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LIPOID PROTEINOSIS OF URBACH WIETHE- A CASE REPORT

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Abstract : A 9 year old male child born of non consanguineous parents presented with complaints of hoarseness of voice, multiple skin lesions and thickening of eyelid margins. On examination the child had hoarseness of voice, multiple acneiform(pock like) and atrophic scars over face, trunk, elbows and knees, beaded papules over the eyelid margins and few hyperkeratotic lesions over trunk, elbows and knees. The tongue was indurated on palpation. On indirect laryngoscopy vocal cords were found to be thickened. Histopathological examination of the skin with PAS stain was consistent with lipoid proteinosis

Keyword : Lipoid proteinosis, acneiform scars, beaded papules.

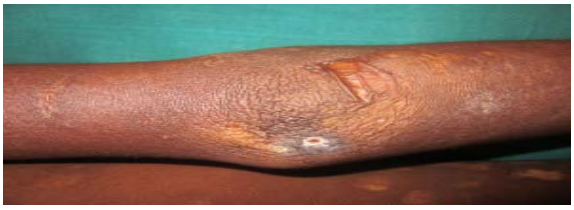
INTRODUCTION:

Lipoid proteinosis is a very rare autosomal recessive disorder that present in early childhood with hoarseness of voice, skin infiltration with beaded papules along the eyelid margins and facial acneiform or pocklike scars.(1) Respiratory system, upper gastrointestinal tract, CNS, lymph nodes and stratified muscles are sometimes involved.(2) This disorder was first described by Weithe and Urbach in 1929. This disorder is attributed to extracellular matrix protein 1 gene mutation located on chromosome 1q21.(3)

CASE REPORT:

A 9 year old male child born of non consanguineous marriage presented with complaints of hoarseness of voice since infancy, multiple skin lesions and thickening of eyelid margins. The skin lesions on face appeared as recurrent episodes of spontaneous vesicle formation which on healing left acneiform or pocklike and linear atrophic scars. Child also gave history of developing scars spontaneously and after mild trauma. The child had no history of epilepsy. Clinical examination revealed hoarseness of voice, multiple acneiform and linear atrophic scars on face. Beaded papules along the eyelid margins were present. The skin appeared thickened over the back of trunk, elbows and knees. Few hyperkeratotic lesions and atrophic scars were present over the back, both elbows and knees. The frenulum and tongue were thickened with only minimal restriction of movements. On palpation the tongue was found to be indurated. Vocal cord thickening was identified on indirect laryngoscopy. CT scan of the brain was normal.

Fig (a) - Beaded papules along eyelid margins.
Fig (b) - Linear and pock like scars over the face.



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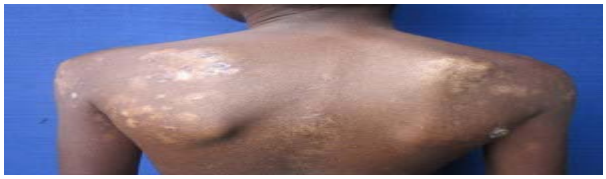
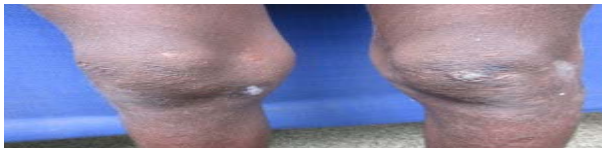


Fig (e)



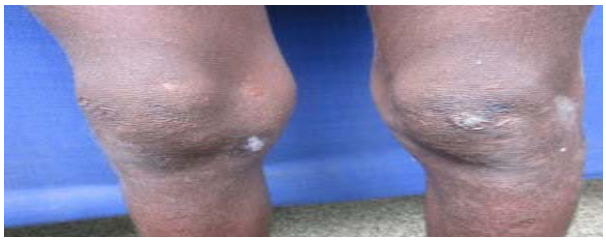


Fig (c,d and e) -Hyperkerotatic lesions and atropic scars over knee, elbow and back.

Fig (d)

Histopathological examination of the skin lesion revealed abundant deposition of PAS positive, diastase resistant eosinophilic hyaline like material in the papillary dermis, surrounding the blood vessels and sweat glands.

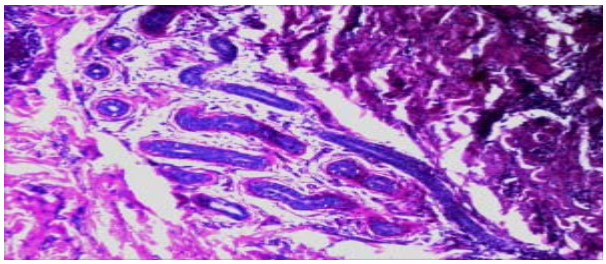
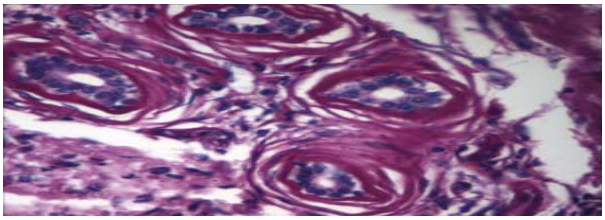


Fig (g) low power fig (h) - high power microscopic view shows hyalin like material around sweat glands (PAS positive-Diastase resistent)



DISCUSSION:

Urbach Weithe disease also known as lipoid proteinosis and hyalinosis cutis et mucosae is a rare autosomal recessive genetic disorder caused by loss of function mutation of the extracellular matrix protein 1 (ECM 1) gene on chromosome 1q21.(3) It was officially reported in 1929 by Erich Urbach and Camillo Wiethe.(4) This disorder has a strong predliction for white races and has no sex predliction. The dermatological symptoms are caused by deposition of hyaline like material in the dermis and thickening of basement membrane in the skin. The symptoms of the disease vary greately from individual to individual.The first symptom is often a weak cry or a hoarse voice due to thickening of vocal cord.

The hoarse voice can be one of the most striking clinical manifestations of the disease. Other manifestations include episodes of blistering in early childhood which become eroded and crusted after minor trauma.(5) Acneiform or pocklike atrophic scars appear on face and elsewhere after minor trauma or spontaneously. Infiltration of skin causes waxy papules, hyperkeratotic or warty plaque and characteristic beaded papules along the eyelid margins. (6) These eyelid lesions are pathognomic and the appearance of these lesions are variously described as string of beads or eyelid beading and is also known as moniliform blepharosis.(7) There may be loss of eyelashes or patchy alopecia due to scalp involvement. The mucosa of the lips, tongue,pharynx become indurated and firm. Epilepsy and psychiatric problems occur in a number of patients and may be associated with intracranial calcification. Visceral involvement has also been reported .(8) The problems progress until early adult life but subsequently stabilize. Histopathologically lipoid proteinosis is characterised by deposition of PAS positive diastase resistant material at the level of the basement membrane, papillary dermis, surrounding the blood vessels and around adnexal epithelia especially the sweat glands and its ducts. (9) Ultrastructural examination reveals concentric ring of basement membrane surrounding blood vessels and irregular reduplication of lamina densa at derma epidermal junction resulting in onion skin ap pearance.(10) Treatment is usually limited. Oral dimethylsulphoxide was reported to help in one patient.(11) Beneficial effect of etretinate and D-penicillamine(12) have also been reported. Dermabrasion, chemical peeling and cabondioxide laser therapy may be helpful for the skin lesions. We report this case for its rarity.

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