

Birth Defects and Childhood Disability Toolkit

Section 3 - Developmental disabilities

Birth Defects Research Foundation, Pune, India

A research NGO working for evidence based policies and advocating for the rights of children with disabilities caused by congenital, developmental and genetic disorders

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3.1 Developmental disabilities: What are they?

Down syndrome

Intellectual disability

Cerebral Palsy

Hearing Impairment

Autism

Vision Impairment

Attention Deficit
Hyperactivity
Disorder

1. What are developmental disabilities?

- Group of disorders causing lifelong disability. The impairment affects movement, emotional, behavioral, cognitive, sensory and motor functioning
- Common causes of childhood disability
- For example: Down syndrome, intellectual disability, cerebral palsy, autism spectrum disorders, attention deficit hyperactivity disorder, congenital vision impairment and blindness, and congenital hearing impairment
- Usually detected early in childhood.

Worldwide prevalence of developmental disabilities among children below 5 years of age – estimated to be **52.9 million**

2. How are developmental disabilities identified ?

Developmental disabilities affect child development.

Development is progress in functioning and acquiring motor, emotional, behavioral, cognitive and sensory skills

(In contrast, **growth** refers to increase in physical size as determined through anthropometric measures)

Typically developing children achieve

Age-specific developmental milestones – i.e. sets of behaviors, skills and abilities at specific ages during infancy and early childhood. (*Check Developmental milestones poster*)

3. What is the difference between developmental delays and developmental disabilities?

Delayed developmental milestones: when a child does not achieve age specific milestones; Caused by factors like poverty, under-nutrition, lack of immunization, lack of nurturing care; may be reversible

Developmental disabilities: irreversible, lifelong disability, frequently accompanied by medical conditions

Rehabilitation,
initiated early (before
3 years of age) can
improve functioning
and quality of life

4. What are the needed public health activities?

- Monitoring early childhood development to identify at risk infants
- Screening for early identification of developmental delays and disabilities
- Early intervention, that is rehabilitation services and long term medical care
- Referral linkages to social welfare services for children with disabilities
- Education, awareness about disability, rights, and routes to accessing available care
- Strengthening perinatal care, ensuring care for small and sick newborns
- Strengthening service delivery through primary health care

Reference Olusanya, B. O. et al (2018). Developmental disabilities among children younger than 5 years in 195 countries and territories, 1990–2016: a systematic analysis for the Global Burden of Disease Study 2016. *The Lancet Global Health*, 6(10), e1100-e1121.; Choo, YY et al (2019). Developmental delay: identification and management at primary care level. *Singapore medical journal*, 60(3), 119. Ansari, H (2021) Magnitude of Developmental Disabilities in India. . In: Kar, A. (eds) Birth Defects in India. Springer, Singapore. https://doi.org/10.1007/978-981-16-1554-2_1

3.2 Early intervention for developmental disabilities

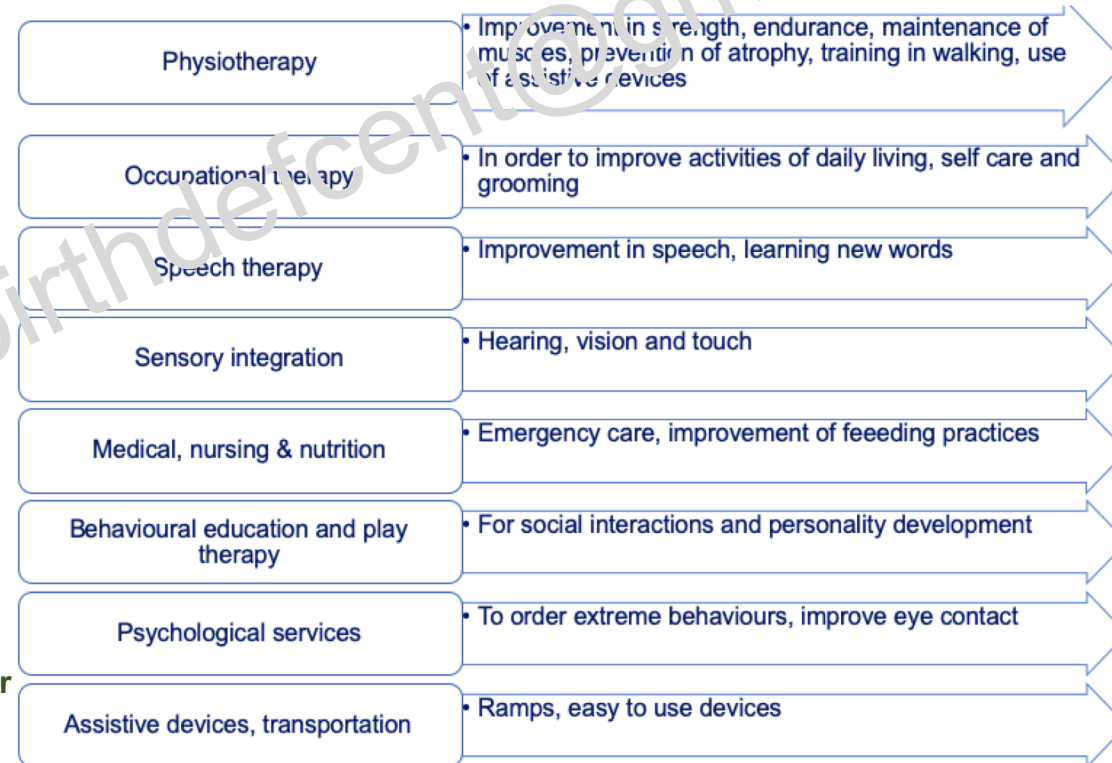
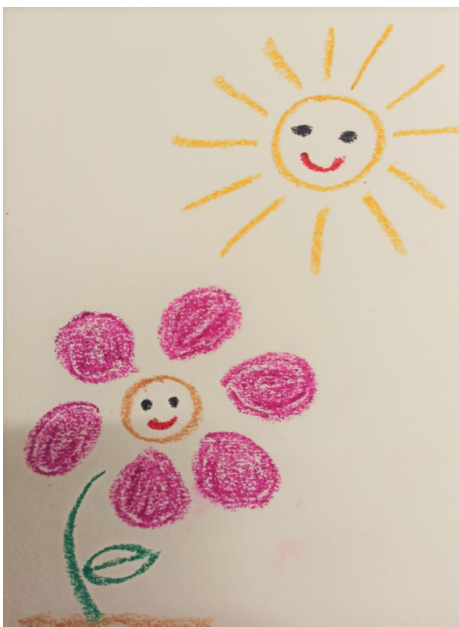
1. What is early intervention?

- Early intervention refers to a set of coordinated services and support systems for infants and young children with developmental disabilities and delays
- Services include therapies, counselling and guidance, better caregiving practices
- Aim is to maximize developmental and health outcomes, improve quality of life & decrease out of pocket expenditures

2. Why is monitoring of child development important?

Monitoring child development is important for early identification and referral for intervention

- ☐ Checklists can assist parents in case of concerns about development of the child
- ☐ Developmental screening by healthcare providers can determine if further evaluation is needed
- ☐ Developmental evaluation by developmental pediatrician/child psychologist to determine the need for specific treatment/early intervention



References: Smythe, T. et al. (2021). Early intervention for children with developmental disabilities in low and middle-income countries—the case for action. *International Health*, 13(3), 222-231. Lipkin, PH et al. (2020). Promoting optimal development: identifying infants and young children with developmental disorders through developmental surveillance and screening. *Pediatrics*, 145(1).

1. What is cerebral palsy? 2. How does it affect children?

- Group of conditions affecting motor skills, causing difficulty in movement, maintaining balance and posture, communicating, learning and performing daily activities
- Symptoms of cerebral palsy are different for each person
 - Spastic syndromes (affect 80%, most common type) muscle stiffness, movement difficulties and difficulty in holding objects, swallowing food and speech difficulties
 - Athetoid or dyskinetic cerebral palsy (affects 20%) : involuntary movements, unusual postures and repetitive movements
 - Ataxic syndromes (affects 5%) : lack of coordination, tremors and walking with feet wide apart, unsteady and shaky movements, difficulty in maintaining balance
- Children may have unusual body postures, delayed developmental milestones, epilepsy, intellectual disability, vision, hearing and speech impairment

Depending on the parts of the body affected

- **Quadriplegia** : both arms and legs, muscles of the trunk and face are affected
- **Diplegia** : both legs are affected, fine motor functions of the arms may be affected to a lesser extent
- **Hemiplegia** : one side of the body is affected

Prevalence 2-3 per 1000 live births

3. How is it diagnosed?

Identified by developmental screening. The average age of diagnosis is 18 months.

3.3 Cerebral Palsy



This YouTube video from the Cerebral Palsy Alliance from Australia provides an excellent overview of the condition.
<https://cparf.org/what-is-cerebral-palsy/>



4. How is it treated?

Rehabilitation therapies, medications for comorbidities

6. What is the risk/recurrence risk?

Only 1% of people with cerebral palsy is likely to have an affected sibling

5. What are the risk factors? Can it be prevented?

- Preterm birth (birth at 28 weeks increases the risk by 50%)
- Low birth weight, multiple births (twins, triplets or more)
- Placental pathology
- Rh or ABO incompatibility
- Maternal infections during pregnancy (eg rubella, Zika virus infection), pregnancy obesity, maternal pre-eclampsia, intrauterine disorders
- Fetal complications like fetal stroke
- Birth related complications, like traumatic head injury, bleeding into the brain, prolonged oxygen deprivation during pregnancy, during or shortly after birth
- Central nervous system infection

Prevention - Antenatal care, strengthening of maternal and child health services especially perinatal services

7. What is the public health role?

- Implementation of medical and rehabilitation services, social welfare measures
- Strengthening antenatal and perinatal care
- Providing care for small and sick newborns
- Medical care, referral for early intervention therapies
- Ensuring referrals to social welfare systems for disability benefits
- Increasing awareness among medical providers
- Ensuring education of parents, teachers, caregivers about cerebral palsy
- Psychosocial support for caregivers, caregiver skills programmes
- Increasing community awareness about cerebral palsy
- Disability sensitization
- Supporting parent-patient organizations/NGOs

3.4 Down syndrome

1. What is Down syndrome?

- An intellectual disability, accompanied by other co-morbidities, caused by an extra copy of whole or part of chromosome 21
- Down syndrome affected children have distinctive physical features; small head, characteristic facial features, flat nasal bridge, epicanthal folds skin fold covering the inner corner of the eye, upwardly slanting eyes, protruding tongue, low set small ears, short neck, extra skin on the back of the neck, short stature, single transverse palmar crease (single line across the palm), short and curved fifth finger, wide space between first and second toes, excessive

2. How does Down syndrome affect children?

- Intellectual disability ranging from mild to severe, but most have mild or moderate intellectual disability
- Increased risk of comorbidities like congenital heart defects, seizure disorders, obesity, thyroid disease, gastrointestinal atresia, leukemia, sensory impairments and complications, attention deficit hyperactivity disorder, autistic behavior
- All children do not have all these comorbidities

3. How is it diagnosed?

- Prenatal ultrasound conducted between 11th and 14th week of pregnancy
- Maternal triple and quadruple screening test which measures maternal serum biomarkers
- Chorionic villus sampling and amniocentesis, followed by karyotyping
- At birth, by typical physical features

References Bull, M. J. (2020). Down syndrome. *New England Journal of Medicine*, 382(24), 2344-2352.
 Agarwal Gupta, N., & Kabra, M. (2014). Diagnosis and management of Down syndrome. *The Indian Journal of Pediatrics*, 81(6), 560-567.

4. How is it treated?

- Advanced maternal age – risk increases with maternal age (1 in 2000 at age 20, 1 in 1000 at age 30, 1 in 365 at 35 and 1 in 100 by 40)
- Carrier of a chromosome 21 translocation
- **Prevention** - prenatal diagnosis, genetic counselling

5. What are the risk factors? Can it be prevented?

Only 1% of Down syndrome have a hereditary component, risk and recurrence risk related to maternal age, carrier status

6. What is the risk/ recurrence risk?

- Intellectual disability cannot be cured
- Symptomatic treatment e.g. surgery for congenital heart defects, and medical treatment for complications and co morbidities
- Rehabilitation therapies (physical, occupational, speech therapies) and special education initiated early as possible, before 3 years of age

7. What is the public health role?

- Implementation of medical and rehabilitation Implementation of medical and rehabilitation services to mitigate complications, prevent progression of disability, social welfare measures
- Ensuring compulsory newborn screening, developmental monitoring
- Referral for intervention services
- Ensuring referrals to social welfare services for disability benefits
- Increasing awareness among medical providers
- Psychosocial support for caregivers, caregiver skills programmes
- Promoting family planning, making contraceptives more accessible
- Increasing community awareness about Down syndrome
- Disability sensitization activities
- Supporting parent-patient organizations/NGOs



Here is a Youtube video that demonstrates the abilities and challenges of living with Down Syndrome
 Running A Business With Down Syndrome In India | EVERYDAY BOSSES #42
https://www.youtube.com/watch?v=1c7x_G8x8oM

3.5 Intellectual Disability

1. What is intellectual disability?

- Previously referred to as mental retardation
- Condition marked by limited intellectual ability (difficulty in understanding, comprehending and learning) and adaptive behaviors (difficulty in conceptual, social and practical skills)
- Categorized into four categories, mild, moderate, severe and profound intellectual disability

2. How does it affect children?

- Developmental and growth delay
- Difficulty in independently doing daily life activities (like eating, bathing, toileting, and dressing)
- Immature behaviour, poor social skills, and poor communication ability
- Higher chances of epilepsy and seizure disorders, allergies, ear problems, digestive problems, menstrual problems, sleep disturbances, vision and hearing impairment and constipation, associated with autism spectrum disorder, cerebral palsy, attention deficit hyperactivity disorder, other congenital disorders

3. How is it diagnosed?

- Cannot be diagnosed during pregnancy, unless associated with a specific disorder that can be diagnosed prenatally.
- Laboratory tests such as chromosome microarray, fragile X test, metabolic tests such as serum amino acids and urine organic acids



Prevalence – 1% globally

4. How is it treated?

Associated medical conditions can be treated, but the intellectual disability cannot be cured

Rehabilitation therapies (physical, occupational, speech therapies), initiated early as possible, before 3 years of age

5. What are the risk factors? Can it be prevented?

- Genetic causes (like Down syndrome and other genetic syndromes), metabolic disorders, congenital brain malformation
- Injury during labor and delivery or insufficient oxygen to the brain, maternal infections, alcohol intake during pregnancy (Fetal Alcohol Syndrome)
- Specific medications during pregnancy
- Acquired after birth by head trauma, exposure to toxic substances like lead and mercury, or infections like meningitis, measles or whooping cough, stroke

Prevention – Antenatal care, newborn screening, immunization,

6. What is the risk/recurrence risk?

Dependent on the etiology, could be as high as 3% - 9% for specific conditions

7. What is the public health role?

- Implementation of medical and rehabilitation services to mitigate complications, prevent progression of disability, social welfare measures to improve quality of life, limit out of pocket expenditure
- Ensuring compulsory newborn screening, developmental monitoring
- Referral for intervention services
- Ensuring referrals to social welfare services for disability benefits
- Increasing awareness among medical providers
- Psychosocial support for caregivers, caregiver skills programmes
- Promoting family planning, making contraceptives more accessible
- Increasing community awareness about intellectual disability
- Disability sensitization activities
- Supporting parent-patient organizations/NGOs

References Patel et al (2020). A clinical primer on intellectual disability. *Translational pediatrics*, 9(Suppl 1), S23.

Morisse, F., Vandemaële, E., Claes, C., Claes, L., & Vandevelde, S. (2013). Quality of life in persons with intellectual disabilities and mental health problems: An explorative study. *The Scientific World Journal*, 2013.

1. What is Autism Spectrum Disorder (ASD)?

- Neurodevelopmental disorder causing social and communication impairments and repetitive behavior
- Occurs due to atypical neurodevelopment
- More common and strongly presented in boys than in girls

2. How does it affect children?

- A range of behaviors e.g. poor social behavior, reduced eye contact, selective interests, stereotypic repetitive behavior, intense dislike to specific smells, taste, colors or textures, sensitivity to ordinary sights, sounds, smells, not looking or responding to people, not wanting to be held, need for predictable routine, lack of verbal and non-verbal communication
- Irritability, hyperactivity
- Speech problems
- Increased risk of health complications like seizures, mental illness and sleep problems IQ level either high or low

3. How is it diagnosed?

Diagnosed using Autism specific screening and diagnostic tools.

Autistic behavior can be identified within the FIRST THREE YEARS of life. Early diagnosis and initiation of treatment can make a very large difference in the child.

4. How is it treated?

There is no cure for autism. Behavioral and occupational therapy, sensory integration therapy helps in enhancing positive behaviors. Medications to manage complications like seizures, constipation, insomnia. Risperidone for difficult to manage behaviors.

3.6 AUTISM



Prevalence 5.05 per 1000 children

Autism : here are some personal stories Autism Symptoms and Behaviours - Home Video

<https://www.youtube.com/watch?v=6eS2>

[CBM24E](https://www.youtube.com/watch?v=6eS2)

Stranger In The Family (Autism Documentary) | Real Stories

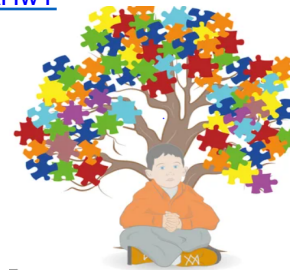
<https://www.youtube.com/watch?v=HZO>

[fjMXNPik](https://www.youtube.com/watch?v=HZO)

Journey to Diagnosis

<https://www.youtube.com/watch?v=O29>

[7E5XAHwY](https://www.youtube.com/watch?v=O29)



5. What are the risk factors? Can it be prevented?

- Genetic factors
- Associated with Down syndrome or Fragile X syndrome
- Advanced parental age
- Exposure to specific drugs or rubella infection during pregnancy
- Low birth weight, abnormally short gestation, birth asphyxia
- Post-natal auto-immune diseases, viral infections or hypoxia
- ASD cannot be prevented

6. What is the risk/recurrence risk?

Genetic etiology identified from 90% increased risk in identical twins, and 20% increased risk in non-identical twins. 50-100 times increased risk.

7. What are the public health implications?

Implementation of medical and rehabilitation services, social welfare measures

- Ensuring screening, evaluation, medical care, referral for intervention therapies
- Ensuring referrals to social welfare systems for disability benefits
- Increasing awareness among medical providers
- Ensuring education of parents, teachers, caregivers
- Psychosocial support for caregivers, caregiver skills programmes
- Increasing community awareness about autism
- Disability sensitization
- Supporting parent-patient organizations/NGOs

References Chiarotti, F., & Venerosi, A. (2020). Epidemiology of autism spectrum disorders: a review of worldwide prevalence estimates since 2014. *Brain sciences*, 10(5), 274. Hyman, S. L et al (2020). Identification, evaluation, and management of children with autism spectrum disorder. *Pediatrics*, 145(1).; Thabtah, F., & Peebles, D. (2019). Early autism screening: a comprehensive review. *International journal of environmental research and public health*, 16(18), 3502

3.7 Attention Deficit Hyperactivity Disorder (ADHD)

1. What is ADHD?

- Neurobehavioral condition
- Characterized by inattention, hyperactivity and impulsiveness.
- More common among boys than girls

Prevalence – 5% -8 % among school going children

2. How does ADHD affect children?

- Range of behavioral extremes; temper tantrums, high demand for attention, aggressiveness, disobedience, impulsiveness, poor social skills that might affect making friends, inability to follow instructions, getting distracted easily, forgetfulness and restlessness, difficulty in learning and paying attention

3. How is it diagnosed?

Diagnosis of ADHD is difficult, especially in children under five years of age. Criteria for ADHD diagnosis is :

- Behavior issues since young age, and prior to 12 years of age
- Behavior should be present for at least 6 months prior to consultation
- Complaints or behavior issues from two independent sources, such as parents and school; parents should be very concerned about the child's uncontrollable behavior; complaints of indiscipline and poor academic performance from school



Youtube video ADHD: Out of Control Kids (Medical/Parenting Documentary) | Real Stories
<https://www.youtube.com/watch?v=yRYI9Bf0yhs>

4. How is it treated?

- Lifelong condition
- A combination of medications and behavioral therapy can reduce the symptoms

References Gnanavel et al (2019). Attention deficit hyperactivity disorder and comorbidity: A review of literature. *World journal of clinical cases*, 7(17), 2420.; Thapar, A. et al (2012). What causes attention deficit hyperactivity disorder?. *Archives of disease in childhood*, 97(3), 260-265.

5. What are the risk factors? Can it be prevented?

- Genetic factors (70-80% heritability)
- Developmental issues in brain regions involved in focus and paying attention
- Deficiency in neurotransmitter levels
- Frontal lobe injury
- Exposure to toxins like lead
- Alcohol and tobacco intake during pregnancy

Prevention – antenatal care, child care

6. What is the risk/recurrence risk?

Recurrence risk - 5 times higher risk among siblings and 1-2% increased risk among cousins.

7. What are the public health implications?

- Implementation of medical and rehabilitation services to mitigate complications, improve behaviors, social welfare measures to improve quality of life, limit out of pocket expenditure
- Ensuring education of parents, teachers, caregivers about ADHD
- Referral for medical care and therapies
- Ensuring referrals to social welfare services for disability benefits
- Increasing awareness among medical providers, teachers
- Psychosocial support for caregivers, caregiver skills programme
- Increasing community awareness about ADHD
- Disability sensitization
- Supporting parent-patient organizations/NGOs

3.8 Congenital Hearing Impairment

1. What is congenital hearing impairment?

- Inability/difficulty to hear since birth. Caused by the incapacity of the ear to convert vibratory mechanical energy of sound to electrical energy of nerve impulses.
- Sensorineural hearing loss affects inner ear or the auditory nerve that connects the ear to brain, causes permanent hearing loss, that deteriorates progressively.

Estimated prevalence - 1.33 per 1000 live births

2. How does it affect children?

- Inability to hear (deaf, hard of hearing)
- Difficulty in social interactions
- Delayed speech and language development
- Challenges schooling, employability if systems are not in place

3. How is it diagnosed?

Cannot be identified during pregnancy. Newborn and developmental screening can identify babies needing further evaluation.

References — Renauld, J. M., & Basch, M. L. (2021). Congenital deafness and recent advances towards restoring hearing loss. *Current protocols*, 1(3), e76.; Kar A (2021) Some common birth defects. In: Kar, A. (eds) Birth Defects in India. Springer, Singapore. https://doi.org/10.1007/978-981-16-1554-2_1 Korver, A. et al (2017). Congenital hearing loss. *Nature reviews Disease primers*, 3(1), 1-17.

4. How is it treated?

Congenital hearing impairment cannot be cured. Hearing aid, cochlear implants, rehabilitative therapies for communication, learning and education such as auditory-verbal therapy, and non-verbal communication i.e. sign language

- **Devices (hearing aids) give best results if fitted before 6 months of age and used continuously.**
- **Early identification before 2 weeks of age is most beneficial as it improves hearing amplification and quality of life**
- **Delayed diagnosis increases the risk of speech impairment**

6. What is the risk/recurrence risk?

Condition specific, 1.43% increased risk in case of family history of congenital hearing impairment

7. What is the public health role?

- Introduction of newborn hearing screening
- Rehabilitation, and access to assistive devices like hearing aids, cochlear devices, verbal and non-verbal communication therapies (sign language)
- Special education, ensuring employment opportunities, appropriate social welfare schemes
- Ensuring accessibility for deaf people
- Psychosocial support, counseling and teaching parents on caregiving
- Community sensitization about disability



Here is a video from the NHS, England on newborn hearing screening.
<https://www.youtube.com/watch?v=85GBPNvABQ8>

Hearing and speech milestones

(Absence of or poor response indicates need for screening for hearing impairment)

0-4 months	Baby is startled at a loud noise, turns head or moves eyes to see the source of the noise, is calmed down by parent's voice
4-8 months	Baby notices nearby sounds, smiles when spoken to, makes baby sounds, understands simple words
8-14 months	Baby responds to name, says simple words, calls to get attention
14-24 months	Puts 2 words together, understands and follows simple instructions

5. What are the risk factors? Can it be prevented?

- Prematurity, low birth weight, admission to neonatal intensive care units for complications
- Infections during pregnancy (cytomegalovirus infection or other viral infections like rubella, syphilis, herpes simplex virus, Zika virus and *Toxoplasmosis gondii* infection)
- Use of certain drugs during pregnancy like streptomycin or gentamicin
- Developmental delays, craniofacial anomalies, single gene disorders like Usher and Waardenburg syndromes
- Consanguineous marriages

Prevention Antenatal care, neonatal screening, rubella vaccination, genetic counselling

3.9 Blindness and Vision Impairment

1. What is vision impairment?

- Blindness - absolute inability to see
- Vision impairment - difficulty to see clearly
- Most common cause of childhood vision impairment is near sightedness (myopia) and far sightedness (hyperopia). These can be corrected with eyeglasses
- Congenital ocular anomalies include congenital cataract, congenital glaucoma, eye malformations (microphthalmos, anophthalmos, coloboma), retinopathy of prematurity, strabismus, coloboma, retinitis pigmentosa, and amblyopia, ptosis

Prevalence of childhood blindness in LMICs – 0.2 to 7.8 per 10 000 children

2. How does it affect children?

- Inability to see, or see clearly
- Increased risk of falls, injuries
- Poor opportunities in life without referral to special education
- Depression and anxiety

Reference Solebo, A. L., & Rahi, J. (2014). Epidemiology, aetiology and management of visual impairment in children. *Archives of disease in childhood*, 99(4), 375.; Kar A (2021) Some common birth defects . In: Kar, A. (eds) Birth Defects in India. Springer, Singapore. https://doi.org/10.1007/978-981-16-1554-2_1; Keil, S et al (2017). Management of children and young people with vision impairment: diagnosis, developmental challenges and outcomes. *Archives of Disease in Childhood*, 102(6), 566

3. How is it diagnosed?

Vision related issues cannot be detected during pregnancy.

Can be recognized if

- The eye looks different. (Misaligned eyes (cross eyed), eyes appear moving (wandering eyes, nystagmus), swelling or bulging of the eye (exophthalmos), droopy lids (ptosis), cloudy eyes with a white haze in the centre of the eye (congenital cataract)
- Child may appear clumsy and bump into objects, hold books, toys close to his face, sit close to the television, loose interest and avoid work that requires good vision, repeatedly shut or close one eye, squint, blink, rub the eye, try to avoid light etc. when there is no bright light

4. How is it treated?

Eyeglasses to correct vision problems. For congenital defects, may need surgery, lens implant, medications, eye patches, and use of therapeutic and low vision aids, or special education

5. What are the risk factors? Can it be prevented?

- Maternal infections (cytomegalovirus, herpes simplex and rubella), alcohol consumption, birth asphyxia, prematurity, low birth weight, oxygen therapy following delivery, neonatal conjunctivitis

Prevention - Antenatal care, rubella vaccination, newborn screening, genetic counselling

6. What is the risk/recurrence risk?

Recurrence is only possible in case of rare hereditary conditions. Genetic counselling will be required if child has genetic disorder related vision impaired.

7. What is the public health role?

- Compulsory eye screening services
- Medical care and rehabilitation, including access to assistive devices like eye glasses, canes etc.
- Special education, employment opportunities for visually impaired
- Referral for appropriate social welfare schemes
- Creating accessible spaces
- Psychosocial support, counseling and teaching parents on caregiving
- Community sensitization about disability

Vision development in babies

- By first month – babies can focus on objects dangling in front of them
- By 3 months – babies can focus on and follow objects
- By 5 months – babies develop depth perception and get better at reaching for objects

Early identification and referral is important. Delay in treatment of vision impairment can lead to blindness



Check out this video :

<https://raisingchildren.net.au/disability/videos/vision-impairment-overview>