CASE REPORT

Iris coloboma in one eye and pigment dispersion syndrome in the fellow eye

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SUMMARY

We report a case of a 43-year-old patient with coloboma of the iris, zonule, ciliary body, choroid and retina in the right eye and pigment dispersion syndrome in the left eye. Considering the hypothesis of the pigment dispersion syndrome pathogenesis in which a difference of pressures in the anterior and posterior chambers creates a posterior convexity of the iris leading to reverse pupillary block, iris touch and consequently causing pigment dispersion, we suggest that the presence of an iris coloboma, by equalising the pressures in the two chambers, prevented the onset of syndrome in that eye.

BACKGROUND

Ocular coloboma is the product of an error in the fetal fissure closure, normally occurring between the fifth and sixth weeks of gestation. It may involve the cornea, iris, zonula, ciliary body, choroid, retina and optic nerve.¹ The incidence of this syndrome is 0.7 per 10 000 live-births.¹ Other inherited autosomal dominant and recessive syndromes and non-inherited syndromes have been linked to this finding. Other ocular conditions like cataracts, lens subluxation, secondary glaucoma, amblyopia, strabismus and anisometropia have been related to this developmental defect.¹

On the other hand, the pigment dispersion syndrome is characterised by the accumulation of pigment in various structures of the anterior segment: in the corneal endothelium, where it takes the shape of a spindle, owing to aqueous humour convection currents and endothelial cell phagocytosis (Krukenberg's spindle); in the posterior lens capsule (Zentmayer's line); and in the trabecular meshwork above Schwalbe's line (Sampaolesi's line). Furthermore, iris stromal defects are evident at retroillumination.² These eyes have a posterior iris insertion with a concave peripheral iris configuration and under certain conditions (accommodation and possibly blinking) a posterior bowing of the iris occurs, which leads to contact with the zonules or anterior lens capsule causing the to slough off of the iris pigment epithelium.²

A hypothesis proposed 30 years ago exposes that a different pressure between the anterior and posterior chambers, higher in the anterior chamber, caused by a reverse pupillary block acting like a one-way valve, leads to posterior iris bowing.^{3–5}

We present a patient in whom iris coloboma seemed to protect her right eye against pigment dispersion syndrome that she presented in her left eye.

Figure 1 Total thickness iris coloboma in the right eye.

CASE PRESENTATION

A 43-year-old female patient presented to our office for a routine eye examination. No known personal or familial diseases were reported neither traumas nor surgeries in her eyes. Best spectacle corrected visual acuity in the right eye with -0.75 D was 20/40 and 20/20 in the left eye, with -0.75 $-0.25 \times 165^\circ$. Biomicroscopy revealed an inferior, total thickness iris coloboma in the right eye; inferior zonules were absent and an inferior lens cleft was evident (figure 1). The intraocular pressure was 12 mm Hg, gonioscopy grade 4 in all the quadrants other than inferior (Shaffer's classification), with inferior iris synechiae without any evidence of the trabecular meshwork in that area. Fundoscopy revealed inferior retinochoroidal coloboma (sparing the optic nerve). A central 0.55 cup-to-disc ratio, a neuroretinal ring without notching, mild peripapillary atrophy (zone α), and a healthy macula were also found (figure 2A). The left eye examination showed no iris coloboma, the presence of Krukenberg's spindle (figure 3) and pigment deposits in the lens' anterior surface. Gonioscopy was grade 3 in all the quadrants (Shaffer's classification), with Sampaolesi's line and pigmentary deposits in the anterior chamber angle. In addition a posterior iris convexity was found. Intraocular pressure was 14 mm Hg. Fundoscopy showed a

Figure 2 Inferior choroidal and retinal coloboma (sparing the optic nerve) in both the eyes: (A) right eye and (B) left eye.

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Figure 3 Absence of iris coloboma and presence of Krukenberg's spindle in the left eye.

partial retinochoroidal inferior coloboma, a central 0.45 cup-to-disc ratio, a neuroretinal rim without notching, mild peripapillary atrophy (zone α) and a healthy macula (figure 2B).

Diagnoses of right eye iris coloboma, retinochoroidal partial coloboma in both the eyes, pigment dispersion syndrome in the left eye and physiological cupping, were made.

TREATMENT

Iridotomy was performed in the left eye, to diminish the possibility of future pigmentary glaucoma.

OUTCOME AND FOLLOW-UP

At 13 months after the laser procedure, iridotomy was patent in her left eye. Intraocular pressure was 13 mm Hg in the right eye and 10 mm Hg in the left eye. The cup-disc-ratio was similar to the initial examination (0.55 right eye and 0.45 left eye). The patient was advised to have regular eye examinations.

DISCUSSION

Association of complete iris coloboma in one eye and pigment dispersion syndrome/pigmentary glaucoma in the fellow eye is very rare. According to our knowledge only one case has been previously published by Tesser⁶ 10 years ago. His patient had pigmentary glaucoma and partial iris coloboma in the right eye, and a total iris coloboma in the left eye without pigmentary dispersion syndrome or pigmentary glaucoma.

Between 15% and 50% of patients with pigmentary dispersion syndrome might progress (10 to 20 years after diagnosis) to pigmentary glaucoma with high intraocular pressure and optic nerve damage, a condition very difficult to treat. Genetic factors related with structural abnormalities possibly inherited as autosomal dominant disorders have been identified.¹⁰

Our patient showed clear signs of pigment dispersion syndrome in the left eye but not in the right eye and we think ocular coloboma in the right eye could have been key to the prevention of the reverse pupillary block, avoiding posterior bowing of the iris and contact with zonule or lens, like iridotomy would do.

These cases (ours and the one reported by Tesser⁶), having the fellow eye as natural control group, support the theory of 'reverse pupillary block' and the use of prophylactic iridotomy, which is still an unsettled issue.^{4 9 11–13}

Learning points

- ▶ Pigment dispersion syndrome is generally an asymptomatic disorder discovered as an incidental finding upon routine ophthalmological evaluation.
- ▶ Owing to its possible progression to a glaucoma of difficult control, pigment dispersion syndrome must be in the mind of every ophthalmologist when examining any patient.
- ▶ Iris coloboma seems to protect the eye against pigment dispersion syndrome, supporting the use of prophylactic peripheral laser iridotomy, to eliminate posterior bowing of the iris preventing the friction with the zonules, the disintegration of iris pigment epithelial cells and the release of pigment granules.

Contributors All authors were actively involved with planning, writing and reviewing the paper. We confirm that we all read the manuscript and agreed to its publication in this form.

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