The Congenital Low Vision Patient- A Complete Picture

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As optometrists, we have the great opportunity to assist people with vision impairment through low vision services. The need for optometrists to provide low vision care is growing. There are two basic categories of low vision patients: those with congenital vision impairment and those with acquired vision impairment. The most prevalent congenital causes, low vision exam basics and illustrative case examples, will be discussed here. Having an understanding of the congenital disorders causing vision impairment, and the basics of a low vision evaluation, will allow more optometrists to participate in low vision rehabilitation.

Causes of Congenital Vision Impairment

There is a vast array of disorders responsible for congenital vision impairment. The most common include retinopathy of prematurity, optic nerve hypoplasia, albinism, aniridia and achromatopsia. To best serve any low vision patient, it is important to understand the disease and the symptoms the patient is experiencing. This is especially critical for children with congenital low vision. For them to be successful academically and maintain their workload effectively and efficiently, they need the right tools, environmental adaptations and a broad support system.

Retinopathy of Prematurity (ROP)

Retinopathy of Prematurity occurs exclusively in premature infants, particularly those who weigh less than 3 pounds, 5 ounces at birth and those born before 28 weeks' gestation

At approximately16 week's gestation, retinal blood vessels begin to bud from the center of the retina near the optic nerve. By 38-40 weeks gestation, the retinal blood vessels have reached the periphery. When an infant is born prematurely, this process is incomplete. ROP occurs when the development of the retinal vasculature is arrested and then proceeds abnormally with disorganized retinal vessel branching and anomalous interconnections.

In approximately 90% of affected infants, ROP spontaneously involutes. But within the other 10% of patients, ROP can lead to bleeding, scarring of the retina, retinal detachment and vision loss. Oftentimes, ROP babies need laser treatments in the peripheral retina to minimize the risk for late retinal detachment. Many children with ROP have varying levels of reduced VA in one or both eyes and the reduction in acuity can range from mild loss to total blindness. Even in cases in which ROP changes cease or regress spontaneously, affected children may have an increased risk of myopia, strabismus, amblyopia and/or future retinal detachment.

ROP is a leading cause of visual impairment and blindness in infants in many industrialized countries. In the 1940s and 1950s, premature babies were given too high concentrations of supplemental oxygen. Since then, oxygen blood levels have been more carefully monitored. In recent years, we have seen an increase in ROP due to advances in modern healthcare because more low birth weight infants are able to survive. In the United States, the incidence of ROP increased from 14.70% in 2000 to 19.88% in 2012. This trend is inversely related to a simultaneous decline in newborn mortality and substantiates the fact that supplementary oxygen alone is not the cause of ROP development. In the US, approximately 1100 -1500 infants per year develop ROP that is severe enough to require surgical intervention and 400-600 become legally blind.

Optic Nerve Hypoplasia (ONH)

Optic nerve hypoplasia is a congenital disorder condition that results in underdevelopment of the optic nerves**.** This occurs during the second month of pregnancy when the optic stalk develops and the nerves fail to develop fully. Most cases of ONH have no clearly identifiable cause. There are no known racial or socioeconomic factors in the development of ONH, nor is there a known association with exposure to pesticides. Optic nerve hypoplasia is thought to affect males and females in equal numbers. Optic nerve hypoplasia is approximately found in 1 in 10,000 children.

Optic nerve hypoplasia is present at birth but many symptoms may not appear until childhood or adolescence. Optic nerve hypoplasia is generally stable and non-progressive. Most people with optic nerve hypoplasia have nystagmus and vision can range from good functional vision or even full vision in one eye to no light perception.

Children with optic nerve hypoplasia may have brain malformations and pituitary problems. Abnormalities of structures of the brain may include hypoplasia of the corpus callosum, underdeveloped white matter in any other location, and cortical heterotopia. The common association of absence of the septum pellucidum has no known functional consequence, and may occur with or without other brain malformations. The hypothalamus is also frequently abnormal.

Some affected children have learning disabilities and developmental delays while others have normal intelligence. Deficiencies of certain hormones may result in slowed growth and/or poor development and may be life-threatening without treatment. Hormone deficiencies can be controlled with daily hormone replacement therapy and close monitoring by an endocrinologist.

In the United States, optic nerve hypoplasia is the third most prevalent cause of vision impairment and the most likely to cause legal blindness in children age three years or younger.

Figure 1: Optic nerve hypoplasia

Albinism

Albinism is a rare genetic disorder in which melanin pigment is reduced or absent in the eyes, skin and/or hair. Disruption in the tyrosinase/melanin biochemical cascade results in decreased melanin production. Since melanin is essential for the full development of the retina, decreased or absent melanin results in reduced visual acuity between 20/50- 20/800. There are two major types of albinism: Oculocutaneous albinism (OCA) and Ocular Albinism (OA). Worldwide albinism is found in 1 in 5,000 to 1 in 40,000. In the US, it is estimated to be 1 in 17,000.

Oculocutaneous Albinism (OCA)

Oculocutaneous albinism (OCA) is group of autosomal recessive disorders occurring in both males and females. OCA is characterized by a reduction or complete lack of melanin pigment in the skin, hair and eyes. This is caused by mutations in enzymes or membrane proteins that contribute to melanin synthesis. Traditionally oculocutaneous albinism was classified into two broad categories based on phenotype: tyrosinase positive (mild/moderate) and tyrosinase negative (severe). The phenotype depends on whether or not residual enzyme activity is present. If tyrosinase is present but has reduced activity, the ability to acquire pigmentation of skin and hair is possible. If tyrosinase has no activity, affected persons will have white skin and hair their entire life. With advances in molecular genetics, oculocutaneous albinism classification has been refined by genotype into seven different subtypes. In addition, syndromes with systemic manifestations have also been delineated.

Visual changes in oculocutaneous albinism include pendular nystagmus, iris transillumination, photophobia, foveal hypoplasia, abnormal decussation of the visual pathway, strabismus, loss of stereopsis and refractive error. Visual acuity can range from 20/60 to 20/400 and is best in individuals with more pigment. All individuals with OCA have the above visual changes but the amount of skin, hair and iris pigment can vary depending on the gene and mutation involved.

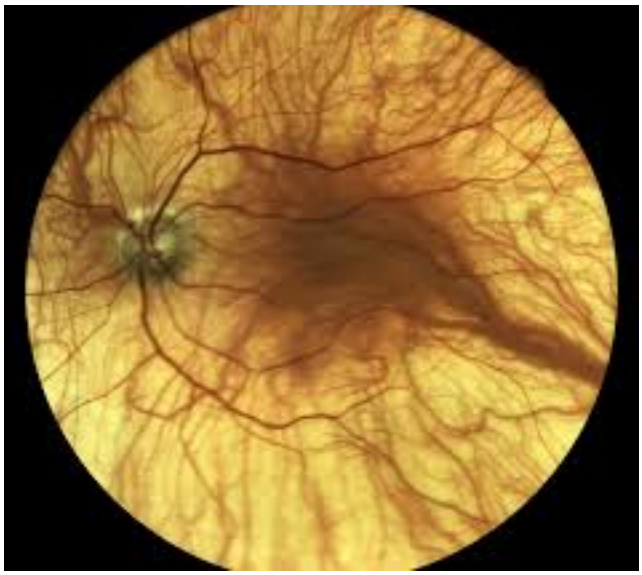

Figure 2: Oculocutaneous albinism

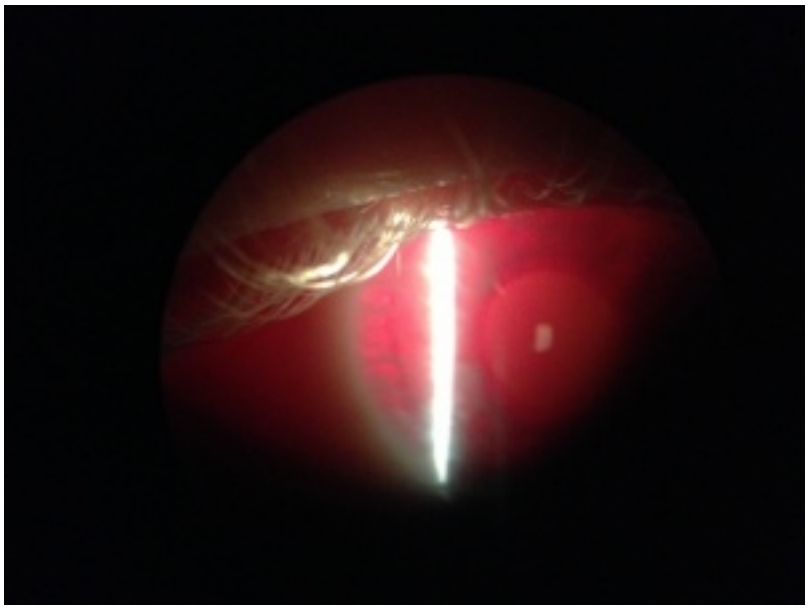

Figure 3: Iris transillumination

Ocular Albinism (OA)

Ocular albinism is an X-linked recessive form of albinism, which primarily affects the eyes unlike oculocutaneous albinism, which affects the skin, hair, and eyes. In a person with ocular albinism, the skin and hair may be only slightly less pigmented than unaffected siblings. Since the inheritance pattern of ocular albinism is X-linked, the disorder primarily affects men. Females may be gene carriers of the condition.

There are three categories of ocular albinism: Ocular albinism type 1 (OA1), Ocular albinism type 2 (OA2) and Ocular albinism with sensorineural deafness.

Ocular albinism type 1 is the most common form of ocular albinism. OA1 is inherited as an X-linked recessive genetic condition caused by mutations in the G protein-coupled receptor 143 gene. Vision impairment is present at birth and does not become more severe over time. Affected individuals have normal skin and hair pigmentation. They often have nystagmus, reduced iris and retinal pigment and photophobia. Ocular albinism has been reported to be one male in 20,000 births.

Ocular albinism with sensorineural deafness is a condition that includes the vision abnormalities of ocular albinism as well as deafness and balance problems. Some affected individuals have different colored eyes and a white forelock of hair. Ocular albinism with sensorineural deafness is inherited as an autosomal dominant genetic condition.

Achromatopsia

Achromatopsia is a congenital hereditary disorder characterized by partial or total absence of color vision. It results from mutations in one of several genes: CNGA3, CNGB3, GNAT2, PDE6C and PDE6H. These gene mutations prevent cones from functioning properly thereby affecting phototransduction. In persons with complete achromatopsia, cones are missing or nonfunctional and therefore have a total lack of color vision and severe visual acuity impairment. In persons with incomplete achromatopsia, some cone function is retained and therefore these people have limited color vision and less severe visual acuity loss. Some people with achromatopsia do not have mutations in any of the known genes. In these individuals, the cause of the disorder is unknown.

There are two primary forms of achromatopsia. Rod monochromatism and blue cone monochromatism. In both forms, there is impaired color vision, reduced visual acuity, nystagmus, photophobia and hemeralopia (day blindness). Achromatopsia is present 1 in 33,000 to 1 in 50,000 births.

Rod Monochromatism

Rod monochromatism is an autosomal recessive condition and the most common form of achromatopsia . It occurs in both males and females. Rod monochromats have more severe vision loss ranging from 20/120- 20/400, more color vision loss and greater light sensitivity. They can be complete with no color vision or incomplete with traces of color vision.

Blue cone monochromatism

Blue cone monochromatism is an X-linked recessive disorder and is a form of incomplete achromatopsia with blue cone function retained. Blue cone monochromats have less profound vision loss typically ranging from 20/60-20/200 and may retain some color vision in the blue and yellow spectrums. Blue cone monochromatism is seen to occur in 1 in 50,000 to 1 in 100,000 in males and could be as rare as 1 in 10 billion in females.

Aniridia

Aniridia is a rare genetic disorder characterized by partial or complete hypoplasia of the iris. There is debate as to whether aniridia is a form of coloboma or failed

development of the optic vesicle rim. Aniridia affects the cornea, iris, intraocular pressure, lens, fovea and optic nerve.

The majority of the time, aniridia has an autosomal dominant inheritance pattern and occurs without systemic involvement due to mutations or deletions of the paired box gene-6 (*PAX6*).

In a minority of patients it can occur as the Wilms tumor-aniridia-genital anomalies-retardation (WAGR) syndrome in which the adjacent *PAX6* and Wilms tumor genes are both deleted.

Persons with aniridia have early onset nystagmus, varying degree of iris and foveal hypoplasia, photophobia, strabismus and reduced visual acuity of 20/100-20/200. Some patients with aniridia have optic nerve coloboma and hypoplasia. Late onset development of cataracts, glaucoma, lens subluxation and corneal opacification and vascularization often occur. Four different forms of aniridia have been identified.

The prevalence of aniridia is 1 in 40,000 to 1 in 100,000. There is no significant predilection for race or gender.

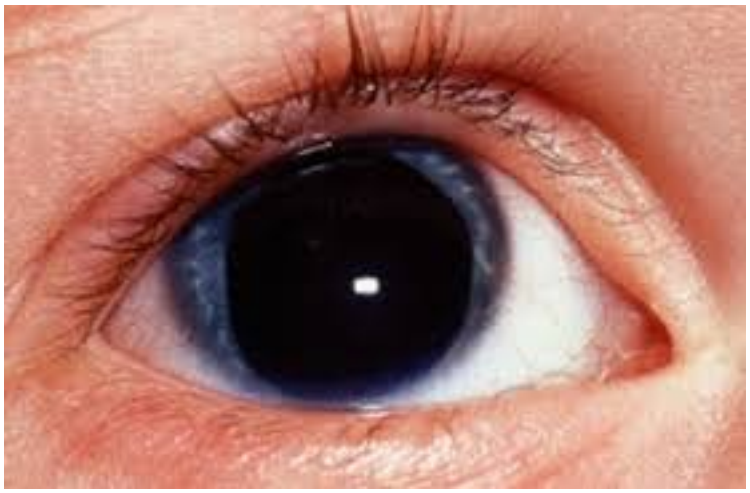

Figure 4: Aniridia

Low Vision Defined

The term low vision covers a broad range of people who have a vision loss that is severe enough to interfere with everyday tasks and that cannot be corrected to normal vision by regular eyeglasses or contact lenses. Normal vision is 20/20 to 20/25 visual acuity. Anyone with *non-correctable* reduced vision is visually impaired or has low vision.

The World Health Organization uses the following classifications of visual impairment.

When the vision in the better eye with best possible glasses correction is:

- 20/30 to 20/60 is considered mild vision loss or has mild low vision.
- 20/70 to 20/160 is considered moderate visual impairment or has moderate low vision.
- 20/200 to 20/400 is considered severe visual impairment or has severe low vision. This category and worse considered legal blindness in the United States.
- 20/500 to 20/1,000 is considered profound visual impairment or profound low vision.
- Less than 20/1,000 is considered near-total visual impairment or near-total blindness.
- No light perception is considered total visual impairment or total blindness.

In addition to visual acuity, there are also levels of visual impairment based on visual field loss.

In the United States, any person with vision that cannot be corrected to better than 20/200 in the best eye, or who has 20 degrees or less of visual field remaining, is considered legally blind. The key to remember here is 'vision that cannot be corrected'. Everyday, patients say they were told they are legally blind. In reality, they simply need glasses or contact lenses and they are correctable to 20/20. These patients are not legally blind and should never be told they are.

Visual impairments take many forms and exist in varying degrees. Visual acuity alone is not a good predictor of a person's vision problems or ability to function. Someone with relatively good acuity (20/40) can have great difficulty functioning, while someone with worse acuity (20/200) might not experience much difficulty performing daily activities.

LOW VISION EXAM

Goal

The goal of optometrists who specialize in providing low vision rehabilitation is to make good use of any sight the patient has and to improve their quality of life. This includes optimizing the patients' reading, activities of daily living, safety, community participation, and psychosocial well being. Low vision rehabilitation

includes optical and non-optical device recommendations and ideally should address the broader impact of vision loss on patients' lives.

Exam Basics

The most important fundamental of a low vision evaluation is to keep it simple. We manipulate 3 variables in a low vision exam:

1. Magnification

The image must be large enough for the child to see it. Children with congenital vision loss naturally hold reading material very close, sometimes only centimeters away. This is not a sustainable practice as it causes back and neck strain. In conjunction with magnification, children are often given a reading stand to promote good ergonomics.

Figure 5: Reading Stand

Examples of magnification

Pocket and stand magnifiers

These are convenient for quick reading of things like price tags, labels and instrument dials and can be illuminated or non-illuminated.

Video magnifier for continuous reading

Handheld, desktop or head-mounted systems enlarge reading material on video display. Image brightness, image size, contrast, foreground/background color are customizable.

Figure 6: Handheld video magnifier

Monocular telescope for distance spotting.
These telescopes are useful for seeing longer distances, such as across the room to see what the teacher is projecting. They can also be modified for near tasks, such as reading.

Figure 7: Monocular telescope

2. Enhance contrast and control glare

Many children with congenital vision loss prefer white or yellow letters on a black background (reverse contrast). As well, children with albinism, achromatopsia and aniridia are photophobic and function best with filters and hats. Photochromic lenses are great options for these children. Two separate filters may be used as well with a higher light transmission filter being worn indoors and darker or lower light transmission filter for outdoors.

Examples of Enhancing Contrast

Appropriate task lighting
Figure/ground relationship

Contrast enhancing filters
Glasses
Acetate sheets which can be placed on top of reading material

3. Modify patient behavior and/or the environment

This is often the biggest challenge. Children with congenital low vision must learn to adapt to the dynamics of their school environment. This includes classroom lighting, navigating the school and changing classrooms, using and managing magnifiers and other devices and proper utilization of technology. As children with congenital vision loss progress to higher grade levels and workload and academic rigor increases, their visual needs must be met in order for them to complete their work effectively and efficiently.

Examples of simple environmental changes

Glare free environment
Desk color (figure ground)
Paper & ink color
Preferential seating
Large print

Case Example #1

ECO is a sweet 10 year old Hispanic female with oculocutaneous albinism, nystagmus, hyperopia and astigmatism. She is in first grade, uses a monitor with a DocuCam to help her see what her teacher is doing, sits in the front of the class and uses a reading stand. Font size in first grade is large enough for ECO to see it most of the time.

Visual Acuities
Distance with Feinbloom
OD: 20/240+
OS: 20/160+
OU: 20/160+

Near
OU: 20/40 at 25 cm (single letters)

Current RX:
OD +6.50 -2.00X 014
OS +8.50 -2.00X175
Current Rx is adequate.

Devices prescribed:
2.8X illuminated stand magnifier

2.5X25 monocular telescope

Case Discussion
The goal is to keep it simple as ECO is only in first grade. ECO has several successful adaptations put in place already. Using a 2.8X illuminated magnifier allowed her to see 20/30 continuous text fluidly. ECO appreciated the light source on the magnifier as it increased contrast. For distance, a 2.5X25 monocular telescope was demonstrated. This allowed ECO to see 20/50 single letters in the distance and she was excited to use it. She immediately began looking at all of the things in the room pointing them out in detail. ECO should continue using large print, preferential seating, the monitor with the DocuCam. Follow up low vision examination in one year is recommended.

Case Example #2

KL is a delightful 15 year old Caucasian female with optic nerve hypoplasia, nystagmus, esotropia, and astigmatism. She also has schizencephaly, seizure disorder and has limited use of her left hand and arm. KL is in 8th grade and concerned about her transition into high school. She prefers 20 pt large print in class and uses a large touch screen Chrome Book. She greatly appreciates using the Chrome Book because it allows her to read things herself. An example is being able to read the morning announcements instead of having to rely on her partner, as she has had to do in the past. She sits in the front of most classes and prefers to use a pen although most of her middle school classes use pencil. She notes she can see pen better. Seeing peer writing in pencil while doing peer grading is extremely difficult. She has access to an iPad and is exploring the camera functions for her transition into high school next year. KL has used a dome magnifier in the past but felt as if it mashed the words together and made it more difficult to read. She is an avid reader but fatigues easily as she only has vision in her left eye.

Visual Acuities
Distance (uncorrected)
OD: Light perception with projection
OS: 20/200-1
OU: 20/200-1

Near
20/50 OU at 10 cm (single word)

Retinoscopy
OD: +0.50 -0.50 X 175
OS: +0.75 -0.75 X 160
No improvement with vision and no Rx given

Devices prescribed

- Prism reading glasses:
  OD: +3.50 -0.50 X 175 with 5 prism diopters Base In prism
  OS: +3.75 -0.75 X 160 with 5 prism diopters Base In prism
- 4X illuminated pocket magnifier
- 6X16 one hand slide monocular telescope
- Hand held video magnifier

Case Discussion
KL was extremely successful with several devices that could be immediately incorporated at school. The prescription reading glasses with prism will help KL to read peer writing more easily. A 4X illuminated pocket magnifier allowed her to see 20/20 single words at near and provided a needed light source. The video magnifier allowed KL to most easily see 20/20 continuous text and to change the contrast to white letters on a black background that she greatly preferred. Using a 6X16 single hand monocular telescope allowed KL to see 20/50 single numbers in the distance. Also, it is operable using only her good right hand. We discussed the future possibility of KL using a bioptic telescope for driving. KL is excited to add each of these devices into her school routine and has applications for each one already mapped out. KL should continue large print in the classroom, the Chrome Book, iPad, preferential seating and using pens. A bioptic evaluation is recommended at the next low vision exam.

Conclusion

Having a big picture overview is the best way to provide low vision rehabilitation to children with congenital vision impairment. Optometrists should have a foundational understanding of the most prevalent congenital disorders and be able to perform a basic low vision evaluation when serving this segment of the population. Low vision rehabilitation is extremely impactful for both the patient and doctor.