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Case Presentation

PHACES syndrome with moyamoya vasculopathy – a case report

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Introduction

"PHACES" (OMIM 606519) is a neurocutaneous disorder, and facial hemangiomas are the hallmark of this syndrome. The syndrome encompasses posterior fossa brain malformations, facial hemangiomas, arterial anomalies, aortic coarctation, cardiac anomalies, eye abnormalities, and sternal defects.

Case Report

A baby girl born of a non-consanguineous marriage presented at 31 months of age with a segmental hemangioma (segments V1&2) involving the right side of the face, retroauricular area, neck, chest, and posterior palate (Figure 1 A,B,C,D), which developed 3-4 weeks after birth. There was mechanical ptosis, strabismus, and amblyopia of the right eye. She also had a sternal cleft at birth, which was corrected surgically (Figure 1 E).

Figure 1

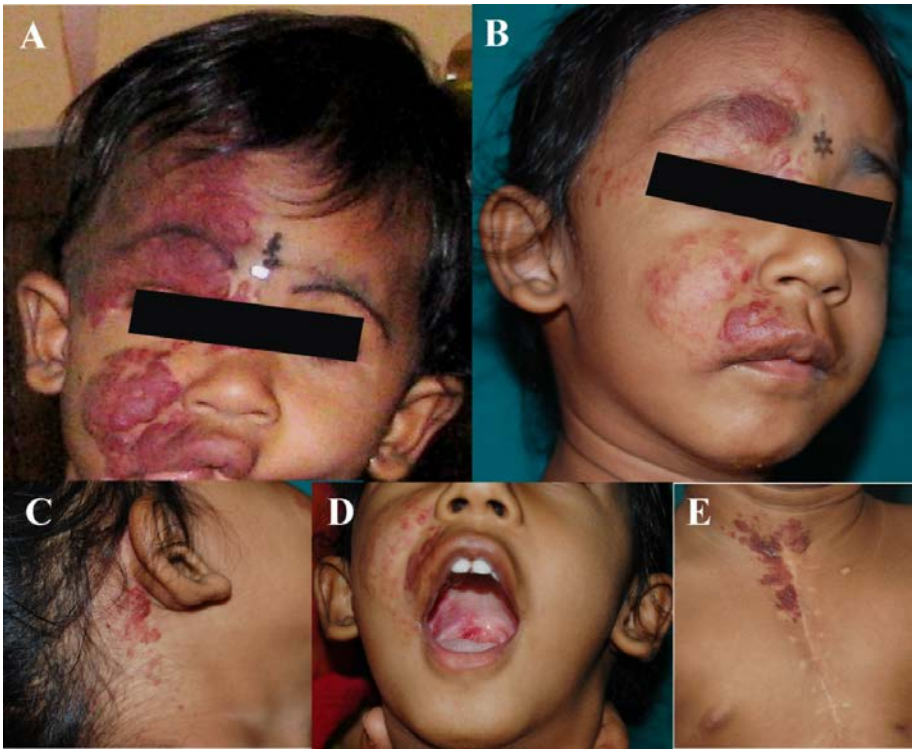

Figure 1. A large segmental facial haemangioma involving the right frontotemporal and maxillary segments at one year (A) and (B) showing mild regression at two and a half years of age. Haemangioma involving the retroauricular area (C), palate (D) neck and sternal area (E). Post-operative scar following sternal repair (E)

Three months prior, she had tonic posturing followed by monoparesis of the right lower limb, which recovered within 2 hours. Magnetic Resonance Imaging (MRI) of the brain with angiography done at that time showed lacunar infarcts, ischemic lesions in the periventricular white matter and centrum semiovale, bilaterally. The right internal carotid artery (ICA), distal basilar artery, and right posterior cerebral artery were occluded. Terminal ICA stenosis was seen on both sides with collaterals in the circle of Willis region giving the 'puff of smoke' (moyamoya) appearance. Flair sulcal hyperintensity was seen, which is proposed to be owing to slow flow in the leptomeningeal collaterals (Ivy sign) (Figure 2 A,B,C). She was started on aspirin and phenytoin, which was discontinued by the parents and she was lost to follow up.

At 4 years of age she presented with left acute hemiparesis and brief left facial focal seizures. A CT scan then showed right anterior cerebral artery infarct (Figure 2 D). She was restarted on aspirin and is asymptomatic currently, with average scholastic performance. The parents were also given the option of revascularization surgery, which they declined. Echocardiography showed trace tricuspid regurgitation and three pulmonary veins draining into the left atrium.

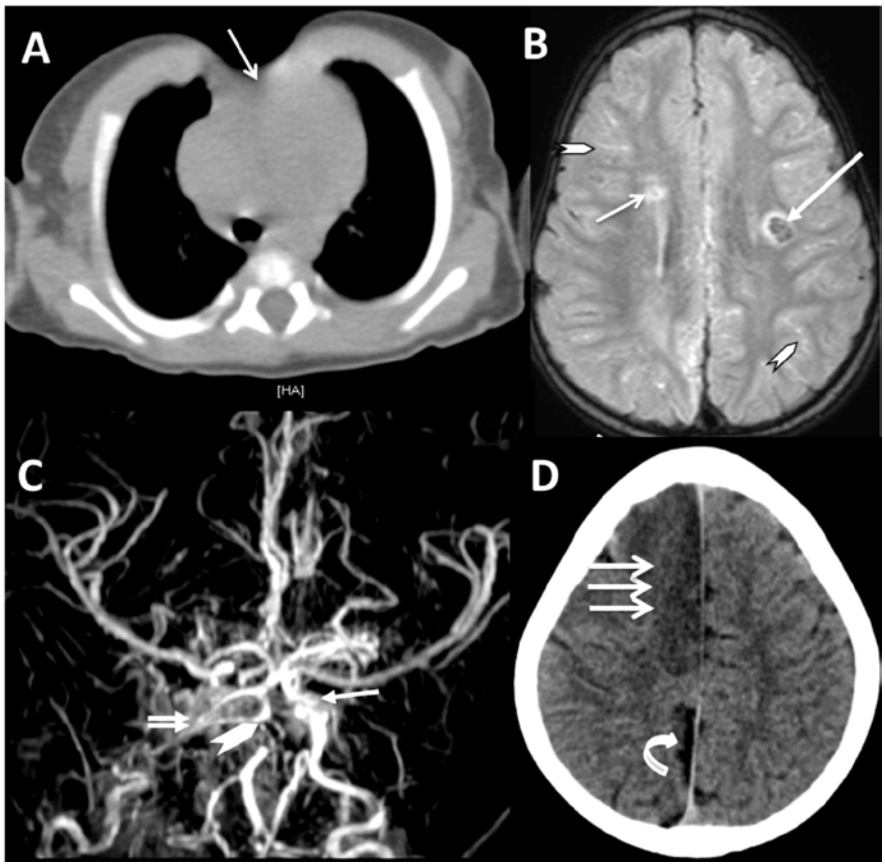

Figure 2 Noncontrast CT thorax (A) shows the sternal cleft (arrow in A). FLAIR axial image (B) of the brain, above the level of the ventricles, showing ischemic changes and lacunar infarcts in the centrum semiovale on both sides (arrows in B). FLAIR sulcal hyperintensity (chevrons in B) is seen due to slow flow in the leptomeningeal collaterals. MR angiogram (C) shows occlusion of right ICA (double arrows show the expected location of right ICA which is not seen as it is occluded), stenosis of terminal left ICA (arrow in C) and occlusion of basilar artery (chevron in C). 'Puff of smoke' is the increased multiple fine linear opacities seen around the circle of Willis. Axial non-contrast CT of the brain (D), at age of 4years, in supraventricular region, showing a subacute infarct in right ACA territory (triple arrows in D) and old infarct in right parietal parasagittal region (curved arrow in D).

Discussion

PHACES syndrome is present in 2% of children with facial hemangiomas and 20% with "segmental" facial hemangiomas. It is more frequent in females [1,2]. Our patient fulfilled the criteria for PHACES as described by Metry (facial haemangioma >5cm, arterial stenosis with moyamoya vasculopathy, sternal cleft) [3].

PHACES is an under-recognized cause of pediatric acute ischaemic stroke (AIS). The average age of presentation of pediatric AIS in PHACES is 8.8 months [4]. Moyamoya vasculopathy, characterized by progressive narrowing of ICA and abnormal vascular networks at the base of the brain (moyamoya vessels) in PHACES was first reported by Burrows et al [5]. Its reported prevalence in PHACES is 7% [4] and affected patients are at risk for ischaemic stroke. Our patient was slightly older than the reported usual age at onset of neurological sequelae which is between 9-18 months. Serial neuroimaging may be justified in

patients with cerebrovascular anomalies, because neurosurgical revascularization procedures such as pial synangiosis could potentially reduce AIS-related morbidity and mortality.

Conclusion

Our case illustrates the clinical course of PHACES syndrome with moyamoya vasculopathy. All patients with a large facial hemangioma should be screened for PHACES syndrome. Early recognition of the associated vascular pathology may enable presumptive interventional treatment before potentially devastating and irreversible sequelae occur.

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