Isolated Polycystic LiverDisease:A Case Report with Review of Literature

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Abstract

Isolated polycystic liver disease is a rare condition that was not reported in Libya or North Africa as we found when reviewing literature available on it. In this paper we present a case of a 70-years-old Libyan male that attended Sebha Medical Center witha history of right hypochondrial swelling and pain of three months' duration. Oninvestigation, ultrasound showed numerous liver cysts with no renal involvement. The following is a review of literature pertaining cystic liver disease.

Introduction

Simple liver cysts are common finding on ultrasound examination of the abdomen[1]. When the patient has more than 20 liver cysts, he is diagnosed with polycystic liver disease (PLD).Since 1856, when PLD was first described in patients with autosomal dominant polycystic kidney disease (ADPKD)[2], it was believed that it can only develop in association with ADPKD. This belief remained dominantuntil 2003, when it was found that there were many types of polycystic liver disease and some of them can occur alone [3].Now it is known that (PLD) can develop in three forms: the first is isolated polycystic liver disease (PCLD), the second is in association with ADPKD and the third form is as a part ofVonMeyenburg complexes[4].

Studies have shown that Isolated polycystic liver disease (PCLD)is linked to mutations in PRKCSH and SEC63 genes. Both of these genes have an autosomal dominant mode of inheritance[5,6,7].A study conducted in Netherland in 2010 by Van Keimpema et al. estimated the prevalence of PCLD to be around 1 per 158000[8], but Lantinga et al. in a review published in 2013, think that this

figure is an underestimation. For comparison, the prevalence of autosomal polycystic kidney disease (ADPKD), which is the most common inherited renal disorder [9],was estimated by various studies to be between 1/400 and 1/1,000[10,8]. Their belief is based on the fact that the Dutch study was conducted on 137 patients from five tertiary referral centers, and that the patients who present to these centers are the symptomatic patients [11] who constitute about 20% of PCLD case, as this disease is commonly asymptomatic[12].

When we reviewed the literature available in online medical databases on this condition, we found no reported cases from North Africa including Libya. Our belief is that this might be due to under reporting of this condition, and the goal of this article is to report the case we encountered, and provide a brief review of the literature regarding PCLD.

Case presentation

A 70-years-old Libyan male, came to Sebha Medical Center complaining of right hypochondrial swelling and pain for the last three months. On further questioning, he admitted to have weight loss, shortness of breath on exertion, generalized fatigability and fever.

On examination he was conscious and oriented but had severe pallor. His blood pressure was normal. On cardiac auscultation, he had normal first and second heart sound with to a systolic murmur. His breath sounds were decreased bilaterally and he had bilateral crepitations which were more on the left side. Examination of the abdomen revealed soft lax abdomen with hepatomegaly. No abnormalities were detected on neurological examination.

Routine investigations were ordered in addition to ultrasound examination for abdomen and pelvis. CBC showed normocytic normochromic anemia with HB= 7.1g/l, MCV= 94fl, MCH=26.4pg. GUE showed urinary tract infection with 5-6 pus cells in HPF while urobilinogen was +, Bilirubin in urine was ++. ESR value was above 100mm in the first hour. Blood glucose, renal functions tests and electrolytes were all normal and virology examination for HIV, HBsAg and HCV was negative. Liver functions tests showed abnormal results in the form of total bilirubin value of 1.4 mg/dl, direct bilirubin was 1 mg/dl, while GPT was 127 U/L, GOT = 153 U/L, ALP = 630 U/L.

Echocardiography showed signs of ischemic heart disease, mild LV enlargement and moderate TR. Ultrasound examination of the abdomen revealed hugely enlarged liver that its dimensions could not be measured, with numerous cysts of different sizes in both lobes of liver. The largest cyst measured about 9.9cm x 8.2 cm (Fig.1), and the smallest one was about 4.2 cm x 4.1 cm. One of the large cysts contained thick fluidwhich was consistent with hemorrhage (Fig.2). The patient was evaluated with CT scanwhich confirmed these findings (Fig.3). The relative took the patient to Tripoli Medical Center after the diagnosis wasmade.

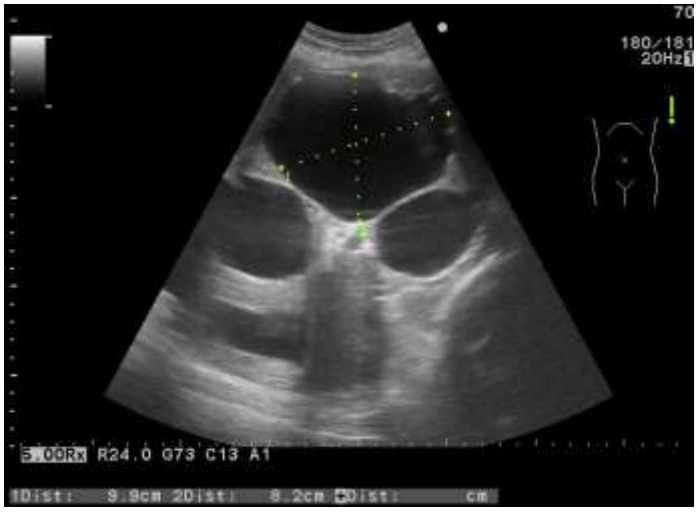

Fig. 1: Ultrasound image showing the patient's liver with some of its multiple cysts, with the measurements of the largest one.

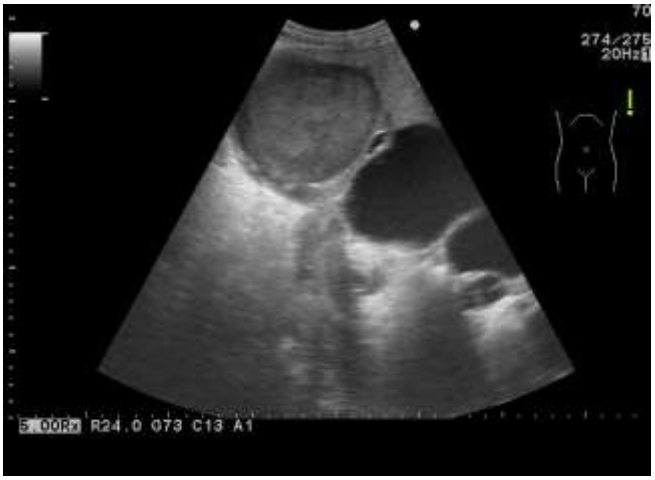

Fig. 2: Ultrasound image showing hemorrhage in one of the liver cysts.

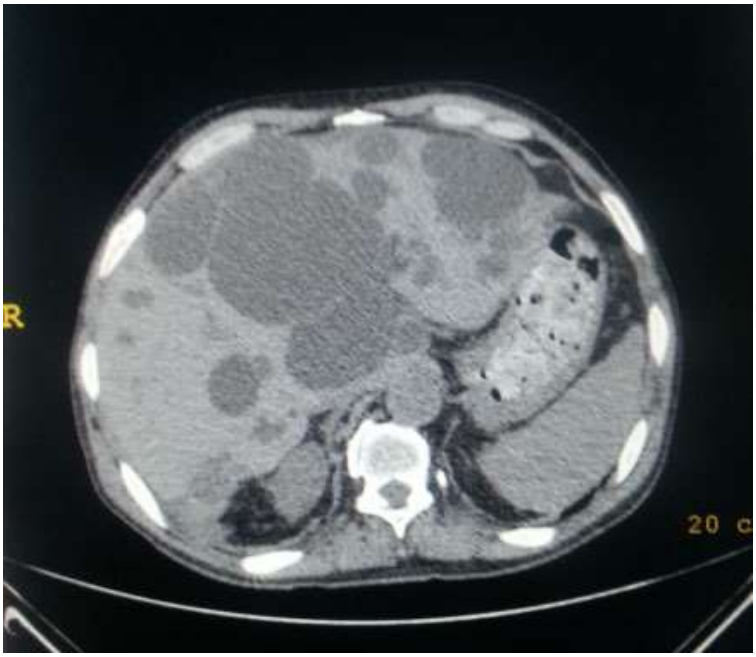

Fig. 3: Axial section from non-contrast enhanced CT scan of the patient abdomen showing his enlarged liver that contains multiple cysts.

Discussion

PCLD is a rare disease worldwide, and our search of literature showed that there are no previous reported cases from Libya or North Africa. This disease does not cause any symptoms in more than 80% of its patients [12]. In the remaining 20%, the uncomplicated cysts can lead to hepatomegaly which causes abdominal distension and discomfort in some patients. The complications of liver cysts can add more symptoms to the patient presentation. Patients having hemorrhage in the cysts can experience acute pain, while cyst infection leads to fever. The liver cysts can also compress the liver vessels and inferior vena cava leading to portal hypertension [13], and in some cases they exert pressure on the surrounding structures leading to early satiety, nausea and vomiting [14]. Obstructive jaundice is a rare complication of PCLD [13].

When complications of PCLD were studied, it was found that about 10% of untreated patients developed complications. The most common one was cyst bleeding, which was found in 8% of the untreated. The second was portal hypertension, which was found in 3% of the patients. Each of ascites and inferior vena cava compression was found in 2% of the untreated cases [8].The case we encountered fits the classical presentation of

the condition and its most common complication, as it presented with symptoms of liver enlargement and had hemorrhage in one of the cysts on ultrasound. The imaging findings of our case were classical and agreed with the described literature with the presence of the cysts and there effect on the liver size and the investigations showed the abnormality in liver functions that can result from this condition. Symptomatic patients with PCD have slight increase in Alkaline phosphatase (ALP) and total bilirubin levels [12], and elevated levels of gamma-glutamyltransferase (GGT) was found in 60-70% of symptomatic cases [15], but these abnormal values are commonly found in the symptomatic patients only, and asymptomatic patients usually have normal liver functions [16].Elevation of serum levels of the gastrointestinal tract tumor marker CA19-9 was found in about 45% of patients with PCLD. On further evaluation, the level of this marker was found to be high in cyst fluid of simple and polycystic liver disease, with no evidence of tumors[17].Treatment is given to cases with severe symptoms or with complication. [18].There are both medical and surgical options for managing patients with PCLD. The medical approaches depend on using analogs for cimetidine and somatostatin to reduce fluid secretion inside the cysts [15,19,20].The surgical options are aspiration and sclerotherapy, fenestration, liver Resection and liver transplantation[14].

Conclusion

Isolated polycystic Liver disease (PCLD) is a rare genetic disorder that is usually asymptomatic and presents with consequences of hepatomegaly and complications of the cyst in symptomatic patients. Its prevalence is estimated to be around 1 per 158000in Netherlands [8], but there are no reported cases or major studies about its prevalence or features in the literature we reviewed from Libya and North Africa. Our case report can be the beginning of efforts to fill this gap in literature.

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