Case Report

Cardiac footprints of a neonatal vein of Galen malformation

Kambiz Norozi^{a,b,*}, Rohit Arora^a, Henry Roukema^a, Victor Han^a and Ralf Rauch^c

^a*Department of Pediatrics, University of Western Ontario, London, Canada*

^b*Department of Pediatric Cardiology and Intensive Care Medicine, Medical School Hannover, Hannover, Germany*

^c*Department of Pediatrics, Rems-Murr Hospital, Waiblingen, Germany*

Received 13 December 2012

Revised 8 January 2013

Accepted 9 January 2013

Abstract. Vein of Galen arterio-venous malformation is a rare congenital anomaly, which presents in the neonatal period. High output heart failure in the absence of an underlying structural heart defect points towards the condition and is an important contributing factor for morbidity and mortality. We describe a case of a term newborn who presented with respiratory distress and cardiomegaly. The characteristic echocardiographic findings of high output cardiac failure were found and eventually led to the diagnosis of an underlying vein of Galen arterio-venous malformation. The relevant literature is discussed.

Keywords: Vein of Galen arterio-venous malformation, echocardiography, neonate

1. Introduction

Vein of Galen arterio-venous malformation (VGAM) is a rare congenital anomaly with a reported incidence of 1: 25,000 live births [1,2]. Besides hydrocephalus and seizures, high output heart failure with severe multi-organ failure are important contributing factors of morbidity and mortality in this condition. Management in the neonatal period remains a challenge. As many untreated newborns die during the first week of life, early diagnosis is of utmost importance. This report describes almost pathognomonic echocardiographic findings in VGAM.

2. Case report

A term infant boy was born at a primary care hospital. Antenatal ultrasound performed in the 2nd trimester had been uneventful. Birth weight was 4.64 kg (97% percentile), length 54 cm (50% percentile) and head circumference 40 cm (>97% percentile). Physical examination revealed a hyperactive precordium and a harsh systolic murmur (grade 3/6) over the left upper sternal border, and chest X-ray suggested cardiomegaly (cardio-thoracic ratio >0.6). At 20-hours of age, the child showed signs of congestive heart failure including respiratory distress, tachycardia, low blood pressure and decreased oxygen saturation as well as an enlarged liver size. As the condition deteriorated further, the boy was intubated and ventilated. In this setting, echocardiography showed a mild biventricular enlargement, with a large patent ductus arteriosus (PDA)

*Corresponding author: Kambiz Norozi, Division of Pediatric Cardiology, Department of Paediatrics, London Health Sciences Centre, Western University, 800 Commissioners Road East, PO Box 5010, London, ON, N6A 5W9, Canada. Tel.: +1 519 685 8500; extension 56062; Fax: +1 519 685 8156; E-mail: kambiz.norozi@lhsc.on.ca.

Fig. 1. (A) High parasternal view, large patent ductus arteriosus (9 mm) with significant right to left shunt (RPA = Right pulmonary artery; LPA = Left pulmonary artery; PDA = Patent ductus arteriosus). (B) Coronal subcostal view, dilated right atrium, interatrial septum bulging towards left atrium (RA = Right atrium; LA = Left atrium). (C) Parasternal short axis view, enlarged right ventricle, interventricular septum bulging towards left ventricle (RV = Right ventricle; LV = Left ventricle). (D) Suprasternal short axis, dilated superior vena cava as sign for increased venous return towards the enlarged right atrium (SVC = Superior vena cava).

(significant right to left shunt, Fig. 1A) and moderate sized ventricular septal defect (VSD) and atrial septal defect (ASD), both with bidirectional shunting. The interatrial and interventricular septums were bulging towards the left side, indicating elevated right ventricular volumes and pressures (Figs. 1B and 1C). In spite of persistent cyanosis, no other structural cardiac abnormality could be detected indicating cyanotic heart defect. Large innominate vein and superior cava veins were present (Fig. 1D) as a hint of increased cranial perfusion. Color Doppler of the aortic arch showed retrograde blood flow through the distal arch into the carotid vessel (Figs. 2A and 2B). Head ultrasound demonstrated widened ventricles with prominent bilateral choroid plexuses, but no hemorrhage. Cerebral Doppler imaging revealed VGAM as the actual cause of high output cardiac failure (Fig. 2C). An magnetic resonance angiogram displayed VGAM, marked enlargement of the vein of Galen, adjacent sinuses and of both internal jugular veins (Fig. 2D).

In addition to the heart failure, an obstructive hydrocephalus with marked ventriculomegaly developed in the course of 5 d. The boy underwent an endovascular embolization of the feeding cranial vessels with concentrated glue (lipiodol and n-butyl cyanoacrylate 1:1), and the immediate post-interventional echocardiogram showed a much improved antegrade diastolic blood flow over the distal aortic arch. Unfortunately, the child died due to massive intra-ventricular hemorrhage occurring 24 h after the procedure. Autopsy findings confirmed VGAM, hydrocephalus and intracranial hematomas with acute hypoxic ischemic encephalopathy.

3. Discussion

VGAM is a rare congenital anomaly comprising of cerebral arterio-venous shunt with multiple feeding arteries. Embryologically, VGAM arise as result of communications between the arterial network and the median

Fig. 2a

Fig. 2b

Fig. 2c

Fig. 2d

Fig. 2. (A) Suprasternal long axis, widened head and neck vessels branching from aortic arch. (B) Color Doppler demonstrates retrograde diastolic blood flow (red) from the distal aortic arch into the head and neck vessels. (C) Cerebral ultrasound, angled parasagittal view, Vein of Galen arterio-venous malformation with ventriculomegaly (VGAM = Vein of Galen arterio-venous malformation). (D) Magnetic resonance imaging angiogram of the head, Vein of Galen arterio-venous malformation with marked enlargement of the Vein of Galen, the straight, transverse and sigmoid sinuses as well as of both internal jugular veins. Vein of Galen arterio-venous malformation with arterial supply from posterior cerebral artery branches with some posterior communicating arteries (IJV = Internal jugular vein).

prosencephalic vein. The insult leading to formation of VGAM is thought to occur after the stage of the 21–23 mm embryo (between the 6th and 11th wk of intrauterine life) [3]. The principal feeders of the malformation include the anterior or prosencephalic group (the anterior cerebral, anterior choroidal, middle cerebral and the posterolateral choroidal arteries), and the posterior or mesencephalic group (the posteromedial choroidal, posterior thalamo-perforating, quadrigeminal and superior cerebellar arteries) [4]. All of them are supplied with arterial blood from either the vertebral arteries or the internal cerebral arteries, which again are fed via the aortic arch. This explains the retrograde diastolic flow in the descending aorta and a systolic-diastolic ante grade flow over the ascending aorta and the prominent head and neck vessels. Both caused the entire arch to appear “red” on color Doppler in diastole which represents one characteristic echocardiographic feature of VGAM (Figs. 2A and 2B). It also elucidates the widening of both the innominate

vein and superior vena cava (Fig. 1D) by an increased venous return. The latter is also the reason for the dilation of right atrium and ventricle with enhanced pressures leading to bi-directional shunt across ASD and VSD (Figs. 1B and 1C). In contrast to patients with a large feeding and a small draining vessel, the VGAM in our patient represented with a large cerebral left-to-right shunt with consecutive low systemic resistance. Together with a physiologically elevated pulmonary resistance in this neonate, the right ventricular volume overload further contributed to the right-to-left shunt across the PDA (Fig. 1A). It nourished the vicious circle of high-output heart failure that could not be adequately controlled by medication and ultimately caused the child’s death, as it led to a desperate attempt of embolization. Lasjaunias et al. [5] have proposed the Bicêtre neonatal evaluation score as a therapeutic guideline for the management of VGAM. Our patient obtained a very low score, being unstable despite medical treatment and

desaturation under assisted ventilation. Thus, therapeutic abstention might have been a better option. In this case, intracerebral hemorrhage was possibly due to occlusion of the venous outlet while the remaining flow into the VGAM was reduced insufficiently. Alternative treatment options have been published, comprising transvenous application of glue, arterial coiling, balloon occlusion or surgical ligation of the feeding vessel [5].

Echocardiographic differential diagnoses are scarce, but include coarctation of the aorta or interrupted aortic arch type-C for the unusual arch blood flow in the absence of hypoplastic left heart, or persistent fetal circulation for the elevated pulmonary artery resistance with consecutive right-to-left shunt over ASD, VSD or PDA [6]. Antenatal diagnosis of VGAM nowadays must be the goal, and it has been described as early as at 25 wk of gestation [7]. Yet, there are no cases of a prenatal treatment of VGAM described in the literature so far, and abortion has only been suggested for in utero cardiac failure or cerebral damage [5]. Postnatal imaging of the cerebral vasculature with magnetic resonance scans including magnetic resonance angiography and magnetic resonance venography should be performed especially in newborns exhibiting clinical criteria for high-output heart failure or presenting with characteristic echocardiographic features for VGAM as documented here [8,9].

References

- [1] Frawley GP, Dargaville PA, Mitchell PJ, Tress BM, Loughnan P. Clinical course and medical management of neonates with severe cardiac failure related to vein of Galen malformation. *Arch Dis Child Fetal Neonatal Ed* 2002;87(2):F144–9.
- [2] Bhattacharya JJ, Thammaroj J. Vein of Galen malformations. *J Neurol Neurosurg Psychiatry* 2003;74(1 Suppl):i42–4.
- [3] Raybaud CA, Strother CM, Hald JK. Aneurysms of the vein of Galen: embryonic considerations and anatomical features relating to the pathogenesis of the malformation. *Neuroradiology* 1989;31(2):109–28.
- [4] Casasco A, Lylyk P, Hodes JE, Kohan G, Aymard A, Merland JJ. Percutaneous transvenous catheterization and embolization of vein of Galen aneurysms. *Neurosurgery* 1991;28(2):260–6.
- [5] Lasjaunias PL, Chng SM, Sachet M, Alvarez H, Rodesch G, Garcia-Monaco R. The management of vein of Galen aneurysmal malformations. *Neurosurgery* 2006;59(5 Suppl 3):S184–94; discussion S3–13. Review. Erratum in: *Neurosurgery* 2007; 60(4 Suppl 2):393.
- [6] Deverall PB, Taylor JF, Sturrock GS, Aberdeen E. Coarctation-like physiology with cerebral arteriovenous fistula. *Pediatrics* 1969;44(6):1024–8.
- [7] Ballester MJ, Raga F, Serra-Serra V, Bonilla-Musoles F. Early prenatal diagnosis of an ominous aneurysm of the vein of Galen by color doppler ultrasound. *Acta Obstet Gynecol Scand* 1994;73(7):592–5.
- [8] Jones BV, Ball WS, Tomsick TA, Millard J, Crone KR. Vein of Galen aneurysmal malformation: diagnosis and treatment of 13 children with extended clinical follow-up. *AJNR Am J Neuroradiol* 2002;23(10):1717–24.
- [9] Smilari P, Incorpora G, Sciacca P, Marletta M, Saporito A, Mazzone L, et al. Vein of Galen aneurysmal malformation. Different clinical expressiveness. Three case reports. *Minerva Pediatr* 2000;52(1–2):55–62.