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Summary

BRIC2 OR PFIC2: A NEW BSEP MUTATION OF UNCERTAIN ALLOCATION

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Hepatobiliary

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A 17-year-old girl with recurrent severe jaundice and itching was diagnosed with familial intrahepatic cholestasis due to compound heterozygous ABCB11 gene mutations, including a novel variant, with complete resolution after medical treatment.

KEY POINTS

- The patient presented with severe jaundice and pruritus, with total bilirubin 24.5 mg/dl, direct bilirubin 18.9 mg/dl, elevated transaminases (AST 307 IU/L, ALT 238 IU/L), and normal GammaGT, having experienced a similar self-resolving episode 2 years prior.
- Genetic testing identified compound heterozygous ABCB11 mutations: c.1460G>A (p.Arg487His) from the father and c2599C>T (Gln867*) from the mother, with the latter being a novel variant not previously described in familial intrahepatic cholestasis.
- Liver biopsy showed hepatocellular and intracanalicular cholestasis with cholestatic rosettes, inflammatory activity, mild fibrosis, and weak BSEP expression on immunohistochemistry.
- Treatment with ursodesoxycholic acid, cholestyramine, and cetirizine for 4 months resulted in complete resolution of jaundice and pruritus with normalized bilirubin levels, and the patient remained asymptomatic without therapy at 1-year follow-up.
- The authors suggest the temporal test will help classify this new variant as BRIC2 rather than PFIC2, relevant for clinicians managing genetic cholestatic liver diseases.

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