gutflix
GI learning, personalised

Summary

CIRRHOSIS DUE TO GLYCOGEN STORAGE DISORDER IXC WITH A NOVEL PHKG2 MUTATION AND CONCURRENT HEPATITIS B INFECTION: A RARE OCCURRENCE

Siddharth Srivastava

UEG Week Vienna 2024 | Poster

Hepatobiliary

October 13, 2024

A 13-year-old boy with cirrhosis, hypoglycemia, and hepatosplenomegaly was diagnosed with glycogen storage disease type IXc after molecular analysis revealed a novel PHKG2 gene mutation despite positive hepatitis B serology.

KEY POINTS

- The patient presented with abdominal distention, recurrent hypoglycemia, self-resolving jaundice, growth retardation, and hepatosplenomegaly with laboratory findings of anemia, thrombocytopenia, elevated transaminases, and raised cholesterol and triglycerides.
- Etiological work-up was positive for HBV surface antigen and antibodies for HBV core and envelope antigens but HBV DNA was undetectable, making HBV an unlikely cause of cirrhosis given the patient's young age.
- Molecular analysis revealed a novel mutation in exon 3 of the PHKG2 gene (c.98G>T, p.Gly33Val), leading to a diagnosis of cirrhosis due to GSD IXc with inactive chronic hepatitis B.
- Symptoms resolved with an optimized diet of corn starch, high-protein and vitamin supplementation, and liver transplantation was discussed with the family.
- This case is relevant to clinicians managing pediatric cirrhosis with metabolic presentations and highlights the importance of genetic testing when standard etiologies are inconclusive.

VIEW THIS CONTENT ON GUTFLIX

https://gutflix.eu/search/d/321378da-9112-11ef-bb57-0242ac140004

AI-generated summary

This summary was generated by an AI large language model based on the content transcript. It is for informational purposes only and should not be considered a substitute for clinical judgment. Always rely on your professional expertise and the full clinical context when making clinical decisions.