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Case Study

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UNUSUAL OCULAR MANIFESTATIONS IN PATIENT OF PSEUDOXANTHOMA ELASTICUM: A CASE REPORT

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We describe a case of 28 year old female of pseudoxanthoma elasticum with angioid streaks in left eye and disc coloboma in right eye. This is an unusual presentation of pseudoxanthoma elasticum and not commonly seen.

A 28 year old patient was referred from department of surgery in view of ptosis in her right eye. She was admitted in department of surgery for splenomegaly with bleeding varices and hepatic adenomas.

On ocular examination the patient had congenital ptosis right eye.[image 1]

Her VA in right eye was 6/24 and left eye was 6/9. Anterior segment was normal in both eyes. On dilated fundus examination the patient had large well-defined excavation in the optic disc in right eye suggestive of disc coloboma and pigmented grayish irregular choroidal crack-like linear dehiscence, forming a network-like pattern, originating at the optic disc from all sides and extending radially suggestive of angioid streaks in left eye.[image2,3]

There was no evidence of choroidal neovascularization or retinal hemorrhages.

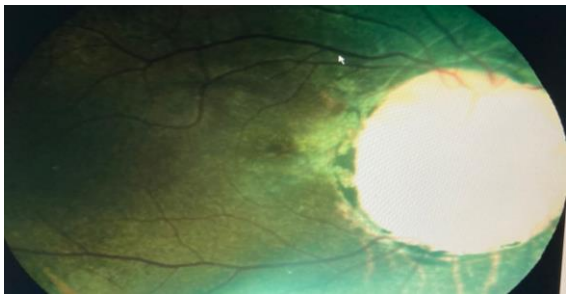
She was hereby referred to department of dermatology for hyperpigmented patches on neck resembling those of pseudoxanthoma elasticum.

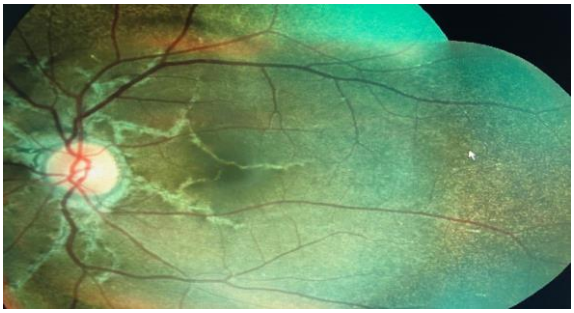
A final diagnosis of pseudoxanthoma elasticum was made.

The patient was warned regarding complications like choroidal rupture and choroidal neovascularization.

The patient was kept under regular followup to rule out progression of disease.

1. Congenital ptosis right eye.



2. Disc coloboma right eye. 

3. Angioid streaks left eye.

DISCUSSION

Pseudoxanthoma elasticum, or Gronblad-Strandberg syndrome, is an inherited disorder that involves multiple organ systems. The characteristic of the disease is degeneration and calcification of the elastic fibers. The incidence of PXE is 1:1,600,000 individuals. It affects females more than male with a female to male ratio of 2:1 and a mean onset of the disease around 13 years.[1]

Clinically, three organ systems are mainly affected by PXE: the eyes, skin and cardiovascular system. The skin is the furthermost and is usually the first organ affected by this disease. The clinical findings of PXE usually become apparent when the affected individuals reach the second and third decade of life. The ocular characteristic finding of this disease is called angioid streaks. They are often bilateral and consist of visible, irregular, linear, crack-like dehiscence of the calcified and brittle Bruch membrane.[2]

The usual sequence of developing eye abnormalities is peau d'orange or mottled hyperpigmentation of the retina, angioid streak. Other retinal lesions: pattern dystrophy, drusen of the optic nerve, peripheral “comet tails” lesions, subretinal fibrosis, and autofluorescent perimacular white spots. The risk of neovascular complications increased with age.[3]

Bruch membrane is rich in elastin and collagen and functions as a transporter of important nutrients and metabolites between the choriocapillaris and RPE. The mineralization of this membrane due to PXE is the underlying pathogenesis of angioid streaks. In PXE, the lack of anti-mineralization factor leads to calcium deposition in all elastic tissues all over the body. This calcification of the elastin-rich Bruch membrane leads to rupture of the blood vessels with subsequent deterioration of vision clinically.[2]

Treatment of angioid streaks depends on multiple factors including most importantly the location of the lesions near to or away from the fovea and whether the patient developed CNV or not yet.[1]

In the above case the patient also presented with unilalteral congenital ptosis which either can be an incidental finding or due to weaking of elastic fibres of levator muscle.

Additionaly our patient had only unilateral angioid streaks but disc coloboma in the other eye which is not commonly seen or reported in pseudoxanthoma elasticum.

CONFLICTS OF INTEREST: NONE.

FINANCIAL DISCLOSURE: NONE.

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