Aortic arch anomalies in PHACE syndrome: An individualized approach to an unusual problem

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PHACE syndrome is a relatively uncommon disease characterized by Posterior fossa anomalies, Hemangiomas, Arterial lesions, Cardiovascular abnormalities, and Eye anomalies (ie, PHACE).¹ One of the most salient characteristics of this disease is its variable presentation, in particular as it relates to the spectrum of aortic and brachiocephalic vascular anomalies (Figure 1). Patients with PHACE syndrome tend to have longer segments of aortic stenosis in unusual locations (eg, mid-transverse arch); areas of aneurysmal dilation; aberrant origin, stenosis, or agenesis of brachiocephalic vessels; and increased vascular tortuosity. All of these characteristics make surgical planning and intervention challenging.

In this issue of the *Journal*, Caragher et al² describe in detail the clinical presentation, surgical intervention, and

Aortic coarctation and brachiocephalic stenoses in a patient with PHACE syndrome.

Central Message

Aortic arch and brachiocephalic vessel anomalies in PHACE syndrome are widely heterogeneous. An individualized approach is needed for surgical repair of aortic coarctation in these patients.

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FIGURE 1. Computerized tomography in a patient with PHACE syndrome shows a long-segment coarctation of the aorta and severe ostial stenoses of the left carotid and left subclavian arteries.

outcomes of 7 patients with PHACE syndrome, the largest series of aortic surgery in this patient population to date. The authors used a variety of different surgical techniques to address this complex problem, including reverse subclavian flaps, patch aortoplasties, as well as interposition and extra-anatomic grafts. Even though aortic resection and direct anastomosis have been used in some patients with PHACE syndrome,^{3,4} the long-segment stenosis and complex nature of the anatomy usually precludes the use of native-tissue traditional repairs. A recent report from our institution described a patient with PHACE syndrome with right-sided aortic coarctation and a vascular ring who underwent reimplantation of the aberrant left subclavian artery with patch enlargement and translocation of the descending aorta into the posterior ascending aorta.⁵ Despite the increased complexity, techniques such as this may avoid the use of interposition or extra-anatomic grafts in these patients, avoiding the obligatory recurrent arch obstruction as the result of somatic growth.

The incidence of recurrent aortic arch obstruction in PHACE syndrome (which seems to be greater than in non-PHACE patients)^{2,4} is likely not only related to the use of prosthetic material but also to intrinsic abnormalities of the aortic wall. Histopathologic analyses

of explanted aortic tissue demonstrate mural zones of scarring and necrosis with decrease in smooth muscle cells and elastic fibers.^{4,6} The long-term effects of these abnormalities on outcomes is unknown.

Given the rarity of this disease, it is worth highlighting a few important points:

- One should suspect PHACE syndrome when faced with an unusual pattern of aortic coarctation and brachiocephalic artery anomalies. Although a large facial hemangioma is present in most patients, it is not a requirement for possible PHACE.
- All patients with suspected PHACE syndrome should undergo cross-sectional imaging of the aortic arch and head and neck vessels, in addition to magnetic resonance imaging of the brain.
- Patients may have airway hemangiomas that can become life-threatening if unrecognized.⁷ Caragher et al² used magnetic resonance imaging as the sole airway diagnostic modality; however, laryngobronchoscopy at the time of surgery should be strongly considered.
- Management of these complex patients requires a multidisciplinary approach.

As illustrated by Caragher et al,² an individualized and creative approach is necessary to address the complex arch anomalies in patients with PHACE syndrome. Long-term follow-up of these patients through case series and the International PHACE Syndrome Registry⁶ is necessary to better determine the optimal management strategy.

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