Arch Argent Pediatr 2023;121(2):e202202570

Portosystemic shunt, a rare cause of neonatal cholestatic hepatitis. A case report

Sofía C. Geffner[a], *Macarena L. Atun*[a], *Sebastián García López*[a], *María P. Carrascal Gutiérrez*[a]

ABSTRACT

Congenital portosystemic shunt is a venous vascular abnormality that connects portal and systemic circulation, resulting in diversion of the blood flow, bypassing the hepatic passage. It is a rare malformation; its incidence varies from 1:30 000 to 1:50 000 newborns. It may be asymptomatic or present with complications in the pediatric age or, less frequently, in the neonatal age.

Upon diagnosis, the need for a surgical or an intravascular intervention for closure should be defined. This decision depends on the malformation anatomical characteristics, clinical manifestations, and complications.

We present the case of a 1-month-old patient referred to our center for the study of neonatal cholestatic hepatitis, with a diagnosis of extrahepatic portosystemic shunt. Intravascular closure of the defect was performed with significant subsequent improvement.

Key words: *portosystemic shunt; cholestasis; vascular malformations; neonatal hyperbilirubinemia; liver.*

doi: http://dx.doi.org/10.5546/aap.2022-02570.eng

To cite: Geffner SC, Atun ML, García López S, Carrascal Gutiérrez MP. Portosystemic shunt, a rare cause of neonatal cholestatic hepatitis. A case report. *Arch Argent Pediatr* 2023;121(2):e202202570.

[a] *Department of Neonatology, Hospital Italiano de Buenos Aires, City of Buenos Aires, Argentina.*

Correspondence to *Sofía C. Geffner: sofia.geffner@hospitalitaliano.org.ar*

Funding*: None.*

Conflict of interest*: None.*

Received*: 1-13-2022*
Accepted*: 6-6-2022*

This is an open access article under the Creative Commons Attribution–Noncommercial–Noderivatives license 4.0 International. Attribution - Allows reusers to copy and distribute the material in any medium or format so long as attribution is given to the creator. Noncommercial – Only noncommercial uses of the work are permitted. Noderivatives - No derivatives or adaptations of the work are permitted.

INTRODUCTION

Congenital portosystemic shunt (PSS) is a vascular malformation that connects portal and systemic circulation, bypassing the hepatic passage. It may be asymptomatic or present clinically with associated complications. In some cases, surgical or intravascular closure may be indicated.

Below, we describe the case of a 30-day-old patient with severe cholestatic hepatitis since birth. Potential differential diagnoses are considered, being extrahepatic PSS the underlying etiology. The incidence of this condition is low and its severe clinical presentation in the neonatal period is rare.

CASE REPORT

Term newborn infant (gestational age of 39 weeks), with a low birth weight for gestational age (2460 grams, percentile 2), with no other relevant perinatal history. The infant was born in San Luis, Argentina, where studies for jaundice, choluria, and acholia were initiated since birth. Cholestatic hepatitis, anemia, thrombocytopenia, and altered coagulation profile (*Table 1*) were confirmed by laboratory tests.

As a first approach, the following tests and exams were carried out:

- Serology tests: negative for adenovirus, cytomegalovirus, Coxsackie B1 and B6 viruses, herpes 1 and 2, parvovirus, HIV, hepatitis B and C, toxoplasmosis, rubella, Chagas disease, and varicella.
- Abdominal Doppler ultrasonography: hepatosplenomegaly with no other findings.

In his place of origin, the infant received phototherapy and phenobarbital for the treatment of hyperbilirubinemia, and ursodeoxycholic acid for cholestasis. Because of the poor response and failure to reach a diagnosis, the decision was made to refer the patient to a tertiary care facility.

At the time of referral, the patient was 31 days of life. Poor weight gain, generalized jaundice, choluria, persistent acholia, and a

Table 1. Patient´s laboratory values. The first two columns correspond to values provided by the hospital of origin in San Luis; the remaining four columns correspond to tests performed at our hospital

	Birth	14 days of life	31days of life (admission to our hospital)	43 days of life (day when the procedure was performed)	73 days of life (30 days after the procedure)	193 days of life (5 months after the procedure)
Hematocrit (%)	29		28	31	29	35
Hemoglobin (g/dL)	9.5		10	10.8	9.8	12.7
White blood cells (cells/mm^3)	15 000		27 300	5500	8500	10 800
Platelets (cells/mm^3)	44 000		72 000	130 000	124 000	386 000
GGT (IU/L)	16	51	75	84	52	92
Total bilirubin (mg/dL)	8.3	42	29	23	15	0.5
Direct bilirubin (mg/dL)	2.9	22	15	12	8	0.2
Alkaline phosphatase (IU/L)	460	345	174	288	556	338
AST (IU/L)	345	1020	446	249	301	242
ALT (IU/L)	99	471	215	109	211	256
Prothrombin time (%)	146		46			119
aPTTK (seconds)	95		36			30
Bile acids in blood (µmol/L)		94	74			
Ammonium (mcg/dL)			191	178	119	104
Alpha-fetoprotein (ng/mL)			67 255		80 000	137
Galactosemia (neonatal screening)			Normal			
Ferritin (mg/mL)			> 15 000			
Albumin (g/dL)			1.9	2.54	3.13	3.79
Proteinemia (g/dL)			3.3	3.24	4.76	5.62
Gammaglobulin (g/dL)			0.23			

GGT: gamma-glutamyltransferase; AST: aspartate aminotransferase; ALT: alanine aminotransferase; aPTTK: activated partial thromboplastin time with kaolin.

slight tendency to irritability were confirmed. Cardiac auscultation revealed a predominantly mesocardial holosystolic murmur. On admission, cholestatic hepatitis with hyperammonemia, increased ferritin, alpha-fetoprotein and bile acids levels in the blood, anemia, and thrombocytopenia persisted (*Table 1*).

Neonatal screening was repeated with 6 normal tests. Potential infectious, metabolic, toxicological, and anatomical causes of cholestatic hepatitis were evaluated. The following results were obtained from complementary studies:

- Color Doppler echocardiogram: *ostium secundum* atrial septal defect, ventricular septal defect, dilation of both ventricles with preserved contractility.
- Abdominal color Doppler ultrasonography: extrahepatic PSS (*Figure 1 A*): venous vessel connecting the main portal vein to the inferior vena cava. Dilated hepatic artery. Portal vein and small-sized branches. No biliary tract abnormalities were observed.
- Abdominal angiotomography (*Figures 1B* and *C*): tortuous duct (6.9 mm) that originated

Figure 1 A. Doppler ultrasonography showing an anomalous communication between the portal and systemic venous systems (arrow). 1 B and C. Computed tomography, transverse (B) and coronal (C) sections showing the malformation (arrow)

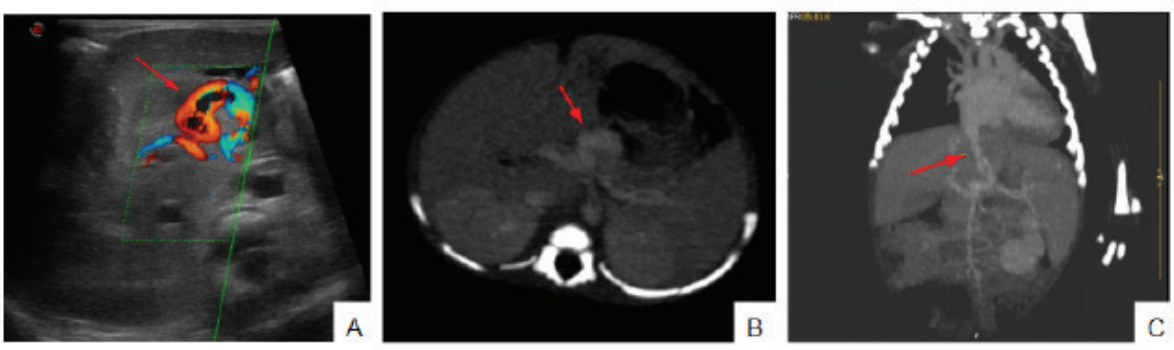
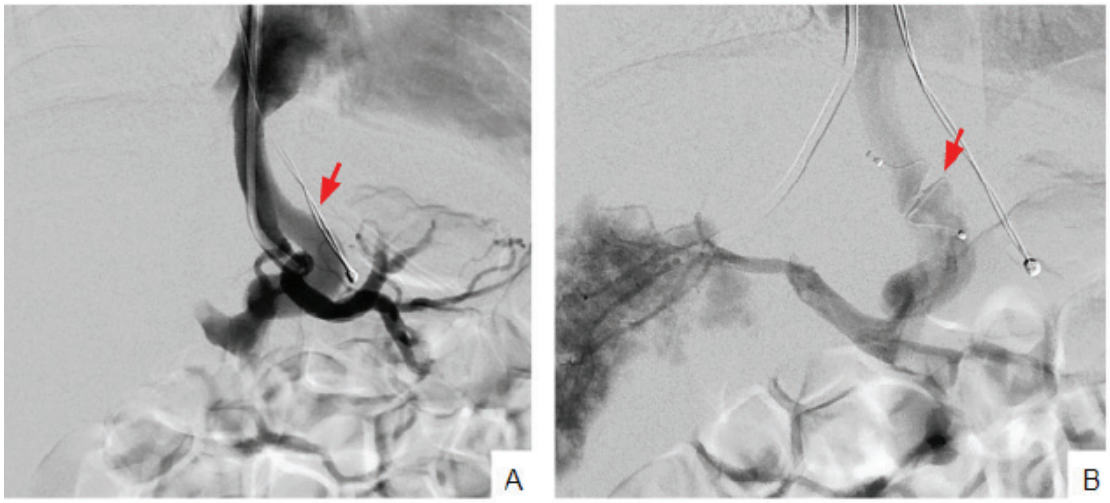


Figure 2. Digital angiography, arrow showing the shunt: before (A) and after (B) placement of an intravascular plug. Blood flow is observed through the vascular defect after placement of the device (B)



cephalad to the splenic and superior mesenteric vein confluence, and drained into the inferior vena cava. Trunk of the portal vein and its patent branches. Increased caliber of the hepatic artery (2.6 mm).

In view of the diagnosis of PSS, with clinical and laboratory implications, the malformation was occluded under digital angiography. Portography confirmed portal vein patency, the shunt was catheterized and embolized using a 4 mm plug, preserving blood flow through it (*Figure 2*). Given the size of the defect, the decision was made to perform a partial closure with the aim of completing it on a second stage. In this way, the risk for complications associated with portal hypertension derived from the system hypoplasia was minimized. After the procedure, nutritional support was initiated with total parenteral nutrition and gradual progression to the enteral route, thus preserving mesenteric blood flow. No complications secondary to portal hypertension were observed.

In the subsequent follow-up, clinical and laboratory improvement was noted, with a decrease in serum bilirubin levels (*Figure 3*). *Table 1* shows blood test results 30 days and 5 months after the procedure. The persistence of hepatitis is highlighted, with elevated transaminases, gamma-glutamyltransferase (GGT), alkaline phosphatase, and ammonium, and normalization of serum bilirubin, hematocrit, and platelet count.

At 85 days of life (42 days after the procedure), the liver Doppler ultrasonography showed a well-positioned plug in the PSS, with no blood flow inside and patent portal veins. After 3 and a half months, the patient was discharged back to his place of origin.

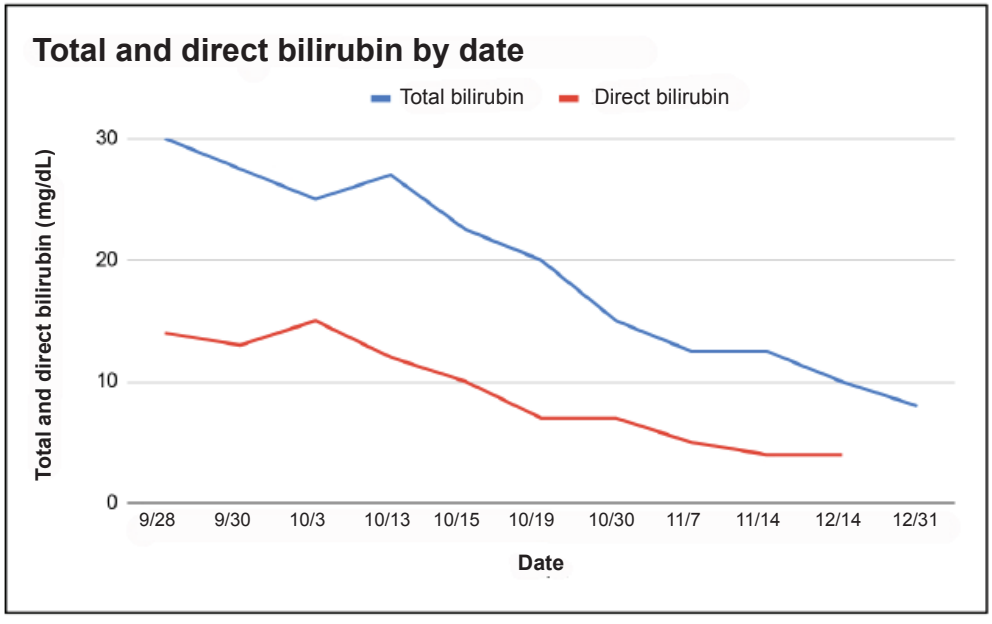
In the subsequent follow-up, he showed improvement in weight and height. He is being followed-up by the Departments of Neurology and Ear, Nose and Throat for altered otoacoustic emissions and automated brainstem auditory evoked potentials.

DISCUSSION

In fetal liver development, there are numerous intra- and extrahepatic connections between portal and systemic veins, which later resolve.

Congenital PSS is a rare vascular malformation that consists of persistent anomalous communications between the portal and systemic venous systems.[1] One or more intra- or extrahepatic vessels may partially or totally divert the portal blood flow to the systemic circulation (bypassing the hepatic passage) and are associated with various complications.[2]

Figure 3. Graph showing serum total bilirubin (in blue) and direct bilirubin (in red) values. A significant decrease is observed since the closure of the malformation (on October 10th.)



The incidence is estimated to be 1:30 000/50 000 newborns.[1,3,4] Diagnosis may be prenatal or postnatal in the context of a patient under study because of increased galactose levels detected during newborn screening tests, cholestatic hepatitis, delayed growth or other associated complications. Occasionally, it may be an incidental finding. It is usually detected in childhood or adulthood, while its diagnosis secondary to complications present in the neonatal period (such as those present in the clinical case described) is more infrequent.

Given this diagnosis, a comprehensive search should be performed to detect vascular, renal, visceral, skeletal, and cardiac abnormalities.[2–5] The most frequently described malformations are atrial or ventricular septal defect, persistent ductus arteriosus, patent foramen ovale, aortic coarctation, tetralogy of Fallot, and valvular heart disease.[6] These associations suggest a prenatal defect during cardiac and abdominal vascular development, or the presence of adaptive changes in the heart in response to the hyperdynamic effect of PSS. In the clinical case described, atrial and ventricular septal defects are described as findings.

It has also been associated with genetic syndromes (e.g., trisomy 21).[3]

Complications related to PSS include neonatal cholestasis, liver failure, polysplenia with cytopenias, hepatic tumors, hepatopulmonary syndrome, and hepatic encephalopathy.

The diagnosis is made through imaging studies, being liver Doppler ultrasonography the first-line modality. Among the possible ultrasonographic findings, Ponziani et al.,[6] describe the following as signs compatible with the presence of PSS: 1) focal hepatic solid lesions; 2) abnormal communications between the portal and the systemic venous systems; 3) hypoplasia of portal vasculature; 4) compensatory dilation of the hepatic artery.

To obtain more information about the anatomy of the defect and the extent of portal system development, a CT scan with intravenous contrast is recommended.[2] Magnetic resonance imaging may be ordered instead, but it usually renders less information. Digital angiography is useful to assess the presence of communications not detectable by other methods. With this method, a balloon shunt occlusion test can be performed to measure portal venous pressure (which reflects the degree of hypoplasia) and to evaluate whether an eventual complete closure of the malformation will be well tolerated.[1,2] Portal venous pressure should not exceed 18–25 mmHg.[1]

The need for treatment is based on two characteristics: the anatomy (dimension and location) and the pressure measured in the occlusion test. In malformations of smaller caliber –intrahepatic and not associated with complications– an expectant management can be adopted, since they may resolve spontaneously within the first 2 years of life.[2] Larger extrahepatic shunts associated with complications or those that have not closed spontaneously may require a percutaneous or surgical closure. In these situations, it is suggested to perform an occlusion test and, if high pressures are present, to perform a 2-stage closure to decrease the risk of developing postoperative acute portal hypertension.

Regarding the mid- and long-term course, intrahepatic portal system plasticity favors liver revascularization after the shunt closure, even when no portal structures are detected in previous imaging studies. This leaves little or no room for liver transplantation in the management of these children.

CONCLUSION

PSS is a rare malformation and can be diagnosed as a finding in asymptomatic patients or associated with complications. The patient described in our clinical case, in the neonatal period, had multiple manifestations associated with the diagnosis: cholestatic hepatitis, jaundice, cytopenias, coagulopathy, and failure to thrive. Early presentation is very unusual, so diagnostic suspicion is critical to request a thorough ultrasonographic search (an unexperienced operator might not recognize it) and reach a diagnosis. ■

REFERENCES

1. McLin VA, Abella SF, Debray D, Guérin F, et al. Congenital Portosystemic Shunts: Current Diagnosis and Management. *J Pediatr Gastroenterol Nutr*. 2019; 68(5):615-22.
2. Bernard O, Franchi-Abella P, Branchereau S, Pariente D, et al. Congenital portosystemic shunts in children: recognition, evaluation, and management. *Semin Liver Dis*. 2012; 32(4):273-87.
3. Tang H, Song P, Wang Z, Han B, et al. A basic understanding of congenital extrahepatic portosystemic shunt: incidence, mechanism, complications, diagnosis, and treatment. *Intractable Rare Dis Res*. 2020; 9(2):64-70.
4. Franchi-Abella S, Gonzales E, Ackermann O, Branchereau S, et al. Congenital portosystemic shunts: diagnosis and treatment. *Abdom Radiol (NY)*. 2018; 43(8):2023-36.

5. Massin M, Verloes A, Jamblin P. Cardiac anomalies associated with congenital absence of the portal vein. *Cardiol Young*. 1999; 9(5):522-5.

6. Ponziani FR, Faccia M, Zocco MA, Giannelli V, et al. Congenital extrahepatic portosystemic shunt: description of four cases and review of the literature. *J Ultrasound*. 2019; 22(3):349-58.