Corkscrew aortic arch in PHACES syndrome: Multimodal imaging of an unusual morphology of tortuous aortic arch in a rare but well-defined syndrome

Ronak Sheth, Arvind Sahay Singh, Sreeja Pavithran, Kothandam Sivakumar
Department of Pediatric Cardiology, Madras Medical Mission, Chennai, Tamil Nadu, India

ABSTRACT

PHACES syndrome, a diffuse aortocraniocerebral vasculopathy, is a neural tube migration disorder, characterized by aortic coarctation and aberrant arch branches. Clinical diagnosis, echocardiography, and surgical management of coarctation in this syndrome are challenging due to peculiar morphological differences. Corkscrew aortic arch, an extreme tortuosity of the aortic arch described in arterial tortuosity syndrome, is not reported in PHACES syndrome so far. Multimodal imaging of this unusual corkscrew aortic arch in two patients with PHACES syndrome is presented.

Keywords: Aortic arch anomaly, corkscrew aorta, hemangioma, neural crest disorder, PHACES syndrome, tortuous aorta

INTRODUCTION

Neural crest migration plays a role in embryogenesis of aortic arch. An early embryonic insult on neural crest-derived structures leads to a vasculopathy named PHACES syndrome, an acronym for Posterior fossa malformations, Hemangiomas, Arterial, Cardiac, Eye, and Sternal abnormalities.[1] Cardiovascular malformations, often aortic coarctation, occur in 40% of patients.[2,3] The peculiarity of aortic arch abnormality in this syndrome includes tortuosity of the arch vessels, involvement of transverse arch, stenosis or agenesis of branches, and frequent aberrancy of subclavian artery, which makes clinical and echocardiographic diagnosis as well as surgical management very challenging.[4] Corkscrew aorta is a term used to describe extreme tortuosity and often seen in arterial tortuosity syndrome, an entirely different single-gene disease involving SLC2A10 gene in chromosome 20q13 that encodes GLUT10 transporter.[5,6] Multimodality imaging of corkscrew aorta in two different patients with PHACES syndrome is presented.

CASE REPORTS

A 3-month-old infant born of nonconsanguinous marriage with extensive left frontal facial hemangioma and smaller right frontotemporal hemangiomas was referred for a cardiac evaluation [Figure 1]. Her growth was normal, and there was no history of seizures, respiratory distress, or stridor. There was no cranial bruit. There were no sternal or midline abdominal defects. Magnetic resonance imaging of the brain showed a severely narrowed left internal carotid artery collateralized through the circle of Willis [Figure 2]. There were no intracranial aneurysms or infarcts. Clinical cardiovascular examination, chest radiograph, and electrocardiogram were normal. There was no major intraocular pathology.

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Address for correspondence: Dr. Kothandam Sivakumar, Department of Pediatric Cardiology, Madras Medical Mission, 4A, Dr. J J Nagar, Mogappair, Chennai - 600 089, Tamil Nadu, India. E-mail: drkumarsiva@hotmail.com

Echocardiogram revealed mild concentric left ventricular hypertrophy and a left aortic arch with a marked tortuosity and corkscrew appearance of its distal part [Figure 3 and Video 1]. The left common carotid artery was not well visualized; the left subclavian artery originated from the tortuous segment that gave a peak and mean Doppler gradient of 35 and 11 mmHg. A computed tomographic aortogram confirmed a tortuous distal aortic arch with coarctation and extremely tortuous coiling right subclavian artery [Figure 4 and Video 2].

A conservative follow-up was suggested in view of preserved femoral pulses and ventricular function. Oral propranolol for the next 10 months facilitated the regression of facial hemangioma [Figure 1]. The growth and development were adequate on follow-up. The aortic arch tortuosity and gradients did not progress, and the ventricular function was preserved.

Aortogram of another historic patient 15 years ago, a young girl with similar facial hemangioma, brisk left arm pulses, and weak pulses in other limbs showed a corkscrew aorta in the right aortic arch associated with severe hypoplasia of the right carotid artery and significant arch gradients [Figure 5 and Video 3]. Her parents declined surgery after explanations about the complexities in the anatomy and surgical approach.

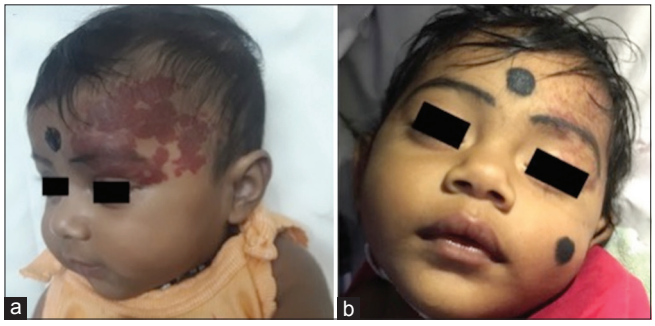

Figure 1: A large left frontal cutaneous hemangioma at 3 months of age (a), regressed on follow-up on oral propranolol therapy (b)

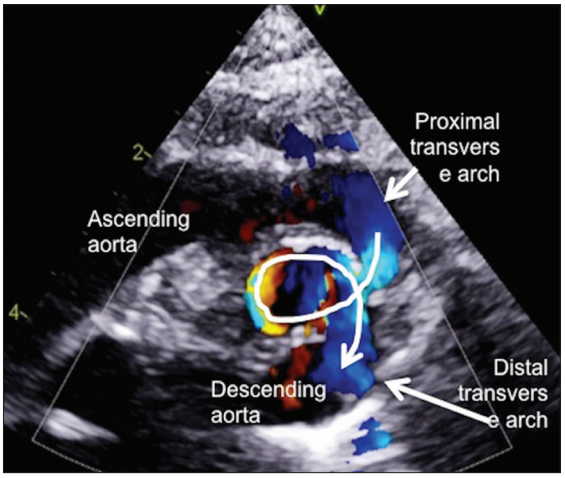

Figure 3: Suprasternal long-axis view of the aortic arch shows marked corkscrew tortuosity of the distal aortic arch with turbulent color Doppler flows

DISCUSSION

Neural crest cells migrate to cranium, pharyngeal arches, and heart and lead to the formation of neural, vascular, cardiac, bony, and other structures.[1] PHACES syndrome refers to a spectrum of developmental field defects with a common denominator of a large segmental craniocervical hemangioma.[2,3] This diffuse aortocraniocerebral vasculopathy results from abnormal neural crest cell migration and peculiarly common in females.[1,7]

A "corkscrew aortic arch" refers to extreme tortuosity occasionally encountered in arterial tortuosity syndrome, but not previously reported in PHACES syndrome.[5,6] The aortic arch anomalies, namely

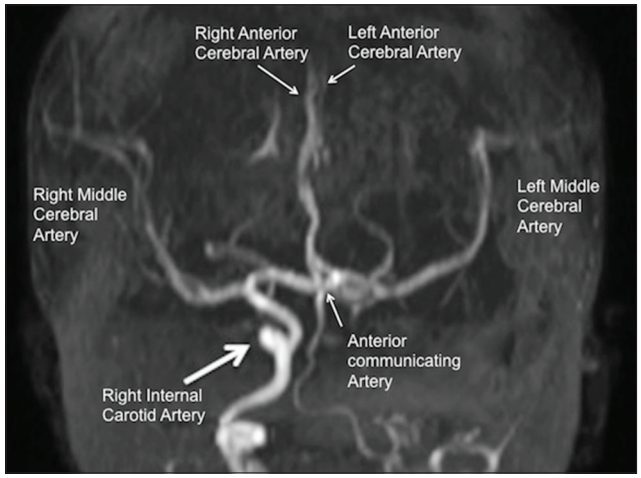

Figure 2: Magnetic resonance angiography of cerebral vessels shows nonvisualization of the left carotid artery and its collateralization from the circle of Willis

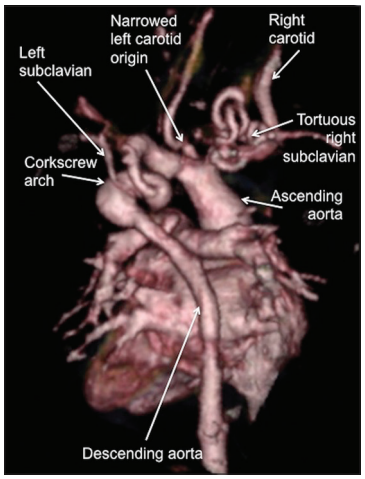

Figure 4: Volume-rendered computed tomographic aortogram viewed from back shows the marked corkscrew tortuosity of the aortic arch and right subclavian artery, marked stenosis of the left carotid artery

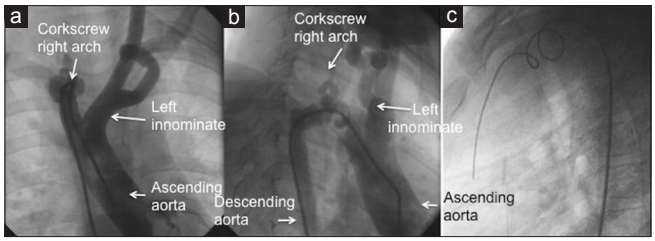

Figure 5: Aortic arch angiogram of the second patient in anteroposterior (a) and right anterior oblique view (b) shows right aortic arch, tortuous left subclavian branch of the left brachiocephalic artery, very stenotic right carotid artery, and stenotic right subclavian artery. A guidewire in lateral view (c) across the arch shows the corkscrew tortuosity

long-segment transverse arch involvement, tortuosity of vessels, aberrancy of subclavian origin in 60% of patients, and stenosis of arch branches, offer unique challenges in diagnosis and management and increase threshold for surgical interventions. The disease severity is underestimated both clinically and on echocardiography.[4] Unlike usual coarctation, bicuspid aortic valve, left heart hypoplasia, and mitral valve anomalies are not seen in PHACES. Surgical techniques are individualized based on arch morphology, especially when there is corkscrew aorta.[8] In patients with right-sided descending thoracic aorta as noted in both our patients, a descending aortic translocation surgery with or without aberrant subclavian reimplantation is adopted as it avoids prosthetic material.[9] Surveillance is crucial as the natural history of PHACES syndrome is less known, though they do better than arterial tortuosity syndrome. In spite of an international registry and broad consensus guidelines, these patients present unique challenges.[10]

Declaration of patient consent

Complete informed appropriate written consent has been obtained from the patient's parents including consent for reporting of patient images and clinical information to be published in the journal. The consent has been granted by them understanding fully that names/initials will not be published and due efforts will be made to conceal identity, but anonymity cannot be guranteed.

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Conflicts of interest

There are no conflicts of interest.

REFERENCES

1. Castillo M. PHACES syndrome: From the brain to the face via the neural crest cells. AJNR Am J Neuroradiol 2008;29:814-5.
2. Frieden IJ, Reese V, Cohen D. PHACE syndrome. The association of posterior fossa brain malformations, hemangiomas, arterial anomalies, coarctation of the aorta and cardiac defects, and eye abnormalities. Arch Dermatol 1996;132:307-11.
3. Metry DW, Dowd CF, Barkovich AJ, Frieden IJ. The many faces of PHACE syndrome. J Pediatr 2001;139:117-23.
4. Bayer ML, Frommelt PC, Blei F, Breur JM, Cordisco MR, Frieden IJ, *et al.* Congenital cardiac, aortic arch, and vascular bed anomalies in PHACE syndrome (from the international PHACE syndrome registry). Am J Cardiol 2013;112:1948-52.
5. Kasar T, Kafalı HC, Türkvatan A, Ergül Y. A rare case of right corkscrew cervical aortic arch associated with retrotracheal aberrant left brachiocephalic vein. Kardiol Pol 2018;76:812.
6. Fernández-Doblas J, Tauron M, Blasco A, Abella RF. Right corkscrew cervical aortic arch. Eur J Cardiothorac Surg 2012;42:903.
7. Bijulal S, Sivasankaran S, Krishnamoorthy KM, Titus T, Tharakan JA, Krishnamanohar SR. Unusual coarctation-the PHACE syndrome: Report of three cases. Congenit Heart Dis 2008;3:205-8.
8. Caragher SP, Scott JP, Siegel DH, Mitchell ME, Frommelt PC, Drolet BA. Aortic arch repair in children with PHACE syndrome. J Thorac Cardiovasc Surg 2016;152:709-17.
9. Herbert J, Guzmán-Pruneda FA, Sumner EE, McKenzie ED. Simultaneous repair of right-sided coarctation and vascular ring. Ann Thorac Surg 2015;100:334-6.
10. Garzon MC, Epstein LG, Heyer GL, Frommelt PC, Orbach DB, Baylis AL, *et al.* PHACE syndrome: Consensus-derived diagnosis and care recommendations. J Pediatr 2016;178:24-3300.