INVESTIGATION OF ABCC6 GENETIC VARIANTS IN A COHORT OF CHOLESTATIC LIVER DISEASE PATIENTS USING WHOLE EXOME SEQUENCING

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Analysis of whole exome sequencing data from 247 patients with suspected cholestatic liver disease identified nine rare heterozygote ABCC6 gene variants in fifteen patients, all classified as variants of uncertain significance.

KEY POINTS

- Nine different rare heterozygote ABCC6 variants were identified in fifteen of 247 whole exome sequencing samples from patients with suspected cholestatic liver disease
- Variants were found in patients with low phospholipid-associated cholelithiasis, intrahepatic cholestasis of pregnancy, elevated transaminases, cholestasis, cholangiocarcinoma, and liver disease of unknown etiology
- All identified variants were classified as variants of uncertain significance using ACMG-AMP guidelines
- The study was prompted by identification of a heterozygote ABCC6 variant in a male patient in his twenties with primary sclerosing cholangitis, chronic cholestatic liver disease, and hypothyroidism
- The findings suggest ABCC6 may have a broader functional involvement in hepatic phenotypes beyond its established association with pseudoxanthoma elasticum

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