Neonatal cholestasis due to primary sclerosing cholangitis

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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

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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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. 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. 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. 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. The definitive treatment is liver transplant.

Discussion

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.

References

1. Dick MC, Mowat AP. Hepatitis syndrome in infancy – An epidemiological survey with 10 year follow up. Arch Dis Child 1985;60:512-6.
2. Giannattasio A, Ranucci G, Raimondi F. Prolonged neonatal jaundice. Ital J Pediatr 2015;41 Suppl 2:A36.
3. Lykavieris P, Bernard O, Hadchouel M. Neonatal cholestasis as the presenting feature in cystic fibrosis. Arch Dis Child 1996;75:67-70.
4. Feldman AG, Sokol RJ. Neonatal Cholestasis. Neoreviews 2013;14:e63.
5. Charlesworth P, Thompson R. Neonatal sclerosing cholangitis: Kings College Hospital experience. J Pediatr Gastroenterol Nutr 2006;42:E77.
6. Floreani A, Rizzato ER, Ferrara F, Carderi I, Caroli D, Blasone L, et al. Clinical course and outcome of autoimmune hepatitis/primary sclerosing cholangitis overlap syndrome. Am J Gastroenterol 2005;100:1516-22.
7. Amede-Manesme O, Bernard O, Brunelle F, Hadchouel M, Polonovski C, Baudon JJ, et al. Sclerosing cholangitis with neonatal onset. J Pediatr 1987;111:225-9.
8. Lindor KD, Kowdle KV, Luketic VA, Harrison ME, McCashland T, Befeler AS, et al. High-dose ursodeoxycholic acid for the treatment of primary sclerosing cholangitis. Hepatology 2009;50:808-14.
9. Goldberg DS. Liver transplant in patients with primary sclerosing cholangitis. Gastroenterol Hepatol (NY) 2016;12:127-9.

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.