CASE REPORT

Pseudoxanthoma elasticum: Case report

Lisseth Stefanía Ramírez Velasco,* Julian David Echeverria Davila, Bertha Bolivia Bayancela Guerrero,*** Mónica Salazar Malliquinga****

* *Postgraduate in Dermatology, Equinoctial Technological University, Quito, Ecuador.*
** *Postgraduate in Dermatology, Equinoctial Technological University, Quito, Ecuador.*
*** *Dermatologist at the Hospital de Especialidades de las Fuerzas Armadas del Ecuador.*
**** *Dermatopathologist at the Hospital de Especialidades de las Fuerzas Armadas del Ecuador.*

Correspondence:
liss1994chete@outlook.com | 0998368319
https://orcid.org/0000-0003-4609-9029
San Fernando, Av Occidental and Antonio Román

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ABSTRACT

Pseudoxanthoma elasticum, also known as generalized or systemic elastorrhexis, is a hereditary connective tissue disorder that affects the skin, heart, and eyes. It presents with an autosomal recessive pattern in which abnormal glycosaminoglycans are produced by pathological fibroblasts and deposited in the elastic fibers of tissues, leading to fragmentation and calcification of the fibers.

This pathology is rare and often associated with systemic involvement, particularly affecting the eyes, heart, and intestines. Therefore, timely diagnosis and proper management are crucial to prevent complications and comorbidities in patients.

INTRODUCTION

Pseudoxanthoma elasticum can present in both autosomal recessive and dominant forms, with the recessive presentation being the most frequent. The locus for pseudoxanthoma elasticum has been located on the short arm of chromosome 16p13:1, in the ABCC6 (MRP6) gene, which affects connective tissue.[1,2]

The condition can manifest from childhood as yellowish papules with a tendency to coalesce and areas of skin that appear loose, hyperelastic, and redundant, known as "chicken skin," "orange peel," or "cobblestone" skin. The lesions are initially located on the neck and later extend to the folds and the periumbilical region.[3,4]

Ocularly, "angioid streaks" appear, corresponding to tears in the Bruch membrane, which can lead to hemorrhages and scarring in the retina. Systemically, intermittent claudication, arterial aneurysms, coronary heart disease, strokes, ecchymosis, hemarthrosis, and uterine and gastrointestinal hemorrhages may occur.[2-4]

Histopathologically, the literature shows elastic fibers with degenerative alterations in the mid-dermis. These elastic fibers are typically swollen and fragmented, or granular. They have a more bluish-gray coloration than usual, appearing twisted, wavy, and broken, or resembling curly wool. The calcification of the altered elastic fibers is characteristic. The elastic fibers are stained using von Kossa, Verhoeff methods, and intense blue staining with phosphotungstic acid, hematoxylin, and orcein.[2]

The differential diagnosis includes dermal and actinic elastolysis, eosinophilia-myalgia syndrome induced by L-tryptophan, amyloid elastosis, dermatofibrosis lenticularis (Buschke-Ollendorf syndrome), Ehlers-Danlos syndrome, serpiginous perforating elastosis,

and cutis laxa.[2,7,8] It is important to note that cases of pseudoxanthoma elasticum induced by D-penicillamine have been reported in patients with Wilson's disease or homocystinuria.

No effective treatment is known, so regular follow-up is crucial to detect serious systemic complications that may arise during the course of the disease.[5] Plastic surgery is useful for treating redundant skin.[2,3,5,6]

CASE REPORT

This is a 76-year-old female patient with a history of Ebstein's anomaly, hypothyroidism treated with levothyroxine, and breast cancer. The patient presents with yellowish papules that coalesce and form plaques, predominantly located on the neck and upper limbs (Figures 1A, 1B, 1C). These lesions have been present since childhood and have increased in size with her development, without additional symptoms. An incisional skin biopsy from the arm was performed, which revealed the presence of basophilic short, curly, and frayed elastic fibers, with calcium deposits between them (Figures 2 and 3). Based on these findings, the diagnosis of pseudoxanthoma elasticum was made. No ocular involvement was observed.

DISCUSSION

Pseudoxanthoma elasticum, also known as generalized or systemic elastorrhexis, is a hereditary connective tissue disorder that affects the skin, heart, and eyes. It presents with an autosomal recessive pattern, where abnormal glycosaminoglycans, produced by pathological fibroblasts, are deposited in the elastic fibers of tissues, leading to their fragmentation and calcification.[1-3]

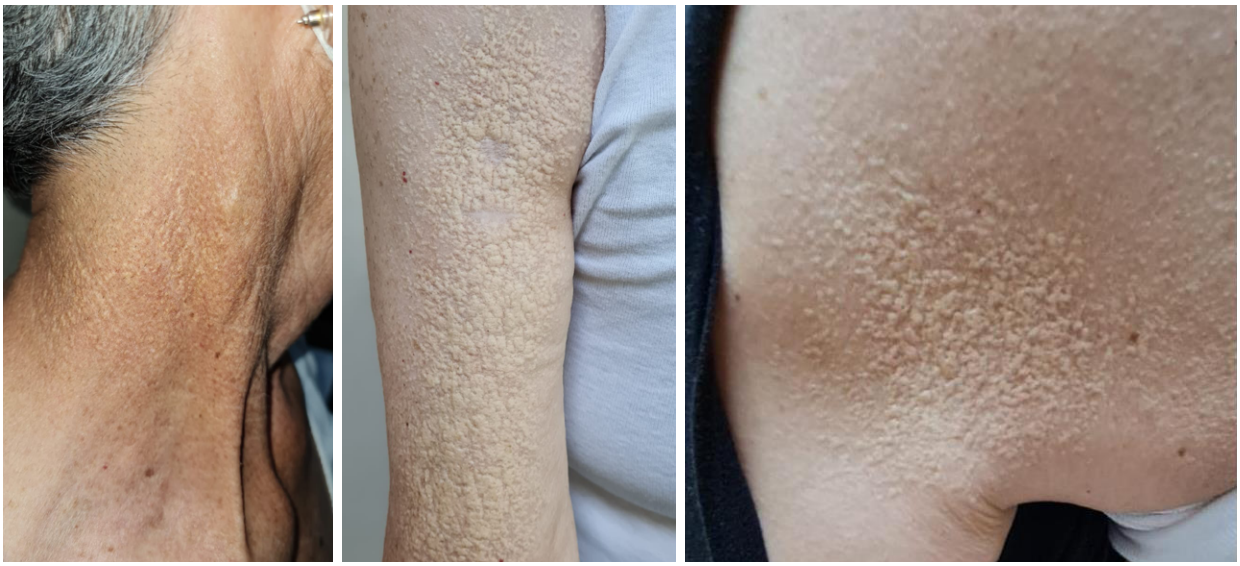

Figure 1A: Yellowish plaques on the neck. Figures 1B-1C: Yellowish plaques on the arms.

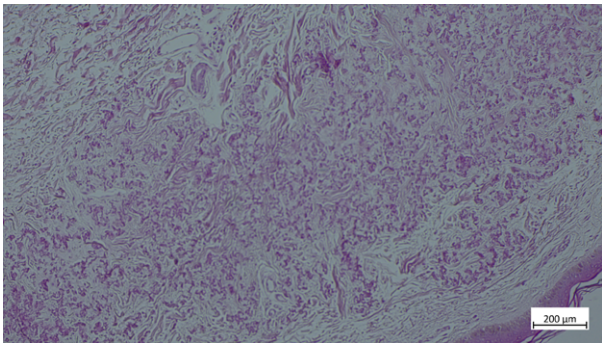

Figure 2: H&E staining highlights the presence of short, coiled, and frayed basophilic elastic fibers, with calcium deposits between them.

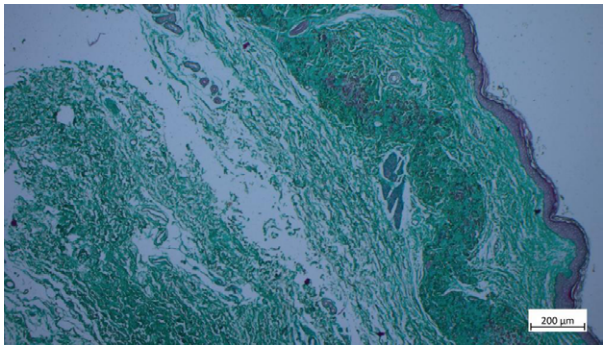

Figure 3: Masson's Trichrome Staining, which shows enhanced accentuation of elastic fibers.

The prevalence of this condition is rare, with a higher predisposition in women. Its early knowledge and diagnosis are crucial in preventing future complications, as it is a disease with systemic involvement.[1]

The purpose of this article is to report the first case of Pseudoxanthoma Elasticum in Ecuador. As a hereditary disease, the management in this case focused on educating the patient, providing genetic counseling, dietary and lifestyle changes, and appropriate follow-up to achieve early detection of possible complications.

CONCLUSION

Pseudoxanthoma Elasticum is a rare condition that has not been reported previously in our country. We report a patient with cutaneous manifestations confirmed by histopathological examination. In our patient's case, no ocular involvement was found, but the presence of Ebstein's anomaly and hypertension could be associated with the underlying pathology.

Given that the disease is chronic and involves systemic involvement, it is essential to provide the patient with the appropriate education to avoid future complications and ensure early diagnosis of any associated conditions.

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