Section: Anatomy (Embryology)

Review Article

AN OVERVIEW OF BURDEN OF DEATH DUE TO CONGENITAL ANOMALIES IN INDIA

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ABSTRACT

Background: Congenital anomalies are a significant but often overlooked cause of morbidity and mortality in India. These birth defects ranging from structural to functional abnormalities contribute notably to neonatal and under-five deaths, especially in low and middle income countries. Objective of this study is to assess the burden, mortality patterns and regional variations of congenital anomalies in India and to highlight actionable strategies for prevention, early detection and management.

Materials and Methods: This review synthesizes national and regional data from published studies, reports from the World Health Organization and Government health surveillance documents. Mortality statistics by anomaly type and age group were analyzed using data from 2015 to 2021. Regional prevalence patterns were compiled from hospital-based studies conducted across different Indian states.

Results: Among congenital anomalies, highest mortality observed in children under five years of age. Congenital heart anomalies were the leading cause, followed by neural tube defects and chromosomal disorders such as Down syndrome. Survival rates vary widely depending on the condition, with Down syndrome showing high survival and congenital heart diseases showing lower survival within the first year of life.

Conclusion: Congenital anomalies remain a major public health concern in India. Strengthening antenatal screening, neonatal diagnosis, birth defect surveillance, and health worker training are critical to reducing the associated mortality and long-term disability. Public awareness, integrated care systems, and national level registries are essential to address the current gaps in prevention and management.

Keywords: Congenital anomalies India, Congenital heart defects, Neural tube defects, Birth defect surveillance, Antenatal screening.

INTRODUCTION

Congenital anomalies are of antenatal origin due to abnormalities in embryogenesis. They may appear before birth, just after birth or late in life. They are structural or functional.[1] Premarital counselling is strongly recommended, particularly for couples who are closely related or have a family history of congenital anomalies. Such counselling provides vital information about the increased risks associated with consanguineous marriages, including a higher likelihood of genetic disorders in offspring. By understanding these risks, couples can make informed decisions, potentially reducing the incidence of inherited conditions.[2] Over 90% of infants with serious birth defects are born in low- and middle-income countries, where access to adequate antenatal diagnostics and postnatal care services is often lacking.[3] In contrast, in high income countries, affected individuals often undergo palliative interventions and continue to survive with long-term functional impairments.[4] Though uncommon, birth defects pose a serious public health challenge due to their collective impact.

Many are entirely preventable with timely primary prevention. Examples include congenital rubella syndrome (avoided via vaccination), neural tube defects like spina bifida and anencephaly (preventable with folic acid), fetal alcohol syndrome (avoided by abstaining from alcohol), Down syndrome, and Rh hemolytic disease.[5]

Impact and importance of problem - Every time globally, within 28 days of birth, 240,000 of newborn die due to birth defects. Birth defects are responsible for a further 170,000 deaths of children between the periods of 1 month and 5 years. Birth defects not only responsible for tremendous drain on natural resources especially in the low-income countries but also there is a significant impact on individuals, families and society.[6] Birth deformations are caused by multiple factors. Both inheritable and environmental factors can play a part in causing birth defects. Causes of birth defects include chromosomal diseases (1in 263 births) and single gene diseases (1 in 81 births). Environmental factors include nutritive scarcities, infections, medical conditions of mother, alcohol and teratogenic agents.[7] A newborn may be with single birth defect or with multiple birth defects and these are of major or minor clinical significance. Single minor defects are present in roughly 14% newborns. Major defects are much more common in early embryos (10 – 15%).[8] A recent global burden of disease report ranked birth defects as the 17th leading cause of disability-adjusted life years (DALYs), accounting for approximately 39 million DALYs worldwide.[9] A recent report highlights that approximately 5,400 to 5,700 infants are born with Down syndrome in the U.S. each year which is approximately 1 in 700 births. In low- and middle-income countries, higher rates persist due to limited prenatal screening and later-age pregnancies, resulting in roughly double the incidence compared to developed nations.[10,11] This is in contrast to countries with effective family planning promotion tend to have earlier and more evenly spaced pregnancies, which significantly reduces the incidence of births among women of advanced maternal age.[12]

Aim of this review is to assess the burden, mortality patterns, and regional variations of congenital anomalies in India. By comparing the burden of different congenital anomalies in different age groups, we can help to develop strategies for prevention, early detection and management of birth defects which can predict the future need of the national surveillance programme and to highlight actionable strategies for prevention.

MATERIALS AND METHODS

This review used publicly available data from WHO reports, National Health Mission databases, and hospital-based studies from 2010 to 2023. Literature was sourced from PubMed and Google Scholar using terms like ‘congenital anomalies India’, ‘birth defects mortality’, and ‘neonatal malformations. All databases were searched from inception. From the searches we reviewed the title and abstract of each paper and retrieved potentially relevant references.

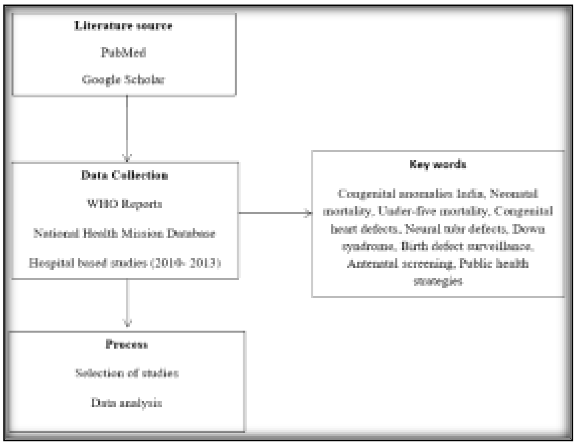

Figure 1: Flow of study.

Global Burden of deaths due to congenital anomalies: According to WHO (2016), birth defects are structural or functional anomalies that occur during intrauterine life and can be linked prenatally, at birth or later in life. An estimated 240,000 neonates die worldwide within 28 days of birth every year due to congenital malformations. Birth defects account a further 170,000 deaths of children between the periods of 1 month and 5 years. Birth defects can contribute to long term disability which poses a significant impact on individuals, families, health care systems and societies. Nine of ten children born with a serious malformations are in low- and middle-income countries. As neonatal and under-5 mortality rates decline, birth defects form a larger proportion of the cause of neonatal and under-5 deaths. The most common severe birth defects are heart malformations, neural tube defects and Downs syndrome.[6] World wide about 246000 cases of Folic acid-preventable spina bifida and anencephaly (FAPSBA) occur annually.[10]

Bell and Oakley have estimated that approximately 75% of neural tube defects (NTDs), including spina bifida and anencephaly, are preventable through adequate folic acid intake. Despite this, global prevention efforts have been insufficient. In 2006, only about 9% of FAPSBA cases were being prevented which increased to 15% in 2012.[13] This underscores a significant gap in global public health initiatives, as the majority of preventable NTD cases continue to occur due to the lack of widespread implementation of effective folic acid fortification strategies.[14] There is an urgent need for those countries with the majority of these cases, such as India,[15] and China,[16,17] to implement mandatory folic acid fortification policies. A comprehensive systematic review and meta-analysis covering perinatal births across China between 2000 and 2021 found that the prevalence of birth defects is 208.9

per 10,000 births . This highlights the critical need to expand access to genomic testing at all stages whether it is preconception, prenatal, or neonatal, to better identify and manage congenital anomalies.[18]

National burden of deaths due to congenital anomalies-

Congenital anomalies constitute the fifth largest cause of death in the neonates in our country.[19] India would account to an estimated 1.7 million babies (6% of all births) born with congenital defects annually (July 2023).[20]

In India, congenital anomalies represent a significant burden with varying incidence rates and survival outcomes. Central nervous system anomalies such as spina bifida and encephalocele occur in approximately 0.5% of live births, with survival chances ranging between 72% and 90% in the first year of life. Cardiovascular anomalies are similarly prevalent, with critical congenital heart diseases (like tetralogy of Fallot and transposition of the great arteries) affecting 0.5% of births and survival rates in the first year of life between 55% and 85%, while common heart defects like atrial and ventricular septal defects generally have survival rates comparable to the general population. Orofacial defects, including cleft lip and palate, have a slightly higher incidence of 0.7%, with survival rates of 92% for cleft lip and 91% for cleft palate in first year of life. Gastrointestinal anomalies seen in 0.4% of births, include conditions like tracheoesophageal fistula and anal anomalies, with first year survival rates of 85% and 87%, respectively. Musculoskeletal defects such as congenital diaphragmatic hernia and omphalocele also have an incidence of 0.7%, with first-year survival ranging from 71% to 90%. Chromosomal abnormalities, notably Down syndrome, are observed in 0.4% of live births, with a relatively high survival rate of 94% in the first year of life.[20]

Congenital heart anomalies and neural tube defect are most common cause of deaths among congenital anomalies. [Table 1].[6]

RESULTS

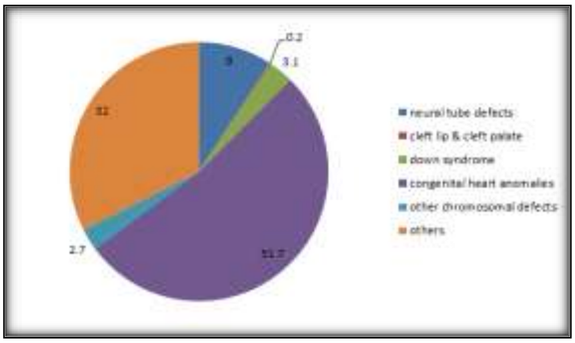

Figure 2: Mortality in all ages in per 1000 deaths due to various congenital anomalies in India (2021).[6]

Table 1: Mortality in all ages per 1000 death due to various congenital anomalies in India.[6]

Congenital anomalies	**Neural tube defects**	**Cleft lip & cleft palate**	**Down syndrome**	**Congenital heart anomalies**	**Other chromosomal defects**	**Others**
2015	10.1	0.4	2.8	54.3	2.6	31.9
2019	9.3	0.6	2.0	37.4	2.1	31.4
2021	9.0	0.2	3.1	51.7	2.7	32.0

Approximately, 1% of deaths among all cause deaths were due to congenital anomalies. It ranges from 0.9% to 1.13%. [Table 2].[6]

Table 2: Mortality in India due to congenital anomaly.[6]

Year	**Population of India**	**Total all cause death in all ages (in thousand)**	**Deaths due to congenital anomaly (in thousand)**
2015	1328024	8976.3	102.1(1.13%)
2019	1366418	9171.3	82.8(0.9%)
2021	1389030	9296.5	98.8(1%)

[Table 3] shows that majority of deaths occur in the 0-4 years age group, with congenital heart anomalies (43.3) being the leading cause, followed by "Others" (27.5) and neural tube defects (8.6). The number of deaths decreases sharply in the 5-14 years age group, with congenital heart anomalies (3.4) remaining the most prevalent. The 15-29 years and 30-49 years age groups show a further decline, with congenital heart anomalies (1.8 and 2.1 respectively) still being the leading cause, followed by "Others" (0.9 and 1.4). Deaths due to congenital anomalies become negligible in people aged 50 years and above, with almost no recorded cases beyond 70 years. Across all age groups, congenital heart anomalies are the most common cause of death due to congenital conditions.[6]

Table 3: Number of deaths (in thousand) in India in various age groups due to congenital anomalies in 2021.[6]

Death due to congenital anomalies in 2021 (in thousand)						
Age group	**Neural tube defects**	**Cleft lip & cleft palate**	**Down syndrome**	**Congenital heart anomalies**	**Other chromosomal defects**	**Others**
0-4years	8.6	0.2	1.9	43.3	2.7	27.5
5-14 years	0.1	0	0.1	3.4	0	1.3
15-29 years	0.1	0	0.1	1.8	0	0.9
30-49 years	0.1	0	0.5	2.1	0	1.4
50-59 years	0	0	0.3	0.4	0	0.4
60-69 years	0	0	0.1	0.6	0	0.3
>70years	0	0	0	0	0	0

Table 4: Common congenital anomalies reported by various regional studies in India.[7,21-27]

Study Name	**Study period**	**Type of Study**	**Place**	**Number of births**	**Number of anomaly**	**Birth defect prevalence percentage**	**Common anomalies**
Qurieshi M A et al. (2016).[21]	2016	Cross-sectional descriptive, hospital based study	Kashmir	1129	17	1.5	Central nervous system (29.41%)
							Cardiovascular system (17.64%)
							Genitourinary system (17.64%)
Narayan J, Narayan S et al. (2019).[22]	1990-2018	Retrospecti-ve, hospital based study	Rajasthan	241,848	6623	2.7	Cardiovascular system (21.65%)
							Central nervous system (19.95%)
Kumar J et al. (2021).[23]	1998-2017	hospital based study	Chandigarh	86,850	1578	1.8	Circulatory system (28.0%)
							Musculoskeletal system (18.6%)
							Urinary system (14.3%)
Jayasree S, Smitha D'Couth (2018).[24]	2009-2015	Cohort study	Kerala	1,08,024	911	0.84%	Urinary system (29.75%)
							Musculoskeletal system (24.15%)
							Nervous system (21.73%)
Nayak V, Bharathi A et al. (2023).[7]	2016-2019	hospital- based observation-al study	Karnataka	11,469	77	0.67%	Craniospinal system (53.2%)
							Musculoskeletal system (19.5%)
							Multiple anomalies (14.3%)
Pagolu KR, Tamanam RR. (2022).[25]	2016-2020	District early intervention centre	Andhra Pradesh	26,423	962	3.6	Congenital Deafness (22.16%)
							Cleft Lip & Palate (22.04%)
							Club Foot (21.93%)
Padmanabhan R et al. (2019).[26]	2017	Hospital records	Tamil Nadu	2132	87	4.018%	Cardiovascular system (35.6%)
							Musculoskeletal system (26.4%)
							Genitourinary system (13.8%)
							Multiple anomalies (14.3%)
Rao B et al. (2014).[27]	2008-2012	Birth registry & hospital records	Karnataka	28373	344	1.21%	Central nervous system (33%)
							Cardiovascular system (24%)
							Gastrointestinal system (17%)

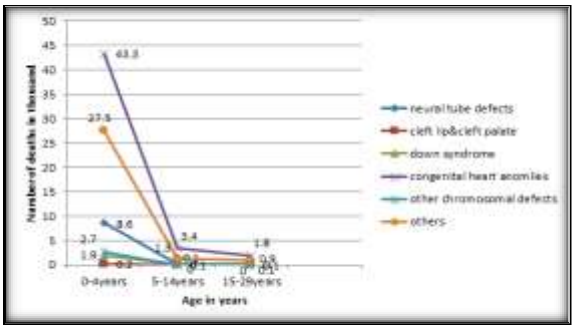

Figure 3: Number of deaths in thousand in India in age group (0–29) years due to congenital anomalies in 2021.[6]

DISCUSSION

Congenital birth anomalies are an important public health problem, and this problem is not decreasing in India. Many studies conducted across India have demonstrated the increased trend of birth defects in the country. In 70% of states classified under the high and middle Socio-demographic Index (SDI) category, birth defects emerged as the third leading cause of neonatal deaths. This highlights the urgent need to establish dedicated services for the management and prevention of birth defects across the country. The analysis also underscores the value of modelled estimates from the Global Burden of Disease study as the most reliable data currently available for understanding the epidemiology of birth defects in low and middle income countries.[28] Keeping in mind the currently observed gaps, the following strategies are proposed-

Health system strengthening: Routine antenatal screenings such as ultrasounds, blood tests and genetic screenings are crucial for the early detection of birth defects. Implementing neonatal screening programs in hospitals can help detect birth defects soon after delivery, ensuring early interventions. While some states have initiated newborn screening programs, there is a need to scale these efforts nationwide. The wide variation in incidence of birth defects may be attributed to geographical, racial, and ethnic differences, variations in survey methodologies, and other contextual factors. Additionally, hospital-based data are often inadequate and may not accurately reflect the true incidence of birth defects within the community.[2] So, coordination is very important among all level of health care.

Capacity building of human resources - Widespread health education can play a pivotal role in raising awareness about preventable causes of fetal malformations and promoting timely interventions.[2]

Strengthening the capacity of healthcare workers, particularly in primary health centres, is critical for improving the detection of birth defects. Training programs focusing on prenatal care, early diagnosis, and referral systems can ensure that even the most remote regions are prepared to address congenital anomalies. Regular training of doctors at PHC as they are the first person of contact for patients so that they can diagnose the birth defect & refer to higher centres. There should be more number of centres with genetic counsellors so that chances of birth defects can be predicted in case of positive family history.

Awareness among parents: The primary risk factors associated with congenital malformations included consanguineous marriage, adverse drug exposure, substance abuse, diabetes mellitus and polyhydramnios. Ensuring the provision of periconceptional vitamins and folic acid supplementation to all women of reproductive age is essential for prevention.[2] Avoiding exposure to harmful environmental agents such as heavy metals, pesticides, and certain medications during pregnancy is crucial for reducing the risk of congenital anomalies.[29] Educating parents, especially mothers, about the importance of regular check-ups, vaccinations (like the rubella vaccine to prevent congenital rubella syndrome), and nutrition during pregnancy can reduce the risk of birth defects.

Surveillance: Most children born with major congenital malformations who survive infancy often experience physical, cognitive or social impairments, and may face an elevated risk of morbidity from associated health conditions. Therefore, implementing primordial and primary prevention strategies is essential to reduce both the incidence of congenital malformations and the burden of related morbidity.[2] Surveillance at the state and district levels, including secondary and primary level, will help to utilize the data for preparation of policy for the state and district levels.

Operational Research: Collaboration between government, Non-Government Organisations and the private sector can help in the development of innovative solutions and ensure that healthcare reaches the most marginalized communities.

All the neonates should be examined with scrutiny for overt as well as occult congenital anomalies and Paediatric surgical care should be considered as an essential component of child health programmes in developing populations. There is regional variability in diagnostic criteria and reporting so it is necessary to establish a registry system for congenital anomalies.[29] Encouraging research into the causes, prevention and treatment of birth defects in the Indian context can provide more tailored solutions to the problem.

Rashtriya Bal Swasthya Karyakram is a Government of India initiative to diagnose and intervene in four critical problems in children less than 18 years they are Defects, Deficiencies, Developmental Delays & Diseases.[30]

Among the 1% of live-born infants affected by congenital heart disease, contributing to approximately 10% of overall infant mortality, fewer than 2% undergo life-saving surgical intervention.[31] This highlights the urgent need for a parallel program focused on capacity building to strengthen diagnostic, surgical, and post-operative care services.[32]

CONCLUSION

Congenital anomalies contribute to approximately 1% of all deaths in India (2021), with 98.8 thousand deaths, making it a significant public health concern. In India, congenital birth defects are emerging as a significant public health issue, contributing to infant mortality, disability, and long-term morbidity. Birth to four years age group bears the highest burden of mortality due to congenital anomalies, particularly: Congenital heart anomalies (43.3%), neural tube defects (8.6%) and other unspecified anomalies (27.5%). Congenital heart defects are the leading cause of deaths across all age groups, especially in early childhood. Neural tube defects and Down syndrome follow with lesser but significant contributions. India reports an estimated 1.7 million babies born annually with congenital anomalies, constituting ~6% of all births (July 2023 estimate). Prevalence rates in Indian hospital-based studies range from 0.67% to 4.01%. Common anomalies vary regionally- cardiovascular, neural tube, musculoskeletal and genitourinary defects predominate. Mortality sharply declines in older age groups, becoming negligible after age 50. Survival rates within the first year of life differ: Cleft lip: 92%, Cleft palate: 91%, congenital heart diseases: 55%–85%, CNS anomalies: 72%–90% and Down syndrome: 94%. India has made some progress in addressing the issue of birth defects. Key programs include Rastriya Bal Swasthya Karyakram, Janani Suraksha Yojana and National Health Mission. Despite the efforts, there are several challenges in creating awareness and preparedness for birth defect identification in India. Rastriya Bal Swasthya Karyakram is a Government of India initiative to diagnose and intervene four critical problems in children less than 18 years they are Defects, Deficiencies, Developmental Delays & Diseases (four "D"s). Under defects, Neural tube defect, Downs Syndrome, Cleft Lip & palate, Developmental dysplasia's of hip, Congenital cataract Congenital deafness, Congenital Heart Disease and Retinopathy of prematurity are to be screened.

Many birth defects are preventable with improved maternal health, folic acid supplementation, and adequate antenatal care. Awareness campaigns focusing on maternal nutrition, vaccination and avoidance of harmful substances like alcohol and tobacco can reduce the incidence of birth defects. Awareness helps parents and healthcare providers recognize symptoms early. Conditions like congenital heart defects or cleft lip can be identified early in pregnancy through routine ultrasound screenings, ensuring timely intervention and better outcomes. Among the 1% of live-born infants affected by congenital heart disease, contributing to approximately 10% of overall infant mortality, fewer than 2% undergo life-saving surgical intervention. This highlights the urgent need for a parallel program focused on capacity building to strengthen diagnostic, surgical, and post-operative care services. Raising awareness about the role of genetics in birth defects can help families seek genetic counseling, particularly if there is a family history of congenital anomalies. This can help with family planning and inform decisions on prenatal testing. To address birth defects effectively, preparedness at multiple levels—individual, community and healthcare system is essential.

Limitations: Since it's a review, interpretation, and synthesis of results can be subjective. The lack of a national registry limits comprehensive burden estimation. Hospital-based studies may underrepresent rural and home births.

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