LIPOID PROTEINOSIS
(Urbach - Wiethe Disease)
A Case Report

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Summary

A case of LIPOID PROTEINOSIS is reported.

KEY WORDS: Urbach-Wiethe Disease, Hyalinosis cutis et mucosae, Lipoglycoproteinosis.

Lipoid proteinosis is an extremely rare, autosomal recessive[1] condition showing widespread papules, nodules, indurated plaques and ulcerated lesions involving the skin and the mucous membranes. Hoarseness is a prominent symptom, due to involvement of vocal cords and the upper respiratory tract. The lipoid material, protein and carbohydrates have been reported in the extra cellular spaces and walls of the blood vessels. Since 1929, when Urbach-Wiethe first reported the disease, only 150 cases have appeared in the world literature[1]. Five cases have been reported from India[2-4] but none from Rajasthan.

The following case emphasizes the principal clinical and histopathological picture of the disease.

Case History

A fifteen year old muslim male from Pali (Rajasthan) was admitted to the Medical College Hospital, Udaipur in the Department of Dermatology, S.T.D. and Leprosy for evaluation of his unusual chronic skin disease. He was the eldest of 4 children born to consanguinous parents (uncle-niece marriage). The patient's mother had noticed the baby having a hoarse cry at the age of three months and since then the hoarseness had been gradually worsening. The mother noticed erythematous maculopapular cutaneous lesions at the age of nine months, which generally healed with the formation of a thin scar. Since then the patient had been repeatedly getting such lesions which were often complicated with secondary infection. Itching of variable intensity had been present from time to time.

At the time of admission the patient complained of loss of hair in the occipital region, repeated ulcerations in the oral cavity with inability to open the mouth fully, difficulty in swallowing solids and dyspnoea on even slight exertion. There was no history of convulsions at any time. There was no family history of similar problem.

General physical examination revealed enlarged and tender cervical, axillary and inguinal lymph glands. Few of the glands in the inguinal region were matted. Detailed examination of cardiovascular system, respiratory system

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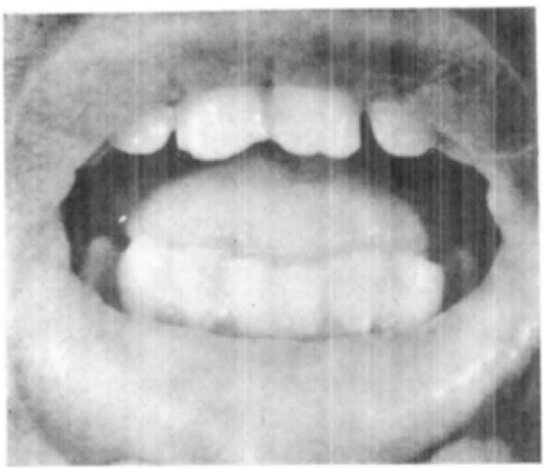

Fig. 1
Photograph showing mucous and oral lesion. Tongue could not be protruded.

and central nervous system revealed no abnormality. Indirect laryngoscopy was not possible because of the gross thickening of the pharyngeal wall and consequent inability to open the mouth fully. Direct laryngoscopy under general anaesthesia revealed a yellowish, pale and swollen epiglottis. Arytenoepiglottic fold was thickened and pale yellowish in color. A swelling was present at the inter-arytenoid region which was also pale yellowish in color and measuring one centimeter in diameter. The oral mucosa was eroded, yellowish and pale. A small pearly nodule was seen over the inner aspect of the lower lip. Marked restriction in mobility of the tongue was seen (ankyloglossia, Fig. 1). Chronic fissures were present at the angles of the mouth.

Generalized papulonodular induration was seen over the skin. The surface at some places was covered by verrucous and hyperkeratotic material specially on the buttocks, back, knees, elbows and nape of the neck. At numerous places there was impetiginization. Many areas on the skin showed atrophy (Fig. 2). Palms and soles were conspicuously uninvolved. There was patchy alopecia on the occipital region (Fig. 3) and partial loss of eye lashes. While in hospital, patient had an attack of acute right sided parotitis with fever.

Investigations

Routine examination of urine and stool did not reveal any abnormality. Blood counts showed leukocytosis of 19,150 c/mm. Urine, stool and blood were negative for protoporphyrins. Serum cholesterol was 200 mg% total serum proteins 6.2 gm% (Albumin 3.5 gms and globulins 2.7 gms). Serum alkaline phosphatase, S.G.O.T., S.G.

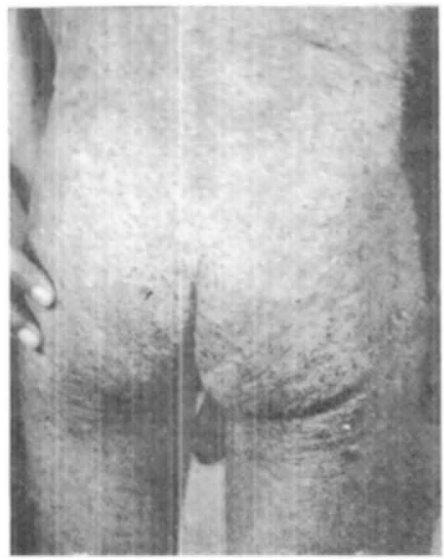

Fig. 2
Photograph showing skin lesions.

P.T., serum bilirubin, creatine, creatinine and blood sugar were within normal limits. X-ray skull showed suprasellar calcification (Fig. 4). Fluoroscopic examination of chest and barium meal studies of G.I.T. were normal. X-rays of teeth were normal. The biopsy taken from skin and the swelling at the interarytenoid region showed PAS positive hyaline material in dermis and submucosa respectively.

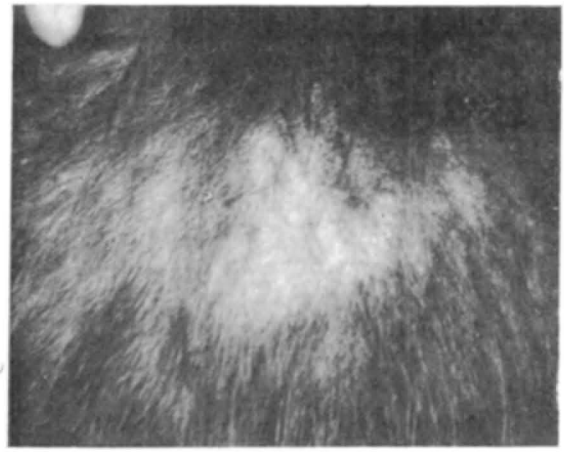

Fig. 3
Photograph showing Alopecia in occipital region.

Discussion

Lipoid proteinosis (Urbach-Wiethe disease) is an extremely rare inherited metabolic disorder. The disease is characterized by nodules, papules and plaques, involving primarily skin and mucous membranes. It has now been recognized as a systemic disorder with generalized visceral involvement[6-9] and because of significant involvement of the skin, the disease is of interest to the dermatologists. The disease is usually first noticed by parents because of child's inability to cry. As the speech develops the child only whispers because of the laryngeal involvement, [3,4,6,10-13]. Some workers have reported lesions on palms and soles but these were absent in our case[14]. The dental abnormalities reported are congenital absence of upper lateral incisors, lower lateral incisors and second bicuspids[8]. Though the two lower lateral incisors

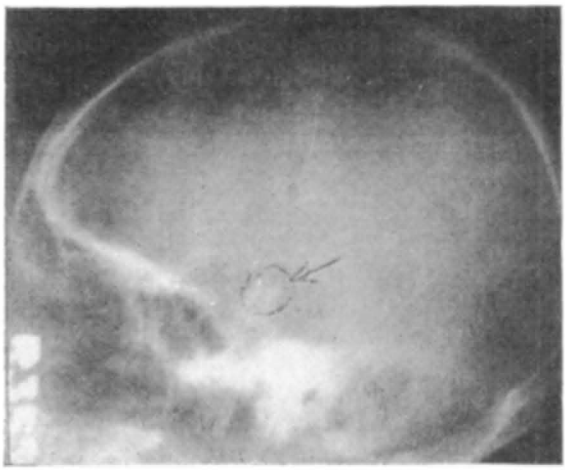

Fig. 4
Photograph showing suprasellar calcification in lateral view of X-ray skull.

were missing in our patient, these had fallen off after erupting normally. The characteristic papulonodular infiltration of lid margins were present in our patient but other reported ocular manifestations like trichiasis, corneal ulceration[12], eccentric pupils[15] and colloid degeneration of retina were not noted.

The oropharyngeal and laryngeal involvement is one of the important clinical signs reported by almost all the workers. These were seen in our case in the form of ankyloglossia, repeated stomatitis, dysphagia, and hoarseness of voice. These symptoms are due to the infiltration in the oral and laryngeal mucosa as pale and pink to yellow papulonodules which imparted a cobblestone appearance. Lesions in oesophagus, stomach, rectum, labia minora and vagina have been reported by Grosfeld et al[1]. Repeated parotitis is reported and is attributed to infiltration at the mouth of the Stensen's duct[10,11]. The additional signs and symptoms like retarded hair growth and associated partial alopecia, were seen in our case. No photosensitivity was recorded in our case. An important diagnostic clue, the intracranial calcification, reported by several workers [1,4,7-9,10-13,16-18] was present in our case. This calcification is suprasellar and is easily seen in radiographs. In suprasellar region the calcification seen may be present in hippocampal gyrus or tentorium. Epilepsy, secondary to this calcification has been reported[1].

The histopathological finding in our case showed PAS positive material in the perivascular region.

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