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Beyond the Hemangioma: Imaging Findings of PHACE Syndrome in Adolescence

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ABSTRACT

PHACE syndrome is an uncommon neurocutaneous disorder in which a segmental infantile hemangioma may reveal occult congenital brain and cerebrovascular abnormalities. We report the case of a 16-year-old girl with a persistent right facial hemangioma, first noticed at two months of age, with no neurological, developmental, or cardiorespiratory symptoms. Examination showed a segmental plaque larger than 5 cm involving the right frontotemporal, periorbital, upper eyelid, and nasal regions. CT confirmed a vascular soft-tissue lesion without intra orbital extension. MRI demonstrated right cerebellar hemispheric and vermian hypoplasia with communication between the fourth ventricle and the retrocerebellar cerebrospinal fluid space, while time-of-flight MR angiography identified a persistent primitive right dorsal ophthalmic artery arising from the cavernous internal carotid artery. These findings fulfilled the criteria for a definite PHACE syndrome. This case highlights the value of integrated neurovascular imaging in detecting clinically silent abnormalities and illustrates an uncommon cerebrovascular manifestation of PHACE syndrome.

KEYWORDS

PHACE syndrome, infantile hemangioma, posterior fossa malformation, cerebellar hypoplasia, magnetic resonance angiography

MAIN ARTICLE

INTRODUCTION

PHACE syndrome is a developmental disorder in which an infantile hemangioma may serve as an external marker of clinically occult congenital abnormalities. Because neurological and cardiovascular examinations may remain normal, targeted MRI and MR angiography plays a central role in detecting associated structural and vascular lesions [1,2]. We report the case of an adolescent with a persistent segmental facial hemangioma associated with ipsilateral posterior fossa abnormalities and a rare persistent primitive dorsal ophthalmic artery.

CASE PRESENTATION

A 16-year-old girl was referred for evaluation of a right facial infantile hemangioma, first noticed at two months of age and still present during adolescence. She had no neurological, developmental, cardiorespiratory, feeding, or swallowing symptoms. Neurological and cardiovascular examinations were normal, with no associated midline anomalies.

Physical examination revealed a large, right-sided, segmental erythematous vascular plaque involving the frontotemporal, upper eyelid, periorbital, and nasal regions. The lesion extended over 5 cm and predominantly involved the right frontotemporal and frontonasal facial segments. No scalp, cervical, or oral cavity involvement was observed, and there were no symptoms suggestive of upper airway involvement. (Figure 1).

Contrast-enhanced CT of the face showed irregular thickening of the right frontotemporal, nasal, upper eyelid, and periorbital soft tissues, measuring up to 9 mm and extending over 5 cm. The lesion contained enhancing serpiginous vascular channels and showed no intra orbital extension (Figure 2).

Brain magnetic resonance imaging revealed a congenital posterior fossa malformation including hypoplasia of the right cerebellar hemisphere and cerebellar vermis. A retrocerebellar cerebrospinal fluid-signal space communicated with the fourth ventricle, without significant associated mass effect (Figure 3).

Time-of-flight (TOF) MR angiography revealed a persistent primitive right dorsal ophthalmic artery arising from the cavernous segment of the right internal carotid artery and coursing

anteriorly toward the orbit (Figure 4). No additional intracranial arterial abnormality was identified.

The association of a segmental facial infantile hemangioma measuring more than 5 cm with a major structural posterior fossa anomaly fulfilled the consensus diagnostic criteria for definite PHACE syndrome. The persistent primitive dorsal ophthalmic artery represented an additional associated cerebrovascular anomaly.

At the time of manuscript preparation, no specific treatment had been initiated. Multidisciplinary follow-up was planned to assess the neurological and vascular findings and determine the management of the persistent cutaneous lesion according to its functional and cosmetic impact.

DISCUSSION

PHACE syndrome is a rare multisystem neurocutaneous disorder characterized by a large segmental infantile hemangioma, usually involving the face or scalp, associated with congenital abnormalities of the posterior fossa, cervicocerebral arteries, cardiovascular system, eyes, or ventral midline structures. Its clinical presentation is heterogeneous, and some extracutaneous abnormalities may remain asymptomatic, therefore, recognition of the characteristic hemangioma should prompt systematic clinical and radiological assessment [1,2].

Definite PHACE syndrome is diagnosed when a facial or scalp hemangioma larger than 5 cm is associated with one major criterion, or when a large segmental hemangioma of the neck or upper trunk is associated with two major criteria. In our patient, the large segmental facial hemangioma with ipsilateral cerebellar and vermian hypoplasia fulfilled these criteria, while the persistent primitive ophthalmic artery provided an additional supporting finding (Table 1).

Table 1 : Diagnostic Criteria for PHACE Syndrome

System	Major criteria	Minor criteria
Brain (vascular)	Arterial dysplasia, stenosis, or occlusion; arterial absence or hypoplasia; aberrant arterial origin or course; persistent trigeminal artery; saccular aneurysm	Persistent embryonic arteries, including the proatlantal intersegmental, primitive hypoglossal, and primitive ophthalmic arteries
Brain (structural)	Posterior fossa anomalies, including the Dandy-Walker complex and unilateral or bilateral cerebellar hypoplasia or dysplasia	Extra-axial lesion compatible with an intracranial hemangioma
Cardiovascular	Aortic arch anomalies, including coarctation, aneurysm, or aberrant subclavian artery with or without a vascular ring	Ventricular septal defect; right-sided or double aortic arch
Ocular	Posterior segment abnormalities, including persistent fetal vasculature, retinal vascular anomalies, optic disc anomalies, optic nerve hypoplasia, coloboma, or peripapillary staphyloma	Anterior segment abnormalities, including sclerocornea, cataract, coloboma, or microphthalmia
Midline	Sternal cleft; supraumbilical raphe	Other sternal defects; hypopituitarism; ectopic thyroid

Adapted from the consensus diagnostic criteria proposed by Metry et al. [3]. PHACE: posterior fossa anomalies, hemangioma, arterial anomalies, cardiac anomalies, and eye anomalies.

Infantile hemangiomas are benign vascular tumors characterized by rapid proliferation during infancy followed by gradual involution over several years. In PHACE syndrome, they are usually large, segmental, and located on the face or scalp, with a distribution that may predict associated abnormalities: frontotemporal (S1) and frontonasal (S4) hemangiomas are more often linked to structural brain, cerebrovascular, and ocular anomalies, whereas mandibular (S3) lesions are more commonly associated with airway, cardiovascular, and ventral midline abnormalities. In our patient, the frontotemporal and frontonasal distribution was consistent

with the associated posterior fossa and arterial abnormalities. The main differential diagnoses include capillary malformations associated with Sturge-Weber syndrome, venous or lymphatic malformations, arteriovenous malformations, and, less commonly, aggressive soft-tissue tumors. The onset age, characteristic clinical course, segmental distribution, and vascular imaging findings usually allow its distinction [4,5].

Structural brain abnormalities in PHACE syndrome most commonly involve the posterior fossa and range from unilateral cerebellar hypoplasia to abnormalities within the Dandy-Walker spectrum, including vermian hypoplasia and communication of the fourth ventricle with a retrocerebellar cerebrospinal fluid space. Less frequent supratentorial findings include cortical dysplasia, neuronal migration abnormalities, and callosal agenesis. These structural abnormalities often occur on the same side as the facial hemangioma and may coexist with cerebrovascular anomalies. In our patient, right cerebellar and vermian hypoplasia with fourth ventricular communication was therefore consistent with the recognized neuroradiological spectrum of PHACE syndrome [3,6].

Cerebrovascular abnormalities in PHACE syndrome reflect disturbed embryonic arterial development and may range from incidental anatomical variants to hemodynamically significant stenotic or occlusive lesions. In our patient, persistence of a primitive right ophthalmic artery arising from the cavernous internal carotid artery supports this developmental vascular spectrum and constitutes an additional minor diagnostic criterion. Although an isolated persistent embryonic artery is generally considered a low-risk finding, its presence reinforces the need for complete vascular assessment to exclude associated arterial dysplasia or flow-limiting lesions [1,7,8].

Management of PHACE syndrome should be individualized according to the hemangioma's activity, associated abnormalities, and cerebrovascular risk. Propranolol is primarily considered for proliferating or function-threatening infantile hemangiomas, whereas persistent cutaneous changes in adolescence may be evaluated for laser therapy or surgery. In our patient's case, the therapeutic approach was deferred pending multidisciplinary assessment of the functional and cosmetic impact of the residual lesion [1].

CONCLUSION

This adolescent presentation illustrates the long-term phenotypic variability of PHACE syndrome and the coexistence of a posterior fossa malformation with an uncommon ophthalmic arterial variant. Therapeutic management should consider primarily the presence of a cerebrovascular risk and the functional or cosmetic impact of the residual hemangioma. Long-term multidisciplinary follow-up and further observations may help clarify the clinical significance of rare embryonic arterial variants in this population.

FIGURES :



Figure 1 : Clinical appearance of the right facial hemangioma : large segmental erythematous vascular plaque involving the right frontotemporal, supraorbital, upper eyelid, periorbital, and nasal regions.

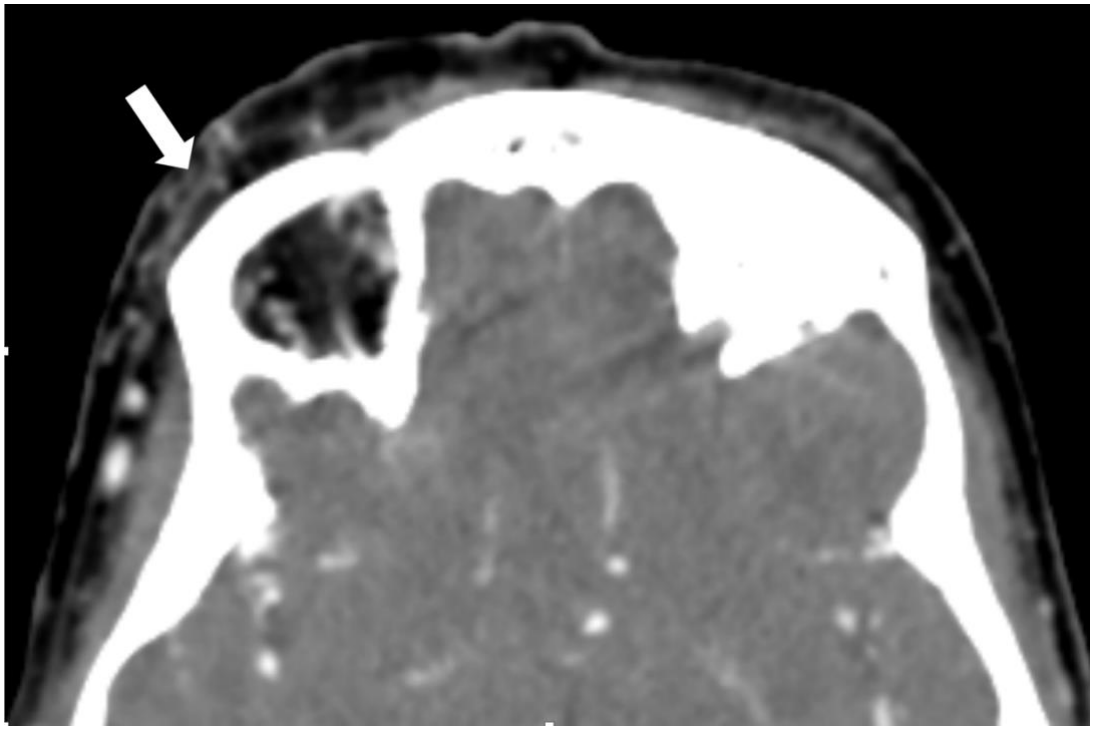


Figure 2: Axial contrast-enhanced CT demonstrates irregular right facial soft-tissue thickening containing enhancing serpiginous vascular channels (arrow).

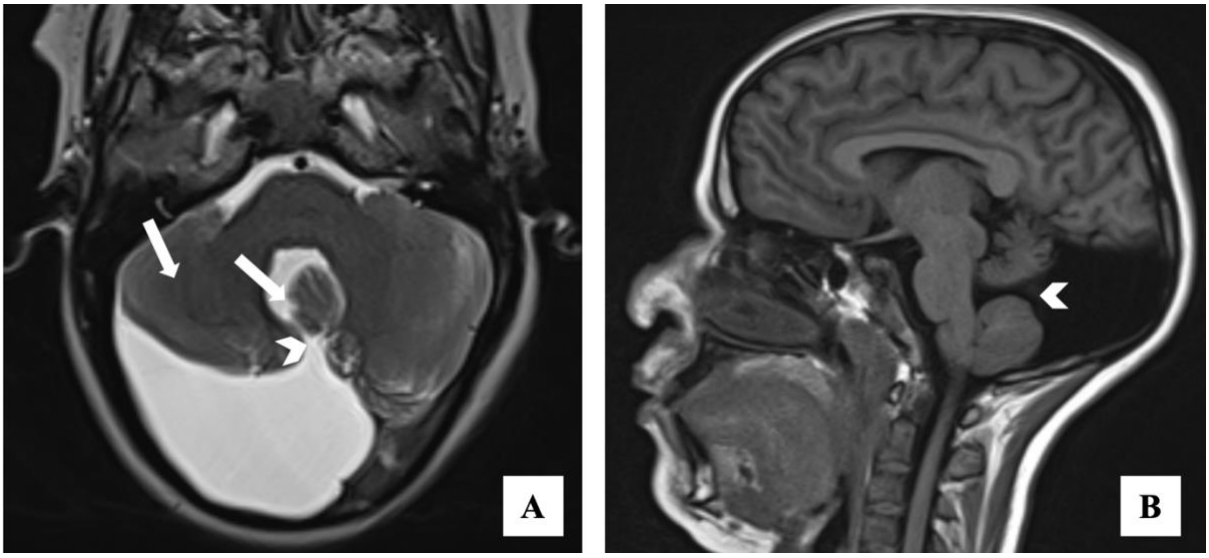


Figure 3 : Axial T2 (A) and midsagittal T1(B) weighted images show right cerebellar and vermian hypoplasia (arrow) with communication between the fourth ventricle and the retrocerebellar cerebrospinal fluid space (arrowhead).

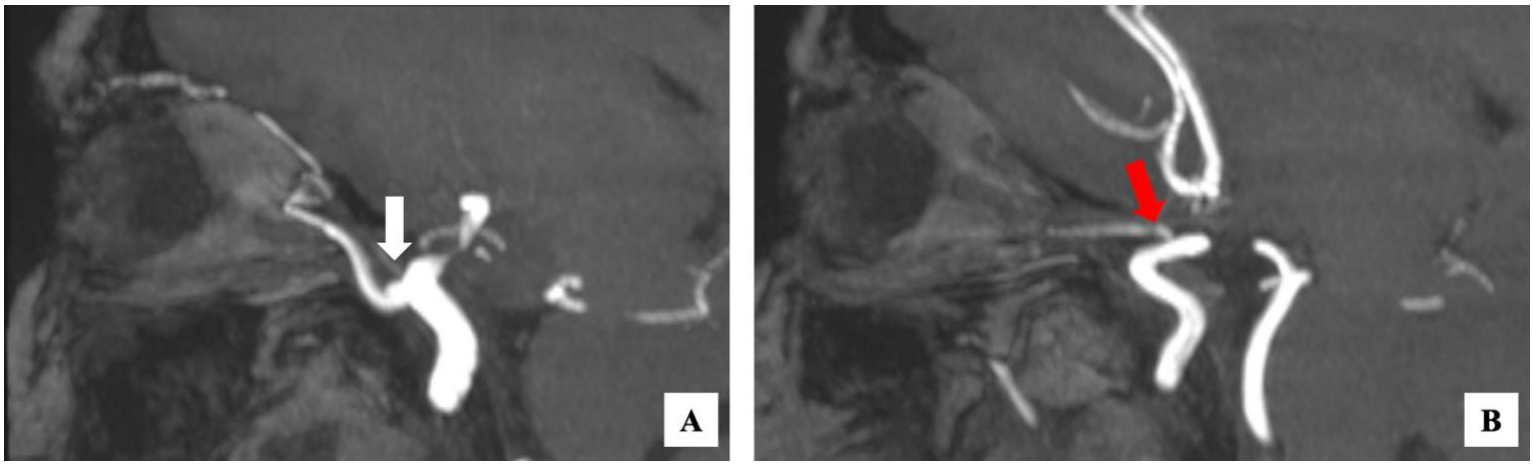


Figure 4 :
A : Sagittal 5-mm maximum intensity projection TOF MR angiographic images show a persistent primitive right dorsal ophthalmic artery arising from the cavernous segment of the internal carotid artery (white arrow).
B : Note the normal origin of the left ophthalmic artery from the supraclinoid internal carotid artery on the contralateral side (red arrow).

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