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Case Report

Familial Granular Corneal Dystrophy (Type 1) Involving four Members of a Single Family: A Case Report

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Abstract

Granular corneal dystrophy (GCD), Type 1 also known as corneal dystrophy Groenouw type I is a type of stromal corneal dystrophy. We report a series of four affected members of a family diagnosed with (GCD type 1). Our index case was a 45-year-old female with bilateral diminution of vision for the past 5 years. The diagnosis was established using clinical examination and anterior segment optical coherence tomography. Further screening of her family members revealed similar disease in her father and two siblings. The patient was not willing for deep anterior lamellar keratoplasty. Hence, she was managed conservatively with best-corrected visual acuity of 6/36 in the right eye and 6/18 in the left eye.

Key words: Anterior segment optical coherence tomography, Autosomal dominant, Granular corneal dystrophy

INTRODUCTION

Granular corneal dystrophy (GCD) (GCD type 1, also known as corneal dystrophy Groenouw type I) is a rare subtype of stromal corneal dystrophy, generally starts around puberty with a slow progression.[1,2] The patient may be asymptomatic for a prolonged period, though some may present with glare, photophobia, and/or recurrent erosions.[1] In this classic variant, there is bilateral involvement showing white, discrete, granular deposits of hyaline in anterior, and deep stroma with clear intervening stroma and sparing of the limbus.[1] Histopathological examination shows bright red amorphous deposits of hyaline material when stained with Masson trichome.[3,4] Definitive treatment for visually significant lesions includes deep anterior lamellar keratoplasty (DALK) or penetrating keratoplasty (PK) with good visual outcomes.[5]

A 45-year-old female presented in the eye clinic with chief complaints of diminution of vision in both eyes for the past 5 years. There was no trauma or associated ocular or systemic illness. Her uncorrected visual acuity was 6/60 in the right eye and 6/36 in the left eye. Slit-lamp biomicroscopy revealed mild epithelial erosion involving central cornea in both eyes (right eye > left eye). There were multiple, white granular deposits in the anterior and deep stroma of the central cornea with sparing of the limbus [Figures 1 and 2]. Rest of the anterior and posterior segment examinations were unremarkable.

Keratometry readings of the right eye (K1 = 43.75 D, K2 = 47.00 D; axis 94°) and left eye (K1 = 43.25 D, K2 = 44.50 D; axis =67°) were noted. Pachymetry revealed increased central corneal thickness readings (right eye =599 μm and left eye =564 μm). Anterior segment optical coherence tomography of both eyes revealed hyperreflective granular lesions extending from the bowman's layer to deeper stromal lamellae in both eyes (right > left eye) as shown in Figures 1 and 2. Patient was advised DALK but patient was not willing for surgical intervention, therefore was managed conservatively. Best-corrected visual acuity (BCVA) was 6/36 and 6/18 in the right and left eye, respectively.

In view of the autosomal dominant transmission of GCD type 1, a thorough screening of her family members was performed as shown in family chart [Figure 3]. We examined a total of 11 patients among which 4 (index patient, her father, and two younger

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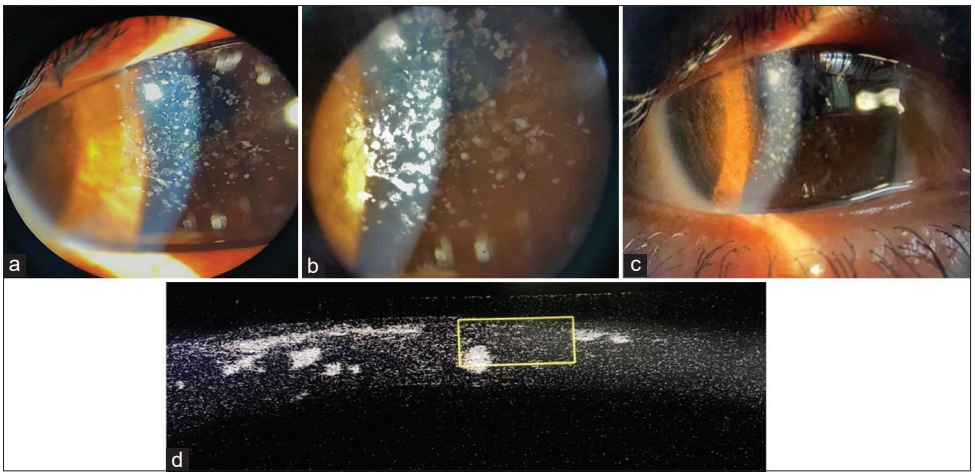

Figure 1: Slit-lamp photograph of the right eye (a-c) showing multiple, whitish granular deposits in the corneal stroma. Anterior segment optical coherence tomography (d) showed the presence of multiple anterior to mid stromal hyper-reflective opacities

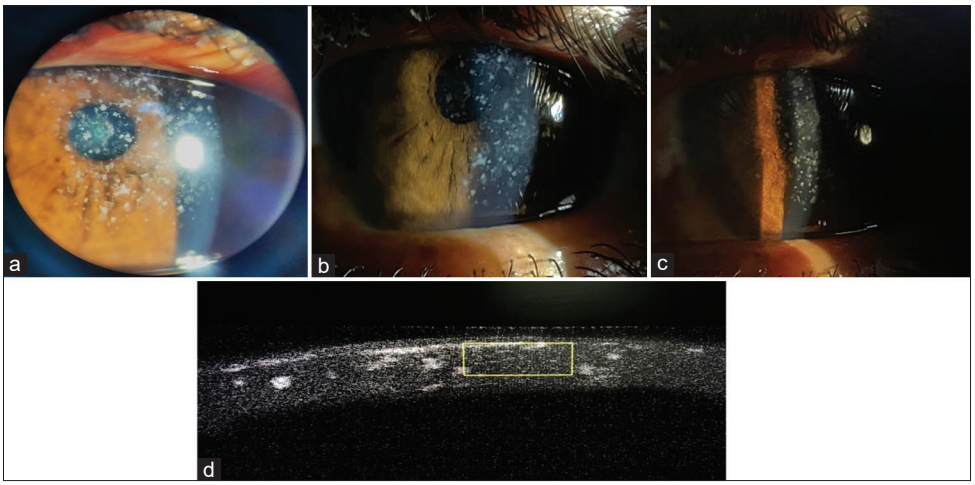

Figure 2: Slit-lamp photograph of the left eye (a-c) showing multiple, whitish granular deposits in the corneal stroma (less dense compared to right eye). Anterior segment optical coherence tomography (d) multiple anterior to mid-stromal hyper-reflective opacities in the central cornea

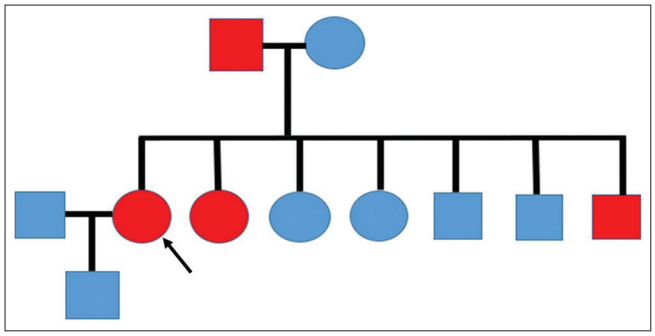

Figure 3: Family chart showing four affected family members shown as red square and red circle. These included the index case (female, 45 years), her father, and two siblings (one male and one female)

siblings) were detected to have GCD type 1 [Figure 4]. Her father had a presence of significant cataract in the right eye along with corneal stromal opacity and was planned for DALK and cataract surgery. The left eye was pseudophakic with BCVA of 6/12. Other two siblings also had complaints of diminution of vision for distant objects for which they were prescribed spectacles to correct the refractive error along with artificial lubricating eye drops [Figure 4].

DISCUSSION

GCD type 1 is an autosomal dominant inherited corneal stromal dystrophy.[1,4] We screened a family with GCD and found four members within varying stages of disease severity. Among 11 members, 4 (36.4%) were found to have GCD type 1. Genetic analysis forms the mainstay to identify and classify the disease.[1] However, in view of financial constraints, this was not performed in our study cohort. Although the majority of patients are asymptomatic

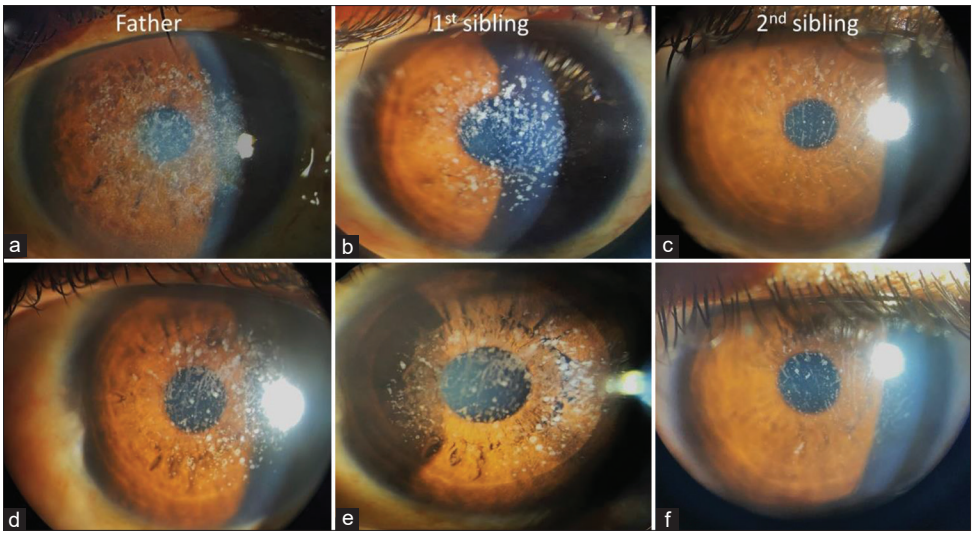

Figure 4: Slit-lamp photograph (diffuse illumination) of three other family members showing similar stromal opacities. Father (right eye – [a], left eye – [d]), 1st sibling (right eye – [b], left eye – [e]), 2nd sibling (right eye – [c], left eye – [f])

in initial stages, insidious progression heralds a multitude of ocular symptoms (diminution of vision, glare, and photophobia due to recurrent erosions). Whereas asymptomatic individuals benefit from refractive error correction and artificial tear substitutes alone, advanced cases may benefit from surgical procedures such as DALK or PK.[5,6]

CONCLUSION

To conclude, high index of suspicion and subsequent family screening is essential to diagnose corneal dystrophies such as GCD type 1 for early visual rehabilitation.

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