Pseudoxanthoma elasticum: under-recognized cause of early onset peripheral arterial disease?

Frank Schröder, Ingrid Hausser, Christiane Szliska, Holger Lawall, Curt Diehm

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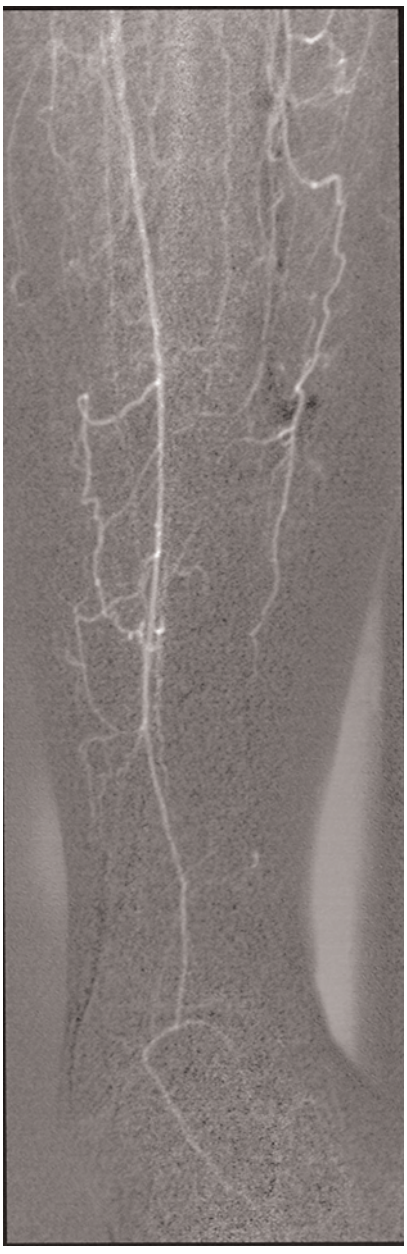
Images in vascular medicine

Pseudoxanthoma elasticum: under-recognized cause of early onset peripheral arterial disease?

Frank Schröder[a], Ingrid Hausser[b], Christiane Szliska[c], Holger Lawall[a] and Curt Diehm[a]

A 50-year-old female patient with critical limb ischemia was referred for evaluation and treatment of peripheral arterial disease (PAD). She had a history of stroke in 2002; however, no classical cardiovascular risk factor was detected. Angiography revealed a severe generalized PAD (Panel A). Small, yellowish, flat, soft papules were present in the neck (Panels B and C) and both antecubital fossae. A skin biopsy showed typical calcified elastic fibres (Panel D: part C) and disintegrated and fringed collagen fibrils (Panel D: parts A and B). These findings are consistent with pseudoxanthoma elasticum (synonym: PXE, Grönblad-Strandberg syndrome). It is a systemic inherited disease with a prevalence of 1:25 000–1:100 000 in white individuals.[1] The defect has been mapped to the ABCC6 gene on chromosome 16 encoding multidrug resistance-associated protein.[2] It causes fragmentation and calcium deposition in elastic fibres of skin, blood vessels, Bruch's membrane of the eye, and other organs. It is not known how this mutation causes these changes in multiple organs. One hypothesis is chronic oxidative stress.[3] Characteristically, patients suffer from skin lesions, cardiovascular manifestations such as early PAD, myocardial infarction or stroke, and ophthalmologic features (i.e. angioid streaks, peau d'orange and maculopathy). The anticipated prevalence in Germany is at least 1000 patients. However, only a few hundred are diagnosed. Therefore, the disease seems to be under-recognized. Pseudoxanthoma elasticum should be considered as a possible diagnosis in patients with early onset of PAD.



Panel A

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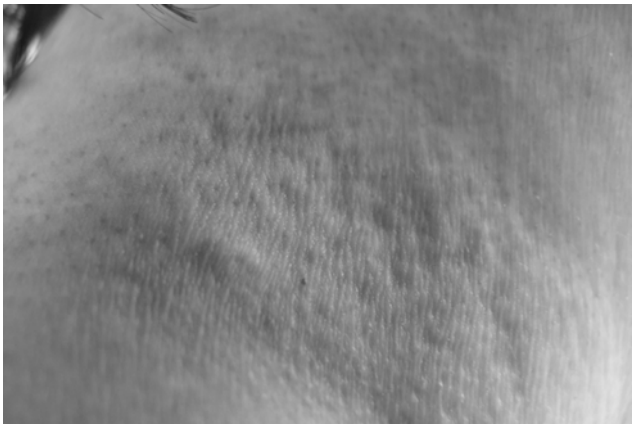
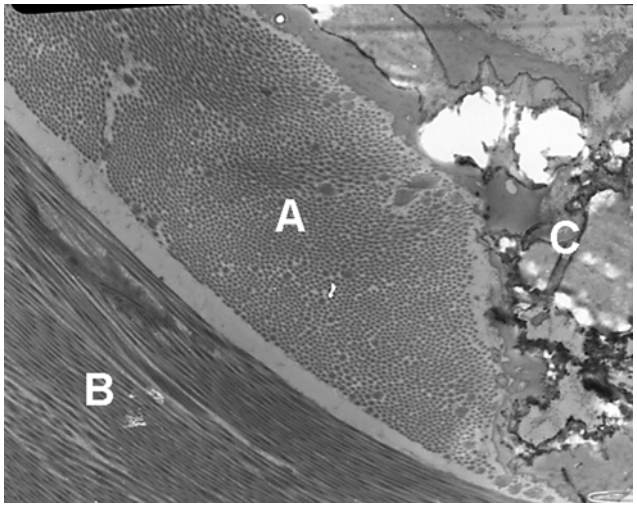
Panel B



Panel C



Panel D

References

1 Terry SF, Bercovitch L, Boyd CD. Pseudoxanthoma elasticum. *Gene Reviews* June 2001; updated November 2003. www.genetests.org

2 Le Saux O, Urban Z, Tschuch C et al. Mutations in a gene encoding an ABC transporter cause pseudoxanthoma elasticum. *Nat Genet* 2000; **25**: 223–27.

3 Pasquali-Ronchetti I, Garcia-Fernandez MI, Boraldi F et al. Oxidative stress in fibroblasts from patients with pseudoxanthoma elasticum: possible role in the pathogenesis of clinical manifestations. *J Pathol* 2006; **208**: 54–61.

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