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Case Report

Neonatal Presentation of Vein of Galen Aneurysmal Malformation: A Case Report

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Abstract

Background: Vein of Galen Aneurysmal Malformation (VGAM) is a rare vascular malformation that is commonly diagnosed prenatally. Given the requirement of complex neurosurgical interventions and poor morbidity associated with most cases, adequate preparation is the key. Non-specific manifestations as the initial presentation can cause a delay in making the diagnosis.

Case Presentation: We present a case of a choroidal type of VGAM diagnosed after birth in a neonate who rapidly progressed to cardiovascular decompensation despite timely recognition and management in a level IV neonatal intensive care unit (NICU).

Conclusion: There is a paucity of literature on neonatal presentation of VGAM without prior diagnosis and the present case highlights the necessity of having a high index of suspicion in order to diagnose and treat the patient effectively.

Keywords: *Vein, Galen, Intracranial, Malformation, Case, Report*

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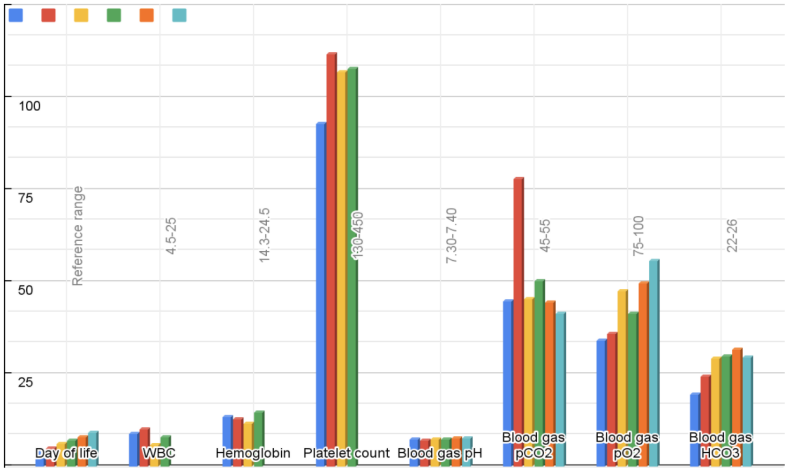
Background

Vein of Galen Malformation is a rare intracranial arteriovenous (AV) malformation (16) with an incidence of 1 in 25000[1]. Although rare, they represent approximately 30% of pediatric vascular malformations.[2] They are distinguished by the shunting of arterial flow into a dilated cerebral vein usually dorsal to the cerebral aqueduct at the tectum. It can present both in fetal and postnatal periods with variable clinical manifestations. Prenatal detection occurs in the third trimester by ultrasound, with better delineation identified by fetal magnetic resonance imaging (MRI). In the neonate and young child with a high degree of suspicion, diagnosis can occur with a range of modalities such as head ultrasounds to more sophisticated imaging such as MRI, magnetic resonance angiography (MRA), and magnetic resonance venography (MRV). Management of VGAM usually involves a multidisciplinary approach with many new endovascular therapies available. Prognosis usually depends on the age of onset and clinical presentation. The Bicetre score can be used to stratify the risk and benefits of endovascular approaches to embolization. The total score range is 0 to 21. A score of <8/21 means fatal prognosis and recommend no treatment, 8-12 needs emergent embolization, and >12 needs embolization at 5 months of age with continued medical management (1).

Case Presentation

A male infant was born at 37 weeks gestation to a 32-year-old G1P0 mother with limited prenatal care. Her pregnancy was complicated by pre-eclampsia and the initial urine drug screen was positive for marijuana. The mother's GBS status was unknown; antibody screen and infection panel were negative and prenatal ultrasound was unremarkable including an anatomy scan. He was born via induced vaginal delivery without any complications, with a birth weight appropriate for gestational age (AGA) at 2.71 kg (30th percentile) and a head circumference of 32cm (18th percentile). Apgar scores were 9 at 1 minute and 9 at 5 minutes. He was found to have a holosystolic murmur on the first day of life, otherwise, his physical exam was unremarkable. It was presumed to be a closing patent ductus arteriosus (PDA) or a peripheral pulmonary stenosis (PPS) murmur, both of which are not pathological in an otherwise stable neonate with normal vital signs who was feeding well, voiding, and active. However, over the next three days, he had audible breathing with subcostal retractions, poor latching, and low urine output prompting a sepsis workup and empiric antibiotic coverage with ampicillin and gentamicin. A chest radiograph revealed cardiomegaly. Feedings were held, and he was placed on a 2L high flow nasal cannula during transfer to a Level II NICU for hypoxia, feeding intolerance, and suspected sepsis. An echocardiogram was suggestive of right heart failure, tricuspid regurgitation, and persistent pulmonary hypertension. A summary of serum lab values in a graphical representation in Table 1.

Table 1| Graphical representation of the laboratory values.



Given this progressive worsening of his clinical status, he was transferred to our tertiary care center's Level IV NICU for cardiology evaluation on Day 4 of life. During transport, he was stabilized on a 2L high flow nasal cannula for mild tachypnea. After admission, he rapidly escalated to non-invasive ventilation for a few hours, then required intubation and ventilator support for worsening respiratory status. Over this time, he had reduced urine output and gentamicin was replaced with cefepime due to concern for acute kidney injury. Subsequent physical examination was significant for hepatomegaly. Blood gas, complete blood count, electrolytes, and urinalysis identified normal anion gap metabolic acidosis, elevated lactate, thrombocytopenia, mild hyponatremia, and trace blood with the hyaline cast on urinalysis. An abdominal ultrasound (US) showed non-specific changes which may relate to renal sinus fat or Tamm-Horsfall proteins.

A repeat echocardiogram was consistent with pulmonary hypertension, with high right ventricular systolic pressure (RVSP), and there was a worsening decrease in left ventricle (LV) function with aortic valve (AV) regurgitation. Physical exam was now notable for a large anterior fontanelle, and despite a change in measured head circumference, this prompted the head US which revealed an arteriovenous malformation on the fifth day of life.

Since the cause of the congestive heart failure was not apparent from the multiple echocardiograms, other differentials like renal failure and arteriovenous malformations were investigated in our patient. Sepsis resulting in shock was also considered. However, with progressive fontanelle bulge, a head ultrasound and subsequent MRI/MRA/MRVs determined arteriovenous malformations as the inciting cause for the volume overload resulting in cardiac failure.

Mildly prominent ventricles with possible intraventricular hemorrhage versus significantly enlarged choroid plexus were seen on the head US. Subsequently, MRI, MRA, and MRV confirmed enlarged (1.5 cm width) abnormal venous varix which was draining into the embryonic falcine sinus with associated multiple abnormal filling arteries consistent with an arteriovenous fistula between deep choroidal arteries and significantly enlarged embryonic median prosencephalic vein. Figure-1(A-F) depicts various magnetic resonance images delineating the patient's VGAM.

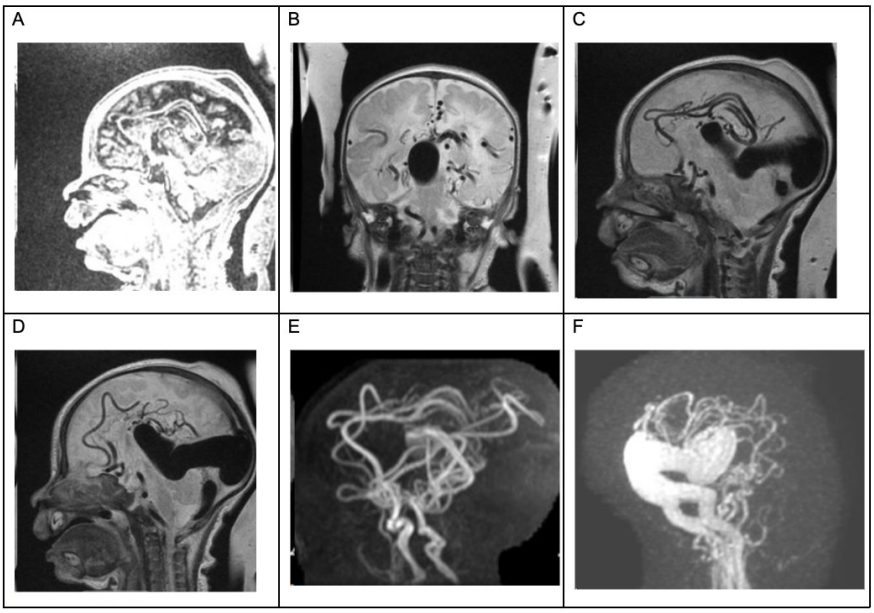


Figure 1. Magnetic resonance images:

A. Sagittal view of the brain MRI showing the AV Malformation in the cerebral hemispheres.
B. coronal view of the brain MRI showing multiple large AV malformations in bilateral cerebral hemispheres
C. Sagittal view of the brain MRI showing contiguous vein of Galen malformation and enlarged vein involving the occipital lobe
D. Sagittal view of the brain MRI showing the draining area of the vein of Galen
E. MRA/MRV image of the Arteriovenous malformation
F. MRA/MRV sagittal image of the enlarged Vein of Galen

There were additional findings of profound white matter necrosis and parenchymal petechial hemorrhages. EEG monitoring showed no epileptogenic waveform. During his course of stay, he persistently had multiple episodes of supraventricular tachycardia (SVT) which did not respond to vagal maneuver but did respond to IV Adenosine.

A family meeting with the team was held and long-term outcomes were discussed. The palliative care team was involved in providing the parents with adequate support, education, and resources. On reviewing his physical examination, labs, imaging, and Bicêtre score of <8 due to multisystem involvement, it was inferred that available interventions would not provide a satisfactory quality of life. After counseling and education, his parents opted for comfort care measures. After compassionate extubation, he passed away due to cardiorespiratory failure within 4 hours and 18 minutes.

Discussion and Conclusions

Vein of Galen aneurysmal malformation is a rare intracranial vascular anomaly with high morbidity. By the 11th week of intrauterine life, the median prosencephalic vein regresses to form the two internal cerebral veins and the vein of Galen. Vein of Galen Malformations encompasses a rare disorder with a spectrum of congenital arteriovenous malformations all of which have a common dilated venous pouch usually from the persistence of the fetal median prosencephalic vein of Markowski.[3,4]

Multiple classification systems categorize VGAM and the most widely cited are the Yasargil and Lasjaunias classifications. The simpler Lasjaunias classification[5] has 2 types, Type I or choroidal type which consists of direct, high-flow shunts located within the wall of the venous aneurysm, and Type II or mural type which involves interposition of an arterial network between the feeders and the venous aneurysm. MRI/MRA/MRV showed a choroidal type of VGAM in our case. Yasargil[6] classified VGAM into four categories: Type I is a pure AV fistula between the arteries and the vein of Galen with an ampulla of the vein of Galen. Type II has a thalamo perforator that travels partly intrinsic and traverses through the brain parenchyma to form a fistula with the vein of Galen extrinsically. Type III is a mixed form involving both types I and II. Type IV consists of proximal AVM with draining veins that drain into the vein of Galen. Amacher and Shillito[7] categorized the age and clinical presentation. Group I include neonates with severe high-output cardiac failure and cranial bruit. Group II consists of neonates with mild heart failure and macrocephaly. Group III includes infants with macrocephaly and cranial bruit without cardiac presentation. Group IV largely consists of older children presenting after 3.5 years with headaches, exertional syncope, and calcified mass in the pineal region.

Evaluation of VGAM in utero and in neonates is predominantly via ultrasound.[8] Additional use of color doppler provides adequate information about patency and the vessels that are feeding and draining the AVM. On computed tomography, VGAM appears as a rounded mass often displacing adjacent ventricles. We used MRI/MRA/MRV to confirm the diagnosis after a positive head US. The gold standard diagnostic test is angiography which is often used while treating the AVM with coil embolization.[7,9,10] However, if diffuse parenchymal brain injury or hemorrhage is seen on fetal imaging, comfort care rather than embolization can be recommended.[11]

Clinical presentation ranges from intrauterine to late neonate and sometimes even in childhood and depends often on the type of VGAM. The typical pathophysiological consequence that increases morbidity and mortality is high-output cardiac failure followed by neurological symptoms subsequent to increased cerebral venous congestion and abnormal cerebrospinal flow leading to hydrocephalus. The spectrum of severity is variable due to age at presentation and the differential angioarchitecture of the VGAM. Newborns usually present with signs of congestive heart failure with respiratory distress and cyanosis often mimicking other common presentations of sepsis or pneumonia. Hydrocephalus can ensue with macrocephaly, prominent scalp veins, and sunset eyes. In older children with chronic states, signs of developmental delay may be seen.[12] Headaches and convulsions may also occur.

In the neonatal period, the clinical presentation often depends on the size of the shunt. [9,13,14] A smaller shunt has mild to no cardiac symptoms and the only presentation is macrocephaly and hydrocephalus[14]. Most infants with bigger shunts usually have cardiovascular and respiratory distress due to the AV shunt[3] as seen in our patient. Our initial differential diagnoses were wide and predominantly focused on structural congenital cardiac anomalies, myocarditis, cardiomyopathies, conductive heart disorders, and toxins resulting in congestive heart failure.

The low vascular resistance of the AV shunts in the brain leads to increased venous shunting to the right atrium causing pulmonary hypertension, subsequently increasing preload, and eventually leading to congestive heart failure. This becomes apparent after birth as there are two competing low vascular resistance systems in-utero, namely the placental circulation and this VGAM.[13] At birth, with the removal of the placental circulation, the VGAM has increased flow leading to overt symptoms of heart failure.

Similarly, our patient presented with heart failure on arrival at our tertiary care center. Increased cardiac demand and decreased diastolic flow increase the risk of myocardial ischemia. Pulmonary hypertension favors right to left shunting increasing cyanosis at presentation. Additionally, this can lead to renal hypoperfusion resulting in renal failure. Ultimately, all these lead to multiorgan failure and result in a poor prognosis. Due to the tremendous blood flow via the fistula, there is a 'steal' phenomenon from the cerebral parenchyma leading to brain ischemia and stroke. Additionally, the altered hemodynamics and angioarchitecture result in high cerebral venous pressure and cerebral edema altogether resulting in rapid brain parenchymal loss, developmental delay, and regression which in its most severe neonatal form is called 'Melting Brain'.[15] The increased venous pressure also leads to the development of hydrocephalus which can cause enlargement of the fontanelle as seen in our patient on physical exam.

Management of VGAM is usually with a multidisciplinary team consisting of pediatric neurosurgeons and neuro-interventional radiologists, neonatologists, and developmental follow-ups as needed by pediatricians. When infants present with severe cardiac failure, staged embolization is the best approach to avoid complications of decompensated cardiac failure[16].

Neurosurgery and gamma knife procedures are reserved for lesions not amenable to the endovascular approach.[3,5,14] The prognosis of this rare disorder often rests on the age of presentation as well as the severity. When VGAM is identified on prenatal ultrasound or at birth, it is often more severe with higher chances of heart failure and subsequent neurological impairment as we saw in our patient, this resulted in a poor and fatal outcome. Additional risk stratification can be made by measuring the maximal mediolateral diameter and cross-sectional area at the narrowest point of the straight or falcine sinus. [11]

Given the complexity and high morbidity of VGAM, timely diagnosis and management remain vital to optimal patient management. A normal physical examination of the fontanelles does not exclude VGAM and there needs to be a high degree of suspicion in any neonate presenting with worsening cardiac failure and pulmonary hypertension with early imaging and transfer to a tertiary care center for interdisciplinary management.

Abbreviations

Vein of Galen Aneurysmal Malformation (VGAM), arteriovenous (AV), neonatal intensive care unit (NICU), magnetic resonance imaging (MRI), magnetic resonance angiography (MRA), and magnetic resonance venography (MRV), appropriate for gestational age (AGA), patent ductus arteriosus (PDA) or peripheral pulmonary stenosis (PPS), ultrasound (US), right ventricular systolic pressure (RVSP), left ventricle (LV), aortic valve, supraventricular tachycardia (SVT).

Availability of data and materials

The data used to support the findings of this study are available from the corresponding author upon request.

Consent to publish

Written consent has been obtained from the parents of the child.

Declarations

Ethics approval and consent to participate: Not applicable.

No funding was obtained for this study.

Conflicts of Interest: No potential conflicts of interest relevant to this article are reported.

Authors' Contributions: AS, MB, and NF conceptualized the report. AS and MB curated patient data. NF carried out project administration, and AS and MB wrote the original draft of the manuscript which was reviewed and revised by NF and LA.

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