

Case Report

A Rare Case of Concurrent Granular Corneal Dystrophy and Keratoconus

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Article Notes:

Received: jun. 27, 2016

Received in revised form: Jul. 21, 2016

Accepted: Sept. 2, 2016

Available Online: Sept. 24, 2016

Keywords:

Corneal dystrophy

Keratoconus

Iran

Abstract

A 23-year-old female presented with progressive bilateral visual loss for the past 5 years. The patient had no history of systemic disease, surgery or medications. Complete ophthalmologic examination and topography were performed. On ophthalmic examination, uncorrected visual acuity was 20/30 in the right eye and 20/50 in the left eye. Both corneas appeared hazy on gross examination. On slit-lamp biomicroscopy, few well circumscribed gray white discrete granular deposits in the central corneal stroma of both eyes were observed. Both corneas were thin and bulging. Corneal topography showed a pattern consistent with keratoconus. The patient was clinically diagnosed as a case of concurrent granular dystrophy and keratoconus, which is a very rare presentation.

How to cite this article: Gohari M, Shojaei A, Rahmatkha S, Salehi H. A Rare Case of Concurrent Granular Corneal Dystrophy and Keratoconus. *Journal of Ophthalmic and Optometric Sciences*. 2016;1(1): 37-41.

Introduction

Keratoconus is a progressive non-inflammatory corneal stromal thinning disorder, which leads to corneal ectasia with irregular myopic astigmatism and visual impairment, as early as in the second decade of life. Although the hereditary pattern is unpredictable, positive family history can be detected in 6 - 8 % of patients ⁽¹⁾. However, despite multiple single reports of an association with other systemic or ocular disorders, it is most often an isolated sporadic condition ^(1,2). Although, keratoconus and corneal dystrophies are rare in the general population, there are reported cases of both occurring in the same eye ⁽³⁾.

It is important to report the association of other conditions with keratoconus since many of these combinations could be manifestations of a common pathogenetic mechanism or may represent adjacent genetic defects ^(1,3). In this report, we present a case with concurrent keratoconus and granular corneal dystrophy.

Case Report

A 23-year-old female presented with progressive bilateral visual loss for the past 5 years. The patient denied any history of systemic disease, surgery, or medications. There was no consanguinity between her parents and no history of a similar disease in her family or pedigree. General physical examination was unremarkable. At the first visit, uncorrected visual acuity (UCVA) was 20/30 in the right eye and 20/40 in the left eye. Best spectacle visual acuity (BSCVA) was 20/25 in the right eye and 20/30 in the left eye. The objective refraction was (+ 2.00 - 5.75.10) in the right eye and (+ 2.50 - 9.25 . 70) in the left eye. Pupillary reactions and ocular motility were normal. The corneas were hazy bilaterally. In slit-lamp biomicroscopy, few well circumscribed gray white discrete granular deposits in the central corneal stroma were observed

bilaterally. There was diffuse haziness which was more pronounced in the central cornea with greater severity in the left eye. Both eyes were thin and bulging. Fleischer's rings and Vogt's striae were not present (Figure 1). Intraocular pressure was 12 and 10 mmHg in the right and left eyes, respectively. The Scissors reflex was unremarkable in both eyes because of central corneal haziness. Despite the hazy media, funduscopy was normal. The patient was clinically diagnosed with granular dystrophy and probable keratoconus (KCN). Corneal topography showed a characteristic pattern of KCN (Figure 2).



Figure 1: In slit-lamp biomicroscopy Fleischer's rings and Vogt's striae were not present.

Discussion

Granular corneal dystrophy (GCD) is transmitted as an autosomal trait. The corneal opacities usually become apparent in the first decade of life as small, discrete, sharply demarcated, and grayish white opacities in the anterior axial stroma ⁽⁴⁾. GCD can be divided into at least three types based on clinical appearance: the classic form of GCD, Avellino corneal dystrophy and the superficial variant of GCD. These dystrophies share identical corneal deposits that stain red after the application of Masson trichrome stain and that appear as rod-shaped bodies in transmission electron microscopy ⁽⁵⁾.

The association of keratoconus with a relatively

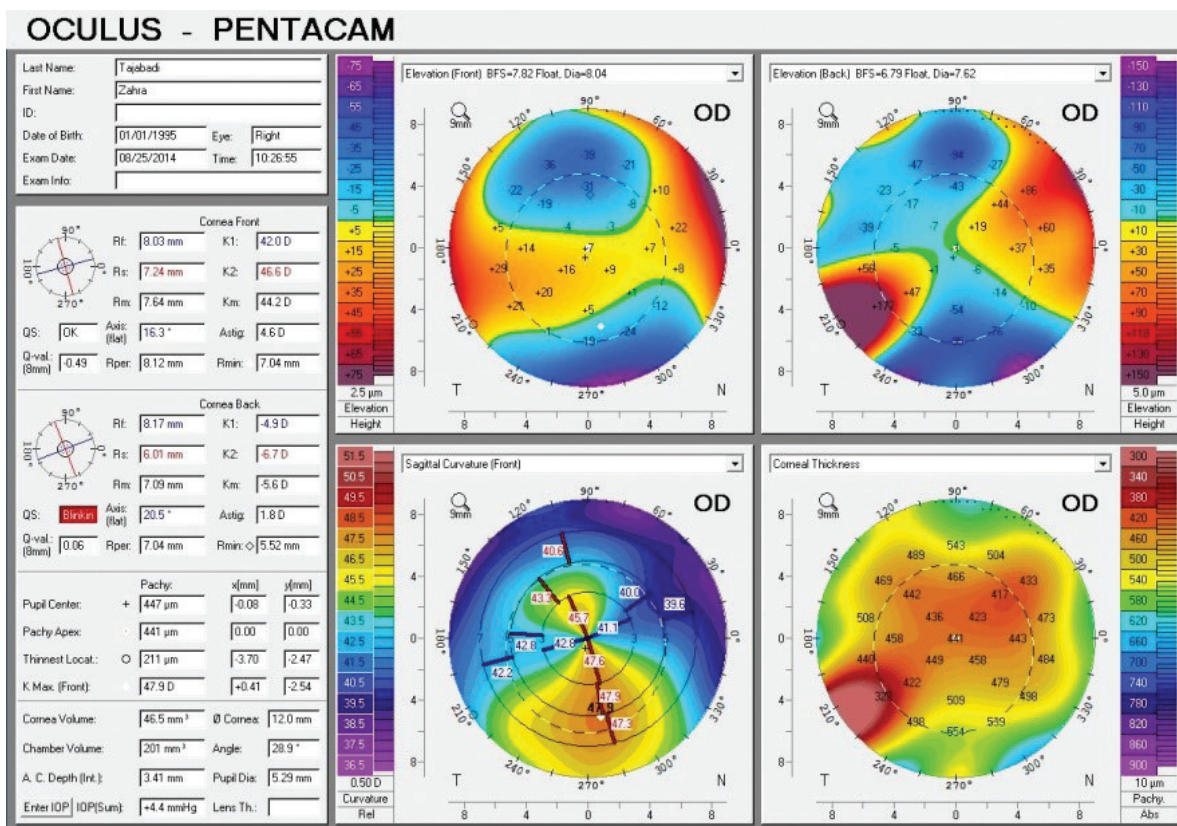
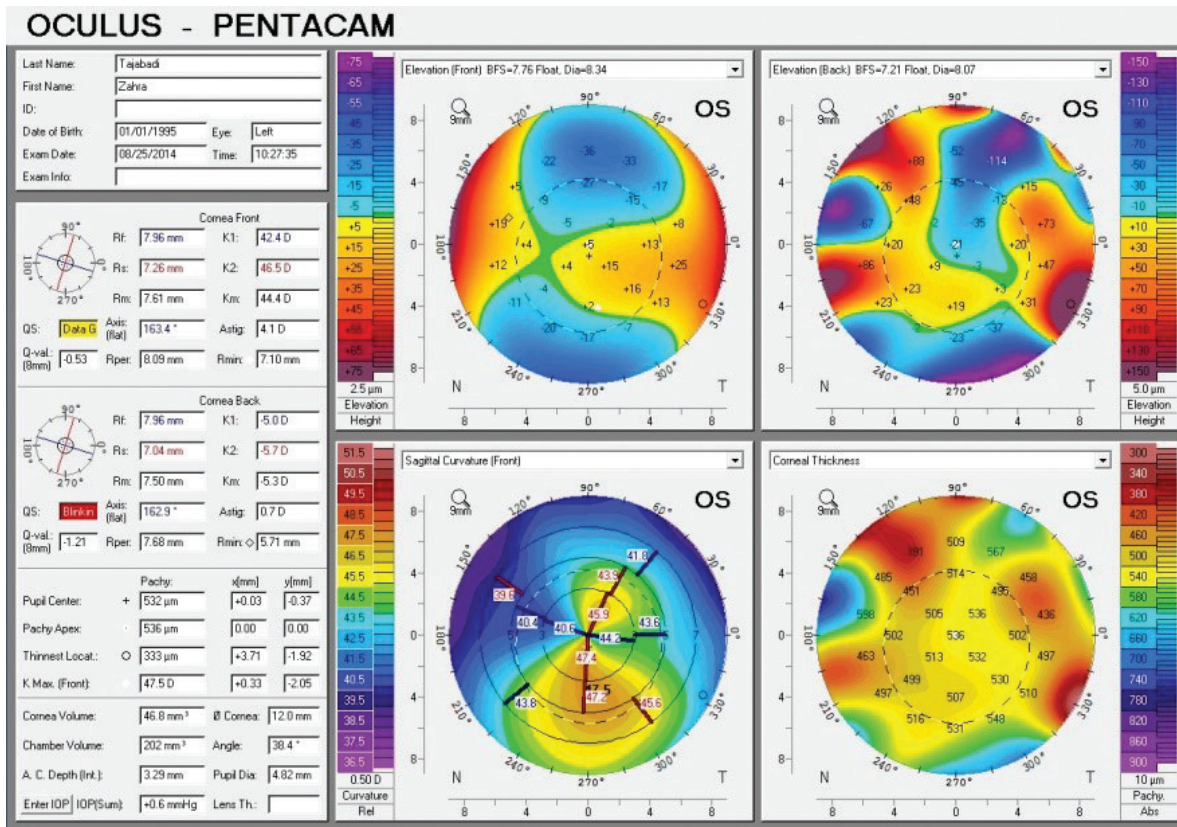


Figure 2: Corneal topography showing significant corneal thinning, asymmetric bow-tie pattern and abnormal elevation maps in both eyes.

rare disorder like granular dystrophy suggests that there may be a genetic linkage between the two diseases. Therefore, chromosome 5, which has been identified for granular, lattice type 1, and Avellino dystrophy, may be a possible gene locus for at least one form of hereditary keratoconus as has already been shown for chromosome 21, although a chance association cannot be excluded⁽⁵⁻⁷⁾.

The incidence of combined keratoconus and granular dystrophy may be higher than suggested by the few case reports in the literature because early stages of keratoconus can be overlooked on routine examination. Computerized corneal topography should help to reveal more cases and to further elucidate its pattern of inheritance.

From a clinical point of view, this case shows the importance of excluding additional keratoconus in patients with granular dystrophy, because this disorder can be the main reason for the decreased vision and can often be treated with contact lenses, so that keratoplasty can be at least delayed or even avoided. Conversely, excimer laser treatment, which can be used in some cases with granular dystrophy, should be applied with caution because of the corneal thinning caused by additional keratoconus^(8,9).

Conclusion

Our case of concurrent granular dystrophy and keratoconus shows the importance of excluding additional keratoconus in patients with granular dystrophy.

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Footnotes and Financial Disclosures

Conflict of Interest:

The authors declare no conflict of interest with the subject matter of the present manuscript.