Clinical Case Reports International

Case Report
Published: 28 Oct, 2023

Long-Term Follow-Up of a Patient with Congenital Hepatic Fibrosis: A Case Report and Review of the Literature

Surkov AN[1,2], Namazova-Baranova LS[1,2], Baranov AA[1,3], Bessonov EE[1], Arakelyan AL[1], Maleto EM[2] and Firumyanc AI[1]*

[1]*Petrovsky Russian Scientific Center of Surgery, Russia*

[2]*Pirogov Russian National Research Medical University, Russia*

[3]*Sechenov First Moscow State Medical University, Russia*

Abstract

Congenital Hepatic Fibrosis (CHF) is a rare, autosomal recessive disease caused by a mutation in the ARPKD gene. It is based on dysgenesis of the biliary tract resulting from a malformation of the ductal plate of the interlobular bile ducts, as a result of impaired epithelio-mesenchymal inductive interactions with the formation of cystic extensions. In patients, clinical manifestations range from asymptomatic hepatosplenomegaly to severe manifestations such as acute liver failure and cirrhosis. Bleeding from esophageal varices, due to portal hypertension, is a serious complication of the disease, threatening death in the absence of urgent surgical intervention. Endoscopic sclerotherapy of esophageal varices is possible, but the most effective method of correcting portal hypertension is portosystemic shunting. This article presents a clinical case of a 24-year-old man with CHF.

Keywords: Congenital hepatic fibrosis; Morphological changes; Portosystemic shunting; Esophageal varices; Stenosing papillitis

Case Presentation

A 24-year-old man. From an early age, the child had a belly enlargement, but no additional examination was carried out. At the age of 6 years, the parents noted the appearance of veins on the chest and abdomen.

The general physical examination revealed hepatosplenomegaly, and laboratory investigations showed thrombocytopenia (134 × 10^9/l) and minimum increase in AST (76.3 IU/l). There were no data for coagulopathy. Viral hepatitis B and C, autoimmune hepatitis, primary sclerosing cholangitis, alpha-1-antitrypsin deficiency, Wilson's disease and hereditary hemochromatosis were excluded. According to abdominal ultrasound: Hepatomegaly (111 mm × 48 mm) with diffuse parenchymal changes, splenomegaly (137 mm × 54 mm), an obliterated umbilical vein with a diameter of 7 mm to 8 mm with active blood flow and continuing parallel to the epigastric superficial vein with the mouth into the basin of the internal iliac vein is visualized. According to the results of gastroesophageal fibroscopy: Grade 2 esophageal varices with potential risk of bleeding. Angiography showed an extrahepatic form of portal hypertension.

Laparotomy, "side-to-side" splenorenal shunting, liver biopsy was performed. Postoperative period without negative dynamics.

Morphology of liver specimens (Figure 1A-1D): Liver architectonics is impaired, wide, connective tissue septa with a large number of cystic dilated bile ducts surrounding unchanged hepatic lobules are detected, central veins are present, hepatocytes with fine-grained cytoplasm. Morphological changes correspond to CHF.

After shunting, the patient was stable and underwent regular examination. He received hepatoprotective therapy (ursodeoxycholic acid). Hepatosplenomegaly, thrombocytopenia persisted, AST was not increased, there were no data for esophageal varices according to the results of control gastroesophageal fibroscopy. According to ultrasound, the anastomosis is complete. At 17 years of age, according to transient elastography, the median value was 39.7 kPa (F4 – on the METAVIR scale).

According to the abdominal MRI (Figure 2A-2D), significant structural changes in the liver parenchyma, signs of cholangitis were revealed. The course of hepatoprotective therapy was

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Correspondence:
Surkov Andrey Nikolayevich, Petrovsky Russian Scientific Center of Surgery, Russia

Received Date: *07 Oct 2023*
Accepted Date: *24 Oct 2023*
Published Date: *28 Oct 2023*

Citation:
Surkov AN, Namazova-Baranova LS, Baranov AA, Bessonov EE, Arakelyan AL, Maleto EM, et al. Long-Term Follow-Up of a Patient with Congenital Hepatic Fibrosis: A Case Report and Review of the Literature. Clin Case Rep Int. 2023; 7: 1618.

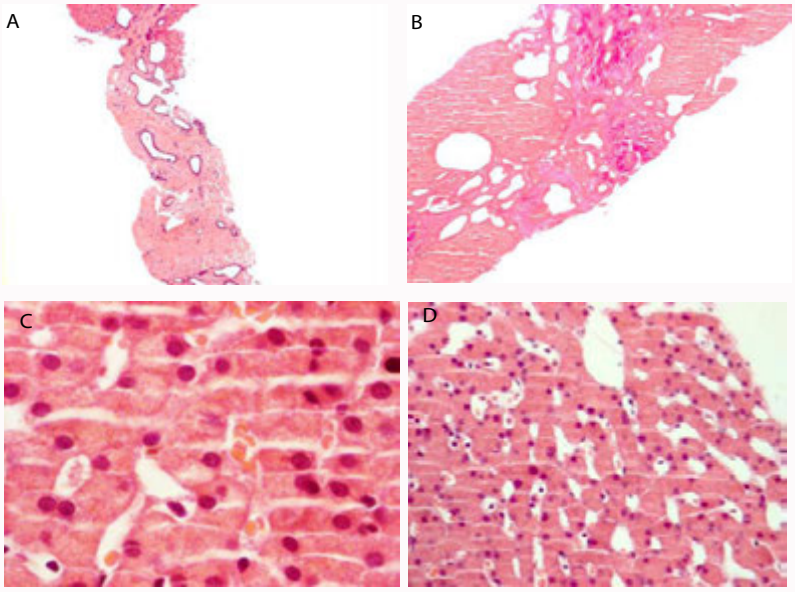

Figure 1 (A-D): Morphological changes in liver in patient with CHF. A) Multiple cystic dilated bile ducts, H&E stain, magnification x100. B) Cystic dilated bile ducts with periductal fibrosis, Van Gieson's stain, magnification x100. C) Intrahepatic cholestasis, H&E stain, magnification x1000. D) Sinusoidal expansion and fullness, H&E stain, magnification x400.

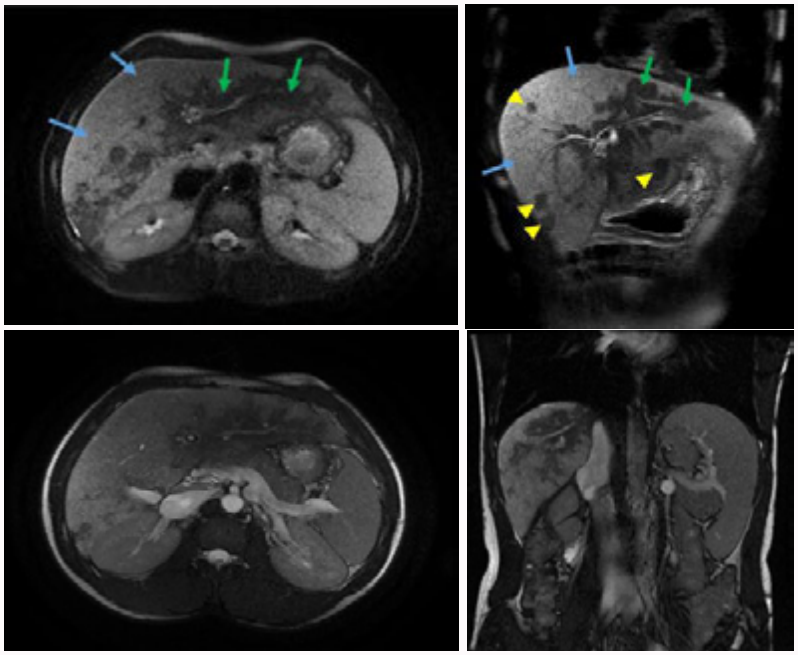

Figure 2 (A-D): MR imaging appearance in patient with CHF. A) Axial T2-weighted images of the liver show diffuse hyperintense fibrotic reticulations (blue arrows) predominantly at peripheral areas. Spared liver parenchyma is visualized at perivascular area (green arrows). B) Coronal T2-weighted images of the liver show diffuse hyperintense fibrotic reticulations (blue arrows) and spared liver parenchyma at perivascular area (green arrows). Also note variable-sized predominantly hypointense regenerative nodules (yellow arrowheads). C-D) Radiological features of portal hypertension in patient with cirrhosis. Axial and coronal 2D Fast Imaging Employing Steady-State Acquisition (FIESTA) show splenomegaly, dilated splenic, left renal and inferior cava veins.

continued.

At the age of 24, the patient was hospitalized with complaints of febrile fever, jaundice, abdominal pain. Laboratory investigations showed negative dynamics of thrombocytopenia (96 × 10^9/L), increased AST (204.1-748.3 IU/L), ALT (101.9-1164.0 IU/L), total bilirubin to 45.9 mmol/L, direct fraction – 20.7 mmol/L and C-reactive protein (64.03 mg/L). Abdominal ultrasound revealed thickening of the walls and dilation of the bile ducts, biliary sludge. MR-signs of choledochoectasia.

Due to the revealed signs of stenosing papillitis, biliary hypertension, mechanical jaundice, endoscopic atypical papillosphincterotomy, choledochal stenting was performed. Patient received antibacterial, hepatoprotective and gastroprotective therapy with positive dynamics – complaints, cytolysis syndrome, cholestasis and inflammatory activity were stopped. He was discharged in a satisfactory condition.

Discussion

The problem of diagnosing CHF is a different clinical course, similar to liver cirrhosis and other diseases that are associated with portal hypertension. The distinctive features of CHF are the relative preservation of liver function, despite its severe structural changes. With CHF, in contrast to cirrhosis, severe portal hypertension initially dominates in preschool and primary school age, whereas with cirrhosis, hepatitis, coagulopathy and liver dysfunction are first present [1,2].

Therefore, morphological examination of liver specimens is of particular importance, which makes it possible to verify the correct diagnosis.

The main causes of mortality in CHF, which is 30% to 50%, are massive bleeding from esophageal varices, as well as uremia, liver failure and cholangitis. Late complications include cholangiocarcinoma and amyloidosis.

The most effective surgical method of treatment is selective portocaval shunting by applying a distal splenorenal anastomosis "side-to-side" without splenectomy, which should be performed already at the grade 2 esophageal varices, and at the risk of bleeding is carried out immediately.

The long-term results of surgery shunting indicate a significant regression of the degree of esophageal varices and the absence of relapses. Therefore, the prognosis of the disease can be considered relatively good [3].

The purpose of therapy for CHF is to prevent or eliminate the consequences of portal hypertension. Treatment with antibacterial drugs is indicated for the development of acute or recurrent cholangitis. The effect of choleretics, which increase the outflow of bile, can be used as an additional method of therapy in cases of recurrent and refractory cholangitis. In this case, ursodeoxycholic acid is preferred [4].

Conclusion

Thus, for the correct diagnosis of CHF, a multidisciplinary approach is needed with the participation of pediatricians, hepatologists, radiologists, endoscopists and pediatric surgeons for timely detection of portal hypertension and decision-making on surgical intervention, which will improve the prognosis of the disease and prevent the development of life-threatening complications. It is important to remember that despite the success shunting, episodes of biliary hypertension may occur with VFP, which should be taken into account in clinical practice.

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