2017 Low Vision Conference: Students with Progressive Vision Loss
May 11, 2017
Austin, TX
Progressive Hereditary Conditions

Presented by
David Lewerenz, OD, FAAO
david.lewerenz@ucdenver.edu

Developed for
Texas School for the Blind & Visually Impaired
Outreach Programs
Progressive Hereditary Conditions

Topics We'll Cover

- Genetics
- Electrodiagnostics
- Specific conditions
  1. Batten Disease
  2. Best Disease
  3. Cone Dystrophy (progressive CD, not to include achromatopsia)
  4. Cone-Rod Dystrophy
  5. Glaucoma in Children
  6. Retinitis Pigmentosa
  7. Stargardt Disease

- Common Rehabilitation Options
- Other / Wrap-Up

Genetics
**Autosomal Dominant (AD) Inheritance**

Only one dominant gene required for inheritance. *No gender difference.*

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- 1 heterozygous affected parent
- 1 homozygous non-affected parent
- 50% chance of affected child

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- 2 heterozygous affected parents
- 75% chance of affected child
Autosomal Dominant Inheritance

![Autosomal Dominant Inheritance Diagram]

Figure 3 In an autosomal dominant disorder, the mutated gene is a dominant gene located on one of the nonsex chromosomes (autosomes). You need only one mutated gene to be affected by this type of disorder. A person with an autosomal dominant disorder — in this case, the father — has a 50 percent chance of having an affected child with one mutated gene (dominant gene) and a 50 percent chance of having an unaffected child with two normal genes (recessive genes).

(Mayo Clinic web site)

Autosomal Recessive (AR) Inheritance

Two recessive genes required for inheritance. **No gender difference.**

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- 2 heterozygous non-affected "carrier" parents
- 25% chance of affected child
- 50% chance of carrier child
• 2 homozygous affected parents
• 100% chance of affected child

Autosomal Recessive Inheritance

Figure 4 To have an autosomal recessive disorder, you inherit two mutated genes, one from each parent. These disorders are usually passed on by two carriers. Their health is rarely affected, but they have one mutated gene (recessive gene) and one normal gene (dominant gene) for the condition. Two carriers have a 25 percent chance of having an unaffected child with two normal genes (left), a 50 percent chance of having an unaffected child who also is a carrier (middle), and a 25 percent chance of having an affected child with two recessive genes (right).

(Mayo Clinic web site)
X-Linked Recessive (XLR) Inheritance

- Passed on through sex chromosomes rather than autosomes
- Recessive gene on X chromosome is expressed because it is unopposed on the Y chromosome
- **Carrier females, affected sons**

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<td>Y</td>
<td>XY</td>
<td>X^M Y</td>
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</table>

- Heterozygous non-affected "carrier" mother and unaffected father
- 25% chance of affected son
- 25% chance of carrier daughter
- 25% chance of unaffected son
- 25% chance of unaffected daughter

**Figure 5 Sex Chromosomes X and Y**
X-Linked Recessive Inheritance

- Passed on through sex chromosomes rather than autosomes
- Recessive gene on X chromosome is expressed because it is unopposed on the Y chromosome
- **Carrier females, affected sons**

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<th>Mother→</th>
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</tr>
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<td>XX</td>
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</tr>
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<td>XY</td>
<td>X⁰MY</td>
</tr>
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- Heterozygous non-affected "carrier" mother and unaffected father
- 25% chance of affected son
- 25% chance of carrier daughter
- 25% chance of unaffected son
- 25% chance of unaffected daughter

Figure 6 Sex chromosomes X and Y
X-Linked Recessive Inheritance

Figure 7 Women can pass down X-linked recessive disorders. A woman who is a carrier of an X-linked recessive disorder has a 25 percent chance of having an unaffected son, a 25 percent chance of having an affected son, a 25 percent chance of having an unaffected daughter and a 25 percent chance of having a daughter who also is a carrier.

(Mayo Clinic web site)

Electrodiagnostics

Figure 8 Strand of DNA
Electroretinogram (ERG)

- Diffuse, biphasic response of the entire retina from light stimulation
  - \textit{a-wave} is the initial response of photoreceptor hyperpolarization
  - \textit{b-wave} is the secondary response of the Müller and bipolar cells
- A diffuse "Ganzfeld" light bowl is the stimulus used
- Corneal (contact lens) electrode, ground electrode placed on earlobe and reference electrode on skin near the eye

![Corneal electrode](image)

\textbf{Figure 9 Corneal electrode}

![a-wave and b-wave](image)

\textbf{Figure 10 a-wave and b-wave}
ERG

- Scotopic (Rod) ERG can be obtained following 20 minutes of dark adaptation
- Photopic (Cone) ERG can be obtained by
  - Light adaptation for 10 minutes and background light in the Ganzfeld
  - Flicker stimulus of 30 Hz
    - Rods drop out at about 20 Hz
- ERG is evaluated for
  - Amplitude of the a- and b-waves
  - Latency between light stimulus and retinal response
  - Implicit time = time it takes the wave to reach peak response

mfERG

- 61 or 103 points in the central 40-50 degrees of the visual field are stimulated with a discrete light and the response is recorded
- Much better for evaluating macular function
  - Macular loss alone will not show up in a regular ERG because such a small portion of the retina is affected

Figure 11 3-dimensional topographic plot
Electro-Oculogram (EOG)

- Measures the difference in electrical potential between the front of the eye and the back of the eye
- The layer of the retina chiefly responsible is the retinal pigment epithelium
- Electrodes placed on the skin of the medial and lateral canthi and a ground electrode on the forehead
- Measurements taken as the eyes are moved back and forth horizontally under scotopic and photopic conditions

Figure 12 Image of a woman wearing an EOG device.

EOG

- Primary measure is the *Arden Ratio*, which is the ratio of the highest amplitude in light and the lowest amplitude in the dark
- Normal Arden Ratio is >1.5
- Most helpful in diagnosing Best disease, and sometimes choroideremia

Figure 13 Image of a retina
Visually Evoked Potential (VEP)

- A specialized EEG where the remaining EEG signal is filtered out from that created by the visual stimulus
- Evaluates the entire visual system, from the retina to the occipital cortex
- Electrodes are placed on the occipital scalp, near the inion, with a ground electrode on the forehead or earlobe

Figure 14 Two images showing the placement of electrodes on the head for a VEP.

Progressive Hereditary Conditions

Figure 15 A strand of DNA

Progressive Hereditary Conditions

1. Glaucoma in Children
2. Cone Dystrophy
3. Cone-Rod Dystrophy
4. Retinitis Pigmentosa
5. Stargardt Disease
6. Best Disease
7. Batten Disease
Glaucoma in Children

- Glaucoma can occur in children and is classified as:
  - Primary Congenital / Infantile Glaucoma (PCG)
    - "Congenital" onset birth to 2 months
    - "Infantile" onset 3 months to 3 years
    - "Juvenile" onset childhood to early adulthood
  - Can be unilateral (20-30%) or bilateral (70-80%)
  - Photophobia, excessive tearing
  - Descemet's breaks / corneal edema
  - Variable VA and VF loss
  - ~1 in 10,000 in US
  - Vision loss can be severe or even total NLP

Figure 16 Image of the eye with Glaucoma.
Glaucoma in Children

- Buphthalmos (large eye) & megalocornea (large cornea) if < 2yo
- Most often sporadic: "Congenital" usually AR, "Juvenile" usually AD
- Most important (of many) genes are CYP1B1 and LTBP2 (AR)
- Congenital trabecular meshwork dysgenesis usually AR
- Iris-TM posterior embryotoxin anomalies, such as Axenfeld-Rieger, are usually AD & develop in children or young adults

Figure 17 Image of a child with buphthalmos (large eye).

Figure 18 Image of a child with megalocornea (large cornea).
Cone Dystrophy (Progressive)

- Overall progressive decline of cone function throughout the retina
  - Not limited to the macula/fovea
  - Blurred distinction between cone d. and cone-rod d. because there is some collateral rod damage when cones deteriorate
  - Cone-rod d. may resemble cone d. in early stages
  - In cone dystrophy photopic ERG is abnormal and scotopic ERG is normal
  - Vision loss varies greatly, but usually results in worse than 20/200 eventually

Figure 19 Contrast with achromatopsia.

Cone Dystrophy ERG

Figure 20 Ten ERG graph images shown in pairs left and right with the headings Cone Dystrophy on the left and Normal on the right. Along the side of each pair of images are the titles: Scotopic (Rods), Max, 30 Hz Flicker (cones), Photopic (cones), and Pattern ERG Cones. From Taylor and Hoyt, Pediatric Ophthalmology and Strabismus, 2005.
Cone Dystrophy (Progressive)

- Develops in childhood or early adulthood
  - Usually no nystagmus
- Affects about 1 in 30,000 people
- Retina appears normal early
  - Bull's eye maculopathy late
  - Loss of foveal reflex
  - Foveal atrophy
- In some cases there can be a glistening green appearance to the retina

![Image showing the green glistening appearance to the retina.](image)

**Cone Dystrophy Inheritance**

- Color vision defect (usually red-green) will sometimes precede loss of visual acuity
- Peripheral visual fields are normal
- Inheritance can be variable
  - Autosomal dominant – GUCA1A gene
  - Autosomal recessive – RDH5 gene
  - X-Linked recessive – COD2 gene
- It's not known why mutations in some of these genes, which encode proteins in both rods and cones, affect cones only
- There is no family history in many cases
**Cone-Rod Dystrophy**

- Cones affected early, rods affected later
- Affects about 1 in 40,000 people
- Symptoms
  - Reduced visual acuity
  - Photophobia
  - Reduced color vision
  - Later – Reduced night vision
  - Later – Reduced visual field
- There's a wide variety of expression, from mild to very severe

**Cone-Rod Dystrophy**

- Signs
  - Retina can appear normal early in the disease
  - Macular degeneration, sometimes bulls eye maculopathy
  - Attenuation of retinal arterioles
  - Pigmentary degeneration in some cases
  - Usually no nystagmus
- Early ERG profoundly reduced in cones and moderately reduced in rods
  - Rod ERG affected more later
Cone-Rod Dystrophy

Figure 22 Retinal images of moderate Cone-Rod Dystrophy. From Taylor and Hoyt, *Pediatric Ophthalmology and Strabismus*, 2005.

Figure 23 Image of advanced Cone-Rod Dystrophy. From Taylor and Hoyt, *Pediatric Ophthalmology and Strabismus*, 2005.

Cone-Rod Dystrophy

- Inheritance can be variable
  - Autosomal dominant – CRS, GYCY2D genes
  - Autosomal recessive – ABCA4, CERKL genes
  - X-Linked recessive – RPGR, CACNA1F genes
- Many genes involved in more than one disorder
  - The ABCA4 gene (autosomal recessive) is also linked to Stargardt disease, cone dystrophy and retinitis pigmentosa
Retinitis Pigmentosa (RP)

- Family of diseases with dozens of genes implicated
- 1 in 4,000 (1 in 1,878 in Navajo)
- Multiple inheritance patterns: AD, AR, XLR, digenic, sporadic
- Syndromal forms often affect cones = rods
- ERG abnormal or extinguished, rods > cones
- Attenuated vessels, optic atrophy, CME, cataracts

![Retinal image of Retinitis Pigmentosa (RP)](image)

Retinitis Pigmentosa

- Symptoms
  - Night blindness
  - Reduced dark adaptation
  - Mid-peripheral visual field loss
  - Reduced peripheral visual field
  - Loss of central vision late in the disease
  - Some are functionally blind by age 30, most are legally blind by age 60
Retinitis Pigmentosa

- Associated clinical signs
  - Pigmented "bone spicule" pattern degeneration, mid-periphery first
  - Attenuated vasculature – may appear relatively early
  - Cystoid macular edema – ~38% of patients with RP on OCT
    - Differences between regular CME and that seen with RP
    - Cataracts – especially PSC, 35-51% of adults with RP have cataracts

Figure 25 cystoid macular edema showing the large ‘cyst-like’ collections of fluid in the retina.

Figure 26 Eye with cataracts
RP and the ERG

• ERG can show presence of RP earlier than other findings
• When RP is suspected due to family history, if ERG is normal at age 6, it is unlikely that patient will develop RP
• Scotopic ERG is lost so early that cone ERG is often used in studies

Figure 27 Woman looking into an ERG device.

Figure 28 Drawing of how an ERG works.

Retinitis Pigmentosa

• Inheritance modes
  • Autosomal dominant – expected to be least severe
    • 30% of cases
  • Autosomal recessive – expected moderate severity
    • 20% of cases
  • X-Linked recessive – expected to be most severe
    • 15% of cases
  • Digenic – rare, two different mutations combine
Retinitis Pigmentosa

- ~30% of patients with RP have no known history of it in their family
- Simplex - one family member w/o family history
- Multiplex - >1 family member w/o family history

Figure 30 Retina of a patient with RP.

Figure 31 Image of a building depicted as it would be seen by someone with RP.
RP Syndromes

- Usher's syndrome (AR) = RP + deafness is one of many associated syndromes, 3 main subtypes
  - About 1 in 25,000 in US
  - Type 1 (1B, 1C, 1D, 1E, 1F, 1G) – severe hearing loss at birth
  - Type 2 (2A, 2C, 2D) – less severe hearing loss
  - Type 3 (3A, 3B) – progressive hearing loss/vestibular loss
- Bardet-Biedl syndrome (AR, 1 in 100,000; 1 in 18,000 in Canada)
  - RP with polydactyly or short/stubby digits, obesity, learning disability, hypogonadism in males, renal abnormalities
- Refsum disease (AR, PHYH or PEX7 genes)
  - RP with loss of sense of smell, hand/feet abnormalities, muscle weakness, ataxia, heart problems
- Bassen-Kornzweig Syndrome – RP + prog. neuro problems, abnormal RBCs
- Alström Syndrome – RP + obesity, hearing loss, diabetes, heart problems
- NARP Syndrome – Neuropathy, Ataxia, and RP
**RP and Vitamin A**

Vitamin A slows down ERG decline

Is vitamin E directly harmful or does it prevent absorption or utilization of vitamin A?

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<th>↓ERG</th>
<th>All 601 subjects</th>
<th>354 high amplitude</th>
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<td>6.1%</td>
<td>8.3%</td>
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<tr>
<td>A+E</td>
<td>6.3%</td>
<td>8.8%</td>
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<tr>
<td>Trace</td>
<td>7.1%</td>
<td>10.0%</td>
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<tr>
<td>E</td>
<td>7.9%</td>
<td>11.8%</td>
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VF shows same trend as ERG, but not statistically significant

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<th>↓VF</th>
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<th>354 high amplitude</th>
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Retinitis Pigmentosa

- Berson 1993 study conclusion - "It is recommended that most adult patients with the common forms of RP take a supplement of vitamin A, 15,000 IU/d…and avoid the use of high dose supplements of vitamin E."

- Controversies - Editorial in same issue has 3 concerns
  - No significant preservation of VF or VA
  - Role of "noise" in low amplitude ERGs and other concerns about ERG interpretation
  - Risk/benefit ratio of high dose vitamin A

Retinitis Pigmentosa

- Analyzed dietary questionnaires of 357 adults from 3 previous trials of who took 15,000 IU vitamin A for 4-6 years
  - Those with high (≥0.20 gram/day) omega-3 consumption from a diet high in oily fish showed
  - 40% slower rate of decline in visual acuity
  - Nearly 50% slower rate of decline in central visual field
  - Is there something synergistic about vitamin A and consumption of oily fish?

Retinitis Pigmentosa

• How much fish does one have to eat to average 0.20 gram/day of omega-3?
  • Fresh salmon, herring, blue fin tuna or sardines: 1 three-ounce serving per week
  • Canned salmon, anchovies, mackerel, swordfish: 1½ three-ounce servings per week


Figure 32 A platter of salmon.

Figure 33 A plate with fish and asparagus.
Retinitis Pigmentosa

- Bottom line concerning nutrition?
  - If vitamin A is recommended, use 15,000 IU/d of vitamin A palmitate, not beta carotene
  - Avoid vitamin E
  - Eat oily fish at least twice per week
  - AMD AREDS formulas are not appropriate for RP, yet many with RP use them
  - Annual blood tests for liver function and vitamin A levels recommended
  - No use in women who are or might become pregnant
  - Take body size into account in dosage?

Figure 34 A box of medicine (PreserVision) with a large red "X" across it.
Famous People with RP

Figure 35 Steve Wynn, Wynn Hotels, Stephen A. Wynn Institute for Vision Research.

Figure 36 Willie Brown, Mayor of San Francisco.
**Stargardt Disease**

- "Fundus flavimaculatus"
- Develops ~7-15 yo, often goes from 20/40 to 20/200 in ~ 5 year period
- Pisciform flecks to "beaten bronze"
- Eventual bilateral visual acuity loss, often to 20/200 or worse
- Central scotoma begins relative, then absolute
- Color vision is normal early in disease, red-green loss later
- Dark choroid due to lipofuscin is diagnostic on FA
- ERG: flash normal is often normal, especially early

![Figure 37 Retinal images showing "fundus flavimaculatus".](image1)

**Figure 37 Retinal images showing "fundus flavimaculatus".**

![Figure 38 Karl Stargardt (Germany) described the disease in 1909, at the age of 34. He lived to be only 52.](image2)

**Figure 38 Karl Stargardt (Germany) described the disease in 1909, at the age of 34. He lived to be only 52.**
Stargardt Disease

- Often there is loss of visual acuity in both eyes during teens
  - Sometimes there are no visible changes in the retina when vision loss begins, resulting in accusation of malingering
  - Visual acuity often declines from 20/40 to 20/100 in about 5 years and often stabilizes at about 20/200
  - No nystagmus, since later onset
- Can develop irregular scotoma with foveal sparing
- The most common inherited macular degeneration – about 1 in 8,000 to 10,000 people worldwide

Stargardt Disease

- Two conditions or one?
  1. Fundus flavimaculatis: White-yellow irregular flecks scattered throughout the retina
  2. Atrophy of macula: Slightly oval bulls-eye pattern, later may resemble "beaten bronze" appearance and later still pigmentary degeneration
    - These often occur together, but can have either or both
    - Disagreement in the past about classification into two disorders or one
    - Genetic basis appears to be the same

Figure 39 Retinal image showing atrophy of the macula in Stargardt Disease. From Basic and Clinical Science Course: Retina and Vitreous, AAO, 2008
Stargardt Disease

Figure 40 Four retinal images, moving from top left clockwise: Early pigment mottling, "snail slime" macula + flecks, fundus flavimaculatus, and bulls eye maculopathy. From Kanski and Bowling, Clinical Ophthalmology: A systematic approach, 7th ed., 2011
Stargardt Disease

- Caused by lipofuscin accumulation in retina pigment epithelium
- "Dark choroid" on fluorescein angiography from lipofuscin
  - Present in about 62% of Stargardt cases
  - Brightness of the choroid background is masked by lipofuscin

Figure 41 Reginal image. From *Basic and Clinical Science Course: Retina and Vitreous*, AAO, 2008

Figure 42 Dark choroid on fluorescein angiography from lipofuscin. From *Basic and Clinical Science Course: Retina and Vitreous*, AAO, 2008

Figure 43 Lipofuscin. From *Basic and Clinical Science Course: Retina and Vitreous*, AAO, 2008
Stargardt Disease

- Usually autosomal recessive
- ABCA4 gene
  - Also implicated in autosomal recessive forms of cone-rod dystrophy and retinitis pigmentosa
- Rarely autosomal dominant, ELOVL4 gene
- Advanced Cell Technologies is performing clinical trial w/ stem cells

Figure 44 Retinal image. From Yanoff and Duker, Ophthalmology, 2008.
Best Disease

- AKA "vitelliform dystrophy" (vitelliform = "resembling egg yolk")
- A primary disturbance of the RPE
- Autosomal dominant - VMD2 (aka BEST1) gene
- Classic egg yoke due to accumulation of lipofuscin in retina
- Variable presentation, from age 4 to 15
- Variable penetrance, even in same family
- VA loss is usually mild to moderate
  - 88% will have at least one eye ≥ 20/40
  - Only 4% will have visual acuity ≤ 20/200

Figure 45 Retina as seen with Best Disease with vitelliform dystrophy.
Best Disease

- About 20% may develop choroidal neovascularization of the macula ("Stage VI"), with much greater loss of vision
- EOG Arden (light/dark) ratio is reduced to ≤1.5
- Sometimes EOG is normal
- Full-field ERG usually normal
- Foveal (multifocal) ERG is usually reduced centrally
- Often hyperopic
- Adult onset is "adult vitelliform maculopathy"
  - Looks similar but different gene locus

Figure 46 choroidal neovascularization of the macula from Basic and Clinical Science Course: Retina and Vitreous, AAO, 2008
Best Disease

- Prevalence 1 to 9 in 100,000
- Stages I through VI

Figure 47 Six images of the retina beginning with top left Vitelliform Stage II, top right Blocked Choroidal background, middle left Material in RPE, middle right Multifocal disease, bottom left Pseudohypopyon III and bottom right Vitelliruptive stage IV, the “scrambled egg”. From Kanski and Bowling, *Clinical Ophthalmology: A systematic approach*, 7th ed., 2011
Batten Disease

• Caused by a mutation in the CLN3 gene, on chromosome 16
  • CLN3 involved in lysosome function
  • Lysozomes are organelles in cells which digest and dispose of waste
  • Cells have buildup of lipofuscin - Neuronal Ceroid Lipofuscinosis (NCL)
  • Neurons seem to be affected more than other tissues
• There are at least eight forms of NCL, including
  • Infantile NCL – Early (first year onset) and late (onset age 2 to 4) subtypes
  • Juvenile NCL (JNCL) – Commonly called Batten Disease (onset 4 to 8 years)
  • Adult NCL – Onset often about 30 years old
• Typically autosomal recessive inheritance
  • Each form caused by a different gene
Batten Disease

- Onset of JNCL (Batten) about age 4 to 7 years old, with progressive
  - Visual impairment
  - Speech difficulties
  - Movement disorders
  - Intellectual disability
- Often initially misdiagnosed as
  - Autism
  - Seizure disorder
  - Epilepsy
  - Many other conditions

Figure 48 A young boy and girl with Batten Disease sit in wheelchairs and play with balloons.
**Batten Disease**

- Vision loss
  - Gradual, often the first symptom of the disease
  - Usually begins age 4 to 7, severe vision loss eventually
  - Complete retinal degeneration with optic nerve atrophy
  - Sometimes bulls eye maculopathy

- Other signs and symptoms
  - Progressive encephalopathy
  - Cognitive and motor decline
  - Blindness
  - Life expectancy 15-35 years

*Figure 49 Images of two retinas showing bulls eye maculopathy*
Batten Disease

- Treatments of NCLs – No cure at this time
- Enzyme replacement therapy
  - BMN-190 by BioMarin
  - NtBuHA
- Stem cell therapy
- Gene therapy
  - Virus vector
- Immunosuppression
  - CellCept
- Symptomatic relief – E.g. anti-seizure drugs

Figure 50 Dr. Frederick Batten 1865-1918
Common Rehabilitation Options

Magnification for Near

- Reading glasses or regular glasses with bifocal
- Optical magnifiers: Hand, stand, illuminated, non-illuminated
- Electronic magnification: Portable or desktop

Figure 51 Strand of DNA

Figure 52 A boy uses reading glasses.

Figure 53 Three images showing various optical magnifiers.

Figure 54 Two images of electronic magnifiers: on the left a portable magnifier and on the right a desktop magnifier.
Magnification for Distance

- Hand held telescope
- Spectacle mounted telescope
- Some electronic options

Figure 55 A young woman uses a hand held telescope.

Figure 56 A boy uses spectacle mounted telescope.

Figure 57 A classroom scene with a student using an iPad to magnify math problems the teacher is showing on the board.
Text to Speech

- Spectacle mounted
- Phone and tablet apps
- Portable and desktop readers

Figure 58 Spectacle mounted device.

Figure 59 Smart phone with app.

Figure 60 Portable and desktop computers used for converting text to speech.
**Tactile**

- Traditional Braille
- Refreshable / electronic Braille

![Figure 61 Finger moves along paper braille page.](image1)

![Figure 62 Child using refreshable braille device.](image2)

![Figure 63 Refreshable braille device.](image3)
Technological Interfaces

- Magnification
- Speech / Audio
- Voice recognition
- Electronic Braille

Figure 64 Computer using a magnification software.

Figure 65 Electronic braille devise attached to a computer.

Figure 66 Computer screen with JAWS.
Poor Night Vision (Nyctalopia)

- Premium flashlights from LED Lenser
- P7R, ~$140
  - 1,000 lumens / LED
  - Battery life 2-40 hours
  - Weight 7.4 oz., Length 6.5"
  - Rechargeable with regulated intensity
  - Adjustable beam
- F1R, ~$140
  - Not adjustable beam, but smaller (4.5")
  - 1,000 lumens / LED
- H14R.2, ~$140
  - 1,000 lumens head lamp
Glare Management

• Filters

• Visors

Figure 70 Four pictures of glasses with different colored filters.

Figure 71 Woman wearing a visor.
Peripheral Visual Field Loss

- Visual field loss
- Field can be expanded with use of minus lens or reverse telescope
- Can be hand held or spectacle mounted
- Minification also occurs

Figure 72 Two pictures of telescopes. The one on the left is mounted on the spectacle and the other is hand-held.
Mobility, Training, Education

- Low vision specialist – OD or MD
- Certified Orientation and Mobility Specialist
- Certified Low Vision Therapist
- Certified Vision Rehabilitation Therapist
- Teacher of the Visually Impaired
- Assistive Technology

Figure 73 A young woman travels with a cane during an orientation and mobility lesson with her instructor.

Wrap-Up

Figure 74 Strand of DNA
## Relative Prevalence

<table>
<thead>
<tr>
<th>Disorder</th>
<th>1 in</th>
<th>USA</th>
<th>Texas</th>
</tr>
</thead>
<tbody>
<tr>
<td>Retinitis Pigmentosa</td>
<td>4,000</td>
<td>80,000</td>
<td>7,600</td>
</tr>
<tr>
<td>Stargardt Disease</td>
<td>9,000</td>
<td>35,000</td>
<td>3,400</td>
</tr>
<tr>
<td>Glaucoma in Children</td>
<td>10,000</td>
<td>32,000</td>
<td>3,000</td>
</tr>
<tr>
<td>Best Disease</td>
<td>20,000</td>
<td>16,000</td>
<td>1,500</td>
</tr>
<tr>
<td>Cone Dystrophy</td>
<td>30,000</td>
<td>11,000</td>
<td>1,000</td>
</tr>
<tr>
<td>Cone-Rod Dystrophy</td>
<td>40,000</td>
<td>8,000</td>
<td>750</td>
</tr>
<tr>
<td>NCL Disorders (all forms)*</td>
<td>100,000</td>
<td>3,200</td>
<td>300</td>
</tr>
</tbody>
</table>

## Resources

- For Clinical Trials - [https://clinicaltrials.gov/](https://clinicaltrials.gov/)
- Orphanet - [http://www.orpha.net/consor/cgi-bin/index.php](http://www.orpha.net/consor/cgi-bin/index.php)
- National Organization for Rare Disorders - [https://rarediseases.org/](https://rarediseases.org/)
- Batten Disease Support and Research Organization - [http://bdsra.org/](http://bdsra.org/)
- Beyond Batten Disease Foundation - [http://beyondbatten.org/](http://beyondbatten.org/)
- Foundation Fighting Blindness - [http://www.blindness.org/](http://www.blindness.org/)
Thanks for your attention!

David Lewerenz, OD, FAAO david.lewerenz@ucdenver.edu
Texas School for the Blind & Visually Impaired Outreach Programs

Figure 63 TSBVI logo.

Figure 64 IDEA logo