

Case Report

Overlapping syndromes: keratosis follicularis spinulosa decalvans with overlapping features of ichthyosis follicularis with alopecia and photophobia

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Abstract We report a case of a 20-year-old male who presented in our dermatology clinic with congenital alopecia totalis, ectropion, bilateral cataract, follicular prominence, plantar keratoderma, ichthyosis on legs, cheilitis, xerosis and photophobia. He had overlapping features of both keratosis follicularis spinulosa decalvans and ichthyosis follicularis with alopecia and photophobia. Or was it a different entity altogether?

Key words

Keratosis follicularis spinulosa decalvans, ichthyosis follicularis, alopecia, photophobia.

Introduction

Keratosis follicularis spinulosa decalvans is a rare disorder that was originally described by Siemens.¹ It is characterized usually in childhood by photophobia, then by corneal dystrophy, widespread follicular hyperkeratosis and scarring alopecia of scalp, eyebrows and eyelashes.

On the other hand the syndrome of ichthyosis follicularis with alopecia and photophobia, a rare ectodermal disorder, first reported by Macleod in 1906,⁴ is characterized by non-inflammatory follicular keratosis, persistent non-cicatricial scalp and body alopecia and severe photophobia since birth. Generalized

xerosis, cheilitis and pruritus are common.

Our patient had partial features of both the syndromes that is why we report this case.

Case report

The patient, a 20-year-old male, presented in the dermatology clinic of the Pakistan Institute of Medical Sciences, Islamabad with the complaints of thickening of the skin of soles, rough dry skin and generalized pruritus since very young age. He had congenital absence of body hair including scalp hair, eyebrows and eyelashes. He also noticed roughness of the skin on his arms and thighs.

He did not have any complaints regarding any other systems. He could go about his daily routine activities. He complained that photophobia had bothered him more when

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Figure 1 Ichthyosis on patient's leg.



Figure 2 Ectropion and blepharitis visible in patient's eye.



Figure 4 Patient's scalp showing alopecia totalis.



Figure 3 Accessory tragus.



Figure 5 Plantar keratoderma.

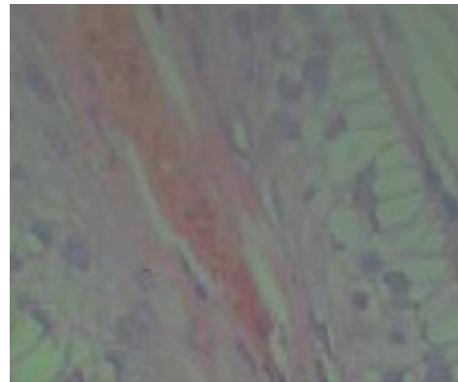


Figure 6 Keratin filled follicles.

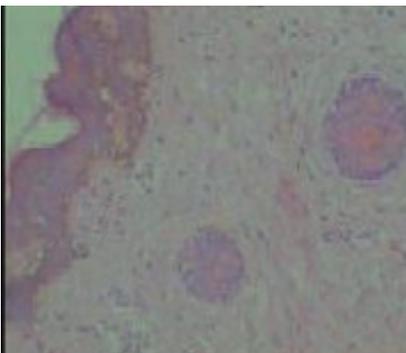
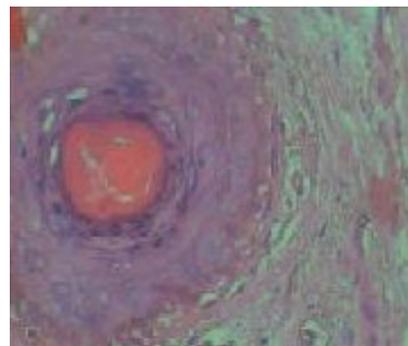


Figure 7 Rudimentary hair follicles in dermis.



A rudimentary hair follicles in dermis under higher magnification.

he was younger but it was not a major complaint at present. Enquiry about family history revealed that he had 4 brothers and 3 sisters and that a 7-year-old brother had the same features as he did. His parents, who were first cousins, were normal, alive and healthy.

On examination patient was a young male with alopecia universalis, follicular prominence, palmoplantar keratoderma, nail dystrophy, ichthyosis on legs, angular cheilitis, xerosis and photophobia (**Figures 1-5**). He had an accessory tragus on the left side. Eye examination revealed diminished visual acuity with myopia, bilateral cataract and ectropion.

Skin biopsy was taken from front of chest and sent for horizontal and vertical sectioning. Histological findings were that the epidermis showed slight hyperkeratosis, dipping, irregular surface, slight acanthosis, and hyperpigmentation while the dermis showed only a few hair follicles which were devoid of hair and keratin filled (**Figures 6 and 7**). A rudimentary hair follicle was also seen (**Figure 8**). There was fibrosis, edema, mucinosis and pigmentary incontinence. Granulation tissue was also seen with mild perivascular chronic inflammation. There was only one erector pilorii muscle showing edema. Sebaceous glands were absent.

Discussion

Many sporadic cases of keratosis follicularis spinulosa decalvans have been reported. It has been defined as a hereditary condition of sex linked recessive inheritance mapped to a locus at Xp21.13-p22.2.¹ Female

heterozygotes may exhibit a milder phenotype as reported by Hollowood *et al.*³

Ichthyosis follicularis with alopecia and photophobia is much rarer for which an X-linked recessive inheritance was proposed by Macleod⁴ when he first reported the syndrome in 1909 but later this pattern was evident in an affected Japanese mother and daughter,⁵ suggesting autosomal dominant transmission. Genetic mapping has not yet been reported.

In our patient the fact that no relation in previous generations is affected suggests sporadic mutation in the mother who could have transmitted the disease to her sons and only two sons are affected, further suggesting an X-linked recessive inheritance.

Keratosis follicularis spinulosa decalvans has been characterized by progressive scarring, alopecia, following variable degrees of inflammatory changes while in ichthyosis follicularis with alopecia and photophobia there is generalized non-inflammatory follicular keratosis and persistent non-cicatricial scalp and body alopecia but with severe photophobia. Generalized xerosis, cheilitis and pruritus as well as the nail changes as present in our patient are features of ichthyosis follicularis with alopecia and photophobia but have not been reported in keratosis follicularis spinulosa decalvans.

The ocular signs including blepharitis, ectropion and corneal dystrophy are all part of keratosis follicularis spinulosa decalvans but not of ichthyosis follicularis with alopecia and photophobia.

Eramo *et al.*⁶ in 1985 reported that cutaneous and ocular features of keratosis follicularis spinulosa decalvans may overlap with ichthyosis follicularis and photophobia in which palmoplantar involvement is absent.

Rand and Baden⁷ in 1983 suggested that keratosis follicularis spinulosa decalvans and ichthyosis follicularis are the terms used to describe more extensive involvement of the same process as occurs in keratosis pilaris atrophicans fasciei where it is more or less confined to the eyebrows. The classification of these disorders into distinct clinical entities is difficult and controversial as the cases are rare and always in a way unique.

We feel that our patient's features fit in an overlapping manner in both keratosis follicularis spinulosa decalvans and ichthyosis follicularis with alopecia and photophobia or it can be a distinct entity altogether which can only be ascertained by gene studies and genetic mapping.

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