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Case Report

An Interesting Case of Granular Corneal Dystrophy Type 1

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Abstract

Corneal epithelial-stromal and stromal dystrophies are a group of inherited disorders of the cornea that are caused by progressive accumulation of deposits within the layers of the cornea. Most dystrophies previously considered stromal are now classified as either epithelial-stromal dystrophies or stromal dystrophies. These deposits are not caused by inflammation, infection, or trauma, but by genetic mutations that lead to transcription of aberrant proteins resulting in the accumulation of insoluble material within the cornea. Hereby author intends to report an interesting case of granular corneal dystrophy type 1.

Keywords:
Granular corneal dystrophy; Bread crump pattern; Epithelial stromal level

Introduction

A 20 yrs. old female came to eye opd for routine eye examination complained of progressive painless visual loss since last five years. Visual acuity in right eye was 6/12 and left eye was 6/9. She had never experienced redness, floaters, trauma, eye pain or watering eyes though occasional history of photophobia, glare and foreign body sensation was present. No history of wearing spectacles, allergy or other systemic illness could be elicited. She denied having long term medications or surgical history

Intraocular pressure and confrontation fields were normal. Visual fields revealed no abnormality. On SLE small, white, sharply demarcated deposits resembling crumps in central anterior stroma were noticed (OD>OS). The bread crump opacities were located at level of bowmans plane and 1/3 of corneal stromal depth in both eyes (Figure 1).

Overall pattern of deposition was disc shaped with limbal sparing pattern. The stroma between the opacities was clear. On direct illumination, the opacities appeared white, but on indirect illumination, they were composed of small translucent dots with vacuoles and a glassy splinter appearance (Figure 2).

Clinical Findings

Onset occurs early in life with crumb like opacities that may broaden into a disk like appearance as the patient reaches the teens. Initially the stroma between the opacities is clear. Gradual increase in number and size of deposits occurs with deeper and outward spread. Corneal sensation is impaired.

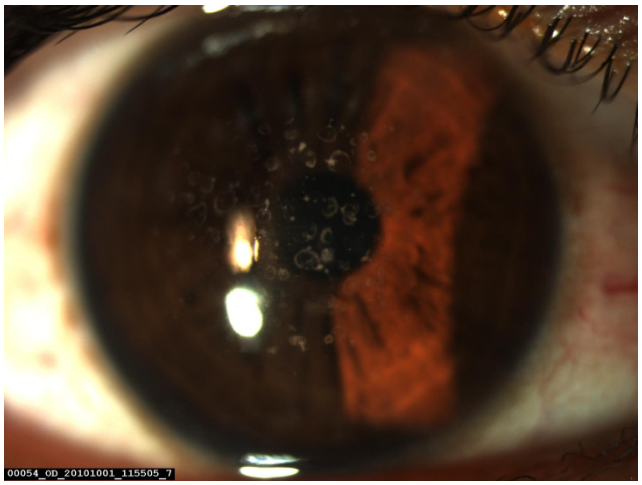

Figure 1: Small, white, sharply demarcated deposits resembling crumps in central anterior stroma OD

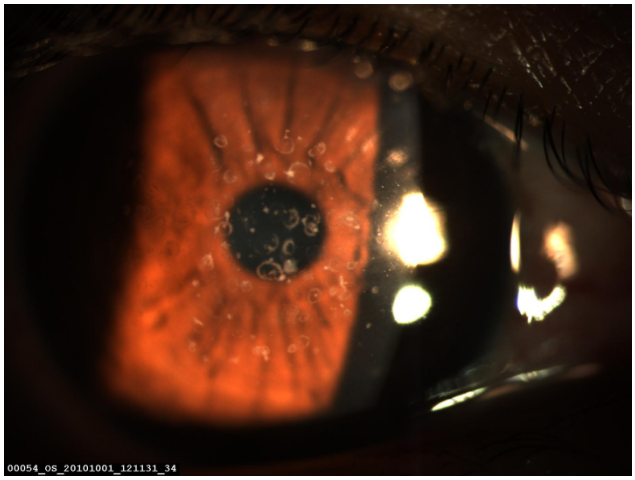

Figure 2: On direct illumination, the opacities appeared white, but on indirect illumination, they were composed of small translucent dots with vacuoles and a glassy splinter appearance.

The lesions do not extend to the limbus but can extend anteriorly through focal breaks in Bowman layer. Recurrent erosions occur and vision decreases as the opacities become more confluent.

The dystrophy is slowly progressive, with vision only rarely dropping to 20/200 after age 40. Patients complain of glare and photophobia.

Histopathologically the opacities are eosinophilia deposits often described as "rock candy like" in the anterior stroma made of a hyaline-like material. The hyaline material stains bright red with Masson trichrome stain.

The 2015 International Committee for Classification of Corneal Dystrophies (IC3D) classification system has divided corneal dystrophies into 4 categories: epithelial and sub epithelial dystrophies, epithelial-stromal dystrophies, stromal dystrophies, and endothelial dystrophies.

Epithelial-stromal corneal dystrophies

- Reis bucklers corneal dystrophy
- Thiel-behnke corneal dystrophy
- Lattice corneal dystrophy type 1 and variants
- Granular corneal dystrophy type 1
- Granular corneal dystrophy type 2

Stromal corneal dystrophies

- Macular corneal dystrophy
- Schnyder corneal dystrophy
- Congenital stromal corneal dystrophy
- Posterior amorphous corneal dystrophy
- Predescemet corneal dystrophy;

GCD is a Category 1, Stromal, TGFBI-associated corneal dystrophy. Although it is classified as a stromal dystrophy, research suggests the possibility that the granular opacities have an origination to the corneal epithelium with a migratory effect to the corneal stroma. When vision is significantly affected or recurrent corneal erosion occurs, despite first and second-line treatments, viable management options thereafter include photokeratectomy and other new surgical treatments such as femtosecond deep anterior lamellar keratoplasty and femtosecond laser-assisted keratoplasty. Future advancements in diagnostic technology, immunohistologic and genetic testing, medications, and surgery will allow for advancements in treating and managing patients with GCD.

Early in the disease process, no treatment is needed. Recurrent erosions may be treated with bandage contact lens and topical antibiotic (erythromycin ointment or 3rd-4th generation fluoroquinolone), therapeutic contact lenses, superficial keratectomy, or PTK. When visual acuity is affected, DALK or PK has a good prognosis. Recurrence in the graft (anteriorly and peripherally) may occur after many years as fine subepithelial opacities varying from the original presentation

Discussion

The light microscopic and TEM appearance and staining attributes of the corneal deposits in GCD are diagnostic. Eosinophilic lesions deposit in the cornea in GCD. The corneal opacities consist predominantly of an extracellular deposition of mutant transforming growth factor beta induced protein (TGFBIp), which stains a brilliant red with the Masson trichrome stain [1]. With the Wilder reticulin stain, the accumulations contain tangles of argyrophilic fibers. The deposits react with histochemical methods for protein as well as with antibodies to TGFBIp. The granules stain positively with luxol fast blue and are reported to stain positively with antibodies to microfibrillar protein.

By TEM characteristic electron dense, discrete, rod-shaped or trapezoid bodies are evident [2-4]. Cross-sectional profiles of the corneal deposits are usually irregularly-shaped, but sometimes hexagonal measuring 100–500 nm in diameter. Clusters of these elongated bodies occur particularly in the superficial corneal stroma and they may be present in the epithelial intercellular space or within degenerated basal epithelial cells [5-7]. Some rod-shaped structures appear homogeneous without a discernible inner structure; others, however, are composed of an orderly array of closely packed filaments (70–100 nm in width) orientated parallel to their long axis, while others appear moth-eaten with variable-shaped cavities containing fine filaments. Some superficial and most deep stromal deposits do not all possess the rod-shaped configuration. Descemet membrane and the corneal endothelium are unremarkable, and so is the cornea between the deposits.

Conclusion

Most dystrophies previously considered stromal are now classified as either epithelial-stromal dystrophies or stromal dystrophies. It has been observed that Granular opacities have an origination to the corneal epithelium with a migratory effect to the corneal stroma. The disorders may or may not affect vision and may or may not be symmetrical. Glare, photophobia or recurrent foreign body sensation must be evaluated meticulously

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