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. No other abnormality is detected.

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 changes. There are no telangiectasias.
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.

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REFERENCES