CASE REPORTS

LIPOID PROTEINOSIS

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A classical case of lipoid proteinosis in a 17-year-old girl presenting with hoarseness of voice, thickened tongue, bullous lesions predominantly on pressure areas and varioliform scarring on the face and the extremities is reported. Her x-ray skull showed typical bean-shaped calcification in the limbic area. Histopathology revealed PAS stained amorphous deposits in the dermis and around the sweat glands.

Key words : Lipoid proteinosis, Urbach-Weithe disease, Hyalinosis cutis et mucosae.

Lipoid proteinosis is a rare genetic disease primarily involving the skin and mucous membranes. It has been described under different names such as hyalinosis cutis et mucosae, Urbach-Wiethe disease etc. Urbach[1] in 1929 first described the comprehensive clinical and histopathological features of this disorder. Thambiah et al[2] recorded the first case report from India. There have been a few other reports[3-7] of lipoid proteinosis in the Indian literature. We are presenting another case with all the classical features of the disease.

Case Report

A 17-year-old female had recurrent bullous lesions all over body since 5 months of her age, hoarseness of voice and inability to open her mouth widely and protrude the tongue out since 5 years of her age. She was born to non-consanguineous parents after full-term normal gestation by normal vaginal delivery. The mile stones were normal. She used to get vesicular lesions which first occurred on the legs over apparently normal looking skin and ruptured in 7-10 days and healed by leaving behind atrophic scars. Lesions occurred predominantly on the pressure areas such as the elbows, the knees, the buttocks and the hands. There was no itching either preceding or after the appearance of the lesions. There was no difficulty in breathing or deglutition. One year later, she developed difficulty in opening the mouth widely and protrude the tongue out. There was no history of photosensitivity or epilepsy or red coloured urine. No other member in the family had similar complaints.

Examination revealed a single flaccid bulla, 1 cm in size, containing hemorrhagic fluid with an erythematous halo around it over the lateral aspect of right leg. Multiple varioliform scars were seen all over her body including the face. There were skin coloured, discrete, firm papules over the malar areas, medial canthi and the eyelids giving rise to a string of beads appearance (Fig. 1). Similar papular lesions were present over the back of the trunk and the interscapular regions. The skin over the cubital fossae was thickened with prominent skin lines (Fig. 2). The mucous membrane over the lower lip was thickened. Discrete indurated papules were seen over the angles of the mouth, the lowr lip and on both sides of the frenulum of tongue. The oral mucosa was pale. Tongue was enlarged,

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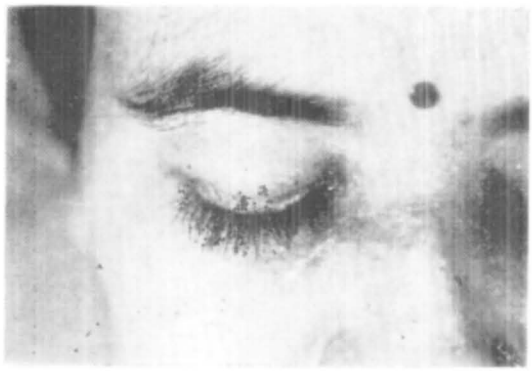

Fig. 1. Beaded papules on upper eyelid, varioliform scars on nose and cheek.

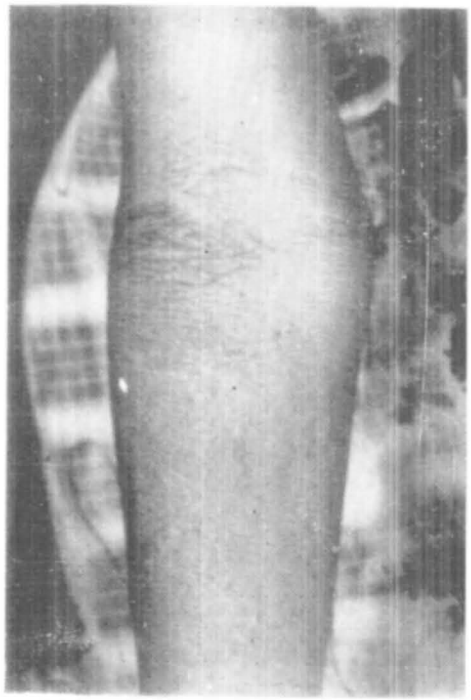

Fig. 2. Thickened, rugose skin in cubital fossa. Note pitted scars on the forearm.

firm in consistency and mobility was restricted. The genital mucosa was normal. Microlaryngoscopy showed plaque like lesions over the left lateral pharyngeal wall, the posterior part of the left vocal cord and the anterior part of the right vocal cord and on the anterior commissures. The subglottic mucosa was grossly thickened. Hair, nails and teeth were normal.

Routine investigations such as haemogram, serum electrolytes, blood urea, blood sugar, liver function tests and lipid profile were within normal limits. Tests for porphyrins in the urine, the faeces and the blood were negative. Fundoscopy revealed no abnormality but for a persistent pupillary membrane in both eyes. The lateral view of the skiagram of skull showed a bean shaped calcification over the limbic area (Fig. 3).

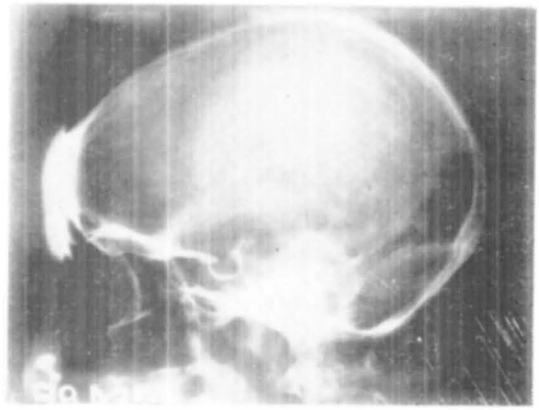

Fig. 3. Bean shaped calcification in the limbic area, lateral skiagram of skull.

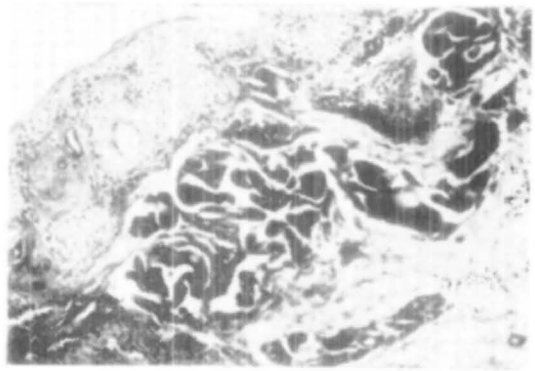

Fig. 4. Massive deposits of amorphous material in the upper dermis (H & E x 400).

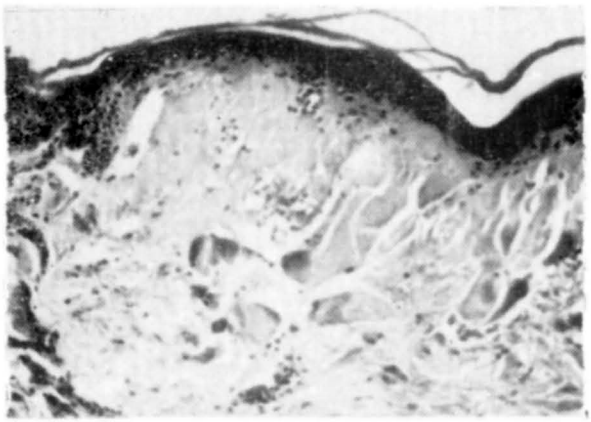

Fig. 5. Mucosal biopsy showing similar deposits in the upper dermis, around blood vessels (FAS x 400).

The skin biopsies from the forearm, the neck and from the lip mucosa all showed typical features of lipoid proteinosis (Figs. 4 and 5). There were large, amorphous deposits in the upper dermis, more so in the dermal papillae. Similar material was also seen surrounding the capillaries and sweat glands. The hyaline material was positive for PAS and congo red stains. Biopsies from the larynx and the lateral pharyngeal wall also showed deposits of similar material.

Comments

The present case has all the classical features such as hoarseness of voice, discrete firm papules over the free margin of eyelids giving rise to the string of beads appearance, the thickening of skin in the flexural areas, discrete firm papulonodular lesions over the laryngeal mucosa, a bean shaped calcification in the limbic area and sub-epidermal hyaline deposits. The pathogenesis of the disease is unknown. The earliest reports described it as a systemic metabolic derangement of lipid.[8] Later, it was suggested that it was due to combined disturbances of protein and fat metabolism.[9] Bauer[10] postulated that it is a lysosomal storage disease due to a single or multiple enzyme defects.

Fleischmajer et al[11] described three major histopathological alterations in the upper dermis, i.e. massive deposition of amorphous material between the collagen fibrils and around the blood vessels, a considerable thickening of the basal laminae, and marked reduction in the number and the size of collagen fibrils. Similar changes were described in erythropoietic protoporphyria (EPP). However, these patients lack hyaline deposits. Hyaline bundles were seen perpendicular to the epidermis and these surrounded the eccrine sweat glands and characteristically replaced them.[12] Hair follicles and arrectores are also surrounded by hyaline. The chemical nature of hyaline is unknown, though it has been suggested to be a glycoprotein, a glycolipid, an acid mucopolysaccharide or altered collagen/elastic tissue.[13] Their histochemical characteristics vary from site to site.

Autosomal recessive transmission has been recorded.[14,15] However, in our case, there was no evidence to suggest the same. Parotid swellings due to parotitis have been described in some cases. This has been directly linked to infiltration of the buccal mucosa, stenosis of Stensen's duct and consequent retrograde infection.[14] However, in our study no such swellings were noted.

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