
Etiologies and Characteristics of Deaf-Blindness

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Preface

There are several disorders, syndromes, infectious diseases, and adventitious conditions that may result in an individual being deaf-blind. As a result of these various etiologies, an individual who is deaf-blind may exhibit a range of vision and hearing losses. Sensory loss may range from mild impairment to total loss of the ability to see and/or hear. Usually, one sensory impairment is more prominent than the other. At times, the condition may be a progressive one. Due to the subtly and progressive nature of some visual and auditory impairments, it is paramount that individuals working with persons with deaf-blindness have an understanding of these various disorders in order to effectively observe and assess for sensory loss. By being aware of the possibility of auditory and visual impairments and the characteristics associated with them, appropriate assessment, educational interventions and adaptations may be implemented in the classroom, home and community.

This monograph has been developed to assist State 622C Coordinators, administrators, educators, related staff and parents to gain a better understanding of the terminology and major causes of deaf-blindness. This monograph has been divided into two sections. The first section covers three chapters that provide basic information relating to definitions, anatomy and common disorders. The first chapter on Definitions and Terminology for Deaf-Blindness, will provide information on who is considered deaf-blind. Additional information will be given on terms commonly found in the vision and hearing literature that are used to describe specific visual and auditory impairments. In the second chapter, Normal Anatomy of the Eye and Ear, the different anatomical parts of the eye and ear and how they function are explained. This should assist the reader in understanding impairments that are discussed in later chapters. In Chapter 3, Common Disorders of the Eye and Ear, various disorders that frequently are referred to in Section II of the monograph are explained in detail.

In Section II of the monograph (Chapters 4 through 8) information is provided on specific conditions and syndromes that may result in visual and auditory impairments. For those individuals who are involved in maintaining the state census on students with deaf-blindness, the chapters are set up to closely resemble the categories found in the federal census. (The census refers to the annual identification and counting of individuals with deaf-blindness which occurs in each state and qualifies the student with deaf-blindness to additional technical assistance activities). Conditions referred to in the census can be located in this section as well as other possible etiologies (see Table 1). This monograph divided these conditions and syndromes into five chapters: Rare Syndromes, Congenital Infections and Other Teratogens, Prematurity & Small for Gestational Age, Adventitious Conditions, and Other Causes of Deaf-Blindness. Each chapter contains information regarding etiology, general characteristics, specific visual and hearing impairments and outcomes for each specific syndrome and condition.

Major Causes of Deaf-Blindness

A. Syndromes

1. Down's
2. Trisomy 13
3. Usher's
4. Other

B. Multiple Congenital Anomalies

1. CHARGE association
2. Fetal alcohol syndrome
3. Hydrocephaly
4. Maternal drug abuse
5. Microcephaly
6. Other (specify)

C. Prematurity as sole known cause

D. Congenital Prenatal Dysfunction

1. AIDS
2. Herpes
3. Rubella
4. Syphilis
5. Toxoplasmosis
6. Other (specify)

E. Post-natal Causes

1. Asphyxia
2. Encephalitis
3. Head injury/trauma
4. Meningitis
5. Stroke
6. Other (specify)

F. Other (specify)

Table 1 Major causes of deaf-blindness

For readers who are using this monograph to assist them with the census, there are a few deviations regarding the categorization from the federal census categories. All syndromes on the census are located in Chapter 4 on syndromes in this monograph. However, CHARGE association, which is located under multiple congenital anomalies on the census, is located under syndromes (although it is not technically a syndrome, it is commonly grouped with them). On the census, congenital prenatal dysfunction and multiple congenital anomalies are grouped together in Chapter 5 on Congenital Infections and Other Teratogens. Prematurity on the census is found in Chapter 6 on Prematurity and Small for Gestational Age. All post-natal causes on the census are found under Adventitious Conditions (Chapter 7).

Section I

Foundations

Prior to learning about various etiologies and educational considerations of teaching individuals who are deaf-blind, it is important to have a solid understanding of the definitions, key terms, anatomical structures, and disorders which are encountered in the deaf-blind field. This section will provide a foundation of information to assist the reader in understanding basic terminology and disorders.

Chapter 1

Definitions and Terminology Pertaining to Deaf-Blindness

A broad array of definitions is used across the nation in identifying infants, toddlers, children and youth as having concomitant visual and hearing impairments. Many of these definitions are based on a functional orientation which permits examination of the learner's sensory impairments in regard to their impact in particular educational environments, such as a program designed for students with a vision or hearing loss. Most states have adapted a definition similar to the one cited in IDEA (Individuals with Disabilities Education Act). This definition states:

...the term "deaf-blind", with respect to children and youth, means having auditory and visual impairments, the combination of which creates such severe communication and other developmental and learning needs that they cannot be appropriately educated in special education programs solely for children and youth with hearing impairment, visual impairment, or severe disabilities, without assistance to address their educational needs due to these dual, concurrent disabilities (IDEA).

Other states use definitions that are medically oriented and often include such descriptive diagnostic information as acuity and field loss for describing visual loss, and the particular frequencies and decibel levels of loss for an auditory impairment. Other technical terms (e.g., sensorineural loss, low vision) may be included as well.

To more fully understand these definitions and how they apply to individuals with deaf-blindness, a discussion regarding the terminology and definitions used in the field of vision and hearing ensues.

Terminology Describing Visual Impairments

Several terms may be used to describe the type of visual impairment a person with deaf-blindness experiences. Some of these are legal definitions while others are derived from the educational field.

Legal Definitions

Legal blindness

Central visual acuity of 20/200 or less in the better eye after correction or central visual acuity of more than 20 / 200 if there is a visual field defect in which the peripheral field is reduced to an angle of 20 degrees or less in the better eye (Koestler, 1976, p. 45).

Partially sighted (or partially seeing): Visual acuity between 20/70 and 20/200 after correction in the better eye. (Although this term is used in many state definitions, it is considered outdated and is not being used by practitioners).

To understand these definitions, two terms may need clarification:

Visual acuity

This refers to the ability to clearly distinguish forms, objects or symbols at specific distances (Gothelf, Rikhye, & Silberman, 1988). The term 20/200 means that the person with the visual impairment can see an object or symbol at 20 feet that a person with unimpaired vision can see at 200 feet. (For a more in-depth description of visual acuity, see Chapter 2, page 11).

Field of vision

The ability to see objects in the periphery of one's vision when looking straight ahead. Individuals with unimpaired vision can usually see objects within 180 degree arc when looking straight ahead. (For more information on fields of vision, see Chapter 2, page 11).

Definitions used in Education

Other definitions have been developed by educators which are more functional and meaningful for programming. Many of these definitions have been adopted into state and federal definitions.

Visual Impairment (visual handicapped)

This term encompasses a wide range of vision loss which can include deficits in acuity, field loss, ocular motility, or color perception. The visual impairment may be permanent or temporary. The term visual handicapped is often used synonymously with the term visual impairment to refer to a vision loss which even with correction, adversely affects a child's educational performance (P.L. 94-142).

Blind

Individuals who are totally without vision or who have light perception only. In the educational field, this term refers to children who use other senses (i.e. hearing and touch) as primary channels for learning or receiving information.

Light perception only

Individuals who are without vision, but can perceive light.

Low vision

This is a broad term which is used to refer to individuals who have significant visual impairments, but still have usable vision. Vision is still used as a primary channel for learning or receiving information. Although the visual impairment continues after correction, visual functioning may increase with the use of optical aids, environmental modifications and/or training (Corn, 1980; Scholl, 1986).

Visual functioning

This refers to how well a person uses the vision he or she has. Visual functioning is considered a learned behavior which is not necessarily reflected by visual acuity. It is possi-

ble for a student to have poor visual acuity and good visual functioning, or visa versa (Gothelf, Rikhye & Silberman, 1988). Individuals who fail to use their vision in an efficient or meaningful way have poor visual functioning and are often included in functional definitions describing visual impairments.

Cortical Visual Impairment (Cortical Blindness)

The visual cortex of the brain or the visual pathways to the brain is damaged. This results in varying visual impairments, depending upon the location of damage in the brain. The anatomy and physiology of the eye is not impaired.

Terminology Describing Auditory Impairments

There are several terms that are used to describe the type and severity of hearing loss a person may be experiencing. Some terminology is legally used to define an auditory impairment (i.e. deafness, hard of hearing), other terminology classifies the hearing loss by the location of damage. Auditory impairments can be further classified by the severity of loss. Other terms can be found as well.

Legal and Educational Definitions

Educators have adopted the terms deaf and hard-of-hearing to denote the student's ability to function in the classroom. These terms are found in federal and state definitions. In IDEA, they have been defined as follows:

Deaf

A hearing impairment that is so severe that the child is impaired in processing linguistic information through hearing, with or without amplification, and which adversely affects educational performance.

Hard of hearing

A hearing impairment whether stable or progressive, which affects a child's educational performance, but is not included under the definition of deaf.

Hearing impairments may also be described in terms of the location of the impairment in the auditory system. The hearing loss may be described as a conductive, sensorineural, mixed, and/or central loss. These terms are defined as follows:

Conductive hearing loss

An obstruction, infection, structural abnormality or other condition in the outer or middle ear which results in a hearing loss.

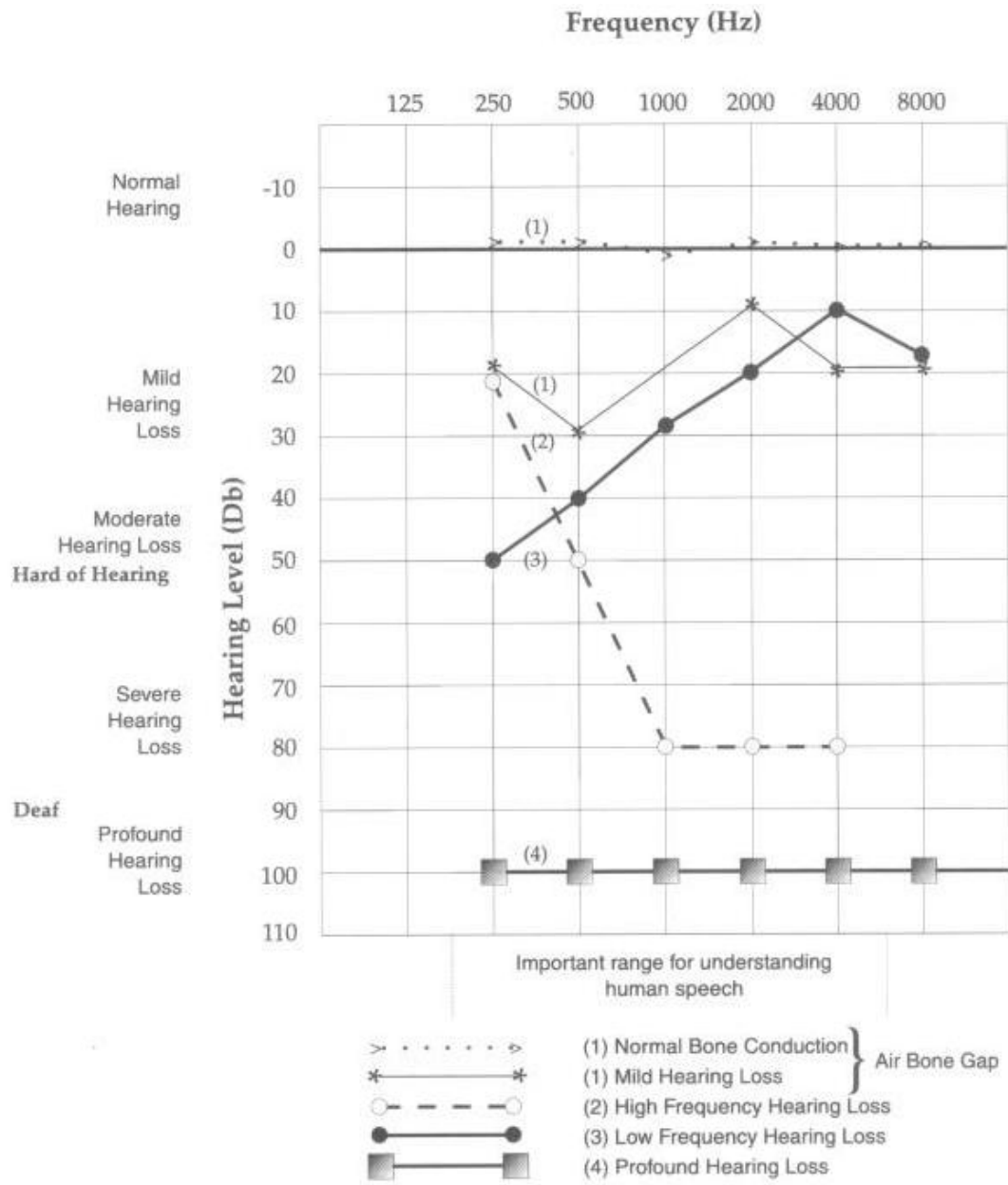
Sensorineural hearing loss

Impairments in the inner ear or auditory nerve that extends from the inner ear to the brainstem which result in a hearing loss.

Mixed hearing loss

A combined conductive and sensorineural hearing loss.

Central hearing loss



Adapted from, Keele (1983), *Developmentally disabled child*, p. 142. New Jersey: Medical Economic Books.

Figure 1 Audiogram

A hearing loss resulting from deficits in the areas of the brain (auditory cortex) that receive and process auditory input, or the pathways going from the brainstem to the auditory cortex.

Individuals who fail to use their hearing in an efficient or meaningful way are often included under a functional definition of sensory impairment.

Degree of Hearing Loss

The severity of a hearing impairment is often described in terms of loudness (decibels) and pitch (frequency or Hertz). An audiometer (electronic instrument) permits measurement of hearing levels at varying decibel levels across low and high frequencies (which are recorded as Hertz or cycles per second). From these results the following levels of severity have been delineated:

Normal

Hearing level 0-20 decibels

Mild hearing loss

Hearing level 21-40 decibels (Can hear conversational speech, but will have difficulty hearing distant or faint sounds. Amplification may be needed.)

Moderate hearing loss

Hearing level 41-60 decibels (Can hear conversational speech 3 to 5 feet away. Will probably need a hearing aid and auditory training)

Severe hearing loss

Hearing level 61-80 decibels (May hear a loud voice at about one foot and be able to identify environmental noises. May be able to determine vowels, but not consonants.)

Profound hearing loss

Hearing loss 80 decibels (May hear loud sounds, but hearing is not a primary modality used for receptive communication) (Hamre-Nietupski, Swatta, Veerjusen and Olsen, 1986)

Hearing losses can be depicted on an audiogram (see Figure 1). As the decibel level required for an individual to initially hear a sound at a certain frequency level increases, the hearing loss increases. The amount of hearing loss may also vary across frequencies. A person may have a low frequency hearing loss in which he or she is unable to hear low sounds or more frequently, a high frequency loss may be present.

Terminology Describing Age of Onset

Further classification may be made in terms of the age of onset of the hearing impairment. This has important implications since the earlier the hearing loss manifests itself in a child, the more disabled he or she is likely to be in terms of language development (Hallahan & Kauffman, 1988).

Congenitally deaf

Born deaf.

Adventitiously deaf

Deafness acquired after birth.

Prelingual deafness

Deafness prior to speech or language development.

Postlingual deafness

Deafness occurring after speech and language is developed. The age that differentiates prelinguistic from postlinguistic deafness has changed through the years due to the shifting emphasis on the importance of receptive language. Presently 18 months is commonly used as dividing point, although some argue that it should be changed to 12 or 6 months (Meadow-Orlans, 1987).

Visual and Auditory Impairments Combined

Due to the broad range of both visual and auditory losses any combination of sensory impairment is possible. Individuals may have:

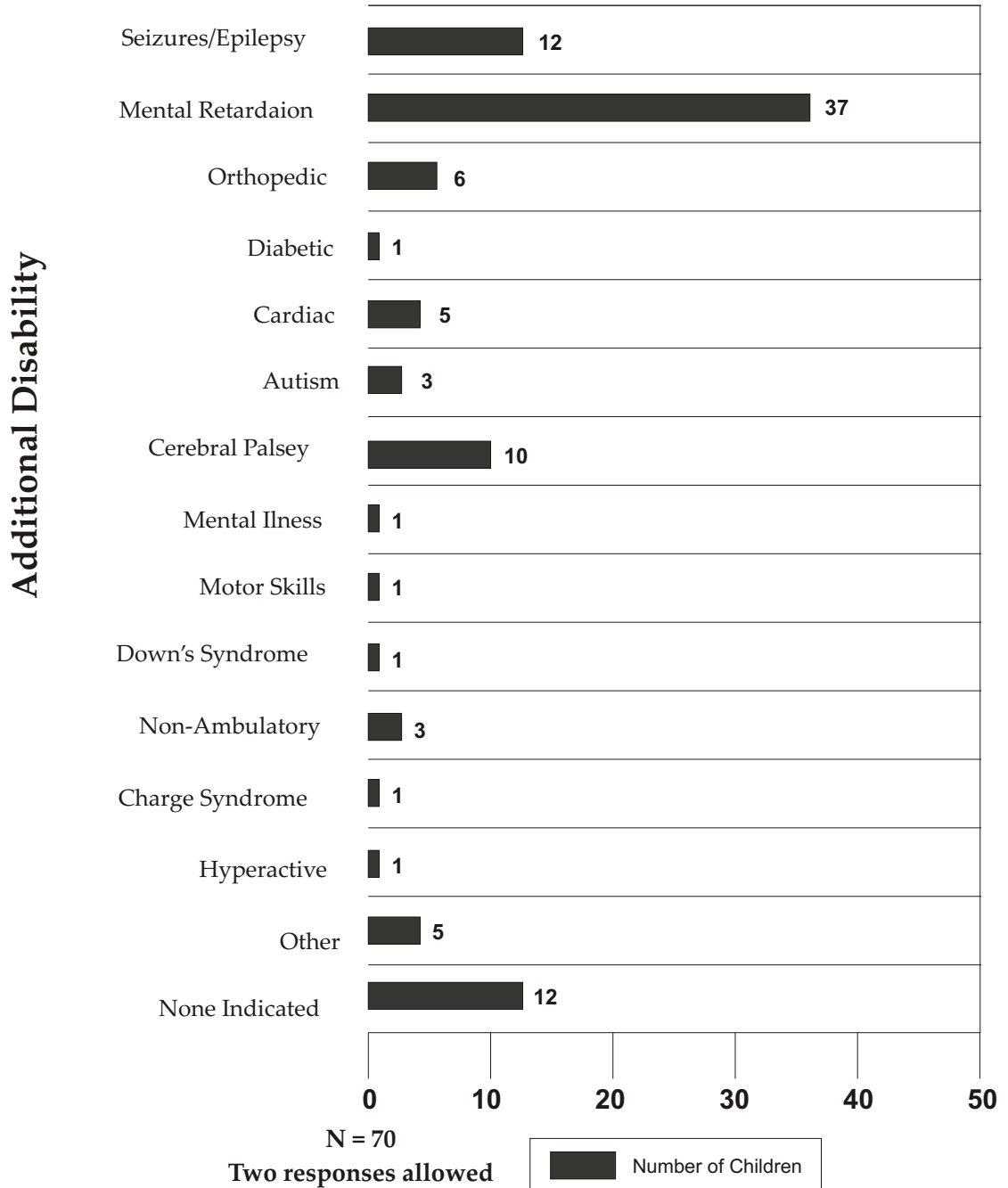
auditory impairment and visual impairment with vision loss being the primary disability; auditory impairment and vision impairment with auditory impairment as the primary disability; auditory impairment and blindness; deafness and visual impairment, and deaf-blindness (Roberts, Helmstetter, Guess, Murphy-Herd & Mulligan, 1984).

The majority of those who are classified as having concomitant visual and hearing impairments have some useful residual vision or hearing. Total absence of vision and hearing is very rare. The following graph depicts most of the possible combinations that may occur with an individual who is deaf-blind (see Figure 2). Concomitant vision and hearing loss may also be conceptualized as occurring congenitally or adventitiously (see Figure 3, page 8).

Often individuals who are deaf-blind also have other impairments as well. These may be additional physical disabilities, mental retardation or other types of disabilities (see Figure 4 page 9).

Overall, there are many different uses of terminology associated with describing the specific visual and auditory impairments of an individual who is deaf-blind. By better understanding these terms, administrators, educators, and related staff will be better equipped to identify those who are deaf-blind and meet their needs.

Children With Deaf-Blindness Extent of Additional Disabilities



Reprinted with permission from Morgan, B. (1984). The INSITE Model. U.S. Office of Education, Bureau of Education for the Handicapped. Washington, D.C.

Figure 2 Combinations of Vision and Hearing Impairments

Individuals with Dual Sensory Impairments

Onset

- Congenitally Deaf-Adventitiously Blind
- Congenitally Deaf-Blind
- Adventitiously Deaf-Blind
- Adventitiously Deaf-Congenitally Blind

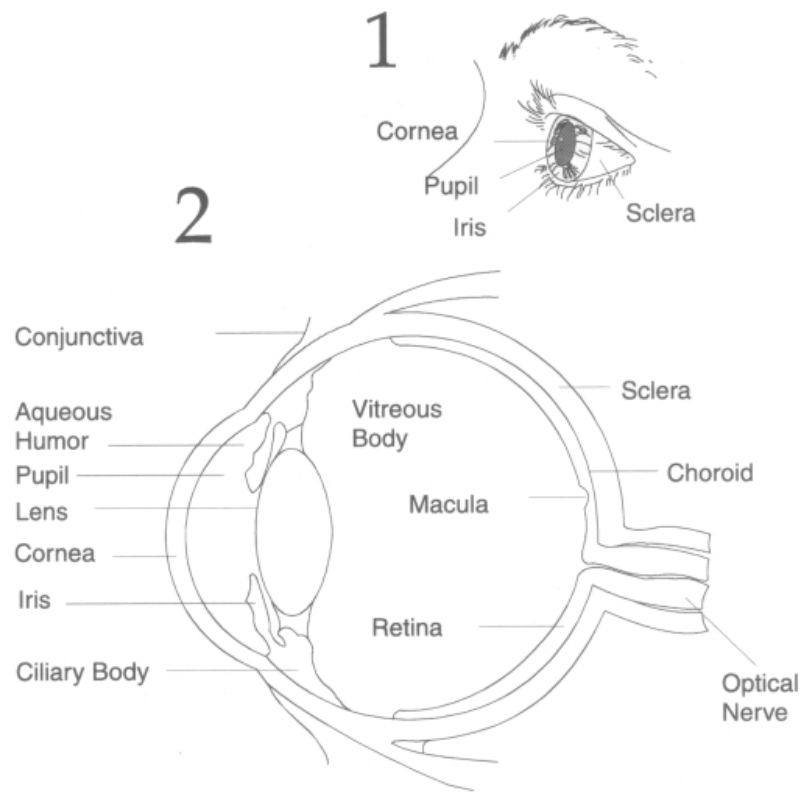
Degrees

- Hard of Hearing-Blind
- Hard of Hearing-Visually Impaired
- Deaf-Visually Impaired
- Deaf-Blind

Conditions

- Stable, Progressive, Temporary

Figure 3 Differences in characteristics



(1) Surface anatomy of the eye. (2) Internal structure.
From Fig 35-1, Bleck & Nagel (1982) reprinted with
permission of Allyn and Bacon.

Figure 5 Anatomy of the eye

Figure 4 Additional Disabilities

Chapter 2

Normal Anatomy of the Eye and Ear

The eyes and ears have complex anatomical structures and physiological processes that are needed for proper functioning of these sensory organs. An understanding of the basic anatomy and physiology of the eye and ear is needed as a foundation for later discussion regarding various abnormalities in the eye and ear that may result in visual and auditory impairments.

The Visual System

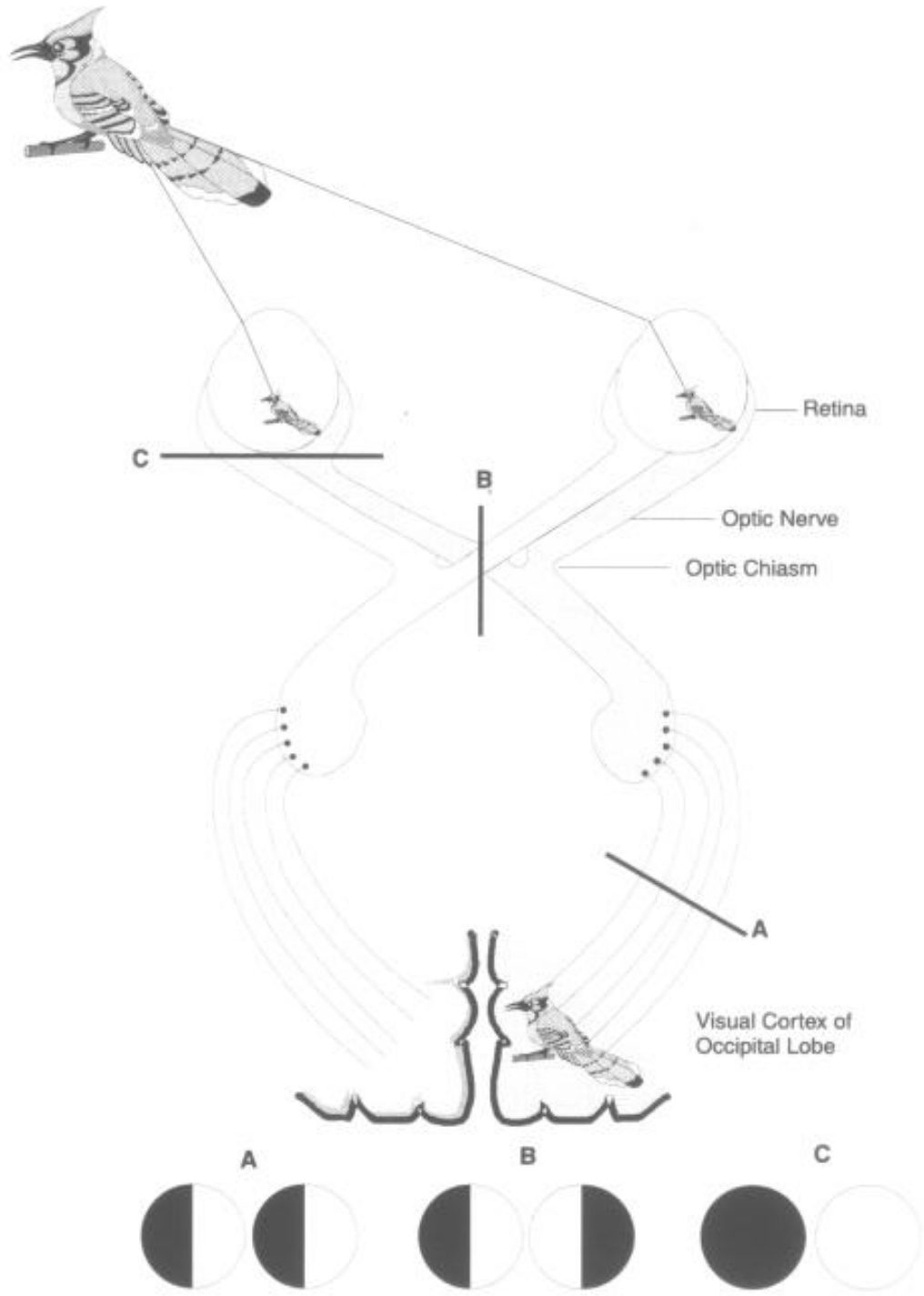
The visual system may be divided into three areas: (1) the eye (and its muscles), (2) the visual pathways to the brain and (3) the visual cortex. The structures of the eye collect and focus light energy into an image, and then convert the image into an electric-chemical impulse. The visual pathways transmit the electric-chemical impulses to the visual cortex. The visual cortex (located in the brain) interprets the impulse and re-lays it to other higher cerebral centers (Biglan, Van Hasselt & Simon, 1988).

The Eye

The eye can be divided into three layers consisting of the outer layer (containing the sclera and cornea), the middle layer (containing the choroid, iris, and ciliary body) and the inner layer (the retina) (see Figure 5). Within these three layers are located internal structures and fluids which refract the light to achieve a clear image.

The outer layer of the eye consists of the cornea, which covers one sixth of the anterior outer portion of the eye, and the sclera that covers the remaining five-sixths of the eyeball. The cornea is clear and refracts light rays as they enter the eyes. It also protects the inner portions of the eye. The sclera is the white part of the eye. It wraps around the entire eyeball except for where the cornea is located. It also serves as protection to the inner contents of the eye (Jose, 1983).

The middle layer of the eye is the uveal tract that consists of the iris, the ciliary body and the choroid. The iris is the colored part of the eye that has an opening in its center, known as the pupil. The iris controls the amount of light entering the eye by regulating the size of the pupil. In dim light, the pupil dilates (enlarges) to allow more light to enter the eye. In bright light, the pupil constricts (becomes smaller) to allow less light into the eye.



An abnormality at various points along the route (upper figure) will lead to different patterns of visual loss (lower figure). These are illustrated:
 A) abnormality at the cortical pathway;
 B) damage to the optic chiasm;
 C) retinal damage.

Figure 5 Anatomy of the eye

Directly continuous with the iris is the ciliary body. Part of the ciliary body is known as the ciliary muscle. It controls the curvature (shape) of the lens by relaxing and contracting the fibers that maintain the tension on the crystalline lens (Ward, 1986). The crystalline lens is located directly behind the pupil. Through the actions of the ciliary muscle, the eyes accommodate (focus for near and distant objects). The other major function of the ciliary body is the production of aqueous humor through the action of the ciliary processes. The aqueous humor is continually produced, fills the anterior portion of the eye, and is reabsorbed through the anterior chamber angle (angle between the iris and cornea). The aqueous humor brings nourishing materials to the cornea and lens and removes waste products.

The choroid is in the posterior part of the eye, located between the sclera and the retina. It contains many blood vessels and provides nutrients to the retina.

The inner (or neural) layer of the eye consists of the retina. It is a light sensitive membrane which converts the light rays to electrical impulses. The retina is composed of approximately 125 million photoreceptors categorized into rods and cones. The rods and cones are responsible for the conversion of light rays to electrical-chemical impulses (Ward, 1986). The rods are sensitive to low intensity illumination (dim light), and can detect gross form and movement. Rods are primarily responsible for peripheral vision. The cones are sensitive to high intensity illumination (day light), and detect fine detail and colors. On the retina there is a small central area in which most of the cones are located. This area is known as the macula. In the center of the macula is the fovea (the area of most distinct vision). The fovea and the macula together are responsible for most central vision.

Between the lens and the retina is a chamber that is filled with vitreous humor. Vitreous humor is clear gelatinous substance that aids in keeping the shape of the eyeball.

Several of the structures are responsible for refracting the light onto the retina and producing a focused image. The light rays pass through the cornea that produces the greatest amount of refraction of light rays. The light rays then pass through the aqueous humor and the pupil to the lens. The lens provides the second greatest amount of refraction to further refine and focus the light. The light rays pass through the vitreous humor and are further refracted. Finally the image appears upon the retina. From that point the image is carried to the brain along the optic nerve and through optic pathways.

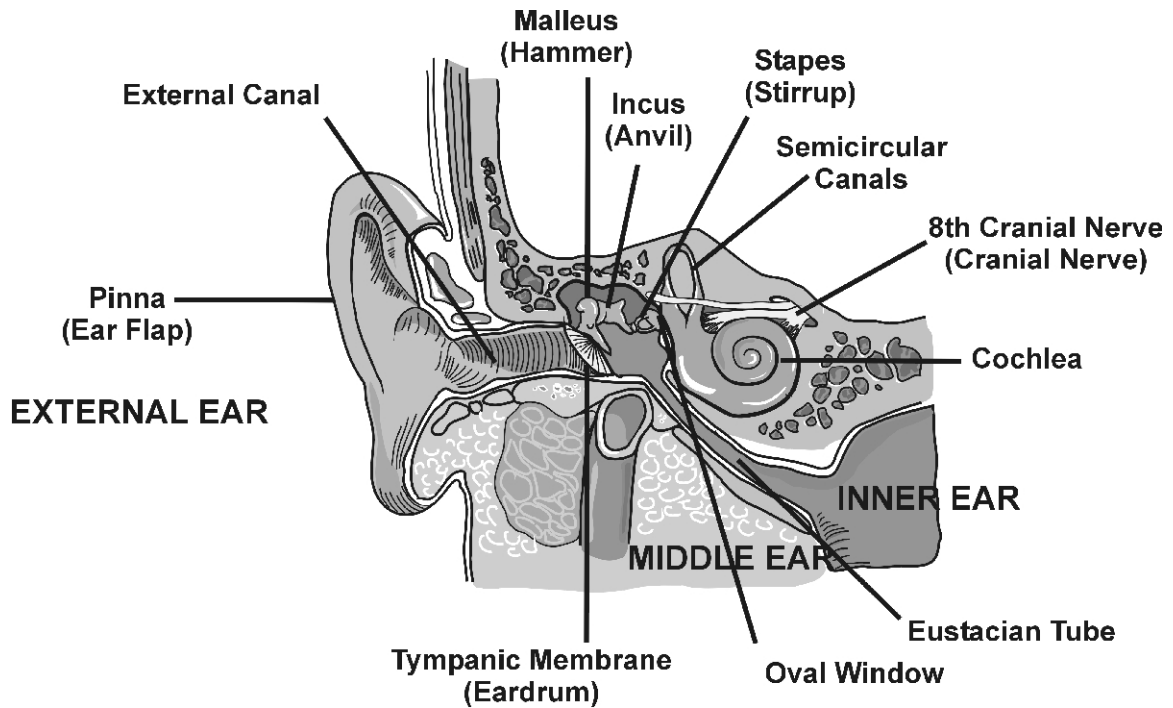
The Extraocular Muscles

Each eye movement is controlled by six muscles surrounding the eyeball (extraocular muscles). These muscles can move the eye up, down, diagonally and sideways. One of the requirements for single binocular vision is the coordinated movement of both eyes by the extraocular muscles (Rosenthal, 1982). Additionally, depth perception is facilitated by alignment of two eyes.

The Visual Pathways

After the image has been converted into electro-chemical impulses by the retina, the impulses travel down the optic nerves that are located posteriorly behind each eyeball. Part

of the fibers of each optic nerve cross each other at a point known as the optic chiasm. The impulse continues through the rest of the optic tracts, optic radiations and to the



posterior part of the brain (occipital lobe). This occurs in such a way that the visual field on the right side of the body is transmitted to the left side of the brain. The visual field on the left side of the body is transmitted to the right side of the brain (see Figure 6).

The Visual Cortex

In the posterior portion of the brain is an area known as the visual cortex (located in the occipital lobe). This area of the brain permits recognition of the transmitted impulses. From this area, the impulse is transmitted to higher cerebral centers. In these higher cerebral centers, visual input is integrated with other sensory input to allow a person to recall or remember perceived images.

Figure 6 Visual pathway

The Auditory System

The auditory system may be divided into four areas: the outer ear, the middle ear, the inner ear and the central auditory system. The ear collects sound waves and converts them into electro-chemical impulses. These impulses exit the ear via the eighth cranial nerve (auditory nerve) and are transmitted to the auditory cortex of the brain.

The Outer Ear

The outer ear is composed of the auricle (pinna) that is the visible part of the ear, and the external auditory canal (see Figure 7). The auricle collects sound waves and transmits them through the external auditory canal. The external auditory canal is lined by skin and contains hair and cerumen (wax). The function of cerumen is to lubricate and prevent infection of the skin, and to decrease sensitivity of the canal to wind and cold.

The Middle Ear

The middle ear is bordered on one side by the tympanic membrane (eardrum) and on the other by the oval and round window membranes, eustachian tube orifice, and the bony medial, superior, and inferior walls of this space. Contained in this air filled space within the temporal bone are three connected bones (or ossicles) known as the malleus (hammer), incus (anvil) and stapes (stirrup). The malleus is attached to the tympanic membrane and the stapes is directly attached to the oval window. When a sound occurs, air vibrations enter the outer ear and cause the tympanic membrane to move. The tympanic membrane converts sound waves to mechanical vibrations by causing the malleus, incus and stapes to mechanically vibrate. The vibrations of these three bones (ossicles) result in moving the oval window membrane, the inner ear fluids, and the round window membrane.

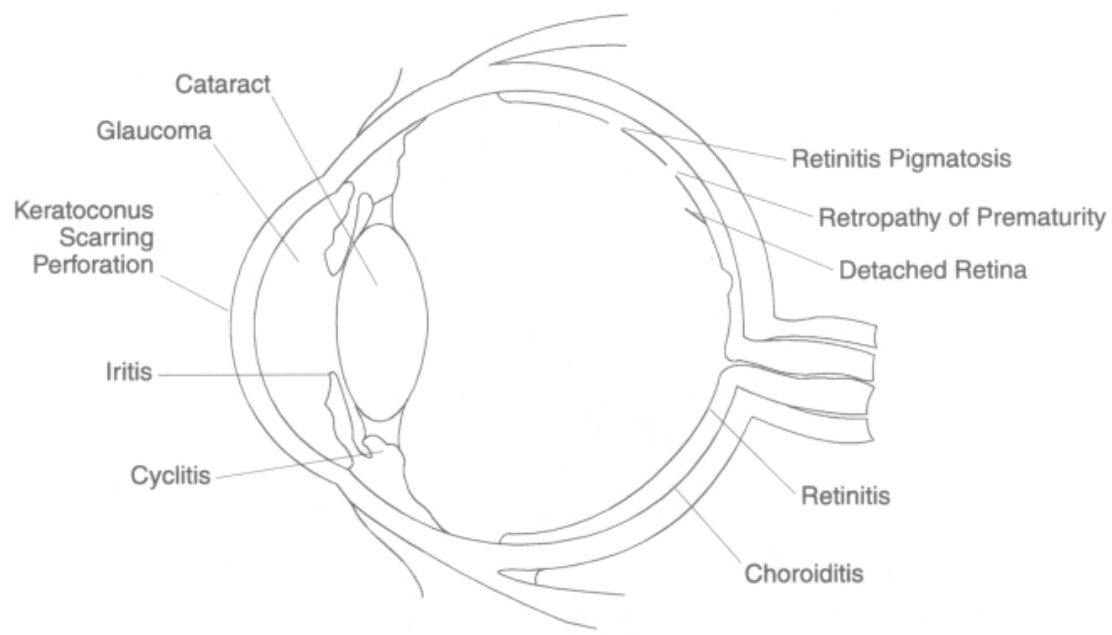
The Inner Ear

The inner ear consists of two main sections: the cochlea (responsible for hearing) and the vestibular mechanism (responsible for sense of balance). The cochlea has the appearance of a snail shell and is a system of coiled tubes that are filled with fluid. In the cochlea is the Organ of Corti which contains auditory receptor cells, known as hair cells. When the oval window membrane vibrates from the motion of the stapes, waves are transmitted within the fluid of the cochlea to the round window membrane. This wave motion moves the hair cells that then stimulate electro-chemical impulses. The type and location of movement of the hair cells determines the auditory information that is transmitted out of the inner ear by the cochlear division of the VIII cranial nerve (auditory nerve).

The Central Auditory System

Once the electrical impulses via the auditory nerve are transmitted to the first synapse at the brainstem, other pathways of the central auditory system transmit the electrical impulses to the auditory cortex. Some of these impulses cross over to the opposite side of the brain while others stay on the same side. The information is received in an area of the brain known as the temporal lobe. From here, other higher centers of the brain will process the sounds.

Figure 7 General view of the ear



*Common Disorders of the
Eye and Ear*

There are several common disorders of the eye and ear that are frequently encountered in children and youth with deaf-blindness. This chapter will discuss some of the most frequently occurring disorders and their impact upon vision and hearing. This will assist the reader in understanding the visual and auditory impairments of persons who are deaf-blind as well as provide a reference for the various etiologies described in the second section of the monograph.

Common Visual Disorders

Visual disorders can be grouped together by their structure and function. Categories of disorder by structure include: disorders of the outer layer (cornea, and sclera), middle layer and surrounding chambers and fluids (aqueous humor, iris, ciliary body, lens and vitreous humor) and inner layer (retina). General eye malformations, disorders of the visual pathway and disorders of the visual cortex follow. A section titled "Disorders affecting functional vision" includes problems of visual acuity, visual field deficits, ocular motility abnormalities, deficits in reflexive visual processes, and color blindness (see Figure 8).

Disorders of the Outer Layer of the Eye

Diseases of the cornea

Diseases of the cornea can result in serious visual impairment ranging from blurring of vision to total blindness. Since the cornea refracts light rays, a lesion on the cornea may result in blurred vision. If scarring or perforation from ulceration occurs, blindness can result. Usually prompt medical treatment permits avoidance of permanent damage (Jose, 1983).

Keratoconus

Keratoconus is a cone shaped cornea that results in a distortion of the entire visual field. This rare disorder usually begins during adolescence and is slowly progressive. Initially, a mild vision loss with myopia (nearsightedness) and astigmatism occurs. Bilateral thinning, scarring and stretching of the center of the cornea occurs and visual acuity then worsens (Apple & Rebb, 1991). Frequent changes of glasses or contacts are needed as the visual acuity worsens. If left untreated, this condition can slowly progress to the point that the cornea ruptures, resulting in blindness. However, contact lenses may improve visual acuity in the early stages and surgery can be performed. This disorder has been found to occur in individuals with congenital Rubella and Down syndrome (Lucas, 1989).

Figure 8 Common disorders of the eye and their location

Disorders of the Middle Layers of the Eye

There are several disorders of the middle layers of the eye and surrounding structures and fluids. Some of these include glaucoma, cataracts, and uveitis.

Glaucoma

Glaucoma is an abnormal increase in intraocular pressure which can damage the eye and visual functioning. There are several types of glaucoma which all involve an imbalance between production and outflow of the aqueous humor. One category of glaucoma is known as primary glaucoma in which there is now other causative agent other than the glaucoma, itself. This category of glaucoma is typically die to a blockage in the area of the eye that the aqueous humor exits. The blockage of the outflow of aqueous humor results in increased intraocular pressure. If this increase in intraocular pressure is permitted to progress, the optic nerve will be damaged, resulting in loss of visual acuity, field loss, and/or blindness (Berkow, 1987; Martyn & DiGeorge, 1987). Congenital glaucoma, which is glaucoma occurring from birth, is considered a primary type of glaucoma.

When the glaucoma develops from another eye disorder (such as uveitis or cataracts) or disorders involving other organ systems, it is known as secondary glaucoma. This form of glaucoma is often caused by blockage of the flow of aqueous humor.

As discussed in subsequent chapters, glaucoma may be associated with other conditions such as congenital infections (STORCH), adventitious conditions (e.g. trauma, tumors), retinopathy of prematurity and some syndromes.

The major signs of glaucoma in infants include eye enlargement, tearing, photophobia, corneal clouding, twitching of the eye muscles and intense eye pain. After age three, the sclera and cornea are less expandable and signs of pressure elevation are different. The child or adolescent may have no symptoms even with pressure high enough to cause visual loss.

When the individual is born with glaucoma (congenital glaucoma), surgical intervention is necessary to correct the blockage and prevent optic nerve impairment. For glaucoma which develops after birth, eye drops can usually control the condition and may be all that is required. In a few cases, surgery may be necessary. For individuals who have secondary glaucoma, the other condition will need to be treated for glaucoma treatment to be successful. In this case, medication or surgery may be indicated.

Cataract

A cataract is any opacification (clouding) of the crystalline lens of the eye. Cataracts may be present at birth (congenital cataracts) or develop later. Its effect on vision varies depending on the size, position and density of the cloudy area. Some cataracts involve pinpoint areas which do not interfere with visual acuity. Other cataracts result in decreased visual acuity with blurred vision. If the opacity is more centrally located, near vision may be affected and vision may be worse in bright light. Those opacities located in the outer portion of the lens may result in poor color discrimination due to the abnormal

scattering of light rays (Jose, 1983). As the cataract progresses, it may become so dense that blindness results.

As the cataracts are developing, frequent changes in eyeglass prescription may help assist with maintaining useful vision. When cataracts progress to the point that useful vision is gone, surgery is performed to remove the lens. When an infant has congenital cataracts, surgery within a few months of birth is often advised to allow for proper development of visual responses. After cataract surgery, the individual will need to wear glasses, contact lenses or may have a prosthetic lens implant.

Cataracts are present in a wide range of developmental disorders. Some of these include congenital infections (e.g. CMV, toxoplasmosis, rubella, and herpes) and syndromes (Trisomy 18, Trisomy 13, Down syndrome, Cockayne, Crouzon, Refsum and Usher syndromes). Cataracts may also occur from trauma or drugs (steroids) and are associated with metabolic disorders. A special type of lens abnormality called "cataract of prematurity" may occur in newborns, but the cloudy areas usually disappear after a few weeks (Martyn & DiGeorge, 1987).

Uveitis

Uveitis is an inflammation of the uveal tract (which consists of the iris, ciliary body and choroid). It includes iritis (inflammation of the iris), iridocyclitis (inflammation of the iris and ciliary body), cyclitis (inflammation of the ciliary body) and choroiditis (inflammation of the choroid). Posterior uveitis includes chorioretinitis (inflammation of the choroid and retina) and retinitis (inflammation of the retina). (These two conditions are typically included under uveitis although they are anatomically posterior to the uveal tract.) Uveitis may be chronic or acute, often resulting in diminished or hazy vision. Black floating spots may be present. Severe pain, redness, and photophobia can occur in iritis and iridocyclitis. Complications such as cataracts, glaucoma and retinal detachment may result (Berkow 1987).

Uveitis has been present in children with such congenital infections as herpes, rubella, syphilis, toxoplasmosis, and CMV. Individuals with AIDS who have acquired one of these infections may also develop uveitis (Knox, 1987). It is important to diagnose uveitis early. Treatment of uveitis consists of corticosteroid medication.

Disorders of the Inner Layer

There are several disorders of the retina which negatively impact on vision. These can be divided into retinal dystrophies, retinopathy of prematurity, and retinal detachments.

Retinal dystrophies (retinitis pigmentosa)

Retinal dystrophies are lesions of the retinal cones, rods and pigments. The main group of retinal dystrophies are known as retinitis pigmentosa (RP). In retinitis pigmentosa, retinal degeneration occurs and melanin pigment migrates into the retina and deposits itself. The condition first begins with the rods being slowly destroyed, starting at the midperiphery. This results in night blindness and progressive loss of the peripheral field of vision. This continues to worsen and contracts from the periphery, leading to tunnel vision. Cone degeneration also occurs and as it progresses, the tubular vision further constricts to the point that central vision is reduced and difficulties occur seeing in the

day as well. Both eyes are usually affected with this hereditary condition and onset is between ages of ten and twenty (Lucas, 1989). Vision loss is gradual with adolescents often exhibiting difficulty traveling at night, difficulty moving from outdoors to indoor lighting as well as doing certain activity such as playing sports due to a loss of peripheral vision. As the condition progresses, total blindness can result later in life (Apple & Rabb, 1991).

Several atypical forms of retinitis pigmentosa have been found in which the symptoms and course of the condition are different than the classic form just described. In some forms of atypical retinitis pigmentosa, degeneration begin centrally. With these, the macula is involved which results in deficits of central vision, poor acuity and color vision abnormalities. These may progress to the periphery and result in degeneration in that area as well.

Certain syndromes may have typical retinitis pigmentosa (e.g. Usher syndrome) or atypical retinitis pigmentosa (e.g. Alstrom syndrome). There is no effective treatment at this time for either typical or atypical retinitis pigmentosa.

Detached retina

When an individual has a detached retina, the retina separates from its supporting structures and atrophies. Depending on the cause of the detached retina, the early symptoms are usually minimal. The individual may see a bright flash of light and possibly floaters that look like black dots or spider webs. As the detached retina passes over the macula, vision is blurred (Apple & Rabb, 1991). A scotoma (blind area) will develop in the field of vision which corresponds to the area of detachment (Jose, 1983). Typically, the detachment occurs from fluid accumulating under the retina which push it away from the choroid. This may occur from other ocular conditions such as retinopathy of prematurity, congenital infections (such as toxoplasmosis), general physical conditions (such as diabetes or head trauma) or after cataract surgery. If the individual is prone to retinal tears or detachment, rough physical activity (trampoline play) must be avoided.

When a retinal detachment occurs, the individual will need to have surgery as soon as possible to reattach the retina.

Retinopathy of prematurity (ROP)

ROP (previously known as retrolental fibroplasia) consists of an abnormal growth of blood vessels which occurs in the immature retina (Biglan, Van Hasselt, & Simon, 1988). About 90% of the cases are mild and spontaneous regression of these abnormal blood vessels may occur with minimal scarring and little to no visual loss (Flynn, 1987). In more severe causes, the abnormal blood vessels extend into the vitreous and may cause retinal detachment, severe visual loss and/or blindness (Biglan, Van Hasselt, & Simon, 1988).

Retinopathy of prematurity is thought to develop from the premature infant being exposed to high levels of oxygen in incubators. However, the condition has been present in premature infants not exposed to these oxygen levels (Berkow, 1987). Children with retinopathy of prematurity have a higher risk of myopia, strabismus and glaucoma.

Eye Malformations

Microphthalmus, anophthalmus, aniridia

Eye malformations can also occur and may affect vision. Some children may be born with microphthalmos, which is extremely small eyeballs. It is common in children who had congenital rubella and is associated with poor visual acuity and nystagmus. Two other eye malformations include anophthalmus (absence of eyes), or aniridia (partial or complete absence of the iris).

Coloboma

A lesion or defect in the eye, known as coloboma can occur when there is a cleft, notch, gap or fissure in the iris, ciliary body, choroid or optic nerve. This occurs as a failure of the cleft to close during early prenatal development (about 6 weeks after conception) which prevents formation of an intact, complete eyeball. If the retina or optic nerve is involved, there is usually a scotoma (blind spot) or field loss corresponding to the site of the defect (Apple & Rebb, 1991). Often the field loss is located in the upper fields of vision causing a person difficulty seeing low overhanging tree limbs, signs or cabinets (Jose, 1983). There may be a decrease in visual acuity, as well as such concomitant visual abnormalities as strabismus or nystagmus.

Colobomas may occur unassociated with other abnormalities or may occur in such conditions as Trisomy 13 and CHARGE association.

Disorders of the Visual Pathways

The visual pathways of the optic nerve, optic tract and optic radiations may be affected and result in visual impairment.

Atrophy of the optic nerve

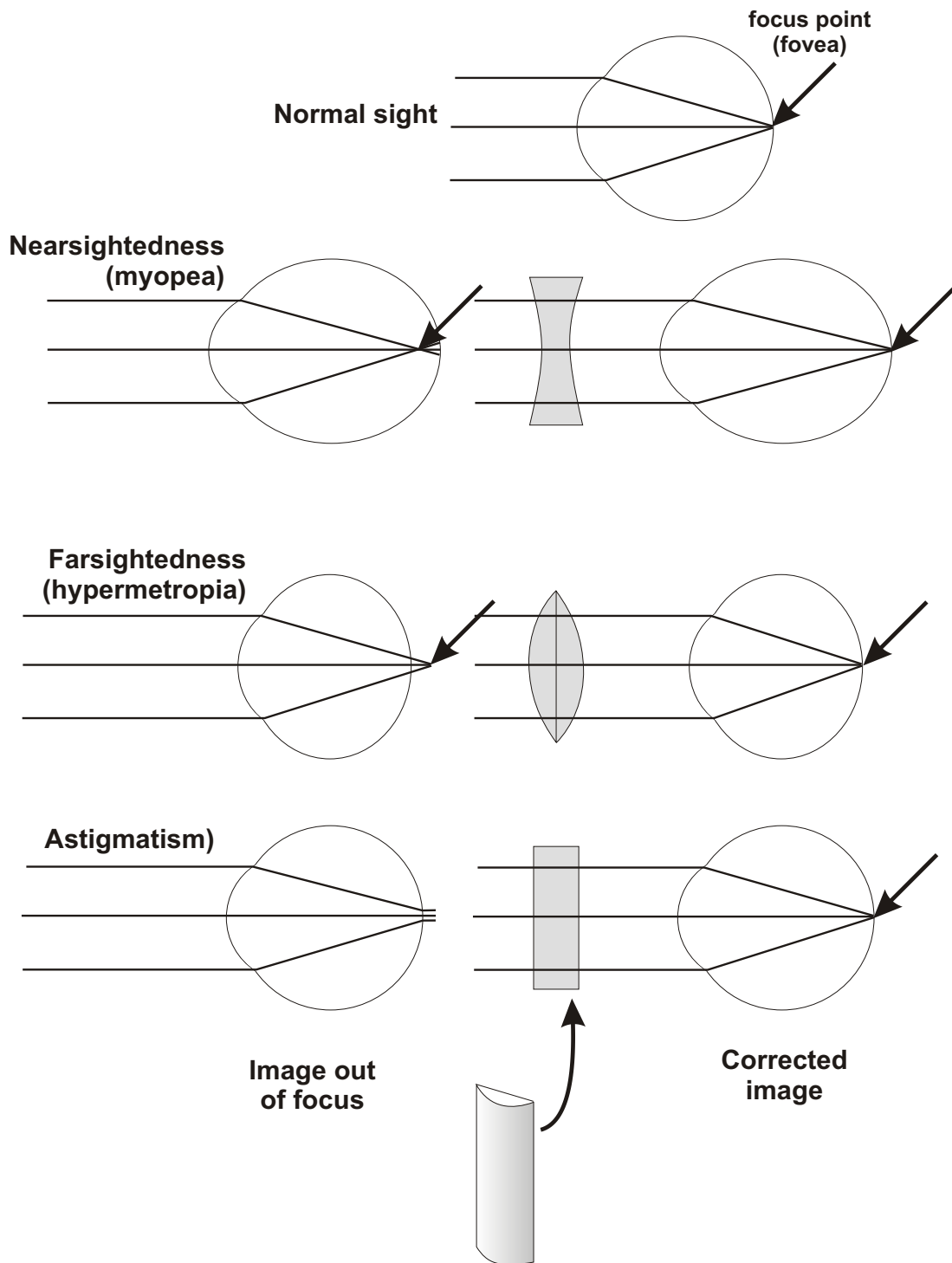
Atrophy of the optic nerve is the most common disorder of the visual pathways. It can be hereditary and/or can be caused by numerous diseases and disorders (e.g. retinitis pigmentosa, tumors, hydrocephalus, and head trauma). Central visual loss and field losses are often present with the visual loss typically being roughly proportional to the amount of nerve atrophy. Total blindness can result (Berkow, 1987).

Congenital optic nerve hypoplasia

Another optic nerve disorder which causes visual impairment is congenital optic nerve hypoplasia (CONH). In this disorder there is incomplete development of the optic nerve. It causes variable degrees of visual impairment. It is often associated with neurological disorders and endocrine problems.

Disorders of the Visual Cortex

Cortical visual impairment (also known as cortical blindness) is a term used to describe damage to the visual pathways or cortex of the brain. In these instances, the eye shows no pathology, however the brain is unable to process the incoming visual information. The resulting visual impairment may range from partial loss of visual acuity to blind-



If the eyeball is too long, images are focused in front of the retina (myopia). A concave lens deflects the rays, correcting the problem. If the eyeball is too short, the image focuses behind the retina and is again blurred (hypermetropia). A convex lens corrects this. In astigmatism, the eyeball is the correct size, but the cornea is misshapen. A cylindrical lens is required to compensate.

ness, depending on the exact location of the damage. Visual field defects may be present as well.

There are several causes of cortical visual impairment. Some of these include: closed head injury, drowning, prolonged convulsion, meningitis, and hypoxia resulting in brain damage. With some of these, visual improvement may occur over time. Hydrocephalus, which is not adequately treated with shunting, may also result in a visual loss. Some improvement in vision may occur after shunting, but this is not always the case (Buncic, 1987). Sometimes cortical visual impairment occurs in children with retardation.

Disorders of Visual Functioning

There are several disorders which may affect proper functioning of the eye in regard to: visual acuity, fields of vision, ocular motility, reflexive visual processes and color discrimination (McLaughlin & Lewis, 1986). These visual processes may be impaired from numerous causes and need careful assessment to determine proper function.

Visual acuity

Visual acuity refers to how clear or sharp an image is in regard to forms or patterns (Cress, 1988). It is described in terms of near and far vision. Near vision is considered the ability to clearly perceive objects at about 14 inches from the eyes (Ward, 1986). When individuals can not clearly see near objects due to an active error, this is known as hyperopia (farsightedness). In hyperopia, the eye is either smaller than normal, or its active power is weaker than the normal system (Jose, 1983). When the light rays enter the eye, they do not properly focus on the retina. The light rays fall behind the retina, resulting in the ability to only see far objects clearly, but not near objects. A convex lens (in glasses or contacts) is needed to focus the light rays on the retina to allow for clear near vision. (see Figure 9).

Far vision describes how well someone sees objects or symbols at a distance. It is typically reported in ratios such as 20/20, 20/70, etc. Normal visual acuity is considered 20/20. The first number refers to how many feet the letters, symbols or objects are from the person. The second number refers to the size of the symbols which represent the distance a person with normal vision could read them. For example, a person with 20/70 vision sees at a distance of twenty feet what a person with unimpaired vision sees at a distance of 70 feet. Individuals with an active error affecting distance vision are considered to have myopia (nearsightedness). In myopia, the eyeball is either longer than normal or the eye has greater than normal active power. Due to either occurrence, the light rays focus in front of the retina causing difficulty seeing objects far away. A concave lens (in glasses or contacts) is needed to focus the light rays on the retina.

Another condition which may affect visual acuity is astigmatism. In this condition there is an unequal curvature of the cornea or lens. In this condition, light does not come to a single point of focus on the retina. Astigmatism results in blurred or distorted images by itself or in combination with farsightedness and nearsightedness. When the stigmatism is moderate or severe, cylindrical lenses are prescribed.

Defects in visual fields

Defects in visual fields is another type of visual impairment. Normal visual fields include areas of peripheral and central vision. Visual field defects or blind spots can result in peripheral vision losses, central vision losses or losses in specific quadrants (see Figure 10). Peripheral vision losses include losses in the outer portions of the visual field (e.g. retinitis pigmentosa). Peripheral field loss results in a reduced angle of vision, or limits how much a person can see at one time. Since the rods, which assist in seeing in dim light, are located in the periphery, a deficit in this area may be accompanied by loss of night vision. A person with a peripheral loss will find it difficult to see in dim light and travel independently at night.

Figure 9 Myopia, hyperopia,, and stigmatism

If the eyeball is too long, images are focused in front of the retina (myopia). A concave lens deflects the rays, correcting the problem. If the eyeball is too short, the image focuses behind the retina and is again blurred (hyperopia). A convex lens corrects this. In astigmatism, the eyeball is the correct size, but the cornea is misshapen. A cylindrical lens is required to compensate.

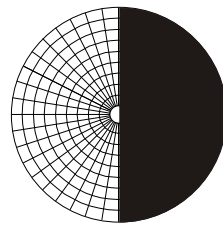
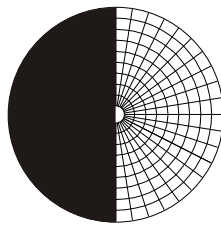
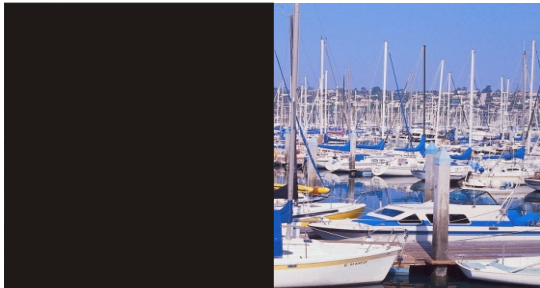
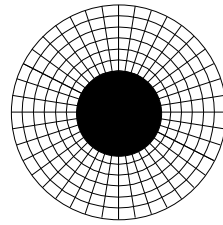
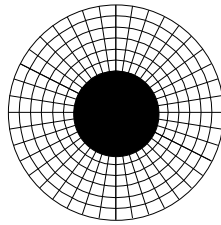
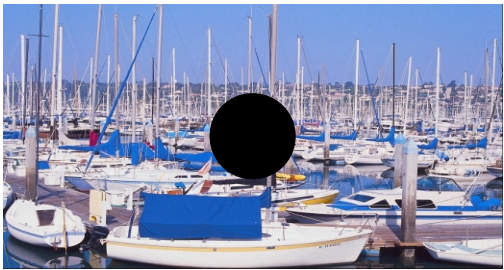
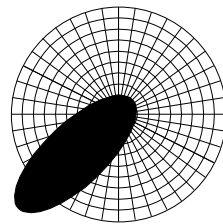
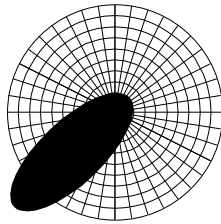
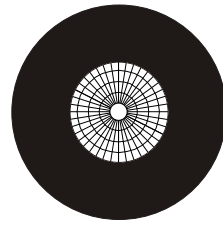
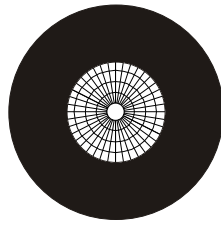
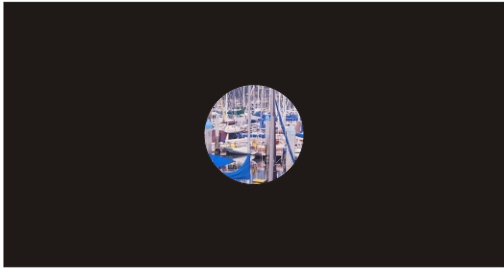
Central vision loss refers to visual losses in the central field of vision. It is due to disorders affecting the macula portion of the retina (e.g. macular degeneration, or atypical retinitis pigmentosa). A person with a central vision loss may miss seeing academic material or objects in their path unless proper programming is in place.

A person may also have scotoma (blind spots) which affect the visual fields. Both central and peripheral vision may be affected with scotomas.

Specific visual field deficits also include hemianopsia (one half of the visual field in both eyes missing) (see Figure 10) and quadrant loss (one fourth of the visual field is missing). These occur more commonly due to a tumor along the visual pathways or brain trauma. This is also present in children with cerebral palsy.

Disorders of ocular motility

Ocular motility refers to the movement of the eye by any of the six muscles surrounding each eye (i.e., extraocular muscles). Difficulties in ocular motility may occur in individu-



als with facial paralysis, eye muscle imbalances, cranial nerve damage or other conditions which may result in diminished or loss of eye muscle movement.

Strabismus

Impairments in eye muscle movement can result in strabismus (which is when one or both eyes deviate from correct alignment). Types of strabismus are: esotropia (where one or both eyes turns in (i.e. nasally)), exotropia (where one or both eyes turn out (i.e. temporally)), and hypertropia where the eye deviates upward. When one eye deviates, diplopia (double vision) can occur. Often the brain will suppress one image so that only one visual image will be seen instead of double.

Amblyopia

The suppression of one image by the brain in individuals with strabismus is the leading cause of amblyopia. Amblyopia is a reduction of corrected central visual acuity in the absence of ophthalmoscopically visual abnormalities. It can also occur due to unequal active errors or other less common causes (Friendly, 1987). The suppression of images transmitted by one eye can lead to eventual blindness in the unused eye (Rosenthal, 1982). This can be avoided with appropriate intervention implemented at an early age.

Impairments in ocular motility may also result in difficulties with tracking, convergence, gaze shift and scanning.

Nystagmus

Another abnormal ocular movements is nystagmus. Nystagmus consists of involuntary, rhythmic eye movements, primarily in the horizontal plane. However, movement may be vertical, diagonal, or rotary and can be fast or slow. Drifting eye movements may be present and present as slow searching movements with no evidence of fixation. When nystagmus is present during the first year of life, it may be indicative of the presence of a bilateral vision loss. It can also be due to a neurological impairment (i.e. hydrocephalus). When nystagmus occurs later, the individual may have poor visual acuity in the affected eye, although binocular vision may be unimpaired. Nystagmus is usually associated with congenital visual abnormalities (Hoyt, 1987).

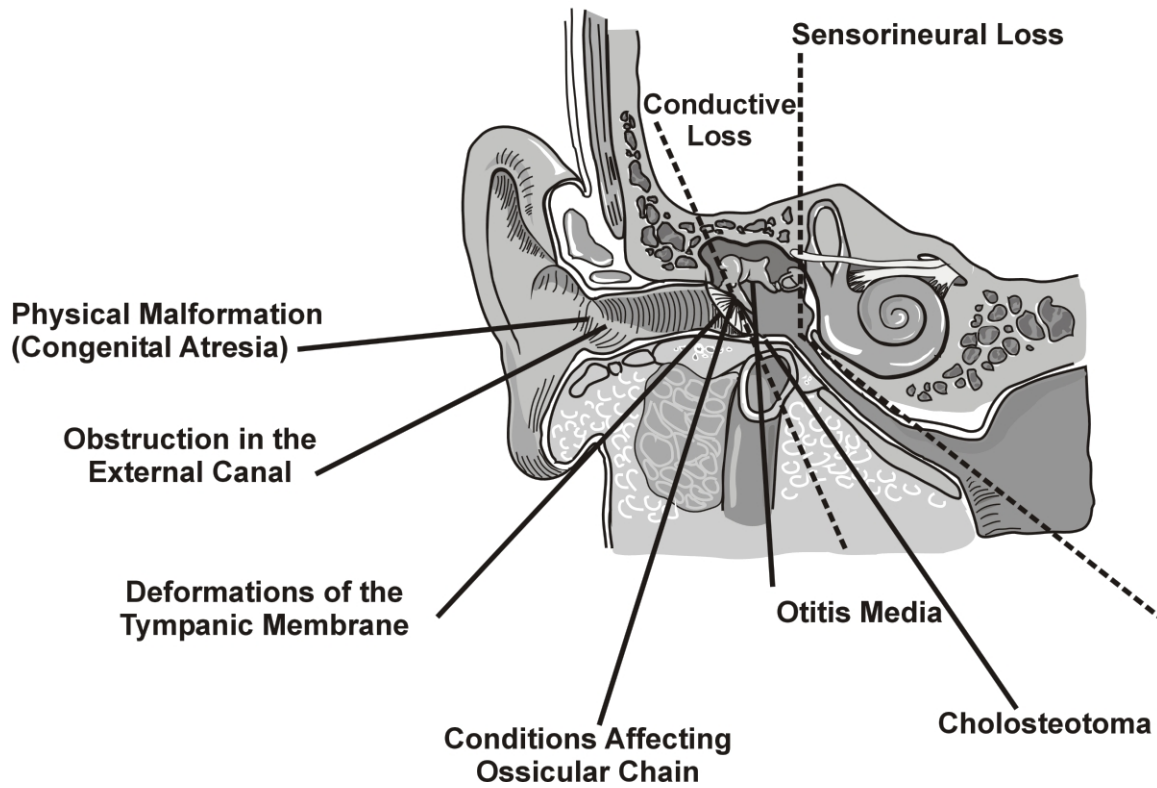


Figure 10 Vision field loss

Disorders of pupillary response and blink reflex

The pupillary response and the protective blink reflex are two reflexive visual processes. The pupillary response is a change in the size of the pupil in response to light. In bright light, the pupil appears to constrict to allow less light into the eye. In dim light, the pupil is enlarged to allow more light into the eye. In some disorders (and under the effects of certain medications), the pupil may be more dilated or constricted than normal or may not change in response to changes in illumination.

The protective blink reflex refers to the ability of the eye to close when a threat to the eye is imminent (Cress, 1988). Assessment of this reflex assists in determining if the person has any functional vision. If the person has vision, but nonfunctioning reflexes, bright lights may be uncomfortable for some individuals whose pupils do not constrict and an increase risk of eye injury may be present when the blink reflex is absent.

Disorders in color perception

Some individuals may have difficulty perceiving color. Individuals who are color blind usually can not see certain colors due to missing or damaged cones (color receptors) in the eye. This is typically a sex-linked genetic abnormality and results from absence of color genes on the X chromosome. The most common type of color blindness are red-green blindness (in which red and green appear the same color), red blindness, and green blindness. Very rare is blue-yellow color blindness (Vaughan, Asbury, & Riordan-Eva, 1992).

The functional vision assessment will assist in determining how well the individual sees, as well as what adaptations are needed. Often environmental arrangements, such as controlling the amount of light, and presenting materials with textures or contrast will be helpful.

Disorders of Hearing

There are several types of common hearing disorders classified by location of impairment. They can be divided into disorders of the outer ear (conductive loss), middle ear (conductive loss), outer and/or middle ear together with inner ear (mixed loss), inner ear and cochlear eighth cranial nerve (sensorineural loss), and central auditory mechanism (central loss) (see Figure 11).

Disorders of the Outer Ear: Conductive Loss

When there is a disorder of the outer ear which affects hearing, it is classified as a conductive hearing loss. There are several types of conditions which result in conductive hearing losses. There can be physical malformations such as congenital atresia (absence or closure of the external auditory canal) or obstruction in the external auditory canal from impacted cerumen (wax), foreign bodies, or a tumor. Infections such as external otitis (an infection of the skin of the external auditory canal, also known as swimmers ear) may also impede hearing.

The tympanic membrane (eardrum), bordering between the outer and middle ear, may become perforated due to an infection or a blow to the head. This results in a conductive loss as well.

Disorders of the Middle Ear: Conductive Loss Otitis media

One of the most frequent causes of a conductive hearing loss is otitis media. Otitis media is an inflammation of the middle ear that is frequently caused by a bacterial or viral infection. Often otitis media is accompanied by fluid in the middle ear which restricts movement of the tympanic membrane. This restriction of movement can result in a moderate hearing loss and possible rupture of the tympanic membrane. Prompt treatment with antibiotics and much less frequently, Myringotomy (lancing of the eardrum) should clear the infection and allow return of normal hearing. Another form of otitis media can occur (serous otitis media) in which there is no infection, but negative pressure occurs in the middle ear, secondary to the poor eustachian tube formation with resultant accumulation of fluid in the middle ear cavity. This can also result in a conductive hearing loss that may require myringotomy and possible insertion of a ventilation tube into the tympanic membrane.

Figure 11 Common disorders of the ear

Cholesteatoma

There are several other abnormalities of the middle ear which can cause hearing impairment. Cholesteatoma is a congenital or acquired benign skin-like mass which can occur in the middle ear and the other areas of the temporal bone. It typically develops as a complication of chronic or recurrent otitis media. Perforation of the tympanic membrane often occurs. One danger of this condition is the possible erosion of auditory or cranial structures, such as the destruction of the middle ear bone. Depending on the location of the cholesteatoma, there may be no hearing loss, progressive conductive hearing loss, or sensorineural hearing loss. Recurrent drainage from the ear (otorrhea) is common. Complications of a cholesteatoma such as meningitis, from abscess and facial paralysis are uncommon today but still occur.

Conditions affecting the ossicular chain

Conditions affecting the ossicular chain (middle ear bones) include discontinuity of the ossicular chain and fixation of one or more bones. Discontinuity of the ossicular chain refers to disruption of the connections between the malleus, incus and stapes. This may occur from congenital defects, head trauma or middle ear disease. Fixation of a middle ear bone may be a congenital or acquired condition in which there is an unusually firm attachment between one or more bones and other portions of the middle ear (e.g. malleus fixation to the roof of the middle ear). Stapes fixation to the rear of the oval window is usually caused by a condition called otosclerosis. These conditions generally produce a conductive hearing loss and frequently can be improved with surgery.

Disorders of the Inner Ear & VIII Cranial Nerve: Sensorineural Loss

A hearing loss due to a dysfunction of the inner ear on the pathway from the inner ear to the brainstem is referred to as a sensorineural loss. In this type of hearing loss, sound reaches the inner ear, but is not completely transmitted to the brain. Typically the individual can not hear the high frequencies; the frequencies needed to understand speech. Most babies born with a hearing loss have a sensorineural loss. This is the case in such conditions as Usher syndrome and congenital infections which result in hearing impairment. However a sensorineural loss may be acquired as well from such causes as ototoxic drugs, trauma, and infections (meningitis) (Ludman, 1988).

Disorders of Outer, Middle, Inner Ear: Mixed Loss

In some conditions and syndromes, a hearing loss is present in which there is both conductive and sensorineural losses. This is referred to as a mixed loss.

Disorders of Central Auditory System: Central loss

A central hearing loss occurs if there are abnormalities in the auditory cortex or the pathways going from the brainstem to the auditory cortex. This may be caused by a brain tumor, vascular changes in the brain or congenital or acquired brain damage. In central deafness, the mechanics of the ear are working correctly, but the individual can not interpret the auditory messages due to damage in the area of the brain which receives the transmissions (Beadle, 1982).

Disorders of Functional Hearing

As with visual impairments, a careful assessment of functional hearing is necessary to accurately determine what the individual can hear. A functional description will inform the teacher, service provider or parent how well and in what situations the individual can hear best. Some students will benefit from use of environmental adaptations and auditory aides.

Section II

Causes of Deaf-Blindness

There are many different syndromes and disorders, which result in visual and auditory impairments. This chapter will present a basic overview of these, stressing possible visual and auditory impairments. These various syndromes and disorders have resulted in children having deaf-blindness.

Although it may be surprising that these two sensory organs would both be impaired, this can be explained by their similar embryonic development. Both eyes and ears develop primarily during the first twelve weeks of pregnancy. Also, both the eye and ear originate from some of the same type of embryonic cells and tissue and have several anatomical similarities (Regenbogen & Coscas, 1985). Because of these similarities, various diseases and conditions result in damage to both organs.

Down Syndrome

Down syndrome, also known as Trisomy 21, is one of the most common chromosomal abnormalities. In this condition, there is an extra chromosome (or extra part of a chromosome) in the cells of the body. This results in several distinct physical characteristics and abnormalities.

Three primary causes of Down syndrome have been identified. In most cases, Down syndrome results from non disjunction which is a failure of the chromosome 21 to separate. Another cause known as translocation, is for an extra chromosome 21 to be attached to another chromosome. The smallest percent of individuals with Down syndrome have Mosaicism where an abnormal separation of chromosome 21 occurs sometime after conception. Only some of the cells in mosaic Down syndrome would have an abnormal number of chromosomes. In this type of Down syndrome, the child may not have all the typical characteristics of Down syndrome. This would depend upon the percentage of body cells with the extra chromosome (Blackman, 1990).

There are several typical physical characteristics in individuals with Down syndrome. Some of these are: a small stature, decreased muscle tone (hypotonia), hyperflexibility of the joints, flattened bridge of nose, small head (microcephaly), protruding tongue, wide gap between first and second toe, flat feet, and broad hands with single palmar crease (simian crease). Children with Down syndrome also are usually mentally retarded, although some with mosaic Down syndrome have been found to have normal intelligence (Berkow, 1987). Congenital heart defects and blockage of the small intestine (duodenal atresia) are often present.

Some children with Down syndrome will have instability between the first two cervical vertebra (atlantoaxial subluxation) (Blackman, 1990).

Visual and Auditory Impairments

Children with Down syndrome often have eye abnormalities. When a child is born with Down syndrome, Brushfield spots (gray to white spots around the periphery of the iris) are often present. This usually disappears during the first year of life. The eyes are typically slanted with extra skin folds at the inner corners of the eye (epicanthal folds). Inflammation of the eyelids (blepharitis) may also occur. Problems in visual acuity (nearsightedness and far-sightedness) and strabismus (cross-eyes) are often present (Diamond, 1982). Keratoconus (cone shaped cornea) may also occur (Lucas, 1989) as well as cataracts (Martyn & DiGeorge, 1987).

Children with Down syndrome are often at risk of having hearing impairments. Difficulties in auditory perception have been found (MacMillan, 1977) and mild to moderate hearing loss may be present (Blackman, 1990). Conductive hearing losses may occur from recurrent and persistent middle ear infections.

Outcomes

Vision and hearing problems may remain present. Major medical problems such as congenital heart defects and blockage of the small intestine (duodenal atresia) are typically medically and surgically managed in infancy.

Trisomy 13

Trisomy 13, also known as Patau's syndrome, is an autosomal aberration in which there are three number 13 chromosomes in a cell instead of a pair. This syndrome is characterized by severe physical abnormalities with many individuals dying in infancy.

Some of the more frequent abnormalities include a small head (microcephaly), gross anatomic defects of the brain (such as holoprosencephaly or failure of the forebrain to divide properly), cleft lip and palate, and congenital heart abnormalities. A Simian crease and extra fingers or toes are common (Berkow, 1987). Typically the child is severely mentally retarded.

Visual and Auditory Impairments

Children with Trisomy 13 frequently have microphthalmia (abnormally small eyes), colobomas (fissures) of the iris and retinal dysplasia (abnormal development of retinal tissue) (Berkow, 1987). Cataracts may also be present (Martyn & DiGeorge, 1987). This results in varying degrees of visual impairment. Hearing impairment and deafness may be present.

Usher Syndrome

Usher syndrome is the leading cause of combined deaf-blindness after childhood. It is autosomal recessive and consists of a combined congenital hearing loss and slowly progressive retinitis pigmentosa. This syndrome has been divided into four types, with type

III and IV being very rare. The most common type, Type I, consists of profound congenital deafness with retinitis pigmentosa. There is no mental or neurological impairment. In Type II, there is a moderate-severe hearing loss and retinitis pigmentosa. Normal intelligence is present. In Type III, complete congenital deafness combined with retinitis pigmentosa occurs. Mild motor incoordination is typically present. (See Hallgren syndrome). Type IV is similar to Type I, but mental retardation is present (Regenbogen & Coscas, 1985).

Visual and Auditory Impairments

Retinitis Pigmentosa usually begins around adolescence/early adulthood. The first symptom, night blindness, may be present earlier and result in difficulty seeing in dim light. As retinitis pigmentosa progresses, visual loss in the peripheral fields begins with eventual tunnel vision. Blindness may not occur until middle or late adult life.

The congenital hearing loss is typically sensorineural with a severe to moderate loss in both ears. Usually the loss is present in the higher frequencies.

Alstrom Syndrome

Alstrom syndrome is an autosomal recessive disorder in which affected children usually have obesity (very fat), diabetes mellitus, profound blindness and progressive deafness. Skeletal abnormalities and chronic renal impairment may also occur. Individuals with Alstrom syndrome usually have normal intelligence.

Visual and Auditory Impairments

The first signs of eye disease begin with the presence of nystagmus and the infant/child showing sensitivity to light. Blindness occurs from an atypical retinitis pigmentosa in which there is first a loss of central vision. Vision loss progressively worsens and by age seven, very little vision remains. Mild to moderate cataracts develop in the teens for the majority of individuals with this syndrome. Glaucoma and dislocated lens have been reported as secondary complications (Regenbogen & Coscas, 1985).

A mild sensorineural hearing loss usually occurs in both ears around seven to ten years of age. This progresses to a moderate to severe hearing loss in the second and third decades of life (Regenbogen & Coscas, 1985).

CHARGE Association

CHARGE Association refers to the presence of several concomitant malformations. The mnemonic stands for: coloboma, heart disease, atresia choanae, retarded growth, genital hypoplasia and ear anomalies /deafness. In order to identify an individual as having CHARGE Association, four of these abnormalities must be present. Some individuals suggest that there may be a less severe form of this syndrome.

Coloboma is a slit or groove in the iris, ciliary body, choroid or retina. It is due to a failure of the optic cleft to completely close at about six weeks of fetal life. It may range from a slit or groove in the iris which causes no visual impairment to a missing eye (anophthalmos).

In CHARGE Association, heart disease refers to several possible defects in the heart which are present from birth. (Some of these include ventricular septal defects, patent ductus arteriosus, tetralogy of Fallot and bicuspid aortic valve.)

Choanal atresia is a closure between the nasal cavity and the passageway for air from the nasal cavity to the windpipe (nasopharynx). If it is unilateral (one sided), symptoms are minor. However, if it bilateral (both sides), the infant will show severe respiratory distress and most infants will die from asphyxiation unless corrected.

Other abnormalities are often present in individuals identified as having CHARGE Association. These include mental retardation, small head (microcephaly), cleft lip and palate, and facial nerve palsy. Central nervous system malformations have been present as well (Lin, Siebert, & Graham, 1990). Feeding difficulties may be present. Abnormalities of the genitourinary system have occurred as well as malformations of the vertebrae and ribs.

Visual and Auditory Impairments

As previously mentioned, coloboma is the typical eye abnormality. It may involve one or both eyes. The effect on the person's vision may be minimal or may result in visual field losses (often occurring in the upper field of vision) as well as decreases in visual acuity. Total loss of vision will be present if anophthalmos occurs.

Other eye abnormalities may be present in CHARGE Association. These include microphthalmus (small eyes), optic nerve hypoplasia (defective development of optic nerve), cataract, retinal detachment, nystagmus and disorders of refraction and ocular movement (Russel-Eggitt, Blake, Taylor, & Wyre, 1990).

The mixed hearing loss often results from combination of sensorineural loss and structural deformities in the outer ear. External auditory abnormalities range from an absence of the external auditory canal opening to deformities of the pinna. Chronic otitis media (middle ear infection) may be present. Hearing loss ranges from mild to profound loss (Regenbogen & Coscas, 1985).

Cockayne Syndrome

Cockayne syndrome is an autosomal recessive disorder which consists of dwarfism, mental retardation, retinitis pigmentosa with optic atrophy and deafness. Individual faces are usually thin with prominent noses and large ears. The face usually has the appearance of an old man/woman (progeria). Humpback (kyphosis), microcephaly (small head), enlarged liver and spleen, growth deficiency, unsteady gait and tremors may also be present.

There may possibly be two forms of the syndrome. In one form, the symptoms appear after birth and result in death at five or six years. In the second type, symptoms are not present until the second year of life and the syndrome slowly progresses for twenty years (Regenbogen & Coscas, 1985).

Visual and Auditory Impairments

Individuals with Cockayne syndrome have retinitis pigmentosa with optic atrophy. This results in night blindness, peripheral field deficits and eventually blindness occurring in the second decade of life. Other eye abnormalities include enophthalmos (sunken eyes), cataracts, nystagmus and sensitivity to light (Apple & Rabb, 1991).

Hearing is usually normal at birth, but a progressive sensorineural loss becomes present. It is usually in both ears and results in a moderate to severe hearing loss (Regenbogen & Coscas, 1985).

Crouzon

Crouzon syndrome, also known as craniofacial dysostosis involves skull and facial deformities, with visual and auditory impairments often being present. As the infant's head begins to grow, the skull expands in abnormal directions resulting in various facial and skull deformities. An increase in intracranial pressure may develop as the brain growth resulting in headaches, seizures and injury to the nervous system. Mental retardation may be present (Regenbogen & Coscas, 1985).

Visual and Auditory Impairments

There are several possible eye abnormalities with Crouzon syndrome. Individuals often have exophthalmos (abnormal protrusion of the eyeball) and an abnormal width between the eyes. The exophthalmos may lead to an inability to completely close the eyelids, inflammation of the cornea and in extreme cases, dislocation of the eyeball.

Other eye abnormalities involve strabismus and optic atrophy (Regenbogen & Coscas, 1985). In strabismus, the affected eye deviates from the line of vision. In this syndrome, the eye often deviates outward. In optic atrophy, there is a wasting away of the optic nerve in which visual impairment may occur around six or seven years of age. The vision impairment may be mild, involving an upper visual field deficit, or more severe. Cataracts and retinal detachments may also occur with this syndrome.

When a hearing impairment is present, it is usually nonprogressive with a conductive or mixed loss (although sole sensorineural losses have occasionally been present). The conductive loss is usually due to malformation of the three bones in the middle ear or due to the external auditory canal being narrowed or closed. Other ear abnormalities include the ears being lower set and the pinna being large (Regenbogen & Coscas, 1985).

Outcome

Surgical intervention may be needed during infancy to provide space for the growing brain and prevent further injury. When the individual reaches eight, if there has been no increase in intracranial pressure, vision and mental development are usually good.

Goldenhar Syndrome

Goldenhar syndrome (also known as oculo-auriculovertebral dysplasia) consists of several defects found at birth. There are skin-like cysts on the eyeball (epibulbar dermoids), appendages in front of the ear and ear malformations. Usually these defects only occur on one side. Other abnormalities to the face, teeth and mouth area may be present (e.g. receding chin, small jaw, and cleft lip and palate). Mild spinal abnormalities have been found in about half of individuals with this syndrome. Congenital heart disease and renal abnormalities may be present as well (Regenbogen & Coscas, 1985).

Visual and Auditory Impairments

The eye has several physical abnormalities which may impair vision. The skin like cysts (epibulbar dermoids), which is the most frequent feature of this syndrome, may cause irregular stigmatism. Coloboma of the eyelid, iris or choroid may occur. Cataracts, nystagmus, strabismus, and retinal detachment have also been present, adversely affecting vision. Central visual pathway abnormalities may also occur, further impairing vision.

Physical malformation of the ear in the form of appendages near the ear or malformed outer ear is common in this syndrome. Conductive hearing loss may occur due to the external ear canal being absent or narrowed. Abnormalities in the middle ear may also be responsible for the conductive loss. Sensorineural hearing loss may occur from abnormalities in the inner ear. A mixed loss may be present as well. Hearing loss is usually only in one ear.

Hallgren Syndrome

Hallgren syndrome is an autosomal recessive disorder which has been considered as a third type of Usher syndrome. It consists of ataxia (muscular incoordination), congenital deafness and retinitis pigmentosa. Ataxia is usually mild and presents itself as a swinging gait. Children will usually walk late because of it. Mental retardation has been associated with some individuals with this syndrome. Less common symptoms include skeletal abnormalities (club foot, hunchback) and seizures (Regenbogen & Coscas, 1985).

Visual and Auditory Impairments

The primary visual problem in Hallgren syndrome is retinitis pigmentosa. Night blindness is usually present by preschool age and a diagnosis of retinitis pigmentosa occurs between seven and fifteen years of age. The retinitis pigmentosa found in individuals with Hallgren syndrome usually has a better prognosis than typical retinitis

pigmentosa. There is usually a moderate rate of progression of the condition with blindness occurring between 41 and 70 years of age (Regenbogen & Coscas, 1985).

The majority of individuals with this syndrome have total or almost total sensorineural deafness of both ears. Other individuals do have enough hearing to understand words. The hearing loss is not progressive.

Kearns-Sayre Syndrome

Kearns-Sayre Syndrome is an autosomal syndrome in which there is a defect in one of the (mitochondrial respiratory chain) enzymes which leads to structural abnormalities of the energy producing element of cells (mitochondria) (Gota, Itami, Kajii, Tochimaru, Endo & Horai, 1990). This syndrome is characterized by a progressive external ophthalmoplegia (ocular muscle paralysis), atypical retinitis pigmentosa and sensorineural hearing loss. Heart problems often develop later in this syndrome and range from mild abnormalities (e.g., bundle branch blocks) to serious blocks (e.g., AV heart block) which may require a pacemaker (Regenbogen & Coscas, 1985).

Several other symptoms may be present. Cerebellar ataxia (muscular incoordination) can occur and it is often mild at first. However, it can progressively worsen, adversely affecting movement. Growth may be impaired in children who have this syndrome before puberty. Mental retardation may be present. Weakness in the face, neck and extremities may also occur.

Three forms of the syndrome have been identified. There is an infantile form which tends to have a severe and rapid course. There is also a juvenile form. The adult form is usually less severe and progresses slowly.

Visual and Auditory Impairment

The first visual impairment is the external ophthalmoplegia. This becomes progressively worse over months or years until complete paralysis of the ocular muscles occur.

The atypical retinitis pigmentosa begins soon after the presence of external ophthalmoplegia and is usually in both eyes. It can cause a wide range of visual impairments. In many cases the abnormal retinitis pigmentosa may cause only minor to no loss of visual acuity. However, in some cases the retinitis pigmentosa is more typical and cause visual field deficits. This may take the form of some peripheral loss of vision, central scotoma (blind spot in the center of vision), ring scotoma or blind spots in other areas. Some individuals can compensate for the blind spots by tilting their heads backwards. Other eye abnormalities may include pupil abnormalities and cataracts (Regenbogen & Coscas, 1985).

In terms of auditory problems, sensorineural hearing loss has been found in about half of the individuals with this syndrome (Regenbogen & Coscas, 1985). The hearing loss is greater in the higher frequencies, but can range overall from mild to profound. The sensorineural loss may be progressive and may begin between three and ten years of age.

Mucopolysaccharidosis

The mucopolysaccharidosis (MPS) are inborn errors of mucopolysaccharides metabolism in which there is a defect or deficiency in (lysosomal) enzymes that degrade certain mucopolysaccharides. The incompletely degraded mucopolysaccharides accumulate in several organ systems and lead to progressive abnormalities. Five types of MPS have both visual and hearing abnormalities and most are autosomal recessive. The five types include: Hurler (MPS-I H), Scheie (MPS I-S), Hunter (MPS II), Morquio (MPS IV), Maroteaux-Lamy (MPS VI).

Hurler Syndrome (MPS-I H)

The major characteristics of Hurler's syndrome consist of dwarfism, large head, nasal discharge, skeletal abnormalities, enlarged liver and spleen and ape-like posture. The facial features have led to the name of "gargoylism" to refer to this entire syndrome. The progressive mucopolysaccharides deposits in the windpipe (trachea) can lead to airway obstruction if not treated (Adachi & Chole, 1990). The full features of the syndrome develop between one and two years of age. After two years, mental development deteriorates and often severe mental retardation is present. Neurological symptoms appear as degeneration occurs. These symptoms include seizures, spastic paraplegia, nystagmus and ataxia (muscular incoordination) (Regenbogen & Coscas, 1985).

Visual and Auditory Impairments

The most common ocular feature of Hurler's syndrome is corneal clouding which often appears at three year of age and progresses slowly, affecting visual acuity. Visual acuity may be severely affected as the individual reaches 30 years of age or older. Occasionally the eyes may have painful episodes with the sensation of a foreign body begin in the eye as well as photophobia (avoidance of light). Other abnormalities include atypical regional pigmentary degeneration and optic atrophy (Collins, Traboulsi, & Maumence, 1990).

Most individuals with Hurler's syndrome have some degree of mild mixed hearing loss which is progressive. The conductive loss may be due to repeated otitis media (middle ear infections) and deformity of the ossicles (three bones in the middle ear). Deformity of the inner ear may also exist resulting in sensorineural loss (Regenbogen & Coscas, 1985).

Scheie Syndrome (MPS-I S)

Scheie syndrome is caused by the same enzyme defect as Hurler's syndrome, but it is less severe and individuals with this syndrome are of normal intelligence. Some characteristics which individuals with Scheie may have is corneal clouding, skeletal abnormalities (hand and face deformity), broad faces (but not gargoylism), and heart abnormalities. Respiratory infections are common (Keith, Scully & Weidman, 1990).

Visual and Auditory Impairments

For individuals with Scheie syndrome, slowly progressive corneal clouding occurs which interferes with vision. The corneal clouding may be present at birth or not until the second decade of life. Diminished vision in bright light occurs with decrease in field of vision. Glaucoma may also be present. In some cases there is atypical retinitis pigmentosa (Apple & Rabb, 1991).

There is typically a mixed hearing loss which is progressive. It is usually present in middle age and rarely develop into a total loss (Regenbogen & Coscas, 1985).

Hunter Syndrome (MPS-II)

Two forms of Hunter's syndrome occur with differing outcomes. The common severe form (MPS II A) results in mental deterioration and death, usually prior to the fifteenth birthday. The other form (MPS II B) is milder and the individual usually has normal (or near normal) intelligence with a normal life expectancy.

Many of the symptoms of MPS II are similar to MPS I, but milder. During the first year of life, respiratory infections, noisy breathing and hernia may be present. Not until two years of age do the more prominent features occur. These include: growth retardation, joint stiffness, enlarged liver and spleen and protruding abdomen. Two to six year old boys are typically hyperkinetic. After five to six years of age, physical and mental abilities deteriorate. The individual has night blindness, mixed hearing loss, dwarfism and gargoyle-like features. Complications include heart disease, mucoid diarrhea, upper airway obstruction and hip disease (Regenbogen & Coscas, 1985).

Visual and Auditory Impairments

Optic atrophy and atypical retinitis pigmentosa are often present in individuals with Hunter's syndrome. Severe night blindness is often present, interfering with vision in dim light. Other eye abnormalities include buphthalmos (infantile glaucoma in which there is an enlargement of the eye), abnormal width between the two eyes, glaucoma and exophthalmus (protrusion of the eyeball). Vision loss and blindness may occur (Regenbogen & Coscas, 1985).

A progressive hearing loss is usually present. It may be mixed or sensorineural. Recurrent otitis media may also occur.

Morquio Syndrome (MPS IV)

This form of mucopolysaccharidosis disease may be present in a severe form (MPS IV A) or a mild form (MPS IV S). The main features consist of dwarfism, prominent lower face, thin enamel on the teeth, knock knees and joint laxity. A lack of endurance may occur about 4 to 6 years of age with increased difficulty walking. There is a tendency for respiratory infections. Intelligence is usually normal. Some additional abnormalities include: heart valve defects (aortic regurgitation) and enlargement of liver and spleen (Regenbogen & Coscas, 1985).

Visual and Auditory Impairments

Corneal clouding occurs in this syndrome around ten years of age. However, this cloudiness is milder than other forms of MPS resulting in only slightly impaired visual acuity. Hearing impairment begins in adolescence and progresses. It is usually a conductive loss, but becomes mixed as the person ages. The mixed loss is usually bilateral with a mild-moderate loss in all frequencies. An ossicular chain (three bones in the middle ear) deformity may cause the conductive hearing loss (Regenbogen & Coscas, 1985).

Maroteaux-Lamy Syndrome (MPH VI)

Maroteaux-Lamy Syndrome is another mucopolysaccharide defect which resembles Hurler syndrome. There are three forms: severe, intermediate and mild. General characteristics include growth retardation resulting in short stature and severe disabilities humpback, flared rib cage, and breastbone protrusion. Deformities of the knees, hips, face and hand may occur. Enlarged liver and heart abnormalities may be present. With the milder form of this syndrome, gross skeletal deformities and cataracts are absent. In all forms of Maroteaux-Lamy syndrome, individuals are of normal intelligence (Regenbogen & Coscas, 1985).

Visual and Auditory Impairments

Bilateral corneal clouding also occurs in this MPS syndrome and is progressive. As the corneal clouding advances, visual acuity may be severely impaired. Optic atrophy may occur.

Conductive, sensorineural and mixed hearing losses in both ears occur (Regenbogen & Coscas, 1985).

Refsum Syndrome

Refsum syndrome is an autosomal recessive syndrome which affects the nervous system, eye, ear, heart and skin and is progressive. The major symptoms are chronic peripheral polyneuropathy, cerebellar ataxia, retinitis pigmentosa and progressive deafness. The neuropathy usually affects the feet and legs resulting in motor weakness and wasting away of muscles (muscle atrophy). A loss of deep sensation, superficial sensation to pain and parathesis may occur. In ataxia, there is an unsteadiness of gait.

Other symptoms include cardiac abnormalities (enlarged left ventricle of the heart, fast heart rate and irregular rhythms) and skeletal malformations. Age of onset varies from child to adult life. Sudden exacerbations and remissions may occur. Sudden, unexpected death has occurred in some cases (Regenbogen & Coscas, 1985).

Visual and Auditory Impairments

Retinitis pigmentosa is the primary visual abnormality found in Refsum syndrome. Night blindness is usually the first symptom in childhood. A loss in peripheral vision occurs usually followed by concentric ring scotomas. Eventually only tunnel vision remains. Congenital cataract and nystagmus may also occur.

A sensorineural hearing loss begins in the second decade of life with the higher frequencies especially being affected. Progression may be slow or sudden. A complete loss may result (Regenbogen & Coscas, 1985).

Turner Syndrome

Turner syndrome is one of the most common chromosomal abnormalities, in which there is a missing (or partially missing) sex chromosome. Due to this, it is also called the XO syndrome. Individuals are typically short in stature with height under five feet and there is usually a failure to develop secondary sexual traits during puberty. About half of those with this syndrome will have a webbing of the neck and peculiar "heart-shaped" faces. Other symptoms include low neck hairline, high palate, dental malocclusion, and congenital heart disease. Other additional abnormalities may include skeletal deformities, high blood pressure, and renal and urinary abnormalities. Also, poor visual memory, deficits in spatial ability and poor motor coordination may be present (Regenbogen & Coscas, 1985).

Visual and Auditory Impairments

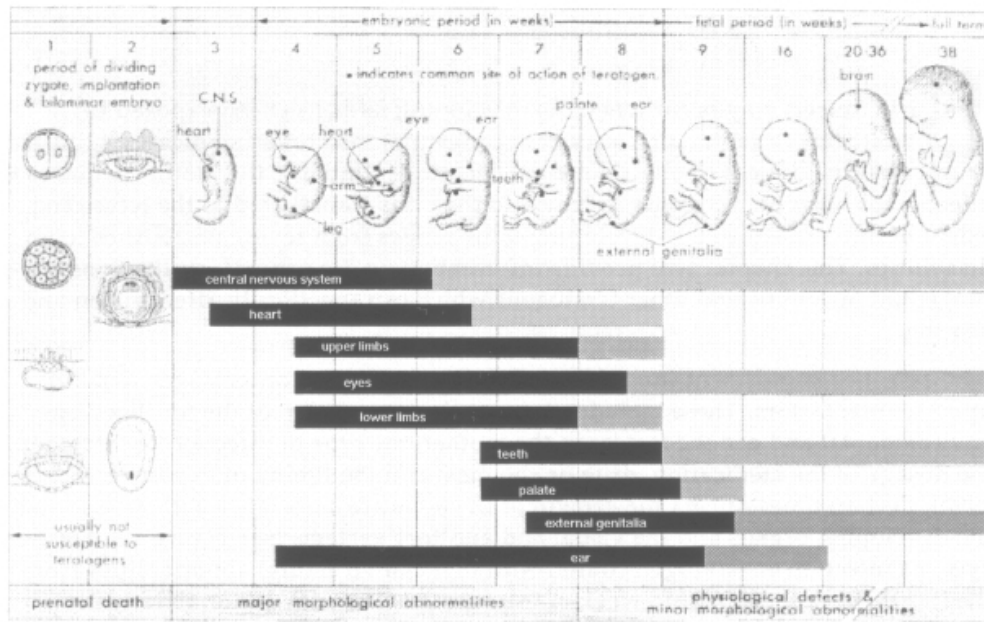
Although eye abnormalities are not common in Turner syndrome, eye abnormalities have been found. Congenital epicanthus (fold of skin over eye), strabismus, and ptosis (drooping of eyelid) have been found to occur. Other eye abnormalities have been occasionally found. These include strabismus, cataracts, farsightedness, amblyopia, and color blindness.

Hearing abnormalities have been found in Turner syndrome but not frequently. Conductive, sensorineural and mixed hearing losses have all been present. Conductive losses may occur from outer and middle ear abnormality. Recurrent otitis media (middle ear infections) are very common and often occur in early childhood (Sculerati, Ledesma-Medina, Finegold & Stool, 1990). When conductive loss occurs, it is often moderate with more difficulty hearing in the higher frequencies. When sensorineural loss occurs, a particular pattern of the hearing loss is found on the audiogram with a symmetrical dip in the middle frequency range. (The maximum dip is between 500 and 2000 HZ with a loss of 20 - 70 Db.) Hearing loss is not progressive (Regenbogen & Coscas, 1985).

Waardenburg Syndrome

Waardenburg syndrome is usually an autosomal dominant mutation producing a range of characteristics. Major characteristics include, hypopigmentation, deafness, optic abnormalities, alterations of the skeletal system (especially a wide distance between the eyes), dystopia canthorum medialis (malposition of the eyelids) and a white forelock or early graying of the hair (Asher, Morell & Friedman, 1991).

Three types of Waardenburg syndrome exist. Waardenburg syndrome Type 1 (WS-1) occurs the most frequently and has the characteristic facial abnormalities. Type 2 is more



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associated with deafness. Type 3 is the most severe form with many musculo-skeletal abnormalities in the face and arms.

Visual and Auditory Impairments

The cardinal sign of Type 1 Waardenburg is the increased distance between the eyes, but visual acuity is usually normal. Abnormal coloration of the iris, drooping of the eyelid and cataracts may also be present and affect vision.

Sensorineural deafness often occurs in this syndrome and it ranges in severity. Three distinct patterns of hearing loss are usually present. The first pattern consists of almost total deafness with slight residual hearing present in the low frequencies. The second type is severe deafness in the low frequencies, but normal in 6000-8000Hz range. The third type is unilateral (one sided) moderate hearing loss. Hearing loss may be progressive (Newton, 1989; Regenbogen & Coscas, 1985).

Other Syndromes

For a listing of other syndromes associated with deaf-blindness, See Table 2.

Other Syndromes Associated with Deaf-Blindness

| | |
|-------------------|------------------------|
| Alport | Moebius |
| Apert | Norrie |
| Bardet-Biedel | Nyssen-van Bogaert |
| Cogan | Pendred |
| Diallinas-Amalric | Rosenberg-Chutorian |
| DIDMOAD | Sarcoidosis |
| EEC Eldridge | Small |
| FOAR | Spranger-Wiedemann |
| Garcin | Strickler |
| Gradenigo | Strachnan |
| Harboyan IVIC | Vogt-Koyanagi-Harada |
| Kniest Dysplasia | Wegener Granulomatosis |
| LADD | Wildervanck |
| Marshall | Ziprkowski-Margolis |

Table 2 Other syndromes associated with deaf-blindness

*Congenital Infections
and Teratogens*

Birth defects may occur when the developing fetus has been exposed to teratogenic agents, such as certain congenital infections, medications, illegal drugs, maternal illness or environmental chemicals. Initially, the mother acquires the infection or takes the teratogenic drug and then it is transmitted to the fetus. The extent of impairment to the developing fetus ranges from severe disabilities to no disabilities. This chapter will provide information on the general implications of congenital infections and other teratogens, which can specifically affect vision and hearing.

The extent of impairment caused by a congenital infection or teratogen depends upon several factors. These include: the genetic composition of the fertilized egg, environmental and genetic factors in the mother, the type of infection or teratogen, the dosage of the medication, drug or chemical and the timing of exposure (Schor, 1990). Certain times during the growth of the fetus are associated with greater risk to the developing organs and the visual and auditory systems (see Figure 12). For the eyes, an infection or teratogen could result in major structural abnormalities from the fourth week to the eighth week of intrauterine life, with minor abnormalities possible from the eighth week to full term. For the ears, an infection or teratogen could result in major structural abnormalities from the fourth week to the ninth week, with minor abnormalities occurring from the ninth week to about the twentieth week.

Schematic illustration of the sensitive or critical periods in human development. The dark band denotes highly sensitive periods, the light band indicates stages that are less sensitive to teratogens. Note that each organ or structure has a critical period during which its development may be deranged and that physiological defects, functional disturbances, and minor morphological changes are likely to result from disturbances during the fetal period. Severe mental retardation may result from exposure of the developing human to high levels of radiation during the 8 to 16 week period.

Congenital Infections

A group of the most common congenital infections resulting in birth defects are referred to by the mnemonic "STORCH". This stands for: Syphilis, Toxoplasmosis, Other, Rubella, Cytomegalovirus, and Herpes. Most infections which the mother acquires during pregnancy rarely result in any serious fetal infection. However, a STORCH infection may affect or damage the placenta or directly injure fetal organs. The extent of vision and hearing loss as well as other defects depend on the type of infection and the trimester in which the fetus was infected. Infections which occur early in pregnancy are more likely to result in more serious birth defects (Anderson, Bale, Blackman, & Murph, 1986).

With congenital infections, there is not only possible damage to the ears and eyes, but other common abnormalities often occur. These other abnormalities are summarized in Table 3. Similar complications to these infections are also found. Some of these complications include mental retardation, shortened life span, poor body growth, motor abnormalities, and glandular disturbances (e.g. thyroid and diabetes). For a better understanding of each of these congenital infections and their effect on vision and hearing, each infection will be described.

Frequently Found Impairments of TORCH Congenital Infections

| | Syphilis | Toxoplasmosis | Rubella | CMV | Herpes |
|--------------------------|----------|---------------|---------|-----|--------|
| Eye/vision impairments | X | X | X | X | X |
| Ear/auditory impairments | X | X | X | X | X |
| Anemia | X | X | X | X | X |
| Brain calcifications | | X | | X | |
| Bone abnormalities | | | X | | |
| Congenital heart defects | | | X | | |
| Encephalitis | X | X | X | X | X |
| Hydrocephalus | | X | X | X | X |
| Jaundice | X | X | X | X | X |
| Liver/spleen enlarged | X | X | X | X | X |
| Low birth weight | X | X | X | X | X |
| Low platelet count | X | X | X | X | X |
| Microcephaly | X | X | X | X | X |
| Pneumonia | | | X | X | X |
| Seizures | X | X | X | X | X |
| Skin rash | X | X | X | X | X |

Table 3 Frequently found impairments of TORCH

Congenital Syphilis

Congenital syphilis is an infection (by the *Treponema pallidum* bacteria) which is transmitted from the untreated infected mother to the fetus. Although it is one of the oldest and most easily preventable infections of the TORCH group, there has been an increased incidence of congenital syphilis occurring in the United States (Rofls & Nakashima, 1990).

The mother acquires this infection through sexual contact with another person who has the infection. The infection progresses through a series of stages. In the first stage, primary syphilis, a localized lesion develops at the site of the initial infection. Often the

pregnant mother who has typically acquired this infection through sexual contact with an infected individual does not realize the significance of the disease and may not seek treatment. If left untreated, secondary syphilis will develop in which the disease may extend to the skin, mouth, genital area and central nervous system. The infection may progress into the third stage, known as tertiary syphilis in which the infection spreads to the adult's organ system. During any of these stages, the syphilis infection could be transmitted to the fetus through the placenta.

Congenital syphilis occurs in two forms. In the first form, congenital syphilis may present itself at birth or early infancy. This is known as early congenital syphilis or infantile syphilis. The infant may have skin lesions, lymph node involvement, blood stained nasal discharge, and have an "old-man" look. Multiple organ involvement may be present. Organs especially affected include: the central nervous system, liver, lungs, bones, skin and kidney (Regenbogen & Coscas, 1985; Williamson & Demmler, 1992). Some infants also have seizures, hydrocephalus, meningitis, and mental retardation (Berkow, 1987). Approximately 25% of infants die in first few months (Anderson, Bale, Blackman, & Murph, 1986).

Many infants born with congenital syphilis remain in a latent stage where symptoms are not present at birth. This second form is known as congenital syphilis tarda. Approximately one third of these individuals develop symptoms in childhood, adolescence or adult life. If symptoms develop, there may be ulcers, vision and hearing impairments, paralysis or wasting away of tissue (atrophy) as well as other neurological abnormalities. For some individuals, symptoms may never develop.

Visual and Auditory Impairments

The most frequent finding in congenital syphilis is eye abnormalities. Eye conditions such as interstitial keratitis, astigmatism, chorioretinitis, iridocyclitis, glaucoma and optic atrophy may be present.

Forty percent of individuals affected with the latent form of congenital syphilis develop what is known as interstitial keratitis. Typically this occurs between five years of age and late teens. Usually both eyes are involved. This condition typically follows three stages: onset, full development and recovery. During the onset stage (1-2 weeks), there is a cloudiness on the cornea with impairment of visual acuity. For more than fifty percent of affected individuals, this begins in the periphery and advances centrally. The full development phase results in the cornea becoming very red and inflamed. A ground glass appearance is present on the cornea, obscuring the iris. There is usually impairment of vision during this time. This phase may last from 8 to 16 weeks. During the recovery stage, which may last from 1 to 2 years, clearing begins at the periphery and moves towards the center. Some cornea opacity may remain or it may clear. Even if it clears completely, vision may remain impaired. Often a central corneal haze will continue to persist. From this disorder, the cornea often has some thinning and irregularities. This results in astigmatism, which interferes with visual acuity. This disorder may also result in severe interference with vision when a membrane involving the eye splits.

In congenital syphilis there is often inflammation of various parts of the eye. In approximately 20 -25% of the cases of congenital syphilis, chorioretinitis is present which is an

inflammation of the choroid and retina (Regenbogen & Coscas, 1985). This may result in diminished and hazy vision (Berkow, 1987). Iridocyclitis (inflammation of the iris and ciliary body) may be present as a wide spread inflammation of the anterior portion of the eye. From these inflammations, scarring, secondary glaucoma, atrophy (wasting away of tissue) of the optic nerve, and retinal detachment may occur.

A sensorineural hearing loss is often present in individuals with congenital syphilis. This hearing impairment may occur in early childhood (as in approximately one third of the cases), or later in life. There is a wide fluctuation in degree of hearing impairment present. For mild cases, there may be difficulty in only hearing the lower frequencies. A severe loss often results in poor speech discrimination with the audiogram often being flat. (A flat audiogram indicates a similar level of loss across all frequencies.) For about half of the individuals who exhibit hearing loss, it is progressive (Regenbogen & Coscas, 1985).

Outcomes

Outcomes depend upon when treatment was started and the amount of tissue involvement. When therapy is started prior to the second year of life, there is usually no permanent eye damage. Medication can be given to clear the cornea with a 85-90% success rate (Regenbogen & Coscas, 1985). When treatment is begun late, deficits in vision may still occur. As for hearing, aggressive treatment may decrease or stop the progressive loss of hearing. However, a hearing impairment typically remains.

Congenital Toxoplasmosis

Toxoplasmosis is an infection (with the *Toxoplasma gondii*) by a parasite which is transmitted to an adult from contact with infected cat feces or meat (especially pork) which has not been completely cooked. In the healthy adult, symptoms are very mild or nonexistent. However, if a pregnant woman contracts this infection, the infection may be transmitted to the fetus.

There is a large range of symptoms which can occur to the child born with congenital toxoplasmosis. Congenital toxoplasmosis acquired during the first trimester of pregnancy typically results in the worst symptoms with the possibility of brain abnormalities (including hydrocephalus, microcephaly, and intracranial calcification). Enlargement of the liver and spleen, anemia and decreased platelets may also occur. On the other extreme, congenital toxoplasmosis contracted during the third trimester of pregnancy often results in the infant having no symptoms.

Neurodevelopmental abnormalities from the infection may occur months or years after birth. Some possible abnormalities include seizures, psychomotor disturbances, cerebral palsy, and mental retardation. (Anderson, Bale, Blackman, & Murph, 1986; Berkow, 1987; Williamson & Demmler, 1992).

Visual and Auditory Impairments

Visual impairment from congenital toxoplasmosis ranges from a mild vision loss to blindness. Chorioretinitis (an inflammation of the retina and choroid area) may occur and result in diminished and hazy vision. Complications of this can result in blindness. Retinal detachment (Lucas, 1989) and cataracts have also been present in children with congenital toxoplasmosis (Martyn & DiGeorge, 1987). Actual death of tissue in the retina (necrosis) may be present (Berkow, 1987).

A sensorineural hearing loss resulting in damage to the inner ear may also occur. This may be present at birth or develop later when no other symptoms are present. Degree of hearing loss varies. A profound hearing loss can occur (Berkow, 1987).

Outcomes

If blood tests are done and the mother is found to have toxoplasmosis, drug treatment will be given to try to reduce the risk of transmission to the fetus (Daffos, Forestier, Capell-Pavlovsky, et. al., 1988). Although medication will be given to the infant with congenital toxoplasmosis to kill the organism and prevent further damage, prognosis remains poor when it was acquired during the first trimester. Routine eye exams will be necessary due to the risk of eye impairment occurring later after birth.

Congenital Rubella

Rubella (also known as German measles) was the most common viral cause of birth defects until a vaccination was developed (Anderson, Bale, Blackman, & Murph, 1986). In children and adults who have acquired Rubella, only mild symptoms are typically present. However, when a pregnant mother is infected with the Rubella virus, transmission of the virus to the fetus can result in multiple impairments.

Infants with congenital rubella may present with several abnormalities or may be symptomless. Abnormalities which may be present include cardiac defects, (patent ductus arteriosus, ventricular septal defects), vision and hearing defects and some organ involvement. Additional abnormalities include: low birth weight, liver and spleen enlargement, inflammation of the brain, microcephaly, decreased platelets (thrombocytopenia), jaundice, anemia, swelling of the lymph nodes (adenopathy), abnormalities with balance (from vestibular involvement), and inflammation of the lungs (pneumonitis). Other organs may be involved as well (kidney, skin and bones).

Long term outcomes for children born with symptoms of the Rubella virus consist of a continuation of some degree of long term impairment. Hearing and vision losses continue. Seizures and cerebral palsy may occur. Although most children with congenital rubella have normal intelligence, mental retardation has been found in some children.

Disorders may emerge long after the acute original rubella infection is over, even in children who had no symptoms of the infection at birth. Children may develop encephalitis (inflammation of the brain) with concomitant learning deficits and behavioral disturbances (Desmond, Wilson & Vanselman, 1985). Other problems such as lack of motor co-

ordination may occur. These changes may take a long time to manifest themselves, occurring from age 2 to 7, with an increase in severity occurring up to twelve years of age (Wolff, 1985). In some cases, hearing losses have been found to occur later in adolescence. Visual impairments have been found to progressively worsen. Close monitoring is necessary throughout the person's life for possible complications occurring from having congenital rubella (Williamson & Demmler, 1992).

Visual and Auditory Impairments

Multiple abnormalities may occur to the eye in infants born with congenital rubella. This involves cataracts, abnormalities to the cornea, iris, ciliary body and retina, glaucoma, microphthalmus (small eyes) and various ocular motor disorders.

Cataracts are the most common eye manifestation. It is a clouding of the lens of the eye and it is usually present at birth, occurring in one or both eyes. Cataracts may be surgically removed. However the combination of cataracts and other abnormalities of the eye often result in poor visual acuity.

The cornea of the eye may present with clouding which may clear or remain as a haze. The cornea may also possibly change shape resulting in poorer refractory power of the eye (keratoconus). Swelling of the cornea may also be present (acute hydrophs of the cornea) (Wolff, 1985).

Abnormalities of the iris and ciliary body may be present. This may include inflammation, atrophy (wasting away), or tissue death in parts of the iris and ciliary body. This may result in pain, redness, photophobia (intolerance of light) and diminished vision. Absence of the muscle which dilates the eye may also be present.

A disorder of the retina (pigmentary retinopathy) is often present and may be progressive. There is often good visual acuity with this, although there have been some cases with a decrease in visual acuity to 20/60. One or both eyes may be involved with central, peripheral irregularities distorting vision (Wolff, 1985).

About ten percent of the children with congenital rubella will develop glaucoma (Wolff, 1985). Glaucoma is a disorder which has increased pressure in the eye due to more eye fluid in the anterior chamber being present than can be absorbed. The increased pressure can cause visual impairments ranging from a slight loss of vision to complete blindness. Typically there is poor visual acuity. Unfortunately in congenital rubella, glaucoma may occur long after the virus is thought to be unable to produce any new disorders.

Physical abnormalities of the eyes may be present such as microphthalmos (small eyes). This may be accompanied with a small brain and small head. Several disorders involving the movement of the eye may occur in twenty percent of children with congenital rubella. Strabismus, which is when the eye(s) deviate from correct alignment, are often present. It can result in diplopia (double vision) or the lack of use of one eye. Nystagmus, which is involuntary cyclical movements of the eyeball, may occur. Ocular torticollis, may also be present (Wolff, 1985).

Optic nerve atrophy and deficits in the visual cortex may also occur in congenital rubella. In both of these conditions, minimal visual loss to a total loss of vision can occur.

Severe refraction errors, especially myopia (near-sightedness) may also be present. It is not uncommon to have acuity worse than 20/200.

As for hearing, a sensorineural loss is the most common long term problem. It is typically present in both ears, but it can involve only one. Degree of hearing impairment varies from mild hearing impairment to profound deafness. Usually the individual is not totally deaf, but can hear high-pitched sounds. Audiogram often show a “belly-up” curve with the greatest loss at 1000 Hz. Poor speech discrimination can occur (Regenbogen & Coscas, 1985). A mild conductive loss may also be present. Many infants who were born without symptoms of Rubella later developed deafness (Anderson, Bale, Blackman, & Murph, 1986). Often the hearing loss is progressive.

Outcomes

At this time, there is no effective antiviral treatment (medication) for congenital rubella. Surgery may be performed on the eye when cataracts and glaucoma are present, with varied success. Prescriptive lenses and hearing aids may be prescribed. The infant with congenital rubella may excrete the virus for six months or much longer. Nonimmunized pregnant women should exercise caution with interaction with infants still shedding the virus.

Cytomegalovirus (CMV)

Cytomegalovirus is an infection that is commonly acquired with little to no symptoms by most healthy individuals before they are fifty years old (Anderson, Bale, Blackman & Murph, 1986). There are typically no impairments resulting from this infection in healthy individuals. However, if CMV is transmitted from the pregnant mother to the fetus, the unborn child may be severely affected.

There are a wide range of symptoms in the infected infant, ranging from no symptoms or impairments to severe or fatal involvement. Possible symptoms and complications include damage to numerous organs (including brain, liver, spleen, heart, and kidney), decreased platelet count (thrombocytopenia), skin rash, and microcephaly (decrease growth of head and brain). Stillbirth, miscarriages and death shortly after birth have occurred.

Long term outcome of children born with symptoms of CMV include mental retardation, motor abnormalities and seizures (Anderson, et. al., 1986). Cerebral palsy, hearing and vision losses may occur as well. Specific learning disabilities may occur in those individuals with normal intelligence (Williamson, Desmond, LaFevvers, Taber, Catlin & Weaver, 1982).

Children who were exposed to CMV congenitally, but were born without symptoms, are at risk of developing problems later. Sensorineural hearing loss may occur later. Some studies suggest that mental retardation and cerebral palsy may also occur (Hanshaw, Scheiner, Mosley, Gas, Abel, & Scheiner, 1976; Coboy, Pass, Stagno, et.al., 1986).

Visual and Auditory Impairments

Several eye disorders may be present causing visual impairments and blindness. Retinitis (inflammation of the retina) is common and may result in blurred vision and vision loss. Unlike toxoplasmosis, if retinitis is not present at birth, it will not typically occur later in life even when the virus is present.

Associated with retinitis may be optic atrophy which is a wasting away of the optic nerve. Vision loss is proportional to amount of nerve damage. Total blindness is possible.

Several other eye abnormalities may occur. Anophthalmia (absence of the eyeball) may occur. Coloboma may also be present. Iridocyclitis, or inflammation of the iris and ciliary body may also occur in which the person has severe pain, redness and photophobia (intolerance of light). Other problems include defects in tissue development of the optic nerve (Egbert, 1985) and cataracts (Martyn & DiGeorge, 1987).

A sensorineural hearing loss is the most common late developing symptom of the congenital infection. The amount of hearing loss ranges from mild to profound. More often it will occur in both ears and may be progressive (Morgan, 1987).

Outcomes

There is no effective medical treatment available for congenital cytomegalovirus.

The virus may be shed by the infant with congenital CMV for years. CMV may be present in the urine, saliva, blood, tears, stool, cervical secretions and semen of individuals with congenital cytomegalovirus. Unless proper infection control measures are used (e.g. proper handwashing technique, proper disposal of wastes, cleaning environmental surfaces) persons working with the infant or young child may acquire the infection. If this is the case, symptoms are usually minimal, but the pregnant women may pass it on to her fetus.

Neonatal Herpes Simplex Virus (HSV)

Neonatal herpes simplex virus causes serious illness and high death rate in newborns. There are two types of this virus: Type 1 which causes most cold sores and Type 2 which results in most genital lesions. Approximately 80% of the neonatal herpes simplex virus infections are caused by Type 2 (Berkow, 1987).

Most STORCH infections are acquired from the mother by transmission of the virus through the bloodstream to the placenta and then to the fetus. Neonatal herpes however, is typically acquired during birth as the infant passes through the infected birth canal, or by the infection ascending upwards to the fetus after the amniotic membranes have ruptured.

Symptoms usually appear between the first and second week after birth, but may not appear until the fourth week. Initially skin vesicles appear and often lead to a more serious form of the disease unless treatment is begun. (Approximately 15% of the infected

infants will have no skin lesion, but will develop encephalitis (inflammation of the brain). Other symptoms which develop include drowsiness, respiratory problems, hypotonia (low tone), hepatitis (inflammation of the liver) and diseases of coagulation. Seizures and coma are possible with brain involvement (Berkow, 1987).

Infants with this infection fall into three classifications. The first classification is known as neonatal herpes simplex. In neonatal herpes simplex, clotting disorders (disseminated intravascular coagulation) and organ abnormalities are present (especially the liver and lungs). In this category, inflammation of the brain or skin abnormalities may or may not be present. The mortality rate for this category is the highest.

The second and third categories are both called localized disease. In one of these, inflammation of the brain (encephalitis) with neurological abnormalities are present. The other type of localized disease is where only the skin, eye, and mouth are involved (Gammon & Nahmias, 1985).

Visual and Auditory Impairments

There are a wide range of eye problems in children with neonatal herpes. On the one extreme there may be a mild eye problem such as conjunctivitis (inflammation of the membrane which lines the eyelid and goes around the eyeball) which causes no vision problems. On the other extreme there can be severe optic nerve atrophy (wasting away of the optic nerve) or retinitis (inflammation of the retina) which can lead to blindness (Anderson, Bale, Blackman, & Murph, 1986; Gammon & Nahmias, 1985).

Several other eye disorders may be present. Inflammation, lesions, and cloudiness of the cornea may be present (keratitis) which may lead to a vision loss. Uveitis, inflammation of the iris, ciliary body and/or choroid, may also occur. When the inflammation is only to the back portion of the eye containing the choroid and retina, a condition known as chorioretinitis occurs in which diminished vision and blindness may result. If inflammation of the optic disk is present, optic atrophy and possible blindness may occur (Gammon & Nahmias, 1985).

Occasionally there may be retina detachment where the retina tears and partially separates from the back of the eye resulting in vision loss unless surgically replaced. Surgery must be done immediately or serious vision loss will occur. Cataracts (a clouding of the lens of the eye) may result in which vision is blurred.

The eye muscles may be affected resulting in strabismus. One or both eyes deviate inward, outward up or down directions. Double vision may result or one eye may not be used.

The spread of herpes simplex virus into the brain may result in defects of the visual pathway. Affected individuals may have visual field deficits where the right or left field of vision is gone in one or both eyes. Vision loss in one fourth of the visual field (quadrants) may also be involved.

Other possible eye problems include microphthalmos (small developed eye) and microcornea (small cornea).

Children infected with neonatal herpes simplex are in the high-risk category for developing hearing loss (Morgan, 1987).

Outcomes

Prompt treatment of this infection increase the likelihood of survival as well as increasing the number of infants who will develop normally (from 10 % to 35%) (Berkow, 1987). Prognosis depends on the extent of the disease. Some children with the disseminated form will have 20% to 30% chance of having multiple impairments including poor growth, mental retardation and blindness (Anderson, Bale, Blackman & Murph, 1986).

Neonatal herpes simplex can often be prevented if the infant is delivered by caesarean section within four hours of amniotic membranes rupture (Nahmias, Josey, Naib, Freeman, Fernandez & Wheeler, 1971).

Other Infections: HIV (AIDS Virus)

Since the original description of TORCH infections, other agents have been linked to congenital infections which do not share the distinctive characteristics of the TORCH infections. One of these infections is the HIV virus (human immunodeficiency virus) which attacks the immune system and can lead to AIDS (acquired immune deficiency syndrome). The majority of infants or children under age 13 who have AIDS acquired the HIV virus from their infected mother. This can occur one of the following ways: (1) during gestation where the virus crosses the placenta, (2) during delivery by contact with maternal blood, or (3) by breast feeding. (Other less common means of transmission in children under 13 is associated with contaminated blood in a blood transfusion and child abuse (Caldwell & Rogers, 1991). Adolescents typically acquire the infection through sexual intercourse with an infected person or through contaminated needles typically used for illegal drugs. Since the blood is now screened, a very small percent received the infection through contaminated blood.

Several strains of the HIV virus have been found (HIV-1, HIV-2) (Levy, 1990). A child may be HIV-positive (i.e. have HIV), but not develop symptoms for years. The child may then develop the AIDS syndrome in which symptoms of an injured immune system arise. Infants and young children with AIDS have variable and non specific symptoms such as: failure to thrive, developmental delay, enlarged liver and spleen, chronic diarrhea, lymphadenopathy, upper respiratory infection, ear infection, thrush and recurrent pneumonia (Anderson, Bale, Blackman & Murph, 1986).

Infants and children with AIDS do not usually have the early opportunistic infections and cancers which adults with AIDS acquire. Instead, infants and children develop secondary infections and recurrent bacterial infections which can lead to sepsis and meningitis. The most common secondary infections in children include Pneumocystis carinii pneumonia (PCP) and Lymphocytic interstitial pneumonitis (LIP) (Caldwell & Rogers, 1991). Infections such as cytomegalovirus and toxoplasmosis may occur in the infant or child with AIDS and adversely affect various organ systems (Levin, Zeichner, Duker,

Starr, Augsburger, Kronwitch, 1989; Mitchell, Erlich, Mastrucci, Huto, Parks, Scott, 1990).

Children with congenital AIDS are also at risk of acquiring infections which immunizations are supposed to provide protection. This is because a child with AIDS is unable to mount proper antibody response to immunizations. Therefore, immunizations do not afford any protection from the infections, and a child could die of measles, for example (Rubinstein, 1989).

Children with AIDS often have central nervous system involvement. Some have microcephaly, progressive encephalopathy, progressive dementia and severe spasticity (Lyman, Dress, Dore, Rashbaum, Rubinstein & Soeiro, 1990; Rubinstein, 1989). Developmental problems such as mental retardation, developmental delays, cognitive deterioration and other neurological impairments and motor abnormalities have been found in some children with AIDS typically within the first five years of life (Diamond, 1989).

Individuals who acquired AIDS in later childhood or early adulthood have some differing symptoms and secondary diseases. Symptoms usually reflect the injured immune system and may include: fatigue, weight loss, intermittent fever, malaise, lethargy, chronic diarrhea, enlarged lymph nodes, dry cough, thrush and a tendency to bruise easily. Neurologic involvement may occur as well with seizures, hallucinations and progressive dementia (Berkow, 1987).

Adolescents and young adults with AIDS acquire opportunistic infections. These are infections which are not a threat to a healthy immune system, but can be fatal to a person with AIDS due to their impaired immune system. Cytomegalovirus and toxoplasmosis are two opportunistic infections which when acquired after birth in a person with AIDS, may have devastating results. Two other acquired diseases include pneumocystis carinii pneumonia and Kaposi sarcoma which may be fatal (Berkow, 1987).

Some children and adults develop AIDS Related Complex (ARC). In this condition, many of the same symptoms are present as with those with AIDS, but the individual does not usually have the typical opportunistic infections or Kaposi sarcoma.

Whether this condition always progresses at some point to full blown AIDS, has not been conclusively determined.

Risk of transmission of the HIV virus is extremely low. Transmission typically occurs from sexual encounters with infected individuals, IV drug use with a contaminated needle or congenital transmission. AIDS can not be transmitted by casual contact. One study examining preschool children with HIV found no transmission of the virus to their household contacts when sharing beds, toilets, utensils, toothbrushes occurred or by hugging or kissing (Rogers, White & Sanders, 1990). Other studies have supported the lack of transmission by casual contact (Berkow, 1987).

Visual and Auditory Impairments

Individuals with AIDS are at risk of having visual and auditory impairments. Several visual impairments have been found (often in conjunction with secondary infections such as those found in the STORCH mnemonic and meningitis). Some eye abnormalities

include chorioretinitis, CMV retinitis, ocular motor nerve palsy, cortical visual impairment, gaze palsy and visual field defect (Mansour, 1990). These conditions may result in mild to severe visual impairment, including blindness. The individual with AIDS is also at risk of having a hearing impairment due to general neurologic impairments and acquisition of such infections as meningitis.

Other Teratogens

Other teratogenic agents exist which may adversely affect the fetus and cause impairment. These other teratogens which can effect vision and hearing include medication and illegal drugs taken by the mother during pregnancy and alcohol.

Medications/Drug Abuse

Prescription medications or illegal drugs which have been taken during pregnancy may affect the fetus in several different ways. One way is that the drug may directly affect the embryo which results in birth defects, toxic effects or death to the embryo. Another possibility is that the drug may interfere with the efficient functioning of the placenta. A decrease in the placenta's ability to exchange nutrients and oxygen with the fetus may occur. The drug could also upset the biochemical balance of the mother, adversely affecting the fetus (Bekow, 1987).

Multiple birth defects may occur from some medications and illegal drugs. However, most of them do not cause damage to the eyes or ears. Of those that do, the antibiotics have been especially linked to visual and auditory impairments. The tetracyclines, for example, have been considered a possible cause of congenital cataracts. Other antibiotics such as streptomycin, gentamycin, and karamycin may damage the fetal labrythine and result in a hearing loss (Berkow, 1987). Developing deaf-blindness from this cause is rare.

Fetal Alcohol Syndrome (FAS)

Alcohol consumption during pregnancy places the fetus at risk of being born with multiple abnormalities. The combined effects of maternal (and possible paternal) alcohol consumption on the infant/child has been referred to as Fetal Alcohol Syndrome (FAS).

The characteristics of FAS depend upon the timing and amount of alcohol consumption. Some of the major characteristics include prenatal and postnatal growth deficiency, delay of gross and fine motor development, congenital malformation and microcephaly. Additionally, FAS is the most common cause of mental retardation. Some less frequent abnormalities resulting from FAS include: joint abnormalities, cardiovascular defects and failure to thrive (Berkow, 1987; Blackman, 1990). Behavior problems and hyperactivity may be present.

Visual and Auditory Impairments

Both vision and hearing impairments have been found in FAS. Congenital optic nerve hypoplasia has been described as occurring as well as myopia. Both conductive and sensorineural hearing disorders are more frequent in children with this syndrome.

Outcomes

As the child grows older, he may continue with cognitive and behavioral difficulties. An unstable home environment may attribute to these problems continuing.

Possible Effects of Teratogens: Hydrocephaly and Microcephaly

Hydrocephaly and microcephaly may occur due to prenatal exposure to teratogenic agents (such as CMV and toxoplasmosis) or from other causes. Both may cause brain damage which can severely affect the child.

In hydrocephaly, there is an excessive accumulation of cerebral spinal fluid in the ventricles (small cavities) in the brain. This is typically due to a blockage of the flow of spinal fluid which circulates through the ventricles, spinal column and around the brain. A blockage of the cerebral spinal fluid results in an accumulation of the fluid in the ventricles that gives the infant or very young child an abnormally large head. The excess fluid causes the ventricles to distend in the infant, toddler, child or youth and results in compressing parts of the brain. (This occurs whether or not the head enlarges.) The compressing of the brain can damage the brain and may result in spastic paralysis of the legs, seizures, or mental retardation (Bleck, 1982).

Microcephaly is an abnormally small head due to impaired brain growth and development. There are many conditions which cause microcephaly, some of which have been described earlier in this monograph. Isolated microcephaly can also occur which may be familial. Microcephaly is often associated with varying degrees of cognitive impairment and may be associated with other signs of dysfunction such as cerebral palsy, and seizures.

Visual and Auditory Impairments

Children with hydrocephalus are at risk of developing visual impairments such as optic atrophy. Hearing impairments are more rare. Many of the conditions that cause microcephaly are associated with vision and hearing problems.

Outcomes

In hydrocephalus, a permanent drainage system is surgically put in place. This drainage system is a long small tube known as a shunt that is placed in a ventricle in the brain and usually ends in the abdominal cavity. The shunt is able to drain off excess cerebral spinal fluid from the brain to another part of the body. This prevents pressure from building up on the brain and will prevent further damage from occurring.

In microcephaly, nothing is usually specifically done for the size of the brain, but the symptoms are treated.

Visual and hearing impairments usually continue unchanged.

General Outcomes of Teratogens

The outcome for the infant who has been exposed to teratogens varies greatly. Those infants who have been exposed to STORCH congenital infections have a higher risk of visual and auditory impairments as opposed to some of the other types of teratogens.

Prematurity and Small for Gestational Age

Newborns who are premature or small for gestational age are at risk of having multiple impairments and complications. The immature or developing visual and auditory system may be negatively impacted in both premature infants and infants born small for gestational age. The difference between these two conditions, and their varying affect on vision and hearing will be discussed.

Prematurity

In the past, definitions of prematurity included the timing of the birth and the weight of the infant. Usually an infant who was born before 37 weeks of gestation or who weighed less than 5.5 pounds was considered premature. Several authorities have shifted away from these definitions since it does not differentiate between a baby born after 37 weeks of gestation and weighing less than 5.5 pounds and one who is born early. Both may have very different problems and outcomes. To address this difference, the term premature is being used to refer to infants who are born before the 37 weeks of gestation.

A variety of causes result in infants being born premature. Early contractions result in the birth of a premature infant and this occurs more frequently in adolescent pregnancies (Block, Saltzman & Block, 1981). Women carrying multiple fetuses or who have had many pregnancies and weak cervical muscles are at greater risk of having premature infants (Batshaw & Perret, 1986). Prematurity also occurs more often in women who develop an infection during the third trimester or who have taken such illegal drugs as cocaine. Women who have such chronic illness as diabetes are also more likely to have a premature child (Niswander & Gordon, 1982). Most frequently the cause is unknown.

Multiple problems and complications may be present for an infant who is premature. Most problems of prematurity are due to immature functioning of organ systems. Often the lungs, heart, central nervous system, GI tract, and kidneys are affected with metabolic problems. The lungs are at risk of developing respiratory distress syndrome (formerly known as hyaline membrane disease) in which the small air sacs (alveoli) in the lung collapse resulting in inadequate air exchange. A heart condition known as patent ductus arteriosus may occur in which a blood vessel (vascular connection between the aorta and pulmonary artery known as ductus artrioses) which usually closes at birth remains open. This may result in heart failure. An immature central nervous system may result in apnea (periods of not breathing) and an increase incidence of sudden infant death. Also, an immature central nervous system may result in inadequate sucking and swallowing reflexes which leads to poor nutrition. If the kidney is not completely functioning, it may be unable to adequately excrete unneeded acids without medication.

Metabolic problems may also be present in prematurity. The infant is at risk of being hypo or hyperglycemic (too little or too much sugar) as well as having

hyperbilirubinemia (too much bilirubin). Typically the infant can not adequately regulate his/her own temperature and is more at risk of developing infections (Yu, 1987).

Vision and Auditory Impairments

Greater incidences of visual impairments are present in infants who are premature. In one regional survey, approximately 4.3% of the premature infants had serious visual defects as opposed to 1.5 per 1000 children in the community (Alberman, Venson, & Evans, 1982).

The visual system is incomplete at birth, with the peripheral retina of each eye being incompletely developed. Infants who are born prematurely may have an interruption in the normal development of the peripheral retina, and vascular abnormalities can occur (Biglan, Van Hasselt, & Simon, 1988). The most common cause of blindness associated with prematurity is retinopathy of prematurity. In this condition, there is an abnormal growth of blood vessels which occur in the immature retina. The effects of this may range from no impairments with spontaneous regression of these abnormal blood vessels, to possible retinal detachment and blindness. Glaucoma may also occur with the retinopathy of prematurity.

Other visual conditions associated with prematurity are optic atrophy, squints, refractive errors and cataracts of prematurity. Optic atrophy is often associated with severe cerebral palsy. Squints and refractive errors may be related to retinopathy of prematurity (Yu & Wood, 1987). Cataracts of prematurity present as cloudy areas on the lens of the newborn's eyes which usually disappear after a few weeks (Martyn & Di George, 1987).

There is a greater incidence of serious hearing impairments in infants born prematurely. Decades ago the incidence as reported as high as 9%, but more recently has decreased to closer to 2% (Yu, 1987).

Several causes may be responsible for the hearing impairments. In prematurity, the underdeveloped blood vessels of the inner ear can easily hemorrhage (bleed), which would decrease the blood supply to the Organ of Corti. Toxic and irreversible effects can occur on the cells in the area (Yeates, 1986). Conditions that are associated with premature infants have been associated as attributing to sensorineural deafness. This includes hypoxia and hyperbilirubinemia. Incubator noise has been debated as contributing to sensorineural deafness. However, most authorities have ruled this out (Morgan, 1987).

If the birth is traumatic, the temporal bone may break which result in bleeding in to the inner ear or brainstem that may interfere with hearing (Morgan, 1987).

Small for Gestational Age

The term small for gestational age (SGA) (also termed intrauterine growth retardation) is used to refer to any infant below the 10th percentile for gestational age. The infant is considered small for gestational age when meeting this criterion regardless if the infant was premature, full-term or post-term (Berkow, 1987). There are also several factors that may result in a baby being small for gestational age. One causative factor may be inade-

quate nutritional intake during pregnancy. Chronic maternal illnesses, congenital infections and maternal drug use have also been associated with an infant being born small for gestational age. The pregnant woman, who is over forty years of age, has an increase incidence of giving birth to an SGA infant. This may be due to inadequate blood supply to the placenta resulting in an unsatisfactory nutritional supply to the fetus (Scott & Jordan, 1972). Genetic and inherited factors may also result in this condition as well.

Infants who are small-for gestational age typically exhibit the same developmental delay as the same aged normal sized infants. If the SGA infant is also born premature, those described conditions resulting from immature systems may be present. The SGA infant is attributed as having a greater incidence of major congenital abnormalities (Blackman, 1990) (although a few researchers disagree with this conclusion, but results are based on small sample size (Grogaard, Linstrom, Parker Colley & Stahlman, 1990). Often the SGA infant does not catch up in terms of size as a premature infant would, but may remain small and underweight throughout his or her life (Blackman, 1990; Starfield, Shapiro, and McCormick, 1982). A higher incidence of developmental disabilities, learning disorders, attention deficit disorders, mental retardation and cerebral palsy has been found (Allen, 1984).

Vision and Hearing Impairments

Infants who are born SGA are at greater risk of having visual and auditory impairments. Since infants who are SGA may also be premature, they are at risk of having the visual and auditory impairments described above. Additionally, the infant may be SGA due to maternal infection or drug abuse. As discussed earlier, the STORCH syndromes and drug abuse can be the cause of several types of visual and auditory impairments. The reader is referred to the previous chapter for more information regarding these.

Outcomes of Prematurity and Small for Gestational Age

Infants who are born prematurely or SGA are at risk for vision and hearing impairments. However, due to the multiple problems, associated conditions and variety of impairments, one single cause cannot often be conclusively identified as the etiology. Outcomes would depend upon the specific condition and degree of impairment.

Chapter 7

Adventitious Conditions

Development of concomitant visual and auditory impairment may be acquired after birth from accidents, child abuse, tumors, stroke, and post-natal infections. Accidents and child abuse may cause direct trauma to the eye and ear, affecting the external or internal mechanism. Accidents and child abuse may also result in severe head trauma or asphyxia which results in brain damage. This in turn may cause impairment in the visual and auditory processes. Tumors may arise in the eye and ear or along the pathways leading to the brain or in the visual and auditory cortex. A stroke may also occur affecting vision and hearing. Post-natal infections of meningitis and encephalitis also may occur causing vision and hearing impairments.

Direct Trauma to the Eye and Ear

Accidents or child abuse may cause direct damage to the eye and ear. The range of impairments varies greatly as to the type of damage caused by the accident. Injury directly damaging the eye or ear may be minor or severe. An object may puncture the eye or the retina may detach from the accident. Cataracts and glaucoma may also occur from the trauma as well. Damage to the retina may result in scotomas (blind spots in the field of vision). Decreases in visual acuity and blindness are possible.

In battered baby syndrome, hemorrhages (bleeding) in the eye, often around the macula and optic nerve may occur in both eyes. Damage to the retina in this syndrome may cause blindness (Buncic, 1987). Hearing loss may occur from a displaced ossicular chain, perforation of the eardrum, or temporal bone fracture from a severe blow to the ear or head.

Severe Head Injury

Severe head injury results in injury to the brain and may cause both vision and hearing impairments. This occurs when the areas of the brain, which interpret visual and auditory input or their connecting pathways are damaged. The type of accident may be classified as an open or closed head injury. In an open injury, the skull has been penetrated or crushed. Examples of an open injury would include a gunshot wound to the head or a heavy object falling on the head. In closed head injury, the skull remains intact and the damage occurs from movement of the brain against the skull. This type of injury more commonly results in visual and auditory impairments. Most car accidents and child abuse (which involves severe shaking of the infant or striking the infant/child's head), results in closed head injury.

To understand the mechanics of closed head injury, the reader must be aware that the brain is "floating" or surrounded by a fluid known as cerebral spinal fluid. The brain is then protected by the skull. When a car accident, bicycle accident, fall, or violent shaking occur, the brain is displaced forward, hitting against the interior of the skull (this site of

damage is known as the coup). The brain then bounces back and hits the opposite side of the skull. (This area of damage is known as the contra-coup). The brain then shifts back and forth hitting these two areas of the brain against the skull several times, resulting in damage at the two sites. However, more diffuse brain damage occurs as the brain rotates as the impact takes place. Damage also occurs as nerve pathways throughout the brain break. The rough bones at the skull base cause further brain damage to that area. Often there will be swelling (edema) and bleeding (contusion, hematoma or hemorrhage) in the brain which can increase the pressure on the brain, causing pressure on the blood vessels and decreasing their flow. When that occurs, further tissue death occurs due to the inadequate blood flow. Several problems may occur as a result of head trauma (see Table 4). Included in this is frequent vision and hearing impairments.

Traumatic Head Injury

Disorders in the cognitive area may include:

1. attention and concentration problems
2. memory problems, especially new learning
3. perceptual problems, including spatial disorientation
4. communication and language disabilities
5. poor judgment; inability to make decisions; decreased abstraction; illogical thoughts
6. confabulation; distractability; perplexity
7. decreased learning abilities; deficits in processing information
8. rigidity and inflexibility; perseveration; inability to adjust to changing contingencies

Disorders in the psychomotor and sensory/physical areas may include:

9. Reduced motor speed
10. visual, auditory, and tactile deficits
11. quadriplegia; hemiplegia; spasticity; ataxia; rigidity states; tremors
12. reduced eye-hand coordination
13. spatial disorientation
14. increased risk seizures

Disorders in the social/affective areas may include:

15. lack of goal-directed behavior, initiative and planning
16. poor self-image; reduced self-worth
17. disinhibition; impulsivity
18. apathy; social withdrawal
19. loss of concern for others, self centeredness
20. indifference; poor grooming; denial of disability
21. family stress; disturbances in sexual behavior
22. aggressive behavior; inappropriate childlike behavior

Table 4 Problems occurring in head injury

Visual and Auditory Impairments

Several types of visual impairments have been associated with head injury (see Table 5).

The most common eye problems are eye muscles palsies. These are paralysis or impairments of the movements of the eye due to impairment of the eye muscles or the nerve innervating them. These eye muscle palsies may result in diplopia (double vision) from strabismus (deviation of one or both eyes in different directions) or the lack of use of one eye (the suppression of one image by the brain). Often these extraocular palsies are reversible (Rosenthal, Griffith, Bond, & Miller, 1984).

Those eye abnormalities that cause the greatest functional impact include perceptual disorders, tracking disorders and central or peripheral types of blindness (Rosenthal, Griffith, Bond, & Miller, 1984). Perceptual disorders may result in difficulties in recognizing consistencies in shapes and color, as well as difficulty in perceiving objects against a background.

Tracking disorders may result in an inability to follow an object through space. Difficulties with vertical, horizontal and diagonal tracking may occur.

Abnormalities of field of vision may be present, especially when the optic tracts have been affected. There may be a loss of half of the right or left field of vision for one or both eyes. (hemianopsia). Loss of quadrants may also occur.

Cortical visual impairments (cortical blindness) may result in which injury occurs to the portion of the brain which receives input from the eyes (occipital lobe). If the blood vessels serving this area have been compressed, cortical visual impairment may also result. Cortical visual impairment may involve a total lack of vision or inconsistent vision. If there is sudden injury, the loss is typically irreversible. Gradual injury due to brain edema (swelling) may not lead to tissue death so vision may return after a few days or months (Rosenthal, Griffith, Bond, & Miller, 1984).

| Common Visual Impairments Found in Traumatic Head Injury | |
|---|---|
| Injury to the eye and/or its muscles | Neurological impairments affecting visual pathways and visual cortex |
| Decreased acuity | Field cuts |
| Blindness | Visual perceptual deficits |
| Scotoma | Diplopia |
| Diplopia | Nystagmus |
| | Gaze palsies |
| | Tracking disorders |
| | Decreased acuity |
| | Blindness |
| | Global stereopsis impairments |
| | Color vision defects |

Table 5 Visual impairments associated with head injury

Optic nerve atrophy is a wasting away of the optic nerve which may result in visual loss, proportional to the amount of nerve damage. There may be decrease visual acuity to total blindness with a lack of response to direct light.

Other vision problems due to neurological impairment may exist, such as problems in perceiving color (color blindness) to nystagmus (fine rapid involuntary movements of the eye). Impairment due to head injury may result in decreased acuity, blindness and scotomata.

Hearing disorders often occur due to trauma to the ear, nerve pathways or brain. Damage in the middle ear may result from tears in the eardrum or displacement of the three bones in the middle ear. Both of these result in a conductive hearing loss. Damage to the inner ear can also occur, resulting in a sensorineural hearing loss. Although a sensorineural loss occurs frequently, it is typically mild and rarely causes functional impairment (Chamovitz, Chorazy, Hanchett, & Mandella, 1987). Damage to the auditory nerve or portion of the brain which receives and interprets messages are more likely to cause permanent hearing losses (Rosenthal, Griffith, Bond, & Miller, 1984).

Outcomes

Typically there continues to be a progressive return of function within the first five years after the injury. During that time, the individual is learning how to compensate for what has been lost. Structural changes also occur in the brain (such as decrease swelling) which result in improved functioning.

The extent of vision and hearing impairment depend on the type and extent of damage.

Accidents Involving Asphyxia

Asphyxia is a decrease in the amount of oxygen in the body. Some of the causes include choking, carbon monoxide poisoning, drowning, strangulation, electric shock, crushing chest injuries and drugs.

Infants, toddlers, children and youth who have an accident involving asphyxia may have a wide range of symptoms. Cerebral palsy, mental retardation and seizures may occur. Accidents involving asphyxia places the child at risk of having visual and hearing impairments.

Tumors

Tumors may arise after birth which adversely affects the normal functioning of the eye and ear. For the eye, losses in visual acuity and losses in fields of vision may occur depending upon the location and type of the tumor. Consequences are more serious when the tumor is retinoblastoma. Retinoblastoma is a cancerous (malignant) tumor which develops from an immature retina. It may be inherited or develop as a mutation. This tumor can result in loss of life unless early surgical intervention occurs.

Tumors may also result in conductive or sensorineural hearing losses. This depends upon the location and severity of the tumor.

Stroke

A stroke (also known as a cerebral vascular accident or CVA) is a disturbance in the blood flow in the brain. This results in a sudden loss of some neurological functioning. The term stroke may include transient, brief strokes (known as transient ischemic attacks (TIA) or a permanent complete stroke (Berkow, 1987; Karanjia & Smigielski, 1988).

A transient stroke (TIA) has an abrupt onset, but resolves itself completely within twenty-four hours. It may be caused by a drop in blood pressure in the arteries of the brain or a temporary occlusion of a cerebral artery (e.g. by a clot). Symptoms of TIA depend upon the area of the brain affected. Motor weakness and numbness may occur. If the area of the brain that comprehends speech is affected, he/she may have difficulty understanding language and may appear confused. Inability to initiate speech or slurred speech may also occur when the motor area for speech is affected. Vertigo (the sensation of moving around in space or having objects move about the person) may be present as well as difficulty walking. A headache may also occur.

In a complete stroke, deficits continue past twenty-four hours. This indicates that death (infarction) of brain tissue has occurred resulting in permanent brain damage. There are several causes of a complete stroke. The major ones include obstruction of a blood vessel (by a clot or foreign substance), long periods of time with low blood pressure, and vascular spasm from a cranial bleed (subarachnoid hemorrhage) or migraine. Some less common causes include leukemia, sickle cell anemia, dehydration and trauma (Karanjai & Smigielski, 1988).

As with TIAs, the symptoms of a complete stroke depend upon the location, size and type of stroke. There may be disturbances in motor function (such as paralysis or weakness) and minor degrees of sensory loss (such as an inability to discriminate objects placed in the hand with the eyes closed). In some cases, there may be disturbances in perceiving body image where a person denies that a body limb belongs to him (anosognosia). Disturbances in language may occur where the youth may be unable to comprehend language or be unable to speak. Difficulties in recalling names of common items or problems in articulation may occur as well. Disturbances in consciousness and behavior can occur. Memory loss, apathy, lack of motivation, mental retardation and seizure disorders may also be present as well as other symptoms.

Visual and Auditory Impairments

In both TIA and complete stroke, visual and auditory impairments may occur. In TIAs, a sudden temporary visual loss (amaurosis fugax) may occur which appears as a curtain descending over one eye. With both TIA and stroke, visual loss may occur in one or both eyes. Field defects, diplopia (double vision), and disturbances in ocular motility can occur. Both eyes may be deviated toward the side of the brain where the injury is located,

with an inability to gaze to the opposite side (Karranjia & Smigielski, 1988). Although hearing impairments may occur, they are not as common.

Outcomes

Progressive improvement in functioning is present up until 3 to 5 years after the injury.

Infections

Two of the most common infections resulting in vision and hearing impairments are meningitis and encephalitis. These two infection will be discussed in terms of their general characteristics and specific visual and auditory impairments.

Meningitis

Meningitis is an infection resulting in inflammation of the membranes (meninges) which cover the brain and spinal cord. Impairments acquired from this infection range from no impairments to possible death within hours. Prognosis depends upon what has caused the infection.

There are several different organisms which can cause meningitis. If the cause is a virus (such as from mumps), the symptoms are usually mild and pose no threat to healthy children. However if the meningitis is caused by a bacteria (such as streptococcus pneumoniae or Hemophilus influenza type B (HIB) or meningococcus), severe damage and/or death can occur. (Less often fungi, parasites, tuberculosis and spirochetes may cause meningitis with varying prognosis.)

For children and adults, typical symptoms include a stiff neck, sore throat, vomiting, headache, fever, lethargy and sensitivity to bright light. A nerve that controls the lateral movement of the eye may be affected. A progression through irritability, lethargy, drowsiness, stupor and coma is possible. For infants, symptoms vary, although fever, vomiting, seizures, high pitched cry and bulging fontanel (soft spots on top of the head) may be present.

Some affected infants or children may be left with brain damage, mental retardation, seizures and motor impairment.

Visual and Auditory Impairments

Visual impairments, such as cortical visual impairments, may occur when neurological damage is present from a bacterial infection.

Hearing loss occurs in approximately 10% of those who survive bacterial meningitis and is typically permanent. Hearing loss may be present in one or both ears. The loss is usually due to damage to the eighth cranial nerve which send impulses from the ear to the brain, or damage to the nerve endings in the inner ear. Deafness may occur, but is more likely seen in individuals who have other neurological deficits (Anderson, Bale, Blackman, & Murph, 1986).

Outcomes

Antibiotics and sometimes corticosteroids are used to treat bacterial meningitis. Immunization with HIB vaccine in early infancy should protect more children from developing HIB meningitis. For viral meningitis, symptoms will be controlled by medications, but the infection usually subsides on its own.

Some individuals who do acquire meningitis, are left with long term conditions such as deaf-blindness.

Encephalitis

Encephalitis is an inflammation of the brain. Encephalitis may result as a primary manifestation of a virus, or occur as a complication of another infection such as measles, chicken pox or rubella. In some cases the encephalitis is carried by mosquito and tick to infected individuals.

Symptoms of this infection vary. At times there may be a decrease in alertness. Other times more severe symptoms may be present such as seizures, paralysis of an arm or leg, or coma.

Depending upon the type of virus, the condition is usually mild with no lasting disabilities. However permanent brain damage can occur with mental retardation or learning disabilities, especially in infants. In a few cases children may die. Other problems that can occur include neurological impairments, seizures, behavior changes, mental retardation and motor deficits.

Visual and Auditory Impairments

When brain damage results from encephalitis, blindness, visual impairments and sensorineural loss may result.

Outcomes

Prognosis depends upon the type of virus, but even very ill individuals may completely recover. Permanent damage is more likely to occur in infants.

APPENDIX

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