



## PSEUDOXANTHOMA ELASTICUM: CASE REPORT

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### ABSTRACT

Pseudoxanthoma elasticum is a rare multisystem disease that can affect the skin, eye, cardiovascular system and gastrointestinal tract. A female patient at the age of 18 applied with the complaint of skin lesions on the neck and on both antecubital areas. Yellow papular lesions were determined in these areas during a physical examination. The punch biopsy material taken from the neck skin was assessed to be compatible with pseudoxanthoma elasticum. As the prognosis of pseudoxanthoma elasticum depends on extracutaneous organ involvement, early diagnosis of the disease gains importance in terms of reducing and even preventing systemic complications.

**Keywords:** Pseudoxanthoma elasticum, skin

### ÖZET

Pseudoxanthoma elasticum nadir görülen, deri, göz, kardiyovasküler sistem ve gastrointestinal sistemi etkileyebilen multisistem hastalığıdır. 18 yaşındaki kadın hasta boyun ve her iki antekubital bölgede cilt lezyonları şikayeti ile başvurdu. Fizik muayenesinde bu bölgelerde sarı papüler lezyonları saptandı. Boyun cildinden alınan punch biyopsi materyali pseudoxanthoma elasticum ile uyumlu olarak değerlendirildi. Pseudoxanthoma elasticumun prognozu ekstrakutanöz organ tutulumuna bağlı olduğu için hastalığın erken tanınması, sistemik komplikasyonların azaltılması ve hatta önlenmesi açısından önem kazanmaktadır.

**Anahtar Kelimeler:** Pseudoksantoma elastikum, cilt

### INTRODUCTION

Pseudoxanthoma elasticum (PXE) is a connective tissue disease characterised by the progressive fragmentation of the elastic fibers at a frequency of 1/100.000 (1). PXE may be autosomal dominant, autosomal recessive, or sporadic (2). Not a definite geographic and racial distribution has been specified. Patients apply with the complaints of skin lesions, decrease in the visual acuity, gastrointestinal system (GIS) bleeding and cardiac complaints (3).

### CASE REPORT

The female patient at the age of 18 applied with skin complaints existing for approximately three years. The patient had a history of occasional nose bleeds. Yellow papular lesions were observed in the neck area and both antecubital areas during physical examination (PE) [Figure 1]. Mild hyperkeratosis and irregular acanthosis were observed in the epidermis during the histopathological examination of the punch biopsy material taken from the neck area. Irregularities in elastic fibres, granular eosinophilic material and mild perivascular lymphocytic infiltration were observed in the middle dermis [Figure 2]. Colouring was observed in the granular material with Elastic Von Gieson (EVG) stain [Figure 3]. In the light of these findings, the case were reported to be compatible with PXE. No kind of a problem was detected in the cardiologic examination of the patient.



Figure 1: Papular and partially atrophic looking lesions on the neck area

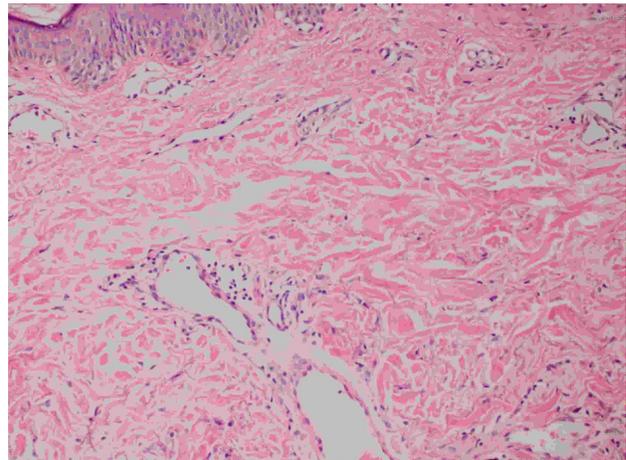


Figure 2: Irregularity in the elastic fibres and eosinophilic granular material in middle dermis (HEX200)

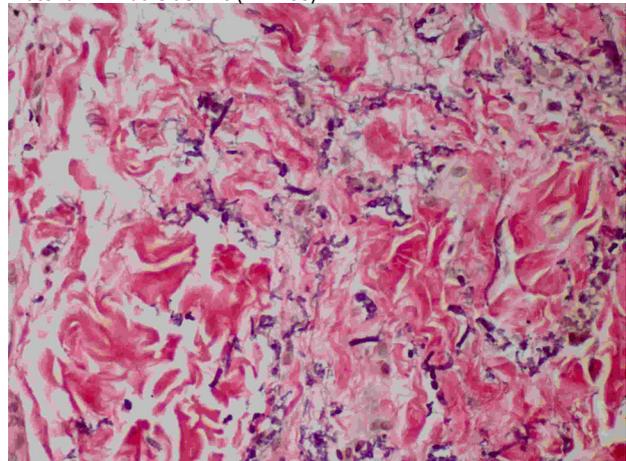


Figure 3: Calcification areas in middle dermis (Elastic Von Gieson x40)

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## DISCUSSION

Pseudoxanthoma elasticum is the rare degenerative disease of the elastic tissue. Although its beginning is most common in the second decade, it can also be seen among the children. It is twice as frequent among women (4). Our case is compatible with the literature in terms of age and gender. The skin lesions are in the form of yellow papules and they especially hold the skin of flexural areas, neck, axilla, umbilicus, ankles, groin, antecubital and popliteal fossa. The lesions may also rarely hold the rectum, soft palate, vagina, nasolabial folds and nasal mucosa(5). Our case also complains of nose bleeding. This makes us think about the nasal mucosal involvement. Skin lesions are generally seen in the early period of the disease. The skin with the lesion looks loose and creased. The Koebner phenomenon in PXE is positive. Degeneration and calcification are observed in the elastic fibres of the middle dermis in PXE; papillary and deep dermis are not affected. The collagen fibres look irregular, and scattered granular material is observed among these fibres (6). The irregularity in collagen fibres in the middle dermis and granular material among these were observed in our case. Positive staining was observed in this material with EVG stain. Ocular changes may occur in the form of decrease in visual acuity. Angioid streaks may be observed among the findings related to the eye. These are irregular, generally in the form of bilateral jagged radiating lines in the perimacular area of the retina. Ophthalmoscopic examination findings are haemorrhages, peripapillary atrophy and subretinal neovascularisation (2). In PXE cases, it is easier to diagnose the cases where the skin and eye findings are observed together. Ophthalmoscopy features were not found in our case. Vascular involvement includes the degenerative change and fragmentation of the internal and external elastic lamina. Consequently, the blood vessel wall becomes prone to rupture and aneurysm. Hypertension, angina, myocardial infarction, claudicatio, aneurysm, stenosis and intracerebral haemorrhage can be observed in vascular system involvement (7). No cardiological problem was found in our case. Hematemesis and melena are observed as gastrointestinal system lesions. The organ that is most affected in GIS is the stomach (8). There are no GIS findings in our case. Family members should be examined as pseudoxanthoma elasticum cases can exhibit genetic transmission. Early diagnosis is important in terms of preventing systemic complications.

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### Conflict of Interest

The author declares that he has no conflict of interest