Original Article

Ophthalmic and Systemic Manifestations in Congenital Optic Disc Anomaly: A Ten-year Retrospective Case Series

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Abstract

Introduction: Congenital optic disc anomaly is a broad spectrum of ophthalmic manifestations frequently associated with congenital visual impairment.

Objectives: To report the ophthalmic and systemic manifestations of congenital optic disc anomaly cases over ten-year.

Methods: The authors retrospectively reviewed the medical records of patients diagnosed with congenital optic disc anomalies and followed up from January 2011 to 2021.

Results: Eight patients were included in the study. The ages ranged from 1 to 35 years. Five patients were female, and three patients were male. Initial visual acuity ranged from 20/20 to counting fingers. Five patients were diagnosed with optic nerve hypoplasia, three were found with mid-line brain defects, one patient with panhypopituitarism, and isolated ONH in two patients. One patient with a morning glory disc found an ipsilateral carotid artery narrowing. One patient with optic nerve pit and another with optic disc melanocytoma found only ophthalmic manifestations. The clinical course was stable in all patients. One patient had a good visual prognosis, three patients had a fair visual prognosis, and four patients had a poor visual prognosis.

Conclusions: Some disease entities, namely, optic nerve hypoplasia and morning glory disc, are related to systemic manifestations that should be investigated and managed. The prognosis for the visual function is usually relatively stable. Neuroimaging can be instrumental in diagnosing neurological abnormalities, a rare but life-threatening condition. Young patients must be evaluated in a multidisciplinary approach with a pediatrician.

Keywords: Congenital optic disc anomalies, Optic nerve hypoplasia, Morning glory disc, Optic nerve pit, Optic disc melanocytoma

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Introduction

Congenital optic disc anomaly is a broad spectrum of ophthalmic manifestations frequently associated with congenital visual impairment. The optic disc has two main characteristics: optic nerve hypoplasia and excavated optic disc anomalies.[1] The excavated optic disc abnormalities comprise optic disc coloboma, morning glory disc, optic disc nerve pit, and peripapillary staphyloma.[2] Congenital optic disc anomaly can be unilateral or asymmetrical bilateral involvement. Bilateral involvement usually occurs with systemic manifestations or inherited genetic disorders with high genetic heterogeneity and different transmission patterns.[3,4] A structural optic disc anomaly related to optic nerve dysfunction often leads to reduces visual acuity and organic amblyopia.[5] Congenital optic disc anomaly may be associated with significant refractive error, strabismus, and infantile nystagmus.[6] Congenital optic disc anomalies are a leading cause of visual impairment in children and adults. However, ophthalmic manifestations may not be the only problem in these patients. Some patients often have systemic manifestations, primarily neurological or endocrine abnormalities, which may be life-threatening. The present study aims to describe the clinical findings and some visual prognosis of these rare diseases which consist of the associated ophthalmic and systemic manifestations among patients with congenital optic disc anomalies in the past ten-year retrospectively reviewed that will provide the physician the information and predict a possible visual outcome and further management decisions.

Methods

The Medical Ethics Committee approved the study of Thammasat University (MTU-EC-OP-1-251/64), Pathum Thani, Thailand, and was conducted by tenets of the Declaration of Helsinki. The authors retrospectively reviewed the medical records of patients diagnosed with congenital optic disc anomalies in the Department of Ophthalmology, Thammasat Hospital, Thammasat University, Thailand, from January 2011 to 2021. All the patients were informed and consented to the research methodology.

Optic nerve hypoplasia (ONH) is diagnosed by small optic disc size, double-ring sign, thinning of the retinal fiber layer, and vascular tortuosity.[7] Small optic disc size is defined by the ratio of disc-macula distance (DM) to disc-diameter (DD) was more than 3.[8] Optic disc coloboma is diagnosed by a demarcated optic disc excavation located inferiorly. Morning glory disc is diagnosed by a funnel-shaped excavation of an enlarged optic disc with central glial tuft, elevated peripapillary retinal pigmentation, and an abnormally increased number of retinal vessels at the disc margin.[9] Optic nerve pit is diagnosed by a round or oval, grayish or yellowish defect in the inferotemporal quadrant or central portion of the optic disc with/without serous macular detachment.[10] Peripapillary staphyloma is diagnosed by a posterior bulging of the sclera encompassing the optic disc. Characteristic fundus findings of each disease diagnose other congenital optic disc anomalies.[11]

Results

A total of ten patients were enrolled. Two patients with missing data were excluded. Consequently, eight patients were included in the study. They had ages from 1 to 35 years. Five were female, and three were male. Initial BCVA ranges from 20/20 to counting fingers. Five patients were diagnosed with ONH, one patient with Morning glory disc, one patient with optic nerve pit, and one patient with optic disc melanocytoma. The authors analyzed the best-corrected visual acuity (BCVA), and the visual prognosis was categorized into good (BCVA 20/40 or better), fair (BCVA 20/50-20/200), and poor (BCVA 20/200 or worse).

The five patients with ONH presented with sensory nystagmus in three patients and strabismus in two. All five patients were assessed for systemic manifestations, including neuroimaging and endocrine workup. Neuroimaging found mid-line brain defects in three patients and isolated ONH in two patients. An endocrine workup found panhypopituitarism in one patient. A Morning glory disc patient presented with unilateral decreased vision and sensory exotropia. Neuroimaging found an ipsilateral internal carotid artery stenosis, the patient with optic nerve pit and an optic disc melanocytoma presented with reduced vision without systemic manifestations (Table 1).

Table 1 Summary of the data of all reported eight patients with congenital optic disc anomaly

No.	Age (years)	Gender	Laterality	Diagnosis	Initial BCVA	Ophthalmic manifestations	Systemic manifestations	Latest BCVA
1	1	F	Bilateral	ONH	F and F	Nystagmus, High myopia, Esotropia	Agenesis of corpus callosum, Absent septum pellucidum, Panhypopituitarism, Schizencephaly, Delay development	20/200, 20/200
2	1	M	Bilateral	ONH	Not F and F	Nystagmus	Absent septum pellucidum, Hypoplasia of corpus callosum, Bilateral colpocephaly, Epilepsy, Delay development	20/100, 20/100
3	2	M	Right eye	ONH	Fc 1 foot	Exotropia	None	Fc 1 foot
4	4	M	Left eye	ONH	Fc 2 feet	Exotropia	None	PL
5	11	F	Left eye	ONH	20/100	None	Absent septum pellucidum	20/100
6	3	F	Left eye	Morning glory disc	20/150	Exotropia	Left ICA stenosis, Antiphospholipid syndrome	20/200
7	33	F	Right eye	Optic disc melanocytoma	20/50	None	None	20/50
8	35	F	Right eye	Optic nerve pit	20/20	None	None	20/20

BCVA: best-corrected visual acuity, Fc: counting finger, F and F: fix and follow, ICA: internal carotid artery, ONH: optic nerve hypoplasia, PL: light perception

Case example

Case 1

A 1-year-old child presented with crossed eyes. The mother noticed her child had crossed eyes since birth. The child can fix and follow with both eyes. External eye examination revealed horizontal pendular nystagmus. Krimsky test showed esodeviation of 45 prism diopters (PD) with normal ocular motility. The pupil size was 3 mm both slow reaction to light. Anterior segments were unremarkable in both eyes. Dilated fundus revealed optic nerve hypoplasia in both eyes. Cycloplegic refraction revealed high myopia in both eyes. Magnetic resonance imaging (MRI) of the brain showed right open lip schizencephaly with absent septum pellucidum and agenesis of the corpus callosum (Figure 1). Endocrine workup found hypopituitarism, including growth hormone, thyroid-stimulating hormone, and adrenocorticotropic hormone. The patient was diagnosed with septo-optic dysplasia.

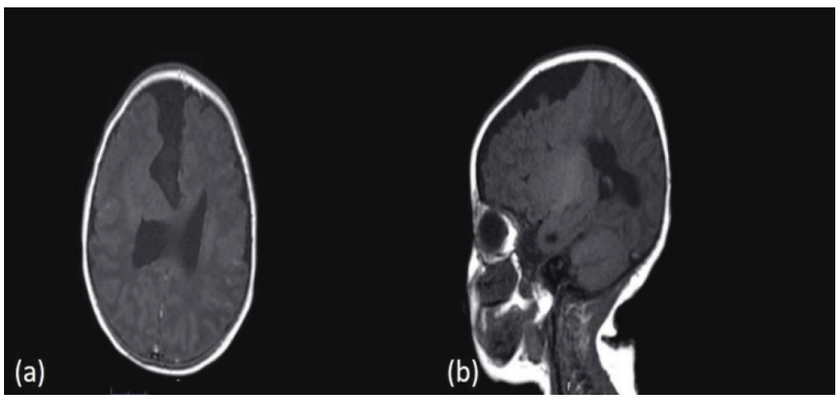

Figure 1 (a) MRI brain axial view shows right open lip schizencephaly with absence of septum pellucidum, (b) MRI brain sagittal view shows agenesis of the corpus callosum.

Case 4

A 4-year-old boy presented with a right eye drifting outward. BCVA in the right eye was counting finger 2 feet and 20/40 in the left eye. Modified Krimsky test revealed exodeviation of 30 PD with normal ocular motility. The pupil size was 3 mm both reacted to light with a relative afferent pupillary defect (RAPD) in the right eye. Anterior segments were unremarkable in both eyes. Dilated fundus revealed optic nerve hypoplasia in the right eye (Figure 2). The left eye fundus was within normal limits. Cycloplegic refraction revealed myopic astigmatism in the right eye. An MRI of the brain showed no significant intracranial abnormalities. Endocrine workup was negative. The patient was diagnosed with isolated optic nerve hypoplasia.

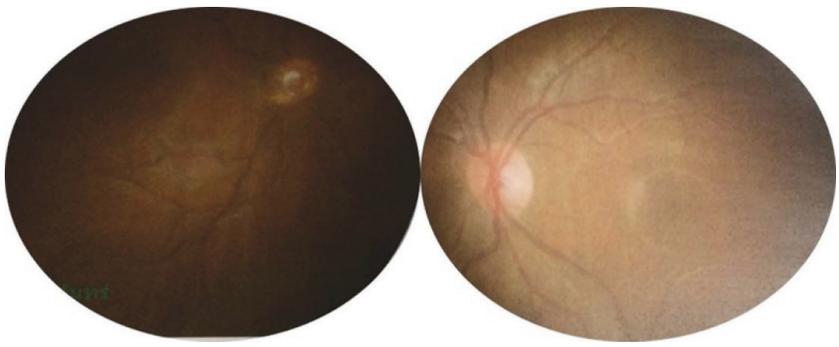

Figure 2 shows the double-ring sign in the right eye with optic nerve hypoplasia.

Case 6

A 3-year-old girl presented with a left eye drifting outward. BCVA in the right eye was 20/40 and 20/150 in the left eye. The Krimsky test revealed exodeviation of approximately 30 PD with normal ocular motility. The pupil size was 3 mm both react to light. Anterior segments were unremarkable in both eyes. Dilated fundus revealed a funnel-shaped excavation of an enlarged left optic disc with a central glial tuft, elevated peripapillary retinal pigmentation, and an abnormal retinal vascular pattern (Figure 3). The right eye fundus was within normal limits. Cycloplegic refraction revealed normal in both eyes. Magnetic resonance angiography (MRA) brain showed narrowing of the supraclinoid and petrous part of the left internal carotid artery with the normal brain parenchyma (Figure 4). The patient had supraventricular tachycardia and was positive for anti-beta 2 glycoprotein. The patient was diagnosed with Morning glory disc and antiphospholipid syndrome, which requires long-life antiplatelet therapy.

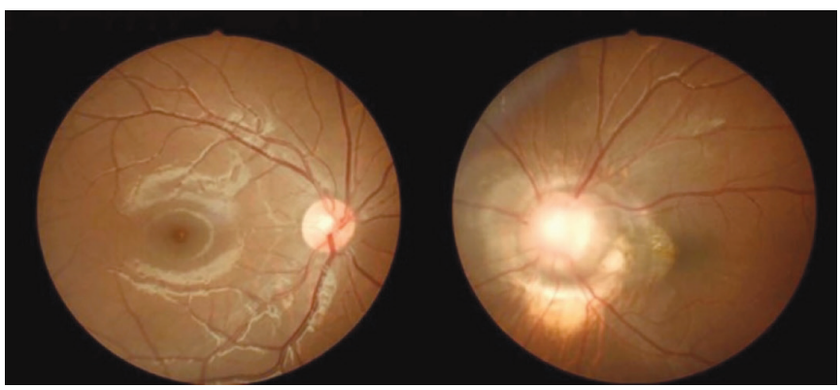

Figure 3 showed a funnel-shaped excavation of an enlarged left optic disc with a central glial tuft, elevated peripapillary retinal pigmentation, and an abnormal retinal vascular pattern.

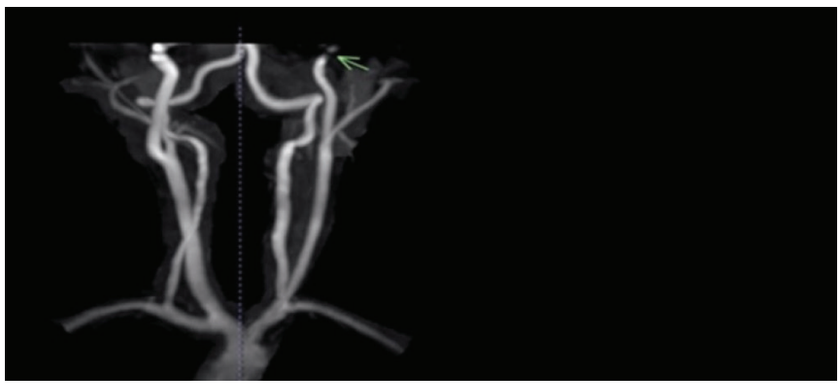

Figure 4 shows the narrowing of the supraclinoid and petrous part of the left internal carotid artery (arrow).

Case 7

A 33-year-old female presented with dry eye in the right eye for two weeks. She received lubricant from an eye clinic when there was a chance of detection of decreased vision in the right eye. She had not had a recent trauma. Her ocular and medical history were unremarkable. BCVA in the right eye was 20/50 and 20/20 in the left eye. The pupil size was 3 mm both react to light with an RAPD in the right eye. Anterior segments were unremarkable in both eyes. Dilated fundus revealed a black elevated mass lesion about 3 disc areas in size that obscured the optic disc (Figure 5). The left eye fundus was within normal limits. There was no serous retinal detachment. B scan ultrasonography of the right eye showed a hyperechoic dome-shaped lesion over the optic disc without a juxtapapillary choroidal lesion. The automated visual field 30-2 showed constriction of the visual field with a residual central island in the right eye and a normal visual field in the left eye. The patient was diagnosed with optic disc melanocytoma and was monitored annually.

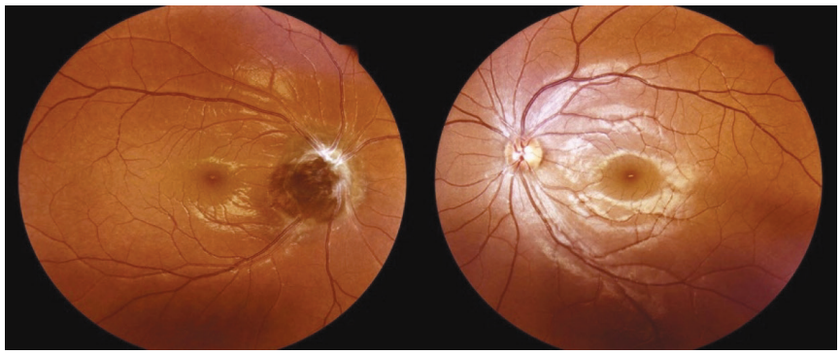

Figure 5 shows a black elevated mass lesion that obscured the right optic disc.

Case 8

A 35-year-old female visited for a routine eye checkup. She had no visual complaints. She was healthy and had no other pertinent past ocular or medical history. BCVA was 20/20 in both eyes. The pupil size was 3 mm both react to light without an RAPD. Anterior segments were unremarkable in both eyes. Dilated fundus revealed a round yellowish defect in the temporal portion of the right optic disc without serous macular detachment (Figure 6). The cup-to-disc ratio was estimated at 0.6 in the right eye and 0.3 in the left eye. The left eye fundus was within normal limits. The macula was flat in both eyes. The patient was diagnosed with an optic nerve pit and was monitored for maculopathy.

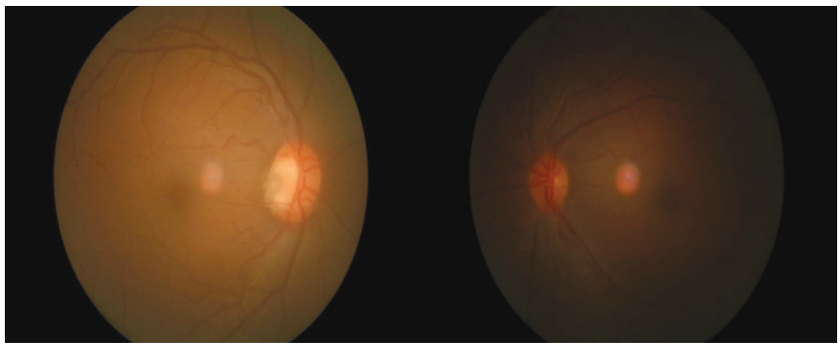

Figure 6 shows a round yellowish defect in the temporal portion of the right optic disc without serous macular detachment.

Discussion

Congenital optic disc anomalies can cause a wide range of clinical presentations. Most patients are accompanied by visual impairment. Signs of visual impairment in young patients are often presented with nystagmus or strabismus. The present study found strabismus in 3 patients, nystagmus in 1 patient, and strabismus with nystagmus in 1 patient. Therefore, young children presented with infantile nystagmus or strabismus should look for abnormalities in the visual sensory pathway, especially the congenital optic disc anomaly.

Visual function was stable in all patients as the clinical course in the disease entities is usually relatively stable. One case had a good visual prognosis, three had a fair visual prognosis, and four had a poor visual prognosis. Patients with ONH had the youngest age because visual impairment leads parents to bring them to see a physician sooner, allowing for earlier diagnosis. In this study, some patients may be diagnosed in adulthood because they are asymptomatic or have mild visual impairment as a patient with optic nerve pit and optic disc melanocytoma. These patients are more likely to have an excellent visual prognosis than ONH and Morning glory disc.

In the present study, we found ONH most commonly seen with congenital optic disc anomaly as in the previous studies.[12] The diagnosis of ONH is based on the characteristic abnormal small optic nerve size. Visual impairment did not depend on the size of the optic nerve but the decreased number of axons in the involved nerve and integrity of the papillomacular bundle, causing this group to have a wide range of vision from 20/20 to no light perception (NPL).[13] Visual prognoses may still be challenging to evaluate in the young patient, but we found that majority of ONH have a poor prognosis. ONH has been associated with systemic manifestations such as septo-optic dysplasia (de Morsier syndrome), which is composed of ONH, panhypopituitarism, and mid-line brain defects such as absent septum pellucidum and agenesis of the corpus callosum.[14] A brain MRI is recommended to avoid radiation exposure in children, and endocrine workup includes fasting glucose, morning cortisol, prolactin, thyroid-stimulating hormone (TSH), free triiodothyronine (T3), and free thyroxine (T4) in all cases.[15] Most of the brain abnormalities were mid-line brain defects. We found two patients who have isolated ONH and one patient who has panhypopituitarism. We also found that the two patients with bilateral involvement had delayed development. Borchert et al. found that bilateral ONH is associated with a higher risk of pituitary dysfunction and delayed development, which corresponds with our patients.[16]

Our patient with Morning glory disc had a poor visual prognosis as the previous study found that visual acuity is usually poor, with only 30% achieving 20/40 or better. Morning glory disc has been associated with systemic manifestations such as encephalocele, corpus callosum agenesis, endocrine abnormalities, and Moyamoya disease.[17] Neuroimaging and vascular imaging should be performed in all patients. We found our patient has ipsilateral internal carotid artery narrowing without associated collateral vessels or cerebral ischemia, as in Moyamoya disease. This patient has a concomitant autoimmune disorder, but fortunately, the patient has been stable without signs of progression in subsequent follow-ups.

Our patient with optic nerve pit was asymptomatic and detected in adulthood. The previous study found that the optic nerve pit will be symptomatic when an associated serous macular detachment is presented and frequently occur in the third and fourth decade of life. The extension of maculopathy affects visual impairment and visual prognosis.[18] No systemic manifestations have been reported. Most tests are limited to eye investigations. Investigations are usually performed to differential diagnose with other optic neuropathy and monitor macular complications. For this reason, our patient had an excellent visual prognosis and can subsequent examinations to look for macular complications.

Our patient with optic disc melanocytoma presented with decreased vision in adulthood. The previous study found that patient with optic disc melanocytoma was diagnosed at the mean age of about 50 years (range 1-91 years). At birth, the optic disc is usually an amelanotic lesion and becomes pigmented over a long time. Therefore, lesions may not be clinically detected at an early age. The diagnosis is based on the characteristic fundus findings. Optic disc melanocytoma is an isolated condition, but an association with an intracranial tumor has been reported.[19] Neuroimaging may help exclude other intracranial lesions. The patient should be follow-up annually to confirm its benign nature.

A limitation of the study was the retrospective method. There was a limited number of patients. Some patients were excluded due to incomplete medical records. There is not enough variety for each disease. In conclusion, some disease entities, namely, optic nerve hypoplasia and morning glory disc, are related to systemic manifestations that should be investigated and managed. The prognosis for the visual function is usually relatively stable. Neuroimaging can be instrumental in diagnosing neurological abnormalities, a rare but life-threatening condition. In addition to ophthalmic manifestation, systemic involvement should be evaluated. The young patient must be evaluated in a multidisciplinary approach with a pediatrician.

Acknowledgments

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Conflict of interest. All authors report no conflicts of interest relevant to this article.

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