Net Quiz

# Papules, pock-like scars, and hoarseness of voice

## CASE REPORT

A 6-year-old boy presented with hoarseness of voice and recurrent episodes of blisters and erosions on the scalp, face, trunk, and extremities since 6 months of age. The erosions healed with scarring. Most lesions were spontaneous in onset although some were post-traumatic. There was no developmental delay. There was neither history of photosensitivity nor of similar family history. His parents were non-consanguineous.

**Figure 1: Multiple pearly papules over eyelid margins of both eyes**

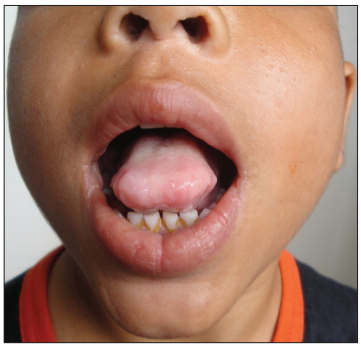

**Figure 2: Enlarged tongue with difficulty in protrusion**

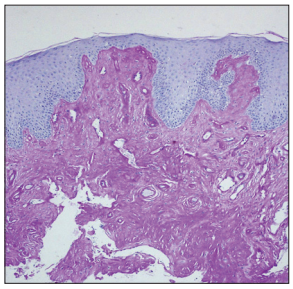

**Figure 4: Marked atrophy of adnexae with few blood vessels showing onion skinning (H and E, ×40)**

Mucocutaneous examination revealed multiple pearly beaded papules on the margins of bilateral upper and lower eyelids [Figure 1]. Multiple "pock-like" scars were present on the face, trunk, and extremities. Few crusted erosions were present on the scalp and extensors of forearms. Lips were enlarged, their mucosal surface revealing yellowish indurated papules. Tongue was enlarged, firm on palpation and the boy had difficulty in protruding it [Figure 2]. Patches of scarring alopecia were present on the occiput and vertex of scalp.

Histopathological examination of the lower lip revealed periodic acid Schiff (PAS)-positive deposits, but diastase-resistant pale eosinophilic hyaline material in the papillary and deep dermis [Figure 3]. Congo red stain was negative. There was marked atrophy of adnexal structures [Figure 4] with few blood vessels showing onion skinning [Figure 5]. Direct laryngoscopy revealed thickened but mobile vocal cords with surface nodulations.

What is your diagnosis?

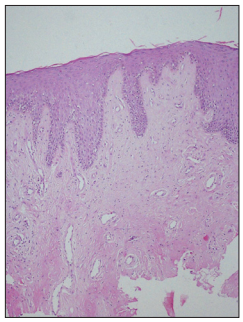

**Figure 3: PAS-positive and diastase-resistant deposits in upper and deep dermis (H and E, ×100)**

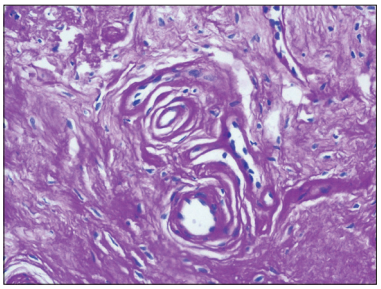

**Figure 5: Onion skinning of blood vessels with PAS–positive and diastase-resistant deposits in deep dermis (H and E, ×200)**

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Diagnosis: Lipoid proteinosis

## DISCUSSION

Lipoid proteinosis is a rare autosomal recessive disorder, first described by Urbach and Wiethe[1] in 1929. It occurs with equal frequency in both males and females.[1] The signs and symptoms of the disease are due to hyaline deposits in skin, mucosae, and internal organs. The underlying defect is in the extracellular matrix protein 1 (ECM1) gene present on chromosome 1q21, which encodes a secretory glycoprotein.[2] Frameshift or non-sense mutations, commonest at exon 7 followed by exon 6, result in loss of function of ECM1 gene.[2,3] Thus, reduced or absent expression of ECM1 glycoprotein leads to aberrant deposition of hyaline material in various tissues.

Clinical manifestations usually develop in infancy or may appear later. Deposits in the vocal cords lead to hoarseness of voice, which is often the first clinical manifestation. Hoarseness may present at birth as a weak cry or appear later but persists throughout life. Cutaneous lesions start as vesicles and crusts on face and extremities that heal with scar formation. Scars may be pock-like, linear, or cribriform. Flesh-colored beaded papules (moniliform blepharosis) are an early finding which can be useful in the diagnosis.[3] Other cutaneous findings include papules and nodules, yellowish discoloration, verrucous thickening of extensors of extremities and pruritus. Mucosal changes include thickened frenulum, tongue, and gingival hypertrophy. Another frequent finding in the eyes is drusen formation in the Bruch's membrane, which has been observed in one-third to half of the patients.[4] Other associations include epilepsy which is probably due to intracranial calcification within the temporal lobe.[5]

The differential diagnoses include erythropoietic protoporphyria (EPP), amyloidosis, and papular mucinosis. EPP can be excluded based on a history of photosensitivity, involvement of photo-exposed areas, and sparing of mucous membranes. Histopathology in EPP reveals deposition of homogenous pale eosinophilic PAS-positive, diastase-resistant material around blood vessels with sparing of adnexa. Amyloidosis and papular mucinosis can be differentiated histologically by special stains.

Lipoid proteinosis has a benign course compatible with a normal life span. Currently, no therapeutic modality is uniformly effective in all patients. Treatment is mainly symptomatic. Vocal cord lesions have been successfully removed with CO2 laser therapy or mucosal stripping although recurrences are possible. Eyelid lesions have been treated surgically with blepharoplasty and CO2 laser.[6] Pock-like scars have been treated with dermabrasion and deep chemical peeling.[6] Systemic agents including glucocorticoids, d-penicillamine, dimethyl sulfoxide and more recently, retinoids are reported to be useful.[7]

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