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Caroli Syndrome Associated with Polycystic Kidney Disease: A Case Report

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Abstract **Case Report**

Introduction*:** Caroli disease is a fibrocystic liver disease characterized by congenital nonobstructive cystic dilatation of the intrahepatic bile ducts. Its association with congenital hepatic fibrosis defines Caroli syndrome. The more common Caroli syndrome is associated with cystic renal dilatations in 60-80% of patients with polycystic kidney disease and Cacchi-Ricci disease as the most frequently noted renal anomalies. ***Objective*:** We want to define this uncommon entity and to describe the radiological characteristics which can, in the absence of their knowledge, be a source of confusion for the radiologist and pose problems of diagnostic delay. ***Case Report: The patient was a male infant, 1 year and 10 months old, with a history of 2 episodes of urinary tract infections at the age of 3 months and 6 months, and a 13-year-old sister with polycystic hepatorenal disease. The patient was admitted to the pediatric emergency department for acute fever associated with fluid diarrhea evolving for 3 days and irritability and crying at the time of urination. A cytobacteriological examination of the urine showed a turbid appearance of the urine with isolation of Escherichia Coli on culture. Abdominal ultrasound revealed bilateral nephromegaly, with diffuse microcystic hyperechogenicity of medullary pyramids generating multiple comet tail images with a "sponge kidney" appearance as well as a homogeneous splenomegaly and a normal sized liver of heterogeneous echostructure with cystic dilatation of the intrahepatic bile ducts which are centered by hyperechoic punctuations realizing the "Dot Sign". The individualization of this sign at an early age associated with splenomegaly allowed the diagnosis of a Caroli syndrome. While the diffuse medullary hyperechogenicity in the kidney allowed to evoke its association with polycystic kidney disease, especially the recessive one. ***Discussion and Conclusion***: Caroli syndrome is a rare entity of autosomal recessive transmission. It poses a double diagnostic and therapeutic problem. Its rarity and the absence of specific clinical signs often lead to misdiagnosis and thus to a delay in treatment. The diagnosis must be made in the presence of a non-obstructive communicating dilatation of the bile ducts and the demonstration of the "Dot sign" on ultrasound. MRI is essential to characterize and specify the topography of the dilatation and to show the communication with the bile ducts. The clinical data, in particular the young age of discovery and family history, as well as morphological abnormalities such as congenital hepatic fibrosis, PH and associated renal anomalies, allow the differentiation of Caroli syndrome from Caroli disease.

Keywords: Caroli disease, fibrocystic, cytobacteriological examination, hyperechogenicity, fibrosis.

INTRODUCTION

Caroli disease is a fibrocystic liver disease characterized by congenital nonobstructive cystic dilatation of the intrahepatic bile ducts. Its association with congenital hepatic fibrosis defines Caroli syndrome [1].

They are rare entities (<1/10000 in the general population) with autosomal recessive transmission [2]. They are due to an embryologic abnormality in the development of the ductal plate.

The more common Caroli syndrome is associated with cystic renal dilatations in 60-80% of patients with polycystic kidney disease and Cacchi-Ricci disease as the most frequently noted renal anomalies [3].

We report the case of an infant, in whom the diagnosis of Caroli syndrome associated with polycystic kidney disease was first retained in view of the clinical data and the characteristic ultrasound appearance.

We want to define this uncommon entity and to describe the radiological characteristics which can, in the absence of their knowledge, be a source of confusion for the radiologist and pose problems of diagnostic delay.

CASE REPORT

The patient was a male infant, 1 year and 10 months old, with a history of 2 episodes of urinary tract infections at the age of 3 months and 6 months, and a 13-year-old sister with polycystic hepatorenal disease.

The patient was admitted to the pediatric emergency department for acute fever associated with fluid diarrhea evolving for 3 days and irritability and crying at the time of urination.

A cytobacteriological examination of the urine (ECBU) showed a turbid appearance of the urine with isolation of Escherichia Coli on culture.

Abdominal ultrasound revealed bilateral nephromegaly, with diffuse microcystic hyperechogenicity of medullary pyramids generating multiple comet tail images with a "sponge kidney" appearance (Figure 1 and 2), as well as a homogeneous splenomegaly and a normal sized liver of heterogeneous echostructure with cystic dilatation of the intrahepatic bile ducts which are centered by hyperechoic punctuations realizing the "Dot Sign" (Figure 3).

The individualization of this sign at an early age associated with splenomegaly allowed the diagnosis of a Caroli syndrome. While the diffuse medullary hyperechogenicity in the kidney allowed to evoke its association with polycystic kidney disease, especially the recessive one.

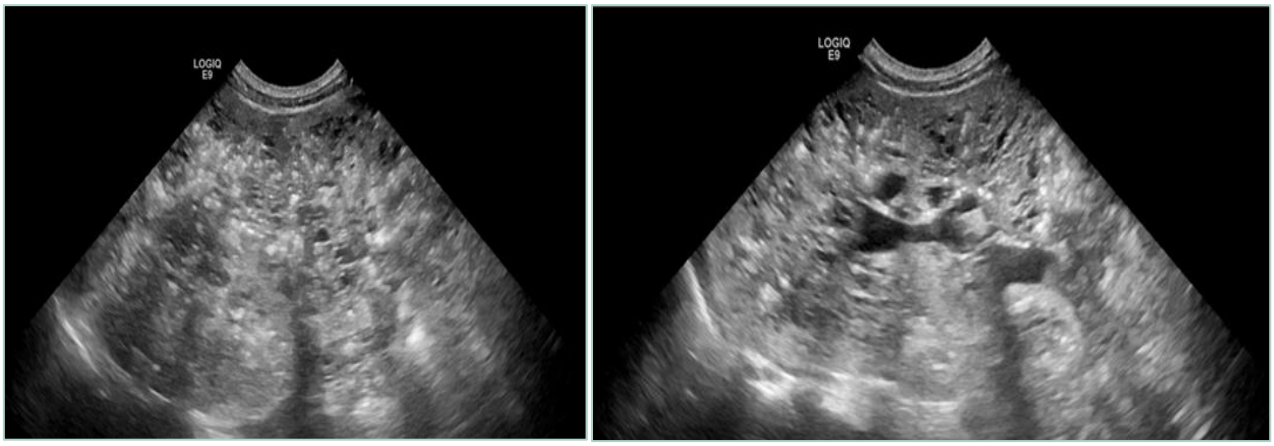

Figure 1: Longitudinal sections of the right kidney: Nephromegaly with diffuse microcystic hyperechogenicity of medullary pyramids generating multiple comet-tail images creating a "sponge kidney" appearance

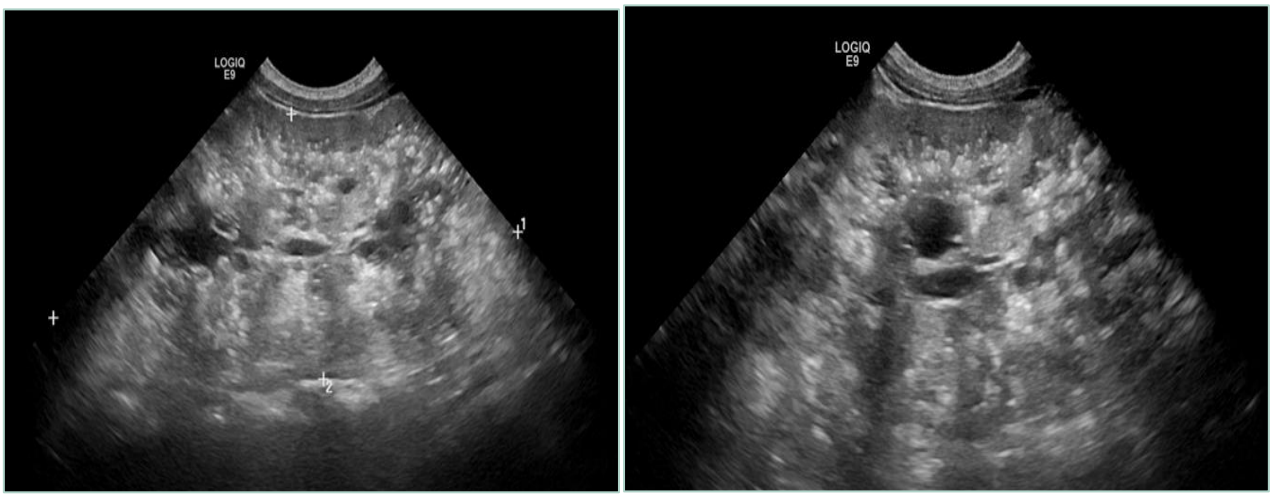

Figure 2: Longitudinal sections of the left kidney: Nephromegaly with diffuse microcystic hyperechogenicity of medullary pyramids generating multiple comet tail images giving a "sponge kidney" appearance

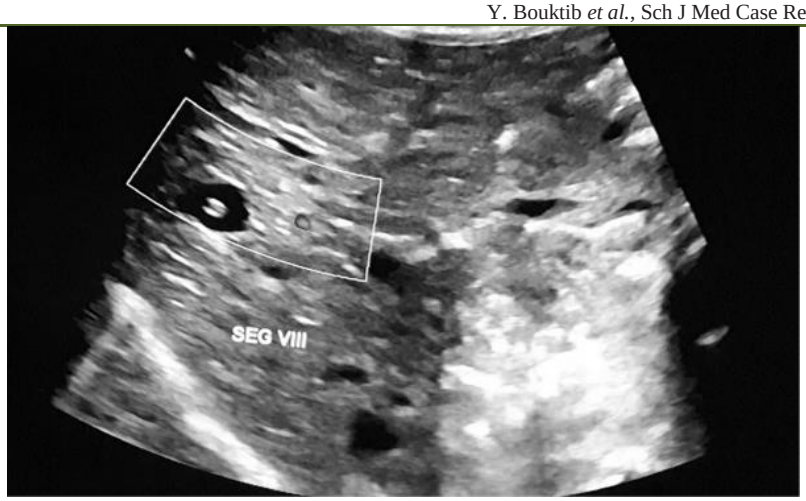

Figure 3: Cystic dilatation of the intrahepatic bile ducts centered by a hyperechoic punctation realizing the "Dot Sign" aspect

DISCUSSION

First described in 1958 by Caroli *et al*., Caroli disease (CD) is a rare congenital fibrocystic liver disorder defined by diffuse (in 75% of cases) or localized (most often left lobe) segmental dilatation of the intrahepatic bile ducts [4, 5]. When the diffuse form is associated with congenital liver fibrosis complicated by cirrhosis or portal hypertension, it is called Caroli syndrome (CS) which is autosomal recessive unlike CD which is non-hereditary. The syndrome can be associated with polycystic kidney disease and Cacchi-Ricci disease. Its association with a bile duct cyst is uncommon, unlike CD [1, 2].

Despite its presence at birth and its familial form, the diagnosis is often made late because of the absence of specific clinical signs. The discovery of HC may be made less frequently at the stage of complications, such as intrahepatic lithiasis, angiocholitis, hepatic abscesses, PH and complications of associated diseases including chronic renal failure. A formidable but rare complication is the degeneration into cholangiocarcinoma, which can sometimes be revealing [3]. It occurs in 7 to 10% of cases in less than 14 years according to Etienne *et al*., [6] and patients with this entity have a risk of developing cholangiocarcinoma that is 100 times higher than that of the general population [7].

The diagnosis of this entity can be suggested by abdominal ultrasound data showing communicating, non-obstructive cystic saccular or fusiform dilatation of the intrahepatic bile ducts. It also allows to search for intrahepatic lithiasis with a high sensitivity. The most characteristic but inconstant sign is the "Dot sign" which corresponds to the image of a portal vessel protruding into the lumen of the ectatic bile duct. It can be visualized also by CT and MRI. It is mainly encountered in Caroli disease or syndrome but it is non-specific. Indeed, it is pathognomonic of ductal plate abnormalities and it also appears sporadically in other conditions such as peribiliary cysts, periportal lymphedema.

On ultrasonography, it appears as echogenic spots or lines (depending on the cross-sectional plane) surrounded by hypoechoic dilatations of cystic intrahepatic bile ducts [8]. On CT and MRI, the sign is usually best seen in axial section. The "dot" represents the cross section of a branch of the portal vein, completely surrounded by the abnormally dilated bile ducts. If intravenous contrast is used, the central branches of the portal vein are enhanced.

Bili-MRI, a non-invasive examination, is the reference examination which has the great advantage of avoiding direct opacifications of the bile ducts, which are often the source of serious iatrogenic angiocholitis, and of not requiring the use of an intravenous contrast product, which allows it to be performed even in cases of renal insufficiency. It visualizes the entire biliary tree and gives a better biliary cartography, highlights cystic dilatations of the proximal intrahepatic bile ducts as well as communications of cysts with the bile ducts, "Dot sign", and intrahepatic stones [9].

The various imaging modalities also demonstrate associated renal lesions. These abnormalities include Cacci-Ricci disease (dilatation of the medullary collecting tubes in the pericalicalical regions.), recessive polycystic disease and, rarely, autosomal dominant polycystic disease [10, 11].

Recessive polycystic kidney disease is defined by the association of bilateral nephromegaly, cystic dilatation of the collecting tubes in a corticomedullary radial arrangement and hepatic involvement with periportal fibrosis and ectasia of the intrahepatic bile ducts. It is most often lethal in the neonatal period due to pulmonary hypoplasia, renal failure and hepatic fibrosis.

There is no curative treatment for Caroli syndrome. Management depends on the clinical presentation: it can be conservative (antibiotics, ursodeoxycholic acid, biliary drainage) or surgical (including, as a last resort, liver transplantation) especially in diffuse forms.

The prognosis is variable and depends on the frequency and severity of episodes of cholangitis, the presence of associated diseases and the increased risk of biliary cancer.

CONCLUSION

Caroli syndrome is a rare entity of autosomal recessive transmission. It poses a double diagnostic and therapeutic problem. Its rarity and the absence of specific clinical signs often lead to misdiagnosis and thus to a delay in treatment.

The diagnosis must be made in the presence of a non-obstructive communicating dilatation of the bile ducts and the demonstration of the "Dot sign" on ultrasound. MRI is essential to characterize and specify the topography of the dilatation and to show the communication with the bile ducts. The clinical data, in particular the young age of discovery and family history, as well as morphological abnormalities such as congenital hepatic fibrosis, PH and associated renal anomalies, allow the differentiation of Caroli syndrome from Caroli disease.

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