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RESEARCH ARTICLE

PSEUDO-XANTHOMA ELASTICUM (PXE) IN A 30-YEAR-OLD PRIMIGRAVIDA: A CASE REPORT

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Abstract

Introduction Pseudo-xanthoma elasticum (PXE) is a rare inheritable condition (affects 1 in 25000 to 1 in 100000) with autosomal recessive inheritance. Skin changes may appear in childhood but due to asymptomatic and benign look of the condition, the diagnosis is frequently not made until serious systemic or ocular complications develop in the third or fourth decade of life. **Case report** A 30-year-old primigravida in her 20 weeks of pregnancy reported to Dermatology OPD, RIMS, Imphal with complaints of red colored, itchy skin eruptions over her abdomen since the last two months. The lesions were insidious in onset and gradually increasing in size and numbers since onset of pregnancy. She was diagnosed as Pseudo-Xanthoma Elasticum in 2005, when she presented with asymptomatic skin lesions over neck .On ophthalmological examination she was found to have Angioid streaks in the retina.There were no other concurrent complaints suggestive of systemic or ocular involvement. On examination, there were multiple erythematous papules, few coalescing to form erythematous plaques with ill defined borders on both sides of her flanks and peri-umbilical area. The skin over the anterior part of neck showed characteristic 'plucked chicken skin' appearance of PXE. The nape of neck and both lateral surfaces of the neck showed 'sagging of the skin'. Histopathological examination revealed characteristic fragmented elastic fibers in mid-dermis. Patient reported after normal vaginal delivery with no aggravation of skin lesions and onset of any unfavorable systemic manifestations.**Conclusion** Diagnosis of PXE is usually delayed due to its benign nature. Dermatologists should have high degree of suspicion to diagnose at initial stages of the disease to avoid serious complications. This case is reported for its rarity and pregnancy outcome.

INTRODUCTION

Pseudo-xanthoma elasticum (PXE) is a rare inheritable condition (affects 1 in 25000 to 1 in 100000) with autosomal recessive inheritance. Skin changes may appear in childhood; but due to asymptomatic and benign look of the condi tion, the diagnosis is frequently not made until serious systemic or ocular complications develop in the third or fourt h decade of life.

Case report

A 30-year-old primigravida in her 20 weeks of pregnancy reported to Dermatology OPD, Regional Institute of Medi cal Sciences, Imphal with complaints of red colored, itchy skin eruptions over her abdomen since the last two month s. The lesions were insidious in onset and gradually increased in size and numbers since onset of pregnancy. She had been diagnosed as Pseudo-Xanthoma Elasticum in 2005, when she presented with asymptomatic skin lesions over t he neck .On ophthalmological examination, she was found to have Angioid streaks in the retina. She had not receive d any specific treatment for her current skin problem. There were no other concurrent complaints suggestive of syste mic or ocular involvement. On examination, there were multiple erythematous papules, few coalescing to form eryth ematous plaques with ill defined borders on both flanks (Fig.1). Few papules around umbilical area were also noted. The skin over the anterior part of neck showed characteristic 'plucked chicken skin' appearance of PXE. The nape o f neck and both lateral surfaces of the neck showed 'sagging of the skin' (Fig.2). No other significant skin lesions no ted over other flexures or non-flexural sites. General physical and systemic examinations were within normal limits. Histo-pathological examination revealed characteristic fragmented elastic fibers in mid-dermis(Fig.3). The patient re ported after normal vaginal delivery with no aggravation of skin lesions or onset of any unfavorable systemic manife stations.

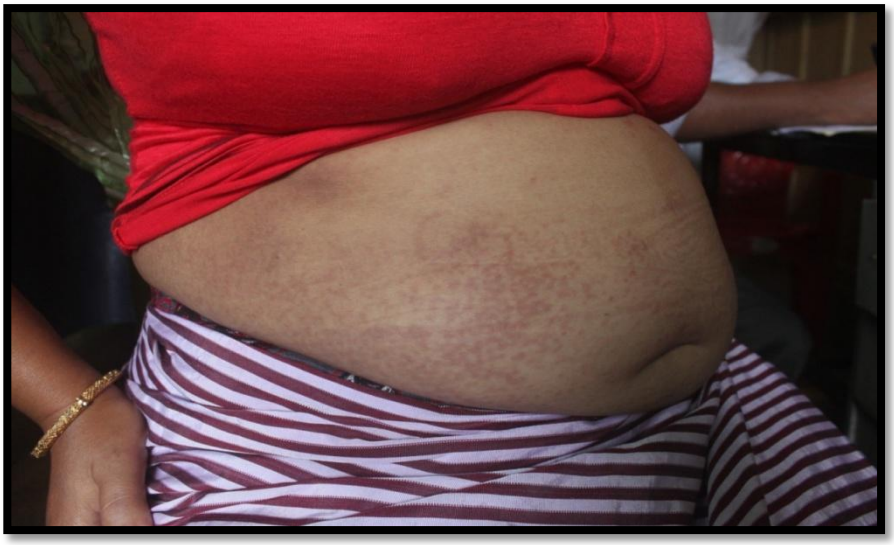

Fig. 1.Erythematous papules and plaques on anterior abdomen, both flanks and peri-umbilical area

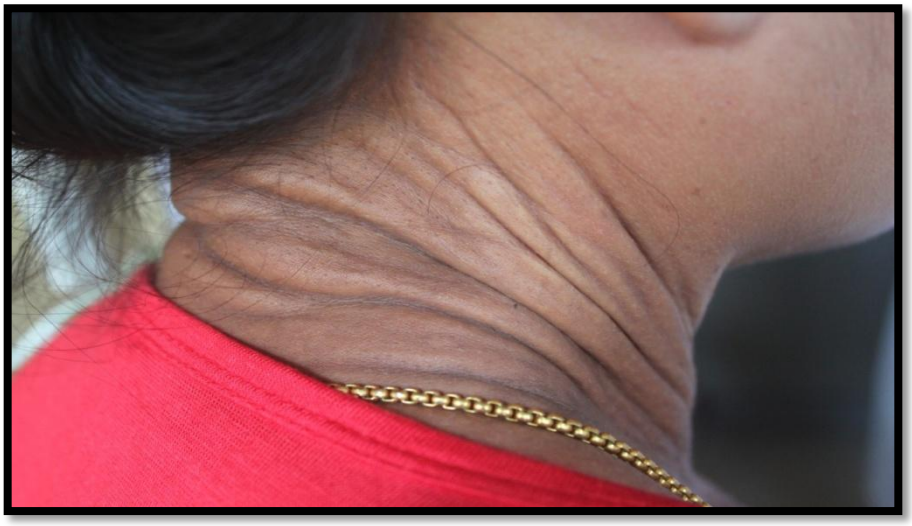

Fig.2. Sagging of skin on lateral aspects of neck

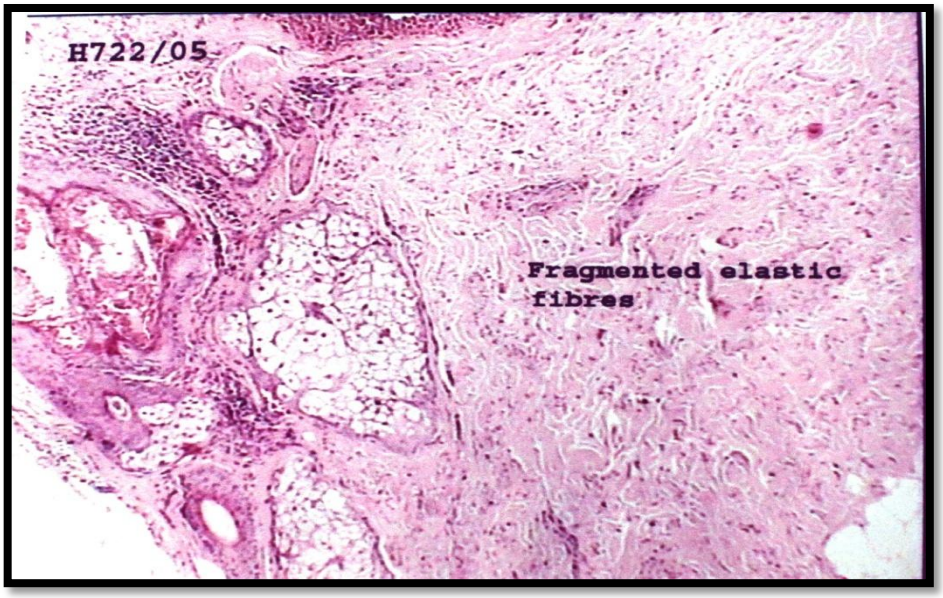

Fig. 3. Histopathology findings showing fragmented elastic fibers in mid -dermis

Discussion

PXE is due to loss of function of ABCC6 gene. This defect results in the decreased plasma anti-mineralization capacity due to reduced matrix gla protein and /or fetuin-A activity allowing progressive mineralization of elastic fibers in target organs like the skin, eye and cardio-vascular system. Calcification of elastic fibers in the dermis, the media and intima of medium sized arteries and Brusch's membrane of the eye produces the characteristic clinical and histo-pathological changes of PXE.[1] Only cutaneous manifestations and Angioid streaks are noted in our case report which is not uncommon.

PXE has characteristic thin, yellowish papules typically appearing in the flexural areas in the first or second decade of life with the lateral neck being the first area to be classically affected.[1] Other flexural folds of axilla, groin and popliteal fossa are frequently involved.[2] Trunk involvement is rare. Periumbilical pseudoxanthoma elasticum (PXE) presenting in a distinctive clinical pattern has been reported in six middle-aged, multiparous women. Other sites usually affected in PXE were spared. The clinical picture in these cases differed from classic PXE.[3]

As seen in our case, PXE characteristically has papules coalescing to form cobble-stone like plaques giving rise to 'plucked chicken skin' appearance over time. The patient also has characteristic sagging of the skin in the neck due to loss of recoil as a result of impaired function of the elastic fibre network. A classical ocular finding, but not pathognomonic of PXE , is Angioid streaks in the retina. It can cause progressive loss of vision by age 40 and legal blindness in few cases.[1] Angioid streaks were detected in our case when she was diagnosed in 2005. However, she does not have any symptoms suggesting visual impairment presently.

PXE can have cardiological manifestations mainly due to formation of atheromatous plaques in elastic media and intima of primarily mid-sized arteries due to progressive calcification. Sequelae may include intermittent claudication, loss of peripheral pulses, reno-vascular hypertension, angina pectoris, myocardial infarction and stroke. There is also increased prevalence of mitral valve prolapse.[1,5] Our case did not have signs and symptoms suggesting cardio-vascular involvement.

Histo- pathological features of PXE show distorted, fragmented elastic fibers in the mid and deep reticular dermis with calcium deposits on the altered elastic fibers on routinely stained sections in advanced cases. Both findings were present in the skin biopsy of our case.

PXE is not associated with markedly increased fetal loss or adverse reproductive outcomes. Although a few pregnancies were associated with worsening of skin manifestations, there was no correlation pregnancy with ultimate severity of skin, ocular or cardiovascular manifestations. There is no basis for advising women with PXE to avoid becoming pregnant, and most pregnancies in PXE are uncomplicated.[4,6]

There is no specific treatment for the condition, but regular follow-up should be done to identify and treat the compli cation early.

Conclusion

Diagnosis of PXE is usually delayed due to its benign nature and initial symptoms may mimic many other papular s kin eruptions. As serious complications are associated with it dermatologists should have high degree of suspicion to diagnose at initial stages of the disease. In females, complications related to pregnancy and possible outcomes of pr egnancy should be discussed with the patient along with genetic counseling as it is an inheritable disorder. This case is reported for its rarity and pregnancy outcome.

References:

1. Ringpfeil F, Uitto J. Heritable Disorders of Connective Tissue. In : Bolognia JL, Jorizzo JL, Rapini RP, editors. Dermatology. 3rd Ed. Spain: Elsevier Saunders publishing ;2012. p. 1612-5.
2. Weedon D. Disorders of elastic tissue. In : The Skin (Symmers W StC, ed), 3rd ed. London : Churchill Livingstone, 1992: 366-7.
3. Hicks J, Carperter CL, Reed RJ. Periumbilical perforating pseudoxanthoma elasticum. Arch Dermatol 1979; 115: 300-3.
4. Bercovitch L, Leroux T, Terry S, Weinstock MA. Pregnancy and obstetrical outcomes in pseudoxanthoma elasticum. Br J Dermatol. 2004 Nov;151(5):1011-8.
5. Mendelsohn G, Bulkley BH, Hutchins GM. Cardiovascular manifestations of Pseudoxanthoma elasticum.Arch Pathol Lab Med. 1978 Jun;102(6):298-302.
6. Valenzano M, Corticelli A, Podestà M, Nicoletti L, Saffioti S, Derchi L.Pseudoxanthoma elasticum and pregnancy: a case report. Clin Exp Obstet Gynecol. 2000;27(3-4):215-7.