Phenotypic Expression of the Optic Disc in Primary Open Angle Glaucoma

Grassi, Lourdes¹; Salazar, Diana²; De Gainza, Agustina³; Bouris, Ella¹; Morales, Esteban¹; Caprioli, Joseph¹

1. UCLA Stein Eye Institute, Los Angeles, CA, United States. 2. W K Kellogg Eye Center, Ann Arbor, MI, United States. 3. Hospital Italiano, Buenos Aires, Argentina

Purpose

We performed a retrospective study to identify demographic and ocular characteristics associated with six identifiable optic disc phenotypes in Primary Open Angle Glaucoma (POAG) patients.

Methods

Inclusion Criteria

Diagnosis of POAG, one disc photo within two years of a visual field (VF), ≥ 2, VFs with Mean Deviation (MD) > -10dB, Pattern Standard Deviation (PSD) probability < 0.05, and a Cirrus Optical Coherence Tomography (OCT) with optic disc area between 1.14 and 3mm².

Phenotypic Classification

The photos were classified independently by three masked glaucoma specialists into disc phenotypic subgroups, according to standard reference photographs:

Agreement:
The three graders agreed on 89% of the total images (1083). All remaining discrepancies (11%) were resolved by a consensus between the graders to decide the final phenotype.

Results

Table 1. Summary Statistics showing statistically significant variables based on ANOVA (continuous) and Chi-Squared test (categorical)

<table>
<thead>
<tr>
<th>Phenotype</th>
<th>Gender (%)</th>
<th>Race (%)</th>
<th>Age</th>
<th>IOP</th>
<th>LogMAR</th>
<th>CCT</th>
<th>Refraction</th>
<th>MD</th>
<th>PSD</th>
<th>Disc Area</th>
<th>RNFL</th>
</tr>
</thead>
<tbody>
<tr>
<td>Concentric Th.</td>
<td>Male 54 (13.1) Female 46 (16.9)</td>
<td>Caucasian 122 (48.4) Hispanic 122 (48.4) Other 122 (48.4)</td>
<td>70.2 (15.6)</td>
<td>85.1 (6.2)</td>
<td>0.42 (0.3)</td>
<td>11.5 (1.0)</td>
<td>-3.6 (2.3)</td>
<td>1.8 (0.4)</td>
<td>0.000</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Focal Thinning</td>
<td>Male 18 (12.9) Female 30 (20.1)</td>
<td>Caucasian 30 (14.3) Hispanic 30 (14.3) Other 30 (14.3)</td>
<td>70.2 (15.6)</td>
<td>85.1 (6.2)</td>
<td>0.42 (0.3)</td>
<td>11.5 (1.0)</td>
<td>-3.6 (2.3)</td>
<td>1.8 (0.4)</td>
<td>0.000</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Tilted</td>
<td>Male 10 (12.9) Female 9 (12.2)</td>
<td>Caucasian 10 (12.9) Hispanic 10 (12.9) Other 10 (12.9)</td>
<td>70.2 (15.6)</td>
<td>85.1 (6.2)</td>
<td>0.42 (0.3)</td>
<td>11.5 (1.0)</td>
<td>-3.6 (2.3)</td>
<td>1.8 (0.4)</td>
<td>0.000</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Extensive PPA</td>
<td>Male 47 (13.4) Female 4 (3.0)</td>
<td>Caucasian 47 (13.4) Hispanic 47 (13.4) Other 47 (13.4)</td>
<td>70.2 (15.6)</td>
<td>85.1 (6.2)</td>
<td>0.42 (0.3)</td>
<td>11.5 (1.0)</td>
<td>-3.6 (2.3)</td>
<td>1.8 (0.4)</td>
<td>0.000</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Broad Thinning</td>
<td>Male 109 (10.4) Female 109 (10.4)</td>
<td>Caucasian 109 (10.4) Hispanic 109 (10.4) Other 109 (10.4)</td>
<td>70.2 (15.6)</td>
<td>85.1 (6.2)</td>
<td>0.42 (0.3)</td>
<td>11.5 (1.0)</td>
<td>-3.6 (2.3)</td>
<td>1.8 (0.4)</td>
<td>0.000</td>
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</tr>
</tbody>
</table>

Summary results

- Tilted phenotype was more frequently associated with Asian race, myopia, lower age and lower Disc Area.
- Broad Thinning phenotype has a thinner Retinal Nerve Fiber Layer.
- Focal Thinning has better Visual Acuity compared to Broad Thinning.
- Focal Thinning and Broad Thinning are more associated with female gender.

Conclusion

This study reports six phenotypic classifications of POAG patients, with emergence of some ocular and systemic differences between phenotypes. Future refinement of phenotypes should allow improved individualization of patient care and enhance the ability to interpret a multitude of genetic associations with POAG.