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PSEUDOXANTHOMA ELASTICUM (GRONBLAD-STRANDBERG DISEASE) WITH CORONARY ARTERY CALCIFICATION

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The literature on pseudoxanthoma elasticum (PXE) has recently been well reviewed.^{1,2} The purpose of this paper is to describe a case of PXE in a female aged 31, showing the unusual features of calcification of the coronary arteries and falx cerebri and flushing of the cutaneous lesion associated with the episodes of haematemesis.

The fundamental defect in PXE is a degeneration of collagen fibres throughout the body. Degeneration of collagen fibres in the media of the muscular arteries has been shown in the limb, coronary, renal, pancreatic, uterine, cutaneous, mesenteric, carotid, thyroid and choroid-plexus arteries. Calcification of the degenerated material in some of these sites has been demonstrated. Superimposed advanced and premature atherosclerosis and arteriosclerosis occurs in all the arteries. In the skin there is fragmentation of collagen fibres in the deeper and middle zones of the corium. Histological calcification of the degenerated material in the skin also occurs. In the eyes degeneration and tears in the lamina elastica of Bruch's membrane of the choroid appears to be the basis of the angioid streaking. Colloid bodies, choroido-retinitis and fundal arteriosclerosis have been described.

Although vascular calcification has been well documented, as far as we are aware there is no previously described case of calcification of the coronary arteries or falx cerebri in this disease.

CASE HISTORY

European female, aged 31. At the age of 10, a skin condition appeared in the neck, axillae, groins and popliteal and antecubital fossae. The condition has persisted and slowly progressed.

At the ages of 15, 16 and 29 she had fairly severe haematemeses. Barium meal and oesophagoscopy have repeatedly been negative. At the age of 30, she had a 4th haematemesis. During this haematemesis, the rash in the groins became bright red for 10 days and then faded. A 5th haematemesis occurred when she was 31. Three days before this haematemesis a well-marked flush, which lasted for a few hours, appeared between the 'xanthomatous' plaques in the groins and collar area.

Since the age of 12, she has had typical migraine headaches occurring about 7-10 times each year. For 4 years she has had mild intermittent claudication in both calves. There is no history of angina pectoris.

Family History. Her brother aged 34 has had a similar condition

since the age of 3. His skin lesions are very extensive. There has never been gastro-intestinal bleeding. He has recently had a coronary thrombosis from which he has recovered.

On Examination

There are typical PXE lesions in the collar area of her neck, in the groin, and in the popliteal and antecubital areas. There is a similar lesion inside the lower lip (Fig. 2). The skin lesions are crepe-like, raised, yellowish, flat papules 1-3 mm. in diameter on an erythematous skin base (Fig. 3). We found that rubbing or stretching the skin made the skin lesions more prominent. In the areas affected the skin is easily stretched, redundant and lax (Fig. 4), and the folds in the axilla are excessive (Fig. 5).

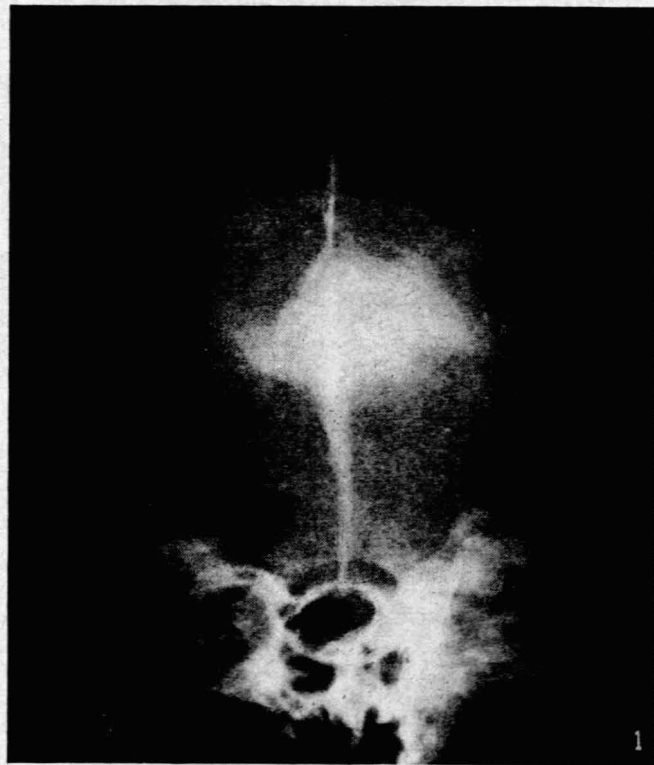


Fig. 1. Radiograph of skull, showing calcification of the falx cerebri.

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The radial pulses are felt with difficulty. Blood pressure in the brachial artery 130/80 mm. Hg. Neither the dorsalis pedis nor the posterior tibial arteries are palpable. The femoral and popliteal pulses are present and there is no delay.

There are typical angioid streaks in the fundi. The fundal arteries and maculae are normal. Visual acuity and visual fields are normal. On physical examination there are no cardiac or neurological abnormalities.

Radiography. The X-ray of the skull shows well-marked calcification of the falx cerebri (Fig. 1). The pineal gland is calcified and normal in situation. There is an area of calcification, slightly larger than the pineal, seen in the lateral view which lies on the same horizontal plane as the pineal and appears to be in the choroid plexus. In addition there is a small annular calcific shadow in the fronto-occipital view suggesting calcification of the right internal carotid artery. The left anterior oblique radiograph of heart shows calcification of the posterior descending branch of the right coronary and possibly the circumflex branches of the left coronary artery (Figs. 6(a) and 6(b)). There is also calcification along the major length of both femoral and post-tibial

arteries. X-ray of the gastro-intestinal tract has repeatedly failed to reveal any cause for the bleeding.

On plethysmographic examination of the lower limbs there is grossly diminished pulsation, diminished response to body heating, and venous occlusion in the vessels supplying the big toes.

Skin biopsy shows typical features of pseudoxanthoma elasticum.

DISCUSSION

This patient presents the classical features of PXE. There are lesions in the skin, fundus oculi, and cardiovascular system. She also illustrates the familial incidence.^{1,3} A preponderance of females has also been noted.^{1,3}

Her presenting symptom on 4 occasions has been haematemesis. As is usual in this condition, barium meals failed to reveal any abnormality. The haematemesis is a local manifestation of the widespread vascular involvement. It is frequently severe and may be fatal.⁴

The patient's skin lesions are typical in their appearance,

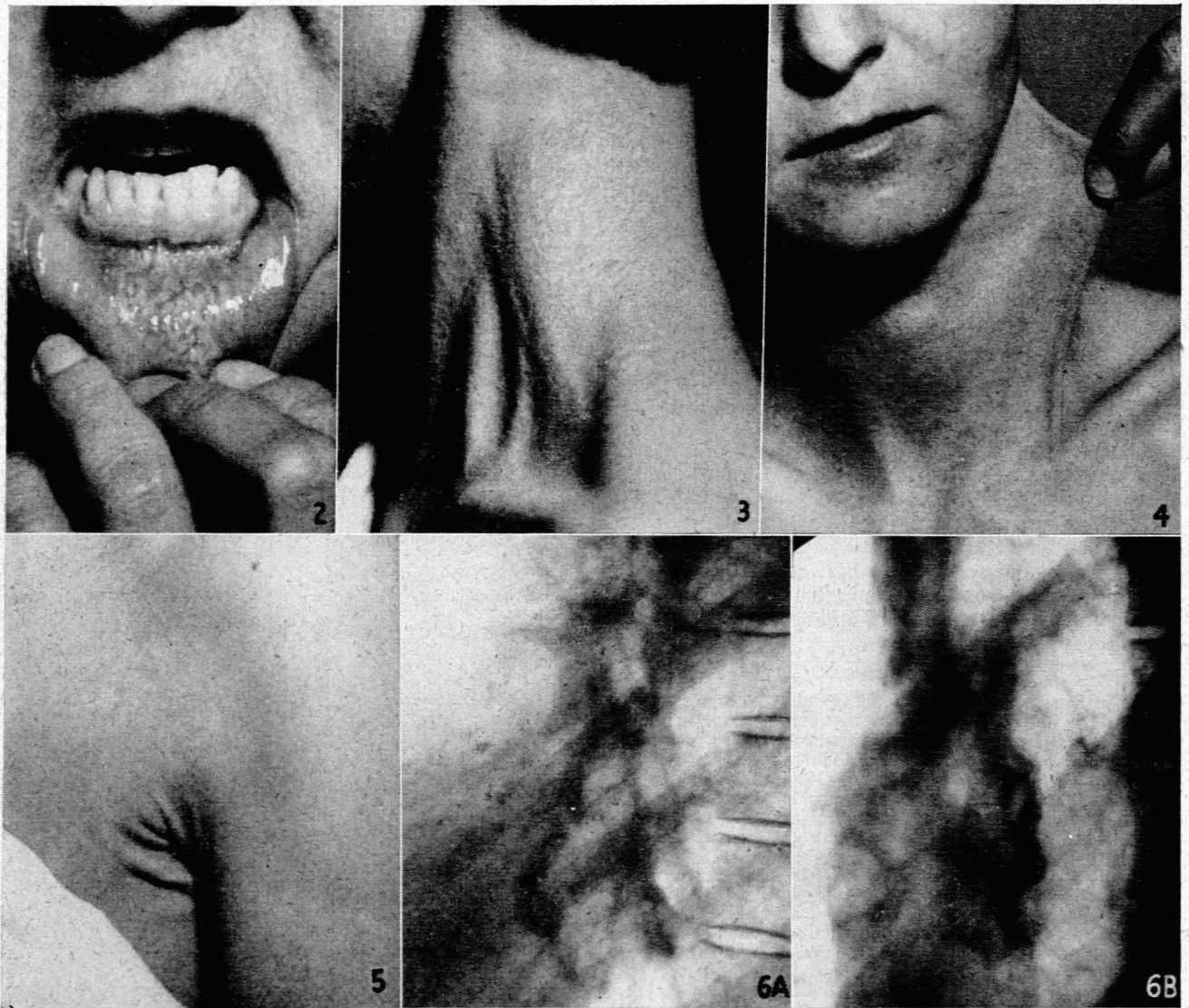


Fig. 2. Buccal mucosa of lower lip, showing typical crepe-like lesion of pseudoxanthoma elasticum. Fig. 3. Left side of neck showing the typical lesion. Fig. 4. Abnormal laxity of skin of the affected area of neck. Fig. 5. Excessive folds of lax skin of left axilla. Fig. 6a & 6b. Left anterior oblique radiographs of heart showing calcification of posterior descending branch of right coronary artery, and possibly the circumflex branch of the left coronary artery.

site and histology. The soft intradermal yellow crepe-like plaques on a slightly erythematous base conform in all respect to Chauffard's classical description.⁵

Flushing of the skin in the affected areas occurred during the episodes of haematemeses. This phenomenon, to our knowledge, has not been described since Chauffard's original reference to it. He described an episode of flushing in which there appeared violaceous rose networks which were not elevated and formed a congested halo around the yellow plaques; but he does not mention the relationship of this flushing to haematemeses. The flushing in our case was directly related to haematemeses and we suggest may serve as a premonitory sign of gastro-intestinal bleeding.

We found that rubbing the affected areas and at the same time slightly stretching the very lax skin, which is present in this disease, made the lesions much more obvious.

Funduscopy showed typical angioid streaking, but no choroidoretinitis or other changes that have previously been described.^{6,7}

There were symptoms of arterial insufficiency in the legs, weak or absent peripheral pulses, gross plethysmographic abnormalities, and radiological vascular calcification. These findings are in keeping with previously described vascular calcification in the peripheral arteries of the arms and legs,^{1, 2, 7} and in the choroid plexus⁸ and internal carotid artery⁹. In our case well-marked calcification was shown in both femoral and post-tibial arteries, in the left internal carotid artery and the choroid plexus.

There was calcification of the falx cerebri; the calcification presumably occurring in the collagen tissue present in this structure. Calcification of this structure does of course occur in normal people.

The calcification in the coronary arteries is a unique feature in this case and, as far as we are aware, has not been previously described. In spite of radiological evidence of gross calcifications there were no symptoms of coronary

insufficiency and the electrocardiograph was normal. Coronary insufficiency and increased incidence of hypertension has been reported in PXE.^{1, 2, 7} The typical arterial collagen degeneration of PXE has been shown in the coronary arteries by numerous workers but there is no report of calcification radiologically or histologically, either in the heart or coronary arteries. Whitish plaques on the atrial and ventricular endocardium and pericardium is well documented² but these were not calcified.

SUMMARY

1. A typical case of PXE pseudoxanthoma elasticum is reported.
2. As far as we are aware radiological calcification of the coronary arteries and falx cerebri are described in this condition for the first time.
3. It is shown that the skin lesions are made more prominent by rubbing.
4. Attention is drawn to flushing of the skin associated with the episodes of haematemeses.
5. The term 'crepe-like' is suggested as a good descriptive term for the skin lesions.

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