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CASE IMAGE

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Polycystic liver disease: An uncommon genetic condition

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Key Clinical Message

Timely recognition, accurate diagnosis, and proper management are vital for preventing complications and improving outcomes in polycystic liver disease.

Abstract

Polycystic liver disease is an uncommon genetic condition characterized by the presence of over 20 liver cysts. It is symptomatic in only 5% of cases. Surgical intervention remains the primary treatment approach for managing symptoms in affected patients. Herein, we report a case of PLD revealed by severe abdominal pain.

KEYWORDS

hepatomegaly, liver, liver cysts, pathology, polycystic liver disease, surgery

1 | CASE IMAGE

A 47-year-old nulliparous woman with a history of hypertension, presented with severe abdominal pain, nausea, and discomfort. She had been previously diagnosed with polycystic liver disease (PLD) associated with sporadic autosomal dominant polycystic kidney disease 5 years ago. Notably, her father had also suffered from PLD, which was identified as the cause of his death. Upon admission, the patient's physical examination revealed significant abdominal distension and hepatomegaly, with multiple cysts palpable. Laboratory investigations showed normal liver and renal function results, indicating the absence of any abnormalities. Ultrasound examination confirmed the presence of multiple cystic formations with uniformly homogeneous content, without signs of adjacent fluid, cystic rupture, or hemorrhage (Figure 1A). The abdominal CT scan revealed the nearly complete replacement of hepatic parenchyma by cysts of different sizes, with the largest cyst measuring 11 × 5 cm (Figure 1B). No evidence of ascites was observed. Additionally, both kidneys exhibited enlargement and were extensively affected by multiple cysts of varying sizes. The patient subsequently underwent left hepatectomy with fenestration. Gross examination of the resected liver segment revealed significant enlargement and predominant replacement by multiple cysts (Figure 1C). On cut section, the liver displayed numerous cysts filled with clear fluid (Figure 1D). The cysts had a smooth lining without any papillary excrescences or masses (Figures 1D and 2A).

Histologically, examination of the liver parenchyma revealed numerous diffuse cystic lesions resembling solitary cysts. These cysts were lined by cuboidal to flat biliary epithelium (Figures 2B–D and 3A,B). The lining of cuboidal biliary cells was supported by a thin wall of fibrous tissue. The patient's postoperative course was complicated by hemorrhagic shock, resulting in their unfortunate demise on the second day after the operation while in the intensive care unit.

2 | DISCUSSION

PLD is a rare genetic disorder that can occur independently or in conjunction with polycystic kidney disease.[1] It is characterized by genetic mutations affecting proteins involved

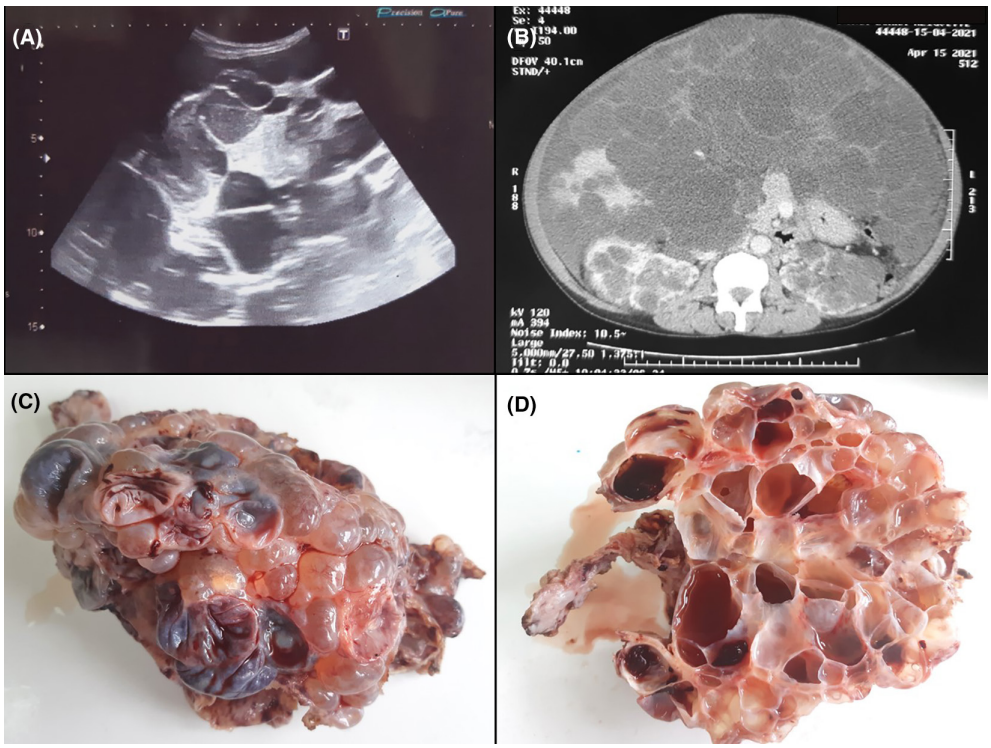

FIGURE 1 (A) Preoperative abdominal ultrasound demonstrates the multiple hepatic cysts of varying size. (B) Preoperative abdominal computed tomography scan shows nearly complete replacement of the hepatic parenchyma by cysts of varying size. (C) Gross findings of polycystic liver disease. The resected liver segment demonstrated marked enlargement with involvement by multiple cysts of various diameters. (D) Cut section of the liver demonstrating cysts of various sizes which contain clear fluid. They have a smooth lining without papillary excrescences or masses.

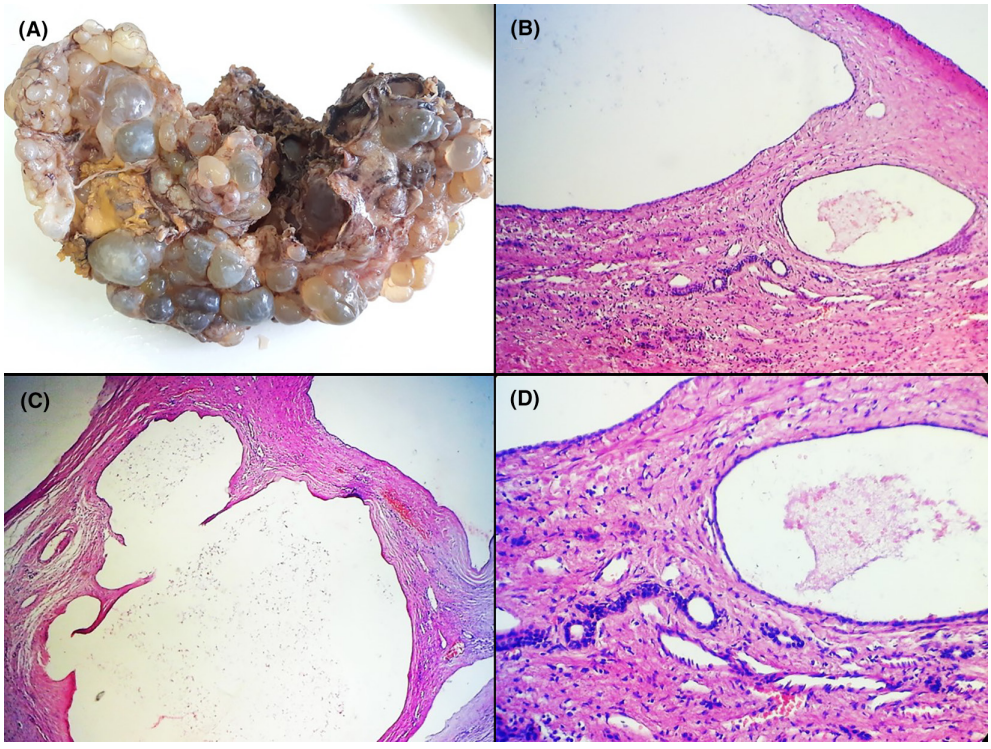

FIGURE 2 (A) The gross findings of polycystic liver disease. The resected liver segment demonstrated marked enlargement with involvement by multiple cysts of various diameters filled with clear fluid replacing most of the hepatic parenchyma. (B) Microscopic examination of liver parenchyma showing cystic lesions lined by cuboidal to flat biliary epithelium, (hematoxylin and eosin, magnification ×40**)**. (C) Cystic lesions resembling solitary cysts, lined by cuboidal to flat biliary epithelium, (hematoxylin and eosin, magnification ×40). (D) Cystic lesions resembling solitary cysts, lined by cuboidal to flat biliary epithelium, (hematoxylin and eosin, magnification ×100)

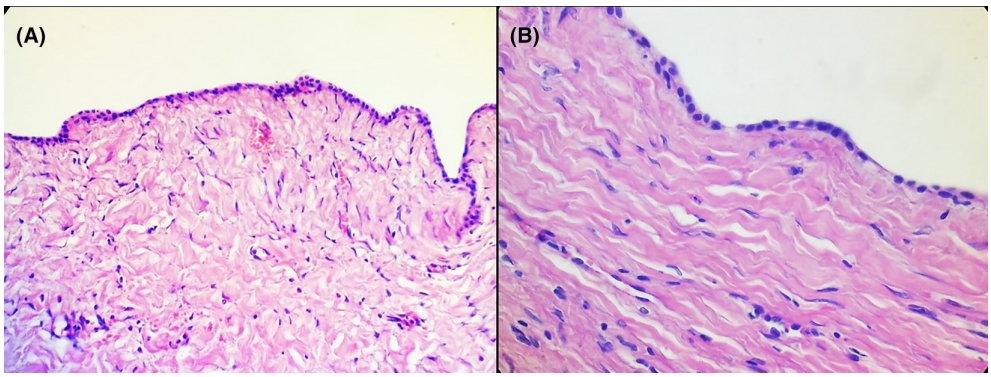

FIGURE 3 (A) Microscopically, the cysts are lined by simple, cuboidal-type epithelium overlying a fibrous wall (hematoxylin and eosin, magnification ×200). (B) High-power view of a cyst wall. Note the lining of cuboidal biliary cells at the top of the image, which rests on a thin wall of fibrous tissue. No atypia and no mitotic figures were noted (hematoxylin and eosin, magnification ×400).

in fluid transport and epithelial cell proliferation within the liver.[1] Diagnosis of PLD is based on factors such as the patient's age, imaging evidence of liver cysts, clinical symptoms, and family history of hepatic and/or renal polycystic disease. While PLD is a progressive condition, only a small number of patients experience severe symptoms. Most individuals with asymptomatic PLD do not require treatment, but a subset of symptomatic patients may benefit from interventions.[1] Surgical intervention is indicated for PLD in cases of symptomatic disease, compromised quality of life, progressive liver enlargement, complications such as cyst infection, hemorrhage, rupture, or biliary obstruction, as well as impaired liver function. Several surgical options are available, including cyst fenestration, liver resection, and liver transplantation. Additionally, ongoing research is investigating medical treatments for PLD.[2,3] Liver transplantation is considered the most effective treatment option for PLD. Given the complexity of the disease, a multidisciplinary team comprising hepatologists, radiologists, and surgeons should collaborate to determine the most suitable management strategy. Regular follow-up visits are essential to monitor disease progression, manage symptoms, and address patient concerns.

AUTHOR CONTRIBUTIONS

Faten Limaiem: Conceptualization; data curation; formal analysis; investigation; methodology; project administration; supervision; validation; visualization; writing – original draft; writing – review and editing. **Mohamed Hajri:** Conceptualization; data curation; formal analysis; project administration; resources; software; supervision; validation; visualization; writing – review and editing.

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CONFLICT OF INTEREST STATEMENT

The authors declare no conflicts of interest.

DATA AVAILABILITY STATEMENT

The data that support the findings of this study are available from the corresponding author upon reasonable request.

ETHICS STATEMENT

All procedures performed were in accordance with the ethical standards. The examination was made in accordance with the approved principles.

CONSENT

Written informed consent was obtained from the patient to publish this report in accordance with the journal's patient consent policy.

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