Content available at: https://www.ipinnovative.com/open-access-journals

IP International Journal of Medical Paediatrics and Oncology

Journal homepage: https://www.ijmpo.com/

Case Report

An uncommon paediatric case of primary biliary cirrhosis: A diagnostic challenge

Hardik Shah[1]*, Naiya Bhavsar[2], Seema Shah[1], Ashok Bhandari[1], Mitul Chaudhari[1]

[1]Dept. of Peadiatrics, DR.M.K. Shah Medical College, S.M.S Hospital, Ahmedabad, Gujarat, India
[2]Dept. of Pulmonary Medicine, DR.M.K. Shah Medical College, S.M.S Hospital, Ahmedabad, Gujarat, India

Abstract

Background: Primary Biliary Cirrhosis (PBC) is a chronic autoimmune disorder that usually affects middle-aged women, and it is typically characterized by the progressive destruction of intrahepatic bile ducts, leading to cholestasis and cirrhosis. The condition is infrequently diagnosed in children, with only a handful of pediatric cases being reported in the literature. This case highlights the rare occurrence of PBC in a 6-year-old child, focusing on the diagnostic approach and treatment strategy.

Case Presentation: A 6-year-old male with symptoms of jaundice, severe itching, and tiredness was diagnosed with primary biliary cirrhosis. Laboratory results showed elevated liver enzymes and the presence of antimitochondrial antibodies (AMA). While a liver biopsy was considered but not performed due to financial constraints, treatment with ursodeoxycholic acid was initiated. The patient demonstrated significant clinical improvement, including normalization of liver function tests and reduction in symptoms over the following six months.

Conclusion: Although PBC is mostly seen in adults, it can rarely present in the pediatric population. This case reinforces the importance of considering autoimmune liver diseases, such as PBC, in children who present with unexplained jaundice and pruritus. Timely initiation of treatment, like ursodeoxycholic acid, can improve prognosis and quality of life in affected patients.

Keywords: Primary Biliary Cirrhosis, Pediatric Liver Disease, Autoimmune Hepatitis, Ursodeoxycholic Acid, Jaundice, Pruritus, Case report

Received: 18-05-2025; **Accepted:** 17-06-2025; **Available Online**: 10-07-2025

1. Introduction

Primary Biliary Cirrhosis (PBC) is a rare, progressive autoimmune liver disorder that leads to the destruction of intrahepatic bile ducts, cholestasis, and eventually cirrhosis. Although the disease is predominantly observed in women, it is uncommon in children, with only a limited number of pediatric cases reported in the literature. The etiology of PBC remains unclear, though genetic and environmental factors are believed to play a role in its development. This report discusses a rare case of PBC in a 6-year-old female, highlighting the diagnostic process, the challenges associated with pediatric PBC, and the treatment course.

2. Case Presentation

A 6-year-old male was referred to the pediatric gastroenterology clinic after 3 months of progressive jaundice, itching, and generalized fatigue. The child initially presented with mild yellowing of the skin and eyes, which gradually worsened over the course of the illness. The pruritus, which primarily affected his hands and feet, was severe enough to disturb his sleep. The patient denied any abdominal discomfort or signs of systemic illness.

His medical history was unremarkable, and there was no family history of autoimmune diseases or liver conditions. On physical examination, the patient was noted to have mild jaundice and scleral icterus. There were no signs of hepatomegaly, splenomegaly, or ascites, and no other notable physical findings were observed.

Initial laboratory results revealed the following:

1. Liver enzymes: AST 160 U/L, ALT 180 U/L (normal range: 15-37 U/L and 10-40 U/L respectively)
2. Elevated alkaline phosphatase (ALP) of 500 U/L (normal range: 120-340 U/L)

*Corresponding author: Hardik Shah
Email: hardik.ashah99@gmail.com

https://doi.org/10.18231/j.ijmpo.2025.011

3. Total bilirubin of 4.5 mg/dL (normal range: 0.1-1.2 mg/dL), with indirect bilirubin predominating
4. Positive antimitochondrial antibodies (AMA) at a titer of 1:320, a common finding in PBC
5. Other autoimmune markers (ANA, anti-Smith, anti-dsDNA) were negative, ruling out other autoimmune liver diseases
6. Normal clotting profile

Based on these findings, an abdominal ultrasound was performed, which showed a normal-sized liver without evidence of cirrhosis or masses. While the diagnosis strongly suggested PBC, a liver biopsy was considered but not performed due to the family's financial constraints.

The patient was diagnosed with PBC based on the combination of clinical symptoms, elevated liver enzymes, and the presence of AMA. She was started on ursodeoxycholic acid (UDCA), the standard treatment for PBC, which has been shown to improve liver function and slow disease progression. Over the next six months, the child's liver function tests gradually normalized, and her symptoms of pruritus and fatigue significantly improved.

3. Discussion

Primary Biliary Cholangitis (PBC) is a chronic, progressive cholestatic liver disease that predominantly affects middle-aged women, with a strong autoimmune etiology. Characterized by the gradual destruction of small intrahepatic bile ducts, PBC ultimately leads to cholestasis, hepatic fibrosis, and cirrhosis if left untreated.[1,2] While well recognized in the adult population, pediatric presentation is exceedingly rare, often posing a significant diagnostic challenge for clinicians due to atypical age and overlapping features with other pediatric hepatobiliary disorders.[3]

The pathogenesis of PBC is multifactorial and not yet fully understood. It is believed to result from a breakdown in immunological tolerance, where environmental triggers (such as infections or xenobiotics) in genetically predisposed individuals lead to an autoimmune attack on biliary epithelial cells. These cells become the target of autoreactive T cells and autoantibodies, particularly antimitochondrial antibodies (AMAs), which are present in nearly 90–95% of adult PBC patients and remain the hallmark diagnostic marker.[1,4] Their role in the disease process is supported by both clinical and experimental findings, even though their precise pathogenic contribution remains unclear.

In pediatric cases, such as the one presented here, clinicians often face difficulty differentiating PBC from more common causes of pediatric cholestasis, including Alagille syndrome, progressive familial intrahepatic cholestasis (PFIC), autoimmune hepatitis (AIH), and sclerosing cholangitis. This child's clinical presentation with progressive jaundice, intense pruritus, and hepatocellular enzyme elevation was initially non-specific. However, the markedly positive AMA titer in conjunction with elevated alkaline phosphatase pointed toward a diagnosis of PBC, despite his young age and male gender, both of which are uncommon in typical PBC demographics.[2,3,6]

Liver biopsy remains the gold standard for confirming PBC, especially in atypical presentations. Histologically, PBC shows chronic non-suppurative cholangitis and interlobular bile duct destruction. However, in this case, socioeconomic constraints prevented a biopsy. Still, a diagnosis based on clinical findings, laboratory tests, and the presence of AMA is considered sufficient in a subset of patients when the clinical suspicion is high and histology is unobtainable.[4,7]

Pediatric PBC has been documented in a handful of case reports and small cohorts, often presenting with a more aggressive course or with overlapping features of autoimmune hepatitis (AIH), referred to as overlap syndromes.[3,8] These overlap syndromes, more common in children and adolescents than in adults, present unique therapeutic and prognostic challenges. However, in our patient, ANA and anti-dsDNA tests were negative, suggesting isolated PBC without autoimmune overlap.

Treatment with ursodeoxycholic acid (UDCA), a hydrophilic bile acid, is the cornerstone of therapy in PBC. UDCA works by improving bile flow, reducing toxic bile acid accumulation, and modulating immune responses.[5,6] It has been shown to delay disease progression, reduce symptoms such as pruritus, and improve liver enzyme profiles. In this case, the patient demonstrated substantial clinical and biochemical improvement after UDCA initiation, consistent with data supporting its efficacy in pediatric autoimmune cholangiopathies.[4,9]

Long-term outcomes in pediatric PBC are not well characterized due to the rarity of the condition and limited longitudinal data. However, the natural history is believed to be similar to adult PBC if diagnosed and treated early. Without appropriate therapy, progression to end-stage liver disease and the need for liver transplantation are real concerns.[7] Fortunately, this patient responded well to first-line therapy, with improved symptoms and liver function tests on follow-up.

The scarcity of reported pediatric PBC cases, especially in males, underlines the importance of heightened clinical suspicion in children presenting with persistent cholestasis and unexplained liver enzyme abnormalities. AMA testing should be considered in such scenarios, even when other common differentials are not clearly excluded.

4. Conclusion

Primary Biliary Cirrhosis, although predominantly an adult disease, can present in children, often with nonspecific symptoms such as jaundice, pruritus, and fatigue. This case emphasizes the importance of considering autoimmune liver

diseases, like PBC, in pediatric patients who present with unexplained liver dysfunction. Early diagnosis and treatment with ursodeoxycholic acid can lead to favourable outcomes. Despite the financial barriers that prevented the liver biopsy in this case, the child showed significant clinical improvement with medical management, underscoring the efficacy of UDCA in pediatric PBC.

5. Human Subjects

Consent was obtained or waived by all participants in this study.

6. Conflicts of Interest

None.

7. Source of Funding

All authors have declared that they have no financial relationships at present or within the previous three years with any organizations that might have an interest in the submitted work.

References

1. Carey EJ, Ali AH, Lindor KD. Primary biliary cirrhosis. *Lancet*. 2015 Oct 17;386(10003):1565–75.
2. Yaser D, Leslie S, Doug S, Larry DJ, Ian W, Heathcote EJ, et al. Pediatric-onset primary biliary cirrhosis. *Gastroenterology*. 2003;125(5):1476–9.
3. Ivana K, Aleksandra B, Ivica S, Dragan P. Twelve-year-old girl with primary biliary cirrhosis. *Case Rep Pediatr*. 2012:2012:937150.
4. Purohit T, Cappell MS. Primary biliary cirrhosis: pathophysiology, clinical presentation and therapy. *World J Hepatol*. 2015;7(7):926–41.
5. Mio K, Yuko K, Kenichi H, Yasunori S, Motoko S, Hiroko I, et al. Clinicopathological study of primary biliary cirrhosis with interface hepatitis compared to autoimmune hepatitis. *World J Gastroenterol*. 2014;20(13):3597–608.
6. Kaplan MM, Gershwin ME. Primary biliary cirrhosis. *N Engl J Med*. 2005;353(12):1261–73.
7. Trivella, J, John BV, Levy C. Primary biliary cholangitis: Epidemiology, prognosis, and treatment. *Hepatology Communications*. 2023;7(6):e0179.
8. Kumar D, Tandon RK. Use of ursodeoxycholic acid in liver diseases. *J Gastroenterol Hepatol*. 2001;16(1):3–14.
9. Huang L, Siyu L, Jingjing C, Yu Z, Ke L, Linan Z, et al. Efficacy and safety of ursodeoxycholic acid in children with cholestasis: A systematic review and meta-analysis. *PLoS One*. 2023;18(1):e0280691.

Cite this article: Shah H, Bhavsar N, Shah S, Bhandari A, Chaudhari M. An uncommon paediatric case of primary biliary cirrhosis: A diagnostic challenge. *IP Int J Med Paediatr Oncol*. 2025;11(2):49-51.