Congenital absence of portal vein in the fetus: a case report

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ABSTRACT
Congenital absence of portal vein (CAPV) is a rare abnormality, which may be associated with other abnormalities. We report a case of prenatal diagnosis of absent portal vein confirmed on postnatal ultrasonography and computed tomography scan. The ultrasound features of CAPV include dilated intra-abdominal segment of umbilical vein, dilated inferior vena cava and the presence of hyperechogenic areas in the liver. Blood coming from the umbilical vein directly drains into the inferior vena cava. The dilated intra-abdominal segment of the umbilical vein shows high velocity pulsatile flow, resembling that of ductus venosus. These findings should prompt a careful search for the portal vein and any associated anomalies.

INTRODUCTION
Congenital absence of portal vein (CAPV) is an extremely rare abnormality and is usually discovered accidentally following investigation for metabolic disturbance or intra-abdominal tumors in children or adults1–3. We report a case of the prenatal finding of CAPV associated with dilated intra-abdominal portion of umbilical vein (UV) and dilated inferior vena cava (IVC).

CASE REPORT
A 20-year-old primigravida, with no significant previous medical history, was referred to the Fetal Medicine unit at St Mary’s Hospital, with the provisional finding of a dilated IVC at a routine 20-week scan. Ultrasound dating of pregnancy was consistent with menstrual dates and the fetal abdominal circumference was on the 50th centile for gestation. Ultrasound examination showed normal cardiac anatomy: four chambers including interatrial and interventricular septa, great vessels and cardiac connections, arterial duct, aortic arch and superior vena cava. The pulmonary veins drained normally into the left atrium. The UV appeared normal up to the point of cord insertion into the abdomen and demonstrated a normal monophasic waveform (Figure 1). Shortly after entering the abdomen, the UV showed a biphasic pattern with forward flow throughout diastole, a Doppler signal compatible with that of ductus venosus (DV) (Figure 2). From this point, the UV was dilated and joined the IVC without any clear-cut demarcation between the UV and the DV.
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Figure 3 Cross section of the fetal abdomen (at 26 weeks' gestation) at the level of stomach showing the dilated intra-abdominal segment of the umbilical vein (shown as 'x'). Note the absence of normal portal sinus. ANT, anterior; ST, stomach; AO, cross section of the abdominal aorta; IVC, cross section of the inferior vena cava; SP, spine.

Figure 4 Oblique section of the fetal abdomen (at 26 weeks' gestation) and chest showing the dilated umbilical vein (UV) joining the IVC directly.

Figure 5 Parasagittal section of the fetal abdomen (26 weeks' gestation) and chest showing dilatation of the inferior vena cava (IVC) distal to its junction with the dilated umbilical vein (UV) (shown as ↑). RA, Right atrium; LA, left atrium; DAO, descending aorta.

Figure 6 Schematic representation of the in utero appearance of absent portal vein showing dilated umbilical vein with a straight on course to a dilated inferior vena cava (IVC) across the liver, which shows echodense areas (secondary to calcification from ischemia).

Figure 7 Cross section of the fetal abdomen (26 weeks' gestation) demonstrating hyperechogenic areas in the liver.

(Figures 3–6). This long dilated vessel had a biphasic Doppler waveform with forward flow throughout diastole along its entire length. The IVC distal to its junction with this dilated vessel was also dilated, but had a normal triphasic flow velocity waveform with reverse flow during atrial contraction. The proximal portion of IVC appeared normal (Figures 5 and 6). No portal system was visualized in the abdomen (Figure 3). The UV was medial to the gall bladder.

Doppler signals obtained from the uterine artery, umbilical artery and fetal peripheral vessels (middle cerebral and thoracic aorta) were normal. The remainder of the fetus appeared normal and there were no markers of aneuploidy. Prenatal diagnosis to determine fetal karyotype was offered, but declined. Maternal blood for infection screen (toxoplasmosis, cytomegalovirus and parvovirus) was negative. A subsequent sonogram at 24 weeks' gestation showed multiple echogenic areas scattered throughout the liver.

Serial scans were performed at regular intervals. Late in the third trimester, the following additional features were noted: dilatation of the right atrium (no significant ventricular disproportion), widespread hyperechogenic areas in the liver (Figure 7), especially the left lobe of liver and enlarged spleen. There was a fall off in the growth velocity of fetal abdominal circumference, but with no other features of intrauterine
growth restriction (normal amniotic fluid volume, umbilical artery and fetal middle cerebral artery Doppler). There was no evidence of fetal hydrops.

There were no maternal complications during pregnancy. Labor was induced at 41 weeks of gestation by amniotomy followed by oxytocin infusion. The intrapartum period was uneventful and resulted in a spontaneous vaginal delivery of a live male infant weighing 2990 g with normal Apgar score (9 at 1 min and 10 at 5 min), normal cord blood gases (pH of 7.29 and base deficit of –4.6) and normal cord glucose.

Examination of the newborn revealed mild microcephaly in relation to weight, but no dysmorphic features. There was moderate hepatosplenomegaly. Shortly after admission the neonate was hypoglycemic, requiring dextrose and glucagon infusions to maintain blood glucose levels. In addition, there was derangement of extrinsic coagulation with an INR of 2.7 (normal range 0.6–1.4) and a prothrombin time of 32 s (normal range 18–25 s). Fresh frozen plasma and additional vitamin K were given to correct the coagulation parameters. No other abnormalities in liver function test were noted in the immediate neonatal period, ALT being 15 U/L and serum ammonia 49 µmol/L. The karyotype was normal. The infant was transferred to a ‘tertiary liver unit’. There was an unexplained upper gastrointestinal hemorrhage, which was treated conservatively. Over the week the neonate’s condition was stabilized and he was discharged after the formula feeds were established.

Postnatal echocardiography was normal. Cranial ultrasound demonstrated bilateral small choroid cysts with no other abnormality. Abdominal ultrasound confirmed absence of intrahepatic portal venous system. The left and middle hepatic veins were attenuated, but appeared to drain directly into the IVC. The right hepatic vein drained retrogradely via the long dilated aberrant vessel, thought to be the patent DV, into the IVC (Figures 8 and 9). The hepatic artery was large and hyperdynamic. The splenic and superior mesenteric veins were patent with normal anatomy, although the precise point of drainage could not be identified on ultrasound examination. The liver showed widespread hyperechogenic areas, thought to be due to intrahepatic calcification. A subsequent computed tomography scan of the abdomen confirmed ultrasound findings and showed that the splenic and superior mesenteric veins drained into the IVC via the patent ductus venous.

At the 6-month review, the liver function tests were mildly deranged but stable. The infant was thriving with normal milestones. Curiously he has developed cutaneous hemangiomata. At the postnatal age of 12 months, developmental milestones were normal, excepting slight delay in speech thought to be secondary to understimulation because of mother’s deprived socioeconomic status. The liver function tests have normalized although the splenic venous pressure remains elevated. The head circumference currently runs along the 5th centile while the length and weight are on the 25th to 50th centile. The ultrasound and magnetic resonance imaging (MRI) of the brain are normal.

**DISCUSSION**

The portal vein perfuses the liver, and in its absence the splenic and superior mesenteric vein (SMV) do not unite but establish direct communication with systemic circulation, by-passing the liver. Most commonly, this results in the SMV joining either the infrahepatic IVC (or rarely the suprahepatic) or the left renal vein. Occasionally splenic and SMV form a confluence but there is no connection between the mesenteric circulation and the liver. As the portal vein develops in close proximity to the developing IVC, the finding of caval abnormalities in conjunction with CAPV is not unexpected.

In the present case, CAPV in the fetus was associated with a dilated IVC, UV and DV. The SMV and the splenic vein (which normally unite to form the portal vein) drained into the IVC via the dilated aberrant vessel in the abdomen (thought to be the patent ductus venosus).

There are very few reports of prenatal diagnosis of abnormalities of the intra-abdominal venous system. Moore et al. report a case of prenatally diagnosed extrahepatic insertion...
of the UV in association with absent intrahepatic portal and umbilical vein. IVC and the right iliac vein (into which the UV drained) were both dilated. Reviewing the literature, the authors categorize the abnormalities of intra-abdominal UV into three groups: (i) persistent right umbilical vein, (ii) absent ductus venosus with extrathoracic insertion of the umbilical vein, and (iii) abnormally dilated umbilical vein, but with normal course.

Persistent right UV can be recognized by ultrasound (in the section used to measure the abdominal circumference) and should be excluded as it is associated with numerous and occasionally lethal malformations. The intrahepatic portion of the UV is lateral to the gall bladder (instead of being medial), the portal vein curves toward the stomach (instead of parallel to stomach) and the UV connects abnormally with the right portal vein instead of the left portal vein.

A clinically useful classification of the abnormalities of intra-abdominal fetal venous system has been proposed by Hofstaetter et al. in a recent report of 16 cases. Two important subgroups with very different outcome were identified. The group with abnormal course of caval veins had a very high (six out of eight fetuses) proportion of major cardiac defects and demonstrate other anomalies. These include cardiac malformations (ventricular septal defect, atrial septal defect, dextrocardia, patent foramen ovale, patent ductus arteriosus), IVC anomalies such asazygos or hemiazygos and left IVC, skeletal abnormalities such as hemivertebra and Goldenhar syndrome, and hepatic tumors and Superina reported on two cases of CAPV and proposed a classification system for portasystemic vascular anomalies.

Two types were recognized based on whether liver was perfused with portal blood (partial shunt, type II) or not (total shunt, type I). CAPV is an example of type I shunt, and this is further classified into a or b depending on whether the two tributaries of portal vein, superior mesenteric vein and splenic vein join to form a confluence (b) or do not (a). Reviewing the literature the authors found that with type I shunts, which are clearly congenital, the patients are typically young at the time of presentation, usually with little or no encephalopathy, despite high serum ammonia levels, and demonstrate other anomalies. These include cardiac malformations (ventricular septal defect, atrial septal defect, dextrocardia, patent foramen ovale, patent ductus arteriosus), IVC anomalies such asazygos or hemiazygos and left IVC, skeletal abnormalities such as hemivertebra and Goldenhar syndrome, and hepatic tumors.

Curiously, this infant developed cutaneous hemangiomata that persisted despite the liver function tests reverting to normal. At 12 months of age, the neonatal head circumference was growing along the 5th centile, although his length and weight were on the 25th to 50th centile. Essentially, we consider the small head size as a variant of normal, especially in view of the fact that the ultrasound and MRI of the brain were normal. However, a new syndrome of cutaneous hemangiomata in association with small head and absent portal vein is a possibility.

The present case has some similarities to the one reported by Laverdier et al., namely prenatal finding of dilated IVC and the aberrant vessel (thought to be ductus venosus) along with intra-abdominal calcifications and the postnatal ultrasound showing absence of portal vein. In this case, the SMV drained into dilated IVC via the dilated left renal vein. We propose that the prenatal finding of dilated IVC, dilated intra-abdominal segment of the umbilical vein and hyperechoic areas in the liver are ultrasound features of an absent portal vein, and this observation should prompt a search for the intra-abdominal portal vein and associated anomalies.

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