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CASE REPORT PEER REVIEWED | OPEN ACCESS

Faced with PHACES: A photo series of its progression and overview of clinical management

Kiana Banafshay, Brooke Walterscheid, Michelle Tarbox

ABSTRACT

Introduction: Posterior fossa anomalies, hemangioma, arterial anomalies, cardiac anomalies, eye anomalies, and sternal anomalies (PHACES) syndrome is a rare, multi-system disorder.

Case Report: We illustrate a case of a 1-month-old female who presented emergently with an enlarging, large, left-sided hemangioma segmentally distributed over the ophthalmic branch of the trigeminal nerve (V1) region prompting clinical concern for PHACES syndrome. Further workup was recommended to evaluate for extracutaneous involvement, which subsequently revealed coarctation of the aorta and multiple arterial abnormalities.

Conclusion: The vast array of potential, devastating complications linked to PHACES syndrome highlights the importance of prompt identification when confronted with hemangiomas, especially in the pediatric population. We further present a photo series demonstrating the evolution of our patient's infantile hemangioma from its subtle initial stages through its rapid evolution leading to her emergent presentation, emphasizing the importance of early clinical suspicion and intervention.

Keywords: Infantile hemangioma, PHACE, PHACES, Propranolol, Segmental hemangioma

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INTRODUCTION

Posterior fossa anomalies, hemangioma, arterial anomalies, cardiac anomalies, eye anomalies, and sternal anomalies (PHACES) syndrome is a rare, multi-system disorder that stands for posterior fossa anomalies, hemangiomas, arterial anomalies, cardiac anomalies, eye anomalies, and sternal anomalies. It can also be associated with hearing and endocrine abnormalities. Its prevalence is remarkably low, as it is seen in less than one per million individuals. Though its etiology is unknown, a segmental infantile hemangioma is often the presenting finding and warrants further investigation. We illustrate a case of a 1-month-old female with high clinical suspicion for PHACES syndrome. Furthermore, we provide an illustrative series of photographs that chronicles the progression of our patient's segmental hemangioma. This case highlights the syndrome's complexity and underscores the significance of having a high index of clinical suspicion, educating providers on its earliest and most subtle signs, and reviewing its overall management.

CASE REPORT

A 1-month-old female with no prior medical history presented to the emergency room for an enlarging, left-sided hemangioma associated with obstruction and swelling of the left upper eyelid. Per the patient's mother, she developed a small pinkish-red spot on the left temple

at one week old, which progressed in size and density over the next several weeks (Figure 1). She also had pectus excavatum, noted by her pediatrician, but it had been less prominent as the patient properly gained weight. Upon obtaining a review of systems, it was elicited that the patient recently exhibited shortness of breath with prolonged feeding times. Physical examination showed a well-demarcated red-violaceous, telangiectatic, dense plaque segmentally distributed over the V1 region. Differential diagnoses at the time included infantile hemangioma, Sturge-Weber syndrome, and PHACES syndrome. Given the large size of the lesion (>5 cm), the concern for PHACES syndrome was high, and further workup was recommended to evaluate for extracutaneous involvement, including an echocardiogram, chest X-ray, and abdominal X-ray. An electroencephalogram was pursued in lieu of magnetic resonance imaging (MRI) to rule out acute posterior fossa abnormalities as the patient was hemodynamically stable, with the intent to pursue an MRI at a later time. An echocardiogram later revealed coarctation of the aorta with a reduced lumen of less than 2 mm, patent foramen ovale, and a small muscular ventricular septal defect. The echoencephalogram, chest X-ray, and abdominal X-ray returned with normal findings. Her complete extracutaneous findings are documented (Figure 2). She was transferred to a pediatric cardiothoracic surgical center to undergo open heart surgery for repair of the aortic coarctation. While undergoing care at that facility, she was initiated on oral propranolol therapy with weight-based dosing; her hemangioma has shown promising involution since its initiation.

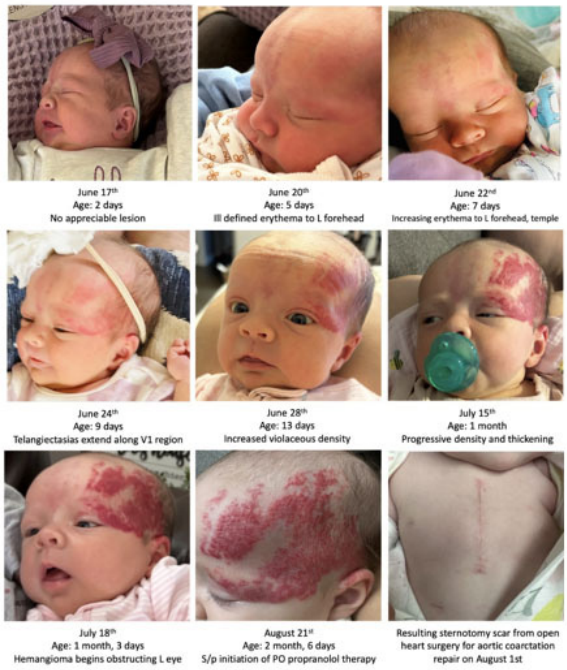

Figure 1: Patient's clinical course illustrating development and advancement of the hemangioma over one month's time, followed by beginning stages of involution upon initiating oral propranolol therapy.

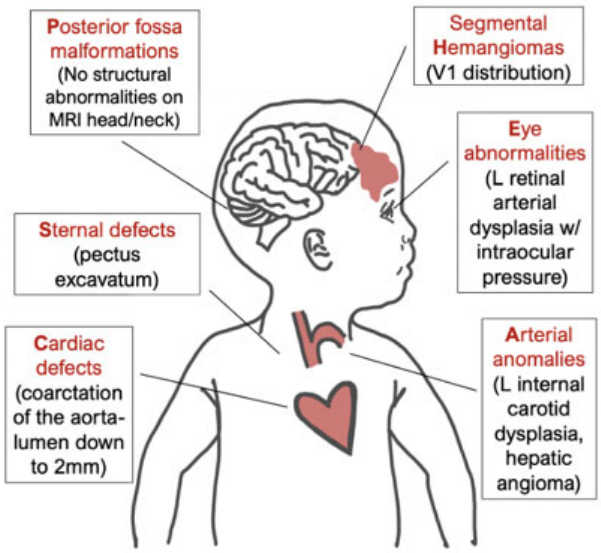

Figure 2: PHACES acronym (in red) with our patient's corresponding findings (in black).

Involvement	Findings
Cranial	Unilateral cerebellar hypoplasia
Vascular	Cerebrovascular accidents, stenosis
Cardiac	Coarctation of the aorta Aortic arch abnormalities
Other	Headaches Sternal pits, clefts, and supraumbilical raphe Eye anomalies (optic nerve hypoplasia, coloboma, cataracts) Auditory deficiency, speech delay Endocrine abnormalities (thyroid dysfunction, hypopituitarism, and adrenal insufficiency)

Figure 3: Extracutaneous involvement in PHACES syndrome.

DISCUSSION

Posterior fossa anomalies, hemangioma, arterial anomalies, cardiac anomalies, eye anomalies, and sternal anomalies (PHACES) syndrome is a neurocutaneous disorder with neurological, arterial, cardiothoracic, and ocular implications [1]. The acronym "PHACE" was first described by Frieden and colleagues and later changed to PHACES to include sternal defects, as identified by Boulinguez and colleagues in 1996 [2]. Its etiology is not fully understood, though it is known to have female predominance with a 9:1 ratio [3].

In 2009, the diagnostic criteria for PHACES were established, encompassing a segmental hemangioma >5 cm on the face, neck, or scalp, plus one major criterion or two minor criteria (Table 1). Our patient met diagnostic criteria for PHACES given her V1 segmental infantile hemangioma >5 cm plus the presence of three major criteria, namely coarctation of the aorta, pectus excavatum, and left retinal arterial dysplasia. Her full involvement is outlined (Figure 2).

While infantile hemangioma is the most common benign tumor of infancy, PHACES syndrome

compromises only 2–3% of infantile hemangioma cases [2]. According to one retrospective cohort study, higher-risk features that can hint at a potential PHACES patient include infantile hemangiomas involving 3 or more locations and a surface area of 25 cm² or greater [4]. The comprehensive list of extracutaneous findings associated with PHACES is extensive and outlined (Figure 3), with the most common being cerebrovascular (91%), brain (52%), and cardiovascular (91%) [2]. Coarctation of the aorta, as seen in our patient, is responsible for 19–30% of cerebrovascular accidents and is associated with an accordingly increased risk [3]. One study attempted to stratify the risk of acute ischemic stroke (AIS) in patients diagnosed with PHACES into 3 different groups—high risk, intermediate risk, and low risk. Low risk for AIS involves arterial anomalies that can be commonly found in the normal population, such as anomalous arterial origins. Intermediate risk includes findings of nonstenotic dysgenesis or narrowing of arteries proximal to the Circle of Willis, while high risk includes greater than 25% narrowing of principal cerebral vessels within or above the Circle of Willis [5]. Our patient will be closely monitored for stroke-like symptoms, and we expect a gradual improvement of her hemangioma with weight-based propranolol dosing.

Table 1: Diagnostic criteria for PHACES syndrome

Rotter et al. (2018) [3]	1. Segmental IH > 5 cm on the face, scalp, or cervical region associated with **1 major criterion or 2 minor criteria** 2. Possible PHACES = IH + 1 minor criterion
Chamli et al. (2023) [2]	**Major criteria:** • Arterial: anomalies of major cerebral or cervical arteries • Brain: posterior fossa anomalies • Cardiovascular: aortic arch anomalies, aneurysm, an aberrant origin of the subclavian artery • Ocular: posterior segment anomaly, retinal vascular anomalies • Sternal defect or cleft **Minor criteria:** • Arterial: cerebral artery aneurysm • Brain: midline anomaly or disruption of cortical development • Cardiovascular: VSD, right aortic arch • Ocular: anterior segment anomaly, cataracts • Midline: hypopituitarism, ectopic thyroid

Abbreviations: MRI: Magnetic resonance imaging (MRI), AIS: Acute ischemic stroke, MRA: Magnetic resonance angiography, PHACES: Posterior fossa anomalies, hemangioma, arterial anomalies, cardiac anomalies, eye anomalies, and sternal anomalies.

Long-term outcomes for PHACES syndrome are poorly described. Due to the variable prognosis, it is important to appropriately screen patients who present with a large hemangioma, especially in a segmental distribution. Initial workup should involve obtaining a comprehensive history and physical examination, with particular attention to ocular abnormalities and sternal midline defects. A screening echocardiogram should also be standard, with subsequent cardiac MRI/magnetic resonance angiography (MRA) if abnormalities are identified [5]. Additional workup includes MRI/MRA of the head and neck. Treatment should be multidisciplinary, as it is highly dependent on the organ system of involvement. For instance, for children with a high-risk arterial anomaly, daily aspirin prophylaxis against acute ischemic stroke can be considered. For infantile hemangiomas, oral propranolol remains the mainstay of therapy. Additional treatment options, such as systemic steroids, surgical excision, or laser therapy, can also be considered. Specialists who may be involved include dermatologists, ophthalmologists, neurologists, cardiologists, endocrinologists, cardiothoracic surgeons, neurosurgeons, and plastic surgeons, to name a few.

CONCLUSION

Posterior fossa anomalies, hemangioma, arterial anomalies, cardiac anomalies, eye anomalies, and sternal anomalies (PHACES) syndrome is a rare neurocutaneous disorder associated with various major organ system complications and portends varying prognoses. Early identification and intervention are crucial in optimizing outcomes and preventing severe cerebrovascular and cardiovascular events. Notably, dermatological manifestations can be the initial indicators of this syndrome, highlighting the critical role of dermatologists in early detection and prevention. Therefore, the presence of a segmental hemangioma should raise suspicion for PHACES syndrome and prompt a thorough evaluation. Proactive consideration can lead to more effective treatments and improve long-term outcomes for this patient population.

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Author Contributions

Kiana Banafshay – Conception of the work, Design of the work, Drafting the work, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Brooke Walterscheid – Conception of the work, Drafting the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Michelle Tarbox – Design of the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

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Conflict of Interest

Authors declare no conflict of interest.

Data Availability

All relevant data are within the paper and its Supporting Information files.