



Here is the answer to last month's case

Meet Nancy

- Nancy is a 35-year-old woman who first noted cream-coloured, pebble-like changes on her neck during her teens
- Her chin creases and mental fold deepened during her twenties
- Epistaxis and claudication have been problems
- Her father is vision impaired



What is your diagnosis?

- | | | |
|---------------------------|--------------------|-----------------------------|
| a) Fibroelastotic disease | c) Type 1 diabetes | e) Pseudoxanthoma elasticum |
| b) Ehlers-Danlos syndrome | d) Cutis laxa | |

Answer: E

Pseudoxanthoma elasticum (PXE) is an inherited, usually autosomal recessive disorder primarily of the elastic fibres. It is slightly more common in females. It may first present with dermatologic, ophthalmologic, or cardiac signs or symptoms.

Involvement of the neck or flexural folds with small, yellow to cream-coloured papules before the age of 30 is diagnostic for this condition.

There may be lax tissue changes of other areas in the skin, oral mucosa, stomach, or genitals.

Angioid streaks of the retina are seen in 85% of patients with PXE and may be the first finding. Macular deposits, hemorrhage, exudates, and drusen-like spots may be present. Progressive blindness may follow.

As elastic fibres degenerate, hemorrhage and epistaxis occur. More serious than this is the weakening of cardiac valves and the myocardium. Mitral valve prolapse is particularly common. Claudication, calcification of peripheral arteries, and premature hypertension in

younger individuals should also suggest this diagnoses. There is a great deal of variation in initial findings and in the progression of PXE.

There is no specific therapy. Care requires a multidisciplinary approach.

Stanley Wine, MD, FRCPC

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Congratulations
to our winner for the month of
May 2012
Dr. S. Chudasama
Winnipeg, Manitoba