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A CASE OF BILATERAL MACULAR COLOBOMA

Dr G. Lavanya Sri Ramya Gowthami* Resident, Department of Ophthalmology, Santhiram Medical College And General Hospital, Nandyal. *Corresponding Author

Dr A. Gowri Priya Resident, Department of Ophthalmology, Santhiram medical college and general hospital, Nandyal.

ABSTRACT Macular coloboma is a rare eye condition that affects around 0.5-0.7/10,000 live births. This is a case of a 13-year-old boy, who presented to our OPD with complaints of diminution of vision in both eyes since childhood. His visual acuity was 6/60 in the right eye and 6/24 in the left eye. There was no improvement with refraction. Slit lamp examination was normal in both eyes. IOP was normal in both eyes. Fundus examination of both eyes revealed B/L chorioretinal lesions in the macula which was larger in the right eye than the left eye .OCT picture of both eyes revealed a defect in the neurosensory area and choroid.

KEYWORDS : Coloboma, macula.

INTRODUCTION

Coloboma is an ocular congenital defect that can affect different areas of the eye such as the iris, the retina, the choroid, the lens or the optic nerve (1). Meanwhile, macular coloboma is an atypical coloboma that is hypothesized to be the result of incomplete differentiation of the arcuate bundle along the horizontal raphe during development (2). Macular coloboma is a rare eye condition that may affect about 0.5-0.7/10,000 live births (3). It is usually sporadic, although autosomal dominant or other inheritance patterns may be followed. Furthermore, the condition may be associated with various syndromes such as patau syndrome, Fraser syndrome, franceschetti syndrome, cat eye syndrome and many others (1). The abnormality of development resulted by intrauterine inflammation was considered to be the main cause of macular coloboma. However, genetic changes were also reported in some literature recently.

Case Presentation:-

A 13-year-old boy resident of Nandyal, a student by occupation came with chief complaints of diminution of vision in both eyes (RT>LT). Diminution of vision was undetected till his school age, which was detected by her school teacher when she observed the child copying notes from other children & advised for a check-up. No syndromes or associations were detected. His Best corrected visual acuity in RE was 6/60 and in the LE was 6/24. Intraocular pressure (IOP) on the non-contact tonometer was 16 mmHg and 12 mmHg in the right and left eye, respectively. The anterior segment examination was unremarkable in both eyes. Eye alignment by the Hirschberg test and cover-uncover test showed orthotropic alignment in both eyes. Extraocular muscle movement showed a full range of movement without limitation bilaterally. Fundus examination of RE showed a tilted disc and a Single, round, well-circumscribed excavated lesion of 3disc diameter size involving retina and choroid with underlying pigmentation and visible choroidal vessels temporal to disc. (Figure 1). Fundus examination of LE revealed a single, oval, well-circumscribed excavation of more than 1 disc diameter area involving the retina and choroid with underlying pigmentation temporal to disc. (Figure 2). OCT picture of BE revealed a defect in the Neurosensory retina and choroid. Based on the clinical examination and OCT imaging, the patient was diagnosed with bilateral macular coloboma.

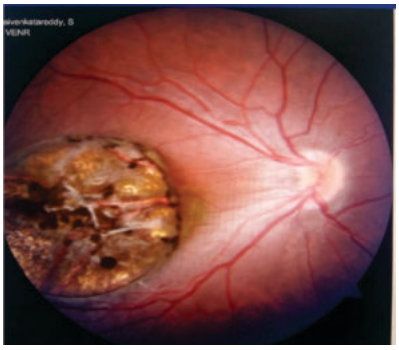

Figure 1

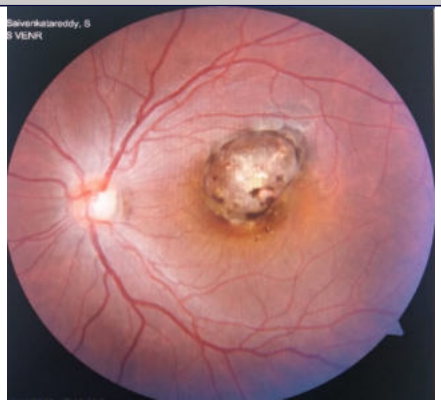

Figure 2

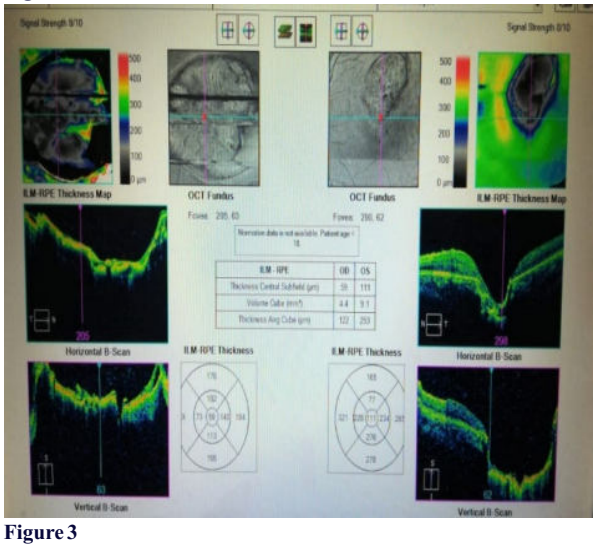

Figure 3

DISCUSSION:-

Macular coloboma results from incomplete or poor differentiation of arcuate bundles along the horizontal raphe, which normally occurs during fetal development (4, 5). As such, these are atypical colobomata as they do not fall in areas that originate from the embryonic cleft. On the other hand, ocular toxoplasmosis leads to post-inflammatory macular scars that can mimic the presentation of macular coloboma (6). Those scars are very similar in morphology to the scars of the congenital macular coloboma making it difficult to differentiate between congenital macular coloboma and post-inflammatory scars. The treatment is usually determined based on the differentiation between different macular scars causes. OCT is the most commonly used tool for the diagnosis of macular coloboma (4,7).

The absence of the retina and choroid layers was reported using OCT in our patient. Many macular coloboma cases are associated with other congenital syndromes and disorders (1). However, our patient did not have any other congenital abnormality or any family history of genetic diseases.

Macular coloboma, especially those located at the posterior pole, can lead to retinal detachment leading to an increased risk of developing complete vision loss (8). Therefore, regular follow-up for patients with macular coloboma is indicated. During each follow up a dilated fundus examination should be done for the patient to elicit any breaks at the coloboma edges (8). Prophylactic laser photocoagulation may be indicated for colobomas not involving the macula and papillomacular bundle. In addition, patients with macular colobomas should be treated carefully for any refractive errors to maximize and optimize the patient's visual acuity (8).

CONCLUSION:-

Congenital coloboma is a rare ocular condition that leads to a non-progressive decrease in visual acuity and if not followed up regularly may cause retinal detachment and vision loss. Optical coherence tomography is the method of choice in diagnosing macular coloboma while treatment should be done by laser photocoagulation if the coloboma is away from the macula with careful observation and follow-ups. Low vision rehabilitation services are also to be borne in mind during rehabilitative management. The presence of a macular coloboma not only warrants an ophthalmic evaluation and management but also a multi-disciplinary approach for the appropriate management of the associated abnormalities.

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