Coloboma of Optic Disc
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History
5 year old male with blurred vision.

Diagnosis
Coloboma of Optic Disc

Additional Clinical
Normal MR of the brain.

Discussion
Coloboma is a rare congenital abnormality related to incomplete closure of the intraocular fissure (choroid fissure or optic cleft) and as such may involve the optic nerve, retina, choroid, iris and/or lens. Typical colobomas are inherited as an autosomal dominant trait with variable penetrance and expression. Colobomas are associated with various syndromes (trisomy 13, Goldenhar, Rubenstein-Taybi, Lenz, Waardenburg, and CHARGE).

Morning glory disc anomaly is a type of coloboma that incorporates the optic disc but also often involves the adjacent retina and choroid. The genetics of morning glory disc are unknown. Morning glory disc is usually unilateral compared to typical colobomas which are bilateral in about 60% of patients. Morning glory disc is associated with coloboma-renal syndrome, trisomy 4q, optic nerve hypoplasia and poor vision, midline intracranial malformations (agenesis of the corpus callosum, pituitary insufficiency, and encephalocele) and vascular malformations (Moya-moya, carotid stenoses, carotid aplasia).

Findings
MR-Funnel-shaped excavation of posterior fundus at the level of the optic disc of the right eye. Additionally, the right oculus is flattened posteriorly and slightly smaller than the left.

Reference
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