Obstructive jaundice in a patient with polycystic liver disease complicated with polycystic kidney and polycystic lung: A case report

1. Introduction

Polycystic liver disease (PLD) is a rare genetic disease caused by mutations in the PKD1, PKD2, or PRKCSH gene.[1] The characteristic manifestation of PLD is liver cysts that originate from bile-duct epithelial cells owing to PLD-related gene mutations. As the liver cysts grow, the hepatic volume increases at a rate of 0.9% to 1.6% per year; in some patients, the total hepatic volume can exceed 10 L.[2] The symptoms of PLD are non-specific and are mainly attributable to the increase in hepatic volume; they include indigestion, loss of appetite, nausea, vomiting, and right upper abdominal pain. Obstructive jaundice is a rare manifestation of PLD, and is usually a consequence of bile-duct compression by external cysts, especially, in patients with numerous hepatic cysts.[3,4]

PLD can not only occur alone as an autosomal-dominant genetic anomaly, but it can also be accompanied by autosomal-dominant or autosomal-recessive polycystic kidney disease (PKD).[5,6] However, the combination of PLD with polycystic lung is rare. In this report, we present an extremely rare case of obstructive jaundice in a patient with PLD complicated by PKD as well as polycystic lung.

2. Case report

A 72-year-old man was admitted to our hospital due to the rupture of some renal cysts. He had first been admitted to our hospital 3 years ago because of hematuria associated with PLD complicated with PKD and secondary renal dysfunction (estimated glomerular filtration rate: 40 mL/minute/1.73 m²).
At that time, hemostasis was successfully achieved, and the hematuria disappeared. The patient was discharged and advised to take detoxification drugs, including Niaoduqing particles and activated charcoal tablets. He was followed up at our hospital for 1 year, during which time his renal function was stable; however, he stopped attending the follow-up exams after 1 year. At 3 months before the present hospitalization, he developed hematuria again, followed by progressive jaundice, loss of appetite, nausea, and weight loss. He had a family history of PKD; his 2 sons have PLD complicated with PKD.

A physical examination during the current hospitalization revealed severe and extensive yellowing of the skin and sclera, and a large, non-tender abdominal mass, with no signs of chronic liver disease, such as ascites and splenomegaly. The results of liver-function tests were indicative of obstructive jaundice: total bilirubin, 12.29 mg/dL (normal range, <1 mg/dL); direct bilirubin, 9.22 mg/dL (normal range, <0.4 mg/dL); alkaline phosphatase, 198.4 U/L (normal range, 45–125 U/L); γ-glutamyl transpeptidase, 180.2 U/L (normal range, 10–60 U/L); aspartate transaminase, 45.2 U/L (normal range, 15–40 U/L), and serum creatinine, 7.69 mg/dL (normal range, 0.67–1.18 mg/dL). Additionally, serological tests for hepatitis were negative.

Computed tomography (CT) demonstrated numerous cysts in the liver and kidneys with a very small proportion of normal residual parenchyma, and also revealed numerous cysts in the lungs (Fig. 1A–D). The findings of magnetic resonance imaging (MRI) and magnetic resonance cholangiopancreatography (MRCP) were indicative of obstructive jaundice resulting from the repression of numerous liver cysts, especially, 2 large cysts in the liver (Fig. 2A–B).

Because of the diffuse distribution of cysts in the liver and the severe renal dysfunction, the patient could not undergo surgical treatments such as percutaneous drainage of hilar cysts, open or laparoscopic cyst fenestration, combined hepatic resection and fenestration, and liver transplantation. Furthermore, medical treatment failed to improve the patient’s jaundice. As the excessive bilirubin could have led to life-threatening bilirubin encephalopathy, we decided to perform dialysis to remove bilirubin from the circulation and relieve the symptoms. Bilirubin adsorption and continuous veno-venous hemofiltration (CVVH) were selected to ameliorate the hyperbilirubinemia and uremia. Surprisingly, the serum bilirubin level dropped from 12.29 mg/dL to 5.01 mg/dL and the levels of urea and creatinine decreased after a single session of combined CVVH-bilirubin adsorption treatment. However, the patient and his family chose hemoperfusion and hemofiltration for further treatment, as this combination was less expensive, though also less effective, than combined CVVH-bilirubin adsorption treatment. The patient has provided informed consent for the publication of this case report.

Figure 1. Computed tomography scans show multiple cysts (arrows) in the lungs, liver, and kidneys, with a very small proportion of normal residual parenchyma.
Unfortunately, 4 days after being discharged, the patient developed sudden shock and unconsciousness, which led to his demise. The sudden deterioration of the patient’s condition may be attributable to spontaneous cyst rupture and bleeding.

3. Discussion

We have reported an extremely rare case of PLD complicated with PKD, polycystic lung, and obstructive jaundice in a 72-year-old man. The results of liver-function tests in our patient indicated obstructive jaundice: total bilirubin, 12.29 mg/dL and direct bilirubin, 9.22 mg/dL. MRI also suggested dilatation of the bile ducts adjacent to 2 large cysts, with no stones, sludge, or other lesions. Obstructive jaundice in a patient with liver cysts is not necessarily due to extrinsic compression of the bile ducts due to the cysts, but may be secondary to congenital cystic malformation of the ducts. Khoonsari et al and Dumonta et al each reported a case of obstructive jaundice secondary to compression of liver cysts. In both cases, exploratory laparotomy and surgical decompression were performed. Unfortunately, our patient could not undergo surgery due to multiple liver cysts of varying sizes and severe renal failure. Although initial treatment with bilirubin adsorption-CVVH dramatically reduced the serum bilirubin level, the patient was unable to continue this treatment due to financial reasons. He eventually succumbed to his disease, and the final cause of death was most likely diffuse cyst hemorrhage.

PLD is more frequent in females than in males. In female patients with PLD, the hepatic cysts can markedly increase in size and number during childbearing age. Studies have shown that PLD is clinically silent in the early stage; however, the symptoms of persistent abdominal pain, abdominal distension, gastroesophageal reflux, back pain, and hepatic vein and portal vein compression appear in later stages, and severe complications such as cyst hemorrhage, rupture, and infection occur in nearly 50% of patients by the final stages. A fatal case of hepatic cyst rupture in a patient with PLD complicated with autosomal-dominant PKD has been reported in the literature. In our patient, the cause of death was most likely diffuse cyst hemorrhage in the liver or kidney.

Complications of advanced liver disease and liver failure rarely appear in patients with PLD, and the liver function is well preserved despite the development of portal hypertension and hepatosplenomegaly. Furthermore, the incidence of jaundice is quite low among PLD patients, unless concomitant choledocholithiasis or cholangiocarcinoma cause obstructive jaundice. In our patient, however, MRI and MRCP showed severe obstructive jaundice related to PLD.

The treatment strategy for the complication of polycystic kidney and polycystic lung is aimed at reducing the cysts or organ volume, relieving the symptoms, and improving the quality of life. To treat PKD, cell proliferation and cyst growth are inhibited using drugs and recently, using gene therapy. Laparoscopic surgical treatment is mainly used to decompress the cysts to reduce the development of renal failure. The treatment of polycystic lung mainly includes preventing lung infections, enhancing immunity, improving the living environment, and keeping the air fresh. If respiratory failure and renal failure occur, the most effective treatment is a combined lung and kidney transplantation.

The therapies for PLD can be roughly divided into medical and surgical strategies, and for the with the latter being the main treatment alternative despite the high cost, and the occurrence of postoperative complications and relapse. Unfortunately, our patient was considered to be unable to tolerate surgery due to the presence of numerous diffuse hepatic and renal cysts and renal failure. In recent years, blood purification has been applied to...
treat hyperbilirubinemia when medical and surgical treatments fail. These include bilirubin adsorption, plasma exchange, hemoperfusion, hemodialysis, hemodiafiltration, and molecular adsorbtion recycling systems. Bilirubin has a molecular weight of 584.67 Da. Clinical studies have shown that hemodialysis, hemoperfusion, or hemodiafiltration combined with plasma exchange can provide good clearance of substances with medium molecular weights.\[16\] Furthermore, CVVH has the advantage of regulating the acid-base balance, water and electrolyte balance, and increasing the clearance of water-soluble toxins, and is thus especially suited for critically ill patients with renal failure.\[19,20\]

4. Conclusion

PLD is a rare genetic anomaly. It is usually accompanied with autosomal-dominant PKD and only infrequently with polycystic lung. Nevertheless, imageological examination of the lungs is necessary for patients with PLD. Although infrequent, jaundice can occur in this group of patients and lead to severe hyperbilirubinemia. Additionally, when liver transplantation and other surgical methods are contraindicated, blood purification methods may be considered an alternative treatment for patients with obstructive jaundice related to PLD.

Author contributions

Conceptualization: Santao Ou, Qi Liu.
Data curation: Santao Ou, Jiaru Lin, Ying Li, Linwang Gan
Formal analysis: Santao Ou.
Writing – review & editing: Liling Zhang.

References

[1] Delis SG, Bakoyiannis A, Triantopoulou C, et al. Obstructive jaundice in polycystic liver disease related to coexisting cholangiocarcinoma. Case Rep Gastroenterol 2008;2:162–9.
[2] Gevers TJ, Drenth JP. Diagnosis and management of polycystic liver disease. Nat Rev Gastroenterol Hepatol 2013;10:101–8.
[3] Ergun H, Wolf BH, Hisyong SL. Obstructive jaundice caused by polycystic liver disease. Radiology 1980;136:435–6.
[4] Wirig JH, Burns R, Longmire WP. Jaundice associated with polycystic liver disease. Am J Surg 1978;136:383–6.
[5] Bae KT, Zhu F, Chapman AB, et al. Magnetic resonance imaging evaluation of hepatic cysts in early autosomal-dominant polycystic kidney disease: the consortium for radiologic studies of polycystic kidney disease cohort. Clin J Am Soc Nephrol 2006;1:64–69.
[6] Everson GT, Taylor MR, Doctor RB. Polycystic disease of the liver. Hepatology 2004;40:774–82.
[7] Jones WL, Mountain JC, Warren KW. Symptomatic non-parasitic cysts of the liver. Br J Surg 1974;61:118–23.
[8] Khoonsari M, Zamani F, Asoubar M, et al. Obstructive jaundice in a patient with polycystic liver. Middle East J Dig Dis 2018;10:117–20.
[9] Dumonta P-N, Rabe A, Mabrut J-Y, et al. Polycystic liver disease complicated by obstructive jaundice. J Vasc Surg 2016;153:149–51.
[10] Raynaud P, Carpentier R, Antonioi A, et al. Lemaigre, Biliary differentiation and bile duct morphogenesis in development and disease. Int J Biochem Cell Biol 2011;43:245–56.
[11] Zhang KH, Yang Y, et al. polycystic kidney complicated with Polycystic Liver disease and polycystic lung. China J Modern Med 2005;15:3680–13680.
[12] Van Keimpema L, De Koning DB, Van Hook B, et al. Patients with isolated polycystic liver disease referred to liver centres: clinical characterization of 137 cases. Liver Int 2011;31:92–8.
[13] Fang T, Yue L, Lin Z, et al. Fatal liver cyst rupture in polycystic liver disease complicated with autosomal dominant polycystic kidney disease: a case report. Forensic Sci Int 2016;262:e5–8.
[14] Zhang Y, Zhang H. Research progress on autosomal dominant polycystic kidney, China Pract Med 2016;11:287–8.
[15] Jia-Ming S, Urology DO, Hospital JC. Application of laparoscopic polycystic kidney decompression. Med Inf 2018;31:91–5.
[16] Wang DJ, Zhang ZX, Wang Y. Efficacy of different blood purification methods in the treatment of hyperbilirubinemia. J Prim Med 2014;475–7.
[17] Zhu DD, Gong DH, Xu B, et al. Combined CVVH-bilirubin adsorption therapy on patients with hyperbilirubinemia. Nephrol Dialy Transplant 2011;20:204–11.
[18] He QP, Gong DH, Wu BY, et al. Treatment of hyperbilirubinemia using a new bilirubin absorber-BS330. Chin J Nephrol Dialy Transplant 2014;23:229–34.
[19] Lee PA, Matson JR, Pryor RW, et al. Continuous arteriovenous hemofiltration therapy for Staphylococcus aureus-induced septicemia in immature swine. Critical Care Med 1993;21:914–24.
[20] Bellomo R, Tipping P, Boyce N. Continuous veno-venous hemofiltration with dialysis removes cytokines from the circulation of septic patients. Crit Care Med 1993;21:522–6.