

Case Report

Caroli disease in a 23-month-old boy: a rare paediatric presentation with an inconclusive cystic renal phenotype

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ABSTRACT

Caroli disease is a ductal plate malformation defined by non-obstructive, communicating dilatation of the intrahepatic bile ducts without congenital hepatic fibrosis; when fibrosis is present, the condition is termed Caroli syndrome. We report the case of a 23-month-old boy who presented with hepatomegaly and jaundice. Abdominal ultrasound showed an enlarged liver with saccular cystic dilatation of the intrahepatic bile ducts and bilaterally enlarged kidneys with poor corticomedullary differentiation, prompting further cross-sectional evaluation. Magnetic resonance cholangiopancreatography confirmed non-obstructive, communicating intrahepatic bile duct dilatation consistent with Caroli disease, while renal imaging revealed a diffuse bilateral microcystic pattern with cortical thinning, most consistent with medullary sponge kidney. Imaging was thus central both to establishing the hepatic diagnosis and to characterizing the associated renal phenotype, sparing the child invasive investigation. This case illustrates that a hepatic ductal plate malformation may present in early childhood and unmask an otherwise silent renal cystic phenotype, and adds to the limited pediatric reports of this association.

Keywords: Caroli disease, Medullary sponge kidney, Lenarduzzi–Cacchi–Ricci disease, Autosomal recessive polycystic kidney disease, Differential diagnosis, Cholestasis

INTRODUCTION

Caroli disease belongs to the ductal plate malformations, the congenital hepatobiliary disorders that follow incomplete remodeling of the embryonic ductal plate around the developing portal tracts.^{1,2} It is defined by segmental, non-obstructive, saccular or fusiform dilatation of the large intrahepatic bile ducts that remain in continuity with the biliary tree, and it predisposes to biliary stasis, intrahepatic lithiasis, recurrent cholangitis and, over decades, cholangiocarcinoma.^{2,3} The distinction from Caroli syndrome, in which the same biliary abnormality is accompanied by congenital hepatic fibrosis with portal hypertension, carries direct prognostic weight and is often blurred in the literature.^{1,2}

Medullary sponge kidney, also known as Lenarduzzi-Cacchi-Ricci disease after the Italian authors who

characterised it in the 1930s, is a developmental disorder of the distal nephron defined by ectasia of the medullary and papillary collecting ducts.⁴⁻⁶ It is usually diagnosed in the third or fourth decade, after renal colic, hematuria, recurrent urinary tract infection or the discovery of nephrocalcinosis; recognition in childhood is uncommon and rests mainly on isolated reports and a small pediatric series.⁵⁻⁷

The renal lesion classically associated with ductal plate malformations is autosomal recessive polycystic kidney disease (ARPKD).^{1,2} Coexistence with medullary sponge kidney is comparatively rare and has been described almost exclusively in adolescents and adults, most often in the syndromic hepatic form.⁸ We report a 23-month-old boy in whom Caroli disease without congenital hepatic fibrosis was identified during a first febrile illness, together with a bilateral cystic renal phenotype whose

appearances pointed to medullary sponge kidney without allowing ARPKD or other ciliopathy-related nephropathies to be formally excluded. We set out the imaging reasoning behind this provisional renal attribution and the uncertainty that remains attached to it.

CASE REPORT

History and clinical findings

A 23-month-old boy was brought to the emergency department with a two-day history of fever, food refusal and marked irritability with persistent crying. There was no vomiting, diarrhea, or respiratory or urinary symptom. He was born to non-consanguineous parents, with no family history of liver disease, kidney disease or nephrolithiasis. The pregnancy and delivery were uneventful and no antenatal abnormality had been detected. Growth and development had been normal, and he had had no previous jaundice, abdominal pain, hematuria or urinary tract infection. On admission he was febrile at 39.2 °C, irritable but consolable, adequately hydrated and hemodynamically stable. Weight and height were not recorded. The liver edge was firm, smooth and non-tender, palpable four fingerbreadths below the right costal margin; there was no ascites and no abdominal collateral circulation. Jaundice of the skin and mucous membranes was present, mild in intensity but unequivocal. There were no cutaneous stigmata of chronic liver disease, no dysmorphic features and no hemihypertrophy. No alternative focus of infection was found.

Laboratory investigations

Blood tests showed conjugated hyperbilirubinemia with a cholestatic enzyme pattern and a marked inflammatory response. The conjugated fraction accounted for the majority of the total bilirubin, consistent with the clinical jaundice. Gamma-glutamyl transferase was elevated out of proportion to the transaminases, indicating a predominantly cholestatic disturbance with only mild

hepatocellular injury. Inflammatory markers were markedly raised, with a neutrophil-predominant leukocytosis. Serum albumin was reduced, and hemoglobin was mildly low with a normochromic, normocytic pattern. Kidney function was preserved, with normal creatinine and urea. The platelet count and prothrombin time were both normal. Blood and urine cultures, obtained before the first antibiotic dose, were subsequently negative. Plasma electrolytes, the calcium-phosphate profile and urinary calcium and citrate were all within normal limits, with no biochemical evidence of distal renal tubular acidosis, hypercalciuria or hypocitraturia (Table 1).

Imaging

Abdominal ultrasound showed an enlarged liver, with a craniocaudal span of 11 cm on the mid-clavicular line, homogeneous parenchymal echotexture and saccular cystic dilatations of the intrahepatic bile ducts localized on segments VII and VIII. Both kidneys were enlarged for age with poor corticomedullary differentiation. The spleen measured 9 cm, marginally above the upper limit of 8 cm proposed at two years of age.⁹ The portal vein was patent with hepatopetal flow, and there was no ascites and no portosystemic collateral circulation. The extrahepatic bile duct was not dilated and the gallbladder was unremarkable. Liver MRI with MR cholangiopancreatography was performed at 1.5 T, comprising fat-suppressed axial and coronal T2-weighted HASTE sequences, diffusion-weighted imaging with apparent diffusion coefficient maps, axial T1-weighted in-phase and opposed-phase acquisitions, a dedicated heavily T2-weighted biliary acquisition with maximum-intensity-projection reconstruction, and fat suppressed T1-weighted sequences after gadolinium enhancement. MRI confirmed multifocal saccular and fusiform dilatations of the intrahepatic bile ducts in the right lateral segments (segment VII and segment VIII) hyperintense on T2 and hypointense on T1 (Figure 1).

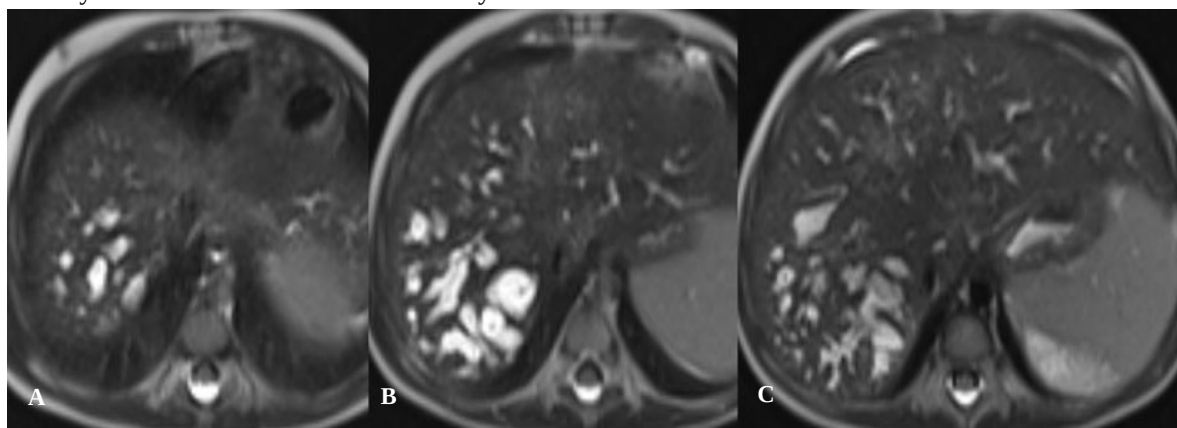


Figure 1: Liver MRI, T2-weighted images. Axial fat-suppressed T2-weighted images showing multifocal saccular and fusiform markedly hyperintense dilatations of the intrahepatic bile ducts in right lobe (yellow arrows) localized in segment VIII (A) and segment VII (B and C). The liver contour is regular, with no periportal fibrous thickening and no portosystemic collateral vessel.

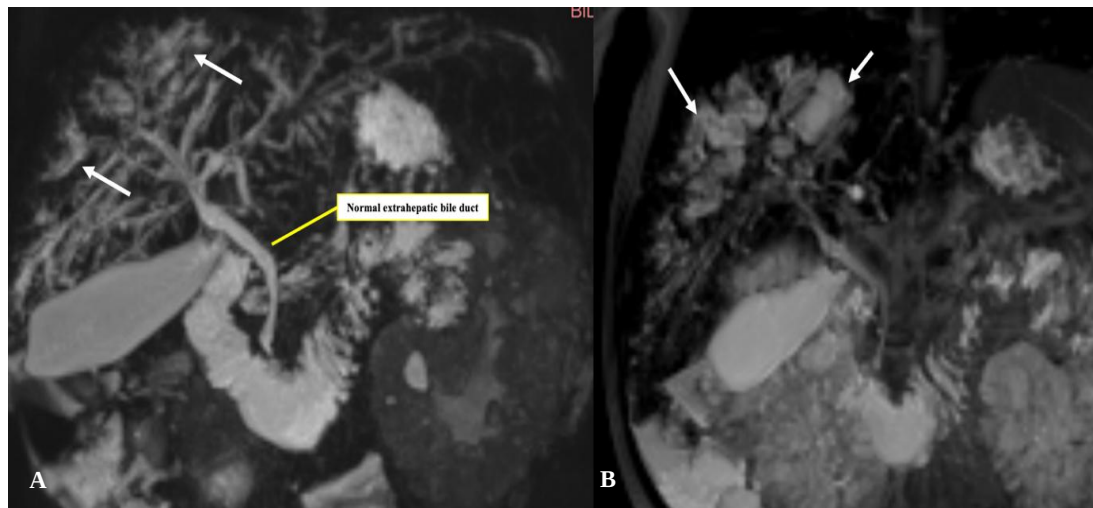


Figure 2: MR cholangiopancreatography. Maximum-intensity-projection reconstruction demonstrating saccular dilations of the intrahepatic bile ducts (A and B: white arrows), in direct continuity with an otherwise normal-caliber biliary tree. The extrahepatic bile duct is normal in caliber (A: yellow annotation), confirming the non-obstructive, communicating nature of the dilatation. No intraductal calculus is identified.

MR cholangiopancreatography demonstrated their direct continuity with an otherwise non-dilated biliary tree and a normal-caliber extrahepatic bile duct, establishing the non-obstructive and communicating nature of the abnormality (Figure 2).

Small enhancing intraluminal foci corresponding to portal venous radicles surrounded by dilated duct, the central dot sign, were identified after contrast. There was no intrahepatic calculus and no biliary wall thickening. The liver measured 11.2 cm craniocaudally, consistent with the sonographic span, and showed no periportal fibrous thickening and no dysmorphic contour.

The portal vein was patent and of normal caliber, and no ascites, varices or portosystemic collateral vessel was seen.

Relatively symmetrical bilateral nephromegaly with preserved reniform morphology but poor corticomedullary differentiation. The renal parenchyma contains numerous small tubular and microcystic T2-hyperintense structures, predominantly involving the medullary and papillary regions and arranged radially towards the papillae.

The cortex appears extremely thinned (Figure 3). No macroscopic calculus and no pelvicalyceal dilatation were identified.

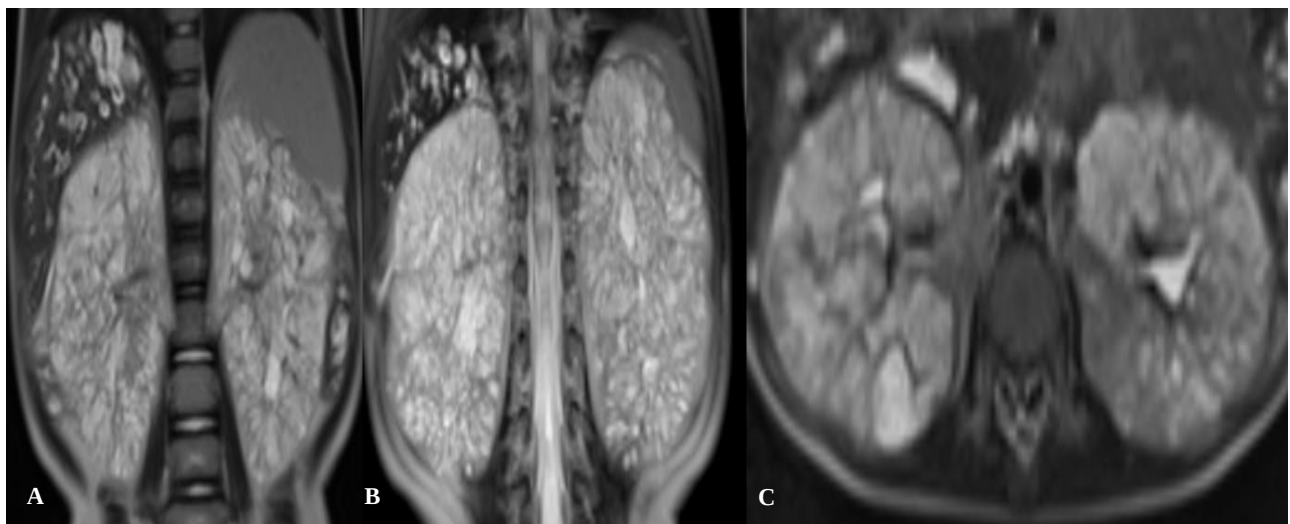


Figure 3: Renal MRI. Coronal (A and B) and axial (C) T2-weighted images of both kidneys, enlarged with preserved reniform outline. Diffuse clustered small rounded hyperintense foci. The cortex is thin and linear a larger cyst is present in the right kidney; neither finding belongs to the classical description of medullary sponge kidney. No macroscopic calculus and no pelvicalyceal dilatation are identified.

Diagnoses retained

Communicating, non-obstructive intrahepatic bile duct dilatation with the central dot sign, in the absence of imaging features of congenital hepatic fibrosis, established the diagnosis of Caroli disease.^{1,2} The renal appearances were most consistent with bilateral medullary sponge kidney, but the cortical thinning cysts precluded a definitive attribution; the renal phenotype was therefore recorded as undetermined, autosomal recessive polycystic kidney disease and other ciliopathy-related nephropathies remaining possible in the absence of genetic testing.

Fever with cholestasis was attributed to a presumed acute cholangitis, retained as a working diagnosis since blood cultures were sterile and no bile was obtained. The alternatives considered for each of these three diagnoses, and the limits of the diagnostic criteria applied, are set out in the discussion.

Treatment and outcome

Empirical intravenous piperacillin-tazobactam was started after cultures were obtained, at 75 mg/kg of the piperacillin

component every 6 hours; no additional anti-anaerobic agent was given, the combination covering the enteric Gram-negative organisms and anaerobes expected in biliary infection. Biliary drainage was discussed at the

multidisciplinary meeting and not undertaken: the intrahepatic dilatation was cystic and non-obstructive, the common bile duct was of normal caliber, and no obstructing lesion was demonstrated, so no target for decompression existed.

The child defervesced within 48 hours and resumed feeding. On day 3, treatment was switched to oral amoxicillin-clavulanate (60 mg/kg/day of amoxicillin in three divided doses) to complete a 14-day course. Inflammatory markers decreased progressively during treatment and had normalized by day 14.

Liver function tests also improved substantially, although the conjugated bilirubin level remained marginally elevated. Kidney function and blood pressure remained normal, and the child was discharged on day 6 (Table 1, Figure 4).

Table 1: Laboratory findings at admission and at the end of treatment.

Parameters	Patient values		Reference range for age
	Day 0	Day 14	
Temperature	39,2	37,2	36.5–37.5 °C
Haemoglobin	9.0 (normochromic, normocytic)	9,2	10.5–13.5 g/dl
White cell count	15,5	7,8	6,000–17,000/mm ³
Neutrophils	9	4	1,500–8,000/mm ³
Lymphocytes	2	2,2	3,000–9,500/mm ³
Platelet count	230	210	150,000–450,000/mm ³
C-reactive protein	82	<5	<5 mg/l
Total bilirubin	10	1	<1.2 mg/dl
Conjugated bilirubin	7	0,6	<0.3 mg/dl
Gamma-glutamyl transferase	123	27	<25 U/l
AST	89	12	<50 U/l
ALT	41	17	<40 U/l
Serum albumin	25	—	35–50 g/l
Serum creatinine	0.3	—	0.2–0.4 mg/dl
Serum Urea	23	—	15–40 mg/dl
Blood glucose	1	—	0.7–1.1 g/l
Sodium	138	—	135–145 mol/l
Potassium	3.6	—	3.5–4.5 mmol/l
Calcium	2,5	—	2.2–2.7 mmol/l
Chloride	98	—	98–107 mmol/l
Phosphate	1,5	—	1.3–2.3 mmol/l
Prothrombin time	98%	—	70–100 %
Urinalysis	Normal	—	Normal
Urine culture	Sterile	—	Sterile
Blood cultures	Sterile	—	Sterile

ALT, alanine aminotransferase; AST, aspartate aminotransferase; PT, prothrombin time; N, Neutrophils; L, Lymphocytes. Reference ranges are indicative for a child of 23 months. A dash indicates an investigation not repeated.

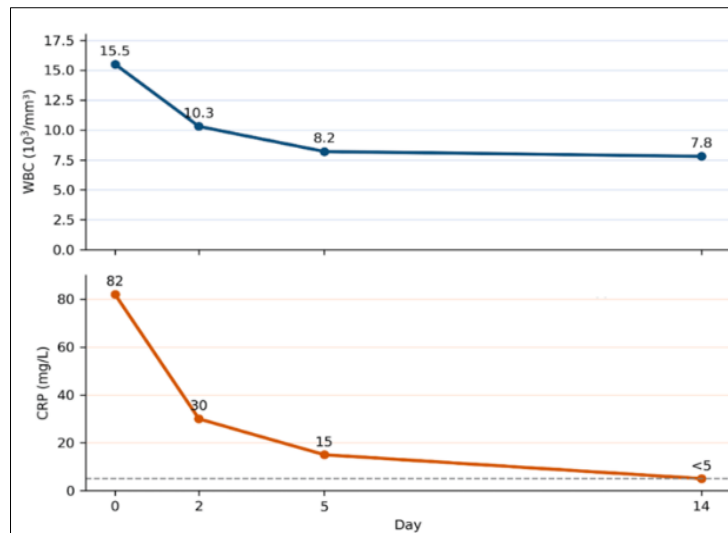


Figure 4: Changes in inflammatory markers during treatment. Progressive decrease in c-reactive protein (CRP) levels and white blood cell count from admission (day 0) to the end of treatment (day 14), with normalization of both parameters by day 14.

Follow-up ultrasound showed persistence of the hepatomegaly, of the cystic biliary dilatation and of the nephromegaly with loss of corticomedullary differentiation. These appearances constitute the baseline for future imaging. The child was enrolled in a structured surveillance program for the periodic monitoring of liver and kidney function, coordinated between pediatrics, pediatric hepatology, pediatric nephrology and pediatric radiology. The family was counselled about the symptoms requiring urgent reassessment, in particular fever with abdominal pain or jaundice, and about maintaining an adequate fluid intake.

DISCUSSION

Pathophysiology

Caroli disease originates in a defect of ductal plate remodeling. Hepatoblasts adjacent to the portal mesenchyme form a cylindrical ductal plate that is progressively remodeled into the intrahepatic bile ducts; failure of this process leaves persistent ectatic embryonic structures around the portal branches.^{1,2} The level of the biliary tree involved determines the phenotype: the large central ducts in Caroli disease, the small interlobular ducts in congenital hepatic fibrosis, and both in Caroli syndrome.^{1,2} These disorders are understood as ciliopathies, which accounts for their frequent renal associations and for the overlap with ARPKD.²

The pathogenesis of medullary sponge kidney is less well defined. Disturbance of the interaction between glial cell line-derived neurotrophic factor and the RET receptor tyrosine kinase, which governs reciprocal induction between ureteric bud and metanephric blastema, has been proposed as the mechanism of collecting duct ectasia, supported by the association with other developmental

anomalies and by reports of familial clustering.^{5,6} This remains a hypothesis, and no diagnostic genetic test is available in routine practice.⁵ The shared framework of disordered epithelial tubulogenesis offers a plausible, though unproven, explanation for the coexistence of the two conditions.

Differential diagnosis of the biliary lesion

Obstructive dilatation, a choledochal cyst and a biliary stricture were excluded by the normal-caliber extrahepatic duct and the absence of any obstructing lesion. Primary sclerosing cholangitis produces multifocal strictures with a beaded appearance rather than discrete saccular dilatations and would be exceptional at this age; recurrent pyogenic cholangitis requires intraductal stones and repeated episodes, neither present here. Peribiliary cysts and biliary hamartomas do not communicate with the biliary lumen, which distinguishes them at MR cholangiopancreatography.¹

Contribution of imaging

Ultrasound is the appropriate first-line examination but cannot establish communication with the biliary tree, which is the defining feature of the disease.² MR cholangiopancreatography is the reference non-invasive technique: maximum-intensity-projection reconstruction demonstrates continuity between the dilated segments and an otherwise normal biliary tree, simultaneously establishing the communicating nature of the lesion and excluding an obstructive cause.^{1,2} A comparative study has shown that the technique also distinguishes diffuse from localized forms, a distinction with direct surgical consequences.¹⁰ The central dot sign, an enhancing intraluminal portal venous radicle surrounded by dilated duct, is characteristic though neither invariably present nor entirely specific.^{1,2} Avoiding ionizing radiation is decisive

in a patient who will require repeated imaging over decades.

The morphological substrate of medullary sponge kidney is ectasia of the medullary and papillary collecting ducts, producing microcysts confined to the pyramids with a fan-shaped orientation towards the papillary tip.^{4,6} Hyperechoic pyramids on ultrasound usually reflect the medullary nephrocalcinosis that complicates the disease rather than the ectasia itself; in its absence the sonographic appearances may be limited to renal enlargement with poor corticomedullary differentiation, as here.⁷ No nephrocalcinosis and no calculus was present in this child, and it is important not to report these complications when they are absent: they are consequences of the malformation, not diagnostic criteria, and their absence at 23 months is expected.

Mild splenomegaly and the question of portal hypertension

The spleen measured 9 cm, marginally above the upper limit of 8 cm proposed at two years.⁹ Splenomegaly in a patient with intrahepatic bile duct dilatation is the finding that would ordinarily point towards congenital hepatic fibrosis with portal hypertension, and therefore towards Caroli syndrome. Three considerations led us to retain the non-syndromic designation. The enlargement was slight, exceeding the reference limit by about a centimeter, whereas the splenomegaly of established portal hypertension is generally more pronounced. No other feature of portal hypertension was present on either modality. Additionally, the child presented with an acute febrile illness and a C-reactive protein of 82 mg/l, a context in which reactive splenomegaly is common at this age; the concurrent normochromic normocytic anemia is equally compatible with an inflammatory cause. The platelet count (230 000/mm³) and the prothrombin time were both normal, arguing against hypersplenism and against any appreciable impairment of hepatic synthetic function, the hypoalbuminemia being readily attributable to the acute inflammatory episode.

Characterizing the renal phenotype: medullary sponge kidney or ARPKD?

Because ARPKD carries a risk of early kidney failure and of significant liver disease, its exclusion was the central issue in this case.^{2,10} Three arguments supported medullary sponge kidney. The cystic change predominated in the medullary and papillary collecting ducts and extended towards the papillary tips, whereas ARPKD produces fusiform ectasia radiating through cortex and medulla together with a diffusely hyperechoic parenchyma. Congenital hepatic fibrosis, present in the great majority of patients with ARPKD, was absent on both modalities. Kidney function was preserved and blood pressure normal, whereas ARPKD presenting in infancy is generally accompanied by early functional impairment and hypertension.¹¹ Medullary nephrocalcinosis was excluded

by the absence of any echogenic focus with acoustic shadowing, renal dysplasia by the preserved renal shape, and nephronophthisis by the normal kidney function; the absence of consanguinity is supportive of a non-recessive phenotype but weak in isolation.

The renal cortex was thin rather than of normal thickness, a finding that temper this conclusion. It doesn't belong to the classical description of medullary sponge kidney, in which the cortex is spared, and it's compatible with a nephropathy involving the whole parenchyma. Cortical thinning at 23 months, in a child whose kidney function is currently normal, warrants close surveillance rather than reassurance. Other ciliopathy-related phenotypes, in particular HNF1B-related renal disease, in which cysts may be associated with an abnormal parenchymal architecture, were not formally excluded either, no genetic study having been performed. In the absence of PKHD1 analysis the distinction rests on the balance of imaging and clinical arguments alone, and ARPKD remains in the differential diagnosis. We therefore report medullary sponge kidney as the most probable interpretation of the renal appearances rather than as an established diagnosis, and the renal phenotype is presented here as undetermined.

Reports combining the Caroli spectrum with medullary sponge kidney are few and concentrated in adolescence and adulthood. The best documented is that of Sinha and colleagues, who described an adult man in whom Caroli disease, medullary sponge kidney, nephrocalcinosis and chronic renal insufficiency were identified during the investigation of a urethral stricture, and who noted that the frequency of the association is unknown and that only a handful of cases had been published.⁸ Further observations exist in journals not indexed in MEDLINE, Scopus or Web of Science and in a congress educational exhibit; they are of limited evidential weight and are not cited here. In the retrievable literature the hepatic lesion was, in most instances, the syndromic form, and the diagnosis followed a complication, whether hemorrhagic, lithogenic or renal.

Recognition of the Caroli spectrum itself in infancy has been reported: a 20-month-old with abdominal distension and hepatosplenomegaly was found to have Caroli syndrome, the authors emphasizing that clinical latency is the principal reason for diagnostic delay, although no medullary sponge kidney was documented in that patient.¹² Taken together, these observations suggest that the ages usually quoted for the two conditions reflect the age at which their complications become symptomatic rather than the age at which the malformations can be detected.

Management and follow-up

No clinical practice guideline is devoted specifically to Caroli disease, to medullary sponge kidney or to their association, and none of the reviews cited here should be read as one. Management is assembled from recommendations addressing each complication, most of

which derive from adults and require adaptation before being applied to a small child.

In the present child the Tokyo Guidelines 2018 diagnostic criteria were, applied literally, satisfied: systemic inflammation (39.2 °C, white cells 15 500/mm³, C-reactive protein 82 mg/l), cholestasis (jaundice with conjugated hyperbilirubinemia and raised gamma-glutamyl transferase) and abnormal biliary imaging; four of the five prognostic factors were present, corresponding to the moderate grade for which early drainage is advised.¹ Acute cholangitis was nevertheless retained as a working hypothesis only. The biliary imaging item is satisfied by a congenital and permanently present malformation and therefore carries no discriminating value in this patient; blood cultures were sterile and no bile was obtained; and the criteria were developed and validated in adults with acquired obstruction. Urinary tract infection was excluded by a sterile urine culture, and an intercurrent infection of another origin cannot be excluded.

For suspected cholangitis, the Tokyo Guidelines 2018 remain the reference framework in adults.¹³ The present patient illustrates two difficulties in transposing them: the diagnostic criteria may be satisfied by default, because the biliary imaging item is permanently fulfilled by the malformation, and the severity criteria may likewise assign a moderate grade to a child in whom leukocytosis and fever reflect the response to any infection and in whom hypoalbuminemia may be chronic. Taken literally they would have mandated early drainage in a patient who recovered on antibiotic therapy alone. In practice, cultures should precede empirical intravenous therapy directed at enteric Gram-negative organisms and anaerobes, with drainage reserved for failure to respond or a demonstrable obstruction.

Ursodeoxycholic acid is widely prescribed with the aim of improving bile flow, but the supporting evidence is limited to uncontrolled observations; it should be presented to families as a measure of uncertain benefit rather than an established treatment. When Caroli disease is confined to one lobe, hepatic resection can be curative and removes both the septic risk and the long-term risk of malignant transformation; when it is diffuse and complicated by refractory cholangitis or secondary biliary cirrhosis, liver transplantation is the only definitive option, and surgical series support timely referral rather than surgery as a last resort.¹⁴ Lifelong surveillance is required, combining clinical assessment, liver biochemistry and cross-sectional imaging, with attention to intrahepatic stones, to any feature suggesting emerging hepatic fibrosis and portal hypertension, and, in adulthood, to the risk of cholangiocarcinoma, which has been quantified in a systematic review.³ Repeated MRI is preferable to computed tomography over such a timescale.

Medullary sponge kidney has no specific treatment, and management is directed at preventing and treating its complications.^{5,6} Surveillance should include periodic

measurement of kidney function, blood pressure against age- and height-specific references, urinalysis, and renal ultrasound for the emergence of nephrocalcinosis or calculi. Distal renal tubular acidosis, hypercalciuria and hypocitraturia should be sought specifically, since they are treatable and drive stone formation.⁶ Should a calculus develop, the EAU Guidelines on Urolithiasis recommend a complete metabolic evaluation in all children, stone analysis whenever material is available, and ultrasound as first-line imaging; alkali citrate therapy has been used in adults with medullary sponge kidney but pediatric dosing must be supervised by a pediatric nephrologist.¹⁵ Adequate fluid intake is the cornerstone of stone prevention, but adult target volumes have no pediatric equivalent and must not be transposed numerically; in a child of this age the objective is age-appropriate maintenance intake, increased during febrile illness and hot weather.

Originality of the observation

Three features give this case its interest. The hepatic lesion was Caroli disease without congenital hepatic fibrosis, whereas the published reports of this combination describe, in most instances, the syndromic hepatic form; the renal component, by contrast, remained a presumptive rather than a confirmed medullary sponge kidney. It was recognized at 23 months, well below the age at which either condition is usually diagnosed, and before any lithogenic, hemorrhagic or renal complication had developed. And it was revealed by a hepatic presentation, so that a renal malformation which would otherwise have remained silent for decades was identified incidentally.

Learning points

Caroli disease is distinguished from Caroli syndrome by the absence of congenital hepatic fibrosis, a distinction with important prognostic and therapeutic implications. MRCP is the preferred imaging modality because it demonstrates communication between the dilated intrahepatic bile ducts and the biliary tree, may reveal the central dot sign, and avoids ionizing radiation during long-term surveillance. In children with associated bilateral renal cystic abnormalities, cyst distribution, hepatic fibrosis, and renal function help differentiate medullary sponge kidney from autosomal recessive polycystic kidney disease; genetic testing may be required when findings overlap. Because congenital bile duct dilatation may fulfil the imaging component of the Tokyo Guidelines at baseline, acute cholangitis should be diagnosed by integrating clinical, inflammatory, and microbiological findings rather than imaging alone.

Limitations

This is a single observation. No histological confirmation was obtained for either organ, so the exclusion of congenital hepatic fibrosis rests on imaging alone and is provisional: the mild splenomegaly means that low-grade fibrosis with early portal hypertension cannot be ruled out

without a liver biopsy, and if the spleen fails to regress the diagnosis would have to be revised to Caroli syndrome. The platelet count and the prothrombin time were normal, which argues against portal hypertension and against impaired hepatic synthesis but does not exclude low-grade fibrosis. PKHD1 analysis was not performed either, and the cortical thinning weakens the exclusion of ARPKD, so that the renal diagnosis remains presumptive throughout this report. The diagnosis of medullary sponge kidney was made on cross-sectional imaging, whereas the historical reference standard was excretory urography, no longer acceptable in children. The suspected biliary infection was never microbiologically confirmed. Follow-up is short, and the natural history of this association from infancy is unknown.

CONCLUSION

The identification of Caroli disease and bilateral medullary sponge kidney in a 23-month-old boy investigated for fever, hepatomegaly and cholestasis. MR cholangiopancreatography demonstrated communicating, non-obstructive intrahepatic bile duct dilatation with the central dot sign and no congenital hepatic fibrosis, while the renal appearances were most consistent with medullary sponge kidney without allowing ARPKD or other ciliopathy-related nephropathies to be formally excluded. The observation illustrates that a hepatic presentation may reveal a renal malformation that would otherwise have remained silent for decades, and that the characterization of the renal cystic phenotype, which carries direct prognostic and management consequences, may remain provisional on imaging alone and require genetic testing and prolonged follow-up.

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