CASE REPORT

4 CASES OF PSEUDOXANTHOMA ELASTICUM - CASE REPORTS

By

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Pseudoxanthoma Elasticum is a hereditory degenerative process in the elastic tissues, manifested mainly by cutaneous and eye ground lesions, angioid streaks and circulatory disturbances. In few cases history of relevant consanguinity is noted. There is much to suggest that the disease is inherited as autosomal recessive trait.

Extensive histopathological studies have led to the conclusion that deposition of calcium on elastic fibres otherwise normal in appearance is the earliest detectable change in the histogenesis in pseudoxanthoma elasticum.

There are a few authors who believe that along with the elastic tissue the collagen fibres are also involved though this is disputed by others.

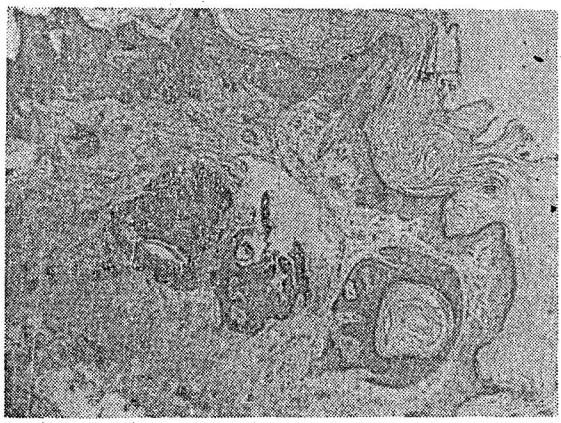
Here we present 4 cases of pseudoxanthoma elasticum of which 2 cases are sisters.

Case 1

Case 1

CASE No. 1 (Laj)

A female aged 21 years complained of papules on the skin of the neck–duration 7 years. i. e. the lesions started at the age of 14 years. The papules increased in size and numbers, some of them coalasced to form elongated raised ridges. The



Bioposy Case No. 1.

papules were later noted in front of elbows and lower abdomen. It was also noticed that the skin of the neck was becoming loose and was producing folds. Apart from the disfigurement these gave no trouble to the patient.

HISTORY

Personal history. Nothing relevant.

Family history. Patient has two sisters. One sister has similar lesions on the neck. The youngest sister did not show any lesions. Father and mother healthy, no abnormality on the skin. According to father and mother nobody in their families showed any such disease. Consanguinity not present.

ON EXAMINATION

Skin. Pin head papules of the same colour as the skin, three or four in a row placed in line with the dermatomes on the neck. Some of them have fused to form elongated ridges. The skin of the neck showed exaggerated folds and was so loose that it could be pulled to the distance of 2 inches from the neck without causing discomfort to the patient. There were multiple atrophic spots on both sides of the neck.

Similar lesions were present in the anti-cubital fossae and in this location the colour of the papule was yellowish. The linear lesions in this area were situated obliquely in the creases of the anticubital fossae. Linea atrophica were present in the popliteal fossae, but no papular lesions were seen.

The lesions were also seen on the lower abdomen, axillae and groins.

No calcification detected.

2. *Eyes* :—

Eyesight. Normal.

Fundus examination. Showed thin black wavy lines starting from near the margin of the disc and radiating towards the periphery lying deep to the retinal vessels. The lines in this case were so thin that differences in thickness could not be appreciated. Fundus as a whole presented a mottled appearance more marked towards the equator. No haemorrhages or retinal degeneration seen.

3. *General examination.*

Clinically heart and lungs were normal.

Pulse at both the wrists. Volume & Tension diminished. Rate 84/mm. *Rhythm* regular. pulse felt at dorsalis pedis, similarly showed reduced tension and volume.

B. P. 110/80 mm. of Hg.

B. C. G. within normal range.

X-Ray of Chest. No abnormality detected.

X-Ray of extremities. No calcification seen.

All other systems. Showed no abnormality.

The patient was mentally alert and did not complain of any constitutional symptoms except cramps in calf muscles. No history of any haemorrhages.

Blood picture. Within Normal limits.

Blood flow values & pulse tracing. Not done.

Biopsy report. No. 5 - 123 - 63 dated 10.1.63. Pseudoxanthoma elasticum associated mild calcification.

CASE No. II. (Prem).

Younger sister of case No I aged 16 years showed lesions similar to her sister which started at the age of 13 years which increased in size and number. The skin of the neck showed few extra folds and could not be pulled away from the neck. No calcification noticed in the skin.

EYES

Eyesight. Normal.

Fundus examination. Showed the presence of small wavy black lines of varying lengths isolated in the different parts of fundii. The angioid streaks were not as prominent as in the case of her elder sister. The fundii as a whole showed mottled appearance. No haemorrhages or degeneration of the retina seen.

GENERAL EXAMINATION

Heart and lungs. Normal.

Pulse. At the *wrist* weak.

Volume and tension diminished especially on the left side. At the *dorsalis pedis* weak volume and tension diminished much more as compared to the radial pulse.

B. P. 110/70 mm of Hg (Left arm)
120/80 mm of Hg (Right arm)

X-Ray of the extremities. No calcification seen.

E. C. G. Normal.

Blood picture. Within normal limits.

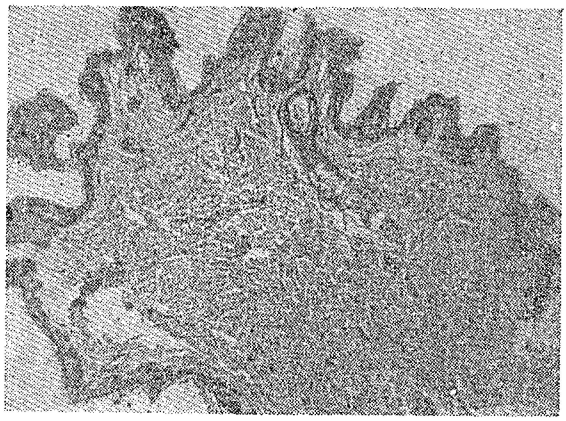
Patient shows no psychic disturbances and is intelligent. Patient does not complain of any constitutional symptoms except ischemia of toes and fingers due to exposure to cold.

Biopsy report. =S-124/63 dated 10-1-1963.

Pseudoxamthoma elasticum associated with mild calcification.

CASE No III (J. K. DUA)

Young male patient aged 24 years was referred to us by the eye department for his skin lesions on the neck. This patient had gone to the eye department for routine medical examination before entering the service and the eye specialist had suspected angioid streaks.



Biopsy Case No 3.

The patient first noticed the lesions on his neck 10 years back when he was 14 years old. The skin of neck showed an advanced picture of pseudoxanthoma elasticum with numerous typical lesions extending on both sides of neck from ears to the clavicular areas. Atrophic spots and atrophic linear lesions were seen through out the skin of the neck giving an appearance of a generalised thinning of the skin. The skin was thrown into multiple folds and could be stretched away from the neck for about 2½ inches without causing any discomfort. Similar lesions were seen around the axillae. No lesions were present on other parts of body. No calcification in the skin.

FAMILY HISTORY

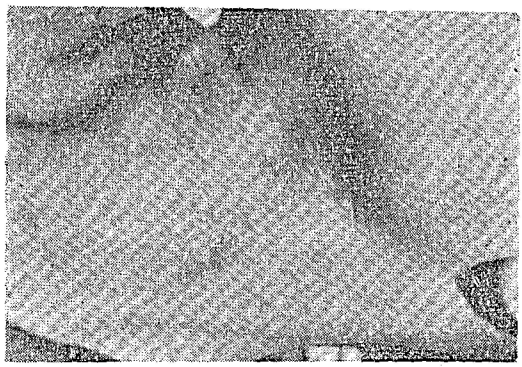
According to the patient no body in his family both on father's and mother's side had suffered from such a disease. The patient has four brothers and two

sisters. No history of consanguinity. None of the family members were available for examination.

EYES

Eye sight. Corrected with glasses upto 6/12, J_2.

Fundoscopy. Showed a fully developed picture of branching, anastomosing, greyish black lines varying in breadth from place to place lying deep to retinal vessels starting from the disc margin and radiating towards the periphery. (Angioid streaks). In addition the central area of the fundus showed pigmentary stippling which denotes the beginning of degenerative changes in the retina.



Case 3

GENERAL EXAMINATION

Heart and lungs. Normal.

Pulse. Both at the wrist and dorsalis pedis normal.

B. P 130/80 mm of Hg.

E. C. G. Normal.

X-Ray chest. Normal.

X-Ray of extremities. No calcification seen.

All other systems. Normal.

The patient has no psychic problems and is intelligent. No constitutional symptoms. No History of haemorrhages.

Biopsy report. S-1173/63 dated 22-3-1963. compatible with pseudo-xanthoma elasticum.

CASE IV (SURJIT SINGH)

A male patient of 17 years of age came to us for a rash on his neck. He first noticed this rash 5 years back i. e. when he was 12 years old. This rash became prominent a year back.

FAMILY HISTORY

History of consanguinity present. Maternal grand mother and paternal grand mother were first cousins. None of the family members showed any such disease. Mother and father were examined. No lesions were detected.

Skin. Patient showed reddish papular lesions in linear groups of 5 to 6 on the front of the neck. Similar lesions yellowish in colour were seen on the sides and lower parts of the back of the neck. Some of the lesions formed brownish raised ridges in the folds of the neck. Very few atrophic spots were present. The skin of the neck was slightly loose and formed few folds. Stretching of skin away from neck was not very marked. Some lesions were also present on the supra clavicular area and also in the supra sternal notch. Similar lesions yellowish in colour were present around the axillae, lower abdomen, groins and antecubital, popliteal fossae. No calcification felt in the skin.

Eyes – Eyesight normal.

Fundoscopy. showed few thin angioid streaks which were not well-developed. No other retinal changes were seen.

GENERAL EXAMINATION

Heart and Lungs. Normal.

Pulse. Very poor in volume and tension both at the radial and dorsalis pedis.

B. P. 120/70 mm of Hg.

E. C. G. Not done.

X-Ray chest. Not done.

X-Ray of extremities. No calcification seen.

All other systems. Normal.

Patient is intelligent but very nervous and unsteady and lacks in concentration. The patient does not complain of any constitutional symptoms. No history of haemorrhages.

Biopsy report. Not yet received.

DISCUSSION

In all the four cases history of heredity could not be determined. Consanguinity was present in one case. Two cases were present in the same family and the youngest sister who is 5 years old though did not show any lesions may develop them later, in view of the fact that both these sisters noticed the lesions at the age of 13 to 14 years. All the four cases have developed the lesions around the age of puberty and as mentioned by some authors endocrinal disturbances my be a contributory factor.

All these cases range from 16 to 24 years of age and hence very mild circulatory disturbances are noticed through the skin and eye lesions show comparatively a well developed picture. None of these cases showed any systemic disturbances. None of the cases reported haemorrhages of any type. Mucous membrane lesions were not seen in any case.

Lastly due to lack of facilities detailed investigations like blood flow, pulse tracing and histopathological studies with different stains and histochemical studies could not be undertaken.

We wish to observe these patients as long as possible and would try to get some of the investigations repeated from time to time.