The Minority Report: Pediatric Low Vision Management of Cone-Rod Dystrophy

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Abstract: Congenital retinal disorders may cause severe vision loss in childhood. Although low vision aids and services are widely accepted to increase the quality of life of visually impaired children, their benefits require further in-depth research.

I. Case History

- **Patient demographics**
  - 14 year old black male, currently in 9th grade.

- **Chief complaint**
  - He presented for a low vision exam with a history of mobility training and use of adaptive technology. He initially presented to the UEC at age 3 with complaints of extreme photophobia and nystagmus, first noticed by a kindergarten teacher. Due to his age and his parents’ limited English and understanding of his visual difficulties, it was difficult to assess his visual symptoms further at that time.
  - His main complaints included difficulty with near tasks such as reading small/regular print, mail, and handwritten materials. However, he was able to read large headlines and the computer at a close working distance.

- **Ocular, medical history**
  - An unknown congenital retinal condition in both eyes since 3 years old.
  - No allergies to medication and no systemic conditions.

- **Other salient information**
  - Parents were first cousins who immigrated to the United States from Ethiopia.
  - His younger sister was previously seen at UEC with profoundly reduced vision to hand-motion in both eyes. She knew Braille. His older twin sisters were normally-sighted.
  - The patient did not know Braille and was not interested in pursuing it.
  - As he had grown up with reduced vision at an early age, he had fully accepted his vision loss and was highly motivated to not let it hinder his future.
  - Current Low Vision Aids included:
    - Mouse-operated closed Circuit television (CCTV) at home. Device recently broke.
    - Desktop closed Circuit television (CCTV) provided by school.
    - Plano wrap-around sunglasses.
    - Patient has used single vision glasses with transitions in the past but did not find them helpful.

II. Pertinent findings

- **Clinical findings**
  - VA was stable to vision from previous visits in both eyes:
    - **Right Eye**:
      - Distance VA: 2/40M (ETDRS chart, Snellen Equivalent 20/400)
      - Near VA: 2.0M@5cm
    - **Left Eye**:
      - Distance VA: 6/200ft (Feinbloom chart, Snellen Equivalent 20/667); Near VA: unable to perform
    - Predicted ADD is +20D
  - Retinoscopy findings:
    - OD: +5.50-3.00x090 (No improvement to vision)
    - OS: +2.50-0.50x090 (No improvement to vision)
  - Trial Frame Refraction: No improvement to vision in either eye
  - Pupils: Equal, round, reactive to light, no APD
Extraocular motility: right jerky nystagmus in primary and extreme gazes horizontally; worse in left gaze OU

Confrontational Visual Field: poor fixation, appears restricted 360 OD, OS to visual axis to finger wiggling

- **Demonstration of Low Vision Devices:**
  - Portable CCTV:
    - Near VA: 0.50M with reverse contrast and 10x magnification at 20cm; reads fluently and efficiently until 1.0M, then slower reading speed until 0.50M.
  - Desktop CCTV:
    - Near VA: 0.50M with reverse contrast and 40x magnification at 26cm; reads fluently and efficiently.
  - Patient demonstrated ease of use and adequate motor manipulation of both devices. He was able to use both devices satisfactorily but reported more comfort with desktop device.

- **Physical findings**
  - Anterior Segment: unremarkable OU
  - Posterior Segment:
    - Optic Nerve: 0.35R OD; 0.40R OS
    - Macula:
      - OD: 3DD flat, round macular scar with hypo and hyperpigmentation
      - OS: >4DD hypopigmentation with hyper-reflective sheen
    - Periphery: Attached 360 OU
  - Macular Optical Coherence Tomography
    - OD: loss of photoreceptor integrity line 1-2DD around the macula, overall thinning of retinal layers with progression compared to previous visit, loss of foveal contour
    - OS: loss of photoreceptor integrity line; disruption of all retinal layers
  - Fundus Auto Fluorescent Imaging
    - OU: focalized central region of hyporeflective area at macula with surrounding ring of hyper-reflectivity; denser OD versus OS

III. Differential diagnosis
- **Primary/leading**
  - Cone-Rod Dystrophy
- **Others**
  - Leber’s congenital amaurosis
  - Achromatopsia or rod monochromatopsia
  - Retinitis pigmentosa (rod cone dystrophy)
  - Stargardt disease

IV. Diagnosis and discussion
- **Elaborate on the condition**
  - Visual impairment in children is rare: 0.2 per 1,000 children in developed areas versus 1.0 per 1,000 children in impoverished areas. Regional differences account for differing prevalence of eye disorders in children from study to study.
  - Cone-rod dystrophy has a prevalence of 1/40,000. The clinical course is rapid and severe, affects cone function prior to rod function, and leads to early legal blindness and disability. Symptoms include: decreased visual acuity, color vision defects, light sensitivity, decreased sensitivity in the central field, progressive loss of peripheral vision, and progressive night blindness.
  - Diagnosis requires integration of clinical history, fundus examination, and ERG.
  - Patients with cone-rod dystrophy are usually severely visually disabled or legally blind by the end of the second decade of life.

V. Treatment, management
• **Treatment and management of patient**
  o Retinal consult within the UEC to determine progression, conduct ERG, and referral to Columbia University for genetic testing. Patient’s mother reported this process was started 5 years ago but was discontinued before results were known.
  o Continue wrap-around sunglasses for glare control and protection. A spectacle prescription was not released as there was no increased visual improvement and transition/tint provided inadequate glare control.
  o Continue use of desktop CCTV at school and at home, and requested coverage of replacement CCTV for home use by the NYS Commission for the Blind.
  o Return to Low Vision Clinic in 1 year for re-assessment of functional needs. The recommended evaluation period is 6 months for young children, even if low vision aids were rejected at initial visit; the recommended evaluation period is 1 year for older children and in times of increased magnification needs or changes in school institutions or situations.iii

• **Low Vision Aids (LVA) with visually impaired children**
  o Visually impaired children with poorer unmagnified reading acuity and central field defects more frequently use LVAs. Those enrolled in special schools are also more likely to have worse reading acuity and use LVAs.v
    - Near LVAs are twice as likely to be prescribed compared to distance LVAs because near vision is crucial to success in school.
    - More than half of visually impaired children can read 12-point font without LVAs. Many children are able to decrease their working distance and utilize relative distance magnification along with accommodation.iv
    - Unfortunately, many are not prescribed appropriate LVAs, have not had a LVA within a year, and/or experience difficulties with LVAs that could be easily remedied by proper recommendations and discussion with low vision specialists.v The type of LVA prescribed to children should factor in the relationships between LVA, child, and task.vi
  o Older children more frequently use LVAs.v The age at which the symptoms first appear/change also guide recommendations of LVAs.
    - Children may benefit from stand magnifiers as early as 3.5 years with training.vii
    - LVAs should be considered early before children start fearing the use of such devices as a stigma.viii
    - The need for LVAs increases with age as the size of print encountered and amplitude of accommodation decreases. In addition, some ocular conditions are progressive, further requiring increased magnification.ix
  o Additional, non-visual disabilities can impact the recommendations of LVAs.
    - Other sensory, developmental, or intellectual disabilities, as well as co-morbid systemic conditions may present simultaneously with impaired vision. iv,viii,x
    - According to the Metropolitan Atlanta Developmental Disabilities Surveillance Program, the majority of children with visual impairment also had one or more developmental disabilities. The more severe the vision impairment, the greater associated risk of an additional disability.xi
  o A Cochrane Review suggests that no current literature adequately studies the effect of LVA with children with visually impairments.xii
  o Another Cochrane Review also suggests no adequate studies exist on the effects of adaptive technology on children’s reading speed and accuracy.xiii

VI. Conclusion
  • Because the incidence of low vision increases with the aging population due to more common diseases such as macular degeneration, glaucoma, and cataracts, most of the current literature focuses on the benefits of LVA for adults. Although LVAs are widely accepted advantageous to visually impaired children, there is little research available on the benefits of LVAs for children.
• Children and adults are not developmentally or emotionally similar. Visually impaired children may also have perceptual delays, poor fine motor skills, or have other developmental delays. These impact the successful use of LVA. To extrapolate the use of low vision devices from research with adults does not adequately address these issues.
• Therefore, increased research into LVA as used with children is necessary to construct a guideline of prescription recommendations of optical and electronic devices. Increased research is also necessary to document the specific types of benefits LVA provide (i.e. increased reading speed or increased comprehension scores.)

VII. References