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(CASE REPORT)

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Decompensated cirrhosis in an adult revealing probable congenital galactosemia: A case report

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Abstract

Congenital galactosemia is an autosomal recessive inborn error of metabolism, most commonly caused by a deficiency of galactose-1-phosphate uridyltransferase (GALT). It typically presents during the neonatal period with acute liver failure, cataracts, and neurological impairment. Diagnosis in adulthood is exceptional.

We report the case of a 30-year-old woman admitted for upper gastrointestinal bleeding revealing cirrhosis with portal hypertension. Medical history included bilateral cataract surgery performed twice, psychomotor delay, bilateral hearing impairment, and a history of unexplored neonatal jaundice. A comprehensive etiological workup for chronic liver disease was negative. Plasma galactose level was elevated at 1.2 mg/dL (normal < 0.7 mg/dL). In the absence of enzymatic and genetic confirmation due to socioeconomic constraints, the diagnosis of probable congenital galactosemia was retained based on a consistent set of clinical and biological findings.

This case highlights the rare possibility of late presentation in adulthood and emphasizes the importance of considering a metabolic etiology in unexplained cirrhosis associated with suggestive systemic manifestations.

Keywords: Congenital galactosemia; Liver cirrhosis; Portal hypertension; Late diagnosis

1. Introduction

Classical galactosemia (type I) is a rare metabolic disorder caused by deficiency of galactose-1-phosphate uridyltransferase (GALT), a key enzyme in galactose metabolism. Its incidence is estimated at 1 in 40,000 to 1 in 60,000 live births in Europe[1]. Accumulation of galactose-1-phosphate and galactitol leads to multisystem involvement, predominantly affecting the liver, central nervous system, and eyes.

In the absence of neonatal screening and early galactose-restricted diet, the disease may progress to hepatic fibrosis and cirrhosis [2]. Adult diagnoses remain rare and are usually related to attenuated forms or delayed recognition [3].

We report a case of cirrhosis revealed by upper gastrointestinal bleeding in a 30-year-old adult woman in whom probable congenital galactosemia was suspected.

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2. Case Presentation

We report the case of a 30-year-old woman born to first-degree consanguineous parents who was admitted for hematemesis and melena revealing portal hypertension.

On physical examination, she was conscious and hemodynamically stable, without jaundice or ascites. Abdominal examination revealed mild collateral venous circulation and massive splenomegaly extending to the umbilicus. No hepatomegaly was palpable.

Upper gastrointestinal endoscopy demonstrated esophageal varices requiring endoscopic ligation, associated with portal hypertensive gastropathy.

Abdominal CT angiography showed a dysmorphic liver with irregular contours and homogeneous parenchymal density, without focal lesions. There was hypertrophy of the left lobe and segment I (caudate lobe) with hypotrophy of segment IV, consistent with established cirrhosis. The portal vein and hepatic veins were patent and non-dilated. Serpiginous peri-splenic and peri-pancreatic venous structures were observed, corresponding to porto-systemic collateral circulation. The spleen measured 21 cm in longitudinal diameter. No ascites or biliary duct dilatation was noted.

Laboratory findings revealed anemia (hemoglobin 10.4 g/dL), leukopenia (2,930/mm^3), and thrombocytopenia (62,000/mm^3), consistent with hypersplenism. Prothrombin time was 60%, and serum albumin was 30 g/L.

Liver enzymes were elevated, with AST at 2 times the upper limit of normal and ALT at 1.9 times the upper limit of normal. Total bilirubin was 7 mg/L with predominance of the conjugated fraction. No significant cholestatic pattern was observed. Serum protein electrophoresis showed hypergammaglobulinemia at 1.6 times the normal value (22.2 g/L). The Child-Pugh score was A6.

Medical history included bilateral cataract surgery performed at the age of 7 years and again at 25 years, psychomotor delay with speech impairment, bilateral hearing loss, and a history of unexplored neonatal jaundice.

Comprehensive etiological investigation for cirrhosis was negative, including viral hepatitis B and C serologies, complete autoimmune workup, evaluation for Wilson disease (normal ceruloplasmin, normal serum and urinary copper levels, absence of Kayser-Fleischer rings), and hemochromatosis screening.

Given the association of unexplained cirrhosis and systemic manifestations suggestive of a congenital metabolic disorder, congenital galactosemia was suspected. Plasma galactose level was elevated at 1.2 mg/dL (normal < 0.7 mg/dL). Enzymatic testing (GALT activity) and genetic analysis could not be performed due to socioeconomic limitations. A galactose-restricted diet was initiated.

3. Discussion

Congenital galactosemia is an inborn error of metabolism caused by deficiency of galactose-1-phosphate uridyltransferase (GALT), leading to toxic accumulation of galactose-1-phosphate and galactitol. It typically presents during the neonatal period with acute liver dysfunction, prolonged jaundice, and gastrointestinal symptoms. Without treatment, the disease may be rapidly fatal [2]. Early hepatic injury may progress to fibrosis and cirrhosis.

Adult diagnoses are exceptional. As early as 1965, Ulbricht reported persistent liver damage in adults with enzymatic deficiency of galactose metabolism [4]. In 1980, Vogt et al. described a 52-year-old man presenting with decompensated liver cirrhosis secondary to galactosemia, demonstrating that the disease may remain clinically silent for decades before presenting with severe hepatic decompensation [6]. More recently, Quelhas et al. (2024) reported late-diagnosed patients, including an adult with documented cirrhosis [3]. Cases of liver transplantation in adult galactosemic patients have also been described [5].

These reports suggest that some forms of galactosemia may follow a milder or partially compensated course, possibly due to residual enzymatic activity, or may remain unrecognized in the absence of neonatal screening programs.

In our case, early-onset bilateral cataracts strongly support the diagnosis, as galactitol accumulation in the lens is a hallmark of galactosemia. Psychomotor delay and bilateral hearing impairment are recognized long-term complications.

Parental consanguinity further increases the likelihood of an autosomal recessive disorder. The absence of an alternative etiology after comprehensive evaluation reinforces the suspicion of a hereditary metabolic cause.

The main limitation of this case is the absence of enzymatic and genetic confirmation. Nevertheless, the combination of consistent clinical history, imaging findings, and biochemical abnormalities supports the diagnosis of probable congenital galactosemia. Similar to the case reported by Vogt et al., our observation illustrates the rare possibility of late presentation in adulthood with portal hypertension-related decompensation.

4. Conclusion

Congenital galactosemia should be considered in cases of unexplained cirrhosis associated with early-onset cataracts and neurodevelopmental impairment, even in adulthood. This case emphasizes the importance of neonatal screening and a broad etiological approach in cryptogenic cirrhosis.

Compliance with ethical standards

Disclosure of conflict of interest

The authors declare that they have no conflict of interest.

Statement of ethical approval

Ethical approval was not required for this case report according to institutional policy. The study was conducted in accordance with the principles of the Declaration of Helsinki.

Statement of informed consent

Written informed consent was obtained from the patient for publication of this case report and any accompanying clinical information.

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Authors' contributions

All authors contributed to the conception, drafting, revision, and approval of the final manuscript.

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