Rieger's anomaly: a 42-year follow-up

Z. GREGOR AND R. A. HITCHINGS

From the Institute of Ophthalmology, Moorfields Eye Hospital, London

SUMMARY A report is given on the follow-up examination of a patient 42 years after Rieger described her case as the first of a newly identified syndrome.

Rieger (1935) described two patients with striking anomalies of iris and angle. Each patient had glaucoma associated with hypoplasia of the anterior leaf of the iris, corectopia, and pseudopolycoria. There were iris adhesions both to the posterior surface of the cornea and to posterior embryotoxon, together with mesodermal tissue in the angle (Rieger, 1935). Later a number of systemic abnormalities were added, including oligo- or hypodontia, with abnormal enamel formation and maxillary hypoplasia (Rieger, 1941). Rieger called the condition dysgenesis mesodermalis cornae et iridis, to which his name was linked.

This report presents the findings for the first patient in Rieger's report who presented for re-examination 42 years later.

Case report

Our patient presented to the casualty department at Moorfields Eye Hospital in January 1977 with a 3-month history of gradual deterioration of vision in her right eye. Measurements of intraocular pressure had been limited to digital tonometry over the past 4 decades and had always been said to be 'normal'. Fifteen years ago she lost light perception in her left eye. She was on no medication.

OCULAR FINDINGS AT ORIGINAL PRESENTATION

At the age of several weeks she was noticed to have had 'changes' in both her eyes. When she was examined at the age of 10 (Rieger, 1935) her vision was 6/8 in each eye, intraocular pressures were raised in both eyes (between 40 and 50 in the left eye, and between 50 and 60 in the right), the right disc and field were normal, and the left disc showed

'flat' excavation and there was a corresponding nasal step in the left visual field.

The anterior segment showed striking changes which may be summarised as follows: The limbus was not well demarcated, and the change from sclera to cornea was very gradual. There were multiple areas of iris adherence to the posterior surface of the cornea at about 1.5 mm from the limbus, and the deeper corneal layers in that area were opacified. The iris itself appeared as a 'delicate membrane of dull brown colour' covered for the most part by yellowish grey tissue, which extended there from the angle. The usual iris structures (crypts, furrows, etc.) were absent. The pupils were abnormal, with corectopia, dyscoria in both eyes, and pseudopolycoria in the right (Figs. 1 and 2). The anterior chamber angles were open but mostly filled by homogeneous yellowish grey tissue. The remainder of the examination showed no abnormality apart from pigment deposits on the anterior capsule of the lenses.

The same year she underwent bilateral cyclo-dialyses. In addition a trephine operation was performed on her left eye.

OCULAR FINDINGS AT RE-PRESENTATION

The corrected visual acuity was 6/36 in the right eye and no light perception in the left. The intraocular pressures were 32 mmHg in the right eye and 68 mmHg in the left, together with epithelial oedema of the left cornea. The right optic disc was grossly cupped. There was no view of the posterior pole in the left eye because of a dense lens opacity. The right peripheral visual field was constricted above and nasally. In both eyes the anterior segment showed an indistinct limbus together with prominent embryotoxon, to which the iris stroma was adherent in a number of places. The iris was grossly deformed with corectopia, dyscoria, and symmetrical polycoria (Figs. 3 and 4). The abnormalities were similar to but more marked than they had been

Address for reprints: Mr R. A. Hitchings, FRCS, Ophthalmic Department, King's College Hospital, Denmark Hill, London SE5 9RS

Fig. 1 A painting of the anterior segment of the right eye; patient aged 10. Corectopia and a small defect in the iris stroma at 11 o'clock. Iris adherent to the cornea in the 7 o'clock position. Indistinct limbus.

(Courtesy of the Editor, Albrecht von Graefes Archiv für Ophthalmologie)

Fig. 2 A painting of the anterior segment of the left eye, patient aged 10. Diagonal orientation of the ectopic pupil, no other stromal defect. Iris adherent to the cornea in the 7 and 1 o'clock positions

(Courtesy of the Editor, Albrecht von Graefes Archiv für Ophthalmologie)

Fig. 3 A painting of the anterior segment of the right eye, patient aged 52. The stromal defect in the 11 o'clock position is complete and much larger. Marked ectropion uveae with uniform radial striations of the iris stroma. The anterior synechiae are more extensive

Fig. 4 A painting of the anterior segment of the left eye, patient aged 52. A new stromal defect in the 9.30 o'clock position, uniform radial striations. The anterior synechiae are more extensive.

42 years ago. There were concentric furrows in a uniformly light brown atrophic anterior leaf of the iris, ectropion uveae, and areas of frank stromal atrophy. In both eyes the anterior chamber angles were completely closed throughout their circumference. There were no abnormalities on systemic examination.

The patient's intraocular pressure was controlled with drops of guanethidine 1%, neutral adrenaline 0.25%, and pilocarpine 4%, and her visual field has not deteriorated further. The patient was not aware of any eye trouble in other members of her family. Her son was found on examination to have normal irides and angles.

Comment

This case report is remarkable for its time span and is also of historical interest. We agree with Rieger that this patient suffers from the anomaly that bears his name, because of the early onset, the bilateral involvement, and the typical appearances despite the absence both of a positive family history and of the systemic abnormalities commonly associated.

Two aspects of the management of this disorder deserve a comment. They are (1) progression of iris changes with Rieger's syndrome, and (2) the preservation of vision in eyes with uncontrolled glaucoma.

PROGRESSION

Rieger's anomaly has been described as a congenital deformity of iris morphology that does not progress (Alkemade, 1969). However, in a number of cases progression has been reported. Cross and Maumenee (1973), in a critical review, stated that progression when noted occurred before the end of the first decade, whereas other long-term follow-up reports suggesting lack of progression were for patients seen after the first decade. In our patient the slit-like defects in the irides which developed over the 42-year period are unlikely to have been caused by

the intraocular surgery or by iris stromal 'rarefaction' secondary to uncontrolled, raised intraocular pressure. We consider these changes to be the result of continuing dissolution of the iris.

MANAGEMENT

Alkemade (1969), in an extensive review of 163 patients with Rieger's syndrome, said that control of intraocular pressure was not easy to achieve. Patients did often not respond to traditional medical treatment, and a large number of operations had been employed in the past. Greater success was reported by Rice in a recent series. He controlled the glaucoma in 5 out of 6 eyes with Rieger's anomaly by means of goniotomy, though the same procedure failed to do so in 17 out of 19 eyes with the closely related syndrome of Axenfeld (Rice, 1977).

Our patient had uncontrolled glaucoma for an unknown but probably extensive period, but preserved useful vision in one eye for 52 years. This long-term visual prognosis should be borne in mind when surgical procedures are being considered for eyes with Rieger's syndrome.

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