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Case Report

Propranolol-responsive cranial nerve palsies in a patient with PHACES syndrome

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ABSTRACT

PHACES syndrome is a neurocutaneous disorder characterized by the presence of segmental hemangiomas with associated anomalies of the posterior fossa, cerebral vasculature, cardiovascular system, eyes, and ventral or midline structures. We present the first case of propranolol-responsive congenital trigeminal and facial nerve palsies secondary to an intracranial hemangioma in a patient with PHACES syndrome.

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1. Introduction

PHACES syndrome is a neurocutaneous disorder characterized by the presence of posterior fossa anomalies, hemangioma (specifically segmental hemangiomas), arterial lesions, cardiac defects or coarctation of the aorta, eye abnormalities, and sternal cleft or supraumbilical raphe. The term was coined by Frieden et al. in 1996 after he noticed this association of anomalies is a series of patients [1]. We present the first case of propranolol-responsive congenital trigeminal and facial nerve palsies secondary to an intracranial hemangioma in a patient with PHACES syndrome.

2. Case description

A 5-week old infant presented to the OHSU Hemangioma and Vascular Birthmark Clinic with a large segmental hemangioma involving the left temporoparietal scalp with extension onto the face and chest (Fig. 1). The parents had first noticed the lesion around 1 week of life and since that time it had progressively darkened and thickened. Further examination demonstrated a near complete left-sided facial paresis (Fig. 2).

The presence of a segmental hemangioma, especially one that involved the head and neck region, had our team concerned for the diagnosis of PHACES syndrome and therefore the patient was admitted to the hospital for PHACES syndrome work-up. Ophthalmology evaluation was notable for decreased corneal sensation consistent with a trigeminal nerve palsy. Magnetic resonance imaging (MRI) of the brain was obtained which revealed intracranial extension of the patient's hemangioma coursing along the left 5th and 7th cranial nerves (Figs. 3 and 4). Additionally, magnetic resonance angiography (MRA) of the chest was significant for an aberrant retroesophageal subclavian artery and redemonstrated the patient's known ventricular septal defect.

Although our patient had passed her newborn hearing screen, the patient was screened by otoacoustic emissions and due to extension of hemangioma within the internal auditory canal, an auditory brainstem response (ABR) was obtained to evaluate the integrity of the patient's auditory nerve (Fig. 5). Despite poor wave morphology in the left ear, a repeatable wave V was obtained down to 20 dB nHL indicating normal hearing sensitivity. Interestingly, however, a wave latency shift was present in the left ear suggesting a neural transmission delay.

Based on the patient's work-up, a diagnosis of PHACES syndrome was confirmed and the patient was started on propranolol therapy at 1 mg/kg/day divided TID in addition to appropriate eye care for corneal protection. After tolerating the initial two doses of propranolol, dosing was escalated to 2 mg/kg/day divided TID.

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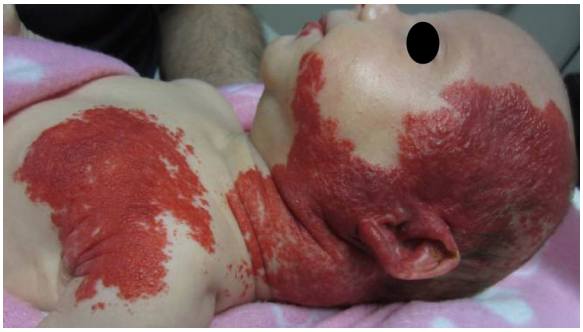

Fig. 1. Patient at presentation.

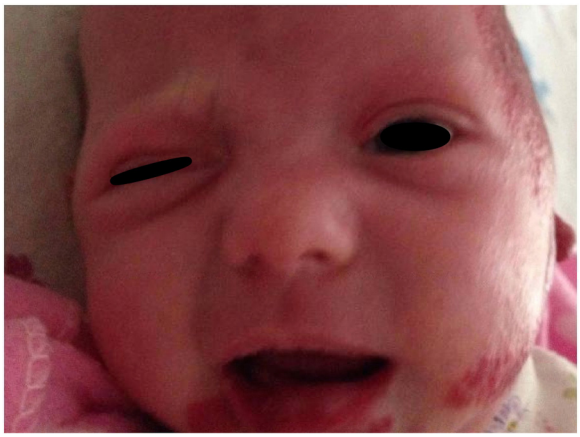

Fig. 2. Evidence of left facial nerve palsy with incomplete eye closure and flattened melolabial crease.

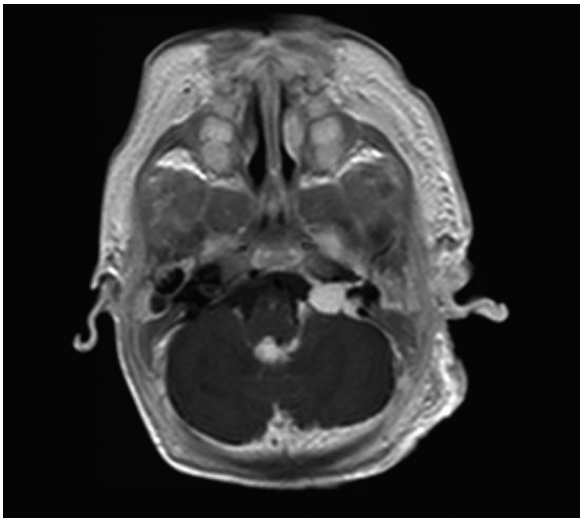

Fig. 3. Axial T1 weighted post-gadolinium MRI of the brain demonstrating intracranial hemangioma within the cerebellopontine angle (CPA) coursing along the facial nerve within the internal auditory canal (IAC) toward the geniculate ganglion.

Weight based dosing was subsequently adjusted by the patient's pediatrician on a monthly basis.

By 3 months of age, there was significant improvement in the appearance of the patient's hemangioma with resolution of the patient's facial nerve palsy (Fig. 6). At 9-month follow-up, the patient's hemangioma had faded significantly (Fig. 7) and interim evaluation by ophthalmology identified near normal corneal sensation. Although the patient is being followed closely by audiology, repeat ABR testing has not been performed since initiation of propranolol therapy due to adequate results on behavioral testing.

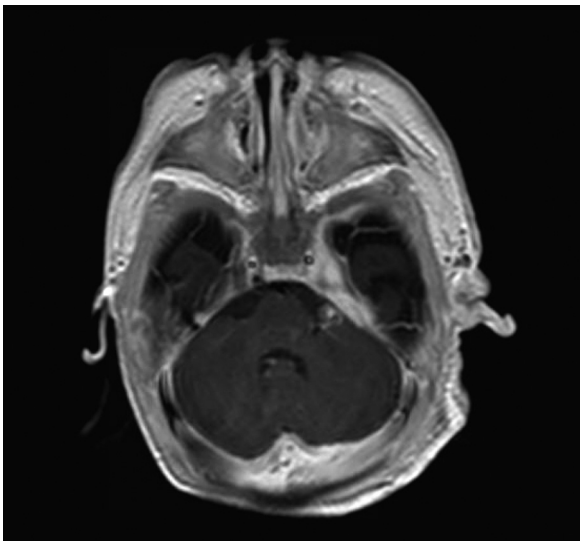

Fig. 4. Axial T1 weighted post-gadolinium MRI of the brain demonstrating asymmetry within Meckel's cave secondary to hemangioma involvement.

3. Discussion

In 2009, diagnostic criteria for PHACES syndrome were established in a consensus statement supported by a multidisciplinary group of specialists with the diagnosis centered around the presence of a segmental infantile hemangioma with an additional major extracutaneous anomaly or two minor extracutaneous anomalies [2]. No standardized work-up exists for the evaluation of PHACES syndrome however imaging of the brain, head, neck, and chest as well as ophthalmology and dermatology consultations have been recommended. At our institution, our work-up of PHACES syndrome involves magnetic resonance imaging (MRI) of the brain, magnetic resonance angiography (MRA) of the head and neck, echocardiogram or MRA of the chest, and an ophthalmology consult. We also obtain a TSH level as endocrine dysfunction, although uncommon, has been described and carries significant implications on growth and development [3].

By definition, patient's with PHACES syndrome have large facial hemangioma involvement given their segmental nature and thus systemic treatment of the hemangioma is typically indicated. Although corticosteroids were traditionally used to treat infantile hemangioma, propranolol therapy has quickly become the standard of care due to its greater efficacy and an improved side-effect profile [4–9]. An important consideration prior to the initiation of propranolol therapy in patients with PHACES syndrome is the risk for acute ischemic stroke (AIS) associated with concomitant cerebrovascular and aortic arch abnormalities [10,11]. Due to this risk, it is recommended that patients receive appropriate imaging prior to initiating propranolol therapy. If a cerebrovascular or aortic arch abnormality is subsequently identified, consultation with a neurologist for risk-stratification and co-management has been suggested. If the benefits of propranolol therapy are felt to outweigh the risks, propranolol therapy can still be initiated. In this scenario, however, it is advised that dosing begin at the lowest possible dose and be divided TID with slow up-titration to avoid abrupt blood pressure changes [12]. For patient's felt to be high risk, close monitoring in an inpatient setting should be considered.