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CASE REPORT

Caroli syndrome: a clinical case with detailed histopathological analysis

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Abstract

Herein we present a clinical case of the Caroli syndrome caused by the compound heterozygous mutation in the *PKHD1* gene. Histopathological assessment of liver detected biliary cirrhosis, numerous dilated bile ducts of various sizes, hyperplastic cholangiocytes containing a large amount of acid mucopolysaccharides, decreased ß-tubulin expression and increased proliferation of cholangiocytes. A significant proportion of hepatic tissue was composed of giant cysts lined with a single layer of cholangiocytes, containing pus and bile in its lumen and surrounded by granulation tissue. An accumulation of neutrophils in the lumen of the bile ducts was observed, as well as an infiltration of the ducts and cysts surrounding connective tissue by $CD4^+$ and to a lesser extent $CD8^+$ lymphocytes. This may be caused by the expression of HLA-DR by cholangiocytes. Atrophy and desquamation of the epithelium of collecting tubules with the formation of microcysts were detected in the kidneys without a clinically significant loss of renal function. Morphopathogenetic mechanisms of the Caroli syndrome can be targets for a potential pathogenetic therapy and prevention of its manifestations and complications.

Keywords Caroli syndrome · Fibrocystin · Ciliopathy · Histopathology

Introduction

The Caroli syndrome (CS) is a rare (prevalence 1:1,000,000) autosomal recessive hereditary disease characterized by a cystic dilatation of the intrahepatic biliary ducts (IHBD) in combination with congenital portal fibrosis. The disease is caused by mutations in the *PKHD1* gene, which contains 67 exons and is localized in chromosome 6p21.1-p12. These mutations lead to a defect in the fibrocystin protein—a structural component of the primary cilia of the cholangiocytes—and cystogenesis [1].

There is a distinction between the CS and Caroli disease, which is characterized by isolated dilatation of large IHBD, the absence of congenital liver fibrosis, in which only one portion of the liver may be affected [2]. The syndrome and the disease were named after the French physician J.Caroli, who was the first to describe the manifestations of this condition without concomitant congenital fibrosis in detail (1958) [3].

Case report

A male patient at the age of 37 was hospitalized with complaints of severe epigastric pain, nausea and vomiting. The patient suffered cirrhosis of the liver of unclear etiology since childhood (upon admission: Child–Pugh class B, Model for End-Stage Liver Disease (MELD) Score 43), and had manifestations of portal hypertension. The patient was infected by the human immunodeficiency virus (HIV), and his Hepatitis B, C was negative, while drug and alcohol use

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