

# Pseudoxanthoma Elasticum: A Report of Three Cases in a Family

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Received: June 5, 2008  
 Accepted: June 21, 2008

## Abstract

Pseudoxanthoma Elasticum is an inherited disorder characterized by generalized fragmentation and progressive calcification of the elastic tissues in the dermis, blood vessels and Bruch's membrane of the eye. Herein, we report 3 patients in a family with pseudoxanthoma elasticum: a father and his two sons. (*Iran J Dermatol 2008;11: 173-175*)

**Keywords:** pseudoxanthoma elasticum, angioid streaks, elastic fibres

## Introduction

Pseudoxanthoma Elasticum (PXE) is an inherited systemic disease of the connective tissues primarily affecting the skin, retina, and cardiovascular system. It is pathologically characterized by elastic fiber mineralization and fragmentation.<sup>1-5</sup> Biopsy of clinically normal skin or scar tissues in patients with angioid streaks may show the histological features of PXE.<sup>4</sup>

The inheritance of PXE is often autosomal recessive but autosomal dominant and sporadic cases have also been reported.<sup>5-7</sup> Here we report a male patient with his two affected sons.

## Cases Report

### Case 1

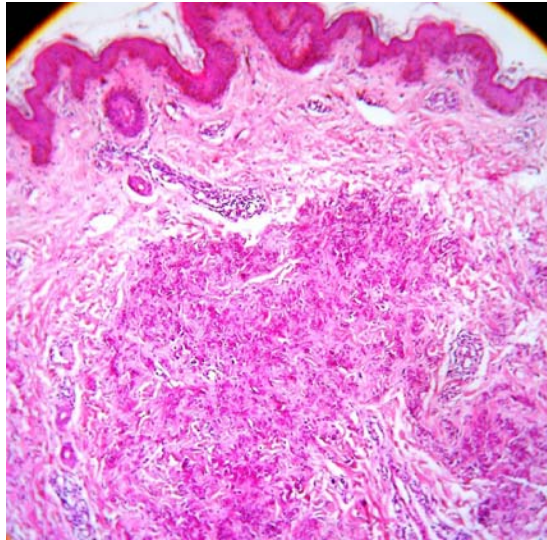
The patient was a 42-year-old male presented with yellowish papules around his neck. The lesions had been present since 15 years ago. The patient also complained of blurred vision from 5 years ago. On physical examination,

1-2 mm yellowish papules were observed around the neck together with skin laxity in this area (figure1). Ophthalmologic examination revealed bilateral retinal angioid streaks for which the

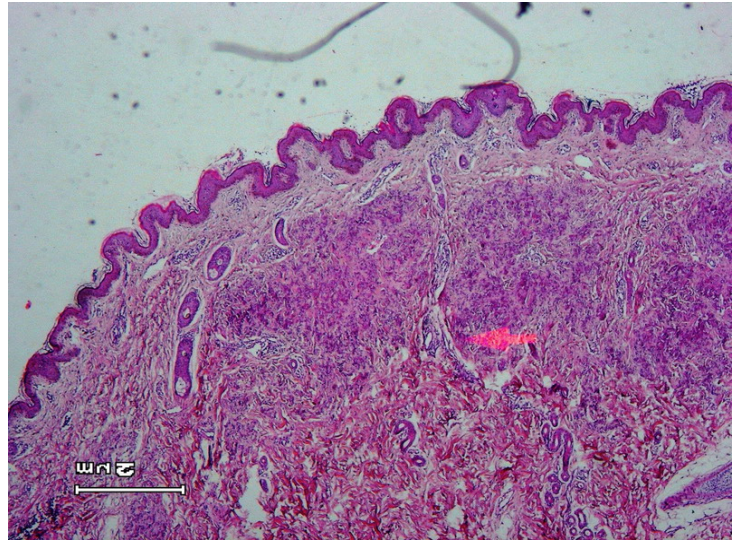
patient had undergone laser therapy 5 years ago although he was still suffering from blurred vision. A skin biopsy was performed which showed degenerated and irregular elastic fibers in the dermis confirmed by Orcein staining and the diagnosis of PXE was made (figure 2,3). Cardiovascular and pulmonary systems and blood pressure were normal. Routine laboratory tests



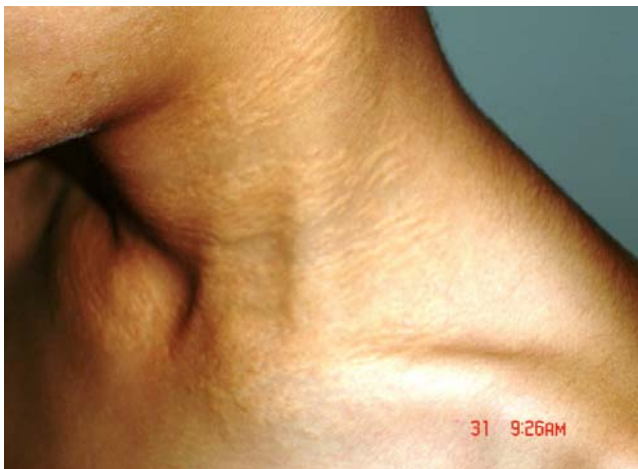
**Figure 1:** PXE lesions and skin laxity around the neck of a 42-year-old patient were within normal limits.



**Figure 2:** Degenerated elastic fibers are observed in the upper and mid dermis.( H&E staining 100X)



**Figure 3:** Degenerated elastic fibers are stained black. (Orcein staining x40)



**Figure 4:** PXE papules around the neck of a 12-year-old patient



**Figure 5:** PXE lesions around the neck of an 18-year-old patient

### Case 2

The case one's son was a 12-year-old male presented with yellow lesions in the neck, axilla and groins. The patient had these lesions since 1 year ago. On physical examination, lots of small yellow papules were observed symmetrically distributed in the mentioned areas (figure 4). The diagnosis of PXE was pathologically confirmed. Ophthalmologic, cardiovascular, and pulmonary examinations and blood pressure were normal. Routine laboratory tests were also normal.

### Case 3

The case one's other son was an 18-year-old male who presented with skin lesions in the neck and axilla since a few years ago. On physical

examination, 1-2 mm yellowish papules were noted in these areas which were distributed in a bilateral fashion (figure 5). Skin biopsy confirmed the clinical diagnosis of PXE. All examinations and routine laboratory tests were normal.

### Discussion

Pseudoxanthoma elasticum is a progressive disorder of elastic fibers in skin, eyes and arterial walls caused by mutations in the *ABCC6* gene.<sup>5</sup> The inheritance pattern in PXE is usually autosomal recessive, but autosomal dominant inheritance and sporadic cases have been reported.<sup>5-9</sup> The incidence of PXE is about 1/160000.<sup>9</sup>

The skin lesions of PXE consist of small yellowish linear papules or reticulate patterns in confluent plaques. The skin is soft, lax and slightly wrinkled and may hang in folds. The sites of predilection are the sides of the neck, below the clavicles, the axilla, abdomen, groins, perineum and thighs.<sup>7</sup> A person is considered to have definite PXE if they have two of the following three criteria: characteristic ophthalmologic signs, characteristic dermatologic signs, and a positive skin biopsy.<sup>5</sup> There are some cases of PXE that have been presented with gastrointestinal bleeding.<sup>8</sup> Our first case had ophthalmologic and dermatologic signs of PXE together with a positive skin biopsy. The other cases, who were the sons of case one, only had dermatologic signs and a pathologic confirmation without ophthalmologic signs but there are some reports of eye involvement in children.<sup>10</sup> In the literature, three families with definite PXE in two successive generations have been reported.<sup>5</sup> Our cases showed PXE in two successive generations.

PXE with angioid streaks has been reported in beta thalassemia patients.<sup>11</sup> Our patients did not have any hematological abnormalities. The patients with PXE should be periodically examined by an ophthalmologist because there is risk of retinal hemorrhage and blurred vision.<sup>12</sup> In patients who do not have typical skin lesions for PXE but have angioid streaks, biopsy of axillary skin or scars should be performed.<sup>13</sup> Every patient with angioid streaks who presents with premature cardiovascular diseases may have histological evidence of PXE, whether or not skin lesions are present.<sup>13</sup> The only other conditions that are regularly associated with angioid streaks are sickle cell anemia and Paget's disease of the bone, and they have occasionally been seen in patients with hyperphosphatemia, Ehlers-Danlos syndrome, lead poisoning, trauma, pituitary disorders and intracranial disorders.<sup>13</sup>

In the absence of cardiac risk factors, patients with myocardial infarction or other signs of atherosclerotic vascular diseases at an early age should be evaluated for PXE.<sup>13</sup>

In two large case series, 13% and 52% of PXE patients had chest pain. In some cases, chest pain and myocardial infarction appeared before the age of 30.<sup>14</sup> Diagnosing PXE is important, because some of the complications can be prevented by avoiding platelet inhibitors such as aspirin. Arterial grafts should not be used for coronary artery bypass surgery in patients with PXE because of the possible calcification of the internal elastic lamina<sup>13</sup>; therefore, cardiovascular and ophthalmologic

evaluations should be done periodically in PXE patients.

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