

Introduction :-

This booklet describes the causes, syndromes and genetic conditions that may cause deafblindness. Knowledge of conditions that cause deafblindness provides teachers with important clues to students health status, physical stability, potential development as well as understanding of symptoms and the progression of students conditions. Educators, professionals and medical practitioners who work with deafblind children will find this information useful. This booklet is a compilation of materials / literature which is already available.

This booklet is a small contribution from Sense International (India) to deafblind field.

For easy accessibility of information for people with sensory impairments this booklet is also available in Braille and Tape on request.

Causes and Syndromes Leading to Deafblindness

What is deafblindness?

Deafblindness is a combination of visual and hearing impairments. The degree of deafness or blindness varies in individuals but the important factor is that this combination (of both sensory impairment) creates unique problems in communication, mobility and their ability to learn about the world. It may seem that deafblindness refers to a total inability to see or hear but many deafblind people have remaining vision or hearing that they can learn to use better. A deafblind child cannot be thought of as blind and also deaf, nor as deaf and also blind. She is deafblind and deafblindness is a unique disability; it has its own concepts and terminology, its own methods of assessment and education, and its own modes of communication which distinguishes it from blindness and deafness understood separately.

It is not a medical concept, the ophthalmologist and the audiologist observe within their respective spheres, and the strictly medical literature never refers to deafblindness. It's a developmental concept, and without it the nature of the disability cannot be understood.

Characteristics of Deafblindness :-

- Have a distorted perception of the world.
- Appear to be withdrawn and isolated.
- Lack the ability to communicate with his or her environment in a meaningful way.
- Lack curiosity and be deprived of many of the basic motivations.
- Are defensive to being touched.
- Have extreme difficulty in establishing and maintaining interpersonal relationship with others.
- Lack the ability to anticipate future events or the results of their actions.
- Have feeding difficulties and/or unusual sleep patterns.
- Have medical problems that lead to serious developmental delays.
- Exhibit frustration, discipline problems, and delays in social, emotional, and cognitive development because of the inability to communicate.

Why are certain children born with “Special Needs”?

The main cause can be divided as;

The **PRENATAL STAGE** - the period before a child is born.

The **NEONATAL STAGE** - the time around baby's birth

The **POSTNATAL STAGE** - the period of baby's infancy, i.e. One week from birth to three years.

Besides this, there are many genetic syndromes.

PRENATAL CAUSES :-

- A mother who is under 18 years or over 35 years may be at higher risk for having a child with special needs
- Mother could have had German measles, tuberculosis or other infections (TORCH)
- Mother could have suffered from very high fever when she was carrying baby
- Mother could have got insufficient nutrition when she was carrying baby
- Mother could have taken X-rays in the early stages of pregnancy
- Mother could have suffered from chronic ailments like hypertension, diabetes etc.

- Rh incompatibility in mother and baby's blood
- Mother could have taken the wrong drugs or excessive drugs
- Physical and/or emotional trauma during pregnancy
- Attempts for abortions
- Mother had convulsions (fits) during pregnancy

NEONATAL CAUSES :-

- Child could have been born prematurely, i.e. before 37 weeks or 259 days or post-maturely, i.e. after 42 weeks
- Mother had a prolonged labour, or a very difficult time while giving birth
- The umbilical cord could have been tightly wrapped around child's neck
- Child could have had bleeding in brain due to injury during birth
- Birth cry could be delayed and child could have taken time to breathe
- Child was deprived of oxygen during or immediately following birth
- Skilled people were not available during delivery to take care.

POST-NATAL CAUSES :-

- Child could have high fever or convulsions (fits) which could have damaged child's brain
- Child could have received a head injury due to fall
- Child could have had insufficient nutrition, which could lead to malnutrition & hamper child's development
- Child could have had a metabolic disorder, which could make the child unable to assimilate fat, protein, or carbohydrates.
- Child could have suffered a brain fever which could have lead to brain damage
- Early childhood jaundice or jaundice at the time of birth
- Child could have suffered or had been improperly treated for tuberculosis.
- Brain infections like Meningitis or Encephalitis

Some common infections that leads to Deafblindness :-

Cytomegalovirus (CMV) :-

CMV is a common virus that infects most people at some time during their lives but rarely causes obvious illness. It is a member of the herpes virus family. Manifestations of this disease may be minimal (e.g., rash and fever) or severe (e.g., microcephaly, mental retardation, and profound sensorineural hearing loss). In most cases, there is no treatment. An effective vaccine has not yet been developed.

Meningitis :-

Meningitis is a condition in which inflammation of the lining (meninges) of the brain and spinal cord occurs due to a bacterial or viral infection. Meningitis is usually bacterial or viral, but in rare cases it can be caused by fungus. In its bacterial form the condition is life threatening. The viral form is usually less severe.

The central nervous system may be affected, and some loss of sight or changes in vision may occur (e.g., strabismus, decreased acuity, cortical visual impairment, and photophobia due to changes in the sizes of the pupils.) Babies born prematurely or with low birth weight have a higher risk of all forms of neonatal meningitis. Prolonged labour after rupture of membranes also increases the risk of neonatal meningitis.

Toxoplasmosis :-

Toxoplasmosis is caused by a parasite called *Toxoplasma gondii*. It forms cysts, which are passed in the feces of its primary or main host, the cat. Cats alone are not the only reason for the incidence of this condition. Undercooked meat and the increasing consumption of unpasteurized goat's milk are two other potential causes.

Luckily the infection that can result is usually very mild. It can be a glandular-like illness or produce symptoms of a mild flu. However, in a pregnant woman the infection, although not a risk to her, can cause congenic abnormality in the unborn child. Lesions of the central nervous system occur, which lead to blindness brain defects, and other serious conditions.

Genetic Causes and Syndromes :-

There are more than 50 syndromes which has associated condition of deafblindness. The conditions described in following syndromes may cause multiple disabilities, including deafblindness. Some of them are;

Congenital Rubella Syndrome:-

Rubella, or German measles, is a mild disease caused by a virus. Usually rubella causes a slight fever which lasts for about 24 hours, and a rash on the face and neck that lasts two or three days. Most people recover quickly and completely from rubella. However, the greatest danger from rubella is not to children or adults, but to unborn babies. If a woman gets rubella in the early months (first trimester) of her pregnancy, her chance of giving birth to a deformed baby may be as high as 80%. These babies may be born deaf or blind or deafblind. They may have damaged hearts or unusually small brains. Many are mentally retarded. Miscarriages are also common among women who get rubella while they are pregnant

Rubella can be prevented by a rubella vaccine, which is usually given to children at 12 to 15 months as part of the scheduled Measles-Mumps-Rubella (MMR) immunization. A second dose of MMR is generally given at 4 to 6 years of age, but should be given no later than 11 to 12 years of age. The rubella vaccine should not be given to pregnant women or to a woman who may become pregnant within 3 months of receiving the vaccine.

Charge Syndromes :-

CHARGE Association is a multi – featured disorder characterized by a unique combination of diverse abnormalities. The name “CHARGE” is made up from the initial letters of some of the most common features seen in this condition.

Coloboma is cleft or failure of the eyeball to close resulting in abnormalities of the retina or optic nerve. This may result in significant loss of vision, defects in visual acuity resulting in near or farsightedness, and oversensitivity to light.

Heart defects include Tetralogy of Fallot, the most frequent type of heart defect reported in the CHARGE association. The tetralogy is “an anatomic abnormality” with severe or total right ventricular outflow tract obstruction and a ventricular septal defect allowing right ventricular unoxygenated blood to bypass the pulmonary arteries and enter the aorta directly.

Atresia choanae is the blocking (atresia) of the airways (choanae) from the back of the nose to the throat that would allow breathing through the nose.

Retardation of growth and development is usually due to heart problems, nutritional problems, or growth hormone deficiency. The developmental delay often is associated with sensory deficits (vision and hearing loss). Some children with CHARGE will be mentally retarded.

Genitourinary problems, in boys include genital hyperplasia and possibly undescended testes. The girls may have small labia. Reflux of the urinary tract or kidneys is common.

Ear abnormalities include a common finding of unusually shaped ears (short and wide with very little or no earlobe). Hearing loss, conductive and/or nerve, ranges from mild to deafness. Evidence exists of other anomalies associated with this condition in addition to those above:

- abnormal tongue size
- cleft lip and /or palate
- facial palsy
- renal abnormalities
- malformations of the larynx
- atresia of the oesophagus
- skeletal abnormalities

Although some cases appear to be influenced by heredity, environmental factors have not been ruled out. There is no laboratory test that can diagnose CHARGE Syndrome. Usually the diagnosis is made because of the presence of a number of these typically unrelated anomalies. At least four of these abnormalities should be present if a child is to be diagnosed as having CHARGE Association. In any child suspected to have CHARGE, cardiorespiratory, ophthalmological, and audiological evaluations may be performed as well as abdominal ultrasound and chromosome evaluation. The exact cause of CHARGE Association is still unknown but research suggests a variety of catalysts may induce the irregularities present in the CHARGE sequence of anomalies.

Usher Syndrome :-

Usher syndrome is a genetic disorder that is characterized by hearing impairment as well as an eye disease called Retinitis Pigmentosa (RP) in which vision degenerates (gets worse) over time. Some people with Usher syndrome also have balance problems. There are three different clinical types of Usher syndrome (US). They are called Usher syndrome type 1 (US1), Usher syndrome type 2 (US2), and Usher syndrome type 3 (US3). Types 1 and 2 are more common than type 3. All types of the syndrome are inherited in the same pattern—as autosomal recessive traits.

Usher Syndrome type 1 (US1) :- People with US1 are profoundly deaf from birth and have severe balance problems. Many of these individuals obtain little or no benefit from hearing aids. Most use sign language as their primary means of communication. Because of the balance problems, children with US1 are slow to sit without support and rarely learn to walk before they are 18 months old. These children usually begin to develop vision problems by the time they are ten. Visual problems most often begin with difficulty seeing at night, but tend to progress rapidly until the individual is completely blind.

Usher Syndrome type 2 (US2) :- People with US2 are born with moderate to severe hearing impairment and normal balance. Although the severity of hearing impairment varies, most of these children perform well in regular classrooms and can benefit from hearing aids. These children most commonly use speech to communicate. Retinitis pigmentosa, which is a degeneration of the retina (the part of the eye that receives images of objects), is characterized by blind spots that begin to appear shortly after the teenage years. The visual problems in US2 tend to progress more slowly than the visual problems in US1. When an individual's vision deteriorates to blindness, his or her ability to read speech from the lips is lost.

Usher Syndrome type 3 (US3) :- Children born with US3 have normal hearing and normal to near-normal balance. Hearing worsens over time. Children develop noticeable hearing problems by their teenage years and usually become deaf by mid to late adulthood. Retinitis pigmentosa in the form of night blindness usually begins sometime during puberty. Blind spots appear by the late teenage years to early adulthood. By mid adulthood, the individual is usually blind.

Norrie's Disease :-

Norrie's Disease typically affects only males, who may be blind and deaf and have cognitive disabilities. Not all children have mental retardation however; in some children the retardation is severe

Down Syndromes :-

Down syndrome is related to a specific type of a chromosome abnormality. Smaller stature along with slower physically and mentally development are typical of the child with Down syndrome. Although some children with Down syndrome are not mentally retarded, most of these children function in the mild to moderate range of mental retardation. Children with Down syndrome may have associated condition of deafblindness.

Turner's Syndrome :-

Only females are affected by this congenital disorder whose major features stem from a missing X chromosome or an alternation in one of the female's two X chromosomes. Those affected are typically of short stature and fail to develop secondary sex characteristics. Hearing loss, Mental retardation, abnormalities of the eye, and heart and kidney disease may be present.

Crouzon's Syndrome :-

Although Crouzon's syndrome is also a type of craniostenosis, it is differentiated from Alpert's syndrome by severe visual impairment and the lack of fused hands or feet. Infants have an abnormal skull shape because of the premature closure of the cranial sutures. Abnormalities of eye are caused by increased cranial pressure. Hearing loss is conductive, with significant middle ear anomalies, although some mixed type hearing losses are possible. Mental retardation may be present, as well as some problems with the spine.

Goldenhar Syndrome :-

A variety of terms have been used to describe this extremely variable disorder. According to medical literature, when malformations primarily involve the jaw, mouth, and ears and, in most cases, affect one side of the body (unilateral), the disorder is often referred to as Hemifacial Microsomia. If abnormalities of the vertebrae and the eyes are also present, the disorder is often called Goldenhar Syndrome. Scanning may identify the condition in certain cases where facial or skeletal anomalies are present. Pre-natal screening and genetic advice may be offered for future pregnancies.

Rett Syndrome :-

Rett syndrome is a complex neurological disorder. It affects mainly girls. Although present at birth, it becomes more evident during the second year. People with Rett syndrome are profoundly and multiply disabled and highly dependent on others for their needs throughout their lives. It is a complex neurological disorder, genetic in origin. A few boys also have the condition. At least one in every 10,000 females born has Rett syndrome. It is believed to be the second most common cause of severe and profound learning disability in girls. Rett syndrome is a progressive neurological disorder in which individuals exhibit reduced muscle tone, autistic-like behaviour, hand movements consisting mainly of wringing and waving, loss of purposeful use of the hands, diminished ability to express feelings, avoidance of eye contact, a lag in brain and head growth, gait abnormalities, and seizures. Hypotonia (loss of muscle tone) is usually the first symptom.

Turner Syndrome :-

Turner syndrome is a disorder caused by the loss of genetic material from one of the sex chromosomes. Turner syndrome affects approximately 1 out of every 2,500 female live births worldwide. It embraces a broad spectrum of features, from major heart defects to minor cosmetic issues. Some individuals with Turner syndrome may have only a few features, while others may have many. Almost all people with Turner syndrome have short stature and loss of ovarian function, but the severity of these problems varies considerably amongst individuals.

Cri du chat :-

Cri du chat syndrome is a group of symptoms that result from missing a piece of chromosome number 5. The syndrome's name is based on the infant's cry, which is high pitched and sounds like a cat. Between 1 in 20,000 and 1 in 50,000 babies are affected. This disease may account for up to 1% of individuals with severe mental retardation. Infants with cri du chat syndrome commonly have a distinctive cat-like cry. They also have an extensive grouping of abnormalities with severe mental retardation being the most important.

Symptoms :-

- high-pitched cry sounds like a cat (for which the syndrome was named)
- low birth weight and slow growth
- small head (microcephaly)
- wide-set eyes (hypertelorism)
- downward slant to the eyes (palpebral fissures)
- small jaw (micrognathia)
- low-set ears (may be malformed)
- skin tags just in front of the ear
- partial webbing or fusing of fingers or toes
- single line in the palm of the hand (simian crease)
- mental retardation
- slow or incomplete development of motor skills

Refsum's Syndromes :-

Recent research has identified the group of enzymes missing in this metabolic disorder. Refsum's syndrome occurs both as an infantile disease causing early death and as a later onset adult condition. As the disease progresses, the body is unable to metabolize, phytanic acid which build up and causes pathological changes in the myelin sheath.

Alport's Syndrome :-

Alport's Syndrome affects either sex and is asymptomatic at first. Kidney disease is often present, with sensorineural deafness and abnormalities of the eye. Hearing loss usually affects both ears but is asymmetrical in degree. This syndromes produces more serious kidney problems in males, leading to a higher mortality rate than with females.

Bardet-Biedl Syndrome :-

Children with Bardet-Biedl syndrome have progressive vision loss, obesity, extra fingers or toes, kidney and liver problems, mental retardation and pituitary abnormalities. Hearing impairment is generally sensorineural and progressive. Poor night vision is often the first complaint related to vision, followed or accompanied by loss of visual acuity.

Hunter's Syndrome :-

Hunter's and Hurler's syndromes are closely associated and result from a deficiency of enzymes that are needed to metabolize mucopolysaccharides. Hurler's syndrome often results in lens opacities, mental deficiency, skeletal abnormalities, and progressive hearing loss. Hunter's syndromes affects males only.

Marfan's Syndrome :-

Marfan's syndrome a disorder of connective tissue, has recently been linked to a single gene on chromosome 15. This disorder is not always associated with mental retardation, yet it does cause skeletal abnormalities, including an inward-curving chest, spider like fingers and toes, and an exceptionally tall, thin body profile.

Neurofibromatosis :-

Neurofibromatosis often shows up as tumors involving the nerves of the skin and internal organs. Any portion of the central nervous system may be involved. Abnormalities of the eye vary and may include ptosis, muscle palsy and atrophy.

Trisomy 13 -15 syndromes :-

An extra chromosome is responsible for trisomy 13-15 syndromes. A significant percentage of children born with this condition die during infancy. Most of those who survive have severe mental retardation and marked physical characteristics. Major abnormalities affect the brain, heart, liver and kidneys. Varying degrees of auditory and visual complications may be present.

Trisomy 18 syndromes :-

Trisomy 18 syndromes is recognizable in the infant, as the head is narrow and elongated, with a bulge at the back of the skull. The chin is extremely small and are the eyes. The ears are often malformed and low set. These children have low birth weight and fail to thrive. Females are three times more likely to experience this syndromes than males.

Wildervanck Syndromes :-

Wildervanck described the first case of what he labeled the cervico-oculo-acoustic dysplasia syndrome. This syndromes has three manifestation : 1) Fused cervical vertebrae 2) an eye movement disorder 3) severe congenital sensorineural or conductive hearing loss. This is an extremely rare syndrome, with only 21 cases reported in the literature worldwide. A syndrome characterized by the combination of congenital deafness, Duane syndrome (eye retraction), and fusion of the neck vertebrae (Klippel-Feil anomaly). Wildervanck syndrome is limited, or almost completely limited, to females. The syndrome is probably due to polygenic inheritance with limitation to females and lethality for males.

Acquired Deafblindness :-

Acquired deafblindness refers to occasions where a person becomes deafblind later in life.

Prevention of Disabilities :-

Pre Natal Stage :-

- Eat a well balanced and nourishing diet supplemented with leafy vegetables, proteins and vitamins
- Ensure weight gain of at least 10 kg.
- Have regular medical check ups during pregnancy
- Take medications only if prescribed by a doctor. Even the normally safe and recommended drugs and medicines may under certain conditions cause serious defects in an unborn child.

- Avoid pregnancies before the age of 18 years and after the age of 35 years consult a doctor before planning pregnancy.
- Provide vaccination against rubella for all women before reaching child bearing age.
- Avoid exposure to illness like measles, mumps etc. especially during the first three months of pregnancy
- Avoid X-rays and exposure to any kind of radiation
- Avoid smoking, chewing tobacco, consuming alcohol and narcotics
- Avoid hard physical work such as carrying heavy loads especially in fields, and other accident prone activities such as walking on slippery ground or climbing stools and chairs.
- Pregnant women should not be subjected to undue emotional stress and strain
- There should be a minimum gap of 3 years between consecutive pregnancies.
- Avoid induced or illegal abortions.
- Delivery must be conducted by trained persons preferably in a hospital where all facilities are available
- Breast feeding by mother immediately after birth will protect the baby from infections.

High Risk Pregnancies :-

- If there is incidence of birth defects in the family
- If there has been difficulty in conceiving or there has been a series of miscarriages
- If the mother has “Rh -ve” blood type
- If the previous pregnancy was multiple pregnancy

High Risk Infants :-

- Premature baby (before 28 weeks), low birth weight babies (less than 2.5 kg), babies born after difficult labour, babies with congenital disease, babies were asphyxiated during birth or who cried delayed (1-3 minutes) may need neonatal intensive care.
- If the baby does not cry immediately after birth (birth cry is considered “absent” after 3 minutes) resituation should be undertaken at once.
- If the baby’s head appears to be abnormally small or large, have it measure and consult a doctor. The approximate head size for a male child at birth is 35 cms. and female child is 34.5 cms.

Post Natal Period :-

- Take the baby to a doctor immediately if ;
 - a. Jaundice persists even after three days of birth (baby appear yellow)
 - b. Baby is blue or even if the tips of the fingers and toes appear blue
 - c. The child has persistent diarrhoea
 - d. The baby has difficulty in breathing or appears to be choking
 - e. Do not allow your child’s temperature to remain above 101⁰F at any time
 - f. If your child gets fits take him to a doctor immediately
 - g. Your child should be immunized against infectious disease like Polio, Measles, Tetanus, Tuberculosis etc.
 - h. Do not allow your child to have too much contact with paint, newsprint ink, lead etc.
 - i. Take precautions against head injury, and other accidents
 - j. Ensure that your child gets a well balanced diet and clean drinking water.

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