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PSEUDOXANTHOMA ELASTICUM WITH NEPHROCALCINOSIS AND SPLENIC CALCIFICATION - A RARE CASE REPORT

Dermatology

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ABSTRACT

Pseudoxanthoma elasticum (PXE) is a rare inherited, multisystem disorder affecting skin, eyes and cardiovascular system. It is characterized by generalized fragmentation and progressive calcification of elastic tissues. We report a 28 year old female who presented with asymptomatic distinctive lesions over her neck and with intermittent pain over both legs.

KEYWORDS:

INTRODUCTION

Pseudoxanthoma elasticum (PXE) is a disease of genetic inheritance due to the mutation of ABCC6 gene. It's characterized by progressive calcification and fragmentation of elastic fibers predominantly in the skin, eyes and cardiovascular system.

CASE REPORT

A 28-year- old female presented with complaints of multiple pale yellow raised skin lesions present over her neck which were asymptomatic and intermittent pain over her both lower limbs since last 6 months duration. She was apparently fine till she developed these symptoms. She had no history of similar complaints in her family. Dermatological examination revealed multiple small (1-3mm) papules, in confluent plaques present on the lateral sides of the neck. (Fig. 1) Nails, oral mucosa and hair were normal. A cardiologist and ophthalmologist opinion was obtained to outlook for systemic involvement.

Routine blood investigations (including Serum calcium & phosphate levels) were done and were normal. Histopathology examination revealed clumped, fragmented and swollen elastic fibers. (Fig. 2) Funduscopy examination showed bilateral angioid streaks. (Fig. 3) Arterial doppler was done for bilateral lower limb and upper limb which revealed nil colour flow noted in bilateral distal posterior tibia artery, no colour flow in the right distal radial artery and low resistance flow seen in the left distal radial artery. Computed topography abdomen was done and revealed bilateral nephroca lcinosis (Fig. 4) and splenic calcification. (Fig. 5)

DISCUSSION

PXE is an inherited disorder, being described as autosomal dominant or recessive patterns. Mutations have been identified on the chromosome 16p13.1 & subsequently on MRP6/ABCC6 gene.[1-3] It typically involves the three organ system; skin, eyes and the cardi ovascular, resulting in skin laxity, retinal haemorrhage and arterial insufficiency respectively.

Cutaneous lesions may arise during the second and third decades of life, which presents as yellowish papules and plaques associated with loose and redundant skin in flexural sites giving an appearance of plucked chicken skin.[4] Mucosal involvement can also occur, most commonly effected site is the inner aspect of the lower lip. Other classical signs of disease are angioid streaks in the eye resulting in retinal haemorrhage and finally progressive loss of vision.[5,6] Vascular calcifications, leads to the formation of atherosclerotic plaques in arteries of medium caliber which causes changes leading to outcomes like claudication pain, hypertension, angina, myocardial infarction.[7] Diagnosis is based on clinical and histological changes.

Histological changes of the involved skin include accumulation of swollen and irregular elastic fibers in the mid and lower dermis. [8]

Management includes excision of cosmetically unacceptable lesions although there is no specific treatment for cutaneous lesions. Ophthalmic care is to prevent retinal hemorrhage is done by avoiding head trauma, heavy straining etc. Laser photocoagulation for choroid revascularization and macular translocation (if bleeding has occurred due to revascularization) can be done. Pentoxifylline is given to improve the blood circulation in the extremities. The prognosis depends on the degree of involvement of the cardiovascular system.

CONCLUSION

Pseudoxanthoma elasticum doesn't only manifest with cutaneous lesions but also an internal screening for other manifestation has to done so as to avoid future complications. A proper counseling has to be given to such patients to avoid systemic complications.

Fig 1: Small papules in confluent plaques over her lateral side of her neck

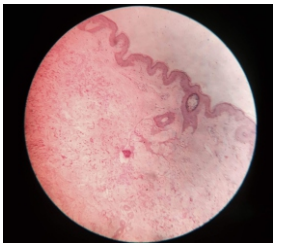

Fig. 2 : Histopathology changes showing clumped, fragmented and swollen elastic fibers

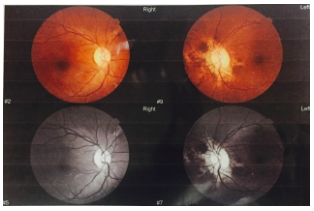

Fig. 3: Bilateral angioid streaks

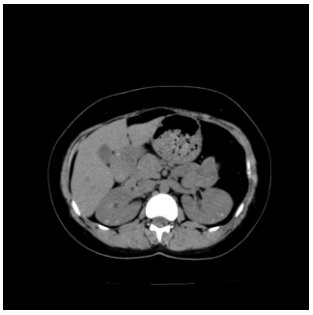

Fig. 4: CT scan showing bilateral nephrocalcinosis

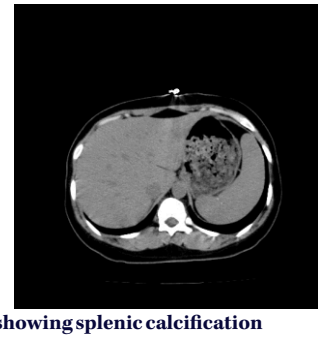

Fig. 5: CT scan showing splenic calcification

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