Vein of Galen arteriovenous malformation mimicking coarctation of the aorta

Mohammed Firdouse · Arnav Agarwal ·
Tapas Mondal

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Abstract Arteriovenous malformation of the vein of Galen is a rare congenital intracranial anomaly lacking a capillary bed and subsequent aneurysmal enlargement of the arterial and venous system, warranting careful management due to associated morbidity and mortality. Coarctations of aorta demonstrate similar neonatal echocardiographic signs to the vein of Galen arterial malformation (VGAM). We present a boy at 37 weeks of gestation whose initial ultrasound and echocardiographic investigations showed a dominant right ventricle and isthmal hypoplasia, suggestive of coarctation of aorta. Follow-up ultrasound and echocardiography revealed an arteriovenous malformation involving middle and posterior cerebral artery branches, eliminating coarctation of aorta. VGAM was confirmed by further ultrasound and

angiographic investigation, which demonstrated a tangle of cerebral and choroidal arterial branches centrally feeding into an enlarged vein of Galen. The boy's hemodynamic and neurological statuses were confirmed to be stable despite increased venous pressure. Elective embolization at 7 months of age was complicated by a cerebrovascular accident, resulting in right hemiparesis despite no residual cardiac issues. This case demonstrates that rarely, arteriovenous malformations such as the vein of Galen malformations may be the primary cause of patients presenting with coarctation of aorta. The rarity of this condition and its guarded prognosis make our case of special interest to cardiologists and the perinatal care team.

Keywords Vein of Galen · Cerebral arteriovenous malformation · Coarctation of aorta · Isthmal hypoplasia

M. Firdouse · A. Agarwal
Faculty of Health Sciences, McMaster University, Hamilton,
ON, Canada
e-mail: mohammed.firdouse@learnlink.mcmaster.ca

A. Agarwal
e-mail: arnav.agarwal@learnlink.mcmaster.ca

M. Firdouse
Bachelor of Health Sciences (B.H.Sc. Honours) Program,
McMaster University, 4077 Tea Garden Circle, Mississauga,
ON L5B 2W5, Canada

A. Agarwal
Bachelor of Health Sciences (B.H.Sc. Honours) Program,
McMaster University, 252 Macedonia Crescent, Mississauga,
ON L5B 3R7, Canada

T. Mondal (✉)
Department of Pediatrics, McMaster Children's Hospital,
McMaster University, 1280 Main Street West, Hamilton,
ON L8S 4K1, Canada
e-mail: mondalt@mcmaster.ca

Riassunto Le malformazioni artero-venose della vena di Galeno sono rare anomalie intracraniche congenite caratterizzate da assenza del letto capillare e successiva dilatazione aneurismatica dei sistemi arterioso e venoso, che richiedono un'attenta gestione, per le associate morbidità e mortalità. Le coartazioni dell'aorta presentano segni ecocardiografici neonatali simili alla malformazione artero-venosa della vena di Galeno (VGAM). Presentiamo il caso di un ragazzo che, alla 37 settimana di gestazione, mostrava all'esame ecocardiografico un ventricolo destro dominante e un'ipoplasia dell'istmo, suggestivi per coartazione dell'aorta. Il follow-up ecografico ed ecocardiografico hanno rivelato una malformazione artero-venosa che coinvolgeva rami delle arterie cerebrali media e posteriore, eliminando l'ipotesi di coartazione dell'aorta. La diagnosi di VGAM è stata confermata da ulteriori indagini ecografiche e angiografiche, che hanno dimostrato un groviglio di rami arteriosi cerebrali e dei plessi corioidei alimentato da

una vena di Galeno ingrandita. I parametri emodinamici e neurologici del ragazzo erano stabili nonostante l'aumento della pressione venosa. A 7 mesi di età l'embolizzazione elettiva è stata complicata da un incidente cerebrovascolare, con conseguente emiparesi destra, nonostante non vi siano state complicanze cardiache. Questo caso dimostra che rare malformazioni arterovenose, come le malformazioni della vena di Galeno, possono essere la causa primaria di pazienti che si presentano inizialmente con la diagnosi di coartazione dell'aorta. La rarità di questa condizione e la sua prognosi severa rendono il nostro caso di particolare interesse per i cardiologi e per i team di assistenza perinatale.

Introduction

Many vascular congenital abnormalities in neonates in the Intensive Care Unit warrant timely diagnosis and careful attention due to the high risk of morbidities and mortality associated with their prognosis. Recognition of the clinical symptoms associated with these anomalies is integral to the differential diagnoses of such conditions. Arteriovenous malformations refer to anomalous connections between arteries and veins in the body without an intervening capillary bed. A vein of Galen arterial malformation (VGAM) is one such anomalous intracranial vascular connection where arteries from the circle of Willis directly drain into a deep cerebral vein. Steinheil et al. was the first to describe this anomalous intracranial arteriovenous shunt as a "varix aneurysm" [1]. However, the terms "vein of Galen malformation", "vein of Galen aneurysmal formation", "arteriovenous aneurysms of the vein of Galen" or "aneurysms of the vein of Galen" have been used interchangeably to describe this condition. This is a medical misnomer as the pathology affects the embryonic median prosencephalic vein and not the vein of Galen [1]. The vein of Galen, a derivative of the embryonic median prosencephalic vein, is actually formed by the union of the left and right internal cerebral veins and eventually drains into the straight sinus.

VGAM represents approximately 1 % [2] of intracranial vascular malformations and account for about 30 % of all arteriovenous malformations in the pediatric age group [3]. The lack of a capillary bed between the cerebral arterial and venous system results in the aneurysmal enlargement of the cerebral vein. Compression caused by this dilated vein may lead to cerebral hypoplasia, atrophy or hydrocephalus [4]. The direct shunting of blood also results in congestive heart failure, which is the most common form of clinical presentation in neonates with VGAM.

On the other hand, coarctations of the aorta account for approximately 8 % of all congenital heart defects [5]. It is

characterized by the narrowing of the isthmus of aortic arch, which is most commonly pre-ductal (juxta-ductal) and very rarely post-ductal. Coarctations of the aorta increase the load on the right ventricle resulting in right ventricular hypertrophy and dilation, as seen in antenatal and early neonatal echocardiographic, which is similar to neonates with VGAM [5].

Although advancements in sonographic and Doppler techniques have made antenatal diagnoses of VGAM and coarctation of aorta possible, their presentations remain similar and often hard to discriminate. Here, we report a neonate with VGAM, who was initially suspected as having a coarctation due to aortic isthmal hypoplasia. The infrequency of these conditions and difficulties in differentiating antenatal clinical findings associated with their diagnosis make our case of special interest to pediatricians and imaging specialists.

Case report

We present a baby boy who was born by a cesarean section with a weight of 3 kg at 37.5 weeks of gestation. Routine fetal ultrasound and echocardiography done at 36 weeks of gestation raised suspicions of coarctation in the aorta due to isthmal hypoplasia with a dominant right ventricle. Parents were accordingly counseled. A follow-up ultrasound and echocardiography at 37 weeks of gestation revealed a cerebral arteriovenous malformation involving branches of the middle and posterior cerebral artery. The nature of this finding remained inconclusive. The right heart continued to remain dilated; however, there were no obvious signs of pericardial effusion. The baby boy was born uneventfully with normal Apgars scores of 8 at 1 min and 9 at 5 min, respectively. A neonatal echocardiography confirmed dilated right atrium and ventricle, mild right ventricular hypertrophy with good biventricular function and ruled out coarctation of the aorta (Fig. 1a, b). No murmurs were audible; however, a bruit was detected over the anterior fontanelle upon auscultation. The baby remained hemodynamically and neurologically stable. Consequently, parents were relieved that their baby was healthy and had no significant disease.

As coarctation in the aorta was ruled out in this baby, focus of attention was directed towards other causes of right ventricular overload leading to further imaging studies including cerebral ultrasound which revealed an enlargement of the straight sinus as well as the vein of Galen, raising the suspicion of a cerebral arteriovenous malformation (Fig. 1c). Therefore, the baby underwent magnetic resonance angiography and venography of the brain, which confirmed the diagnosis of a VGAM. The magnetic resonance imaging showed a tangle of tortuous vessels involving the branches of the left middle cerebral

Fig. 1 **a** Four-chamber echocardiographic image showing ventricular size disproportion. Right ventricle (RV) is significantly dilated and larger than the left ventricle (LV). Also, $RA \gg LA$. The interatrial septum is deviated to the left suggesting high RA pressure. **b** In a ductal view, there is isthmal hypoplasia without any definite coarctation. The brachiocephalic vessels seem to be slightly enlarged. AV aortic valve, LA left atrium, MPA main pulmonary artery, DA

descending aorta. **c** Color flow mapping of cerebral ultrasound showing extensive involvement of the major cerebral arteries: A parasagittal view showing interconnecting anterior and posterior cerebral arteries; B transverse view showing bilateral posterior cerebral arteries; C transverse view showing bilateral middle cerebral arteries; D 3-dimensional reconstruction of the cerebral arteriovenous malformation

artery, left posterior cerebral artery and left choroidal artery. All the aforementioned feeding vessels coursed into the enlarged confluence of the straight sinus and the enlarged vein of Galen where a venous pouch existed. There was no evidence of hydrocephalus, intracerebral or intraventricular hemorrhage, ischemia or increased T1 hyperintense signal in the brain. In fact, the brain appeared very healthy and normal despite the increased venous pressure (Fig. 2a, b). The baby also underwent electroencephalographic testing which showed no seizure activity. As the baby continued to remain clinically stable, the neurology team was consulted to follow the baby.

The baby underwent elective surgery for correction of his vein of Galen malformation, at 7 months of age, which unfortunately led to a cerebrovascular accident resulting in right hemiparesis. Over the next 2 years, his motor skills and developmental milestones increased with physiotherapy and occupational therapy. There were no residual cardiac problems as well.

Discussion

Vein of Galen malformation is a rare cause of cardiac failure in neonates. In some cases, VGAM patients have

Fig. 2 **a** Sagittal magnetic resonance image (angiography) with contrast showing the anterior cerebral artery (ACA) and middle cerebral arteries (MCA) draining into a large pouch (P) behind the third ventricle and above the tentorium cerebelli. No signs of hydrocephalus and intracerebral or intraventricular hemorrhage can be seen. **b** Transverse magnetic resonance image (angiography) with

contrast showing tangled and tortuous feeding vessels involving left middle (MCA) and left posterior cerebral arteries (PCA) draining into a dilated venous confluence of the vein of Galen (GV), straight sinus, and bilateral transverse sinuses. No signs of hydrocephalus and intracerebral or intraventricular hemorrhage can be seen

been noted to show signs of coarctation in the aorta, either being associated with it [6–8] or mimicking the disease (pseudocoarctation) [9–11].

The exact etiology and pathophysiology of coarctations in the aorta remain unclear; however, there are two widely accepted theories—the ductal tissue theory and the reduced forward flow theory. According to the ductal tissue theory, tissue from the ductus arteriosus invades into the distal aorta and the isthmus. Following birth, as the ductus arteriosus closes, the part of the aorta with infiltrating ductal tissue also undergoes constriction [12]. According to the reduced forward flow theory, if cardiac chambers or large vessels do not receive sufficient blood, it will not develop. Normally, the aortic isthmus receives only about 10 % of the combined cardiac output [13]. In conditions such as VGAM, blood is redirected superiorly to areas of low vascular resistance, thereby further reducing the amount of blood being delivered to the aortic isthmus. Consequently, isthmal growth may be compromised which might manifest itself as a coarctation [12]. Once the absence of an aortic coarctation after birth (following the closure of ductus arteriosus) has been confirmed, then there will not be any further risk of developing coarctations, as the patient continues to wait for treatment of the arteriovenous malformation.

One of the most common antenatal presentations of coarctation in the aorta is right–left ventricular size discrepancy upon echocardiographic investigation. However, this diagnostic criterion has a high false positive rate of up to 80 % especially after 34 weeks of gestation [14]. This can primarily be attributed to the fact that the right

ventricle is physiologically larger than the left ventricle and this discrepancy is more prominent towards the end of the gestational period (normal: LV/RV <1.5) [15]. Moreover, other causes of prominent right ventricle include constriction of ductus arteriosus, a spectrum of hypoplastic left ventricle, tricuspid regurgitation (for example: Ebstein’s anomaly), and more rarely arteriovenous malformations such as the vein of Galen malformation.

As previously reported, the prognosis of patients undergoing repair only for aortic coarctation without any treatment for their vein of Galen malformation is dismal [16]. Therefore, it would be vital to consider the presence of arteriovenous malformations in the upper extremities and the head and neck region as a primary cause for the coarctation of aorta. It must be noted that treatment of one condition will not address the other. If both the lesions were to be present, patients would need separate interventions for the coarctation and the arteriovenous malformations.

Accordingly, parents of such patients must be counseled to ensure that they have the right understanding of two seemingly different conditions in their child, of which, one may be the cause of the other. It must be emphasized that both conditions might need completely different treatments (one treatment will not improve the other).

Conclusion

Our case demonstrates that rarely, arteriovenous malformations such as the vein of Galen malformations may be

the primary cause of patients presenting with symptoms of coarctation of aorta. Thus, fetal–maternal medicine teams should be aware of non-cardiac etiologies of such presentations, and include a vein of Galen arteriovenous malformation in patients presenting with coarctation of aorta, despite the relative rarity of the condition. As cerebral arteriovenous malformations are a known rare cause of isthmal hypoplasia or definite coarctation, fetal ultrasound should regularly include examination of the superior vena caval distribution, particularly in the brain.

Conflict of interest Mohammed Firdouse, Arnav Agarwal, Tapas Mondal declare no conflicts of interest.

Informed consent All procedures followed were in accordance with the ethical standards of the responsible committee on human-experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000 (5). All patients provided written informed consent to enrolment in the study and to inclusion in this article of information that could potentially lead to their identification

Ethics standards The authors confirm that all human and animal studies have been approved by the appropriate ethics committee and have therefore been performed in accordance with the ethical standards laid down in the Helsinki Declaration of 1975 and its late amendments. Additional informed consent was obtained from all patients for which identifying information is not included in this article.

Human and Animal Studies The study was conducted in accordance with all institutional and national guidelines for the care and use of laboratory animals

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