Congenital portosystemic shunts: experience of a tertiary Tunisian referral center

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Congenital portosystemic shunt is a rare condition in which communications between the systemic venous circulation and the portal veins drain blood directly into the systemic circulation. Diagnosis may occur from the prenatal period to adulthood. Nevertheless, diagnosing and treating a congenital portosystemic shunt, particularly in the perinatal stage, remain challenging, as multiple complications can occur. This study aimed to describe the clinical characteristics of 3 cases of congenital portosystemic shunts diagnosed during pregnancy or the neonatal period, the diagnostic procedures, and their outcomes. This study reported 3 cases of full-term newborns with a congenital portosystemic shunt diagnosed at neonatal age. Case 1 was antenatally diagnosed with a congenital portosystemic shunt, which was confirmed postnatally via computed tomography and was associated with malformed ductus venosus and hypoplasia of the right portal vein. Cases 2 and 3 were siblings: a boy who had diffuse hemangiomatosis and a congenital portosystemic shunt complicated with severe persistent pulmonary hypertension and a girl who presented with a congenital portosystemic shunt and Kell alloimmunization. Congenital portosystemic shunts can be detected on prenatal ultrasounds during the etiologic workup of one of its complications or may be incidentally identified later in life. Children with congenital portosystemic shunts may develop various biological abnormalities, such as pulmonary hypertension, hypoxemia, encephalopathy, and liver tumors. A multidisciplinary approach and standardized protocols are required to optimize the management of congenital portosystemic shunts and minimize the short- and long-term consequences of congenital portosystemic shunts.

Key words: congenital portosystemic shunts, hepatology, newborn, prenatal diagnosis

Introduction

Congenital portosystemic shunt (CPSS) is a rare congenital malformation.[1] This condition consists of an abnormal communication between the portal and systemic veins, leading to a complete or partial diversion of the portal flow away from the liver to the systemic circulation, mainly the inferior vena cava (IVC).[2] This congenital condition is still incompletely understood. Specifically, CPSS results from the persistence of fetal vessels, which leads to abnormal communications between any vein of the portal system and any vein of the IVC system.[2]

The incidence of CPSS is estimated to be 1 in 30,000 live births.[3] However, as light increase in the incidence of CPSS has been reported, probably because of the enhancements in imaging technologies.[4,5] This condition may be asymptomatic and is sometimes presented with complications, although less frequently in the neonatal period.[6] Because of these abnormal connections, severe outcomes may occur, leading to biochemical and metabolic disorders, pulmonary vascular diseases (hepatopulmonary syndrome or pulmonary arterial hypertension), liver tumors, and even encephalopathy.[6] The management of CPSS remains a significant concern, and the multidisciplinary approach sheds light on the multifaceted and persistent challenge of handling this rare condition.

Here, we reported 3 cases of newborns diagnosed with CPSS and a

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This study was performed in line with the principles of the Declaration of Helsinki. Approval was obtained from the ethical committee of the Maternity and Neonatology Center of Tunis in September 2024. All patients' parents received explanations of the purposes, procedures, risks, and benefits of this study.

Informed consent was obtained from all the participants' parents included in the study.

The authors affirm that all the participants' parents provided informed consent.

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review of the literature to provide a better understanding of this condition and its management, especially in a resource-constrained country.

Materials and methods

We conducted a monocentric observational retrospective study over 5 years (January 1, 2019, to January 1, 2024) in the neonatology department of the Maternal and Neonatal Center of Tunis. We collected all cases diagnosed with CPSS. The inclusion criteria required at least 1 imaging study demonstrating CPSS. Demographic, clinical, and imaging data at diagnosis and follow-up data and therapeutic approaches were recorded retrospectively.

Medical investigations were performed in line with the principles of the Declaration of Helsinki. Approval was obtained from the ethical committee of the Maternity and Neonatology Center of Tunis in September 2024. Informed consent was obtained from all the participants' parents included in the study.

Results

Case 1

The case involves a full-term male infant delivered via cesarean delivery because of fetal distress, born to a mother who had COVID-19 during pregnancy. Prenatal ultrasound (US) at 25 weeks of amenorrhea revealed umbilical-portal-systemic venous shunts, agenesis of the ductus venosus, hepatic calcifications, and mildly dilated intestines. Abernethy syndrome anomalies were stable in the third trimester of pregnancy, as shown in Figure 1, with subsequent fetal echocardiography at 34 weeks of gestation, indicating significant cardiomegaly and biventricular hypertrophy.

Postnatally, the infant exhibited respiratory distress and hepatosplenomegaly but no facial dysmorphia. Echocardiography confirmed cardiac chamber dilation, pulmonary arterial hypertension (PAH), and a restrictive ductus arteriosus. Abdominal computed tomography (CT) scan confirmed a type I extrahepatic shunt and revealed a portocaval fistula through a malformed ductus venosus, hypoplasia of the right portal vein, dysmorphic liver with left hypertrophy and right atrophy, calcifications, and splenomegaly.

Treatment included 25 days of oxygen therapy and sildenafil for PAH. At 2.5 years, the child showed normal growth and development but still exhibited persistent PAH and portal hypertension, with occasional wheezing episodes. Follow-up US revealed a persistent 10-mm portosystemic shunt with intrahepatic circulation, signs of portal hypertension, and splenomegaly. Surgical intervention is now being considered because of ongoing complications.

Case 2

A male newborn was delivered via cesarean delivery at 37 weeks of gestation because of fetal macrosomia and polyhydramnios. Initial examination revealed central cyanosis, SpO2 of 88% on room air, severe respiratory distress, hepatosplenomegaly, and extensive hemangiomatosis covering >70% of his body. Early assessments showed hepatosplenomegaly with a vascular malformation involving multiple hepatic veins, including a dilated right hepatic vein. Transfontanellar US detected grade I intraventricular hemorrhage, whereas echocardiography revealed a large patent ductus arteriosus with significant PAH but no sign of heart failure. The newborn required mechanical ventilation and received inhaled nitric oxide and sildenafil to manage severe pulmonary hypertension. Despite treatment efforts, the newborn developed pulmonary hemorrhage and high-output heart failure because of the portosystemic shunt, necessitating vasoactive medications. Sadly, the infant passed away on the 21st day of life.

Case 3

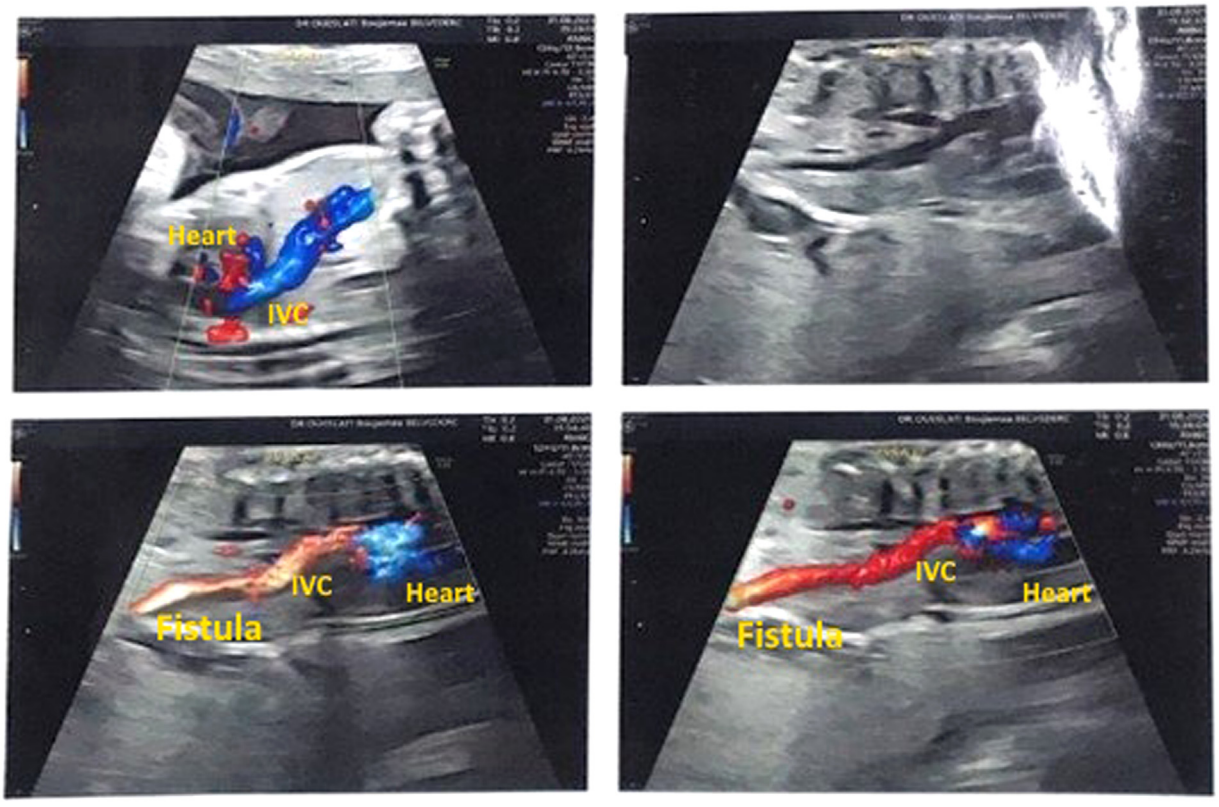
A female infant, the sister of case 2, was delivered via emergency cesarean delivery at 38 weeks of gestation because of fetal distress. The pregnancy was complicated by gestational diabetes mellitus. At 22 weeks of amenorrhea, fetal US revealed a ventricular septal defect, a portoumbilical shunt, and dilation of an umbilical vein collateral. In addition, an elevated uterine artery resistive index was noted. The newborn, weighing 3900 g, was admitted to the neonatal intensive care unit for monitoring. Initial physical examination revealed a heart murmur, clear respiratory sounds, and no sign of hydrops fetalis. Abdominal US and CT (Figure 2) on the second day of life revealed a fistula between the umbilical vein and the patent left portal branch, measuring 34 mm in length and 2.4 mm in diameter, along with homogeneous hepatomegaly (major axis of 48 mm). This shunt would likely fall under the category of type II extrahepatic shunts. A follow-up US at 22 days of life showed persistent dilation of the right hepatic vascular structure (30 mm), continuity with the left portal vein branch, and discrete homogeneous hepatomegaly. The patient was asymptomatic at the last clinical checkup at the age of 13 months.

Discussion

Knowledge regarding CPSS is scarce given that the low incidence of this malformation has prevented the realization of large studies. Here, we enrolled 3 cases of CPSS, representing the only Tunisian series to date. In line with previous studies in which approximately 75% of CPSSs were diagnosed during childhood, all cases of CPSSs had neonatal onset.[7]

The diagnosis of CPSS was suspected antenatally in 2 of 3 described cases. Previous studies have indicated that prenatal diagnosis is possible through US imaging, which can detect abnormal vascular connections between the portal and peripheral venous systems or an enlarged umbilical vein.[8,9] To date, CPSS is more frequently diagnosed prenatally, especially intrahepatic shunts, on US scanning. Extrahepatic portosystemic shunts, also known as "Abernethy malformations," are further classified into types I and II. Intrahepatic portosystemic shunts are not restricted to a single liver lobe and may consist of multiple portosystemic connections.[10] Almost 42% of CPSSs are diagnosed in utero with direct visualization of the shunt or indirect signs.[11] In a systematic review published in 2023, prenatal

FIGURE 1
Third-trimester ultrasound (Case 1): Umbilical-portal shunt branching into the inferior vena cava



IVC, inferior vena cava.

Jerbi. Congenital portosystemic shunts. Am J Obstet Gynecol Glob Rep 2024.

diagnosis happened mostly in the third trimester of pregnancy, with a median gestational age of 33 weeks.[12] In contrast with these findings, prenatal diagnosis was made earlier for cases 1 and case 3 at 25 and 22 weeks of amenorrhea, respectively.

Shunting-induced alterations in fetal venous circulation may cause reduced liver perfusion, intrauterine growth restriction, and cardiac failure, which was the case for the first case of the growth-restricted newborn reported in this series.[13] In accordance with previously reported cases and series, fetal heart failure and ventricular septal defect were antenatally detected in cases 1 and 3, respectively. Heart failure, with or without a congenital heart malformation, because of high cardiac output, was present in 27 of 168 patients in the Lambert review published in Archives of Cardiovascular Diseases.[14] Others reported associated malformations or syndromes in 30% to 45% of cases.[6] Many studies have described CPSS as a part of genetic syndromes, such as Down syndrome, trisomy 18, Noonan syndrome, and Turner syndrome.[10]

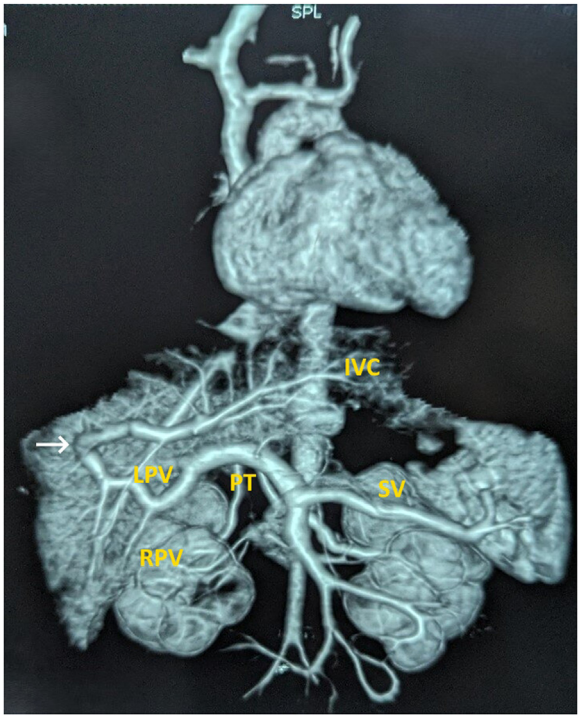
Patients with CPSS present with a wide spectrum of symptoms and complications that may occur during life, although asymptomatic cases, discovered incidentally on imaging, are not uncommon. Our study highlights the highly variable clinical manifestations and outcomes of this rare congenital malformation.

Cutaneous manifestations, although less common, have been reported in 5% of cases.[2] In the cases reported above, 1 patient had severe hemangiomatosis, and another patient had a positive family history of diffuse hepatic hemangiomatosis. A possible relationship between angiomas and CPSS has been highlighted, and many cases have been reported. Nevertheless, whether the presence of such an association represents a distinct syndrome is not yet known.

PAH was reported to be one of the most common cardiac complications of CPSS.[15] Although the exact cause is not fully understood, it is believed that poor hepatic clearance of vasodilator substances affecting the pulmonary endothelium plays a role. Neonatal presentation may include subtle hypoxia.[16] Of note, 2 of our patients presented with this condition, and one of our patients died from severe PAH diagnosed on the first day of life.

Finally, the management of CPSS needs a multidisciplinary approach to manage systemic manifestations and

FIGURE 2
Postnatal Abdominal CT (Case 3): Fistula between umbilical vein and patent left portal branch with homogeneous hepatomegaly



IVC, inferior vena cava; *LPV*, left portal vein; *PT*, portal trunk; *RPV*, right portal vein; *SV*, splenic vein.
Jerbi. Congenital portosystemic shunts. Am J Obstet Gynecol Glob Rep 2024.

the development of further complications. A close follow-up is very important to identify possible complications as soon as possible, and new guidelines are now available to help clinicians manage this condition.[17]

Although intrahepatic shunts have been reported to close spontaneously, especially in newborns,[18] authors suggest referring incidentally diagnosed CPSS after the age of 2 years or symptomatic CPSS at any age to specialized centers for better evaluation and treatment.[19]

Conclusion

Our 3 cases illustrate the clinical manifestations and evolutionary aspects of CPSS in the neonatal population. CPSSs are associated with severe complications, which can be challenging to manage. Few neonatal studies have been conducted, and even fewer studies have been reported in low-income countries. Although antenatal diagnosis is more common in our country with the widespread use of US, standardized care and specialized centers are not yet available.

Glossary

CPSS: Congenital portosystemic shunt
CT: Computed tomography
IVC: Inferior vena cava
LPV: Left portal vein
PHA: Pulmonary arteria hypertension
PT: Portal trunk
RPV: Right portal vein
SV: Splenic vein
US: Ultrasound

CRediT authorship contribution statement

Bilel Jerbi: Conceptualization. **Hajer Chourou:** Data curation. **Rim Ben Aziza:** Formal analysis. **Omar Jelassi:** Methodology. **Yosra Sdiri:** Writing – original draft. **Wafa Belhadj Ammar:** Investigation. **Samia Kacem:** Validation. **Nadia Aloui:** Formal analysis. **Radhouane Achour:** Supervision.

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