INTRAHEPATIC CYSTIC DISEASE WITH CONGENITAL FIBROSIS (CAROLI’S COMPLEX DISEASE). A CASE REPORT AND REVIEW OF THE LITERATURE

Demetrio Tamiolakis¹, Panagiotis Prassopoulos², Athanasia Kotini³, Kiriaki Avgidou⁴, Constantine Simopoulos³ and Nikolas Papadopoulos¹

¹Department of Cytopathology, General Hospital of Alexandroupolis, ²Department of Radiology and ³Department of Surgery, Democritus University of Thrace, Alexandroupolis, Greece

SUMMARY – A female patient affected by Caroli’s disease with congenital fibrosis (Caroli’s complex), aged 27 years, is described. Caroli’s disease had been asymptomatic to the present. It was recognized as an intraoperative finding during the left hepatectomy procedure after an acute abdominal crisis episode. The main reason for this surgery was the incidence of malignant transformation to cholangiocarcinoma of the cells of the cystic walls. The complex Caroli’s disease is more common than other forms. The case report is supplemented with literature review and discussion on the etiopathogenetic mechanisms hypothesized.

Key words: Caroli’s disease, etiology; Caroli’s disease, diagnosis; Caroli’s disease, complications; Hepatectomy; Case reports

Introduction

Although Caroli’s disease generally involves the entire liver, it may be segmental or lobar. The inheritance is autosomal recessive. Clinically, patients suffer from bouts of recurrent fever and pain. Jaundice occurs only when a stone blocks the common bile duct°. Leukocytosis is observed typically when acute cholangitis develops. Liver tests are generally normal except during the episodes of obstructive jaundice. The diagnosis is established by cholangiography (intravenous, transhepatic), endoscopic retrograde cholangiopancreatography, ultrasonography, and computed tomography (CT). The complications include recurrent cholangitis, abscess formation, septicemia or pyemia, intrahepatic lithiasis, and amyloidosis. Adenocarcinomas, including some arising in cases with a lobar distribution, have also been reported³,⁴. Surgical treatment is by internal or external drainage procedures. Segmental or lobar forms of Caroli’s disease can be treated by partial hepatectomy.

According to Desmet³, the pathogenesis of Caroli’s disease seems to involve total or partial arrest of remodeling of the ductal plate of the larger intrahepatic bile ducts. In Caroli’s syndrome (Caroli’s disease with congenital hepatic fibrosis) the hereditary factor causing the arrest of remodeling seems to exert its influence not only during the early period of bile duct embryogenesis, but also later during development of the more peripheral biliary ramifications (the interlobular bile ducts).

Case Report

A 27-year-old female was referred to our hospital with an acute abdominal crisis. On clinical examination there was hepatomegaly. Serologic testing showed leukocytosis and normal levels of the tumor associated antigens CEA and CA 19-9. Indirect hemagglutination test was negative.

Ultrasound revealed multiple small and large unilocular cysts involving the left hepatic lobe. Most appeared anechoic due to clear fluid content with posterior acoustic enhancement, and had a smooth regular wall. There were also some cysts containing internal echoes, and the largest ones produced mass effect with pericystic biliary dilatation. One cyst showed a discontinuity in the wall...
(ruptured cyst). CT appearance of the cysts with respect to ultrasonography was as follows: anechoic ones consisted of a well-circumscribed, homogeneous mass with no discernible wall. They had attenuation value (range from -5.8 HU to 20 HU), and showed no enhancement after intravenous contrast medium administration. Those producing mass effect showed contrast enhancement (evidence of pericystic biliary radicle dilatation). Left hepatectomy was subsequently performed and the surgical specimen was submitted to routine histologic analysis.

On gross examination multiple cysts (of greatest diameter ranging from 1 to 4.5 cm) were found (Fig. 1). The cysts were round to oval and were encapsulated. They contained several liters of clear fluid. Some of them were hemorrhagic and one was ruptured. The lining of the cysts consisted of a single layer of flat or cuboidal epithelium (Fig. 2). The cells rested on a basement membrane and were supported by a variably dense fibrous connective tissue. The hepatic parenchyma was intersected by sinuous bands of dense fibrous connective tissue that involved portal areas, isolating single or several lobules (Fig. 3). The lobular architecture was partly disturbed. The hepatocytes showed no abnormalities. Our findings were consistent with congenital cystic disease and hepatic fibrosis (Caroli’s syndrome).

Discussion

Cystic and dysplastic lesions of the liver are a complex group of disorders, the relating classification and terminology being quite confused. The dilatations may be extrahepatic or intrahepatic but may affect the entire biliary system, either diffusely or focally. Some consist of cystic dilatations of the biliary tract that communicates with the bowel (e.g., choledochal cyst, Caroli’s disease). The classifications proposed by Longmire et al. in 1971 and by Hadad et al. in 1976, which in turn was partly based on the classification of Alonso-Lej et al. from 1959, have helped
clarify the issue. Whether the solitary (non-parasitic) bile
duct cyst fits into this group of disorders remains to be
determined. The cystic dilatations, however, are by no
means `pure`, i.e., limited to dilatation of one form or an-
other, since some may also be associated with proximal and/
or distal atresia, or with congenital hepatic fibrosis. Landing
has proposed a unifying hypothesis for neonatal hep-
atitis, biliary atresia and choledochal cyst.

Congenital polycystic disease clearly exists in two
forms, the infantile and the adult, which have different
modes of inheritance; von Meyenburg complexes fall into
the spectrum of adult polycystic disease. Congenital he-
aptic fibrosis is regarded by some as morphologically indis-
tinguishable from infantile polycystic disease, although
others consider the renal changes to be different. Formi-
dable arguments have been advanced against the very ex-
istence of congenital hepatic fibrosis as an entity
The relationship of congenital hepatic fibrosis to other dis-
eses such as Ivemark’s familial dysplasia and Meckel’s syn-
drome requires clarification, as pointed out by Lieberman
el al. and Murray-Lyon et al. The hepatic changes in
Potter’s type III cystic disease of the kidney also appear
equivalent to those of congenital hepatic fibrosis. A clas-
sification based on the renal abnormalities may perhaps
prove more useful than the one emphasizing liver chang-
es. A unifying concept of all congenital cystic and dysplas-
tic liver diseases has yet to be formulated.

The current classification includes five types:
type 1 – choledochal cyst, a localized cystic dilatation of
the extrahepatic bile duct;
type 2 – diverticulum of the common bile duct or gallblad-
der;
type 3 – choledoche, a lesion that extends into the wall
of the duodenum;
type 4 – multiple dilatations of extra- and intrahepatic
ducts (Caroli’s syndrome); and
type 5 – fusiform extra- and intrahepatic dilatation.

Our case was consistent with type 4 lesion (Caroli’s syn-
drome). Hypertension in these patients is said to be
presinusoidal and may be related to the hypoplasia of the
portal veins. It can be treated by portal system shunt. In a
series of 27 children with congenital hepatic fibrosis, por-
tasystemic shunt was carried out in 16 children between
3 and 16 years of age. No impairment of hepatic function
or sign of hepatic encephalopathy was apparent during
follow-up examinations over periods ranging from 3 months
to 12 years. The infantile form has a favorable prognosis.
As far as the adult form is concerned, the treatment of
choice is hepatic resection due to the high incidence of
malignant transformation to cholangiocarcinoma of the
cells of the cystic walls. Simple Caroli’s disease (with-
out congenital fibrosis) is less common than other forms

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Sažetak

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D. Tamiolakis, P.Privopoulos, A. Kotini, K. Avgidou, C. Simopoulos i N. Papadopoulos


Ključne riječi: Carolijevo bolest, etiologiju; Carolijeva bolest, dijagnostika; Carolijeva bolest, komplikacije; Hepatektomija; Prikaz slučaja