Arteriovenous shunting as a new feature of PHACES

Case report

HUAN WANG, M.D.,1 ALBERT K. OH, M.D.,3 AND DARREN B. ORBACH, M.D., PH.D.1,2

Departments of 1Neurosurgery and 2Radiology, Neurointerventional Radiology, Children’s Hospital Boston, Brigham & Women’s Hospital, Harvard Medical School, Boston, Massachusetts; 3Department of Surgery Plastic Surgery, Hasbro Children’s Hospital, Providence, Rhode Island

Patients with the congenital neurocutaneous disorder PHACES are at a markedly increased risk of ischemic infarction during childhood. Although intracranial arterial anomalies have been well described, venous abnormalities have not been documented. The authors report on a unique case of a 3-month-old girl with PHACES and a skull base osteodural arteriovenous fistula. A separate arteriovenous shunt at T-5 may also have been present. Imaging findings and treatment strategies are discussed. (DOI: 10.3171/2008.10.PEDS08169)

Key Words • angiography • arteriovenous fistula • embolization • hemangioma • PHACES

PHACES is a congenital neurocutaneous disorder first described in 1978 by Pascual-Castroviejo12 and later expanded on by Frieden and colleagues.4 The spectrum of clinical findings includes posterior cranial fossa malformations, hemangiomas, arterial anomalies, coarctation of the aorta and cardiac defects, eye abnormalities, and sternal defects (PHACES). Although hemangioma is the unifying clinical feature, partial phenotypical expression and variegated manifestations often add uncertainty to the diagnosis, resulting in an incomplete clinical and radiological evaluation.

Extracutaneous manifestations of PHACES most frequently involve CNS structural and cerebrovascular abnormalities.5 Representing the greatest potential source of complications,2 these CNS findings warrant focused attention because affected children are at risk for developmental delay, mental retardation, seizures, and acute ischemic stroke. Neurological sequelae have been reported in 50–90% of patients.7

Virtually all reported intracranial vascular anomalies associated with PHACES have been arterial in nature;10 consisting of agenesis of major cervical arteries, embryological persistence of arteries, arterial stenoses, dilatative arteriopathy, and aneurysms. We report on a unique case of a 3-month-old girl with PHACES, a skull base osteodural AVF, and MR imaging findings suggestive of a separate spinal AVF at T-5.

Case Report

This 1-month-old infant girl presented with an ulcerated hemangioma of the right posterior scalp, measuring 8 × 8 cm on physical examination. Ultrasonographic evaluation of the surrounding area was nondiagnostic. Magnetic resonance imaging was recommended for further workup, but the child’s parents declined. The patient presented again at 3 months of age with an aggressive subglottic hemangioma that was causing stridor and respiratory distress. Imaging studies of the hemangioma revealed a skull base osteodural AVF with encroachment over the inferolateral right cerebellar hemisphere (Fig. 1). Classic intracranial artery anomalies were also documented, including an atretic right internal carotid artery (Fig. 2) and an azygous anterior cerebral artery.

The skull base osteodural AVF was supplied by ectatic BA branches, in particular an enlarged dural branch of the right AICA (Fig. 3), multiple branches of the right external carotid artery (Fig. 4 left), and musculoskeletal

Abbreviations used in this paper: AICA = anterior inferior cerebellar artery; AVF = arteriovenous fistula; BA = basilar artery; PHACES = posterior cranial fossa malformations, hemangiomas, arterial anomalies, coarctation of the aorta and cardiac defects, eye abnormalities, and sternal defects; VA = vertebral artery.
branches of the right VA (Fig. 4 right). The right occipital bone and mastoid portion of the right temporal bone showed marked osteolysis (Fig. 1). The right AICA dural feeding vessel demonstrated arteriovenous shunting into the right lateral sinus and filling of cerebellar dural veins. The drainage of this complex AVF involved a mastoid bone venous lake communicating with the right sigmoid sinus through multiple intraosseous channels (Fig. 5).

Despite aggressive steroid therapy, the patient had recurrent respiratory distress and underwent resection of the subglottic hemangioma. When she was 8 months old, the AVF was partially embolized transarterially through the BA feeding vessels (primarily the right AICA) using coils and Onyx. The right AICA supply terminated in a cluster of dysplastic vessels that abutted the inferior aspect of the cerebellar hemispheric convexity, and represented the largest inflow to the fistula (Fig. 3); this pedicle posed a risk of intracranial hemorrhage and thus was addressed first. Targeted embolization here achieved a significant reduction in overall arteriovenous shunting (Fig. 6). Five months later, follow-up MR images demonstrated near normalization of the ventricle size and extraaxial subarachnoid space. Staged embolization with Onyx via the right middle meningeal artery was started when the patient was 13 months of age. Because the intracranial component shows virtually no opacification, and the patient remains neurologically intact without evidence of intracranial venous hypertension, the risk of intracranial hemorrhage or neurological developmental impediment is low. Further staged extracranial embolization is planned for definitive closure.

We examined the caudal drainage of the fistula fluoroscopically, and saw no significant intraspinal venous outflow. The T-5 lesion observed on MR imaging (Fig. 7) thus probably represents a second arteriovenous shunt; further angiographic exploration is pending.

**Discussion**

Approximately 50% of patients with PHACES have a cerebrovascular anomaly. Reported anomalies include persistent embryonic vessels, agenesis/hypoplasia of major
cervical arteries, anomalous vasculature, arterial stenoses, segmental dolichoectasia, and aneurysms. It is striking that all of the currently reported variations are arterial in nature, while the underlying abnormalities for other neurocutaneous syndromes, such as cerebrofacial arteriovenous metameric syndrome and Sturge-Weber syndrome, involve abnormalities of the capillaries and veins.

The present study is the first detailed description of arteriovenous shunting in a patient with PHACES. In a recently published clinical series of 17 patients, intracranial arteriovenous shunting was described briefly in 1 patient. Because arteriovenous malformations are believed to be of venous origin, the existence of these cases suggests that the underlying vascular abnormalities of PHACES may not be purely arterial.

Dural arteriovenous fistulas presenting in adulthood have been well characterized with regard to their risk of hemorrhage, and a classification system stratifying this risk based on angiographic features such as cortical venous reflux is in wide use. Due to the extreme rarity of dural fistulas in young infants, the risk of intracranial hemorrhage posed is more difficult to assess. However, given the additional risks of cerebral venous thrombosis and diffuse venous hypertension, fistulas presenting in infancy are associated with very high overall morbidity and mortality rates.

As a concrete manifestation of clinically relevant venous hypertension, our patient had increasing prominence of the ventricular cavities and extraaxial subarachnoid space, as well as increases in head circumference, during the time that fistula treatment was limited by airway issues. The patient's head circumference at 3 months was 42 cm (90th percentile) and 48.5 cm at 10 months, well off of the growth chart. The initial transarterial embolization

![Image](image1.png)

**Fig. 4.** Angiography. *Left:* Lateral view of a right common carotid artery injection demonstrates supply to the AVF via the right middle meningeal and posterior auricular branches. *Right:* Frontal view of right VA injection demonstrates supply to the AVF from the musculoskeletal branches of the right VA.

![Image](image2.png)

**Fig. 5.** Frontal view of the venous phase of a left VA injection demonstrates bone lakes in the right petrous apex (large arrow) communicating with the right sigmoid sinus in draining the AVF via multiple intraosseous channels (small arrows).

![Image](image3.png)

**Fig. 6.** Frontal (left) and lateral (right) views of a left VA injection after embolization of the BA supply to the AVF demonstrates that the fistula is now exclusively extracranial, with improved intracranial runoff.

![Image](image4.png)

**Fig. 7.** Sagittal T2-weighted (left) and 2 consecutive axial inversion recovery (right) MR images showing a posterior right intrathecal flow void suggestive of thoracic spinal arteriovenous shunt.
bolization through the right AICA resulted in significant flow reduction of the arteriovenous shunt. Interestingly, 5-month follow-up MR images demonstrated near normalization of ventricle size and extraaxial subarachnoid space (Fig. 8), probably reflecting the effects of reduced venous hypertension (head circumference at 14 months was 49 cm, having markedly slowed its increase). Similar resolution of hydrocephalus and stabilization of neurological symptoms in the setting of vein of Galen malformations embolization has been reported.\(^5\)

The etiology of PHACES remains elusive, with work underway to elucidate possible genetic links,\(^4\) developmental field defects, or teratogenic influences.\(^3,13\) Although currently considered sporadic in nature, PHACES has a striking female predominance, with a reported female-to-male ratio as high as 9:1.\(^1\) Our experience with an infant with PHACES and arteriovenous shunting may shed further light on the timing of the onset and its underlying pathogenesis.

**Disclaimer**

The authors report no conflict of interest concerning the materials or methods used in this study or the findings specified in this paper.

**Acknowledgments**

The authors gratefully acknowledge the leadership and staff of the Vascular Anomalies Center at Children’s Hospital Boston, through whom this case came to our attention.

**References**


Accepted October 21, 2008.
Address correspondence to: Darren B. Orbach, M.D., Ph.D., Neurorinterventional Radiology, Brigham & Women’s Hospital, 75 Francis Street PBB 356, Boston, Massachusetts 02115. email: dorbach@partners.org.