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Case Report

The Eye That Revealed the Hidden Storm: A Neonatal Vein of Galen Aneurysmal Malformation—A Case Report and Comprehensive Literature Review

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Abstract

Background: Vein of Galen aneurysmal malformation (VGAM) is a rare congenital cerebrovascular anomaly that predominantly presents in the neonatal period with cardiovascular compromise rather than primary neurological deficits. Diagnostic delays are common because initial neurological examinations are typically reassuring, while high-output cardiac failure and persistent pulmonary hypertension of the newborn (PPHN) mimic common cardiopulmonary disorders.

Case Presentation: A preterm female neonate born at 36 weeks' gestation presented with escalating respiratory distress, severe PPHN (estimated pulmonary artery systolic pressure [PASP] of 90 mmHg), high-output cardiac failure, and prominent right unilateral proptosis. Fetal ultrasound had previously demonstrated a middle cranial fossa "probable arachnoid cyst". Non-contrast cranial CT and subsequent contrast-enhanced MRI, MRA, and MRV revealed a 4.5 × 4.2 × 5.0 cm right temporo-frontal extra-axial aneurysmal venous sac with arterial supply from external carotid branches and direct drainage into the distal right sigmoid sinus. Repeated neuroimaging abroad confirmed the diagnosis of VGAM.

Management & Outcome: Medical stabilization with nasal continuous mandatory ventilation, sildenafil, and aggressive furosemide diuresis relieved pulmonary venous congestion and enabled successful weaning to room air by day 11 of life. Regional neurointerventional services were unavailable; the infant was discharged on day 12 for planned elective embolization abroad. Staged transarterial embolization (3 sessions) was subsequently performed internationally.

Conclusions: Unilateral proptosis in a neonate with high-output heart failure or refractory PPHN is a critical red flag for dural sinus involvement. Postnatal vascular neuroimaging (MRI/MRA/MRV) should be performed urgently for any antenatally reported intracranial cyst. Geographic gaps in pediatric neurointerventional services remain a major determinant of clinical outcome.

Keywords: *Vein of Galen aneurysmal malformation; Persistent pulmonary hypertension of the newborn; High-output cardiac failure; Neonatal proptosis; Transarterial embolization.*

Introduction

Vein of Galen aneurysmal malformation (VGAM) accounts for approximately 1% of pediatric intracranial vascular anomalies, with an estimated incidence between 1 in 25,000 and 1 in 100,000 live births [2,8,11]. Unlike adult cerebrovascular malformations, VGAM in neonates manifests primarily as a systemic cardiovascular crisis driven by low-resistance arteriovenous shunting rather than focal neurological deficits, seizures, or hemorrhage [4,5,9].

Antenatal ultrasonography frequently misidentifies VGAM as an arachnoid cyst, choroid plexus cyst, or dilated cavum veli interpositi [27,28]. Postnatally, rapid shunting overloads the neonatal myocardium, precipitating PPHN and high-output cardiac failure that closely resemble primary cardiac or pulmonary disease [3,4,17]. Bedside cranial bruits are diagnostic when present, but are documented in only a minority of cases [2,8].

Although advances in neurointerventional radiology and staged transarterial embolization have improved survival rates to over 60% in modern series, outcomes remain heavily contingent upon early diagnostic recognition, effective bridging medical therapy, and rapid access to specialized pediatric neurointerventional centers [6,14,18,19,23,29]. We report a diagnostic case of VGAM presenting with severe PPHN and unilateral proptosis following an antenatal misdiagnosis of an arachnoid cyst, highlighting key pathophysiological mechanisms and structural gaps in regional neurointerventional capacity.

Case Presentation

Patient History & Delivery

A female neonate was delivered at 36 weeks' gestation via elective lower-segment cesarean section. The delivery was indicated by a previous cesarean section and an antenatally detected cystic lesion in the middle cranial fossa (reported differential: probable arachnoid cyst). The mother was a 29-year-old gravida 2, para 1 with an uncomplicated pregnancy and unremarkable antenatal screening.

At birth, Apgar scores were 8 and 9 at 1 and 5 minutes, respectively. Growth parameters were appropriate for gestational age: birth weight 2.615 kg, length 47 cm, and head circumference 35 cm. Physical examination in the neonatal intensive care unit (NICU) demonstrated an active neonate with a soft fontanelle, normal tone and primitive reflexes, mild dysmorphism, high-arched palate, small ankyloglossia, soft cardiac systolic murmur, asymmetric right facial swelling, and mild right ocular proptosis.

CLINICAL TIMELINE

Antenatal (3rd Trim)	Fetal scan: Cystic lesion in middle cranial fossa
Birth (Day 0)	LSCS at 36 wk; Apgars 8/9; right proptosis noted
Day 0 (8 Hours)	Respiratory distress (SpO_2 88-90%); nasal CMV
Day 1 of Life	Echo: PASP 90 mmHg (PPHN), hyperdynamic state Cranial CT: 4.5 x 4.2 x 5.0 cm extra-axial mass
Day 2 of Life	Brain MRI/MRA/MRV: Aneurysmal sac, ECA feeders
Day 4 of Life	Echo: PASP 70 mmHg; Sildenafil initiated
Day 7 of Life	Congestion deterioration; Furosemide started
Day 9-11 of Life	Diuresis success; weaned to room air
Day 12 of Life	Discharged; regional transfer attempts fail
Post-Discharge	Traveled abroad; 3 staged embolizations done

Cardiopulmonary Collapse and Clinical Course

The infant remained stable on room air for the first 8 hours of life before developing progressive tachypnea, subcostal retractions, and desaturations (88–90%). Nasal continuous mandatory ventilation (CMV) with a fraction of inspired oxygen () up to 0.30 was initiated. Chest radiograph demonstrated cardiomegaly and diffuse bilateral pulmonary haziness.

Echocardiography on day 1 revealed severe PPHN with an estimated PASP of 90 mmHg, a large patent ductus arteriosus (PDA), a patent foramen ovale (PFO), hyperdynamic circulation, and preserved biventricular contractility. Repeat echocardiography on day 4 showed persistent severe PPHN (PASP 70 mmHg); oral sildenafil was started. On day 7, escalating oxygen requirements and pulmonary congestion prompted the addition of furosemide. Furosemide diuresis produced rapid clinical improvement, allowing weaning off respiratory support to room air by day 11. Neurological status remained consistently normal throughout the NICU stay.

Diagnostic Imaging

- **Laboratory Investigations:** Septic screens, blood cultures, renal function, liver function, metabolic screens, and newborn screening panels were normal. Neonatal hearing screening yielded a "pass" on the left and a "refer" on the right.
- **Non-Contrast Cranial CT (Day 1):** Identified a well-circumscribed, iso- to hyperdense extra-axial mass in the right temporo-frontal region (). The mass encroached upon the right orbital apex with thinning of the right sphenoid wing, mild right proptosis, effacement of cortical sulci, and right lateral ventricle compression.
- **Brain MRI / MRA / MRV (Day 2):** Confirmed an intensely enhancing extra-axial aneurysmal sac connecting directly to the distal right sigmoid sinus. MRA demonstrated supply from terminal branches of the right external carotid artery (ECA). Flow voids on T2 and FLAIR sequences confirmed high-velocity arteriovenous shunting. A subacute 10-mm right frontoparietal subdural hematoma was noted adjacent to the lesion.

Imaging Modality	Key Radiological Findings	Clinical Significance
Cranial CT (Non-contrast)	4.5×4.2×5.0 hyperdense extra-axial mass; orbital apex thinning.	Excludes simple arachnoid cyst; indicates acute vascular lesion.
Brain MRI (T2 / FLAIR)	Prominent T2/FLAIR flow voids; splaying of right MCA; 10-mm subdural rim.	Confirms high-flow shunt and mass effect on cerebral tissue.
MRA / MRV	ECA arterial feeders; direct drainage into distal right sigmoid sinus.	Establishes angioarchitecture and venous drainage pathway.
Echocardiography	PASP 90 mmHg; PDA/PFO shunting; preserved biventricular function.	Pathognomonic signature of extracardiac AV shunt.

Management & Transfer Attempts

Neurosurgical evaluation confirmed that the lesion was unamenable to direct surgical resection and required staged transarterial embolization at 4 to 5 months of age. Multiple regional tertiary centers declined transfer due to lack of specialized pediatric neurointerventional capacity. International air transport was infeasible due to medical, transport, and financial barriers.

The infant was discharged on day 12 of life on room air following cessation of cardiac medications. The family subsequently traveled abroad, where repeat neuroimaging formally established the diagnosis of VGAM. Three staged transarterial embolization sessions were successfully performed. The patient was subsequently lost to follow-up, leaving long-term neurodevelopmental outcomes unknown.

Radiological Gallery

Cranial Computed Tomography

Figure 1. Non-contrast cranial CT on day 1 of life. Axial section shows a large, well-circumscribed, iso- to hyperdense right temporo-frontal extra-axial mass () compressing the right lateral ventricle and effacing local sulci. Thinning of the greater wing of the sphenoid and orbital apex encroachment are visible.

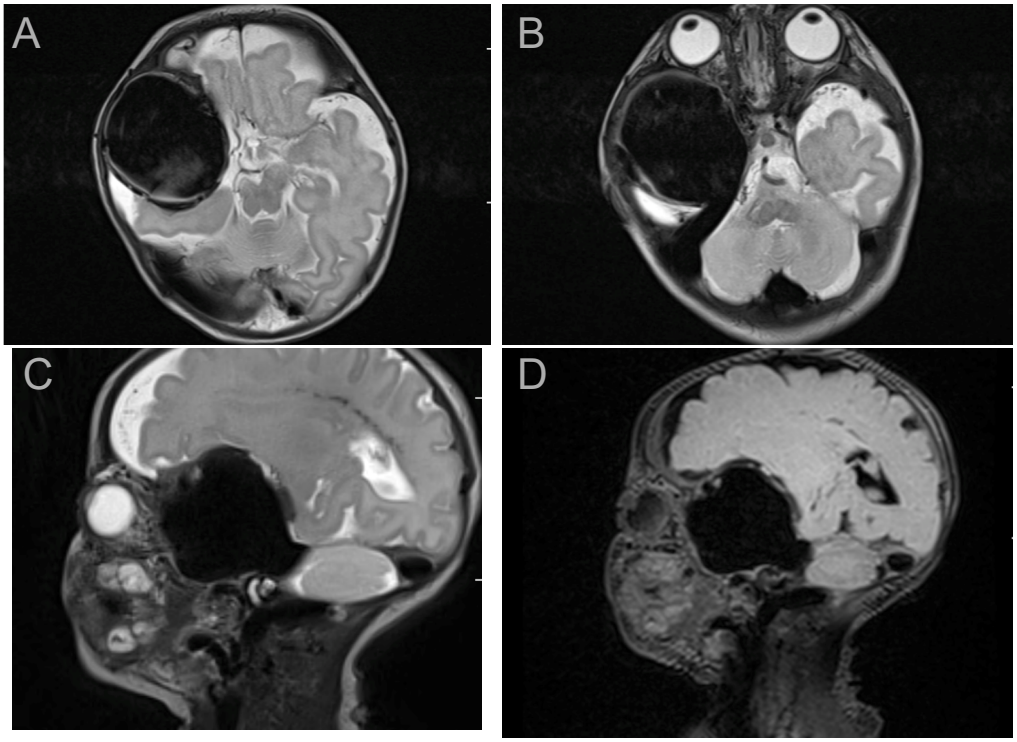
Magnetic Resonance Imaging

Figure 2. Brain MRI on day 2 of life. (A) Axial T2-weighted sequence displaying marked flow voids within the aneurysmal sac. **(B)** Axial T2 section at orbital level demonstrating right proptosis. **(C)** Sagittal T2 and **(D)** Sagittal FLAIR sequences illustrating craniocaudal extent and perilesional vascular collaterals.

Angiography & Venography

Figure 3. MRA and MRV angioarchitecture. (A) Sagittal MRA MIP showing terminal right ECA arterial feeders. **(B)** Coronal MRV MIP confirming direct drainage into the distal right sigmoid sinus. **(C)** Axial MRA/MRV MIP demonstrating dynamic shunt flow.

Figure 1. Non-contrast cranial CT on day 1 of life.



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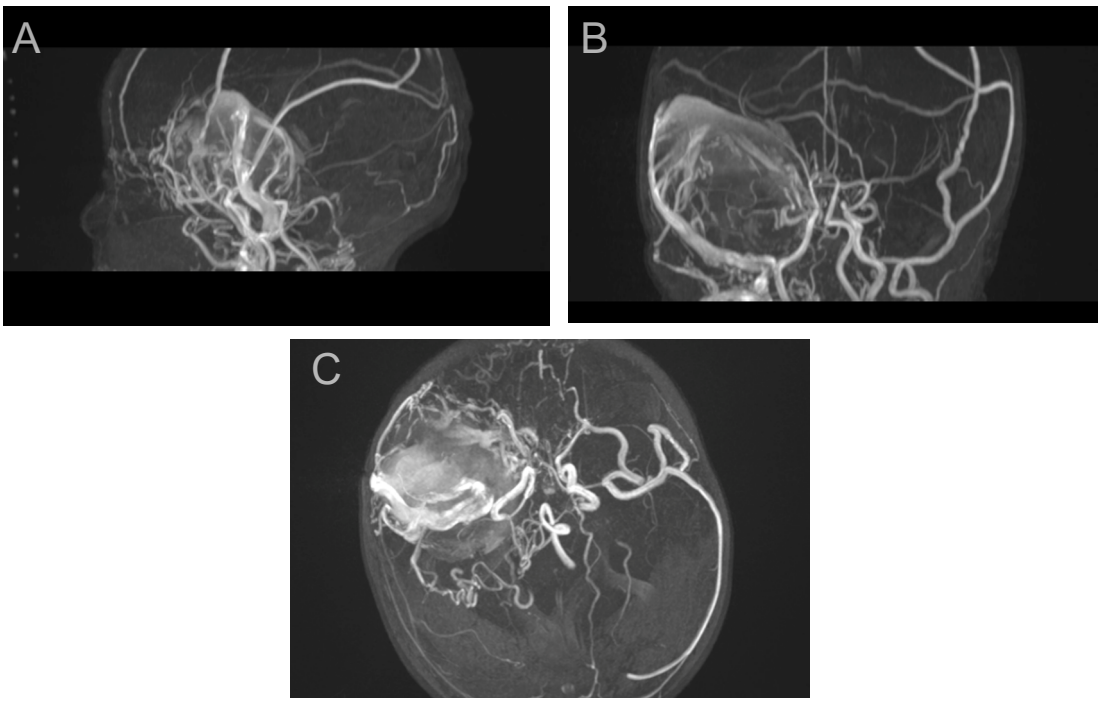


Figure 3. MRA and MRV angioarchitecture. (A) Sagittal MRA MIP showing terminal right ECA arterial feeders. **(B)** Coronal MRV MIP confirming direct drainage into the distal right sigmoid sinus. **(C)** Axial MRA/MRV MIP demonstrating dynamic shunt flow.

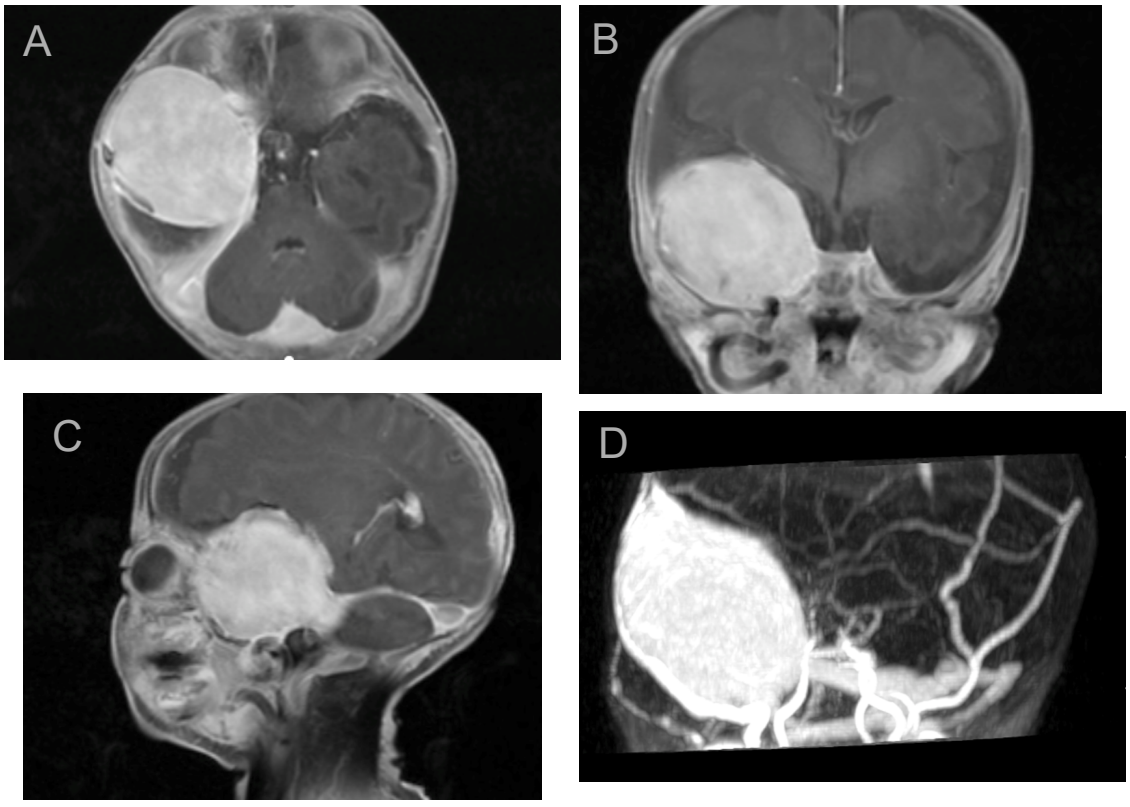


Figure 4. Contrast-enhanced T1 MRI and 3D volume rendering. (A) Axial. **(B)** Coronal. **(C)** Sagittal post-contrast T1 images displaying intense, uniform enhancement of the venous sac. **(D)** 3D volume-rendered reconstruction illustrating spatial relationships with skull base vasculature

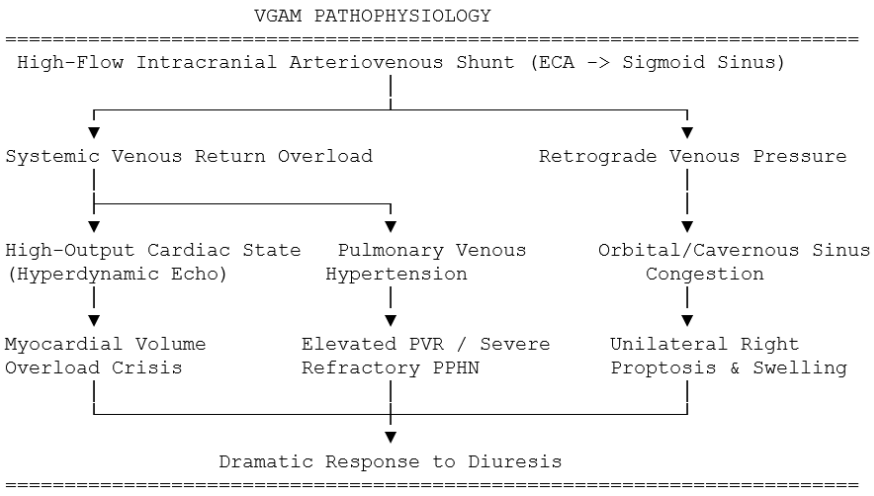
Post-Contrast Studies

Figure 4. Contrast-enhanced T1 MRI and 3D volume rendering. (A) Axial, **(B)** Coronal, and **(C)** Sagittal post-contrast T1 images displaying intense, uniform enhancement of the venous sac. **(D)** 3D volume-rendered reconstruction illustrating spatial relationships with skull base vasculature.

Discussion

Pathophysiologic Integration

The systemic manifestations of VGAM stem directly from low-resistance arteriovenous shunting that diverts systemic cardiac output into the low-pressure venous circuit. [1,5,11,24].



Persistent Pulmonary Hypertension (PPHN): Shunt flow markedly increases venous return to the right heart, exposing the transitioning neonatal pulmonary vascular bed to excessive volume and flow. Secondary pulmonary venous hypertension prevents normal postnatal vasodilation, resulting in elevated pulmonary vascular resistance (PVR), severe pulmonary artery hypertension (PASP 90 mmHg), and persistent right-to-left shunting through the PDA and PFO [3,17]. Diuretic administration (furosemide) reduces pulmonary venous congestion, breaking the cycle of elevated PVR and enabling respiratory weaning.

High-Output Cardiac Failure: Neonatal myocardium operates near maximal capacity on the Frank-Starling curve and possesses limited compliance . The sustained volume load from the shunt exhausts cardiac reserve. Preserved biventricular contractility alongside hyperdynamic flow is the classic echocardiographic hallmark of an extracardiac vascular shunt [4,5,9].

Orbital Proptosis & Facial Swelling: Drainage into the right sigmoid sinus raises venous pressures within connected cavernous and sphenoparietal channels. Retrograde venous stasis transmitted to ophthalmic veins leads to orbital soft tissue edema, venous plexus engorgement, and unilateral proptosis [30].

Embryological Context & Classification

During early embryonic development (6–11 weeks' gestation), deep venous drainage is handled by the transient median prosencephalic vein of Markowski. Failure of this embryonic vein to involute due to direct arterial fistulous connections results in VGAM [10,11]. The Lasjaunias classification divides VGAM into choroidal (multiple feeders, severe early neonatal cardiac failure) and mural (few fistulous connections, late infant presentation with macrocephaly) subtypes [1,6,10].

The present case displayed an atypical right temporo-frontal extra-axial location draining directly into the distal sigmoid sinus. This anatomy lies within the spectrum of VGAM variants or dural sinus malformations with secondary venous aneurysms, sharing an identical embryological basis and management approach [32,38,39].

Differential Diagnosis

In a neonate presenting with cardiac failure, pulmonary hypertension, and an extra-axial mass:

Arachnoid Cyst: Non-vascular structure; does not enhance or produce flow voids, cardiac failure, or proptosis [12,13].

Dural Sinus Malformation (DSM) with Venous Aneurysm: Involves lateral/dural sinuses; presents with identical high-output cardiac failure and orbital venous congestion.

Pial Arteriovenous Malformation: Typically intraparenchymal rather than extra-axial, draining primarily into cortical veins.

Congenital Brain Tumors (e.g., Teratoma, Choroid Plexus Papilloma): Produce mass effect and hydrocephalus, but lack high-velocity flow voids and hyperdynamic AV shunt physiology [12,13].

Clinical Pearls: Diagnostic Framework

To avoid diagnostic delays, clinicians should apply five core pearls when evaluating unexplained neonatal cardiorespiratory distress:

1. Refractory Cardiac Failure with Normal Contractility: Unexplained cardiomegaly and hyperdynamic cardiac function indicate an extracardiac shunt [4,5,9].
2. Disproportionate PPHN: Severe PPHN despite minimal parenchymal lung disease should prompt cranial and abdominal vascular imaging [3,17].
3. Unilateral Proptosis or Facial Swelling: A critical red flag pointing toward intracranial dural sinus hypertension and retrobulbar venous stasis [30].
4. Antenatal "Arachnoid Cyst": Any fetal intracranial cystic lesion requires postnatal cross-sectional vascular imaging (MRI/MRA/MRV) [26].
5. Cranial Bruit or Macrocephaly: Auscultation over the fontanelle or temporal regions can reveal pathognomonic continuous vascular bruits [2,8].

Modern Management & Outcome Predictors

Definitive management relies on staged transarterial embolization using liquid embolic agents (N-butyl cyanoacrylate [NBCA] or Onyx) [14,22]. Triage is guided by the Bicêtre Neonatal Evaluation Score (0–21 scale based on cardiac, cerebral, hepatic, respiratory, and renal function) [6,20]:

Score < 8: Severe multiorgan failure; high mortality; indicates emergency embolization.

Score 8–12: Urgent embolization within the first weeks of life.

Score > 12: Stable on medical management; elective staged embolization deferred to 4–5 months of age.

Our patient achieved stabilization (Bicêtre score > 12), enabling safe deferral to elective embolization. Modern series report favorable neurodevelopmental outcomes in 60–70% of treated infants [18,19,23,29].

Learning Points / Key Messages

- **Postnatal Vascular Imaging Mandatory:** Any antenatally reported middle or posterior cranial fossa "cyst" requires early postnatal MRI/MRA/MRV to rule out high-flow vascular malformations.
- **Orbital Red Flags:** Unilateral proptosis in a neonate with high-output cardiac failure is a powerful pointer to dural sinus malformation and orbital venous engorgement.
- **Echocardiographic Fingerprint:** Severe PPHN combined with preserved biventricular contractility and a hyperdynamic state should immediately trigger cranial ultrasound with color Doppler.
- **Diuretic Bridging:** Aggressive furosemide therapy relieves pulmonary venous congestion, providing an effective physiological bridge to elective endovascular treatment.
- **Health System Infrastructure:** Developing regional pediatric neurointerventional networks is essential to reduce disparities in access and prevent lost follow-up in rare cerebrovascular diseases.

Conflict of Interest

The authors declares no conflict of interest.

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