

## LIPOID PROTEINOSIS

Mayuri K Shah, Priti P Shah, R C Rawal, F E Bilimoria

A 20-year-old male presented with hoarseness of voice since birth, beaded papular lesions on both eyelids, multiple hypopigmented atrophic scars on trunk and extremities and yellowish infiltrated plaques on tongue, palate and buccal mucosae. Patient was diagnosed clinically as a case of lipid proteinosis. It was confirmed by mucosal biopsy.

**Key Words :** Lipoid proteinosis, Hyalinosis cutis et mucosae

### Introduction

Lipoid proteinosis (hyalinosis cutis et mucosae) is a rare autosomal recessive disorder characterised by deposition of hyaline material in skin, mucous membranes and internal organs. Urbach and Wieth first described this condition in 1929.<sup>1</sup> First noticeable sign is hoarseness of voice starting as early as at birth which worsens later on. Oropharyngeal mucosa shows extensive yellowish infiltration. The tongue is firm and enlarged with restricted movement. There is hypodontia or anodontia. Papulovesicular lesions are found which heal with depressed scars. Tiny papular lesions occur along the margins of upper and lower eyelids giving beads in string appearance. Cicatricial alopecia is also a common association.

Hyaline deposits are also found in the internal organs.<sup>2</sup> Intracranial calcification of hippocampal gyri causes convulsions.<sup>3</sup>

Histopathology suggests accumulation of hyaline material beneath the basement membrane of capillaries and adnexal structures.

### Case Report

A 20-year-old male born to a nonconsanguineous parents presented with

hoarseness of voice since birth. He had thickened yellowish plaques on tongue and oral mucosae for past 10 years.

Clinical examination revealed multiple tiny papular lesions on upper and lower eyelids giving beads in string appearance.

Multiple hypopigmented atrophic scars were found on face, back and extremities. Oral mucosae revealed yellowish infiltrated plaques on tongue, palate and buccal mucosae. The tongue was enlarged and woody hard with restricted movements. The scalp revealed 3 patches of cicatricial alopecia. Systemic examination was normal. No neurological changes were found.

Routine investigations were normal. Biopsy of oral mucosae revealed PAS-positive hyaline material around capillaries and adnexal structures.

### Discussion

Hoarseness of voice since birth, typical 'beads in string' appearance of eyelids, cicatricial alopecia, oral mucosal lesions and multiple atrophic scars over the body were the diagnostic features of lipid proteinosis. Biopsy of oral mucosae confirmed the diagnosis. This case is presented for its rarity and very typical diagnostic features.

### References

1. Rook A. Lipoid proteinosis. Urbach-Weithe disease. *Br J Dermatol* 1976; 94: 341-2.

From the Department of Dermatology, BJ Medical College, Civil Hospital, Ahmedabad-16.

Address correspondence to : Dr Mayuri K Shah

2. Caplan RM. Visceral involvement in lipid proteinosis. Arch Dermatol 1967; 95: 149-55.
3. Hofer P.A. Urbach-Weithe disease: a review. Acta Derm Venereol 1973; 53 (Suppl 71) : 1-56.

# Surgery in Dermatology

**Authors :** Dr Harvinder Singh Marwah &  
Dr Mansukh P. Ghalla.

**Comprises of :** 230 pages, 24 illustrations &  
130 coloured photographs.

**Price Rs 500/-** (copy will be sent by V.P.P.).

**Doctors interested:** please write to Dr Marwah's  
skin & V.D. centre,  
1<sup>st</sup> floor, 214 B/7 Kamal Kunj  
Irla Bridge, S.V. Road,  
Andheri (W), Mumbai 400058,  
India.



**FIG. I** Shows 'Beads on String' Appearance on Left Eyelid.



**FIG. II** Shows Yellowish White Infiltrated Plaques in Oral Mucosa.