



Generalized arterial calcification of infancy and pseudoxanthoma elasticum: two sides of the same coin

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Résumé en anglais	Generalized arterial calcification of infancy (GACI) is associated with biallelic mutations in ENPP1 in the majority of cases, whereas mutations in ABCC6 (ATP-binding cassette subfamily C number 6) are known to cause pseudoxanthoma elasticum (PXE). However, ABCC6 mutations account for a significant subset of GACI cases, and ENPP1 mutations can also be associated with PXE lesions. Based on the considerable overlap of GACI and PXE, both entities appear to reflect two ends of a clinical spectrum of ectopic calcification rather than two distinct disorders. ABCC6 and ENPP1 mutations might lead to alterations of the same physiological pathways.
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[1] [http://okina.univ-angers.fr/publications?f\[author\]=13985](http://okina.univ-angers.fr/publications?f[author]=13985)

[2] [http://okina.univ-angers.fr/publications?f\[author\]=13986](http://okina.univ-angers.fr/publications?f[author]=13986)

[3] <http://okina.univ-angers.fr/publications/ua8259>

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