

Case Report



Caroli'S Syndrome Associated with Medullary Sponge Kidney: A Case Report and Review of The Literature

Mehmet İlkin NAHARCI^{a1}, Gökhan ERDEM¹, Cemal Nuri ERÇİN², Teoman DOĞRU¹, İlker TAŞÇI¹,
Fatih ÖRS³, Serkan TAPAN⁴, Alper SÖNMEZ¹

¹Gülhane Askeri Tıp Akademisi, İç Hastalıkları Anabilim Dalı, ANKARA, Türkiye

²Gülhane Askeri Tıp Akademisi, Gastroenteroloji Anabilim Dalı, ANKARA, Türkiye

³Gülhane Askeri Tıp Akademisi, Radyoloji Anabilim Dalı, ANKARA, Türkiye

⁴Gülhane Askeri Tıp Akademisi, Biyokimya Anabilim Dalı, ANKARA, Türkiye

ABSTRACT

Herein, we presented a case of Caroli's syndrome associated with medullary sponge kidney and reviewed the literature. A 21-year-old male was admitted to our internal medicine department with the complaints of fatigue, anorexia and recurrent right upper abdominal pain. Abdominal ultrasonography showed hepatomegaly and multiple dilated intrahepatic bile ducts. Computed tomography of the abdomen indicated cystic images distributed in both lobes of the liver and multiple medullary cysts within the kidneys. The magnetic resonance cholangiopancreatography (MRCP) pointed out multiple cystic dilatations of intrahepatic bile ducts. The main bile duct and extrahepatic bile ducts were appeared to be normal. The patient had a brother who had been diagnosed of Caroli's syndrome 12 years ago. Radiologic investigation of the other family members revealed no abnormality.

Key words: Caroli's syndrome, medullary sponge kidney

ÖZET

Medullar Sünger Böbrek İle İlişkili Karoli Sendromu: Olgu Sunumu ve Literatürün Gözden Geçirilmesi

Biz burada medullar sünger böbreği ile beraber Caroli sendromu olan bir olguyu sunup literatürü gözden geçirdik. 21 yaşındaki bir erkek iç hastalıkları kliniğimize yorgunluk, iştahsızlık ve tekrarlayan sağ üst kadran ağrısı ile başvurdu. Batın ultrasonografisinde hepatomegali ve intrahepatik safra kanallarında birçok dilatasyon görüldü. Batının bilgisayarlı tomografisi ile incelenmesinde karaciğerin her iki lobunda kistik oluşumlar, ayrıca böbrekte medullar kistler görüldü. Manyetik rezonans kolanjiopankreatografi tetkikinde intrahepatik safra kanallarında birçok kistik dilatasyon tespit edildi. Ana safra kanalı ve ekstrahepatik safra kanalları normaldi. Hastanın 12 yıl önce Caroli sendromu tanısı konulan bir erkek kardeşi vardı, ancak ailenin diğer üyelerinin radyolojik araştırmalarında anormallik görülmedi.

Anahtar kelimeler: Karoli sendromu, medullar sünger böbrek

Caroli's disease is a congenital disorder characterized by multifocal, segmental dilatation of large intrahepatic bile ducts. Caroli's syndrome includes both of Caroli's disease and congenital hepatic fibrosis (1). Caroli's disease and Caroli's syndrome are extremely rare disorders (1 case per 1,000,000 populations), with autosomal recessive inheritance pattern (2). The disease may emerge anytime in life but is unusual after the fifth decade. Its clinical course is characterized by repeated episodes of bacterial cholangitis due to biliary stagnation within the dilated ducts (3,4). Caroli's syndrome is associated with renal cystic dilatations in 60-80% of patients. Medullary sponge kidney is most frequently noted kidney disorder (5). Herein, we present a case of Caroli's syndrome associated with medullary sponge kidney.

CASE REPORT

A 21-year-old male was referred to our internal medicine department with the complaints of fatigue, anorexia and recurrent

right upper abdominal pain. His complaints had been present for 8 years. His family history was evident for a male sibling that had been diagnosed of Caroli's syndrome 12 years ago. Radiologic investigation of the other family members revealed no abnormality.

Physical examination revealed an enlarged liver, palpable 3 cm below the right costal margin. Pertinent laboratory investigation was as follows: Complete blood count and routine biochemical analysis revealed as; hemoglobin: 14.1 g/dl, leukocyte count: 12.700 /mm³, platelet count: 220.000 /mm³, blood urea nitrogen: 34 mg/dl, creatinine: 0.7 mg/dl, total bilirubine: 0.6 mg/dl, alanine aminotransferase: 8 U/l, aspartate aminotransferase: 20 U/l, alkaline phosphatase: 121 U/l. Urin analysis was normal.

Plain x-ray of the abdomen was unremarkable. Abdominal ultrasonography showed hepatomegaly, multiple dilated intrahepatic bile ducts and multiple cystic dilatations within both kidneys. Computed tomography of the abdomen indicated

^a Corresponding Adress: Dr. Mehmet İlkin NAHARCI, Gülhane Askeri Tıp Akademisi, İç Hastalıkları Anabilim Dalı, ANKARA, Türkiye
e-mail: drnaharci@yahoo.com

cystic images distributed in both lobes of the liver and multiple medullary cysts within the kidneys (Figure 1). Magnetic resonance cholangiopancreatography (MRCP) was performed, which pointed out multiple cystic dilatation of the intrahepatic duct (Figure 2). The main bile duct and extrahepatic bile ducts were normal.

Based on these clinical and laboratory findings, he was diagnosed as Caroli's syndrome with medullary sponge kidney, and then he has currently being followed up at our out-patient department for the complications of Caroli's syndrome.



Figure 1. CT scans showing cysts in liver and in kidneys.

DISCUSSION

The disease was firstly described by Caroli et al. and has autosomal recessive inheritance (3). Two types of the disease have been identified, type 1 (Caroli's disease) and type 2 complex form (Caroli's syndrome) which is associated with congenital hepatic fibrosis, portal hypertension and cirrhosis (6). Caroli's disease may be localized to one lobe of liver or may be diffuse.

The clinical manifestations of Caroli's syndrome are related to the biliary abnormalities and portal hypertension (7). There are several clinical presentations depending on the age of onset and the predominance of hepatic or renal involvement. In both Caroli's disease and syndrome, the saccular or fusiform dilatation of bile ducts predisposes to stagnation of bile leading to the formation of biliary sludge and intraductal lithiasis. Bacterial cholangitis occurs frequently and may be complicated by septicemia and hepatic abscess (2). Patients with Caroli's syndrome can present with portal hypertension and its sequelae, such as ascites and esophageal variceal hemorrhage. Other patients present only with intermittent abdominal pain. Pruritus is common.

On physical examination, the liver is frequently enlarged. Patients with renal involvement may also have enlarged kidneys, which may be palpable. Laboratory studies typically show an elevation of serum alkaline phosphatase, direct bilirubin, and a leukocytosis with a predominance of neutrophils. Hepatic synthetic function is well preserved initially, but may be affected by progressive liver damage due to recurrent cholangitis and biliary obstruction. Coagulopathy from vitamin K malabsorption may occur in cholestatic patients (8,9).

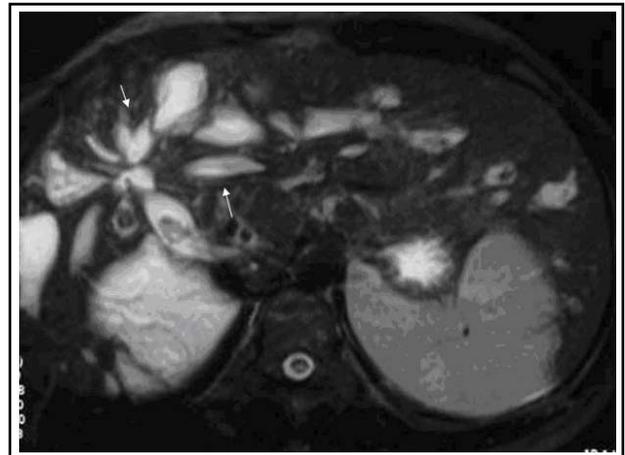


Figure 2. MRCP showing cystic dilatations of the intrahepatic bile duct.

The diagnosis of Caroli's disease and Caroli's syndrome is established by imaging studies that demonstrate bile duct ectasia and irregular, cystic dilation of the large proximal intrahepatic bile ducts with a normal common bile duct. These findings can readily be identified with ultrasonography, endoscopic retrograde cholangiopancreatography, or magnetic resonance imaging which can also demonstrate the renal features of cystic disease (2,6). A liver biopsy is rarely required for the diagnosis.

Renal lesions and choledocal cysts are the associated conditions with Caroli's syndrome. Renal anomalies include renal tubular ectasia (medullary sponge kidney, cortical cyst), adult recessive polycystic kidney disease, and rarely autosomal dominant polycystic kidney disease (10,11). Medullary sponge kidney is described by ectatic and cystic malformations of the collecting ducts and tubules of one or more papillae of one or both kidneys. These anatomic abnormalities produce no symptoms. The morbidity of this disorder is the result from nephrolithiasis and urinary tract infection, both of which are thought to be secondary to the anatomic abnormalities (12).

There is no curative treatment for Caroli's disease. Treatment is mainly supportive and should be individualized. Cholangitis and sepsis are treated with appropriate antibiotics and biliary stone extraction whenever feasible (5). Endoscopic sphincterotomy and stone extraction can be used to remove common duct stones. In contrast, the extraction of intrahepatic stones is far more difficult (5). Patients, who have recurrent attacks of biliary infection, particularly those who also have complications related to portal hypertension, may need liver transplantation (7). Patients who have developed esophageal varices should receive prophylaxis with a non-selective beta blocker (13). A selective shunting procedure can provide relief from portal hypertension if liver function may be well preserved (14). Unexplained clinical deterioration or the appearance of a new biliary stricture should raise a concern that cholangiocarcinoma has developed (15).

Asymptomatic patients with medullary sponge kidney require no specific therapy except to maintain high fluid intake to reduce the risk of nephrolithiasis. Infection should be treated aggressively, and instrumentation of the urinary tract should be minimized to avoid infection. The prognosis is variable depending on the severity of liver disease and the presence of coexisting renal dysfunction. Since the risk of cholangiocarcinoma is increased up to 7 percent, screening for this complication important (16).

Our patient exhibited clinical and radiologic findings for Caroli's syndrome with medullary sponge kidney. The absence of symptom and laboratory finding of hepatic or renal dysfunction enabled us to keep track progress and observe him until the development of complication.

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