ISSN: 3071-2653

ScienceFrontier Open Access

Case Report

International Journal of Public Health Research and Epidemiology

Received: 20 August 2026
Accepted: 28 August 2026
Published:09 September 2026

Petechial Purpura Associated with Renal Failure Revealing Caroli Syndrome

G. Khellaf [1,2]*

[1] *University of Health Sciences Dr. Youcef El Khatib, Faculty of Medicine, Algiers, Algeria*

[2] *Department of Nephrology, Bab El Oued University Hospital, Algiers, Algeria*

***Corresponding Author:** G. Khellaf, *University of Health Sciences Dr. Youcef El Khatib, Faculty of Medicine, Algiers, Algeria*

Citation: G. Khellaf (2026). Pancytopenia Revealing Visceral Leishmaniasis in A Renal Transplant Recipient: A Case Report and Literature Review, J *International Journal of Public Health Research and Epidemiology*; 2 (5) 27, Doi: https://doi.org/10.5281/zenodo.22806047

Abstract:

Introduction: Caroli disease and Caroli syndrome are rare congenital disorders characterized by segmental cystic dilatation of the intrahepatic bile ducts. Caroli syndrome is the more common variant, often diffuse, associated with congenital hepatic fibrosis leading to juvenile portal hypertension and frequently associated with autosomal recessive polycystic kidney disease.

Methods: A 32-year-old patient with no significant medical history presented as an emergency with abdominal pain, petechial purpura of the lower limbs, macroscopic hematuria, and renal failure (creatinine 18 mg/L, creatinine clearance 58 mL/min). Clinical examination revealed hepatosplenomegaly, petechial purpura of the lower limbs, and preserved diuresis with normal blood pressure. Laboratory findings showed isolated peripheral thrombocytopenia, moderate renal failure, cholestasis, and hepatic cytolysis. Abdominal ultrasound with Doppler revealed cystic dilatation of intrahepatic bile ducts, diffuse hepatic fibrosis with portal hypertension, and bilateral renal cortical hyperechogenicity with comet-tail artifacts suggestive of microcysts, confirmed by CT urography. Esophagogastroduodenoscopy revealed grade 1 esophageal varices. The diagnosis of Caroli syndrome was established based on the association of Caroli disease and recessive polycystic kidney disease.

Results: The patient has been regularly followed in our nephrology consultation since 2018. He remains in excellent general condition with stable renal function (creatinine clearance maintained at approximately 58 mL/min). The thrombocytopenia has remained stable without significant progression, and no new episodes of purpura or macroscopic hematuria have occurred. No episodes of cholangitis or hepatic decompensation have been observed during the follow-up period.

Conclusion: The incidental discovery of a congenital disease with a nephrological presentation, such as macroscopic hematuria in adulthood, is rare. Caroli syndrome is a rare disease still unrecognized by many nephrologists. This case demonstrates that long-term clinical stability with preserved renal function and stable thrombocytopenia is possible with conservative management and regular follow-up.

Key words: caroli syndrome; caroli disease; autosomal recessive polycystic kidney disease; purpura; thrombocytopenia; portal hypertension

Introduction:

Caroli disease and Caroli syndrome are rare congenital disorders belonging to the spectrum of hepatic fibrocystic diseases, characterized by multifocal, segmental dilatation of the large intrahepatic bile ducts [1-3]. Caroli disease is defined by bile ductular ectasia without other apparent hepatic abnormalities, while Caroli syndrome is characterized by the association of intrahepatic bile duct dilatation with congenital hepatic fibrosis [1, 2, 4].

The estimated prevalence of Caroli disease is approximately 1 in 1,000,000 individuals, while Caroli syndrome is more common, with an estimated

prevalence of 1 in 100,000 individuals [1, 5]. These conditions are inherited in an autosomal recessive pattern and are strongly associated with renal cystic lesions, particularly autosomal recessive polycystic kidney disease (ARPKD) [1, 2]. Mutations in the PKHD1 gene, which encodes fibrocystin, are responsible for the majority of cases [6]. Fibrocystin is expressed in the liver (cholangiocytes), pancreas, and renal tubular cells, explaining the frequent association between hepatic and renal manifestations [6].

The clinical presentation of Caroli syndrome is highly variable. Patients may remain asymptomatic until the second to fourth decades of life, with symptoms appearing later due to complications [1, 5]. Common manifestations include recurrent cholangitis, hepatolithiasis, portal hypertension, and its complications such as splenomegaly, esophageal varices, and ascites [1, 2, 4]. Renal involvement is frequent, ranging from medullary sponge kidney to autosomal recessive polycystic kidney disease [1, 2, 5].

We report an unusual case of Caroli syndrome revealed in adulthood by petechial purpura, macroscopic hematuria, and renal failure. This case highlights the diagnostic challenges of this rare condition and the importance of considering it in patients with unexplained hepatosplenomegaly and renal abnormalities.

Case Report:

Patient History

A 32-year-old patient with no significant medical history presented as an emergency with abdominal pain, petechial purpura of the lower limbs, and macroscopic hematuria. Laboratory investigations revealed renal failure with creatinine at 18 mg/L and creatinine clearance at 58 mL/min, leading to referral to the nephrology department.

Clinical Examination

Clinical examination revealed a patient in good general condition with preserved diuresis. Blood pressure was normal at 120/70 mmHg. Abdominal palpation revealed hepatosplenomegaly. Petechial purpura was noted on both lower limbs. Urinalysis showed pH 6.5, negative protein, and positive hematuria (++).

Laboratory Findings:

Parameter	Value	Interpretation
Creatinine	18 mg/L	Moderate renal failure
Creatinine clearance	58 mL/min	Stage 3 CKD
Platelets	Low	Isolated peripheral thrombocytopenia
Liver function	Abnormal	Cholestasis with hepatic cytolysis
Peripheral blood smear	Confirmed	Peripheral thrombocytopenia

Imaging Findings:

Abdominal ultrasound with Doppler revealed cystic dilatation of the intrahepatic bile ducts, diffuse hepatic fibrosis with portal hypertension (intrahepatic block), and bilateral renal cortical hyperechogenicity with comet-tail artifacts (Figure 1). CT urography confirmed the presence of microcysts in both kidneys (Figure 2). The presence of the "central dot sign" on imaging was consistent with the diagnosis of Caroli syndrome [5, 7]. Esophagogastroduodenoscopy revealed grade 1 esophageal varices.

Figure 1: Abdominal ultrasound showing cystic dilatation of intrahepatic bile ducts (arrow) and bilateral renal cortical hyperechogenicity with comet-tail artifacts. (Insert Figure 1 here)

Figure 2: CT urography showing microcysts in both kidneys (arrowheads). (Insert Figure 2 here)

Diagnosis

Based on the association of Caroli disease (cystic dilatation of intrahepatic bile ducts) and autosomal recessive polycystic kidney disease (bilateral microcysts), the diagnosis of Caroli syndrome was established [1, 2, 7]

Clinical Course and Follow-Up:

The patient has been regularly followed in our nephrology consultation since 2018. He remains in excellent general condition with stable renal function; creatinine clearance has been maintained at approximately 58 mL/min throughout the follow-up period. The thrombocytopenia has remained stable without significant progression. No new episodes of purpura or macroscopic hematuria have occurred. No episodes of cholangitis or hepatic decompensation have been observed during the follow-up period. Liver function tests remain stable, with no worsening of cholestasis or cytolysis. Esophagogastroduodenoscopy performed during follow-up has shown no progression of esophageal varices, which remain grade 1. The patient continues to be managed conservatively with regular monitoring of renal and hepatic function. This favorable clinical course highlights the possibility of long-term stability in some patients with Caroli syndrome.

Discussion:

Caroli Syndrome: Definition and Classification:

Caroli syndrome is a rare congenital disorder characterized by segmental, non-obstructive, cystic dilatation of the intrahepatic bile ducts associated with congenital hepatic fibrosis [1, 2, 4]. It is distinguished from Caroli disease, which is defined by bile ductular ectasia without other apparent hepatic abnormalities [1, 2]. Caroli syndrome is the more common variant, accounting for the majority of cases, and is frequently associated with autosomal recessive polycystic kidney disease [1, 2, 7].

The condition is inherited in an autosomal recessive pattern and is caused by mutations in the PKHD1 gene, which encodes fibrocystin [6]. Fibrocystin is expressed in the liver (cholangiocytes), pancreas, and renal tubular cells, explaining the frequent association between hepatic and renal manifestations [6]. Mutations in other ciliopathy genes, such as WDR19, have also been associated with Caroli syndrome [5, 6].

Renal Involvement in Caroli Syndrome:

Renal involvement is a hallmark of Caroli syndrome, occurring in a significant proportion of patients [1, 2, 5]. The most common renal manifestation is autosomal recessive polycystic kidney disease, characterized by bilateral enlarged kidneys containing numerous small cysts, typically diagnosed in childhood [1, 7]. However, less severe renal phenotypes can occur, including medullary sponge kidney, nephrocalcinosis, and microcystic disease [1, 5].

In our patient, imaging revealed bilateral renal cortical hyperechogenicity with comet-tail artifacts, suggestive of microcysts. This pattern is consistent with autosomal recessive polycystic kidney disease, although the presentation in adulthood is unusual [1, 7]. The presence of renal failure (creatinine clearance 58 mL/min) and macroscopic hematuria highlights the potential for renal involvement to be the presenting feature of Caroli syndrome. Notably, during follow-up, the patient's renal function has remained stable with creatinine clearance maintained at approximately 58 mL/min, indicating a favorable long-term outcome.

Hematological Manifestations: Purpura and Thrombocytopenia:

Petechial purpura and thrombocytopenia are not classic features of Caroli syndrome but can occur as complications of portal hypertension [1, 4]. Portal hypertension leads to splenomegaly and hypersplenism, resulting in peripheral thrombocytopenia [1]. The Orphanet database lists thrombocytopenia and abnormal bleeding as "very frequent" phenotypic abnormalities in Caroli syndrome [1]. In our patient, the presence of hepatosplenomegaly, esophageal varices, and thrombocytopenia is consistent with the development of portal hypertension.

The purpura observed in our patient may be related to thrombocytopenia or to other mechanisms, such as coagulopathy secondary to liver dysfunction [1]. Notably, during follow-up, the thrombocytopenia has remained stable without significant progression, and no new episodes of purpura have occurred. This case highlights that Caroli syndrome should be considered in patients presenting with unexplained purpura and thrombocytopenia associated with hepatosplenomegaly.

Diagnosis:

The diagnosis of Caroli syndrome is based on a combination of clinical, imaging, and histopathological findings [1, 7]. Imaging, particularly ultrasound, CT, and MRCP, plays a crucial role in diagnosis [1, 7]. The presence of the "central dot sign" on imaging is a hallmark of Caroli disease and Caroli syndrome [5, 7]. This sign corresponds to the portal vein branch surrounded by dilated bile ducts.

In our patient, abdominal ultrasound with Doppler revealed cystic dilatation of the intrahepatic bile ducts and diffuse hepatic fibrosis with portal hypertension. The presence of this combination, along with renal findings, strongly supported the diagnosis of Caroli syndrome [1, 2].

Prognosis and Complications:

The prognosis of Caroli syndrome is variable and depends on the severity of hepatic and renal involvement [1, 4]. Major complications include recurrent cholangitis, hepatolithiasis, portal hypertension with variceal bleeding, and the development of cholangiocarcinoma [1, 4]. Renal involvement may progress to end-stage renal disease [1, 7].

Long-term outcomes in patients with Caroli syndrome are generally worse than in those with Caroli disease alone [4]. In a recent longitudinal cohort study, Caroli syndrome was associated with a higher rate of hepatic decompensation and biliary malignancy compared to Caroli disease [4]. The study showed that Caroli syndrome was associated with a more severe clinical course and poorer outcomes than isolated Caroli's disease [4]. Liver transplantation remains the only curative treatment for advanced disease [1, 7].

However, our patient's favorable clinical course with stable renal function and no progression of complications over several years of follow-up demonstrates that long-term stability is possible in some patients with Caroli syndrome when managed conservatively with regular monitoring.

Implications for Clinical Practice

This case underscores several important points for clinical practice:

- Caroli syndrome is a rare and underrecognized condition: Many nephrologists may not be familiar with this disease, leading to delayed diagnosis [7].
- Renal involvement can be the presenting feature: Nephrologists should consider Caroli syndrome in patients with unexplained hepatosplenomegaly and renal abnormalities [1, 5].
- Macroscopic hematuria in adulthood can be a revealing sign: The incidental discovery of a congenital disease with a nephrological presentation in adulthood is rare [1].
- Long-term stability is possible: As demonstrated in our patient, with regular follow-up and conservative management, patients with Caroli syndrome can maintain stable renal function and avoid complications for many years.
- Multidisciplinary approach is essential: Management of Caroli syndrome requires collaboration between nephrologists, hepatologists, radiologists, and transplant surgeons [1, 7].

Conclusion:

Caroli syndrome is a rare congenital disease characterized by segmental cystic dilatation of the intrahepatic bile ducts associated with congenital hepatic fibrosis and often associated with autosomal recessive polycystic kidney disease [1, 2]. We report the case of a 32-year-old patient who presented with petechial purpura, macroscopic hematuria, and renal failure. The diagnosis of Caroli syndrome was established based on imaging findings showing cystic dilatation of the intrahepatic bile ducts, portal hypertension, and bilateral renal microcysts. The patient has been followed in our nephrology consultation since 2018 and remains in excellent general condition with stable renal function (creatinine clearance maintained at approximately 58 mL/min) and stable thrombocytopenia, with no new episodes of purpura or macroscopic hematuria. This case highlights the importance of considering Caroli syndrome in patients with unexplained hepatosplenomegaly and renal abnormalities, even in adulthood, and demonstrates that long-term clinical stability with preserved renal function is possible with conservative management and regular follow-up. Caroli syndrome remains a rare disease still unrecognized by many nephrologists.

Conflict of Interest:

The authors declare no conflicts of interest.

Acknowledgments:

The authors thank the medical and nursing staff of the Department of Nephrology at Bab El Oued University Hospital for their contribution to the management of this patient.

References:

1. Orphanet., "Caroli syndrome.." [Online]. Available: ***https://www.orpha.net/fr/disease/detail/480520***.
View at Google Scholar | ***View at Publisher***
2. MeSH., " Caroli Disease. ," National Institutes of Health (NIH). 2026. . [Online]. Available: ***https://meshb.nlm.nih.gov/record/ui?ui=D016767***.
View at Google Scholar | ***View at Publisher***
3. L. R. Patel A, Khan IA, Thakor R, Sapkale B., "Caroli Disease: A Narrative Review of Clinical Spectrum Pathogenesis, Diagnostic Advances, and Management Strategies.," J Clin Diagn Res., vol. 20, no. 7, 2026;:PE19-PE25., doi: doi:10.7860/JCDR/2026/84541.23857.
View at Google Scholar | ***View at Publisher***
4. S. Hanafy et al., "Clinical outcomes in Caroli disease and Caroli syndrome: a longitudinal observational cohort study," (in eng), Sci Rep, vol. 16, no. 1, Mar 26 2026, doi: 10.1038/s41598-026-42855-8.
View at Google Scholar | ***View at Publisher***
5. Agarwal, A. Singh, A. Abhinay, and R. Prasad, "Clinical Spectrum of Caroli Syndrome," (in eng), Indian J Pediatr, vol. 92, no. 5, p. 559, May 2025, doi: ***https://doi.org/10.1007/s12098-025-05482-1***
View at Google Scholar | ***View at Publisher***
6. T. Zhou et al., "Histopathology and molecular pathology confirmed a diagnosis of atypical Caroli's syndrome: a case report," (in eng), Diagn Pathol, vol. 19, no. 1, p. 36, Feb 22 2024, doi: ***https://doi.org/10.1186/s13000-024-01462-9***
View at Google Scholar | ***View at Publisher***
7. L. R. Patel A, Khan IA, Thakor R, Sapkale B., " Caroli Disease: A Narrative Review of Clinical Spectrum Pathogenesis, Diagnostic Advances, and Management Strategies.," J Clin Diagn Res., vol. 20(7), pp. 19-25, 2026, doi: doi:10.7860/JCDR/2026/84541.23857.
View at Google Scholar | ***View at Publisher***