Case Report

Optical Coherence Tomographic Features in a Case of Bilateral Macular Coloboma with Strabismus

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Abstract

Purpose: To report the optical coherence tomography (OCT) findings in a patient with bilateral macular coloboma with strabismus.

Methods: A 21-year-old male presented with macular coloboma in both eyes. Fundus photography, fundus fluorescence angiography (FFA) and OCT were performed.

Results: Color fundus photography showed a sharply-demarcated, round macular defect, approximately 4×4 disc diameters with bare sclera at the base and pigment clumping in both eyes. FFA showed hypofluorescence at the macula corresponding to the size of the lesion bilaterally. OCT revealed a crater-like depression at the macula, demonstrating atrophic neurosensory retina, and an absence of retinal pigment epithelium (RPE) and choroid in the lesion.

Conclusion: OCT can be a beneficial tool to confirm the diagnosis of macular coloboma. (***Eye Science 2011;26:244-246***)

keywords: optical coherence tomography; macular coloboma

Macular coloboma is characterized by a sharply defined, rather large defect in the central area of the fundus that is oval or round and coarsely pigmented. The condition is often bilateral and familial and has been associated with ocular or systemic abnormalities and may result from either a developmental abnormality or intra-uterine inflammation[1,2]. Consideration of the differential diagnosis is essential, because many diseases mimic a coloboma in the macula; optical coherence tomography (OCT) may be a beneficial tool to make a diagnosis of macular coloboma, because it can show a subtle defect of the neurosensory retina and choroid at a high resolution[3,4]. This study examines a case of bilateral macular coloboma with strabismus and its OCT characteristics.

Case report

A 21-year-old patient was referred to our clinic with poor vision in both eyes. The patient had suffered from extropia in the left eye since early childhood. His family history for ocular and systemic diseases was unremarkable. Ophthalmic examination showed that visual acuity of both eyes was CF/20 cm; best-corrected visual acuity was 0.06 in the right eye with $-1.00+3.00\times90$ and 0.02 in the left eye with $-2.00+3.00\times90$. Intraocular pressure (IOP) on non-contact tonometry was 17 mmHg and 16mmHg in the right and left eye, respectively. The anterior segments examination was unremarkable in both eyes.

The left eye showed exotropia of around 20°(Figure 1). No foveal fixation was presented in either eye. Horizontal nystagmus appeared when looking at the left side and was relieved when looking at the right side. Binocular movement was not limited. The fundus examination showed bilateral small optic discs, of oval shape and reddish hue. There was bilateral pigmented macular coloboma around 4×4 disc diameter in size with a clear margin. The coloboma was surrounded by a dark rim of the retinal pigmented epithelium (RPE). A few large choroidal vessels were visible against the white scleral background of

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the defect. The retinal vessels were narrow and crossed the defect (Figures 2A; 3A). FFA showed hyper-fluorescence at the macula, corresponding to the size of the lesion in both eyes (Figures 2B; 3B). OCT (3D OCT-1000, TOPCON, Japan) shows that the neurosensory retina is abnormally atrophied and there is an absence of RPE and the choroid coloboma in the lesion (Figures 2C; 3C). Based on clinical examination, FFA and OCT imaging, the patient was diagnosed as having bilateral macular coloboma, sensory strabismus, and ametropia.

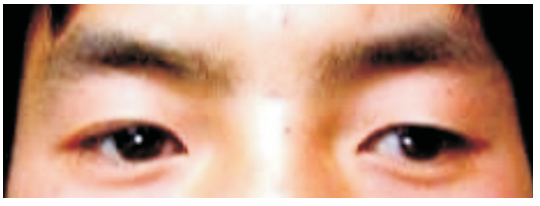

Figure 1 The patient's left eye appeared to show exotropia of around 20°.

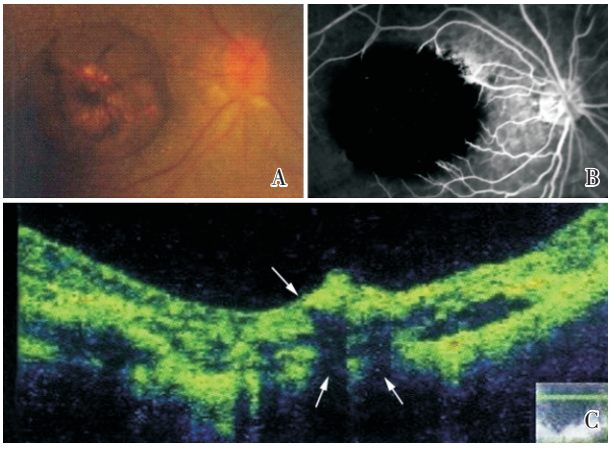

Figure 2 (A) Fundus photograph showed a sharply demarcated, round-shaped defect, approximately 4×4 disc diameter and a pigment clump was found at the macula in the right eye. (B) FFA showed a darkly hypofluorescent area in the macula of the right eye, corresponding to the size of the lesion. (C) An OCT line scan through the lesion of the right eye showed atrophy of the neurosensory retina and the RPE and choroid are absent.

Discussion

Macular coloboma is thought to result from intra-uterine inflammation or an abnormality of development. Most cases are now recognized as being owing to an intra-uterine infection with Toxoplasma gondii. A developmental abnormality seems to be the cause

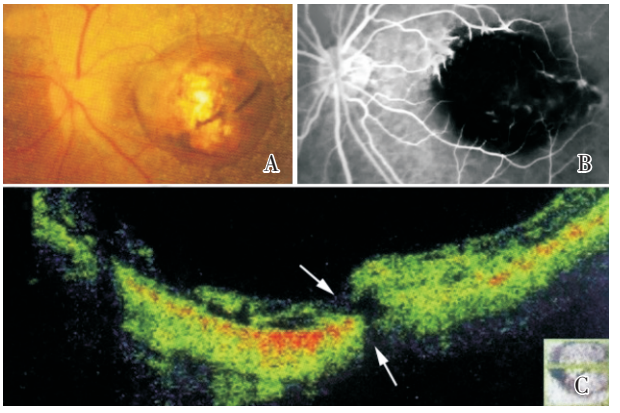

Figure 3 (A) Fundus photograph of the left eye showed a similar lesion at the macula with a thickened margin. (B) FFA showed hypofluorescence with minimal leakage and relative hyperfluorescence at the edge of the coloboma in the macula of the left eye. (C) A horizontal section scan on OCT of the left macula showed a full thickness defect involving the foveal, RPE and the choroid.

in those patients with a hereditary or family origin, and those with other ocular or systemic abnormalities[1,4,5].

Mann divided macular coloboma into three types on the basis of the ophthalmoscopic appearance. The first type is the pigmented macular coloboma. The macula was covered with dense irregularly arranged masses of pigment. The capillary layer of choroid was absent and the sclera and a few large choroidal vessels can be seen deep in the pigment. There was no ectasia of the base. The overlying retinal vessels were normal.

The second type is the non-pigmented coloboma. It is a round or oval patch with a punched-out appearance, with ectatic sclera at the base. The base is always slightly hollowed out and may be measurably ectatic. Around the edge of the white base is a band of pigment. A few large choroidal vessels were attenuated. Sometimes, the retinal vessels stop abruptly at the margin of the defect.

The third type is the macular coloboma associated with abnormality of blood vessels. This type is very difficult to see. The vascular anomaly may apparently take the form either of an abnormal anastomosis of vessels in the defect with choroidal or retinal vessels, or of a vessel passing forward from it into the vitreous.

Several previous studies have described patients

with macular coloboma combined with Down Syndrome; adult vitelliform dystrophy; retinitis pigmentosa; Leber's congenital amaurosis, etc[1,6,7,8]. Aziz et al[3] reported a three-year-old girl with Down syndrome with intraoperative OCT confirmation; the patient was diagnosed as having bilateral macular coloboma. Panagiotidis et al[1] described a case of macular coloboma in a patient with adult-onset vitelliform dystrophy in the fellow eye. OCT revealed a full thickness defect involving the foveal retina, RPE, Bruchs membrane and choroids; the defect in the Bruchs membrane was the smallest, and the diameter of the choroidal defect was the largest, exceeding the retinal defect. There was a sharply shelving edge with posterior bowing and increased reflectivity corresponding to the sclera.

Oh et al[4] reported the OCT findings in a patient with unilateral macular coloboma. OCT shows that the neurosensory retina is abnormally atrophied, and RPE and choroid are absent in the lesion. A thin membranous structure of atrophied neurosensory retina is outlining the bare sclera and large choroidal vessels. Hussain et al[6] reported a case of unilateral macular coloboma with macular dystrophy and its OCT characteristics. An OCT line scan through the lesion showed an absence of foveal contour with a bowl-shaped deformity. A sharp shelving edge could be seen, with posterior bowing high reflectivity suggestive of the sclera.

In our study, we reported an OCT finding in a case of macular coloboma with strabismus, where we found an abnormal thinning of the neurosensory retina, an absence of RPE and choroidal tissue, leaving bared sclera in the lesion. In our patient, the changes were typical, both clinically and on FFA; OCT confirmed the diagnosis of macular coloboma. The diagnosis of macular coloboma can potentially be made solely on the basis of clinical findings; however, OCT imaging is a useful objective way of confirming the diagnosis.

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