PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS TYPE 3: A NEW LIKELY PATHOGENIC GENETIC VARIANT OF ABCB4 GENE

Luís Maia

UEG Week Copenhagen 2023 | Poster

Hepatobiliary

October 15, 2023

A new likely pathogenic ABCB4 variant (p.I472T) is identified in three siblings with biliary stone disease and elevated liver enzymes, supporting reclassification from variant of unknown significance based on genotype-phenotype co-segregation.

KEY POINTS

- Three siblings (two sisters and one brother) presented with biliary complications including gallstones, choledocholithiasis, intrahepatic bile duct stones, and acute pancreatitis, along with elevated liver enzymes in one patient
- Genetic testing identified the variant c.1415T>C (p.I472T) in heterozygosity in all three siblings; two siblings also carried the pathogenic variant p.S320F
- The p.I472T variant was previously classified as a variant of unknown significance with one entry in ClinVar and is not described in the literature
- The authors propose reclassifying p.I472T as likely pathogenic based on genotype-phenotype co-segregation observed in this family
- This case illustrates how heterozygous ABCB4 variants are associated with increased risk for cholesterol stones, microlithiasis, or sludge rather than the severe cholestasis and progressive liver failure seen in autosomal recessive PFIC3

VIEW THIS CONTENT ON GUTFLIX

https://gutflix.eu/search/d/7e95e22c-743c-11ee-a8f7-0242ac140004

AI-generated summary

This summary was generated by an AI large language model based on the content transcript. It is for informational purposes only and should not be considered a substitute for clinical judgment. Always rely on your professional expertise and the full clinical context when making clinical decisions.