Case Report

A novel case of congenital hepatic arterio-veno-portal shunts with umbilical vein aneurysm

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ABSTRACT

A term male infant presented with congenital hepatic arterio-veno-portal shunts. A mass-like lesion in the left lobe of the liver received blood supply from not only the umbilical vein, but also the hepatic and inferior intrahepatic arteries, communicating with the hepatic and portal veins in a complicated manner, with an umbilical vein aneurysm. The blood flow of the arterio-veno-portal shunts spontaneously and gradually declined from the neonatal period to six years of age. Although mild high-output cardiac failure had developed, no life-threatening events or health problems originating from portosystemic shunts, such as pulmonary artery hypertension and hepatopulmonary syndrome, were observed. However, this report shows that scrupulous follow-up to identify pulmonary artery hypertension and hepatopulmonary syndrome should be continued because complete resolution of the arterio-veno-portal shunts was not obtained in this case.

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Introduction

Congenital hepatic vascular shunts are generally classified into the following types: arteriovenous (AV), arterioportal (AP), portosystemic (PS), and mixed [1]. Classification is clinically important because different types of vascular shunts cause different clinical manifestations. In general, portosystemic shunts do not cause any acute signs or symptoms during the neonatal period. However, AV and AP shunts may cause high-output cardiac failure, persistent pulmonary arterial hypertension, respiratory distress, and consumptive coagulopathy in the acute stage, and may require catheter intervention in the neonatal period [2,3]. Thus, pediatricians should be aware of the possibility that these conditions can be life-threatening. The mixed type consists of at least two types of communications: AV, AP, and PS shunts. However, the involvement of all three vascular systems creates arterio-veno-portal (AVP) shunts. Here, we report a novel case of congenital hepatic AVP shunts and a prenatal umbilical vein aneurysm, with particular emphasis on the clinical and imaging findings.

Case presentation

A 34-year-old woman was pregnant and referred to our hospital with fetal cardiomegaly in the 22 week of gestation. No signs of cardiac failure were observed on fetal echocardiography. Magnetic resonance imaging (MRI) in the 37 week of gestation showed a huge aneurysmal vascular dilatation, 5 cm in diameter, along the round ligament of the liver (Fig. 1). The lesion was supplied from a thick tortuous umbilical vein and drained into the ductus venosus, suggesting an umbilical vein aneurysm. No arterial blood supply was detected.

A term male infant with a birth weight of 3.0 kg was delivered vaginally. Apgar scores were 8 and 9 at 1 min and 5 min, respectively. Physical examination revealed no abnormal findings such as cardiac murmur, subcutaneous edema, cutaneous lesions, or hepatomegaly. Mild cardiomegaly was observed on chest radiography (Fig. 2), although echocardiography showed normal cardiac function without any cardiac anomalies. Abdominal ultrasound (US) performed immediately after birth showed a low echoic mass-like lesion in the liver with intralesional massive turbulent vascular flow (Fig. 3). The lesion received blood supply from not only the umbilical vein but also the hepatic and inferior epigastric arteries, and communicated with the portal and hepatic veins. The intrahepatic portal veins were well developed and showed hepatoportal blood flow. Abdominal computed tomography (CT) with contrast enhancement performed on the first day of life confirmed the US findings (Fig. 4). Based on the fetal MRI and postnatal US and CT findings, a diagnosis of multiple hepatic AVP shunts with an umbilical vein aneurysm was made.

An umbilical vein catheter was placed immediately after birth in case of an emergency intervention. On the sixth day of age, blood sampling and pressure measuring from the umbilical vein catheter was performed. As a result, intralesional oxygen partial pressure of the blood sample was 60 mmHg on room air breathing, indicative of the presence of arterial blood supply. Additionally, an intracatheter mean blood pressure of 4 mmHg suggested favorable venous drainage without congestion. Oral diuretics were administered for mild high-

Fig. 1 – Magnetic resonance imaging of the fetus (at 37 weeks of gestation)
Balanced field echo T2-weighted image in the coronal section shows a round mass-like lesion (arrowhead) in the liver (A) and a dilated umbilical vein (arrowhead) along the round ligament of the liver in the sagittal section (B)
output cardiac failure due to hepatic AV shunting. In contrast to the significant imaging findings, the clinical course was uneventful. Laboratory findings showed transient abnormalities: mild elevation of serum galactose (4.6 mg/dL) and bile acid (62 μmol/L) at the first week of age, which decreased to the normal range at 1 and 6 months of age, respectively, without any medication. Hyperammonemia was not observed during the course. The patient was discharged from the hospital at 1 month of age, without any significant clinical complications.

Since the imaging abnormalities persisted throughout infancy, close observation was conducted based on the clinical signs and symptoms, as well as laboratory and imaging examinations. Diuretics were discontinued at the age of 4 months. Oxygen saturation on pulse oximetry (SpO₂) was checked every 3 to 4 months to detect hypoxemia as an initial manifestation of pulmonary artery endothelial damage and subsequent pulmonary AV fistula caused by a hepatic PS shunt. The value of SpO₂ was always >95% on room air breathing. Lung perfusion scintigraphy using 99m technetium-macroaggregated albumin (99mTc-MAA) performed at 0 months and 1 year of age revealed the absence of a pulmonary AV shunt. Abdominal US and CT showed a gradual regression in the size of the aneurysmal and tortuously dilated shunt vessels, as well as early abnormal filling of contrast media in the portal and hepatic veins in the arterial phase at 1 year of age (Fig. 5). Even when CT showed an abnormal intravascular contrast filling indicative of a PS shunt, sequential transrectal portal scintigraphy using 99mTc-pertechnetate consistently showed a PS shunt ratio of less than 20%. Abdominal CT at 5 years of age revealed a marked regression of the vascular abnormalities, whereas hepatic parenchyma resulted in significant atrophy of the lateral segments of the left lobe (Fig. 5).

During the 6 years of follow-up, the patient had consistently remained healthy without any hepatic, cardiac, or respiratory problems. At 5 years of age, the total developmental score of the Wechsler Intelligence Scale for Children-Fourth (WISC-IV) was 87 (within normal range). Brain MRI performed at 1 and 5 years of age did not show any abnormalities, including manganese deposition in the basal ganglia.

Discussion

A mixed type of congenital hepatic vascular shunt is defined as comprising at least two types of abnormal vascular communications (AV, AP, and PS) and is considered to be uncommon. The vascular abnormalities observed in our patient were rare because the abnormality is multiple, complex, and involves all three hepatic vascular systems (arterial, venous, and portal).

The mechanism underlying the development of these complex vascular abnormalities remains unclear. However, there is an imaging clue to its pathophysiologic mechanism: an umbilical vein aneurysm. An umbilical vein aneurysm, also known as an intra-abdominal umbilical vein varix, is a rare vascular abnormality of the fetus and is considered to be a developmental disease rather than a congenital malformation [4]. It is commonly observed in fetuses with multiple congenital anomalies, such as trisomy 21, congestive heart failure, and vascular abnormalities, such as prenatal agenesis of the ductus venosus [4,5]. In our patient, an umbilical vein aneurysm was detected in the fetal period, but there was no evidence of multiple congenital anomalies, other systemic diseases, or agenesis of the ductus venosus on fetal US and MRI. However, cardiomegaly developed immediately after birth, and the postnatal echocardiographic examination revealed high-output cardiac failure without any cardiac malformations. Therefore, the most probable cause of the umbilical vein aneurysm may be the prenatal development of a hepatic AP (AV) shunt and subsequent congestive heart failure.

It remains uncertain whether complex AVP shunts developed simultaneously at that time or were associated with perinatal hemodynamic conversion from fetal circulation to neonatal circulation.

Another uncertainty is the mechanism of spontaneous regression of vascular abnormalities without any particular treatment. In general, hepatic AV and AP shunts are categorized as fast-flow shunts [6], and cause high-output cardiac failure at an early age (mean 2.2 months) in 58% of the affected infants [7]. Currently, congenital hepatic AV and AP shunts

Fig. 2 – Chest radiograph immediately after birth
Mild cardiomegaly (cardiothoracic ratio = 55%) without definitive pulmonary hypervascularity. An umbilical vein catheter has been placed
are believed to not regress spontaneously [8-10]. In contrast, spontaneous regression has been described in congenital hepatic slow-flow PS shunts at the age of 2 years [6]. There is no literature describing the spontaneous regression of congenital hepatic vascular shunts of the mixed type. Agha et al. described the spontaneous regression of a large congenital hepatic AV malformation [11]. They speculated that the reduction of shunt flow accompanied by treatment with diuretics and captopril might have contributed to the spontaneous regression of the lesion. In this article, they referred to their experience of spontaneous involution of a vein of Galen aneurysmal malformation in succession to thrombus formation. These descriptions led us to hypothesize that not only a hepatic slow-flow shunt, but also an AV and AP shunt may spontaneously regress when intralesional blood flow is significantly low in velocity.

In our patient, although the vessels appeared thick on US and CT, we speculate that the AP shunt was mild and the intralesional blood flow was slow. This speculation is supported by two clinical evidences: one is that high-output cardiac failure remained mild throughout the clinical course, and the other is that the direct measurement of the intracatheter mean blood pressure of an umbilical vein catheter was low (4 mmHg).

Various types of complications are described as hepatic vascular shunts, exemplified by portal vein hypoplasia/atrophy, hepatic parenchymal abnormalities (cirrhosis, hyperplastic nodules, atrophy, etc.), pulmonary AV fistula to PS shunt, patent ductus venosus, portal hypertension, mesenteric congestion to AP shunt, high-output cardiac failure, consumptive coagulopathy, and hydrops fetalis to AV shunt [12]. The clinical course of our patient is particularly unique because no complications were observed or remained milder than those in patients with a simple AV, AP, or PS shunt.

We speculate that the degree of arterial blood supply plays an important role in the unique clinical presentation of the patient. First, the presence of arterial involvement in the fetal period was the main cause of the significant dilatation of the shunt vessels. Second, the “appropriate” degree of AV and AP shunts caused only mild cardiac failure but no other serious complications. The appropriateness also aided hepatoportal portal flow and subsequent development of the portal vein, and prevented the most serious complications associated with portal vein hypoplasia/atrophy. Third, a small amount of arterial fast flow allowed spontaneous regression of the vascular shunts. In addition, focal atrophy of the lateral segments of the left lobe as a late sequela may also be associated with hemodynamic changes in arterial blood flow. The above-mentioned appropriate portal flow might occur only in the right lobe, but not in the left lobe, which results in focal atrophy in the left lobe.

The most important issues for clinical healthcare workers are the assessment and management of patients with hepatic AV shunts. In the neonatal period, the most significant concern is the development of cardiac and hepatic failure. In cases with severe AP and/or AV shunts, emergency catheter intervention is the most reliable and least invasive treatment of choice. Therefore, we inserted an umbilical vein catheter immediately after birth, through which emergency transcatheter intervention (arterial embolization) could be performed promptly. Fortunately, the catheter was not necessary in our patient because the high-output cardiac failure remained mild. For the management of cardiac failure, we gave our patient oral diuretics. Inotropic agents may also be useful for severe cases. This medication could be discontinued at a few months of age after the radiologic improvement of cardiomegaly. Careful monitoring of serum or filter paper blood galactose and ammonia is recommended because they reflect...
the severity of the PS shunt and overall hepatic insufficiency, respectively [13,14]. In our patient, the filter paper blood galactose level had normalized at 1 month of age; hyperammonemia did not develop throughout the course.

After the acute phase, accurate estimation of the severity of the PS shunt and early detection of pulmonary arterial hypertension (PAH) and hepatopulmonary syndrome (HPS) are important [15]. Transrectal scintigraphy is the most reliable examination for assessing the PS shunt ratio. PS shunt ratios of more than 60% generally require shunt closure to avoid encephalopathy and liver dysfunction, and PS shunt ratios of 30% to 60% are at increased risk of those illnesses [4]. We performed the examination approximately once a year, which revealed only a minimal abnormality of 10% to 20% of the PS shunt ratio (almost equivalent to background accumulation). PAH and HPS are the most severe complications of PS shunts and can be life-threatening. The mean age of onset of PAH and HPS due to congenital PS shunts was reported to be 4 to 5 years of age; however, both could be observed during the neonatal and early infantile periods [15]. In addition, the shunt vessel size did not correlate with the emergence of PAH [16,17]. The mortality rate of PAH caused by congenital PS shunts is thought to be approximately 40% to 50% [15,16]. Taken together, scrupulous follow-up for PAH and HPS screening is essential. Several methods are considered clinically useful for the early detection of PAH and HPS. One of the simplest methods for PAH screening is the measurement of SpO₂ using a pulse oximeter. Echocardiography is a reliable and less invasive modality for the early detection of PAH and should play a major role in clinical practice. Lung perfusion scintigraphy using 99mTc-MAA is the modality of choice for the assessment of HPS. Extrapulmonary accumulation (brain, kidney, etc.) is always an abnormal finding and suggests the presence of pulmonary AV shunt/fistula. In our patient, serial assessment including the measurement of SpO₂ (at every hospital visit), echocardiography (1-2 times a year), and scintigraphy (twice/5 years) did not show any significant abnormality.

In the chronic stage, hepatic shunts have a potential risk of vascular insult and encephalopathy. Fortunately, our patient showed favorable psychomotor development, which was sup-

Fig. 4 – Dynamic contrast-enhanced CT of the liver in the neonatal period
Dynamic CT of the arterial phase demonstrates multiple significantly dilated tortuous hepatic vessels involving the hepatic artery, hepatic vein, and portal vein, suggestive of arterio-veno-portal (AVP) shunting

Fig. 5 – Contrast-enhanced CT of the liver at 1 year (A) and 5 years of age (B)
Multiple dilated AVP shunts almost disappeared, except for a residual aneurysmal dilatation in the left lobe. Aneurysmal dilatation has decreased in size (arrowhead). Focal atrophy in the lateral segments of the left lobe (B)
ported by the normal result of WISC-IV (at 6 years of age) and the absence of manganese deposition in the basal ganglia on brain MRI (at 5 years of age). However, further careful observation is recommended because the minimal shunt remains at 6 years of age.

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