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Case Report

# Evaluation of a Case with Biliary Atresia and Heterotaxy Syndrome (Left Isomerism) in Terms of Liver Transplantation: A Case Report

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**Abstract**

Left isomerism, a subtype of heterotaxy marked by multiple spleens and abnormal organ positioning, poses additional complexity when coexisting with biliary atresia, the leading cause of neonatal cholestasis and pediatric liver transplantation. We report a 5-month-old male infant with biliary atresia and left isomerism, presenting with persistent jaundice, acholic stools, and dark urine. Despite undergoing Kasai portoenterostomy at one month, clinical and laboratory parameters did not improve. Cardiac assessment revealed left atrial isomerism, atrioventricular septal defect, pulmonary atresia, and patent ductus arteriosus. Abdominal CT showed a midline liver, polysplenia, absent inferior vena cava with azygos-hemiazygos continuation, preduodenal portal vein, intestinal malrotation, and visceral heterotaxy. Laboratory tests indicated severe cholestasis, hepatocellular injury, and impaired liver function. The patient was evaluated and listed as a liver transplant candidate. The coexistence of biliary atresia and left isomerism complicates both diagnosis and treatment. Early imaging, multidisciplinary management, and timely transplantation are essential to improve outcomes. This case underscores the importance of a comprehensive, individualized approach in managing biliary atresia with complex congenital anomalies.

**Keywords:** Biliary Atresia, Heterotaxy Syndrome, Liver Transplantation

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Left isomerism, also known as polysplenia syndrome, is a form of heterotaxy characterized by the presence of multiple spleens and other deviations from normal organ positioning. This condition is classified as situs ambiguus or heterotaxy, where the organs do not follow the typical left-right arrangement seen in standard anatomical structures. In left isomerism, the body may exhibit bilateral left-sidedness; that is, structures usually found on the left side of the body may be mirrored on both sides.[1] This is often associated with variations in the arrangement of major blood vessels and internal organs.[2]

Biliary atresia is a rare neonatal disease caused by obstruction of the bile ducts, leading to severe cholestasis, fibrosis, and cirrhosis. Although it is the primary cause of neonatal cholestasis, it is also the most common reason for liver transplantation in children. Although the exact etiology remains unknown, early surgical intervention and disease management through the Kasai procedure are of great im-

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portance to control the disease and delay the need for liver transplantation.[3, 4]

The coexistence of left isomerism and biliary atresia is clinically significant because of the complexity created by having to manage both conditions simultaneously. Additional congenital defects are observed in approximately 10% of infants with biliary atresia, often involving left-right axis abnormalities, such as situs inversus and polysplenia. This association suggests a possible underlying genetic factor in the disease.[1] The presence of left isomerism can complicate the diagnosis and treatment of biliary atresia, as it may mask typical signs of anatomical abnormalities or affect surgical outcomes.

The incidence of biliary atresia is estimated to be between 1 in 5,000 and 1 in 20,000 live births, with higher incidence rates observed in Asia.[5] When seen in conjunction with conditions such as left isomerism, early and accurate diagnosis is crucial; timely intervention and prevention of serious complications help achieve the best possible surgical outcomes.[3]

In children with biliary atresia, especially when accompanied by complex conditions such as left isomerism, liver transplantation is a vital treatment option. In cases where biliary atresia coexists with left isomerism, additional anatomical variations, such as the absence or abnormal structure of the portal veins, may render traditional liver transplantation techniques unfeasible.[6] These vascular anomalies require special surgical approaches and preoperative imaging to ensure successful liver transplantation.[7] Surgical management of these patients requires a multidisciplinary approach to optimize outcomes and address preoperative challenges, such as malnutrition and portal hypertension.[8]

In conclusion, liver transplantation is a vital option for the treatment of biliary atresia, especially in complex clinical situations in which concurrent left isomerism is present.

## Case Report

A five-month and fourteen-day-old male patient was evaluated at an outside center for persistent jaundice during the first weeks after birth, acholic stools, and dark-colored urine. Imaging and biochemical tests led to a diagnosis of biliary atresia. Kasai portoenterostomy was performed in the first month of life. However, since there was no improvement in clinical and laboratory findings postoperatively and jaundice persisted, the patient was referred to our center for further evaluation.

On evaluation of the cardiac system, transthoracic echocardiography revealed left atrial isomerism, complex atrioventricular septal defect (AVSD), pulmonary valve atresia, and patent ductus arteriosus (PDA). The pulmonary arteries were perfused by means of a stent placed in the PDA, and there was mild regurgitation in the atrioventricular valves. On contrast-enhanced thoracic CT, a right aortic arch and an AVSD are observed (Fig. 1). The patient's overall hemodynamic status was stable, and cardiac surgery was postponed to a later date. Given the presence of cardiac and major vascular structural anomalies, recognition of cardiac anomalies before liver transplantation may be important.

Abdominal CT tomography showed that the liver was displaced toward the midline and shifted to the left. In addition, polysplenia consistent with multiple small nodular splenic structures, absence of the inferior vena cava (IVC), and, secondary to this, azygos-hemiazygos vein dilatation, preduodenal portal vein, short pancreas, intestinal malrotation, loss of visceral symmetry (Fig. 2) were detected. These findings support the presence of heterotaxy syndrome (left isomerism) in this patient.

Laboratory evaluations revealed that the total bilirubin level was 17.84 mg/dL and the direct bilirubin level was 8.78 mg/dL, indicating significant cholestatic jaundice.

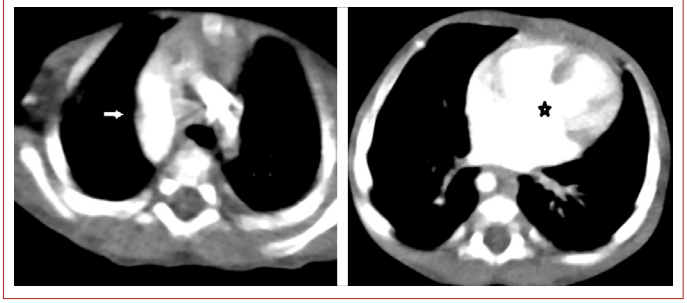

**Figure 1.** Contrast-enhanced thoracic CT images show a right-sided aortic arch (white arrow) and an atrioventricular septal defect (AVSD) (asterisk).

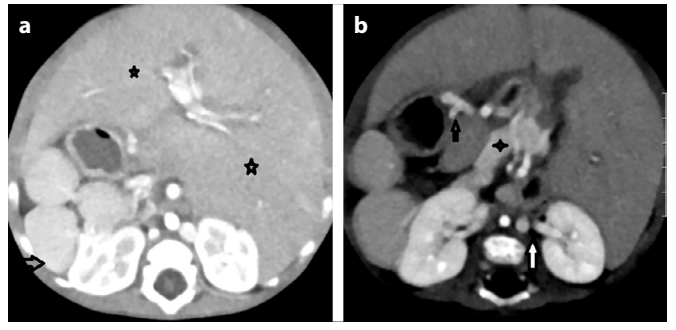


**Figure 2.** Contrast-enhanced abdominal CT images. **(a)** The liver is located in the midline, and both lobes exhibit a left-lobe configuration (asterisks). Multiple splenules consistent with polysplenia are seen (arrow). **(b)** A right-sided stomach, a preduodenal portal vein (black arrow), a shortened pancreas (asterisk), and a dilated azygos vein (white arrow) are observed.

There was a notable elevation in transaminase levels (AST, 647 U/L; ALT, 300 U/L), which was consistent with active hepatocellular injury. The LDH level was 281 U/L, indicating nonspecific cellular damage. The serum albumin level was below the reference values of 3.0 g/dL, showing decreased protein synthesis capacity of the liver. There was also a disturbance in coagulation parameters: INR was 1.61, and prothrombin activity was 38.4%. These findings indicate that the patient experienced advanced hepatic dysfunction and impaired liver reserve.

In light of these clinical, laboratory, and radiological findings, the patient was evaluated by a multidisciplinary council for liver transplantation and has been placed under follow-up as a transplant candidate.

## Discussion

Left isomerism is mostly associated with polysplenia syndrome and presents unique challenges when accompanied by biliary atresia. Left isomerism encompasses a spectrum of cardiac and extracardiac anomalies, including[2] interruption of the inferior vena cava, complete atrioventricular septal defect, and complete heart block. These anomalies complicate the diagnosis and treatment of biliary atresia, which, due to progressive fibrosis of the biliary tree, leads to cholestasis and liver damage, and is the most common cause of neonatal cholestasis and pediatric liver transplantation.[4]

The presence of laterality defects, such as left isomerism, increases the complexity of genetic and environmental factors in the management of these cases. Studies suggest that genetic mutations affecting the determination of the left-right axis, such as the CFC1 gene, may predispose patients to both left isomerism and biliary atresia; however, other genetic or environmental factors may also contribute to this phenotype.[1]

Early diagnosis of biliary atresia is crucial for effective treatment, with interventions such as hepatoportoenterostomy ideally performed within the first 45 days of life yielding the best outcomes. This surgical procedure aims to restore bile flow from the liver to the intestine, potentially slowing the progression of liver damage. However, despite early intervention, the majority of patients with biliary atresia eventually require liver transplantation because of ongoing liver deterioration.The progressive nature of biliary atresia means that even with successful initial treatment, long-term liver health remains a significant concern. As patients age, the cumulative effects of cholestasis and fibrosis often lead to cirrhosis and liver failure. This necessitates close monitoring of liver function and regular follow-up throughout childhood and adolescence. Liver transplantation, when required, offers these patients the best chance for long-term survival and improved quality of life, addressing both biliary obstruction and the resultant liver damage.[3, 4]

In such complex cases, radiology, a multisystemic approach involving pediatric surgeons, hepatologists, cardiologists, and geneticists, is required. This comprehensive approach enables the early detection of various complications arising from the coexistence of left isomerism and biliary atresia and facilitates the development of personalized treatment strategies.[9]

Improvements in management may include the development of screening protocols for the early detection of biliary atresia and associated anomalies, as well as ongoing research on genetic factors and potential environmental triggers involved in the etiology of these combined conditions.[10]

## Conclusion

Heterotaxy syndrome accompanied by biliary atresia (especially left isomerism) can lead to significant anatomical and physiological challenges in liver transplant planning. In such cases, early diagnosis, detailed imaging, multidisciplinary evaluation, and timely transplantation decisions play decisive roles in reducing mortality and morbidity. This presentation provides a remarkable example of the approach to liver transplantation in cases of biliary atresia associated with complex congenital anomalies.

### Disclosures

**Informed Consent:** Written, informed consent was obtained from the patient's family for the publication of thís case report and the accompanying images.

**Conflict of Interest:** None declared.

**Financial Disclosure:** None.

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## References

1. Jacquemin E, Cresteil D, Hadchouel M, Raynaud N (2002) CFCI gene mutation and biliary atresia with polysplenia syndrome. Journal of Pediatric Gastroenterology and Nutrition 34:326.
2. Tawfik AM, Zaky MM, Eladalany MA, Elmokadem AH, Batouty NM (2013) Polysplenia syndrome: a review of the relationship with viscero-atrial situs and the spectrum of extra-cardiac anomalies. Surgical and Radiologic Anatomy 35:647–653.

3. Schreiber RA, Harpavat S, Hulscher JBF, Wildhaber BE (2022) Biliary Atresia in 2021: Epidemiology, Screening and Public Policy. Journal of Clinical Medicine 11:999.
4. Feldman AG, Mack CL (2015) Biliary Atresia: Clinical Lessons Learned. Journal of Pediatric Gastroenterology and Nutrition 61:167–175.
5. Tam PKH, Wells RG, Tang CSM, Lui VCH, Hukkinen M, Luque CD, De Coppi P, Mack CL, Pakarinen M, Davenport M (2024) Biliary atresia. Nature reviews Disease primers.
6. Ledesma-Medina J, Bron KM, Dominguez R, Bowen A, Young LW (1985) Pediatric liver transplantation. Part I. Standardization of preoperative diagnostic imaging. Radiology 157:335–338.
7. Barshes NR, Lee TC, Goss JA, Carter BA, Balkrishnan R, Karpen SJ (2005) Orthotopic Liver Transplantation for Biliary Atresia. Liver Transplantation 11:1193–1200.
8. Sundaram SS, Mack CL, Sokol RJ, Feldman AG (2016) Biliary atresia: Indications and timing of liver transplantation and optimization of pretransplant care. Liver Transplantation 23:96–109.
9. Bezerra JA, Karpen SJ, Hoofnagle JH, Mack CL, Sokol RJ, Wells RG, Doo E (2018) Biliary Atresia: Clinical and Research Challenges for the Twenty-First Century. Hepatology 68:1163–1173.
10. Wang KS, Karpen SJ, Holcomb G, et al (2015) Newborn Screening for Biliary Atresia. Pediatrics 136:e1663–e1669.