CASE REPORT

Vein of Galen malformation presenting as persistent pulmonary hypertension of newborn (PPHN)

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SUMMARY

Arteriovenous malformation is a recognised cause of persistent pulmonary hypertension in a newborn (PPHN). Vein of Galen malformation (VOGM) is a rare vascular malformation which can be life-threatening if not diagnosed and treated early. We describe a case of a term baby who presented at day 4 of life with PPHN secondary to VOGM. The neonate underwent two transarterial interventional embolisation procedures on day 9 and then another one due to developing ventricular dilation on day 44. He remains stable since and was doing well at clinical review at 10 weeks and 4 months of age. VOGM usually presents in the neonatal period with high-output cardiac failure. In a baby who presents atypically with pulmonary hypertension, a cranial ultrasound scan should be considered to look for extracardiac shunting in the brain, especially, VOGM.

BACKGROUND

Vein of Galen malformation (VOGM) is a very rare condition affecting the blood vessels of the brain with a prevalence of 1/25 000 live-births.¹ It usually presents with high-output cardiac failure in the neonatal period and it can also present with pulmonary hypertension. The presenting features in our neonate were persistent tachypnoea, lethargy and poor feeding. A chest X-ray (figure 1) showed cardiomegaly with clear lung fields and an echocardiogram showed a structurally normal heart with evidence of pulmonary hypertension. The baby was transferred to our neonatal unit for ongoing care. A cerebral ultrasound examination (figure 2) was carried out and a provisional diagnosis of VOGM was made which was later confirmed on MRI. Early diagnosis of this type of malformation is crucial as the delay in treatment often results in poor outcome.²

CASE PRESENTATION

This was a term baby boy delivered by elective caesarean section at a local district hospital. Birth weight was 3650 g (75th centile) and head circumference 37.5 cm (91st centile). The pregnancy was uncomplicated with normal antenatal anomaly scan at 20 weeks of gestation. This baby was born in good condition and did not need any resuscitation at birth. He was discharged home at 24 h of age. The baby presented to the accident and emergency on day 4 with feeding difficulties, breathlessness and lethargy. On examination, the baby was noted to be tachycardic, breathless with marked chest recessions, an overactive precordium, a loud systolic murmur over tricuspid area, normal preductal

Figure 1 Chest X-ray.

oxygen saturation 95%, low postductal oxygen saturation (85%) in air and cardiomegaly on the chest X-ray. The rest of the clinical examination was essentially normal (no hepatomegaly) and there was no evidence of dysmorphism. He was transferred urgently to a regional cardiac centre with a provisional diagnosis of congenital heart disease. Echocardiogram showed a structurally normal heart with marked tricuspid regurgitation (high systolic velocity jet of 4 m/s implying an elevated right ventricular pressure), right-to-left shunting at both atrial and ductal level with suprasystemic pulmonary pressures suggestive of persistent pulmonary hypertension of newborn (PPHN). The baby was transferred to our neonatal unit for further management. He needed continuous positive airway pressure support and 35% FiO₂ to maintain oxygen saturations above 95%.

Figure 2 Cranial ultrasound.

To cite: Tiwary S, Geethanath RM, Abu-Harb M. *BMJ Case Rep*. Published online: [please include Day Month Year] doi:10.1136/bcr-2013-200425

Figure 3 MRI scan.

INVESTIGATIONS

In view of persistent tachypnoea and no improvement in clinical status, a repeat echocardiography was performed after 48 h, which confirmed the previous finding. There was evidence of volume overload of the right heart resulting in right heart failure. This led to performing a bedside cranial ultrasound scan which revealed bilateral dilated lateral ventricles with a midline cystic vascular abnormality in vein of Galen area suggesting VOGM. This baby underwent an MRI scan of the brain, which confirmed the diagnosis (figure 3).

TREATMENT

The baby was started on oral diuretics and later underwent two transarterial embolisation procedures by an interventional radiologist at the specialist centre on day 9. He developed seizures post-treatment needing treatment with phenobarbitone. He was discharged on day 13 from the specialist centre with follow-up arrangements at our centre. He, however, had to undergo another embolisation procedure on day 44 following progressive ventricular dilation. This time around his recovery was uncomplicated and was discharged home.

OUTCOME AND FOLLOW-UP

On follow-up at 4 months of age, he is doing well and his head size is now growing along the 25th centile and the ventricular indices stabilised. He does not have any signs of cardiac failure and he has normal neurological examination.

DISCUSSION

VOGMs are rare congenital vascular malformations resulting from the development of arteriovenous (AV) connection between primitive choroidal vessels and the median prosencephalic vein of Markowski.³ The malformation develops between 6th and 11th week of fetal development.⁴ It comprises less than 1% of cerebral vascular malformations at any age, but in the paediatric vascular malformation group it might account up to 30%. High-output cardiac failure in the newborn period is a common presenting feature. However, it can also result in mass effects causing progressive neurological impairments. Alternatively, the malformation can also cause obstruction to the flow of cerebrospinal fluid and result in hydrocephalus. The natural history of these lesions is mostly depressing with a mortality of 42–91% if left untreated.⁵ There are many classification systems of VOGM and the two most widely used systems are provided by Yasargil and Lasjaunias. Yasargil classified VOGM

into four types. Types 1–3 lesions involve a direct fistulous communication with the vein of Galen. Type 4 lesions represent parenchymal AV malformations, which drain into vein of Galen.

Lasjaunias classified VOGM as choroidal and mural types depending on the location of fistula. Choroidal type has multiple fistulas communicating with the anterior end of the median prosencephalic vein. In mural type the fistula is in the wall of the median prosencephalic vein. The outcome for choroidal type is worse than for those with mural type.⁶ The detailed classification of VOGM is not within the scope of this article and has been described elsewhere.⁷ There are many published case reports on VOGM presenting in neonatal period with high-output cardiac failure, but very less has been mentioned about its presentation with PPHN apart from a single case report by Seher *et al*⁸ who described a baby presenting at 24 h of age with cardiac failure who had VOGM and died at 22 days of age. The predominant accompanying feature in this baby was PPHN.

PPHN is defined as a syndrome resulting from maladaptation to extra-uterine life characterised by an elevation in pulmonary vascular resistance, right-to-left shunting and hypoxaemia. Its prevalence is 2–6/1000 live-births. The typical echocardiographic findings demonstrate right-to-left shunting at both the patent foramen ovale and the patent arterial duct. There is often severe tricuspid regurgitation suggesting raised pulmonary pressure. PPHN is frequently associated with underlying pulmonary parenchymal abnormalities including meconium aspiration, congenital pneumonia, sepsis, lung hypoplasia and alveolar capillary dysplasia. Sometimes there is no evidence of lung disease and the aetiology is unknown. It can be associated with conditions affecting the left side of the heart and congenital cardiac malformations must be sought particularly those affecting the mitral valve. Though PPHN is multifactorial in origin, most of the time, the cause of the pulmonary hypertension can be explained by careful history and clinical examination. In the absence of any abnormal finding or setting in a neonate with pulmonary hypertension, one should actively look for vascular malformations of the liver and brain before diagnosing PPHN.

Intracardiac shunting during the newborn period usually does not result in right-sided volume overload. In the presence of significant right heart volume overload, we recommend to rule out extracardiac shunts, predominantly AV malformations in the liver and brain. It is possible that the pulmonary hypertension masked the right heart failure in the early days in our baby.

Learning points

- ▶ Early diagnosis and treatment of VOGM is crucial as delay in treatment results in poor outcome.
- ▶ In the absence of any abnormal finding/setting in a neonate with persistent pulmonary hypertension of newborn, we should actively look for arteriovenous malformations particularly in the liver and brain (vein of Galen malformation).
- ▶ Endovascular embolisation to occlude the shunt is the treatment of choice and is associated with better outcome.

Acknowledgements The authors would like to thank Professor Bhattacharya, Royal Hospital for sick children, Yorkhill, Glasgow for permitting them to use MRI scan picture for this report.

Contributors RMG and MA-H were involved in the management of the patient. RMG and ST performed the literature search and wrote the manuscript. All authors were involved in editing the manuscript.

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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