

Birth Defects and Childhood Disability Toolkit

Section 2 - Common congenital anomalies



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

www.birthdefectsindia.com

Contact us : birthdefcent@gmail.com WhatsApp+91 9172959236

 anita_kar

2.1 CONGENITAL ANOMALIES: WHAT ARE THEY?

1. What are congenital anomalies (congenital malformations, birth defects)?

Abnormalities that occur during anatomical development of the foetus for example:

1. **Congenital heart defects** (defects in the structure of the heart)
2. **Cleft lip and palate** (defect in development of the face of the baby)
3. **Clubfoot** (defect in development of the feet causing locomotor disability)

2. What are their health implications?

- Mild, moderate or severe disability caused by impairment in movement, cognition, hearing, speech, vision, learning
- Frequently associated with medical complications (e.g. congenital rubella syndrome causes cataracts resulting in blindness, hearing impairment, intellectual disability and developmental delay)
- May cause major cosmetic effects eg cleft lip and palate, limb deformity
- Pregnancy loss (miscarriage, stillbirth)
- Preterm births and low birth weight
- Childhood mortality (neonatal, infant, child)
- Premature adult mortality
- Stigma, isolation, emotional distress, poor quality of life, out of pocket expenditure

3. How are they caused?

Etiology of most congenital anomalies is unknown:

1. **Constitutional**
 - a) Mostly gene-environmental factors
 - b) Less frequently, may be caused by single gene mutations and chromosomal abnormalities
2. **Acquired**
 - c) Teratogenic exposures



Birth prevalence- affects 2-3% pregnancies

Selected references: 1.Christianson A, Howson CP, Modell B (2006) March of Dimes Global Report on Birth Defects: the hidden toll of dying and disabled children. March of Dimes Birth Defects Foundation, White Plains, New York; 2. Sixty-third World Health Assembly WHA63.17 Agenda item 11.7 21 May 2010 Birth defects. Available via https://apps.who.int/gb/ebwha/pdf_files/WHA63/A63_R17-en.pdf?ua=1 Accessed March 2018 ; 3. Kar, A. (2021). Birth Defects: A Public Health Approach. In: Kar, A. (eds) Birth Defects in India. Springer, Singapore. https://doi.org/10.1007/978-981-16-1554-2_1

4. What are the needed public health activities?

1. Ensuring that birth defects services are included in the MCH programme

2. Developing capacity to diagnose, refer, treat, manage common conditions

3. Prevention

4. Surveillance

5. Increasing awareness about birth defects, prevention, available services for children

6. Disability sensitization

7. Strengthening rehabilitation workforce services, implementing screening and early intervention

8. Regulation of fetotoxic environmental exposures; ensuring evidence-based interventions for children with disabilities and chronic medical conditions, ensuring the rights of children with disabilities

9. Partnerships with NGOs, professional organizations

2.2 Congenital Heart Defects (CHD)

1. What are congenital heart defects?

- Malformations in the structure of the heart and/or in the blood vessels carrying blood to and from the heart.
- Causes intermixing of deoxygenated and oxygenated blood or reduction in blood flow.
- Most prevalent birth defect

Birth prevalence – 9.3 per 1000 live births

CHD subtype	Prevalence of CHD subtype per thousand (95% confidence interval)
Ventricular septal defect	3.071 (2.845–3.305)
Atrial septal defect	1.441 (1.215–1.687)
Patent ductus arteriosus	1.004 (0.803–1.228)
Pulmonary stenosis	0.546 (0.485–0.611)
Tetralogy of Fallot	0.356 (0.326–0.387)
Transposition of the great arteries	0.295 (0.269–0.322)
Atrioventricular septal defect	0.290 (0.265–0.316)
Coarctation of the aorta	0.287 (0.261–0.314)
Pulmonary arteriovenous aneurysm	0.272 (0.253–0.425)
Congenital heart block	0.268 (0.028–0.752)

2. How does CHD affect children, adults?

Babies with CHD

- Unable to suckle, irritability, poor feeding and poor weight gain, rapid breathing, grey or bluish tinge to skin, edema of face, legs

In older children

- Shortness of breath, fainting after exertion, easily fatigued, edema of hands, feet, legs, repeated episodes of cough, not responding to routine treatment

In children and adults

- Neurodevelopmental, neurocognitive and psychosocial complications, affects schooling and adult employment
- Non cardiac complications in adults

3. How are they diagnosed?

- May be detected during pregnancy, by ultrasound (fetal echocardiogram).
- After clinical suspicion, by echocardiography, chest X-ray and other tests

4. How is it treated?

70% CHDs require surgeries (done within the first year of life for critical CHDs); minor CHDs get repaired spontaneously

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

- 15-20% linked to chromosomal anomalies e.g. Down syndrome, other chromosomal disorders, few with single gene disorders
- Maternal characteristics: obesity, pre-existing diabetes, tobacco use, certain medications during pregnancy, advanced maternal age (>35 years), use of assisted reproductive techniques, prior pregnancies, maternal chronic conditions such as epilepsy, use of medications, such as valproate, phenytoin, and isotretinoin may be associated with some types of CHDs
- Folic acid deficiency, Infections like rubella, influenza, family history,
- No specific method of prevention

6. What is the risk/recurrence risk?

CHD in a firstborn child associated with increased risk in subsequent children; three-fold increased risk compared to general population for a family in which the mother, father, or sibling has CHD

7. What is the public health role?

- Developing capacity to identify, treat and manage CHDs
- Providing access to surgery
- Ensuring early identification through newborn screening using pulse oximetry, or through school health checkups
- Increasing awareness among general practitioners about signs, symptoms and availability of public services
- Preconception screening to identify women at risk (e.g. women with existing diabetes, family history), further providing appropriate management
- Strengthening caregiver skills for managing children till and after surgery, psychosocial support and counselling services for parents
- Ensuring referrals to social welfare services to ensure access to financial support
- Increasing community awareness
- Supporting NGOs providing services

Women with congenital heart defects are at a higher risk of complications during pregnancy

Pregnancy can be high risk for women with congenital heart defects

2.3 Orofacial Clefts (OFCs)

1. What are OFCs?

- Cleft (gap) in the roof of the mouth (cleft palate) or upper lip (cleft lip) occurring due to errors in development of the craniofacial bones.
- Most prevalent craniofacial congenital malformation.
- Can be unilateral, bilateral, complete, incomplete, cleft lip only, cleft palate only or cleft lip and palate.
- Generally (70 -80%) isolated 'non-syndromic' cases
- Boys have a higher risk of cleft lip/palate and girls have a higher risk of only cleft palate

Prevalence 1 per 700 newborns, ethnic and geographic variations

2. How does OFC affect children?

- Difficulty while feeding
- Trouble speaking clearly
- Hearing problems
- Increased risk of ear infections
- Dental problems
- Psychological issues



3. How are they diagnosed?

- Physical examination of newborn,
- Can be detected in the prenatal period through ultrasound scan

4. How are they treated?

Reconstructive surgery to close the clefts, within the first two years of life, completed in two stages.

7. What are the public health issues?

- Early identification of OFCs through compulsory newborn exam prior to discharge
- Developing strategies to ensure access to surgery, e.g. strengthening capacity, collaboration with international NGOs like the SmileTrain
- Implementation of medical and rehabilitation services, social welfare measures
- Increasing awareness among medical providers about signs, symptoms and referral pathways to public services
- Strengthening caregiver skills for managing infants, young children till and after surgery
- Psychosocial support and counselling for parents
- Ensuring referrals to social welfare services
- Increasing community awareness

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

- Genetic causes, parental consanguinity
- Folic acid deficiency
- Maternal obesity, alcohol and tobacco use during pregnancy, high fever in early pregnancy, pre-gestational and gestational diabetes, certain types of seizure medications; sedatives, folate antagonists
- **Prevention**: moderately strong evidence for reduced risk in women reporting preconception folic acid intake

6. What is the risk/recurrence risk?

Recurrence risk estimated to be 3.5% for first, 0.8 % for second-, and 0.6% for third degree relatives. Significantly higher recurrence risk for offspring and siblings of severe cases, eg sibling recurrence risk of isolated bilateral cleft lip with cleft palate was 4.6% versus 2.5% for a case of unilateral defect.

References: Kadir A. et al., (2017). Systematic review and meta-analysis of the birth prevalence of orofacial clefts in low-and middle-income countries. *The Cleft palate-craniofacial journal*, 54(5), 571-581; Wehby, G. L., & Cassell, C. H. (2010). The impact of orofacial clefts on quality of life and healthcare use and costs. *Oral diseases*, 16(1), 3-10. Grosen et al. (2010). A cohort study of recurrence patterns among more than 54 000 relatives of oral cleft cases in Denmark: support for the multifactorial threshold model of inheritance. *Journal of medical genetics*, 47(3), 162-168.

2.4 Spina Bifida



1. What is spina bifida?

- Spina bifida is a neural tube defect (NTD)
- Caused by incomplete closure of the spinal chord
- Anencephaly is another NTD where there is incomplete formation of the brain. It is a severe condition leading to stillbirth, or early neonatal death
- Spina bifida may present as an isolated case, or may be associated with other birth defects like hydrocephalus, clubfoot, contractures, scoliosis, hip dislocation

Birth prevalence 4 per 1000 live births

References Avagliano L et al (2019) Overview on neural tube defects: From development to physical characteristics. Birth defects research. 15;111(19):1455-67.; Atlaw, D et al (2021). Magnitude and determinants of neural tube defect in Africa: a systematic review and meta-analysis. BMC pregnancy and childbirth, 21(1), 1-14.

2. How does it affect children?

- Spina bifida may cause
- Paralysis of the legs, locomotor impairment
- Loss of urinary and bowel control,
- Urinary tract infection, constipation,
- Development of pressure sores in wheel chair bound children

3. How is it diagnosed?

- Prenatal ultrasound
- Prenatal triple marker test in the 2nd trimester

4. How is it treated?

- Surgery for repair of the spinal lesion
- Physiotherapy
- Additional treatment for complications, for example, SB may be accompanied with hydrocephalus

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

- Folic acid deficiency
- Gestational diabetes
- Maternal obesity
- Hyperthermia
- Use of certain medications
- Alcohol and tobacco consumption

6. What is the risk/recurrence risk?

Recurrence risk is 20-50% for the next pregnancy.

Prevention

- **70% of folate sensitive NTDs are preventable by maintaining adequate folate levels during the preconception period and pregnancy.**
- 400 micrograms of folic acid for at least 90 days **prior to conception**
- For women with a history of an affected birth, 4mg of folic acid per day for a minimum period of 90 days **BEFORE PREGNANCY**
- Ensuring availability and access to folate fortified foods

7. What are the public health issues?

- Implementing prevention, that is ensuring folic acid supplementation for women in the preconception period, access to folate fortified foods, identifying and targeting at high risk women
- Educating all women about folic acid and spina bifida, and other birth defects
- Ensuring access to emergency surgery for neonates with spina bifida
- Referral for early intervention services
- Strengthening caregiver skills, psychosocial support and counselling services for parents
- Increasing awareness among medical services, alerting the need to counsel eligible women about preconception folate supplementation
- Ensuring referrals to social welfare services for disability benefits
- Supporting NGOs disseminating information on folic acid, and support and care for children with spina bifida

1. What is hydrocephalus?

- Larger than normal head size
- Caused by collection of cerebrospinal fluid in the brain
- Congenital hydrocephalus associated in 70% of cases with other anomalies, most commonly spina bifida.
- Develops later in pregnancy, when elective termination of pregnancy may not be possible
- Hydrocephalus may be acquired in the post-natal period, caused by conditions like prematurity.
- Affects more males than females.

Prevalence 1 per 2000 live births

2. How does hydrocephalus affect children?

Disabling condition

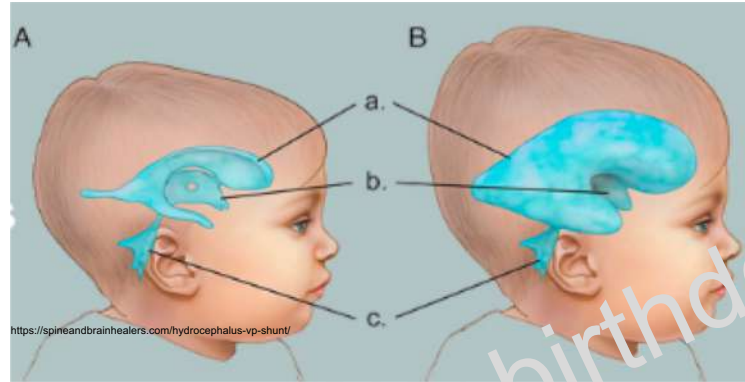
- increased pressure inside the brain, causes irritability, listlessness, high pitched cry, problems sucking or feeding, recurrent vomiting and seizures, eyes fixed downward (sunsetting eyes),
- Headache, poor appetite, sluggishness,
- Intellectual disability, visual impairment, unable to speak, difficulties in movement and balance; impacts learning and activities of daily living

3. How is it diagnosed?

Recognized by obvious increase in head size; bulging fontanelle
Can be detected by ultrasound during pregnancy

Reference: Kahle et al (2016) Hydrocephalus in children. Lancet 387(10020):788–799

2.5 Hydrocephalus



Here are two contrasting stories about living with hydrocephalus: Rooting for Roona

<https://www.facebook.com/SocioStoryOfficial/videos/rooting-for-roona/330922407093916/>

Hydrocephalus - Rhys's Story

<https://www.youtube.com/watch?v=RnD2cgrLGmc>

4. How is it treated?

- Surgical insertion of a shunt to drain out extra fluid into the abdomen where it can be absorbed
- Other symptomatic treatment
- Rehabilitation therapies to cope with the daily life activities and improve functioning

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

- Overproduction of cerebrospinal fluid inside the brain caused by infections during pregnancy, or by defects in the structure of the brain, so that the fluid cannot be reabsorbed/cannot drain out from the brain.
- No single method of prevention
- Surgical care, medical care, rehabilitation therapies, counselling and support for parents

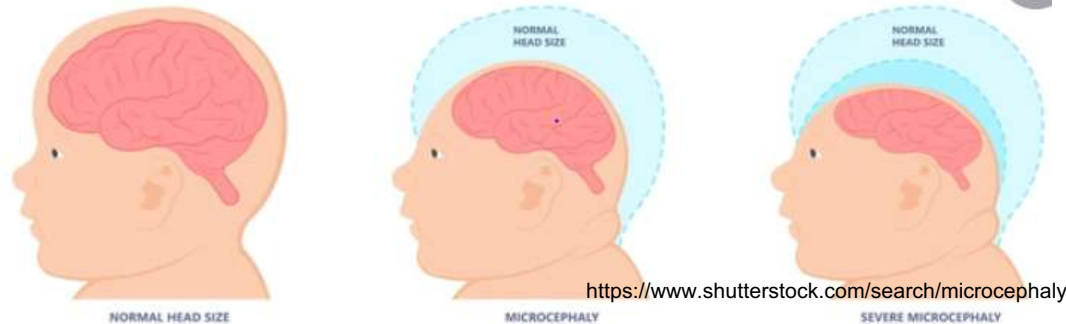
6. What is the risk/recurrence risk?

Risk increased with specific conditions e.g. spina bifida

7. What are the public health issues?

- Access to medical services
- Ensuring compulsory newborn examination and appropriate referral to specialist care
- Increasing awareness among medical providers and information on availability of public services
- Referral for rehabilitation therapies
- Psychosocial support for caregivers
- Ensuring referrals to social welfare services for disability benefits
- Increasing community awareness about hydrocephalus and other birth defects
- Supporting NGOs providing care

2.6 Microcephaly



<https://www.shutterstock.com/search/microcephaly>

1. What is microcephaly?

- Small head size compared to head size of typically growing babies of same age and gender
- Defined as head circumference more than two standard deviations (SD) below the mean for gender and age, (includes about 2% of the population; severe microcephaly (greater than three SD below the mean, affects 0.1% of the population)
- Present at birth (congenital microcephaly) or develops later in life (postnatal or acquired microcephaly)

Prevalence 2 - 12 per 10,000 births

2. How does microcephaly affect children?

Severe microcephaly is a debilitating condition, characterized by intellectual disability, frequent seizures, developmental delay, visual, hearing impairment, locomotor difficulties and intellectual impairment

3. How is it diagnosed?

Detected in ultrasound scans by the end of the second trimester or early in the third trimester.

4. How is it treated?

- Symptomatic treatment for seizures, hyperactivity and neuromuscular symptoms.
- Rehabilitation therapies (including physical, occupational and speech therapies)

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

- Maternal infections (like cytomegalovirus, herpes simplex virus, Zika virus infection); genetic abnormalities (chromosomal or single gene disorders), metabolic disorders, teratogens; prenatal, perinatal, and postnatal injuries

6. What is the risk/recurrence risk?

Recurrence risk is dependent on the underlying cause

7. What are the public health issues?

- Ensuring compulsory newborn exam for microcephaly
- Assuring access to medical and rehabilitation services
- Increasing awareness among medical providers, and about the availability of public medical and rehabilitation services
- Psychosocial support for caregivers
- Increasing community awareness about microcephaly, Zika virus risk, and other birth defects
- Ensuring referrals to social welfare services for disability benefits
- Supporting parent-patient organizations /NGOs

References Hanzlik, E., & Gigante, J. (2017). Microcephaly. *Children*, 4(6), 47.; Devakumar et al (2018). Infectious causes of microcephaly: epidemiology, pathogenesis, diagnosis, and management. *The Lancet infectious diseases*, 18(1), e1-e13.; Nawathe, A., et al (2018). Fetal microcephaly. *Bmj*, 361.

2.7 Congenital Zika Syndrome (CZS)

1. What is Congenital Zika Syndrome?

- A severe birth defect caused by Zika virus disease during pregnancy, ZKV disease symptoms are *fever, skin rashes, conjunctivitis, body pain, weakness*
- CZS characterized by a spectrum of structural and non-structural birth defects – microcephaly, intellectual disability, locomotor difficulties, hearing impairment and blindness.
- Can be transmitted from **asymptomatic individuals**

Prevalence Occurs in **6-10%** of pregnancies with laboratory confirmed prenatal infection

2. How does it affect children?

- Severe intellectual disability, poor neurological and language development
- Epileptic seizures
- Mild congenital heart defects
- Diaphragmatic paralysis

3. How is it diagnosed?

Singleplex RT-PCR, CDC Trioplex real time RT-PCR, and serological tests.

4. How is it treated?

- Treatment of medical conditions, rehabilitation services
- Psychosocial support of caregivers
- Social welfare measures



Understanding how CZS affects mothers, and their babies

[CBSN Originals S3 E5 Zika: Children of the Outbreak | Full Documentary](https://www.youtube.com/watch?v=ywPEj1IU7pM)
<https://www.youtube.com/watch?v=ywPEj1IU7pM>



John Ragai from Petaling Jaya, Malaysia, CC BY 2.0
 <<https://creativecommons.org/licenses/by/2.0/>>, via Wikimedia Commons

5. What are the risk factors?

Zika virus disease caused by bite of infected mosquito (*Aedes aegypti*, *A. albopictus*).
 (Mosquitoes have white markings on their body, and bite during the day (generally during the early morning hours and late afternoon hours).
 Settings with poor vector control, lack of surveillance, existing case in the community enhances the risk of CZS

6. What are the prevention measures?

- Vector (mosquito) control measures
- Insecticide treated bed nets, indoor residual spraying,
- Avoiding travel to areas with ongoing transmission

7. What are the public health implications

- Preventing vector breeding
- Increasing awareness among women about CZS and prevention
- Offering testing, ultrasound scans to women in areas with ongoing transmission
- Screening and early intervention for all newborns and infants
- Ensuring access to medical and rehabilitation services, social welfare services
- Counseling and support for parents; education on caregiving skills
- Increasing awareness among medical providers
- Increasing community awareness about CZS and other birth defects
- Supporting parent-patient organizations / NGOs

Reference: Baud, D et al. (2017). An update on Zika virus infection. *The Lancet*, 390(10107), 2099-2109.; Rasmussen, S. A., & Jamieson, D. J. (2020). Teratogen update: Zika virus and pregnancy. *Birth Defects Research* 112(15):1139-1149. Des, A. E. et al. (2021). Vertical transmission of Zika virus and its outcomes: a Bayesian synthesis of prospective studies. *The Lancet Infectious Diseases*, 21(4), 537-545.

2.8 Clubfoot (CONGENITAL TALIPES EQUINOVARUS, CTEV)

1. What is clubfoot?

- Congenital defect affecting muscles of legs and feet
- Feet turned inwards, pointing to other leg.
- May affect one or both feet.
- More common in boys than girls.
- May be isolated, or associated with other congenital syndromes
- Shows ethnic variation

Prevalence 1 in 1000 live births

2. How does it affect the children?

Lifelong locomotor disability if left untreated

3. How is it diagnosed?

Can be detected in the prenatal period through ultrasound
Newborn examination



<https://www.indiamart.com/productdetail/denis-browne-bar-db-split-leather-clubfoot-shoes-for-ctev-correction-2469511048.html>



Birth Defects Centre, Pune

Check this story about a child with clubfoot.

<https://www.youtube.com/watch?v=yxjbw-Qd8r8>

4. How is it treated?

- Treated by Ponseti method,
- Foot placed in a cast for 2 to 3 months
- Involves weekly visit to the orthopaedic surgeon for positioning
- Followed by bracing, using a foot brace (shoe with a bar which stops the foot from twisting back to its original position).
- Needs to be worn all the time for 3 months, and then at night till 4 years of age.
- Ponseti method has a success rate of 90%, and treated children show no signs of walking problems.

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

- Risk increased when
- There is another family member with clubfoot,
 - Associated with maternal lifestyle factors (smoking, alcohol use, recreational drug use during pregnancy,
 - pregnancy complications such as less amniotic fluid during pregnancy, uterine fibroids etc
 - No specific preventive method
 - can be detected by ultrasound by 20th week of pregnancy, providing opportunity for counseling parents about benefits of Ponseti treatment

7. What are the public health issues?

- Ensuring compulsory newborn exam for clubfoot
- Providing medical and rehabilitation services
- Supporting caregivers with information especially on the importance of treatment compliance
- Increasing awareness among medical providers and knowledge about availability of treatment
- Increasing community awareness about clubfoot and other birth defects
- Supporting parent-patient organizations / NGOs

6. What is the risk/recurrence risk?

Risk of clubfoot increases by 2-3% when one parent is affected and by 15% when both parents are affected.

References Dave et al (2020). *Pediatric Rheumatology*, 18(1), 1-7.; Werler et al. (2013) *American journal of medical genetics Part A*, 161(7), 1569-1578.

2.9 Developmental Dysplasia of the Hip (DDH)

1. What is DDH?

- Malformation of bones at the hip joint resulting in poor articulation of the thigh bone in the socket of the hip joint.

Prevalence 1–2 per 1000 through physical examination, increases to 5–30 per 1000 with ultrasound screening of hips

4. How is it treated?

- Treated by placing the child in a Pavlik harness, a brace which stabilizes hip joints.
- Keeps baby's hips spread outwards and upwards towards the chest.
- Baby needs to stay in the harness for at least 12 weeks, compliance important
- Surgery needed in severe cases

7. What are the public health issues?

- Ensuring newborn and child screening for DDH
- Implementation of medical and rehabilitation services, social welfare measures
- Supporting caregivers with information and the importance of treatment compliance
- Referral for any intervention services for children with disabilities
- Increasing awareness among medical providers, and availability of public services
- Increasing community awareness about DDH and other birth defects,
- Supporting NGOs providing services

2. How does DDH affect children?

- Causes difficulty in walking as one leg is shorter than the other
- In adulthood chronic knee and back pain, limp because of shortened leg length

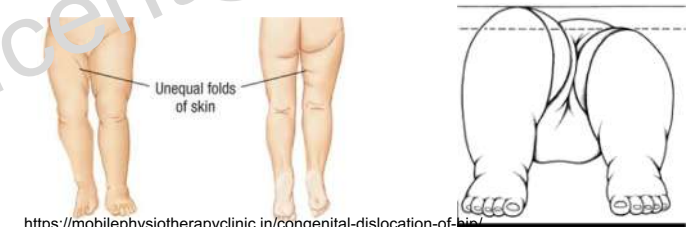
Here is Anushka's story. Her Little Steps | Dysplasia of Hip: Story of Anushka
<https://www.youtube.com/watch?v=CwbNvDSSIQk>



https://www.rch.org.au/kidsinfo/fact_sheets/Pavlik_Harness_for_DDH/

3. How is it diagnosed?

Can be identified during newborn examination or in infancy as asymmetric skin creases in the groin and thigh regions, limited hip movement, unequal knee height, or child waddles and limps while walking or walks on toes.



<https://mobilephysiotherapyclinic.in/congenital-dislocation-of-hip/>

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

- Family history
- Baby in breech position
- Low amniotic fluid volume
- multiple pregnancies
- other skeletal deformities of the spine (torticollis, spinal /foot deformities)
- No specific prevention method
- Immediate initiation of treatment can prevent disability

6. What is the risk/recurrence risk?

- Risk increased 12 times in case of family history
- 6% increase if a sibling has DDH
- Risk increased by 12% if one parent has DDH, 36% if both parents have DDH.
- Girl babies with a family history of DDH are at a higher risk of having DDH.

References: Yang, S. et al. (2019). Developmental dysplasia of the hip. *Pediatrics*, 143(1); Jorgensen, M. D. et al. (2021). Experiences of living with developmental dysplasia of the hip in adults not eligible for surgical treatment: a qualitative study. *BMJ open*, 11(12), e052486.

2.10 Congenital Limb Defects

What are congenital limb defects?



- Conditions where the development of the arm or leg is affected
- Limb may be deformed, or whole or part of limb may be missing
- May be isolated defects, or as part of genetic syndromes such as Adams–Oliver syndrome, Holt–Oram syndrome, Fanconi anemia and VACTERL



Upper limbs (4 in 10,000 live births) are more frequently affected than lower limbs (2 in 10,000 live births)

Children with congenital limb defects have problems in writing, walking and doing other daily life activities

Causes



- Defects during development.
- In very rare cases, may be associated with family history
- may also be caused by amniotic bands, (strands of tissues from the amniotic sac in a twine with the limb of the developing fetus, restricting further development of the structure)
- Historically associated with a severely teratogenic drug, thalidomide, that caused phocomelia or amelia in the early 1960's

Syndactyly, polydactyly



What is syndactyly and polydactyly?

- Extra finger or toe (polydactyly) or fingers or toes are fused together (syndactyly.)
- Isolated cases do not cause major problems to the health and well-being of the baby
- Surgery for polydactyly, in order to remove the extra finger or toe. For syndactyly, surgery separates the fused fingers and toes.



Public health issue :



- Surgery and rehabilitation, access to prosthetic devices, orthotics
- Psychosocial support, referral for disability benefits
- Disability sensitization of communities

References

- Wilcox (2015) Congenital limb deficiency disorders. Clin Perinatol 42(2):281–300
- Vasluian E, van der Sluis CK, Johansen (2016) Health-related quality of life in adults with congenital unilateral upper limb deficiency in Norway. A cross-sectional study. Disabil Rehabil 38(23):2305–2314