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ORIGINAL RESEARCH

A Case Report on Congenital Biliary Atresia with Ventricular Septal Defect

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ABSTRACT

Biliary atresia, an obstructive cholangiopathy with an idiopathic etiology, is a congenital disorder that eventually destroys liver parenchyma. In some circumstances, it is associated with other abnormalities such as congenital heart diseases, intestinal, and spleen anomalies. Diagnosis of biliary atresia is based on clinical manifestations and specific diagnostic tests. Prolonged pathological jaundice is a critically important sign in newborns that needs to be recognized early in order to reduce the chances of further complications. The mainstay of treatment is the Kasai procedure (portoenterostomy), and it is eighty percent successful if performed before or within the first two months of life.

Here, we report a case of a two-month-old baby who had biliary atresia with a ventricular septal defect (VSD). The baby had a prolonged history of jaundice, pruritus, yellow-colored urine, and pale stools. He was initially brought to a local clinic for his jaundice, where pharmacological treatment was commenced. However, the symptoms persisted, and he was referred to a hospital where he was placed under blue light phototherapy with no marked improvement in his symptoms. For a definite diagnosis and treatment, he was then referred to a multispecialty hospital where diagnostic tests were ordered. The HIDA scan showed no uptake of contrast in the bile ducts and gallbladder, which is suggestive of congenital biliary atresia. An intraoperative cholangiogram showed a definite diagnosis of biliary atresia, which was performed, and the Kasai procedure was done successfully. All his family members were healthy with no history of congenital disease.

The essential outcome in this case is to consider biliary atresia as a cause of prolonged jaundice in infants, and occasionally it is associated with other congenital anomalies. The diagnosis and treatment should be urgent in order to prevent complications.

KEYWORDS: Biliary Atresia, Kasi procedure, Cholangiogram, Intrahepatic, Extrahepatic.

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INTRODUCTION

Biliary atresia (BA) is a rare congenital anomaly characterized by the obliteration of intrahepatic and extrahepatic bile ducts with fibrous tissues. It results in the blockage of bile flow from the liver to the small intestine and progresses to end-stage liver disease [1]. The overall incidence is low, approximately 1 in 15,000-20,000 births [2]. The exact etiology of this condition is not well recognized. However, multiple causative factors have been attributed to its pathogenesis, including certain gene mutations, various infectious agents like Cytomegalovirus [3, 4], and immunological factors [5].

Diagnosing biliary atresia remains challenging and cannot easily differentiate from other causes of pathological jaundice. The initial symptom is prolonged jaundice in newborns, which does not resolve with pharmacotherapy and phototherapy. Other associated symptoms include dark urine, irritability, and pale stools, which can vary on a daily basis [3, 5, 6]. In rare occasions, approximately 15-20% of cases of biliary atresia coexist with other congenital anomalies, while approximately 10-20% are isolated cases of BA [2]. Biliary atresia can coexist with polysplenia, asplenia, intestinal malformation, situs

inversus, and cardiovascular abnormalities. In some cases, as in this patient, BA coexisted with a ventricular septal defect.

The diagnosis of biliary atresia can be made based on clinical manifestations, liver function tests (LFT), liver biopsy, and HIDA scan, but intraoperative cholangiogram remains the gold standard diagnostic test. Infants suspected of having biliary atresia should be evaluated as soon as possible because optimal outcomes depend on early referral and timely Kasai procedure (hepatoportoenterostomy). About eighty percent of children who undergo this procedure before two months of age will reach adolescence without complications and enjoy a good quality of life [5]. If the condition is diagnosed at a late stage, liver transplantation is the only remedy to overcome this condition [7].

CASE REPORT

A two-month-old male baby presented with jaundice, pruritus, dark yellow-colored urine, and pale stools. His symptoms started on the third day and progressed to the second month of life. A physical examination of the abdomen revealed no abnormality, but a pansystolic murmur was heard in the lower left sternal border. His heart rate was 130 bpm, respiratory rate 25 cpm, temperature 98.8°F, and SpO^2 was 98 percent.

Initially, the patient was taken to a local clinic for evaluation where vitamin D, K, and phenobarbital were prescribed. However, the symptoms deteriorated, and he received phototherapy in a local hospital with no marked improvement in his symptoms. The patient was then referred to a multispecialty hospital for observation and a definite diagnosis. The following tests were performed:

Total Serum bilirubin was 5.6 mg/dl; direct Serum bilirubin was 5.3 mg/dl, while indirect Serum bilirubin was 0.3 mg/dl. In addition to direct hyperbilirubinemia, SGPT was 189 IU/L, Alkaline phosphatase was 1343 IU/L, Gamma GT was 90 IU/L, PT was 51 sec, INR was 4, APTT was 91 sec, and Plasma Ammonia was 97 ug/dl. The level of alpha-1 antitrypsin (AAT) was normal. TSH was 4.3 mU/L. Serological tests for Cytomegalovirus, hepatitis A, B, and C were negative.

Abdominal Ultrasonography was unremarkable. Two-dimensional echocardiography indicated a five-millimeter perimembranous septal defect. Hepatobiliary scintigraphy or HIDA (Hepatobiliary Imino Di Acetic) SCAN revealed fair hepatocyte uptake, CBD and gall bladder were not visualized, and no tracer activity was noted in the gut until 24 hours of the study (fig. 1). Liver biopsy was obtained, and two sections were studied microscopically. Section A from the gallbladder wall showed intact mucosa with columnar cells. Section B examined from the tissue as a liver biopsy showed hemorrhagic material only. During laparoscopic Intraoperative cholangiogram, a shrunken gall bladder was detected. A contrast dye was injected into the gallbladder, but no flow was seen into the extrahepatic biliary duct.

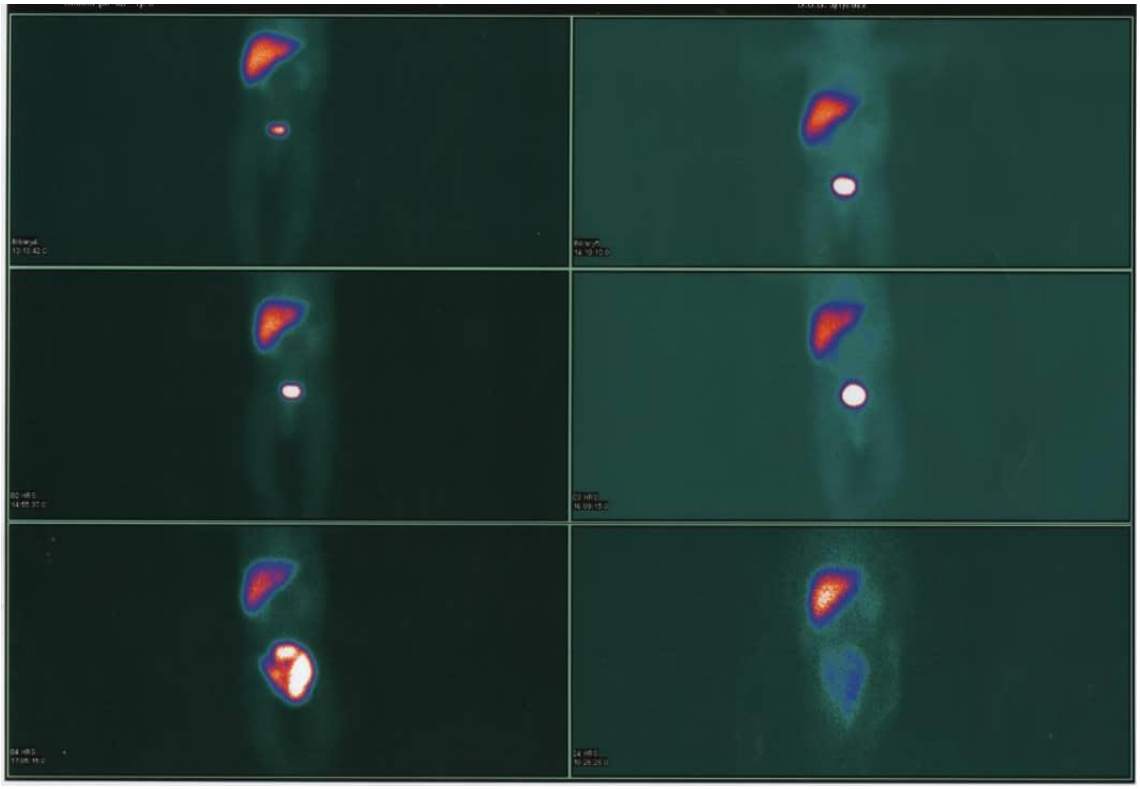

Figure 1: Cholecystography with 1.5 mCi of Tc-99m-DISIDA which shows fair perfusion blush followed by fair tracer uptake by functioning hepatocytes.CBD and gall bladder are not visualized in the study. No tracer activity is noted in the gut until the end of study, excretion of tracer is noted through kidneys.

Clinical manifestations of the patient, along with associated histological, radiological, and intraoperative cholangiogram findings, were all consistent with biliary atresia. The Kasai procedure was successfully performed, and no intraoperative or postoperative complications were noted. Follow-up and repeated laboratory examinations showed significant improvement.

DISCUSSION

Based on the patient's history, physical examination, and laboratory examinations, it was indicated that pathological jaundice was due to persistent direct hyperbilirubinemia. Therefore, common causes of neonatal direct hyperbilirubinemia were considered as a differential diagnosis. In this case, the usual causes of neonatal direct hyperbilirubinemia, which consist of viral hepatitis, cytomegalovirus, alpha1 antitrypsin deficiency, congenital hypothyroidism, congenital biliary atresia, and choledochal cyst, were evaluated [4].

Viral hepatitis is one of the most common infections affecting a high number of people in Afghanistan. However, in this case, serological tests for hepatitis A, B, and C were negative, and hyperbilirubinemia due to viruses has been ruled out. Cytomegalovirus is another infectious cause of pathological jaundice. CMV is a member of the herpes group of viruses. The infection is usually transmitted through contact with infected secretions, particularly saliva and urine, but can also be transmitted transplacentally from a mother to the fetus or through breastfeeding [8]. It is one of the most usual infections, which has various manifestations. It is usually asymptomatic with no obvious symptoms. However, hepatomegaly and cholestatic jaundice are the prominent features of this virus in neonates. This patient's cytomegalovirus PCR and IgM were both negative, indicating jaundice from another etiology.

Another common genetic cause of cholestatic jaundice is alpha-1 antitrypsin deficiency. Children with alpha-1-antitrypsin deficiency can develop liver disease at any age [9]. Itchy skin and pale stools are neonatal symptoms in this disorder, and our patient had the same symptoms. Therefore, genetic testing was performed, which excluded the disease, and the level of alpha1 antitrypsin was normal.

Congenital hypothyroidism and galactosemia can also result in cholestatic jaundice. Several studies have shown that congenital hypothyroidism can result in severe hyperbilirubinemia. One of the most important and earliest signs of hypothyroidism is prolonged jaundice during the neonatal period [10]. Galactosemia is a common metabolic liver disease of childhood, including during the neonatal period, which causes severe hyperbilirubinemia in neonates associated with multiple organ damage [11]. In this case, urinary examination showed no evidence of galactosemia, and newborn screening could not be performed due to financial constraints.

Choledochal cyst, a congenital anomaly of the biliary duct that we suspected as a cause of cholestatic jaundice in this case, was ruled out by physical examination and ultrasonographic findings, which were not consistent with the diagnosis of choledochal cyst. Choledochal cyst is known to be an uncommon cause of neonatal jaundice [12].

Biliary atresia is another cause of cholestatic jaundice. A stepwise approach was carried out in this case to diagnose biliary atresia as the cause of cholestasis. If affected infants are identified with BA within the first months, they undergo the Kasai hepatoportoenterostomy (HPE) attempt to restore bile flow and delay liver cirrhosis [13-14]. In this case, Hepato-biliary scintigraphy or HIDA scan was conducted, revealing no evidence of biliary excretion over a twenty-four-hour period. Furthermore, liver biopsy showed only hemorrhagic material, and finally, Laparoscopic Intraoperative cholangiogram confirmed the diagnosis of biliary atresia as the cause of jaundice in this patient.

CONCLUSION

The essential outcome in this case is to consider biliary atresia as a cause of prolonged jaundice infants and occasionally it is associated with other congenital anomalies. The diagnosis and treatment should begin urgently to prevent complications.

ABBREVIATIONS

VSD	Ventricular Septal Defect
HIDA	Hepatobiliary Imino Di Acetic Acid
BA	Biliary Atresia
SGPT	Serum glutamic pyruvic transaminase
GT	Glutamine Transferase
AAT	Alpha one antitrypsin
CBD	Common Bile Duct
CMV	Cytomegalovirus
INR	International normalized ration
TSH	Thyroid stimulating hormone
PT	Prothrombin time
aPTT	Activated partial thromboplastin time
PCR	Polymerase chain reaction.

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None.

COMPETING INTERESTS

The authors declare no competing interests with this case report.

CONSENT

A written informed consent was obtained from the patient's guardian for publishing the present case report.

AUTHORS' CONTRIBUTIONS

The participation of each author corresponds to the criteria of authorship and contributorship emphasized in the Recommendations for the Conduct, Reporting, Editing, and Publication of Scholarly work in Medical Journals of the International Committee of Medical Journal Editors. Indeed, all the authors have actively participated in the redaction, the revision of the manuscript, and provided approval for this final revised version.

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