IMAGES **in PAEDIATRIC CARDIOLOGY**

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Abstract

PHACE syndrome is a rare neurocutaneous disorder characterised by an association of infantile haemangiomas with structural anomalies of brain, cerebral vasculature, eye, aorta and chest wall.[1] Coarctation of aorta (COA) is most the common cardiac anomaly reported in PHACE syndrome. COA or interrupted aortic arch in PHACE is unique and complex both in location and character compared to the typical coarctation anatomy. Arterial tortuosity of the cerebral vasculature has been well described in literature in PHACE syndrome. We present a rare case of tortuous aortic arch continuing as descending aorta in an infant with PHACE syndrome.

MeSH: PHACE syndrome, coarctation of aorta, interrupted aortic arch.

Case Report

A 2-month-old infant with a large haemangioma over left side of face, neck and shoulder was referred for cardiac evaluation to rule out congenital heart disease. The child was asymptomatic with no signs of cardiac failure, normal oximetry in room air and unremarkable cardiac examination. 2D-Echo revealed a moderate ostium secundum atrial septal defect (OS-ASD) with left to right shunt. The aortic arch distal to the left internal carotid artery was not traceable. Abdominal aorta showed normal pulsatile Doppler flow pattern.

CT angiography for arch delineation revealed a normal calibre ascending aorta (8.6mm) with a hypoplastic proximal arch (4.3mm) and tortuous distal arch continuing as the descending aorta (4.3 mm). The left subclavian artery was hypoplastic and originated from a tortuous segment of aortic arch with a severely hypoplastic left vertebral artery. There were two large haemangiomas, one in left maxillary area and other over left neck and shoulder extending to the superior mediastinum.

Fig 1: Reconstructed image of Arch and descending aorta showing proximal arch hypoplasia with tortuous distal part of arch and hypoplastic left subclavian from tortuous segment of arch. Descending aorta is same as calibre as transverse arch with no discrete coarctation.

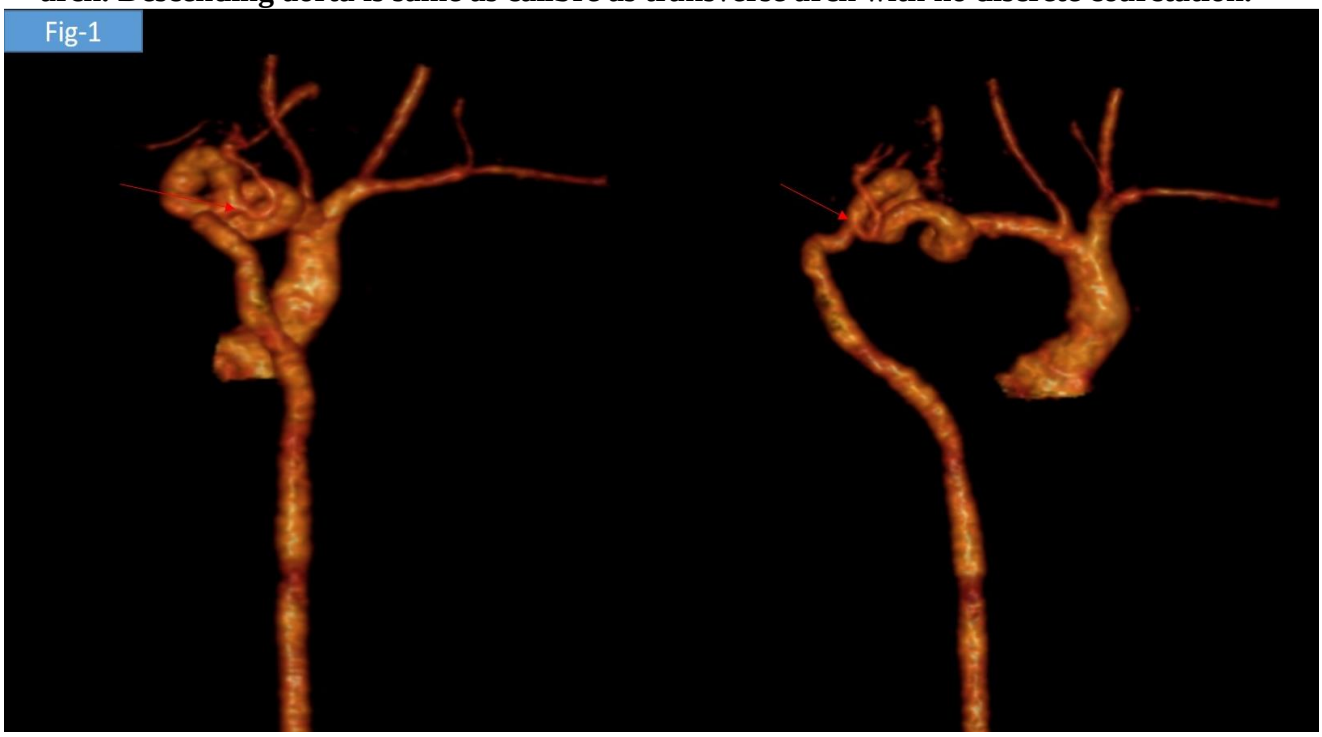
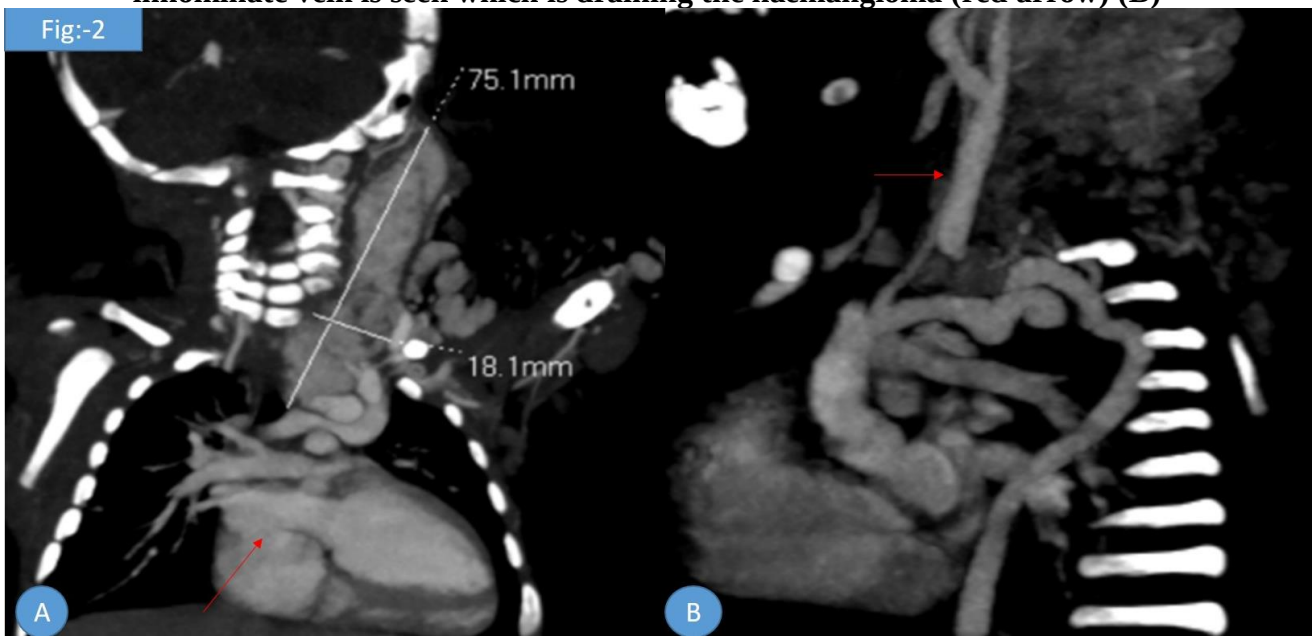
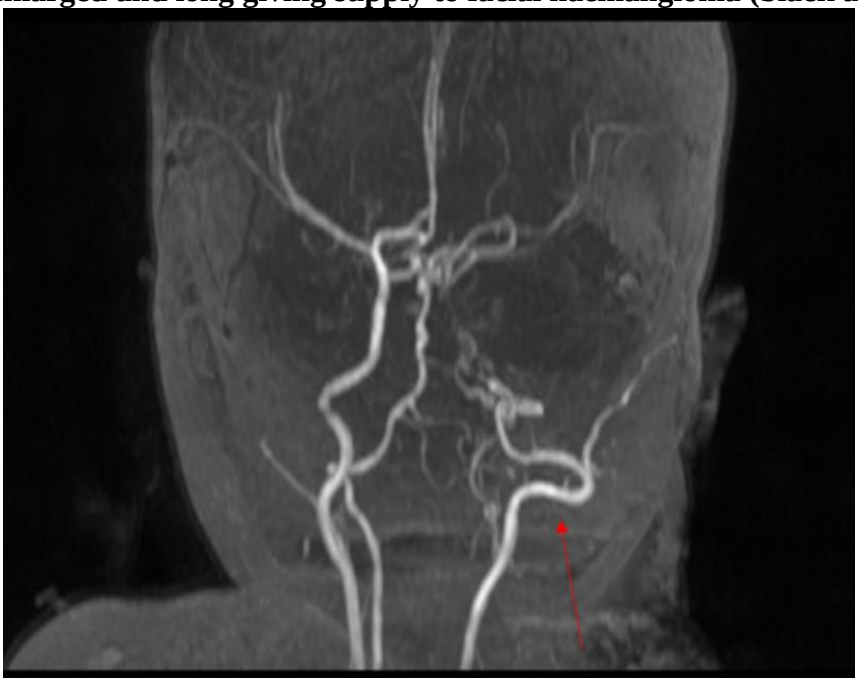


Fig 2: Antero-posterior CT – angiographic image showing large haemangioma extending from neck, shoulder to anterior mediastinum and moderate size ASD (red arrow) (A). Lateral CT angiogram view showing tortuous arch and unobstructed descending aorta. Dilated left innominate vein is seen which is draining the haemangioma (red arrow) (B)



Magnetic Resonance Imaging (MRI) of the brain with angiography revealed hypoplasia of the left sided cerebral circulation. The left anterior cerebral and middle cerebral artery were filling retrogradely from the circle of Willis. The left vertebral artery and its branches to the left pons were also diminutive in size and associated with hypoplasia of the left pons. Ophthalmic evaluation was normal.

Fig 3: MRI Angiogram shows faint vascularity of left half of brain. The left external carotid and is enlarged and long giving supply to facial haemangioma (black arrows).



The child was treated with oral prednisolone and propranolol for the haemangiomas. On follow up after 8 months, the child is developmentally normal with normal blood pressure and there was significant regression of the haemangioma.

Discussion

PHACE syndrome is an association of posterior fossa malformations, facial haemangioma, arterial abnormalities, coarctation of aorta, and eye abnormalities.[1,2] In addition, an "S" has been added to the original acronym, for sternal abnormalities: PHACE (S).[1,2] It is nine times more common in females.[2] The diagnostic criteria for this syndrome requires the presence of a segmental haemangioma > 5cm of head (face or scalp) plus 1 major criteria or 2 minor criteria (involving the cerebrovascular, cardiovascular, or ocular organ systems, the brain structure, or ventral or midline defects).[2]

Cardiovascular anomalies which are considered as major criteria include, a) coarctation of aorta or dysplasia (includes kinking, looping, tortuosity), b) aneurysm, and c) aberrant origin of subclavian artery with or without a vascular ring.[2] Cardiovascular anomalies constituting minor criteria include a) ventricular septal defects (VSD) and, b) right aortic arch/ double aortic arch.[2]

COA constitutes 14.5% of the cardiac manifestations in PHACE syndrome.[2] Unlike isolated COA there is associated unusual dilation and aneurysm formation of the adjacent arch segments in COA observed in PHACE syndrome.[2,3] These anomalies can be associated with abnormality of brachiocephalic vessels (dilation, tortuosity and aberrant subclavian artery).[2]Bayer et al reported aberrant origin of subclavian as the most common cardiovascular anomaly followed by COA.[3] In our case distal arch and descending aorta was tortuous with hypoplastic proximal arch. Despite this hypoplasia and tortuosity, there were no signs and symptoms COA clinically and on

echocardiography. Hence, child was not offered any intervention and we planned to clinically monitor the child at regular intervals.

The pathogenesis of this syndrome is not well understood, however developmental dysplasia of large and medium size arteries has been suggested.[3, 4] Surgical corrections of COA in PHACE have been described in symptomatic infants. Histopathologic study of the diseased aorta shows abnormalities of the intimal and medial layers i.e. fibrosis of inner tunica media and loss of smooth muscle and elastin (arteriopathy).[3,4] Similarly, distortion and aneurysm of aortic large and medium sized vessels can be seen in Aortic Tortuosity Syndrome (ATS) but this condition is an autosomal recessive disorder due to mutation in *SLC2A10* gene. It is also characterised by other features of connective tissue disorder such as hyper-extensile joints, joint laxity, hypotonia and abdominal wall/diaphragmatic hernia.[5]

Cerebrovascular abnormalities are the most common extra cutaneous manifestations and include dysplasia, narrowing, aberrant course or origin of the brain arteries, or persistence of embryonic anastomosis.[2,6] These arterial anomalies tend to occur either ipsilateral to the cutaneous haemangiomas, or bilaterally.[2]

Long-term follow up is required to understand the natural history of these anomalies as both cardiac and cerebrovascular are the major cause for morbidity in these patients.

References

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