Congenital extrahepatic portosystemic shunt

Introduction

This article presents the case of a 42-year-old man who presented with abdominal pain and had an incidental finding of a congenital extrahepatic portosystemic shunt. On computed tomography imaging this was demonstrated to be a type II (splenorenal) shunt. His ammonia level was elevated, but he had no features of encephalopathy. Ultrasound imaging of his liver with Doppler assessment of the portal vein was within normal limits. A thrombophilia screen was found to be negative. He was managed symptomatically and discharged home with regular outpatient follow up. This article reviews the literature regarding this rare phenomenon, its classification and symptom profile, and discusses its sequelae.

Discussion

Portosystemic shunts most commonly occur in the context of liver disease or following venous occlusion in thrombophilic states. Rarely, congenital extrahepatic portosystemic shunts are diagnosed and can be associated with other congenital anomalies as described by Murray et al (2003).

Morgan and Superina (1994) classified these shunts into type I (Abernethy malformations) with complete absence of portal vein, or type II with preserved portal venous flow and with varying clinical and epidemiological consequences (*Table 1*).

Asymptomatic congenital extrahepatic portosystemic shunts are diagnosed as incidental findings on abdominal imaging such as computed tomography. Occasionally, as demonstrated by Venkat-Raman et al (2001), clinically asymptomatic shunts have been diagnosed in children by Doppler ultrasonography during investigation for galactosaemia.

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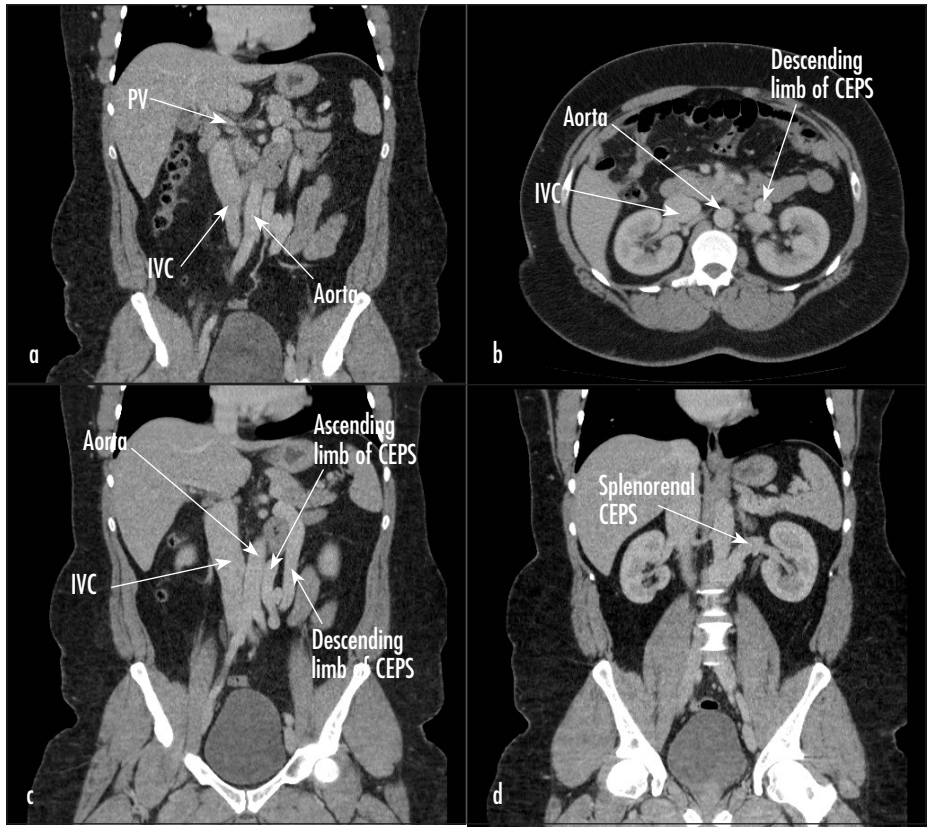
Figure 1. Computed tomography imaging of congenital extrahepatic portosystemic shunt (spleno-renal; CEPS). a. Coronal computed tomography image demonstrating small patent portal vein. b. Axial portal venous phase computed tomography showing descending limb of CEPS joining left renal vein. c. Coronal views of CEPS (ascending and descending limbs). d. CEPS joining left renal vein. IVC = inferior vena cava; PV= portal vein.



Case Report

A 42-year-old man presented with a 1-day history of sudden onset left iliac fossa pain. On deep palpation, he had tenderness in the left iliac fossa, but no features of peritonism. Blood investigations showed a normal full blood count and C-reactive protein level. A computed tomography scan of his abdomen (*Figure 1*) showed subtle sigmoid diverticulosis. An incidental note was made of a tortuous and markedly dilated venous channel to the left side of his spine. This communicated between the portal venous system and the systemic circulation via the left renal vein. In addition, the main portal vein was noted to be rather small.

On revisiting the history there was no history of alcohol excess or family history of thromboembolic conditions. He had no stigmata of chronic liver disease.

An ultrasound with Doppler assessment of the portal vein demonstrated an architecturally normal liver with anterograde portal venous flow. In addition, a full thrombophilia screen was negative.

A diagnosis of congenital extrahepatic portosystemic shunt was made. His serum ammonia level was mildly elevated at 66 μmol/litre (reference range 20–60 μmol/litre), in keeping with this condition. The patient receives yearly follow up in gastroenterology clinic and remains well and without any complications.

Table 1. Congenital extrahepatic portosystemic shunts (Morgan and Superina classification)

Shunt type	Shunt anatomy	Clinical features	Associated congenital anomalies
I	End-to-end communication from the portal system to the inferior vena cava (most common), azygous vein, renal vein, right atrium or iliac veins	Presents more commonly in younger patients	Biliary atresia
		Females > males	Polysplenia
		Usually symptomatic	Situs inversus
			Multicystic dysplastic kidneys
Ia	The splenic vein and superior mesenteric vein each drain separately into the systemic circulation		Goldenhar syndrome (oculoauriculovertebral dysplasia)
			Choledochal cyst
Ib	The splenic and superior mesenteric veins join to form a common trunk and directly drain into the systemic circulation, bypassing the liver		Skeletal anomalies
			Cutaneous haemangiomas
II	Side-to-side anastomosis of the portal vein with the inferior vena cava, or gastrorenal, splenorenal or portorenal shunts	Presents in older age groups	Pulmonary valve atresia
		No gender predominance	Patent ductus arteriosus
		Usually asymptomatic	Goldenhar syndrome
			Polysplenia
			Inferior vena cava anomalies

From Morgan and Superina (1994)

Encephalopathy is the commonest clinical presentation in symptomatic patients. This is more often seen in older patients, possibly because the ageing brain is more susceptible to the effects of harmful metabolic products such as ammonia.

Additionally, Matthews et al (2014) reported symptoms including pain, jaundice and regenerative hepatic nodules, owing to altered hepatic blood flow.

While imaging (ultrasound, computed tomography or magnetic resonance imaging) is the preferred investigation to confirm the presence of a portosystemic shunt, ultrasound Doppler may fail to detect smaller shunts. Nuclear medicine studies such as iodine-123 iodoamphetamine can be used to assess shunt dynamics, as shown by Alonso-Gamarra et al (2011). Furthermore, serum ammonia level is a useful, although non-specific, investigative adjunct.

Definitive treatment of congenital extrahepatic portosystemic shunts is determined by the type of shunt; liver transplantation is the only effective treatment for symptomatic type I congenital extrahepatic portosystemic shunts. However, as Lautz et al (2011) have shown, the various options available to manage type II shunts are dependent on shunt size and symptoms.

Smaller shunts may be observed, and lifestyle changes, including a low protein diet, may be advocated. Surgical options such as occlusion, ligation or embolization have been investigated by Lautz et al (2011) for shunts not responding to conservative management.

Asymptomatic shunts often require no treatment, but their presence still can have clinical implications, such as providing a route for pathogens and toxins to bypass hepatic immune surveillance as well as providing a potential future route for pulmonary metastasis of gastrointestinal cancer as shown by Matthews et al (2014). **BJHM**

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Lautz TB, Tantemsapya N, Rowell E, Superina RA (2011) Management and classification of type II congenital portosystemic shunts. *J Pediatr Surg* **46**(2): 308–14 (doi: 10.1016/j.jpedsurg.2010.11.009)

Matthews TJ, Trochsler MI, Bridgewater FH, Maddern GJ (2014) Systematic review of congenital and acquired portal-systemic shunts in otherwise normal livers. *Br J Surg* **101**(12): 1509–17 (doi: 10.1002/bjs.9619)

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Murray CP, Yoo SJ, Babyn PS (2003) Congenital extrahepatic portosystemic shunts. *Pediatr Radiol* **33**(9): 614–20 (doi: 10.1007/s00247-003-1002-x)

Venkat-Raman N, Murphy KW, Ghaus K et al (2001) Congenital absence of Portal vein in the fetus: a case report. *Ultrasound Obstet Gynecol* **17**(1): 71–5 (doi: 10.1046/j.1469-0705.2001.00312.x)

LEARNING POINTS

- Congenital portosystemic shunts can be asymptomatic or can present with encephalopathy, atypical abdominal pain or jaundice.
- Other potential complications of this phenomenon include psychiatric disturbance as a result of chronic hyperammonaemia.
- Management is dependent on type of shunt and symptoms, but definitive treatment can only be achieved with liver transplantation.
- Awareness of these sequelae may allow the practicing clinician to instigate appropriate investigation and management accordingly.