Congenital hepatic arteriovenous malformation with persistent pulmonary hypertension: an unusual presentation

Congenital hepatic arteriovenous malformations (HAVMs) are rare anomalies, which typically present in infancy with congestive heart failure, anaemia and hepatomegaly. HAVM presenting with persistent pulmonary hypertension of the newborn is extremely rare and associated with high mortality. This case study reports a unique premature infant who developed persistent pulmonary hypertension of the newborn (PPHN) soon after birth. Congenital HAVM was diagnosed by neonatologist-performed echocardiography during evaluation of PPHN and the infant was treated promptly, after which the pulmonary hypertension resolved with excellent outcome.

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Keywords

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Key points

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1. Congenital HAVM presenting with PPHN is rare and carries a poor prognosis – early diagnosis and prompt treatment may improve outcome.
2. Nitric oxide (NO) is the treatment of choice in PPHN. Congenital AVMs presenting with PPHN secondary to congestive heart failure need diuretic therapy and definitive treatment rather than NO.

Congenital hepatic arteriovenous malformation (HAVM) presenting with persistent pulmonary hypertension of the newborn (PPHN) is a rare anomaly and only three cases have been reported in the literature. They usually present with congestive heart failure, anaemia and hepatomegaly in infancy.[1] HAVM presenting with PPHN is reported to have a high mortality. Two of the three reported cases died during infancy and the surviving term infant had an early diagnosis and coil embolisation. To the best of our knowledge none of the reported cases were born prematurely.[2] We report the first case of a preterm infant presenting with PPHN who had a prompt diagnosis and early intervention for an extensive congenital HAVM, with excellent outcome.

Case presentation

A 32-week gestation male infant with a birth weight of 1.87kg was delivered by elective caesarean section for worsening Doppler and absent end diastolic flow. Cardiomegaly and increased middle cerebral artery Doppler were detected on the previous antenatal scan, which was thought to be from severe anaemia although no potential cause of anaemia was found. No other anomalies were detected. He was born in good condition with normal Apgar scores. He required continuous positive airway pressure for respiratory distress soon after birth, however, at four hours of age he was ventilated for progressively worsening respiratory distress and an increased oxygen requirement. Surfactant was given and he was treated for suspected sepsis. A chest X-ray revealed bilateral white out and cardiomegaly. His haemoglobin was noted to be 19.8g/L. There was a significant difference between pre- and post-ductal oxygen saturations and a clinical diagnosis of PPHN was made at four hours of age.

The echocardiogram demonstrated a grossly dilated right side of the heart with moderate tricuspid regurgitation (velocity of 3.3m/s; estimated pulmonary artery pressure of above 50mmHg), right to left shunt across the ductus arteriosus and bidirectional shunt across the foramen ovale. This confirmed a diagnosis of PPHN with suprasystemic pulmonary artery pressure in an otherwise structurally normal heart. While performing the echocardiogram the neonatal team incidentally noted a large HAVM. By abdominal ultrasound, the radiologist confirmed the diagnosis of a large HAVM involving almost the whole right lobe of the liver at five hours of age.

The initiation of inhaled nitric oxide (iNO) is the therapy of choice in the management of PPHN to decrease the pulmonary vascular resistance. However, given the clinical diagnosis of PPHN and congestive heart failure secondary to congenital HAVM, iNO was not initiated

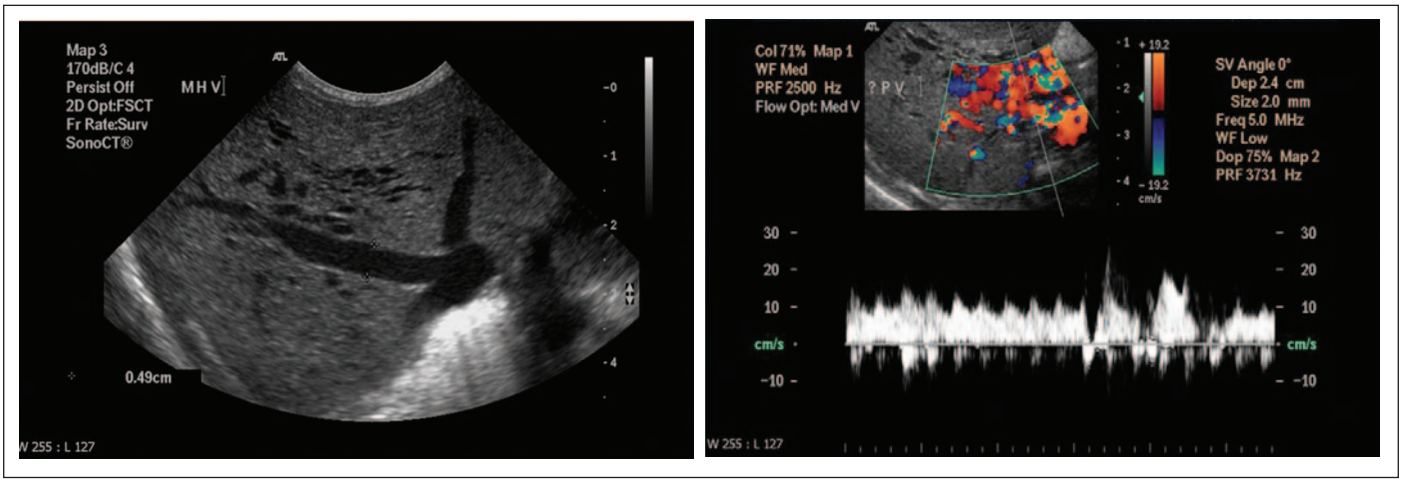

FIGURE 1 Angiography on day four showed a complex, extensive HAVM fed predominantly by an enlarged right hepatic artery. Left: Two-dimensional ultrasound image showing the extensive malformation. Right: Colour flow mapping of the arteriovenous shunt on Doppler ultrasonography.

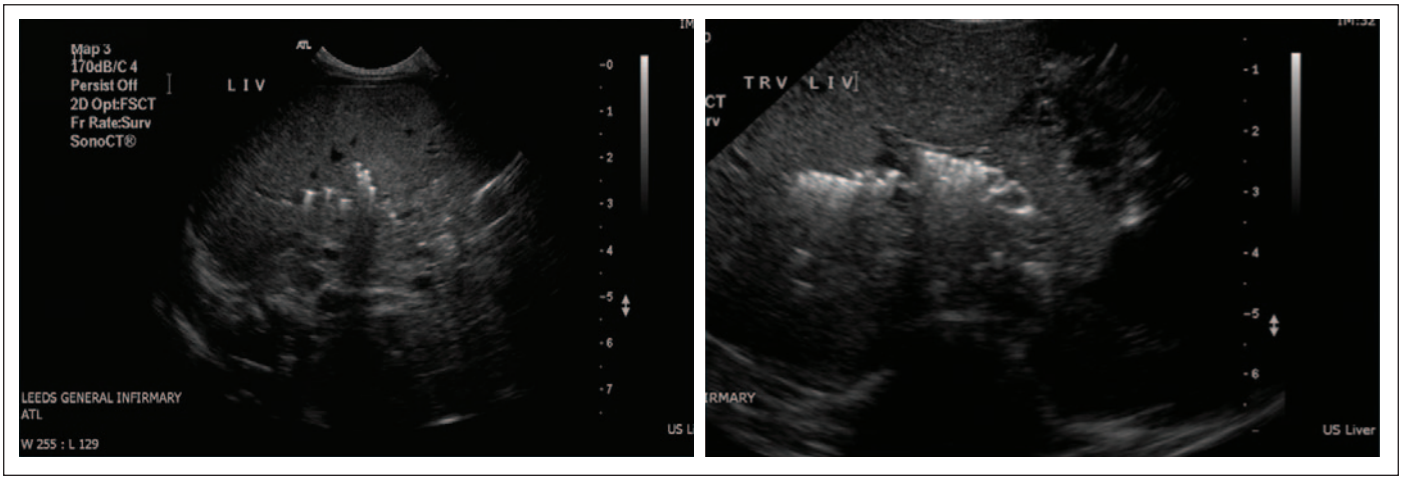

FIGURE 2 Two-dimensional ultrasound images showing the *in situ* embolisation coils for treating the HAVM. Left: Coils in the posterior right hepatic lobe. Right: Further coils in the middle and anterior parts of the right hepatic lobe.

because it may have worsened heart failure by decreasing the pulmonary vascular resistance hence increasing pulmonary blood flow. The infant was started on inotropes (dopamine 10µg/kg/min) to maintain his blood pressure. The clinical condition improved after starting diuretics (furosemide and spironolactone), although he remained ventilated. Cardiorespiratory management was optimised, guided by neonatologist-performed echocardiography.

Angiography on day four of life showed a complex, extensive HAVM fed predominantly by an enlarged right hepatic artery (**FIGURE 1**). Coil embolisation of the HAVM was accomplished using multiple coils resulting in a marked reduction in flow through the HAVM (**FIGURE 2**). Repeat ultrasound performed the day after the procedure showed a persistent small amount of flow to the HAVM and decreased hepatic size. Following the procedure, mechanical ventilation was successfully weaned and the infant was extubated two days after treatment.

The diuretics were continued and the child made excellent progress – within a week of birth he was off of all respiratory support and was monitored for signs of heart failure. Serial echocardiograms demonstrated complete resolution of pulmonary hypertension and the dilatation of the right side of the heart returned to normal size in due course. Diuretics were stopped and he was discharged home at term-equivalent age. He continues to make good progress with normal growth and neurodevelopmental parameters at two years of age.

Discussion

PPHN is commonly associated with pulmonary parenchymal pathology, sepsis, lung hypoplasia, congenital diaphragmatic hernia or congenital cardiac defects, and commonly seen in term infants.[3] The key pathophysiologic component of PPHN is either increased pulmonary vascular resistance or increased pulmonary blood flow. Almost all cases of PPHN are associated with elevated pulmonary vascular resistance, however, there are very few conditions in which increased pulmonary blood flow is an important component of PPHN and one of these conditions is peripheral arteriovenous malformation (AVM).

The symptoms of congenital AVM during infancy are seen primarily in the central nervous system, lung and liver. Cerebral AVMs have been reported to present with PPHN during the neonatal period and there are only three reported cases in the literature of congenital HAVM associated with PPHN.[2] One case of HAVM had primary presentation as PPHN[4] but PPHN has not been reported as the primary presentation in a preterm infant with HAVM. The case presented here is unique because the baby was born prematurely, had a prompt diagnosis (at

four hours of age) and early intervention (on the fourth postnatal day), which resulted in an excellent outcome.

The pathogenesis of HAVM is not understood. Congenital HAVMs are rare anomalies that most commonly (69-100%) present with congestive heart failure at an early age (mean 2.2 months). Other presenting features include hepatomegaly, consumptive coagulopathy, anaemia, portal hypertension and hydrops fetalis.[5]

The treatment of PPHN is primarily dictated by the underlying pathophysiology. Pulmonary vasodilators are commonly used to decrease pulmonary vascular resistance; iNO is the drug of choice. Treatment of PPHN due to congenital HAVM involves medical management of congestive heart failure and definitive treatment of HAVM. The options for definitive treatment include hepatic artery embolisation or ligation, partial hepatic resection and orthotopic liver transplantation.[1,5]

Successful transcatheter coil embolisation is probably the intervention of choice, especially in patients with a single arteriovenous fistula. Even in infants with multiple collaterals, embolisation may improve outcomes in severely ill patients. Surgical resection or ligation of the feeding artery is associated with significant risk in sick infants, especially with extensive HAVM and multiple collaterals. Orthotopic liver transplant may be reserved for infants with HAVM and multiple collaterals who do not respond to transcatheter embolisation and whose lesion is not amenable to surgical resection.[1,5]

Summary

Congenital HAVM presenting with PPHN is extremely rare and reported to have a very high mortality. This case had an early diagnosis and was treated with coil embolisation four days after birth, which resulted in an excellent outcome despite the infant being premature and very sick at birth. This emphasises the importance of a high index of suspicion for early diagnosis and definitive treatment in infants with congenital AVMs. As a matter of course, infants with PPHN should undergo a comprehensive structural echocardiographic assessment to rule out a structural congenital heart defect.[6,7] The authors suggest this should also include a careful evaluation of the liver and brain to rule out congenital AVM, especially when an echocardiogram shows a significantly dilated right side of the heart with signs of heart failure.

Patient consent

The authors received consent to publish this report from the patient's parents.

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