Cerebral Arteriovenous Malformation Detected by Newborn Congenital Heart Disease Screen with Echocardiography

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INTRODUCTION

The newborn critical congenital heart defect (CCHD) screen has been implemented in the United States since 2011. It prevents newborns with ductal-dependent congenital heart disease from being discharged from the hospital without echocardiography. The "false-positive" rate of the CCHD screen is 0%–1.8%,[1–4] but 27%–80% of these false positives arise from important noncardiac pathology.[1–3,5–7] Cerebral arteriovenous malformations (AVMs) are often not detected on prenatal ultrasound, but early detection improves outcomes.[8,9] We describe a case of cerebral AVM detected by abnormal newborn CCHD screen and echocardiography. This case demonstrates the importance of considering noncardiac pathology in "false-positive" CCHD screens.

CASE PRESENTATION

A 1-day-old term male infant was transferred to our facility after he failed his CCHD screen. He was born with no complications, weighed 3.4 kg (56th percentile), and was breastfeeding well. His gravida 3 para 3 mother received consistent prenatal care, with normal screening ultrasound findings. Family history was negative for congenital heart disease. At the time CCHD screening was performed, the infant reportedly had a soft heart murmur and no other symptoms of structural cardiac disease. Skin and mucus membranes did not appear cyanotic, and four extremity blood pressures were normal.

Per American Academy of Pediatrics guidelines, a newborn passes the CCHD screen if oxygen saturation is ≥95% in the upper and lower extremities, and right upper and lower extremity saturation is within 3 percentage points. A newborn fails the screen if upper or lower extremity saturation is <90% or the difference between right arm and right foot saturation is >3%. The test is repeated three times to ensure accuracy. This patient failed all three tests, with lower extremity saturation <90%, and two of his tests showed differential saturation >3%. Initial echocardiography at the referring facility suggested aortic coarctation, with a large patent ductus arteriosus (PDA) and

small ostium secundum atrial septal defect. Umbilical central lines were placed, alprostadil was started at 0.05 μg/kg/min to maintain patency of the ductus arteriosus, and the infant was intubated for transport by helicopter.

Investigations

Physical examination upon arrival at our institution demonstrated a hyperdynamic precordium with a grade 1/6 short systolic ejection murmur at the upper left sternal border. Echocardiography on day of life 2 showed no evidence of aortic coarctation (Figure 1A). However, there was right ventricular enlargement with supersystemic right ventricular systolic pressure of 76 mm Hg (simultaneous systemic blood pressure 66/40 mm Hg), with dilation of the superior vena cava (Figure 1B) and innominate vein (Figure 1C) and a moderate-sized PDA with bidirectional shunt. The left ventricular ejection fraction was 65%. The finding of innominate vein and superior vena cava dilation raised concern for increased cerebral vascular return. This prompted further imaging through the anterior fontanelle. Color Doppler imaging demonstrated high-velocity continuous cerebral flow concerning for an AVM (Figure 1D). Auscultation over the anterior fontanelle at this time disclosed a prominent bruit. A formal cranial Doppler ultrasound confirmed a vein of Galen malformation (VGAM).

Management

Magnetic resonance imaging and magnetic resonance angiography on day of life 3 revealed a large choroidal type VGAM (Figure 2A). Numerous deep feeding vessels from the thalamic and choroidal system (Figures 2B and 2C) created a fistula into the prosencephalic vein that ultimately drained into a central venous collecting system. Head computed tomography was negative for any calcifications. Calcifications are associated with >30% of VGAMs and suggest severe intracranial hypertension.[8,10] Clinically, the infant had no neurologic deficits and returned to oral feeding after he was extubated. The bruit was still audible through the fontanelle, but now with decreased intensity, suggesting a minimal arteriovenous pressure gradient. Neurosurgery consultation recommended delaying intervention until the infant was older, to decrease risks associated with endovascular embolization of such small vessels. Repeat echocardiography on day of life 4 was unchanged, with no overt heart failure and persistent elevation of right ventricular systolic pressure. Oral furosemide was initiated to reduce risk for volume overload and cerebral hypertension. The infant was monitored closely, with serial echocardiograms every 2 to 3 days. Echocardiographic findings continued to be stable, and he was discharged home on day of life 13, with frequent cardiologic and neurologic outpatient follow-up.
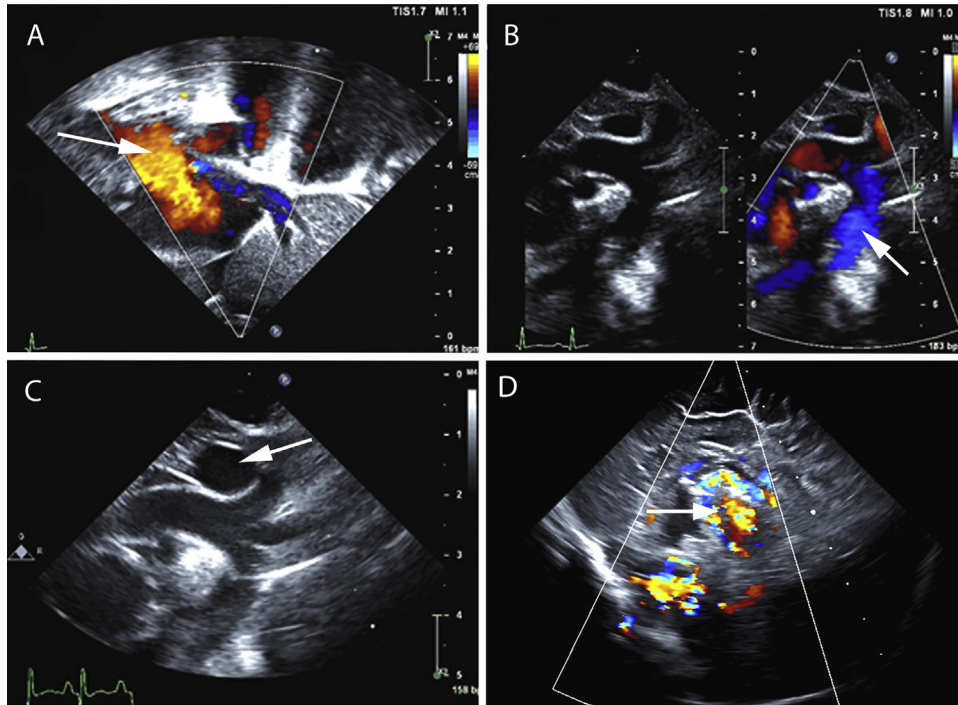
Figure 1 Echocardiogram images. **(A)** Subcostal sagittal plane bicaval view with color Doppler demonstrating increased flow from the superior vena cava (*red flow, arrow*). **(B)** Suprasternal aortic arch view with side-by-side color imaging demonstrating laminar flow and no evidence of aortic coarctation (the *arrow* points to widely patent aortic isthmus at the site of previous concern for coarctation). **(C)** Suprasternal two-dimensional arch view reveals an enlarged innominate vein (*arrow*). **(D)** Cranial imaging through the anterior fontanelle during echocardiography depicts high-velocity, turbulent venous flow in the vein of Galen and draining vessels (*arrow*).
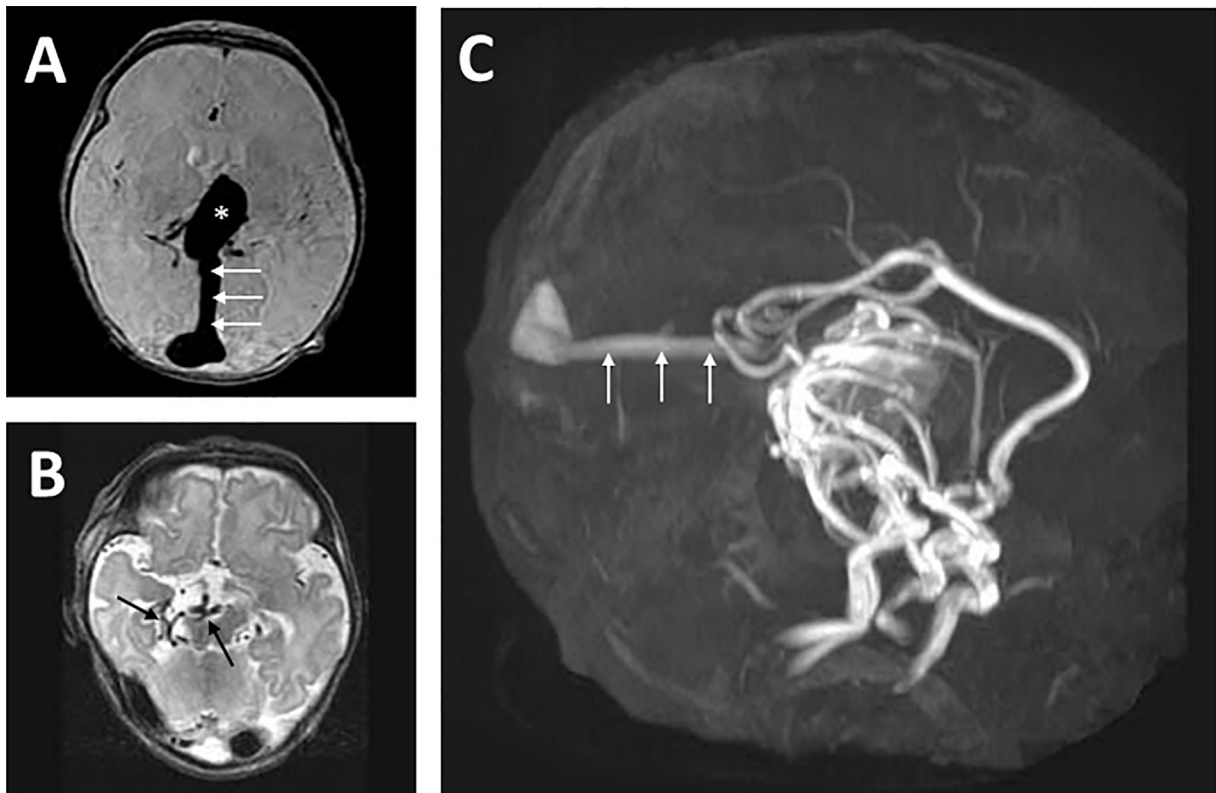
Figure 2 Magnetic resonance images. **(A)** Axial magnetic resonance image demonstrates a dilated prosencephalic vein (*asterisk*) draining into a persistent embryonic falcine sinus (*arrows*). **(B)** Further imaging superior to the AVM reveals several choroidal and thalamic arteries (*arrows*) contributing to the fistula. **(C)** Magnetic resonance angiography illustrates the magnitude of the malformation, with a persistent falcine sinus (*arrows*) and torturous choroidal and thalamic vessels.

Outcome

The patient is now 9 months old and doing well. Growth and development are normal. He underwent two coil embolizations of the VGAM (at 3 and 6 months of age), which were performed at another tertiary children's hospital. Results of cardiovascular examinations remain stable, now with normal pulmonary pressures and no evidence of heart failure. He is not receiving any cardiac medications.

DISCUSSION

To our knowledge, this is the first published case of VGAM detected by CCHD screen. Although a vascular malformation is considered noncardiac pathology, signs suggestive of an AVM may be seen on echocardiography. The fistula created by the malformation lacks capillary connections between the cerebral arteries and veins to reduce blood return to the venous system. This creates a high-velocity shunt that leads to increased cerebral venous return and increased cardiac preload. The dilated superior vena cava, dilated innominate vein, and right ventricular dilation were evidence of high cardiac preload. In addition, there was a small amount of diastolic runoff from the PDA toward the brachiocephalic arteries by color Doppler, potentially indicative of a cerebral shunt. In this case, increased venous return, coupled with elevated pulmonary vascular resistance, likely led to right-sided volume overload and pressure and right-to-left shunting at the level of the atrial septum and the PDA. Right-to-left shunting through the PDA would also explain his differential upper and lower saturation on CCHD screening. AVMs typically do not cause cyanosis and are purely left-to-right shunts, but the early timing of the CCHD screen captured the combination of volume overload to the right side and elevated pulmonary resistance causing right-to-left shunting. This would explain why the patient initially failed his CCHD screen, yet his saturation normalized as the pulmonary resistance dropped.

CCHD screening has been routinely performed in the United States since 2011, and VGAMs are very rare.[9,11] Additionally, this particular case was severe. Smaller cerebral AVMs might not dramatically affect cardiac preload after birth. Most failed CCHD screens result in a primarily cardiac workup. Smaller health care centers do not have pediatric cardiology teams to recognize subtle echocardiographic findings, and this may limit noncardiac diagnoses arising from echocardiographic findings. The ability for smaller centers to follow up positive CCHD screens has been cited as one concern for implementing universal screening.[1,6]

As many as 80% of CCHD "false-positive" screens occur because of important noncardiac pathology.[1–3,5–7] This case highlights the importance of considering working through a broad differential that includes infection, pulmonary, cardiovascular, or neurologic pathology[3,4,12–14] before dismissing positive test results. Although prenatal ultrasound technology continues to improve, noncardiac congenital malformations may be missed using routine screening parameters at 20 weeks. Unfortunately, most VGAMs are first identified when the patient experiences a serious consequence, such as stroke, hydrocephalus, seizures, or heart failure.[15] Early detection in this case provided time for the patient's family and health care team to evaluate multiple treatment approaches and to institute close surveillance for symptoms. Detecting noncardiac pathology is an additional benefit of CCHD screening.

Finally, this case also illustrates the importance of the newborn physical examination. In retrospect, the infant had a hyperdynamic precordium, detection of which should prompt a more thorough cardiovascular evaluation. More important, auscultation of the fontanelle would expose a prominent bruit. Listening for a cranial bruit is a very specific and easily implemented technique that should be part of all neonatal physical examinations. In this case, the "old-fashioned" echocardiographic probe (a stethoscope) could make the diagnosis even before the formal echocardiographic examination.

CONCLUSION

This case illustrates the importance of considering noncardiac causes for failed CCHD screening. Bedside echocardiography is an essential component of the CCHD screening program. Echocardiographers should remember that imaging beyond the chest is needed to explain intracardiac abnormalities. The open fontanelle in newborns is a window to important extracardiac pathology, during both physical examination and imaging.

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