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XERODERMA PIGMENTOSUM WITH OCULAR COMPLICATIONS*

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XERODERMA pigmentosum was first described by Kaposi in 1870 (quoted by Duke-Elder, 1938). Jensen (1962) estimated that up to that date about 900 cases had been recorded. The condition is said to be rare in Negroes because of the presence of the cutaneous pigment, but has been described in about eight cases (Targowsky and Loewenthal, 1956).

The condition appears in the first years of life, characteristically with pigmented naevi on exposed parts of the skin. These occur mainly on the face, arms, and exposed parts of the body surface, and develop into warty growths, ulcerate, and may become carcinomatous. This assumed actinic sensitivity may be transferred as a recessive gene and occur simultaneously in several members of a family; consanguinity of the parents is an important factor and the condition appears to be sex-linked (François, 1961). The dermal manifestations of xeroderma pigmentosum may be confused with those of arsenical poisoning (Hopkins and van Studdiford, 1934).

The eyelids are frequently involved in xeroderma pigmentosum, and the disease usually becomes malignant in adolescence or early adult life. The skin atrophies and ectropion may occur; the bulbar conjunctiva may become thin and atrophic, with inflamed patches resembling phlyctens; the lesions may become malignant. Blepharospasm and photophobia are common symptoms when the cornea becomes infiltrated.

The skin changes characteristic of xeroderma pigmentosum are hyperkeratosis, atrophy of the Malpighian layer, perivascular lymphocytic infiltration, scattered melanophages in the underlying connective tissue, and accumulation of melanin granules in the basal layer. This disease is complicated by basal cell carcinoma, more rarely by squamous cell carcinoma.

Case Report

A 2-year-old Bantu girl, seen at the St. John Ophthalmic Hospital, complained of severe lacrimation. The history revealed no parental consanguinity and three siblings were healthy. No history of arsenical poisoning could be elicited from the mother.

Examination.—Marked photophobia and blepharospasm were evident. The hands, face, neck, feet, thorax, and abdomen showed pigmented naevi not raised above the

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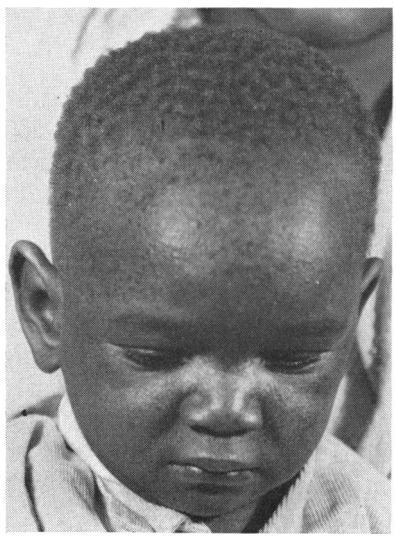
surface of the skin, of average size 3 mm. diameter (Fig. 1). A few eyelashes were absent, and there was ectropion of the lower lids; the left lower lid showed a squamous carcinoma. The conjunctiva was dry and atrophic and slightly pigmented (a sign described by Markowitz, 1935); the lesions had the appearance and distribution of the bulbar form of spring catarrh. A pannus-like formation was observed at 7 o'clock in the left eye (Fig. 2). At the limbus (in relation to this pannus) there was a phlyctenular ulcer with associated corneal oedema; it was raised, vascular, friable, with a roughened surface, and invaded the cornea.



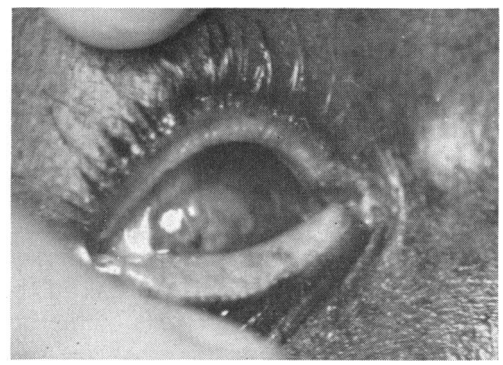


FIG. 2.—Limbal tumour in the left eye.

FIG. 1.—Facial distribution of pigmentation, in an infant aged 2 years, with xeroderma pigmentosum.

Laboratory Investigations.—There was no evidence of leukoplakia or of a lingual tumour (Loewenthal and Trowell, 1938). The cardiovascular, pulmonary, abdominal, and nervous systems showed no abnormality.

Under general anaesthesia biopsies were taken of a naevus of the skin of the right upper eyelid, of the warty growth on the left lower lid, and of the tumour-mass at the limbus.

The skin biopsy showed a slight degree of hyperkeratosis; the interpapillary pegs were normal; the pigment was enormously increased and almost obscured the deeper layers of epidermis; the corium showed fragmentation of the connective tissue and increased pigmentation; the pigment occurred in coarse clumps; there was perivascular lymphocytic infiltration.

The warty mass showed the changes of squamous carcinoma. It consisted of atypical, pleomorphic prickle cells and epithelial pearls, composed of concentric layers of keratinized tumour cells (Fig. 3, opposite).

The phlyctenoid area near the limbus showed early signs of malignant degeneration.

Treatment of Xeroderma Pigmentosum

Various creams containing titanium dioxide have been suggested as a protection against sunlight (Smithers and Wood, 1952); steroids have also been used with varying success.

The malignant tumours may be excised or treated with radiation.

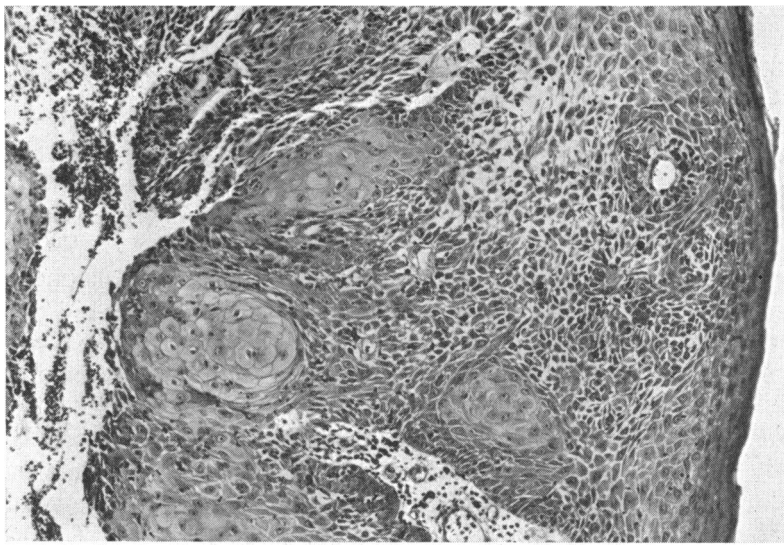


FIG. 3.—Section of skin from right upper lid, showing squamous carcinoma. × 90.

Discussion

The Negro is believed to be immune to xeroderma pigmentosum, because his pigmentation protects him from light, but this has been shown to be incorrect (Targowsky and Loewenthal, 1956; Loewenthal and Trowell, 1938).

It is customary for Bantu children to be exposed completely to the sun's rays, and this accounts for the interesting feature in the case reported here of the presence of pigmented naevi on the thorax and abdomen.

Xeroderma pigmentosum is probably an inborn abnormality which includes hypersensitivity to light of the skin and possibly of the eyes as well.

The prognosis of xeroderma pigmentosum is poor, death occurring most often from meningitis or haemorrhage.

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