Distinctive neck swelling in a patient with a vein of Galen arteriovenous malformation

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Abstract A vein of Galen arteriovenous malformation (VGAM) is a rare intracranial shunt lacking a capillary bed and resulting in subsequent aneurysmal enlargement. VGAM has been previously reported to present as cardiovascular abnormalities, including increased right-sided cardiac load and pressure, cardiac dilatation, pulmonary hypertension and retrograde flow into the aortic arch. We report the first case of VGAM presenting as a gross right-sided neck swelling in a 39-week-old immediately at birth. Transthoracic echocardiography detected dilatation of the superior vena cava and innominate vein, increased right ventricular and pulmonary arterial pressure, and retrograde flow into the aortic arch. Follow-up neck ultrasound revealed tortuous carotid artery and bilateral dilatation of neck veins. Transcranial ultrasound revealed abnormally dilated vessels posterior to the third ventricle, confirming VGAM. Our case demonstrates that while the

differential diagnoses of neck swelling are extensive, such a presentation upon Doppler and echocardiographic investigation, may be suggestive of extracardiac causes such as VGAM. The management of such a condition remains critical due to risk of hemorrhage and extensive cerebral involvement.

Keywords Vein of Galen · Cerebral arteriovenous malformation · Pulsatile neck swelling · Gross neck mass

Riassunto La vena di Galeno (VGAM) è una malformazione arterovenosa rara a livello intracranico, caratterizzata da shunt senza un letto capillare e conseguente, successivo, allargamento aneurismatico. La VGAM è stata segnalata in precedenza come anomalia cardiovascolare, con sovraccarico del cuore destro, dilatazione cardiaca, ipertensione polmonare e flusso retrogrado nell'arco aortico. Riportiamo il primo caso di VGAM caratterizzata da gonfiore e tumefazione del lato destro del collo in neonato nato alla 39 settimana di gestazione. L'ecocardiografia transtoracica ha rilevato dilatazione della vena cava superiore e della vena anonima, aumentato di volume del ventricolo destro e della pressione arteriosa polmonare e flusso retrogrado aortico. Il follow-up ecografico del collo ha rivelato tortuosità della carotide e dilatazione bilaterale delle vene del collo. L'ecografia transcranica ha rivelato un'anomala dilatazione dei vasi posteriori al terzo ventricolo, confermando la presenza di VGAM. Il nostro caso dimostra che, anche se le diagnosi differenziali delle tumefazioni del collo sono numerose, una presentazione Doppler ed ecocardiografica come quella riportata, può essere indicativo di causa extracardiaca come la VGAM. La gestione di tale condizione è critica per il rischio di emorragia e di ampio coinvolgimento cerebrale.

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Introduction

Diagnosis and management of intracranial arteriovenous shunts in the pediatric population is often challenging. A vein of Galen arteriovenous malformation (VGAM) is the most common type of intracranial arteriovenous shunt in neonates and infants [1], accounting for up to 30 % of all paediatric vascular abnormalities [2, 3]. Anatomically, this embryonic arteriovenous shunt is located medially in the choroid fissure, where the prosencephalic and mesencephalic arterial groups drain directly into the cerebral venous system [1, 4]. Raybaud et al. [5, 6] were the first to recognize this dilated and ectatic venous structure as the median prosencephalic vein of Markowski. Thus, VGAM is a medical misnomer as the pathology affects the embryonic median prosencephalic vein of Markowski and not the vein of Galen [7]. The vein of Galen, a derivative of the embryonic median prosencephalic vein, is formed by the union of the left and right internal cerebral veins and eventually drains into the straight sinus. Lacking a fibrous wall, the median prosencephalic vein typically balloons as a consequence of the direct arteriovenous connection, resulting in an aneurysmal enlargement [6, 8]. Numerous complications, including increased cardiac output, myocardial ischemia, congestive cardiac and multiple organ failures, cerebral venous hypertension and associated hydrocephalus, cerebral edema, and developmental or neurological deficits, have been associated with VGAMs [1, 9–15].

VGAMs are typically detected on antenatal ultrasonographic scans as cystic midline brain lesions after 25 weeks of gestation. Diagnosis often relies on colour flow Doppler imaging, with magnetic resonance imaging as a confirmatory investigation. When detected postnatally (more typical in clinical practice), the malformation is often diagnosed by transcranial ultrasound after the initial suspicion is raised by cardiac investigations [1].

Neonatal clinical diagnoses of VGAM may be made based on different symptoms and signs, and may be incidental or associated with significant sickness. Here, we report the first case of VGAM presenting as a gross right-sided neck swelling. The timeliness and diagnostic value such a presentation might add to differential diagnoses makes this case of special interest to pediatricians, cardiologists and neurologists.

Case report

We present a 4,080-g baby girl born at 39 weeks and 4 days of gestation with a head circumference of 35.5 cm (50th percentile). The baby girl immediately presented with a right-sided neck swelling and no respiratory effort at

Fig. 1 Four chamber echocardiographic view showing deviation of interatrial septum to the left suggesting pressure loaded right atrium, and a significantly dilated right ventricle

birth. The patient needed significant resuscitation, including positive pressure ventilation, chest compressions and intubations. A chest radiograph was taken following intubation, revealing cardiomegaly. The baby was then transferred to the neonatal intensive care unit for further care.

Initial echocardiographic investigation showed increased right ventricular and pulmonary arterial pressure (Fig. 1) with right-to-left patent ductus arteriosus. Retrograde flow into the aortic arch from the neck vessels, tortuous right carotid artery, and dilatation of superior vena cava and innominate vein were also observed (Fig. 2a, b). A normal S1 and a loud S2 with a grade 4 out of 6 systolic murmur radiating to the neck were noted on auscultation. A follow-up neck ultrasound demonstrated a tortuous right carotid artery and bilateral dilatation of neck veins indicative of a cerebral arteriovenous malformation. Further transcranial ultrasound investigation revealed abnormally dilated vessels posterior to the third ventricle. Occasional jerking of the arms and legs and a plethoric facial appearance were observed, but no dysmorphic features were noted.

The patient suffered intracranial hemorrhage during follow-up, resulting in convulsion and extensive cerebral involvement. Consultation with the family was then followed with withdrawal of care.

Discussion

VGAM is a rare condition, and its diagnosis is typically based on the presentation of adverse symptoms or the use of imaging investigations to identify abnormal vasculature (Bhattacharya, 2003). The differential diagnoses of patients presenting with a neck mass or swelling may be challenging, considering the proximity of numerous vital structures in this region. Midline neck masses that move

Fig. 2 **a** Transthoracic echocardiograph showing significant dilatation of the brachiocephalic artery (BCA), left common carotid artery (LCCA) and left subclavian artery (LSCA) from the ascending aorta (AsAo). **b** Doppler image showing severe flow reversal in the descending aorta (DAo) in relation to cerebral vasodilation as expected in Steal Syndrome

while swallowing or tongue protrusion may be indicative of thyroidal masses, thyroglossal duct cysts, dermoid cysts or thymic cysts. Multiple independent masses, especially in the posterior triangle of the neck, could be indicative of lymphomas or metastases. The most commonly diagnosed lateral neck lesions include cystic hygromas and branchial cysts in pediatric and adult populations, respectively. A solid neck mass may be indicative of a salivary gland tumor, sternocleidomastoid tumor (torticollis) or carotid body tumor. Neck swelling lesions that differ from these characteristics might be suggestive of vascular abnormalities such as aneurysms, hemangiomas and ectasias [16].

The aforementioned differential diagnoses remain primary considerations with the presentation of a congenital neck lesion. However, the specific presentation seen in our case differs from the usual differential diagnosis in several ways. Thyroidal mass, thyroglossal duct cysts, dermoid cysts and thymic cysts are midline, whereas the swelling observed in our case was right-sided. Enlarged lymph nodes represent a plausible differential diagnosis, but are commonly associated with malignancy, infection or an immune reaction as well as fever-related symptoms, which were absent in our case. Branchial cleft cysts are commonly a result of acute infection, making it an unlikely

differential diagnosis for a presentation noted immediately at birth. Last, laryngoceles are typically associated with coughing, while hemangiomas are characterized by discolored red-to-blue masses, neither of which were noted in our case [17].

Increased right-sided cardiac load and pressure, cardiac dilatation, pulmonary hypertension and retrograde flow into the aortic arch have been previously reported to be strongly suggestive of VGAM [1]. A right-sided gross neck swelling at birth with the presence of a pulse or bruit in the neck might warrant further investigation for a diagnosis of VGAM. This neck swelling, in conjunction with confirmatory transcranial and cardiovascular ultrasonographic observations, should be indicative of VGAM as a potential diagnosis to pediatricians, neonatologists and cardiologists.

Conclusion

We report the first gross presentation of VGAM in a patient born with a pulsatile right-sided neck swelling. Our case demonstrates that while the differential diagnoses of neck swelling are extensive, Doppler and echocardiographic investigations revealing retrograde flow into the aortic arch from the neck vessels, tortuous right carotid artery, and dilatation of superior vena cava and innominate vein may be indicative of extracardiac causes such as VGAM. Due to the high risk of cardiac and systemic failures, developmental deficits or neurological complications associated with VGAM, this clinical observation warrants greater attention from pediatricians, neonatologists and cardiologists due to the timeliness of its presentation and its diagnostic value.

Conflict of interest Arnav Agarwal, Mohammed Firdouse, Claudia Lace De Almeida, and Tapas Mondal declare that they have no conflict of interest.

Ethical standard 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.

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