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Case Report

Asymptomatic presentation of a congenital malformation of the portal vein with portosystemic shunt

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ABSTRACT

Malformations of the portal venous system consist of congenital and acquired anomalies. Congenital portosystemic shunts represent rare vascular developmental anomalies that allow partial or complete diversion of blood flow from the portal venous system to the systemic venous system, bypassing the liver. Congenital portosystemic shunts may be associated with malformations or congenital absence of the portal vein, and it was first described by John Abernethy in 1793. Most cases are diagnosed in early childhood, but some congenital shunts may remain asymptomatic and are encountered incidentally because of the widespread use of computed tomography and magnetic resonance imaging. In this report, we discuss the case of a 40-year-old female who presented to the Emergency Department with right upper quadrant pain, nausea, and vomiting. Clinical presentation and abdominal computed tomography angiography were consistent with the diagnosis of calculous cholecystitis and congenital absence of portal vein with intrahepatic portosystemic shunts. We discuss the importance of radiology in diagnosing such incidental malformations, coupled with a review of the current literature on this topic.

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Introduction

Congenital portosystemic shunts (CPSS) are rare vascular developmental anomalies that result in a complete or partial diversion of the portal flow away from the liver into the systemic venous circulation [1]. They are commonly classified into extra-hepatic portosystemic shunts (EHPS) and intra-hepatic portosystemic shunts (IHPS) [2]. The overall incidence of CPSS is 1:30,000 at birth and 1:50,000 for those who survive into adulthood [3]. An IHPS may either seal spontaneously within the first 2 years of life or may persist [4]. Persistent shunts are usually asymptomatic and go undetected for years, as presented in this case report. The prevalence of intrahepatic shunts is estimated to be 0.0235%, as reported from a random ultrasonography screening of asymptomatic adults [5].

Embryology

There are 3 groups of embryonic veins that constitute the primordial venous system which gives rise to the hepatic venous architecture. These primary embryological veins are the cardinal, vitelline, and umbilical veins [1,2,6]. The 2 cardinal veins and the 2 vitelline veins develop into systemic and portal venous systems, respectively, whereas the umbilical veins drain the yolk sac and placenta before regressing [4].

The development of the hepatic venous system occurs between the fourth and 10th week of gestation [1,6]. Initially, the 2 vitelline veins emerge from the anterior surface of the yolk sac and drain into the sinus venosus [4]. By the end of the fourth week of gestation, cross communicating veins develop between the vitelline veins, and they anastomose with each other to form a figure of eight pattern around the developing duodenum (termed the vitelline venous network) [2]. At the eighth week, the basic structure of the hepatic sinusoids has already developed. Through selective involution, the final configuration of intrahepatic branches of portal and hepatic venous systems is yielded [4]. Between the 10th and 12th week, the left vitelline vein disintegrates, while the cranial part of the right vitelline vein and the portion that lies inferior to the liver develop into the terminal branches of the inferior vena cava, and portal and superior mesenteric veins, respectively [4]. Clamping the umbilical cord at birth leads to collapse of the umbilical vein, decreasing blood flow through the ductus venosus. Both structures close and give rise to the ligamentum teres and ligamentum venosum, respectively [6].

Incomplete involution of the vitelline venous system following development of the hepatic sinusoids is thought to be

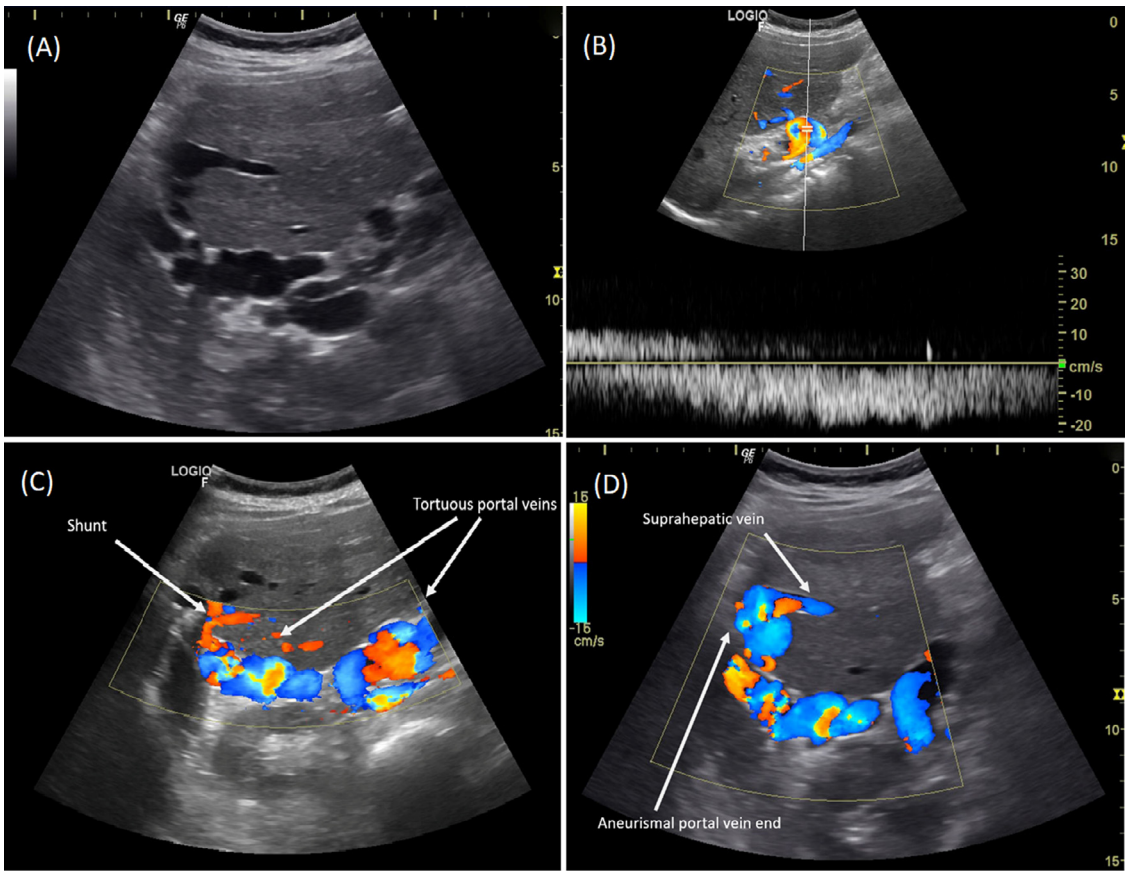

Fig. 1 – (A) Ultrasound showing tortuous structures under the liver; (B) color Doppler images showing continuous flow of the tortuous portal veins; (C) ultrasound at the level of the suprahepatic veins showing tortuous veins originating from the portal system joining the suprahepatic veins; (D) color Doppler images showing the aneurysmal portal system vein communicating with the right suprahepatic vein

the main cause of shunt formation, and the location of the shunt depends on the exact point at which the vitelline veins fails to differentiate [4].

Histology and classification

Histologic findings may include the absence of portal veins in small portal tracts, absent or hypoplastic portal veins in medium and large-sized portal tracts, isolated capillaries and arterioles in the lobules, hypertrophy of hepatic artery branches, and aneurysmal dilatations of the portal venous branches [7].

One of the first classifications of CPSS was proposed by Park et al [8], and it is as follows:

Type 1 - single tube-like vessel connecting the right branch of the portal vein (PV) to the inferior vena cava.

Type 2 - localized peripheral shunt, in which one hepatic segment contains anomalous communications between the peripheral branches of the PV and the hepatic veins.

Type 3 - Aneurysmal communication between the peripheral branches of the PV and the hepatic vein.

Type 4 - Multiple intrahepatic shunts in both lobes of the liver.

Several other classifications exist; the most commonly used classification was put forth by Stringer [2], who categorized CPSS into either intrahepatic or extrahepatic.

Complications

Portosystemic shunts are associated with numerous congenital anomalies including cardiac, urogenital, and other vascular malformations [9–11]. Concomitant liver abnormalities may also be present, such as malformations, hypoplasia or complete agenesis of the portal vein, congenital choledochal malformations, and biliary atresia [12].

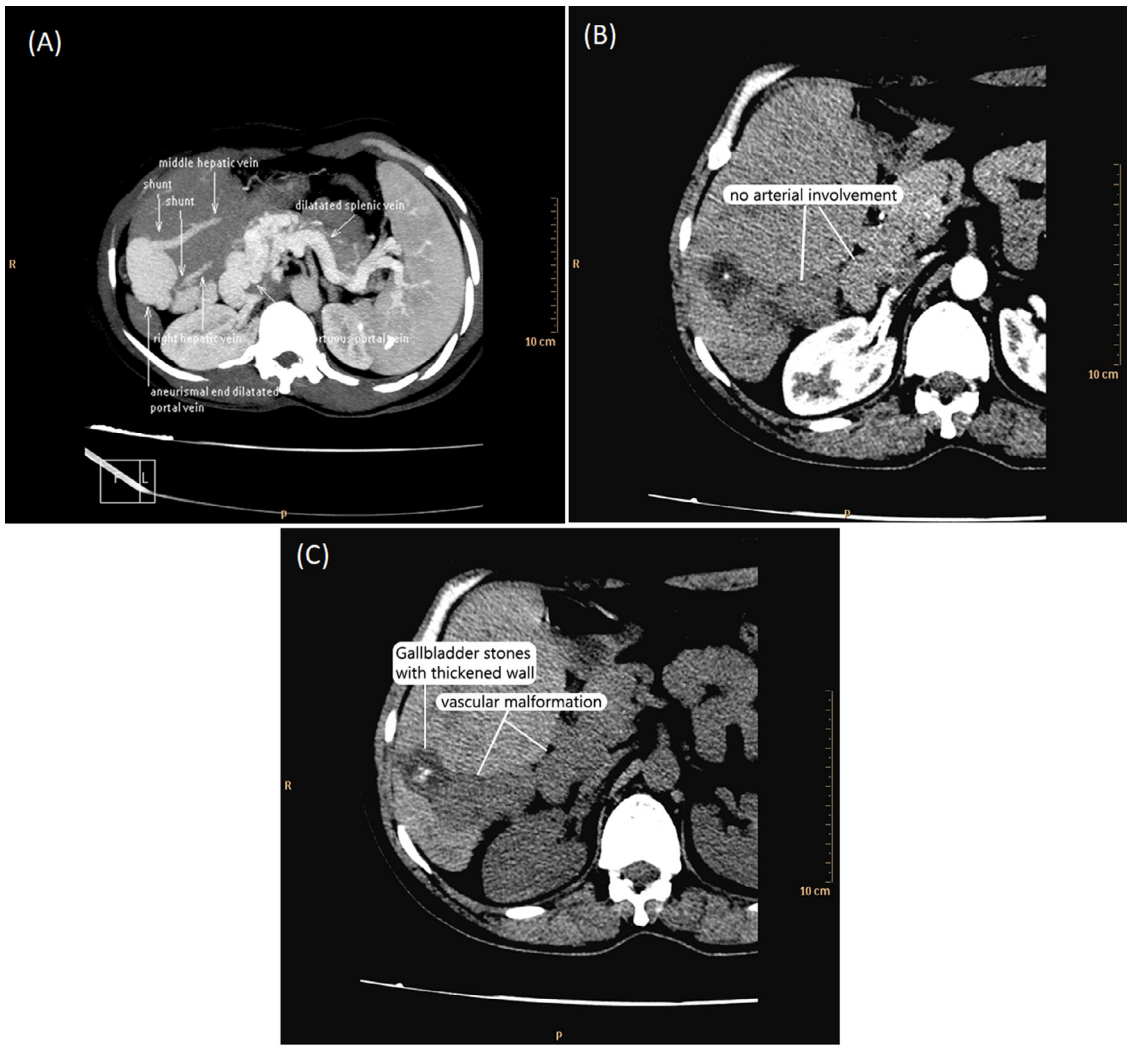

Fig. 2 – (A) Portal phase CT angiography shows dilated splenic vein, tortuous anomalous portal veins ending in an aneurysmal vein joining the middle hepatic vein; (B) axial CT in arterial phase showing no arterial involvement; (C) axial CT without contrast showing calculus cholecystitis and tortuous structures under the liver

In the pediatric population, important clinical manifestations of CPSS include hepatopulmonary syndrome, pulmonary hypertension, and hepatic encephalopathy [4,13,14]. They usually manifest as cyanosis, digital clubbing, and dyspnea. Other manifestations include neurocognitive decline, behavioral issues, learning disabilities, seizures, and failure to thrive [2,4].

In the adult population, a noteworthy complication of CPSS is the development of regenerating liver nodules due to altered local hemodynamics, which commonly occurs with extrahepatic shunts [15]. Hepatic malignancies are also more prevalent in these patients and are almost exclusively associated with EHPS [16,17]. Other less frequently encountered complications include gastrointestinal bleeding, vaginal bleeding, and renal complications such as membranoproliferative glomerulonephritis [18–20]. Endocrine disorders have also been reported, including hyperinsulinism, hypoglycemia, hypothyroidism, hyperandrogenism, amenorrhea, and precocious puberty [21,22]. Given these associated abnormalities, a comprehensive evaluation is essential for all patients with newly-diagnosed CPSS.

Management

Awareness of concomitant abnormalities is essential for treatment planning. Due to the high complication rate, computed tomography angiography should be performed whenever a vascular malformation is suspected to establish an accurate diagnosis. Management entails observation, correction of metabolic abnormalities, surgical correction of the shunt, local excision of the shunt, or in critical cases, liver transplant [1,6].

Case Report

A 40-year-old female with no past medical history presented to the Emergency Department complaining of right upper quadrant pain, nausea, and vomiting, especially after heavy meals. Physical examination revealed right upper quadrant tenderness with a positive Murphy's sign. Her lab results showed increased levels of ALP (135 U/L), AST (60 U/L), and ALT (55 U/L).

An abdominal ultrasound confirmed calculous cholecystitis. It also revealed a CPSS between branches of the portal and intrahepatic venous systems. The patient had no symptoms from the shunt. She did not show any signs of hepatic encephalopathy or hepatopulmonary syndrome. Further imaging studies were subsequently obtained.

Imaging findings

Ultrasound

Abdominal ultrasound showed gallbladder wall thickening measuring up to 5 mm and gallbladder stones up to 10mm in size. Subhepatic tortuous anechoic structures were also visualized with low continuous flow on color Doppler (Fig. 1).

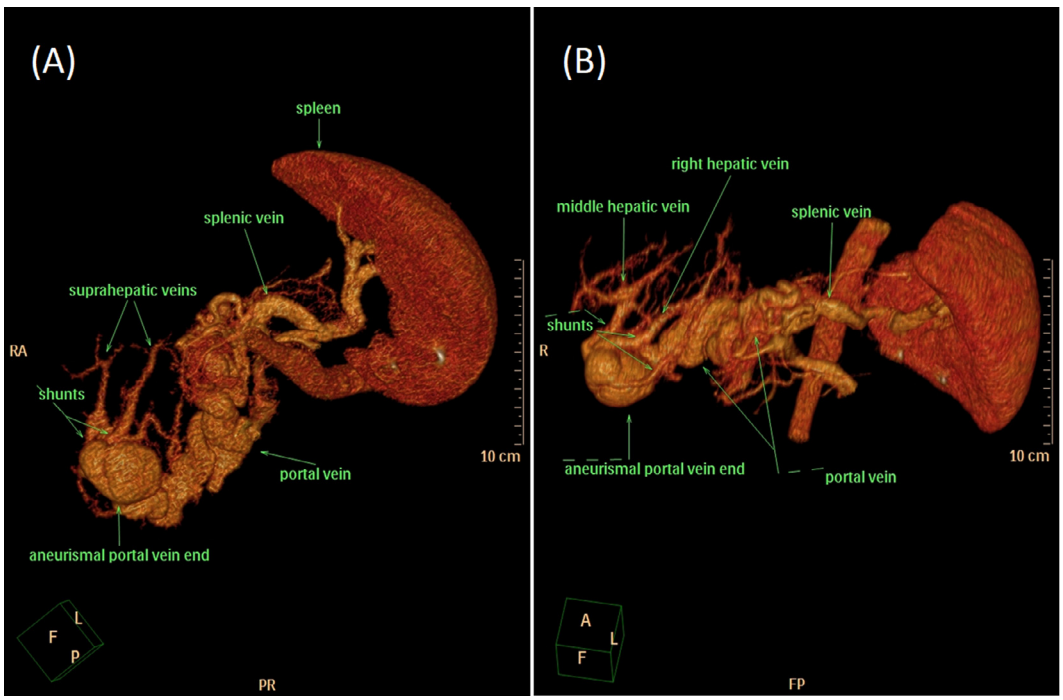

Fig. 3 – 3D reconstruction of abdominal CT angiography shows tortuous portal veins ending with an aneurysmal shunt into the suprahepatic veins

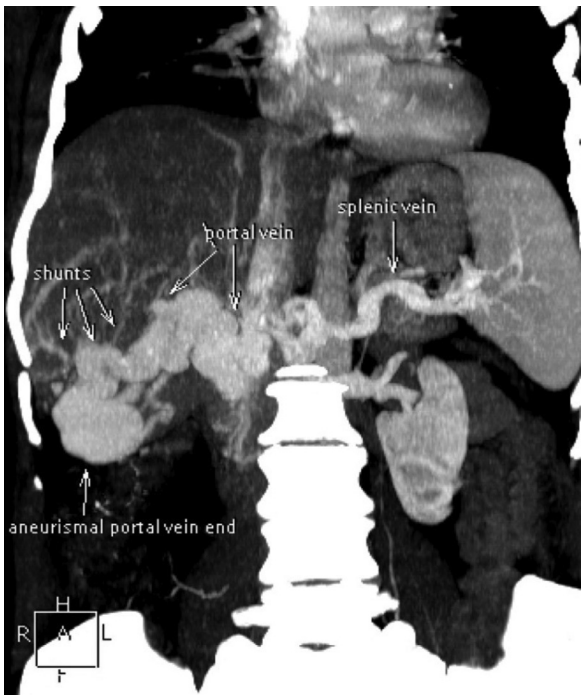

Fig. 4 – Coronal view in the portal venous phase

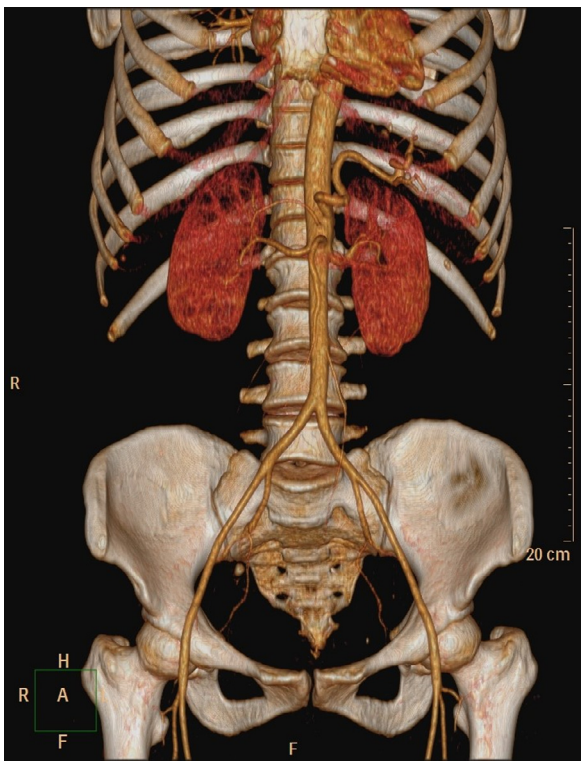

Fig. 5 – CT scan VR in arterial phase showing no arterial involvement in the tortuous structure

CT Angiography

Abdominal CT angiography confirmed calculous cholecystitis. The portal vein could not be visualized in the portal venous phase. Instead, tortuous varicose portal system veins with aneurysmal dilatation of portal venous branches were evident (Figs. 2–5). No anomalies of the abdominal aorta or its branches were seen.

Diagnosis

The patient was diagnosed with calculous cholecystitis. The secondary diagnosis was that of congenital absence of portal vein with Type 4 intrahepatic portosystemic shunts between portal branches and the suprahepatic vein.

Discussion

The pathogenesis of CPSS involves deviations from the normal embryological development of the portal and systemic venous circulation. The development of the hepatic venous system is a complex process that occurs between the fourth and 10th week of gestation [1,6]. Anomalies during these developmental events lead to the persistence of abnormal communications between the portal and systemic venous systems, the so-called CPSS.

IHPS is believed to arise mainly from incomplete involution of the vitelline veins, abnormalities that occur between the third and eighth week of gestation [2,4]. Studies suggest that type 1 IHPS arises from persistence of the right vitelline vein, while persistent communication between the vitelline venules within the newly formed hepatic sinusoids results in IHPS types 2-4 [2,4].

Due to the common association of CPSS with congenital heart disease, it has been speculated that the persistence of an embryonic vitelline vein may be caused by underlying anomalies with angiogenesis [1]. These anomalies then contribute to alterations of normal hemodynamics that might predispose to persistence of abnormal venous communications [1].

Despite the wide spectrum of clinical manifestation and complications from longstanding shunts, we present a case of an asymptomatic patient with intrahepatic CPSS, emphasizing the increasing incidence of CPSS as an incidental finding secondary to the widespread use of imaging for other indications. IHPS is more predominant in males as compared to females [13] and most patients are diagnosed during childhood [13], making our patient, a 40-year old previously asymptomatic woman, an uncommon presentation.

Conclusion

As cross-sectional imaging modalities such as ultrasound, CT, and magnetic resonance imaging become widely available, we anticipate an increase in the incidental diagnosis of rare congenital malformations such as CPSS. These incidental findings

further emphasize the variability and broad clinical manifestations of this condition.

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