# A RARE CASE OF LIPOID PROTEINOSIS

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**ABSTRACT**

**BACKGROUND**

Lipoid proteinosis is a rare autosomal recessive disorder that presents early in infancy with hoarseness of voice followed by pox like and acneiform scars with infiltration and thickening of skin and certain mucous membranes. Mutations in the gene encoding extracellular matrix protein 1 (ECM1) on chromosome 1q21 is the cause in lipoid proteinosis. On histopathology, cutaneous deposition of amorphous eosinophilic material and thickened capillary dermis was noted. On immunofluorescence, anti-type 4 collagen antibodies were seen. No definitive treatment for disease was present.

**KEYWORDS**

Lipoid Proteinosis, Extracellular Matrix Protein 1, Anti-Type 4 Collagen Antibody.

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## BACKGROUND

Lipoid proteinosis is also known as hyalinosis cutis et mucosae, Urbach-Wiethe disease. It is a monogenic autosomal recessive inherited disorder characterised by infiltration of hyaline material into the skin, oral cavity, and larynx.

## Case Report

A 9-year-old female child born out of non-consanguineous marriage, student by profession presented with eyelid beading, generalised depressed scars on face, upper extremities, yellowish spots on tongue, buccal mucosae and inner aspect of lower lip, inability to protrude tongue completely, difficulty in speaking since early childhood, loss of scalp hair. Child had a change in quality of voice since 2 years. Child had never experienced complete symptomatic relief from the day it started. There was no history of prior eye infections, similar illness in family members or neurological symptoms.

General and systemic examination was within normal limits except for mild pallor. Cutaneous examination revealed bilateral beaded eyelids giving moniliform blepharosis appearance. Generalised pox like atrophic scars were seen on skin of face Fig. 1 and bilateral upper limbs Fig. 6. Yellowish, waxy multiple papules were seen on mucosa of lip and tongue, oral hygiene was poor. Tongue Fig. 2 & 3 was enlarged, with woody feel showing indentation at margin with short frenulum Fig. 5. Diffuse alopecia of scalp is present Fig. 7.

After history and general examination, routine investigations were within normal limits. Biopsy revealed hyperkeratosis and irregular acanthosis of epidermis, homogenous eosinophilic hyaline material deposition in arteriocapillary wall, adnexal basement region, sweat coils & dermis Fig.4.

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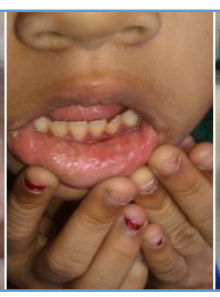


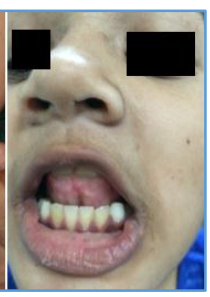


***Figure 1*** ***Figure 2*** ***Figure 3***

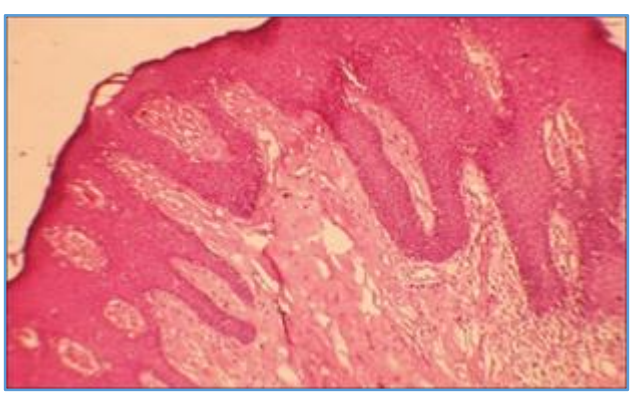
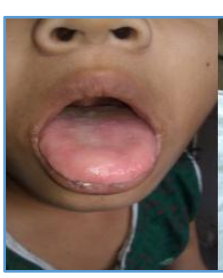


***Figure 4***



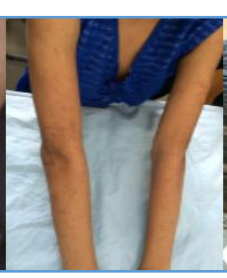


***Figure 5*** ***Figure 6*** ***Figure 7***

## DISCUSSION

Lipoid proteinosis is a rare autosomal recessive disorder that presents early in infancy with hoarseness of voice followed by pox like and acne form scars with infiltration and thickening of skin and certain mucous membranes. Approximately 250

cases have been described in the literature with a higher prevalence in South Africa and Sweden.[1,2,3]

The pathogenesis lies in mutation of gene encoding extracellular matrix protein 1 (ECM1) on chromosome 1q21, human ECM1 codes for glycoprotein of unknown function.

Histopathology features consisted of extensive cutaneous deposition, amorphous eosinophilic material which is PAS+ and diastase resistant deposits seen in surrounding capillaries, sweat coils and in the thickened capillary dermis. Immunofluorescence shows presence of anti-type 4 & anti-type 7 collagen antibodies.

Present early in infancy with hoarseness of voice due to infiltration of vocal cords[4], followed by pox like acne form scars, along with thickening of skin, mucous membranes and organ involvement. First clinical sign observed is hoarseness.

The classical and most recognisable sign is beaded eyelid papules (Moniliform blepharosis). Cutaneous changes include waxy, yellow papules and nodules with generalised skin thickening. Hyperkeratosis may occur in region of mechanical friction. Scalp involvement may lead to hair loss which is not a significant finding. Mucosae of tongue, soft palate, tonsils and lips are infiltrated leading to respiratory difficulty. Inability to protrude tongue is a diagnostic sign.

Extracutaneous features consist of epilepsy and neuropsychiatric abnormalities, calcification in temporal lobes or hippocampus, dental abnormalities, widespread visceral involvement (Lungs, Kidneys, Bladder, and Gastrointestinal Tract)[5]

There is no definitive treatment available for it, but therapeutic trials including oral steroids, oral dimethyl sulfoxide (DMSO) are rarely useful. CO2 laser was used for thickened vocal cords surgery and beaded eyelids.

**CONCLUSION**

Patients with lipoid proteinosis are likely to present first to a dermatologist because of the appearance of their skin; therefore, it is important that the dermatologic diagnosis is not to be missed. We described patients with lipoid proteinosis and discussed the salient features of this disease.

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