Necrotising enterocolitis in a full-term infant with reversed diastolic flow in the descending aorta: what is the diagnosis?

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SUMMARY

Vein of Galen malformations (VGM) are rare intracranial vascular anomalies that constitute 1% of all intracranial vascular malformations. Untreated VGM have a very poor prognosis. A high proportion of patients who present in the neonatal period rapidly deteriorate and succumb to congestive cardiac failure. The prenatal diagnosis and possible in utero referral to highly specialised centre of postnatal care have resulted in considerable improvement in prognosis. The authors present a case of a postnatally diagnosed VGM in a full-term infant presenting with progressive cardiac failure and necrotising enterocolitis secondary to gut hypoperfusion. They emphasise the importance of prenatal diagnosis by pulsed wave Doppler and colour-velocity imaging for subsequent referral to a centre of excellence in a multidisciplinary approach involving neonatologists, paediatric cardiologists and interventional radiologists in order to optimise the management and limit the neurological sequelae in children with this type of malformation.

BACKGROUND

Vein of Galen malformations (VGM) are rare intracranial vascular anomalies that constitute 1% of all paediatric congenital anomalies. They represent 15–30% of vasculature malformations presenting in the paediatric age group. These lesions are characterised by the presence of multiple arteriovenous shunts draining into a median cerebral venous collector, the median prosencephalic vein of Markowsky (MProSV), corresponding to an embryonic vessel normally absent in the adult stage.^{1–4}

The authors present a case of an aneurysmal VGM, with postnatal diagnosis, manifested by a rapid cardiovascular deterioration and marked systemic hypoperfusion with early necrotising enterocolitis. The aim of this case report is to alert the clinician that in a newborn with a quick cardiovascular deterioration, with a structurally normal heart, it is important to think about an extra-cardiac aetiology, such as a high-volume arteriovenous shunt. The early recognition of this entity and its judicious and timely management are important in the prognosis.

CASE PRESENTATION

A female infant was delivered vaginally, at 40 weeks gestation, after an uneventful pregnancy. Maternal serological tests and fetal ultrasound scans (12, 22 and 33 weeks gestation) were normal. The infant's Apgar scores were 6, 8 and 9 at 1, 5 and 10 min, respectively. At 12 h of life she was transferred to

the neonatal intensive care unit (NICU) presenting with acrocyanosis and hypotonia with feeding difficulties.

At admission in the NICU, physical examination revealed haemodynamic stability, without four limbs blood pressure differential and a grade II/VI systolic murmur at the left parasternal region. The remaining physical examination was unremarkable.

INVESTIGATIONS

Septic work-up: normal haematological findings, C reactive protein 0.22 mg/dl; mild hyperlactacidaemia (4 mmol/l) and hypoglycaemia (28 mg/dl).

Chest x-ray: cardiomegaly.

Echocardiography: structurally normal heart with right cavities dilatation and high ventricular contractility, pulmonary artery systolic pressure ± 40 mm Hg, patent *ductus arteriosus* and patent *foramen ovale* with left–right shunt.

DIFFERENTIAL DIAGNOSIS

- ▶ Early-onset sepsis
- ▶ Inborn errors of metabolism
- ▶ Cardiac heart failure due to an extra-cardiac cause, such as a high-volume arteriovenous shunt

TREATMENT

- ▶ Antimicrobial therapy (combination of ampicillin and gentamicin)
- ▶ Intravenous fluids (glucose 4–5 mg/kg/min)
- ▶ Bowel rest by stopping enteral feeding

OUTCOME AND FOLLOW-UP

During the following 12 h, there was a rapid and progressive deterioration of the general state, with tachypnoea and intercostal retractions, poor peripheral perfusion and abundant bile gastric draining. The abdomen became progressive distended and painful through palpation.

At 24 h of life, the septic work-up was repeated, showing a persistent hyperlactacidaemia (lactate 5.2 mmol/l) and a slightly elevated C reactive protein 2.69 mg/dl, with normal glucose, urea, creatinine, ionogram, transaminases and ammonia. Abdominal x-ray images were suggestive of necrotising colitis stage II (figure 1a,b). After evaluation by paediatric surgery department, it was decided to maintain surveillance and start metronidazole.

Through time, the newborn developed recurrent apnoea and need for invasive ventilation. A control abdominal x-ray at 48 h of life revealed dilated bowel loops, paucity of gas and a 'fixed loop' (unaltered gas-filled loop of bowel), but no pneumoperitoneum (figure 1c). At the 72 h of life, the

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Figure 1 (a-c) Abdominal x-ray demonstrating intestinal pneumatosis, characterised by linear radiolucent images, visible on the walls of intestine loops.

echocardiography revealed an accentuated dilatation of the right heart chambers, moderate pulmonary hypertension and a reversed diastolic flow in the descending aorta.

With all these features, we suspected an extra-cardiac problem for the cardiac failure, with a high index of suspicion for an arteriovenous malformation aneurysm. A bedside cranial ultrasound was done and demonstrated a midline vascular malformation (figure 2a), with reduced flow in the anterior cerebral artery. A screening cerebral angiotomography was performed diagnosing an aneurysmal VGM with $30 \times 15 \times 18$ mm of dimensions (figure 2b). An exploratory laparotomy was performed on the same day demonstrating pneumotosis coli and colic perforation.

A peritoneal drainage and subsequent ileostomy were necessary. The congestive heart failure was managed with enalapril, furosemide, dopamine and digoxine, but, over time, there was a progressive deterioration of the heart function, unresponsive to medical treatment.

After contact with several hospitals, a multidisciplinary team agreed that an endovascular approach offered the best possible outcome and a life-saving endovascular technique was attempted at day 13. Perioperative angiography revealed a mixed-type VGM. The right anterior and posterior choroidal pedicles were catheterised and, after injection of cyanoacrylate glue, there was a significant slowing of venous filling. Subsequent to the

Figure 2 Vein of Galen malformation images: (a) Cranial ultrasound showing a median anechoic lesion, with a pulsatile flow in the Doppler study, (b) Contrast-enhanced axial tomography scan displaying a well-defined, intensively enhancing, median lesion.

procedure, the infant showed marked haemodynamic instability, mixed acidosis and severe hypoxaemia (arterial blood sample: pH 6.9, pCO₂ 74 mm Hg, pO₂ 14.6 mm Hg, HCO₃ 16.1 mmol/l, BE—17.6 mmol/l), requiring aggressive ventilator parameters. One hour after admission into the NICU, the infant suffered cardiorespiratory arrest unresponsive to resuscitation measures.

Postmortem cranial ultrasound revealed an intraventricular haemorrhage with bleeding within the brain parenchyma. No autopsy was done.

DISCUSSION

VGM arise as a result of direct arteriovenous communications between the arterial network and the MProSV. The mechanism of this abnormal development remains unknown, probably occurring during the embryonic period between 6th and 11th weeks of fetal life. As a consequence of the shunts, the anterior segment of the MProSV, instead of regressing, progressively enlarges under the stress produced by high-pressure inflow from the choroidal feeders.²

VGMs are occasionally detected on antenatal ultrasound scans (from about 25 weeks gestation) as apparently cystic midline brain lesions, colour-flow Doppler, suggesting a VGM. Antenatal MRI confirms the diagnosis and allows treatment planning with delivery at a centre with the appropriate facilities and expertise, principally fetal medicine, neonatology, paediatric cardiology and intensive care, and interventional neuroradiology. More commonly, VGMs are diagnosed after birth. The presentation is related to the size of the shunt itself, imposing elevated preload on the right side of the heart leading to cardiac failure. Often delivery and the first 24 h are unremarkable. Larger shunts may then show rapid deterioration with progressively worsening cardiac failure evolving into multiorgan failure. A smaller shunt may present later with mild cardiac failure and failure to thrive. Occasionally, children present later in childhood with macrocrania or prominent facial veins secondary to venous outflow obstruction.^{5 6}

Untreated VGMs have a poor prognosis and are almost always fatal. Management of children with high-flow arteriovenous shunts of the brain is among the most challenging areas in modern medicine and combines the symptomatic treatment of heart failure and hypoperfusion complications with the treatment of endovascular injury. Ideally, when the haemodynamic conditions allows, the endovascular procedure should be deferred until 5–6 months of life. By decreasing flow to the VGM, perfusion to the kidneys and heart improves, venous hypertension in the pulmonary circulation can be decreased, and persistent right-to-left shunts can close. Management options to decrease flow include diuretics, inotropic agents and vasodilators.⁴ Their use offers stabilisation before intervention and contributes in reducing neurological injury. However, as in the presented case, a congestive heart failure refractory to medical treatment is an indication for emergency embolisation.^{1–3 7–11}

To the best of our knowledge, this is the first case of VGM presented as an NEC that has been described so far. High-volume arteriovenous shunts, such as VGM, typically result in prominent reversal of forward flow in the abdominal aorta during diastole and a resultant low-diastolic pressure. The mesenteric circulatory insufficiency secondary to this 'diastolic

steal phenomenon' probably predispose to NEC development.¹² With the presented case, the authors intend to alert the clinician to the possibility of this diagnosis in any newborn with rapid and progressive heart failure with no obvious cause in order to make an early diagnosis. The vein of Galen can be visualised using ultrasound with Doppler. Aggressive management of heart failure is essential in these children aiming to limit the neurological sequelae and postpone an endovascular intervention.

Learning points

- ▶ Vein of Galen malformations are rare intracranial vascular anomalies.
- ▶ Untreated vein of Galen malformations (VGM) have a very poor prognosis.
- ▶ Early diagnosis and aggressive medical treatment of cardiac failure may postpone endovascular intervention.
- ▶ Routine ultrasound screening in prenatal care can lead to identification of a VGM; the newborn should be born in a centre with a possible multidisciplinary team approach (including neurology, neurosurgery, interventional neuroradiology, cardiology and neonatal intensive care).

Competing interests None.

Patient consent Obtained.

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