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Case Report

Bilateral Optic Disc Coloboma with Coexisting Unilateral Retinochoroidal Coloboma in an Adolescent from Northwestern Nigeria: A Case Report

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Abstract

Optic disc coloboma is a rare congenital anomaly of the optic nerve. We report a case of bilateral optic disc coloboma with coexisting unilateral retino choroidal coloboma in an adolescent from Northwestern Nigeria, who presented with complaints of difficulty seeing distant objects for 3 months, associated with ocular pains, tearing, and headache while reading. She had never worn spectacles. No previous history of systemic illnesses or trauma. Developmental milestones were optimal for age. Ocular examination revealed a best corrected visual acuity of 6/5 and 6/6 in the right and left eyes respectively. Dilated fundoscopy revealed an enlarged bilateral optic disc with a white bowl-shaped excavation, absent inferior neuro retinal rim, thin superior rim, and an area of well-defined oval retino choroidal defect, located inferior and slightly temporal to the right optic disc. No syndromic features or any ocular complications associated with optic disc and retino choroidal colobomas were seen.

Keywords: Optic Disc Coloboma; Retinochoroidal Coloboma; Adolescent; Northwestern Nigeria.

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Quick Response Code:

Introduction:

Coloboma is derived from the greek word meaning mutilation [1]. It occurs usually as a result of partial or complete defective closure of embryonic or choroidal fissure during embryonic life and could be associated with other congenital abnormalities[2].Optic disc coloboma (ODC) is visible clinically as a bowl-shaped depression usually situated inferiorly with an absent inferior neuroretinal rim with bilateral and unilateral presentations having an equal frequency[3].ODC can occur as a sporadic case or as an autosomal dominant inheritance[3].Prevalence of ODC from population-based studies in adults ranges from 3.7/100,000 to 8/100,000[4].ODC can be isolated or associated with some ocular disorders such as microphthalmia or part of a syndrome such as CHARGE (Colobomas, Heart defect, choanal Atresia, Retardation of Growth, Ear abnormalities)syndrome[3].ODC may coexist with coloboma of other ocular structures like the iris, ciliary body, zonules, lens, choroid, and retina[4].Visual impairment in patients with coloboma may be due to involvement of the fovea, optic nerve, and maculopapular bundle or complications like retinal detachment, choroidal neovascular membrane, and cataract formations. Refractive errors have been associated with coloboma and can cause amblyopia [4-5]. Optic disc and Retinochoroidal colobomas are rare in Nigeria. Few cases have been reported in Southwest and North central parts of the country [6-7-8]. The case that was reported in the North Central was that of a 16-year-old male student with unilateral ODC and associated myopia. There was a history of childhood seizure disorders [7]. To the best of our knowledge, this is the first case of ODC reported from the Northwestern part of Nigeria.

Case Report:

A 13-year-old female was referred from paediatric outpatient department to the ophthalmology department of the same tertiary hospital, on account of difficulty seeing distant objects for 3 months duration, with associated ocular pains, tearing, and headache while reading. No history of ocular deviation or nystagmus. She had never worn spectacles. No previous history of systemic illnesses or trauma. The patient is the first child of five children from a monogamous non-consanguineous family. Pregnancy, Labour, and Antenatal period were uneventful. Developmental milestones were optimal for age. No family history of ocular diseases or congenital anomalies. No history of similar problems among siblings or other relatives.

General and systemic examinations were normal. Ocular examination revealed a best corrected visual acuity of 6/5 and 6/6 in the right and left eyes respectively. Cycloplegic refraction with tropicamide and cyclopentolate was OD: +300DS and OS: +325DS. Anterior segment examination with a Slit lamp biomicroscope was normal, with no pupillary abnormality. Extraocular muscle motility was full in all directions of gaze, with no diplopia, and no nystagmus. Intraocular pressure was 12mmHg and 13mmHg in the right and left eye respectively using Goldmann applanation tonometry.

A dilated fundoscopy of the right eye with +78DS lens revealed an enlarged optic disc with a white bowl-shaped excavation occupying the whole inferior aspect of the disc, absent inferior neuroretinal rim, and intact superonasal rim, macula was normal, and the retina was flat. Inferior and slightly temporal to the optic disc was an area of well-defined oval retinochoroidal defect, clearly separated from the disc, with absent retinal pigment epithelium, anomalous vessels within the defect, and an area of choroidal sclerosis, appearing as greyish rim superiorly (Fig.1). The left eye revealed an enlarged optic disc with bowl-shaped excavation and small area of superior neuroretinal rim and pigmentation surrounding the disc margins. The macula was normal, and the retina was flat (Fig.2). Patients were prescribed the spectacles. Verbal and written assent and consent were obtained from the Patient and the father respectively.

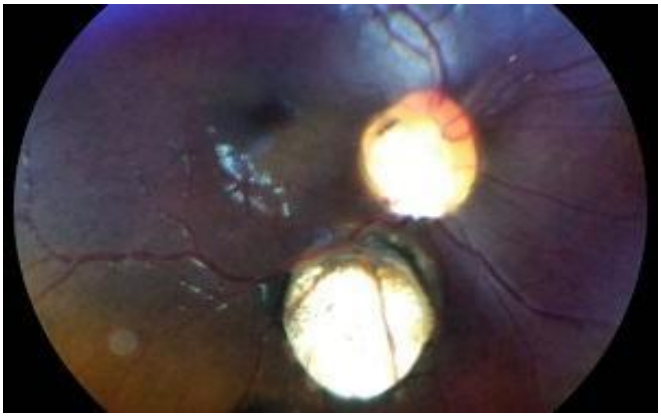

Figure 1: Right eye: Optic disc and Retinochoroidal colobomas

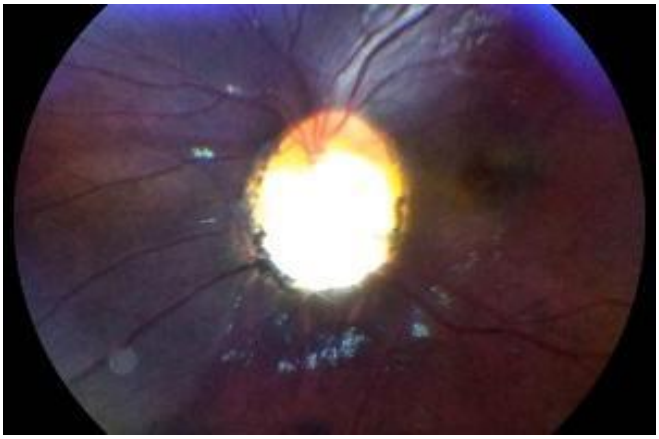

Figure 2: Left eye: Optic disc coloboma

Discussions

Optic disc coloboma is a rare congenital anomaly that occurs as a result of partial or complete defective closure of the embryonic fissure and can be associated with other congenital anomalies [2]. It usually involves the inferior part of the optic disc with the absence of an inferior neuroretinal rim and can be either unilateral or bilateral [3]. Optic disc coloboma (ODC) can occur as an isolated case or may coexist with colobomas of the other parts of the eye like retinochoroidal, iris, ciliary body, zonules, or lens colobomas [4]. Our patient has bilateral ODC coexisting with right eye retinochoroidal coloboma. Cases of bilateral optic discs coloboma with chorioretinal extension have been reported, and most of the bilateral cases have been associated with systemic abnormalities such as CHARGE (Colobomas, Heart defect, choanal Atresia, Retardation of Growth, Ear abnormalities) syndrome, Dandy-walker syndrome, Goldenhar syndrome, Aicardi syndrome, and renal coloboma syndrome [9-10]. Our case has no systemic associations or abnormalities. The majority of cases of ODC are sporadic, occasionally autosomal dominant inheritance may occur, and less frequently autosomal recessive or x-linked inheritance [11]. It is pertinent that all patients with ODC undergo chromosomal analysis. Our patient could not do this because it was not available in our centre. The severity of visual impairment in ocular coloboma depends on the involvement of the fovea and optic disc and associated complications like Retinal detachment, choroidal neovascular membrane formation, cataract, and amblyopia due to uncorrected refractive errors [1]. Rhegmatogenous retinal detachment can occur in a patient with ODC and is usually a result of retinal breaks close to the margin of the coloboma and the escaping of fluid into the subretinal space [8]. Patients with retinochoroidal coloboma can develop non-rhegmatogenous retinal detachment especially when coexistent with ODC[8]. Choroidal neovascular membrane has also been associated with poor vision, especially in the case of retinochoroidal coloboma. This is a result of the distortion of the retinal pigment epithelium at the margin of the coloboma and the growth of the blood vessels from the choroid into the

subretinal space [6]. None of these complications were present in our patient, but the patient needs to be on lifelong follow-up because of the high risk of developing retinal detachment and choroidal neovascular membrane. Refractive errors usually accompany coloboma and can cause anisometropic amblyopia, especially in unilateral cases. The role of amblyopia in bilateral coloboma has not been adequately evaluated [12], although it has been reported in some patients [5]. Myopia is more frequently associated with ocular coloboma although other types of refractive errors have been reported [5-12]. Our patient had hypermetropia with the best corrected visual acuity of 6/5 and 6/6 using +3.00DS and +3.25DS in the right and left eye respectively. Other ocular disorders like strabismus and nystagmus that have been reported in patients with coloboma [2] were not found in our patients.

ODC should be differentiated from glaucomatous optic neuropathy because it was misdiagnosed as a case of glaucoma [13]. In ODC, the Optic disc is whitish, very large, with excavation located inferiorly and the absence of an inferior neuroretinal rim and usually thin superior rim. Other differential diagnoses are Megalopapilla, Morning glory disc anomaly, and Optic disc pit.

The reported case should be subjected to a lifelong follow-up so that signs of possible complications like retinal detachment and choroidal neovascular membrane formation can be detected for early interventions.

Conclusion:

ODC is rare in Nigeria and the rest of the world. This case was reported because of its rare occurrence and to highlight the importance of binocular dilated fundoscopy and critical disc assessment in any child with unilateral and, or bilateral visual impairment. This will help in identifying this rare asymptomatic condition early so that appropriate interventions can be instituted to avoid the occurrence of complications. It also highlights the importance of a multispecialty approach to the management of this condition because of its frequent multi-systemic involvement and the possibility of associated genetic and inherited syndromes. It is pertinent for physicians and general practitioners to be aware of the affectation of the eye by these syndromes for prompt referral to ophthalmologists.

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