

## Case Report

# Polycystic liver disease: a rare case study

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### ABSTRACT

Polycystic liver disease is a rare inherited disorder which affects 1 in 1,00,000 population. There are two forms of polycystic liver disease (PLD) i.e. isolated PLD and autosomal dominant PLD which is associated with renal cysts. PLD is usually asymptomatic and incidentally detected. Some may present with complaints of abdominal distension, bloating, early satiety, weight loss. Computed tomography (CT) or magnetic resonance imaging is the investigation of choice to diagnose polycystic liver disease. Below we report you a case of 55-year-old female who came with the complaints of pain abdomen since 8 days and was diagnosed as polycystic liver disease on CT, who received treatment by laparoscopic fenestration surgery of cysts and showed symptomatic improvement.

**Keywords:** Laparoscopic fenestration surgery, Polycystic liver disease, Radiographic findings

## INTRODUCTION

Polycystic liver disease (PLD) is characterized by development of multiple cysts in liver parenchyma due to genetic mutations leading to ductal plate malformations.<sup>1</sup> The disease is usually asymptomatic and an incidental finding but in case of large cysts, there may be symptoms like early satiety, abdominal discomfort, dyspnea, nausea and vomiting due to pressure effect.<sup>2</sup> There are many surgical treatment options available for PLD like percutaneous aspiration, fenestration, hepatic resection and liver transplantation. Treatment is considered only if the disease is severe and the type of treatment depends on the size, number and location of the cysts.<sup>3</sup> The aim of the treatment is to reduce the liver volume and give symptomatic relief to the patient. The objective of this report is to outline the treatment of PLD by laparoscopic fenestration.<sup>4,5</sup>

## CASE REPORT

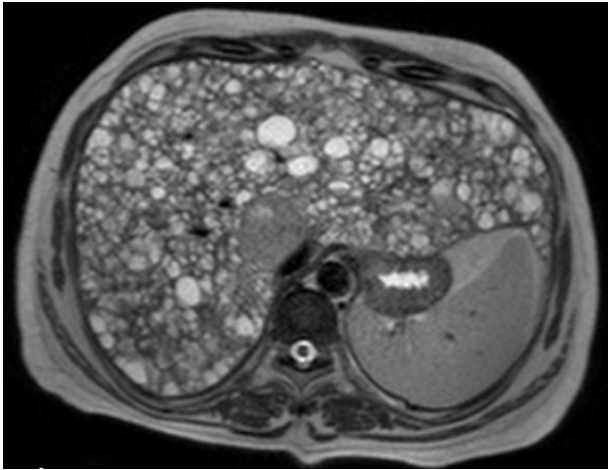
Authors hereby report you a female patient aged 39 years without previous known morbidities with hepatic cysts

which are diagnosed on computed tomography (CT) scan of abdomen. She had family history of autosomal dominant PLD where her mother is a known case. After the diagnosis, patient was under regular follow up.

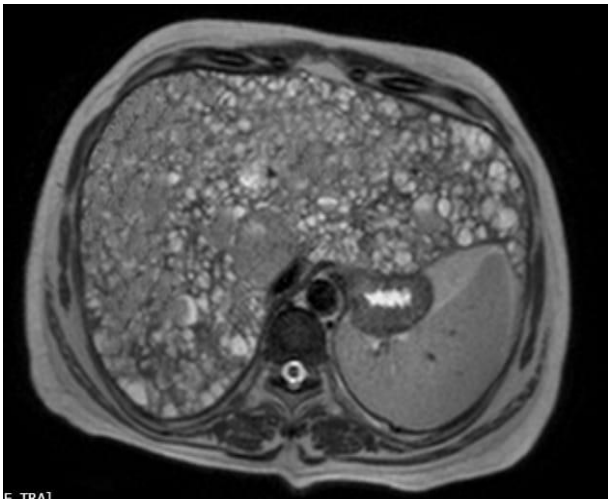
Patient presented to surgery outpatient department with complaints of pain abdomen over right upper quadrant associated with significant weight loss and with no other complaints. Abdominal CT scan was done to confirm the above findings which are suggestive of PLD. Patient was being conservatively managed and advice for monthly close follow-up. After 6 months of follow-up, she had barely maintained her activities of daily living with progressive worsening of fatigue and dyspnea on exertion. Abdominal CT revealed large cysts.

### Investigations

All blood tests and tumor markers were unremarkable. Abdominal CT revealed multiple studded lesions in liver with gross hepatomegaly with small cysts in right kidney. On abdominal CT after 6 months, cyst size has been increased extensively.



**Figure 1: Radiograph before laparoscopic fenestration surgery.**



**Figure 2: Radiograph after laparoscopic fenestration surgery.**

### **Treatment**

Laparoscopic fenestration of cysts was done where multiple cysts in the enlarged liver were opened up and the cysts walls were excised with electrocautery. No surgical complication occurred, and the patient was discharged after 4 days of the surgery. The total liver volume before the surgery was 2400 ml and 4 days after the surgery showing a volume reduction of 10% as calculated on CT volumetry. After surgery, significant improvement in quality of life and daily activity performance was achieved. She is on follow up for 6 months with no complaints and increase in size of the cysts.

### **DISCUSSION**

PLD is a rare inherited disorder where cysts grow 0.9-3.2% annually. The hepatic cysts are rarely diagnosed before puberty and women are frequently diagnosed with symptomatic cysts than men.<sup>6</sup>

Genetic studies confirmed that mutations in SEC63 and PRKCSH genes are associated with defects in functioning of sec63 and hepatocystin which are involved in protein processing, which led to formation of hepatic cysts and mutations in these genes affect proteins such as polycystin-1 and polycystin-2, which are associated with the transport of fluid and growth of epithelial cells, which are the two dominant mechanisms for formation and growth of cysts.<sup>7</sup>

The main risk factor for PLD is having a family member with the disease. Because this inheritance is autosomal dominant, if one family member is affected, there is a chance of other family member being affected is 50%. The most significant factor contributing to massive PLD is female sex. It is believed that female steroid hormone may influence several levels of factors responsible for secretion and growth of liver cysts. The risk may also be related to pregnancy, exposure to exogenous steroid hormones such as contraceptives, female hormonal replacement therapy by post-menopausal women.<sup>8,9</sup> The main symptoms of polycystic liver disease is abdominal distension due to large mass of cysts in the liver. Other symptoms include shortness of breath, early satiety, abdominal discomfort. Patient should undergo ultrasound (USG) or CT imaging of the abdomen, which would reveal the diagnosis. Patients with liver cysts can also experience impingement on the venous drainage of liver, causing pseudo-Budd Chiari syndrome.<sup>3</sup> The diagnosis is considered when Author find >20 isolated cysts in the liver.<sup>10</sup> In case of family history of PLD, presence of >4 cysts is the criteria to diagnose. Liver cysts are usually identified by USG/CT but MRI being most sensitive. Laboratory tests are unspecific and are usually normal. Tumor markers such as CA19.9 can be elevated in 45% of cases due to increased production of biliary epithelium but without associated malignancy.<sup>2</sup> Asymptomatic cysts do not require any active management except close follow up monthly. The current treatment options available for symptomatic cysts are mainly surgical or radiological. Surgical intervention is chosen based on the criteria defined by Gigot et al-taking into account type-I- the size of cysts, type-II- number of hepatic cysts, type-III- area of free hepatic parenchyma in 3 types.

- 1) Patients with less than 10 large cysts (>10 cm), 2) Patients with multiple cysts of medium size but sparing large areas of liver, 3) Massive spread of cysts affecting almost the entire liver.

Type I and II- aspiration and sclerotherapy, laparoscopic fenestration and partial hepatectomy. Type III- procedures are ineffective.<sup>11</sup> The most widely used treatment is reducing the bulk of liver which can be achieved by laparoscopic fenestration/cyst aspiration. Hepatectomy and liver transplant surgery are the final treatment options available. Liver transplantation is not a simple procedure, as the size of the liver and the impingement of the adjacent structures in these patients makes transplantation difficult. The early mortality rate

from performing the operation ranges from 10%-20%, though once patient recovers from immediate postoperative period (approximately 3 months), their long-term survival after transplant is excellent.

Medical therapies are currently under investigation in clinical trials but are not preferred because of its high cost and no proven efficacy.

## CONCLUSION

PLD is a rare disorder and most of the patients are asymptomatic. This patient was symptomatic hence surgical intervention was planned. Laparoscopic fenestration is a safe and effective procedure in the treatment of symptomatic PLD. PLD if managed appropriately will render the patient asymptomatic and therefore deserves mention.

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## REFERENCES

1. Pantè S, Di Dio V, Putorti A, Salvo A, Barbera N, Catalfamo G, et al. Laparoscopic cyst fenestration in the treatment of polycystic liver disease. *Ann Ital Chir.* 2014;85(3):298-303.
2. Arnold HL, Harrison SA. New advances in evaluation and management of patients with polycystic liver disease. *Am J Gastroenterol.* 2005;100(11):2569.
3. Everson GT. Polycystic liver disease. *Gastroenterol Hepatol.* 2008;4(3):179.
4. Szabó LS, Takacs I, Arkosy P, Sapy P, Szentkereszty ZS. Laparoscopic treatment of nonparasitic hepatic cysts. *Surg Endos Other Inter Techniq.* 2006;20(4):595-7.
5. Van Keimpema L, Ruurda JP, Ernst MF, Van Geffen HJ, Drenth JP. Laparoscopic fenestration of liver cysts in polycystic liver disease results in a median volume reduction of 12.5%. *J Gastrointest Surg.* 2008;12(3):477-82.
6. Grantham JJ, Torres VE, Chapman AB, Guay-Woodford LM, Bae KT, King Jr BF, et al. Volume progression in polycystic kidney disease. *New Engl J Med.* 2006;354(20):2122-30.
7. Temmerman F, Missiaen L, Bammens B, Laleman W, Cassiman D, Verslype C, et al. Systematic review: the pathophysiology and management of polycystic liver disease. *Aliment pharmacol Therapeut.* 2011;34(7):702-13.
8. Qian Q, Du H, King BF, Kumar S, Dean PG, Cosio FG, et al. Sirolimus reduces polycystic liver volume in ADPKD patients. *J Am Soci Nephrol.* 2008;19(3):631-8.
9. Qian Q. Isolated polycystic liver disease. *Advan Chronic Kidney Dis.* 2010;17(2):181-9.
10. Gevers TJ, Drenth JP. Diagnosis and management of polycystic liver disease. *Nature Rev Gastroenterol Hepatol.* 2013;10(2):101.
11. Gigot JF, Hubert C, Banice R, Kendrick ML. Laparoscopic management of benign liver diseases: where are we?. *HPB (Oxford).* 2004;6(4):197-212.

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