A Rare Optic Disc Anomaly with Unique Fundoscopic and MRI Findings

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An eleven-month-old boy presented to the ophthalmology clinic with a history of strabismus and severe visual impairment of the right eye. There was no history of seizures, vomiting, loss of consciousness, or neurological deficits. He was developmentally appropriate for age. Family history was not contributory. Specifically, there was no history of neurofibromatosis type 1 (NF1) or similar condition in the family. On examination, there were no dysmorphic features or skin changes. His neurological examination was unremarkable. Ophthalmic examination revealed a large angle esotroopia in the right eye. The child did not take up fixation with this eye. Anterior segment examination showed a central dot-like opacity in the posterior surface of the lens. Dilated fundus examination revealed a large dysplastic optic nerve with peripapillary pigmentation and limited elevation of the retina. This along with the radial distribution of vessels, and a tuft of glial tissue on the optic disc **(Figure 1).** A stalk of glial tissue extended from the central part of the optic disc to the posterior surface of the lens and was identified as Cloquet's canal, a remnant of fetal vasculature. Examination of the left eye revealed persistent pupillary membrane in the anterior segment. Posterior segment examination was unremarkable. Magnetic resonance imaging (MRI) of the brain and orbit were done and revealed funnel-shaped excavation of the right optic disc associated with elevated adjacent retinal surface and discontinuity of the uveoscleral surface. A thin and faint band of tissue was seen extending from the posterior surface of the lens to the head of the optic nerve along the Cloquet canal which correlated with the fundoscopic findings. Absence of fluid signal intensity within the right optic nerve sheath around the right optic nerve was observed **(Figure 2).** Focal thickening and faint T2 signal intensity of the ipsilateral optic chiasm were noted without abnormal enhancement **(Figure 3).** There was no evidence of other midline abnormality. Magnetic resonance angiography was unremarkable and there was no evidence of Moyamoya phenomenon.

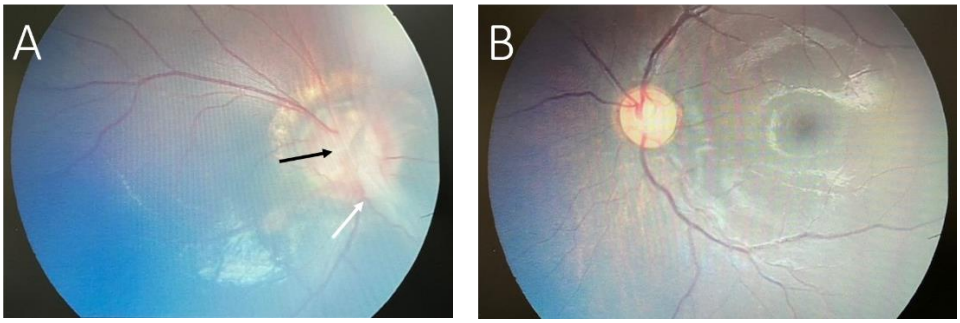

Figure 1. Color fundus photographs of the **(a)** right eye showing a large dysplastic optic nerve with peripapillary pigmentary disturbance. The retinal vessels are radially distributed on the optic disc. Note the tuft of glial tissue on the optic disc (black arrow) and a fibroglial stalk (white arrow) extending anteriorly from the center of the optic disc. This is a remnant of the Cloquet's canal. The macula is hypoplastic. **(b)** The fundus photograph of the left eye shows a normal optic disc, macula, vessels, and retinal background.

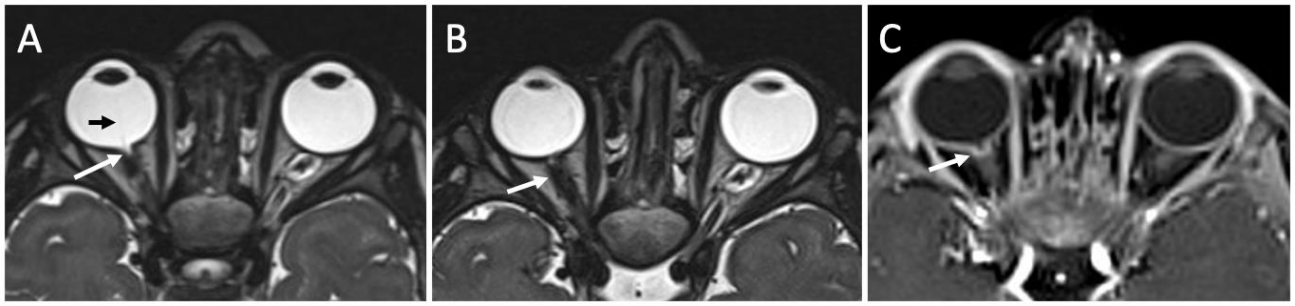

Figure 2. (a) Axial T2-sampling perfection with application optimized contrasts using different flip angle evolutions (SPACE) image shows funnel-shaped excavation of the right optic disc associated with elevated adjacent retinal surface and discontinuity of the uveoscleral surface (white arrow). A thin and faint band of tissue extending from the posterior surface of the lens to the head of the optic nerve along the Cloquet canal (black arrow). **(b)** Axial T2-SPACE image demonstrates effacement of the fluid signal in the right optic nerve sheath around the right optic nerve (arrow). **(c**) Axial contrast-enhanced T1-weighted image with fat saturations shows marginal enhancement of the distal right optic nerve (arrow).

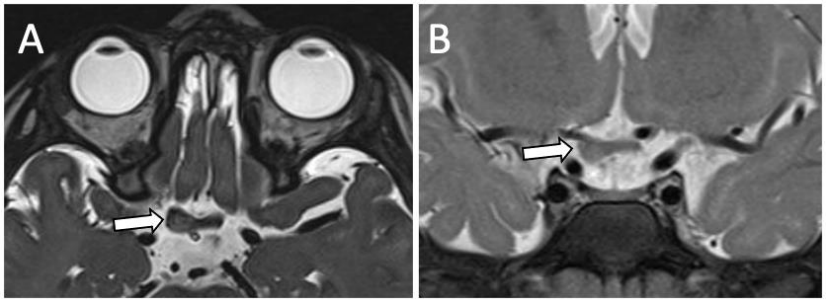

Figure 3. (a) Axial T2-SPACE image and **(b)** coronal T2-weighted image with fat saturation show focal thickening and faint T2 hyperintensity of the ipsilateral optic chiasm (arrows).

Question

What is the diagnosis?

a. Coloboma
b. Peripapillary staphyloma
c. Morning glory disc anomaly
d. Optic disc pit
e.

Answer

c. Morning glory disc anomaly

Discussion

Morning glory disc anomaly (MGDA) is a rare congenital anomaly of the optic disc with a typical fundoscopic and radiological appearance.[1,2] It is manifested as funnel-shaped excavation of the optic disc with chorioretinal pigmentary changes and radially oriented vessels resembling a morning glory flower.[3] Patients may present initially with reduced visual acuity, leukocoria, and strabismus.[3,4] Other ocular abnormalities that are described with MGDA include hypertelorism, persistent fetal vasculature, retinal detachment, congenital cataract, and drusen.[3,5] Visual pathway abnormality is an important association of MGDA and includes ipsilateral thickening and contralateral hypoplasia of the optic chiasm secondary to axonal degenerations.[4,5,6,7]

Although previous literature about MGDA labeled the observed abnormal thickening of the optic pathway as optic pathway glioma (OPG), recent studies considered this finding as a developmental malformation associated

with MGDA that should not be confused with OPG.[4,8,9] For instance, in a large retrospective study that included 32 patients with MGDA, 21 of them had mild segmental thickening of the ipsilateral optic nerve.[8] Likewise, Nguyen DT et al. included a total of nine confirmed cases of MGDA. In eight of them, the thickness was varied and irregular along the optic pathway, including thick, thin, and normal. Serial MRI scans redemonstrated optic nerve abnormalities with no evidence of progression or evolution.[4] Having said that, it is worth mentioning that in our patient, the thickened ipsilateral optic chiasm shows also mild T2 hyperintensity which is not a consistent feature in the reported cases. In fact, Firouzabadi et al., who studied a large number of MGDA with optic pathway thickening in their cohort, they reported normal T2 signal intensity of the optic nerves and optic chiasm in all subjects.[8] Therefore, a follow- up MRI has been planned for our patient.

MGDA is commonly associated with other facial and midline abnormalities that necessitate further MRI imaging of the brain and orbits.[1,2] Examples of midline abnormalities are cleft lip and palate, basal encephalocele, and corpus callosum agenesis.[1,3] Other reported associated midline anomalies include persistent craniopharyngeal canal, pituitary gland and infundibular deformity, and tubular or nodular nasopharyngeal lesions.[8] Intracranial vascular abnormalities like segmental aplasia/hypoplasia of the circle of Willis vessels or the Moyamoya phenomenon have also been described with MGDA.[1,3,10]

Although the majority of MGDA cases are unilateral, cases with bilateral MGDA have been reported in literature.[8,9] In addition, despite most cases of MGDA being sporadic, there are few cases that were found to be familial and associated with other genetic mutations or other syndromes like PHACE syndrome, which is characterized by combinations of congenital anomalies including posterior fossa malformations, hemangiomas, arterial anomalies, coarctation of the aorta and cardiac anomalies, and eye abnormalities.[1]

The cardinal MRI features of MGDA are funnel shaped morphology of the posterior optic disc with elevation of the retinal surface, defect and discontinuity of the uveoscleral coat, and abnormal tissue associated with the ipsilateral distal intra-orbital segment of the optic nerve.[2,4] These findings differentiate between MGDA and other differential diagnoses and ocular anomalies like coloboma and peripapillary staphyloma.[2] Coloboma is a term used to describe any focal discontinuity in the structure of globe.[2,3] MGDA was described in some literature as a focal subtype of coloboma of the optic nerve head.[2] Staphyloma occurs due to choroidal thinning and stretching without a defect.[2] MGDA is also differentiated from both coloboma and staphyloma by the presence of abnormal central glial tuft, pigmentary changes, and radially oriented vessels, which are best seen during fundoscopic examination.[1,3] Optimization of visual acuity with refractive correction and occlusion therapy of the good fellow eye is crucial to prevent amblyopia.[3]

In conclusion, MGDA is a rare disc anomaly which has typical fundoscopic and radiological appearance. Orbital and brain MRI is recommended in such cases as a screening tool to look for other midline anomalies and vascular abnormalities.

Disclosure Statement

The authors declared no conflicts of interest. An informed consent was obtained from the patient's parent.

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