LIPIDPROTEINOSIS (URBACH-WIETHE SYNDROME)*

BY

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SIEBENMANN (1908) described lipidproteinosis, and the condition was subsequently reported as a clinical entity by Urbach and Wiethe (1929). During infancy and early childhood a structureless eosinophilic substance develops under the epithelium of the skin and upper respiratory tract. Hoarseness is followed by a skin eruption giving a pale, yellow-brown, pock-marked appearance, especially on the face and scalp, with the extensor surfaces (e.g. the elbows) showing hyperkeratotic lesions. Localized deposits appear as white plaques on the pharyngeal wall and on the tongue which becomes more and more immobilized. Respiratory distress due to laryngeal stenosis in later life may require tracheostomy, but otherwise the prognosis is good.

The lid margins usually show discrete nodules resembling solid beads of yellow-brown wax among the lashes. In time these can reach up to 5 mm. in diameter and approach the posterior borders of the lid edges, but the conjunctiva is not involved.

Case Report

An intelligent boy aged 14 years, 4' 9" in height, came of unrelated parents. His father died of "heart trouble" and his mother after a perforated "gastric ulcer". His brother and other relations are normal and do not bruise easily (Cowan, Alexander, Vickers, and Cawdell, 1961). Hoarseness and skin lesions have been present since infancy.

At the age of 9 years trichiasis caused ulceration of the right cornea which healed with scarring (Fig. 1), the visual acuity being reduced to 6/36.

Epilation of ingrowing lashes prevented similar injury to the left eye, and the visual acuity is 6/6. The lid margins show typical beading (Fig. 2), and lashes are present, being even hypertrophic on the upper lids.

FIG. 1.

FIG. 2.

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The fundi are normal without angioid streaks. The facial skin shows the typical depressed pale scars in a light-brown translucent surround, giving a pock-marked appearance (Fig. 3).

FIG. 3(a).

FIG. 3(b).

The skin elsewhere is clinically normal, apart from an appearance of atrophy and premature ageing in the creases of the neck, axillae, and antecubital fossae, while the extensor surfaces of the hands and elbows show hyperkeratosis (Fig. 4). It is also easy to see typical changes in the tongue (Fig. 5).

FIG. 4.

FIG. 5.

Laryngoscopy revealed that the edges of the vocal cords were irregular and white. Biopsy from a white plaque on the posterior pharyngeal wall demonstrated amorphous eosinophilic material beneath the epithelium, replacing the connective tissue even into the papillae (Fig. 6a, b, overleaf). No foam cells or giant cells were to be seen.

FIG. 6(a).

FIG. 6(b).

Two trials of steroids in therapeutic dosage at 9 and 14 years did not improve the voice or skin lesions (Cowan and others, 1961).

Laboratory Investigations.—X rays of chest, hands, feet, and skull were negative. Serum lipoproteins (beta): normal; serum lipids: 708 mg. per cent.; serum cholesterol: 332 mg. per cent.; serum electrophoresis: normal protein pattern. Blood sugar (fasting): 93 and 115 mg. per cent.; glucose tolerance: normal; serum creatinine: 0.8 mg. per cent.; blood count: normal.

Discussion

This combination of hoarseness, pock-marked skin with beading of the lid margins, and thickened mucosa of the tongue is easily recognized despite its rarity. The lid lesions are usually regarded as being of no significance apart from diagnosis (Blodi, Whinery, and Hendricks, 1960), and no instance of corneal ulceration due to trichiasis appears to have been reported.

The importance of examining the lid edges for ingrowing lashes is not realized, but is clearly demonstrated in the present case.

Isolated reports of the following ocular lesions in association with lipid-proteinosis are quoted by Blodi and others (1960): degeneration and drusen at the macula, chorio-retinitis, and a conjunctival nodule.

Inheritance is by a recessive gene (François, 1960), familial occurrence being more likely in consanguineous marriages.

Various changes in blood chemistry are described. The serum lipids and cholesterol are usually normal but phosphatides and lipoproteins may be increased. Hyperglycaemia has been reported but diabetes mellitus seldom develops. Although Blodi and others (1960) describe anomalies of the serum proteins, these are not always found. The only abnormal blood finding in the present case was the raised serum cholesterol level.

Histochemical staining fails to give reactions typical of amyloid, but various staining reactions suggest the presence of lipids. P.A.S. gives marked staining which is not affected by digestion with amylase (Cowan and others, 1961).

These authors describe intracranial calcification with or without epilepsy. In the patient described there was neither a history of epilepsy nor x-ray findings of calcification within the skull.

The possibility of unsuspected ocular abnormalities was checked. The anterior chamber angles were within normal limits and ectopia lentis was not present. The latter was suggested because of a recent report in which striae albicantes accompanied ectopia lentis (Kachele, 1960).

A disease which may cause difficulty in differential diagnosis is disseminated xanthomatosis (Montgomery and Osterberg, 1938). This usually begins in adult life and affects the upper respiratory mucosae and skin; papules, at first reddish but later yellowish-brown, may appear at the lid edges (Spiegel, 1938), but especially in the flexural creases and axillae; conjunctival infiltration and diabetes insipidus can also occur. On the other hand, lipidproteinosis begins in infancy, prefers the extensor surfaces of joints, does not involve the conjunctiva, and is occasionally accompanied by diabetes mellitus, but never by diabetes insipidus.

Summary

Corneal ulceration due to trichiasis in a case of lipidproteinosis (Urbach-Wiethe syndrome) is described. The syndrome is reviewed and its differential diagnosis from disseminated xanthomatosis briefly discussed.

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