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Case report

Keratoglobus with ARCL1B (EFEMP2 gene) cutis laxa

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ABSTRACT

Purpose: To report a case of keratoglobus in a patient with autosomal recessive (AR) cutis laxa.
Observations: A 38 year old male presented with decreased vision in both eyes uncorrectable with spectacles and a history of corneal rupture in the left eye from incidental trauma a decade prior. His ocular exam was consistent with keratoglobus. His medical and family history indicated AR cutis laxa.
Conclusions and importance: We believe that this is the first reported case of keratoglobus associated with cutis laxa.

1. Report of a case

The patient is a 38 year old male who presented with a history of reduced vision in each eye from birth. He had been diagnosed with Cutis laxa. His parents were unaffected but his mother was a carrier of this gene mutation. His older sister was born with the disease and died during childhood due to gastrointestinal complications. Genetic testing indicated a mutation at the EFEMP2 gene. The patient had a history of sagging facial and arm skin as well as abdominal wall hernias. A corneal rupture occurred in the left eye ten years prior to his visit due to a minor injury and was repaired.

Visual acuity on presentation was 20/50 in the right eye and 20/70 in the left eye with best spectacle corrected visual acuity. Visual acuity with scleral contact lenses was 20/20 in each eye. Slit lamp evaluation demonstrated diffuse thinning of each cornea that was more prominent peripherally (Fig. 1). Corneal opacity was present temporally in the left eye at the location of the previous rupture. The corneal topography demonstrated diffuse corneal protrusion and corneal thinning (Fig. 2). The corneal diameter measured in the horizontal meridian was approximately 11.6 mm in each eye measured with the ocular coherence tomogram (OCT).

2. Discussion

Keratoglobus is a rare condition that is grouped with other non-inflammatory corneal ectasia and include keratoconus and pellucid marginal corneal degeneration PMCD). Keratoglobus is characterized by diffuse thinning of the cornea from limbus-limbus associated with protrusion of the cornea.[1] The corneal diameter is normal which differentiates it from megalocornea and buphthalmos. Keratoglobus is a bilateral condition that is usually present at birth. It is usually minimally progressive. Keratoglobus has been associated with systemic connective tissue disorders such as Ehlers-Danlos syndrome,[2] Marfan syndrome, and Rubenstein-Taybi Syndrome.[3] Keratoglobus is not

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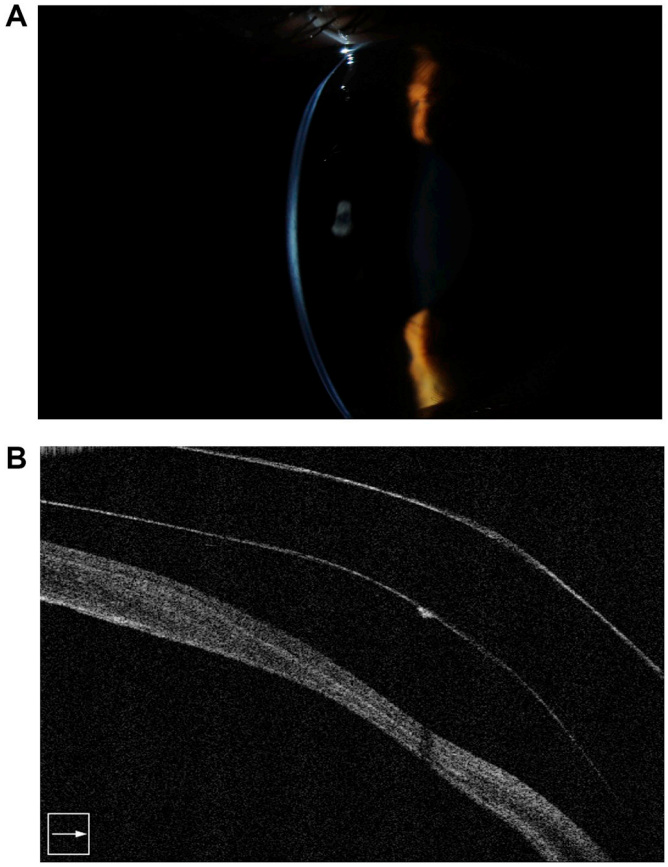

Fig. 1. Slit lamp photos (A) and OCT with scleral contact lens in place (B) of the right cornea noting generalized and focal thinning and protrusion.

usually amendable to current corneal crosslinking techniques due to the extent and diffuse nature of corneal thinning and the non-progressive nature of the condition. Surgical management is challenging and may involve a staged approach with an initial partial thickness corneoscleroplasty followed by a smaller diameter penetrating keratoplasty.

Cutis laxa is a connective tissue disorder characterized by loose skin particularly on the face, arms, and groin. Affected individuals often have an aged appearance. Cutis laxa has a wide range of clinical presentation including isolated skin manifestations to severe life-threatening systemic complications. Cutis laxa may involve the gastrointestinal, pulmonary, musculoskeletal, and vascular systems. Inguinal and umbilical hernias develop as well as vascular aneurysms. Chronic obstructive pulmonary changes and diaphragmatic defects as well as cardiac complications may contribute to early deaths. Generalized muscular hypotonia and joint laxity may occur.[4]

Various patterns of inheritance have been reported. The condition may present as an autosomal dominant, autosomal recessive (as in this case), X-Linked recessive inheritance, or acquired.[4] Abnormal formation of elastic fibers is the source of the findings with Cutis laxa. The ELN gene produces elastin, a critical component of elastic fibers. Other genes (EFEMP2, FBLN5, and ATP6V0A2) code for important proteins in the formation of elastic fibers and have been associated with cutis laxa. Entropion associated with the dermal features of cutis laxa has been reported.[5] Corneal findings have not been reported with Cutis laxa. Elastic fibers are important functional elements in a normal cornea and their absence may be associated with corneal changes.[6]

We believe that this represents the first case of corneal ectasia found in a patient with documented cutis laxa. Due to the importance of elastic fibers in the cornea it is expected additional cases will be found. It is important that dermatologists, ophthalmologists, and other healthcare providers be aware of this association so that affected patients can be informed about the possibilities of severe occur injury from minor trauma and also the potential visual rehabilitation through contact lenses.

Patient consent

This report does not contain any personal information that could lead to the identification of the patient.

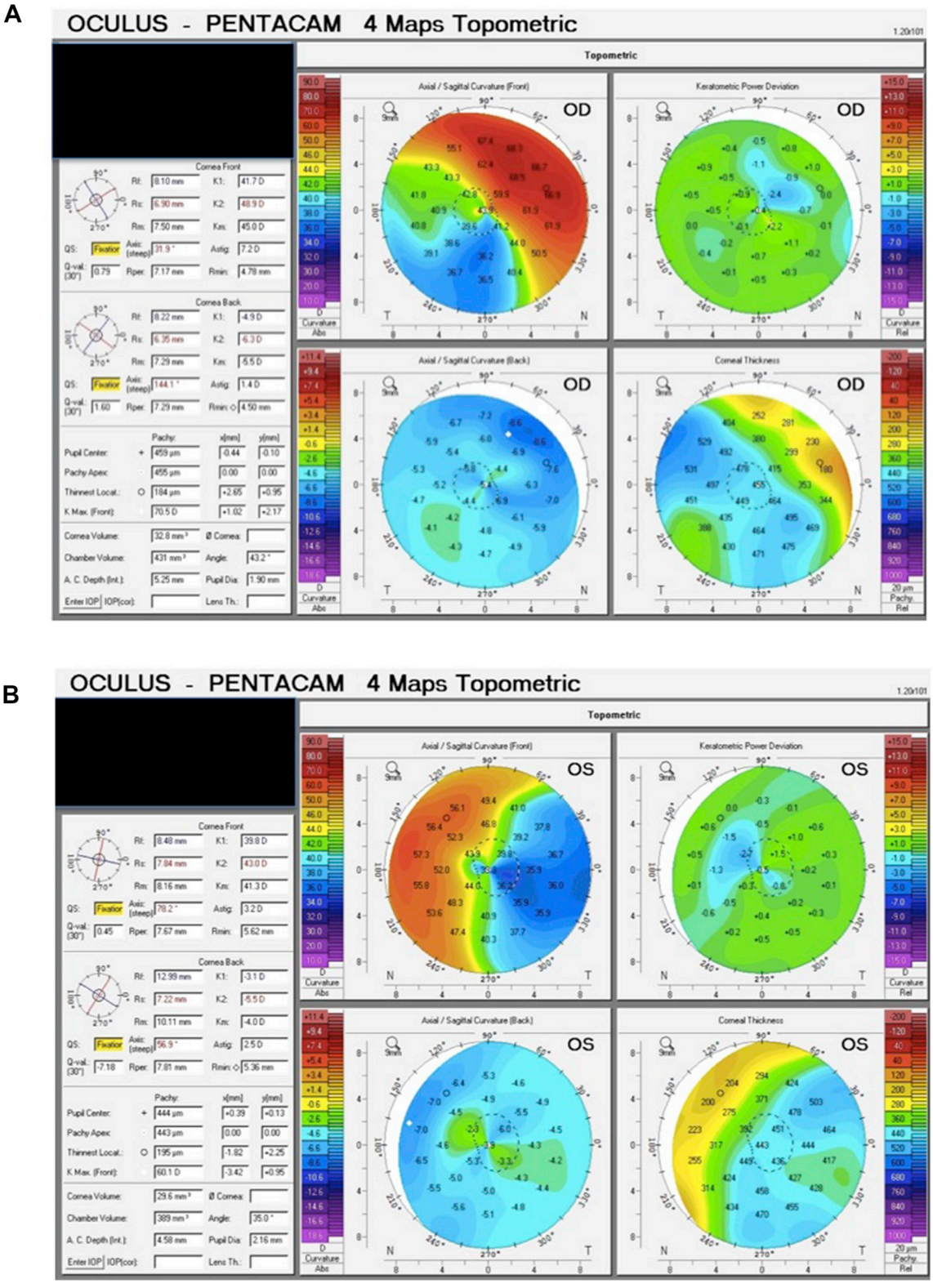

Fig. 2. Corneal topography Right (A) and Left (B) demonstrating bilateral diffuse and focal thinning and superonasal steepening consistent with keratoglobus.

Conflicts of interest

There are no conflicts of interest for any of the authors.

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Conflicts of interest

There are no conflicts of interest.

Authorship

All authors attest that they meet the current ICMJE criteria for Authorship.

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