

CT scan diagnosis of hepatic adenoma in a case of von Gierke disease

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Abstract

Hepatic adenoma is a well-defined, benign, solitary tumor of the liver. In individuals with glycogen storage disease I, adenoma tends to occur at a relatively younger age and can be multiple (adenomatosis). Imaging plays a pivotal role in diagnosing hepatic adenoma and in differentiating adenoma from other focal hepatic lesions. Especially in patients with von Gierke disease, in addition to the associated hepatomegaly caused by steatohepatitis and the diffusely reduced attenuation of the liver parenchyma seen on CT, there may be more than one hepatic adenoma in up to 40% of patients. Malignant degeneration of hepatic adenoma into hepatocellular carcinoma can occur and hence imaging is important for prompt diagnosis of adenoma and its complications. In this case report, we present a case of liver adenoma diagnosed by CT scan in a patient with von Gierke disease.

Key words: CT; Hepatic adenoma; von Gierke disease

Introduction

Hepatic adenoma is the second most common benign tumor of the liver in adults.^[1] In children, hepatic adenoma is rare and, when encountered, is generally seen in association with an underlying glycogen storage disorder.^[1] CT scan is useful to detect adenoma in patients with glycogen storage disorder and helps in differentiating adenoma from other focal liver masses like hepatoblastoma, hepatoma, etc. We present a rare case of a 12-year-old boy with von Gierke disease (glycogen storage disease type I) in whom hepatic adenoma was detected on CT scan and confirmed by histopathological examination.

Case Report

A 12-year-old boy was brought to the hospital by his parents with upper abdominal discomfort for 15–20 days.

On physical examination the child was found to have retarded growth (dwarfism) and a pendulous abdomen. Per abdominal examination revealed hepatomegaly. USG study of the abdomen showed hepatomegaly with diffusely raised echogenicity and a 7.5 × 4.5 cm-sized, well-defined, hypoechoic mass lesion in segment VI of the right lobe of the liver [Figure 1]. CT scan of the abdomen showed hepatomegaly with diffuse low attenuation of the liver and a 7.5 × 4.5 × 4.8 cm well-circumscribed, low-attenuation, subcapsular focal lesion in segment VI of the right lobe; this lesion showed mild to moderate heterogeneous contrast enhancement and had a nonenhancing area within it [Figures 2 and 3]. Multiple small bilateral renal calculi were also noted [Figure 2]. Hepatomegaly with diffusely low attenuation of liver, bilateral renal calculi (which hinted at hyperuricemia – a known feature of von Gierke disease), clinical features of dwarfism, facial dysmorphism, and protuberant abdomen, were all features that favored the diagnosis of von Gierke disease. Moreover, in the pediatric age-group, liver tumors are rare and hepatic adenoma occurs almost exclusively in patients with underlying von Gierke disease. CT scan features like well-circumscribed margins, subcapsular location, and contrast enhancement also supported the diagnosis of hepatic adenoma. Based on these CT scan findings and clinical features, a probable diagnosis of hepatic adenoma in a case of type Ia glycogen storage disease (von Gierke disease) was suggested.

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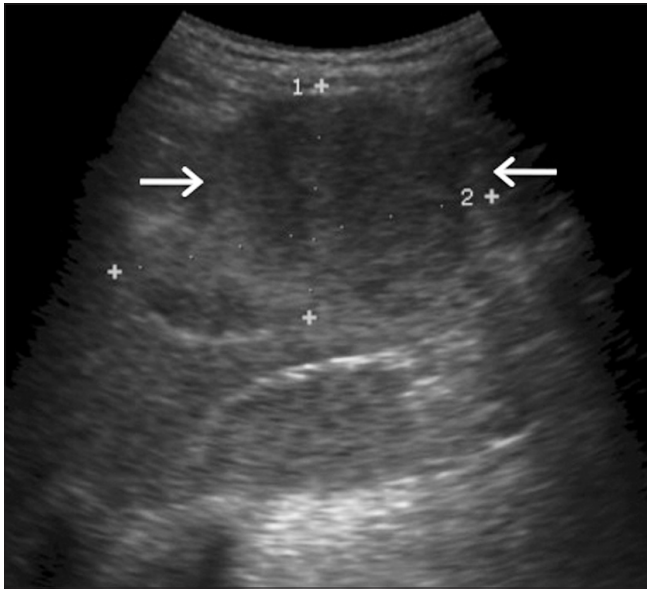


Figure 1: Gray-scale USG shows hepatomegaly with diffusely increased echogenicity and a well-defined predominantly hypoechoic mass (arrows) in segment VI of the right lobe of the liver

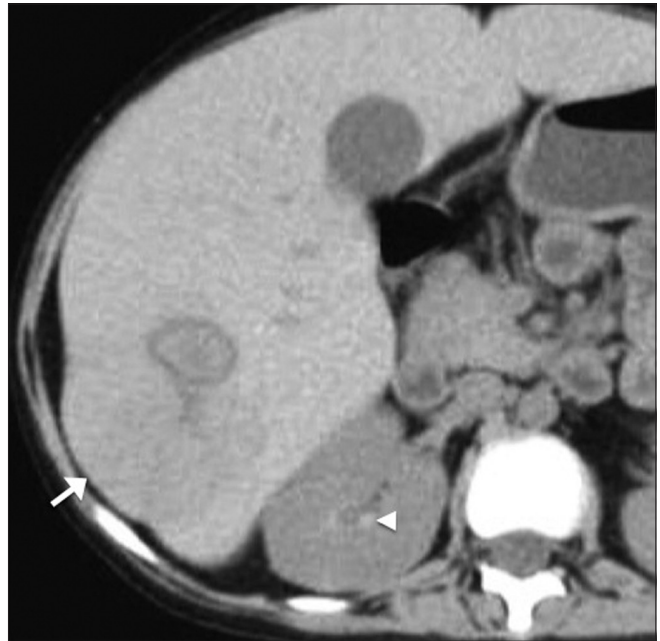


Figure 2: Axial precontrast CT scan of the abdomen shows hepatomegaly with diffusely low attenuation of the liver and a well-circumscribed, low-attenuation, subcapsular focal lesion (arrow) in segment VI of the right lobe. Note the bilateral small renal calculi (arrowheads)

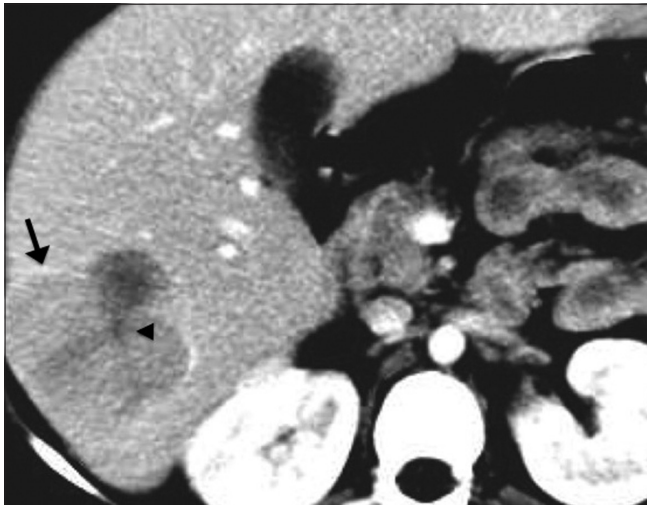


Figure 3: Axial contrast-enhanced CT scan of the abdomen shows capsular enhancement (arrow) with mild to moderate heterogenous contrast enhancement of the tumor (arrowhead); there is a nonenhancing area within the tumor as well

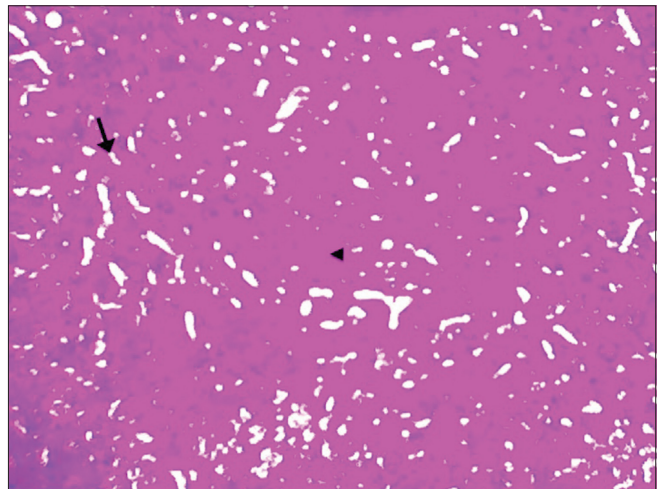


Figure 4: Histopathological picture of the adenoma shows the paucity of hepatocytes and bile ductules (arrows), with the presence of significant fatty change in the background liver (arrowhead), PAS stain

Preoperative laboratory evaluation revealed increased uric acid levels and raised cholesterol. The random blood sugar level was 60 mg%. Liver function tests were within normal limits.

The patient was operated and the mass in the liver was surgically resected. The specimen was sent for histopathological examination along with a small bit of liver tissue, which showed PAS-positive glycogen-rich hepatic cells, confirming glycogen deposition in the liver, while the lesion itself was confirmed to be a hepatic adenoma [Figure 4].

Discussion

Hepatic adenomas are well-defined, benign, focal hepatic masses surrounded by fibrous pseudocapsules that are formed due to compression of the adjacent liver tissue. Hepatic adenomas are generally solitary and occur in females on long-term oral contraceptive pills or in users of anabolic steroids. Association with diabetes mellitus and von Gierke type Ia glycogen storage disease is also known. These adenomas are associated with high

incidence of hemorrhage, necrosis, and fatty change and, especially in pregnancy, increased tumor growth rate and risk of tumor rupture. In von Gierke disease, one or more hepatic adenomas are reported to be present in up to 40% of patients that occur by the second or third decade, and can hemorrhage and, in some cases, become malignant.^[2] Imaging plays a vital role in the timely diagnosis of such lesions. Early diagnosis is important as these tumors are known to transform into hepatocellular carcinoma.^[2,3]

Type I glycogen storage disease (glucose-6-phosphatase or translocase deficiency; von Gierke disease) is a rare autosomal recessive disorder caused by glucose-6-phosphatase deficiency in the liver, kidney, and intestinal mucosa. It has an incidence of approximately 1 in 100000. It can be divided into two subtypes: type Ia, in which the glucose-6-phosphatase enzyme is defective, and type Ib, in which the translocase that transports glucose-6-phosphate across the microsomal membrane is defective. The defects in both subtypes lead to inadequate conversion in the liver of glucose-6-phosphate to glucose and thus make affected individuals susceptible to fasting hypoglycemia.^[2,4]

Usually, a child suffering from von Gierke disease presents with hepatomegaly at around the age of 3–6 months. However, neonates can present with hypoglycemia and lactic acidosis. Dwarfism, doll-like faces, thin extremities, and a protuberant belly due to massive hepatomegaly are other distinct and common clinical features. The hepatomegaly is due to glycogen and fat accumulation in hepatocytes. Despite the hepatomegaly, liver enzymes are usually normal or near normal. Renomegaly is a known feature, but the spleen and heart are not enlarged. Almost all patients older than 20 years have proteinuria, and many have hypertension, kidney stones, nephrocalcinosis, and altered creatinine clearance.^[5]

Hyperuricemia is present. Hyperlipidemia includes elevation of triglycerides, cholesterol, and phospholipids. Type Ib patients have additional findings of neutropenia and impaired neutrophil function, with resultant recurrent bacterial infections and oral and intestinal mucosal ulceration.^[4,5]

Pathologically, the adenoma appears as well-circumscribed mass on the external surface of the liver. Grossly, it is a soft, pale or yellow/tan-colored mass with large areas of hemorrhage or infarction and a pseudocapsule. Microscopic examination of hepatic adenomas reveals sheets or cords of hepatocytes, absence of portal and central veins and bile ducts, increased amounts of glycogen and lipid, and scattered thin-walled vascular channels within the mass.^[6]

On USG, the adenoma tends to be hyperechoic due to the fat and glycogen content. Hemorrhage can also modify its

appearance, with recent hemorrhage being hyperechoic and old blood, hypoechoic (thus mimicking a cyst). Prominent subcapsular vessels if any are detected by color Doppler imaging. Often the USG and Doppler appearance is similar to focal nodular hyperplasia.

On CT scan and MRI, adenomas typically appear as spherical, well-defined, hypervascular and heterogenous masses due to hemorrhage and fat and they may mimic focal nodular hyperplasia (FNH). However, adenoma can be differentiated from FNH by the fact that the latter shows homogeneous enhancement during the arterial phase of imaging and a central scar, while a hepatic adenoma enhances heterogeneously and does not show a central scar. On MRI, hepatic adenomas generally appear predominantly hyperintense on unenhanced T1W images and heterogeneously hypointense on T2W images. On in and out-phase MR images, signal loss is often noted due to fatty components within the adenomas. On contrast-enhanced studies performed using liver specific agents like Gd-EOB-DTPA, hepatic adenomas frequently enhance in the arterial phase, with late washout. On hepatocyte phase imaging by using gadobenate dimeglumine, adenomas characteristically appear hypointense because of the lack of biliary canaliculi that helps to distinguish the adenomas from focal nodular hyperplasia.

Hepatocellular carcinoma (HCC) can also be a differential diagnosis of adenoma, however contrast MRI can help to differentiate HCCs from adenomas. HCCs have increased vascularity and show increased enhancement in the arterial phase and increased washout on delayed images. MRI contrast agents that show biliary tracts (eg, gadoxetic acid), show uptake in the adenoma but not in an HCC as HCCs are hepatocellular in origin.^[7,8]

In conclusion, imaging is helpful in diagnosing hepatic adenoma in patients with von Gierke disease.

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