Lipoid proteinosis in a six-year-old child

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ABSTRACT

Lipoid proteinosis (LiP) (OMIM 247100) is a rare autosomal recessive disease caused by loss of function mutations in the extracellular matrix protein 1 gene, ECM1, on chromosome 1q21. Clinically characterized by hoarseness in early infancy, followed by waxy papules and plaques on the face and body along with pox-like and acneiform scars. We report here a 6-year-old female child with LiP, who presented to our OPD for recurrent vesiculobullous lesions and beaded lesions over eyelid margins.

Key words: Childhood, lipoid proteinosis, moniliform blepharosis

INTRODUCTION

Lipoid proteinosis (LiP) also known as hyalnosis cutis et mucosa or Urbach-Weithe disease (OMIM 247100) is a rare autosomal recessive disorder,[1] characterized clinically by a myriad signs and symptoms that include hoarseness of the voice, beaded eyelid papules (Moniliform blepharosis), yellowish-white mucocutaneous infiltrates, and atrophic pox-like scars.[2,3] Rarely, vesicles and crusted lesions may occur in the early stage of lipoid proteinosis.[4] We report this interesting case of lipoid proteinosis in a small child who presented initially with vesiculobullous lesions and moniliform blepharosis. The index case had a very early mucocutaneous infiltration in lower gingival mucosa looking like small pebbles, a rare and interesting finding.

CASE REPORT

A six-year-old female child born of consanguineous parents was referred to the skin out patient, with complains of beaded deposits over eyelid margins [Figure 1] since last one year and associated with hoarseness of voice. According to her mother, the baby used to have multiple discrete vesicles over elbow and upper arm in her early infancy, which used to stay for only 1–2 days, and subsequently healed with scars. After subsequent 1–2 yrs they gradually stopped appearing. Mother noticed hoarseness of voice since early infancy. She was examined and found to be otherwise healthy and active and never had a history of ic convulsion or any other neurologic symptoms. Elder sibling didn’t have any such complaints. On cutaneous examination, there was multiple waxy white beaded lesions over both eyelid margins (Moniliform blepharosis) and multiple pocks like scars of different sizes over elbows, upper arm and lower back. [Figure 2], without any fresh vesicles. Examination of oral cavity examination revealed tongue with almost of normal consistency and appearance,thickened frenulum showing yellow white infiltration [Figure 3]. Another interesting finding was multiple beaded lesions over mucosal side of lower lip. [Figure 4]. Indirect laryngoscope showed few small beaded deposits over epiglottis and vocal cords. Biopsy was done from oral mucosa, which revealed deposits of periodic acid-Schiff (PAS)-positive hyaline material around capillaries and adnexal structures [Figure 5]. With the clinical and HPE findings a final diagnosis of lipoid proteinosis was made. Examination of other vital systems like cardiovascular system, central nervous system and eye was normal in both. A routine hemogram, renal and liver profile was within normal limits. No radiological abnormalities were noted or X-ray of skull.

Parents were counseled and child was kept under observation.

DISCUSSION

LiP is a rare autosomal recessive disorder, characterized histologically by infiltration of periodic acid Schiff-positive hyaline material into the skin, upper aero - digestive tract and internal
The disease is slowly progressive with a benign course and occurs equally in males and females. In two thirds of patients it presents in newborn period with hoarseness, cutaneous manifestations like skin infiltration and thickening occurs in first 2 years of life with beaded papules on eyelid margins, and facial acneiform or pock-like scars and was first described by Siebenmann in 1908. It was established as a distinct clinical and histological entity by Urbach and Weithe in 1929. Urbach gave the term “Lipoid Proteinosis” in 1932 to this disease. Moreover, the infiltrates in the tongue and its frenulum limit lingual movements and cause speech difficulties. Usually, the hoarse voice is present at birth or in early infancy, as the first manifestation as observed in the index case. Myxedema which in the infancy also present with hoarseness and diffuse non pitting thickening of skin pose a diagnostic dilemma. Thyroid function tests will help to clinch the diagnosis. Few forms of
mucopolysaccharidodes may present with thickened skin and coarse facies resembling LiP but a biopsy will differentiate two easily. In more severe cases respiratory distress can be seen, where there is diffuse infiltration of the pharynx and larynx, at times requiring tracheostomy. The disorder has recently been shown to result from loss-of-function mutations in the extracellular matrix protein 1 gene on chromosome 1q21. The function of the protein extracellular matrix protein 1 gene is still unclear; although an important role in skin physiology and homeostasis has been hypothesized. Histological changes of late infiltrative lesions of lipid proteinosis are well documented in the literature. The most obvious change is the deposition of PAS positive and diastase resistant hyaline material around blood vessels and appendages. However, there is paucity of reports describing the histological characterization of the early stage of lipid proteinosis. Vesiculobullous disorders in a child can be a diagnostic challenge. Common causes of blisters in early childhood include genodermatoses like epidermolysis bullosa and infections like herpes simplex. LiP may rarely present with vesiculobullous lesions in childhood. Most of the cases reported so far have been from South Africa and Central Europe, few cases have also been reported from India.

We report a child, who presented in early childhood with blistering dermatosis. Although this rare autosomal recessive disorder has been described in the literature, its occurrence is rare in India.

A high index of suspicion is required when one deals with blistering dermatosis in a child and various authors have reported many case with vesiculobullous presentation of lipid proteinosis. In few brain damage develops over time and is associated with the development of mental retardation and epileptic seizures. The gene responsible for LiP has recently been identified. It performs an unknown function in the skin related to the production of collagen. There is no cure for LiP. Some dermatologists have had success treating the skin eruptions with oral steroid drugs and oral dimethyl sulphoxide (DMSO). Carbon dioxide laser surgery of thickened vocal cords and eyelid lesions proved helpful in some studies. Dermabrasion may improve the appearance of the skin lesions. But practically no treatment is satisfactory.

Children with LiP may have behavioral or learning difficulties, along with seizures. Obstruction in the throat rarely may require a tracheostomy. Mortality rates in infants and adults are slightly increased because of problems with throat obstructions and upper respiratory tract infection and seizures, if present, may be treated with anticonvulsants.

To conclude, we emphasize the fact that vesiculobullous lesions may be presenting feature without other classical manifestations and can be misdiagnosed as a case of epidermolysis bullosa or any other blistering dermatoses common in infants. Typical lesions of lipid proteinosis may develop later in subsequent years, and so they need to be followed up for long time. Children with this condition in their pre adolescent and adolescent period develop lot of psychological stigma and mental distress, which need to be addressed at earliest and parents be counseled adequately.

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