PHACE syndrome in antenatally diagnosed posterior fossa anomaly

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ABSTRACT

PHACE is a neurocutaneous syndrome, an acronym to describe patients with facial segmental hemangiomas and other malformations. We describe a newborn antenatally diagnosed to have posterior fossa anomaly and subsequently as PHACE syndrome.

Key words: Dandy–Walker spectrum, hemangioma, vascular neurocutaneous disorder

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INTRODUCTION

Infantile hemangiomas are commonly seen birthmarks with an incidence of 4%-5%.[1] Pascual-Castroviejo, in 1978, described the association between infantile hemangiomas and brain anomalies.[2] PHACE syndrome is a neurocutaneous syndrome, an acronym, coined by Freiden *et al.* to include posterior fossa (PF) malformations coexisting with facial segmental hemangiomas, *a*rterial anomalies, *c*ardiac anomalies/arterial coarctation, and *e*ye abnormalities. The diagnosis of definite and possible PHACE syndrome is based on criteria proposed by Metry *et al*.[3] Here we report a newborn with antenatal diagnosis of PF anomaly and subsequently PHACE syndrome.

CASE REPORT

A young primigravida mother delivered a female baby at term by vaginal route with Apgar scores of 8 and 9 at 1 and 5 minutes respectively. Routine fetal scan at 34 weeks had revealed a large posterior fossa cyst. Baby weighed 3140 g at birth and head circumference was 36 cm. Physical examination showed large hemangioma in facial segmental regions 1 and 4 extending from the forehead, including both eyelids on to the maxillary area, and right side of the scalp (over frontal, and parts of parietal and occipital areas) [Figure 1].[3] The widest dimension measured was more than 5 cm. Oropharyngeal examination revealed a hemangioma over the soft palate. The head shape was normal and there was no occipital prominence. Other than that stridor her systemic examination was normal. Flexible video laryngoscopy showed congenital laryngomalacia, and there was no airway hemangioma. Magnetic resonance imaging of the brain confirmed the diagnosis of Dandy–Walker spectrum with a large posterior fossa cyst communicating to fourth ventricle with vermian hypoplasia [Figure 2]. There was no hydrocephalus, midline anomalies, or neuronal migration disorder, and demonstrable vessels at the base of the skull were normal. A diagnosis of PHACE syndrome was considered and detailed evaluation was done. Thyroid screening was normal. (thyroxine : 7 µg/dL, thyroid stimulating hormone: 4.3 mIU/mL). Echocardiogram and Doppler did not show vascular anomalies; aorta and branches were normal. Eye examination did not reveal arteriovenous malformations in the fundus, and disc was normal. Hearing was normal. Postnatal course was uneventful and the baby was discharged with advice regarding continued follow-up. At follow-up visit of 6 weeks the size of hemangioma was regressing; however, clinical photograph could not be taken at 3 months and later as the child was lost to follow-up.

DISCUSSION

The diagnosis of PHACE syndrome in the index case was made in the presence of facial segmental hemangioma measuring >5 cm with structural PF lesion. PHACE is considered to be an error during neural crest development and may be present in approximately

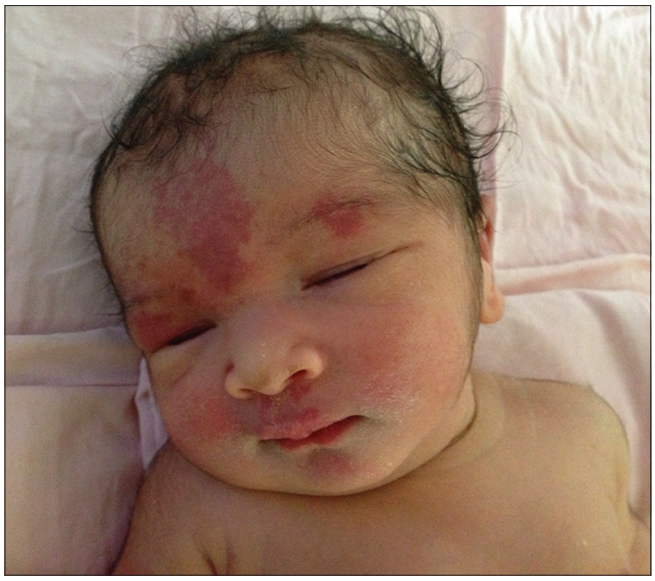

Figure 1: Facial hemangioma over segments 1 and 4

20%-31% of cases with large hemangioma with distribution corresponding to facial developmental unit and has a female preponderance.[4] Cerebrovascular and cardiovascular anomalies are the most common extracutaneous manifestations. The most common structural brain malformations are of cerebellum and PF structures such as Dandy–Walker complex, with or without cerebellar hypoplasia. Supratentorial lesions are seen in less than one-third of patients. Airway hemangioma may be seen in 52% of cases and is an emergency.[5] In such cases systemic steroids may be safer than beta-blockers if cardiac and cerebral vasculature anomalies are not ruled out.[4] Close follow-up visits are advised for growth monitoring and hormonal evaluation as endocrine disturbances are commonly seen. Vascular anomalies can result in cerebrovascular stroke incidents and hence magnetic resonance angiogram is warranted in future. Neurodevelopmental sequelae are known in later life.[6]

To conclude, cerebral anomalies can be part of vascular neurocutaneous disorder, especially when associated with large segmental hemangiomas. These necessitate a detailed work-up.

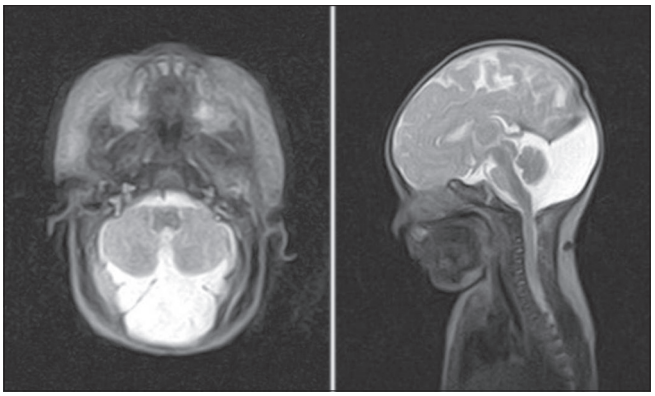

Figure 2: Axial and sagittal T2-weighted images: Large posterior fossa cerebrospinal fluid intensity cyst with inferior vermian hypoplasia and narrow communication of fourth ventricle with the cyst

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