**INTRODUCTION**

Ichthyosis follicularis, alopecia, and photophobia (IFAP) syndrome is a rare genetic, oculocutaneous disorder. The term ichthyosis follicularis was coined by Lesser in 1885 to describe the thin, filiform projections protruding from the hair follicles of a patient who also had alopecia. The association between ichthyosis follicularis, alopecia, and photophobia was first reported by MacLeod in 1909 as a syndrome. This disorder results from mutations in the membrane-bound transcription factor protease site 2 gene that impairs cholesterol homeostasis and the ability to cope with endoplasmic reticulum stress. We report this rare case of IFAP with atypical presentation and it was interesting to note that alopecia in this child was confined to eyebrows; this unique presentation has not been described earlier. This rare oculocutaneous disorder requires proper documentation so that identification of its variants may be possible in the future.

**CASE REPORT**

A 13-year-old male child came to the dermatology outpatient department with complaints of multiple raised lesions over face and body since 3 months of age and loss of eyebrows since his 1st year. History of photophobia and recurrent respiratory tract infection was present. The child was born of a nonconsanguineous marriage, full-term normal vaginal delivery and immunized till date. There was no history of loss of the hair over the scalp, no history of any delayed milestones, no history of similar complaints in the family [Figure 1], and no history of atopy. On examination, multiple, pinpoint, skin-colored papules seen over the forehead, bilateral cheeks, chin, neck, chest, abdomen, back, bilateral arm, bilateral forearm, bilateral upper thighs, and gluteal region [Figure 2]. Madarosis was present [Figure 3]. Palms, soles, oral cavity, dentition, nails, and genitalia appeared normal. Ophthalmic examination revealed bilateral posterior blepharitis and photophobia. Routine hematological examinations revealed elevated eosinophil count, other investigations were normal. Skin biopsy of the spiny papules revealed features suggestive of keratosis pilaris [Figure 4]. Hair shaft examination was normal, dermoscopy over the eyebrows and lesions...
over trunk revealed keratinous plugs in the follicular orifices. Neurological examination was normal. Mutation analysis was not done due to financial constraints in the family. Diagnosis of IFAP syndrome was made based on characteristic cutaneous features, alopecia, photophobia, and histopathology report.

**DISCUSSION**

The IFAP syndrome is a rare genetic disorder characterized by the triad of generalized follicular hyperkeratosis, alopecia, and photophobia. An X-linked recessive pattern of inheritance has been established for IFAP. Therefore, the risk for a female carrier to have an affected son is 50%. The mutation might also arise in the patient de novo. This disorder has also been reported occurring simultaneously in a mother and daughter suggesting an autosomal dominant mode of transmission.

The generalized cutaneous “thorn-like” projections impart an unusual sensation on palpation resembling a “nutmeg grater” or “the prickly surface of a rose leaf.” Alopecia is not associated with inflammation or scarring and in majority of cases and it is confined to the scalp. Eyebrows and eyelashes are also involved in many reports, but involvement of eyebrows alone is unique feature in our patient. Psoriasiform plaques, atopic manifestations, cheilitis, hypohidrosis, dystrophic nails, and inguinal hernia may possibly occur in up to 40% of patients. Photophobia, corneal ulcerations, corneal vascularization, cataract, astigmatism, and loss of vision have been reported. Intellectual disability, hypotonia, seizures, short stature, frontal bossing, choanal atresia, large ears, recurrent infections, and intestinal anomalies may also be present.

The histopathology of this patient showed that hyperkeratosis, acanthosis, mild spongiosis, distended follicular orifice containing horny plug seen extending above the skin surface. Lymphocytic infiltration around the blood vessels of adnexal structure is also a common feature. They were identical to those described in previous cases suggesting features of keratosis pilaris which support the clinical diagnosis of IFAP. IFAP syndrome should be differentiated from keratosis follicularis spinulosa decalvans (KFSD) [Table 1]. KFSD is characterized by scarring alopecia in contrast to IFAP.
Cutaneous lesions usually respond to topical keratolytics, urea containing preparations, and emollients. In few patients, a moderate response to acitretin therapy has been reported. Adequate lubrication of the ocular surface is necessary. Life expectancy in patients with IFAP syndrome can vary from death in the neonatal period to normal survival. Genetic counseling should be given as the syndrome is associated with X-linked recessive or autosomal dominant mode of inheritance.

CONCLUSION

It was interesting to note that alopecia in this child was confined to the eyebrows; this unique presentation has not been described earlier. This rare oculocutaneous disorder requires proper documentation so that identification of its variants may be possible in the future.

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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Conflicts of interest

There are no conflicts of interest.

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Table 1: Comparison between ichthyosis follicularis, alopecia, and photophobia syndrome and keratosis follicularis spinulosa decalvans

| Disease                  | IFAP | KFSD               |
|--------------------------|------|--------------------|
| Cutaneous manifestation  | Spiny follicular papules | Spiny follicular papules |
| Erythema                 | Absent | Present over eyebrows |
| Alopecia                 | Nonscarring | Scarring |
| Ocular manifestation     | Corneal ulceration, conjunctivitis may be present | Corneal dystrophy may occur |
| Photophobia              | Present | Present |
| Plantar hyperkeratosis   | Absent | May be present |

IFAP – Ichthyosis follicularis, alopecia, and photophobia syndrome; KFSD – Keratosis follicularis spinulosa decalvans