Congenital absence of the portal vein (CAPV) is a rare malformation in which intestinal and splenic venous flow bypasses the liver and drains directly into the systemic circulation via a congenital portosystemic shunt. We describe two cases of CAPV presenting as pulmonary hypertension that were initially suspected as primary pulmonary hypertension. However, subsequent ultrasonography and CT detected the absence of a portal vein and the presence of a portosystemic shunt. Pulmonary hypertension is a recognized complication of liver disease and portal hypertension. However, these two cases illustrate that CAPV may result in pulmonary hypertension without liver disease or portal hypertension.

**Index words:** Portal vein  
Portosystemic shunt  
Hypertension, pulmonary  
Angiography, CT  
Liver

Congenital absence of the portal vein (CAPV) is a rare malformation in which intestinal and splenic venous flow bypasses the liver and drains directly into the systemic circulation via a congenital portosystemic shunt. Variable associated anomalies and clinical manifestations caused by the disrupted enterohepatic circulation in CAPV have been reported, including congenital cardiac anomalies, skeletal and visceral anomalies, hepatic neoplasms, liver dysfunction, hemorrhoidal bleeding and portosystemic encephalopathy (1, 2). We describe two cases of CAPV presenting as pulmonary hypertension, in which primary pulmonary hypertension was initially suspected. However, subsequent ultrasonography and CT detected the absence of the portal vein and the presence of a portosystemic shunt. To the best of our knowledge, only three cases of pulmonary hypertension associated with CAPV have been reported since 1974 (3). Pulmonary hypertension is a recognized complication of liver disease and portal hypertension. However, these two cases illustrate that CAPV may result in pulmonary hypertension without liver disease or portal hypertension and that CAPV can be diagnosed with noninvasive imaging techniques such as Doppler ultrasonography or CT angiography.

**Case Report**

**Case 1**

A 3-year-old girl presented with progressive dyspnea of several weeks duration. The medical history and physical examination were nonspecific. No hypoxemia was evident by arterial blood gas analysis ($O_2$ saturation:
97.7%, PaO₂: 79.9 mmHg, PaCO₂: 39 mmHg, pH: 7.49). The patient had anemia and thrombocytopenia (hemoglobin level: 10.8 g/dL, hematocrit: 34.9%, platelet count: 84 × 10⁹/L), but biochemical parameters and the clotting profile were within normal ranges, except for a slightly elevated total serum bilirubin level (1.8 mg/dL). Serological markers for hepatitis B and C viruses were negative. Chest radiography showed cardiomegaly and centroperipheral pulmonary vascular discrepancy suggestive of pulmonary hypertension (Fig. 1A). Echocardiography revealed pulmonary hypertension with right ventricular enlargement and mild tricuspid regurgitation. Cardiac catheterization revealed a pulmonary capillary wedge pressure of 9 mmHg, and pulmonary arterial pressure of 71/37 (mean 54) mmHg. Pulmonary arteriography confirmed the absence of a pulmonary embolism, and no evidence of an intracardiac or intrapulmonary shunt was seen by air-bubble contrast echocardiography or by cardiac catheterization. Primary pulmonary hypertension was initially suspected.

Fig. 1. A 3-year-old girl presented with dyspnea.
A. Chest radiography showing cardiomegaly and centroperipheral pulmonary vascular discrepancy suggestive of pulmonary hypertension.
B. Arterial [a] and portal [b] phase axial images showing the absence of intrahepatic portal veins and a dilated hepatic artery [white arrow], and early inferior vena caval opacification [arrowhead] via a portosystemic shunt. Note the normal hepatic veins [yellow arrowheads].
C. A CT angiography volume rendered image showing the absence of a main portal vein. The superior mesenteric vein (SMV) and the splenic vein (SV) joined and drained through the inferior mesenteric vein (IMV) into the left internal iliac vein (yellow arrowheads) and then into the inferior vena cava (asterisk).
However, abdominal ultrasonography and CT angiography showed that the porta hepatis contained the common bile duct and the common hepatic artery with no main portal vein. Intrahepatic portal veins were not seen, either. The common hepatic artery was found to be dilated (Fig. 1B). Furthermore, ultrasonography revealed a markedly enlarged mesenteric vein draining into the left internal iliac vein. There were no hepatic parenchymal lesions. Splenomegaly was also noted. CT angiography confirmed the absence of both intrahepatic and extrahepatic portal veins, and the superior mesenteric vein and the splenic vein were found to be joined and to drain through the inferior mesenteric vein into the left internal iliac vein (Fig. 1C). These findings were compatible with CAPV, a type I-b portosystemic anomaly as classified by Morgan et al. [4].

After a 19 month-follow-up under conservative management, an approximately 3.5 cm sized hyperechoic hepatic mass was detected by ultrasonography. The mass appeared hyperintense on T2-weighted MR images and mildly hyperintense on T1-weighted images. Strong enhancement of the lesion was observed during the early arterial phase, but during the late portal and venous phases, the periphery of the lesion was slightly hypointense versus the liver parenchyma. This was considered to be due to a central scar of a focal nodular hyperplasia.

Fig. 1. D. 19 months after a diagnosis of CAPV a 3.5 cm sized mass was detected in the right lobe of the liver. The mass was hyperintense on T2-weighted MR images and mildly hyperintense on T1-weighted images. Strong lesion enhancement was observed during the early arterial phase and the lesion periphery was slightly hypointense versus the liver parenchyma during the venous phase. The lesion central portion which showed persistent enhancement (yellow arrow), was considered to be a central scar of a focal nodular hyperplasia.

Strong enhancement of the lesion was observed during the early arterial phase, but during the late portal and venous phases, the periphery of the lesion was slightly hypointense versus the liver parenchyma, whereas its center showed persistent enhancement, which was considered to be due to a central scar (Fig. 1D). There was no evidence of chronic liver disease, and the level of alpha-fetoprotein was not elevated (< 3ng/mL). Although not histologically confirmed, the hepatic mass was considered a focal nodular hyperplasia, which is a relatively common complication of CAPV [2, 3].

The patient remains under conservative management. Liver transplantation in this case is contraindicated due to the severe pulmonary hypertension [5, 6].

Case 2

A 20-year-old female presented with syncope, and complained of progressive dyspnea of several weeks duration. A physical examination showed no abnormalities, and arterial blood gas analysis indicated no hypoxemia ($O_2$ saturation: 98.9%, $PaO_2$: 87.3 mmHg, $PaCO_2$: 37.6 mmHg).
The patient had severe thrombocytopenia (platelet count: $5 \times 10^9$/L), the serum bilirubin level was elevated (total bilirubin: 5.6 mg/dL, direct bilirubin: 1.5 mg/dL), and the serum albumin level was slightly low (2.9 g/dL). Other biochemical parameters and the clotting profile were within normal ranges. Chest radiography showed cardiomegaly and prominent right and left pulmonary trunks with an abnormal centroperipheral pulmonary vascular discrepancy suggestive of pulmonary hypertension (Fig. 2A). Echocardiography revealed right ventricular enlargement and mildly reduced right ventricle contractility, accompanied by mild tricuspid regurgitation. The left ventricle was compressed by an enlarged right ventricle. Pulmonary arterial systolic pressure calculated from tricuspid regurgitation velocity was 110 mmHg, and there was no evidence of an intracardiac or intrapulmonary shunt by air-bubble contrast echocardiography. There was no evidence of a pulmonary embolism on a lung ventilation/perfusion scan. Primary pulmonary hypertension was initially suspected.

However, abdominal ultrasonography showed the presence of no portal vein in the porta hepatis, and no intrahepatic portal vein. CT angiography confirmed these findings, and further revealed that the superior mesenteric and splenic veins joined and drained into the inferior vena cava via the left renal vein (Fig. 2B). Marked splenomegaly was noted. The liver was not cirrhotic as seen by ultrasonography and CT, and a brain MRI revealed symmetric T1 hyperintensity in the bilateral globus pallidi, suggestive of hepatic encephalopathy.

**Fig. 2.** A 20-year-old female presented with syncope.  
**A.** Chest radiography showing cardiomegaly and prominent right and left pulmonary trunks with an abnormal centroperipheral pulmonary vascular discrepancy suggestive of pulmonary hypertension.  
**B.** A CT image showing the porta hepatis containing a dilated hepatic artery without a main portal vein (yellow arrowhead). No cavernous transformation was evident. The superior mesenteric (SMV) and splenic veins (SV) joined and drained into the left renal vein (LtRV) via a splenorenal shunt (yellow arrows), and then into the inferior vena cava (asterisk).  
**C.** A T1 weighted axial image showing symmetric hyperintensity in the bilateral globus pallidi, suggestive of hepatic encephalopathy.
eral globus pallidi, suggestive of hepatic encephalopathy (Fig. 2C).

During conservative management of the severe pulmonary hypertension and thrombocytopenia, the patient suddenly expired due to cardiac arrest on the fourth day of admission.

**Discussion**

During fetal development, abnormal involution of peri-intestinal vitelline venous loops can result in CAPV and extrahepatic portosystemic shunt formation (2, 4). According to the case reports of liver biopsies in CAPV patients, histopathology displays portal tracts with normal arteries and bile ducts but with severe loss of portal vein branches (1, 7). Since the intrahepatic branching of the developing portal vein is a prerequisite for the formation of intrahepatic bile ducts, it has been proposed that CAPV is not aplasia or agenesis of the portal vein, but it is caused by a secondary phenomenon such as the thrombotic occlusion of the extrahepatic portal vein during the prenatal period (1).

CAPV is classified into two types according to the absence (type a) or presence (type b) of the confluent superior mesenteric and splenic veins (4). Previously reported drainage sites of extrahepatic portosystemic shunts in CAPV vary, and have included the inferior vena cava (IVC), renal vein, iliac vein, inferior mesenteric vein, left hepatic vein, right atrium, and azygos vein (1-3).

Various anomalies including cardiac, skeletal, and visceral anomalies have been reported in association with CAPV (1-3). Clinical presentations of CAPV are various, and CAPV can be entirely asymptomatic. Features suggestive of portosystemic shunt like hyperammonemia, hypoalbuminemia, hypergalactosemia, mild liver function test abnormalities, and hepatic encephalopathy may exist, and patients can present with persistent jaundice and growth failure, mental retardation, and hemorrhoidal bleeding (2, 3, 8).

CAPV is often associated with a benign or malignant hepatic neoplasm. Focal nodular hyperplasia is a relatively common complication of CAPV, and an adenoma, hepatoblastoma, or hepatocellular carcinoma may be present (2, 3, 8). These lesions might be caused by an abnormal response of the liver cells to the lack of portal blood flow, increased arterial hepatic flow, and to the abnormal delivery of hepatotrophic factors (2, 3, 8).

In patients with liver disease and portal hypertension, the pulmonary vasculature is exposed to gut-derived substances that bypass hepatic detoxification and enter the lungs through the portosystemic shunt (5). This is believed to provide the mechanism of two distinct pulmonary vasculopathies associated with liver disease and portal hypertension vasoconstriction and pulmonary arteriolar wall damage in portopulmonary hypertension and vasodilatation and arteriovenous shunting in hepatopulmonary syndrome (HPS) (5). In CAPV, enteric venous flow completely bypasses the liver and enters the pulmonary circulation without liver disease or portal hypertension (5, 7). Three cases of pulmonary hypertension and five cases of HPS in association with CAPV have been reported since 1997 (3, 7).

In a recent study on liver transplantation in CAPV patients, it was suggested that prophylactic liver transplantation be conducted in CAPV patients before fatal pulmonary hypertension or HPS develops, as this might complicate or preclude liver transplantation (6). Thus, it is important to first detect the absence of the portal vein and the presence of a portosystemic shunt by imaging studies, regardless of the clinical presentation.

We have described two cases of CAPV that presented as pulmonary hypertension without severe liver dysfunction or portal hypertension. Both cases were initially diagnosed as having primary pulmonary hypertension, but abdominal ultrasonography and CT angiography lead to the identification of pulmonary hypertension, and a diagnosis of CAPV. Anatomical loss of the portal vein accompanied by abnormal venous drainage of the supramesenteric and splenic veins is required for a definite diagnosis of CAPV (9). CT angiography and MR angiography have been proven as reliable and non-invasive diagnostic techniques for imaging the portal system, as in our cases (2, 8, 9). These two cases may be differentiated from acquired portal vein thrombosis by the absence of cavernous transformation of the periporal collateral veins or the absence of other secondary signs of portal hypertension, such as varices and ascites (3).

We conclude that it is important to investigate the possibility of a portosystemic shunt in cases considered as primary pulmonary hypertension, even in the absence of symptoms and signs suggesting liver disease or portal hypertension. Although clinical presentations of CAPV are various, the imaging characteristics of CAPV, i.e., the absence of a portal vein and the presence of a portosystemic shunt can be detected precisely by ultrasonography, CT angiography, or MR angiography regardless of clinical presentation. The resulting early non-
invasive diagnosis allows appropriate treatment to be initiated before the development of fatal pulmonary hypertension or HPS, which might complicate or preclude liver transplantation.

References