Vein of Galen aneurysmal malformation: from fetal diagnosis to perinatal management and long term follow up. A multidisciplinary paradigm for the diagnosis and treatment of a rare and challenging disease

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Objective. Vein of Galen aneurysmal malformation (VGAM) is a high-flow, low-resistance arterovenous malformation of the choroidal arterial system. It represents the most frequent congenital intracranial vascular malformation with an incidence of about 1 in 25,000 deliveries. VGAM results from the abnormal persistence of the median prosencephalic vein of Markowski, which normally involutes by the 11th week of gestation. Morbidity and mortality rates are still high despite a progressive improvement in pathophysiological understanding of the disease and treatment strategies. The diagnostic and therapeutic pathways are complex and based on a multidisciplinary approach, incorporating neuroradiological definition of the lesion, neonatal and pediatric intensive care support, and endovascular treatment (EVT). Few centers worldwide regularly take care of patients with VGAM from fetal diagnosis to long term follow-up and the current literature does not provide sufficient data to define many controversial aspects of this complex condition.

Materials and Methods. Several specialities are involved in the care of patients with VGAM at Gaslini Children's Hospital (IGG VGAM Team): Genetics, Perinatal Pathology and Medicine, Fetal and Pediatric Cardiology, Neurology, Neonatal and Pediatric Intensive Care, Interventional Radiology and Neuroradiology. A diagnostic and treatment paradigm was applied in a case series including all newborns with VGAM admitted to the Neonatal and Pediatric Intensive Care Unit between 2009 and 2021. Main scopes of the multidisciplinary approach in our Institution were: to define the genetic background of VGAM; to study fetal and neonatal ultrasound, echocardiographic, and neuroradiological features and identify prognostic factors; to apply specific intensive care strategies for neonates with VGAM and severe CHF and optimize treatment pathways (palliation, EVT, medical treatment); to define and overcome treatment challenges in endovascular treatment; to study specific pathological features secondary to VGAM: brain damage, lung and heart pathological changes secondary to AV shunt and overflow; to manage pregnancy, delivery, perinatal period and follow up with counseling and family support; to plan a multidisciplinary long term follow up with evaluation of outcomes (functional outcome and quality of life).

We here present a summary of the IGG VGAM Team experience from 2009 to 2022.

Results. Treatment, prognostic factors, and main outcomes are described in **Table 1**. Intensive care support and endovascular treatment prevented refractory cardiac and multiorgan failure in the neonatal period. Overall survival of treated patients was 87% (27/31) and good neurological outcome in survivors was 77%.

Conclusions. The complexity of VGAM pathophysiology requires a multidisciplinary approach in a tertiary care center with specific experience and competencies. Morbidity and mortality rates remain high in newborns; however, the mortality rate in our population was lower than the ones reported in the literature (30-60%). The neuroradiological and hemodynamic risk factors identified are consistent with those described by other authors. International key-experts' collaboration and data sharing are mandatory to finding appropriate answers to the challenges in the diagnosis and treatment of VGAM. The IGG VGAM Team is promoting an international registry through a network of leading researchers in this field to advance research and knowledge, and ultimately improve outcomes.

Table 1.

Total number of patients enrolled at Gaslini Children's Hospital	45
Number of patients enrolled in the neonatal period • inborn (prenatal diagnosis) • outborn (postnatal diagnosis)	38 33 5
Patients treated in the neonatal period	20 (53%)
Patients treated beyond the neonatal period	11 (29%)
Total EVTs • neonatal EVTs	68 28
EVTs/patient	0-8
Patients with EVT complications • newborns • children	11/31 (35%) 10/20 (50%) 1/11 (9%)
Neonatal mortality • procedure related • palliation for congenital severe cerebral damage	8 (21%) 4 (10.5%) 4 (10.5%)
Functional outcome Good overall performance or mild disability • total • survivors	 60% 77%
Factors associated with adverse outcome • neuroradiological • hemodynamic	 - sagittal superior sinus stenosis - jugular bulb stenosis - arterial pseudofeeders - heart failure - right to left PDA shunt
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EVT: endovascular treatment. PDA: persistent ductus arteriosus