Scholars Journal of Medical Case Reports
Abbreviated Key Title: Sch J Med Case Rep
ISSN 2347-9507 (Print) | ISSN 2347-6559 (Online)
Journal homepage: https://saspublishers.com

OPEN ACCESS

Radiology

Complications of Caroli's Disease: Two Case Reports and a Literature Review

B. Slioui[1*], S. Belassri[1], N. Hammoune[1], A. Mouhssine[1]

[1]Radiology Department, Avicenne Military Hospital, FMPM, Cadi Ayad University, Marrakech, Morocco

DOI: https://doi.org/10.36347/sjmcr.2024.v12i12.003 | **Received:** 30.10.2024 | **Accepted:** 04.12.2024 | **Published:** 06.12.2024

***Corresponding author:** B. Slioui
Radiology Department, Avicenne Military Hospital, FMPM, Cadi Ayad University, Marrakech, Morocco

Abstract **Case Report**

Caroli's disease is a rare congenital disorder of the bile ducts, characterized by intrahepatic segmental dilatation. It can remain asymptomatic for many years. Imaging, especially hepatobiliary MRI, plays a central role in diagnosis, identifying pathognomonic signs (Dot-sign, cyst communication with bile ducts), and monitoring complications. Treatment primarily involves surgery, ranging from partial resection to liver transplantation for diffuse forms. This article presents two cases: one involving an association with cholangiocarcinoma and another complicated by intrahepatic stones and cholangitis. These cases highlight the major complications, diagnostic approaches, and therapeutic strategies for this rare condition.

Keywords: Caroli's disease - Hepatic MRI – Cholangiocarcinoma – intrahepatic lithiasis - Cholangitis.

Copyright © 2024 The Author(s): This is an open-access article distributed under the terms of the Creative Commons Attribution **4.0 International License (CC BY-NC 4.0)** which permits unrestricted use, distribution, and reproduction in any medium for non-commercial use provided the original author and source are credited.

INTRODUCTION

Caroli's disease is a congenital anomaly of the bile ducts, defined by focal or segmental dilatation of the intrahepatic ducts and frequently associated with severe complications. It can remain asymptomatic for many years.

Although rare, with a prevalence of approximately 1 in 1,000,000 [1], this condition presents diagnostic and therapeutic challenges due to potentially severe complications, such as intrahepatic stones, cholangitis, and cholangiocarcinoma.

This article reports two illustrative cases, emphasizing the critical role of imaging in recognizing pathognomonic signs and monitoring patients, while discussing clinical implications and therapeutic options, ranging from hepatic resection to transplantation. These observations underscore the complexity of managing this disease and the importance of early diagnosis to improve outcomes.

Case 1

A 40-year-old patient under follow-up for cystic formations diagnosed as hepatic cysts was referred for evaluation of a hepatic mass discovered during routine ultrasound.

Hepatobiliary MRI Findings:

The MRI included T2 FS axial and coronal sequences (Figure 1), diffusion imaging with multiple gradients (400 and 800) (Figure 2) and ADC mapping, Bili-MRI (Figure 3), and T1 FS Gadolinium during portal and delayed phases (Figure 4).

Analysis revealed multiple diffuse cystic formations in the liver, hypointense on T1, strongly hyperintense on T2 (Figure 1), with no restriction on diffusion imaging, thin-walled, and non-enhancing across all phases. Communication with bile ducts was noted on Bili-MRI (Figure 3), and the Dot-sign was observed during the portal phase (Figure 4).

A mass in segment IV was hypointense on T1, hyperintense on T2, with diffusion restriction (Figure 2). It showed peripheral enhancement with internal septal enhancement during the delayed phase, suggesting a fibrous lesion (Figure 4).

Given the communication of the cystic lesions and the presence of the Dot-sign, the diagnosis of Caroli's disease was established. The segment IV mass was likely cholangiocarcinoma based on its fibrous nature. The patient was referred for surgical management.

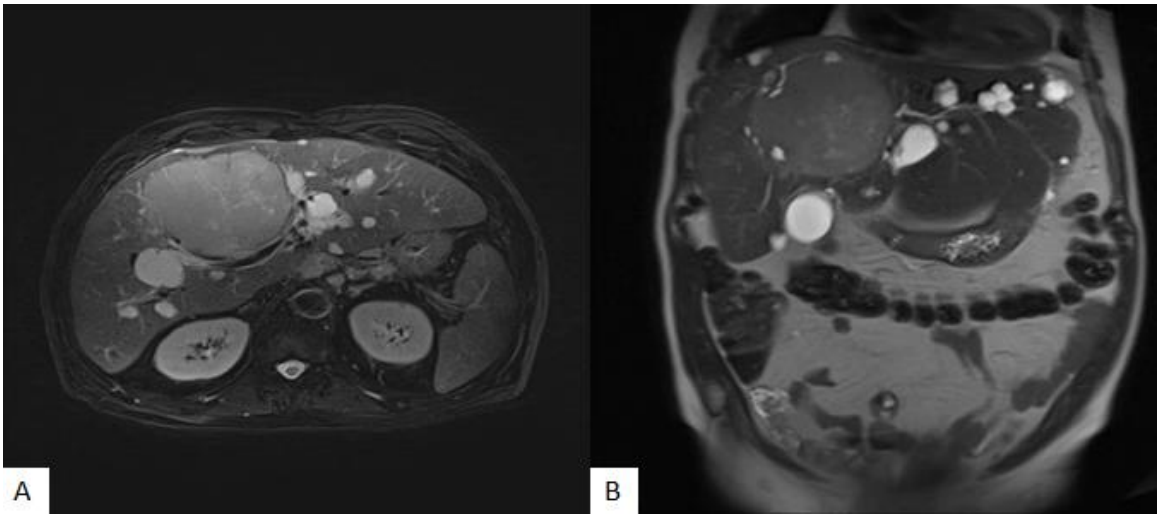

Figure 1: Liver MRI in axial T2 sequences (A) and coronal T2 (B) showing the presence of a tissue mass in segment IV with multiple cystic lesions of the liver

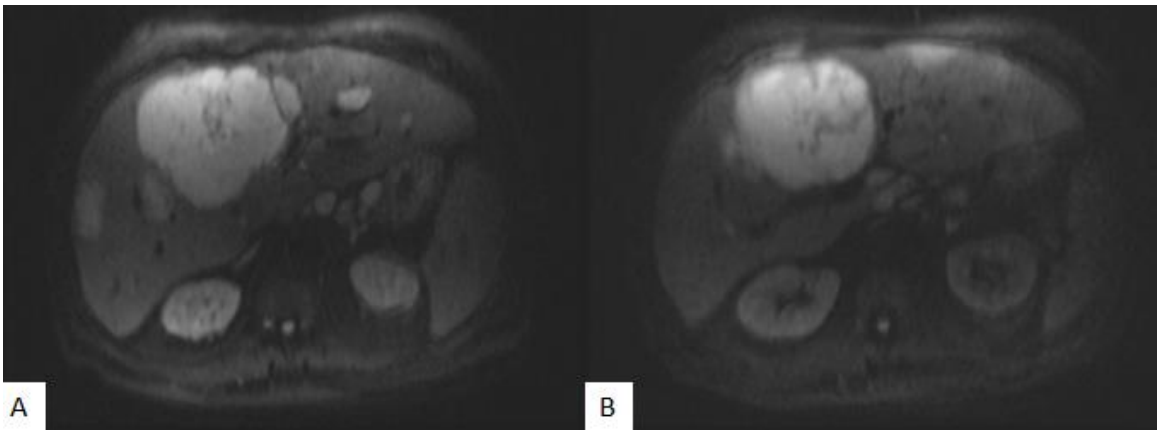

Figure 2: Liver MRI in diffusion sequences B400 (A) and B800 showing restriction of the mass

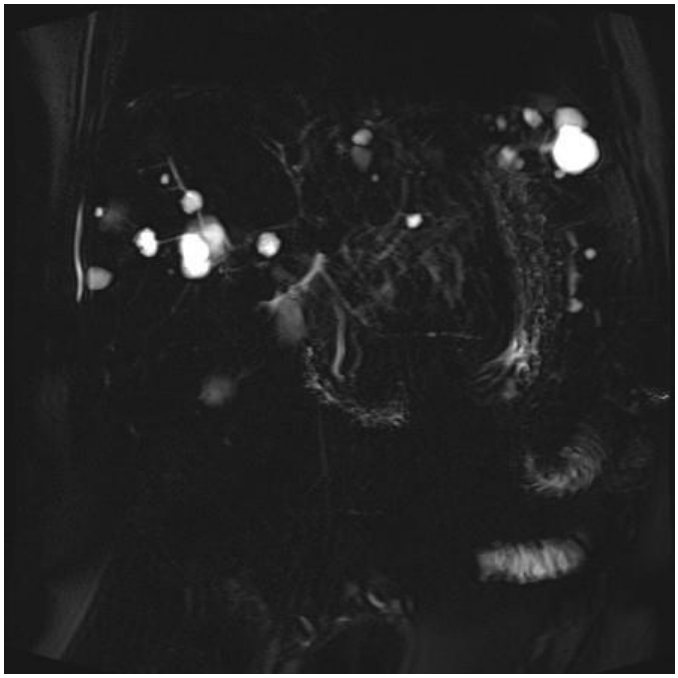

Figure 3: Liver MRI in BILI-MRI sequences demonstraiting cyst communication with the intrahepatic bille ducts (IHBD)

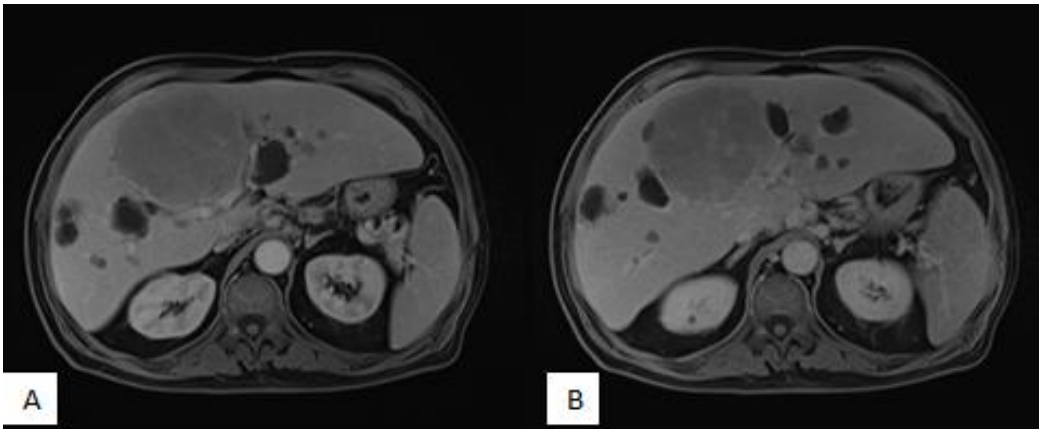

Figure 4: Hepatic MRI in T1 sequences with gadolinium at early (A) and late (B) post-contrast phases showing the Dot-Sign and late-phase enhancement of the mass

Case 2

A 60-year-old patient with no medical history presented with febrile jaundice. Emergency abdominal ultrasound revealed hepatomegaly with multiple cystic formations, associated with dilatation of the intrahepatic bile ducts (IHBD) (Figure 5) and the common bile duct (CBD) (Figure 6).

Hepatobiliary MRI Findings:

MRI included sequences similar to Case 1. Imaging revealed multiple diffuse cystic formations in the liver, hypointense on T1, strongly hyperintense on T2, without diffusion restriction, and thin-walled with no enhancement. Communication with bile ducts and the Dot-sign were identified on Bili-MRI during the portal phase.

Additionally, the CBD was dilated due to a stone in the distal bile duct (Figure 6), and the IHBD showed multiple intrahepatic stones in the left liver (Figure 5). Portal-phase enhancement of the CBD was consistent with cholangitis.

A cyst in segment VIII exhibited T1 hyperintensity, suggesting hemorrhagic changes. The diagnosis of Caroli's disease complicated by intrahepatic stones, migration syndrome, and hemorrhagic changes in the segment VIII cyst was established.

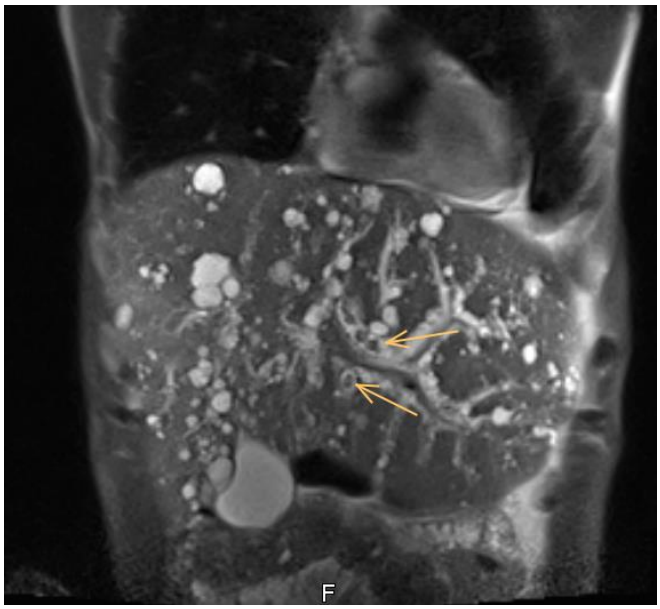

Figure 5: Liver MRI in coronal sequence revealing multiple cystic formations of the liver with dilated intrahepatic bile ducts and intrahepatic stones (arrows)

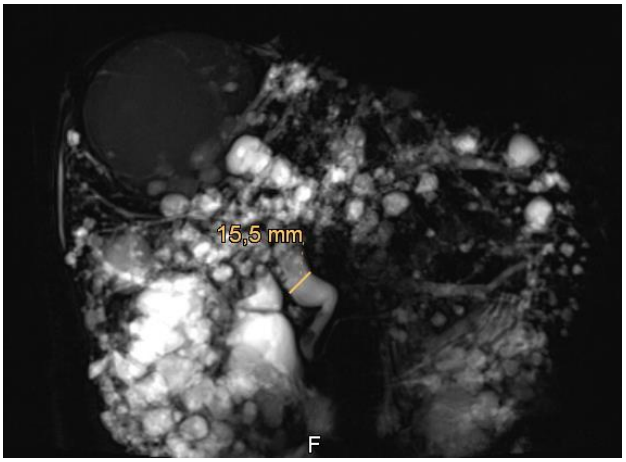

Figure 6: Hepatic MRI with BILI sequences showing dilation of the common bile duct with a stone in the distal commun bile duct

DISCUSSION

Caroli's disease is a rare congenital condition defined by segmental dilatation of intrahepatic bile ducts, which may be diffuse or localized [1].

Two distinct entities are described [2]:

1. Caroli syndrome: Associated with hepatic fibrosis and ectasia of renal tubules, it follows an autosomal recessive inheritance pattern and is the most common form.
2. Caroli disease: True congenital cystic malformations of the bile ducts, not associated with hepatic fibrosis or renal abnormalities.

These cystic malformations are classified into five types by Todani based on location, extent, and type of dilatation [3]. Caroli's disease corresponds to Type V and is the least common (<1%).

Epidemiologically, Caroli's disease has a prevalence of 1 in 1,000,000 with equal sex distribution [1]. Although present from birth, it often remains asymptomatic. In 80% of cases, symptoms manifest before the age of 30 due to secondary angiocholitis from intrahepatic stones, which may progress to abscesses or even secondary amyloidosis [1].

The risk of cholangiocarcinoma is approximately 100 times higher in patients with Caroli's disease compared to the general population, with synchronous prevalence ranging from 7% to 25% in reported series [4].

Imaging is crucial, starting with ultrasound for initial screening and transitioning to MRI as the gold standard for its superior spatial and tissue resolution. Pathognomonic signs include cystic lesion communication with bile ducts, absence of underlying obstruction, and the Dot-sign, indicating arrested remodeling of the ductal plate.

Therapeutic management is surgical: partial hepatectomy for localized forms or liver transplantation for diffuse forms or cases with hepatic fibrosis [5]. Surgery is justified by the risk of malignant transformation, as seen in Case 1.

CONCLUSION

Caroli's disease should be considered in the differential diagnosis of hepatic cysts, which are frequently encountered in clinical practice. Any imaging atypia warrants further investigation, particularly with MRI as the reference modality.

Early diagnosis allows for timely management, reducing the risk of severe complications.

Conflict of Interest: The authors declare no conflicts of interest.

REFERENCE

1. Jarry, J., Leblanc, F., & Saric, J. (2010). Maladie de Caroli monolobaire. *Presse Med*, 39, 847-848.
2. Guettier, C. (2010). Lésions Kystiques biliaires du foie. *Annales de pathologie*, 30, 448-454.
3. Mannai, S., Kraïem, T., Gharbi, L., Haoues, N., Mestiri, H., & Khalfallah, M. T. (2006, July). Les dilatations kystiques congénitales des voies biliaires. In *Annales de chirurgie* (Vol. 131, No. 6-7, pp. 369-374). Elsevier Masson.
4. Mabrut, J. Y., Partensky, C., Jaeck, D., Oussoultzoglou, E., Baulieux, J., Boillot, O., ... & Gigot, J. F. (2007). Congenital intrahepatic bile duct dilatation is a potentially curable disease: long-term results of a multi-institutional study. *Annals of surgery*, *246*(2), 236-245.
5. Ulrich, F., Pratschke, J., Pascher, A., Neumann, U. P., Lopez-Hänninen, E., Jonas, S., & Neuhaus, P. (2008). Long-term outcome of liver resection and transplantation for Caroli disease and syndrome. *Annals of surgery*, *247*(2), 357-364.