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CASE PRESENTATION

Congenital Keratoglobus: a Case Report

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ABSTRACT

Introduction: congenital keratoglobus is a bilateral disease characterized by generalized or diffuse, non-inflammatory corneal thinning. It is associated with high refractive errors and corneal fragility, which may lead to amblyopia and ocular perforation, with negative implications for the quality of life of patients and their families.
Objective: to describe the clinical features, differential diagnosis, and therapeutic options in a patient with congenital keratoglobus.
Case Report: a six-month-old male infant, with no relevant prenatal or perinatal history, was referred to pediatric ophthalmology for ocular screening. The initial examination revealed large corneas with generalized protrusion and bilateral central thinning, deep anterior chamber, and clear media. Retinoscopy showed high refractive errors, confirmed by keratometry and pachymetry, with corneal diameters of 14 mm and normal intraocular pressure. Fundus examination revealed an attached retina and an optic disc with well-defined margins. Differential diagnoses such as isolated megalocornea, keratoconus, and congenital glaucoma were ruled out based on biometry, gonioscopy, and absence of characteristic clinical signs. Genetic studies excluded association with autosomal recessive syndromes. Optical correction with rigid gas-permeable contact lenses and periodic follow-up were indicated, with parents informed about the guarded prognosis and the need for vigilance to prevent amblyopia.
Conclusions: clinical diagnosis of keratoglobus at early ages is challenging due to its similarity with other congenital ocular diseases and the patient's limited cooperation. Early correction of the high refractive error prevents the onset of amblyopia.

Keywords: Astigmatism; Clinical Diagnosis; Diagnosis, Differential; Corneal Diseases; Contact Lenses.

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INTRODUCTION

Keratoglobus is a corneal ectasia characterized by generalized or diffuse, non-inflammatory corneal thinning. Considered a rare bilateral disease, it may appear in a congenital or acquired form, with no predilection for sex or ethnic groups.[1,2]

The congenital variant is present from birth, with progression to more severe forms between 20 and 30 years of age. No hereditary pattern is defined, although some authors describe it as autosomal recessive; associated with Ehlers-Danlos syndrome type VI or brittle cornea syndrome. The acquired form has been described in association with "chronic rubbers" as a result of allergic keratoconjunctivitis, chronic marginal blepharitis, idiopathic orbital inflammation, and thyroid eye disease.[3,4]

The clinical presentation, fundamentally in early ages, is usually confused with megalocornea, keratoconus, and congenital glaucoma.[5] The most frequent complications are usually amblyopia due to the associated refractive defect and corneal perforation.[6] Making an early diagnosis is essential to correct the high astigmatism it produces and to avoid negative sequelae on vision. For these reasons, it was decided to present a case report with the objective of describing the clinical features, differential diagnosis, and therapeutic options in a patient with congenital keratoglobus.

CASE REPORT

A six-month-old male infant, who attended the pediatric ophthalmology consultation for ocular disease screening due to his age. Upon interview, his parents denied prenatal, delivery, and postnatal history.

The initial ophthalmological physical examination with simple illumination detected both eyes with large corneas (Figure 1). Anterior segment biomicroscopy confirmed: increased corneal diameter with generalized protrusion and central thinning in both eyes (OU). Deep anterior chamber, iris without alteration, central, regular and symmetrical pupil, clear media, and normal pupillary reflexes in OU.

Fig. 1 Increased corneal diameter in OU.

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Dilated fundus examination: Oval optic disc with well-defined margins, attached retina OU.
Retinoscopy (under dilation):
OD: -2.00-5.50 x 180°
OS: -2.00-6.00 x 180°

Studies under general anesthesia were planned, with the following results:
Keratometry: OD: K1 46.00 K2: 47.75, OS: K1 46.25 K2: 47.75
Corneometry: 14 mm OU
Intraocular pressure (IOP): 12 mmHg OU
Central corneal thickness: OD: 421 microns, OS: 424 microns
Biometry: OD: 21.49 mm, OS: 21.43 mm
Gonioscopy: chamber angle visible up to the ciliary body band in OU (open angle)

Due to the clinical characteristics of the patient and the results of the examinations performed, isolated megalocornea, keratoconus, and congenital glaucoma were ruled out; congenital keratoglobus was diagnosed. To rule out association with other autosomal recessive inheritance syndromes (Ehlers-Danlos type VI and brittle cornea syndrome), a consultation with genetics specialists was decided. The studies performed at the provincial Medical Genetics Center were negative.

Optical correction with gas-permeable contact lenses was indicated, maintaining periodic follow-up in the pediatric ophthalmology and cornea consultation, to evaluate the degree of progression of the disease, the evolution of the associated refractive defect, and the prevention of amblyopia. The parents were informed that the visual prognosis was guarded.

DISCUSSION

Keratoglobus is a very rare bilateral ectasia. The typical globular protrusion of the cornea is due to generalized or diffuse thinning, more marked in the periphery, adopting the corneal profile a globular shape. The anterior chamber is deep and the corneal diameter is usually normal, although some authors report that it may be increased, as evidenced in the present case.[1,7]

It is described that it can be congenital or acquired. Genetically, it has been related to keratoconus and other general syndromes, which were ruled out. Isolated forms have been described. An early presentation is associated with a higher risk of progression and complications. Acute hydrops and spontaneous or trauma-induced perforation are frequent.[8]

The disease is clinically characterized by symptoms of fluctuating vision, light hypersensitivity, irritation, and diplopia, aspects that may go unnoticed by family members in congenital forms. The diagnosis is generally clinical, once the patient has been examined; however, keratometry, corneometry, pachymetry, corneal topography, and more recently corneal tomography, have improved the possibilities for identifying corneal ectasias in early stages. Other examinations such as biometry, gonioscopy, intraocular pressure measurement, and fundus examination allow differentiation from other conditions.[9]

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The differential diagnosis of keratoglobus must be made with megalocornea, keratoconus, and congenital glaucoma. In megalocornea, there is a corneal diameter greater than 13 mm, an element present in the case, but it is ruled out in isolation since the thickness, curvature, and transparency must be normal to propose the diagnosis. The patient under study presented a greatly decreased thickness and increased curvature.[5]

Keratoconus refers to an axial, bilateral corneal ectasia, generally asymmetric, in which thinning causes a progressive increase in corneal curvature, with myopia and irregular astigmatism. Despite being the most frequent ectasia, in the presented case it is ruled out because the most frequent age of presentation is at puberty, with progression throughout the third and fourth decades of life. On ophthalmological examination, no scissor shadows were observed on retinoscopy, no Charleaux's oil droplet sign by retroillumination with the dilated pupil, nor the classic external signs of Munson or Rizzuti. Furthermore, on slit lamp examination, no other signs were found: Fleischer ring, Vogt's striae, superficial and deep corneal opacities, and prominent corneal nerves.[4,7]

Another essential differential diagnosis to be made is congenital glaucoma, a disease that was ruled out since the patient, despite presenting an increased corneal diameter, did not show edema, the intraocular pressure was normal, the fundus did not express glaucomatous alteration, and on gonioscopy the characteristic signs were not reported: poorly differentiated angle, anterior insertion of the iris, and the presence of a mesodermal membrane that totally or partially lines the angle (Barkan's membrane).[10]

Treatment options vary according to the patient's age and the clinical characteristics of the disease. General treatment is fundamental; the patient must be instructed on the importance of not rubbing their eyes, using topical anti-allergic medication in patients with atopy, and the use of topical ocular lubricants.[1]

Optical correction can be achieved with glasses or contact lenses; rigid gas-permeable and scleral lenses are recommended. Some authors discourage their use due to a higher risk of trauma during insertion and removal maneuvers, as well as a higher probability of infection; however, other specialists report excellent results if the fitting is performed at early ages.[11] In the present case, this was the option considered with good results during the follow-up period.

Surgical options are reserved for patients with complications or those who are not satisfied with corrective treatments using lenses, always assessing the risk-benefit ratio. Among the surgical techniques are intrastromal rings, anterior lamellar keratoplasty, and penetrating keratoplasty with large-diameter corneal grafts (from limbus to limbus), although it entails a higher risk of rejection due to corneal vascularization, loss of limbal stem cells, and anatomical alteration of the chamber angle.[1,4,8]

Through the present case, the importance of an early ophthalmological examination is evidenced. It is valid to highlight the need to comply with blindness prevention programs in Cuba, which make it possible to screen cases with diseases that may go unnoticed by family members and that constitute a cause of low vision or legal blindness due to amblyopia. The timely diagnosis in the present case allowed early treatment, aimed at avoiding this complication or sequel.

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CONCLUSIONS

The clinical diagnosis of keratoglobus at early ages is difficult due to its similarity with other congenital ocular diseases, and the patient's cooperation. A detailed examination and the use of specific diagnostic tests allow an accurate diagnosis and the possibility of early correction of the high refractive defect, in order to prevent the appearance of low vision or blindness due to amblyopia.

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