A case of pseudoxanthoma elasticum

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Pseudoxanthoma elasticum (PXE) is a rare, genetic, multisystem connective tissue disorder primarily affecting the skin, eyes, and the cardiovascular system (CVS) [1, 2]. PXE is characterized by progressive pathologic calcification and fragmentation of the elastic tissue in the affected organs. PXE appears to be present in all over the world's populations. In different reports, the prevalence is given to range between 1:25,000 and 1:100,000 [3]. Due to lack of adequate knowledge among patients and physicians, delay in diagnosis, and genetic heterogeneity, the exact prevalence of PXE might be underestimated. Although autosomal recessive inheritance patterns have mainly been reported, some families show autosomal dominant inheritance and some cases are sporadic without family history [4]. In most cases, the skin is the first organ system affected. Skin changes are usually detected in the second or third decade [5]. The affected skin eventually becomes lax, redundant, and inelastic. PXE also affects ocular fundus due to pathologic changes in Bruch's membrane. Early ocular findings are pigment irregularities of the temporal fundus called "Peau d' orange", angioid streaks due to breaks of Bruch's membrane and later choroidal neovascularisation and disciform scars [1]. CVS manifestations of PXE include arterial hypertension, peripheral arterial disease, angina

pectoris, restrictive cardiomyopathy, mitral valve prolapse, and sudden cardiac death [3, 6].

A 61-year-old female patient was referred to our dermatology outpatient clinic by the ophthalmology clinic because of angioid streaks detected on fundus examination. Patient did not have any subjective complaints. She had had history of diabetes for 11 years and coronary artery disease for 10 years. On dermatologic examination, we detected yellow cobblestone like lesions with lax, redundant, and inelastic skin over the anterior and lateral parts of the neck and axilla (Fig. 1). She had not consulted any dermatologist before because of these lesions.

Patient had applied to the ophthalmology outpatient clinic because of visual impairment. Best corrected visual acuities were 0.2/0.2 and intraocular pressures were 15/15 mmHg respectively for the right and left eyes. Anterior segments were normal. Fundoscopy revealed bilateral angioid streaks originating from the optic disk, radiating outwards especially under the macula as grayish–brownish irregular lines (Figs. 2 and 3). Optical coherence tomography examination revealed increased reflectivity at the level of retinal pigment epithelium–Bruch's membrane–choroid complex. Biopsy taken from the affected skin showed calcified and fragmented elastic fibers in the middle and lower dermis and confirmed the diagnosis.

PXE is a rare connective tissue disorder leading to a broad range of complications including mainly the skin, eyes, and CVS [1]. Clinical findings are due to pathologic calcification and fragmentation of elastic tissue in the dermis of the skin, Bruch's membrane of the retina, and intima of the vascular arteries [5]. These pathologic changes result in characteristic skin lesions, visual problems, various CVS complications, and rare problems such as gastrointestinal system bleeding [7].

Clinical manifestations of PXE are rarely present at birth. The skin is the first organ affected in most cases and the

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Fig. 1 Yellow cobblestone like lesions with lax, redundant, and inelastic skin over the anterior neck

average age of diagnosing skin lesions is 22 years [4]. Early skin changes are small asymptomatic soft papules of yellow–ivory color presenting in a reticular pattern and they usually start over the neck and later affect axilla groin and flexural areas [1]. The affected skin eventually becomes lax, redundant, and inelastic. Skin biopsy is diagnostic. Although it is the skin lesions which are detected first in PXE patients, this was not true for our case. She had not regarded her lesions as a disorder and had not consulted a dermatologist before. Due to delay in diagnosis, ocular and CVS complications had already developed. After the diagnosis of ocular complications, the patient was referred to us and we confirmed the diagnosis by the biopsy.

Fig. 2 Angiod streaks originating from the optic disk, radiating outwards under the macula in the right eye

Fig. 3 Angiod streaks originating from the optic disk, radiating outwards under the macula in the left eye

The earliest changes of PXE in the retina are pigment irregularities called “Peau d’orange” temporal to the fovea. Angiod streaks are irregular linear breaks or dehiscences in thickened and calcified Bruch’s membrane extending from peripapillary area into the peripheral fundus [8]. In later stages, a full thickness defect may develop within Bruch’s membrane with ingrowth of fibrovascular tissue leading to choroidal neovascularisation (CNV). Both angiod streaks and “Peau d’ orange” are usually bilateral lesions. Practically all PXE patients develop angiod streaks within 20 years after the diagnosis [9]. Choroidal neovascularisation is the most serious complication of angiod streaks and occurs in 72–86 % of cases [10]. Our patient has reduced visual acuity bilaterally because of angiod streaks localized under the macula like “spider’s web” but she has not developed CNV yet. She does not have diabetic retinopathy which may also cause visual impairment. She has been advised to come for a periodic control to the retina department of the ophthalmology clinic.

PXE has serious complications leading to morbidity and mortality. In a study, it was shown that vision-specific functioning had a significant impact on emotional well being of the patient whereas cardiovascular disease had less impact. The dermatologists must be aware of this rare disorder and as soon as a case is diagnosed; necessary consultations should be performed and patients should be informed about all possible complications.

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