Keratoglobus in the Rubinstein-Taybi syndrome

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SUMMARY The case of a 20-year-old male with the Rubinstein-Taybi syndrome associated with unilateral acute corneal hydrops is presented. The initial findings were of keratoglobus, but after the corneal oedema had settled the cornea assumed a more conical contour. The relationship between keratoglobus, keratoconus, and acute hydrops is discussed.

Rubinstein-Taybi syndrome is a condition of broad thumbs and toes, facial abnormalities, and mental retardation that was first described in 1963.¹

We describe a patient with this syndrome who developed unilateral acute corneal hydrops. Corneal ectasia has not been described in association with this syndrome previously. Initially the hydropic cornea was globular in contour, but later, as the oedema settled, the cornea assumed a more conical shape. The relationship between keratoglobus, keratoconus, and acute hydrops is discussed.

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Case report

A 20-year-old male with Rubinstein-Taybi syndrome presented to the Ophthalmology Department of the Royal Hallamshire Hospital, Sheffield, with a painful left eye. Six months previously the patient's father had noted the left cornea to be bulging, and two days before presentation the cornea had become opaque. The patient was in pain, extremely distressed, and very difficult to examine.

The diagnosis of Rubinstein-Taybi syndrome had been made when the patient was 2 years old at the Sheffield Children's Hospital. He was mentally retarded, microcephalic, and had typical features of the syndrome with a beaked nose, hypertelorism,

Fig. 1 Lateral view of left eye at presentation showing corneal oedema and keratoglobus.

Fig. 2 Anterior view of left eye at presentation showing diffuse corneal oedema and central bullae.

Fig. 3 Lateral view of left eye six months after presentation. The cornea is clear and has lost its globular contour.

and broad thumbs and toes. He had a convergent squint and underwent surgery, but no other ocular abnormalities were noted.

Examination under ketamine anaesthesia showed that the left cornea was diffusely bulging and oedematous with a globular contour (Figs. 1, 2). The cornea was thinned apart from the central area, which was irregular and markedly oedematous. No view of the anterior chamber was possible. The intraocular pressure was 19 mmHg and the corneal diameter 11 mm. The right eye showed no evidence of corneal thinning or ectasia.

A diagnosis of unilateral keratoglobus with acute hydrops was made and a subconjunctival injection of 4 mg of betamethasone given. Treatment was symptomatic, with topical prednisolone drops and mydriatics. Over the following four weeks the symptoms settled, and he became less distressed but no more co-operative.

Six months later he was re-examined under ketamine anaesthesia. The left cornea had cleared but was markedly thinned and protruding. The peripheral cornea was vascularised and the central stroma scarred, though the overlying epithelium was

intact (Fig. 3). The anterior chamber was quiet and of normal depth. Intraocular pressure was 20 mmHg in both eyes.

Discussion

The syndrome of broad thumbs and toes, facial abnormalities, and mental retardation was described by Rubinstein and Taybi in 1963.¹ They reported on seven children who were severely mentally handicapped, microcephalic, and who had broad thumbs and great toes. Other features since reported include hypotonia and disturbed gait, frequent chest infections, a beaked nose, external ear abnormalities, a high narrow palate, and several skeletal anomalies.²

The ocular features of the syndrome have been described.³ Ptosis, antimongoloid slant of the palpebral fissures, strabismus, refractive error, epicanthal folds, optic atrophy, cataract, coloboma of the iris, and high arching eyebrows were included in the features described by Rubinstein and Taybi in their original report. Roy *et al.*⁴ add long lashes and nasolacrimal duct obstruction to this list.

Corneal ectasia is not listed as an association, though one of Rubinstein and Taybi's initial cases was reported as having a corneal scar.¹ The association of keratoconus with Down's syndrome is well known, approximately 5% of these patients being affected.⁵ Acute hydrops in Down's syndrome is more common than in other patients with keratoconus⁶ and may be associated with eye rubbing. To our knowledge keratoconus has not been reported in association with Rubinstein-Taybi syndrome.

The relationship between keratoglobus and other corneal abnormalities, especially keratoconus, megalocornea, and buphthalmos has been the source of some confusion in the past.⁷ However, keratoglobus is now listed as a corneal ectasia, together with keratoconus, pellucid marginal degeneration, and posterior keratoconus.⁸

Keratoglobus is characterised by thinning and protrusion of the entire cornea, which is otherwise of normal size. Corneal thinning is most marked peripherally. Ruptures of Descemet's membrane leading to acute hydrops do occur,^{9,10} and the cornea is susceptible to rupture owing to its extreme thinness.⁹

Pouliquen recognises two types of keratoglobus.¹¹ The first is a rare bilateral condition that is usually congenital, with an autosomal recessive inheritance. It may also occur as part of a syndrome with joint hyperextensibility, deafness, and blue sclerae.¹² The second type of keratoglobus is less common and may be unilateral. It arises in an abnormal cornea, usually in keratoconus, though it has been described with

hyperthyroidism.¹³ Pouliquen considers this form as severe keratoconus.

As the patient reported here had the appearance of keratoconus after the hydrops had settled, his condition may be classified as the second type of keratoglobus.

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