

# Congenital Absent Portal Vein with Extrahepatic Portosystemic Shunt: A Futile Growth

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## Introduction

Abernethy malformation is a rare congenital malformation defined by an extrahepatic porto-systemic shunt directing splanchnic blood directly into the inferior vena cava, as first described in 1793. Clinical presentation, especially in children, is extremely variable. In some cases, porto-systemic malformations may remain asymptomatic, making the diagnosis difficult. In other cases, they may cause metabolic disorders and damage various organs and systems, such as the liver, central nervous system and respiratory tract.

## Case Report

A 4-year-old girl presented in emergency department with confusion and altered level of consciousness. She had history of increasing jaundice, loss of appetite, and weight loss for last 1 year. She denied abdominal pain, fever, vomiting, diarrhea and night sweats. Her physical examination revealed pallor and hepatomegaly without tenderness. She was mildly anemic with a hemoglobin measurement of 10.0 g/dL. Liver function tests were abnormal. The total protein was 6.9 g/dL with a slightly low albumin of 3.0 g/dL and reversed albumin/globulin ratio. Serological markers for ceruloplasmin and other viral markers were negative. In clinical setting of hepatic encephalopathy; symmetrical hyper intense globus pallidi on T1WI (**Figure 1**) is suggestive of hepato-cerebral syndrome. Triphasic contrast enhanced computed tomography (CECT) of the abdomen show absent intrahepatic branches of portal vein. An abnormal side-to-side communication is seen between extrahepatic portal vein (8.6 mm diameter) and inferior vena cava (IVC) (**Figure 2**). Hepatic arterial tree and hepatic veins are normal in development. Diagnosis of congenital extrahepatic portocaval shunt (Abernathy malformation type Ib) was made. Other associated findings were moderate splenomegaly, left sided infrarenal IVC (**Figure 3**) and block L3-L4 vertebra. Patient refused liver biopsy. Patient referred to higher centre for further management and liver transplant.

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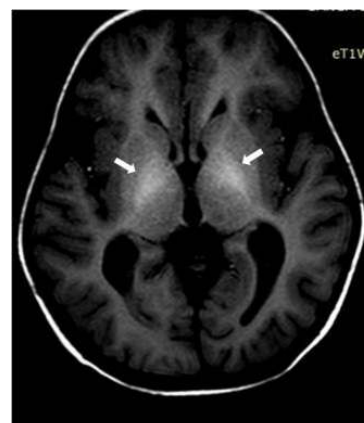
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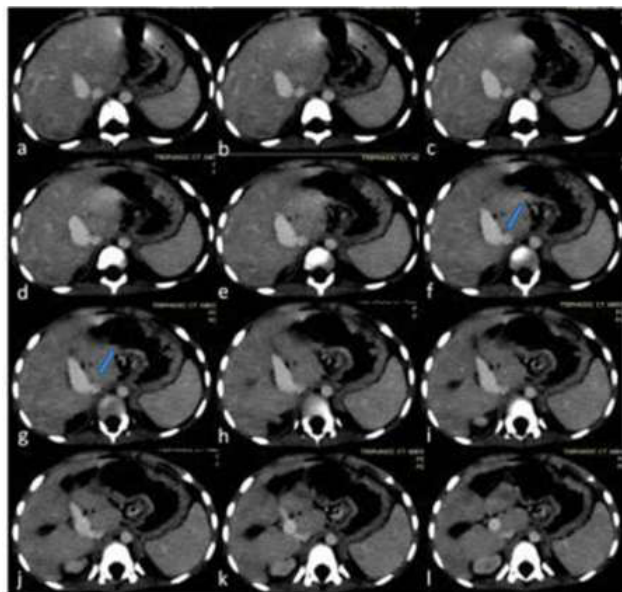
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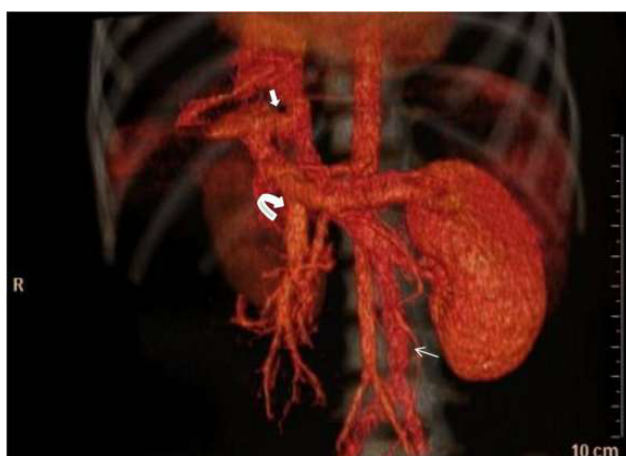
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**Figure 1** Axial T1WI image at the level of basal ganglia reveal symmetrical hyperintense globus pallidi (white arrow).



**Figure 2** Sequential axial sections (a-l) through the upper abdomen (cranio-caudal) show absent intrahepatic portal branches and side-to-side communication (blue arrow) between extrahepatic portal vein and inferior vena cava.



**Figure 3** Volumetric reconstruction image show side-to-side communication (white solid arrow) between extrahepatic portal vein and inferior vena cava, confluence of splenic vein, superior mesenteric vein and main portal vein (curved white arrow) and left side IVC (thin white arrow).

## Discussion

As first construed by John Abernethy in 1793 [1]; congenital absence of portal vein (CAPV) is a rare congenital malformation of spleno-portal axis wherein splanchnic blood is completely or partially diverted directly into the IVC. Majority of affected patients were <18 years age females [2]. Classification of congenital extrahepatic portocaval shunt (Table 1) proposed by Morgan and Superina [3].

Embryologically, between gestational weeks four and five, the paired vitelline veins form three anastomosis that, over the

course of the first trimester, undergo selective involution to produce the portal vein [4]. Similarly, at 6–8 weeks gestation, progressive development and regression of paired posterior cardinal, subcardinal and supracardinal systems gives rise to IVC. Aberrations in this process of involution may result in various combination of portal vein and IVC abnormalities, like preduodenal portal vein and anomalous continuation of inferior vena cava, absence of the portal vein and interruption of the IVC with azygos continuation, left sided IVC [3] and infra-hepatic interruption of the IVC with portal continuation. Other frequently associated congenital abnormalities include cardiovascular and situs anomalies, polysplenia, genitourinary tract and skeletal system have been documented [5, 6].

Liver abnormalities are very common, reported in 48% of published cases and include liver dysfunction, nodular regenerative hyperplasia, hepatic adenoma, hepatoblastoma, hepatocellular carcinoma, choledochal cyst, caroli's disease and biliary atresia, are considered secondary to the absence of portal hepatotrophic factors due to systemic shunting of the visceral venous return and increased arterial hepatic flow [7, 8]. Our patient has no detectable structural hepatic abnormality.

Hepatic encephalopathy; as highlighted in our case is a common presentation. Although hyperammonemia can present without encephalopathy, increasing age and shunt ratio of more than 60% favors the onset of hepatic encephalopathy [6, 9]. Also significantly low level of ammonia in venous blood has been found in cases of CAPV, thought to be due to compensatory alteration in intestinal bacterial flora [9]. Uncommon clinical presentations pulmonary hypertension, hepatopulmonary syndrome and growth retardation [6, 9].

Treatment is only indicated for patients with clinically significant shunting. Definitive treatment includes liver transplantation for type I malformation and surgical or transcatheter shunt occlusion in type II malformation [10].

## Conclusion

Congenital porto-systemic shunt can have variable presentation from no symptoms of portal hypertension to systemic manifestation like encephalopathy or hepatopulmonary syndrome or symptoms related to liver masses. Early diagnosis is must for appropriate and timely management of shunt and related complication.

**Table 1** Classification of congenital extrahepatic portocaval shunt.

<b>Type 1a</b>	End-to-side portocaval anastomosis with independent drainage of the superior mesenteric vein and splenic vein
<b>Type 1b</b>	End-to-side portocaval anastomoses with convergence of the superior mesenteric vein and splenic vein
<b>Type 2</b>	Side-to-side portocaval anastomoses

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