

CASE REPORTS

Ultrasound-guided percutaneous drainage and sclerotherapy in a patient with isolated autosomal dominant polycystic liver disease

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ABSTRACT

Isolated polycystic liver disease (IPLD) is a rare genetic condition characterized by the presence of multiple liver cysts with no association with polycystic kidney disease. Most patients are asymptomatic and acute complications (cyst torsion, bleeding, infection) are uncommon. Imaging techniques, including abdominal ultrasounds, computerized axial tomography, and magnetic resonance imaging, represent a vital diagnostic modality. They are also useful for therapy support in this disease. Below we report a peculiar case of a female patient recently diagnosed with IPLD who, having received treatment with ultrasound-guided percutaneous drainage and sclerotherapy for a giant liver cyst, showed symptom and laboratory improvement.

Key words: Isolated autosomal dominant polycystic liver disease. Ultrasound-guided percutaneous drainage. Sclerotherapy.

INTRODUCTION

Isolated polycystic liver disease (IPLD) is a rare genetic condition recognized as different from polycystic kidney disease (1).

IPLD is a hereditary autosomal dominant disorder that manifests with multiple cysts throughout the liver parenchyma that arise from dilated biliary micro-hamartomas.

Barbado-Cano A, Moreno-López M, Olveira-Martín A, Cerpa-Arencibia A, Fernández-Martos R, Suárez-de-Parga JM. Ultrasound-guided percutaneous drainage and sclerotherapy in a patient with isolated autosomal dominant polycystic liver disease. Rev Esp Enferm Dig 2015;107:175-177.

Received: 12-04-2014
Accepted: 01-07-2014

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Its natural history is characterized by an increase in liver cyst volume and numbers (2).

Within the spectrum of illness, polycystic liver disease is a most silent, unknown condition (1,3,4).

We report the case of a female patient with symptomatic IPLD, with no apparent family history, who had a giant liver cyst treated with percutaneous drainage and sclerotherapy, which resulted in a good sustained response.

CASE REPORT

A 48-year-old woman with a history of endometriosis, uterine myoma treated with embolization and myomectomy, and no toxic habits (smoking, alcoholism). No children. Parents and siblings with no relevant histories. She seeks help for growing abdominal perimeter, epigastric discomfort with heartburn, postprandial bloating, and diffuse, sustained abdominal pain more severe in the right hypochondrium. Physical examination reveals a distended abdomen where an indurated, tender mass may be palpated in the right hypochondrium, epigastrium, and mesogastrium. Neither splenomegaly nor ascites may be identified. Laboratory tests show increased GGT levels in an otherwise uneventful profile (CBC, urinalysis, chemistry, proteinogram, coagulation study, hepatotropic virus and tumor markers). An abdominal computerized tomography (CT) scan reveals hepatomegaly with countless cystic lesions, among which a 18-cm cyst in the right liver lobe (RLL) stands out (Fig. 1); no relevant findings in bile ducts, pancreas or spleen. Pelvic right kidney from liver displacement. Small bilateral renal cysts. Calcified uterine myomas. Abdominal ultrasounds confirmed the diagnosis of polycystic liver disease with a giant cyst in RLL (Fig. 2). The absence of significant kidney cysts and unmet polycystic kidney disease criteria allow a diagnosis of IPLD to be reached. The patient is subsequently admitted to the gastroenterology ward where a percutaneous drainage under sonographic guidance is performed on

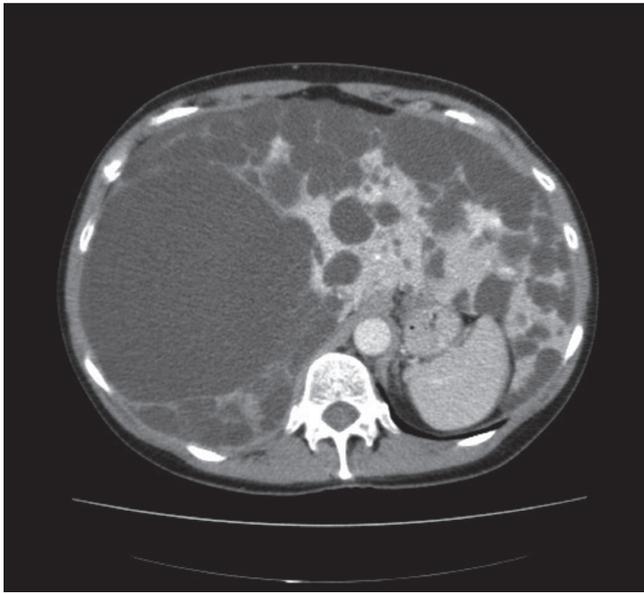


Fig. 1. Abdominal computed tomogram showing hepatomegaly with multiple liver cysts, one of them 18 cm in diameter in the right lobe of the liver.

the giant liver cyst using an 8F pigtail catheter, obtaining a clear fluid, with no immediate complications. On a second stage that same day the radiology department performs a transcatheter cystography (Fig. 3) where no evidence of contrast leakage is seen, and 270 mL of pure ethanol are instilled over 30 minutes, which are then aspirated. The patient was then discharged with both clinical and laboratory improvement and no immediate complications. She is still followed up at the clinic and remains asymptomatic, with no additional sclerotherapy sessions required. Four years after the procedure an abdominal CT scan was performed which showed the decreased size of the previously treated liver cyst (8.4 cm).

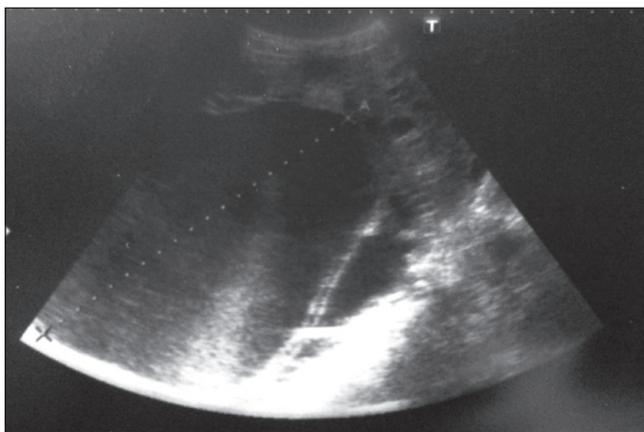


Fig. 2. Abdominal ultrasounds showing multiple anechoic round lesions, the biggest one -18.3 cm in size- located in the right lobe of the liver.

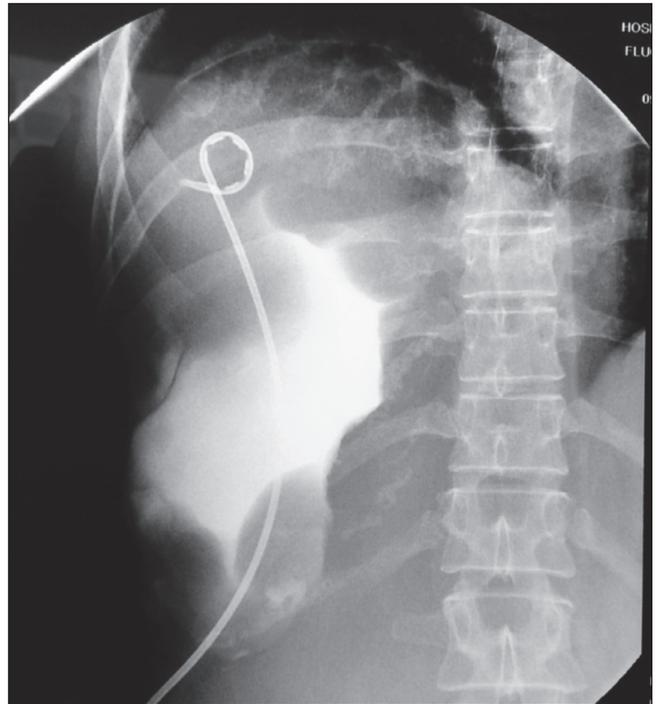


Fig. 3. Transcatheter cystography showing no contrast leaks.

DISCUSSION

Polycystic liver disease represents a group of genetic conditions where liver cysts occur either alone (IPLD) or in association with kidney cysts (autosomal dominant polycystic kidney disease, ADPKD) (2).

IPLD occurs more commonly in the fourth decade of life (4,5), and usually develops earlier and is aggravated by multiple pregnancies (3,4). During childhood its incidence, although not well established (4,6), is estimated to be lower than 0.01 % (1,7). Therefore, it can be considered a rare or minority disease (8).

The underlying genetic origin encompasses mutations in two genes: Gene PRK CSH, in the short arm of chromosome 19 (19p 13.2- 13.1), and gene SEC 63, in the long arm of chromosome 6 (6q21- q23), which code for proteins hepatocystin and Sec 63, respectively (1). However, genetic studies suggest clinical heterogeneity among family members, most of them being asymptomatic. Penetrance is estimated around 80 % (14).

The diagnostic criteria for IPLD in patients with no family history is the presence of over 20 liver cysts (15). The case reported herein has no family history of polycystic liver disease but shows over 20 liver cysts in diagnostic images, and does not meet Ravine criteria for ADPKD (more than two cysts in each kidney according to age) (2). Genetic testing is now only recommended for patients with a positive family history and severe hepatic disease (3). Nevertheless, our patient declined to sign an informed consent for genetic tests.

Most patients with IPLD are asymptomatic (3-6). However, starting in the fourth decade of life, cyst growth may result in clinical manifestations due to a mass effect, including tender hepatomegaly and dyspepsia, as was the case with our patient. Cyst-related complications such as infection, rupture, bleeding, and malignant conversion may rarely occur (1,3,9).

Extrahepatic, non-cystic, vascular manifestations may develop in 6 % of patients, including intracranial aneurysms and heart valve disorders (mitral regurgitation or prolapse) (1,4). This suggests that IPLD is not confined to the liver and may well be considered a systemic condition. The above case report has no extrahepatic manifestations. The fact should be highlighted that indiscriminate screening is not currently recommended to rule out additional manifestations (1).

Therapy for symptomatic IPLD is not well established. Medical treatment is ineffective. Percutaneous aspiration with cyst sclerosis, fenestration, surgery with partial hepatectomy, and rarely liver transplantation are all recommended, but no management protocol has been established (3-6,10-13). Some authors accept that surgical resection is the treatment of choice (6,10). However, in the above-reported case, a minimally interventional procedure such as percutaneous drainage and ethanol sclerotherapy represents an effective therapy. This technique requires an instillation of sclerosing substances to inhibit fluid build-up by damaging the cyst epithelium. Most commonly used substances include ethanol, minocycline and tetracycline. In IPLD, it is recommended when a predominant, symptomatic cyst over 5 cm is present and accessible to percutaneous drainage (2). This technique has a wide safety margin but may bring about complications, including peritoneal irritation from sclerosing agent leaks onto the peritoneum. The reported case had no immediate complications following the procedure, was followed up at the outpatient clinic, and showed symptom remission. Due to the difficulties entailed by the minority nature of IPLD,

only a multicenter, prospective study may protocolize a therapeutic option of choice.

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