

A RARE CASE OF A PATIENT WITH WILLIAMS SYNDROME AND LIVER CIRRHOSIS OF UNKNOWN ETIOLOGY. PREDISPOSITION OR COINCIDENCE?

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Poster

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A case report describes the first documented occurrence of liver cirrhosis in a patient with Williams syndrome, with no identifiable classic etiology after extensive workup.

KEY POINTS

- A 7-year-old male with genetically confirmed Williams syndrome developed transaminasemia, hepatosplenomegaly, and imaging findings consistent with cirrhosis including Grade 1 esophageal varices
- Liver biopsy revealed extensive bridging fibrosis, incomplete regenerative nodules, and macrovesicular steatosis without evidence of a specific chronic liver disease
- Comprehensive testing including serology, genetic mutation analysis, and metabolic disorder screening did not identify a classic cause of cirrhosis
- The authors suggest the mechanism may be genetic or metabolic related to Williams syndrome itself, as liver cirrhosis has not been previously reported in this condition
- This case highlights the variable organ involvement in Williams syndrome beyond its known gastrointestinal manifestations of reflux, constipation, diverticulosis, hernias, and celiac disease

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