Birth Defects and Childhood Disability Toolkit

Section 1 - Birth defects and public health

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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1.1 What are birth defects (congenital disorders)?

1. Birth defects (congenital disorders, congenital anomalies or congenital malformations,) are conditions that affect the development of the fetus causing structural or functional anomalies [1]

2. They are maternal and child health issues of concern ("congenital"= existence at or before birth; prenatal = during pregnancy)

3. Common examples include congenital heart defects, clubfoot, cleft lip and palate, spina bifida, congenital cataract, limb deformities, and Down syndrome.

4. The complications of congenital disorders like congenital rubella syndrome, microcephaly, hydrocephalus, manifest as disability in young children. These include congenital blindness or low vision, congenital hearing impairment, intellectual disability, speech and movement impairments, and behavioural disorders like autism and attention deficit hyperactivity disorder.

5. They may be identified during pregnancy by ultrasound, or at birth (visible birth defects like clubfoot), or in early childhood.

6. Congenital disorders can be suspected when a baby has developmental difficulty, repeated episodes of illness, and disability since birth.

7. Birth defects are included in Chapter XVII of ICD-10 (Congenital malformations, deformations and chromosomal abnormalities). Maternal infections resulting in malformations, and teratogenic syndromes with malformations are included in other chapters.

8. Birth defects cause miscarriage, stillbirth, neonatal and child mortality. Children with congenital disorders have special health care needs. They need to be referred appropriately for specialized investigations, medical care and rehabilitation. Parents and caregivers need counseling and psychosocial support.

Services for children with congenital disorders are available at the District Early Intervention Centers, established through the Rashtriya Bal Swasthya Karyakram

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Reference: 1. World Health Organization Congenital disorders https://www.who.int/news-room/fact-sheets/detail/birth-defects ; 2. Kar, A.(2021). Birth Defects : A Public Health Approach In :Kar,A (ed) Birth Defects in India, Springer, Singapore. https://doi.org/10.1007/978-981-16-1554-2_1 3. Christianson, A., Howson, C. P., & Modell, B. (2006). March of Dimes. Global report on birth defects. The hidden toll of dying and disabled children. Available online.;

1.2 Common congenital disorders can be treated

- If untreated, many congenital disorders can cause lifelong medical conditions and disability
- Families may incur out of pocket expenditure on treatment

Congenital Heart Defects
clubfoot
Neural Tube Defects
Hydrocephalus
Microcephaly
Orofacial cleft
Developmental dysplasia of hip
Congenital limb defects

Down syndrome
Intellectual disability
Cerebral Palsy
Hearing Impairment
Autism
Vision Impairment
Attention Deficit Hyperactivity Disorder

Some congenital disorders can be cured (eg. congenital heart defects)

Treatment can make sure that a child is disability-free for life (eg treatment for clubfoot)

Treatment can prevent stigma and isolation (eg repair of orofacial clefts)

Rehabilitation therapies improve activities of daily living and helps the child and caregivers (like occupational therapy for Down syndrome)

Access to assistive devices (like hearing aids) improves opportunities in life

Remember…A child with a disability is a child

Put the child before the disability

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References: Lemacks J, Fowles K, Mateus A, Thomas K (2013) Insights from parents about caring for a child with birth defects. Int J Environ Res Public Health 10(8):3465–3482; Poley, M. J., Brouwer, W. B., van Exel, N. J., & Tibboel, D. (2012). Assessing health-related quality-of-life changes in informal caregivers: an evaluation in parents of children with major congenital anomalies. *Quality of life research* *21*(5), 849–861. https://doi.org/10.1007/s11136-011-9991-7

1.3 Impact of congenital disorders on children, their parents/caregivers

Untreated and chronic conditions impact children and families

Children

- Poor or suboptimal physical, language, cognitive, or psychosocial development
- Psychosocial distress caused by the cosmetic effect of the disorder (eg orofacial cleft)
- Physical consequences of medical complications
- Increased risk of premature mortality or reduced life expectancy,
- Maltreatment (emotional, physical, sexual abuse, bullying)

→ Poor physical, mental health

→ Perception of stigma, isolation

Caregivers

- Economic hardship
- Anxiety, concern about future
- Guilt, sadness
- Physical consequences of caregiving
- Family relationships, marital relationships, Family/sibling neglect, gender-related issues,

Pregnant women

Detection of a malformation during pregnancy, or a stillbirth or miscarriage causes distress to pregnant women and prospective parents.

Compared with children without disabilities, children with disabilities are:

- 34 per cent more likely to be stunted
- 26 per cent more likely to be wasted
- 53 per cent more likely to have symptoms of acute respiratory infection
- 25 per cent less likely to receive early stimulation and responsive care
- 25 per cent less likely to attend early childhood education
- 16 per cent less likely to read or be read to at home
- 42 per cent less likely to have foundational reading and numeracy skills
- 49 per cent more likely to have never attended school
- 47 per cent more likely to be out of primary school
- 33 per cent more likely to be out of lower-secondary school
- 27 per cent more likely to be out of upper-secondary school
- 32 per cent more likely to experience severe corporal punishment
- 41 per cent more likely to feel discriminated against
- 51 per cent more likely to feel unhappy
- 20 per cent less likely to have expectations of a better life

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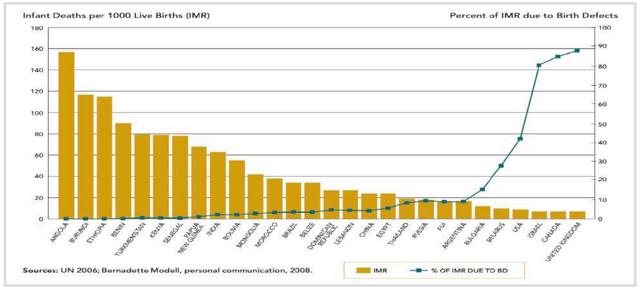
Reference: . Kar, A. (2021). Birth defects stigma. *Birth Defects in India: Epidemiology and Public Health Implications*, 317-333.
Unicef seen, counted, included https://www.google.com/url?sa=t&rct=j&q=&esrc=s&source=web&cd=&ved=2ahUKEwihmO-qw_iEAxVDslYBHewbCOsQFnoECB4QAQ&url=https%3A%2F%2Fdata.unicef.org%2Fwp-content%2Fuploads%2F2022%2F12%2FDisabilities-Report_11_30.pdf&usg=AOvVaw0lwJgBWMXRRfUiSi-_DmBz&opi=89978449

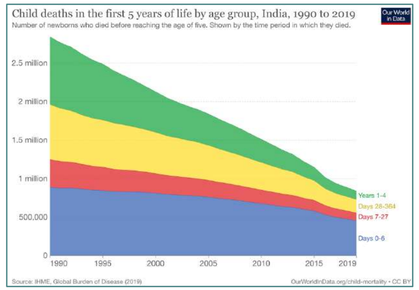
1.4 Global epidemiology of congenital disorders

(a)



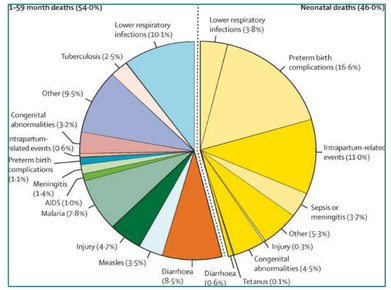
- As infant mortality caused by common infectious conditions of childhood (eg diarrhea, pneumonia) decline, the proportion of deaths caused by congenital disorders increase.[1] (a)

(b)



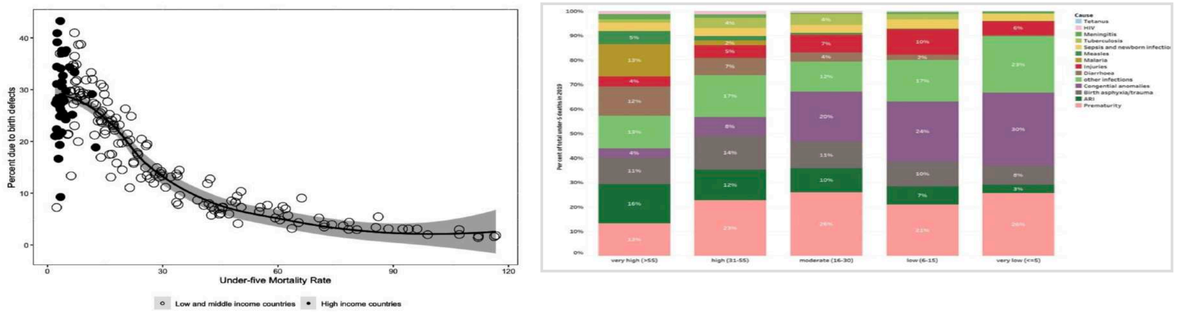
- Child mortality has declined by 53% between 2000 and 2019,[2] brought about by declines in infectious causes of child death. (b)

(c)



- In 2019, congenital anomalies are the fourth leading cause of neonatal mortality globally (c)

(d)



- In 2019, congenital disorders were responsible for an estimated 404 000 deaths of children under five years of age [3] The proportion of under-5 mortality attributable to birth defects has increased from 4.6% in 2000 to 7.6% in 2019.[3]
- The percent of deaths attributable to congenital anomalies is higher in high income countries, where under-5 mortality is low (d,e)[4] Congenital anomaly attributable mortality is lower in LMICs where child mortality from other causes are high (mortality in high income countries 27.7%, as compared to 7.4% in LMICs). [3]

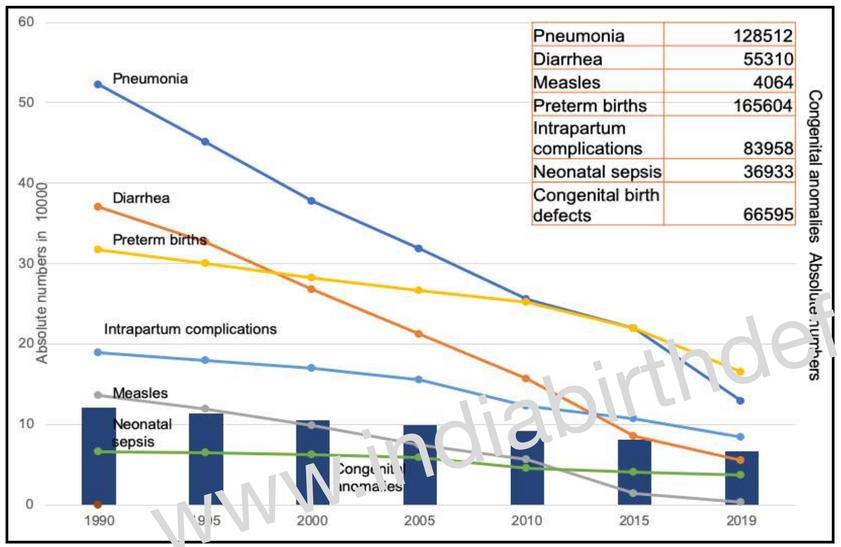
- ***In absolute numbers however, mortality in LMICs was as much as 27 times higher than that reported in high income countries.***[3]

References
1. Christianson, A., & Modell, B. (2004) *Ann Rev Genomics Human Genet* ; *5*: 219–265.
2. Perin, J. et al. (2022) *Lancet Child & Adolescent Health*, *6*(2), 106-115
3 Perin, J. et al. (2023) *BMJ Open*, 13(1)
4. World Health Organisation Congenital disorders. Available at https://www.who.int/news-room/fact-sheets/detail/birth-defects

1.5 Congenital disorders : Epidemiological considerations

1. In India, there has been a 53% decline in child mortality, primarily due to control of infectious diseases. [1] Neonatal mortality due to preterm birth, intrapartum complications, neonatal sepsis are also declining[2,3]. In this situation, the proportion of mortality caused by congenital disorders increase.

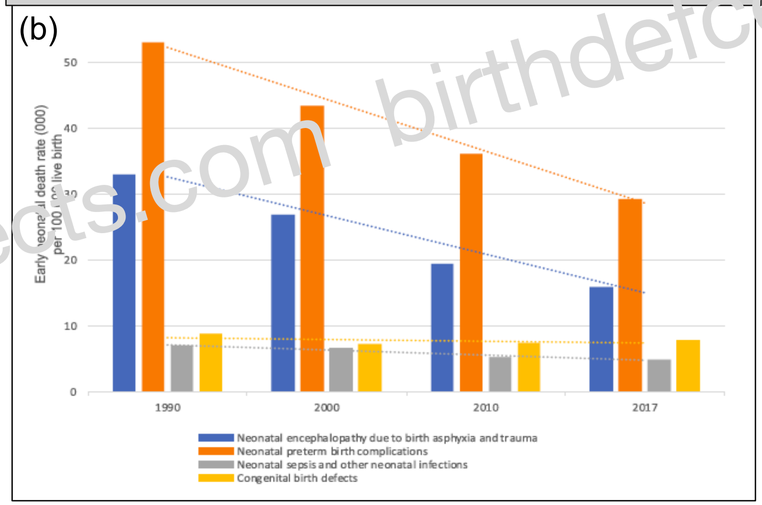
2. There have been significant declines in the major causes of child mortality in India[2]



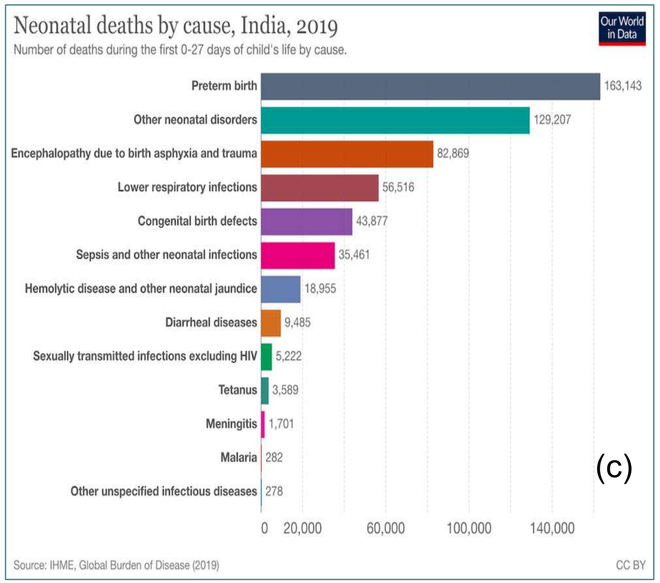
Pneumonia	128512
Diarrhea	55310
Measles	4064
Preterm births	165604
Intrapartum complications	83958
Neonatal sepsis	36933
Congenital birth defects	66595

3. The decline in deaths attributable to congenital disorders has shown a minimal decline of 15%[2]
(b)Proportion of mortality caused by conditions like neonatal sepsis is lower than that of congenital disorders[3]

(b)



4. Although congenital disorders are reported to be the fifth largest cause of neonatal mortality in India[2], this ranking is based on aggregated data from across India. Congenital disorders may account for higher proportionate mortality in states where NMR is low, such as Goa and Kerala [3]



(c)

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1. Perin, J. et al. (2022) *The Lancet Child & Adolescent Health*, *6(2)* :106-115,
2. Dandona R et al. (2020) *The Lancet*; *395*:1640-1658.
3. Ujagare D & Kar A (2021) *J Comm Genet ; 12*(1): 81–90.

1.6 Essential public health activities needed to address congenital disorders (Birth defects service)

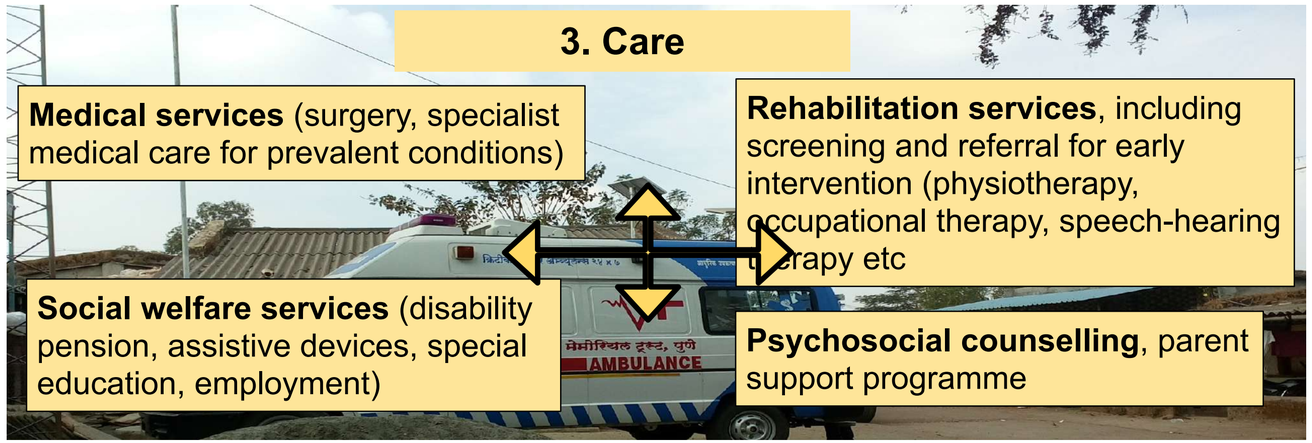
1. Surveillance (Systematic collection of data)

- through routine maternal and child health data reporting systems
- Or through hospital based surveillance systems

2. Prevention

Preconception, antenatal period

- Folic acid supplementation/fortification programmes , rubella immunization
- Awareness programmes about teratogens, behavioral modifications, optimization of body weight
- Screening and management of high risk pregnancies
- Prenatal and genetic screening/testing services
- Genetic services for prevalent genetic disorders



3. Care

Medical services (surgery, specialist medical care for prevalent conditions)

Rehabilitation services, including screening and referral for early intervention (physiotherapy, occupational therapy, speech-hearing therapy etc

Social welfare services (disability pension, assistive devices, special education, employment)

Psychosocial counselling, parent support programme

4. Health workforce, infrastructure, supplies

Developing requisite competency of health service staff; (for diagnosis, referral, management); development of infrastructure and supplies

5. Creating intersectoral linkages, referral pathways

with welfare services for children with disabilities, education, employment opportunities, legal rights

6. Community outreach

- Awareness about children with birth defects and disabilities,
- Disability sensitization
- Awareness about treatment, early intervention services
- Support for community based organizations

7. Regulations

Regulation of foeto-toxic agents, regulations to ensure evidenced care for children

8. **Research** for developing evidence-based birth defects service

Reference: 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

1.7 Prevalence and Recurrence Risk

Prevalence – Total number of cases present at a particular point of time. Expressed as a rate (either birth prevalence (total births i.e. stillbirths and livebirths), live birth prevalence, or population prevalence).
Recurrence risk – the probability that the disorderin a family member will affect (recur) in other family members.

1 Congenital anomalies

Condition	Prevalence	Estimated absolute births* (India)	Recurrence Risk
Congenital heart defects	9.1 per 1000 live births	239 999	2-5% in isolated cases, 50% for autosomal dominant inheritance, associated with several chromosomal anomalies like DS
Orofacial Clefts (Isolated)	1 in 700 births, ethnic variation	37 676	Increased 32 times for cleft lip, 56 times for cleft palate among siblings
Spina bifida	1 in 1000 births, ethnic variation	26 374	Increases by 20 -50% above the general population
Clubfoot	1 in 1000 live births, ethnic variation	26 374	Increases by 2-3% when one parent is affected, 15% when both parents are affected
Developmental dysplasia of hip	1-2 per 1000 through physical examination, increases to 5-30 per 1000 with ultrasound screening of hips	52 747	Positive family history increases the risk by 12 fold, risk to sibling of affected child is 6%, if parent has DDH 12% 36% for subsequent pregnancy if both parent and child have DDH
Congenital limb defects	4 per 10,000 for upper limbs and 2 per 10,000 for lower limbs	10 549, 5275	Recurrent risk if on monogenic origin
Congenital cataract	0.63 – 9.74 per 10,000 births	25688	

*Annual births India – 26 373 560 (2018)

2 Developmental disabilities

Condition	Prevalence	Estimated absolute births* (India)	Recurrence Risk
Down syndrome	1 in 800 births, risk increases with increasing maternal age	32 967	Older women at increased risk
Attention-deficit hyperactivity disorder	7.2% prevalence		Siblings at 5 times increased risk
Autism spectrum disorders	1:2% 3-4 times higher in boys		High, 50-100 times higher for subsequent pregnancies
Cerebral palsy	1.5 – 2.5 per 1000 live births, 3 per 1000 live births in 4-48 year age group	65 934	
Congenital hearing loss	1.33 per 1000 live births for congenital bilateral permanent hearing loss	35 077	18% probability for deafness in children for hearing couple with one deaf child and no family history

3 Genetic conditions

Condition	Prevalence	Estimated absolute births* (India)	Recurrence Risk
Sickle cell anemia	1-5 % of the global population, variation in prevalence by regions and ethnicity		25% risk of affected birth for each pregnancy and 50% risk of carrier
Duchene and Becker muscular dystrophies	1/3300 (DMD) 1/18,000-1/31,000 (BMD) males		X – linked recessive inheriteance
Achondroplasia	1 in 25,000 – 30,000 live births	1055	Autosomal dominant, 0.23% (1 RR per 443 in siblings of a sporadic case)
Hemophilia	1 in 5000 (hemophilia A) 1 in 30,000 (hemophilia B) male births	5275, 879	X – linked recessive inheritance

Prevalence rates and recurrence risk data are mostly from high income countries

Reference in Kar A Some common birth defects. In Birth Defects in India Epidemiology and Public Health Implications; Springer Singapore, 2020 (provides references to articles from which this data is collated

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Drawing attention to the global health issue of birth defects, childhood disability and public health in low and middle income countries

1.8 Birth defects Surveillance

1. What is birth defects surveillance?

•Surveillance is defined as 'the ongoing, systematic collection, analysis and interpretation of data essential to planning, implementation and evaluation of public health practice'

2. Why is surveillance important?

Data from surveillance systems are critical for
- Monitoring the burden of different types of birth defects
- Detecting changes over time and place including outbreaks
- Determining maternal risk factors and identifying at-risk populations
- Evaluation of prevention programmes
- Planning and advocacy for public health actions
- Guiding research and policies on the broader social and economic impacts of birth defects beyond the health domain

3. How is data on birth defects reported?

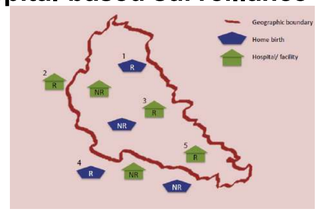
•Reported as prevalence estimates: birth prevalence and/or live birth prevalence reported as total number of affected fetuses/neonates per 10 000 births/live births
•True incidence of congenital anomalies cannot be measured

4. What factors affect the reporting of prevalence data?

•Type of surveillance system: population or hospital-based surveillance system
•Classification and coding of anomalies: ICD-10
•Inclusion of pregnancy outcomes - live births, stillbirths and/or terminations of pregnancies
•Age at diagnosis – prenatal, at birth, during childhood
•Case ascertainment: active/passive, sources and inclusion criteria

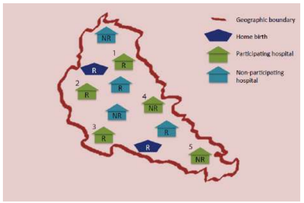
5. What are the types of surveillance systems for birth defects?

(2) Hospital-based surveillance



•Five cases occurring only at participating hospitals are registered, irrespective of whether the mother is a resident (R) or a non-resident (NR)

(1)Population-based surveillance



•Five cases registered are only of resident mothers (R), even if the birth has occurred outside the geographic area under surveillance. Data of non-resident mothers (NR) are not included

Situationally appropriate birth defects surveillance systems have not been established in most LMICs so that there is limited data on the magnitude and types of birth defects in these settings

References: Bhide, P. (2021). Birth Defects Surveillance in India. In: Kar, A. (eds) Birth Defects in India. Springer, Singapore. https://doi.org/10.1007/978-981-16-1554-2_4

Contributed by Dr Prajkta Bhide, PhD