Lipoid proteinosis

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DESCRIPTION

A 25-year-old man, born of a non-consanguineous marriage, presented with a history of progressively increasing, asymptomatic, pinhead-sized small lesions over the eyelid along with hoarseness of voice since early childhood. There was a preceding history of developing small vesicular and crusted lesions over the face which healed spontaneously with scarring. He had no history of photosensitivity, dyspnoea, dysphagia, seizures, visual disturbances, respiratory obstruction or restricted tongue movement. No one else in the family had similar complaints. Mucocutaneous examination revealed multiple, skin coloured, linearly arranged, closely aggregated 1–2 mm-sized papules predominantly over both upper eyelid margins (figure 1A). The facial skin was waxy in appearance with multiple, ill-defined atrophic superficial scars of varying sizes (figure 1B). Corrugated, rough, keratotic, ill-defined plaques were present over bilateral elbows and knees (figure 1C). The tongue was hypertrophied and showed dental impressions (figure 1D). Skin biopsy from elbow revealed homogeneous hyaline, eosinophilic material which was confluent in the papillary dermis and was localised to perivascular and periadnexal structures in the mid-dermis. It was periodic acid-Schiff (PAS) positive and crystal violet and Congo red negative (figure 1E,F). Indirect laryngoscopy revealed mild thickening of aryepiglottic fold along with nodular growth over it in the posterior most part extending to interarytenoid area. On clinicopathological correlation, a final diagnosis of lipoid proteinosis was made and the patient was transferred to otolaryngology unit for evaluation and management of nodular growth.

Lipoid proteinosis, also known as Urbach-Wiethe syndrome (MIM 247100), is an uncommon autosomal recessive genodermatosis characterised by deposition of an amorphous hyaline material predominantly in the skin and mucosa of upper aerodigestive tract.[1 2] Virtually any organ may get involved including lungs and central nervous system. It occurs due to loss-of-function mutation in the extracellular matrix protein 1 (ECM1) gene located on chromosome 1q21. Mutation in the ECM1 gene results in abnormalities in the glycolipids or sphingolipids degradation pathway, decreased production of the fibrous collagens and overproduction of basal membrane collagens leading to deposition of PAS-positive hyaline materials in dermis and submucosa.[1 3] The disease classically manifests in infancy with a hoarse cry due to laryngeal infiltration. Various cutaneous manifestations include multiple atrophic scars over face, eyelid beading, keratotic plaques over elbows and knees and waxy infiltration.[1 2] Life expectancy is usually normal. Various treatment modalities including retinoids, D-penicillamine, oral steroids and dimethyl sulfoxide have been tried with unsatisfactory results.[1 2]

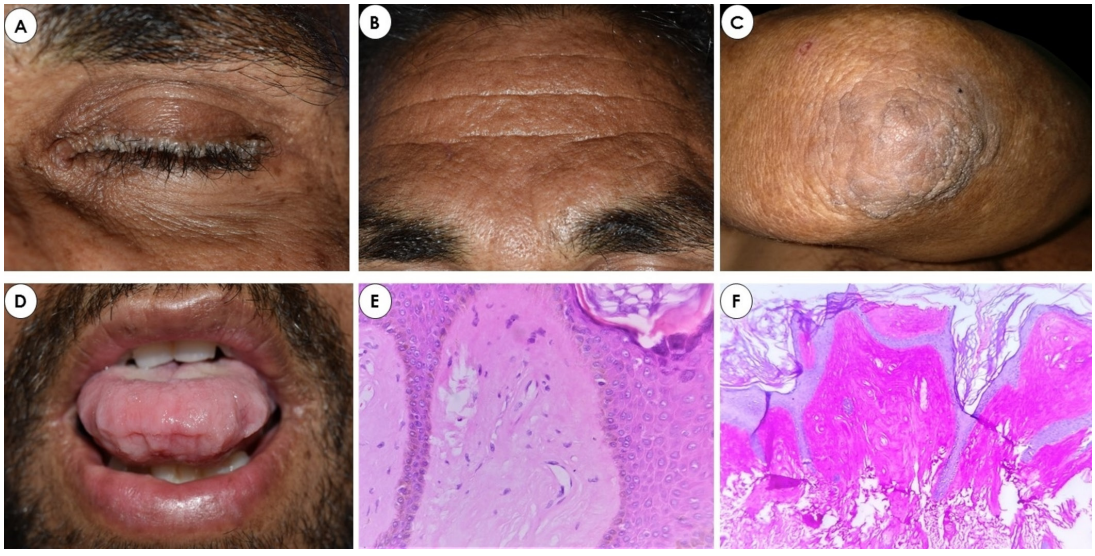

Figure 1 (A) Multiple, skin-coloured, linearly arranged tiny 1–2 mm-sized knobby papules over left upper eyelid margin. (B) Superficial atrophic scars over forehead with waxy infiltrated appearance of the skin. (C) Rough corrugated plaque over the right elbow. (D) Enlargement of the tongue with prominent dental impressions. (E and F) Eosinophilic homogeneous hyaline material in dermal papilla with periodic acid-Schiff (PAS) positivity (H&E, 40×; PAS stain, 10×).

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Learning points

- Lipoid proteinosis is an autosomal recessive genodermatoses.
- Classical clinical manifestations are hoarse cry, multiple atrophic scars, eyelid beading, keratotic plaques over elbows and knees, and waxy infiltration of skin.
- Underlying aetiopathogenesis is deposition of hyaline material in the skin and mucosa of upper aerodigestive tract.

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