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Summary

RECURRENT ACUTE LIVER FAILURE (RALF) IN CHILDREN – UNRAVELING THE MYSTERY

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UEG Week Vienna 2024 | Poster

Hepatobiliary

October 13, 2024

A case series of four children with recurrent acute liver failure identifies novel SCYL1 and RINT1 gene mutations through whole exome sequencing, demonstrating the role of genetic testing in uncovering the etiology of previously indeterminate cases.

KEY POINTS

- Four children with recurrent acute liver failure during febrile episodes were studied at a tertiary care institute in India from January 2023 to January 2024, with genetic diagnoses made after two years of initial presentation.
- Whole exome sequencing identified three novel mutations: two cases had SCYL1 gene mutations causing spinocerebellar ataxia 21 (CALFAN syndrome) and one case had a RINT1 gene mutation.
- SCYL1 mutation cases presented with low GGT cholestasis, neurological manifestations including tremor, ataxia, speech and developmental delay, and progressed to cirrhosis with early portal hypertension despite liver function recovery between episodes.
- RINT1 mutation case showed complete liver function recovery between episodes without progression to chronic liver disease.
- Overall mortality was 50 percent; liver transplantation is life saving but the progressive neurological phenotype in SCYL1 cases is a potential contraindication for transplantation.

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