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Congenital Hepatic Fibrosis: 2 Case Reports

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Abstract **Case Report**

Congenital hepatic fibrosis is a rare disease. It is included in the group of congenital fibrocystic diseases. Its clinical manifestations are variable, but it is most often revealed by the signs of portal hypertension. The diagnosis is based on the histopathological study of the liver biopsy. We report two cases of congenital liver fibrosis observed in two 18-year-old patients.

Keywords: Congenital hepatic fibrosis, fibropolycystic diseases, portal hypertension, liver biopsy, periportal fibrosis, bile ducts proliferation.

INTRODUCTION

Congenital hepatic fibrosis (CHF) is a rare cause of portal hypertension. It is a rare autosomal recessive disease; responsible for a lack of remodeling of the ductal plate resulting in periportal fibrosis associated with a proliferation of bile ducts. CHF is included in the group of congenital fibrocystic diseases which includes a wide spectrum of diseases that are generally accompanied by liver damage. It is usually associated with polycystic kidney disease [1, 2]. However, cases of isolated congenital hepatic fibrosis have been observed [3, 4]. We report 2 cases of congenital hepatic fibrosis observed in two 18-year-old patients.

CASE REPORTS

Case 1

18-year-old female patient with congenital cardiac heart disease made of interatrial communication and spontaneous closure of interventricular communication. At the age of 12; the patient presented with a portal hypertension syndrome revealed by a gastrointestinal bleeding due to a rupture of esophageal varices. During the etiological research; viral serology, autoimmune tests, Wilson's disease and hemochromatosis tests were all negative. Thus a liver biopsy was performed. The pathological study showed collagen fibrosis surrounding portal spaces in favor of congenital liver fibrosis. The patient underwent esophageal variceal ligation and was put under propranolol with regular follow-up.

The evolution was characterized by progressive progression to cirrhosis with the appearance of biological signs of hepatocellular failure and a dysmorphic liver in abdominal imaging.

The condition of the patient worsened at the age of 18 by the occurrence of infected ascites. The analysis of ascites fluid showed: rate of protein at 5.5 g / l, white blood cells count: 1600/ mm³ with neutrophils at 1120 / mm³, absence of germs on direct examination.

The patient was first put on antibiotics and albumin perfusions and afterwards diuretics were also introduced with good clinical and biological progress.

Currently the patient is waiting for a liver transplant and is regularly followed in consultation.

Case 2

Our second case is about an 18-year-old male patient, diagnosed with polycystic kidney disease since the age of 6 months and who's currently in chronic kidney failure stage. At the age of 7, he developed a portal hypertension syndrome revealed by its first bleeding episode due to a rupture of esophageal varices. The patient underwent esophageal varices ligation and put under propranolol with regular monitoring.

As part of the etiological research, abdominal imaging had not noticed any liver cysts. Viral serology and autoimmune tests were negative.

Considering that the patient had a huge splenomegaly responsible for significant hypersplenism with the need for repeated transfusions; a splenectomy was performed. During the surgical procedure, a liver biopsy was also performed. Its pathological study had revealed a periportal fibrosis and irregularly shaped proliferating bile ducts in favor of congenital hepatic fibrosis.

Ten years after splenectomy, the patient was admitted to our center with gastrointestinal bleeding. Esophagogastroduodenoscopy found a Forrest III gastric ulcer with a type I gastric varix with red markings.

The patient was put on proton-pump inhibitor with sclerosis of the gastric varix and continuation of propranolol. The patient has no jaundice nor ascites, no evidence of neither clinical nor biological hepatocellular failure and no liver cysts on imaging.

Currently, the patient is regularly followed-up in hepatology and nephrology consultation.

DISCUSSION

Congenital hepatic fibrosis (CHF) is an autosomal recessive disease defined in terms of histological findings by periportal fibrosis associated with a proliferation of bile ducts.

The CHF is one of the fibropolycystic diseases, which also includes Caroli disease, autosomal dominant polycystic kidney disease (ADPKD), and autosomal recessive polycystic kidney disease (ARPKD) and other rare diseases [2].

These are diseases linked to gene mutations that code for the formation of kidney and bile duct structure leading to embryonic malformations of the ductal plate [5]. These mutations are located on chromosome 6; it is the PKHD1 gene involved in the synthesis of fibrocystin, the protein responsible for major structural abnormalities in the liver and kidneys [5-7].

Although infantile polycystic kidney disease is an autosomal recessive disease, our second patient is a sporadic case and there were no other cases in the family.

Since CHF occurs in association with a range of hereditary and non-hereditary diseases, with the involvement of several organs. Whenever the diagnosis of CHF is made; an investigation of the involvement of other organs must be done.

In most cases, the first manifestations of the disease are signs associated with portal hypertension, especially splenomegaly and esophageal varices, complicated often by gastrointestinal bleeding as was the case for our two patients [8]. Clinical manifestations of CHF are, however, non-specific, rendering the diagnosis extremely difficult. The onset of signs is highly variable, ranging from infancy to the 5th or 6th decade of life, although this disorder is diagnosed in most patients in adolescence or young adulthood [9]. The diagnosis was made in adolescence for our two patients.

In adults, the disease is associated with two major risks: gastrointestinal hemorrhage caused by portal hypertension and cholangitis due to a bacterial infection of the dilated intra- hepatic bile ducts. Luckily, none of our patients developed a cholangitis.

Diagnostic confirmation is based on performing a liver biopsy. The presence of periportal fibrosis associated with biliary duct proliferation in the pathological study is sufficient to confirm the diagnosis [2, 4].

There is no drug available to treat fibrosis and malformation of ductal plaque. Treatment of this condition consists on the management of complications such as cholangitis and portal hypertension. This management can be medical, surgical or endoscopic [2].

Medical treatment involves the use of nonselective β-blockers such as propranolol in an effort to reduce portal pressure. Endoscopic ligation of esophageal varices might be used as primary or secondary prophylaxis of gastrointestinal bleeding by esophageal varices rupture. Medical and endoscopic treatments are often associated. However, the treatment of choice is based on performing surgical portosystemic shunts [10].

Liver transplantation is the only known cure for CHF. It is indicated in advanced stages with signs of severe hepatocellular failure [2].

CONCLUSION

Congenital hepatic fibrosis is a very rare disease that is often associated with other fibropolycystic diseases. The liver biopsy is essential to confirm CHF. Once the diagnosis is made, we must look for the involvement of other organs especially the kidney.

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