Case Report

Neonatal cholestasis due to primary sclerosing cholangitis

Naman Sadanand Shetty[1], Ira Shah[1]

[1]*Department of Pediatrics, Pediatric Liver Clinic, B.J. Wadia Hospital for Children, Mumbai, Maharashtra, India*

Abstract

Neonatal cholestasis is rarely caused due to primary sclerosing cholangitis, which is an inflammatory disease of the bile ducts, which results in obstructive fibrosis of the ducts. A 7-month-old male child presented with jaundice along with high-colored urine and clay-colored stools since birth. Liver biopsy showed mild bile duct proliferation with cholangioles showing bile and thrombi suggestive of primary sclerosing cholangitis.

Keywords: Clay stools, neonatal cholestasis, primary sclerosing cholangitis

Introduction

Cholestasis is defined as a condition in which there is a reduction in the flow of bile through the extrahepatic or intrahepatic bile ducts due to impaired secretion of bile by the hepatocytes and obstruction of the bile duct. Neonatal cholestasis affects 1 in 2500 live births.[1] Jaundice persisting for more than 2 weeks after birth in infants should be evaluated for neonatal cholestasis.[2] The major etiologic factors of cholestasis are extrahepatic biliary atresia, infections such as bacterial, urinary tract infections, cytomegalovirus, and syphilis, toxins such as drugs, genetic disorders including tyrosinemia, galactosemia, α1 antitrypsin deficiency, and cystic fibrosis, and idiopathic neonatal hepatitis.[3,4] Infants suffering from neonatal cholestasis present with jaundice, dark urine, light or alcoholic stools, and hepatomegaly. Primary sclerosing cholangitis is characterized by inflammation and obliterative fibrosis of intrahepatic and extrahepatic bile ducts with dilation of preserved segments. Neonatal sclerosing cholangitis (NSC) is a rare autosomal recessive condition. Histopathologically, it resembles biliary atresia very closely and a distinction must be made by means of biliary imaging.[5] We present a child who had neonatal cholestasis due to primary sclerosing cholangitis.

Address for correspondence: Dr. Ira Shah, 1/B, Saguna, 271/B, Street Francis Road, Vile Parle (West), Mumbai - 400 056, Maharashtra, India.
E-mail: irashah@pediatriconcall.com

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Case Report

A 7-month-old male child presented with jaundice along with high-colored urine and clay-colored stools since birth. There were no antenatal problems and birth was uneventful with birth weight of 3 kg. He was on exclusive breast feeds. At 5 months of age, he had been hospitalized and a liver biopsy had been done that showed cholestasis with vacuolated appearance of hepatocytes and mild bile duct proliferation with cholangioles, showing bile and thrombi suggestive of primary sclerosing cholangitis. His serial liver function tests are depicted in Table 1. On examination, weight was 5.9 kg and height was 67 cm. He was deeply jaundiced and had hepatosplenomegaly and ascites with palmar erythema. Other systems were normal. Ultrasound of the abdomen showed coarse echotexture of the liver, with normal gallbladder and common bile duct. He was started on Vitamins A, D, E, and K along with spironolactone and ursodeoxycholic acid and advised regarding liver transplant.

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Table 1: Serial liver function tests of patient

	5 months	7 months
Bilirubin (mg/dl, direct)	10.7 (9.2)	15.2 (7.7)
SGOT (IU/L)	-	-
SGPT (IU/L)	127	220
Total proteins (g/dl)	6.1	6.4
Albumin (g/dl)	3.8	2.7
GGTP (mg/dl)	686	366
Alkaline phosphatase (IU/L)	822	-
INR	1	1.6

SGOT: Serum glutamic-oxaloacetic transaminase; SGPT: Serum glutamic-pyruvic transaminase; GGTP: Gamma-glutamyl transpeptidase; INR: International Normalized Ratio

Discussion

NSC is a rare autosomal recessive condition. It is characterized by persistent conjugated hyperbilirubinemia clinically and bile plugs, porto-portal bridging fibrosis with copper-associated protein deposition.[5] Diagnosis is made usually by endoscopic retrograde cholangiopancreatogram. It appears to be common among children of consanguineous marriages, suggesting it is inherited as an autosomal recessive trait. It has been associated with two syndromes: Kabuki syndrome (involving facial dysmorphism, developmental delay, growth hormone deficiency, skeletal anomalies, and congenital heart defect) and neonatal ichthyosis-sclerosing cholangitis syndrome which appears to result from a claudin-1 deficiency.[6] It may be associated with a variety of disorders, including Langerhans cell histiocytosis, immunodeficiency, psoriasis, cystic fibrosis, reticulum cell sarcoma, and sickle cell anemia. The distinction between sclerosing cholangitis and obstructive cholangiopathies of infancy such as biliary atresia is vague. Infants with NSC may have pathological and radiological features that are very similar to biliary atresia early in the course of disease and that do not become characteristic of sclerosing cholangitis till the disease evolves.[7] Our patient had normal gallbladder and common bile duct on ultrasound and liver biopsy done at the age of 5 months was suggestive of primary sclerosing cholangitis though clinically he had presented with jaundice and clay stools which are highly suggestive of biliary atresia. He did not have any ichthyosis or dysmorphism ruling out neonatal ichthyosis or Kabuki syndrome. Treatment of sclerosing cholangitis involves providing symptomatic relief to the patient by administration of cholestyramine for pruritus, and fat-soluble vitamin supplements to prevent vitamin deficiency. Long-term, high-dose ursodeoxycholic acid therapy is associated with improvement in serum liver tests but does not improve survival and was associated with higher rates of serious adverse events.[8] The definitive treatment is liver transplant.[9]

Conclusion

NSC may be considered as a potential etiology in a patient with neonatal cholestasis, especially when the clinical presentation mimics biliary atresia, but there is a presence of gallbladder and liver biopsy shows the presence of cholangitis.

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Conflicts of interest

There are no conflicts of interest.

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