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RESEARCH ARTICLE

"Caroli's syndrome associated with medullary sponge kidney disease and nephrocalcinosis-A case report."

Dr Rajesh Kumar Saket, Dr Divyashree, Dr Rashmi Rekha, Dr Pallovie Singh

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Corresponding Author

Dr Rajesh Kumar Saket

Abstract

Caroli's disease is an autosomal recessive condition characterized by non-obstructive dilatation of intrahepatic biliary ducts. The exact etiology is unclear. Two variants of Caroli's disease are- type I is simple form; in which bile ducts are dilated without hepatic fibrosis and the type II which is associated with congenital hepatic fibrosis along with its sequelae, also known as Caroli's syndrome. The importance of recognizing this disease as a cause of biliary stasis lies in fact that it is frequently associated with recurrent cholangitis, liver abscesses, cirrhosis and cholangiocarcinoma.

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INTRODUCTION

Caroli's disease or communicating cavernous ectasia of the intrahepatic bile ducts is a rare autosomal recessive disorder which occurs due to an aberration in the development of the intrahepatic bile ducts secondary to intrauterine vascular insult[1]. The possible cause has been cited an arrest of or a derangement in the normal embryologic remodeling of ducts and causes varying degrees of destructive inflammation and segmental dilatation. If the large intrahepatic bile ducts are affected, the result is Caroli's disease, whereas abnormal development of the small interlobular bile ducts results in congenital hepatic fibrosis. If all levels of the biliary tree are involved, features of both congenital hepatic fibrosis and Caroli's disease are present. This condition has been termed "Caroli's syndrome." Caroli's syndrome is associated with renal cystic dilatations in 60-80% of patients. Medullary sponge kidney is the most frequently noted kidney disorder[2]. We report here a case of Caroli's syndrome with medullary sponge kidneys and nephrocalcinosis. The classical "central dot sign" described on CT is well demonstrated easily on ultrasound.

Case Report:

A nine month old male infant presented with distention of abdomen, failure to gain weight, underdeveloped mile stone and irritability. There was no history of fever or jaundice. On examination, he was afebrile, tachypneic, pale, and irritable with abdominal distension and hepatomegaly. Other systems were within normal limits.

On ultrasound of the abdomen, multiple dilated cystic anechoic structures (Fig-1) were seen in the both lobe of the liver. These were communicating with the biliary tree. A small enchogenic dot representing portal vein branch was seen in the center of some of these cystic dilatations giving the typical 'central dot' sign. On Doppler imaging (Fig-2), the portal vein branches were accompanying these dilated ducts. There was mild hepatomegaly with coarse echotexture of liver suggestive of diffuse liver parenchymal disease. The common hepatic duct, common bile duct and gall bladder were normal. Bilateral kidneys were enlarged in size and echogenic with few discrete calcific foci (Fig-3). There was mild ascites.

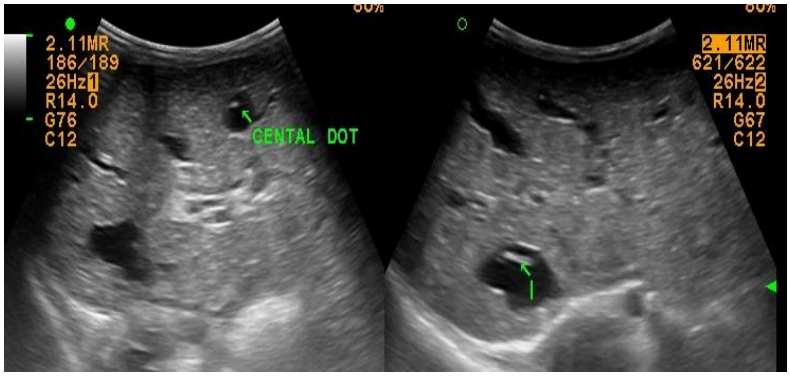


Fig (1) USG abdomen showing multiple dilated cystic anechoic structures were seen in the both lobe of the liver with the typical 'central dot' sign (arrows).

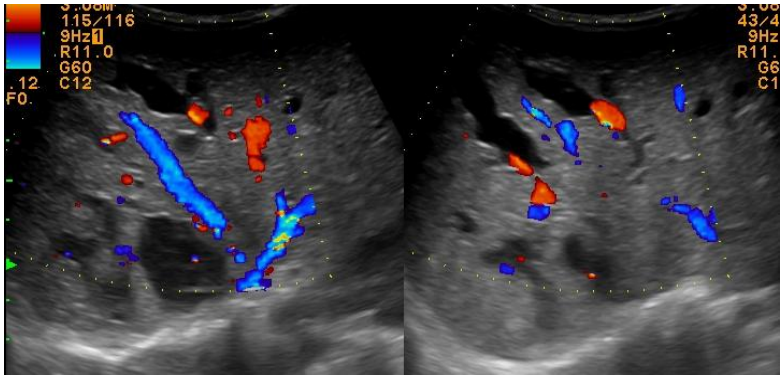
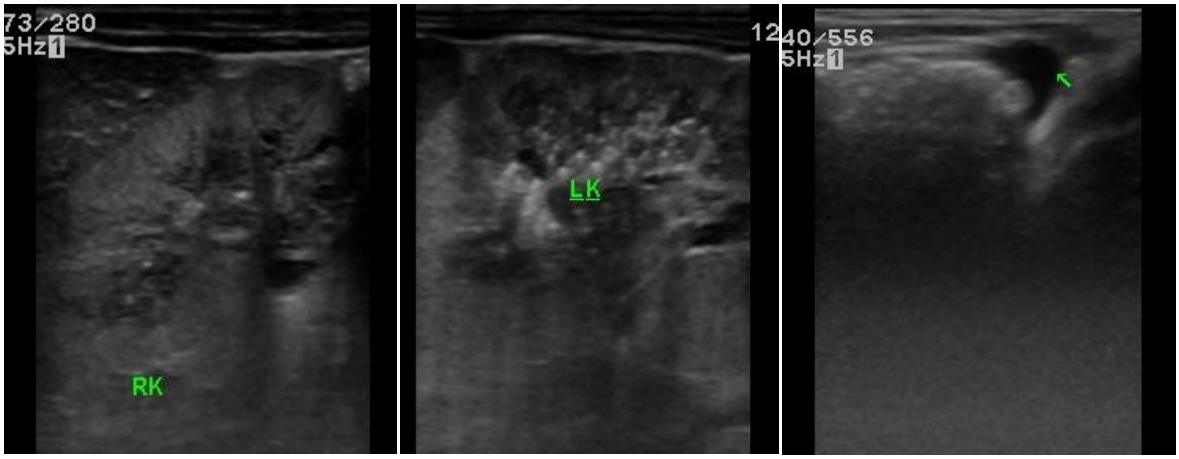
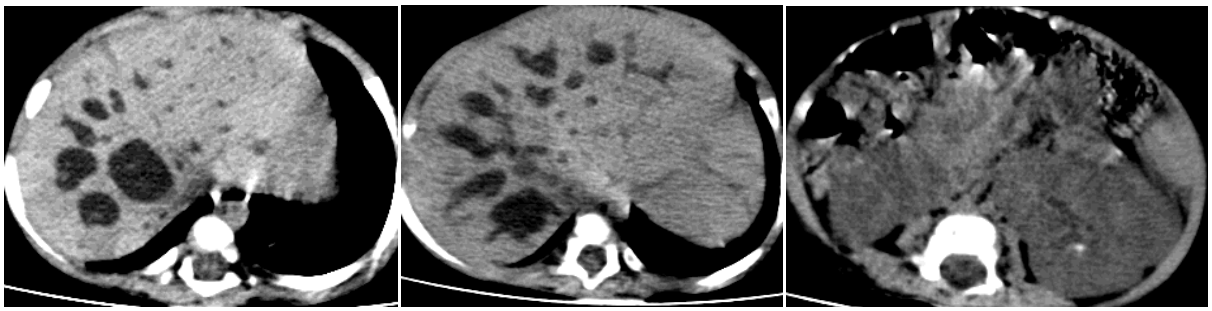


Fig (2) Doppler USG showing, the portal vein branches were accompanying dilated intrahepatic ducts.



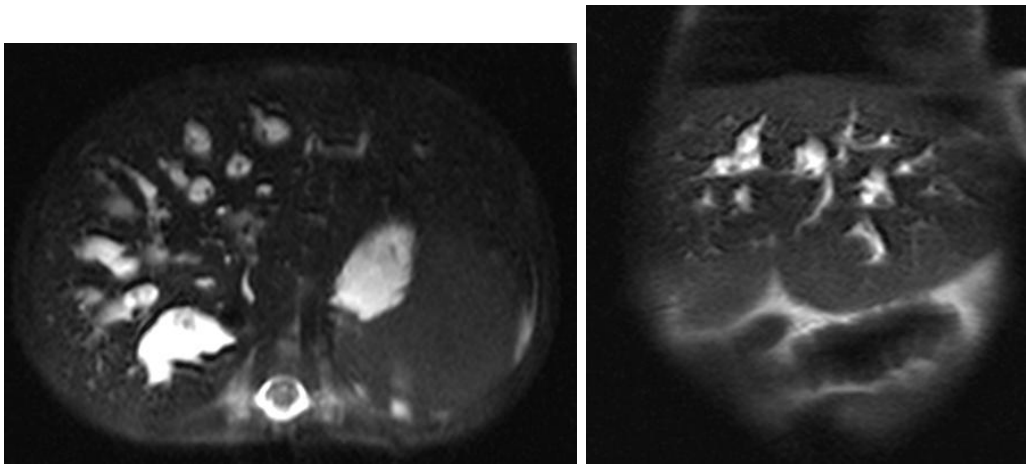
Fig(3) Bilateral enlarged and echogenic kidneys with few calcific foci and mild asites(arrow).

CT scan of abdomen showed saccular dilatation of intrahepatic biliary radicals. A small hyperdense dot representing portal vein branch was seen in the center of some of these cystic dilatations giving the typical 'central dot' sign. Bilateral kidneys were enlarged in size with few calcific foci in the medulla consistent with nephrocalcinosis (Fig-4).

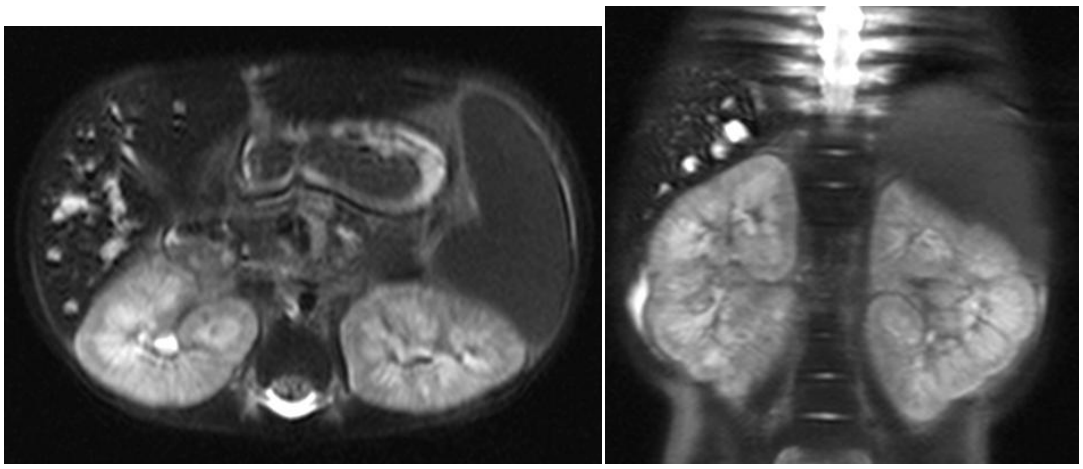


Fig(4)(a,b,c)Non contrast CT abdomen showed saccular dilatation of intrahepatic biliary radicals with 'central dot' sign and nephrocalcinosis.

Magnetic resonance cholangiopancreatography (MRCP) was performed, which showed (Fig-5) multiple cystic dilatation of the intrahepatic duct with 'central dot' sign. The common bile duct and extrahepatic bile ducts were normal. Bilateral kidneys were enlarged with multiple cystic changes(Fig-6).
The final diagnosis Caroli's syndrome with bilateral medullary sponge kidneys and uncomplicated nephrocalcinosis was made.



Fig(5) T2W MR (a-axial and b coronal) images showing multiple dilated intrahepatic ducts with 'central dot' sign.



Fig(6) T2W MR (a-axial and b coronal) images showing enlarged bilateral kidneys with multiple cystic changes.

Discussion:

Caroli's disease is one of the rare and complex autosomal recessive disorders resulting from the aberrant development of the intrahepatic bile ducts. It was first described by Caroli et al. in 1958[3]. The estimated incidence is

1 in 1,000,000 population. There is no sex predilection[4]. 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 [5]. Caroli's disease may be localized to one lobe of liver or may be diffuse.

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[6,7]. 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.

Disease manifestations of Caroli's syndrome usually appear in childhood and young adults, but may also go unrecognized the whole life. The patients with Caroli's disease and congenital hepatic fibrosis may reveal features of portal hypertension like ascites and esophageal variceal hemorrhage[4]. The significance of recognizing Caroli's disease as a cause of biliary stasis is its frequent association with various complications. The common ones are stone formation and cholangitis. Bile duct dilatation leads to bile stagnation and then to lithiasis. Recurrent cholangitis is the primary cause of morbidity and mortality. Less frequent complication is cholangiocarcinoma with an incidence of 7–14%[8].

With rapid advances in non-invasive techniques like Ultrasonography (USG), Computed Tomography (CT) and Magnetic resonance imaging (MRI), Caroli disease is more frequently diagnosed nowadays. USG is the initial investigation of choice. On ultrasonography, the pure form shows diverticulum-like sacculi of intrahepatic biliary tree, more pronounced towards the center which can be segmental or generalized.
The key to the diagnosis of Caroli's disease and Caroli's syndrome lies in demonstrating the communication between the dilated intrahepatic biliary duct (IHBD) segments and the rest of the biliary system. This finding enables us to distinguish Caroli disease from polycystic liver disease and biliary hamartomas. The central dots correspond with fibrovascular bundles surrounded by dilated bile ducts. These bundles contain a portal vein radicle and accompanying hepatic artery branch.
The combination of endoscopic retrograde cholangiopancreatography (ERCP) and USG leads to a more reliable diagnosis to the extent of the disease which is essential for appropriate management[2]. CT serves as a useful preoperative roadmap if insufficient data is obtained on USG, however with advent of MRI and MRCP (Magnetic Resonance Cholangiopancreatography), invasive procedures like ERCP and biopsy are rarely indicated[5].

The differentials to be considered are primary sclerosing cholangitis (PSC), recurrent pyogenic cholangitis, polycystic liver disease, biliary papillomatosis and sometimes obstructive dilatation[4]. Biliary ductal dilatation, stenosis, intrahepatic calculi and malignancy may be associated with Caroli's disease as well as with PSC, however, in PSC the dilatation is rarely saccular and is typically more isolated and fusiform which is not the characteristic of Caroli's disease. All these biliary changes are also seen with recurrent pyogenic cholangitis and this differential is difficult to exclude on imaging alone. However, in recurrent pyogenic cholangitis the patient is septic while it is not so in simple Caroli's disease. . In polycystic hepatic disease, the cysts do not communicate with bile ducts[8].

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[2].

Our patient exhibited clinical and radiologic findings for Caroli's syndrome with medullary sponge kidney.

Conclusion:

Clinical presentation and course of Caroli's syndrome is highly variable and symptoms may appear early or late during life. Because of the slow and usually silent progress of Caroli's syndrome along with its rarity and fatal complications, it should be considered in the differential diagnosis of recurrent cholangitis of unknown cause.

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