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Pseudoxanthoma Elasticum: A Vascular Diagnostic Challenge (P1103)

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INTRODUCTION

Pseudoxanthoma Elasticum (PXE) is an inherited connective tissue disorder that affects the skin, eyes, and cardiovascular system. PXE has an autosomal dominant recessive pattern of inheritance. It is characterized by the calcification of the elastic fibres of the connective tissues. In most cases, it is expressed by mutations of the ABCC6 gene located in chromosome 16p 13.1[1].

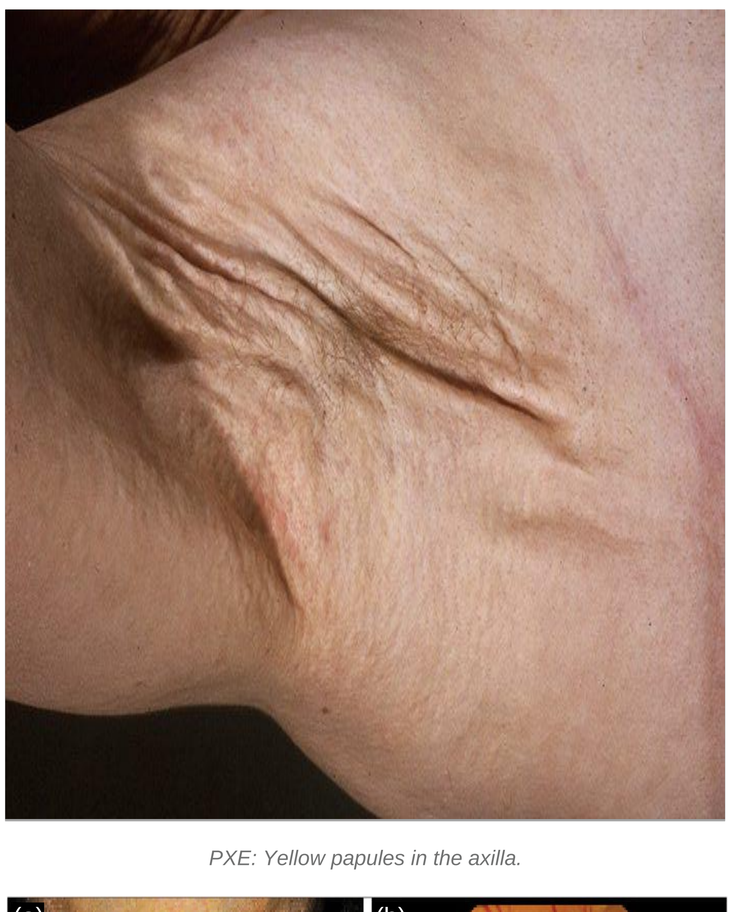
Cardiovascular involvement is mainly characterized by mineralization and fragmentation of elastic fibres of blood vessels and premature atherosclerosis.

PXE can be transmitted as an autosomal dominant trait or autosomal recessive trait with an estimated prevalence ranging from 1 in 70,000 to 1 in million [2][3].

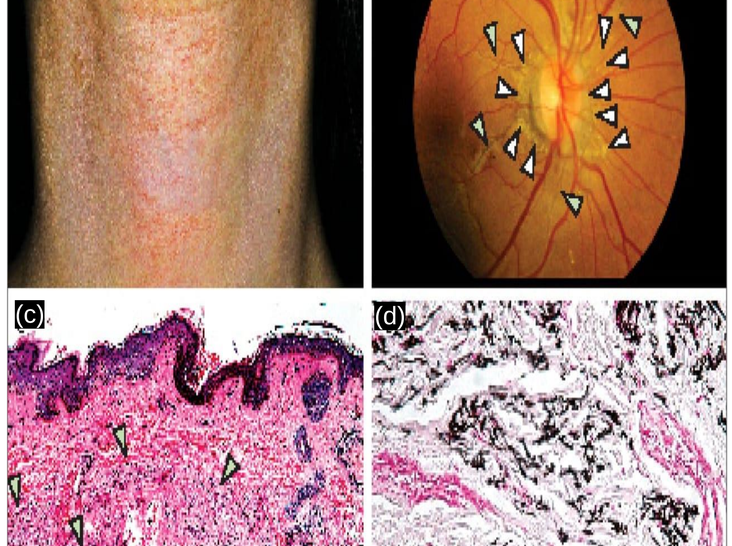
In some families, the cutaneous changes can be predominant with relatively mild eye or cardiovascular involvement, while in other families the involvement of the eye and cardiovascular system may be severe with limited skin findings [4].

The first limited skin lesions to be noted are on the skin in the lateral part of the neck. Skin lesions begin in childhood but they are not usually noted until adolescence. Small yellow papules are seen in a linear or reticular pattern. Clinically, patients show characteristic ocular manifestations including peau d'orange and angioid streaks [5]. Intermittent claudication is the most common cardiovascular symptom (30% of patients) and often represents the first sign of atherosclerosis [5].

This is a case report of a 26-year-old lady with PXE who developed peripheral arterial disease (PAD) with no other identifiable risk factors.



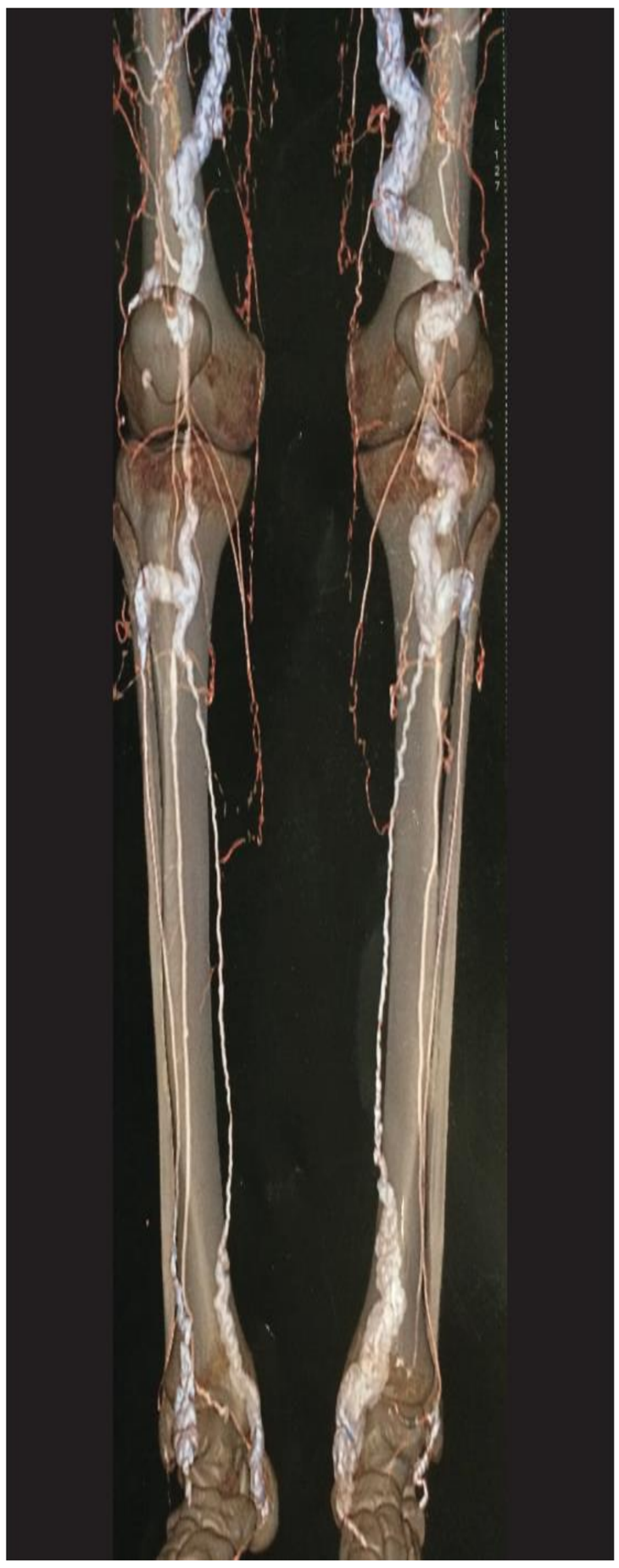
PXE: Yellow papules in the axilla.



PXE: Neck yellow papules (a) ; eye angioid streaks (b) ; histopathology of papules (c) and (d).

Discussion

Although PXE can be associated with considerable morbidity and significant mortality, the phenotypic spectrum is highly variable with both inter-and intrafamilial heterogeneity. Clinical variability is evident by observations that the involvement of all three major organ systems, i.e., skin, eyes, and the cardiovascular system is encountered in patients, whereas others, even within the same family, have a limited involvement of one of these organs. This may indicate that disease expression is possibly influenced by environmental factors [7]. The manifestations are noted during infancy, whereas in most cases the clinical signs are not evident until the second and third decade of life, sometimes more lately. It has been estimated that the average time between the onset of skin lesions and the diagnosis is about 20-25 years. This prolonged time-lapse can be explained by the fact that the majority of patients were not concerned about their skin lesions and didn't seek medical advice until signs of ophthalmic and cardiovascular involvement presented in middle age [8].



PXE: Lower limbs CTA.

Discussion

However, early diagnosis is important if ocular and cardiovascular complications are to be prevented. Cardiovascular manifestations are very varied and include decreased pulse, hypertension, angina pectoris, and intermittent claudication. Gastrointestinal hemorrhage, melena, and hematemesis can also be observed. Patients can develop atherosclerosis, cardiac arrest, and strokes at younger ages. All these alterations are secondary to the mineralization and fragmentation of elastic fibres of the medium-sized arteries and aorta as well as the arterioles and coronary vessels of the endocardium, pericardium, and connective tissue of the myocardium [9].

Unfortunately, there is limited information of PAD associated with PXE. The majority of information available for PXE-related arteriopathies is described in relation to cardiothoracic procedures. There have been many cases that have highlighted the need for individuals undergoing bypass grafting and potentially having PXE to undergo arterial studies prior to surgery [10].

Occlusive peripheral vascular disease relating to connective tissue disorders is rarely discussed in literature.

The management of individuals with PAD attributed to PXE is difficult because it seems that their vascular disease cannot be controlled by lifestyle changes and medications. Revascularization in claudication is contra-indicated because of the diffuse and distal nature of the vascular disease. However, in cases of critical ischaemia, a thorough peripheral vascular evaluation must be performed and revascularization should be attempted even with the poor nature of the disease [11]. It must also be brought to the attention that due to autosomal inheritance of the condition, a thorough family history must be taken and potential arterial disease risks should be explained to the patient. The physician should be aware of the associated systemic involvement of PXE. Diagnosis of the disease might minimize the devastating ocular and cardiovascular complications of blindness, critical ischaemia, cardiac arrest, stroke, and intracranial aneurysm rupture.

There is no specific treatment or effective treatment for cutaneous or systemic manifestation. It's multidisciplinary and consists of ophthalmological, cardiovascular monitoring, and genetic counseling. The abandonment of smoking habits, moderate physical exercise, and proper diet with supplements intake of magnesium, phosphate, and pyrophosphate analogs can reduce the progression of the disease.

CONCLUSIONS

The early diagnosis of PXE may be important to minimize the serious complications and impact on quality of life. This case report may help the vascular surgeon for the early recognition of this rare disorder and to consider it whenever confronted with intermittent claudication patients of a young age group.

The clinical manifestations are late-onset and the early cutaneous findings, frequently the first diagnostic signs of PXE are subtle and usually not recognized by the vascular surgeon. There is no cure for PXE. The management is preventive and the patient should be monitored on regular basis.

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