Retinitis pigmentosa with bilateral irido-fundal coloboma

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A 40-year-old male, presented with gradual painless progressive loss of vision in both the eyes more in right since 1 year. Born out of consanguineous marriage, he gave a history of night blindness since childhood. The best-corrected visual acuity was 20/200 and 20/40 in right and left eye, respectively. Refraction showed an axial myopia of -9.00 DS/-1.50 DC at 100 degree in the right eye. The left eye showed refraction of +1.00 DC at 170 degree. Intraocular pressure with noncontact tonometer was 15 and 16 mm of Hg. Axial length was found to be 24.93 mm and 23.60 mm in right and left eye, respectively.

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Vertical and horizontal corneal diameters were 11 mm in both eyes. Keratometry values in the right eye were K vertical 43.55D, K horizontal 44.06D and in the left eye were K vertical 43.05D, K horizontal 43.49D.

Slit-lamp examination revealed a clear cornea and normal anterior chamber depth. Inferonasal iris coloboma was noted in both eyes [Fig. 1]. The lens showed nuclear sclerosis grade 2 with posterior subcapsular opacification in right and nuclear sclerosis grade 2 in the left eye according to the Lens Opacities Classification System II (LOCS II) The posterior segment showed a normal-looking disc with minimal arteriolar attenuation, boney spicules distributed from the mid periphery throughout the retina in both eyes. Chorio-retinal coloboma was type 3 (separated from the optic disc by a clear area of the retina) in the right eye [Fig. 2] and type 7 (extreme peripheral coloboma) in the left eye [Fig. 3] according to Ida Manns classification. B scan ultrasonography showed an excavated lesion in the inferior aspect suggestive of fundus coloboma in the right eye. In the left eye the coloboma was not well demarcated on B scan ultrasonography.

Phacoemulsification with intraocular lens implantation under topical anesthesia was done in the right eye. Postoperatively at the end of 1 month best-corrected visual acuity was 20/30 in the right eye with a refraction of -0.75 DC at 80 degrees and near vision of N6 with + 3.00 DS.

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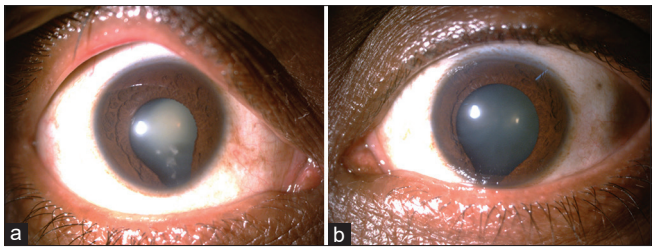

Figure 1: Anterior segment photographs with diffuse illumination (a) Right eye showing inferonasal iris coloboma with cataractous lens. (b) Left eye showing inferonasal iris coloboma

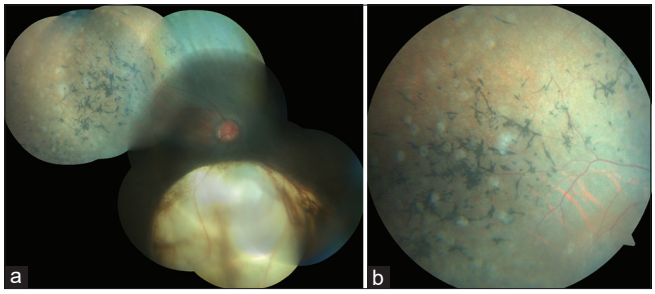

Figure 2: (a) Fundus photograph of right eye showing coloboma in the inferior quadrant sparing disc and macula with bony spicules in the mid periphery. (b) Fundus photograph right eye, superonasal quadrant showing bony spicules in the mid periphery

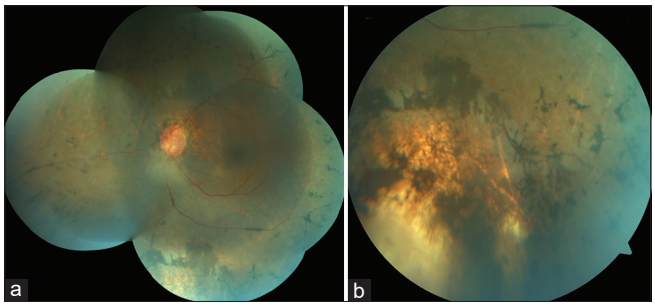

Figure 3: (a) Fundus photograph of left eye depicting bony spicules in mid periphery and extreme peripheral coloboma inferiorly. (b) Peripheral coloboma with bony spicules in inferior quadrant

Discussion

Retinitis pigmentosa is most commonly associated with posterior subcapsular cataract 53%[1] glaucoma 2.3%[2] myopia, keratoconus, vitreous detachment, and optic disc drusen. Association of macular colobomas with retinitis pigmentosa, especially autosomal dominant RP and Ushers syndrome has been extensively reported in the literature.

In 2004, phenotypically different RP with macular coloboma was reported by Parmeggiani in 3 adult siblings[3], which was stated as an autosomal dominant feature.

The association of iris and fundal coloboma with RP has been rarely reported in the literature. Kelly *et al*.[4] described the incidence, ocular findings, and systemic associations of coloboma in a population-based cohort study in the United States, reported one case with unilateral coloboma in association with RP. Population-based studies in India are scanty. Pallavi Agrawal *et al*.[5] reported a similar case of unilateral coloboma from a case in Mumbai, India in 2013.

Bilateral Irido-fundal coloboma and RP can be separate entities occurring together in this case, or it could be a rare association. Similar cases/case series need to be reported to come to a conclusive result.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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