Treatment of Ocular Surface Disease, Secondary to Familial Dysautonomia, with Prosthetic Replacement of the Ocular Surface Ecosystem (PROSE) Devices.

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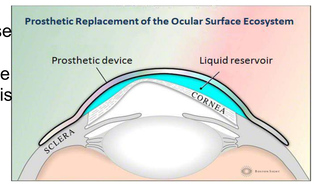
Background

The BostonSight® Prosthetic Replacement of the Ocular Surface Ecosystem (PROSE) treatment is a device created by the Boston Foundation for Sight (BFS) in Needham, MA. The device protects and supports the ocular surface, and can even promote the ocular surface to heal. Starting PROSE treatment is at first labor intensive and involves a number of examinations and follow-ups to customize each PROSE device to each individual patient. Patients must also be trained in the application and removal of the devices, as well as its care and cleaning. These devices have a multitude of uses for many conditions, can provide increased comfort, Improved vision, prevent disease progression and complications, and can significantly improve the quality of a patient's life. Below is an example of the successful management of an ocular condition with PROSE devices.



Case History

19 year old white female

Chief complaint: Blurry Vision.
- Patient denied any complaint of dry eye, irritation, or foreign body sensation
- Referred to BFS by PECP due to recent history of recurrent corneal ulcers OU

Ocular history:
- Neurotrophic Keratitis OU
- Keratoconjunctivitis Sicca OU
- History of corneal ulcers OU
- Corneal scarring OU
- Optic Neuropathy OU
- No previous contact lens wear

Medical history:
- Familial Dysautonomia
- GERD
- Sleep Apnea
- Hyeprtension
- Hypotension

Medications:
- Midodrine 2.5 mg PO QD
- Clonidine 0.1 mg PO QHS
- Fluorinef 0.1 mg PO QOD
- Benadryl 50 mg PO QHS
- Pepcid 20 mg PO QD

Ocular Medications:
- Lubrifresh ung Q2H OU

Allergies:
- NKDA

Pertinent Exam Findings

Entering VAs:
- Distance sc: 20/80, PH 20/70 OD; 20/125, PH 20/100 OS
- Near sc: J10 OD; J7 OS

Pertinent Exam Findings (cont'd)

OSDI Questionnaire:
- No symptoms of dryness reported by patient

Anterior Segment:
Lids/Lashes:
- Trace debris on lashes OU

Conjunctiva:
- Trace injection OU
- Oxford IV Staining OU

Cornea:
- OD: 1mm superficial stromal scar located inferonasally
- OS: 3mm round area of thinning/opacity inferonasally
- Oxford IV Staining OU **(Figures 1 and 2)**
- No epi defects or ulceration OU
- Absent TBUT OU

Anterior Chamber:
- Deep and quiet OU

Treatment and Management

- Initial - Frequent lubrication using PFATs and ung OU
  - Tried and failed due to non-compliance, Patient was neurotrophic and would forget to instill drops
  - Patient developed corneal ulcers while using PFATs and ung. Referred to BFS by PECP due to this
- PROSE Devices:
  Final Device parameters:
  OD: Vault 4.25, BC 7.88, Pwr -0.50, Diam= 18.50, 4 toric peripheral curves: 12:00= 0, 3:00= -0.30, 6:00= -0.35, 9:00= -0.50
  OS: Vault 4.10, BC 7.88, Pwr -1.25, Diam= 18.50, 4 toric peripheral curves: 12:00= 0, 3:00= -0.55, 6:00= -0.45, 9:00= -0.3
- BCVA with devices:
  - OD: 20/80
  - OS: 20/125
- The patient and aunt were successfully trained on the proper application and removal of the devices, and the devices were dispensed to the patient
- Follow-up with the patient was scheduled for three months after dispensing of the devices
- Patient was lost to follow-up until 1 year later

One Year After Start Treatment
- Patient returned for annual PROSE comprehensive
- Devices were comfortable and patient (and aunt) reported that there was no re-occurrence of any corneal ulcers after beginning PROSE treatment

Entering VAs:
- Distance cc: 20/125 OD; 20/125 OS (reduced)
- Referred to Neuro-ophthalmology

Anterior Segment:
Conjunctiva/Cornea:
- Trace injection OU
- Oxford II Staining OU - **Much improved**

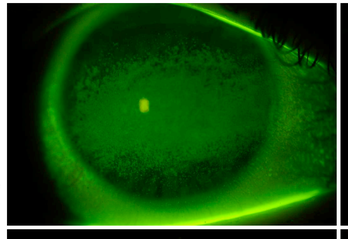


Fig 1: Right eye at initial exam.

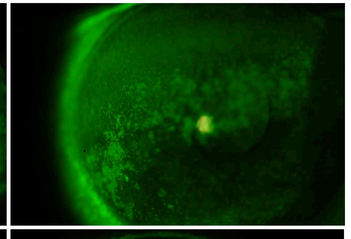


Fig 3: Right eye at 2 years.

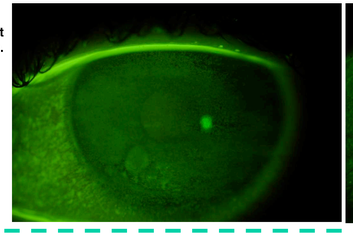


Fig 2: Left eye at initial exam.

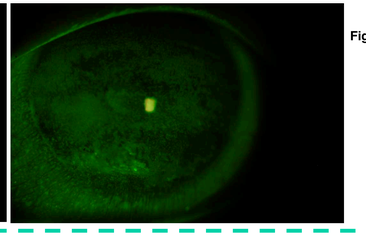


Fig 4: Left eye at 2 years.

Treatment and Management cont'd

Two Years After Start Treatment
- Patient returned again for annual PROSE comprehensive
- Devices were still comfortable and there was still no re-occurrence of any corneal ulcers

Entering VAs:
- Distance cc: 20/125 OD; 20/125 OS
- Patient self-reported that everything was "stable" per the neuro-ophthalmologist. A request for the patients records was submitted

Anterior Segment:
Conjunctiva:
- Trace injection OU
- Oxford II Staining OU - **Stable**
- Photos taken at this exam.

Three Years After Start Treatment – findings same as year previous

Discussion

- Familial Dysautonomia (FD), also known as Riley-Day syndrome, or HSAN III, is a hereditary sensory and autonomic neuropathy.[1–3] It is a rare genetic disorder that mainly affects people of Ashkenazi Jewish decent.[1–3]
- It is characterized by an incomplete development of sensory and autonomic neurons, resulting in a general sensory dysfunction coupled with variable autonomic dysfunction.[1]
- Of all the clinical manifestations of FD, its ocular manifestations are the most prominent. Two manifestations that always occur in FD are: the absence or severely reduced production of tears, and corneal anesthesia.[1,2]
- An additional ocular complication that is now being noted is a form of Optic Atrophy, and is being noted with increased frequency.[1–3]
- Systemic complications of FD can include: susceptibility to multiple bouts of Bronchopneumonia, Cyclical vomiting, GERD, postural hypotension, Pain insensitivity, absent deep tendon reflexes, paroxysmal hypertension, and emotional instability.[1]

Discussion cont'd

- Diagnosis is also often made during ophthalmologic investigation and is based upon a tetrad of ocular signs: Corneal anesthesia, exotropia, diminished or no tear flow, and hypersensitivity to topical methacholine.[1] Genetic based testing has also been developed.

Complications and successes:
- It was determined that all fitting changes were to be made purely objectively due to the corneal anesthesia preventing any subjective responses from the patient.
- FD patients are often non-compliant with their treatments as they usually have no complaint of dry eye or irritation. This often causes traditional dry eye treatment methods to be unsuccessful.
- Since the start of treatment, there was no re-occurrence of any corneal ulcers and the surface staining significantly improved.
- Although the punctate staining was reduced, it was not completely resolved as reported by other cases of scleral lens use in FD patients.[4,5] Further questioning of the patient and aunt revealed that the patient sleeps with her eyes open and uses a CPAP machine for sleep apnea.
- The patient was advised to start using a sleeping mask to help prevent any ocular surface drying while asleep.
- VAs were reduced 1 year after start of treatment, despite improved ocular surface health. FD associated optic atrophy was determined to be a possible cause for the reduced VA, and the patient was scheduled to have a neuro-ophthalmologic consult. Follow-up from Neuro-ophthalmology was self-reported as "stable" by the patient and her aunt. A request for the Neuro-ophthalmology notes was sent.

Conclusions

- Familial Dysautonomia (FD) is a genetic disorder that causes corneal anesthesia and absent tear production leading to severe ocular surface disease.
- Management of this ocular surface disease utilizing scleral lenses is becoming a more common alternative therapy for FD patients, and in many instances can help patients avoid serious and life altering complications, such as corneal ulcers and perforation.
- This specific patient showed improved support of the ocular surface with the use of PROSE device scleral lenses

Acknowledgements

Thank you to Dr. Chirag Patel, OD, FAAO and Dr. Anita Gulmiri, OD, FAAO for their guidance and support during my residency.

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