Correcting Crumby Vision: The Use of Scleral Lenses for Management of the Patient with Granular Corneal Dystrophy Type I

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Abstract

Purpose: To discuss the management of a young patient with granular corneal dystrophy type 1 (GCDI) using specialty scleral contact lenses. Additionally, to demonstrate how scleral contact lenses can be used to delay surgical procedures when managing corneal diseases.

Case Report: A 26-year-old Caucasian male presented to the clinic with chief complaints of glare, haze, intermittent dryness, monocular diplopia, and a history of GCDI. The patient presented with entering unaided acuities of 20/30- OD, 20/40 OS. Habitual scleral contact lenses yielded visual acuities of 20/25- OD, OS. A slit lamp examination revealed granular opacities located in the central anterior stroma, spanning the central 5-6mm of the cornea. The patient returned for a follow-up contact lens fitting to determine if scleral lenses would appropriately manage his condition.

Conclusions: Several considerations should be observed when managing a young patient with GCDI. Scleral lenses are a useful tool to manage aberrations, diplopia, and other visual sequelae of a granular corneal dystrophy diagnosis. Surgery, including phototherapeutic keratectomy (PTK) and deep anterior lamellar keratoplasty (DALK), should be considered with caution due to high rates of recurrence within surgical grafts. Therefore, scleral lenses should be used to correct visual function and comfort of symptoms related to corneal surface disease. The lack of visual acuity improvement, risk of recurrence, and limited repeatability after PTK or DALK procedures should inspire delay of surgery. For the young patient, scleral lenses should be the management of choice, delaying a surgical procedure and inevitable recurrence. This case report highlights clinical findings of granular corneal dystrophy type 1 with subsequent management and treatment considerations.

Key words: Granular corneal dystrophy type 1 (GCDI), Avellino dystrophy, breadcrumb dystrophy, scleral contact lenses, phototherapeutic keratectomy (PTK), deep anterior lamellar keratoplasty (DALK)

Introduction

GCDI is an autosomal dominant stromal dystrophy resulting from mutations in the TGFBI gene and is characterized by well demarcated breadcrumb shaped opacities in the central anterior stroma.[1] The dystrophy appears in adolescence or early adulthood and is accompanied by late diffuse corneal haze which causes reduced visual acuity, glare, and photophobia. As the condition progresses the hyaline deposits coalesce and run deeper into the stromal layer which causes decreased corneal sensitivity.[1] The depth of these deposits may be visualized using an optic section on the biomicroscope or anterior segment optical coherence tomography (OCT). The characteristic deposits may eventually cause painful corneal epithelial erosions. Initial treatment for the potential erosions includes the use of artificial tears and bandage contact lenses. Surgical methods used for the treatment of granular corneal dystrophy include phototherapeutic keratectomy (PTK) and deep anterior lamellar keratoplasty (DALK).[2] This case aims to discuss the use of scleral contact lenses to manage granular corneal dystrophy type 1.

Scleral contact lenses are designed to vault over the entire corneal surface, landing on the sclera. These lenses offer correction of visual symptoms caused by a variety of ocular surface diseases and corneal ectasias. A fluid reservoir forms between the lens and the cornea, smoothing surface irregularities, providing relief for those with ocular surface disease and allowing for healing of the ocular surface. Primary indications for use of scleral lenses includes ocular surfacer disease, corneal irregularity, and severe refractive error.

Case Report

A 26-year-old Caucasian male presented to the clinic for a Contact Lens Scleral Diagnostic exam. He arrived with chief complaints of glare and haze, intermittent dryness, monocular diplopia, and a history of GCDI. The patient reported that he had noticed his vision worsening since his last complete exam. The patient had no significant personal or family history, and no additional ocular history to contribute. The patient had not undergone any genetic testing. At the time, the patient was wearing scleral contact lenses from the previous year. The patient also reported working as a technician at a local ophthalmology practice. Before arriving to the clinic, he had been educated on the need for PTK or high-order aberration (HOA) correcting lenses from a corneal specialist. The patient was adamant about avoiding surgery yet had extremely specific standards for his visual performance. Initial treatment using wavefront guided scleral contact lenses had

been unsuccessful due to patient discomfort and dissatisfaction with glare reduction. The patient presented with entering unaided acuities of 20/30- OD, 20/40 OS. With his habitual contact lens correction, visual acuities were 20/25- OD and OS. Slit lamp examination revealed severe granular opacities located in the central anterior stroma. The breadcrumb shaped opacities spanned the central 5-6mm of the cornea, interfering with the visual axis (*See Figures 1&2)*.

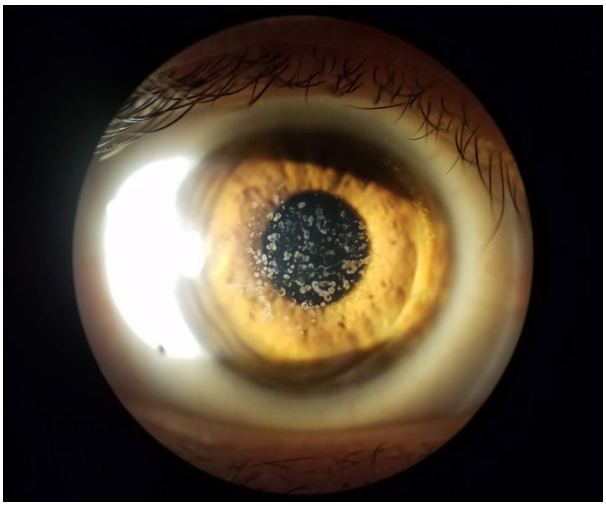

Figure 1. Photo of granular opacities spanning the central cornea, OD. Image Source: *Michigan College of Optometry*

Anterior segment OCT scans confirmed the presence of hyaline deposits in the anterior stromal layer of the cornea. Additionally, signs of dry eye disease and tear film irregularities were observed. The patient also had pingueculas temporally OU and nasally OD. His habitual scleral lenses featured no vaulting or notches to

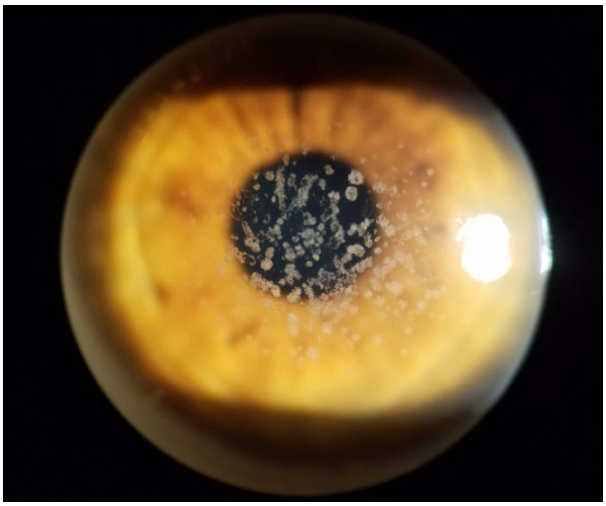

Figure 2. Photo of granular opacities spanning the central cornea, OS. Image Source: *Michigan College of Optometry*

accommodate for these conjunctival findings. Significant tear exchange was noted during the exam. When asked, the patient described removing his lenses generally twice per day to reduce lens fogging. Topographies were taken at this exam to aid in the customization of this scleral contact lens fitting (*See Figures 3&4*).

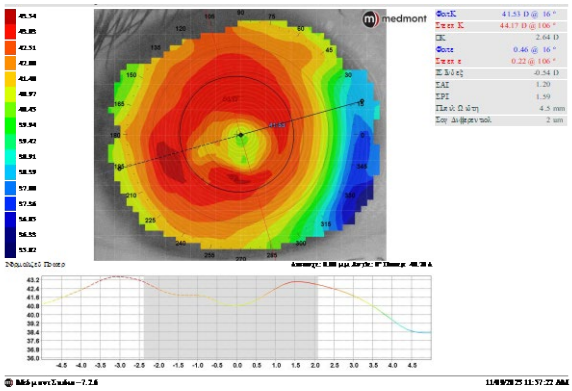

Figure 3. Axial power topography of the right eye showing slight tear film irregularity. However, data was able to be obtained. Image Source: *Michigan College of Optometry*

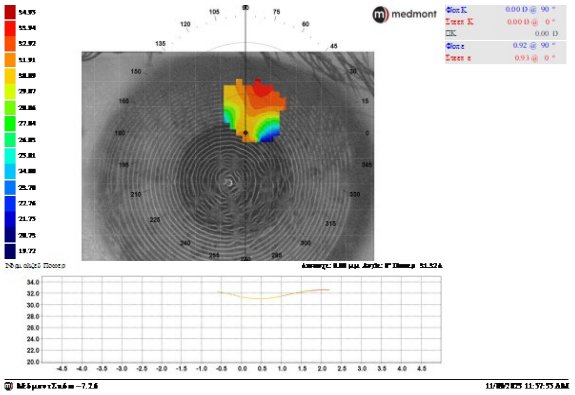

Figure 4. Axial power topography of the left eye showing severe tear film irregularities as sequelae to GCDI. Image Source: *Michigan College of Optometry*

The patient returned for a follow-up fitting and was fit in a 15.80 mm quadrant-specific scleral lens. The quadrant specific design allowed for correction of visual acuity to 20/20 OD and OS, improved landing on the pingueculas and provided patient comfort to treat symptoms of glare, haze, and dry eye disease. The patient returned for another follow-up with complaints of monocular diplopia. A larger optic zone diameter, 9.0 mm, was then ordered to alleviate his symptoms of monocular diplopia. The patient returned for a subsequent follow-up. He presented with a complaint of blur in both eyes, with the left eye being more aggravating. At this follow-up, his visual acuities were 20/25 OD and 20/25-2 OS. The patient was educated that disease progression had occurred and will likely continue. Additional patient education at this visit included the possibility of PTK or DALK in the future, but his best vision and comfort would be found using scleral contact lenses at this time. The patient returned for another follow-up. At this visit, the patient complained of discomfort in the superior area of his scleral lenses in the right eye. To address his concerns, a slit lamp examination was performed to determine if adjustments to the lens fit could be made. At the conclusion of this visit a new scleral lens was ordered. The quadrant-specific design allowed for the superior quadrants to be loosened without compromising the fitting relationship in the inferior zones and the landing vaults over his nasal and temporal pingueculas. A final follow-up was then scheduled. At this visit, the patient had no more complaints of discomfort with the scleral lenses. Visual acuities were found to be stable at 20/25 OD and 20/25- OS. The slit lamp examination revealed adequate fitting lenses in all quadrants for both eyes. It was determined that no further adjustments needed to be made because both doctor and patient goals had been met. The scleral contact lenses were finalized at this visit (*See Figure 5 & Table I.)* The patient was instructed to follow-up with his primary eyecare provider for his annual dilated fundus examination. An annual contact lens exam was also recommended. At this next exam, external photography will again be conducted to monitor the progression of his disease.

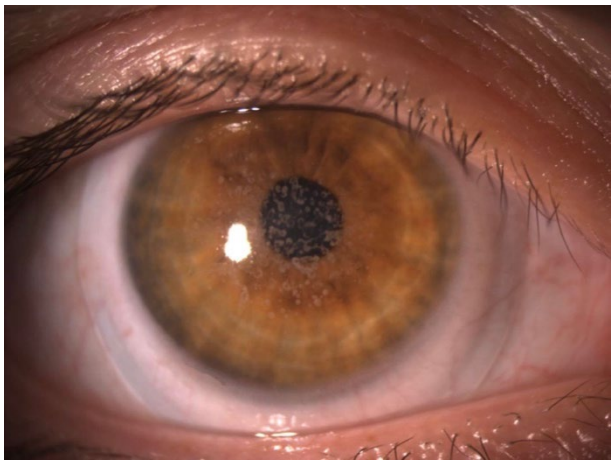

Figure 3. Photo of the finalized scleral contact lens on eye, OD.
Image Source: *Michigan College of Optometry*

Finalized Lens Data

Refractive Data			
	Subjective Refraction	**Visual Acuity**	**Manual Keratometry**
OD	+0.25 -1.25 x 115	20/20-2	42.75 x 020 43.62 x 110
OS	+0.75 -2.00 x 079	20/25-2	43.37 x 157 44.50 x 067

Finalized Lens Parameters		
	OD	**OS**
BC	8.23 mm	8.23 mm
Diameter	15.80 mm	15.80 mm
Optic Zone Diameter	9.0 mm	9.0 mm
Edge Vault Height	0.30 µm	0.30 µm
Edge Vault Width	30.0 µm	30 µm
Edge Vault Angle	45.0	130
Sagittal Depth	4.573 µm	4.520 µm
Power	+4.00 -1.25 x 120	+4.00 -1.00 x 050
Visual Acuity	20/20-	20/20-

Table I. Refractive data and finalized scleral lens parameters.

Discussion

Epidemiology: GCDI is inherited as an autosomal dominant trait with a 100% penetrance of the gene. [3] The stromal dystrophy is caused by a p.Arg555Trp mutation on chromosome 5q31in the *TGFBI* gene.[4] The incidence of GCDI is unknown; however, a Chinese study estimated the incidence to be 0.24% out of a 2,068-cohort study.[5]

Pathophysiology: GCDI is a bilateral, non-inflammatory condition. Lesions characteristic of GCDI appear as white, discrete, irregularly shaped, sharply demarcated spots in the anterior stroma.[4] The lesions consist of aggregated deposits of hyaline. Over time, the deposits tend to coalesce and become more numerous, impairing visual acuity around the fourth decade of life.[6] As the disease progresses, patients may experience recurrent corneal erosions as the corneal epithelial basement membrane becomes involved. Corneal surface irregularities may be confirmed on topographic imaging.

Differential Diagnoses: GCDI can resemble many corneal dystrophies. The first dystrophy to exclude should be granular corneal dystrophy type II (GCDII). GCDII differs from GCDI because it is characterized by hyaline deposits in the anterior stroma, like GCDI, in addition to amyloid deposits in the deeper stroma. The appearance of the opacities in GCDII will feature large and small breadcrumb opacities, in addition to ground glass shaped opacities. Localization of the

deposits using biomicroscopy is key to differentiating between GCDI and GCDII. Additional differentiating tests include histopathology using Masson trichrome staining and Congo red staining, as well as anterior segment optical coherence tomography which reveals the depth of the lesion(s) in question.[7]

Lattice corneal dystrophy, the most common stromal dystrophy, should be another differential diagnosis when caring for the patient with GCDI. Lattice corneal dystrophy is another inherited corneal disorder featuring corneal opacities that may lead to corneal erosions and tear film irregularities. However, the opacities featured in this disease are deposits of amyloid, rather than hyaline deposits like GCDI.[8] A diagnosis of lattice corneal dystrophy is also made clinically through a slit lamp examination. The main differentiating factor between GCDI and lattice corneal dystrophy is the shape and appearance of the corneal opacities seen during the slit lamp examination. Lattice corneal opacities appear like ground glass and span the central cornea, spreading toward the periphery.[9] Lattice corneal dystrophy may also be managed with scleral contact lenses and treated with a DALK or PTK.[10]

Corneal scars should be another consideration when central opacities are seen during a slit lamp examination. Corneal scars are commonly caused by injury, infection, or disease.[11] Corneal scars also appear as white opacities and can be located centrally or peripherally. A scar will have sharply demarcated borders and appear translucent under close observation. Like the other diagnoses, corneal scars are diagnosed via slit lamp examination and corneal topography scans.[11]

Management and Treatment:

Management of GCDI begins with the use of artificial tears and bandage contact lenses when corneal erosions occur. Due to the chromatic aberrations that occur with corneal opacities, patients with GCDI are often unable to function using spectacle correction. Corneal surface and tear film irregularities that occur with GCDI prevent these patients from safely and comfortably using soft contact lenses for correction. Therefore, scleral contact lenses and sometimes even high-order aberration correcting lenses are used to manage the patient with GCDI. Once the opacities decrease visual acuity, PTK surgery may be performed. PTK surgery aims to remove the opacities located in the anterior stroma.[12] DALK surgery may be considered with deep stromal deposits or when PTK is no longer repeatable.[13] Following either surgery, there is no statistical improvement in visual acuity for the patient with GCDI. Additionally, recurrence within the corneal graft material

is almost universal within four years of surgery.[14]

Conclusions: A diagnosis of GCDI typically leads to surgical referral for PTK or DALK procedures. However, scleral lenses with standard optics can correct for HOA resulting from corneal surface irregularities. Therefore, scleral lenses may be used to correct visual function and comfort of symptoms related to the corneal surface disease. Scleral contact lenses allowed this patient to overcome the symptoms of his disease and delay the need for surgery. Surgery does not statistically improve visual acuity, and recurrence within the corneal graft material is almost universal within four years of surgery. Therefore, delay of surgery is crucial to managing the young patient with corneal irregularities. The use of scleral contact lenses to delay corneal surgery is not limited to GCDI; rather, scleral lenses may delay corneal surgery for numerous corneal dystrophies, degenerations, ocular surface diseases, post-trauma, and even post-surgery. For the young patient, the use of scleral lenses for correction of granular corneal dystrophy should be the management of choice.

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