Acquired pseudoxanthoma elasticum presenting after liver transplantation

Submitted by Emmanuel Lemoine on Tue, 02/24/2015 - 15:21

Titre
Acquired pseudoxanthoma elasticum presenting after liver transplantation

Type de publication
Article de revue

Auteur
Bercovitch, L. [1], Martin, Ludovic [2], Chassaing, N. [3], Hefferon, T. W [4], Bessis, D. [5], Vanakker, O. [6], Terry, S. F [7]

Editeur
Elsevier

Type
Article scientifique dans une revue à comité de lecture

Année
2012

Langue
Anglais

Date
2012

Numéro
5

Pagination
873 - 8

Volume
64

Titre de la revue
Journal of the American Academy of Dermatology

ISSN
1097-6787

Mots-clés
Adult [8], Biliary Atresia/surgery [9], Female [10], Craft Rejection [11], Humans [12], Liver Cirrhosis/surgery [13], Liver Transplantation/adverse effects/immunology [14], Multidrug Resistance-Associated Proteins/genetics [15], Pseudoxanthoma Elasticum/etiology/genetics/pathology [16]

Résumé en anglais
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.