Poster ID Number : 2

"Hyaline deposits spark off the scars, beads & growls": Lipoid proteinosis – A report of two cases

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INTRODUCTION

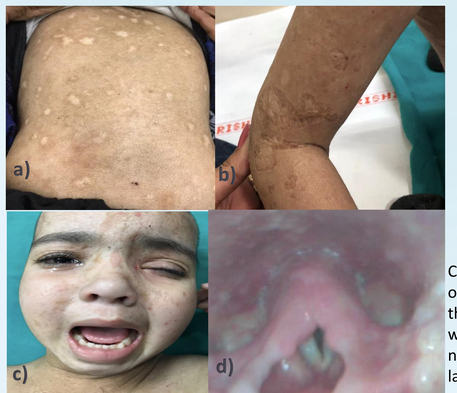
Lipoid proteinosis is a very rare progressive autosomal recessive disorder characterized by deposition of hyaline material in the skin, upper aerodigestive tract, and internal organs.[1] There are less than 500 individuals with lipoid proteinosis that is reported worldwide. Lipoid proteinosis was reported for the first time in 1929 by Urbach, a Viennese dermatologist, and Wiethe, an otolaryngologist. Mutations of the *ECM1* gene that encodes a glycoprotein known as extracellular matrix protein 1 is identified as the cause for this condition.[2] Clinical manifestations vary greatly from individual to individual and may include skin scarring, beaded eyelid papules, and laryngeal and tongue infiltration leading to hoarseness and limited lingual movements.[1]

CASE REPORT

We report two cases of 5 years and 6 years respectively who were both products of non-consanguineous marriage. **Case 1:** She presented with a history of hoarseness of voice and recurrent blistering and erosions that healed by scarring observed since 6 months of age. On examination, there were diffuse waxy infiltration of skin, multiple tiny yellowish colored papules of 0.5 cm in size, multiple oval punched out varioliform and linear scars (BSA: 60-70%). There was subtle beading of eyelid margins (moniliform bhlepharosis). The tongue was slightly infiltrated. Patchy alopecia was noted on the scalp. Laryngoscopy showed hyaline deposits in vocal cords. Rest of the systemic examination was normal. Histopathology showed eosinophilic hyaline PAS +/Diastase- deposits in papillary dermis, perivascular and periadnexal areas**.**

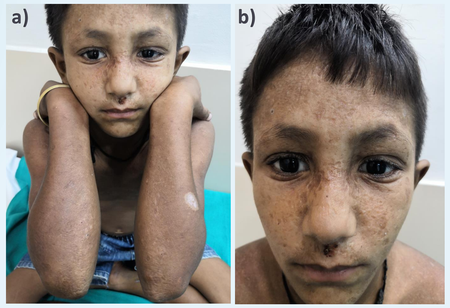
Case 2: presented with hoarse cry and recurrent blistering since 1 month of life and skin lesions healed with pock-like scars. On examination multiple infiltrated pock-like scars were present on the face and limbs predominantly (BSA: 10-12%). Rest of the systemic examination was normal. Histopathology showed subtle hyaline deposits in papillary dermis and perivascular areas. Both of them were treated with oral vitamin A therapy.

CASE 1



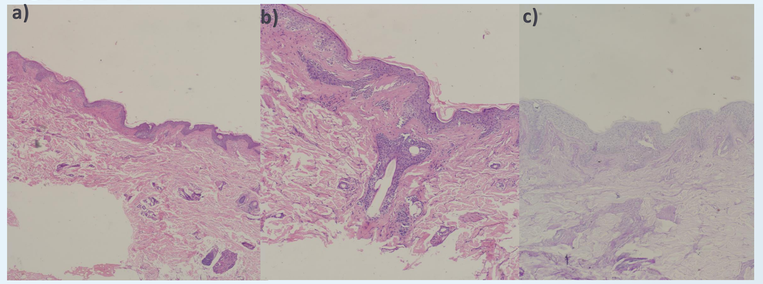
Case.1 Clinical images a) Pock-like scars over the trunk. b) similar pock-like scars on the arms. c) acneiform scars on the face with beaded papules over eyelids. d) nodules on the vocal cords seen in laryngoscopy.

CASE 2



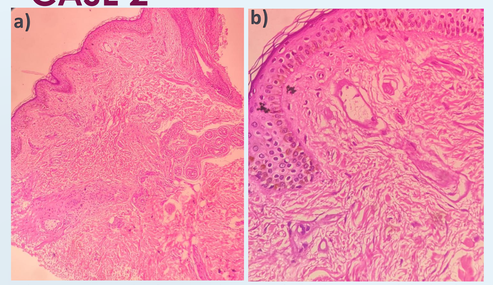
Case.2. Clinical images a) multiple varioliform scars on face and elbows. b) leonine facies with moniliform blepharosis.

CASE 1



Photomicrograph of case 1 showing a) eosinophilic hyaline deposits along the dermo-epidermal junction & vessels (H & E, 10x). b) peri-adnexal eosinophilic hyaline deposits (H & E, 40x). c) and d) PAS-D stain (40x)

CASE 2



Photomicrograph of case 2 showing a) subtle eosinophilic hyaline deposits along the dermo-epidermal junction and vessels (H & E, 10x) b) perivascular eosinophilic hyaline deposits (H & E, 40x).

DISCUSSION

The earliest clinical manifestation of lipoid proteinosis is progressive hoarseness of voice due to deposition of hyaline material in the vocal cords.[3] Both patients described here had hoarseness of voice as the earliest presentation and almost similar clinical picture. Usually cutaneous findings present as two overlapping stages that begins in first two years of life. Histopathologic features of second stage are well known. It is characterized by the deposition of eosinophilic, PAS positive and diastase resistant hyaline material in the basement membrane resulting in thickening of the dermoepidermal junction; and deposition in the papillary dermis surrounding the capillaries, the adnexal epithelia, especially sweat coils are hallmark histological features.[3] Similar material is deposited in erythropoietic protoporphyria, but with limited distribution, being perivascular only without sweat coil involvement. Histopathologically, the first stage shows non-dyskeratotic acantholysis and a cell-poor subepidermal blister containing fibrin and extravasated RBCs.[4] Case 2 had very subtle findings and minimal deposits were found unlike case 1 thus signifying the inter-individual variation in the deposition of hyaline material.

CONCLUSION

Although the clinical feature of hoarseness of voice, beaded waxy papules over eyelids and pock like scarring are characteristic, and observed in both cases but histopathological findings had some variations. Thus, careful clinico-histopathological correlation is essential in establishing the diagnosis and differentiating from porphyrias, amyloidosis, etc. This report provides an insight into the characteristic clinical and histopathological features of this rare disorder and also emphasizes on the inter-individual variation of the disease which can be attributed to the amount in hyaline deposits in the dermis.

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