Clinical Case Report

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A case report on an incidental discovery of congenital portosystemic shunt

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Abstract

Rationale: Congenital portosystemic shunt (Abernethy malformation) is a rare entity causing the portal blood to drain directly into the systemic circulation, eluding the liver. These shunts arise through disturbances in the embryonic development.

Patient concerns: A 9-year-old male patient was referred to our department for further evaluation of a vascular malformation which was diagnosed in another facility when the patient was 2 years old, following a routine abdominal ultrasound. The patient had no complaints and the physical examination was normal at all times.

Diagnosis: Laboratory tests and esogastroduodenoscopy were normal. The abdominal ultrasound showed a side-to-side shunt between a short portal trunk and the inferior vena cava. A hepatic mass suggestive for focal noduar hyperplasia was seen in the left liver lobe. Abdominal angio-computed tomography (angio-CT) was performed and the ultrasonographic anomalies were confirmed. Multiple other vascular malformations were diagnosed—hepatic artery emerging from the superior mesenteric artery, with early division; hepatic veins forming a short common trunk before draining into the inferior vena cava; supranumerary right renal artery emerging from the aorta, tributary for the upper renal pole. Ecocardiography showed left superior vena cava persistence. The final diagnosis was Abernathy malformation type IB. In the meantime the patient was diagnosed with allergic asthma.

Interventions: No surgical cure was pursued because the malformation was an incidental discovery.

Outcomes: The patient was followed-up closely from the final diagnosis (when he was 9 years old) to present (he is currently 10 years old) with no change in his status—he remained asymptomatic.

Lessons: Angio-CT should be the performed whenever a vascular malformation is suspected in order to establish a correct diagnosis, because portosystemic shunts carry a high risk of severe complications. Knowing that patients with portosystemic shunts may have pulmonary hypertension, respiratory complaints should be carefully evaluated—in this particular case, even though the most probable cause for the respiratory symptoms was pulmonary hypertension, it was ruled out by cardiac ultrasonography and further investigations confirmed the diagnosis of allergic asthma.

Abbreviations: angio-CT = computed tomography angiography, cm = centimeter, CT = computed tomography, i.v. = intravenous, sec = seconds, VRT = volume rendering technique, MIP = maximum intensity projection.

Keywords: case report, portosystemic shunt, pulmonary hypertension, vascular malformation

1. Introduction

Congenital portosystemic shunts that continue to exist after the neonatal period are estimated to have an incidence ranging from 1:30,000 to 1:50,000 live births.[1,2] This condition was described in 1973 by Abernethy, and subsequently other anomalies were reported to be associated with the portosystemic shunt.[1]

Congenital portosystemic shunt, also known as Abernethy malformation, is a rare vascular malformation in which the portal blood drains towards the systemic circulation eluding the liver.[1,3,4] These shunts arise through disturbances that intervene in the embryonic development of the splanchnic circulation, or when elements that are specific to the fetal circulation (especially those regarding the ductus venosus) persist beyond the intrauterine life.[1,3,5]

It is important to acknowledge congenital portosystemic shunts in order to closely follow-up these patients and identify any complications that might occur. In case of abdominal surgical interventions, recognizing the risk of hemorrhagic complications might improve management.

We present the case of an asymptomatic 9 years old boy evaluated for a complex vascular malformation which turned out to be Abernethy type Ib syndrome.

We obtained the written informed consent of the patient's legal guardians for publishing this case.

2. Case report

We present the case of an asymptomatic male patient who was first evaluated in our department at age 9. At age 2 the patient

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underwent for the first time a routine abdominal ultrasound in another medical facility and was diagnosed with a congenital malformation of the portal vein (portal cavernoma was considered). The ultrasound showed inhomogeneous liver with hyperechoic diffuse areas in both lobes with a reshuffled aspect, hyperechoic periportal spaces, consistent with periportal edema. The portal trunk and intrahepatic portal branches were not visualized. In the liver hylum multiple vascular Doppler signals were seen. Systolic velocity was of 70 cm/s and diastolic 22 cm/s in the hepatic artery. The resistive index was 0.70, value above normal, suggesting hepatic arterialization.

At that time (age 2) physical examination was normal. Laboratory tests performed at that point were all within normal limits—complete blood count, transaminases, bilirubin, protein level, renal function tests, coagulation tests. An esogastroduodenoscopy was also performed and the aspect was within normal limits.

Next, the patient was referred to the Surgical Department for further evaluation. He underwent a computed tomography (CT) which showed a hypodense image with a semilunar aspect, axial diameters of 2.1/1.9/3 cm, irregular contour, located in the IV's segment. After i.v. contrast administration the image appeared to be enhancing, with a multilocular aspect (comprised of vascular structures). Few other smaller hyperdense images (of vascular aspect) were seen perilesional. The portal trunk and rami were not visualized. Multiple collateral venous structures were seen in the splenic hylum and in the peripancreatic region. Homogeneous splenomegaly was also reported. No other abnormalities were seen. No surgical cure was pursued at the time.

When the patient was 6 years old, he started presenting chronic cough and recurrent wheezing. Laboratory tests displayed high immunoglobulin E levels and spirometry showed distal airway obstruction. At age 7 the diagnosis of allergic asthma was established and he was treated appropriately.

At age 9 the patient was reevaluated in our department and the abdominal ultrasound showed a short portal trunk which drained side-to-side into the inferior vena cava (Fig. 1). In the left liver lobe a nodular hyperechoic well defined lesion was seen, showing a hypoechoic central scar—suggestive for focal nodular hyperplasia (Fig. 1).

We proceeded to an abdominal angio-CT which confirmed the presence of a spleno-porto-mesenteric system draining into the inferior vena cava through a short extrahepatic portal trunk, below the origin of the hepatic veins. The aspect was consistent with Abernethy type IB syndrome (Fig. 2).

In addition, other vascular malformations were found, but none with clinical significance. The hepatic veins were forming a short common trunk before draining into the inferior vena cava. The hepatic artery was emerging from the superior mesenteric artery with early division into 2 main branches, 1 for each hepatic lobe (Fig. 3). The celiac trunk had only 2 branches (the splenic artery and the left gastric artery). There was a supranumerary right renal artery emerging from the aorta, tributary to the upper renal pole.

Considering the complex vascular malformation, we performed an echocardiogarphy which showed persistent left superior vena cava. No pulmonary hypertension was reported.

The final diagnosis of Abernethy syndrome type Ib was confirmed.

During the following year no treatment was pursued and the evolution was uneventful—the patient remained asymptomatic.

3. Discussion

From an anatomic point of view, portosystemic shunts are divided into intrahepatic shunts and extrahepatic shunts.[1,3–5] For the intrahepatic ones the shunt originates from the portal branches, after the portal bifurcation. For the extrahepatic ones the shunt connects the portal trunk or its tributaries to a vessel belonging to the systemic circulation.[4] Type I portosystemic shunts are characterized by the complete absence of the portal vein, the portal blood draining entirely into the inferior vena cava.[5] Type I is further divided into IA—for which the splenic vein and the mesenteric vein separately drain into the inferior vena cava, and IB for which the portal vein itself drains into the inferior vena cava.[4] For type II portosystemic shunts the portal vein is unharmed, but there is an extrahepatic connection between the portal vein and the inferior vena cava.[5]

Reviewing the literature, the malformation we identified in this case should be classified as Abernethy type IB syndrome[4]—the patient has a short portal trunk which drains into the inferior vena cava in a side-to-side manner.

In the embryonic life there is a communication between the portal vein and the inferior vena cava.[6] The Abernethy

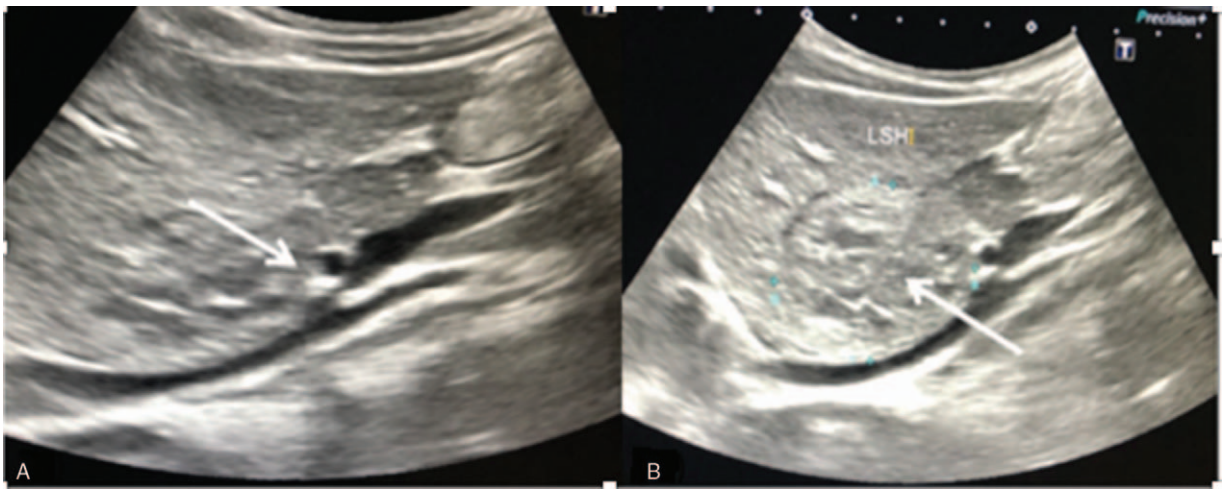

Figure 1. (A) Abdominal ultrasound—short portal trunk, draining side-to-side into the inferior vena cava (arrow). (B) Abdominal ultrasound—left liver lobe with nodular image—focal nodular hyperplasia (arrow).

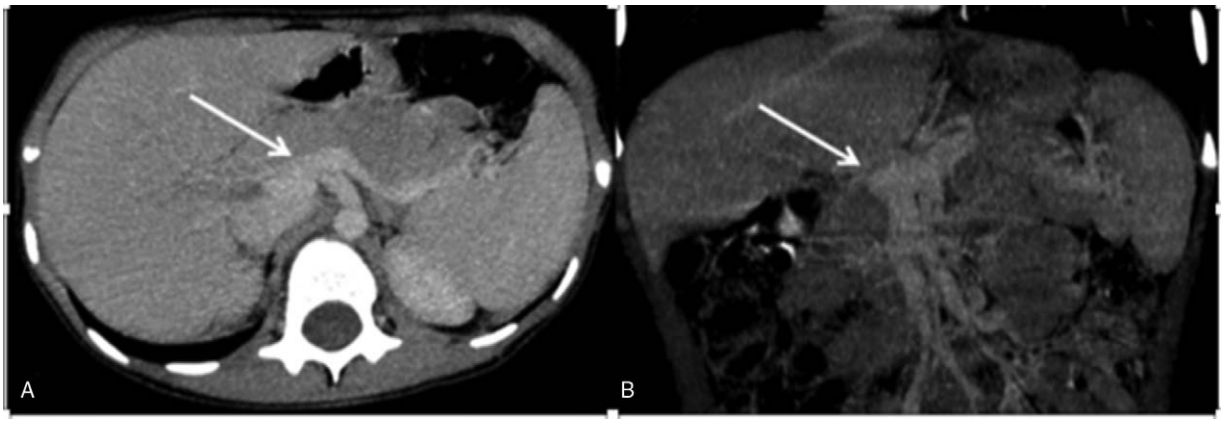

Figure 2. (A) Axial view; (B) coronal view. Short portal trunk draining into the inferior vena cava (arrow).

malformation is secondary to an abnormal embryonic development, an aberrant development or involution of the fetal circulatory system.[1]

The hepatic primordium arises during the 4th week of intrauterine life.[7] There are 3 major vascular systems of the embryonic circulation—the vitellin veins, the cardinal veins, and the umbilical veins.[1,7] The cardinal and the vitellin veins will subsequently form the systemic and the portal circulation, while the umbilical veins will involute.[1]

The liver has 2 distinct afferent vascular systems during the embryonic life:

- the portal vein together with its rami form the functional circulation of the liver (blood that originates from the splanchnic vessels—intestines, pancreas, and spleen) which transports about 75% of the blood that passes through the liver;
- the hepatic artery together with its rami form the nutritional circulation of the liver—arterial blood.[7]

The efferent vascular system of the liver is represented by the suprahepatic veins. Afferent and efferent vessels are connected through an intrahepatic capillary system.[7]

The embryonic development of the portal system is set between the 4th and the 10th week of intrauterine life.[4,7] The vitelline vessels emerge from the Yolk sack. In the 4th week of intrauterine life, cross-communicating channels between the 2 vitelline vessels (left and right) develop, forming a periduodenal loop (vitelline venous network).[1,4] The selective involution of these shunts leads to formation of the final configuration of the portal system.[1,4,8,9] The abnormal involution of this periintestinal loop leads to forming a malformation known as portal vein duplication. The portal vein may be completely absent if the periintestinal loop excessively involutes.[4] Incomplete involution of this vitelin vein system secondary to the development of the hepatic sinusoids is the main cause of portosystemic shunts, and the site of the shunt depends on the anatomical site (right or left) and level (proximal or distal) where these veins fail to differentiate.[1,10]

The persistence of the right vitelline vein leads to the formation of an intrahepatic or extrahepatic side-to-side shunt, while the persistence of the left vitelline vein causes the formation of other extrahepatic shunts, draining into the inferior vena cava above the confluence of the suprahepatic veins or directly into the right atrium.[1,10]

The clinical presentation is reported to be variable, particularly in the pediatric population, ranging from asymptomatic to patients presenting symptoms related to different organs and

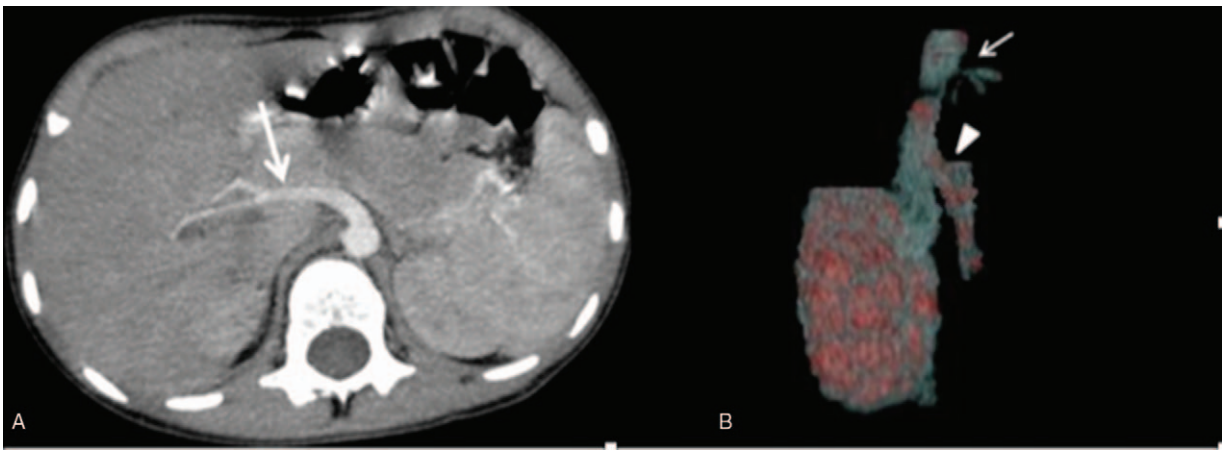

Figure 3. (A) Axial MIP—hepatic artery emerging from the superior mesenteric artery, with early division (arrow); (B) Oblic VRT—arrow—hepatic veins forming a short common trunk before draining into the inferior vena cava (arrow head); short portal trunk draining into the inferior vena cava (arrow). MIP = maximum intensity projection, VRT = volume rendering technique.

systems—liver, respiratory system, central nervous system.[3] Characteristically, Abernethy syndrome patients have hypoxia and hepatopulmonary syndrome.[5]

Our patient was initially evaluated in another medical facility only by abdominal ultrasonography because abdominal angio-CT was not available, as administration of contrast medium injection was not possible, which misled the diagnosis. Abdominal angio-CT is essential in diagnosing this type of malformations.

The abdominal angio-CT, considered the first choice examination,[3] which was performed in our department, showed no evidence of intrahepatic portal vein rami. In spite of the fact that the intestinal blood bypassed the liver and reached the systemic circulation through the shunt, which should have resulted in numerous complications (encephalopathy, pulmonary hypertension),[1,11] the patient was asymptomatic at all times, hence no treatment was performed, but he was followed-up closely in order to promptly identify any change in his status. In young patients with congenital portosystemic shunts hyperammonemia without encephalopathy was reported. As possible explanations decreased sensitivity of the young brain to the effects of ammonia was proposed.[12]

The presented patient was identified to have a nodular image in the left liver lobe, which was not biopsied, but imaging aspect was suggestive for focal nodular hyperplasia. Nodular hepatic lesions have been reported in patients with Abernethy malformation, most frequently benign. They appear to be caused by the increase of the arterial blood flow through the hepatic artery in context of the absence of the portal blood flow. This circulatory malformation provokes the abnormal development and regeneration of the liver, hence the hepatic lesions.[13,14] From a histologic point of view, focal nodular hyperplasia, regenereative nodular hyperplasia, hepatic adenoma, hepatoblastoma, and hepatocellular carcinoma have all been reported to be associated with the Abernethy syndrome.[15]

Portosystemic shunts may result in complications such as portal hypertension, liver failure, encephalopathy (secondary to hyper-ammonaemia), development of hepatocellular tumors (either benign or malignant), hepatopulmonary syndrome, and pulmonary hypertension.[16,17] Asymptomatic presentation, as in the presented case, is rare. Acknowledgment of these shunts is important before abdominal surgical interventions because there is a high risk of catastrophic bleeding when encountering unknown vascular malformations, possibly related to other anatomical malformations.[17]

Treatment options depend on the shunt type and include conservative management or closure of the shunt—either transcatheter or surgical closure after balloon occlusion test, which helps to decide whether there should be a 1-step or 2-steps closure of the shunt (small portal radicals, usually not visualized on ultrasonography, are allowed to enlarge by the 2-step procedure). In some cases liver transplantation may be needed.[12,18]

In our patient, given that he is persistently asymptomatic, we consider the conservative treatment to be the best choice.

4. Conclusion

Abdominal ultrasound is usually the first examination performed that may detect a vascular malformation, but all lesions that have a vascular aspect should be further evaluated by angio-CT for a precise diagnosis. Early correct diagnosis is important for appropriate management, because portosystemic shunts carry a high risk of severe complications. Pulmonary hypertension should always be considered as a cause of respiratory symptoms in cases of complex vascular malformations.

Author contributions

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References

[1] Papamichail M, Pizanias M, Heaton N. Congenital portosystemic venous shunt. Eur J Pediatr 2018;177:185–294.
[2] Bernard O, Franchi-Abella S, Branchereau S, et al. Congenital portosystemic shunts in children: recognition, evaluation, and management. Semin Liver Dis 2012;32:273–87.
[3] Timpanaro T, Passanisi S, Sauna A, et al. Congenital portosystemic shunt: our experience. Case Rep Pediatr 2015;2015:691618.
[4] Ghuman SS, Gupta S, Buxi TB. The Abernethy malformation-myriad imaging manifestations of a single entity. Indian J Radiol Imaging 2016;26:364–72.
[5] Benedict M, Rodriguez-Davalos M, Emre S, et al. Congenital extrahepatic portosystemic shunt (Abernethy Malformation Type Ib) with associated hepatocellular carcinoma: case report and literature review. Pediatr Dev Pathol 2017;20:354–62.
[6] Loomba RS, Frommelt M, Moe D, et al. Agenesis of the venous duct: two cases of extrahepatic drainage of the umbilical vein and extrahepatic portosystemic shunt with a review of the literature. Cardiol Young 2015;25:208–17.
[7] Collardeau-Frachon S, Scoazec JY. Vascular development and differentiation during human liver organogenesis. Anat Rec (Hoboken) 2008;29:614–27.
[8] Guérin F, Blanc T, Gauthier F, et al. Congenital portosystemic vascular malformations. Semin Pediatr Surg 2012;21:233–44.
[9] Tanoue S, Kiyosue H, Komatsu E, et al. Symptomatic intrahepatic portosystemic venous shunt: embolization with an alternative approach. AJR Am J Roentgenol 2003;181:71–8.
[10] Stringer MD. The clinical anatomy of congenital portosystemic venous shunts. Clin Anat 2008;21:147–57.
[11] Matsuura T, Takahashi Y, Yanagi Y, et al. Surgical strategy according to the anatomical types of congenital portosystemic shunts in children. J Pediatr Surg 2016;51:2099–104.
[12] Pathak A, Agarwal N, Mandliya J, et al. Abernethy malformation: a case report. BMC Pediatr 2012;12:57.
[13] Hao Y, Hong X, Zhao X. Congenital absence of the portal vein associated with focal nodular hyperplasia of the liver and congenital heart disease (Abernethy malformation): a case report and literature review. Oncol Lett 2015;9:695–700.
[14] Banz V, Olliff S, Taniere P, et al. Liver tumours in patients with Abernethy malformation. ANZ J Surg 2011;81:640–1.
[15] Yu HC, Kim JH, Murakami G, et al. An anomalous portal vein crossing the lesser sac and ending at the upper part of ductus venosus. Anat Cell Biol 2015;48:218–21.
[16] Malgras B, Guerin F, Dokmak S. Hepatocellular carcinoma in a patient with congenital porto-systemic shunt. J Visc Surg 2017;154:217–9.
[17] Mohtashami A, Kiat A, Cross J, et al. Catastrophic intraoperative bleeding due to congenital extrahepatic porto-systemic shunt anomaly: a surgical case report of two rare anomalies. Int J Surg Case Report 2018;44:161–5.
[18] Sharma S, Bobhate PR, Sable S, et al. Abernethy malformation: single-center experience from India with review of literature. Indian J Gastroenterol 2018;37:359–64.