Visual Conditions and Functional Vision:
Early Intervention Issues

Visual Conditions
in Infants and Toddlers
Session 3

The University of North Carolina at Chapel Hill
Early Intervention Training Center for Infants and Toddlers With Visual Impairments
FPG Child Development Institute
Objectives

After completing this session, participants will

1. identify the most prevalent visual conditions found in young children with severe visual impairments in the United States and Canada and how they differ from those found in adults.
Objectives

After completing this session, participants will

2. identify the three most prevalent visual conditions—cortical visual impairment (CVI), retinopathy of prematurity (ROP), and optic nerve hypoplasia (ONH)—in young children with visual impairments. Describe causes and characteristics of each condition as well as the implications for early development and intervention.
Objectives

After completing this session, participants will

3. discuss the causes, characteristics, and implications of the following visual conditions: structural abnormalities—anophthalmia, microphthalmia, coloboma, albinism, retinal disorders such as retinoblastoma and Leber’s Congenital Amaurosis, congenital cataracts, and delayed visual maturation.
Objectives

After completing this session, participants will
4. describe the characteristics and implications of the following conditions that may occur as primary or secondary diagnoses—strabismus, amblyopia, glaucoma, nystagmus, and refractive errors.
Prevalence of Visual Impairments

• The prevalence of severe visual impairments in developing countries is about 1 in 1,000, as compared to about 1 in 10,000 in wealthy countries.

• The most prevalent visual conditions in adults with severe visual impairments are diabetic retinopathy, macular degeneration, cataracts, and glaucoma.

• Hatton and colleagues (2001) reported that the most prevalent visual conditions in young children in their sample were CVI, ROP, ONH, albinism, and structural abnormalities such as anophthalmia, microphthalmia, and coloboma.
# Critical Events in Visual Conditions: Age of Diagnosis

<table>
<thead>
<tr>
<th>Condition</th>
<th>Diagnosis</th>
<th>Referral</th>
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<tr>
<td><strong>CVI</strong></td>
<td>7.9 months</td>
<td>10.9 months</td>
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<tr>
<td><strong>ROP</strong></td>
<td>2.4 months</td>
<td>11.5 months</td>
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<tr>
<td><strong>ONH</strong></td>
<td>4.3 months</td>
<td>8.1 months</td>
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<tr>
<td><strong>Structural</strong></td>
<td>2 weeks</td>
<td>9.5 months</td>
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<td><strong>Albinism</strong></td>
<td>3.4 months</td>
<td>11.7 months</td>
</tr>
<tr>
<td><strong>Other</strong></td>
<td>5.2 Months</td>
<td>11.3 months</td>
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</tbody>
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*Hatton et al., 2001*
Diagnosis and Referral

- Structural abnormalities may be diagnosed very early because they may be apparent soon after birth.
- Lag time between diagnosis and referral suggests that closer collaboration with eye care specialists and other early intervention programs is needed.
- Earlier referral could lead to more immediate supports for families and facilitation of optimal development of infants with VI.

Hatton et al., 2001
Most Prevalent Conditions in Young Children With Severe VI

Hatton et al. (2001) reported that the most prevalent visual conditions in a sample of 406 infants and toddlers with severe VI were:

- cortical visual impairment (CVI),
- retinopathy of prematurity (ROP),
- optic nerve hypoplasia (ONH),
- structural abnormalities, and
- albinism.

This was consistent with studies reported by Ferrell (1998), Hatton (1991); Hatton et al. (1997), and Steinkuller et al. (1999).
Amount of Vision in Young Children With Severe VI

- It is difficult to determine whether infants and toddlers meet criteria for legal blindness.
- Approximately 63% of children with structural abnormalities and 42% of children with albinism were designated legally blind in the Hatton et al. study (2001).
- Children with diagnoses of legal blindness may have access to more resources, for example, quota funds for developmental resources from the American Printing House for the Blind.
Multiple Disabilities and VI

- Children with albinism are more likely to have a single disability of visual impairment when enrolled in specialized programs (Hatton et al., 2001).

- Children with CVI are most likely to have additional disabilities at time of enrollment in specialized programs for children with VI (Hatton et al., 2001).

- Children with multiple disabilities and their families may require supports and services that are specific to their unique needs based on each child’s combination of disabilities.
Health Conditions and VI

- Children with CVI and ROP are more likely to have co-occurring health conditions.
- Infants and toddlers with CVI and ROP who depend on technology may have unique medical needs that affect early intervention.
- Some sensory stimulation activities may trigger seizures.
- Children with respiratory problems may be sick more often and more likely to catch contagious illnesses.
Cortical Visual Impairment (CVI)

- Ferrell (1998) and Hatton et al. (2001) found CVI to be the most prevalent visual condition in young children with severe VI.

- CVI results from injury to the brain or visual pathways in the brain rather than disorders or abnormal structures of the eye.

- CVI varies in severity from child to child and from environment to environment, and children with CVI may experience improvement in visual function.
Causes of CVI

- Oxygen deprivation (hypoxia, ischemia)
- Prematurity
- Periventricular leukomalacia
- Trauma
- Meningitis
Visual Behaviors and CVI

CVI can be divided into two groups: cortical and subcortical injuries.

**Cortical**
- exotropia
- horizontal conjugate gaze deviation

**Subcortical**
- esotropia
- tonic downgaze
- ONH and other optic nerve abnormalities

Children in both groups have roving eye movements associated with severe visual impairment and similar rates of nystagmus.

*Brodsky et al., 2003*
Visual Behaviors and CVI

Children with CVI typically have

- neurological abnormalities in addition to other ocular disorders,
- fluctuating vision based on fatigue and levels of sensory input,
- limited or no eye contact,
- vision that generally improves over time but does not extend to typical levels of vision, and rates of improvement that are determined by the age at which CVI occurred and the area of the brain that is injured.

*Carden & Good, 2003*
Visual Behaviors and CVI

The following characteristics have been documented in children with CVI:

- additional neurological abnormalities,
- fluctuations in vision,
- preferences for colored objects,
- light gazing, and
- turning head and eyes away from objects while reaching for them.

*Good et al., 1994
Jan et al., 1987*
Visual Behaviors and CVI

The following characteristics have been documented in children with CVI:

• using touch rather than vision to identify objects,
• preference for familiar environments, and
• photophobia in about a third of children with CVI.

Good et al., 1994
Jan et al., 1987
Characteristics of Children With CVI

In a sample of 406 children, 86 had CVI.

• Approximately half of children with CVI were considered legally blind.
• 79% appeared to have developmental delays or multiple impairments.
• 57% had seizures.
• 24% had eating disorders.
• 21% were dependent on technology (e.g., tracheotomies or GI tubes).
• 17% had respiratory problems.

Hatton et al., 2001
Retinopathy of Prematurity (ROP)

• The prevalence of ROP has increased since the 1980s because improved technology has allowed smaller and younger infants to survive.

• ROP is responsible for 500 to 550 new cases of blindness in the U.S. each year (Siatkowski & Flynn, 1998).

• Medical technology constantly evolves, making it challenging to stay abreast of the latest trends in treatment.
Premature Eye With ROP

Classification of ROP

Scheme of retina

Classification of ROP

The location of the disease is denoted by zones.

**Zone I**: The inner zone extends from the optic disc to twice the disc-macular distance, or 30 degrees in all directions from the optic disc.

**Zone II**: The middle zone extends from the outer border of Zone I to the ora on the nasal side and to approximately the equator on the temporal side.

**Zone III**: The outer zone extends from the outer edge of Zone II in a crescentic fashion to the ora serrata.

*Flynn, 1991, p. 64*
Stages of ROP

**Stage 1:** A thin, relatively flat, white demarcation line separates the avascular retina anteriorly, from the vascularized retina posteriorly. Vessels that lead up to the demarcation line are abnormally branched and/or arcaded.

*Ober et al., 2003, p. 602*
Stages of ROP

**Stage 2**: The demarcation line has visible volume and extends off the retinal surface as a white or pink ridge. Retinal vessels may appear stretched locally, and vault off the surface of the retina to reach the peak of the ridge. Tufts of neovascular tissue may be present posterior to, but not attached to, the ridge.

*Ober et al., 2003, p. 602*
Stages of ROP

**Stage 3**: Extraretinal fibrovascular (neovascular) proliferative tissue emanates from the surface of the ridge, extending posteriorly along the retinal surface, or anteriorly toward the vitreous cavity, giving the ridge a ragged appearance.

*Ober et al., 2003, p. 602*
Stage 4: Subtotal retinal detachment. Traction type retinal detachment results from the development of proliferating tissue in the vitreous gel or on retinal surfaces, subdivided into two types.

Ober et al., 2003, p. 602
Stages of ROP

4A. Subtotal retinal detachment not involving the fovea that generally carries a relatively good prognosis because the macula and fovea are not affected.

4B. Subtotal retinal detachment involving the fovea and macula that results in poor vision.

Flynn, 1991
Stage 4A of Retinopathy of Prematurity

Stage 4B of Retinopathy of Prematurity

Severe Stage 5 Retinopathy of Prematurity

**Stage 5:** Total Retinal Detachment is a complete, funnel-shaped retinal detachment with poor visual prognosis. The funnel may have an open or closed form.

Risk Factors for ROP

• ROP is inversely related to birth weight and gestational age.
• In 2001 it was recommended that infants whose birth weight is less than 1500 grams or who are younger than 28 weeks gestational age be screened for ROP.
• It was also recommended that infants with birth weights between 1500 to 2000 grams with unstable clinical courses or who were classified as high-risk be screened.
• The first ROP examination should be conducted at 4 to 6 weeks of chronological age or within the 31st to 33rd week of gestational age.
Who is at risk for ROP?

Infants who develop the most severe ROP have

• more complicated hospital courses
• respiratory distress syndrome
• pneumothoraces
• patent ductus arteriosus
• cerebral intraventricular hemorrhage
• sepsis
• other complications associated with prematurity

Phelps, 1989
Who is at risk for ROP?

The CRYO-ROP study reported the following characteristics associated with higher risk of severe ROP:

- Lower birth weight
- Younger gestational age
- White race
- Multiple births
- Being born in a hospital not involved in the CRYO-ROP study

Ober et al., 2003
Oxygen and ROP

• Since the 1950s, oxygen administration has been associated with the development of ROP.

• The level and length of oxygen administration that results in ROP is still unknown (Ober et al., 2003).

• Recent research shows a decrease in the severity of ROP based on the changes in management implemented by NICU staff and the monitoring of oxygen levels (Chow, Wright, Sola et al., 2003).
ROP and Additional Disabilities

Approximately 70% of children with ROP have additional disabilities (Hoon et al., 1988; Termote et al., 2003).

Disabilities associated with ROP include

- mental retardation,
- cerebral palsy,
- behavioral problems, and
- deafness/hard of hearing.
Surgical Treatments and ROP

Since the 1980s, a number of surgical treatments have been used for ROP to
• prevent the retina from detaching,
• reattach the retina, and
• remove scar tissue that forms within the eye.

These treatments all seek to prevent the loss of vision or to restore useful vision.

If ROP has progressed to stage 4B or 5, successful surgery usually results in light perception or the ability to see hand motions.
Cryotherapy

Cryotherapy involves repeatedly applying a probe to the surface of the eye to freeze through the wall of the eyeball to the retina.

The cold temperature destroys the portion of the retina to prevent the development of abnormal blood vessels and stops the progression of the disease to reduce the possibility of blindness.
Results of Cryotherapy

• Decreases unfavorable outcomes, thereby reducing the number of children who are blind or severely visually impaired as a result of ROP

• Produces higher incidence rates and levels of myopia than laser photocoagulation

Connolly et al., 2003
Laser Photocoagulation

Laser photocoagulation limits the damage to adjacent structures, produces less inflammation and contraction of the vitreous than cryotherapy.

It is less cumbersome and is as effective as cryotherapy.

McNamara et al., 1991, 1992
Ober et al., 2003
Combined Treatments

Eustis et al. (2003) suggest that combined treatment of cryotherapy and laser photocoagulation appears to be as safe and effective as either method alone.

Combined treatments might be useful for infants with small pupils or media opacities or those with anterior disease and for infants with ROP in their posterior area in order to decrease the time required for surgery.
Vitrectomy

This procedure is used for Stages 4B and 5 and is seen as the last hope for restoring vision.

Vitrectomy is a technique in which the lens of the eye is removed, and the vitreous membranes are segmented by making pie-shaped cuts. Preretinal membranes are removed from the retina surface to eliminate traction and allow the retina to be reattached.
Scleral Buckling

Scleral buckling is a controversial surgical technique saved for Stages 4 and 5 of ROP.

Scleral buckling involves implanting a silicone band around the eyeball that supports the structure of the globe and compresses breaks in the retina that might be precursors of retinal detachment.
Optic Nerve Hypoplasia (ONH)

• ONH is considered the most prevalent congenital optic disorder found in young children with severe VI (Phillips & Brodsky, 2003).

• ONH results from the abnormal development of nerve fibers that make up the optic nerve and is present at birth.

• ONH may affect one (unilateral) or both (bilateral) eyes.

• Visual functioning ranges from normal to total blindness.
Risk Factors for ONH

Maternal Risk Factors

• young maternal age
• first pregnancy or fourth or later pregnancy
• smoking

Child Risk Factors

• premature birth
• small gestational age
• low birthweight

Tornqvist et al., 2002
ONH and Congenital Hypopituitarism

Hypopituitarism is associated with impaired growth, hypoglycemia, developmental delay, seizures, and death, making early diagnosis critical.

Brodsky et al., 1997
ONH and Septo-optic Dysplasia (SOD)

• SOD is diagnosed with an MRI and is associated with the absence of the septum pelucidum and a thinning of the corpus callosum accompanied by small optic nerves.

• Children with SOD frequently have hypopituitarism and may exhibit clinical signs that are similar to those of children with ONH alone.

• Vision loss and hypopituitarism are the two most common functional problems associated with SOD.
Structural Abnormalities

**Anophthalmos**—failure of the globe to develop resulting in no eye.

**Microphthalmos**—abnormally small globe

**Coloboma**—gap or cleft in ocular structures that result from failure to develop fully during fetal development. May affect a number of ocular structures such as the optic nerve, retina, choroid, and iris.

These three disorders are usually detected soon after birth and result from a failure of the embryonic fissure to close at about five to seven weeks gestation (Nishal, 2003a).
Albinism

Albinism is the absence of or a reduction in the pigment in the skin, eye, or both (Traboulsi, 2003). Ocular albinism and oculotaneous albinism are genetic disorders that result in

- nystagmus,
- lack of pigment in the iris,
- hypoplasia of the fovea,
- strabismus,
- high stigmatic refractive error,
- reduced pigmentation in the fundus, and
- reduced vision.
Leber’s Congenital Amaurosis (LCA)

• LCA is a congenital, autosomal recessive retinal disorder with an incidence of 1 in 33,000 that results in severe visual impairment (Eibschitz-Tsimhoni, 2003).
• Infants with LCA develop nystagmus and have sluggish pupillary response.
• Visual function can range from 20/200 to no light perception.
• An electroretinogram is required for a definitive diagnosis.
Characteristics of LCA

• Some children with LCA have cognitive impairments, hearing loss, kidney disorders, and growth deficiency.

• Eye poking, nystagmus, and roving eye movements may be present in children with LCA.

• 17-37% of children with LCA have neurological disorders.
Retinoblastoma

• Retinoblastoma is a malignant tumor within the eye that is fatal if not treated.
• It is the most common type of ocular malignant cancer during childhood.
• Signs include a white reflection in the child’s pupil or strabismus.
• Moore (2000) reports that half of the cases are inherited genetic defects and the other half are due to spontaneous genetic mutations.
Congenital Cataracts

Cataracts are opacities in the lens of the eye. They can be
• unilateral or bilateral,
• congenital or acquired, and
• can occur in isolation or co-occur with other impairments.

The impact of cataracts on visual functioning depends on
• age of onset,
• location of cataract in lens, and
• morphology or structure of the cataract.
Types of Cataracts

• Bilateral cataracts may be associated with a systemic disorder and often require additional medical tests unless they are inherited as an autosomal dominant trait.

• Dense cataracts must be removed by 2 months of age to assure that a clear image is focused on the retina (Buckley, 1998; Wright, 2003d).

• Unilateral cataracts present challenges due to risk of amblyopia.
Visual Functioning and Cataracts

• If nystagmus is present prior to surgery, visual function of 20/60 to 20/80 is typical after surgery.

• The larger, denser, and more centrally located the cataract is, the greater the resultant visual impairment will be (Buckley, 1998, p.269).

• Post surgery, corrective lenses must be fitted for near vision because the lenses are no longer present for accommodation.
Strabismus

• Strabismus is a misalignment of the eyes with resulting abnormal eye movements that results from muscle imbalance and produces images that are not focused directly on the fovea.

• Strabismus is common and often associated with refractive disorders. It can co-occur with other visual disorders such as ROP or CVI.
Strabismus

• Abnormal eye movements that occur with strabismus include phorias or tropias. Eyes may turn inward toward the nose (eso) or outward toward the temple (exo).

• Vertical deviations are denoted by the *hyper* prefix (e.g., hypertropia)
Amblyopia

• Amblyopia describes a reduction of visual acuity in the absence of abnormal ocular structures. It results from lack of visual stimulation via clear focused images and is the most common cause of decreased vision in childhood.

• Treatment is more likely to be successful if it is started early and if there is reasonably good visual acuity in the amblyopic eye (Kushner, 1998).

• Treatment options include patching or occluding the good eye until visual functions improves to normal in the affected eye.
Glaucoma

• Glaucoma refers to a group of disorders in which the pressure inside the eye increases and potentially damages the optic nerve and retina.

• Three major types of pediatric glaucoma include primary infantile or congenital glaucoma (open angle), juvenile, and secondary.
Glaucoma

- Secondary glaucoma may co-occur in other visual disorders or syndromes such as aniridia, ROP, juvenile rheumatoid arthritis, or rubella.

- Signs and symptoms include corneal opacities, corneal enlargement, large or bulging eyes, photophobia, optic nerve cupping, amblyopia, strabismus, and anisometropia.
Nystagmus

- Nystagmus is an involuntary oscillation of one or both eyes (Awner & Catalano, 1998; Hertle, 2003).
- Nystagmus is associated with decreased vision within the first two years of life resulting from ocular disorders.
- Nystagmus is the primary diagnosis if no other ocular disorder can be identified.
- Conjugate nystagmus means that the eyes move together synchronously; if disconjugate, then the eyes move separately.
Nystagmus

- **Pendular nystagmus**—movements are of equal speed and in the same direction; often associated with visual acuity of better than 20/200 in at least one eye and with loss of central vision

- **Jerk nystagmus**—movements faster in one direction and slower in the other

- **Searching nystagmus**—roving horizontal movements without fixation; often associated with visual acuity that is worse than 20/200
Treatment for Nystagmus

• A thorough ocular examination is required because most nystagmus is accompanied by other visual disorders.

• Acquired nystagmus that is diagnosed after the first few years of life is almost always associated with neurological disorders.

• Treatment might include surgery on eye muscles to lessen head tilt or eccentric gaze or to treat strabismus.
Treatment for Nystagmus

• Corrective lenses might be used to treat refractive errors, muscle imbalances, or to dampen the oscillating movements that result from nystagmus.

• Children with nystagmus should not be discouraged from using head tilts or eccentric gaze because these behaviors may allow a null point that reduces the involuntary eye movements.
Refractive Errors

Refractive errors occur when the cornea and lens fail to refract (bend) light rays in order to focus images at the optimal location on the retina. If uncorrected, refractive errors can lead to amblyopia, detached retinas, cataracts, opacities of the vitreous, and choroidal hemorrhages.
Refractive Errors

- **Myopia**—nearsightedness; caused by an elongated globe or overly strong bending powers of the lens and cornea

- **Hyperopia**—farsightedness; caused by a shorter globe or weak bending powers of the lens and cornea

- **Astigmatism**—blurred vision in both near and far range; caused by an unevenly rounded cornea
Delayed Visual Maturation (DVM)

- Delayed visual maturation (DVM) has been defined as unexplained decreased vision followed by rapid improvement to normal levels before the 1st birthday.
- DVM is a diagnosis of exclusion that can only be made in retrospect after an infant diagnosed with poor vision shows normal development of vision (Elston, 2000; Russell-Eggitt et al., 1998).
- Children with DVM have normal electroretinograms and visual evoked potentials.
- DVM can be differentially diagnosed from CVI if visual function improves and if the child appears to be developing typically.
Types of DVM

**Type I DVM** (idiopathic or isolated) includes children with normal general/neurological development and no underlying pathology. Between 3-6 months of age, infants with Type 1 experience a rapid and spontaneous improvement in vision to normal or near-normal levels.

**Type II DVM** is associated with systemic disorders or mental retardation. Vision usually improves but may take longer and there may be continued loss of vision.

*Russell-Eggitt et al., 1998*
Types of DVM

Type III DVM is associated with other ocular diseases such as oculocutaneous albinism (Kassmann-Kellner, 1998), cataracts, or aniridia.

• Vision is worse than would be expected from the disease alone and the mean age of visual recovery is 20 weeks (Russell-Eggitt et al., 1998).

• Interestingly, the onset of nystagmus may precede recovery in type III DVM.

• Visual recovery is completed by 8 months of age, but is also determined by the visual abilities and other characteristics of the child.

Russell-Eggitt et al., 1998