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PHOTOLETTER TO THE EDITOR

A new variant of ichthyosis follicularis with alopecia and photophobia (IFAP) syndrome with coexisting psoriasiform lesions and palmoplantar keratoderma. IFAP-PPK syndrome?

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Abstract

IFAP is an acronym for a rare congenital ectodermal disorder characterized by **i**chthyosis **f**ollicularis, **a**lopecia and **p**hotophobia. A recessive X-linked mode of inheritance was initially proposed but recent reports in girls suggested genetic heterogeneity of this syndrome. We herein describe a 1-year-old boy with clinical features typical of IFAP syndrome plus psoriasis-like lesions and palmoplantar keratoderma (PPK).

IFAP syndrome is a distinctive clinical entity defined by the presence of ichthyosis follicularis, alopecia and photophobia. A recessive X-linked mode of inheritance was initially suggested but more recently an autosomal pattern has been proposed.[1] Although the pathogenesis of this disorder remains to be unraveled, a better understanding can be acquired, as more cases are reported in the medical literature.

A 1-year-old boy presented in referral dermatology clinic in Sana'a, Yemen, with dry rough skin, photophobia and total alopecia involving the eyebrows and eyelashes since birth (Fig. 1). He was born as a full-term of normal non-consanguineous parents and had normal growth and developmental milestones. Cutaneous examination revealed hyperkeratotic spinous papules on the scalp, trunk and extremities with thorn-like sensation on palpation (Fig. 1-3). The skin as a whole was dry and rough as in ichthyosis. Non-scarring universal alopecia (Fig. 1) and palmoplantar keratoderma (PPK) (Fig. 4) were also seen.

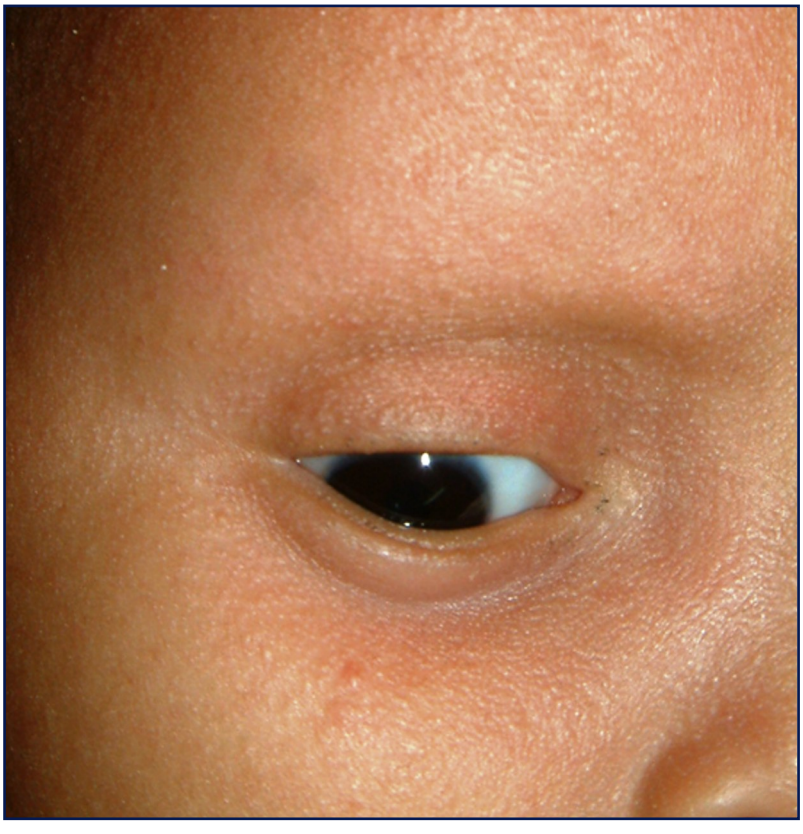

Figure 1

Total atrichia, with thorn-like projections over right temporal area.

Additional features observed include, psoriasiform lesions over the left side of the abdomen (Fig. 5, 6), and atrophic scars over the right side of the abdomen and both arms (Fig. 1, 4, 5). There was a history of photophobia since birth described as irritation, redness and watering of the eyes when exposed to sunlight. No teeth or nails changes were noted. There was no history of any neurological or hearing deficit, sweating defect, collodion membrane, ectropion or eclabium. No similar features were found in other family members.

Patients with IFAP have striking alopecia, photophobia and generalized cutaneous "thorn-like" projections that have been described as "nutmeg grater" or "the prickly surface of a rose leaf".[2] Hyperkeratosis is sometimes present over the elbows and knees. In 2005, Rai and Shenoi reported a case of a 23-year-old man who had PPK with ichthyosis follicularis, alopecia and twenty-nail dystrophy. Nonetheless, their patient did not have typical features of IFAP syndrome because of the absence of photophobia.[2]

Males with IFAP syndrome have an inexorable progression of corneal vascularization and loss of vision. Retinal vascular tortuosity may be a clinical sign of carrier status in females.[3]

IFAP syndrome results from congenital absence of or marked defect in the pilosebaceous apparatus. The follicular hyperkeratosis is believed to be secondary to the obstruction of sebum. Recently, it was shown that IFAP syndrome is caused by functional deficiency of membrane-bound transcription factor protease, site 2 (MBTPS2), a membrane-embedded zinc metalloprotease that activates signaling proteins involved in sterol control of transcription and endoplasmic reticulum stress response.[1]

Recessive X-linked pattern of inheritance was proposed in IFAP syndrome. However, the condition has recently been reported in females, suggesting autosomal inheritance or genetic heterogeneity of the syndrome. Our patient's family history was unremarkable and examination of other family members was not contributory, suggesting that the manifestation of IFAP in this case most likely resulted from a new mutation.

Mégarbané *et al*[4] reported two brothers with IFAP syndrome with features of hyperkeratotic psoriasiform lesion, like in our case. However, our patient has additional features of PPK, hence we are proposing the term IFAP-PPK syndrome.

The mainstay treatment of IFAP syndrome is symptomatic by frequent and regular use of emollients. No studies have been conducted to evaluate the efficacy of other topical treatments, probably because of the rarity of the condition. Acitretin has been described to improve cutaneous features and corneal erosions in one case.[5]

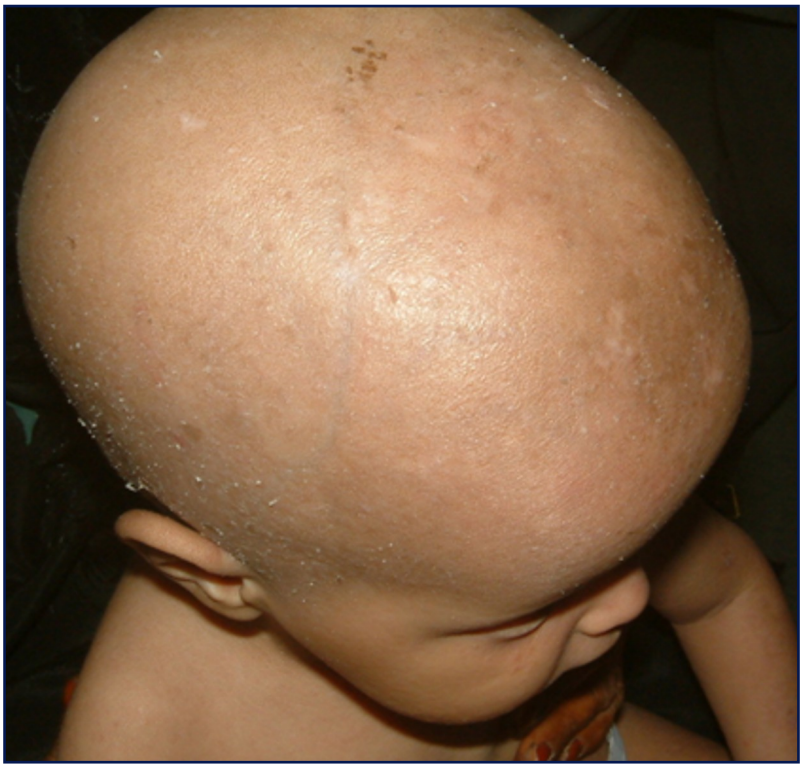

Figure 2

Thorn-like projections over the scalp, with loss of scalp hair.

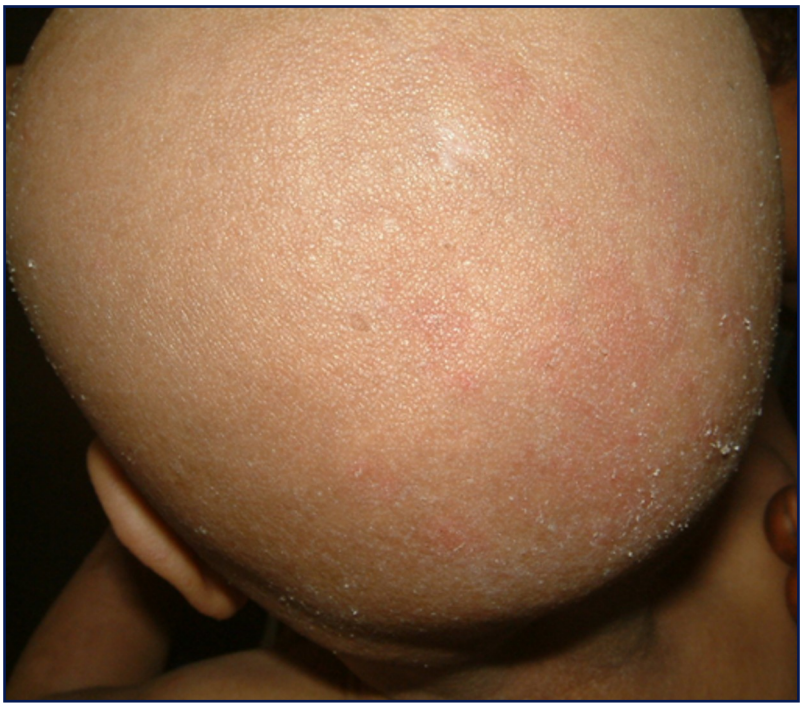

Figure 3

Close up view of the thorn like projections over the scalp.

Ophthalmic consultation is necessary for appropriate intervention for any associated eye problems.

In conclusion, IFAP syndrome in our patient is most likely caused from a new mutation. Our case adds to the literature the association of PPK with classical IFAP syndrome and by this we are suggesting this entity as IFAP-PPK syndrome. We are hoping this report increases clinicians' awareness of this rare congenital syndrome and promotes a broader knowledge of possible associations that may coexist with it.

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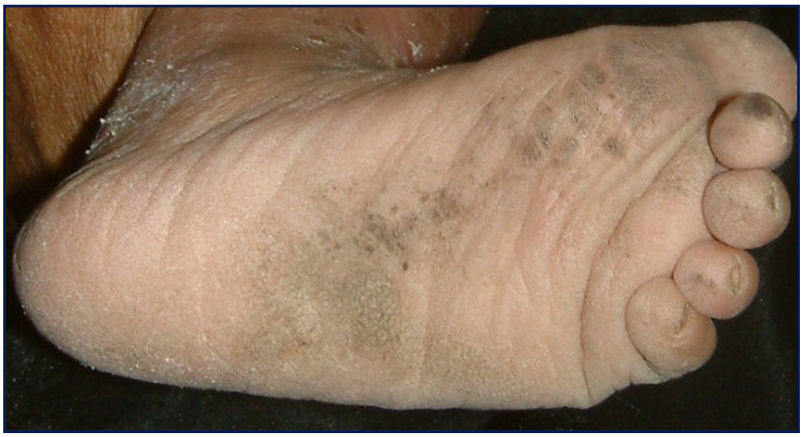

Figure 4

Brown warty hyperkeratosis over the sole and psoriasiform hyperkeratosis over the ankle.

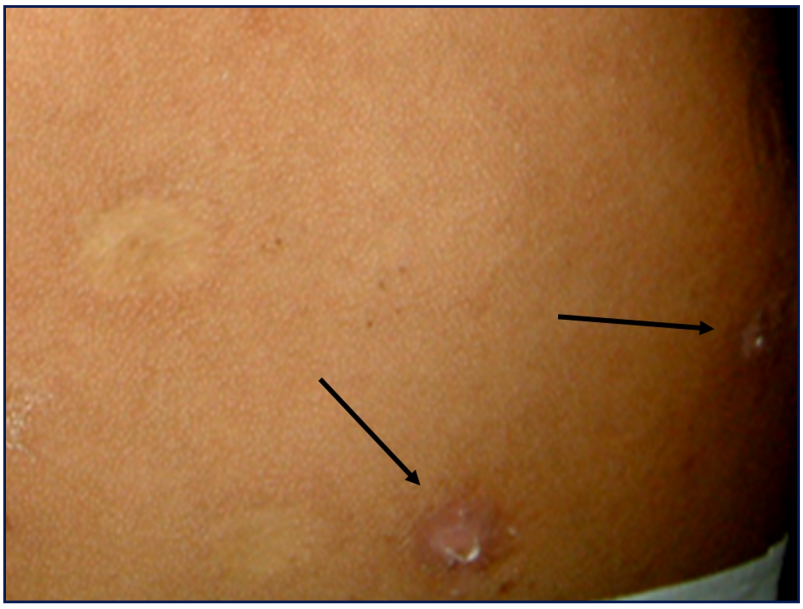

Figure 5

Psoriasiform plaques over left side (arrow) and atrophic scars over right side of abdomen and right arm.

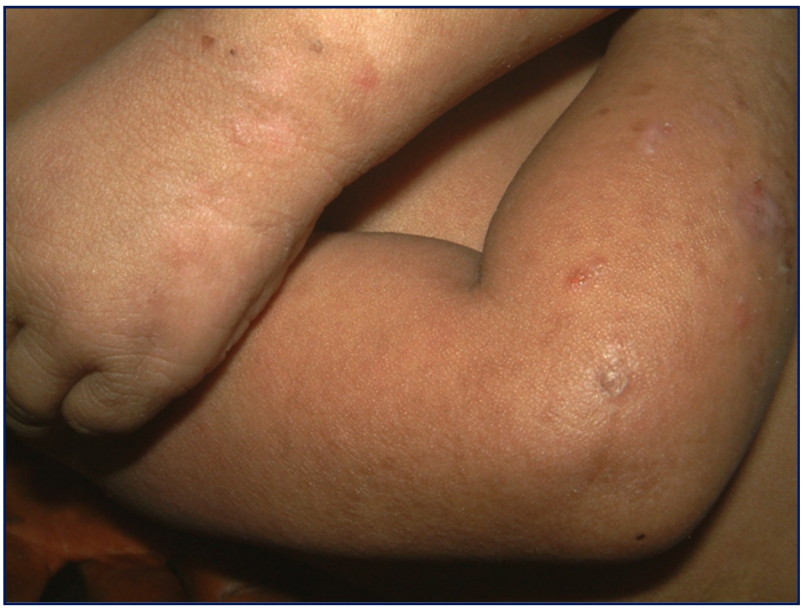

Figure 6

Erosions, excoriations and atrophic scars over right arm and left hand, due to repeated scratching.