



MORNING GLORY DISC ANOMALY & OPTIC DISC COLOBOMA: A JUXTAPOSITION (& CLUE TO AETIOLOGY?)

Logan Mitchell¹, MBChB (Otago), Lionel Kowal^{1,2}, FRANZCO

1.Royal Victorian Eye and Ear Hospital, East Melbourne, Victoria, Australia 2.Private Eye Clinic, East Melbourne, Victoria, Australia

AIMS

- i) Describe a case of a 6 year old girl presenting with a left morning glory disc anomaly and right optic disc and retinochoroidal coloboma, and
- ii) To discuss possible implications for aetiology of these conditions with reference to the literature.

INTRODUCTION

Morning glory disc anomaly (MGDA) is a rare developmental anomaly of the optic nerve head. First called "morning glory" disc anomaly by Kindler in 1970 (due to its likeness to the morning glory flower – see Figure 1), this anomaly has often been erroneously labelled, grouped and confused with optic disc colobomas (ODC) and peripapillary staphylomas in the literature, with unfortunate consequences in regards to clinical descriptions, associations and hypotheses regarding pathogenesis¹.

Children with MGDA often present with strabismus (90%) or leukocoria². The possible presence of ocular and systemic associations must be appreciated, which (critically) are quite distinct from the associations of ODC (such as CHARGE syndrome). Moyamoya disease and trans-sphenoidal basal encephaloceles are two key systemic associations. Moyamoya ("puff of smoke" in Japanese) disease is the result of a primary intracranial vascular agenesis (usually of the terminal internal carotid circulation) which results in collateral formation (hence the "puff of smoke" seen on angiography) and eventual ischaemia. This can cause headaches, seizures and strokes in children, and subarachnoid haemorrhage in adult patients.

The embryological maldevelopment resulting in MGDA is still debated¹⁻³. ODC and retinochoroidal colobomas result from incomplete closure of the embryonic fissure of the optic vesicle. However, MGDA is thought to result from a primary mesenchymal or mesodermal abnormality (eg. abnormal development of posterior sclera and/or lamina cribrosa), due to the incorporation of the whole optic disc into the excavation, and the presence of a glial tuft. There is some histopathological evidence to support this⁴. The associated vascular anomalies (both retinal and intra-cranial) suggest a role for vascular patterning defects, whilst midline signalling is implicated by virtue of the craniofacial associations. It is hypothesised that cases where MGDA is associated with a colobomatous appearance are due to effects of posterior scleral development problems on embryonic fissure closure.

| Table 1. Features of Morning Glory Disc Anomaly | |
|---|--|
| Funnel-shaped excavation of posterior fundus that incorporates disc | |
| Disc markedly enlarged, often orange or pink | |
| Disc may seem recessed or elevated within excavation | |
| Wide annulus of chorioretinal pigmentary disturbance | |
| White tuft of glial tissue overlies central disc | |
| Blood vessels often arise from periphery of disc and run abnormally straight | |
| Usually unilateral, can be bilateral | |
| Visual acuity usually 20/200 or worse, can be 20/20 | |
| More common in females | |
| 26-38% will develop peripapillary retinal detachment in first decade | |
| Sub-retinal neovascular membranes can develop | |
| Disc margin may have unusual regular contractile movements (probably due to transmission of CFS pressure changes) | |

| Table 2. Comparison between MGDA and ODC | |
|---|--|
| Morning Glory Disc Anomaly | Optic Disc Coloboma |
| Optic disc in excavation | Excavation in optic disc |
| Symmetrical excavation | Asymmetrical excavation (inferiorly) |
| Central glial tuft | No glial tuft |
| Severe peripapillary pigmentary change | Minimal peripapillary pigmentary change |
| Anomalous retinal vasculature | Normal retinal vasculature |
| More common in females, less common in blacks | No sex or racial predilection |
| Rarely familial (no known genetic associations) | Often familial (with known genetic associations) |
| Rarely bilateral | Often bilateral |
| Not associated with colobomata | Iris, ciliary body and retinochoroidal colobomata common |
| Distinct systemic associations | Distinct systemic associations |

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.

- Peri-natal 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

Family medical history:

- mother a carrier for cystic fibrosis
- maternal grandfather had 'kidney tumours'
- paternal great-uncle had haemochromatosis
- sister had capillary haemangioma as a baby also

EXAMINATION

- 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.
- Corneal diameters 11 mm both eyes
- Normal anterior segments (in particular no iris or ciliary body coloboma).

WORK-UP

Magnetic resonance imaging and angiography of our case was arranged, and demonstrated no intra-cranial abnormalities. In particular there was no basal encephalocele, nor any moyamoya vessels.

Ocular examination of our case's sister (aged 9), father and mother was performed. All of these examinations were normal, with no iris, ciliary body or optic disc colobomas seen. The mother did have a small peripapillary scleral crescent located inferotemporally on the right, but this fell within normal limits and was not thought to represent a retinochoroidal coloboma.



FUNDAL APPEARANCES

- 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.

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 (both rare) 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 months⁵. Neuroimaging also revealed a sphenopharyngeal meningoencephalocele. She had a right MGDA, with 3/200 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

The authors would like to acknowledge the assistance of Dr Alex Levin in the clinical interpretation of this case, and his invaluable advice.

REFERENCES

1. Taylor D, Hoyt CS. (Ed.). (2005). Pediatric Ophthalmology and Strabismus. Elsevier Saunders.
2. Lee BJ, Traboulsi EI. Update on the morning glory disc anomaly. Ophthalmic Genetics. 2008;29:47-52
3. Traboulsi EI. Morning glory disc anomaly – more than meets the eye. J AAPOS 2009;13:333-4
4. Manschot WA. Morning glory syndrome: a histopathological study. Br J Ophthalmol 1990;74:56-58
5. Bakri SJ et al. Ocular malformations, moyamoya disease, and midline cranial defects: a distinct syndrome. Am J Ophthalmol 1999;122:356-7