Original Article/นิพนธ์ต้นฉบับ

Morning Glory Disc Anomaly with Clinical Bilateral Microphthalmos and Midline Facial Defect

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บทคัดย่อ

ภาวะขั้วประสาทตาผิดปกติตั้งแต่กำเนิดแบบ morning glory disc ที่เกิดขึ้นทั้ง 2 ข้างเป็นภาวะที่พบได้น้อย โดยผู้ป่วยในรายงานนี้ ตรวจพบว่าขั้วประสาทตาผิดปกติทั้ง 2 ข้าง โดย 1 ข้าง มีความผิดปกติของดวงตามากจนทำให้มีขนาดเล็กมากผิดปกติ (microphthalmos) และยังมีความผิดปกติของการสร้างกระดูกใบหน้าและบริเวณส่วนกลางของสมอง (midline facial defect) ซึ่งบริเวณที่สำคัญคือต่อมใต้สมอง (pituitary gland) ที่ต้องใช้ในการสร้างฮอร์โมนที่สำคัญสำหรับการดำรงชีวิต จากรายงานนี้ต้องการเน้นย้ำความสำคัญของในการวินิจฉัยภาวะขั้วประสาทตาผิดปกติที่สามารถพบร่วมกับภาวะการทำงานของต่อมใต้สมองผิดปกติ ซึ่งจักษุแพทย์ควรส่งตรวจเพิ่มเติมและส่งปรึกษากุมารแพทย์ผู้เชี่ยวชาญได้อย่างเหมาะสม เพราะหากขาดความใส่ใจในด้านนี้ ผู้ป่วยอาจไม่ได้แค่สูญเสียการมองเห็น แต่หากการวินิจฉัยและรักษาภาวะการทำงานของต่อมใต้สมองผิดปกติล่าช้า จะส่งผลให้ผู้ป่วยอาจมีปัญหาเรื่องการเจริญเติบโตและพัฒนาการ ซึ่งจะยิ่งทำให้ผู้ป่วยเสียโอกาส

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คำสำคัญ : ขั้วประสาทตาผิดปกติตั้งแต่กำเนิด, Morning glory disc

ผู้นิพนธ์ทั้งหมดไม่มีส่วนเกี่ยวข้องหรือผลประโยชน์ใดๆ กับผลิตภัณฑ์ที่ได้กล่าวอ้างถึงในงานวิจัยนี้

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Original Article/นิพนธ์ต้นฉบับ

Morning Glory Disc Anomaly with Clinical Bilateral Microphthalmos and Midline Facial Defect

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Abstract

Bilateral morning glory disc anomaly (MGDA) is a rare presentation. Here we report the case of a 4-year-old boy with bilateral MGDA and midline facial defect, likely to be the cause of bilateral microphthalmos and pituitary gland function abnormality. This rare entity is of important interest to both pediatrician and ophthalmologist, as an exemplary case of early detection and treatment of pituitary hormone abnormality in childhood. **Thai J Ophthalmol 2018; January-June 32(1): 25-30.**

Keywords: microphthalmos, midline facial defect, morning glory disc anomaly

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Introduction

Morning glory disc anomaly (MGDA) is a non-progressive congenital abnormality of the optic disc and peripheral retina, as previously described by Kindler in 1970.[1] The term "morning glory disc" was derived from the shape of the disc resembling a morning glory flower. The prevalence of this condition is unknown, due to its uncommon occurrence. MGDA usually manifests as a unilateral condition, however, bilateral conditions have also been reported.[2] There are a number of other ocular and systemic abnormalities associated with MGDA.

Ocular abnormalities associated with MGDA include decreased visual acuity, strabismus, peripheral arterio-venous anastomoses, persistent fetal vasculature (PFV), cataract, retinal detachment and microphthalmos.[3-5] The MGDA-associated systemic abnormalities are uncommon, and some could be life-threatening. These include midline facial abnormalities, basal skull defect, trans-sphenoidal basal encephalocele, agenesis of corpus collosum, panhypopituitarism and Moyamoya disease.[3, 6] It is very crucial to detect these neurological abnormalities associating with MGDA with appropriately neuroimaging include magnetic resonance imaging (MRI) and magnetic resonance angiogram (MRA).[3, 7] The neuroimaging not only provides the findings of MGDA-associated neurological abnormalities, but also confirms the diagnosis of MGDA from other optic nerve anomalies.[8]

Case report

A 4-year-old boy came to Ramathibodi Hospital, Bangkok, for second opinion after being diagnosed with congenital blindness at the age of 2 months. His mother noticed that both of his eyes, especially the left one, appeared smaller when compared to other children of the same age. He was born full term in an uncomplicated pregnancy and delivery. He was otherwise a healthy child.

Ophthalmic examination showed wandering eye movement in both eyes. His visual acuity was light perception and no light perception in the right and left eyes, respectively. External eye examination showed that he had microphthalmos in both eyes, which was more severe in the left (corneal diameter of 8.5 x 10 mm in the right eye and 7.5 x 9 mm in the left eye) (Figure 1). Fundus examination revealed obscured view of the left eye due to early phthisis bulbi and enlarged excavated optic disc in the right eye with numerous retinal vessels emerging in a radial pattern. Other retinal findings in the right eye were fibroglial tissue in the central part of the optic disc, peripapillary pigmentary changes, and localized

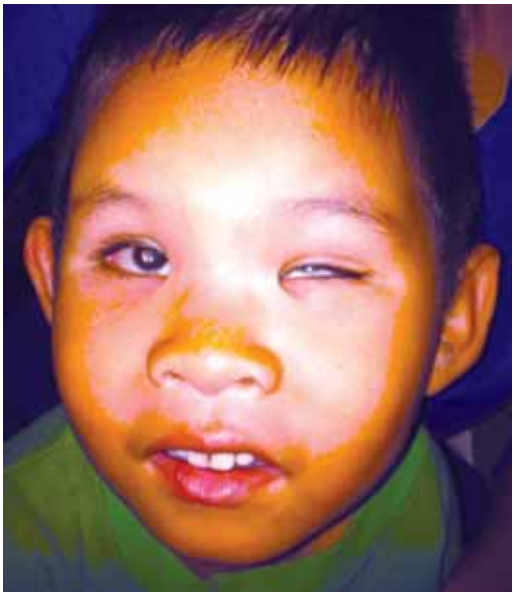

Fig. 1 *Face photography shows bilateral microphthalmos with minor dysmorphic facies including telecanthus, wide nasal bridge, and hypoplasia of mid face. (Colored figure is on page 45)*

subretinal fluid at the peripapillary area with no retinal breaks (Figure 2). On physical examination, he had minor dysmorphic facies including telecanthus, wide nasal bridge, and hypoplasia of the mid face (possible maxillary hypoplasia). The patient had no cleft lip or palate and was otherwise healthy.

MRI of the brain showed midline defect at the floor of the sella with inferior retraction of third ventricle and optic chiasm without herniation of brain tissue (Figure 3A). Presence of funnel-shaped optic discs in both eyes is noted, confirming the diagnosis of bilateral MGDA (Figure 3B). Presence of hyposignal

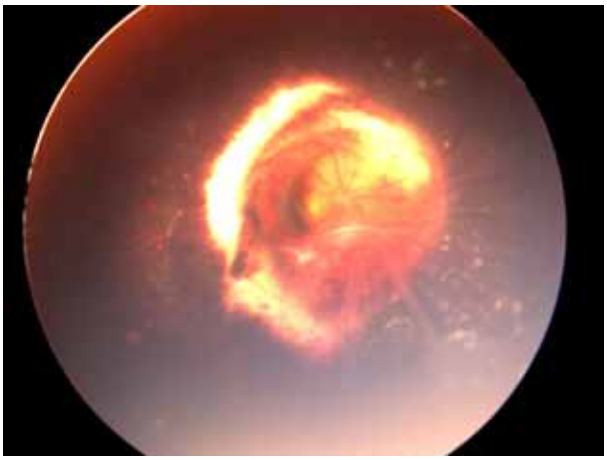

Fig. 2 *Fundus examination of the right shows typical characteristics of MGDA including an enlarged excavated optic disc, numerous retinal vessels emerging in a radial pattern, fibroglial tissue in the center of optic disc and peripapillary pigmentary changes. (Colored figure is on page 47)*

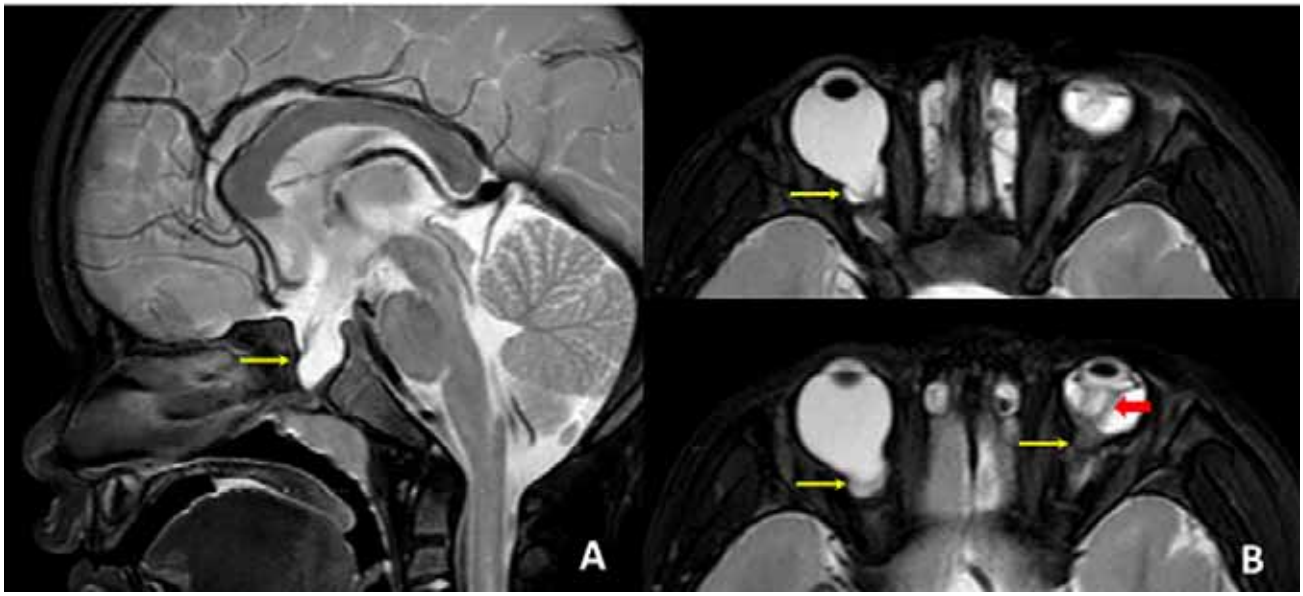

Fig. 3 *MRI of brain and orbit. (A) Sagittal T2 view shows midline defect of the floor of sella (arrow) with widened CSF-filled sellar turcica without definite herniation of brain tissue or meningeal sac. (B) Axial fat-saturated T2 of the orbit (top and bottom) demonstrates a funnel-shaped morphologic pattern of the optic disc (arrows) confirming the clinical diagnosis of bilateral MGDA and presence of hyposignal T2 retrolental soft tissue along the Cloquet's canal in the left orbit likely persistent fetal vasculature (red arrow). (Colored figures are on page 47)*

T2 retrolental soft tissue along the Cloquet's canal in the left orbit indicated likely PFV.

The endocrinology work up revealed the patient had isolated growth hormone deficiency, which was then treated and followed by an endocrine pediatrician.

Discussion

MGDA is one of the congenital abnormalities that normally occur sporadically and its cause is not fully understood. However, genetic association of the abnormality has recently been reported. In 2003, Azuma and coworkers reported 1 in 8 patients with optic nerve anomalies and mutation of *PAX6* gene had bilateral MGDA.[9] However, there is still very limited insight into the genetic components that can explain sporadic and isolated congenital optic disc malformation. Comprehensive genetic testing was not performed in our case.

The pathogenesis of MGDA is largely unknown,[10] however, a number of theories have been proposed, including primary mesenchymal abnormality, abnormal closure of the embryonic fissure and enlargement of the optic stalk during embryonal development.[3, 11, 12] The visual acuity of the MGDA patients is usually poor. However, the visual acuity may vary from 20/20 to no light perception with only ~30% of patients having visual acuity better than 20/40.[3] Most patients with MGDA were referred to pediatric ophthalmologists because of cataract and strabismus findings. The manifestation of MGDA with bilateral microphthalmos and PFV as in our case is uncommon. Unilateral microphthalmos has been reported to be associated with MGDA in up to 41% of the patients.[3] Patients with MGDA have high risk for retinal detachment (up to 33%)[3] and choroidal neovascularization.[13] Fortunately, retinal detachment associated with MGDA usually resolves spontaneously in up to 36% of cases over a period of time.[11, 14]

The differentiation of MGDA from optic nerve coloboma is important because systemic associations are different in these conditions. In our case there was typical characteristic fundus findings of MGDA in the right eye but we could not identify fundus findings in the left eye due to phthisis bulbi. However, the characteristic of MRI findings in the left eye is highly suggestive of MGDA. There are many associated life-threatening systemic conditions in MGDA that should be carefully recognised. In patients with signs of midline facial abnormalities or frontonasal dysplasia including telecanthus, broad nasal tip, cleft lip and cleft palate should be carefully considered because they could be linked to basal encephalocele, agenesis of corpus collosum and pituitary related endocrinological abnormalities.[15] Despite the lack of cleft lip and cleft palate in our case, his minor dysmorphic facies including telecanthus with broad nasal tip were suggestive of the possibility of midline facial abnormalities. Another central nervous system abnormality that could occur with MGDA in up to 45% is Moyamoya disease, which is an abnormal narrowing or hypoplasia of the cerebral arteries. The disease may cause intellectual impairment, transient ischemic attacks, recurrent stroke, and seizure.[7] Due to many life-threatening conditions associated with MGDA, MRI and MRA are recommended imaging modalities in all patients diagnosed with MGDA. In addition to imaging, survey for endocrinological abnormalities should also be performed in all the MGDA patients.[3]

In conclusion, we have herein reported a rare presentation of MGDA, including bilaterality, microphthalmos and midline facial abnormalities. These presentations are highly suggestive of neurological and pituitary hormone abnormalities. Thus, neuroimaging and endocrinological investigations are crucial in cases with these presentations. This case highlights the importance of early recognition of systemic abnormalities associated with MGDA and midline facial abnormalities to both pediatrician and ophthalmologist.

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