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CASE REPORT

Biliary atresia in a 3-month-old infant (case report)

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Abstract

Biliary atresia (BA) is a congenital disease that occurs when extrahepatic bile ducts are either absent or deficient, resulting in liver fibrosis, portal hypertension, and eventually cirrhosis. It is the most common cause of persistent obstructive jaundice in newborns lasting more than two weeks is this condition. Abdominal ultrasound (US) is the primary imaging technique used to diagnose BA, while computed tomography (CT) is reserved for more complex cases. The gold standard for diagnosing BA is still intraoperative cholangiogram with liver biopsy. Treatment for BA usually involves Kasai hepatoportoenterostomy, but some patients still require liver transplantation due to diagnostic delays and advanced disease. In this study, the authors present the case of a 3-month-old infant with biliary atresia and its ultrasound characteristics, who underwent liver transplantation due to advanced disease. The primary objective of imaging is to provide a prompt diagnosis, given the crucial significance of timely surgical intervention.

Keywords Biliary atresia · Pediatric · Radiology · Us · Liver · Infant

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Introduction

Biliary atresia (BA) is a devastating pediatric cholangiopathy that impacts the liver bile ducts. Although it is a rare disease, but it remains the most common indication for pediatric liver transplantation, despite the success of the Kasai hepatoportoenterostomy procedure. Pediatric biliary tract diseases encompass a range of conditions with varying clinical presentations. Consequently is essential to correlate sonographic, clinical, laboratory, and epidemiological findings since there are no effective medical therapies to slow the disease's progression. The definitive diagnosis is achieved only through direct exploration of the biliary tract in the surgical theatre, but abdominal ultrasound (US) plays an important role in the screening of infantile cholestasis. High-resolution real-time US is a simple and non-invasive first diagnostic method for differentiating between obstructive and nonobstructive causes of jaundice in infants and children.

Improved understanding of the disease and its diagnosis may lead to the development of targeted therapies and better outcomes for this young patients.

Case presentation

A 3-month-old patient came to our attention for jaundice (conjugated hyperbilirubinemia) and increased liver function indices. A previous abdominal ultrasound, performed in the first few days of life, documented no significant changes in the abdominal organs, with the exception of a stomach full of ingestions and no visualization of the gallbladder. The patient was hospitalized, and metabolic screening, sweat test, and serologic tests (HAV, HCV, HBV, CMV, EBV, HSV, HHV6, Adenovirus, Toxoplasma) were performed and found to be normal.

An abdominal ultrasound was performed, which showed hepatomegaly with superficial nodularity and non-homogeneous ecostructure, perihepatic fluid effusion and in the region of the falciform ligament (Fig. 1), ecogenic fibrous tissue near the portal vein (Fig. 2), splenomegaly with homogeneous ecostructure (Fig. 3). The color-Doppler integration, showed portal vein increased in caliber, with biphasic flow (Fig. 4a), and a slight increase in peripheral subcapsular vascularization hepatic, predominantly arterial (Fig. 4b). The patient was transferred to a highly specialized transplant center where she performed a CT scan of the upper abdomen with contrast that confirmed the presence of liver with superficial nodularity and signs of portal hypertension for

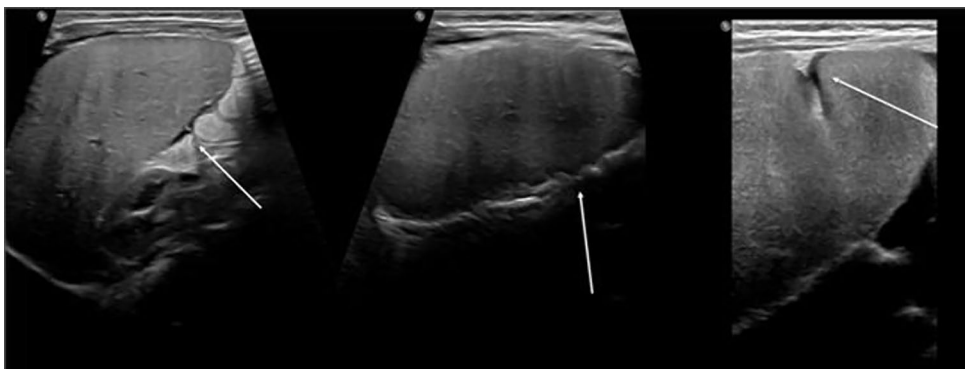

Fig. 1 Ultrasound of the abdomen showed hepatomegaly with superficial nodularity and non-homogeneous ecostructure, perihepatic fluid effusion and in the region of the falciform ligament

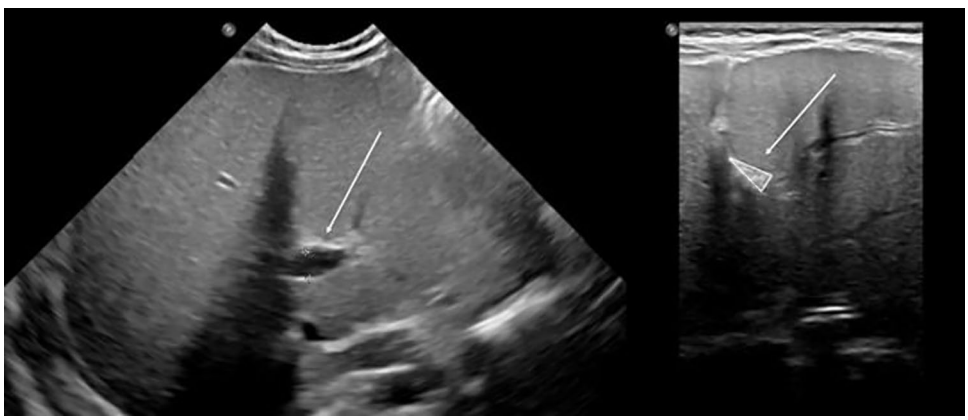

Fig. 2 Ultrasound of the abdomen showed ecogenic fibrous tissue near the portal vein

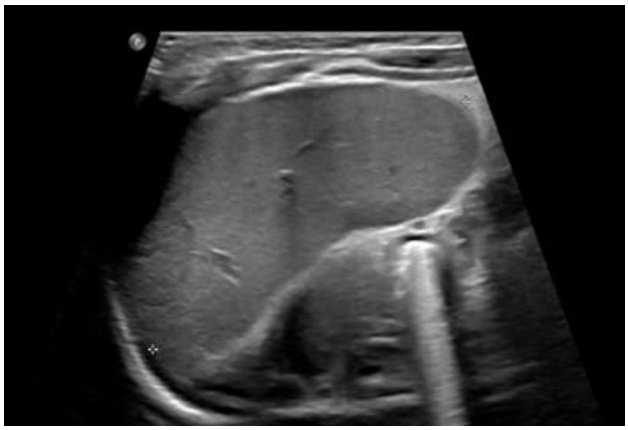

Fig. 3 Ultrasound of the abdomen showed splenomegaly with homogeneous ecostructure

homogeneous splenomegaly, portal vein increased in caliber and packages of perigastric and periesophageal varices. The gallbladder was not displayed and the persistence of the Arantius' duct was also reported. Diagnosis of biliary atresia was made. The little patient had a liver transplant.

Discussion

Biliary atresia (BA) is a congenital disease characterized by an absent or severely deficient extrahepatic biliary tree. It has an incidence of 1/15.000–20.000 live born in mainland Europe, female newborns are slightly more affected than males (M:F = 1:1.5) [1–3]. It's the most common cause of neonatal cholestasis, it occurs within the first three months of life and is characterized by jaundice with conjugate hyperbilirubinemia (direct bilirubin levels > 1.0 mg/dL or > 17 mmol/L), pale stools and dark urine. Other rare presentations may include symptoms like vitamin K-induced bleeding tendencies. However, typical signs of chronic liver disease such as portal hypertension, ascites, and splenomegaly require time to manifest, typically more than three months [1]. The etiology of this condition is unknown, but it is believed to be related to genetic defects in the formation of ductal plaque and bile ducts or ductal inflammation in the uterus by viral or autoimmune causes [4–6]. The underlying process is not known, but hepatocyte function progressively declines, and patients experience portal hypertension and fibrosis, leading to biliary cirrhosis, with a life expectancy of two years. In fact, biliary atresia (BA) has an unfortunate outcome if not recognized and surgically treated promptly (within 45–60 days of birth). If not promptly addressed through surgery within the initial 45–60 days after birth, the outlook becomes grim. This is due to the progressive decline in hepatocyte function, which triggers fibrosis and portal hypertension, ultimately leading to the development of secondary biliary cirrhosis and a life expectancy of only two years [3].

There are two forms of biliary atresia: the non-syndromic form, accounting for about 80% of cases; and the syndromic form, also called biliary atresia splenic malformation syndrome, accounting for about 20% of cases. The syndromic form is associated with polysplenia, intestinal malrotation, preduodenal portal vein, absent inferior vena cava, aberrant hepatic artery and abdominal heterotaxia [7, 8]. Biliary atresia is in differential diagnosis with: neonatal hepatitis**,** Bile-plug syndrome, Alagille syndrome, Choledochal malformation.

The first-level imaging technique for the diagnosis of biliary atresia is abdominal ultrasound, it is important to ensure that the child has been fasting 3–4 h before ultrasound to minimize possible darkening of intestinal gas and allow complete distension and proper evaluation of the gallbladder [9]; failure to fast may misdiagnose an abnormal or absent gallbladder. Abdominal ultrasound may show: absent or abnormal gallbladder, the "gallbladder ghost triad" is characterized by small gallbladder (< 19–25 mm in length) with irregular contour and discontinuous wall; lack of gall bladder contraction after oral feeding; "triangular cord sign" that is echogenic fibrous tissue anterior to portal vein at site of obliterated extrahepatic biliary duct; non visualization/absence of the common bile duct (CBD); presence of microcysts or macrocysts near the porta hepatis; increased hepatic artery diameter, peripheral arterialization (hepatic subcapsular

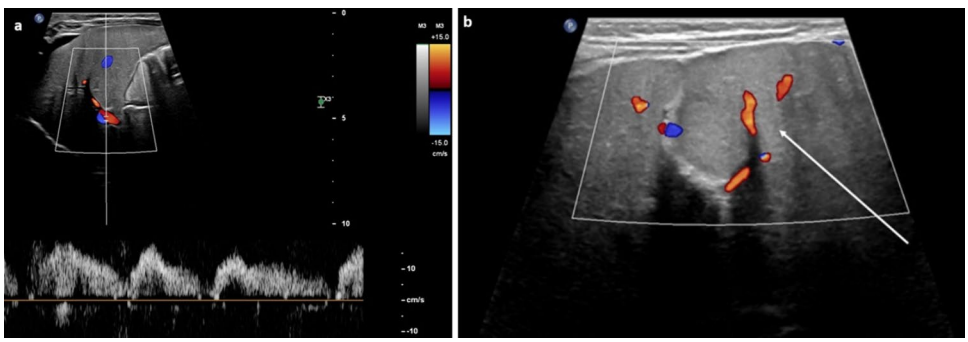

Fig. 4 Ultrasound-color Doppler showed portal vein increased in caliber with biphasic flow (**a**) and slight increase in peripheral subcapsular vascularization hepatic, predominantly arterial (**b**). Origin: © U.O.C. Department of Pediatric Radiology ARNAS Civico-Di Cristina-Benfratelli, Palermo, Italy, 2022

flow); polysplenia [3, 4, 7, 10, 11]. The triangular cord sign has a sensitivity of 78.2%, specificity of 100% and diagnostic accuracy of 90% for biliary atresia [3, 12]. When identified, the combination of triangular cord sign and a gallbladder abnormality is 87% sensitive and 90% specific for biliary atresia [7]. However, it should be remembered that in 20% of biliary atresia cases, the gallbladder may be normal [3, 13, 14]. Moreover, the presence of abdominal heterotaxy, midline liver, polysplenia, asplenia, and preduodenal portal vein further heightens suspicion for biliary atresia accompanied by malformations [4].

Doppler ultrasound, along with hepatic artery measurements, serves as a valuable complement to B-mode ultrasound. Mean hepatic artery diameter and ratio of hepatic arteryto-portal vein diameter are greater in this patients [9].

CT is useful in complicated cases. The gold standard for diagnosis is an intraoperative cholangiogram with concomitant liver biopsy [10]. Other available diagnostic techniques are magnetic resonance cholangiopancreatography (MRCP) and HIDA (hepatoiminodiacetic acid) scan. Frequently, a definitive diagnosis is achieved only in the surgical theatre through direct exploration of the biliary way, preferably combined with intraoperative cholangiography [3].

The primary treatment is surgery, the Kasai hepatoportoenterostomy (HPE), which consists of resection of the choledochal remnants, gallbladder and portal plate and construction of a jejunal Roux-en-Y anastomosis or cholecystostomy to restore biliary drainage [7]. If children with biliary atresia are operated within the first 60 days of age, bile flow can be established in 70% of cases; in those operated after 90 days of age, sufficient bile flow is achieved in only 25% of cases [4, 15]. Liver transplantation is performed if Kasai hepatoportoenterostomy fails or for advanced cirrhosis [7]. Due to her age, advanced liver dysfunction with signs of portal hypertension and the persistence of the Arantius' duct, our patient underwent a liver transplant.

The primary aim of imaging is to achieve a prompt diagnosis, given the critical significance of early surgical intervention. However, distinguishing biliary atresia from other non-surgical causes of neonatal cholestasis poses a significant challenge [7]. Actually, biliary atresia is the most common cause of hepatic transplant in paediatric age (about 75% of hepatic transplant in children aged < 2) [3].

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Declarations

Conflict of interest The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

Consent to publish Informed consent was obtained from patient's parents for the publication of the case report.

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