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A Rare Atypical Presentation of Iichthyosis Follicularis, Alopecia, and Photophobia Syndrome

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Abstract : Ichthyosis follicularis, alopecia, and photophobia (IFAP) syndrome is a rare oculocutaneous disorder of genetic origin. This disorder results from mutations in the membrane-bound transcription factor protease site, two genes that impair cholesterol homeostasis, and the ability to cope with endoplasmic reticulum stress. We report a rare case of IFAP syndrome with an atypical presentation, and it was interesting to note that the child had patchy non-scarring alopecia over the scalp along with unilateral madarosis. To our best knowledge, this unique presentation has not been described earlier. The child presented with photophobia and unilateral ptosis. The child also had short stature and intellectual disability. Skin histopathology was nonspecific and consisted of dilated hair follicles with keratin plugs extending above the skin surface. This rare oculocutaneous disorder requires proper documentation so that identification of its variants may be possible in the future. Early recognition of atypical presentations can help in preventing cardiovascular complications, which remain the major cause of death.

Keywords : alopecia, photophobia, ichthyosis follicularis, IFAP syndrome
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