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Case Report

Bilateral Congenital Retinal Macrovessel with Bilateral Optic Disc Oedema: A Case Report

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Abstract

We aim to report a ten-year-old male presenting with a trivial symptom of headache and photophobia. in turn, revealing to be bilateral congenital retinal macrovessels with bilateral optic disc oedema secondary to cerebral venous malformation. Pediatric and neurology opinion was taken, but it came out to be unremarkable. Unaided visual acuity on Snellen's chart was 6/60 in the right eye, which did not improve on pinhole and refraction and 6/6 in the left eye. Fundus examination of the right eye revealed clear media with poorly defined disc margins at the superonasal and nasal borders, with gliosis noted at the superotemporal borders of the optic disc, and the left eye revealed blurred nasal disc margins with dilated and tortuous vascular arcades. In the macular region, there were aberrant dilated vessels reaching inside the foveal avascular zone but not crossing the horizontal raphe in both eyes. Neurology and paediatrics opinions ruled out any other comorbidity, and a diagnosis of congenital retinal macroaneurysm was made.

Keywords: *Congenital Macroaneurysm; Disc Oedema*

Introduction

Congenital retinal macrovessels (CRM) are a congenital vascular abnormality of the retina first described by Mauthner in 1864, in which aberrant vessels, more commonly veins, cross the horizontal raphe at or over the macular region [1].

During the embryonic period, the mesenchymal cells accumulate towards the base of the hyaloid artery at the 15th to 16th week of gestation, which in turn form the retinal vascular arcades. The high oxygen demand of retinal development usually causes obliteration of primitive vessels supplying the macular and foveal region during gestation, resulting in the formation of the foveal avascular zone [2]. Ischemic insult during the gestational period can result in persistence of these primitive vessels in infancy.

These vascular malformations are mostly unilateral and arise more commonly from venous than arterial circulation [3]. Although mostly asymptomatic, these can be associated with retinal pathologies such as macular haemorrhage, macular telangiectasia, retinal vascular occlusion, and macular oedema [4,5]. Recently, a common association with cerebral vessel malformation has been proposed as well [6].

We aim to report one such case of a patient presenting with a trivial symptom of headache and photophobia in turn revealing to be bilateral CRM with bilateral optic disc oedema secondary to cerebral venous malformation.

Case Report

A 10-year-old male patient presented with complaints of headache for two years, which was mild in nature, diffuse, intermittent and poorly localised. It was associated with photophobia and glare when looking at bright sources of light. Unaided visual acuity on Snellen's chart was 6/60 in the right eye, which did not improve on pinhole and refraction and 6/6 in the left eye.

Anterior segment on slit lamp examination in both eyes showed a clear cornea, quiescent anterior chamber and a clear crystalline lens with normal pupillary reflexes. Vitreous was clear, and no evidence of inflammation was noted. Intraocular pressures using applanation tonometry were 14 mm of Hg and 18 mm of Hg in the right and left eyes, respectively.

Fundus examination of the right eye revealed clear media with poorly defined disc margins at the superonasal and nasal borders, with gliosis noted at the superotemporal borders of the optic disc. Spontaneous venous pulsations were not noted at the time of presentation. Vessel tortuosity of the vascular arcades was noted. In the macular region, there was a dilated and tortuous aberrant vein crossing the horizontal raphe and draining directly into the central retinal vein (Figure 1). Foveal reflex was dull in the right eye, but no intraretinal haemorrhage and intraretinal cyst were noted. Peripheral retinal examination was within normal limits and did not reveal any similar malformations.

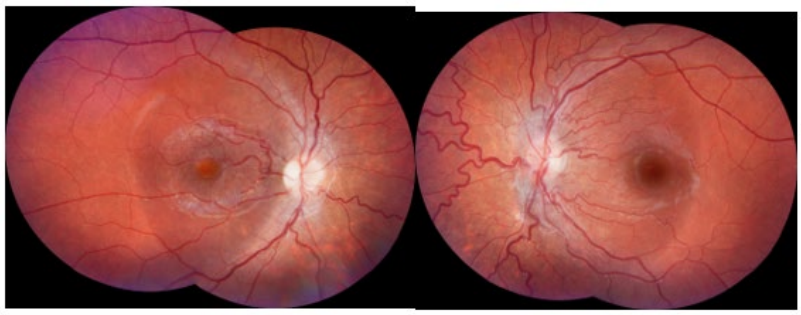


Figure 1a and 1b: *Showing fundus pictures with a dilated and tortuous aberrant vein crossing the horizontal raphe in the right and left eyes, respectively.*

Fundus examination of the left eye again revealed blurred nasal disc margins with dilated and tortuous vascular arcades. Spontaneous venous pulsation was not noted in the left eye as well. In the macular region, there were aberrant dilated vessels reaching inside the foveal avascular zone but not crossing the horizontal raphe in the left eye. There were no retinal haemorrhages and intraretinal cysts visible clinically.

Spectral domain optical coherence tomography of the macula, using a raster scan, revealed an altered foveal contour, with large blood vessels traversing the nerve fibre layer and distortion of the Internal limiting membrane. However, the remaining inner and outer retinal layers were intact with no disruption noted. Choroidal thickness was normal with no abnormality noted. Macula raster scan of the left eye revealed a normal vitreo-macular interface and normal foveal and ILM contours, with aberrant dilated vessels extending into the foveal avascular zone, but these were located near the outer plexiform layer of the retina. The rest of the retinal layers were normal with no disruption. The underlying choroid was normal with normal choroidal thickness (Figure 2a and 2b). Spectral domain optical coherence tomography of the optic nerve head of the right eye revealed increased thickness of the nasal margins of the optic disc with surrounding thickening of the nerve fibre layer, more prominent towards the nasal retina, suggesting gliosis (Figure 3a and 3b).

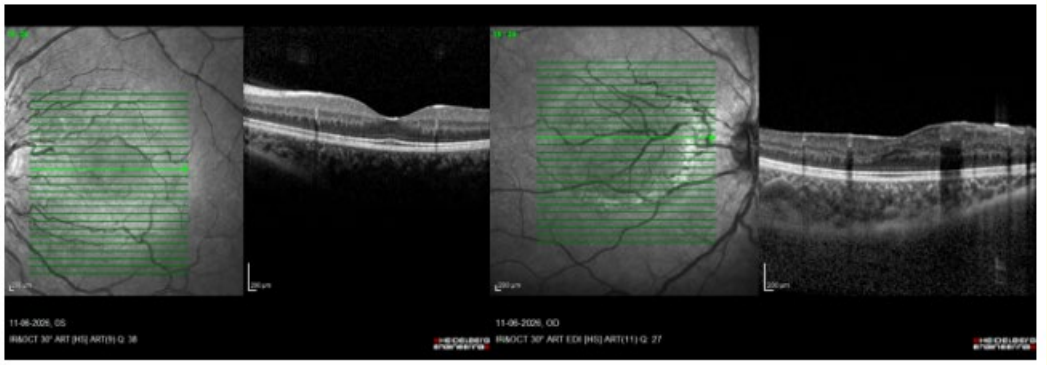


Figure 2a and 2b: *Optical coherence tomography Macula raster scan of the right eye revealed a normal vitreo-macular interface and normal foveal and ILM contour. Left eye shows aberrantly dilated vessels extending into the foveal avascular zone.*

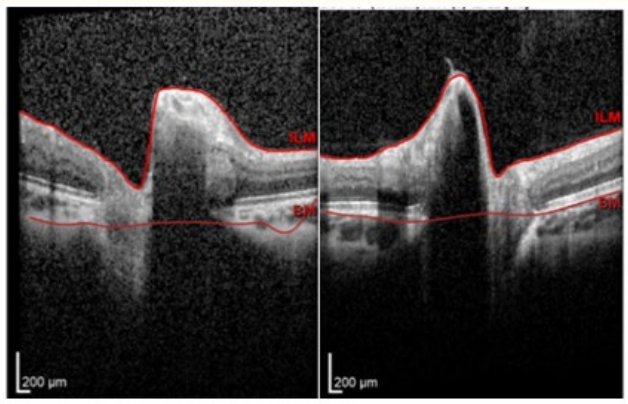


Figure 3a and 3b: *Optical coherence tomography optic nerve head scan of the right eye revealed disc oedema in both eyes.*

The patient was diagnosed with bilateral CRM with optic disc oedema in both eyes. The patient was counselled on the prognosis and the progression of the disease and was advised to get a contrast-enhanced magnetic resonance image of the cerebral circulation to rule out the cause of raised intracranial pressure and any cerebral vascular malformation. Neurology opinion was taken, which was unremarkable.

Discussion

CRM is a rare congenital vascular malformation with an incidence of 1 in 200000, characterized by an aberrant dilated tortuous vessel, more commonly a vein crossing the horizontal raphe [7]. Almost always an incidental finding during routine ophthalmic examination, a few present with complications like macular oedema, etc. Bilateral CRM presenting with unilateral decreased vision and bilateral optic disc oedema in our case, is a rare presentation.

Pathogenesis is postulated to be the result of ischemic insult at the gestational age of 15^{th} - 16^{th} week of gestation, disrupting the normal obliteration of mesenchymal primitive retinal vasculature, resulting in abnormal persistence beyond the period of infancy [2]. These abnormal vessels become anomalous in course and calibre and result in changes in the surrounding normal vasculature and retinal layers.

CRM usually present as an asymptomatic retinal condition, not usually affecting visual acuity and daily activities. Rarely can cause secondary macular oedema, central serous chorioretinopathy, and foveal cyst, which can present as metamorphopsia and blurred vision. Glare and photophobia, although nonspecific and uncommon for retinal conditions, were the presenting complaints of our patient, thought to arise due to the anomalous vessels in and around the foveal avascular zones, resulting in angioscotomas.

Recent advances in multimodal imaging have also improved the prognostication of patients with CRM. Fundus fluorescein angiography shows early filling of the anomalous vessel with no leaks, generally, unless associated with macular oedema or Choroidal neovascular membrane. Optical coherence tomography helps in identifying all these secondary complications and can plan the treatment accordingly. OCT angiography is the newest non-invasive modality that can help distinguish the involvement of superficial, middle and deep venous plexuses as present in outpatients and can predict early secondary complications.

Optic disc oedema is not a direct outcome of CRM, but rather a postulated consequence of its pathogenesis as a systemic association with cerebral vascular malformation, resulting in raised intracranial pressure. Our patient had bilateral optic disc oedema, which was resolving, as revealed by surrounding glial tissue suggestive of chronicity, hinting at a benign developmental anomaly.

Conclusion

Bilateral CRM represent an exceptionally rare developmental retinal vascular anomaly. The coexistence of bilateral optic disc oedema and cerebral venous malformation further underscores the possibility of a shared developmental vascular origin involving both the retina and the cerebral circulation. Recognition of these associations is important, as affected patients may benefit from appropriate neuroimaging and multidisciplinary evaluation. Reporting additional cases will improve understanding of the natural history, systemic associations, and optimal management of this rare condition.

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