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A RARE CASE OF PSEUDOXANTHOMA ELASTICUM

Dermatology

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ABSTRACT

Pseudoxanthoma elasticum (PXE) is also known as Gronblad-Strandberg syndrome. It belongs to the group of connective tissue disorders that affect the elastic tissue of the skin, blood vessels, and the eyes. Both autosomal dominant and recessive patterns of inheritance can be seen. Skin lesions consist of small, yellowish papules in rows or a lacy pattern, which may coalesce to form larger patches. The skin is soft, lax and slightly wrinkled. Common sites affected in PXE are the sides of the neck, below the collar bones, the armpits, abdomen, groins, perineum and thighs. Currently there is no effective treatment for the condition. As it can be passed to next generation genetic counselling can be helpful.

KEYWORDS

Endometrial carcinoma, Quality of life, socio-demographic, clinical determinants.

CASE REPORT:

A 25 year old female patient presented in our department with a 5 year history of multiple asymptomatic raised lesions on the neck. There was no other significant personal and family history. Cutaneous examination revealed multiple, variable sized papules on the neck. They were slightly yellowish in color.(Figure1) Routine laboratory tests like hemogram, liver function tests, renal function tests were within normal limits. Electrocardiography and radiograph of the chest were normal. Ophthalmological examination was also normal. Histopathological examination from one of the papular lesion revealed the presence of fragmented and calcified elastic fibres in the dermis. Thus on the basis of clinical and histopathological findings a diagnosis of Pseudoxanthoma elasticum was made.

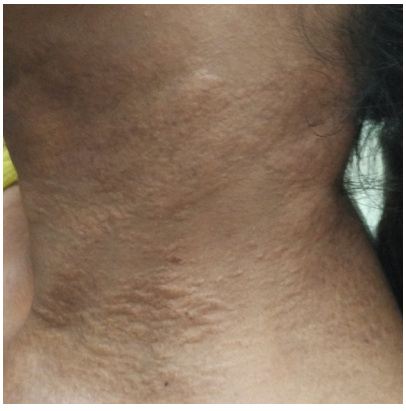

Figure 1: Multiple Yellowish Papules On Side Of Neck

DISCUSSION

Pseudoxanthoma elasticum (PXE) is an inherited connective tissue disorder which is characterized by abnormal calcification of the elastic tissues of the skin, retina and cardiovascular system. Both autosomal dominant and autosomal recessive patterns of inheritance are seen.The prevalence is calculated as 1 in 25,000-70,000. The condition results results from mutations of transmembrane transporter protein adenosine triphosphate binding cassette-C6 (ABC-C6), the gene located at chromosome 16p13. yellowish papules or plaques with an associated increase in skin laxity are the typical cutaneous manifestations of the condition. flexures and periumbilical skin are the commonly affected sites. Angioid streaks, breaks in the Bruch's membrane, with secondary changes of the retinal pigmented epithelium (peau d'orange) and choriocapillaris are the ocular changes which can be seen. These ocular defects are asymptomatic initially, but may be complicated by retinal neovascularization, recurrent hemorrhage, disciform scarring and finally loss of vision. Cardiovascular manifestations are usually last to manifest and result from slow and progressive calcification of elastic arterial walls with an increased risk of accelerated peripheral vascular disease, ischemic heart disease, hypertension and cerebrovascular disease. Calcification, alteration and fragmentation of elastic structures in the mid-dermis are the characteristic histopathological changes seen. The condition is usually diagnosed late in the second or third decade of life.

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