



Acquired pseudoxanthoma elasticum presenting after liver transplantation

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Résumé en anglais	<p>BACKGROUND: Pseudoxanthoma elasticum (PXE) is thought to be a metabolic disorder resulting from mutations in the gene encoding the cellular transporter, ABCC6, which is primarily expressed in liver and kidney. We encountered 3 patients who developed clinical and histopathological evidence of PXE after liver transplantation, suggesting that PXE could have been acquired from the transplanted organ. OBJECTIVE: We sought to delineate the clinical features and screen each patient and samples of donor liver for mutations in the ABCC6 gene. METHODS: Each patient underwent full clinical examination, skin biopsy, and ophthalmologic examination, and whole genome sequencing using standard techniques. Fixed samples of donor liver tissue were available for mutation analysis in two patients and of donor kidney tissue in one. RESULTS: All 3 patients had unequivocal clinical and histopathologic evidence of PXE. No patient (or family member available for screening) had evidence of mutations in ABCC6. Neither liver specimen nor the single available kidney specimen showed evidence of mutations in ABCC6. LIMITATIONS: Liver tissue was not available from one patient and DNA was of poor quality in another, resulting in limited screening. Genetic testing does not detect ABCC6 mutations in 10% of patients with confirmed PXE. CONCLUSION: Although we were unable to demonstrate ABCC6 mutations in limited screening of fixed donor livers, the absence of any PXE mutations in the affected patients, the timing of onset of PXE, and the known acquisition of other metabolic disorders and coagulopathies from donor livers suggest that PXE was likely acquired via liver transplantation.</p>

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