Case Report

An adolescent girl in Hong Kong with type Ib Abernethy malformation complicated by multiple focal nodular hyperplasia ☆,☆☆

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ABSTRACT

Congenital portosystemic venous shunts are developmental anomalies. They represent portal communication with the systemic circulation. The pathogenesis is linked to the complexity of the embryological development of the inferior vena cava and portal vein. We reported a case of an asymptomatic 14-year-old Chinese adolescent girl in Hong Kong with a confirmed congenital portosystemic shunt type 1b. The condition can be diagnosed using contrast-enhanced CT scans and MRIs. Early recognition of the condition is important due to elevated risks of developing hepatocellular tumours. Liver transplantation may be considered curative.

Introduction

Congenital portosystemic shunts within the hepatic parenchyma are not uncommon. Most adult patients with portosystemic shunts present with bleeding or hepatic encephalopathy as first manifestations [1]. It has been hypothesized that patients become more symptomatic with advancing age as the tolerance for high ammonia levels diminishes [2]. Ultrasound may reveal drainage of portal vein into inferior vena cava. Contrast-enhanced CT scan and MRI scan of abdomen were performed which confirmed the diagnosis of type 1b Abernethy malformation without associated major anomalies. The common clinical presentations, associated anomalies, diagnostic workup and treatment options of this disorder were discussed.
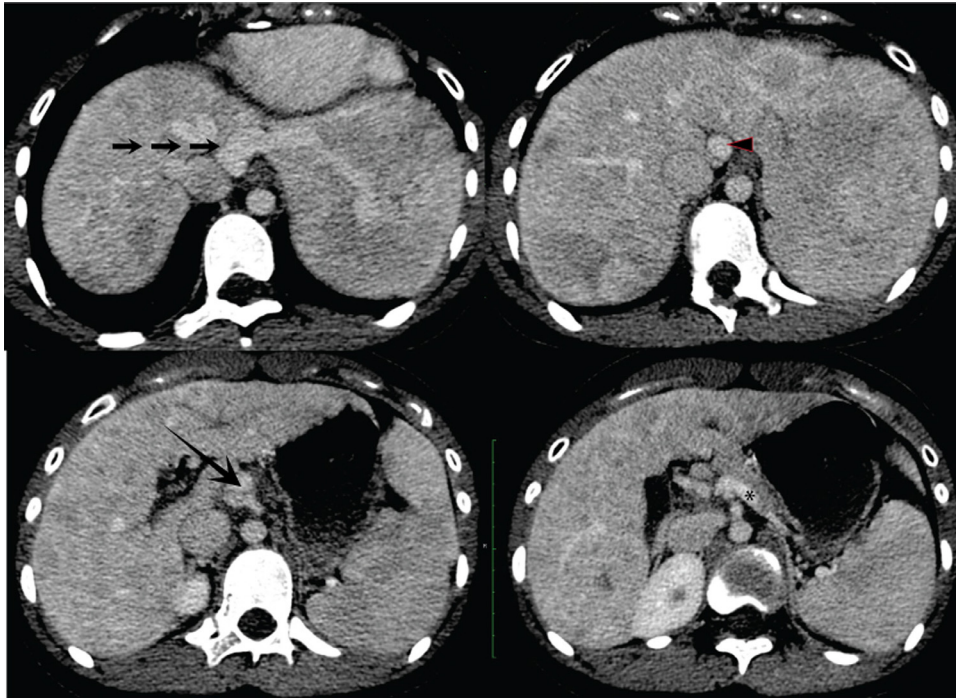
Fig. 1 – Axial images of CT liver. Absence of intrahepatic portovenous system. The splenic vein (asterisk) & superior mesenteric vein (arrow) drain into a common vein (arrowhead) that drains directly into the intrahepatic IVC at the hepatic venous confluence level (arrow array).

Case report

A 14-year-old Chinese adolescent girl was evaluated for deranged liver function test results in Hong Kong. Her past medical history included preterm birth (34 gestational weeks), repaired cleft palate, bilateral conductive hearing loss, spina bifida occuta treated with laminectomy, perimembranosus ventricular septal defect with closure done, treated right cataract and retinal detachment, cerebral atrophy. She had no active use of any medication. She denied alcohol or cigarette use. Physical examination was positive for hepatomegaly, negative for abdominal tenderness, guarding or rebound.

Blood test results were available, ie. Alanine aminotransferase (85 U/L), total bilirubin (22 μmol/L), alkaline phosphatase (206 U/L). GGT 284 IU/L (abnormal), ammonia 87.3 μmol/L (abnormal) AFP 2 SI μg/L. Sodium, potassium, chloride, bicarbonate, creatinine and glucose level were otherwise all within normal limits.

Her CT scan revealed hepatomegaly. Furthermore, the main portal vein was formed by the splenic vein and superior mesenteric vein as usual but neither left or right main portal vein was present, ie, absence of intrahepatic portovenous system. The splenic vein and superior mesenteric vein drained into a common vein that led directly into the intrahepatic IVC at hepatic venous confluence level (Fig. 1 and Fig. 2). Hypertrophic hepatic arteries arising from the celiac axis were noted. The hepatic artery was the single vascular supply to the liver.

Liver showed heterogeneous contrast enhancement. Patchy hypoenhancing hepatic lesions were seen on both lobes in arterial and portal-venous phase, which became more isodense in the delayed phase.

Primovist (Gadoxetate disodium)-enhanced MRI scan detected absence of intrahepatic portal venous system. The splenic vein and superior mesenteric vein joined into a common vein that drained directly into the distal IVC (HV confluence region). Those findings were suggestive of type 1b congenital extrahepatic portosystemic shunt (CEPS).

Multiple focal hepatic lesions were present. Most were T1 hyperintense, T2 hypointense, with mild homogeneous arterial hyperenhancement and hyperintensity on hepatobiliary phase (Fig. 3). No washout or capsule was noted. Some lesions displayed a central feeding vessel and central scar. Findings were suggestive of focal nodular hyperplasia. On the other hand, the patient did not have other risk factors of hepatocellular carcinoma, which could be an important differential diagnosis part from focal nodular hyperplasia. She was not a hepatitis B carrier and has no history of transfusion or body tattoos. The lesions were unchanged in appearances and sizes in subsequent annual follow-up MRI scans and therefore carcinoma is unlikely.

No intervention was performed as the patient remained asymptomatic all along. The aetiology of this shunt was considered to be congenital. This extrahepatic portosystemic shunt was just part of the constellation of congenital anomalies that the patient has experienced as described in the clinical history.
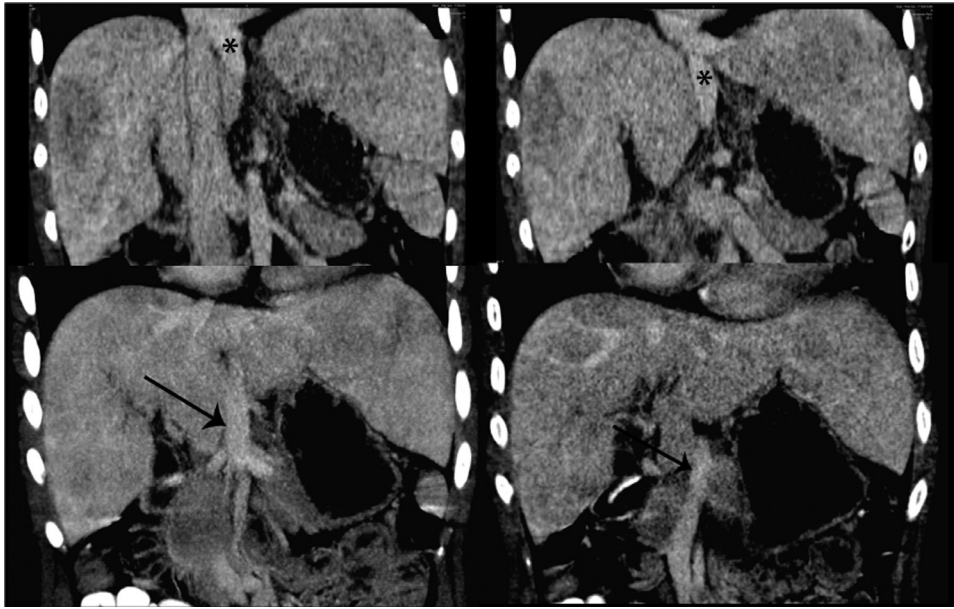
Fig. 2 – Coronal reformat images of CT liver. The common vein (asterisks) that drains directly into the intrahepatic IVC at the hepatic venous confluence level. The superior mesenteric vein before joining the splenic vein to form the common vein (arrows).

In normal individuals, there are no anatomical connections between the components of the portal system and the systemic or hepatic veins within or outside the liver. Abernethy malformation is a rare condition in which portal blood is shunted into the systemic circulation via an abnormal communication of the portal system with the systemic circulation. Abernethy malformation, which was often undiagnosed, was first described in 1793 [1]. Congenital portosystemic shunts are classified into 2 major categories: total shunting with complete absence of intrahepatic portal venous flow (extrahepatic) – type I; type 1 Abernethy malformation can be further divided into subtypes 1a and 1b. In subtype 1a, the superior mesenteric vein and the splenic vein do not connect, whereas in subtype 1b, the superior mesenteric vein and splenic vein connect to form a portal vein, which then drains into a systemic vein, as in our illustrated case; In type 2 Abernethy malformation, the portal vein is intact, but a side-to-side shunting with the inferior vena cava (intrahepatic) [2].

CEPS may be asymptomatic. However, clinical features of portosystemic shunts may be related to the shunting of portal blood, associated congenital abnormalities and hepatic lesions. They are associated with multiple congenital abnormalities, with the most common involving the cardiovascular system, and include ventricular and atrial septal defects [3]. CEPS are also associated with anomalies in the gastrointestinal, genitourinary, skeletal and vascular systems. Most abnormalities are more common in type I CEPS [4], just like in our above reported case.

Symptoms related to the shunt include hepatopulmonary syndrome [5], hepatic encephalopathy of which patients may present with neurological symptoms and symptoms due to portosystemic encephalopathy and increased blood ammonia levels [6]. They can also have metabolic dysfunction, such as hyperammonemia and galactosemia [7].

Early recognition of CEPS is important. Patients with congenital agenesis of the portal vein, due to enhanced arterial blood flow, may develop hepatocellular lesions such as benign focal nodular hyperplasia, hepatocellular adenoma and regenerative nodules [8–11].

Hepatocellular carcinoma has been reported by a number of authors as an association with the Abernethy malformation [12]. There has been an increase in the diagnosis of the condition with modern advances in medical imaging. Early noninvasive cross-sectional images via Doppler ultrasound, computed tomography or magnetic resonance imaging is key to providing an accurate diagnosis and classification, further directing the therapeutic course and clinical outcome.

Imaging plays a significant role in diagnosis, classification, and treatment of these patients. Early noninvasive cross-sectional images via Doppler ultrasound, computed tomography or magnetic resonance imaging are useful in pre- and post-treatment imaging as well as imaging of its complications [13].

There is no consensus guideline for the optimal management of patients with CEPS.

According to the type of shunt, symptomatic patients were divided into 2 groups. Liver transplants may be considered with type 1 shunts, whereas shunt closure (eg, interventional embolization or surgery) may be chosen for those with type 2 shunts [14].

Jaklitsch et al. [15] noted that half of individuals with Abernethy malformation type Ib would develop one or multiple types of tumours, ranging from benign to malignant liver tumours. It could be interpreted as a common finding for a rare disease.

As mentioned earlier, hyperarterialization of the liver could be a postulated cause of the increased risks of developing liver tumours, therefore, curative resection or orthotopic liver
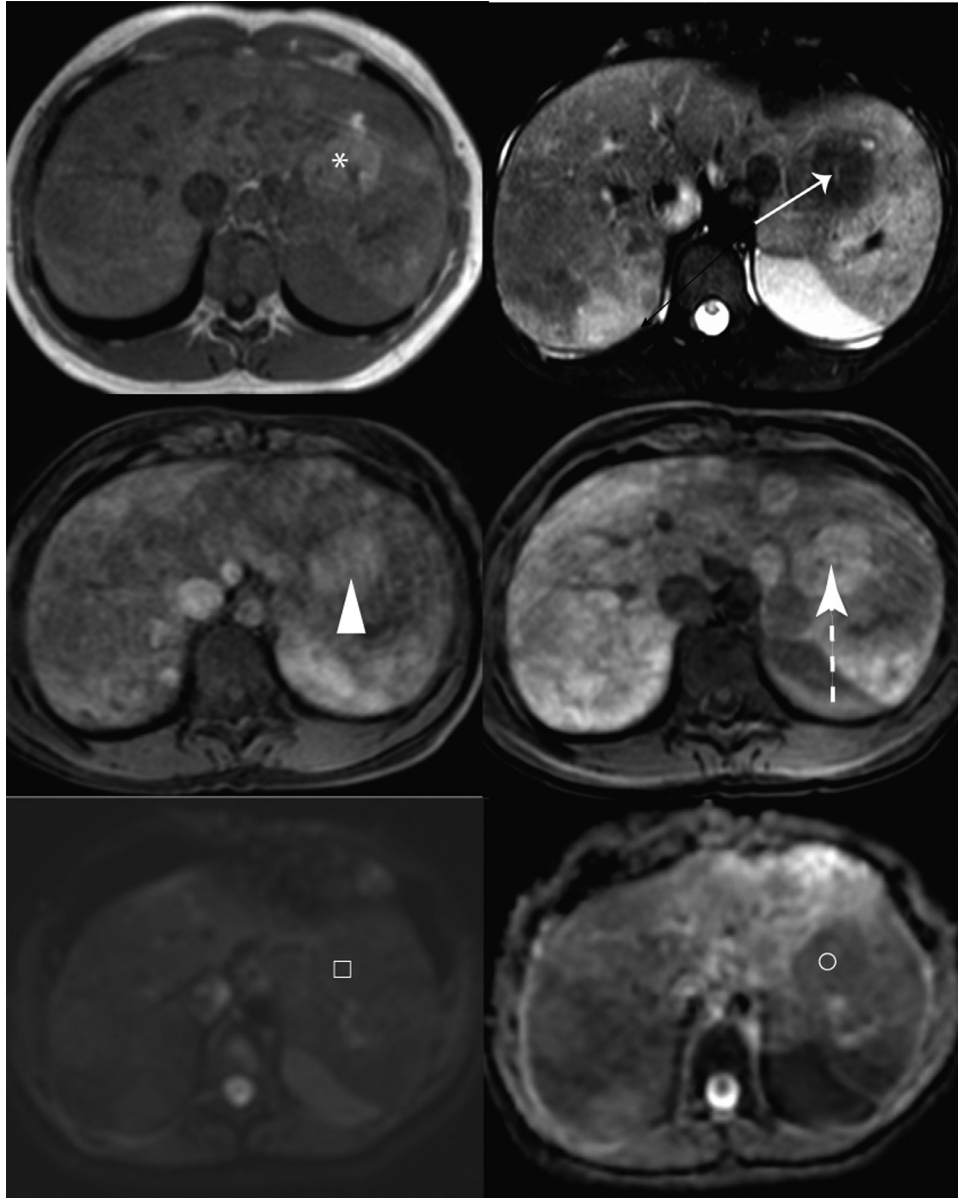
Fig. 3 – Primovist (Gadoxetate disodium)-enhanced MRI Liver. Selected MR images of the focal nodular hyperplasia appear hyperintense in T1-weighted sequence (asterisk) and hypointense but with a hyperintense central scar in T2-weighted sequence (arrow). The aforesaid lesion is hypervascular and displays hyperintense signal in the arterial phase (arrowhead). It also appears to be hyperintense in the hepatobiliary phase (dotted arrow), indicating that they are containing hepatocytes. No washout or capsule was noted. It is hypointense in diffusion-weighted imaging (box) and normal ADC value (circle).

transplantation if recommended might alter the course of disease into better long-term outcome.

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