**Morning Glory Disc Anomaly & Optic Disc Coloboma: A Juxtaposition (& Clue to Aetiology?)**

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**Case Report**

A 6 year old female, of Anglo-Saxon and Eastern European origins, was referred to co-author LK from her optometrist regarding decreased right vision and abnormal fundal appearances.

**Explanation**

- **Facial appearances normal, no hypertelorism or lid/palate notching/clefting.**
- **Best corrected visual acuities 6/24 right, 6/9 left**
- **Cycloplegic refraction +2.25 D right, +1.75 D left**
- **Right relative afferent pupillary defect**
- **Ocular motility full, alignment orthophoric near and distance.**
- **No fundus lesions, visual field tests normal.**
- **Corneal diameters 11 mm both eyes**
- **Normal anterior segments (in particular no iridic or ciliary body coloboma).**
- **Perinatal history: small baby with initial failure to thrive due to a ‘poor swallow reflex’.**
- **Past medical history: notable for small capillary haemangioma on forehead (lasered as baby)**
- **Family ophthalmic history: nil**
- **No sex or racial predilection**
- **Rarely bilateral**
- **No known genetic associations**

**Examination**

- **Right disc has a defect in inferior neuroretinal rim, with a dark ring around most of the disc. Presence of glial tissue in centre. No contractility noted.**
- **Inferior to disc are two patches of chorioretinal atrophy representing coloboma.**
- **Left disc has a dark ring surrounding an excavation which includes a small sliver of peripapillary retinal pigment epithelial atrophy. Cup is central. No contractility noted.**

**Table 1. Comparison between MGDA and ODC**

<table>
<thead>
<tr>
<th>MGDA</th>
<th>ODC</th>
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<tbody>
<tr>
<td>Optic disc in excavation</td>
<td>Excavation in optic disc</td>
</tr>
<tr>
<td>Symmetrical excavation</td>
<td>Asymmetrical excavation (inferiorly)</td>
</tr>
<tr>
<td>Central glial tuft</td>
<td>No glial tuft</td>
</tr>
<tr>
<td>Severe peripapillary pigmentary change</td>
<td>Minimal peripapillary pigmentary change</td>
</tr>
<tr>
<td>Anomalous retinal vessels</td>
<td>Normal retinal vessels</td>
</tr>
<tr>
<td>Moderate deep vascular atrophy, loss of foveal reflex</td>
<td>No severe or foveal predilection</td>
</tr>
<tr>
<td>Family history</td>
<td>Family history</td>
</tr>
<tr>
<td>Not associated with coloboma</td>
<td>May be associated with coloboma</td>
</tr>
<tr>
<td>Distinct systemic associations</td>
<td>Distinct systemic associations</td>
</tr>
</tbody>
</table>

**Conclusion**

The presence of a MGDA and optic disc/retinochoroidal coloboma in fellow eyes in this patient represents an interesting observation and possible insight into the aetiology of these two anomalies. Should these conditions (bilateral) be due to completely different defects in embryological development, then their juxtaposition in the same patient would be extremely rare. Perhaps more likely is our hypothesis that these two anomalies may share a common pathway at some point during development of the posterior sclera and closure of the embryonic fissure.

Bakri and others (including Traboulsi) described the case of a 10 year old girl who developed a stroke due to moyamoya disease, having had a right esotropia and pendular nystagmus from age 6 months\(^3\). Neuroimaging also revealed a sphenopharyngeal meningoencephalocoele. She had a right MGDA, with 3/20 visual acuity, and left optic nerve hypoplasia with an inferior retinochoroidal coloboma and 20/40 visual acuity. This case shares some similarities with ours, however our patient has no midline abnormalities nor moyamoya vessels on MRA.

Our case presents another dimension to the range of presentations of excavatory optic disc anomalies, and in doing so poses some interesting questions regarding the pathogenesis of MGDA. Further detailed assessment and descriptions of patients with MGDA, and use of clear and accurate terminology to avoid diagnostic confusion, may help us further elucidate the developmental pathway responsible for this rare and curious anomaly. This case also serves as a reminder that neuroimaging (MRI and MRA) of children with MGDA is indicated to exclude possibly life-threatening systemic associations.

**Acknowledgement**

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**References**