Dermatology Practical & Conceptual

What is in a Voice? Deciphering Clue in a Case of Facial Varioliform Scars in a Young Girl

Srishti Dabas[1], Tanvi Dev[1], Vaishnavi Modi[1], Yankila Tamang[2], Ditixa Patel[3], Nikhil Arora[3]

1 Department of Dermatology, Venereology and Leprosy, Maulana Azad Medical College and Lok Nayak Hospital, New Delhi, India
2 Department of Pathology, Maulana Azad Medical College, New Delhi, India
3 Department of Otolaryngology, Maulana Azad Medical College and Lok Nayak Hospital, New Delhi, India

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Corresponding Author: Tanvi Dev, MD, Department of Dermatology, Maulana Azad Medical College and Lok Nayak Hospital, New Delhi, India. Phone numbers: +91 99582 25519. E-mail: ivnat2008@gmail.com

Introduction

Lipoid proteinosis (LP), also known as hyalinosis cutis et mucosae is a rare autosomal recessive disorder, characterized by deposition of amorphous hyaline material in skin and other internal organs [1]. Only several hundred cases of LP have been reported in literature [2]. We describe a similar case in young girl.

Case Presentation

A 7-year-old-girl, born from consanguineous marriage, presented with generalized itching and excoriations, since 1 year of age, along with scars on bilateral cheeks and extremities. She was otherwise healthy and had achieved developmental milestones adequate for her age. Examination revealed pale doughy, yellowish-colored skin on the face with atrophic varioliform scars on bilateral cheeks, chest, arms and legs, over a background of post inflammatory hyperpigmentation. There were skin-colored smooth waxy-appearing closely aggregated papules (1x2 mm) over buttocks and gluteal cleft and beaded papules along the eyelid margin (moniliform blepharosis) bilaterally. Dome-shaped erythematous papules were also noted over the tongue and labial mucosa (Figure 1). On interrogation with the child, we noted a shallow, hoarse voice which was present since infancy according to the mother. However, her language and communication skills were intact. Laryngoscopy revealed hypertrophic true and false vocal cords with thickening of inter-arytenoid area, though there were no deposits noted (Figure 2, A and B). There was no history suggestive of neuropsychiatric involvement. There was no family history of similar complains. No cranial calcification was noted in radiology of skull. Histology from the

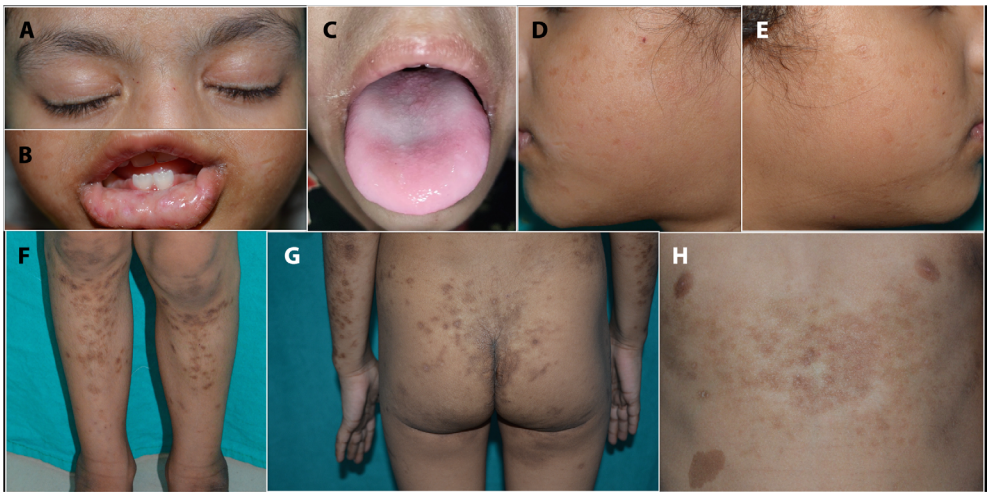

Figure 1. Skin and mucosal manifestations. (A) Beaded papules along the eyelid margin (moniliform blepharosis). (B) Dome-shaped erythematous papules over labial mucosa. (C) Small ill-defined erythematous papules over dorsum of tongue. (D,E) Doughy pale skin on face along with atrophic varioliform scars on left cheek. (F-H) Smooth waxy, closely aggregated papules over buttocks and gluteal cleft on a background of atrophic varioliform scarring and post inflammatory pigmentation (PIH). Atrophic scars and PIH is also noted over bilateral shins and abdomen.

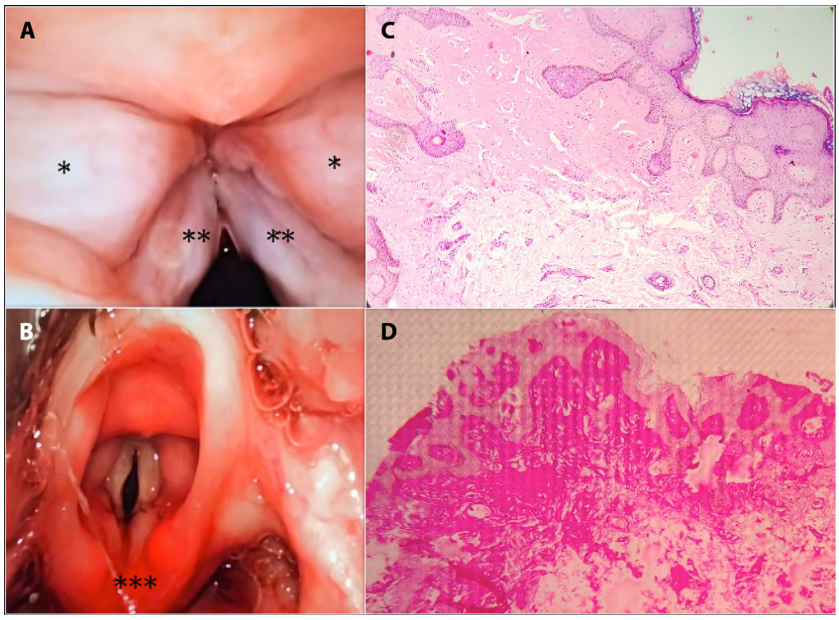

Figure 2. (A) Fibro-optic laryngoscopy showing hypertrophic false (marked as *) and true vocal cords (marked as **). (B) Thickening of inter-arytenoid area (marked as ***). (C) Histopathology from skin biopsy showing confluent homogenous eosinophilic deposits within the papillary and upper reticular dermis with slit-like spaces at 100x magnification (H&E). (D) Histopathology showing Periodic acid-Schiff positive deposits in the upper dermis (100x).

waxy papules on buttocks showed confluent homogenous eosinophilic deposits within the papillary and upper reticular dermis with slit-like spaces, a few blood vessels and fibroblast. Rest of the dermis was unremarkable (Figure 2, C and D). The deposits stained positively with periodic acid-Schiff stain. A diagnosis of lipoid proteinosis was made based on clinical and histological findings.

Conclusions

Lipoid proteinosis is a rare genetic disorder caused by mutation in extracellular matrix – 1 (ECM-1) gene characterised by hyaline-like deposition in various tissues including larynx, skin, eyelids and other internal organs [1]. The disease course is chronic and fluctuating, with gradual progression

until adulthood. LP occurs worldwide with no definitive age, sex, or race predilections [3]. Dermatological manifestations are consistently noted in the form of vesicles and hemorrhagic crusts healing with atrophic pox-like scars. Later, with increasing hyaline deposition in the dermis, the skin becomes diffusely thickened and appears waxy with a yellowish discoloration. Papules, nodules, and plaques appear on the face and lips. Verrucous and keratotic cutaneous lesions have been noted on extensor surfaces like elbows and knees. The classical and most characteristic sign is the beaded eyelid papules (moniliform blepharosis) [4]. The first manifestation, however, is a weak cry or hoarse voice (due to infiltration and deposition of hyaline-like material in the vocal cords) appearing during early infancy. Hoarse voice usually persists lifelong and can lead to complete aphonia [5]. Central nervous system manifestations commonly include epilepsy and behavioral manifestations. Calcification of the temporal lobes or hippocampi have also been reported [6]. None of these manifestations were noted in our patient.

References

1. Shah JS, Shah HA. Lipoid proteinosis: Review of Indian cases. *J Oral Maxillofac Pathol*. 2022;26(2):236-41. DOI: 10.4103/jomfp.jomfp_249_21. PMID: 35968171; PMCID: PMC9364649.
2. Loos E, Kerkhofs L, Laureyns G. Lipoid Proteinosis: A Rare Cause of Hoarseness. *J Voice*. 2019;33(2):155-158. DOI:10.1016/j.jvoice.2017.05.024
3. Jahanimoghadam F, Hasheminejad J. Oral Manifestations and Dental Management Considerations of Lipoid Proteinosis: A Case Report and Review of Literature. *J Dent (Shiraz)*. 2022;23(3):321-326. DOI:10.30476/DENTJODS.2021.89748.1435
4. Vahidnezhad H, Youssefian L, Uitto J. Lipoid Proteinosis. In: Adam MP, Feldman J, Mirzaa GM, et al., eds. *GeneReviews*®. Seattle (WA): University of Washington, Seattle; January 21, 2016.
5. Savage MM, Crockett DM, McCabe BF. Lipoid proteinosis of the larynx: a cause of voice change in the infant and young child. *Int J Pediatr Otorhinolaryngol*. 1988;15(1):33-38. DOI:10.1016/0165-5876(88)90048-1
6. Siebert M, Markowitsch HJ, Bartel P. Amygdala, affect and cognition: evidence from 10 patients with Urbach-Wiethe disease. *Brain*. 2003;126(Pt 12):2627-2637. DOI:10.1093/brain/awg271