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Summary

Rare causes of cholestatic diseases

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The speaker discussed genetic causes of cholestasis in adults, emphasizing that primary cholestasis originates in hepatocytes (BSEP deficiency and related genetic variants) while cholangiopathies affect cholangiocytes, and presented diagnostic approaches and emerging IBAT inhibitor therapy for patients with residual transporter function.

KEY POINTS

- The speaker reported that primary cholestasis from BSEP deficiency presents with elevated serum bile acids, raised AST and ALT, but low or normal alkaline phosphatase and gamma-GT, requiring about 20 to 30 percent residual BSEP function to avoid symptoms.
- A case was presented of a male patient in his 30s with episodic cholestasis since age 12 who had a homozygous USP53 frameshift variant; the speaker stated USP53 works with myosin 5B to traffic BSEP to the canalicular membrane, and without it BSEP remains in recycling endosomes.
- The speaker stated that in ten adult BSEP-deficient patients treated with IBAT inhibitors, serum bile acids decreased from above 500 micromoles and pruritus improved, consistent with meta-analysis data, though the patient presented progressed to liver transplant and developed HCC.
- The speaker recommended genetic workup when presentation does not fit common diagnoses, especially with high serum bile acids but normal gamma-GT, or when patients fail standard treatment such as AMA-negative PBC.
- IBAT inhibitors are approved in Europe for PFIC and Alagille syndrome and work well in ABCB11 patients with residual function, though the speaker noted patients may experience loose stools, transient transaminase elevation from increased bile acid synthesis, and require fat-soluble vitamin monitoring.

– The speaker emphasized that patients with two truncating BSEP or ABCB4 variants typically require liver transplant before adulthood, while missense variants with residual function may reach adulthood, and prospective registries are needed to determine long-term cancer risk and transplant-free survival on IBAT therapy.

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