PSEUDOXANTHOMA ELASTICUM (PXE)

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ABSTRACT:

PXE is a genetic disorder of connective tissue, characterised by progressive mineralisation of elastic fibres [1].The disorder is characterised by skin lesions, CVS manifestations & ocular lesions. It may be inherited as an autosomal recessive or autosomal dominant trait & has a prevalence estimated as 1 in 100,000 [1]. Exact cause of this disorder is unknown. Progressive calcification & fragmentation of elastic fibres in skin, in Bruch's membrane in the eye & in blood vessels appear to be responsible for the clinical manifestations of this disorder.

Our patient had yellowish, sagging, skin folds around neck simulating "Chicken neck". Similar lesions were also noticed over cubital fossae & axillae. She had typical reticulated, whitish lesions over lower labial mucosa. The typical ophthalmic finding of "Angioid Streaks" was not seen in our patient. She was diagnosed as PXE on clinical & histological grounds.

Key word : *Pseudo Xanthoma Elasticum (PXE)*

CASE REPORT :

A thirty year old Omani female patient presented with abnormal skin around the neck for last many years. Patient also attended ENT clinic for pain in the lower part of throat. Otherwise patient is comfortable, without any other signs & symptoms.

Skin around neck, cubital fossae, arms show yellowish discoloration with redundant folds which can be pulled & stretched. Patient also had whitish reticulated lesions over lower labial & adjacent mucosa. Rest of the skin & mucosa do not show any significant abnormality. Her blood pressure was 130/80 mm of Hg, Peripheral Pulse : Normal. Ophthalmic Examination did not show angioid streaks however pigmentary mottling seen over equatorial region. No other systemic manifestations were noted.

The laboratory investigations revealed : Urine Analysis : No hematuria; CBC : Within normal limits; Blood Sugar (Fasting) : 4.2 mmol/litre; TFT : Within normal limits. The ECG showed nonspecific T wave changes only.

The Skin Biopsy from neck region shows :

Epidermis showed thin layer of Keratin, no parakeratosis & minimal acanthosis. Overall the epidermis was thinned out.

Dermis showed altered elastic fibres which were degenerated & basophilic. Few faintly bluish areas suggestive of calcium deposits were seen.

Verhoeff's stain shows bluish black fibres & Orcein Stain showed brownish fibres. The histopathology shows typical findings of Pseudo Xanthoma Elasticum.

DISCUSSION & REVIEW OF LITERATURE :

PXE is a genetically transmitted elastic tissue disorder of skin & other parts of the body like eyes, kidney, CVS. It involves mainly skin around body folds. It shows cream colored yellowish, crepe like redundant patches specially on sides of neck, axillae, anticubital fossae[2]. Redundant folds in the neck is known as chicken neck [3]. Naso labial folds on face are exaggerated known as ' hound dog' appearance. Mucous membrane of mouth, vagina, rectum also show similar types of lesions. Skin changes usually occur in second decade of life. Involvement of skin may be progressive & it may involve the entire skin. Diagnosis is made by skin biopsy which is characterised by fragmentation & calcification of elastic fibres in middle & lower third of dermis. Although normal elastic fibres do not stain with H/E stain but altered elastic fibres in PXE stain blue because of their calcium content.

Angioid streaks [3] (85% of patient) is the characteristic ocular lesion of PXE (red brown curvilinear bands radiating from optic disc). It apparently results from breaks in Bruch's membrane associated with faulty elastic fibres in other portion - lamina elasticum. Fibro vascular in growth may result in retinal haemorrhage, detachment & severe visual loss. Other ocular lesions include characteristic yellowish mottling of the posterior pole called ' leopard spotting' & a reticular pigmentary pattern in retina [4]. The association of skin lesions & angioid streaks is called as " Gronblad - Strandberg Syndrome ".

Calcification of media of blood vessels with sub-

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sequent intimal perforation leads to serious complications. Claudication is the most common problem. Pulse is absent / diminished. Hypertesion is prevelant in adults & it is associated with renal artery involvement [1]. Gastro intestinal haemmorrhage appears due to fragile submucosal vessels. Bleeding may also occur in urinary tract.

Progressive calcification of elastin with fragmentation of elastic fibres in the areas of calcifiation is the basic defect in PXE [3]. In dermal blood vessels, abnormal elastin in the internal elastic lamina may be detected in the absence of mineralisation [5]. Abnormal amounts of proteoglycans have been detected in the skin & urine of patient with PXE. Abnormal proteolytic [6] & elastolytic activity have been reported in dermal fibroblasts. Similar types of skin changes also are reported in patients of cystinuria who are taking D. Penicillmine [7]. Both, autosomal dominant & autosomal recessive forms are reported in PXE [3]. Most of the patients appear to inherit as on autosomal recessive trait. The least form of the disorder involves skin only & presents with widespread skin sagging clinically similar to Cutus laxa[7]. Female outnumbers male in PXE.

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