Meibomian gland dysfunction in a case of ichthyosis follicularis with alopecia and photophobia syndrome

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We are reporting an interesting case of ichthyosis follicularis with alopecia and photophobia syndrome in a daughter and father from the Indian subcontinent associated with Meibomian gland dysfunction. A three-year-old female child presented with pain, redness and severe photophobia in both eyes since one month.

Cutaneous examination revealed ichthyosis, absence of hair all over the body including eyebrows, scalp and angular cheilosis. Ocular examination revealed bilateral severe meibomianitis, multiple superficial punctate keratitis in right eye and corneal epithelial defect in the left eye. Other systemic features were prominent high forehead and large ears. Her father had similar symptoms. Her symptoms improved after adequate treatment of meibomian gland dysfunction. She is asymptomatic at present.

Key words: Ichthyosis follicularis, alopecia and photophobia syndrome, meibomian gland dysfunction, ocular involvement

Ichthyosis refers to a relatively uncommon group of skin disorders characterized by the presence of excessive amounts of dry surface scales. It is regarded as a disorder of keratinization or cornification and it is due to abnormal epidermal differentiation or metabolism. The syndrome of ichthyosis follicularis with alopecia and photophobia (IFAP) syndrome was first described in 1909 by MacLeod 1909 in three boys.[1] Recently, we observed a daughter and father with this syndrome associated with severe Meibomian gland dysfunction (MGD) in the eyes.

Case Report

A three-year-old child presented with complaints of pain, watering and severe photophobia in both eyes since one month. There was history of absence of hair all over her body including eyebrows and eyelashes since birth. There was dryness, itching and scaling of skin since birth, and recurrent self-resolving angular cheilosis. There was no history of delayed milestones, deafness, diarrhea and seizures. There was no history of consanguinity in the family. Vision could not be recorded at presentation due to severe photophobia.

Ocular examination revealed bilateral severe meibomianitis, multiple superficial punctate keratitis in the right eye and a large corneal epithelial defect (7 mm × 7 mm) in the left eye [Fig. 1a-c]. Fundus in right eye was within normal limits and in the left eye was not visible. Cutaneous examination revealed non-cicatricial complete body alopecia with severe angular cheilosis. The palms and soles were unaffected [Fig. 2a and b].

She was given systemic erythromycin for two weeks. Hot fomentation of eyelids, massage of lids with tetracycline eye ointment, liberal lubrication and low-dose steroid fluorometholone BID was advised for two weeks. Antibiotic eye drop tobramycin QID was added in the left eye. One week after this treatment she had improved symptomatically. The epithelial defect in the left eye had healed with minimal scarring and the punctate keratitis resolved over 4 weeks. Visual acuity at final follow-up was 20/20 in right eye and 20/60 in left eye. Meibomian gland treatment was continued. She is asymptomatic at present.

The father had all the systemic features as the daughter [Fig. 3]. Ocular examination revealed bilateral severe meibomianitis. He was started on similar treatment as his daughter.

Discussion

Generalized ichthyosis and alopecia have been reported in very few syndromes. These are ablepharon ichthyosis, alopecia ichthyosis and pseudo-hermaphrodites, alopecia – skeletal abnormalities – mental retardation, deafness ichthyosis, dermotrichic syndrome, ectodermal dysplasia – alopecia – mental retardation syndrome, Hayden syndrome, hereditary mucop epithelial syndrome, IFAP syndrome, ichthyosis deafness, Hirschprung disease, ichthyosis hypertrichosis hyperhidrosis syndrome, KIDS syndrome, keratosis follicularis spinosid decalvans, Sjogren – Larson syndrome, triculodermovertebral syndrome and woodhouse syndrome. Out of these conditions, ocular involvement is primarily seen in ablepharon ichthyosis syndrome, KIDS syndrome, IFAP syndrome and keratosis follicularis spinosid decalvans syndrome. In patients with KID syndrome, nails are often dystrophic, teeth may be small or malformed and ocular changes are usually observed during the second or third decade. In addition, there is a congenital hearing loss, palmoplantar hyperkeratosis with leather grain-like keratoderma.[3] Our patient was a three-year-old female. She did not have deafness, ablepharon or patchy alopecia. She had severe photophobia, non-scarring ichthyosis, total alopecia, high forehead, large ears and angular stomatitis – all features that has been found in cases of IFAP syndromes reported till date.

Forty cases of IFAP syndrome have been reported worldwide with different clinical manifestations.[2] An interesting feature of this syndrome is that patients bear a striking resemblance to each other probably due to the loss of hair and photophobia.[2]

Ocular abnormalities are important for the diagnosis of IFAP syndrome. The ocular features that have been reported up till now are punctate keratopathy, erosion, corneal scarring, atopic keratoconjunctival inflammation, horizontal nystagmus, myopia and absence of eyebrows and eyelashes.[2] The presence
of MGD as the cause of tear film instability has not been noted in the previously reported cases. Embryologically, the Meibomian glands are formed by the ingrowth of the basal epithelial cells at the inner margin and the sweat glands of Moll that arise from the wall of the hair sacs. Being ectodermal in origin these are likely to be affected. Photophobia and punctate keratitis may be due to secondary to tear film instability caused by MGD. Kamo et al. have highlighted the pilosebaceous anomaly in a three-year-old boy with IFAP syndrome. In our patient, both photophobia and punctate erosions resolved with treatment of MGD. Adequate treatment of MGD helped in preventing the recurrent attacks of inflammation and scarring.
in our patient.

Mode of inheritance in IFAP syndrome is not definitely determined.[6] Most of the reported patients were males, and in some families milder effects were noted in possible transmitting female. Recently, a mother and daughter, and two unrelated female patients, with an IFAP syndrome have been reported. However, there are no reports till now, which shows a father to daughter transmission like in our case, which may throw some light to the pattern of inheritance in this disease. The daughter had more severe disease as compared to the father indicating the possibility of variable penetration of the genetic abnormality. In our case it seemed likely that the inheritance was X-linked dominant with variable penetrance.

In conclusion, IFAP syndrome is a rare ectodermal developmental disorder with variable manifestation. A father and daughter with this syndrome are described in this report. Ocular manifestations are an important part of this disease spectrum. MGD may be one of the contributing factors for photophobia seen in this syndrome.

References