

## Urbach-Wiethe Disease

A 6-year-old boy presented with complaints of hoarseness of voice since infancy and multiple scarred lesions over face for past three years. There was a preceding history of recurrent mildly pruritic papulo-vesicular eruptions over face and distal extremities. The child was born of non-consanguineous marriage and no other family members were affected. There were no history of seizures or photosensitivity, and developmental milestones were normal.

On physical examination, multiple pock-like atrophic scarring along with few waxy papules were seen on face (*Fig. 1*) and dorsum of both hands. Lower lip was enlarged and protuberant. The margins of both upper and lower eyelids were beaded with yellowish waxy papules (moniliform blepharitis or string of beads). Loss of eyelashes was also apparent on inner side of right lower eyelid. Histopathologic examination of skin lesions revealed deposition of periodic acid-Schiff (PAS)-positive eosinophilic hyaline materials within the dermis along with adnexal atrophy and onion skinning of blood vessels. All these findings were suggestive of Urbach-Weithe disease.

Urbach-Weithe disease, also known as lipid proteinosis, is an uncommon autosomal recessive disorder caused by loss of function mutation in *extracellular matrix protein-1* gene leading to aberrant deposition of hyaline materials (lipid-protein complexes) within skin, oral mucosa, vocal cords and occasionally intraocular structures. The differential diagnoses are erythropoietic protoporphyria, amyloidosis and papular mucinosis. Absence of photosensitivity, urine and stool tests for porphyrins and absence of mucin and amyloid deposition in tissue samples rule out these entities.



**FIG. 1** Pock-like atrophic scarring and waxy yellowish papules on face, along with lower lip, thickened lower lip, beaded eyelid margins with yellowish waxy papules and loss of eyelashes on inner side of right lower eyelid.

There is no definitive cure of this condition. Oral dimethylsulphoxide, penicillamine and etretinate have been used with variable efficacy. Laryngeal lesions can be treated with microlaryngoscopy and dissection of the vocal cords. Dermabrasion, chemical peeling and laser therapy is helpful for skin lesions.

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