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Summary

Undertstanding genetic causes of liver and bile duct diseases: Treatment implications

Verena Keitel-Anselmino

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Hepatobiliary | Mechanisms & Personalised Medicine

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The speaker presented a diagnostic and management framework for genetic cholestatic liver diseases, distinguishing hepatocyte disorders (primarily BSEP/ABCB11 deficiency) from cholangiocyte disorders (primarily ABCB4/MDR3 deficiency) based on laboratory patterns and clinical phenotypes.

KEY POINTS

- Hepatocyte cholestasis presents with elevated ALT, AST, and bile acids but normal gamma-GT, while cholangiocyte cholangiopathies present with elevated gamma-GT and/or alkaline phosphatase; BSEP deficiency is the most common genetic hepatocyte cholestasis in adults.
- The speaker reported a case of BSEP deficiency triggered by antibiotics, with bile acids 20-30 times normal, that normalized within two weeks on an IBAT inhibitor and remained stable after drug discontinuation at six months.
- Only 20-30% of normal BSEP function is needed to remain healthy; patients with polymorphisms become symptomatic when additional stressors such as drugs, hormones, or infections reduce function below this threshold.
- ABCB4 deficiency causes low phospholipid-associated cholelithiasis, affecting 1% of patients hospitalized for stones; the speaker stated about half of these patients in their cohort carry an ABCB4 variant and have increased hepatobiliary malignancy risk.
- The speaker recommends lifelong ursodeoxycholic acid for LPAC patients with elevated enzymes or high-risk ABCB4 mutations, with yearly ultrasound surveillance for malignancy, and stated IBAT inhibitors should be used early in severe BSEP disease to prolong native liver survival.

– The speaker disclosed advisory board participation for both companies with IBAT inhibitors and positioned these drugs as appropriate for severe genetic cholestasis phenotypes where approved.

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