Sinus Venosus Atrial Septal Defect Associated with Vein of Galen Malformations: Report of Two Cases

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SUMMARY. Two unique cases are presented of infants with signs of vein of Galen malformations, whose unsuspected associated sinus venosus atrial septal defects were detected during routine echocardiography. A conservative approach to cardiac treatment is advocated.

KEY WORDS: Vein of Galen — Sinus venosus — Atrial septal defect — Cerebral arteriovenous malformation

Cerebral arteriovenous malformations involving the vein of Galen are rare, and often fatal, causes of congestive heart failure in infancy. Recent advances in intravascular embolization treatments have resulted in increasing survival rates [3-5]. Occasionally, associated cardiac defects such as patent ductus arteriosus have been described.

Sinus venosus atrial septal defects are an unusual type of atrial septal defect, 5-10% of the total [2], often associated with partial anomalous pulmonary venous return. We describe two cases of the first recorded association between vein of Galen lesions and sinus venosus atrial septal defects. The atypical clinical features require a high index of suspicion and necessitate the highly reliable technique of echocardiography to make the diagnosis. A conservative treatment approach is recommended.

Case Reports

Case 1

This 16-month-old boy weighed 6 lb 7 oz at birth, gestation had been complicated by hyperemesis gravidarum. He was born by primary cesarian section for failure to progress. A routine third trimester ultrasound had demonstrated an intracranial malformation, which was confirmed after birth as a vein of Galen arteriovenous malformation by computerized tomography and angiography. Mild congestive heart failure developed shortly after birth, and he underwent a partial retrograde transvenous embolization of the lesion with n-butyl cyanoacrylate and platinum coils at 1 week of age. No seizures or neurological compromise developed while receiving phenobarbital.

His first 2 months were marked by significant congestive heart failure requiring digoxin and furosemide therapy. The failure and cranial bruit began to resolve at 2 months of age when echocardiography first suggested a sinus venosus atrial septal defect in addition to the high output state.

At 4 months of age, he was readmitted for a second embolization of NBCA into several arterial feeding vessels, which was successful and diminished the intensity of the bruit. Within a few weeks a third admission was necessitated by a transient left hemiparesis and mild hydrocephalus, possibly accompanied by a seizure. A third embolization at 6 months of age resulted in the complete thrombosis of the vein of Galen. This was accompanied by prolonged fever, anemia, and abnormal coagulation tests, with the disappearance of the bruit. The mild hydrocephalus regressed.

Current neurological status includes a minimal left hemiparesis with right-sided electroencephalographic (EEG) abnormalities and mildly delayed speech, but he is walking at 16 months, while on phenobarbital. He is thriving and has not required cardiac medications since 9 months of age.

Examination reveals head circumference of 46 cm with pulse 120 and BP 90/50. The head is normal without bruits. The hemiparesis is barely perceptible, and he says a few words. The precordium is quiet without bulges or thrills. The lungs are clear. There is no clubbing, cyanosis, or edema. The jugular venous hum present in the supravclavicular area, only in the upright position. There is a normally split second heart sound, but a grade 2/6 systolic murmur along with the left sternal border. No diastolic murmurs are heard, and the pulses are equal. There is no hepatosplenomegaly.

The original chest x-rays showed marked cardiomegaly which resolved. The neonatal electrocardiograms (ECGs) showed severe biventricular hypertrophy, which completely nor-
Friedman et al.: Vein of Galen Malformations

A contrast CT which displayed a vein of Galen malformation with outlet stenosis, which was confirmed by magnetic resonance imaging (MRI) and angiography. The palsy resolved. At 8 months of age, a successful endovascular embolization was performed which occluded two afferent arteries, or about 80% of the flow, leaving only one small feeding vessel open. The bruit disappeared. Although neurologically normal, he was treated with phenytoin. The last MRI showed bilateral extra-axial fluid collections (subdural hygromas).

Fig. 1. Two-dimensional subxiphoid echocardiogram in patient 1 in long (A) and short axis (B) views displaying sinus venosus atrial septal defect (arrow). LA, left atrium; LV, left ventricle; RA, right atrium; RV, right ventricle.

An incidental chest x-ray revealed cardiomegaly. Examination revealed weight 8.9 kg, pulse of 144 bpm, and respirations 40 bpm. The head was large and box-like with circumference 52 cm and frontal bossing. There was dilated scalp and facial veins, but no bruit. The fontanelle was soft and open with a nonfocal neurological exam. There was no clubbing, cyanosis, or edema. The lungs were clear. The precordium was hyperactive without thrills, and the apical impulse was displaced to the 6th intercostal space in the left anterior axillary line. There was a prominent S3 gallop and a grade 2/6 midsystolic flow murmur at the left sternal border. There were no diastolic murmurs. The femoral pulses were diminished at the previous puncture sites. There was no hepatosplenomegaly.

An electrocardiogram showed increased right ventricular forces. The echocardiogram showed dilatation of right-sided structures with a large sinus venosus type atrial septal defect and probable partial anomalous pulmonary venous return. Cardiac management is currently deferred until the neurological care is completed.

Case 2

This 8-month-old asymptomatic boy was born after an uncomplicated pregnancy. At 4.5 months of age, a rapidly increasing head size was noted with the appearance of prominent scalp veins, although he remained neurologically normal. Head ultrasound and computerized tomography without contrast were not diagnostic. At 6 months of age, a sixth nerve palsy led to the performance of a contrast CT which displayed a vein of Galen malformation with outlet stenosis, which was confirmed by magnetic resonance imaging (MRI) and angiography. The palsy resolved.

At 8 months of age, a successful endovascular embolization was performed which occluded two afferent arteries, or about 80% of the flow, leaving only one small feeding vessel open. The bruit disappeared. Although neurologically normal, he was treated with phenytoin. The last MRI showed bilateral extra-axial fluid collections (subdural hygromas).

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Discussion

This is the first description of an association between sinus venosus atrial septal defects and vein of Galen arteriovenous malformations. Vein of Galen lesions cause congestive heart failure in infancy [3–5], which may be difficult to recognize because clinical findings are nonspecific until a cranial bruit is recognized. Improving survival rates requires the prompt diagnosis of possible associated lesions, but the signs of infantile atrial septal defect are atypical and may be masked in the presence of a high flow state with congestive failure and other murmurs. The current state of echocardiography is so sensitive and specific [1], especially in the infant, as to allow the reliable diagnosis of an unsuspected sinus venosus defect even in the absence of confirmatory catheterization data. Therefore, now that vein of Galen malformations can be more successfully treated, associated cardiac defects should be considered if the signs of heart failure persist after occlusion of the vein of Galen shunt.

The incidence of the association of sinus venosus defects with vein of Galen lesions is unknown; however, during the past 12 years, we have treated 37 embolization patients and found only these two atrial defects. Two independent computerized literature searches back to 1976 failed to reveal any other reports of an association between congenital