Clinical Images

A rare case of ichthyosis follicularis, alopecia & photophobia syndrome

Fig. 1 (A). Lower limb photograph showing dyskeratotic papules (arrow) more pronounced over extensor extremities and distributed symmetrically with sparing of soles. (B). Photograph of the right side of face showing hyperkeratosis of skin over ear lobe (arrow), (C). Madarosis (arrow) and angular chelosis (arrow). (D). Loss of scalp hair.

Fig. 2. Right eye photograph showing corneal infiltration (arrow) and vascularization (arrow). Other eye had similar features.

Fig. 1 (A, B and C were taken when the child was 3 year old and Fig. 1 D and 2 at age 7 years).

A 3 year old boy presented to Sri Sankaradeva Nethralaya, Guwahati, Assam, India, in February 2008 with complaint of photophobia since past six months. He had widespread non-inflammatory exuberant follicular projections over the skin, more pronounced over the extensor extremities, with sparing of palms and soles (Fig. 1A). He had hyperkeratosis of skin over the ears (Fig. 1B), madarosis (Fig. 1C), scanty eyelashes (Fig. 1C) hyperkeratosis of lid margin with folliculosis, conjunctival congestion and follicular reaction. The child had alopecia (Fig. 1D). Corneal infiltration was present at the level of epithelium and sub-epithelium (Fig. 2). On the basis of above features, he was diagnosed of ichthyosis follicularis alopecia and photophobia (IFAP) syndrome¹ that is caused by mutation in MBTPS2 gene on chromosome Xp22². On a follow up at seven years of age, superficial (terminal loop type) and stromal corneal neovascularization was noted (Fig. 2). Symptomatic relief was obtained with dark goggles/glasses and lubricating eye drops.

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References
