Our Dermatology Online

Case Report

# Ichthyosis follicularis, alopecia, and photophobia syndrome: A case report

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## ABSTRACT

Ichthyosis Follicularis, Alopecia, and Photophobia Syndrome (IFAP syndrome) is a rare X-linked genetic disease. It is characterized by the triad of hyperkeratotic follicular papules, total to subtotal alopecia, and photophobia with varying severity. IFAP syndrome results from mutations in the gene encoding the membrane-bound transcription factor peptidase, site 2 (*MBTPS2*). Histology is non specific. Follicular hyperkeratotic papules can be treated using topical keratolytics and emollients or in severe cases retinoids. The prognosis dependS of the extracutaneous manifestations mainly heart and lungs disease.

**Key words:** Alopecia; Icthyosis; Pediatric

## INTRODUCTION

The ichthyosis follicularis, atrichia, and photophobia (IFAP) syndrome is a rare genodermatosis first described by MacLeod in 1909 [1]. The X-linked recessive mode of inheritance was the first reported and the gene mapped to the 5.4Mb region between DXS989 and DXS8019 on Xp22.11-p22.13 [2]. Recently, the genetic basis of IFAP syndrome has been confirmed to be associated with mutations of the *MBTPS2* (membrane-bound transcription factor protease, site 2) which codes for an intramembrane zinc metalloprotease [3]. Its mode of inheritance is X-linked recessive, thus mostly affecting males. Affected or carrier females may display some of its clinical features [4].

## CASE REPORT

A 05-year-old child from a non-consanguineous marriage presented for consultation for a congenital alopecia of the eyebrows. He also complained of slightly pruritic skin lesions evolving for 02 years and a slight photophobia. He had a history of psychomotor retardation, as well as cryptorchidism operated on with discovery on preoperative transthoracic ultrasound of pulmonary stenosis. On admission, he was found to have short stature and low body weight. The dermatological examination found non-scarring alopecia of the eyebrows with respect of the eyelashes and the hair (Fig. 1). It also showed generalized dry skin with presence of multiple keratotic follicular papules at the level of the trunk and the extension face of the limbs with the appearance of sandpaper (Fig. 2). Teeth and nail exam were normal. Cardio pulmonary examination found no abnormalities. a Ophthalmologic examination revealed photophobia while cornea and ocular fundus were normal. Biopsy of a follicular papule showed dilation of the follicular ostuim by a keratotic plug (Figs. 3 and 4). In the light of these clinical and histological data, we retained the diagnosis of IFAP syndrome in our patient. He was put under keratolytics and emollients with improvement of the keratotic papules and xerosis.

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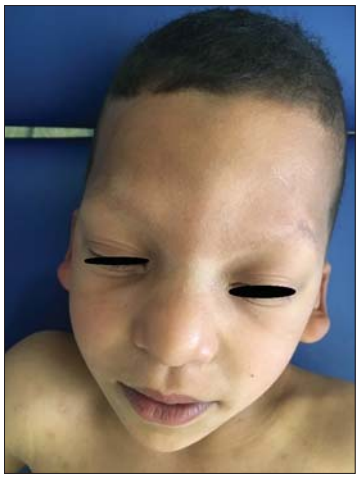

**Figure 1:** Non scarring alopecia of the eyebrows with respect of the eyelashes and the hair.

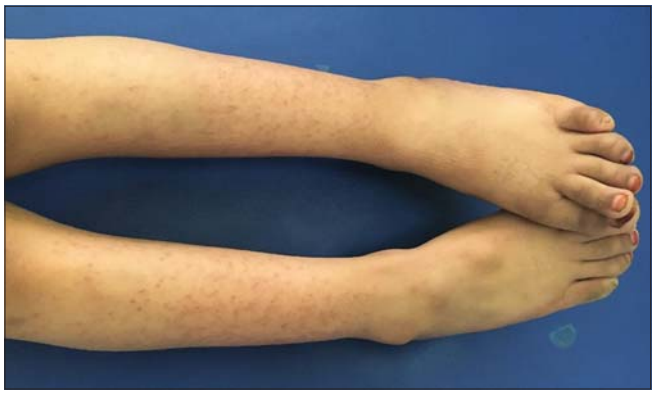

**Figure 2:** Widespread keratotic follicular papules on the extensor surface of the limbs.

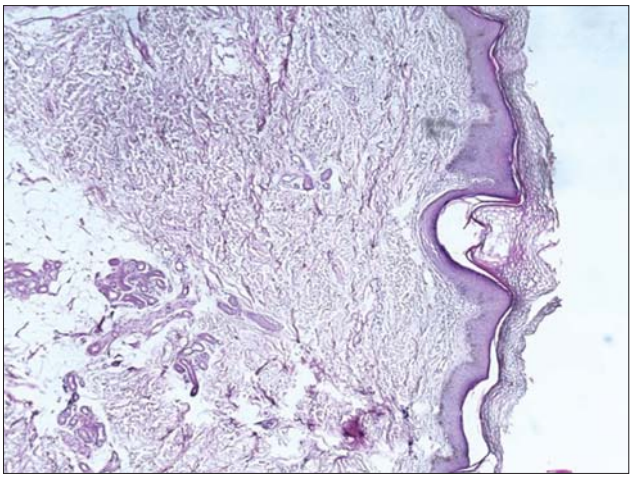

**Figure 3:** Ostial dilation by keratotic plug without hair shaft.

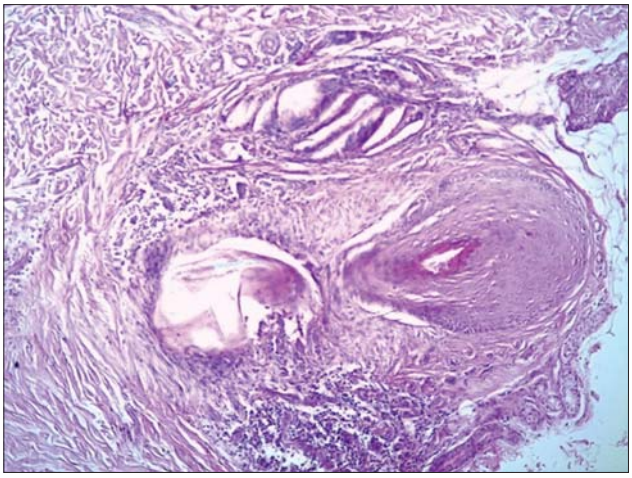

**Figure 4:** Macrophagic granuloma around hair shaft debris.

## DISCUSSION

Ichthyosis follicularis is the most common cutaneous manifestation of the IFAP syndrome. It presents with noninflammatory follicular keratotic papules, mainly involving the scalp and extensor extremities giving the sandpaper texture. Congenital non-cicatricial alopecia involves the scalp, eyebrows, and eyelashes. Most of the cases have complete body hair loss, while some others have thin and sparse hair. Other cutaneous findings include psoriasiform plaques, lamellar scaling, angular cheilitis, periungual inflammation, and nail dystrophy [3,4]. Photophobia may exist since birth or may develop later in childhood [5]. It is believed that a defect in Bowman membrane results in superficial corneal ulceration and progressive corneal scarring . Vision acuity is generally low [6]. The most frequent neurological features in IFAP are intellectual disability, and seizures [7]. The diagnosis of the IFAP syndrome is based on the clinical features and on the presence of a mutation in the MBTPS2 gene. Skin histopathology is non-specific and consists of dilated hair follicles with keratin plugs extending above the surface of the skin, decreased or absent sebaceous glands and normal sweat glands [4]. A moderate response to acitretin therapy at a dose of 0.3 to 1 mg/Kg/day with improvement in cutaneous features and corneal erosions but no changes regarding alopecia and photophobia have been noted in some patients. Follicular hyperkeratosis can be treated using topical keratolytics, urea preparations, and emollients [4]. Life expectancy in patients with IFAP syndrome can vary from death in the neonatal period to normal survival. Cardiopulmonary complications remain the major cause of death [8].

## CONCLUSION

Our observation reports a rather particular variety of ichthyosis. The relative rarity of this syndrome and the inaccessibility of genetic analysis make diagnosis difficult.

## Consent

The examination of the patient was conducted according to the principles of the Declaration of Helsinki.

The authors certify that they have obtained all appropriate patient consent forms, in which the patients gave their consent for images and other clinical information to be included in the journal. The patients understand that their names and initials will not be published and due effort will be made to conceal their identity, but that anonymity cannot be guaranteed.

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