arTs Università degli Studi di Trieste
Archivio della ricerca – postprint

Young boy with a long history of splenomegaly and cytopenia

Laura De Nardi ,[1] Chiara Zanchi ,[2] Luca Basso,[3] Daniela Sanabor,[4] Grazia Di Leo,[5] Egidio Barbi[1,2]

A 15-year-old boy was admitted with a history of cytopenia (white blood cell count 3.170/μm, platelets 90.000/μm) associated with splenomegaly, found during investigations for recurrent mild jaundice due to Gilbert's syndrome.

He was in good general health, without systemic symptoms; therefore, the leading causes of asymptomatic splenomegaly were excluded. Coagulation, liver tests and abdomen ultrasound (US) were normal, showing a hepatopetal portal flow to the colour-Doppler. There was no sign of haemolysis on haematology investigations. The C reactive protein, immune globulins levels and erythrocyte sedimentation rate were normal, excluding both an infective and an immune regulation disorder. We excluded the haematological malignancy and lymphoproliferative disorders through a peripheral blood smear and a bone marrow biopsy.

His history was remarkable for neonatal sepsis, which required umbilical venous catheter during hospitalisation in a neonatal intensive care unit (NICU). The patient follow-up was interrupted for a while, probably due to his good health condition.

At age 17 years, the child accessed our emergency department. for a minor trauma to the limbs, and his physical examination was unremarkable, except for the splenomegaly. We repeated the abdomen US, with colour flow Doppler (figure 1).

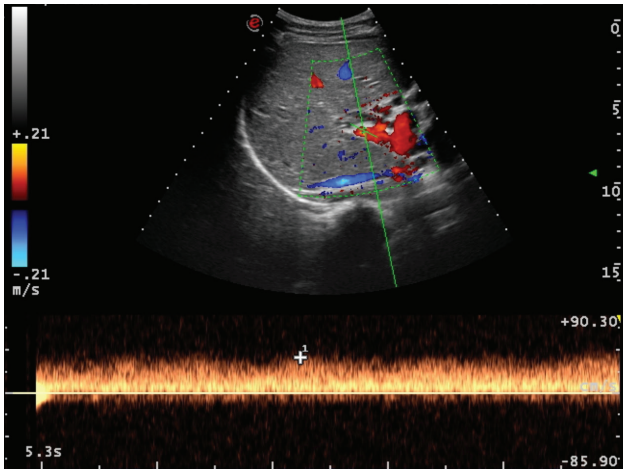

Figure 1 B-mode shows 1.4 cm of maximum calibre of portal vein at hilus with slightly perihilar hyperechogenicity; colour and power Doppler US shows preserved hepatopetal flow and PSV of 41 cm/s (normal range 20–40 cm/s). PSV, peak systolic velocity; US, ultrasound.

QUESTIONS

1. What is the most likely diagnosis?
    1. Portal vein obstruction
    2. Gaucher disease
    3. Autoimmune sclerosing cholangitis
    4. Leukemic hepatic infiltration
2. What is the gold standard imaging for diagnosis?
    1. Abdomen Doppler US
    2. Contrast-enhanced CT
    3. CT without contrast
    4. Ultrasound-based elastography
3. How should this child be managed?
    1. Upper gastrointestinal endoscopy
    2. Beta-blocker therapy
    3. Left-mesenteric portal vein bypass (Meso-Rex bypass)
    4. Transjugular intrahepatic porto-systemic shunt

Answers can be found on page 119.

ANSWERS TO THE QUESTIONS ON PAGE 118

ANSWER 1. A

The US showed numerous tortuous vessels occupying portal vein bed, consistent with a portal cavernoma. Portal vein obstruction (PVO), with secondary portal cavernoma, is one of the most frequent causes of portal hypertension in paediatric age and the leading identified causes are the direct injury of the vein (omphalitis and umbilical vein catheterization) and neonatal sepsis.[1] The first manifestations of PVO are usually upper gastrointestinal bleeding or isolated splenomegaly on routine examination in asymptomatic individuals. Cytopenia related to hypersplenism can be the only initial sign.[2] The absence of hepatomegaly is one of the diagnostic hallmarks of PVO since the liver is even smaller than normal due to the reduced portal blood supply. Gaucher disease is considered in the differential diagnosis as it can first show up just through a splenomegaly with cytopenia and bone pain, expression of bone marrow foam cells' infiltration. The hepatic US can show hypoechoic, hyperechoic or mixed nodules due to Gaucher cell accumulation in the earlier stages, and fibrosis in the later stage. In this case, Gaucher disease was ruled out by normal acid phosphatase and chitotriosidase levels. Leukemic hepatic infiltration is usually associated with hepatomegaly and abnormal liver enzymes and function tests. It was excluded by the normality of both the peripheral blood smear and the bone marrow biopsy. A sclerosing cholangitis was unlikely for the absence of clinical and biochemical evidence of cholestasis.

ANSWER 2. A

When PVO is suspected, US is the first-line imaging method to use, since it achieves an accuracy ranging from 88% to 98%, with high sensitivity and specificity.[3] However, the US suffers from some limitations and may sometimes be inconclusive (40% of patients with PVO require another US to make the diagnosis). This may be due to patient obesity/meteorism or insufficient operator's experience.[4] Contrast-enhanced CT and contrast-enhanced MRI better define the extent of thrombosis, allowing a detailed mapping of portosystemic collaterals, crucial for planning the interventions aimed to recanalise the PV system (figure 2).[5] US elastography is primarily used as an alternative to liver biopsy for the assessment of hepatic fibrosis and to predict complications in patients with cirrhosis. Finally, CT without contrast is not useful in this condition.

ANSWER 3. A

When PVO is suspected, all patients should first undergo upper gastrointestinal endoscopy to check for the presence, degree and status of oesophageal varices and hypertensive gastropathy, which will allow for a better planned therapeutic approach. Prophylactic band ligation or sclerotherapy of varices at risk of bleeding are the most appropriate choices in such patients' management. Beta-blocker therapy, in order to decrease pressure within the portal system, is now very rarely employed, since its use in paediatric age is debated.[6 7] The Meso-Rex bypass is an important therapeutic option in patients with extrahepatic portal vein thrombosis. However, surgical timing has not been well standardised by paediatric liver centres worldwide. Recent consensus guidelines suggest that the Meso-Rex bypass should be offered for primary and secondary prophylaxis of variceal bleeding and other complications (severe hypersplenism, growth retardation and symptomatic portal biliopathy due to biliary stasis) when there is favourable anatomy and suitable surgical expertise.[6 8] Unlike the transjugular intrahepatic portosystemic shunt (TIPSS), a percutaneous image-guided procedure in which a conduit is constructed within the liver diverting the portal blood flow into the systemic circulation, this approach restores the portal flow to the liver, carrying mesenteric and splenic venous blood around the obstructed portal tract, through the interposition of an autologous jugular vein graft between superior mesenteric vein and part of the left portal system.[2 9–12] Nevertheless, the Meso-Rex bypass is very rarely performable in children who underwent umbilical vein catheterization because they often lack the Rex recessus patency. In presence of recurrent and severe variceal bleeding, surgical treatment must be considered. Even if the TIPSS and liver transplant are exceptional options, when the Meso-Rex bypass is not feasible, TIPSS should be considered as a bridge to liver transplant.

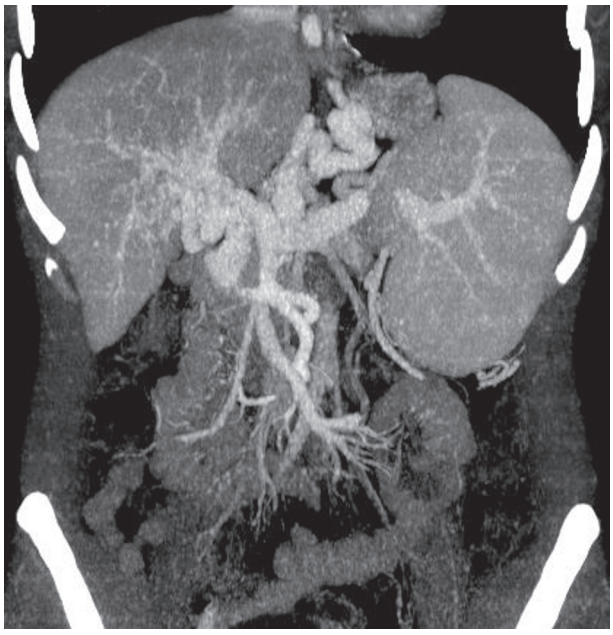

Figure 2 Contrast-enhanced CT shows a portal vein of normal calibre and tortuosity of many collateral hilar veins, suggestive for portal cavernoma.

PATIENT OUTCOME

The boy underwent gastroscopy which revealed degree F1 oesophageal varices. Since his nearly adult age, he started beta blocker therapy and was referred to a specialist liver centre in order to assess Rex recessus patency and perform surgery to restore the normal portal flow. PVO should be suspected and carefully investigated in all children with isolated splenomegaly and cytopenia, especially with a history of admission to NICU, since the prevalence in these patients is between 12% and 40%.[2 10 11]

Laura De Nardi ,[1] **Chiara Zanchi** ,[2] **Luca Basso,**[3] **Daniela Sanabor,**[4] **Grazia Di Leo,**[5] **Egidio Barbi**[1,2]

[1]Department of Medicine, Surgery and Health Sciences, University of Trieste, Trieste, Italy
[2]Department of Pediatrics, Institute for Maternal and Child Health IRCCS "Burlo Garofolo", Trieste, Italy
[3]Radiology, Hospital San Martino, Genova, Liguria, Italy
[4]Department of Radiology, Institute for Maternal and Child Health IRCCS "Burlo Garofolo", Trieste, Italy
[5]Pediatric Gastroenterology, Endoscopy and Nutrition Unit, Institute for Maternal and Child Health IRCCS "Burlo Garofolo", Trieste, Italy

Correspondence to Dr Laura De Nardi, Department of Medicine, Surgery and Health Sciences, University of Trieste, Trieste, Italy; laura.denardi1993@gmail.com

Acknowledgements The authors thank Martina Bradaschia MD for the English revision of the manuscript.

Contributors LDN and CZ drafted the manuscript. DS and LB did the ultrasound and processed images. GDL contributed to the conception of the work. EB critically revised the article for relevant intellectual content.

Funding The authors have not declared a specific grant for this research from any funding agency in the public, commercial or not-for-profit sectors.

Competing interests None declared.

Patient consent for publication Parental/guardian consent obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

© Author(s) (or their employer(s)) 2022. No commercial re-use. See rights and permissions. Published by BMJ.

To cite De Nardi L, Zanchi C, Basso L, *et al*. *Arch Dis Child Educ Pract Ed* 2022;**107**:118–120.

Accepted 6 September 2020

Arch Dis Child Educ Pract Ed 2022;**107**:118–120. doi:10.1136/archdischild-2019-318626

ORCID iDs
Laura De Nardi http://orcid.org/0000-0002-8141-3389
Chiara Zanchi http://orcid.org/0000-0001-8513-1963

REFERENCES

1 Schettino GCM, Fagundes EDT, Roquete MLV, *et al*. Portal vein thrombosis in children and adolescents. *J Pediatr* 2006;82:171–8.
2 Abd El-Hamid N, Taylor RM, Marinello D, *et al*. Aetiology and management of extrahepatic portal vein obstruction in children: King's College Hospital experience. *J Pediatr Gastroenterol Nutr* 2008;47:630–4.
3 Margini C, Berzigotti A. Portal vein thrombosis: the role of imaging in the clinical setting. *Dig Liver Dis* 2017;49:113–20.
4 Ferri PM, Ferreira AR, Fagundes EDT, *et al*. Portal vein thrombosis in children and adolescents: 20 years experience of a pediatric hepatology reference center. *Arq Gastroenterol* 2012;49:69–76.
5 Jha RC, Khera SS, Kalaria AD. Portal vein thrombosis: imaging the spectrum of disease with an emphasis on MRI features. *AJR Am J Roentgenol* 2018;211:14–24.
6 Grammatikopoulos T, McKiernan PJ, Dhawan A. Portal hypertension and its management in children.. *Arch Dis Child* 2018;103:186–91.
7 Gana JC, Cifuentes LI, Gattini D, *et al*. Band ligation versus beta-blockers for primary prophylaxis of oesophageal variceal bleeding in children with chronic liver disease or portal vein thrombosis. *Cochrane Database Syst Rev* 2019;9:CD010546.
8 Shneider BL, de Ville de Goyet J, Leung DH, *et al*. Primary prophylaxis of variceal bleeding in children and the role of MesoRex bypass: summary of the Baveno VI pediatric satellite symposium. *Hepatology* 2016;63:1368–80.
9 Williams S, Chan AKC. Neonatal portal vein thrombosis: diagnosis and management. *Semin Fetal Neonatal Med* 2011;16:e339:329–39.
10 Alvarez F, Bernard O, Brunelle F, *et al*. Portal obstruction in children. I. clinical investigation and hemorrhage risk. *J Pediatr* 1983;103:696–702.
11 Weiss B, Shteyer E, Vivante A, *et al*. Etiology and long-term outcome of extrahepatic portal vein obstruction in children. *World J Gastroenterol* 2010;16:4968–72.
12 di Francesco F, Grimaldi C, de Ville de Goyet J. Meso-Rex bypass--a procedure to cure prehepatic portal hypertension: the insight and the inside. *J Am Coll Surg* 2014;218:e23–36.