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 20091 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

- 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.

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.

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 sequelae 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 pericellular 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.

Results

Box 1: Diagnostic ocular criteria

<table>
<thead>
<tr>
<th>Major criteria</th>
<th>Minor criteria</th>
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<tbody>
<tr>
<td>Posterior segment anomaly</td>
<td>Sclerocornea</td>
</tr>
<tr>
<td>Persistent fetal vascularature</td>
<td>Cataract</td>
</tr>
<tr>
<td>Retinal vascular anomalies</td>
<td>Coloboma</td>
</tr>
<tr>
<td>Optic nerve hypoplasia</td>
<td>Microphthalmia</td>
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<tr>
<td>Peripapillary staphyloma</td>
<td></td>
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</tbody>
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Fig. 1: Posterior segment anomalies

Fig. 2: Anterior segment anomalies

References