PSEUDOXANTHOMA ELASTICUM
(Report of 3 cases with review of literature)

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Summary

Three cases of pseudoxanthoma elasticum are described. One case has features of Gronblad Strandberg syndrome with lepromatous leprosy. The other two cases are reported in siblings. A short review of literature is given.

In 1884 Balzer described the post-mortem findings in a man with advanced pulmonary tuberculosis in whom elastic tissue degeneration was found in the skin and heart which he thought were due to true xanthoma. In 1889 Anatole Chauffard described a 35 years old man with changes in the skin, gastrointestinal haemorrhages, weak peripheral pulses and failing vision. In the same year Doyne described angioid streaks in the retina, so named by Knapp in 1892. In 1896 Darier studied the histopathology of Chauffard's case and named it pseudoxanthoma elasticum. Hallopeau and Laffitte in 1903 described the changes in fundus oculi in Chauffard's case.

In 1929 in Sweden, Gronblad an ophthalmologist and Strandberg a dermatologist together studied and separately reported a patient in whom both pseudoxanthoma elasticum and angioid streaks were present. In 1936 Franceschetti proposed the name Gronblad-Strandberg sydrome for the disease in which both findings were present.

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Angioid Streaks are seen as red, gray or brown, poorly defined streaks radiating along vessels from the optic disc. They represent breaks in the elastic tissue of Bruchs membrane.

Cardiovascular defects are related to changes in the elastic tissue of the blood vessel walls. They include arterial insufficiency in the lower extremities, premature medial calcification of peripheral arteries, reduction in arterial pulses, symptoms of coronary insufficiency and hypertension.

Gastrointestinal haemorrhages also occur in some patients characterised by recurrent often severe episodes of haematemesis and malena. In 1954 Kapalan showed histopathologically that bleeding was due to connective tissue degeneration in the submucosal arteries of the stomach.

It is well established that the disorder is transmitted as an autosomal recessive trait. The prevalence has been estimated to range from 1 in 160,000 to 1 in 1,000,000. Partial sex limitation to females is proposed by all investigators.

More than 50% of the cases reported are in the age group of 30 - 50 years.

Majority of these cases probably had the disorder in an asymptomatic form from a much earlier age.

The earliest detectable change, histopathologically is the deposition of calcium on elastic fibres. In the fully developed lesion the elastic fibres in the mid and lower dermis are degenerated, fragmented and swollen and mucopolysacharides mainly hyaluronic acid is increased. There is reduction in collagen bundles. Electron microscopy shows irregularly shaped elastic fibres with holes of varying sizes either empty or containing fibrillar or granular material. Similar changes in the elastic tissue occur in the media and intima of the blood vessels, Bruchs membrane of the eye and in the endocardium and pericardium.

The diseases of questionable relationship reported along with pseudoxanthoma elasticum include Paget's disease of the bones, diabetes mellitus, thyrotoxicosis, Marfan's syndrome and Ehlers – Danlos syndrome.

Report of Cases

Case I. A 58 years old male, a retired school teacher attended the outpatient department of Medical College Hospital, Kottayam in March 1977 for multiple hypopigmented anaesthetic patches on the back and buttocks of six years' duration and intermittent claudication of one year's duration. There was no history of visual disturbances, chest pain or gastrointestinal bleeding. Patient was taking Dapsone irregularly from 1971 for Hansen's disease. In 1971 ear lobe smears for acid fast bacilli were positive. There was no family history of Hansen's disease, skin lesions suggestive of pseudoxanthoma elasticum or any heart disease. On examination he had nodular infiltrations on the nose and ear lobes and multiple hypopigmented macules with hazy borders on the back and buttocks. The skin of the sides of his neck was soft, lax and wrinkled, hanging in folds with small yellow crepe like patches along the skin folds (Fig. 1). The skin of the neck was coarser and thicker than normal, like coarse grained Moracean leather or a plucked chicken. The anterior axillary folds were also exaggerated. The arterial pulsations in the lower extremities were diminished. Ophthalmoscopy revealed black angioid streaks radiating from the optic disc on the left side. He had a normal blood pressure. Routine blood, urine and motion examinations were normal. Serum calcium was 10.4 mg% and cholesterol 208 mg%. Repeat smear for acid fast bacilli were negative from both ear lobes at the time of examination. X-ray chest and E. C. G. were normal. X-ray of the lower extremities revealed no vascular calcification.

Biopsy of skin from the side of the neck (Fig. 2) showed degeneration of elastic fibres characterised by fragmentation, clumping and swelling of fibres in the mid and lower dermis. Collagen bundles were reduced in number. These histopathologic findings confirmed the diagnosis of pseudoxanthoma elasticum. The diagnosis of lepromatous type of leprosy was confirmed by another biopsy from a patch on the back which showed atrophy of epidermis, a clear-zone in the subepidermal region and infiltration by foam cells in the mid and lower dermis in a diffuse fashion.

Case II. A 22 year old male attended the out patient department of Medical College Hospital, Kottayam in April 1977 for yellow discolouration of the front and sides of the neck of 4 years' duration. He was also having recurrent attacks of chest pain during the last 1 year. There was no history of gastrointestinal bleeding, intermittent claudication or defective vision. His 17 year old younger brother was also having similar complaints for the past 1 year.

Fig. 1 Case I, showing typical features of pseudoxanthoma elasticum on the skin of the side of the neck.

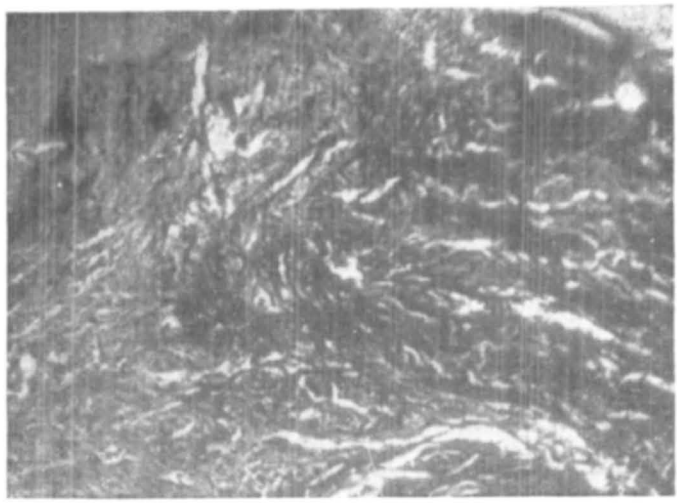

Fig. 2
Photomicrograph of skin section from case I, Verhoeff's Stain showing degenerated, clumped and fragmented elastic tissue in mid and lower dermis.

On examination he had skin lesions typical of pseudoxanthoma elasticum on the sides and front of neck and both axillae. Fundi oculi were normal and so too the peripheral pulses. Blood pressure, haemogram urinalysis, serum cholesterol, serum calcium, x-ray chest, x-ray limbs and E. C. G. were all within normal limits. Skin biopsy from the side of the neck confirmed the diagnosis of pseudoxanthoma elasticum.

Case III. A 17 year old male, the younger brother of case No. II was called for examination and was found to have slight yellow discolouration on the front and sides of the neck of 1 year duration. Careful local examination revealed very small yellow coloured papular lesions arranged along the lines of skin folds on the front and sides of the neck. Fundi oculi, peripheral pulses and blood pressure were normal. Haemogram, serum calcium and cholesterol, x-ray chest and limbs were all normal. Biopsy of skin from the side of the neck showed changes of pseudoxanthoma elasticum.

Comments

Three cases of pseudoxanthoma elasticum are reported of which the first case showed a full blown picture of Gronblad Stranberg syndrome with skin lesions of pseudoxanthoma elasticum and angioid streaks in the fundus oculus. He also had early involvement of the peripheral blood vessels as indicated by intermittent claudication even though there was no calcification of blood vessels in X-rays. He was also suffering from lepromatous leprosy. The co-existence of the 2 diseases seems to be purely coincidental. Second case had skin lesions and angina pectoris. His brother (Case No. III) was also affected showing the hereditary nature of the disease. In all the three cases clinical diagnoses were confirmed by skin biopsies.

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