figures and Tables

Figure 1



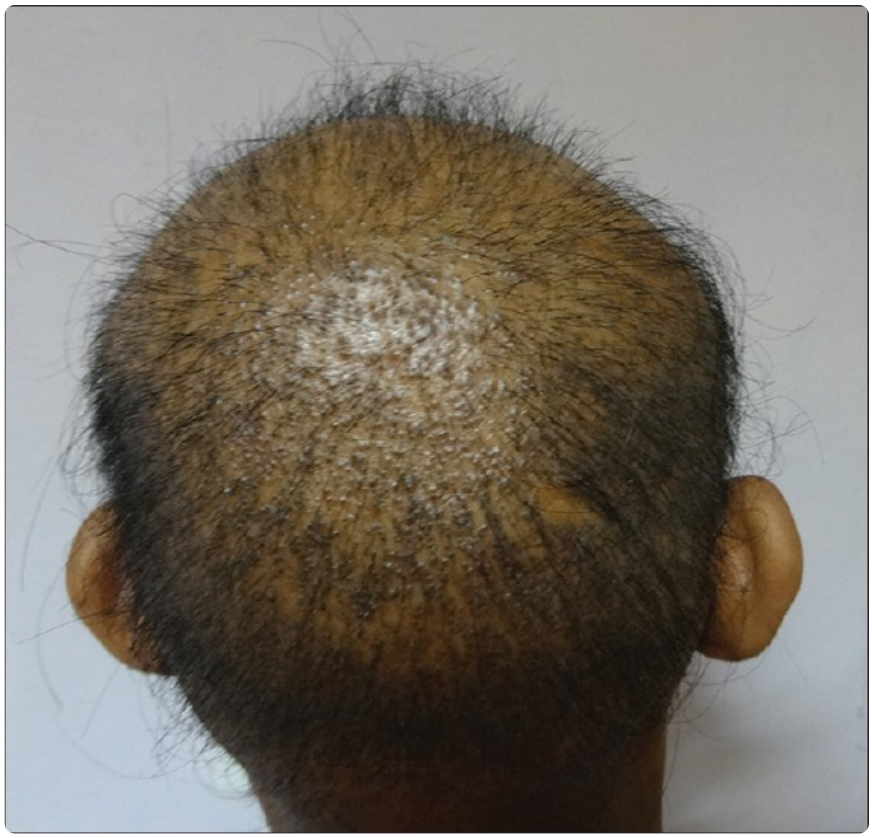
Keratotic papules with sparse hair over the scalp

Figure 2



Follicular plugging, perifollicular fibrosis, and vertical scar (H and E, ×40)

Figure 3



Significant hair growth seen after 3 months of Isotretinoin