Vein of Galen malformation and interrupted aortic arch in a neonate: a previously unreported association

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The arteriovenous malformation of the vein of Galen is a rare entity in neonates. Its association with other major cardiac anomalies is even more uncommon. We report the first case of interrupted aortic arch associated with the vein of Galen malformation in the English literature. The possible etiology, diagnosis and management of this rare combination are discussed.

The location of vein of Galen is under the cerebral hemispheres. The function of the vein of Galen is the drainage of the anterior and central regions of the brain into the sinuses of the posterior cerebral fossa. Originating from the median vein of the prosencephalon, the vein of Galen is formed by the union of the two internal cerebral veins and goes on to drain into the straight sinus. An arterioveneous malformation occurs when either directly or via an interposed angiomatous malformation the vein of Galen receives an arterial input from one or more major intracranial arteries resulting in arteriovenous shunting of blood. In reality, the term vein of Galen malformation should not be used any more. The malformation is due to the fact that the precise defect is now known to be a persistent embryonic prosencephalic vein of Markowski developing during 6 to 11 weeks of gestation, which actually drains into the vein of Galen. The vein of Galen malformation, therefore, is a misnomer.

Advances in diagnostic and therapeutic modalities have considerably altered the perception of vein of Galen malformation manifesting in the neonatal period, from a condition with uniformly grave morbidity and mortality to a favorable one,[1-2] except in the most severe cases. As intractable cardiac failure is a common denominator in a majority of neonatal cases,[3] and the coexistence of any significant congenital heart disease that can imperil the already jeopardized cardiac status portrays a dismal outcome.[3-4] Only twenty-three (23) cases of vein of Galen malformation coexisting with a congenital cardiac defect have been reported, including five patients by McElhinney[5] in his review of 1998. Although coarctation of aorta has been noted in 20% (9/43) of cases of vein of Galen malformation, to our knowledge this is the first case to be reported in a neonate with arteriovenous malformation of the vein of Galen coexistent with the interrupted aortic arch defect.

Case

A full-term female infant was born by normal spontaneous vaginal delivery with Apgar scores of 7 and 9 at 1 and 5 minutes, respectively. The birth weight was 2730 grams, head circumference 32 centimeters and length 50 centimeters. A diagnosis of congenital hydrocephalus was suspected on a prenatal sonogram performed at the referring hospital. The repeat prenatal sonogram at our institution revealed oligohydroamnion, an intracerebral cystic mass and severe cardiomegaly. After delivery, the infant was transferred to the neonatal intensive care unit.

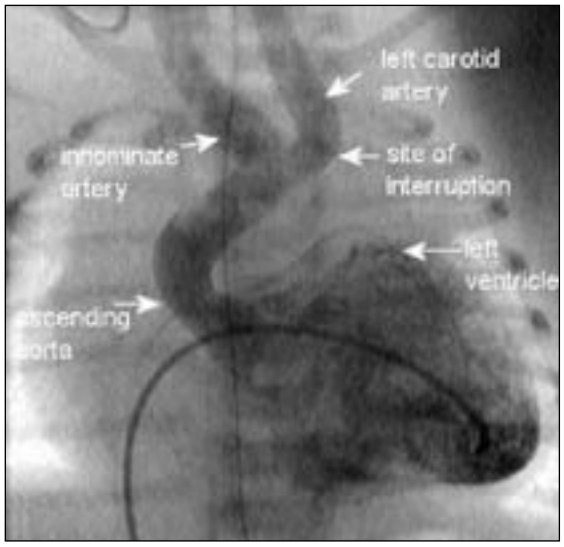

Figure 1. Left ventriculogram in straight antero-posterior view showing aortic arch interruption (type "B") distal to the left carotid artery. A severely dilated superior vena cava and the right atrium are seen in the background.

Within a few hours of birth a significant systolic murmur without central cyanosis was noted in the apical area of the heart, which prompted an urgent cardiology consultation. The echocardiographiz findings were suggestive of critical coarctation of the aorta, but interrupted aortic arch could not be excluded. A continuous infusion of prostaglandin E_1 was started. Type B interrupted aortic arch with intact ventricular septum was confirmed on the first day of life by cardiac catheterization and a CT brain scan was performed on the same day, which revealed a severe type of vein of Galen malformation.

On the second day of life, signs and symptoms of severe congestive heart failure were noted and mechanical ventilation was initiated for impending respiratory failure. Seizure movements of bicycling and rowing leg movements were noted. These were controlled by intravenous phenobarbital. An MRI of the brain was performed on the fourth day of life, which found no intracerebral hemorrhage. A joint meeting of the parents with neurosurgery, interventional radiologist and neonatologists was held in which the parents requested the use of all possible measures even with the highest risk of mortality. On the fifth day of life, cerebral angiography was performed through the right axillary artery approach. The plan was to embolize the vein of Galen malformation. Due to tortousity and spasm of both the right and left internal carotid arteries further advancement of the catheter was not possible. Although the left middle cerebral artery and the distal major feeders of the arteriovenous malformation were accessible, embolization was not possible due to vasospasm. The angiogram, however, showed that the blood flow in the feeding branches of the arteriovenous malformation was very slow and rupture of the arteriovenous malformation was suspected. The procedure was aborted and an immediate CT scan of the brain revealed acute bilateral severe intraventricular hemorrhage with a moderate left subarachnoid hemorrhage and associated hydrocephalus. The patient's neurological condition deteriorated rapidly after the procedure. He developed coma and expired on the 14th day of life.

Discussion

The anomaly of the vein of Galen carries variable prognosis that depends on the time of presentation and the associated severity. In general, the cases diagnosed in the prenatal or in the neonatal age group carry the worst prognosis. This diagnosis alone presents a diagnostic and therapeutic challenge. However, its association with other anomalies, especially with complex congenital heart diseases, significantly influences the prognosis.

McElhinney et al[5] in 1998 reviewed the coincidence of the congenital heart defects with the vein of Galen malformation. Only 18 such cases had been reported prior to his review, which presented 25 patients in total. Only 27% (5/18) of congenital cardiac defects were diagnosed by echocardiography in the case reports prior to McElhinney while 67 % (12/18) were detected by autopsy. This contrasts with the diagnoses in the McElhinney cases, all of which were made by echocardiogram. The coarctation of aorta and or isthmic hypoplasia[6] are one of the most common defects other than sinus venosus atrial septal defects reported[7] in coexistence with the vein of Galen malformation. However, the absence of the vein of Galen malformation in coexistence with the diagnosis of interrupted aortic arch (Type B) in a search of MEDLINE in the English literature from 1960 to date makes our case the first to be reported in the English literature. In addition, our patient had intact ventricular septum, which is a very unusual finding as Type B interrupted aortic arch is almost always associated with mal-aligned ventricular septal defect.

A plausible hypothesis cited for the coarctation of aorta and or isthmic hypoplasia is increased flow across the arterial duct and reduced antegrade flow across the aortic isthmus, leading to an augmented right ventricular output in utero.[8] Decreased antegrade flow from the ascending aorta to the isthmus in

the presence of the vein of Galen malformation therefore seems logical. The cause in our case could be the same or a more severe degree of the same hypothesis.

The clinical presentation in the cases of vein of Galen malformation in neonates depends upon its size, involvement of various vessels and any associated defects. The spectrum of congenital cardiac defects present in the cases of the vein of Galen malformation range from those requiring urgent life saving procedures to careful observation and follow-up. Therefore, the importance of the exclusion of significant defects, accountable for significant therapeutic and prognostic implications needs to be emphasized. Our case underwent an urgent embolization attempt first as this was the main cause of intractable congestive heart failure. The repair of the interrupted aortic arch was contemplated, but was felt deferrable due to widely open ductus arteriosus, absent blood pressure gradient between the upper and lower extremities, and adequate perfusion in the lower extremities.

The complications of high cardiac output failure and intracranial hemorrhage that occurred in our neonate have been well described in cases of vein of Galen malformation. The intracranial hemorrhage could be spontaneous as mentioned by Simpson[9] or could have occurred during the embolization attempt because of the fragile vessels of the aneurysm. The CT brain scan on the fourth day of life in our infant was without any signs of intraventricular hemorrhage. However, the repeat CT brain scan performed immediately after the embolization attempt at the 5th day revealed significant intracranial hemorrhage. In addition, no significant changes in the clinical signs and symptoms were noticed in the time lapsing between these two scans. Both facts suggest an inadvertent perforation of the aneurysm, leading to intraventricular hemorrhage during the diagnostic angiography. Spontaneous rupture of the aneurysm due to fragile vessels and general anesthesia during angiography is another possibility.

The prognosis has considerably changed for cases of vein of Galen malformation.[1-2] The diagnosis, once made more often at autopsy is now possible antenatally[10-11] in most cases. Additionally an increasing number of neonates are now successfully treated with catheterization-embolization technique[12] including detachable balloons, steel (Gianturco) coils and liquid tissue adhesives (cyanoacrolate). Survival rates of 70% to 80% in neonates and young infants and cure rates of approximately 50% have been described. Still the presence of severe cardiac failure has been associated with a poor prognosis. Neonates with cardiac lesions like aortic coarctation in the presence of severe cardiac failure remain at high risk of mortality and morbidity. Physicians and parents alike experience ethical dilemmas in these challenging cases. Initially a case-by-case approach, prioritizing the sequence of clinical procedures according to the clinical course seems more rational. The decisions for continuation or even withdrawal of care are usually required late in the clinical course. A multidisciplinary parental counseling is advisable in these circumstances.

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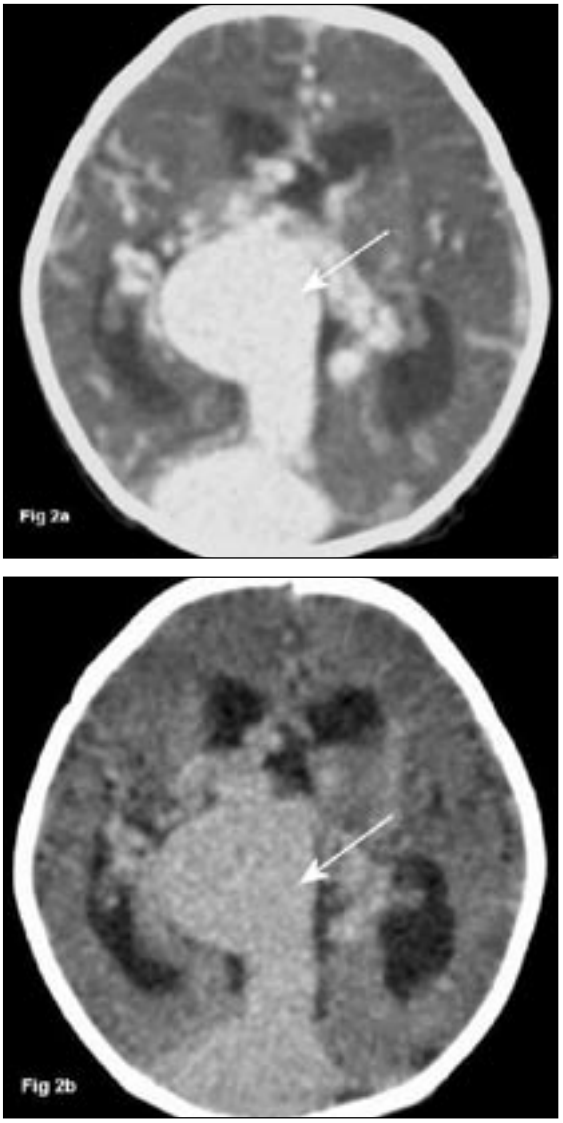

Figure 2. Contrast (2a) and plain (2b) CT scan, coronal section of the brain showing large aneurysm malformation of the vein of Galen.

References

1. Dowd CF, Halbach VV, Higashida RT, et al. Endovascular management of vein of Galen malformations. *New trends in management of cerebrovascular malformations.* New York: Springer-Verlag, 1994;553-558.

2. Wisoff JH, Berenstein A. Interventional neuroradiology. In: Edwards MSB, Hoffman HJ, eds. *Cerebral vascular disease in children and adolescents.* Baltimore: Williams and Wilkins, 1989:139-57.

3. Carroll CPH, Jakoby RK. Neonatal congestive heart failure as the presenting symptom of cerebral arteriovenous malformation. *J Neurosurg.* 1966;25:159-63.

4. Johnson IH, Whittle IR, Besser M, Morgan MK. Vein of Galen malformation: diagnosis and management. *Neurosurgery.* 1987;20:747-58.

5. D B Mc Elhinney, VV Halbach, N H Silverman, et al. Congenital cardiac anomalies with vein of Galen malformations in infants. *Archiv Dis Child.*1998;78:548-551.

6. Comstock CH, Kirk JS. Arteriovenous malformations: locations and evolution in the fetal brain. *J Ultrasound Med.* 1991;10:361-5.

7. Friedman DM, Rutkowski M, Madrid M, et al. Sinus venosus atrial septal defect associated with vein of Galen malformations: report of two cases. *Pediatr Cardiol.* 1994;15:50-2.

8. Rudolph AM, Heymann MA, Spitznas U. Hemodynamic considerations in the development of narrowing of the aorta. *Am J Cardiol.* 1972;30:514-25.

9. Simpson RK Jr, Fischer DK, Haber LM, Mawad ME, Rose JE. Coexistence of multiple arteriovenous malformations and an anomalous aortic arch. *Clin Neurol Neurosurg.* 1988;90(2):169-73.

10. Nyberg DA, Mahony BS, et al. *Diagnostic ultrasound of fetal anomalies.* A Text and Atlas. Boca Raton, Fla: CRC Press 1990.

11. Reiter AA, Huhta JC, et al. Prenatal diagnosis of arteriovenous malformation of the vein of Galen. *J Clin Ultrasound.* 1986;14:623-628.

12. Friedman DM, Verma R, Madrid M, et al. Recent improvement in outcome using transcatheter embolization techniques for neonatal aneurysmal malformations of the vein of Galen. *Pediatrics.* 1993;91:583.