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Case Report

Congenital Optic Disc Anomalies - Diagnosis and Clinical Manifestations -

Anushree Gupta*

Ophthalmology Department, Dr. Radhakrishnan Government Medical College, Hamirpur, Himachal Pradesh

***Address for Correspondence:** Anushree Gupta, Ophthalmology Department, Dr. Radhakrishnan Government Medical College, Hamirpur, Himachal Pradesh, India, Tel: +91-780-749-8219; E-mail: anushree0612@gmail.com

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ABSTRACT

Congenital optic nerve head anomalies are a group of structural malformations of the optic nerve head and surrounding tissues. Visual prognosis depends on the type of anomaly present. Different systemic associations are associated with congenital optic nerve anomalies. Newer ancillary investigations aid in diagnosis as well as detection of complications. Therefore, correct diagnosis and monitoring is important. Congenital optic nerve head anomalies are classified according to abnormalities of optic disc size, conformation or by the presence of abnormal tissue at the nerve head (pseudo-swelling). Here we present two cases of congenital optic disc anomalies with abnormality of conformation- Optic disc pit and Morning glory disc.

Keywords: Congenital; Optic disc; Decreased vision

ABBREVIATIONS

OCT: Optical Coherence Tomography; BCVA: Best Corrected Visual Acuity; MRI: Magnetic Resonance Imaging

INTRODUCTION

Optic nerve anomalies can categorised as congenital or acquired. Congenital optic nerve anomalies are classified according to abnormalities of optic disc size, conformation or by the presence of abnormal tissue at the nerve head (pseudo-swelling). Acquired abnormalities of the optic nerve are classified according to the reaction of the optic nerve to insult: cupping, swelling, and atrophy. Here we present two cases of congenital optic disc anomalies with abnormality of conformation- Optic disc pit and Morning glory disc.

CASE 1

A 34 year old lady presented in the outpatient department with chief complaint of poor vision in the right eye for the past 8 years and progressively decreasing for past 3 months. There was no history of pain, redness, flashes orfloater. General physical and systemic examination was normal. Her best-corrected visual acuity was 6/60 in the right eye and 6/6 in the left eye. Anterior segment and intraocular pressure was normal. Pupils were round and reactive. Dilated fundus examination of the right eye showed an oval, greyish cater like defect 1/4th disc size in the temporal aspect of the disc. Macular detachment is seen confined within the arcades. Retinal pigmentery epithelial changes were present in the central macular area (Figure 1&2). Fluorescein angiography showed early hypofluorescence of the optic pit followed by late hyperfluorescence. A gradually increasing hyperfluorescence with a petalloid pattern of hypo- and hyperfluorescence was seen in the area of serous macular detachment (Figure 3). Stratus optical coherence tomography of the right eye revealed neurosensory macular detachment with splitting of inner and outer retinoschisis like separation in the inner retinal layers. A hyperreflective tissue was seen over the pit area and communication between the macular detachment and pit was clearly visible (Figure 4&5). A diagnosis of optic pit with serous macular detachment was made. Patient refused treatment due to personal reasons.

CASE 2

A 50-year-old woman showed up in outpatient department for eye examination. Her BCVA in the right eye was 6/6 and in left eye were hand movements close to face only. Her left eye vision had been poor since childhood. Examination of anterior segment and intraocular pressure was within normal range. There was a mild afferent pupillary defect in the left eye. Ocular alignment was orthophoric and extra ocular movements were full.

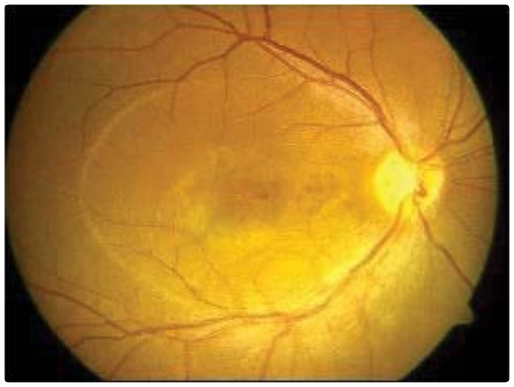

Figure 1: Fundus photograph of the right eye: Optic pit in temporal aspect of disc with macular detachment.

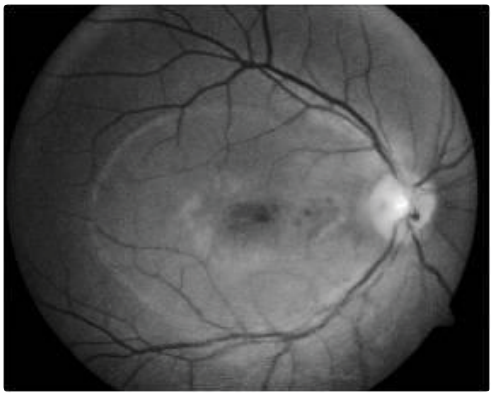

Figure 2: Red free photograph of the right eye.

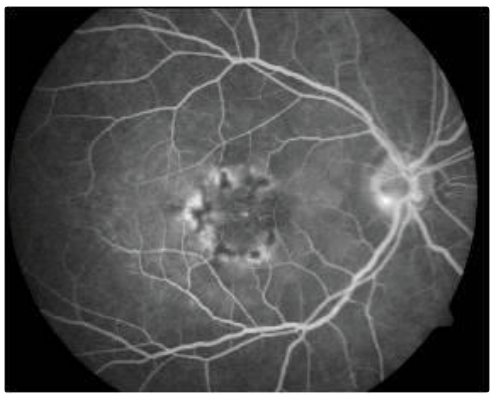

Figure 3: Fluorescein Angiography of the right eye: late phase showing hyperfluorescence of the pit and macular area.

Fundus examination of right eye was normal. Fundus examination of left eye showed an enlarged and excavated funnel shaped optic disc, with multiple anomalous, thin vessels radiating circumferentially. A tuft of white material obscured the central cup. There was a wide area of peripapillary pigment abnormality, with zones of hyperpigmentation and hypopigmentation (Figure 6). Fundus fluorescein angiography was normal. MRI brain and orbit was normal. Rest of her general physical and systemic examination was normal. Diagnosis was made of isolated Morning Glory Syndrome.

DISCUSSION

Optic disc pit

Optic disc pit are rare congenital anomaly first described by Weithe in 1882 [1]. They are localized excavations in the optic disc and are mostly located on the temporal aspect of the optic disc. They occur equally in men and women with an estimated incidence of 1 in 11,000 people.

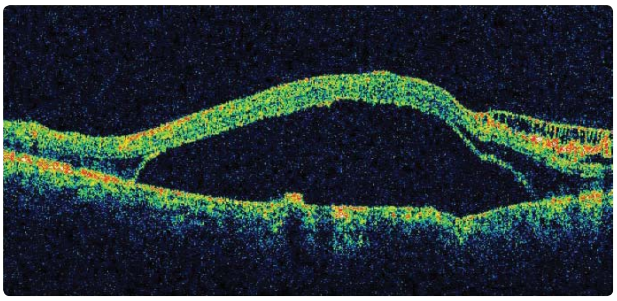

Figure 4: OCT of the right eye: shows serous macular detachment and retinoschisis.

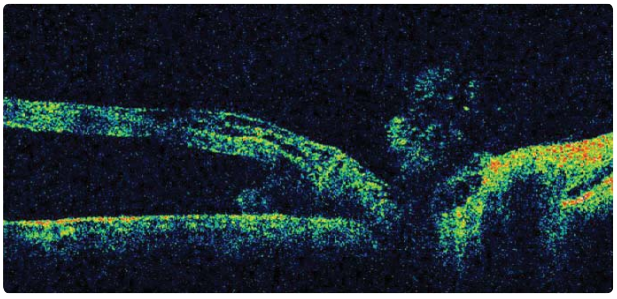

Figure 5: OCT of the right eye: shows communication between macular detachment and optic pit.

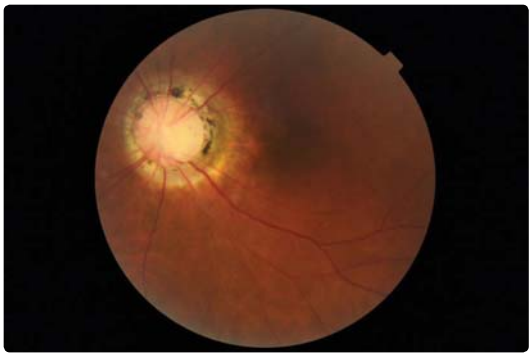

Figure 6: Fundus photograph of the left eye: Morning glory disc.

Pits are unilateral in 95% of cases; vary in color, mostly being grey. Peripapillary retinal pigment epithelial changes and choroidal atrophy is seen most of the eccentrically located pits. Optic disc pit are usually asymptomatic although visual field defects and maculopathy has been demonstrated.

Visual field defects include nasal and temporal steps, altitudinal defects, para-central scotomas, arcuate scotomas, generalized constriction, and localized constriction [2,3]. Systemic associations are usually not seen conjunction with congenital optic pits.

Maculopathy occurs in 25-75% of patients with optic nerve pit usually between third and fourth decade of life leading to visual deterioration [4]. There are different theories regarding source of fluid in maculopathy. It is proposed that vitreous fluid can enter the subretinal space through the formation of a macular hole. Other possible sources are Cerebrospinal Fluid (CSF) through the optic disc pit defect, leakage from blood vessels at the pit and from the choroid, through the Bruch's membrane and peripapillary atrophy [5-7]. Progression of retinal fluid formation has been described in which fluid from the optic disc pit creates a schisis-like inner retinal separation, associated with a mild cecocentral scotoma. Then, an outer layer macular hole develops beneath the inner layer, associated with a dense central scotoma. The fluid then dissects subretinally creating an outer retinal detachment [8].

Optical coherence tomography can demonstrate subretinal fluid, retinoschisis, intraretinal fluid and lamellar macular holes in case of optic nerve with maculopathy [9]. Fluorescein angiography shows early hypofluorescence and late staining in the area of the optic disc pit. A well-delineated round or oval area of late hyperfluorescence corresponding to the area of macular elevation is seen [10].

Management of optic disc pit related maculopathy includes numerous techniques such as laser photocoagulation, intravitreal gas injection, macular buckling and pars plana vitrectomy [11].

Morning Glory anomaly

It is a rare disc anomaly first described by Kindler [12] in 1970. It is an nonprogressive, unilateral condition usually occurring as an isolated ocular anomaly. Vision is usually impaired in the affected eye. PAX 6 gene mutation can be responsible.

It is characterized by funnel-shaped excavation that includes the enlarged optic disc with a surrounding area of peripapillary chorioretinal pigmentary changes. A white glial tuft is present within the centre of the disc and represents persistent hyaloid remnants. Similar to petals on a flower, the blood vessels emanate radially from the disc. Blood vessels are increased in number and difficult to distinguish arteries from veins. If the excavation includes the macula, it is termed as macular capture.

It is common to find relative afferent pupillary defect, visual field defects such as enlarged blind spot, strabismus, and amblyopia. It can be complicated with serous retinal detachment in 30% of cases and rarely choroidal neovascularisation. Systemic associations include frontonasal dysplasia, mid facial anomalies, basal encephalocele, posterior fossa brain malformation, pituitary insufficiency [3].

Isolated Morning Glory syndrome usually does not require treatment unless it is associated with complications [13].

Management of Morning Glory syndrome with a non-rhegmatogenous serous detachment includes Pars Plana Vitrectomy (PPV), gas injection and peripapillary laser [14].

CONCLUSION

Diagnosis of optic disc anomalies is usually made on clinical examination itself and different investigative techniques are useful in confirming diagnosis and early detection of any complications. A proper systemic evaluation should be done wherever any systemic association is suspected.

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