The vascular phenotype in Pseudoxanthoma elasticum and related disorders: contribution of a genetic disease to the understanding of vascular calcification

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Résumé en anglais	Vascular calcification is a complex and dynamic process occurring in various physiological conditions such as aging and exercise or in acquired metabolic disorders like diabetes or chronic renal insufficiency. Arterial calcifications are also observed in several genetic diseases revealing the important role of unbalanced or defective anti- or pro-calcifying factors. Pseudoxanthoma elasticum (PXE) is an inherited disease (OMIM 264800) characterized by elastic fiber fragmentation and calcification in various soft conjunctive tissues including the skin, eyes, and arterial media. The PXE disease results from mutations in the ABCC6 gene, encoding an ATP-binding cassette transporter primarily expressed in the liver, kidneys suggesting that it is a prototypic metabolic soft-tissue calcifying disease of genetic origin. The clinical expression of the PXE arterial disease is characterized by an increased risk for coronary (myocardial infarction), cerebral (aneurysm and stroke), and lower limb peripheral artery disease. However, the structural and functional changes in the arterial wall induced by PXE are still unexplained. The use of a recombinant mouse model inactivated for the Abcc6 gene is an important tool for the understanding of the PXE pathophysiology although the vascular impact in this model remains limited to date. Overlapping of the PXE phenotype with other inherited calcifying diseases could bring important informations to our comprehension of the PXE disease.
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