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Case Report

An Unusual Overlap of Lipoid Proteinosis and Cutaneous Amyloidosis – A Case Report

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ABSTRACT

Lipoid proteinosis is a rare genodermatosis that primarily affects the skin and mucous membranes of the respiratory and digestive systems. Cutaneous amyloidosis is characterised by deposition of amyloid protein in the upper papillary dermis. The present case is a 32-year-old male who presented with asymptomatic multiple raised skin coloured lesions and hoarseness of voice since childhood. On examination, he had multiple skin coloured and yellow waxy papules, some discrete and others coalescing to form plaques over the trunk, neck, upper and lower limbs. He also had multiple hyperpigmented papules and plaques on the back and neck, and ovoid shaped scars on the back. Examination of the eyelids revealed moniliform blepharosis, and there were pock like scars on the face. Indentations were present on the lateral and anterior borders of the tongue. Biopsy confirmed the same with few areas positive for Congo red staining. Overlap of lipoid proteinosis and macular amyloidosis was diagnosed. We report this overlap condition for its rarity.

Keywords: Lipoid proteinosis, Amyloidosis, Congo red

INTRODUCTION

Lipoid proteinosis, also known as Urbach-Weithe disease, is a rare autosomal recessive disorder affecting skin, upper aerodigestive tract and internal organs. There is loss of function mutations in the extracellular matrix protein 1 (ECM1gene) on chromosome 1q21.[1] Primary cutaneous amyloidosis is abnormal extracellular deposition of amyloid, which is limited to skin. They stain positively with Congo red and show apple green birefringence under polarised light.[2]

CASE REPORT

A 32-year-old male, born for a consanguineous couple, came with complaints of skin lesions and hoarseness of voice since 2 years of age. The skin lesions started as blisters on the face and lower limbs, which eventually ruptured to form erosions and healed with scarring. There was history of multiple elevated skin lesions gradually increasing in number and progressive thickening of skin over neck and extremities. No history of any neurologic complaints or visual disturbance. On examination, multiple yellow coloured waxy and hyperpigmented discrete papules present over the arms, forearms, hands, neck, trunk, scrotum and lower legs. Ill-defined hyperpigmented macules and few papules demonstrating rippled pattern at the periphery of the lesions were present on the back and upper limbs [Figure 1]. Moniliform papules were arranged linearly over the free margin of bilateral upper eyelids [Figure 1a]. Multiple pock like scars were present over the face and upper limbs. He had difficulty in protruding the tongue. Few indentations present

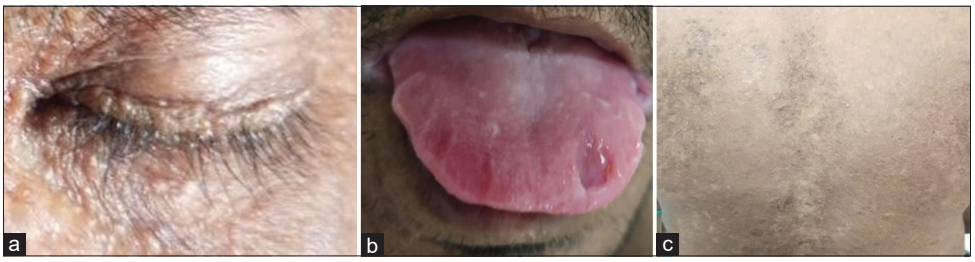

Figure 1: (a) Moniliform blepharosis, (b) Indentations on the tongue and (c) Hyperpigmented macules and papules demonstrating peripheral rippled pattern on the back.

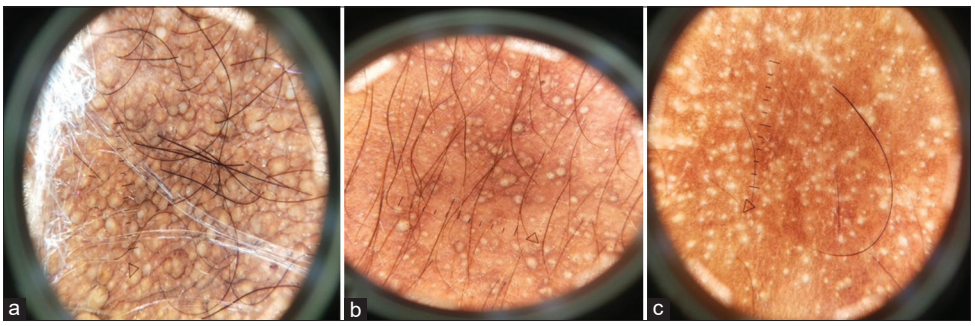

Figure 2: (a) Yellow waxy papules with pulpy appearance on the neck (Heine Delta 30, ×10), (b) Hypopigmented waxy papules on the back (Heine Delta 30, ×10) and (c) Small white central hubs with surrounding brown venation like pattern on the back (Heine Delta 30, ×10).

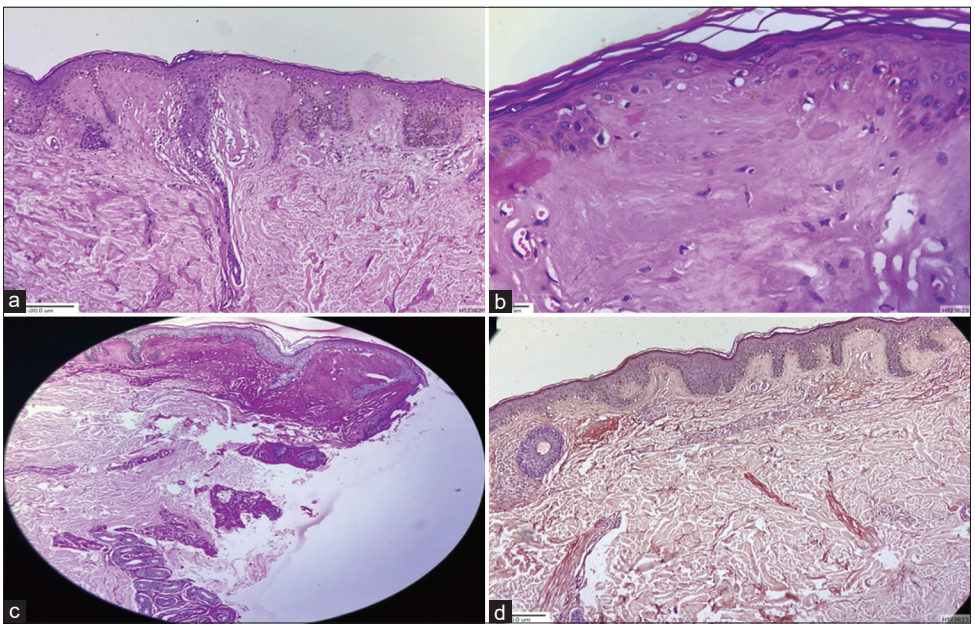

Figure 3: (a) Hyperkeratosis and eosinophilic amorphous deposits in upper dermis [Haematoxylin and Eosin (H&E), ×10], (b) Hyaline deposition in the papillary dermis (H&E, ×40), (c) Periodic acid-Schiff (PAS) positive hyaline material in the dermis and surrounding adnexal structures (H&E with PAS, ×40) and (d) Congo Red positive amyloid deposits in the dermis (Congo Red stain, ×10).

over the borders of the tongue [Figure 1b]. Genital mucosa was normal. Hair and nails were normal. Dermoscopy of the lesions on the neck showed multiple yellow waxy papules, some with pulpy appearance and lesions on the back showed multiple hypopigmented waxy papules and multiple small white central hubs with surrounding venation like extension of brown pigmentation [Figure 2]. Blood count, liver and renal function tests, thyroid profile, urine routine and chest

X-ray were normal. Serology for hepatitis B, C and human immunodeficiency virus were negative. Skin biopsy taken from the lesion on the upper back and right forearm showed mild hyperkeratosis and eosinophilic amorphous deposits in upper dermis [Figure 3a-b]. Periodic acid-Schiff (PAS) positive hyaline material was found in the dermis and around adnexal structures [Figure 3c]. Congo red positive amyloid deposits were distributed in the dermis [Figure 3d]. Apple green birefringence under polarised light could not be demonstrated. Immunohistochemistry and direct nucleotide sequencing of extracellular matrix-1 (ECM-1) were not performed due to limited resources.

DISCUSSION

Lipoid proteinosis usually presents at birth or early infancy. The patients classically present with shiny and waxy facial skin, warty yellowish papules, plaques and nodules. The common sites of involvement are elbows, knees and nape of neck. They also have pock like scars, moniliform blepharosis, scarring alopecia, thickened tongue and frenulum. The first manifestation at birth or early infancy is the weak cry and hoarseness of voice. It can be associated with corneal opacities, secondary glaucoma, intestinal bleeding, neurological manifestations such as seizures, memory loss, schizophrenic behaviour and neuropsychological manifestations. Intracranial calcification in the temporal lobe or hippocampus can be demonstrated in the computed tomography.[1,3] Occasionally, patients can present with multiple cutaneous lesions and mild or no systemic involvement.[4] Dermoscopy of the lesions show sulci and gyri with pale structureless areas, small rounded pinkish white structures in multiple clumps-pulpy appearance.[3] The hallmark finding in histopathology is the deposition of PAS positive and diastase resistant hyaline material in the upper dermis, with localisation around blood vessels and eccrine sweat glands. There is no effective therapy until this date. Patients have a fair life expectancy with regular follow-up.[1,4] Macular amyloidosis presents as poorly delineated hyperpigmented patches of greyish-brown macules with rippled pattern and hyperpigmented papules are found in lichen amyloidosis. The commonly involved sites are interscapular area, shins, forearms, clavicles, breast, face, neck and axilla. Mechanical trauma and ultraviolet exposure are also contributing factors. It can also be called as friction amyloidosis, towel melanosis and nylon clothes friction dermatitis. Amyloid deposits are limited to upper dermis in macular and lichenoid lesions which stain positively with Congo red.[2,5] Amyloid deposits in this case can be attributed to ultraviolet exposure, constant friction and scratching of skin.

CONCLUSION

Lipoid proteinosis and amyloidosis act as close differentials in histopathological study and Congo Red stain is used to differentiate between them. In our case, ironically, both exist together. Only few cases of lipoid proteinosis, about 300 have been reported in the world literature and fewer, that is 30 from India. To the best of our knowledge, there has not been any case reports of lipoid proteinosis associated with cutaneous amyloidosis. Hence, we report this case for its rarity and infrequent association with cutaneous amyloidosis.

Ethical approval

Institutional Review Board approval is not required.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

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Nil.

Conflicts of interest

There are no conflicts of interest.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

The authors confirm that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript and no images were manipulated using AI.

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