Lipoid proteinosis

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Lipoid proteinosis is an extremely rare and striking disorder characterized by a peculiar infiltration of the skin and mucous membranes (Rook, 1968). Siebenmann (1908) was the first to describe the disease in a 19-year-old girl, having hyperkeratosis of skin and affection of the mucosa of the mouth, tongue, palate and larynx.

The nature of the underlying metabolic defect is unknown. The disease affects both sexes equally and is often familial. It is probably determined by an autosomal recessive gene. Dental anomalies and consanguinous marriages were common in a group of 9 cases described by Urbach and Weithe (1929).

The condition develops early in life with hoarseness which becomes prominent within the first few years. The laryngeal changes are often progressive and may necessitate tracheostomy. In addition, the mucosa of the pharynx, tongue and lips develop firm yellow-white infiltrates. The tongue is enlarged and feels firm on palpation and its range of movement is decreased. Occasionally there is similar involvement of the mucosa of the labia and vagina.

Skin changes, in various forms, appear constantly by the age of 12 years. The scalp shows diffuse non-scarring alopecia. Nodular lesions occur on the lid margins, on the face, where they may lead to pock marks; and on the elbows and knees, where they may progress to form warty hyperkeratotic plaques. There are infiltrated plaques at the angles of the mouth and in the gluteal cleft. Atrophic patches may be found on the face and perifollicular atrophy of the neck and upper arms produces an appearance similar to that of pseudoxanthoma elasticum. A 'diabetic tendency' and a family history of diabetes mellitus have been reported. Endocranial calcification with or without the association of epilepsy have also been described (Cowan *et al.*, 1961).

Case report 1

A 35-year-old male, H.I., working as a weaver, came to the E.N.T. Department, Kasr El Aini Hospital, in September 1967, complaining of hoarseness of voice since childhood; the hoarseness was marked in the last few years. No dysphagia was accompanying the condition. No past history suggestive of syphilis, tuberculosis, or epilepsy. No similar condition in the family. Examination, revealed waxy wrinkled face. On the forehead he had slightly atrophic lesion, the eyebrows were normal but the eyelashes were scanty and the upper lid margins were studded with pearly, bead like lesion. The margins of his nostrils were thickened and he had infiltrated plaques at the corners of the mouth with fissuring (Fig. 1). On the neck and chest, there were yellowish follicular papules with slight perifollicular atrophy (Fig. 2). E.N.T. examination revealed crusting of the vestibules with some stenosis of both nostrils. Nasal mucosa was atrophic. Submucosal infiltrations of the hard palate and scarring of the soft palate.

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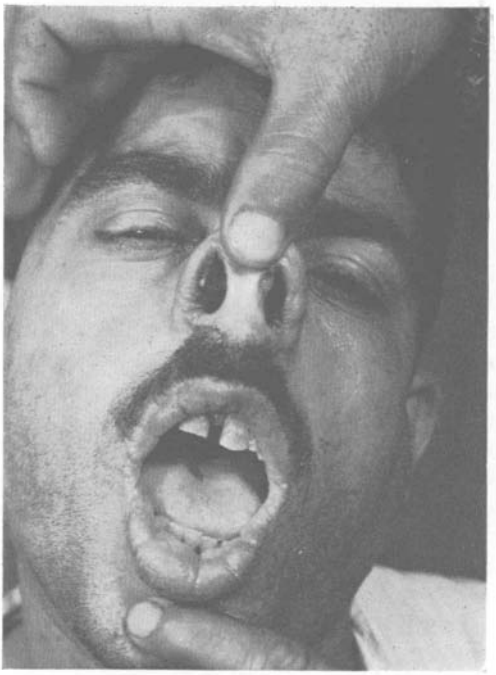


FIG. 1.
Photograph of Patient No. 1, showing thickened lips with fissuring infiltration of the margin of upper eyelid and fissuring of the nostrils.

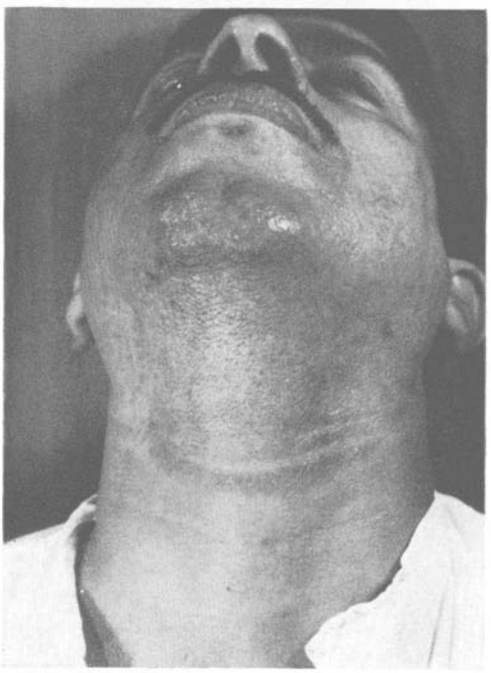


FIG. 2.
Showing infiltrations of the skin of the neck in a collar form.

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Indirect laryngoscopy showed swollen and thickened epiglottis which was folded upon itself. There was thickening of the arytenoids. Both vocal cords were thickened.

Investigations

Blood picture; haemoglobin 73 per cent (Sahli).
Red cell count 4,320,000 per c.mm.
Total leucocytic count 10,200 per c.mm.
Differential.
Basophils 0 per cent; Eosinophils 0 per cent; Staff nucleated 3 per cent; Segmented 72 per cent; Lymphocytes 20 per cent; Monocytes 5 per cent.
Sedimentation rate:
First hour 14; Second hour 32.
Urine examination was normal.
Wassermann: Negative.
Carbohydrate tolerance curve; Fasting 88, after ½ hour 111, after 1 hour 153, after 2 hours 80.
Total serum proteins were 6·5 g./100 ml.
Plasma: total cholesterol 160 mg./100 ml.
Total Lipids: 810 per cent.

Histopathology

Biopsy specimen was taken from an infiltrated area of the skin of forehead, fixed in 10 per cent neutral buffered formalin. Paraffin sections were made. There was flattening of the epidermis with small widely separated rete pegs. Discrete areas of structureless eosinophil material found in the dermis. In the adjacent stroma many of the capillaries were surrounded by a narrow sharply defined concentric layer of similar hyaline material (Fig. 3). The results with congo red and methyl violet were negative for amyloid. The hyaline material showed PAS positive reaction.

Case report 2

A female student named A.M., aged 13-years, came to the E.N.T. Department, Kasr El Aini Hospital, in February 1968, complaining of hoarseness of voice since she was 3 years old. The condition was accompanied by dryness of the mouth and the appearance of brownish skin lesions. No past history suggestive of syphilis or tuberculosis. Family history: not contributary.

Examination

The patient was underweight, brownish skin lesions variable in size mostly small, the size of a pin's head were observed. The skin at the flexures was roughened with brownish discoloration (Fig. 4). The lid margins were infiltrated. The lips were thickened, pale and fissured. The tongue was infiltrated and of limited mobility (Fig. 5). The palate is pale and infiltrated. The tonsils were moderately enlarged with irregular surface and exaggerated crypts.

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FIG. 3.
Biopsy of skin lesion H and E × 100 showing perivascular infiltration of lipoid material.



FIG. 4.
Showing warty hyperkeratotic plaques on elbow.

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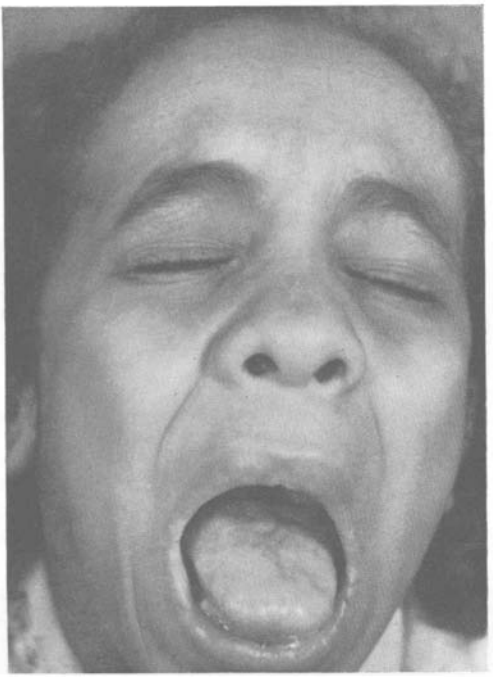


FIG. 5.
Face waxy, tongue infiltrated, mobility impaired.

Indirect laryngoscopy: revealed swollen thickened epiglottis, cords were thickened and freely mobile.

No liver enlargement.

No spleen enlargement.

No glandular enlargement.

Investigations

Blood picture, haemoglobin 71 per cent; white-cell count 7,600 c.mm. (normal differential count).

Wassermann: Negative.

Total protein in blood 6·54 g. per cent.

Urine free.

Histopathology

Biopsy was taken from the skin lesion and revealed the same picture as described before.

Discussion

Most of the classical features of lipoid proteinosis were present in our two patients except parental consanguinity, diabetes mellitus and involvement of the vaginal and anal mucosa in the second case.

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The histological picture is typical with that described by Laymon and Hill (1957). The nature of the hyaline material is not well known but what can be said is that it is not amyloid. The study of the literature indicates that, unlike primary amyloidosis, the prognosis of lipoid proteinosis is good. Some cases may require tracheotomy for their severe laryngeal involvement. Cortisone therapy was tried with little effect.

Summary

Two cases of lipoid proteinosis were presented. Histopathological and histochemical studies supported the belief that the disease is primarily an infiltrative process.

Acknowledgement

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