Iridofundal Coloboma Associated with Vitreous Haemorrhage and an Intraocular Mass: A Case Report

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Introduction: Iridofundal colobomas, being developmental defects, are known to be accompanied by several ocular anomalies but an association with vitreous hemorrhage and an intraocular mass has not been reported earlier.

Case: We report a case of an 18 years old subject having iridofundal coloboma in both eyes and an association of vitreous hemorrhage and an intraocular mass in the left eye. The diagnosis was confirmed with a detailed ocular examination, fundus fluorescein angiography, swept-source optical coherence tomography and ultrasonography. The laser barrage of coloboma caused a regression in the size of the intraocular mass and prevented recurrence of vitreous hemorrhage over one and half years of follow-up.

Conclusion: The source of vitreous hemorrhage in this subject is unclear though some anomalous vessels in relation to the supero-nasal mass, optic disc and coloboma of the left eye might have caused it. The laser barrage of coloboma obscured these anomalous vessels resulting in the regression of the intraocular mass.

Key words: Intraocular mass, Iridofundal coloboma, Ocular anomalies, Persistent fetal vasculature, Vitreous haemorrhage.

INTRODUCTION

The presence of retino-choroidal coloboma has been found to be associated with ocular anomalies like persistent pupillary membranes (Gupta et al, 2020), persistent fetal vasculature (Takkar et al, 2016; Gupta et al, 2020), lens coloboma (Ward, 2012), cataract and subluxation of lens (Ward, 2012; Takkar et al, 2016), microphthalmos (Yagev et al, 2000), and retrobulbar cyst (Yagev et al, 2000). We report an unique instance of a subject having anirido-fundal-coloboma associated with an intraocular mass who presented with vitreous haemorrhage. Such an association has not been described earlier. This case report emphasizes

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that varied ocular anomalies may be present in patients having irido-fundal colobomas and thus detailed examination is required.

CASE

An eighteen-year subject presented with diminution of vision and outward deviation of left eye (LE) since childhood. She had been prescribed glasses for both eyes elsewhere but these did not lead to much improvement. So she was referred to a higher centre. She presented to our hospital with a complaint of a notable decrease in vision of LE since the past 15 days. There was no history of trauma or any underlying systemic disease.

She had best-corrected visual acuity (BCVA) of logMar +0.2 in right eye (RE) and logMar +1.5 with accurate projection of rays in LE.The ocular examination showed infero-nasal iris coloboma in BE. The LE had microphthalmia with 15 degree exodeviation and it did not take fixation on performing alternate cover test. However, there was no nystagmus or restriction of ocular movements. The RE had tilted optic disc with C:D of 0.6:1 along with Type III fundal coloboma sparing fovea. The LE had vitreous haemorrhage.

The intraocular pressure measured 18.9 and 22.4 mmHg; the axial length was 22.73 mm and 22.72 mm; pachymetry was 589 microns and 603 microns; corneal diameter was 10 mm x 11mmand 8 mm x 9 mm in vertical and horizontal meridian; and keratometry was 42.50 x 44 at 135 degree and 41.75 x 43.25 at 10 degree in RE and LE respectively. The patient did not have ocular abnormalities like cataract and heterochromia iridum.

The patient was advised to stay in a propped up position with tablet vitamin C 500 mg two times a day. The ultrasonography B scan of LEshowed an inferior fundal coloboma along with mild to moderate echoes in vitreous cavity and round hyperechoic lesion in supero-nasal quadrant. There was no retinal or choroidal detachment. Two months later, following resolution of vitreous haemorrhage, the BCVA in LE improved to logmar +1.3 with an accurate projection of rays. The fundus examination showed a type II coloboma along with a supero-nasal mass and a vessel extending from the optic disc onto the superonasal mass. The fundus fluorescein angiography (FFA) showed kinked, tortuous vessels and reduplication of veins in the region of superonasal mass in LE. There was staining of margins of mass as well as coloboma which increased in intensity in the later phase. There was a mild leak at the optic disc in the late phase. The retinal vessels extended from the optic disc over to the superonasal mass. **(Figure 1a and 1b)**. The magnetic resonance imaging of head and orbit showed an oblong ocular globe with loss of normal rounded configuration and conical shape of posterior ocular wall suggestive of fundal coloboma with small intraocular mass.

The optical coherence tomography (OCT) showed central macular thickness of 230 microns in RE while it could not be measured in LE as fovea was involved in coloboma.

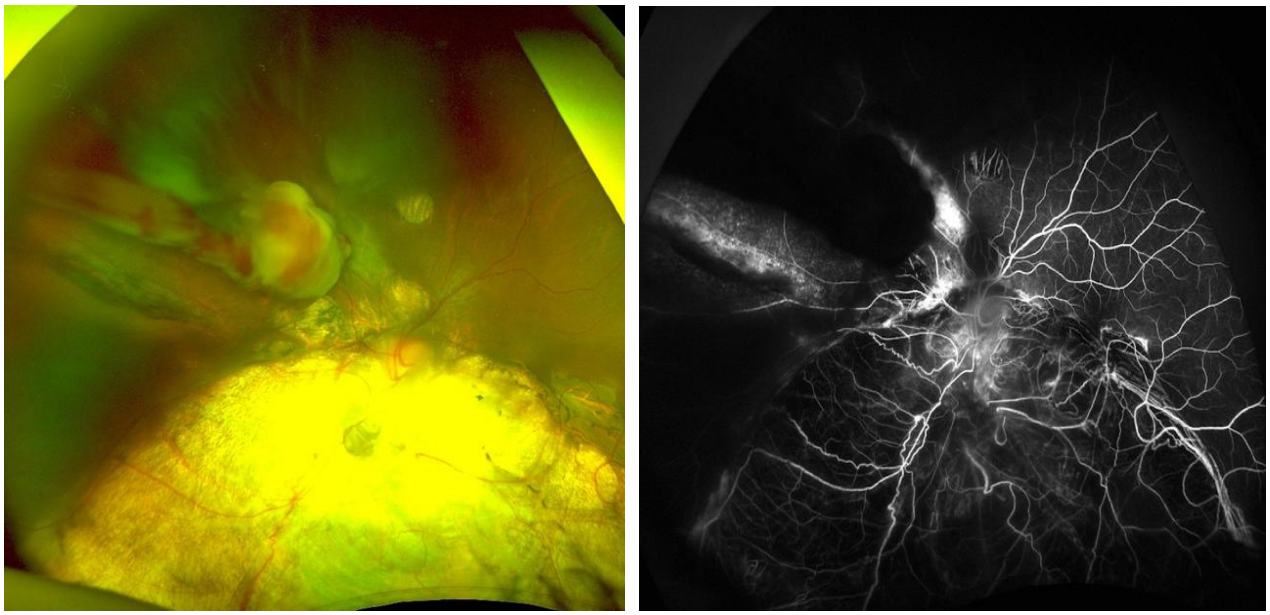

Figure 1a and 1b: Colour fundus photo and fundus fluorescein angiography of LE before laser; arrow points to vessel extending from optic disc onto the superonasal mass.

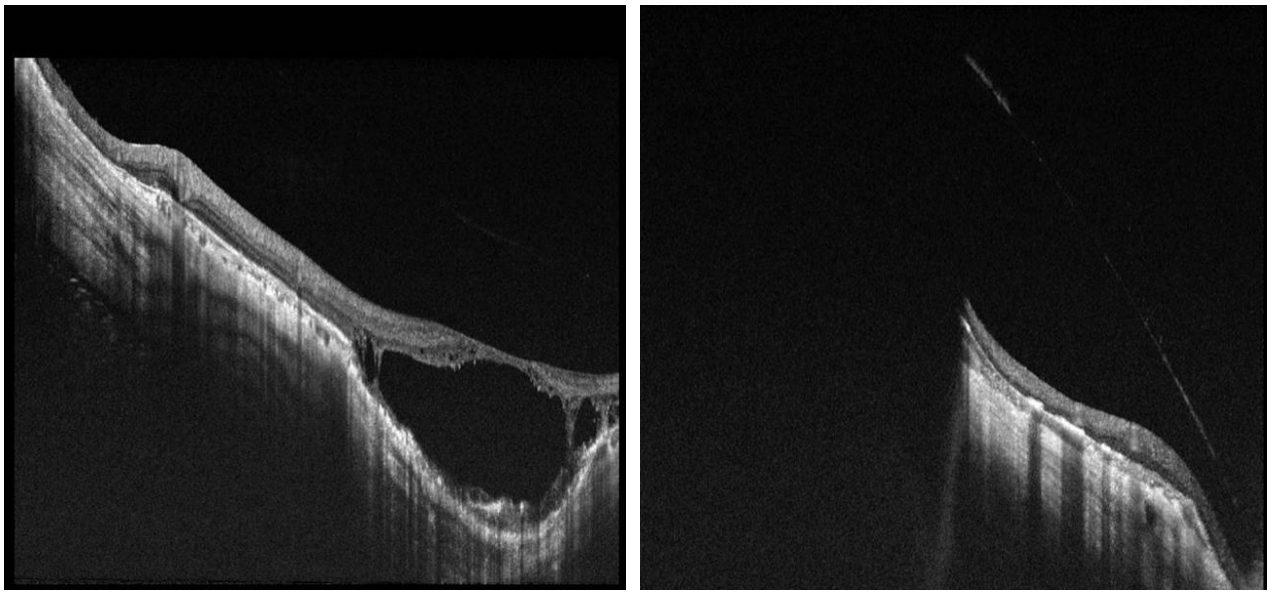

Figure 2a and 2b: Swept source optical coherence tomography scan; arrows showing intercalary membrane detachment over coloboma and back shadowing by supero-nasal mass.

The swept source OCT (SS OCT) scan of LE showed an intercalary membrane detachment in the region of coloboma and back shadowing on the retino-choroidal-scleral tissue by the supero-nasal mass **(Figure 2a and 2b)**. The RE SSOCT scan showed choroidal cavitations and an intercalary membrane detachment in the coloboma region. Amsler's grid showed normal

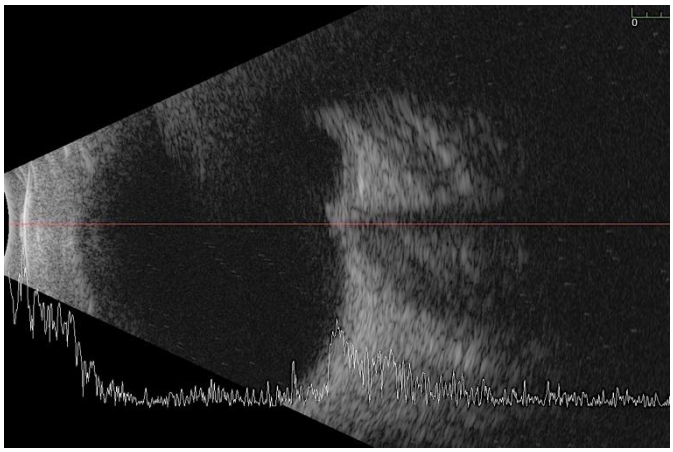

Figure 3: Ultrasound B scan of LE showing an intraocular mass located superiorly.

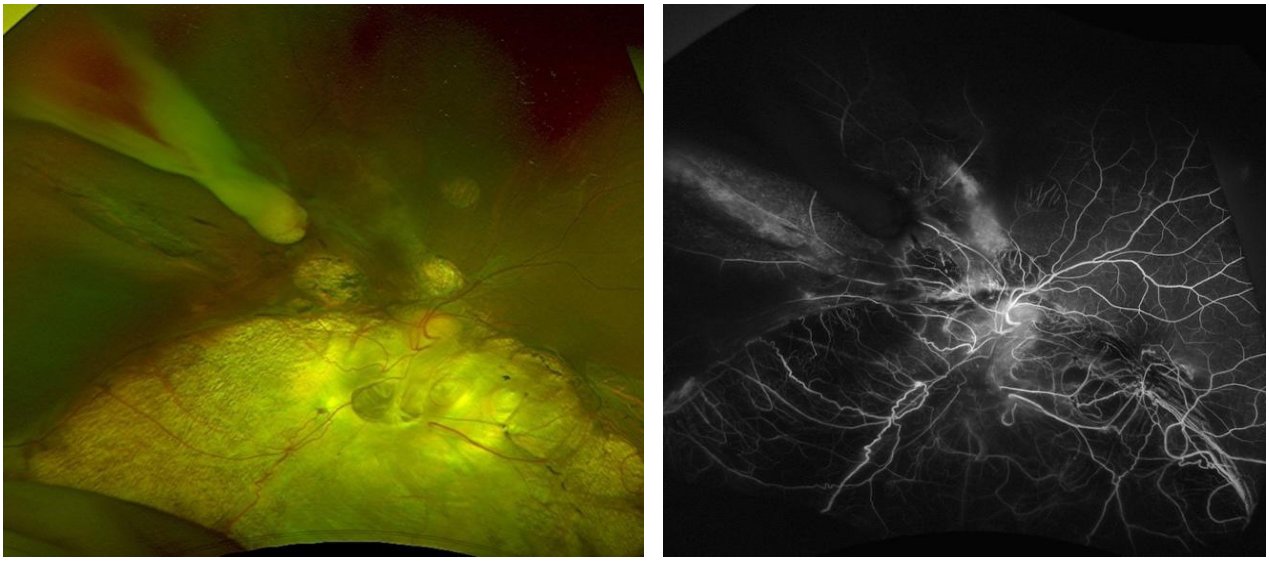

Figure 4a and 4b: Colour fundus photo and fundus fluorescein angiography of LE after laser; arrow points to partially obscured vessel extending from optic disc onto the superonasal mass.

plot in RE and central scotoma in the LE.

The axial ultrasound B scan of LE showed a hypoechoic mass located supero-nasally inside the eyeball showing mild spikes all over the mass on the corresponding A scan **(Figure 3).**

The ocular adnexa was normal and there was no other congenital malformation or neurological abnormality.The antenatal and birth history was unremarkable and we did not notice any ocular or systemic abnormality in parents and her siblings including two sisters and one brother.

The laser barrage of the coloboma caused regression of the mass as well as reduction in leak at the optic disc **(Figure 4a and 4b)**. The examination of fundus and fluorescein angiography photos showed that laser barrage of coloboma on the nasal side partially obscured

blood vessels extending from the optic disc onto the superonasal mass. Probably this abnormal blood vessel had caused vitreous haemorrhage earlier.

There was no recurrence of vitreous haemorrhage over one and half years of follow up.

DISCUSSION

The hyaloid artery is a branch of primitive dorsal ophthalmic artery which originates from the internal carotid artery.It enters the optic cup inferiorly to form the hyaloid vascular system in the fourth to fifth week of gestation. The hyaloid artery gives rise to vasa hyaloidea propria for vitreous cavity and capillary network called tunica vasculosa lentis for posterior surface of lens which connects to choroidal vasculature through irido hyaloid vessels (Goldberg, 1997). During embryonic development, the hyaloid vasculature also supplies oxygen and nutrients to the developing inner retina (Calvo et al, 2013).

Persistent fetal vasculature (PFV), previously known as persistent hyperplastic primary vitreous (PHPV), is a spectrum of disease resulting from failure of normal programmed involution of hyaloid vasculature. PFV typically presents as an idiopathic, unilateral congenital malformation and includes partial or possibly total persistence of all of the fetal intraocular vasculature after birth, rather than just the post-lental vessels. The nomenclature change from PHPV to PFV implies the inclusion of pathology throughout this system: the vasculature, iris, lens, vitreous, retina, macula and optic nerve (Goldberg, 1997).While anterior subtype of PFV is indicated by presence of retrolental opacity, elongated ciliary processes or cataract; posterior PFV includes presence of an elevated vitreous membrane from the optic nerve, retinal fold or dysplasia, retinal detachment or optic nerve hypoplasia (Hunt et al, 2005). Pollard (1997) found that 3 percent of full-term children have some clinically detectable remnants of the hyaloid system, which may lead to spontaneous bleeding, if these vascular remnants are still perfused.

The association of choroidal coloboma with persistent fetal vasculature (PFV) may be due to the fact that the embryonic fissure normally closes by the 6th week of fetal life and it is around this time that the secondary vitreous begins to form (Barishak, 1992). An insult during this period can cause concurrent appearance of both anomalies (Takkaretal, 2016). A “coloboma like iris defect” and persistent fetal vasculature have been identified in a patient with PHACE syndrome (Lasky et al, 2004).Typically, the irido-fundal coloboma and the PFV present in the inferior or the infero-nasal quadrant of eyes which correspond to the site of embryonic fissure. However, atypical presentations of colobomas in different quadrants/sites have also been infrequently noticed; and this atypical location has been attributed to the presence of an accessory embryonic fissure (Gupta et al, 2020). In our subject, a failure of closure of the

infero-nasal embryonic fissure caused Type II coloboma in LE. Either an accessory embryonic fissure at a supero-nasal location in LE caused migration/retention of some tissue along with an abnormal vasculature intraocularly. Or that posterior subtype of PF manifested in the form of an intraocular mass/retinal fold and a persistent vascular remnant. The abnormal vasculature most probably caused spontaneous vitreous hemorrhage.

Persistent fetal vasculature (hyaloid vessels) is usually associated with cataract, microphthalmia and developmental anomalies of retina and optic nerve - all of which hamper vision significantly (MacDonald, 1965). Our patient had diminished vision, microphthalmos, increased corneal thickness and raised intraocular pressure and she presented with vitreous hemorrhage. The examination of fundus and fluorescein angiography photos showed a blood vessel extending from the optic disc included within Type II coloboma, onto the superonasal mass. There was a mild leak at the optic disc in the late phase of FFA. Probably this abnormal blood vessel had caused vitreous haemorrhage earlier. The laser barrage of coloboma on the nasal side partially obscured this vessel, causing a regression in size of this mass.

While the hyaloid artery enters from the inferior side of the optic cup, the nature of vessels located in the supero-nasal mass causing vitreous haemorrhage is not clear. Further studies on embryonic development of the eye specifically with respect to location and closure of embryonic fissures including accessory embryonic fissures and course of migration of the hyaloid artery and persistence of fetal vasculature in different manners are recommended.

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