

Congenital Biliary Atresia

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INTRODUCTION

Biliary atresia is the progressive fibro-inflammatory obliteration or discontinuity of the extrahepatic biliary system, resulting in obstruction of bile flow, presenting within the first three months of life¹. Although an infectious cause seems reasonable, no agent has consistently been found in such cases. Certain Human Leucocyte Antigen (HLA) types may predispose to biliary atresia¹. If not surgically corrected, secondary biliary cirrhosis invariably results. Patients with biliary atresia can be subdivided into two distinct groups: those with isolated biliary atresia (postnatal form), which accounts for 65-90% of cases, and patients with associated situs inversus or polysplenia/asplenia with or without other congenital anomalies (fetal/embryonic form), comprising 10-35% of cases². The pathology of the extrahepatic biliary system widely varies in these patients, and the following classification is based on the predominant site of atresia:

- Type I involves obliteration of the common duct; the proximal ducts are patent
- Type II is characterized by atresia of the hepatic duct, with cystic structures found in the porta hepatis.
- Type III (>90% of patients) involves atresia of the right and left hepatic ducts to the level of the porta hepatis²

Incidence of biliary atresia has been noted to be between 1:10,000 – 1:15,000 live births³ and is highest in the Asian population. The disorder also occurs in black infants, with an incidence approximately two times higher than that observed among white infants. Extrahepatic biliary atresia occurs more commonly in females than in males².

CASE

A 4 month old female child, delivered at term with a birth weight of 3 kg. The mother noticed yellowish discoloration of eyes soon after delivery which was progressive and associated with deep yellow urine and pale stools. No pruritus was reported. For a period of one month, the jaundice deepened. She

has had no blood transfusions. Her developmental milestones had been well attained. No known liver disorders run in the family.

On examination, the child was alert, afebrile, deeply jaundiced and had some palmar pallor. There was enlargement of lymph nodes in both axillae and inguinal regions measuring 0.5 – 1cm in diameter, firm, discreet, mobile and non-tender. Firm subcutaneous nodules were felt over the anterior chest wall.

Her anthropometric measurements were normal for age. The patient had abdominal distension with visible superficial veins. The liver was hard, non-tender and not nodular, with a firm edge palpable 9cm below right costal margin (liver span 12cm along mid-clavicular line). The spleen was also enlarged 5 cm below left costal margin. The abdomen was tympanic to percussion and normal bowel sounds were auscultated. The remainder of the physical examination was essentially normal.

Full blood picture revealed a moderate normocytic anaemia. The white cell line showed leukocytosis (35,100/ uL) with absolute and relative lymphocytosis (65.9%) but the platelet count was normal. The peripheral smear revealed target cells and a left shift in the white cell line. The Liver Function Tests featured elevated total bilirubin (174.5 umol/L), predominantly of the direct type. Aspartate aminotransferase (523 U/L), Alanine aminotransferase (208 U/L) and Gamma Glutamyl Transferase (154 U/L) were moderately elevated. Serum Cholesterol was slightly elevated.

On Abdominal Ultrasound Scan, the liver displayed homogenous echogenicity and the gall bladder could not be visualised despite adequate fasting. There was no ascites and the biliary ducts were not dilated. These findings were consistent with a diagnosis of congenital biliary atresia with a differential diagnosis of idiopathic neonatal hepatitis.

In the ward, the patient began bleeding continuously from the femoral puncture site which was treated with direct pressure and intravenous Vitamin K. She was also receiving a multivitamin preparation, ferrous sulphate and phenobarbitone. A paediatric surgical review revealed that the girl needed a porto-enterostomy (Kasai's procedure), but this could not be performed immediately due to long waiting lists. She was discharged home and enrolled for follow up in the Paediatric clinic.

Prognosis in this child appeared to be guarded due to the late presentation to hospital and delayed diagnosis – a complication of liver cirrhosis may have already developed as manifested by the bleeding diathesis coupled with the laboratory findings which indicate hepatocellular injury.

DISCUSSION

Neonatal cholestasis typically presents with variable degrees of jaundice, dark urine, and light stools. In the case of biliary atresia, most infants are full-term and may manifest normal growth and weight gain during the first few weeks of life (idiopathic neonatal hepatitis is commoner in pre-term infants; our case was born at term). It also has no familial tendency, unlike idiopathic neonatal hepatitis which has a familial incidence of 20%³.

Hepatomegaly may be present early, and the liver is often firm or hard to palpation. Splenomegaly is common, and an enlarging spleen suggests progressive cirrhosis with portal hypertension. Bleeding tendencies are common due to liver dysfunction. The patient may also have multiple xanthomata signifying hyperlipidemia.

In the absence of coagulopathy (which this child suffers from), the gold standard for confirming a diagnosis of obstructive neonatal cholestasis is a percutaneous liver biopsy. The expected findings on histology are bile ductular proliferation, bile plugging, portal-portal fibrosis, and an acute inflammatory reaction².

Once biliary atresia is suspected, surgical intervention is the only mechanism available for a definitive diagnosis (intraoperative cholangiogram) and therapy (Kasai portoenterostomy)². The Kasai procedure is most successful if performed before 8 weeks of life (90%)³, and should generally not be

undertaken in infants older than age 4 months, because the likelihood of bile drainage at this age is very low¹.

Failure to surgically correct the anomaly will eventually result in various late manifestations including growth failure, fat soluble vitamin deficiencies, pruritus, xanthomata, portal hypertension and bleeding diathesis³. Death is usually caused by liver failure, septicaemia, acidosis or respiratory failure secondary to intractable ascites². Liver transplant has dramatically altered the outlook for such patients in the developed world; unfortunately this remains out of reach for most patients in our setting.

CONCLUSION:

The case highlights the importance of early diagnosis for all cases of neonatal jaundice– and in particular the need for early distinction between medical and surgical causes of hyperbilirubinemia. It also points out the need for training of more specialized personnel and expansion of our paediatric surgical facilities.

REFERENCES:

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