Learning to Drive with Congenital Hereditary Endothelial Dystrophy (CHED): A Pediatric Low Vision Case Report

ABSTRACT
Congenital hereditary endothelial dystrophy (CHED) is a rare condition often associated with congenital glaucoma. A young amblyope and talented artist who believed he would never be able to drive learns to use a bioptic telescope.

I. CASE HISTORY

Patient demographics
16-year-old Caucasian male; junior in high school; artist and musician

Chief complaint
Physician directed referral for a low vision evaluation; blurry vision at distance and near with 3-week-old progressive addition spectacle correction

Low vision goals
1. To be able to drive
2. To be able to read comfortably at home and in school

Ocular, medical history
Congenital hereditary endothelial dystrophy OU
Glaucoma suspect OU
Congenital nystagmus OU
Deprivation amblyopia OD>OS
Constant alternating esotropia OU

Surgical history
2009 DSEK OS
2010 DSEK OD, peripheral laser iridotomy OS
2015 PHACO with PCIOL OD, OS
2016 YAG capsulotomy OD

Medications
Cosopt BID OU since 2003
Durezol BID OU since 2016

Previous therapy
History of patching for 2 hours/day, each eye for 2 years post-DSEK

II. PERTINENT FINDINGS

Distance BCVA
OD: 20/80
OS: 20/50-
OU: 20/50

Near BCVA
OD: 0.4/1.2M
OS: 0.4/0.8M
OU: 0.4/0.8M
**Distance cover test**  15° CAET
**Near cover test**  25° CAET

**Stereo acuity**  No forms, alternating constant suppression

**Color vision**  Unable to accurately assess; abnormal findings OD, OS

**Extraocular motility**  FROM, no pain or diplopia OD, OS

**Confrontation visual field and Vision Disc**  FTFC OD, OS

**Pupil testing**  OD: 6mm fixed pupil
OS: 3.5mm in bright, 4.5mm in dim, peaked pupil

**Contrast sensitivity**  1.60 log CS OD, 1.68 log CS OS, 1.68 log CS OU

**Amsler grid**  0 degrees of central scotoma OD, OS

**Anterior Segment:**

**Intraocular Pressures**  9mmHg OD, OS, GAT @ 3:12PM

**Adnexa/lids/lashes**  WNL OD, OS

**Conjunctiva**  Trace papillae OD, OS

**Sclera**  White and quiet OD, OS

**Cornea**  Corneal graft clear and intact OD, OS

**Anterior chamber**  4+ nasal/temp Van Herrick, no cells or flare OD, OS

**Iris/pupil**  OD: Blue iris, fixed pupil
OS: Blue iris, peaked pupil, patent peripheral iridotomy 11 o’clock

**Crystalline lens**  OD: PCIOL centered, s/p YAG capsulotomy with trace fibrotic PCO
OS: PCIOL centered and clear

**Anterior vitreous**  Clear OD, OS

**Low Vision Device Evaluation:**

**Distance**  Ocutech VES-Sport 4X BiOptic TS OS  20/20

**Near**  Optelec PowerMag+ 3.5X/10D HHM with Bright White LED  0.3/0.5M

**Filters**  Eschenbach SmartLux Digital Electronic Video Magnifier  0.4/0.5M

**Lighting**  No significant improvement reported

**Direct incandescent lighting for near tasks**

**III. DIFFERENTIAL DIAGNOSIS**

**Primary**  Congenital hereditary endothelial dystrophy (CHED2)

**Others**
1. Congenital hereditary endothelial dystrophy (CHED1)
2. Congenital glaucoma
3. Posterior polymorphous corneal dystrophy
4. Congenital syphilis
5. Trauma
IV. DIAGNOSIS AND DISCUSSION

- Congenital hereditary endothelial dystrophy (CHED) is a rare corneal dystrophy characterized by bilateral diffuse clouding of both corneas
  - It is caused by a mutation in the SLC4A11 gene (chromosome 20 loci 20p13) that encodes a sodium borate transporter needed in the proper growth and terminal differentiation of neural crest cells during development
  - Abnormal corneal endothelial cells develop and fluid build-up occurs leading to corneal opacities
  - Other structures of the eye are usually normal
  - CHED may be associated with hearing loss, also known as Harboyan syndrome
  - Two variants of CHED have been described:
    - The autosomal dominant variant (CHED1) presents after 1 year of age and is progressive; these patients are born with clear corneas and develop clouding over the first few years of life; they complain of epiphora and photophobia, but do not have nystagmus
    - The autosomal recessive variant (CHED2) is stationary; corneal clouding is present at birth and is accompanied by nystagmus; visual acuity is worse than in CHED1
    - There is controversy over whether or not CHED1 is a distinct entity as there are genetic and histopathological overlaps with posterior polymorphous corneal dystrophy (PPCD)

- Congenital glaucoma can produce corneal clouding from infancy
  - Patients with CHED often have higher intraocular pressures secondary to thicker than normal pachymetry measurements
  - There is evidence of concomitant cases of CHED and glaucoma
  - Signs of congenital glaucoma include megalocornea, corneal haze, increased cup-to-disc ratio, Haab’s striae, and buphthalmos

V. TREATMENT AND MANAGEMENT

- Surgical intervention, such as penetrating keratoplasty (PKP) or Descemet’s stripping endothelial keratoplasty (DESK) is usually required in patients with CHED
  - Prognosis and final visual outcomes depends on the degree of corneal clouding, the level of amblyopia, and the success of the surgery

- In order to address the patient’s goals for driving and reading, a bioptic monocular telescope worn over the left eye and an electronic video magnifier were recommended
- Bioptic training involves localization, focusing, spotting, tracing, tracking, and scanning
  - Activities include Hart Chart column jumping, Hart Chart saccades, wall saccades, hallway spotting, Marsden ball, and descriptive driving

- Vision therapy was also recommended to improve the patient’s binocularity for driving
- The patient returned for six follow-up sessions, including one fitting, one dispense, and four training sessions with vision therapy
- A referral was made for a bioptic driver’s permit and behind-the-wheel training by a Certified Driver Trainer (CDT)
VI. CONCLUSION

- Corneal clouding from congenital hereditary endothelial dystrophy can cause amblyopia of varying severity
- Many young patients are eager to overcome the limitations of low vision and become licensed drivers as getting a driver’s license is a rite of passage for most teenagers
- Bioptic telescope systems (BTS) are great devices for young patients with 20/200 vision or better in either eye and with near normal horizontal fields for driving and use in the classroom
- Patients with nystagmus may benefit from a bioptic telescope with a larger exit pupil and wider field of view
- Bioptic telescopes should be fitted in front of the dominant, non-deviating eye of strabismic patients, or in front of the better seeing eye if the patient alternately tropes
- Strabismic patients with anomalous retinal correspondence are better able to detect objects in the ring scotoma produced by the telescope than those with normal correspondence and adapt by suppressing the deviating eye

VII. BIBLIOGRAPHY