Abstract:
This case report demonstrates the MRI orbit findings in a young female who presented with diminution of vision with fundus findings of morning glory disc. On MRI, funnel shaped morphologic pattern resembling characteristic ‘morning glory’ was revealed for bilateral optic discs. Embryology and genetics of this rare entity are also discussed briefly. Finally, conclusion was made that of bilateral ocular colobomas with characteristic MRI imaging features.

Keywords: Ocular coloboma, morning glory, scotoma, optic fissure, MRI orbit.

INTRODUCTION

Ocular coloboma resulting from failure of closure of the embryonic fissure are reported for 0.5 to 2.2 occurrences of every 10,000 live births. In few instances, it is linked to systemic illnesses and might be sporadic or inherited. A case studied with MRI Orbit was reported in our institute. The term coloboma is derived from a Greek word koloboma, which means a part that was removed by mutilation or is missing. It is a rare congenital defect and is caused by improper closure of the fetal optic fissure along the eye and optic nerve and is made up of small, isolated malformations in the uvea, retina, and optic nerve. The iris, lens, choroid, retina, and optic nerve can all be impacted by colobomas. They are labelled "typical" if located in the inferonasal quadrant of the affected structure and "atypical" if they are located elsewhere. A range of malformations result from variations in the size, location, and number of abnormalities. The most frequent complications include cataract and retinal detachment. The risk of latter is higher in those with coloboma of the posterior pole, which affects 23–42% of individuals. Ophthalmoscopically, the sclera is visible due to the lack of the overlaying uveal tract and retina, which clinically may result in visual field scotoma. Large optic nerve excavations are typically found inferiorly during a routine fundoscopic examination by a clinician.

OBJECTIVE

Objective of this case report is to study the findings and comprehend the location and extent of coloboma with the help of MRI orbit and to describe the MRI characteristic features of bilateral ocular colobomas.

CASE REPORT

We report a case of a 32-year-old female patient who presented with diminution of vision in the bilateral eyes for the past one year. Patient had no complains of hypertension and diabetes. No significant family history was obtained. Patient was referred to the radiology department for MRI Orbit after ophthalmological investigations.

The visual acuity of the patient was 6/6 for right eye and 6/9 for left eye. The visual field of the patient was tested via Humphrey visual field 24-2 test and demonstrated large blind spots or scotomas. Funduscopic findings revealed the classic morning glory disc in left eye, which is, enlarged disc with funnel shaped excavation and increased number of retinal vessels emerging from the rim of excavation and are arranged radially like spokes of wheel. This abnormality gets its name from its striking resemblance to the morning glory flower. Funduscopic findings in the right eye revealed glaucomatous disc. Optical coherence tomography was also done for the patient which showed normal results.

MRI Orbit was then performed for the patient to investigate the cause of scotomas. Images of 2-mm-thick sections were acquired at 2-mm intervals without use of contrast material. T2 Weighted 3D drive sequence is incorporated. It revealed a funnel shaped morphologic pattern of the right optic disc with elevated adjacent...
A Case Report Of Bilateral Ocular Colobomas With Characteristic Morning Glory Feature On MRI

 DOI: 10.9790/0853-2209011113 www.iosrjournal.org 12 | Page

retinal margins measuring ~ 4.7 x 3.4 mm (TRX CC). Effacement of the subarachnoid space was also noted in the distal intra-orbital segment of ipsilateral right optic nerve. The typical features of colobomas could not be appreciated in the Fundoscopic examination of Right eye. However, MRI Orbit study revealed typical funnel shaped morphologic pattern appearing like ‘morning glory’.

The left orbit also shows a funnel shaped morphologic pattern of the left optic disc with elevated adjacent retinal margins measuring ~ 1.6 x 1mm (TR X CC). Discontinuity of the normal uveo-scleral coat was noted.

Figure (a) shows axial section of T2W 3D DRIVE sequence demonstrating posterior excavation of right optic nerve disc with elevated adjacent retinal margins (white arrow), typically resembling ‘Morning glory’.

Figure (b) shows axial section of T2W 3D DRIVE sequence demonstrating posterior excavation of left optic nerve disc with elevated adjacent retinal margins (white arrow).

IV. DISCUSSION

Ocular coloboma is a rare congenital abnormality caused by failed choroidal or embryonic fissure closure during the fifth and seventh weeks of fetal development. Any ocular structure, such as the zonules and ciliary body, choroid, retina, and optic nerve, may be affected by colobomas. Colobomas can occur alone, in combination with other ocular conditions like microphthalmia, or as part of a multisystem disease like CHARGE syndrome (Coloboma, heart defects, choanal atresia, growth retardation, genital abnormalities and ear abnormalities). The fetal optic fissure, which runs infero-nasally along the optic nerve and globe, can fail to fuse, leading to typical colobomas. Fetal optic vesicle serves as the genesis of the eye and optic nerve. Two different types of invaginations occur in the optic vesicles during development. The outer layer—the future neurosensory retina—approaches the inner layer—the future retinal pigmented epithelium—to form the optic cup. A fetal fissure is simultaneously formed by invagination along the optic cup and stalk's inferior medial surface. The two retinal layers oppose one another at the fetal fissure. The layers combine to form the finished neurosensory and pigmented retina. Colobomas likely develop when the inner layer of the retina overgrows and exhibits exaggerated eversion
at the fissure, disrupting normal apposition and fusion. The length and location of the exaggerated protrusion of the inner layer affect the defect's size and location.4

Typical colobomas are autosomal dominant in trait. Bilateral and unilateral defects are found at the same rate. Vision may be unaffected or impacted to variable degrees by scotomas. A keyhole-shaped defect in the iris tissue frequently characterizes anteriorly situated colobomas. The optic nerve, retina, and choroid may be affected by a coloboma that is posteriorly placed. Involved retina is reduced to glial tissue and lacks an underlying retinal pigment epithelium (RPE) or choroid. The intersection of the coloboma and normal retina displays this as a whitening region that frequently has pigment deposition. Optic nerve involvement can occur in a variety of ways, from physiological cupping to severe retinal involvement.6 Microphthalmos, iris and ciliary colobomas, neovascular membranes, macular holes, lens notching, and optic nerve colobomas have all been linked to optic nerve colobomas. A retrobulbar fluid-density cyst may also be present that may communicate with the subarachnoid space.6 The appearance of colobomas on CT and MRI can vary. Broadly, they are divided into two variants: optic nerve head coloboma with associated midline structural abnormalities of the brain and skull and coloboma with a cyst with potential extrusion of the vitreous posteriorly into the cyst. Examination of unilateral optic nerve head colobomas may reveal a wide range of fundoscopic findings ranging from minor physiologic excavations.4 Associated abnormalities include microphthalmia being the most common, optic nerve atrophy, trans-sphenoidal encephalocele, midline cranio-cerebro-facial cleft and corpus callosum agenesis. In our case, MRI revealed right sided ocular coloboma which was not appreciated on fundoscopy. It shows typical morning glory flower appearance on MRI. However, in our case there was no obvious microphthalmos.

V. CONCLUSION

We conclude that fundoscopy is used for the diagnosis of colobomas, however imaging is used for its confirmation and for excluding other abnormalities as described above. The appearance on CT or MRI show that the affected globe is usually small with a focal posterior defect in the globe with vitreous herniation. On MRI Coloboma shows characteristic ‘Morning Glory’ appearance. We conclude that the entire spectrum of colobomas is to be well studied and diagnosed with the help of ophthalmological investigations, imaging findings and genetic correlation for the complete workup and treatment of the patient.

REFERENCES