CLINICAL PATTERN OF PSEUDOXANTHOMA ELASTICUM IN INDIAN SUBCONTINENT

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Summary

Clinical pattern of pseudoxanthoma elasticum in the Indian subcontinent along with report of three fresh cases of pseudoxanthoma elasticum is described.

Pseudoxanthoma elasticum is a well recognised hereditary disorder, which may have protean manifestations. The entity has been found to occur in India and has been reported sporadically from North [1,2,3]; South [4,5,6,7]; East [8,9,10,11]; and West [12]. There has hardly been an endeavour to describe the disease in a systematic manner and to delineate the clinical pattern of the disease. Hence, we are prompted to review 20 cases of the disease thus far available from India, which includes three patients of our own.

Clinical features

Of the 20 cases 13 were males and 7 females. Their ages ranged between 15 to 43 years. Age of reporting was earlier in females as compared to males. Duration of the disease was variable; most patients having had the disease since childhood. A positive family history was elicited in 9 cases. In 5 cases an autosomal recessive pattern of inheritance was suspected.

Cosmetic embarrassment was the main presenting features in the large majority of patients. Two cases had systemic manifestations in the form of giddiness, headache, vertigo or weakness of limb. One of these patients also had repeated haemorrhage per rectum.

Morphology of the skin lesions was similar to those of the cases described below. Purpura and elastosis perforans serpiginosa were seen in one case each. Various sites such as sides of the neck, axillae, groins, supraclavicular and paraumbilical areas and cubital fossae were affected in that order of frequency. In a couple of cases popliteal fossae, cubital fossae and mucous membrane were also affected. Forehead and abdomen were involved in one case each.

Fundus examination revealed angioid streaks in 12 cases, macular atrophy in 6 and choroidal degeneration in 3.

One case had high blood pressure. Subarachnoid haemorrhage with attending hemiparesis / hemiplegia was seen in 2 other cases. One of these showed left ventricular hypertrophy on electrocardiographic examination. Peripheral pulsation was feeble in 5 cases. Calcinosis cutis was demonstrated in one and calcification of dorsalis pedis and posterior tibial arteris were seen in another case. Occult blood, suggestive

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of gastrointestinal haemorrhage, was found in only 2 cases.

Case reports

1. 27 years old unmarried female, reported with wrinkling of the skin on the neck since childhood. Lesions were gradually progressive for the first few years. There was no history of giddiness, headache, blurring of vision, pain in the abdomen, intermittent claudication, palpitation, chest pain or gastro intestinal haemorrhage.

There was no history of consanguinity. Among 3 other siblings, 2 brothers were normal and one sister had problem similar to that of the patient. Twenty one members of the family comprising three generations were altogether studied of which 13 were females and 8 males. Only two females under review were found to be suffering from the disease.

Examination of the skin showed marked hypopigmentation on the neck. The skin was soft, lax and wrinkled. In addition it was studded with multiple yellowish papules of 1-3 mm size arranged in linear and/or reticulate pattern (Fig. 1). Similar lesions were also present on the upper part of the chest below the clavicles. There was no evidence of telangiectasia. Axillae and groins were unaffected.

The cardiovascular, respiratory and central nervous systems were clinically normal. On ocular examination, vision was 6/12 in the right and 6/6 in the left eye. Angioid streaks were seen in both fundii. In addition macular degeneration characterised by rounded, whitish, degenerative spots were also seen. Choroidal degeneration and excessive pigmentation were present.

The blood, urine, stool and radiological investigations were within normal limits. ESR was 42 mm/1st hour.

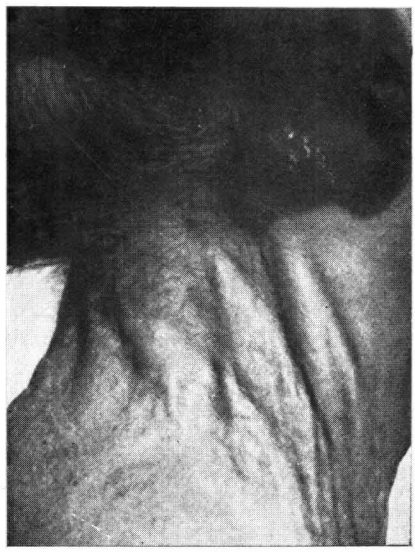

Fig 1 Multiple papules studded over the soft, laxed and wrinkled skin around the neck

2. 25 years old unmarried sister of patient reported with insidiously progressive lesion around the neck and axillae since childhood. They were asymptomatic. The morphological characterstics of the lesions were similar

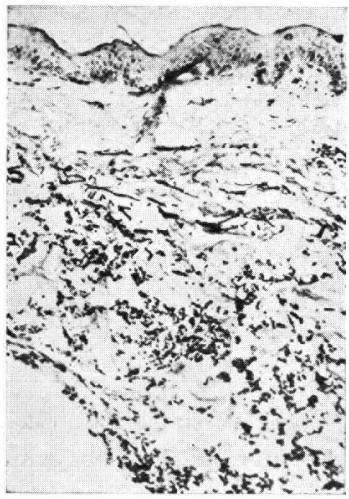

Fig. 2 Swollen, fragmented, irregularly clumped basophilic fibres in the mid and lower dermis (H & E × 100).

to those of the aforesaid case. No clinical abnormality was found on detailed systemic examination. There was a refractive error in the right eye; the vision being 6/9 as compared to 6/6 on the left. Angioid streaks, as well as macular and choroidal degeneration were present in both eyes.

3. 21 years old male had skin lesions similar to those of patients 1 and 2 since childhood. Angioid streaks were demonstrated on ocular examination. No other abnormality was detected.

Histopathology

Haemotoxyline and eosin stained sections from the lesions showed changes characterised by swollen, fragmented and irregulary clumped basophilic fibres (Fig. 2) which were found to be deeply stained by Verhoeff's stain. These were primarily confined to the middle and lower dermis. Von-Kossa stain was negative. The epidermis was normal.

At the time of reports, D. L. Alpha tocopherol, an antioxidant agent, was being administered to these patients in doses of 1000 - 1500 i.u, daily for 6 weeks. The therapeutic effectiveness shall be reported subsequently.

Discussion

Pseudoxanthoma elasticum was first described by Balzar and Darrier. Subsequently, the condition was described under the name Gronblad-Strandberg syndrome in which eye lesions were found in association with skin lesions. Since its first description several reports of this disease have appeared in the literature. There is no doubt regarding the global distribution of the disease. Nevertheless, it is pertinent to study its pattern in different parts of the world. Interestingly, this study has brought forth a few parametres peculiar to Indian subcontinent. Specialists working in this region are probably well aware of this condition [1-12].

The disease manifested at an early age in most of our patients who reported the onset of the disease during childhood. This is in contrast to late onset of the disease reported elsewhere [13]. The higher prevalence of males over females in our study is also at variance with that reported in standard text books [14]. Systemic manifestations excepting for the ocular changes namely angioid streaks, macular atrophy and choroidal degeneration were seen less frequently than in other reports [14]. However the pattern of familial incidence and morphological pattern of the skin lesions in the Indian patients is largely in accordance with that in other series [13].

Acknowledgement

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Book Review

Treatment of Skin Diseases, J. S. Pasricha, 2nd Edn., Oxford & IBH Publishing Co., New Delhi, 1979 pp. 178, Price Rs. 15.25 (LCUE)

The second edition of the book has been found thoroughly revised and rewritten. The first two chapters are devoted to general information, the subsequent chapters deal with the common skin diseases. Also it has an appendix giving names of proprietary medicines which are commonly used in skin diseases. The book highlights the diseases of the skin with simple definition and description of the diseases and the modern line of treatment. Many photographs both black and white and coloured photographs of the diseases are provided, which makes it very easy to be understood by the readers. Majority of the readers will be general practitioners and medical students, who get very little teaching and training of dermatology in their undergraduate studies, though later in life they are to face quite a big percentage of cases suffering from skin diseases in their everyday practice. The book will be of immense help to them. The book has 24 illustrations and 8 coloured plates.

— B. N. Banerjee