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Summary

Management of cholestasis (Complete Session)

UEG Week Barcelona 2026 | Presentation

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Three cases illustrate the expanding role of genetic testing in diagnosing cholestatic liver diseases previously labeled as seronegative primary biliary cholangitis or cryptogenic cholangiopathy, with implications for pruritus management and surveillance strategies.

KEY POINTS

- A young woman with chronic cholestatic enzyme elevation, negative autoantibodies, and intrahepatic cholestasis of pregnancy was found by whole exome sequencing to have heterozygous ABCB4 (MDR3) frameshift variant, meeting criteria for LPAC syndrome; the speaker reported she improved on ursodeoxycholic acid and is followed yearly with ultrasound surveillance for cholangiocarcinoma risk based on retrospective cohort data showing 5% risk in heterozygous carriers.
- A 48-year-old woman diagnosed with AMA-negative PBC and severe treatment-refractory pruritus was found to have heterozygous NOTCH2 variants of uncertain significance and craniofacial abnormalities consistent with Alagille syndrome; the speaker reported near-complete resolution of pruritus three months after starting the IBAT inhibitor Odevixibat, supported by two clinical trials in Alagille syndrome.
- The speakers stated that genetic diagnoses are found in 20 to 30% of cryptogenic cholestasis cases across European cohorts, with higher yield in patients aged 40 or below, and emphasized that AMA-negative PBC diagnoses should be reconsidered case-by-case for alternative etiologies.
- A 58-year-old man with mid bile duct cholangiocarcinoma and insufficient future liver remnant (26% after planned right hepatectomy) underwent staged approach with lymphadenectomy, right posterior vein ligation, and portal vein embolization rather than immediate resection; the speaker emphasized that effective biliary drainage and avoiding cholangitis are imperative for surgical candidates.
- The speaker teaching the Alagille case disclosed that whole exome sequencing was chosen over a cholestatic gene panel partly because research funds were available; the speaker recommended that before IBAT inhibitors became available, bezafibrate or nasobiliary drainage were treatment options for cholestatic pruritus in genetic cholangiopathies.

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