Case study

Cacchi-Ricci and Caroli : Both rare but frequently associated

ABSTRACT

Aims: Caroli and Cacchi Ricci are two rare diseases but frequently associated. We report the case of a young girl who presented these exceptional entities.

Results: We report the case of a 14-year-old girl with no particular pathological history, who presented to the emergency room with rectal bleeding that had been evolving for 1 months. The association of Cacchi Ricci and Caroli diseases was diagnosed on imaging (abdominal ultrasound and computed tomogrphy, bili-MRI), following an assessment carried out in the face of signs of portal hypertension and gastrointestinal bleeding.

Conclusion: Although both rare, Cacchi Ricci disease and Caroli syndrome are frequently associated. This association is to be watched all the more since it increases overall morbidity and mortality and often requires consideration of double hepatorenal transplantation as the only curative therapeutic alternative.

Keywords: [Caroli, Cacchi Ricci, sponge medullar kidney}

1. INTRODUCTION

Cacchi Ricci disease or sponge kidney is a rare genetic pathology, most often sporadic and corresponding to a renal malformation manifested by a dilation, typically bilateral, of the medullary collecting ducts. It is frequently associated with Caroli's disease, which is a congenital hepatic malformation characterized by non-obstructive dilation of the intrahepatic bile ducts with or without congenital hepatic fibrosis.

2. PRESENTATION OF CASE

We report the case of a 14 year-old girl with no particular pathological history, who presented to the emergency room with rectal bleeding evolving since 1 month. The girl was pale, dehydrated, and reported and increasing fatigue. The clinical exam objectified an abdominal distension with collateral circulation and an abdominal mass of 17x25 cm, and bilateral lumbar mass. The digital rectal exam confirmed the presence of blood. Biology objectified an anemia at 7.6 g/dl, low platelets at $85.10^3/mm^3$, an hypernatremia at 149 mEq/l with a normal plasma creatinin with an elevated urea 0.98 g/l. An ultrasoud was requested, indicating an enlarged liver and spleen. An abdomial CTscan was then performed, confirming the hepato and splenomegaly associated with segmental, multifocal dilations of the intrahepatic bile ducts and bilateral enlarged kidneys with multiple cystic formations giving the particular aspect of sponge kidneys. Both of this particular aspects allowed us to evoke Cacchi Ricci and Caroli diseases. A Bili-MRI was then conducted confirming the non-obstructive character of the bile ducts dilatation.

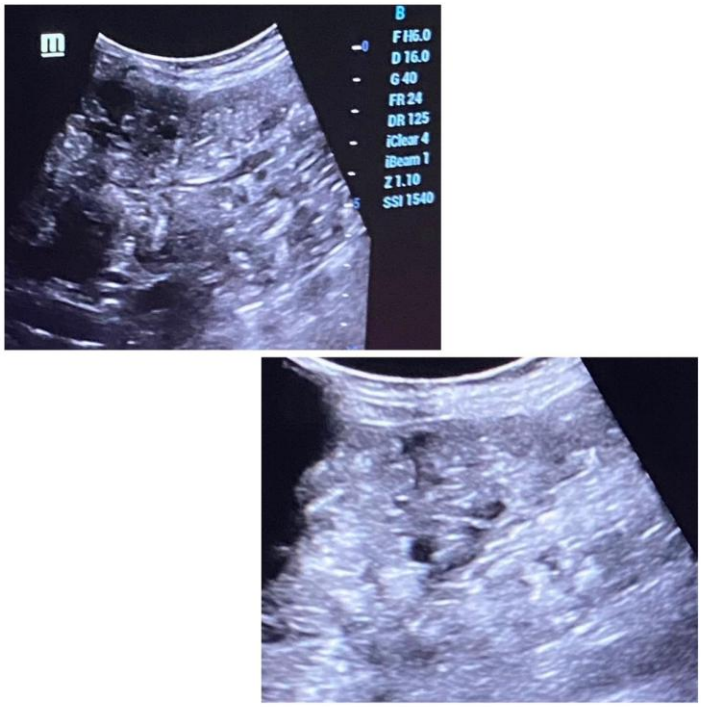

Fig. 1. Echography: Longitudinal section of the kidneys, showing an echogenic cortex with multiple intramedullary cysts of variable size and some microlithiasis.

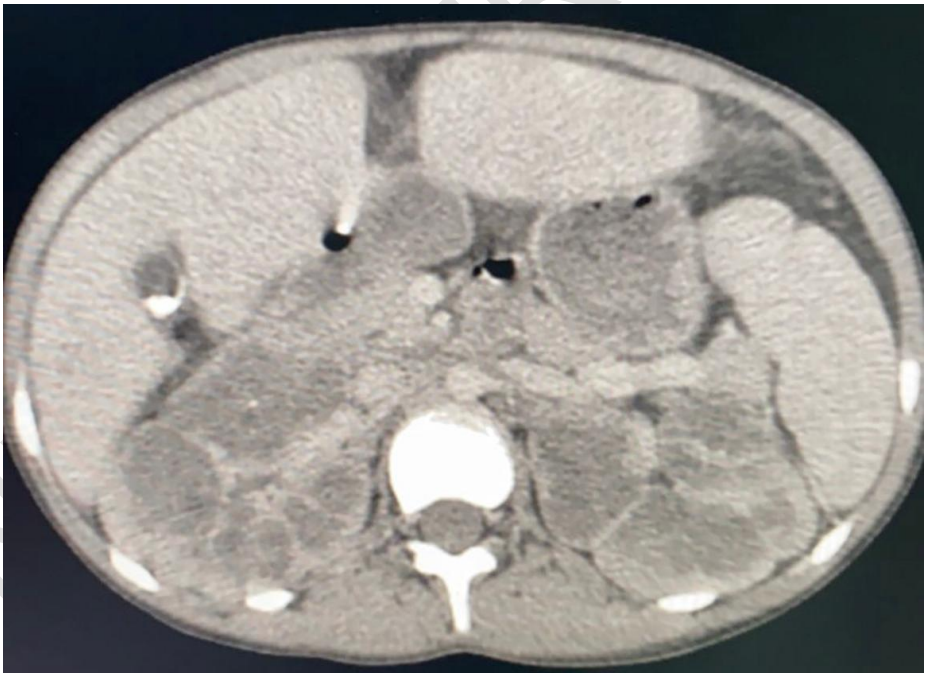

Fig. 2. CT in axial section after injection of iodinated contrast product, portal phase : enlarged kidneys, site of multiple cystic formations of the medulla creating a sponge appearance.

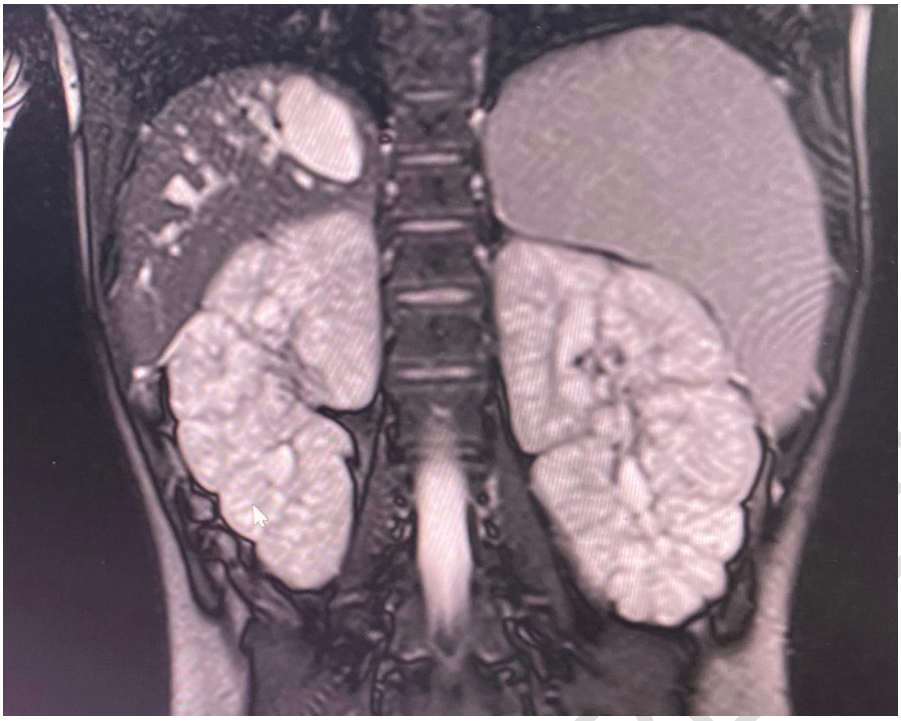

Fig. 3. MRI in coronal section, balanced in T2: highlighting globular kidneys, site of multiple cystic formations of the medulla, realizing a sponge appearance. Presence of a major cystic and monoliform dilatation of the intrahepatic bile ducts falling within the scope of Cacchi-Ricci disease.

3. DISCUSSION

Cacchi Ricci disease also known as medullary sponge kidney is generally sporadic and more rarely hereditary with no known genetic cause. The prevalence of the medullary sponge kidney is 1 out of 5000 person wich makes it a relatively rare disease. The pathogenesis of which remains poorly known to this day. It owes its second name of “sponge” kidney to its typical radiological appearance, in cross section, due to the dilatations of the medullary and papillary portions of the distal collecting ducts which can be up to 8 millimeters in diameter [1]. The injury is classically bilateral but most often remains asymptomatic and evolves slowly with an age of diagnosis generally between 20 and 30 years-old [2]. It is manifested most frequently by repeated episodes of urinary tract infections and lithiasic disease due in particular to tubular dysfunctions [3]. Often complicated by nephrocalcinosis, it owes its prognosis to its evolution towards terminal renal failure [1].

Caroli's disease is considered as rare with an incidence of 1 of 1 million births. It is due to a developmental anomaly during the fetal organogenesis of the bile ducts resulting in the dilation of the intrahepatic bile ducts. Caroli's syndrome is distinguished from Caroli’s disease, with the addition of to liver fibrosis with a darker prognosis linked to complications due to portal hypertension and the evolution, often inevitable, towards terminal liver failure [4]. The cause of the disease is not known at this day but the mutation of PKHD1 gene is largly considered, being responsible of an abnormality in the synthesis of fibrocystin [5]. The treatment of Caroli’s affection is mainly supportive. Liver resection is considered on unilobar diseases, however the transplantation is to be pondered when the hepatic disease is diffuse [4,6].

These two diseases, both rare, are however frequently associated [7].

4. CONCLUSION

Although both rare, Cacchi Ricci disease and Caroli syndrome are frequently associated. This association is to be watched all the more since it increases overall morbidity and mortality and often requires consideration of double hepatorenal transplantation as the only curative therapeutic alternative.

Ethical Approval:

As per international standard or university standard written ethical approval has been collected and preserved by the author(s).

Consent

As per international standard or university standard, Parental' written consent has been collected and preserved by the author(s).

COMPETING INTERESTS

Authors have declared that they have no known competing financial interests OR non-financial interests OR personal relationships that could have appeared to influence the work reported in this paper.

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