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# OCULAR MANIFESTATIONS IN PHACE SYNDROME AND REVIEW OF LITERATURE

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## ABSTRACT

**OBJECTIVES**: To introduce PHACE syndrome (Posterior fossa malformations, Hemangiomas, Arterial anomalies, Coarctation of the aorta and other cardiac defects, and Eye abnormalities) to the ophthalmologic literature; to report the case of PHACE syndrome associated with congenital cataract with lid hypoplasia on lateral half of lid with trichiasis; and to review the ocular and systemic findings that may occur in this group of disorders. **DESIGN:** Case report and literature review. **METHODS:** To examine a child with PHACE syndrome and congenital cataract with lid coloboma and foreign body sensation due to trichiasis and review the ophthalmologic and systemic manifestations of this syndrome. **RESULTS:** We report a case of nine years old female with foreign body sensation in left eye with auricular and nasal hypoplasia and multiple ocular menifestations like-lid coloboma,trichiasis,cloudy cornea with signs of healed iridocyclitis with congenital cataract with partially absorbed lens. Skin showed atrophic scarring with telangia ectasia over face and upper chest. We did epilation of the misdirected lashes with application of sectoral cryo. **CONCLUSIONS:** Ophthalmologists who examine children with large facial haemangioma should consider PHACE syndrome in the differential diagnosis and should obtain appropriate CNS imaging studies and cardiac evaluation when the diagnosis is suspected. Congenital lid hypoplasia with trichiasis and uveitis should be added to the list of PHACE-associated ocular anomalies.

**KEYWORDS:** PHACE syndrome, ocular manifestation.

## INTRODUCTION

PHACE syndrome is the association of a large haemangioma usually on the face or neck, in combination with one or more other abnormalities. Every infant diagnosed with PHACE syndrome has different medical needs. Some have mild symptoms while other have more severe symptoms. PHACE syndrome is uncommon but may have been misdiagnosed in the past. It affects girls nine times more often than boys. The goal to report this case was to better understand the risk of this syndrome and to develop guidelines for its evaluation and management and also to add ocular findings which were under reported previously.

## CASE REPORT

A nine years old female child came to us for ocular foreign body sensation and diminission of vision in the left eye (PL +PR Accurate). Her right eye was absolutely normal. Child had large facial atrophic scarring with telangia ectasia over face on her left half and upper chest with auricular hypoplasia with nasal cleft.MRI brain showed watershed territory chronic infarct in right pariatooccipetal region. Right hemiatrophy of right cerebral hemisphere. MRI also showed left otomastoiditis. Ocular findings on slit lamp examination in left eye were- lid hypoplasia in lateral half, trichiasis, cloudy cornea with superficial vascularisation, with signs of healed uveitis, partially absorbed cataractous lens.Fundus findings in the right eye were normal and in left eye only faint glow was visible. We did epilation of the misdirected lashes with application of sectoral cryo in the left eye. Child responded well to medical line of her treatment for fascial lesion. As cornea was extensively damaged we planned a keratoplaty along with cataract extraction.

**Fig 1-Facial Telangiaectasia**

**Fig 2 Auricular hypoplasia**

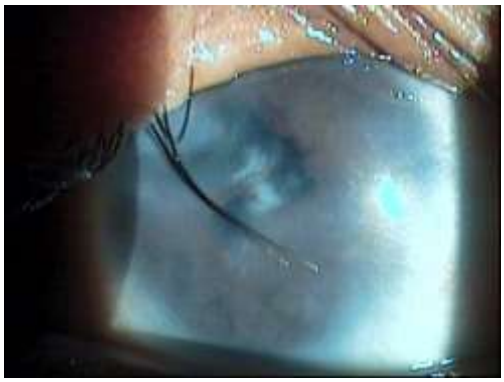

**Fig 3: Trichiasis with anterior segment anamolies**

## DISCUSSION

The term PHACE is an acronym that refers to a group of abnormal medical findings. When these occur in combination, the diagnosis of PHACE syndrome is made. [1, 2]

**PHACE defined as**

***P***osterior fossa- These are brain malformations that are usually present at birth. These brain malformations do not form after the infant is born.

***H***emangioma- The hemangioma usually covers a large area on the skin of the head or neck (greater than 5 cm). The term "segmental" is sometimes used to describe these hemangiomas.

***A***rterial lesions- The abnormalities of the blood vessels in the neck or head.

***C***ardiac abnormalities/aortic coarctation- These are abnormalities of the heart or the blood vessels that are attached to the heart.

***E***ye abnormalities like –

**Posterior segment abnormalities**.

- Retinal vascular abnormality.
- Persistent fetal retinal vessels.
- "Morning-glory" disc.
- Peripapillary staphyloma.
- Optic nerve hypoplasia.

**Anterior segment abnormalities.**

- Microphthalmos.
- Coloboma.
- Congenital cataracts.
- Sclerocornea.
- Iris hypoplasia.
- Exophthalmus.
- Congenital third nerve palsy.
- Horner syndrome.
- Iris vessel hypertrophy.

Till now very few cases were reported in context to ophthalmological findings.

Metry et al [3] reviewed 14 cases of hemangioma and PHACE syndrome and an additional 116 cases in the literature and found the $V_1$ segment of the fifth cranial nerve was the most commonly affected dermatome. Optical findings included increased retinal vascularity, bilateral retinal hyperemia, and Horner syndrome.[3]

Weon et al reported a case of PHACE syndrome without any ocular findings.[4]

There are both major and minor characteristics of PHACE syndrome defined by the published criteria. The criterion is classified by organ system affected. The organ systems include cerebrovascular, structural brain, cardiovascular, ocular, and ventral or midline anomalies.[5-8]

In our case there were findings of both major and minor criterion of PHACE syndrome. To our knowledge there were very few cases in which lid hypoplasia, trichiasis and uveitis were reported. And these findings we can add to the list of PHACE syndrome in minor criterion as all these conditions involved the anterior segment of eye.

## CONCLUSIONS

Ophthalmologists who examine children with large facial haemangioma should consider PHACE syndrome in the differential diagnosis and should obtain appropriate CNS imaging studies and cardiac evaluation when the diagnosis is suspected. Congenital lid hypoplasia with trichiasis and uveitis should be added to the list of PHACE-associated ocular anomalies.

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