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Multidisciplinary care of fetal vein of Galen arteriovenous malformation diagnosed via Doppler ultrasound and magnetic resonance imaging: A case report

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ABSTRACT

Fetal vein of Galen malformation (VOGM) represents a rare congenital anomaly affecting the fetal cerebral vasculature. A 27-year-old Middle Eastern woman was referred due to intrauterine growth restriction (IUGR) and fetal cardiac anomalies identified at 35 weeks of gestation. The diagnosis of fetal VOGM with dilated neck vessels was established through a combination of color Doppler ultrasonography and magnetic resonance imaging. A multidisciplinary approach involving maternal-fetal medicine, neonatology, pediatric cardiology, and interventional radiology was implemented. Given the grave prognosis for the baby, the patient received comprehensive counseling. Subsequent monitoring revealed a non-reassuring fetal heart trace, prompting the decision to perform a cesarean section. The newborn, a girl, was admitted to the neonatal intensive care unit for further management but she died shortly thereafter, with heart failure and intracranial hemorrhage identified as the probable causes of death. In summary, the diagnosis and management of VOGMs demand specialized expertise and a collaborative, multidisciplinary approach to optimize patient care and outcomes.

1. Introduction

Fetal vein of Galen malformation (VOGM) is a rare congenital abnormality of the fetal cerebral vasculature [1,2] that accounts for roughly 1% of all fetal intracranial malformations [3].

From an embryological perspective, the pathophysiology of VOGM is not fully delineated. Nonetheless, the current theory suggests that VOGM takes place during the 6–11th weeks of gestation. This arteriovenous malformation is the consequence of a persistent link between the embryonic choroidal blood vessels and the prosencephalic median vein of Markowski. The lack of regression of this vascular link during embryological development culminates in the emergence of several irregular arteriovenous shunts and the appearance of the vein of Galen [4].

Clinically, fetal VOGM is diagnosed during late pregnancy, most commonly during the third trimester [5]. Nevertheless, diagnosis at earlier time points has been also reported [6]. Fetal brain injury, in the form of intracranial cysts, is a common clinical feature of VOGM at the time of clinical diagnosis [7,8].

Prenatal diagnosis of fetal VOGM can be established through a wide array of imaging modalities. The most common modality is 2D real-time ultrasonography with or without color Doppler [9]. Additionally, magnetic resistance imaging (MRI) can also be employed to examine the extent of intracranial damage [9,10].

Herein, we a present case of fetal VOGM that was diagnosed during the 35th week of gestation, utilizing a combination of ultrasonography with color Doppler and MRI. Moreover, we provide a brief review on the prognosis and management of fetal VOGM.

2. Case Presentation

A 27-year-old Middle Eastern woman, G3P2, was referred to hospital for intrauterine growth restriction (IUGR) with fetal cardiac anomalies. She was estimated to be at 35 weeks of gestation, with a spontaneous singleton pregnancy. Fetal assessment revealed VOGM. She had no previous history of any illnesses or operations. She had had two previous uneventful full-term vaginal deliveries. Her living children were thriving and met all proper milestones. The patient's prenatal care was uneventful. There was no other significant medical or family history.

Her body weight was 47.4 kg, her height 148.4 cm, and her body

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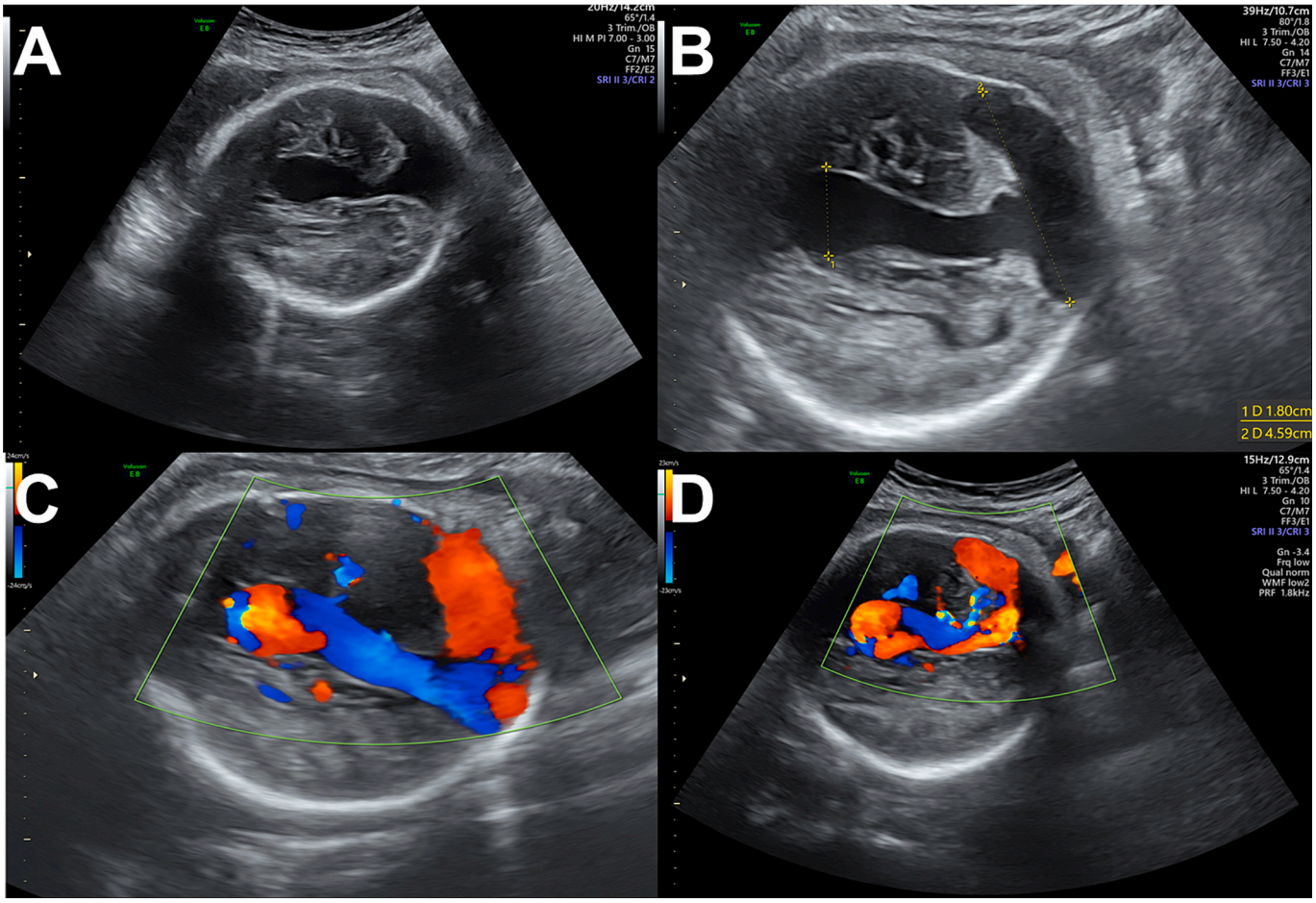

Fig. 1. Axial two-dimensional ultrasound image of the affected fetus with vein of Galen arteriovenous malformation at a gestational age of 35 weeks. A: A large thin-walled anechoic area next to the midline of the brain. B: Marked dilatation of the fetal brain vasculature measuring 1.8 cm in the anterior segment and 4.7 cm in the posterior segment. C and D: Color Doppler shows a full blood flow signal in the brain's anechoic area.

mass index 21.5 kg/m^2. On general examination, the patient did not have apparent features of anemia or jaundice. Abdominal examination demonstrated a uterine fundal height consistent with 30 weeks of gestation with positive fetal heart sound.

Laboratory investigations comprising complete blood count, hepatic, and renal functions were unremarkable. Urine culture results and the serological TORCH infection screening were negative.

An obstetric ultrasound exam at a gestational age of 35 weeks demonstrated a single viable fetus, cephalic presentation, adequate amniotic fluid, and an estimated fetal weight around 1544 g which was equivalent to second percentile. The umbilical artery Doppler ultrasound scan was normal. There was marked dilatation of the fetal brain vasculature, measuring 1.8 cm in the anterior segment and 4.7 cm in the posterior segment (Fig. 1). In addition, the fetal neck vessels were dilated, measuring 1.09 cm (Fig. 2).

Fetal MRI confirmed the diagnosis of VOGM, demonstrating a persistent median prosencephalic vein, which measured up to 1.8 cm, supplied by choroidal arteries and draining into the straight sinus (Fig. 3). There were markedly dilated bilateral transverse sinuses, in addition to distended jugular veins and superior vena cava (Fig. 4). The cerebellum was mildly compressed by the adjacent dilated deep veins. There was evidence of cardiomegaly with pericardial effusion.

A fetal echocardiogram showed cardiomegaly with an abnormal axis of 90 degrees. There was membrane in the left atrium as a supra-mitral ring versus cor triatriatum; however, it was not causing any obstruction.

A multidisciplinary approach with maternal-fetal medicine, neonatology, pediatric cardiology, and intervention radiology was adopted. The patient was counseled regarding the very poor prognosis for this baby. It was noticed that there was a non-reassuring fetal heart trace and consequently cesarean section was performed. A baby girl was delivered, cord pH 7.2, and weight 1.7 kg. Apgar scores were 2 at 1 min, 4 at 5 min, and 7 at 10 min. The neonate was admitted to the neonatal intensive care unit for further management but died shortly after. The most likely cause of death was heart failure and intracranial hemorrhage.

3. Discussion

From a genetic perspective, it is reported that around 10–30% of fetal VOGM cases involve a mutation involving the Ephrin type-B receptor 4 (EPHB4) gene [11,12]. In the present case, genetic testing was not done, and the mother had no risk factors for the pregnancy. Moreover, the fetal cardiomegaly could have been a potential predisposing factor contributing to the non-reassuring fetal heart trace and the performance of an emergency cesarean section.

Prenatal diagnosis of VOGM is increasingly possible, largely owing to the wide array of fetal imaging modalities [13]. Ultrasonography with color Doppler is the mainstay diagnostic modality [9]. MRI plays an important role in delineating the extent of intracranial damage [9,10].

Complications of fetal VOGM include the backlog of blood overload that manifests in the form of distended brain ventricles, dilated brain sinuses, engorged superior vena cava vessels, and distended neck vasculatures [14]. In the present case, a fetal echocardiogram was performed to better gauge the cardiovascular function.

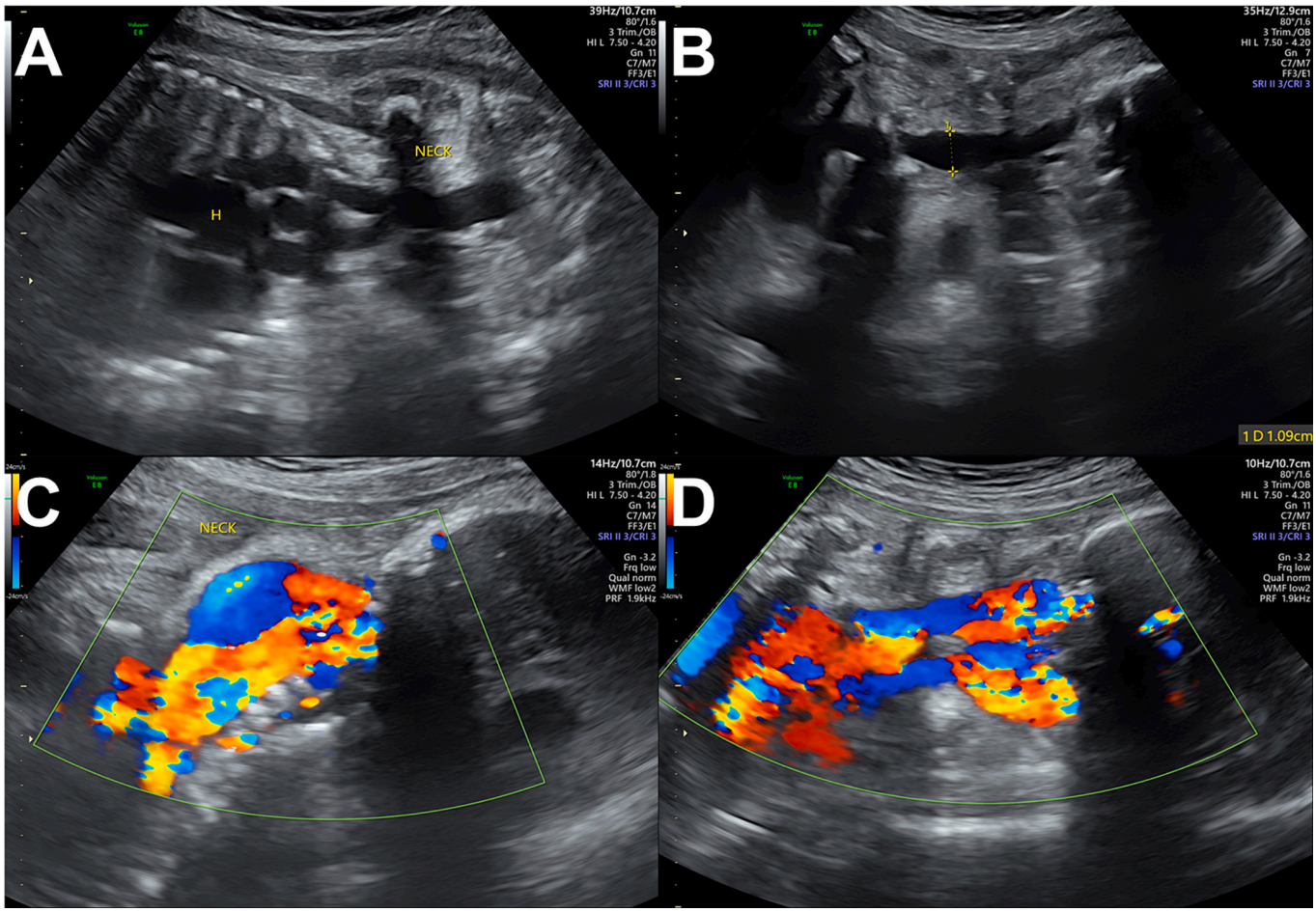

Fig. 2. Sagittal two-dimensional ultrasound view of the fetal neck showing dilated fetal neck vessels at a gestational age of 35 weeks. A and B: the fetal neck vessels dilated, measuring 1.09 cm. C and D: Color Doppler shows a full blood flow signal in the neck area.

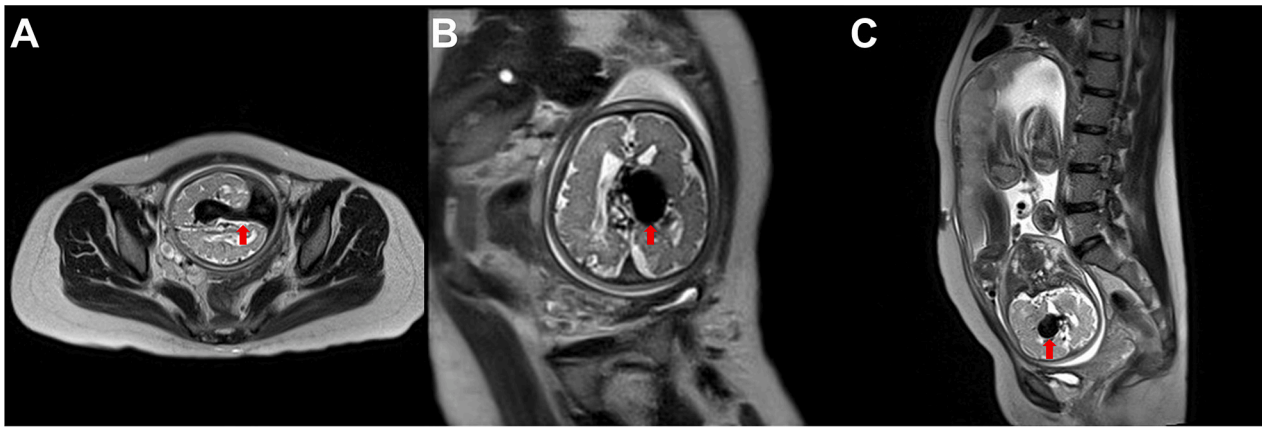

Fig. 3. Magnetic resonance image of the affected fetus with vein of Galen arteriovenous malformation at a gestational age of 35 weeks. A and B: In this coronal view of the head, the vein of Galen is dilated, measuring 1.8 cm in maximum diameter (red arrow). C: This sagittal image shows the dilated vein of Galen (red arrow). (For interpretation of the references to color in this figure legend, the reader is referred to the web version of this article.)

The prenatal diagnosis of VOGM is not an indication for offering termination of pregnancy. A retrospective cohort investigation of 21 prenatally diagnosed cases of VOGM revealed that nearly 43% of mothers elected to terminate pregnancy ($n = 9$) and roughly 29% of cases ($n = 6$) culminated in neonatal death. Only six cases (29%) culminated in live births, three of which incurred abnormal neurological development [15]. In a retrospective multi-center study of 49 cases of fetal VOGMs, 47% of mothers opted for pregnancy termination [16]. In the present case report, the decision to terminate pregnancy was not made; however, preterm delivery occurred.

Large-sized cerebral damage, tricuspid regurgitation, and VOGM volume $\geq$ 20,000 mm^3 have been identified as substantive prenatal factors linked to dismal prognosis among patients with fetal VOGM [16]. A recent systematic review and meta-analysis of 11 studies comprising a

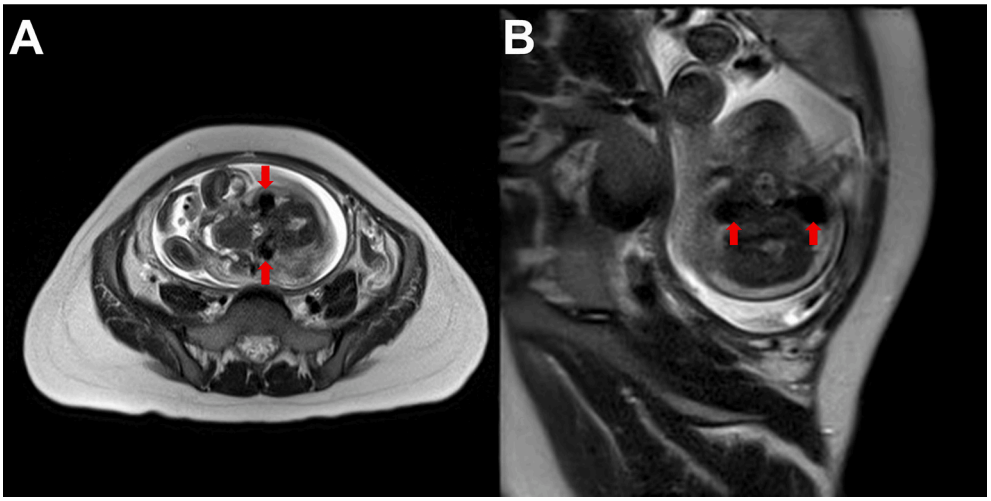

Fig. 4. Magnetic resonance image of fetal neck showing dilated fetal neck vessels at a gestational age of 35 weeks. A and B: In this coronal view of the neck, there are markedly dilated bilateral jugular veins (red arrow). (For interpretation of the references to color in this figure legend, the reader is referred to the web version of this article.)

total of 226 cases with an antenatal diagnosis of VOGM revealed hydrops presented in 7% of cases, whereas cardiomegaly and other features of heart dysfunction were identified in 23% [14]. Up to 13% of cases resulted in preterm delivery. Moreover, abnormal neurodevelopmental complications were noticed in 37% of cases, whereas 61% of the cases featured abnormal postnatal imaging of the brain. Only 30% of cases were free from neurological dysfunction after birth, although this observation was limited by the variable time points at follow-up. Overall, this meta-analysis concluded that there was an association of fetal VOGM with a high occurrence of cerebral insult, cardiovascular dysfunction, and aberrant neurodevelopmental complications post-delivery.

Pregnancy complicated by fetal VOGM should be managed in a multidisciplinary fashion, with healthcare personnel with expertise in neonatology, maternal-fetal medicine, cardiology, interventional radiology, and pediatric neurosurgery [17,18]. There is no in utero treatment. Management strategies generally include pregnancy termination, medical therapy, endovascular treatment, and surgical intervention.

Medical therapy is largely targeted toward managing cardiac failure and pulmonary hypertension. No universally agreed recommendations or clinical trials have been carried out regarding the best medical therapy for neonates with poor cardiac function [19].

Yan and colleagues conducted a comprehensive systematic review and meta-analysis, assessing the outcomes and complications of endovascular embolization in a large cohort of patients with VOGMs. The study included 34 research papers, encompassing 667 patients, among whom 44% were neonates when treatment was administered [20]. Overall, the study showed that close to 70% of patients had a favorable outcome. The frequencies of post-embolization adverse events and deaths were 37% and 10%, respectively. Moreover, a recent review highlighted that lack of undergoing endovascular treatment is linked to poor outcome among neonates/infants with VOGMs [21].

Surgical intervention is often reserved for clearing of intracranial bleeding (hematomas) as well as for management of hydrocephalus [22]. However, it should be noted that shunt placement for management of hydrocephalus is correlated with a higher burden of morbidity and mortality among patients with VOGMs, and thus shunt placement should be considered as a last resort [23].

In conclusion, diagnosis and management of VOGMs need expertise and a multidisciplinary approach. Therefore, the prenatal diagnosis contributes information that may aid physicians in guiding their patients to make optimal clinical decisions together.

Contributors

Afaf Tawfiq contributed to patient care and the literature review, and revised the article critically for important intellectual content.

Zehour Alsabban wrote the original draft, was involved in the management of the patient, revised and submitted the figures.

Saeed Baradwan contributed to the conception of the case report, was involved in the literature review, participated in data acquisition, and drafted the manuscript.

All authors approved the final manuscript.

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Patient consent

Written informed consent was obtained from the patient for publication of this case report and the use of accompanying images.

Provenance and peer review

This article was not commissioned and was peer reviewed.

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Conflict of interest statement

The authors declare that they have no conflict of interest regarding the publication of this case report.

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