Module: Visual Conditions and Functional Vision: Early Intervention Issues

Session 3: Visual Conditions in Infants and Toddlers

Major Points

A. Prevalence of visual conditions in infants, toddlers, and preschoolers

Prevalent visual conditions in infants, toddlers, and preschoolers in the United States are different from those found in developing countries and in adults with visual impairments.

Blindness in children (ages birth to 14) accounts for approximately 4% of the population of persons who are blind throughout the world (Thylefors, Negrel, Pararajasegaram, & Dadzie, 1995). The prevalence of severe visual impairments in developing countries may be as high as 1 in 1,000, as compared to 1 in 10,000 in wealthy countries (Gilbert, Anderton, Dandona, & Foster, 1999). Prevalent causes of severe visual impairments in developing countries include Vitamin A deficiency, corneal scarring, trachoma, and inherited conditions (Gilbert, Anderton, Dandona, & Foster, 1999).

Roodhooft (2002) reports that the World Health Organization lists cataracts, trachoma, and glaucoma as being responsible for more than 70% of global blindness. The prevalent causes of severe visual impairments in adults of developing countries are similar to those found in children and include Vitamin A deficiency, cataracts, glaucoma, trachoma, and onchocerciasis (“river blindness,” caused by a parasitic worm).

The causes of blindness in children vary from those found in adults. According to Prevent Blindness America and the National Eye Institute’s report, the leading causes of visual impairment and blindness in Canada and the United States include diabetic retinopathy, age-related macular degeneration, cataracts, and glaucoma. This report also shows cataracts as the leading cause of blindness worldwide.

In 1999, Steinkuller and colleagues studied the prevalence of visual conditions in residential schools for children who are blind in the United States and found that cortical visual impairment (CVI), retinopathy of prematurity (ROP), and optic nerve hypoplasia (ONH) were the most prevalent conditions in their sample of 2,553 children. These authors noted that residential schools experienced an influx of children with CVI in 1985 and of children with ROP in the early 1990s. Although ROP and CVI are seldom mentioned in studies of visual impairment in developing countries, ROP is present in middle-income countries such as Cuba, Paraguay, Bulgaria, and Thailand (Gilbert, Rahi, Eckstein, O’Sullivan, & Foster, 1997).
In 2001, the Model Registry of Early Childhood Visual Impairment Collaborative Group reported that the most prevalent visual conditions in young children served by specialized agencies for individuals with visual impairments were CVI, ROP, ONH, albinism, and structural abnormalities such as anophthalmia, microphthalmia, and coloboma (Hatton et al., 2001). Out of a sample of 406 children between the ages of birth and 3 years, other visual conditions (in order of prevalence) included retinal disorders, optic nerve atrophy, cataracts, delayed visual maturation, refractive errors, nystagmus, glaucoma, oculomotor apraxia, aniridia, corneal defects, Peter’s anomaly, hemianopsia, and others. Visual condition was listed as unknown for 78 children. The ages at which children in the study were diagnosed, referred for early intervention services, and entered specialized early intervention programs for children with visual impairments are described in Table 1.

<table>
<thead>
<tr>
<th>Visual Condition</th>
<th>Diagnosis of Visual Impairment</th>
<th>Referral to Specialized Program</th>
<th>Entry into Specialized Program</th>
</tr>
</thead>
<tbody>
<tr>
<td>CVI N = 86</td>
<td>7.9 (7.4)</td>
<td>10.9 (8.0)</td>
<td>11.4 (8.0)</td>
</tr>
<tr>
<td>ROP N = 69</td>
<td>2.4 (2.9)</td>
<td>11.5 (8.4)</td>
<td>12.2 (8.1)</td>
</tr>
<tr>
<td>ONH N = 31</td>
<td>4.3 (2.8)</td>
<td>8.1 (6.4)</td>
<td>8.6 (6.5)</td>
</tr>
<tr>
<td>Structural N = 16</td>
<td>0.4 (0.8)</td>
<td>9.5 (8.6)</td>
<td>10.0 (8.4)</td>
</tr>
<tr>
<td>Albinism N = 19</td>
<td>3.4 (3.3)</td>
<td>11.7 (10.5)</td>
<td>12.2 (10.4)</td>
</tr>
<tr>
<td>Other N = 107</td>
<td>5.2 (5.8)</td>
<td>11.3 (9.1)</td>
<td>11.9 (9.3)</td>
</tr>
</tbody>
</table>

Table 1. Average Age in Months (SD) of Critical Events by Visual Conditions (Hatton et. al., 2001, p. 424)

Structural disorders such as anophthalmia (absence of the eyes), microphthalmia (very small eyes), and coloboma (failure of parts of the eye to develop), or those with obvious physical attributes such as albinism (pale skin, white hair) tend to be diagnosed very
early because they are apparent soon after birth. Retinopathy of prematurity, or ROP (a disorder of the retina that is related to prematurity), is also diagnosed relatively early, at 2.4 months, because these children are detected by screening exams in the hospital nursery. Although the average age of diagnosis of children with these disorders was just under two weeks (.4 of a month), the average age of referral to specialized programs was about 9.5 months. However, these children may be enrolled in early intervention programs that do not specialize in visual impairments before that time.

Optic nerve hypoplasia (failure of the optic nerve to develop normally during the prenatal period) was diagnosed at an average age of 4.3 months; however, the average age at which children were referred for specialized services was not until 8.1 months of age. Cortical visual impairment (loss of vision associated with no ocular structure defects) was diagnosed later because eye care specialists are usually uncertain about the degree and cause of visual loss. Once diagnosed, physicians and other professionals make more prompt referrals to specialized early intervention programs.

The lag time between diagnosis of these prevalent causes of visual impairment in very young children and the time that they are referred for specialized early intervention services for children with visual impairments is noteworthy. Practitioners and administrators in local programs need to collaborate more closely with eye care specialists and other early intervention programs in the community to decrease the time between diagnosis and referral. Earlier referral can lead to more immediate support for families in order to facilitate the optimal development of infants with visual impairments.

For the past 6 years, Trista has worked as a ECVC with her local early intervention agency. When she first began, she had a caseload of three children who all had retinopathy of prematurity. Today she supports 16 families of young children with visual impairments or blindness. Her caseload includes children with cortical visual impairment, retinopathy of prematurity, optic nerve hypoplasia, Leber’s congenital amaurosis, high myopia, and albinism.

Recently Trista was asked to support other local ECVCs in an effort to develop child find activities. She thought back to when she first began her work in early intervention. In order to increase her caseload and meet the needs of children in her area, she had informed people about the program, helped others understand the leading causes of visual impairment in young children, and built trusting relationships with other local programs, NICUs, therapists, ophthalmologists, pediatricians, and Part C coordinators. As Trista worked with other ECVCs, she shared ways to collaborate with professionals in order to increase referral rates, thereby increasing the number of children identified with visual impairments in their area.

B. Five most common visual conditions
Cortical visual impairment (CVI), retinopathy of prematurity (ROP), optic nerve hypoplasia (ONH), structural abnormalities of the eye (anophthalmia, microphthalmia, and colobomas), and albinism have been identified as the most common etiologies of severe visual impairments in very young children (Ferrell, 1998; Hatton, 1991; Hatton, Bailey, Burchinal, & Ferrell, 1997; Hatton et al., 2001; Steinkuller et al., 1999).

<table>
<thead>
<tr>
<th>Vision Condition</th>
<th>Legally Blind</th>
<th>Not Legally Blind</th>
<th>Unknown/Missing</th>
</tr>
</thead>
<tbody>
<tr>
<td>CVI n = 86</td>
<td>47 55%</td>
<td>6 7%</td>
<td>33 38%</td>
</tr>
<tr>
<td>ROP n = 69</td>
<td>30 44%</td>
<td>15 22%</td>
<td>24 34%</td>
</tr>
<tr>
<td>ONH n = 31</td>
<td>16 52%</td>
<td>2 6%</td>
<td>13 42%</td>
</tr>
<tr>
<td>Structural n = 16</td>
<td>10 62%</td>
<td>3 19%</td>
<td>3 19%</td>
</tr>
<tr>
<td>Albinism n = 19</td>
<td>8 42%</td>
<td>5 26%</td>
<td>6 32%</td>
</tr>
</tbody>
</table>

Table 2. Vision status at initial referral to specialized agencies for the visually impaired (Hatton et al., 2001, p. 425)

When children are first referred to specialized agencies that provide services for infants and toddlers, it may be difficult to determine if they meet criteria for legal blindness, as demonstrated by the relatively high number with unknown or missing information in Table 2 (Hatton et al., 2001). However, it is noteworthy that more than half of the children with each diagnosis did have information about the status of legal blindness. Visual function may change considerably during the first few years, and many more children are found to meet the criteria of legal blindness over time. Children who have a diagnosis of legal blindness can have access to more resources, such as quota funds for developmental resources from the American Printing House for the Blind.

Children with multiple disabilities and their families require support and services that are tailored to the unique combination of disabilities for each child. To be most effective, early intervention should be individualized with emphasis on the family’s concerns and priorities as well as the results of a multidisciplinary assessment.
<table>
<thead>
<tr>
<th>Eye Condition</th>
<th>Seizures</th>
<th>Respiratory Problems</th>
<th>Eating Disorders</th>
<th>Technology Dependent</th>
</tr>
</thead>
<tbody>
<tr>
<td>CVI N = 86</td>
<td>49 57%</td>
<td>15 17%</td>
<td>21 24%</td>
<td>18 21%</td>
</tr>
<tr>
<td>ROP N = 69</td>
<td>8 12%</td>
<td>21 30%</td>
<td>14 20%</td>
<td>13 19%</td>
</tr>
<tr>
<td>ONH N = 31</td>
<td>3 10%</td>
<td>0</td>
<td>1 3%</td>
<td>1 3%</td>
</tr>
<tr>
<td>Structural N = 16</td>
<td>2 13%</td>
<td>2 13%</td>
<td>3 19%</td>
<td>1 6%</td>
</tr>
<tr>
<td>Albinism N = 19</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
</tr>
</tbody>
</table>

Note: Some children had more than one health condition.

Table 4. Health Conditions Associated with Specific Eye Conditions (Hatton et al., 2001, p. 427)

Children with CVI and ROP are more likely to have co-occurring health conditions than children with ONH, structural abnormalities, or albinism. In fact, none of the 19 children with albinism in this sample had experienced seizures, respiratory problems, eating disorders, or were technology-dependent upon entry into specialized programs for young children with visual impairments. Infants and toddlers with CVI and ROP who depend upon technology for breathing and/or eating may have unique medical needs that may impact early intervention. Close collaboration with medical personnel and occupational therapists is required to assure that the goals for early intervention are congruent with the child’s medical needs. Early interventionists who work with children who have seizures should be aware that some sensory stimulation activities may actually trigger seizures, and they should plan interventions accordingly. Finally, children with respiratory problems may be sick more often and may be more likely to catch contagious illnesses. Therefore, early interventionists must be vigilant about
avoiding contact with these children when the interventionists are ill or think they may be becoming ill with a respiratory illness.

Six-month-old twins Shanu and Tamul were born at 25 weeks’ gestation. Their early interventionist recently submitted a referral to Laurence, a local ECVC, for support and services. Laurence briefly reviewed the referral. He learned that Shanu has bilateral retinal detachment due to Stage 5 ROP and uses a feeding tube and oxygen for most of the day. Tamul has Stage 3 ROP.

When Laurence spoke with the early interventionist, he learned that their mother is very hesitant to have people in the home because the twins are frequently ill and are still medically fragile. He also learned that their mother does not allow other people to handle the twins unless they are sign and symptom free from colds and other illnesses, and then only after they have thoroughly washed their hands. Given their mother’s concern for the health of her children, Laurence knew that he would need to work closely with the intervention team in order to meet the family’s needs.

C. Cortical visual impairment

Cortical visual impairment (CVI) has been one of the two most prevalent visual conditions reported in children with severe visual impairments during the past 20 years.

Cortical visual impairment is caused by lesions along the visual pathway or in the brain in the areas where images are interpreted. Because so many areas of the brain are involved in processing visual information and because of the resiliency of the brain, children with CVI may be very heterogeneous. In large samples of young children with severe visual impairments, Ferrell (1998) and Hatton and colleagues (2001) reported that cortical visual impairment was the most prevalent visual condition. Pediatric ophthalmologists also recognize CVI as a leading cause of visual impairment in children in countries with adequate health care (Brodsky, Fray, & Glasier, 2002; Carden & Good, 2003).

Whiting, Jan, and colleagues (1985) introduced the concept of cortical visual impairment to distinguish it from cortical blindness, a term that had been used to describe blindness in adults that resulted from brain injury. These authors studied CVI in 50 children and concluded that CVI varies in severity from child to child and from environment to environment, and that children with CVI may experience improvements in visual function. Jan, Groenveld, Sykanda, & Hoyt (1987) suggest that the following characteristics may distinguish children with CVI: normal pupillary response, normal appearance of the media and fundus, no optokinetic nystagmus (involuntary eye movement that occurs when striped optokinetic drum is presented near the child’s eyes), no blink to threat, no tracking, normal extraocular movements, lesions appearing between geniculate bodies to occipital cortex, and EEG rhythms correlating with residual vision.
Oxygen deprivation (hypoxia, ischemia) is cited as a common cause of CVI. Increasingly, however, CVI is associated with prematurity and may occur along with retinopathy of prematurity and/or periventricular leukomalacia (PKL), a condition that results from damage to the brain in the ventricle area that is also often associated with oxygen deprivation (Brodsky et al., 2002; Carden & Good, 2003). Other causes of CVI include meningitis, congenital brain malformations, head trauma, infections, and shunt failure (Langley, 1998).

Brodsky et al. (2002) completed a retrospective study of 100 children with CVI and concluded that children with CVI actually could be divided into two groups. The first group consists of children born full-term who have damage to the striate and peristriate cortex and whose damage is cortical in nature. The second group consists of premature children with preterm injuries to subcortical white matter and/or to the optic radiations. Using neuroimaging techniques and detailed descriptions of visual behaviors, these authors reported that

- children with both types of CVI were likely to have strabismus (82%). However, those with cortical vision loss were more likely to have exotropia, and those with subcortical vision loss were more likely to have esotropia.
- roving eye movements were associated with more severe visual impairment in both groups. Children with cortical vision loss tended to have horizontal conjugate gaze deviation (eyes will not move together on a horizontal plane), and children with subcortical visual loss tended to have a tonic downgaze.
- children in both groups had similar rates of nystagmus, or rapid involuntary movements of the eyes, that have been noted as not present in children with CVI by other ophthalmologists.
- children with subcortical vision loss were more likely to have optic nerve hypoplasia and other optic disc abnormalities.

According to Carden and Good (2003), children with CVI typically have other neurological abnormalities and may also have other ocular disorders. Often, children with CVI have poor vision that may fluctuate from day to day based on fatigue and on levels of sensory input, and they usually do not make eye contact or enjoy looking at faces. The vision of many children with CVI often improves over time, consistent with resiliency in brain function, but this improvement usually does not extend to visual function at typical normal levels. The rate of improvement may be determined by the age at which CVI occurs and the area of the brain that is injured, suggesting that the two categories proposed by Brodsky and colleagues (2002) may be helpful in determining the visual potential for children with the two types of CVI. Research will be necessary to confirm the utility and prognostic value of cortical versus subcortical vision loss. However, Carden and Good (2003) note that the prognoses are not as favorable for children with damage of the optic radiations or those who have been diagnosed with periventricular leukomalacia.
Katrien, a 14-month-old with Stage 3 ROP, sat on the lap of her foster mother, Layla, during an exam with the ophthalmologist. Based on his exam, the ophthalmologist felt that Katrien ought to be more visually attentive. During the exam, Layla commented on the fact that even when Katrien wears her glasses, she does not attend visually to people or objects in her environment. When the ophthalmologist questioned her further, he heard familiar comments such as “Katrien never looks at me when I talk to her” and “Sometimes I feel like her vision is different every day.” After completing the clinical exam, reviewing her complicated medical and birth history, and hearing the family’s concerns, the ophthalmologist and Layla agreed to have Katrien further evaluated for a diagnosis of cortical visual impairment.

As can be seen from data from the Model Registry of Early Childhood Visual Impairment (Hatton et al., 2001) found in Tables 1 through 4 in Major Points A and B, children with CVI were typically diagnosed at 7.9 months of age, referred to specialized agencies that provide VI services at 10.9 months, and actually enrolled in those programs at about 11.4 months of age. Approximately half were considered legally blind upon entry into specialized agencies, and 79% appeared to have developmental delays or to be multiply impaired at entry into specialized programs. Children with CVI in the Registry had a number of health disorders—57% had seizures, 24% had eating disorders, 21% were dependent on technology such as tracheostomies (surgical opening into windpipe to facilitate mechanical ventilation) or gastrostomy (artificial opening through abdomen to stomach to facilitate feeding) tubes, and 17% experienced respiratory problems.

In a qualitative study of behavior in nine children between 21 months and 9 years, 8 months of age, Porro and colleagues (1998) reported the following behaviors in children with CVI in response to various objects.

- Head movements to the stimulus and changes in facial expressions were noted in all nine children.
- Eight children followed the movement of the stimulus.
- Seven children reached for the stimulus.
- Six children displayed intermittent fixation toward stimulus and smiled in response to the stimulus.
- Four children appeared to visually avoid the stimulus.
- Three children looked away while reaching for the stimulus, looked past the stimulus, or changed posture to indicate that they saw it.
- Two of the children engaged in stereotypic movements.

In addition, the authors observed that children with CVI avoided those objects with tactile and auditory stimuli.

Jan and colleagues (Good, Jan, DeSa, Barkovich, Groenveld, & Hoyt, 1994; Jan, Groenveld, Sykanda, & Hoyt, 1987) provided detailed descriptions of CVI from a medical and behavioral perspective. In particular, these authors noted that almost all
Children with CVI have additional neurological abnormalities. Behaviors observed in children with CVI included fluctuations in vision that occur from hour to hour or day to day, preferences for colored objects, tendencies to light-gaze, turning the head and eyes away from objects during reach, using touch rather than vision to identify objects, preference for familiar environments that are not noisy or busy, and photophobia in about a third of children with CVI.

Children with CVI and the families in which they live are very heterogeneous, and an individualized, family-centered approach must be taken in planning and implementing early intervention. As noted by Morse (1999), professionals who work with children with CVI must consider a host of variables, such as the environment and the functionality of goals, when planning and implementing intervention. Morse (1990) also provided helpful suggestions about how ECVCs and other early interventionists should recognize that the child’s internal state is critical to visual attention and to involvement in intervention activities. The type, intensity, and duration of visual experiences must be carefully considered when working with children with CVI. Also, using motivating people, objects, and activities, and presenting learning materials carefully and systematically may assist in intervention with young children with CVI.

Levack, Stone, and Bishop (1994) suggest the following strategies for working with children with CVI (pp. 16-17).

- Use other sensory cues to stimulate or support visual information.
- Avoid visual overstimulation by introducing items one at a time in an uncluttered environment.
- Work with only one sense at a time when using other sensory cues to stimulate visual performance.
- Watch for preference in color, shape, size, movement, and field.
- Make changes gradually, recognizing that these students can have difficulties processing information.
- Try shaking or moving objects as they are brought into students’ line of vision.
- Determine the best position for individual students to use their eyes.
- Make visual cues bold and simple, using them consistently throughout programming.
- Present visual stimuli in a simple figure-ground environment.

Langley (1998) notes that input to the vestibular system may help to establish an alert visual state and that stable positioning is critical to efficient use of vision in children with CVI. Other strategies encouraged by Langley for facilitating effective use of vision include

- helping the child to touch objects to integrate vision and touch,
- introducing objects into the peripheral visual fields,
- using a variety of positions and spatial orientations with children with severe multiple impairments,
- avoiding excessive visual stimulation,
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for Infants and Toddlers With
Visual Impairments

- using illuminated toys or light sources to encourage visual attention,
- using toys that move,
- presenting one toy or object at a time,
- using brightly colored objects instead of black-and-white objects,
- using color to highlight objects, shapes, and print,
- leaving plenty of space between objects,
- using books with single or simple pictures,
- using simple pictures that do not have extraneous detail,
- providing sufficient contrast between objects and background,
- using routines,
- clearly noting the beginning and ending of activities,
- encouraging the use of touch to slide objects apart to avoid crowding effect, and
- promoting head control.

The Blind Babies Foundation in San Francisco developed a series of fact sheets on prevalent visual disorders in very young children. The fact sheet on cortical visual impairments, which includes strategies for working with these children, is included as Handout B.

Felix, the occupational therapist, was eager to meet with Carson, the ECVC, and the Romane family. The Romane family had expressed interest in making independent feeding a goal for their son, Dallas. Dallas has cortical visual impairment and mild ataxic cerebral palsy. Felix felt comfortable in assisting the family in creating adaptations for the motor components for feeding goals, but he knew that Carson could help them in facilitating Dallas’s optimal use of vision during mealtime.

During the meeting, Carson discussed the results of Dallas’s latest functional vision assessment (FVA). Based on the FVA, they knew that Dallas responds well to the color purple and that he tends to use his right eye for central fixation and viewing. He also has difficulty with clutter and noisy environments. In order to facilitate Dallas’s feeding goals, the family decided to begin their dinner routine with the television off to reduce noise. Additionally, they decided to use a purple sippy cup placed on the right side of the highchair tray. The family also moved Dallas’s highchair so that it faced the white kitchen wall rather than the open family room. This would assist in decreasing the clutter in the background. Within a few weeks, the family noticed that Dallas was much more visually attentive during meal times and was showing progress in independently using his sippy cup during these routines.

D. Retinopathy of prematurity (ROP)

What is ROP and how does it develop?
Retinopathy of prematurity (ROP), formerly called retrolental fibroplasias, is a retinal disorder that occurs most often in premature infants. Theodore Terry identified ROP in 1942 in an epidemic of blindness among premature infants (Siatkowski & Flynn, 1998). In the 1950s, researchers became aware of an association between the administration of oxygen and the disorder, resulting in a curtailment of the use of oxygen with premature infants. The incidence of ROP dropped significantly; however, professionals subsequently discovered that the reduction of oxygen resulted in increased mortality for premature infants and an increased incidence of neurological impairments such as cerebral palsy (Ober, Palmer, Drack, & Wright, 2003).

With improved medical technology that allowed smaller and younger infants to survive, the incidence of ROP during the 1980s increased. Siatkowski and Flynn (1998) estimate that ROP is responsible for between 500 to 550 new cases of blindness in the United States each year. Because ROP resulted in blindness in hundreds of infants and because of increased technology in the field of ophthalmology, the field of pediatric ophthalmology devoted considerable attention to the treatment of this disorder during the late 1980s and 1990s. Hundreds of studies have been reported in medical journals since that time, making it difficult to stay abreast of the latest developments in treatment.

According to Ober et al. (2003), the development of blood vessels in the retina begins at 16 weeks’ gestation, when the vascular precursor mesenchyme flows from the optic disc and grows across the surface of the retina in a wavelike fashion. The blood vessels reach the edge of the retina on the nasal side at about 32 weeks and the edge of the retina on the temporal side soon after birth. Because the vascular precursor does not reach the temporal side of the retina until 40 weeks, most ROP is located in the temporal retinal region. As this vascular precursor grows in the nerve fiber layer of the retina, “its trailing edge is a delicate, chicken-wire meshwork of capillaries which, by absorption and remodeling, results in mature arteries and veins surrounded by a capillary meshwork” (Siatkowski & Flynn, 1998, p. 61).

Siatkowski and Flynn (1998) note that ROP develops when an unidentified agent destroys vascular endothelium in the most vulnerable area—where the capillary meshwork is located. At that point, two tissues survive and unite through the remaining vascular channels. The structure that these tissues form is called the mesenchymal arteriovenous shunt, and it replaces the capillary bed that was destroyed. This shunt, which forms a demarcation line between the areas of the retina that have blood vessels and those that do not, is nourished by mature arteries and veins.

In the late 1980s a group of ROP specialists from 11 countries developed the classification system for ROP that is currently used, called the International Classification of Retinopathy of Prematurity (ICROP). The system specifies the location in the eye and the extent of the disease and the severity or staging of the disease (Ober et al., 2003).
According to Flynn (1991, p. 64) 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 serrata 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.

The number of clock hours involved (see PowerPoints 3S through 3PP) indicates the vascular involvement or the extent of the disease. Ober et al. (2003, p. 602) describe the first three stages of ROP as follows.

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

**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 may 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.

**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.

Flynn’s (1991) descriptions of Stages 4 and 5 of ROP are provided below.

**Stage 4:** Subtotal retinal detachment is a traction-type detachment caused by the development of proliferating tissue in the vitreous gel or on retinal surfaces. This is subdivided into two types:

- **A.** Subtotal retinal detachment not involving the fovea generally carries a relatively good prognosis because the macula and fovea are not affected.
- **B.** Subtotal retinal detachment involving the fovea and macula results in poor vision and often includes a traction fold through the macula.

**Stage 5:** Total retinal detachment is a complete, funnel-shaped detachment with poor visual prognosis. The funnel may have an open or closed form.
Which children are most at risk for ROP?

According to Ober et al. (2003), ROP appears to be inversely related to birth weight and gestational age. The American Academy of Pediatrics, the American Association for Pediatric Ophthalmology and Strabismus, and the American Academy of Ophthalmology (2001) developed guidelines for screening for ROP. These guidelines recommend that infants whose birth weight is less than 1500 grams or who are younger than 28 weeks’ gestational age be screened for ROP using a detailed protocol developed by the three organizations. In addition, they recommended screening for infants with birth weights between 1500 and 2000 grams who had unstable clinical courses or who are thought to be at high risk. Based on numerous studies that examined the stage at which ROP first develops, these groups recommend that the first examination for ROP should be conducted at four to six weeks of chronological age or within the 31st to 33rd week of postconceptual age (gestational age at birth plus chronological age).

The landmark Multicenter Trial of Cryotherapy for Retinopathy of Prematurity (CRYO-ROP) examined ROP in a cohort of children born in 1986 and 1987 with birth weights less than 1251 grams. In that study, approximately 66% of these infants developed ROP, with 82% of the infants who weighed less than 1000 grams developing ROP and 90% of the infants with birth weights of less than 750 grams developing ROP (Ober et al., 2003).

Phelps (1989) noted that when birth weight and gestational age are matched, infants who develop severe ROP are most often those who have had “the most complicated hospital courses, i.e., asphyxia at birth, respiratory distress syndrome requiring mechanical ventilation, pneumothoraces, patent ductus arteriosus, cerebral intraventricular hemorrhage, sepsis, and other complications commonly associated with prematurity” (p. 418). Other researchers have also reported relationships between ROP and severe illness and complicated clinical courses (Cryotherapy for Retinopathy of Prematurity Cooperative Group, 2002).

Oxygen administration has been associated with the development of ROP since the 1950s; however, the level and length of oxygen administration that results in ROP is still unknown (Ober et al., 2003). A few cases of ROP have been diagnosed in which oxygen was not administered; however, some researchers have found a relationship between incidence and severity of ROP and the amount of time that infants experience certain levels of oxygen tension in the blood (Ober et al., 2003). Chow, Wright, Sola, and colleagues (2003) recently reported significant decreases of severe ROP in infants weighing less than 1251 grams when the neonatal intensive care unit (NICU) staff implemented changes in the management and monitoring of oxygen levels. Specifically, cases of Stage 3 and Stage 4 ROP decreased from 12.5% in 1997 to 2.5% in 2001 as a result of changes in oxygen administration and management, and laser treatment for ROP declined from 4.5% in 1997 to 0 in 2001.
It now seems that a protein called vascular endothelial growth factor (VEGF) may be related to oxygen levels in the brain and may be the mechanism by which oxygen levels affect the development of ROP (Chow et al., 2003). A discussion of the relationship between the genetic expression of VEGF and the vascularization of the retina in premature infants is beyond the scope of this session. Chow et al. (2003) describe the proposed relationship, and we recommend that interested readers review that article for additional information.

The CRYO-ROP group reported that the following characteristics were associated with a higher risk of severe ROP: lower birth weight, younger gestational age, White race, multiple births, and being born in a hospital that was not one of 23 hospitals involved in the CRYO-ROP study (Ober et al., 2003).

**Do children with ROP have additional disabilities?**

Hoon and colleagues (1988) described how retinopathy of prematurity changed between 1951 and 1987. They noted that ROP in the 1980s was a disease of smaller and younger infants who seemed to have very little useful vision; however, the percentage of children who have disabilities in addition to vision remained stable over the course of 37 years (approximately 70 percent). Disabilities that have been associated with ROP include mental retardation, cerebral palsy, behavioral problems, and hearing impairments (Hoon, Jan, Whitfield, McCormick, Richards, & Robinson, 1988).

Termote and colleagues (2003) studied the incidence of children with ROP and their concomitant disabilities and found that there was a statistically insignificant decrease in the number of children with ROP in the Netherlands between 1994 and 2000. However, the authors reported a significant increase in behavior abnormalities in children with ROP, and 68% of the children with ROP had multiple disabilities.

Hatton and colleagues (2001) reported that 45% of their sample of children with ROP appeared to have additional disabilities at the time of enrollment into specialized programs serving children with visual impairments. Because the disability status of children was recorded at the time of enrollment into specialized agencies at an average age of 11.5 months, the prevalence of additional disabilities at this young age is probably an underestimate. Only the most severe disabilities would be diagnosed at 11 months, while more moderate and subtle disabilities might not be evident until kindergarten or later. Approximately 44% of the 69 children with ROP were considered legally blind at entry into specialized programs. Children with ROP were more likely to have respiratory problems than other children (30% of the 69 children had respiratory problems) and approximately 20% had eating disorders or were technology-dependent.

Msall and colleagues (2000) described the neurodevelopmental outcome for children in the CRYO-ROP study and reported that the severity of neonatal ROP was a marker for functional disability at 5.5 years of age. Better functional status as measured by an
instrument called the WeeFIM was associated with favorable visual acuity, favorable neurological score at age two years, absence of ROP, possession of private insurance, and African-American ancestry. The functional status of children with favorable visual acuity was compared to that of children with unfavorable visual acuity (rating visual acuity below 6.4 cycles per degree). Children with favorable visual acuity were not as likely to have neurodevelopmental disabilities as children with unfavorable visual acuity. Regarding “self care,” the percentages were 25.4% versus 76.8%; for “continency,” 4.5% versus 50%; for “motor disability,” 5.3% versus 42.7%; for “communication-cognitive disability,” 22.4% versus 65.9%. This finding is consistent with Hatton et al. (1997), who reported that children with visual function of 20/800 or worse had lower mean levels of development than children whose visual function was 20/500 or better.

Only 5% of the children with ROP enrolled in the CRYO-ROP study at the 5-year follow-up had visual acuity of 20/200 or worse (Cryotherapy for Retinopathy of Prematurity Cooperative Group, 2002). Children with this unfavorable outcome either had ROP in Zone 1 or in Zone 2 (7 or more clock hours).

**What surgical treatments are used for ROP?**
Because approximately 90 percent of cases of acute ROP spontaneously regress (Flynn, 1991), Flynn suggested that “the burden of proof is then on the advocate of the particular therapy to show that the proffered therapy offers a better prospect than the natural history of the disease” (1991, p.73). Even though 90% of ROP cases regress without intervention, glaucoma and late onset retinal detachment have been identified as late complications of regressed ROP by Ober et al. (2003) who note that ROP presents a life-long threat to vision and that “afflicted patients should be examined at least once each year for life” (p. 613).

Since the early 1980s a number of surgical treatments have been used to arrest the progression of ROP, to prevent the retina from detaching, to reattach the retina, and/or to remove scar tissue that forms within the eye. All of these treatments seek to prevent the loss of vision or to restore useful vision. However, if ROP has progressed to Stage 4B or 5, successful surgery usually results only in light perception or the ability to see hand motions. Surgical treatments reported in the literature include cryotherapy, laser photocoagulation, vitrectomy, and scleral buckling.

*Cryotherapy* is an ablative (tissue-destroying) procedure for use in severe active ROP (Stage 3) that was performed to arrest the progression of the disease in the 1980s and 1990s. It involves repeatedly applying a probe to the surface of the eye to freeze through the wall of the eyeball into the retina (Cryotherapy for Retinopathy of Prematurity Cooperative Group, 2002). The cold temperature destroys a portion of the retina that has not yet developed a supply of blood vessels. The destruction of this part of the retina decreases the growth of abnormal blood vessels, halting the progression of the disease and reducing the possibility of blindness.
The value of cryotherapy in arresting the progression of ROP was proven by the CRYO-ROP study that began in 1986 at 24 medical centers throughout the United States (Palmer, Glynn, Hardy, Phelps, Phillips, Schaffer, & Tung, 1991). Infants with threshold disease (five contiguous clock hours or eight noncontiguous clock hours of extraretinal neovasculatization, Stage 3) in Zone 1 or Zone 2 with associated posterior retinal vascular dilation and tortuosity (in addition to the disease) were randomly assigned to cryoablation of the peripheral affected retina. In infants with bilateral ROP, only one eye was treated and the other eye served as a control. The study ended 9 months earlier than scheduled (January 1988) due to the observed treatment benefit. Cryotherapy lowered the rate of unfavorable outcomes (retinal detachment, retinal fold, or retrolental mass) from 51.4 percent in control eyes to 31.1 percent in the treated eyes (three months after cryotherapy), and from 47.4 percent to 25.7 percent of controls versus treated eyes 12 months after cryotherapy. The study found that Zone 1 disease had a considerably worse prognosis than Zone 2 disease. Cryotherapy in Zone 1 eyes failed in 75 percent of the treated eyes; however, unfavorable outcome was found in 91.7 percent of the control eyes. Urrea and Rosenbaum (1989) noted that cryotherapy does not always prevent blindness, and that complications from general anesthesia and cardiac arrhythmias should be considered. Connolly, Ng, McNamara, Regillo, Vander, and Tasman (2002) reported that cryotherapy results in higher incidence rates and higher levels of myopia than laser photocoagulation, while Quinn and colleagues (2001) reported that eyes treated with cryotherapy were more myopic than control eyes that were not treated.

During the 1990s, laser photocoagulation started to supplant cryotherapy for the purpose of arresting blood vessel development that might lead to retinal detachment. In 1992 Sternberg and colleagues began to recommend laser photocoagulation instead of cryotherapy because it limits damage to adjacent structures, lessens the inflammation and contraction of the vitreous that is stimulated by cryotherapy, decreases serious ocular and systemic complications that result from “greater manipulation required from cryotherapy” (p. 200), and is less cumbersome. McNamara and colleagues (1991, 1992) reported that laser photocoagulation was as effective as cryotherapy, and Ober et al. (2003) support that claim after reviewing a number of studies that compared cryotherapy with laser treatment.

Combined treatment using cryotherapy and laser photocoagulation has recently been suggested by Eustis, Mungan, and Ginsberg (2003), who describe the advantages and disadvantages of each method in a retrospective study of 13 infants. These authors noted that cryotherapy might be best suited to anterior ROP and that media clarity is not an issue with this method; however, cryotherapy may cause damage to other tissues in the eye, may be more painful and may result in more inflammation, requires general anesthesia, is difficult to use in posterior ROP, and results in high levels of myopia. Eustis et al. (2003) note that laser photocoagulation is as effective as cryotherapy, results in less tissue damage and less myopia, and is easier to use with ROP that occurs in the posterior area of the eye. However, laser photocoagulation is more difficult
to use in the anterior of the eye, if the media is unclear, or if the pupil is very small, and it might take longer than cryotherapy. Also, laser photocoagulation has been associated with the development of cataracts, as noted by Eustis et al. (2003), Ober et al. (2003), and Paysse, Miller, Brady-McCreery, and Coats (2002). Eustis et al. (2003) report that combined treatment appeared to be as safe and effective as either method alone. They suggested that combined treatment might be useful for infants with small pupils, media opacities, or anterior disease, and for infants with ROP in the posterior area in order to decrease the time required for surgery.

Vitrectomy is a surgical procedure usually reserved for ROP that has progressed to Stage 4B or 5 and is seen as the “last hope” for restoring vision. Urrea and Rosenbaum (1989) describe closed vitrectomy as 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 to allow the retina to be reattached. Sternberg and colleagues (1992) found “dismal visual results for children undergoing vitrectomy for Stage 5 retinopathy of prematurity” (p. 201). More recently, Ober et al. (2003) discussed vitrectomy and other types of vitreoretinal surgery for advanced ROP and concluded that “persistent retinal dysfunction limits vision even after retinal surgery for severe ROP” (p. 624).

Scleral buckling seems to be reserved for ROP at Stages 4 or 5 and may be the most controversial surgical technique (Urrea & Rosenbaum, 1989). It involves surgically 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. The success rate of scleral buckling was reported to be 20 percent or less in the early 1990s (Noorily, Small, DeJuan, & Machemer, 1992). Ober et al. (2003) describe scleral buckling as a possible treatment for advanced ROP but note that a prospective randomized clinical trial is needed to determine its efficacy for treatment of severe ROP.

Information regarding the long-term visual status of children with acute ROP appears to be conflicting. Several studies admitted that surgery did not necessarily prevent blindness, and light perception and the ability to perceive hand motions may be the best outcome of scleral buckling or vitrectomy. Attempts to use preferential looking to obtain more precise measures of visual acuity have been made and may offer promise for more precise measurement of vision. Restrictions in the visual field of children with ROP have been documented.

Literature regarding ROP from the medical field is overwhelming in quantity, scope, and technical detail. There is a tremendous scarcity of information regarding ROP in the educational field, particularly regarding intervention strategies that might be unique to this group of children.

Handout C was developed by the Blind Babies Foundation (1998) and provides a description of ROP and strategies for working with children and families with this
disorder. Handouts D and E were developed by the CRYO-ROP Multicenter Study of ROP for parents of premature babies and for parents of children with severe ROP (Multicenter Trial of Cryotherapy for Retinopathy of Prematurity, n.d., 1996).

Adonia and Jared sat quietly next to the incubator in the neonatal intensive care unit. Piper, their daughter, was born at 25 weeks’ gestation and weighed 1 pound, 2 ounces. It had been quite an ordeal in the NICU—she had surgery for her eyes and her heart, and she experienced occasional seizures.

Piper now weighed just over 5 pounds and would be going home soon. Even though Adonia and Jared were excited about finally taking their baby home, they were nervous. Piper would still need oxygen at night, she would have to see multiple specialists for continuous follow-up, and they were just beginning to understand the impact of her visual impairment. The ophthalmologist in the NICU had explained that Piper had stage 4 retinopathy of prematurity.

At first, Adonia and Jared worried that treating their baby with oxygen had caused her visual impairment. The doctor reassured them that this was not the case, because the retina usually does not finish developing until a baby is about 32 weeks’ gestation and continues through final development even after babies are born. Because Piper was born early and had developed Stage 4 retinopathy of prematurity, the retina had begun to pull away from the back of her eye. The doctors had completed surgery to help save the retina and improve Piper’s vision. The doctor called the surgery successful, but Adonia and Jared realized that they would not fully understand how and what Piper could see until she was home.

E. Optic nerve hypoplasia (ONH)

Optic nerve hypoplasia (ONH) is often one component of a syndrome known as septo-optic dysplasia (SOD) that may be associated with pituitary disorders such as growth hormone deficiency, thyroid dysfunction, or diabetes insipidus.

Optic nerve hypoplasia is probably the most prevalent congenital optic disorder found in young children with severe visual impairments (Phillips & Brodsky, 2003). Unlike retinopathy of prematurity and cortical visual impairment, which may result from multiple factors, optic nerve hypoplasia is caused by abnormal development of the nerve fibers that make up the optic nerve during fetal development. Specifically, there are fewer fibers in the optic nerves of individuals with this disorder. Optic nerve hypoplasia may affect one eye (unilateral) or both eyes (bilateral). Because this disorder is present at birth, it is unquestionably congenital.

To the eye care specialist, ONH may present as a small optic nerve head that is gray or pale in color and that may be surrounded by a yellow halo bordered by a dark ring of pigment (Phillips & Brodsky, 2003). In addition, the blood vessels in the retina may
appear tortuous or twisted. The visual function of children with this disorder ranges from normal sight to total blindness (Phillips & Brodsky, 2003; Tornqvist, Ericsson, & Kallen, 2002). Often, ONH is diagnosed during an ophthalmological exam and is then confirmed through magnetic resonance imaging (MRI).

ONH is often associated with other malformations of the central nervous system (Parker, Hunold, & Blethen, 2002; Phillips & Brodsky, 2003). Absence of the septum pelidicum, with agenesis (thinning) of the corpus callosum accompanied by small optic nerves, is the basis for a diagnosis of septo-optic dysplasia (SOD). Magnetic resonance imaging (MRI) is typically used to diagnose septo-optic dysplasia. Approximately 45% of children with SOD have additional brain abnormalities such as schizencephaly or periventricular leukomalacia. The absent septum pelidicum and the thin corpus callosum do not cause developmental problems; however, other associated abnormalities of the brain may cause such disorders. Like those with ONH, children with SOD frequently have hypopituitarism and may exhibit the same clinical signs of this disorder as children with ONH alone.

Parker et al. (2002) noted that although the absence of the septum pellidicum is stressed in the diagnosis of SOD, vision loss and hypopituitarism are the two most commonly associated functional problems. In a study of 823 children with SOD, they reported a prevalence of 6.3 per 100,000, with similar rates in girls and boys. A group of 65 children who were not being treated with growth hormone was compared to a group of 582 receiving growth hormone treatments. Thyroid hormone deficiency was present in 8% of the untreated group, as compared to 27% of the treated group, while corticotrophic hormone deficiency was present in 3% of the untreated group and in 24% of the treated group. Approximately 85% of the untreated group were referred due to short stature or decreased growth rate. The authors noted that only 12% of the children in the untreated group had endocrine evaluations, despite the fact that there is a known association between SOD and hypopituitarism and that there have been at least five incidences of sudden death in children with SOD associated with hypopituitarism (Brodsky, Conte, Taylor, Hoyt, & Mrak, 1997).

In an epidemiological study of 156 individuals with ONH in Sweden, Tornqvist et al. (2002) reported that 40% had an isolated diagnosis of ONH. Although approximately 43% of the participants also showed signs of cerebral involvement, 63% had additional impairments, with the most common being a combination of mental and motor impairments. According to World Health Organization criteria for blindness, 32% of the participants were blind. Maternal risk factors associated with ONH included young maternal age, first pregnancy or fourth or later pregnancy, and smoking. Child risk factors included premature birth, small size for gestational age, and low birth weight. In this sample in Sweden, ONH occurred at a rate of 7 per 100,000 births. Although few mothers of children in this study reported drug use, the authors suggested that drugs used for fertility treatment and for depression might be associated with ONH.
Dattani and colleagues (Dattani, Martinez-Barbera, Thomas, Brickman, Gupta, Wales, et al., 2000) and Thomas and colleagues (Thomas, Dattani, Brickman, McNay, Warne, Zacharin, et al., 2001) reported that some cases of SOD may result from mutations in the HESX1 gene and that there is a mouse model for the disorder with phenotypic features that are similar to those seen in humans with the disorder.

Phillips, Spear, and Brodsky (2001) studied the co-occurrence of congenital hypopituitarism with ONH in children and found that 26 of 67 children with ONH had both conditions. Because hypopituitarism is associated with impaired growth, hypoglycemia, developmental delay, seizures, and death, early diagnosis is critical. These authors found that an MRI of the neurohypophysis area in the brain can predict congenital hypopituitarism in children with ONH. The 26 children with both disorders displayed the following characteristics.

<table>
<thead>
<tr>
<th>Condition</th>
<th>Number of children</th>
</tr>
</thead>
<tbody>
<tr>
<td>Decreased growth</td>
<td>20</td>
</tr>
<tr>
<td>Extended neonatal jaundice</td>
<td>11</td>
</tr>
<tr>
<td>Neonatal hypoglycemia</td>
<td>10</td>
</tr>
<tr>
<td>Seizures</td>
<td>9</td>
</tr>
<tr>
<td>Polyuria and polydipsia</td>
<td>5</td>
</tr>
<tr>
<td>Fever with no known cause</td>
<td>5</td>
</tr>
<tr>
<td>Small penis</td>
<td>3</td>
</tr>
</tbody>
</table>

**Table 5. Health Conditions Associated with Combined ONH and Congenital Hypopituitarism** (Phillips et al., 2001)

Although growth hormone deficiency is the most common pituitary problem associated with ONH, it is relatively difficult to diagnose and may not be recognized until children reach 4 years of age. Hypothyroidism, or decreased production of thyroid hormones, may cause prolonged neonatal jaundice, slow growth, and developmental delay. Hypocortisolism, or decreased production of corticotropin hormones, may result in hypoglycemia, low blood pressure, frequent infections, seizures, and developmental delay. Corticotropin hormones are needed to maintain blood pressure, blood sugar, and body temperature. Hypogonadism may cause small penis and delayed or precocious puberty. Damage to the anterior pituitary may result in diabetes insipidus, which places children at risk for dehydration during illnesses and which may occur along with deficiencies in temperature regulation. If pituitary problems are detected early, hormone replacement can be used to decrease the severity of clinical symptoms. Hellstrom, Aronsson, Alexson, Kyllerman, Kopp, Steffenburg, et al. (2000) proposed a process to improve the diagnosis of SOD. They noted that individuals with SOD can be organized into four groups: those with abnormal midline cerebral structures, optic nerve, and pituitary abnormalities; those with midline cerebral abnormalities and ONH; those with
midline cerebral abnormalities and pituitary disorders; and those with ONH and pituitary disorders. Additionally, they described a detailed protocol for diagnosing disorders involving the optic nerve, midline structures of the brain, and pituitary function.

Hatton et al. (2001) reported that ONH was diagnosed in a sample of 31 children at an average age of 4.3 months, and that they were referred for specialized services at 8.1 months on average. Approximately half of the children in the sample were considered legally blind upon entry into specialized programs, and 61% of the children were considered to have a single disability of visual impairment at that time.

Handout F from the Blind Babies Foundation describes optic nerve hypoplasia and strategies for working with young children with this disorder. Additional strategies can be found in the suggested reading by Bahar et al. (2003). Levack et al. (1994) note that high illumination, magnification, high contrast, or Braille/tactile materials might be required for children with ONH. Levack, Stone, and Bishop’s other suggestions include avoiding visual clutter, using simple images that are presented in isolation, avoiding busy backgrounds, and modifying expectations due to fluctuating visual performance.

Damon crawled across the floor to his mother, Linda. She scooped him into her arms and kissed him on the cheek as she walked into the kitchen. Linda remembered how she used to dread this part of the morning. It was time for Damon’s medications, and in the past he had always resisted when it was time to take them.

Damon was born with optic nerve hypoplasia (ONH) and was diagnosed at 2 months with septo-optic dysplasia (SOD). He now takes growth hormones and medication for diabetes insipidus. Linda is especially thankful for the support she receives from Damon’s ECVC, Lynn. Lynn has provided the family with information so that they can understand Damon’s diagnosis, visual abilities, and development. Also, she introduced Linda to other families that have children with ONH and SOD. Talking with other moms, Linda learned helpful tips on how they administer medications to their children. With these ideas, Linda has learned how to make this time of the day a positive experience.

F. Anophthalmia, microphthalmia, and colobomas

Structural abnormalities of the eyes such as anophthalmia and microphthalmia are often diagnosed early because they are evident at birth or shortly thereafter. Colobomas may impact different eye structures and may not be as evident at birth.

Developmental abnormalities of the globe such as anophthalmos (failure of the globe to develop resulting in no eyes) and microphthalmos (abnormally small globe) are usually detected soon after birth and result from a failure of the embryonic fissure to close at about five to seven weeks gestation (Nischal, 2003a). Colobomas also result from a failure of the embryonic fissure to close at around the fifth week of gestation and may affect a number of ocular structures such as the optic nerve, retina, choroid, and iris,
and “are part of a continuum that extends to microphthalmia and anophthalmia” (Nischal, 2003b, p. 431). According to Bateman (2003), the prevalence of coloboma is .26 per 1000 live births.

Anophthalmos is a rare disorder in which there is a complete absence of any tissues of the eye. If any of the structures of the eye are present, then severe microphthalmos, rather than anophthalmos, is diagnosed. Because it is difficult to distinguish these two disorders without radiological scans, “most cases encountered are classified under the term clinical microphthalmos” (Nischal, 2003a, p. 386). Nischal (2003a) and Cook, Sulik, and Wright (2003) suggest that environmental factors such as teratogens probably cause anophthalmos. Ophthalmologic treatment of anophthalmus typically involves enlarging the orbit and fitting artificial eyes for cosmetic purposes.

There are three types of microphthalmia. Bateman (2003) reports that the incidence of microphthalmia is .22 per 1000 live births. Simple microphthalmia describes small eyes that are normal. Nanophthalmos describes small eyes with a normal size lens and a crowded anterior segment; this disorder has been linked to a specific hereditary genetic defect. Complex microphthalmos describes an eye that is malformed and abnormally small (Nischal, 2003a). This condition has a relatively high prevalence of .65 per 1000 cases encountered in a pediatric ophthalmologic clinic and probably accounts for two-thirds of all cases of microphthalmia. Complex microphthalmos is usually bilateral, and visual function can range from normal to no vision at all. Microphthalmos may occur alone or as part of a systemic condition such as CHARGE syndrome, in which colobomas and/or microphthalmos occur along with at least two other features that might include heart defects, atresia choanae (structural defect between nasal passage and throat), retarded growth and development, central nervous system abnormalities, hypogonadism, ear abnormalities, and/or deafness.

For children with colobomas, Levack et al. (1994) suggest that magnification, average or bright light with no glare, telescopes, sunglasses, visors, and use of high contrast might be helpful in facilitating optimal use of vision. For children with microphthalmos, Levack et al. recommend average or bright light with no glare, good contrast, and possibly magnification.

Dajah was born at full term without complications to a mother with a typical and healthy pregnancy. Before Dajah left the hospital, she was diagnosed with anophthalmia in her left eye and colobomas of the iris, choroid, and retina in her right eye. The hospital where Dajah was born immediately referred the family to the local division of services for the blind.

When Dajah was 6 weeks old, the family met with a social worker and a ECVC who helped the family understand these two specific visual conditions. The family had fears and concerns about how their daughter would appear to others because she had only one eye. The ECVC and social worker explained to the family that they could find an
ophthalmologist who would refer them to an ocularist to fit Dajah with a conformer until she was old enough for a prosthetic eye. The ECVC explained that colobomas are like having holes in those parts of her eye. The family now understands that colobomas result from failure of parts of the eye to fully develop within the first few weeks of pregnancy. Additionally, the ECVC discussed how she could come to their home and support the family in understanding Dajah’s visual abilities and make suggestions for adaptations, when necessary, to facilitate Dajah’s optimal use of vision as she develops.

G. Albinism

Albinism is often diagnosed at early ages and is typically associated with relatively useful vision and with photophobia. Children with albinism typically do not have additional disabilities.

Albinism is an absence or reduction in the pigment or melanin in the skin, eyes, or both (Traboulsi, 2003). Ocular albinism and oculocutaneous albinism are genetic disorders that may 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. According to McManaway and Bonsall (2003), there are two types of oculocutaneous albinism and three types of ocular albinism. Oculocutaneous albinism type 1 (OCA1) results from a defect in the tyrosinase gene and is also known as tyrosinase-negative oculocutaneous albinism, while oculocutaneous albinism type 2 (OCA2), also known as tyrosinase-positive oculocutaneous albinism, results from a defective P gene located on chromosome 15 that produces a transmembrane protein necessary for normal pigmentation.

Although OCA1 is relatively easy to diagnose due to a lack of pigment in the hair, skin, and eyes, other forms of albinism may be more difficult to diagnose. According to Traboulsi (2003), albinism is the leading cause of nystagmus in young males. Therefore, McManaway and Bonsall (2003) suggested that albinism should be suspected in young males who present with nystagmus. The three types of ocular albinism described by McManaway and Bonsall include OA1, which results from a genetic defect on the short arm of the X chromosome and which can be diagnosed using a skin biopsy. The second form of ocular albinism, OA2, is found primarily in natives of the Aland Islands (Finland), while OA3 is autosomal recessive due to a genetic defect in the P gene on Chromosome 15.

Traboulsi (2003) noted that the overall prevalence of albinism is 1 in 20,000, while the prevalence of OCA1 is 1 in 39,000 in Whites and 1 in 28,000 in Blacks. Approximately 1 in 50,000 individuals has ocular albinism. Photophobia is often identified as a major characteristic of individuals with albinism. However, Traboulsi noted that not all individuals with albinism are photophobic and that sunglasses can further reduce visual acuity. Corrected visual acuity often ranges from 20/100 to 20/200; however, some
individuals with OCA1 may have visual acuity of 20/40 (Traboulsi, 2003). Some individuals with albinism have defects of the optic nerve near the optic chiasm that cause difficulties with depth perception.

Hatton et al. (2001) reported that the 19 children with albinism in their sample were diagnosed at an average age of 3.4 months with referral to a specialized agency at 11.7 months. Approximately 42% of the children were legally blind, only one of the 19 children had multiple disabilities, and none of them had associated health disorders.

Levack et al. (1994) and Bishop (1986) suggest several options for working with children with albinism:

- magnification;
- allowing children to move closer to objects;
- high contrast;
- tinted lenses, sunglasses, visors;
- diffused lighting positioned behind the child;
- reducing glare; and
- telescopes.

Handout G from the Blind Babies Foundation (1998) defines albinism and provides strategies for working with young children with this condition.

Wayne and Gail planned a picnic at the park for their family. Their youngest son, Ross, has oculocutaneous albinism like his father. Before leaving, Gail made sure to pack Ross’s sun visor and sunglasses. Their last trip to the park had not been much fun for Ross because his sun visor had been left at home. Ross was unable to play with his siblings in the grass because the sun was too bright, and he could not open his eyes fully. Gail also made sure that Ross had a lightweight, long-sleeved shirt to cover his sensitive skin. The last thing she packed was a yellow-and-black soccer ball. The ball provides sufficient contrast so that Ross can chase the ball with his siblings.

H. Retinoblastoma and other retinal disorders

Retinal disorders such as Leber’s congenital amaurosis may be difficult to diagnose. Retinoblastoma results from malignant tumors on the retina and presents families with special challenges.

In Wright and Spiegel’s (2003) pediatric ophthalmology textbook, ten chapters are devoted to pediatric retinal disease: heritable disorders; retinitis pigmentosa; disorders of the vitreous; retinal vascular disorders; nonvascular hamartomas (noncancerous tumors); retinoblastoma and malignant intraocular tumors; retinopathy of prematurity; infectious, inflammatory, and toxic diseases of the retina and the vitreous; myopia; and patterns of retinal disease. Retinopathy of prematurity, one of the three most prevalent...
disorders in young children with severe visual impairments, has been discussed in detail in Major Point D. Because an exhaustive description of retinal disorders in young children is beyond the scope of this session, we will confine our discussion to Leber’s congenital amaurosis and retinoblastoma.

**Leber’s congenital amaurosis** (LCA) is a congenital autosomal recessive disorder with an incidence of 1 in 33,000 that results in severe visual impairment (Eibschitz-Tsimhoni, 2003). Infants with LCA typically develop nystagmus in the second or third month and have a sluggish pupillary response. Visual function can range from 20/200 to no light perception. In infancy, the fundus and retina may appear normal, so an electroretinogram is required for a definitive diagnosis. As the children get older, the optic disk may appear pale and there may be changes in the pigment of the retina (McManaway & Bonsall, 2003). Some children with LCA may have cognitive impairments, hearing loss, kidney disorders, or growth deficiency.

According to Drack and Kimura (2003), the prevalence of children with LCA with accompanying neurological disorders ranges from 17% to 37%; however, most have normal intelligence. Eye poking, nystagmus, and roving eye movements may be present in children with LCA.

Levack and colleagues (1994) noted that children with LCA may use peripheral vision to view objects and that magnification or bringing materials closer might be helpful. In addition, she suggests that diffused light, telescopes, tinted lenses, sunglasses, and visors might be helpful, as well as high contrast materials and adjustable lighting.

**Retinoblastoma** is a malignant tumor within the eye that is fatal if left untreated. It is the most common type of ocular malignant cancer during childhood. Parents often detect the condition by noticing a white reflection in the child’s pupil. Strabismus may be another sign of retinoblastoma. According to Buckley (personal communication, October 29, 2003), about one-third of the cases of retinoblastoma are due to inherited genetic defects while two-thirds are due to spontaneous genetic mutations. In the inherited cases, only about 25% have a family history of retinoblastoma. Most cases of unilateral retinoblastoma are not inherited. Individuals with bilateral retinoblastoma have a lifetime risk of 15% to 20% for developing other nonocular malignant tumors.

Retinoblastoma is usually diagnosed within the first one to two years of life, but it can be diagnosed in older children as well (Murphree & Christensen, 2003; Shields & Shields, 1998). Prevalence rates (number of cases diagnosed each year) are approximately 11 cases per one million children under five years of age while incidence rates (number of babies born with a disorder per year) range from 1 in 12,400 to 1 in 20,000 (Moore, 2000; Murphree & Christensen, 2003). Murphree and Christensen suggest that unilateral retinoblastoma may be more prevalent in underdeveloped countries such as Mexico, India, countries in Central America, and countries in central Africa.
If left untreated, retinoblastoma will spread from the eye to the orbital tissue and into the brain. It can also spread to the bone and bone marrow. After retinoblastoma is diagnosed clinically, an imaging study is used to confirm the diagnosis. Murphree and Christensen (2003) note that current treatment typically involves systemic chemotherapy followed by consolidation treatment, in which local surgery—laser therapy or cryotherapy—is used to completely destroy any remaining malignant cells. In some cases in which there are large, advanced tumors, enucleation of the eye may be warranted (Murphree & Chistensen, 2003). Moore (2000) notes that it is not yet clear whether systemic chemotherapy is as effective as radiation therapy or whether chemotherapy will have more side effects. Genetic counseling is important for children and families with retinoblastoma to clarify reproductive risks.

Levack et al. (1994) note that children with retinoblastoma may have difficulties with depth perception. Handout H from the Blind Babies Foundation (1998) describes a number of retinal disorders, including Leber’s and retinoblastoma.

I. Congenital cataracts

_Cataracts_ are opacities or cloudiness in the lens of the eye. They may affect one eye (unilateral) or both eyes (bilateral), and they may be present at birth (congenital) or acquired after birth. Cataracts may occur in isolation or they may occur with other impairments in systemic disorders.

According to Buckley (1998), the impact of a cataract on visual function depends on the age of onset, the location of the cataract, and its morphology or structure. Zonular cataracts occur in a particular area or zone of the lens, and lamellar cataracts are the most common type of zonular cataracts. They often result from damage to the fetus while in utero and commonly affect both eyes. Infant hypoglycemia and galactosemia (metabolic disorders) may cause this type of cataract. This type of cataract may be progressive and require surgery in infancy or early childhood. Nuclear cataracts are usually bilateral, may be very dense, may be associated with microphthalmos and microcornea, and may result from an autosomal dominant genetic disorder. Sutural cataracts are near the periphery of the lens, may be unilateral or bilateral, may be X-linked or autosomal recessive, and typically do not cause severe visual impairments. Subcapsular cataracts occur near the front or back of the lens and usually occur after birth. Subcapsular cataracts that are near the anterior (front) of the lens usually do not impact vision; however, posterior cataracts (near the back of the lens) usually will cause significant loss of vision. Polar cataracts involve the lens capsule or the area under the lens capsule. Anterior polar cataracts are usually in the center of the anterior capsule, are small and white, result from irregularities in the separation of the lens during embryonic development, and may be inherited as an autosomal dominant trait. Some have no impact on vision while others may progress and may occur with strabismus, amblyopia, or anisometropia. Posterior polar cataracts are found at the back of the eye near the lens capsule; they are often associated with aniridia and, due to their central
location, with significant visual loss. A total cataract describes a lens that is completely opaque and that therefore usually requires immediate removal. Any of the previously described types of cataract may become total cataracts. A membranous cataract is a thin, fibrous lens often associated with trauma, congenital rubella, or Lowe syndrome.

Bilateral cataracts may be associated with a systemic disorder. Unless they are inherited as an autosomal dominant trait, they often indicate a need for additional medical tests. 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 special challenges due to the risk of amblyopia. If nystagmus is present before surgery, visual function of 20/60 to 20/80 is typical after surgery. According to Buckley (1998), it is sometimes difficult to determine whether nondense cataracts are significantly impacting vision; however, “the larger, denser, and more centrally located the cataract, the greater the resultant visual impairment” (p. 269). After surgery, corrective lenses must be fitted for near vision because the lens of the eye is no longer present for accommodation. Wright (2003d) recommends contact lenses for children under one year of age and intraocular lenses for children who are 2 years or older. Buckley (1998) notes that, for children from whom the lens of the eye has been removed, aphakic glasses are the safest choice for corrective lenses; contact lenses are the most common method of correction following bilateral or unilateral cataract removal.

The visual prognosis for children with cataracts is not as good as it is for adults with cataracts (Buckley, 1998). However with early surgery, careful optical correction, and aggressive treatment of amblyopia, optimal visual outcome is much more likely.

For children who are aphakic (lens of the eye has been removed), Levack et al. (1994) suggest that magnification, bringing materials closer, and adjustable lighting might be helpful. Strabismus may sometimes be associated with cataracts, and so a patching (penalization) regimen may be prescribed for some children to prevent amblyopia. (See Session 5 for more information on patching.)

Jaedon rested peacefully in her father’s arms. She was born with bilateral posterior cataracts, a condition inherited from the maternal side of her family. She had undergone surgery the day before to remove the cataracts. The doctor explained that Jaedon’s cataracts were near the back of the lenses and were large and very dense, with the potential to cause significant visual impairment if they were not removed. Jaedon is 3 months old and will now wear aphakic glasses to compensate for the lenses that were removed from her eyes. These glasses will help Jaedon focus on objects at close range.

J. Strabismus, amblyopia, glaucoma, nystagmus, and refractive errors
Strabismus is a misalignment of the eyes that results from muscle imbalances. It is relatively common and is often associated with refractive disorders, and has a prevalence of 3% to 5% (Vivian, 2000). Strabismus can occur in association with other visual disorders that result in decreased vision, such as retinopathy of prematurity or albinism.

According to Olitsky and Nelson (1998), strabismus can be constant (tropias) or intermittent (phorias) and can affect near or distance vision. Typically, one eye fixates on objects and the other eye drifts in, out, or vertically. Alignment of the eyes typically cannot be assessed until about 3 months of age; deviations noticed during the first 3 months may not be abnormal.

In horizontal strabismus, the eyes may turn in toward the nose (eso or convergent deviations) or outward toward the temple (exo or divergent deviations). Vertical deviations are denoted by the prefix hyper (Wright, 2003a).

Esotropia is much more common than exotropia and typically requires surgical correction. However, any refractive errors or amblyopia should be treated before surgery (Vivian, 2000). Exotropia diagnosed within the first year of life is rare and may be associated with ocular or neurological disorders. Exotropia usually first appears around the age of 2 or 3 years and begins intermittently. Amblyopia is relatively rare in these cases. If amblyopia is present, it should be treated with patching before surgery. (See Session 5, “Using Assessment Results in Intervention.”) Orthoptic exercises may help in the treatment of intermittent exotropia by increasing motor fusion potential (Vivian, 2000). Corrective lenses can also help some children with exophoria. Olitsky and Nelson (1998) note that surgery, corrective lenses, base in prisms, patching, and orthoptic exercises may be used to treat exotropia.

For all types of strabismus, early treatment is critical to assure that clear images are focused on the fovea. Consequently, aggressive treatment during the first two years of life appears to be recommended universally to assure that optimal visual potential (acuity and binocular vision) is achieved (Olitsky & Nelson, 1998; Vivian, 2000; Wright, 2003b; 2003c). Handout I from Prevent Blindness America (2003b) describes strabismus in detail.

"Come here, my little sweet potato," called Selia. She walked toward her toddler, Klyde, who was smiling as he scooted away from her. He had once again pulled his glasses from his face and was moving away from his mother. Without his glasses, Klyde’s left eye turned in toward his nose. When he was about 6 months old, Selia noticed that Klyde’s right eye appeared to turn out and she mentioned it to her pediatrician, who recommended a visit to the ophthalmologist. The ophthalmologist said that Klyde had exophoria. Selia thought at first that Klyde was too young for glasses, but the ophthalmologist reassured her that wearing glasses now would help prevent amblyopia, which could occur if Klyde continued to be unable to use his right eye efficiently. Even
though it was difficult persuading him to keep his glasses on, Selia knew that treatment was critical to assure that Klyde could see clearly and to prevent delays in his development.

Amblyopia refers to a reduction of visual acuity in the absence of abnormal ocular structures or of other organic conditions. It is the leading cause of monocular (affecting one eye) visual impairments in children and in young and middle-aged adults (Pediatric Eye Disease Investigator Group, 2002). In most cases, amblyopia is not associated with severe visual impairments, so ECVCs and OMSs probably would not have children with a primary diagnosis of amblyopia on their caseloads. Most cases of amblyopia involve decreased vision in one eye, with normal vision in the other. In most states, eligibility for services for individuals with visual impairments is based on visual acuity in the better eye, making most individuals with amblyopia ineligible for services from ECVCs and OMSs.

Amblyopia results from a lack of visual stimulation via clear, focused images, and is the most common cause of decreased vision in childhood (Wright, 2003e). Children are susceptible from birth to 7 years; the earlier the onset, the greater the vision loss. Amblyopia is present when there are at least two Snellen lines of difference in visual acuity between the eyes (e.g., 20/20 and 20/40, 20/30 and 20/50). Kushner (1998) questions the two-line difference criterion, noting that more subtle differences in vision can still result in amblyopia. Children with amblyopia have difficulty reading the letters on line-acuity optotype charts due to a crowding phenomenon, and do much better with single optotypes rather than linear optotypes. Therefore, children with amblyopia should be assessed with linear optotypes so that the amblyopia is not underestimated. Handout J from Prevent Blindness America (2003a) describes amblyopia in detail. Levack et al. (1994) note that children with amblyopia need high contrast and reduced glare, and that depth perception can be a challenge.

According to the Pediatric Eye Disease Investigator Group (2002), amblyopia is typically associated with strabismus such as esotropia in early childhood or, in some cases, with “anisometropia (differences in refractive error between the two eyes), a combination of strabismus and anisometropia, or with visual deprivation. About 25% of patients have a visual acuity in the amblyopic eye worse than 20/100, and about 75% have an acuity of 20/100 or better” (p. 268). Treatment of amblyopia is usually most successful if implemented before 8 years of age.

According to Kushner (2002), functional amblyopia can be treated by correcting any refractive errors, and then patching or occluding the good eye until visual function improves to normal in the affected eye. Treatment is more likely to be successful if begun early, and if there is reasonably good visual acuity in the amblyopic eye.

In 1998, Kushner reported successful treatment of some children with amblyopia with the following co-occurring ocular disorders: partial cataract, iris/choroidal coloboma,
albinism, optic nerve hypoplasia, disk coloboma, and congenital nystagmus. He recommended a gradual tapering of the occlusion after the amblyopia has been corrected, rather than the abrupt cessation of patching, which has sometimes been associated with a recurrence of amblyopia. Strategies for successful occlusion are included in Session 5 of the Visual Conditions module.

Pharmacological penalization is another treatment option for amblyopia that involves blurring the vision in the good eye by administering eye drops such as atropine. Atropine is typically used to dilate eyes during eye exams by impeding accommodation. For atropine to be effective, however, the vision in the amblyopic eye must generally be 20/100 or better. Therefore, pharmacological penalization may not be appropriate for children with severe visual impairments who are typically served by ECVCs and OMSs.

The Pediatric Eye Disease Investigator Group (2002), in a recent multicenter randomized trial of occlusion therapy versus pharmacological penalization, reported that, for children whose corrected visual acuity in the amblyopic eye was between 20/40 and 20/100, both treatments are effective. This group is currently conducting a randomized trial in children whose visual acuity is 20/100 or worse in the amblyopic eye. The results of that trial may be more relevant for children served by ECVCs and OMSs.

Kaye et al. (2002) suggest that combined optical and atropine penalization may be most effective for some children with amblyopia. The Pediatric Eye Disease Investigative Group (2003) also conducted a randomized trial of patching regimens for children with moderate amblyopia. This group reported that patching for two hours each day combined with near-vision tasks during one of those two hours was as effective as patching for six hours each day. In all cases, researchers report that compliance with either patching or atropine penalization is the key to successful treatment of amblyopia.

Glaucoma refers to a group of disorders in which the pressure inside the eye increases, potentially damaging the optic nerve and retina. It usually results from structural abnormalities in the angle structures and the anterior segment that interfere with the drainage of aqueous fluid from the eye. Pediatric glaucoma is quite different from adult glaucoma (Reynolds & Olitsky, 2003). The three major types of pediatric glaucoma are primary infantile, also called open-angle or congenital glaucoma; juvenile glaucoma; and secondary glaucoma. Primary infantile glaucoma results from angle anomalies alone, with no other malformations or associated diseases. Juvenile glaucoma is essentially late-onset (beginning at 5 years of age) primary infantile glaucoma. Any pediatric glaucoma associated with a systemic disease or otherwise acquired is considered a secondary glaucoma. In addition to various diseases and syndromes, secondary glaucomas can be associated with lens-iris abnormalities or persistent hyperplastic primary vitreous; with inflammatory disorders such as juvenile rheumatoid arthritis or rubella; with tumors such as retinoblastoma; with trauma; or with drugs (steroids). Children with glaucoma should be monitored closely to prevent damage to the optic nerve or the retina from high pressure.
Signs or symptoms of glaucoma include corneal opacities and corneal enlargement, large or bulging eyes, photophobia, optic nerve cupping, amblyopia, strabismus, and anisometropia (differences in refractive error in the two eyes). Treatment can include medication to lower the pressure in the eye or surgical intervention to permit the aqueous fluid to drain and to normalize the pressure within the eye (Reynolds & Olitsky, 2003; Walton, 1998).

According to Levack et al. (1994), children with glaucoma may have inconsistent vision. ECVCs should be alert to signs of increased pressure that would require medical intervention. Sunglasses, adjustable lighting, high contrast, and magnifiers might be helpful for some children with glaucoma.

Darby, the ECVC, arrived at Levi’s IFSP meeting with brochures about childhood glaucoma. Levi had previously been diagnosed with glaucoma and strabismus, and the team, including the family, had made great progress in facilitating Levi’s development. Darby wanted to make sure that each team member knew the basic facts about glaucoma, including signs and symptoms of increased pressure in Levi’s eyes. Darby knew that if all the team members were aware of such symptoms, they would be better prepared to prevent further damage to Levi’s eyes that could lead to serious visual impairment or even blindness. Darby also wanted to discuss the importance of continuing to monitor Levi’s use of vision to determine environmental modifications. Glaucoma in addition to strabismus could have further implications for the types of lighting needed and the contrast used in the materials found in his daily routines.

Nystagmus is an involuntary oscillation of one or both eyes (Awner, & Catalano, 1998; Hertle, 2003). According to Awner and Catalano (1998), most nystagmus is associated with decreased vision resulting from ocular disorders within the first two years of life. Therefore, nystagmus is a secondary disorder in most cases. A primary diagnosis of nystagmus is made only when no ocular disorder can be identified.

If nystagmus is conjugate, both eyes move together synchronously; if it is disconjugate, then the eyes move separately. If the nystagmus is pendular, then the movements are of equal speed and direction; movements described as jerky will move faster in one direction and slower in the other. Most clinicians describe jerk nystagmus during its fast phase, indicating the direction of the fast gaze and whether the oscillating movement is horizontal, vertical, oblique, rotary, or a combined form.

Latent nystagmus is a jerk nystagmus that is present only during monocular occlusion. If nystagmus is present in both eyes, it is called manifest latent nystagmus. Strabismus (usually esotropia) is almost always present. Individuals with this type of nystagmus have poor or absent fusion, and may have much worse monocular than binocular visual acuity because of the nystagmus. Manifest latent nystagmus is similar to latent nystagmus and is present without occlusion. It is usually seen when one eye has
significant visual loss early in life. Eye movement recordings may be required to distinguish latent nystagmus/latent manifest nystagmus from infantile nystagmus syndrome (Hertle, 2003).

Hertle (2003) lists more than 40 types of nystagmus. However, he suggests that most types can be classified as either infantile nystagmus syndrome, latent nystagmus, manifest latent nystagmus, or acquired nystagmus. Previously called congenital ocular motor nystagmus, infantile nystagmus syndrome (INS) is typically diagnosed during the first few months of life and may present as a single disorder or may be associated with other visual disorders such as albinism, ainarid, optic nerve hypoplasia, and retinal disorders such as Leber’s congenital amaurosis and achronmatopsis. INS worsens during fixation and when the child is anxious. Children with INS often hold their heads in atypical postures in order to decrease the rate of nystagmus. About half of the children with INS also have strabismus and refractive errors. Typically, INS will decrease, but will persist as the child gets older. INS has an accelerating slow phase of movement that distinguishes it from other types of nystagmus. The most often cited incidence of INS is 1 in 6,550 (Hertle, 2003). Frequently, INS is characterized by a slow phase and a fast phase, and by horizontal movements. Children with this disorder often have a head tilt that is used to keep the eyes in a position to establish and maintain a null point at which the nystagmus is minimized. This head tilt stabilizes the image. The exact cause of this disorder is not known; however, the disorder results from an inability to smoothly calibrate eye movements during fixation. According to Hertle (2003), the defect that causes nystagmus can arise during conception (retinal disorders that are inherited), during early prenatal development, or after birth.

According to Awner and Catalano (1998), individuals with isolated cortical visual impairment do not have nystagmus, while individuals with decreased or absent vision usually do have nystagmus. Searching nystagmus (roving horizontal movements without fixation) is often associated with visual acuity worse than 20/200 and is seen in patients with Leber’s congenital amaurosis or optic nerve hypoplasia. Pendular nystagmus (both eyes move together at about the same speed in each direction) is often associated with visual acuity better than 20/200 in at least one eye and may co-occur with loss of central vision, albinism, achronmatopsis, and congenital stationary night blindness. Latent and manifest latent nystagmus are associated with strabismus.

Because most nystagmus is accompanied by other visual disorders, a thorough ocular examination is required. If no anomalies are detected in the ocular structures during clinical examination, then electroretinograms (to detect retinal disorders such as Leber’s or achronmatopsis) or MRI exams (to detect neurological insults or tumors) should be completed. Acquired nystagmus that is diagnosed after the first few years of life is almost always associated with neurological disorders and therefore requires careful follow-up (Hertle, 2003).
Treatment for nystagmus can include surgery on the eye muscles to lessen head tilt or eccentric gaze, or to correct strabismus (Hertle, 2003). Surgical management of nystagmus is aimed at shifting the null point by manipulating the muscles that control the eye so that atypical head positions or eye gaze would no longer be necessary. Corrective lenses, including special prism spectacles, might be used to treat refractive errors or muscle imbalances (strabismus) or to dampen the oscillating movements that result from nystagmus. In most cases, children with nystagmus should not be discouraged from using head tilts or eccentric gaze because these behaviors may allow them to achieve a null point, reducing the involuntary eye movements associated with nystagmus and results in improved visual acuity. Levack et al. (1994) further recommend high contrast and sufficient lighting.

_Perry, the ECVC, was observing 6-month-old Quinn, who has optic nerve hypoplasia. Perry had worked with Quinn for about 3 weeks and was completing Quinn’s functional vision assessment. As Perry held Quinn in his lap and spoke quietly to him, he observed that Quinn had nystagmus. As he continued to talk and play babbling games with Quinn, he noticed that Quinn would turn his head slightly to the left and downward. Perry made note of this null point on his assessment sheet, drawing Quinn’s eyes and marking the position of his null point. Perry also recorded that the nystagmus was conjugate (both eyes), pendular (equal speed and directions), and horizontal. He described this to Quinn’s mother as Quinn’s eyes moved together horizontally in equal speeds and directions. He also explained that the null point refers to the place where the nystagmus slows and where Quinn is able to optimize his vision._

_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, and may result in _hyperopia_, _myopia_, or _astigmatism_. If the eyeball is too long or the cornea and lens have bending powers that are too strong, the focal point of light entering the eye will be in front of the retina, resulting in myopia (nearsightedness). Children with myopia have difficulty focusing on distant objects; they may squint, ignore things in the distance, and experience frequent headaches. If the eyeball is too short or the bending power of the lens is too weak, the focal point will be behind the retina, resulting in hyperopia (farsightedness). Children with hyperopia have difficulty focusing up close and often experience nausea, headaches, dizziness and rubbing of the eyes. Infants are generally born hyperopic, but as the eyes grow, the hyperopia decreases. Astigmatism occurs when the cornea is not spherical but unevenly rounded, causing blurred vision both at a distance and up close. Only part of an object may be in focus at any one time. Astigmatism can occur in combination with myopia or hyperopia.

Eye care specialists can detect refractive errors during standard examinations. Uncorrected refractive errors can lead to amblyopia (lazy eye), while severe myopia can cause detached retinas, cataracts, opacities of the vitreous, and choroidal hemorrhages. See Handout M for further information on refractive errors.

**K. Delayed visual maturation (DVM)**
Some children who are diagnosed with limited vision during the first few weeks of life experience dramatic improvement in visual function before their first birthday. This unexplained limited vision followed by rapid improvement to normal levels has been called *delayed visual maturation (DVM)*. Interestingly, neither Wright and Spiegel (2003) nor Nelson (1998) include DVM in their pediatric ophthalmology textbooks. Elston (2000) describes DVM in his chapter on visual pathway disorders and notes that it is a diagnosis of exclusion that can only be made in retrospect after an infant diagnosed with poor vision shows normal development of vision. Russell-Eggitt, Harris, and Kriss (1998) provide an overview of DVM and the confusion regarding its diagnosis, and also note that it can be confirmed only retrospectively.

Typically, infants with DVM are seen at 6 to 8 weeks of age, after their parents notice that they do not make eye contact or visually attend. DVM may appear as an isolated disorder or with systemic disorders or developmental delays; the disorder may also accompany albinism. Usually, electroretinograms (ERGs) and visual evoked potentials (VEPs) are normal in children with DVM. Elston notes that delayed visual maturation can be differentially diagnosed from CVI if visual function improves and if the child appears to be developing typically. In true cases of DVM, improvement in vision starts at about 12 to 15 weeks of age.

Hodgins and Harris (2000) describe DVM as a pediatric eye movement disorder and note that it is named in infants who have poor vision in the absence of ocular disorder, but whose vision spontaneously improves to normal or near-normal levels. Consistent with Tresidder, Fielder, and Nicholson (1990), Hodgins and Harris describe three types of DVM among young children.

- **Type I DVM**, called idiopathic or isolated DVM, includes children who have normal general/neurological development with no underlying pathology. Between three and six months of age, infants with Type 1 experience a spontaneous and rapid improvement in vision to normal or near normal levels. If the child’s vision improves to normal levels by 3 to 6 months of age, a diagnosis of delayed visual maturation may be made in retrospect as there was no other reason or rationale for the infant’s lack of visual responsiveness.

- **Type II DVM** is associated with systemic disorders and mental retardation. Vision usually improves but it may take longer and there may be some continued lack of vision (Russell-Eggitt et al., 1998).

- **Type III DVM** is associated with other ocular diseases such as oculocutaneous albinism, cataracts, or aniridia. Vision is worse than would be expected from the existing disease alone and the mean age of visual recovery is 20 weeks (Russell-Eggitt et al., 1998). Interestingly, the onset of nyctagmus 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.

**References for Major Points**


Brodsky, M.C., Fray, K.J., & Glasier, C.M. (2002). Perinatal and subcortical visual loss:


