PSEUDOXANTHOMA ELASTICUM (PXE) - REPORT OF THREE CASES

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Pseudoxanthoma Elasticum (PXE) is a hereditary disorder characterized by generalized defect of elastic tissue. Symptoms are multiple and varied. The disease is most often recognized because of cutaneous lesions. Ocular disturbances are also common. Involvement of cardiovascular system causes a great variety of problems.

Balzer (1884) presented the first autopsy report of PXE in which he described the skin changes. He considered this disorder to be one of the Xanthomatoses. Darier (1896) established this disease as a separate entity by describing the histopathology and offered the name PXE. There have been less than 400 cases of PXE reported in the literature since it was first described by Balzer (Karth et al, 1964). Only a few case reports of PXE are available in Indian literature (Thambiah et al, 1964; Krishnan Kutty and Sadasivam–1965; and Mulay and Bikhachandani, 1965). Thus publication of these three cases will be of interest.

CASE REPORTS

Case No. 1 : Nallu, 39 years male was having skin eruption on sides of neck for the past four years but did not give much attention to this because of its symptomless character. Detailed interrogation revealed no manifestations in other systems.

One sister (case 2) and one brother (case 3) of the patient were having similar eruption. History of consanguinity was present in parents. One sister (30 years of age) and one brother (27 years of age) were not showing any skin lesions suggestive of PXE.

Skin around neck except back was lax, rough, and thickened. This configuration of skin extended to upper part of the front of chest. On flexing the neck, the skin was thrown into folds (Fig No. 1) and on stretching the skin yellowish papules were seen. A similar area of skin was seen in left axilla. Left groin showed discrete yellowish papules.

Examination of systems showed no significant abnormality. Ocular fundi did not show any angioid streaks.

Stools did not show occult blood on repeated examinations. Urine revealed no abnormality. Blood smear did not show any sickle cells. Serum cholesterol was 188 mg. % Serum calcium was 11 mg. % Blood Sugar was 85 mg. % E. C. G. was within normal limits. B. M. R. was + 8% Oscillometry of limbs was normal. Psychiatry check up showed no abnormality. Skin Biopsy:—H & E stain shows thin epidermis

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Fig. 1
Showing laxity of the skin of the neck thrown into folds.

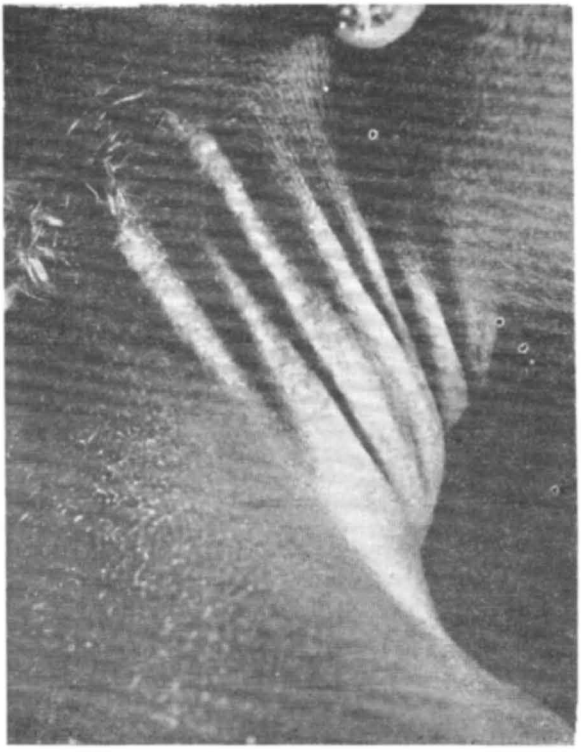

Fig. 2
Showing small yellowish papules over the folds of the skin on the side of the neck.

Fig. 3—Showing small light coloured papules illustrate the changes in the exilla.

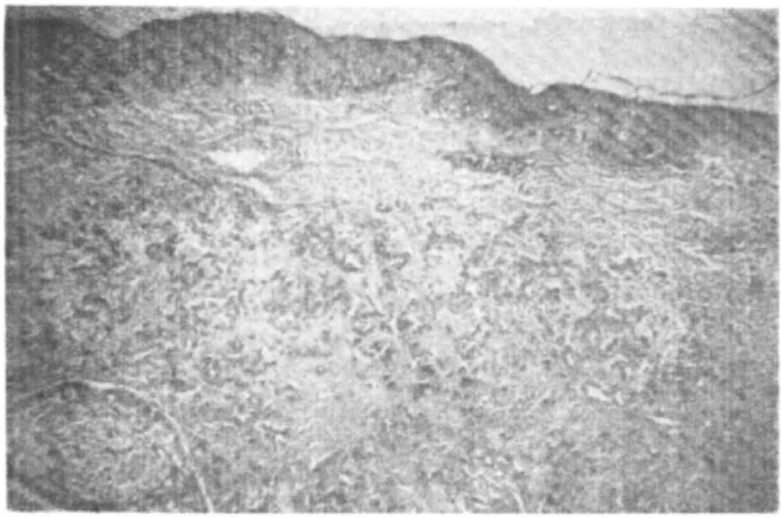

Fig 4—Section of the skin showing spotty calcification in the middle and lower dermis. H & E x C 100.

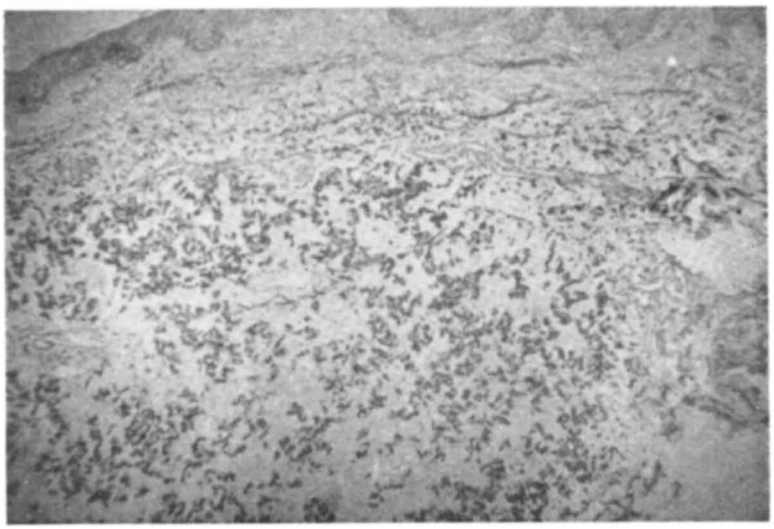

Fig. 5.—Section stained with Verhoeff's stain to show fragmentation of the elastic tissue. x C 100.

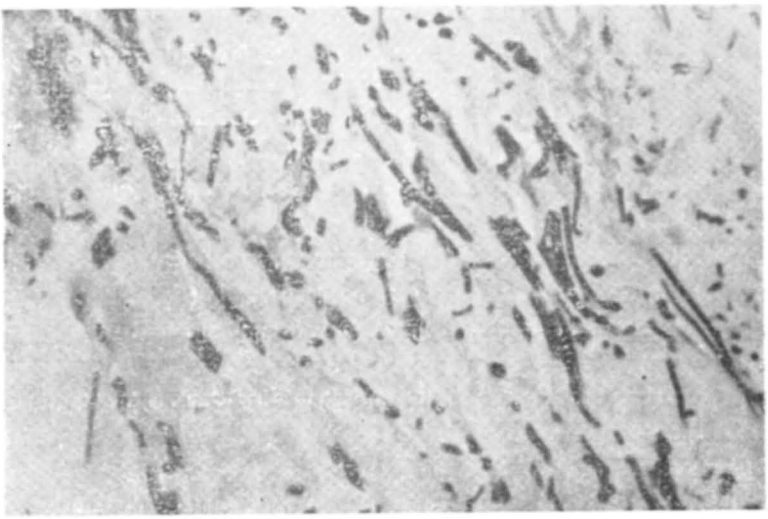

Fig. 6.—Higher magnification of section stained with Verhoeff's stain showing fragmented elastic fibres. x C 500.

with increased pigmentation of the basal cell layer. In the mid-dermis there is collection of blackish granular material (Fig. No. 4). Verhoeff's stain shows increased, fragmented and clumped elastic fibres in the mid and deep dermis. (Fig. No. 5 & 6).

Case No. 2: Panjoli, 41 years female was having skin lesions similar to case 1 on the front and sides of neck (Fig. No. 2), axillae (Fig. No. 3) and groins for the past 7 years. Peripheral blood vessels showed normal pulsations. Examination of eye did not reveal any angioid streaks or any other abnormality in fundus. Laboratory investigations did not reveal any significant abnormality. Histopathology of skin revealed same abnormal features as in Case 1.

Case No. 3: Kumaraswamy 43 years male was having similar lesions on the front and sides of neck only for the past 7 years. Peripheral blood vessel pulsations were normal. Examination of eyes showed definite angioid streaks in the left eye, and peripapillary choroidal sclerosis with degenerative patches in both eyes. Laboratory investigations did not reveal any significant abnormality. Skin biopsy showed same histopathological features as in the first two cases.

DISCUSSION

Various families described in literature provide a strong evidence that PXE is usually transmitted by an autosomal recessive gene. However, Berlyne et al (1961) thought that in given sibships affected persons were usually either all males or all females. On this basis they proposed that the mode of inheritance is that of partial sex-linkage. Our cases support an autosomal recessive inheritance.

Whether or not the basic defect of PXE is truly elastic tissue degeneration or actually a degeneration of collegen which takes on the histological appearance of elastic tissue, is still a controversial issue.

In this disease the skin, particularly in the neck, axillae and groins becomes flaccid, wrinkled into folds and rather loose; when stretched it shows yellowish marks from which the name PXE is derived. Our patients showed these typical lesions. Angioid streaks are considered characteristic fundoscopic signs in PXE and together with skin lesions constitute Gronblad-Strandberg Syndrome. One of our patients showed angioid streaks in left eye.

Although we were unable to find any other abnormalities in our patients, many have been reported and some are likely to occur later in life. Hence such patients should be thoroughly examined and investigated and should be under regular and constant observation.

Recurring upper gastro-intestinal haemorrhages is a serious problem reported in patients with PXE.

SUMMARY

A family of PXE supporting an autosomal recessive inheritance is reported.

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