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Lipoid Proteinosis- A Rare Case Report

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Abstract

Lipoid proteinosis is a rare autosomal recessive disorder, characterized by infiltration of hyaline material into the skin, oral cavity, larynx and internal organs[1]. Here we report a 15 year old male presenting with classical features of lipoid proteinosis.

Keywords : Lipoid, Proteinosis, Hyaline.

Introduction

Lipoid proteinosis was first described by Siebenmann in 1908. Twenty years later, it was established as a distinct entity by two Viennese physicians, Erich Urbach, a dermatologist, and CamilloWiethe, an otorhinolaryngologist[2]. It is due to mutation in ECM1 gene[3].Kowalewski*et al*. have postulated that extracellular matrix gene 1 glycoprotein plays an important role inregulating blood physiology and anatomy of the skin, as evidenced by gross alteration in the microvasculature of mid- and deep dermis in their study on patients with lipoid proteinosis[4]

Case Report

15 year old male second child of third degree consanguineous marriage presented in our OPD with complaints of thickening of the skin and raised skin lesions over the face and trunk. He was apparently normal at birth and since one year of age he started developing vesicles initially over the scalp which progressed to involve all over the body which get eroded and crusted leading to scarring. He had change in voice since one year of age.There was appearance of similar skin lesions on and off over face, trunk, back and extremitiesleading to multiple scars. There is no history of photosensitivity, seizures, mood changes,breathing difficulty. No history of similar complaints in the family. On examination he was short statured with normal IQ. Dermatological examination showed multiple infiltrated papules over forehead, cheek, multiple varioliform scars present all over the face, multiple beaded papules over upper eyelid (figure 1&2)

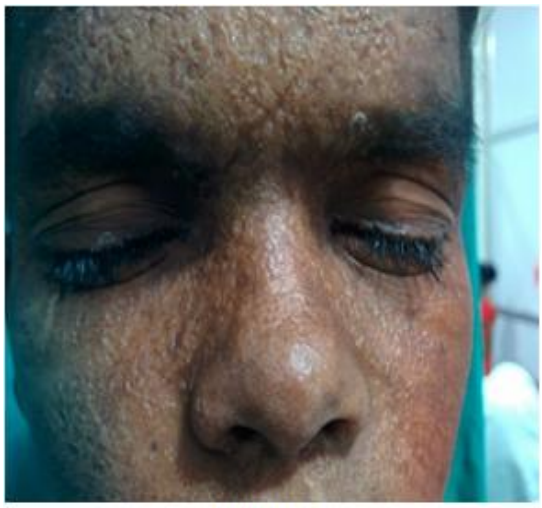


Figure : 1

Figure : 2

A patch of scarring alopecia seen over the parietoccipitalregion (figure 3).Multiple atrophic scars seen over back with mottled pigmentation (figure 4), verrcoushyperpigmented plaques over elbows and knees (figure5), tongue is enlarged and lip was everted

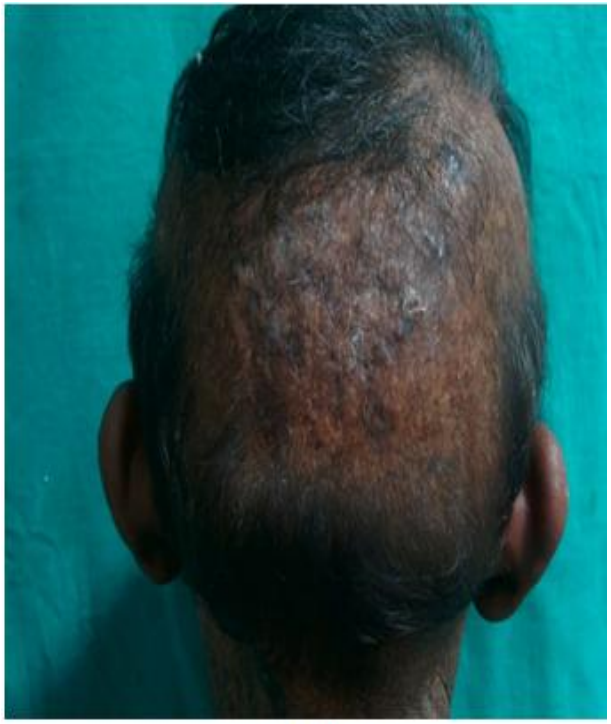


Figure : 3

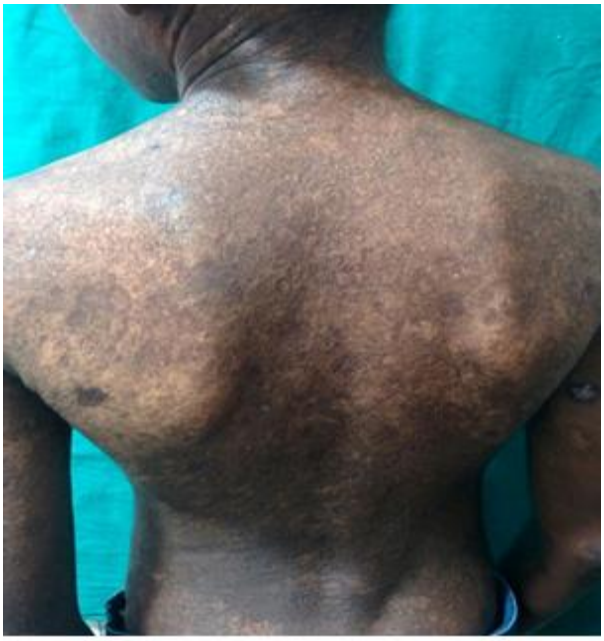


Figure : 4

Figure : 5

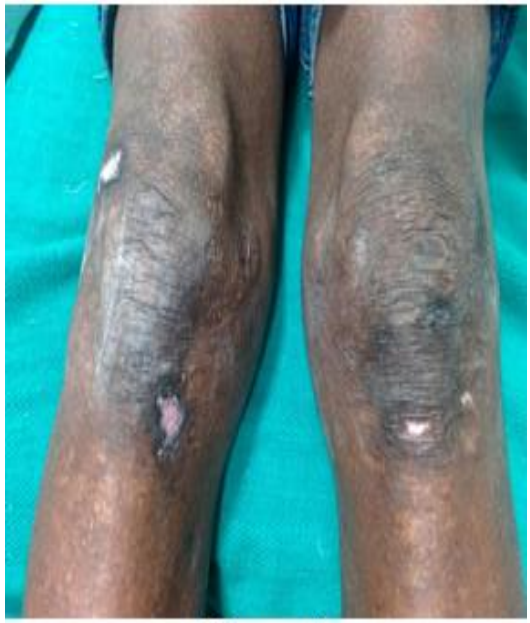
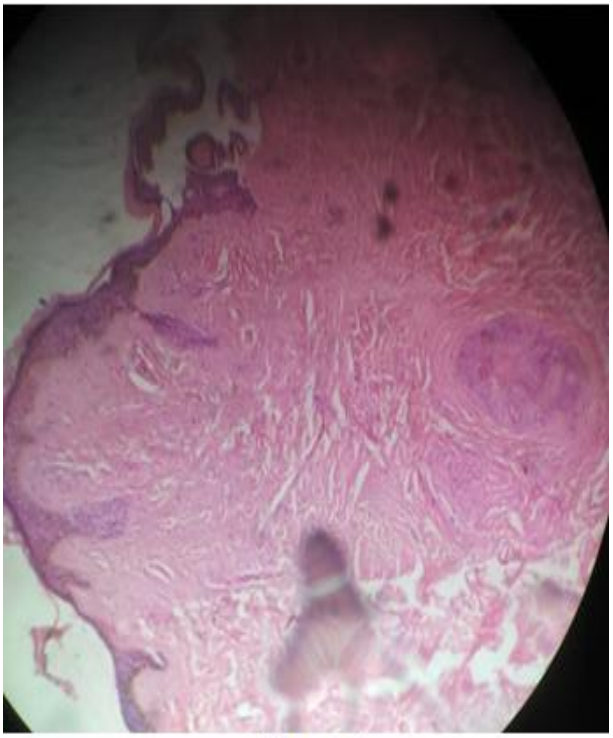


Figure : 6

Skin biopsy was done from waxy papule over the back. Histopathological examination showed hyaline material around the blood vessels, pilosebaceous unit. PAS stain was positive (Hyaline Material). MRI brain showed calcification in the bilateral amygadla. Patient was planned for retinoids. ENT, neurological opinion obtained and followed up.



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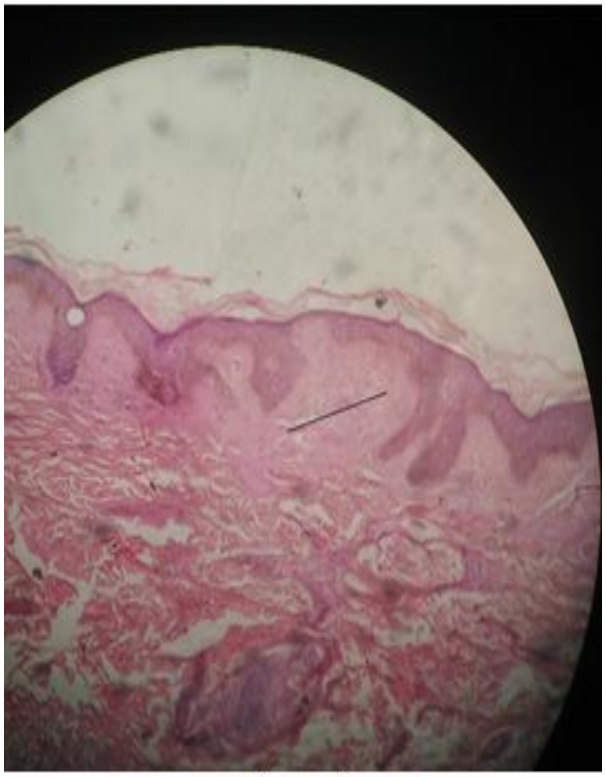

Figure : 8

Discussion

Lipoid proteinosis usually presents in infancy with hoarseness, which can progress to complete aphonia. The vocal cords are Thickened, with nodules here and on the epiglottis. Occasionally, stridor necessitates a tracheostomy. The lips, pharynx, soft palate, uvula and tonsils develop yellow-white submucousinfiltrates. The tongue is enlarged and firm with infiltrates on itsundersurface. The frenulum becomes short and thick, restrictingtongue movement, such that it cannot be protruded. There may berecurrent inflammation of the salivary glands .The first skin lesions are often blisters in early childhood, which become eroded and crusted after minor trauma. Acneform, pock-like scars appear on the face and elsewhere, either following trauma or spontaneously. Infiltration of the skin can cause waxypapules, hyperkeratosis or warty plaques, which may become darker with time. These lesions may affect the palms or backs of the hands, forehead or elbows, where they can be prominent and resemble xanthomas. Characteristic 'beaded' papules are presentalong the margins of the eyelids but they may be subtle (moniliformblepharosis)[1]. There may be loss of eyelashesor patchy alopecia due to scalp involvement. Some patients complain of itching or increased sensitivity to sunlight . Visceral involvement has also been reported. Problems progress untilearly adult life but subsequently stabilize. H&E-stained sections of early lesions reveal pink, hyaline-like thickening of the capillaries within the papillary dermis[2].Many agents, including topical and systemic corticosteroids, oral dimethyl sulfoxide[5]and intra-lesional heparin, have been investigated and used in the treatment of LP. None of these agents has demonstrated any sustained benefits. A recent report describes the use of acitretin[6] in one patient, with improvement in hoarseness but not skin lesions.[3]Life expectancy is usually normal, barring infrequent attacks of respiratory obstruction that rarely require tracheostomy.[7]Parents of affected children should be counseled regarding the risk of having other affected offspring.

Conclusion

This case is reported due to its rarity, classical history and clinical presentation of lipoid proteinosis and the skin biopsy is contributory.

References

1. Burrows N Genetic Disorders of Collagen, Elastin and Dermal Matrix In: Griffiths CE, Barker J, Bleiker T, Chalmers R, Creamer D, editors. Rook's Textbook of Dermatology. 9th ed, Vol. 2. West Sussex: Wiley Blackwell; 2016. p. 1188-1207

2. Dyer J Lipoid Proteinosis and Heritable Disorders of Connective Tissue In: Goldsmith LA, Katz SI, Gilchrest BA, Paller AS, Leffel DJ, Wolff K, editors. Fitzpatrick's Dermatology in General Medicine. 8th ed, Vol. 2. New York: The McGraw-Hill Companies; 2012. p. 296--312.

3. Quist J, Quist S and Gollnick H Deposition diseases In: Bolognia J, Jorizzo J, Schaffer J, editors. Dermatology 3rded, Vol.1.Elsevier sanders: p 712-714

4. T, Wessagowit V, South AP, Ashton GH, Chan I, Oyama N, *et al*. Extracellular matrix protein 1 gene (ECM1) mutations in lipoid proteinosis and genotype phenotype correlation. J Invest Dermatol 2003;120:345-50.

5. Wong CK, Lin CS. Remarkable response of lipoid proteinosis to oral dimethyl sulfoxide. Br J Dermatol 1988;119:541-4.

6. Toosi S, Ehsani AH. Treatment of lipoid proteinosis withacitretin: A case report. J Eur Acad Dermato lV enereol2009;23:482-3

7. Hamada T. Lipoid proteinosis. ClinExpDermatol 2002;27:624-9