Int Ophthalmol (2019) 39:1599–1602
https://doi.org/10.1007/s10792-018-0971-6

PHOTO ESSAY

Granular corneal dystrophy: an enigma resolved

Pranita Sahay · Divya Agarwal · Prafulla K. Maharana · Jeewan S. Titiyal

Received: 25 February 2018 / Accepted: 16 June 2018 / Published online: 25 June 2018

Abstract
Purpose To report the intra-familial phenotypic variation of granular corneal dystrophy (GCD) across different age groups.
Method Two cases of GCD belonging to the same family (mother and daughter) were assessed and clinical findings were noted.
Result An 18-year-old female with complaint of glare, on examination showed brownish granules involving bowman's layer and superficial corneal stroma suggesting a diagnosis of Bowman layer dystrophy. Screening of her mother revealed multiple diffuse white granular opacities with snowflake appearance involving the central cornea. The intervening cornea was clear and limbus was not involved. Focal illumination showed deep stromal involvement. All these findings were typical of GCD. Genetic analysis revealed mutation of TGF beta-1 located on 5q31 which was consistent with our clinical diagnosis of GCD.
Conclusion Variable clinical presentation of GCD in different age groups can lead to diagnostic dilemma. Screening of family members can be helpful especially when dealing with early cases of GCD.

Keywords Granular corneal dystrophy · Corneal dystrophy · TGF beta 1 · 5q31

Manuscript

An 18-year-old female presented with complaints of glare in both eyes and UCVA of 6/6. On slit lamp examination under diffuse illumination, multiple discrete brownish granules arranged in a verticillate pattern involving the central cornea were observed (Fig. 1a). The opacities were scattered with clear intervening areas and a clear periphery. On focal illumination, opacities were noted to be sub-epithelial with no extension into deep stromal layer (Fig. 1b). Multiple hyper-reflective lesions involving the Bowman layer were noted on anterior segment optical coherence tomography (ASOCT) imaging (Cirrus HD-OCT, Carl Zeiss Meditec, Dublin, CA) (Fig. 2).

The superficial location of the lesion led to a provisional diagnosis of Bowman layer dystrophy but on screening the patient's mother, multiple diffuse white granular opacities with snowflake appearance involving the central cornea were observed on diffuse

Electronic supplementary material The online version of this article (https://doi.org/10.1007/s10792-018-0971-6) contains supplementary material, which is available to authorized users.

P. Sahay · D. Agarwal · P. K. Maharana (✉) · J. S. Titiyal
Dr. Rajendra Prasad Centre for Ophthalmic Sciences, All India Institute of Medical Sciences, New Delhi, India
e-mail: drpraful13@gmail.com

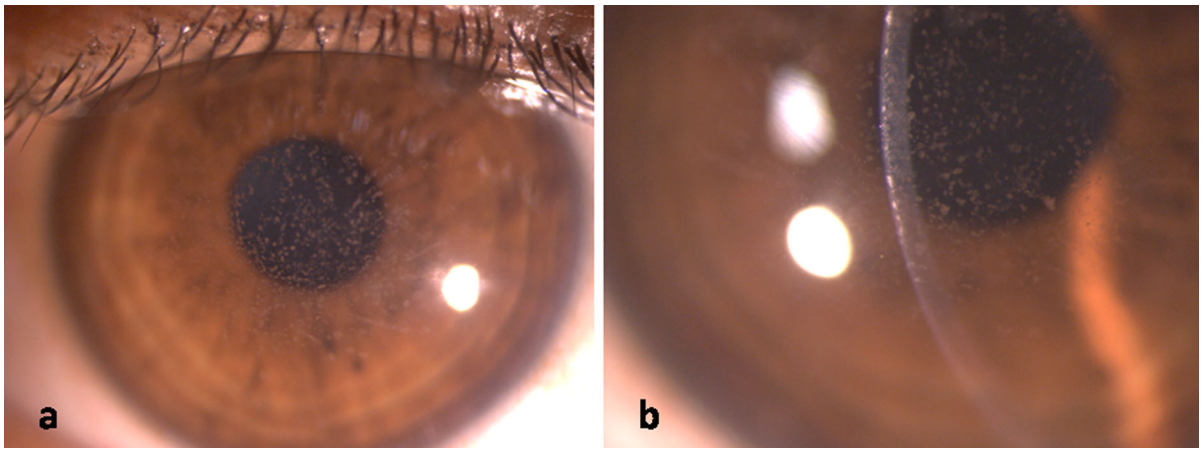

Fig. 1 **a** Slit lamp image in diffuse illumination showing multiple discrete brownish granules arranged in a verticillate pattern involving the central cornea; **b** slit lamp image in focal illumination of the same case showing sub-epithelial opacities with no extension into deep stromal layer

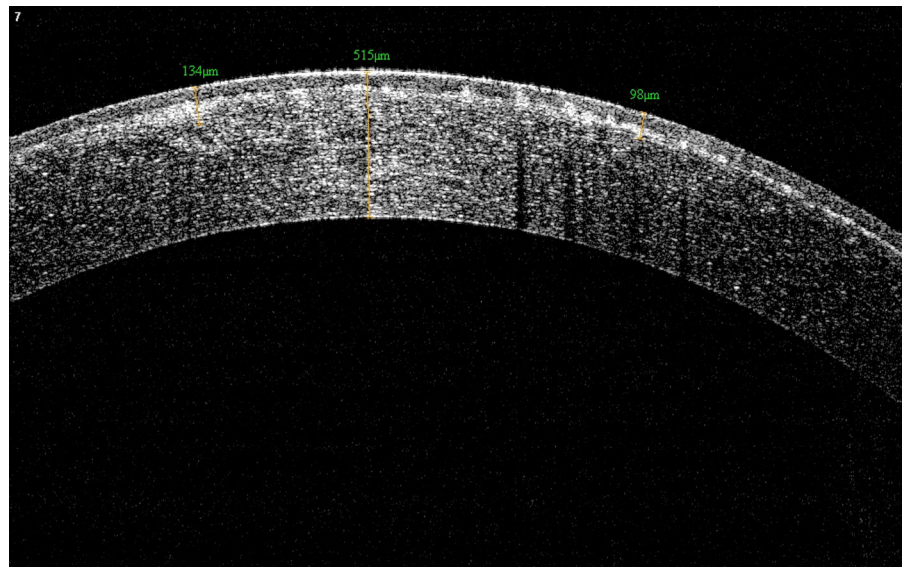

Fig. 2 ASOCT image of the case in Fig. 1 showing multiple hyper-reflective lesions involving the Bowman layer

slit lamp illumination (Fig. 3a). The intervening cornea was clear and limbus was not involved. Focal illumination showed deep stromal involvement (Fig. 3b). Multiple hyper-reflective lesions involving deep stromal layer and extending up to the descemet membrane were noted on ASOCT imaging (Fig. 4). All these findings suggested a clinical diagnosis of granular corneal dystrophy (GCD). Genetic testing of both daughter and mother was done. Sanger analysis, a technique for DNA sequencing, revealed mutation of TGF beta-1 located on 5q31.

Comment

This case highlights the variable presentation and appearance of GCD in different age groups which can lead to a diagnostic dilemma among clinicians, especially when dealing with cases of early GCD [1]. Clinical findings of the daughter's eye led to a dilemma, wherein a Bowman layer dystrophy was initially suspected. However, screening of her mother's eye helped us to come to a final diagnosis of GCD. This case therefore highlights the importance

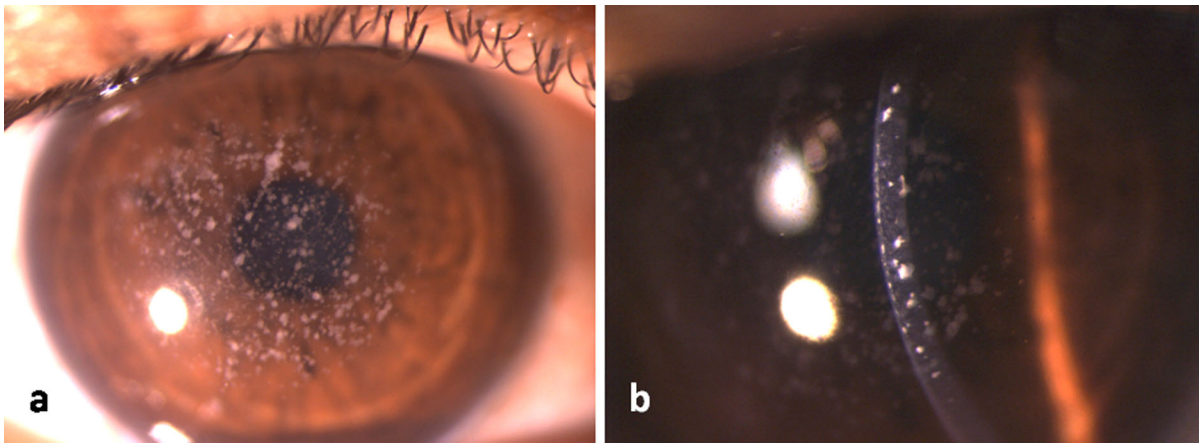

Fig. 3 **a** Slit lamp image in diffuse illumination showing multiple diffuse white granular opacities with snowflake appearance involving the central cornea; **b** slit lamp image in focal illumination of the same case showing granular opacities involving the deep stromal layer

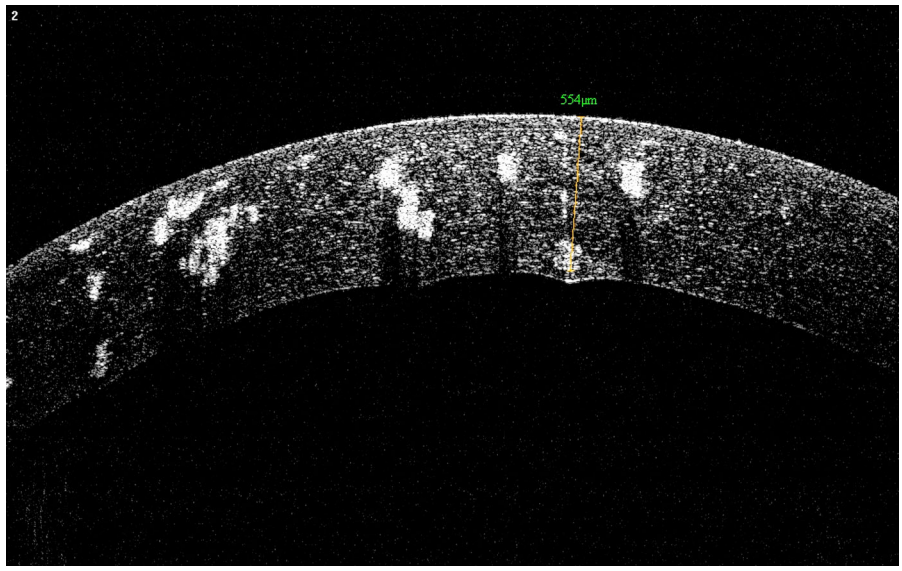

Fig. 4 ASOCT image of the case in Fig. 3 showing multiple hyper-reflective lesions involving deep stromal layer and extending up to the descemet membrane

of assessment of all family members in view of its autosomal dominant inheritance pattern [2].

Conclusion

Granular corneal dystrophy has variable presentation in different age groups. Screening of family members can help resolve the diagnostic dilemma when dealing with early cases of granular dystrophy.

Compliance with ethical standards

Conflict of interest There is no conflict of interest among the authors.

Ethical approval All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Declaration of Helsinki and its later amendments or comparable ethical standards.

Patient's consent Patient's consent was obtained for the purpose for publication.

References

1. Weiss JS, Møller HU, Aldave AJ et al (2015) IC3D classification of corneal dystrophies–edition 2. Cornea 34:117–159
2. Weiss JS, Møller HU, Lisch W et al (2008) The IC3D classification of the corneal dystrophies. Cornea 27(Suppl 2):S1–83