ABSTRACT

Pseudoxanthoma Elasticum is a rare autosomal recessive systemic disease that causes degeneration and fragmentation of elastic fibres with calcification. With the skin manifestations and the simultaneous occurrence of angioid streaks. Pseudoxanthoma Elasticum is also known as Gronblad-strandberg syndrome. It is a multisystem disorder and typically involves skin, eyes and cardiovascular system. Recognition of disease early helps to prevent complications and improves quality of life. We report a case of Pseudoxanthoma Elasticum, to obtain a better understanding of clinical features and early diagnosis of the disease. The case is interesting as patient primarily sought medical consultation due to ocular manifestations of disease.

KEYWORDS: Pseudoxanthoma Elasticum, Elastic Fibres, Angiod Streaks.

INTRODUCTION

Pseudoxanthoma elasticum (Gronblad-Strandberg syndrome) is the most common problem arises in the skin and eyes, and later in blood vessels in the form of premature atherosclerosis. The disease was first described by Rigal in 1881 and the term was adopted by Darier in 1896 as pseudoxanthoma elasticum by observing the specific skin changes in histopathological process. The Eye features was first described by Gronblad and Strandberg in 1929 and early atheromatosis by Carlbord in 1944. Pseudoxanthoma elasticum is two times more common in females. There is major manifestation of skin in few families while other has predominant cardiovascular and eye involvement. The skin involvement starts in second or third decade of life. It is crucial to diagnose disease early to avoid severe systemic complications. We report a case of Pseudoxanthoma Elasticum, to obtain a better understanding of clinical features and early diagnosis of the disease. The case is interesting as patient primarily sought medical consultation due to ocular manifestations of disease.

CASE REPORT

33 years of old male, clerk by occupation, admitted for defective vision on the right side for 15 months and left side for 3 months duration. This was associated with general weakness for 3 months. The patient was healthy till Jan 2013, when he started seeing objects double and distorted. He complained that his right eye vision was dim and words seen were blurred. This persisted till April 2014 when gradually double vision disappeared but blurring and dimness of vision progressed. In Dec 2013, he noted blurring of vision in his left eye also, which is gradually increasing. The patient also complained of body weakness for last 3 months. He says that he gets easily fatigued if he stands for some period or walks a little fast. Systemic review was unremarkable and there was no history of hypertension, diabetes mellitus and bleeding disorder. He was married with six children and all were healthy. On examination, a young man of average built. Vitals were normal and Systemic examination was unremarkable. Ophthalmological examination revealed angioid streaks in retina with neovascularization. Small well defined hemorrhages were present on the medial side of the disc and between disc and macula of the right eye with marked macular degeneration. Left eye also revealed angioid streaks with early retinal exudates and macular degeneration.

No other abnormality is detected.

On Examination of skin: Multiple redundant folds are present on the side of neck as shown in the photograph of this patient. This appearance is known as 'Chicken neck' appearance. Multiple small, circumscribed, yellowish, redundant patches are present on the anterior and posterior axillary folds, and popliteal fossae, as well as groin show skin hanging loosely and similar skin

FIGURE-1: FUNDSCOPY: ANGIOID STREAK IN RETINA IN PSEUDOXANTHOMA ELASTICUM PATIENT SHOWN

Correspondence to:
Shujaat Hussain
Professor of Medicine
Al-Nafees Medical College & Hospital
Isra University, Islamabad Campus, Pakistan

Email: shujaat.hussain@yahoo.com

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changes. There are no telangiectasias. 

As shown in the Figure-2 (loose folds of skin on the side of the neck, this appearance is known as chicken neck). All routine investigations were normal. Histopathology of biopsy taken from right side of neck showed degeneration of elastic fibers in dermis. They were fragmented and distorted consistent with diagnosis of Pseudoxanthoma elasticum. This patient was treated initially empirically with Vitamin A, Vitamin B and Vitamin E but without any beneficial effect. Photocoagulation was tried once a week for six weeks but there was little improvement. Considering the nature of this disease nothing more could be expected.

DISCUSSION

Pseudoxanthoma elasticum is an autosomal recessive systemic disorder of the elastic connective tissue. The determination of inheritance is by mutation in gene ABCC6 resided in the short arm of chromosome 16, erratically expressed in the liver and kidneys.

The disease especially involves the skin, eyes, and gastrointestinal tract and in small and medium arteries. The initial and common clinical feature is discoloration of skin and formation of yellow small papule around the sides of neck and flexural areas. There is absolute loss of central vision due to angioid streak that is caused by slow deposition of calcium in elastic fibers of retina. The cases of eye involvement must be monitored by fluorescein angiography and ophthalmoscope.

There are small and medium arteries by deposition of calcium leads to early athermatosis. Blood vessels involvement can lead to hypertension, heart attacks, stroke, peripheral vascular disease and gastrointestinal bleed.

There is no specific treatment is present to date. However the progression of disease is slowed by smoking cessation, moderate physical exercise, and diet with magnesium and vitamin K. All patients should be monitored by eye check up, blood count, lipid profile, and echocardiogram. Surgery is usually not performed due to risk of complications like formation of keloids, dehiscence and calcium deposition in surgical wound.

Patients with PXE are instructed to adopt healthy life style to delay cardiovascular complications and avoid high risk sports. The early diagnosis of PXE allows accurate provision of information and life style modification to avoid complications.

CONCLUSION

There should be a need to early diagnosis and life style modification with dietary supplementation to slow progression of disease. There should be periodical eye and cardiovascular follow-ups. The prognosis depends upon liability to haemorrhage. Haematemesis and subarachnoid haemorrhage both prove fatal. No definite treatment exists.

Contribution of Author:
Maryam Rashid: Provoking the idea of manuscript and Introduction.
Aysha Babar: Discussion writing.
Syed Saif Ur Rehaman: Collection of Reference for Introduction & Discussion
Kamil Shujaat: Final formatting of entire manuscript.
Shujaat Hussain: Final Proof Reading.

REFERENCES

