Diagnosis and Management of Caroli's Disease along with Autosomal Dominant Polycystic Kidney Disease in a Middle-Aged Male

Aun Ali, Ammara Salam, Madeeha Shahid, Pirhay Fatma and Muhammad Ahmed

Department of Surgery, Fazaia Ruth Pfau Medical College, PAF Base Faisal, Karachi, Pakistan

ABSTRACT

Caroli's disease is a congenital disorder characterised by irregular saccular dilatation of the intrahepatic biliary tree on imaging. Rarely, it has been associated with autosomal recessive polycystic kidney disease (ARPKD) due to a mutation in the *PKHD 1-gene*. We present a case of a middle-aged gentleman who presented with pain in the right-upper abdomen and subclinical jaundice. Magnetic resonance cholangiopan-creatography (MRCP) revealed areas of saccular dilatation of multiple intrahepatic biliary channels along with the central dot sign and bilateral renal cysts. However, the association of autosomal dominant polycystic kidney disease (ADPKD) is extremely rarely reported with Caroli's disease. We propose that the management of Caroli's disease be medically tailored as per the patient's presentation. Surgical intervention is only warranted in case of complications including malignancy.

Key Words: *Autosomal dominant polycystic kidney disease, Caroli's disease, Central dot sign.*

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INTRODUCTION

Caroli's disease is a congenital progressive biliary cystic disease and mostly has an autosomal recessive inheritance. It causes portal hypertension, recurrent cholangitis, intrahepatic stones, cholangiocarcinoma, and liver failure in advanced cases. A potentially effective treatment for advanced Caroli's disease is liver transplantation.[1] Sometimes autosomal recessive polycystic kidney disease (ARPKD) is also present, owing to the genetic mutation in the *PKHD 1-gene,* making the condition more complicated.[2] Even rarer than this condition is Caroli's syndrome, which is congenital hepatic fibrosis, associated with hepatic duct dilatation resulting in early cirrhosis and portal hypertension leading to oesophageal varices and variceal bleeding.[3] Here-in, we present a case of Caroli's disease along with autosomal dominant polycystic kidney disease (ADPKD) in a middle-aged male, an extremely rare association, which was managed medically without surgical intervention.

CASE REPORT

A 42-year healthy gentleman presented with recurrent episodes of non-radiating right hypochondriac pain for the past three months. The pain typically started at night or following a heavy meal.

Correspondence to: Dr. Madeeha Shahid, Department of Surgery, Fazaia Ruth Pfau Medical College, PAF Base Faisal, Karachi, Pakistan
E-mail: madeeha_87s@hotmail.com

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There was no associated weight loss, fever, nausea, or vomiting. His past medical history included a hospital admission six months prior for a similar complaint. No other significant history was elicited. Only mild icterus in the eye was noted on general physical examination. Rest of the systemic examination including the abdomen was normal.

Laboratory findings showed mild anaemia. The haemoglobin was 12.2 g/dl. Total bilirubin level was 40 umol/L, which dropped down to 15 umol/l the following week. Apart from these, other investigations including haematologic, coagulation indices as well as renal biochemistry were normal. Ultrasound abdomen revealed a normal-walled distended gallbladder with calculi, the largest stone measuring 1 cm. Few intrahepatic dilated channels were detected. No feature of hepatic fibrosis was noted. Magnetic resonance cholangiopan-creatography (MRCP) showed areas of saccular dilatation of multiple intrahepatic biliary channels in both hepatic lobes more on the left side. A central dot sign was demonstrated in some of these dilated ducts which confirmed the diagnosis of Caroli's disease (Figure 1). Calculi in the gallbladder were seen. Multiple small cysts were detected in the kidneys bilaterally (Figure 2). Extrahepatic ducts were normal. Based on these findings, diagnosis of symptomatic cholelithiasis with evidence of Caroli's disease and early features of ADPKD was made.

The patient was treated conservatively with intravenous antibiotics, antispasmodic agents, proton pump inhibitors, and fluids for symptomatic relief. He remains in follow-up to monitor any progression of disease and development of complications. To date, he is doing well.

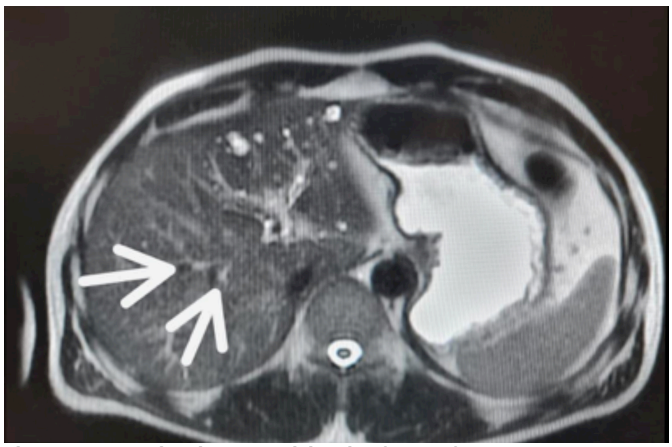

Figure 1: MRCP showing central dot sign (arrows).

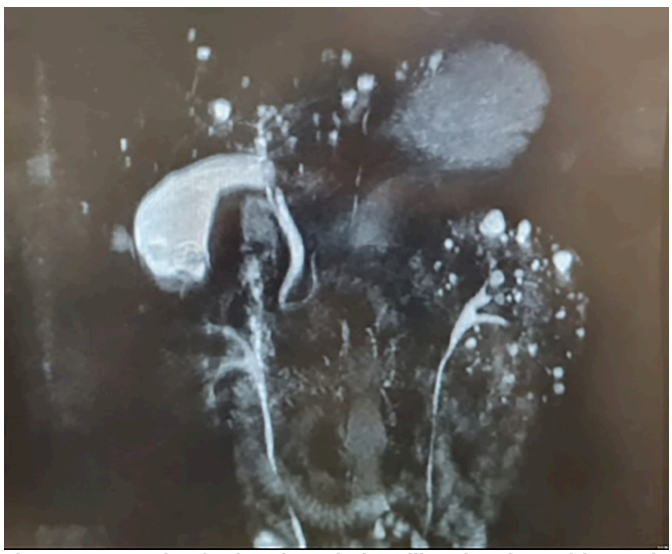

Figure 2: MRCP showing intrahepatic duct dilatation along with cysts in both kidneys.

DISCUSSION

Todd reported the first case of Caroli's disease in 1818. Later, in 1958, Jaques Caroli provided a true description of the Caroli's disease. Type V choledochal cyst in the Todani classification is Caroli's disease. The actual incidence of Caroli's disease is unknown, but estimates range from 1: 100,000 to 1: 1,000,000.[4] The prevalence in males is higher and presents before adulthood.[5]

Caroli's syndrome has often been associated with ARPKD as both are autosomal recessive disorders owing to the genetic mutation in the polycystic kidney and hepatic disease (*PKHD 1) gene*.[2] However, ADPKD has been rarely reported with Caroli's disease. Hasegawa *et al*. reported that only four cases of ADPKD associated with Caroli's disease have been published so far, this case making it to the fifth.[6]

Caroli's disease is a result of ductal plate malformation in the first gestational week, which leads to defects in the biliary channels and is characterised by the segmental saccular dilatations of intrahepatic bile ducts, which can be diffuse or localised to a segment and are connected with the biliary channels.[7] This differentiates it from the simple hepatic cysts present in ADPKD. It is associated with an increased incidence of cholangitis and intrahepatic abscess formation. Many factors are responsible for this malformation including neonatal occlusion of a hepatic artery, aberrant rate of growth of biliary epithelium and surrounding parenchyma, and the inability of the ductal plates to involute causing cyst formation around the portal triads.[8] Certain genetic mutations on chromosome 6p21, *PKHD 1-gene*, that are responsible for fibrocystic protein synthesis, account for these malformations of kidneys and liver.[9]

Early diagnosis of Caroli's disease is critical as partial hepatectomy can cure the patient if the disease is limited to a single lobe. Delayed diagnosis will lead to disease progression, adversely affecting the prognosis. In the present case, the patient presented with biliary colic which settled on conservative management. Non-invasive radiological investigations (CT or MRI) are ideal for the diagnosis of Caroli's disease. These show areas of saccular dilatations of intrahepatic biliary channels along with a central dot sign. While not pathognomonic, the central dot sign is highly suggestive of Caroli's disease. This shows that the dark area is continuous with the remaining bile ducts and appears as a round area inside the liver, inside which is visible a whitish dot representing the entire surrounding of contrast within the portal vein branch by abnormally dilated bile ducts.[5] Early asymptomatic radiological features of ADPKD were also reported so no intervention was needed. The complicated disease needs to be dealt with surgically.[1]

It is concluded that the literature review does not reveal guidelines for the management of cholelithiasis in the presence of asymptomatic Caroli's disease. Prognosis varies according to the severity and associated renal dysfunction. We recommend symptomatic treatment for Caroli's disease with ADPKD.

PATIENT'S CONSENT:

The patient provided written informed consent.

COMPETING INTEREST:

The authors declared no conflict of interest.

AUTHORS' CONTRIBUTION:

AA: Substantial contributions to the conception and design of the work.

AS: Substantial contributions of analysis and interpretation of data for the work.

MS: Drafting of the work and revising it critically for important intellectual content, and agreement to be accountable for all aspect of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

PF: Drafting of the work and revising it critically for important intellectual content.

MA: Final approval of the version to be published.

All authors approved the final version of the manuscript to be published.

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