

Snapshot

Pseudoxanthoma elasticum

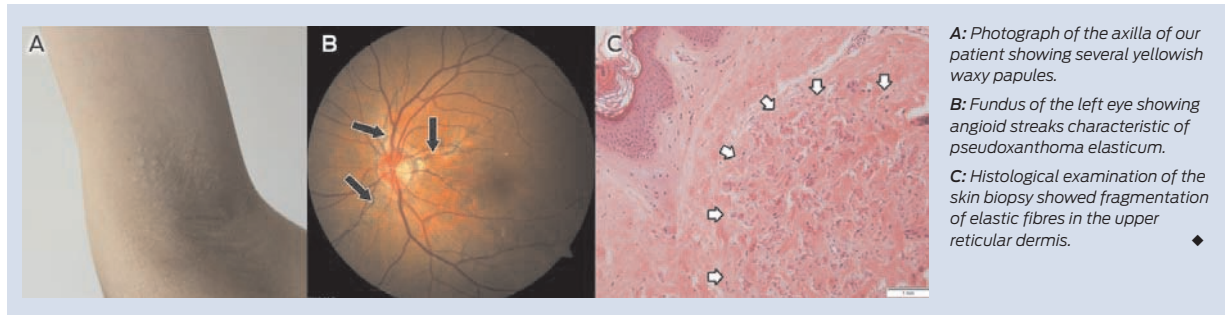
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A 31-year-old woman presented with a 20-year history of lesions on the skin of her neck and axillae. Her family history was unremarkable.

Physical examination revealed multiple pinhead-sized yellowish flat-topped papules on the skin of the axillae and lateral aspects of the neck (Figure, A). These lesions made us suspect pseudoxanthoma elasticum, so we performed a fundoscopic examination which showed obvious angiod streaks (Figure, B). A biopsy of the axillary

lesions showed a band of degenerated clumps of elastic fibres (Figure, C) and calcium deposits in the upper reticular dermis.

A diagnosis of pseudoxanthoma elasticum (PXE) was made. This condition is caused by mutations in the gene that encodes the ABCC6 protein, an ATP-binding cassette, subfamily C member.¹

1 Pfendner E, Uitto J, Gerard GF, et al. Pseudoxanthoma elasticum: genetic diagnostic markers. *Expert Opin Med Diagn* 2008; 2: 63-79. □