Ophthalmic involvement in PHACES syndrome: Prevalence, spectrum of anomalies and outcomes.

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Background

- PHACES Syndrome includes Posterior fossa malformations, infantile Hemangiomas (IFH; 95% facial) with Arterial, Cardiac, Eye (6-7%) and Sternal anomalies.[1-2]
- Major and minor diagnostic criteria were developed in 2009[1] and revised in 2016.[2] (Box 1)
- Ocular anomalies were reported to be 6% and 7% for posterior (major criterion) and anterior (minor criterion) segment anomalies, respectively.
- Ophthalmic involvement other than congenital anomalies was not addressed in the original or revised consensus statements with limited published data. Furthermore, there is no data on long-term follow up.[3]

Objectives

- To present a large cohort of patients with PHACES highlighting prevalence, spectrum of anomalies and outcomes of ophthalmic involvement.

Design/Methods

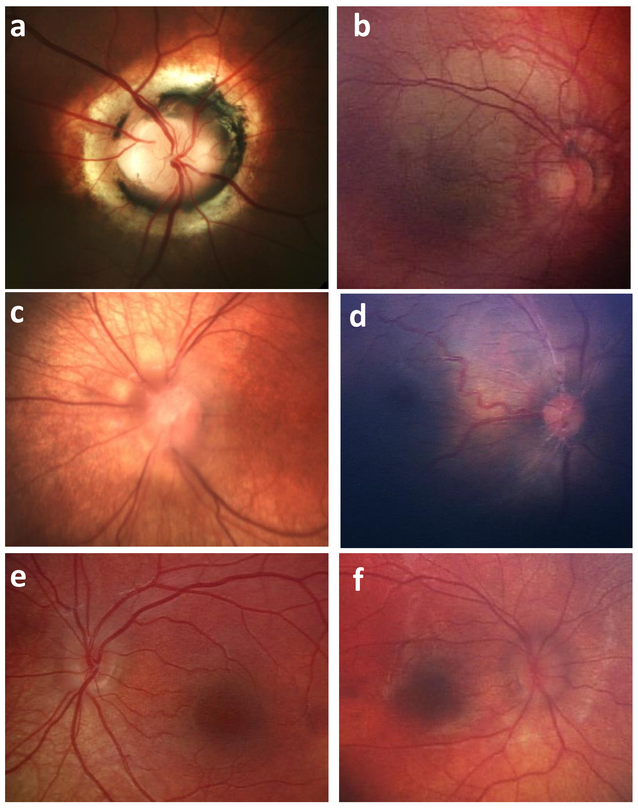
- Retrospective non-comparative single institution observational case series of children with definite PHACES syndrome diagnosed and managed between 2000-2019 at a single tertiary referral center.
- Dermatology database was utilized to reduce selection bias.
- The study included 43 patients with Definite PHACES Syndrome.
- **Ophthalmic involvement:**
  1. Congenital ocular anomalies either diagnostic (major/minor criterion) or non-diagnostic including intraocular hemangioma.
  2. Ophthalmic complications due to IFH.
- **Severe Ophthalmic involvement:** presence of an amblyogenic ptosis, proptosis, or misalignment capable of affecting visual potential and/or requiring surgery.
- **Primary outcomes:** Frequency and spectrum of ocular involvement.
- **Secondary outcomes:** Final visual and long-term ocular outcome.
- **Poor visual outcome:** final visual acuity worse than 20/200.

Results

- **Prevalence and spectrum of ophthalmic involvement:**

1. Congenital anomalies: (Figure 1 & 2)
   - Five children (11.6%) had diagnostic anomalies
   - Four children (9.3%, one without diagnostic criteria) had non-diagnostic anomalies.

Fig. 1: Posterior segment anomalies

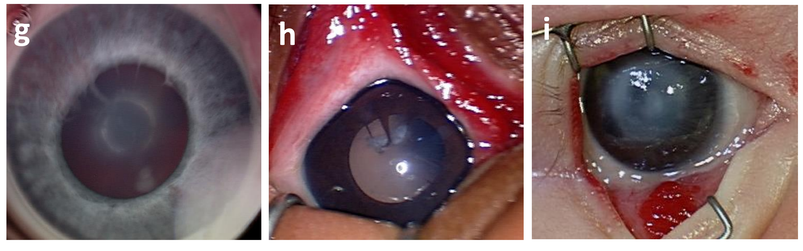


Posterior segment anomalies: a) & b) Peripapillary staphyloma. c) & d) Dysmorphic optic nerve head with gliotic changes that did not fit any major criteria, with retinal vascular anomalies in d. e) & f) Optic nerve hypoplasia with right pseudoedema of the optic nerve head.
Anterior Segment anomalies: g) persistent pupillary membrane and iris hemangioma, h) Persistent pupillary membrane and cataract and i) Peter's anomaly and cataract.

Box 1: Diagnostic ocular criteria

Major criteria
Posterior segment anomaly
Persistent fetal vasculature
Retinal vascular anomalies
Morning glory disc anomaly
Optic nerve hypoplasia
Peripapillary staphyloma
Minor criteria
Anterior segment anomaly
Sclerocornea
Cataract
Coloboma
Microphthalmia

Fig. 2: Anterior segment anomalies



2. Periocular facial hemangioma:

- Twenty-nine children (66%) had periocular IFH involving the eyelids (29), orbit (14), and/or conjunctiva (13). Bilateral in three children
- Severe ophthalmic involvement in 21/29 patients and included severe ptosis (9/21), proptosis (9/21), and strabismus (6/21).
- Long-term sequalae in 14/21 (67%) and included ptosis (11 mild, 2 severe), cicatricial entropion (2) and strabismus (8).
- Seven children required corrective surgery (6 prior to 2011 and initially treated with systemic steroids). Furthermore, 4 of which had > 1 surgery (all initially treated with steroids).
- Orbital/conjunctival periocular IFH involvement was significantly associated with long-term ophthalmic effects and need for surgery (67% and 40%, p=0.01 and 0.03 respectively)
- Sixteen children (67%) developed amblyopia during IFH control. Of which, 13 responded well to amblyopia therapy.

Visual outcome: (At the final follow-up of mean 8.7 years)

- 26/29 eyes with ocular involvement achieved visual acuity better than 20/80. Of which, 21 had vision better than 20/40 and 5 had vision ranging from < 20/40 to 20/80.
- Three children had poor vision, all had posterior segment anomaly.

Conclusions

- Our institutional experience with PHACES syndrome shows 14% had congenital ocular anomalies and, 66% had ocular sequelae of IFH that were severe in 2/3 and required surgery in 1/3 of cases.
- Vision, follow-up frequency and anticipated outcomes are all important concerns for families that are usually raised at presentation. Having frequency figures would help guide their counseling.
- A child without major ocular anomalies has normal visual potential but timely follow-up will help to ensure early detection and treatment of helps early reversal of amblyogenic stimuli.

References

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