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CASE REPORT

Unveiling the mystique: pseudoxanthoma elasticum case report

Revelando o mistério: relato de caso de pseudoxantoma elástico

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Abstract

Pseudoxanthoma elasticum (PXE) is a rare genetic disorder of autosomal recessive inheritance affecting the skin, eyes, and cardiovascular system. We report a case of 17-year-old female with well-defined reticulated, skin-colored plaques over the anterior aspect and in nape of the neck. Punch biopsy from the lesion revealed benign stratified squamous lining with the middle and lower thirds of the dermis showing fragmented and calcified basophilic elastic fibers amidst collagen bundles. Adjacent areas show dense lymphocytic infiltration and giant cell reaction. Verhoff Von Gieson stains showed black-colored fragmented elastic fibers. Ocular examination revealed peau d' orange appearance of retinal blood vessels, which was considered as the early stage of retinal involvement. PXE is currently an incurable disease that on early diagnosis can prevent the Ocular and cardiovascular complications.

Keywords: Autosomal recessive. Elastic fibers. Pseudoxanthoma elasticum.

Resumo

Pseudoxantoma elástico é uma doença genética rara de herança autossômica recessiva que afeta a pele, os olhos e o sistema cardiovascular. Relatamos o caso de uma mulher de 17 anos com placas reticuladas bem definidas, da cor da pele, na área do decote e na nuca. A biópsia por punção da lesão revelou revestimento escamoso estratificado benigno com terços médio e inferior da derme mostrando fibras elásticas basofílicas fragmentadas e calcificadas em meio a feixes de colágeno. A área adjacente mostra infiltração linfocítica densa e reação de células gigantes. A coloração de Verhoff Von Gieson mostrou fibras elásticas fragmentadas de cor preta. O exame ocular revelou aparência de vasos sanguíneos da retina em tons de laranja e peau-d, o que foi considerado o estágio inicial do envolvimento da retina. O PXE é atualmente uma doença incurável que, no diagnóstico precoce, pode prevenir complicações oculares e cardiovasculares.

Palavras-chave: Autossômica recessiva. Fibras elásticas. Pseudoxantoma elástico.

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Introduction

Pseudoxanthoma elasticum (PXE) is a rare genetic disorder with an autosomal recessive mode of genetic transmission affecting multiple organs, such as the skin, eyes, heart, and gastrointestinal system[1]. PXE, also termed as Gronblad Strandberg syndrome, is a rare disorder due to a mutation in the *ABCC6* gene (ATP-binding cassette transporter C6), located in chromosome 16. Prevalence of PXE was found to be 1/25.000 to 1/100.000 inhabitants. It has been found that the prevalence of PXE is 10 times more in women than in men[2,3]. It encodes an ATP-binding driven anion transporter, seen in the cell membrane of the liver and kidney. PXE is a form of genodermatoses, currently an incurable disease associated with serious complications due to elastic fiber fragmentation and calcification[4]. Here we report a rare case of PXE with its clinical, histopathological, and ocular findings.

Case report

A 17-years-old female came to the outpatient department with the complaints of itchy, yellowish papules and plaques in the anterior and lateral aspect of her neck for the past 1 year. No relevant personal or family, or any medical history of dermatosis. No history of any consanguineous marriage in the family. On examination, painless, uneven skin-colored reticulated plaques (Fig. 1) without any surrounding inflammatory signs were seen in the anterior aspect and in the nape of the neck, giving a parchment-like skin appearance.

Complete hemogram and routine blood investigations were found to be normal. The patient was then subjected to a skin biopsy. A punch biopsy was taken from the lesion in the anterior and nape of the neck. Biopsy revealed benign stratified squamous lining with middle and lower thirds of dermis showing fragmented and calcified basophilic elastic fibers amidst collagen bundles. The adjacent area shows dense lymphocytic infiltration and giant cell reaction (Fig. 2).

Verhoff Von Gieson stains showed black-colored fragmented elastic fibers in the dermis (Fig. 3).

Diagnosed as a case of PXE and the patient was then subjected to ocular examination, which revealed peau d' orange appearance of retinal blood vessels, which was the earliest retinal manifestation. Following the investigation, the patient was then subjected to cardiovascular evaluation and the results were normal. The patient is currently on follow-up.

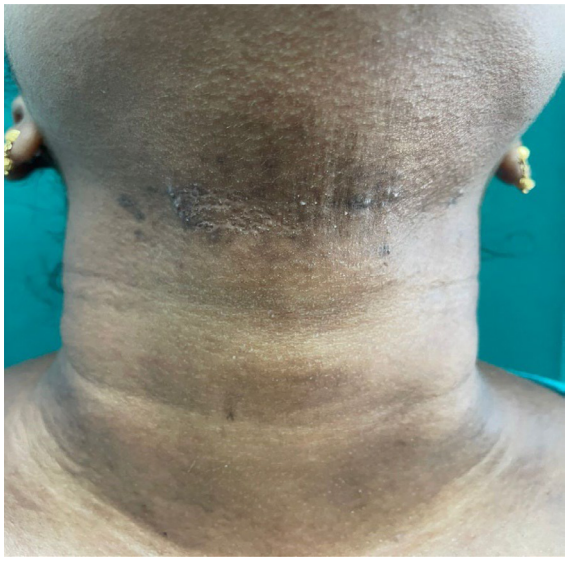

Figure 1. Clinical image of patient showing reticulated plaques over the anterior aspect of the neck.

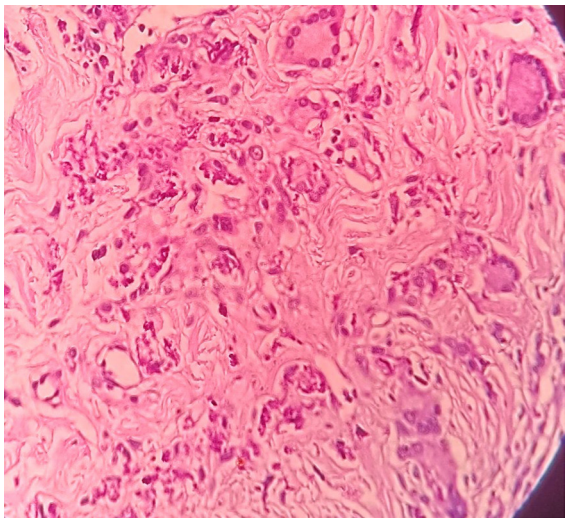

Figure 2. Histopathological image showing fragmented elastic fibers with calcification, lymphocytic infiltration, and giant cell reaction (H&E stain, 40×).

Discussion

PXE is a rare inherited genetic disorder, caused due to abnormal mineralization of the connective tissue with elastic fibre degeneration affecting various organs, such as the skin, eyeballs, and cardiovascular system[5].

Cutaneous manifestations are seen as yellowish macules or papules, or plaques. PXE can occur at any age

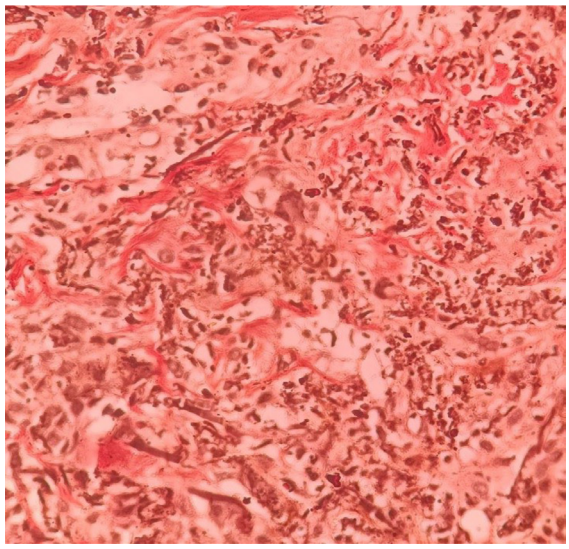

Figure 3. Histopathological image showing fragmented elastic fibers (Verhoeff Von Gieson stain, 40×).

and the skin appears lax and wrinkled. The lesions start appearing over the lateral aspects of the neck, followed by skin creases, armpits, popliteal, and inguinal regions. Oral mucosa and genital region can also be affected[6,7].

Early ocular involvement starts as peau d' orange appearance of retinal vessels and progresses to the development of angioid streaks due to lesions over the Bruch membrane. Progressive retinal pigmentation, macular degeneration, choroidal neovascularization, retinal hemorrhage, and scar formation can occur, leading to the most serious complication of complete ocular blindness[8].

Various cardiovascular abnormalities occur in a case of PXE, such as bradycardia, hypertension, angina pectoris, atherosclerosis, and cardiac arrest at younger ages. Gastrointestinal manifestations, including melena, hemorrhages, and hematemesis, can be seen. Some patients presented with stroke at younger ages. Major pathogenesis behind these complications was found to be fragmentation of elastic fibers in the lining of blood vessels and in the connective tissue of the pericardium, myocardium, and endocardium of the heart[9].

Certain inherited hemolytic disorders, such as beta thalassemia, hereditary spherocytosis, and Sickle cell anemia, can occur in patients with PXE[10].

Plomp et al. proposed guidelines for diagnosing PXE which includes major criteria: (a) skin lesions, such as yellow cutaneous plaques and papules, fragmentation, clumping, and calcification of elastic fibers, (b) ophthalmic lesions, such as Peau d' orange appearance of retina or the presence of angioid streaks, and (c) genetic factors such as mutation of both alleles of *ABCC6* gene or a first degree relative affected with PXE. Minor criteria include ophthalmic lesions, such as one angioid streak shorter than 1 disk diameter, one or more comets in the retina, and one or more wing signs in the retina: genetic factors, such as the presence of a mutation of one allele of the *ABCC6* gene[11].

Major differential diagnosis for PXE includes solar elastosis, PXE-like papillary dermal elastolysis. PXE-like papillary dermal elastolysis occurs in elderly females, where partial or complete loss of elastic fibers were seen but without calcification. Solar elastosis shows irregularly thickened, coarse, disorganized, and tangled elastic fibers[12].

Conclusion

This case report deals with a young female with typical clinical and histopathological features of PXE highlighting a need for a multidisciplinary approach for accurate diagnosis of this rare disorder. This helps us to understand the disease better and to discover the newer therapeutic approach to treat the disease and to prevent the complications as early as possible.

Funding

None.

Conflicts of interest

None

Ethical considerations

Protection of humans and animals. The authors declare that the procedures followed complied with the ethical standards of the responsible human experimentation committee and adhered to the World Medical Association and the Declaration of Helsinki. The procedures were approved by the Institutional Ethics Committee.

Confidentiality, informed consent, and ethical approval. The authors have followed their institution's confidentiality protocols, obtained informed consent from patients, and received approval from the Ethics Committee. The SAGER guidelines were followed according to the nature of the study.

Declaration on the use of artificial intelligence. The authors declare that no generative artificial intelligence was used in the writing of this manuscript.

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