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Lipoid Proteinosis: A Case Report

KEYWORDS: Lipoid proteinosis, Urbach-Weithe disease, acneiform scars, eyelid beading, Periodic- Acid Schiff (PAS) positive hyaline material, temporal lobe calcifications.

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ABSTRACT *Lipoid proteinosis (LP), also known as Urbach Weithe disease, is a rare disorder with autosomal recessive inheritance, resulting in progressive widespread deposition of Periodic- Acid Schiff (PAS) positive hyaline material in the skin, mucous membranes of the upper respiratory tract and other internal organs. Classical clinical presentation includes acneiform pox-like skin scarring with formation of yellowish, waxy papules, beaded eyelid papules and laryngeal infiltration leading to hoarseness of voice. Neurological affection in the form of seizures and behavioural disturbances with associated bean-shaped calcifications in the temporal and hippocampal areas may be present.*

We hereby report a young male with a classical presentation of Lipoid proteinosis.

Introduction

Lipoid proteinosis (LP), also known as Urbach-Weithe disease is a rare disorder with autosomal recessive inheritance, first described in 1929. The disease may display multisystemic involvement, but the skin and mucous membranes of the upper respiratory tract are those primarily affected. The disease is characterized by various cutaneous manifestations such as waxy papules, cutaneous scarring, eyelid beading; so also, noncutaneous manifestations due to infiltration of hyaline material in the skin, larynx and other organs[1]. The disease is characterized histopathologically by inter-cellular deposits of Periodic-Acid Schiff (PAS) positive hyaline material in skin, mucous membranes of the upper respiratory tract and other internal organs[2]. This hyaline material results from deposition of non-collagenous proteins and glycoprotein[3]. Cutaneous involvement is in the form of diffuse skin infiltration followed by gradual thickening, finally resulting in yellowish waxy papules and acneiform pox-like scarring. Infiltration of tongue and frenulum can limit tongue movements and cause speech difficulties. Calcifications in the temporal lobes, hippocampus can occur[4], and may be associated with neurological and psychiatric sequelae.

Case Report

A 15 years old male patient presented to us with a short history of fever and cough. He gave a chronic history of thick and yellowish-appearing skin over the face and the extremities. The skin affection had been present since the patient's childhood. There was an associated chronic history of mild hoarseness of voice, but no history of respiratory difficulty, seizures, psychiatric or visual disturbances. There was a history of second degree consanguinity. None of the other family members were affected. On examination, the patient had thick waxy papules over the face. The facial skin also had small acneiform poxlike scars all over (Image 1).

Image 1: Patient's photograph

He had severe anemia on admission, a slightly hoarse voice, impaired mouth opening and evidence of pharyngitis. An indirect rigid laryngoscopy could not be performed due to restricted mouth opening. He was administered antibiotics and blood transfusions, and was subjected to CT imaging of the brain and a full thickness skin biopsy.

CT scan of the brain revealed bilateral bean-shaped hyperdense areas in the temporal lobes, suggestive of calcification, characteristically seen in lipoid proteinosis (Image 2).

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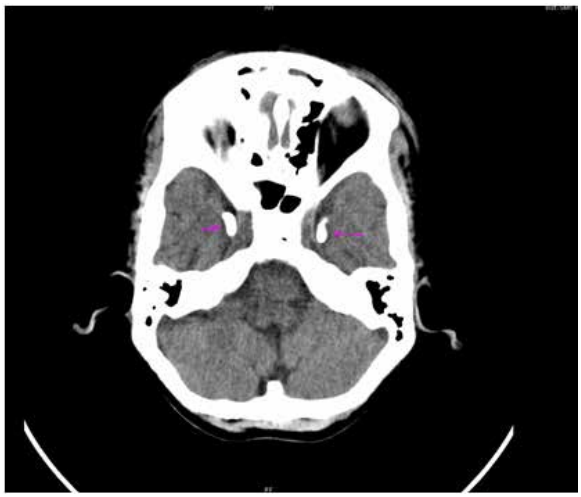
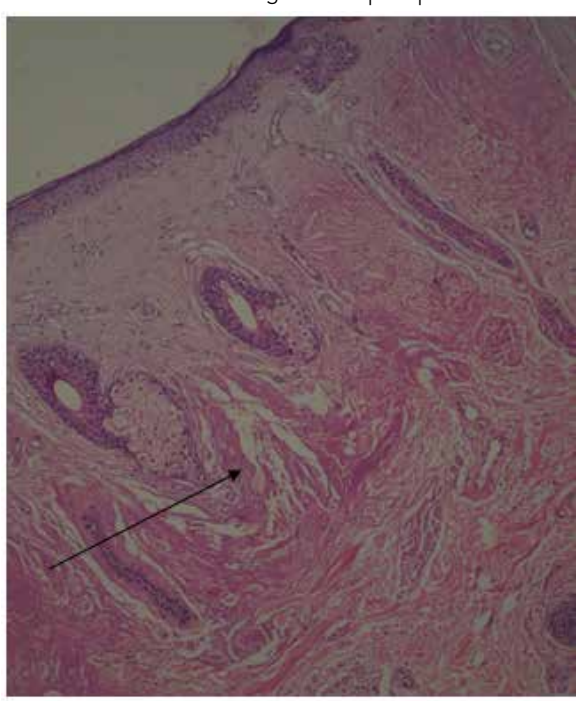
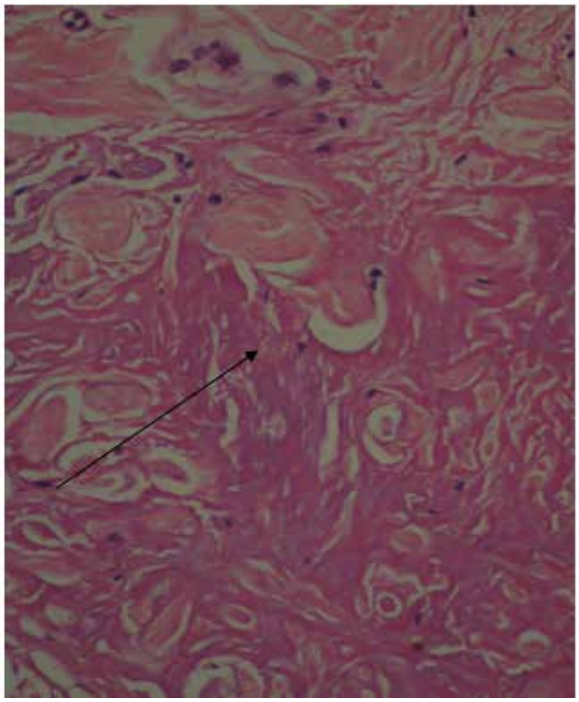


Image 2: CT Brain image

The skin biopsy revealed heavy intercellular deposition of pink eosinophilic material around the blood vessels in the pars reticularis (Images 3 &4). Histopathological findings were consistent with the diagnosis of lipoid proteinosis.





Images 3 & 4: Skin Biopsy images

Discussion

Lipoid proteinosis, a rare genetic disease with autosomal recessive inheritance, is characterized by diffuse deposition of Periodic-Acid Schiff (PAS) positive hyaline material in the skin and mucous membrane, mainly of the upper respiratory tract. The underlying genetic abnormality is due to loss-of-function mutations in the gene encoding extra-cellular matrix 1 (ECM1) on chromosome 1q21[5]. Fewer than 300 cases have been described in medical literature. Most cases have been reported from the African continent[6]. Though rare in the Indian subcontinent, there are reports from hospitals across India[7,8,9].

The first clinical manifestation is usually during childhood with progressive hoarseness of voice due to diffuse laryngeal infiltration[10]. This can occasionally cause significant respiratory distress. Hoarseness may be present at birth or ay present later on in life. Our patient had a past history since childhood of hoarseness of voice, but there was no history of any respiratory compromise or embarrassment.

Skin affection in lipoid proteinosis is in the form of waxy, yellowish papules and acneiform pox-like scars over the face and extremities. Our patient presented with these typical skin lesions.

Other findings include beaded eyelid papules, thickening of the frenulum of the tongue, which can limit tongue movements and cause speech difficulties. Small bowel affection may lead to intestinal bleeding. Hyaline deposits can occur in the conjunctiva, cornea and retina. Neurological symptoms such as memory loss, behavioural problems may be present in some patients. Neurological affection is in the form of intracranial calcifications in the temporal lobes and amygdaloid bodies, detected by brain imaging[3]. CT scan of the brain in our patient revealed classical bilateral temporal lobe calcifications.

Histologically, lipoid proteinosis is characterized by deposition of intercellular Periodic-Acid Schiff (PAS) positive, hyaline material around the capillaries, in the papillary dermis, with thickening at the dermo-epidermal junction. Skin biopsy of our patient demonstrated these characteristic findings of dif-

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fuse intercellular hyaline material deposition in the dermis.

Prognosis in patients with lipoid proteinosis is generally good. Except for occurrence of respiratory obstruction that occurs infrequently and rarely requires tracheostomy, life expectancy is usually normal. There is currently no definitive therapy. Treatment approaches include oral steroids, intralesional heparin, atretinate and dermabrasion of affected skin for cosmetic improvement.

Conclusion

Lipoid proteinosis is a rare genetic disorder with autosomal recessive inheritance. It is characterized by diffuse deposition of Periodic-Acid Schiff (PAS) positive hyaline material in the skin and mucous membranes of the upper respiratory tract and other internal organs. Neurological affection with characteristic occurrence of bean-shaped calcifications in bilateral temporal and hippocampal areas may be present. Although a rare genetic disorder, it should be suspected in any young patient presenting with hoarseness of voice, skin affection in the form of thick papules and scarring affecting the face and the extremities, with occasional neurological and psychiatric manifestations.

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