Asymptomatic esophageal varices in a case of pseudoxanthoma elasticum

Sir,

Pseudoxanthoma elasticum (PXE) is a rare genetic disorder characterized by fragmentation and mineralization of elastic fibers. It can present with classical cutaneous features with extracutaneous involvement of eyes, medium-sized vessels, and gastrointestinal (GI) tract.[1-4] GI manifestations are reported in 13% of these patients, with most reports documenting gastric involvement.[4]

We describe a case of a 33-year-old married woman, born of a second degree consanguineous marriage, presented with sagging of skin on neck since 3 years and wrinkles on face since 2 years. The patient was apparently alright 3 years ago when she noticed gradual sagging of skin on the neck and axilla and wrinkling of face. There were no complaints of intermittent claudication, chest pain, dyspnea on exertion, visual disturbances, hematemesis, or malena. There was no contributory family history. General examination did not reveal any significant abnormality.

Cutaneous examination showed wrinkling on forehead, chin, cheeks, and neck. There was decrease in elastic recoil and sagging of skin on axilla and neck [Figure 1b, 1d]. Wrinkles were more prominent on chin, nasolabial fold, and outer canthus of eyes. Examination of neck had numerous minute yellowish papules arranged in reticular pattern that were apparent on stretching the skin giving a “cobblestone” or “morocco leather” appearance [Figure 1a, 1c].[1,2] Laxity of skin was also present in axilla. There was no evidence of oral or genital mucosal involvement.

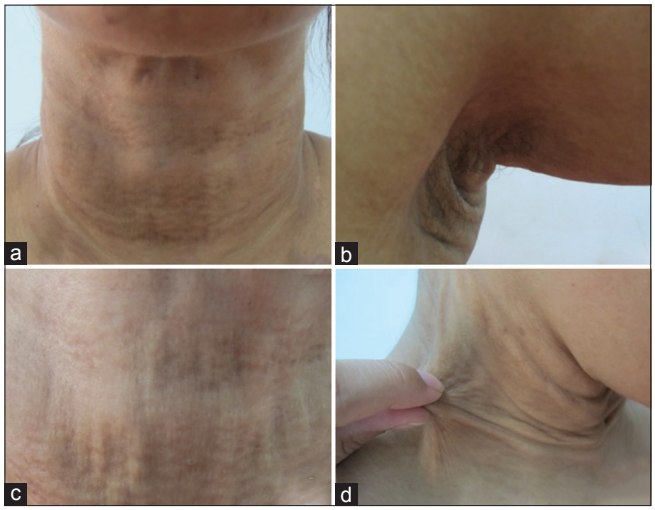

Figure 1: (a) Cobble stone or Morrocan leather appearance of skin on neck.(b) Laxity and sagging of skin on axilla (c) reticular pattern of minute yellow papules on neck (d) sagging of skin and lack of elastic recoil on lateral aspect of neck

Skin biopsy from lateral aspect of neck revealed fragmented elastic tissue in dermis and clumping of the fragmented fibers [Figure 2c], Elastic Van Gieson stain revealed fragmented elastic fibers confirming the diagnosis of PXE [Figure 2d]. Laboratory investigations were within normal limit. Ophthalmic evaluation with fundoscopy showed bilateral angioid streaks [Figure 2a]. Esophagogastroscopy showed grade two esophageal submucosal varices at 12 o’ clock position at esophagogastric junction [Figure 2b]. Chest X-ray and echocardiography showed no abnormality.

We confirm the diagnosis of PXE with esophageal varices.

PXE or Gronblad–Strandberg syndrome is a genetic disorder characterized by fragmentation and mineralization of elastic fibers containing connective tissue, mainly the mid-laminar layer of the dermis, Brusch’s membrane of the eyes, and medium-sized arteries.[1-4]

The disease prevalence is estimated at 1:160,000 and is caused by autosomal recessive mutations in ABCC6gene on short arm of chromosome 16; however, a few autosomal dominant cases are reported.[1-4]

Reports hypothesize that a metabolic disorder where vitamin K metabolites do not reach the peripheral tissue results in clinical features, whereas others document an absence of RP6 that causes severe manifestations.[1,3] PXE has also been described in association with thalassemia and sickle

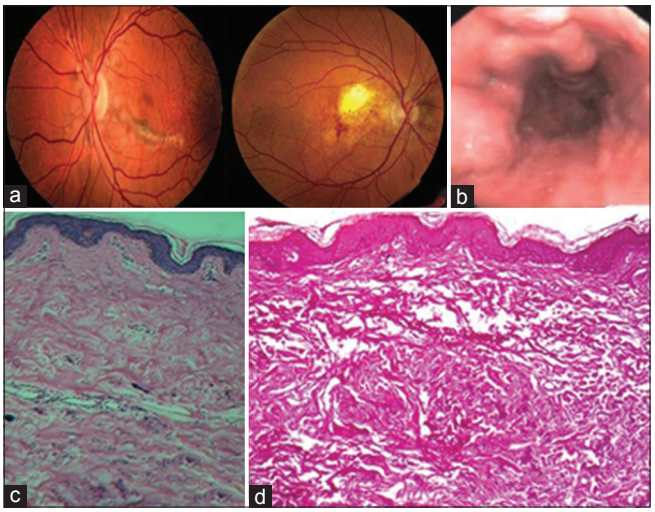

Figure 2: (a) Bilateral angioid streaks on ophthalmologic examination. (b) Endoscopic examination showing esophageal varices. (c) H and E stain slide 10X magnification showing fragmentation and clumping of fibres in dermis. (d)Elastic Van Gieson stain 10X maginification showing fragmented elastic fibers in dermis

cell anemia.[1,3] Gama-glutamyl carboxylase is another gene, which is implicated in a condition phenotypically similar to PXE.[1,3]

Classical presentation consists of discrete yellowish papules that are prominent on stretching skin, which later become confluent to form linear or reticular pattern which is described as “plucked chicken skin” or “Moroccan leather” or “cobble stone” appearance.[1,2] Skin over the flexures become lax and redundant with decreased elastic recoil. Some patients present with oblique mental creases on the chin and puckering of skin over the chin.[3] Ophthalmic manifestations involve peau d’orange appearance of Bruch membrane, mineralization of elastic fibers in Bruch membrane causes fissures, called as angioid streaks seen in 85% of the cases. Reports of retinal hemorrhage and detachment have been documented.[1-4] Medium and small-sized vessel calcification presents as intermittent claudication, myocardial infarction, angina pectoris, and atherosclerosis.[1,2,4]

Gastrointestinal (GI) manifestations are reported in 13% of the patients and usually involve the stomach.[1,4] GI involvement very often can constitute the first clinical manifestation of PXE. The first episode of GI bleeding may occur in the second decade of life. In pregnant patients of PXE, GI tract bleeding may occur due to vessel wall relaxation and vascular congestion of the stomach. An endoscopy performed far from the bleeding may show discrete, raised, submucosal yellowing lesions that macroscopically resemble characteristic PXE skin lesions; endoscopic examination during the bleeding may reveal a petechiae-like picture of diffuse hemorrhagic gastritis, active bleeding of the characteristic submucosal yellowish lesions and frequently a bleeding ulcer.[4] Hitoshi Nishiyama *et al*. have described various endoscopic characteristics of GI tract involvement, which includes small submucosal yellowish nodular lesion, cobblestone appearance, shallow ulcer, erythematous mucosa with numerous petechiae or erosions, and serpentine submucosal vessel.[1] Endoscopy in our patient shows the presence of a serpentine submucosal esophageal vessel at 12 o’ clock position. To our knowledge no such articles have been reported in Indian literature.

CONCLUSION

We report this case, which had classical cutaneous and ophthalmologic findings characteristic of PXE with the clinically asymptomatic finding of submucosal esophageal varices, which was diagnosed coincidentally on endoscopy. This suggest the role of routine endoscopic screening of PXE patients to detect the hidden manifestations and avoid catastrophic complications such as bleeding if followed up regularly.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

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Access this article online

Quick Response Code:

Website: www.idoj.in

DOI: 10.4103/2229-5178.171049

Cite this article as: Hemmady K, Tambe S, Jerajani HR. Asymptomatic esophageal varices in a case of pseudoxanthoma elasticum. Indian Dermatol Online J 2015;6:65-6.