CASE REPORT

Congenital absence of the portal vein and interruption of the inferior vena cava with end-to-end portosystemic shunt, hepatopulmonary syndrome and focal nodular hyperplasia of the liver

C.-T. Wong*, Y.-W. Lee, W.-C. Ho, K.-H. Pay, K.-M. Cheung

Caritas Medical Centre, Shamshuipo, Kowloon, Hong Kong, People’s Republic of China

Introduction

A 9-year-old female patient presented with cyanosis due to an intra-pulmonary right to left shunt. Imaging and biopsy showed congenital absence of the portal vein, interruption of the inferior vena cava and focal nodular hyperplasia in the liver. The authors believe this is the first reported case of end-to-end shunting of portal blood into the inferior vena cava and the second case of congenital absence of the portal vein causing hepatopulmonary syndrome. The imaging findings are described and the literature reviewed.

Case report

A 9-year-old female patient presented to the accident and emergency department with dry cough and decreased exercise tolerance. Obstetric and perinatal history was uneventful and she enjoyed good health in the past. No significant family history noted. Oxygen saturation (SaO2) was 90% on room air. Physical examination revealed central cyanosis. Blood gas investigation revealed central cyanosis. Blood gas investigation showed PaO2 of 62 mmHg, which did not improve significantly after 50% oxygen. Contrast echocardiogram using agitated normal saline confirmed intra-pulmonary shunting. No cardiac abnormality detected by echocardiogram. Blood ammonia level was normal and there was no sign of hepatic encephalopathy.

Chest radiograph was unremarkable apart from a prominent azygos vein. Computed tomography (CT) of the thorax and abdomen (Lightspeed QX/i Four-slice scanner, General Electric Medical Systems, Milwaukee, WI, USA) revealed no macroscopic arteriovenous malformation in the thorax but the pulmonary vessels were prominent. The intra-hepatic portal veins were absent with hypertrophy of the hepatic arteries, compatible with congenital absence of the portal vein (CAPV) (Fig. 1). The infra-hepatic portion of the inferior vena cava (IVC) was totally absent (Fig. 2). The right common iliac vein and right renal vein drained into the azygos vein. On the other side, the left common iliac vein and left renal vein drained into the hemiazygos vein. The hemiazygos system finally drained into the azygos vein in the thorax. The superior mesenteric vein (SMV) joined with the splenic vein and drained directly into the IVC behind the liver in an end-to-end manner. A 4 cm nodule was noted in segment 2 of the liver (Fig. 3). The situs of the internal organs was normal. CT-guided core biopsy of the nodule and adjacent liver parenchyma showed absence of portal venous branches and focal nodular hyperplasia (FNH). No evidence of chronic liver disease noted.

Discussion

Although variants or partial agenesis of the portal system are not uncommon,1 congenital total absence of the intra-hepatic portal veins is a rare malformation, and was first described by John Abernethy in 1793.2 Howard and Davenport3 used the term "Abernethy malformation" to describe patients with congenital diversion of portal blood away from the liver, either by end-to-side or side-to-side extra-hepatic shunt. Morgan and Superina4 have classified portosystemic shunts into two types with emphasis on functional and clinical differences. In type 1 shunts, the liver is not perfused with portal blood and is equivalent to CAPV. Type 1 shunts are further classified into type 1a where the SMV and splenic vein do not join to form confluence and type 1b where the two veins join before entering the systemic circulation. Type 2 shunts are partial shunts due to portal-hepatic anastomoses. The actual prevalence of CAPV is not known as some patients are asymptomatic and only detected incidentally. To date 37 cases of CAPV
are reported in the English literature. The age at diagnosis varies between 1 day and 50 years, most cases being diagnosed in childhood. Most reported cases were female. Various routes of drainage of the portal blood are described in the literature, including entrance into the supra- or infra-hepatic IVC, left-side IVC, left hepatic vein, left renal vein, left iliac vein or through collaterals to the azygos system. Liver anomalies are very common and include liver dysfunction, FNH, 5–7 nodular regenerative hyperplasia, hepatic adenoma, hepatoblastoma, hepatocellular carcinoma and biliary atresia. Other frequently associated congenital abnormalities include cardiovascular and situs anomalies, polysplenia, Goldenhar syndrome and IVC abnormalities.

Figure 1  Transverse CT images of the upper abdomen after intravenous contrast medium administration. (a) The intra-hepatic branches of the portal vein are totally absent. Atretic remnants of the proximal portion of the main right and left portal branches are noted (arrow). (b) The intra-hepatic branches of the hepatic arteries have hypertrophied.

Figure 2  Transverse CT images after intravenous contrast medium administration. (a) At the level of the confluence of the splenic vein and the superior mesentery vein. The IVC is absent. The retrocral azygos vein (arrow) and hemiazygos vein (arrowhead) are grossly distended because they drain the blood from the lower part of the body as a result of inferior vena cava
The relationship between CAPV and nodular hyperplasia has been reviewed by Motoori et al.\textsuperscript{8} It is still uncertain whether circulatory disturbance alone in the liver could cause nodular hyperplasia or not. Nevertheless the association between disturbance of portal blood flow and nodular hyperplasia of the liver appears to be very strong.\textsuperscript{9–11} Hepatic encephalopathy is a common presentation in patients with portal-systemic shunt. Watanabe\textsuperscript{12} reviewed the causes of portal-systemic encephalopathy in non-cirrhotic patients and included CAPV as type V cause. Our patient presented with hepatopulmonary syndrome instead of encephalopathy, which is the second case reported in patients with CAPV.\textsuperscript{13} Hepatopulmonary syndrome is the clinical relationship between hepatic disease and the existence of pulmonary vascular dilatations, which can result in a range of arterial oxygenation abnormalities. The question of why some patients had encephalopathy, a few had hepatopulmonary syndrome and others were asymptomatic is difficult to answer. To date no patient with CAPV and both hepatic encephalopathy and hepatopulmonary syndrome has been reported. As the number of patients with a congenital portosystemic shunt presenting as hepatic encephalopathy is larger, its pathogenesis and prognosis are better studied in the literature.\textsuperscript{12,14,15} Very little is known about the pathogenesis of hepatopulmonary syndrome in patients with CAPV. Intra-pulmonary vascular dilatations due to alteration in the synthesis or metabolism of vasoactive pulmonary substances at the hepatic level have been suggested as the cause of hepatopulmonary syndrome in patients with end-stage liver parenchymal disease complicated by portosystemic shunt. Whether this mechanism can be applied to patients with CAPV is uncertain.

A review by Mayo et al.\textsuperscript{16} indicates anomalies of the IVC are not rare. Anderson et al.\textsuperscript{17} in 1961 suggested the use of “infra-hepatic interruption of the IVC with azygos (hemiazygos) continuation” to describe the condition of failure of fusion of the hepatic and prerenal segments of the IVC combined with persistence of either the right lumbar azygos vein or the left lumbar hemiazygos vein.\textsuperscript{17} This

\textbf{Figure 3} Transverse CT images in the upper abdomen, before and after contrast medium administration. A 4 cm nodule was noted in segment 2 of the liver. (a) It is isodense in the non-contrast study, (b) hyperdense in the arterial phase and (c) hypodense in the portal venous phase. Small central scar, which is hypodense in the arterial phase and hyperdense in the portal venous phase, was noted within the nodule.

\textsuperscript{17} This interruption. (b) At mid-heart level the azygos vein (arrow) is as big as the descending aorta, the basis of the “double vessels” sign for the prenatal sonographic diagnosis of interruption of inferior vena cava. Note the prominent descending branches of the pulmonary arteries on both sides, probably related to the intra-pulmonary shunting. (c) At the level of the azygos arch, which is grossly dilated. The azygos vein drains into the superior vena cava as usual (arrow).
group of patients was first described by Stark in 1835 and later by Dwight in 1900. It is often associated with cardiosplenic syndromes. The present patient represented a rare form of interruption of IVC, in which there are bilateral inferior venae cavae draining into theazygous and hemiazygous systems respectively, similar to a case described by Mayo et al. The present patient represents a rare form of interruption of IVC, in which there are bilateral inferior venae cavae draining into the azygous and hemiazygous systems respectively, similar to a case described by Mayo et al.

Although embryologically the systemic veins in the abdomen are described as intra-embryonic while the portal venous system is extra-embryonic, they are intimately related both in spatial relationship and in temporal development, especially at the region of the developing liver. Anastomoses are present between the two systems during early development of the embryo.

The development of the portal vein occurs between 4–10 weeks gestation, by selective regression of peri-intestinal vitelline venous loops. Abnormal involution of the vitelline veins can lead to malformations such as preduodenal portal vein. Conversely CAPV can be the result of excessive regression of the venous plexus in the vicinity of the developing liver.

The development of the IVC is even more complex and occurs between 6–8 weeks gestation. It also involves progressive development and regression of vessels, namely the paired posterior cardinal, subcardinal and supracardinal systems. Mayo et al. suggested that bilateral IVC with azygos and hemiazygous continuation represents persistence of the entire supracardinal system, without evidence of right-sided dominance.

The association of portal vein anomalies with IVC abnormalities is not rare. The presence of preduodenal portal vein and abnormality of the IVC is well documented. The patient reported by Howard and Davenport had both absence of the portal vein and interruption of the IVC with azygous continuation. However, her preduodenal portal vein drained into theazygous vein and she had other congenital abnormalities including situs inversus, polysplenia and biliary atresia.

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In view of this particular combination, the present patient represents the first reported case of end-to-end portosystemic shunt in Abernethy malformation. It is of interest that both conditions are commonly associated with other congenital cardiovascular, splenic and situs anomalies, but none occurred in the present patient.

References

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