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Original Research Article

Burden and Spectrum of Congenital Malformations in Tertiary Health Facilities: A Retrospective Multicenter Evaluation

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Abstract:

Background: Congenital anomalies are among the leading causes of perinatal morbidity and mortality worldwide. Their etiology is multifactorial and they pose lifelong challenges to affected children, families, and healthcare systems.

Objective: To determine the burden, incidence, and pattern of congenital malformations among deliveries in tertiary care hospitals.

Methods: A retrospective observational study was conducted across two tertiary hospitals in Srinagar and New Delhi—Lal Ded Hospital, Government Medical College, Lady Hardinge Medical College—from January to June 2024. Medical records of all deliveries beyond 24 weeks of gestation were reviewed. Neonates and stillbirths with structural congenital anomalies were included. Data were analyzed for incidence, system involvement, and outcomes.

Results: Among 17,200 deliveries, 412 neonates had congenital anomalies, yielding an incidence of 2.4% (24 per 1,000 live births). Musculoskeletal anomalies were most common (31%), followed by central nervous system defects (23.8%), cardiovascular (15%), genitourinary (12.1%), respiratory (7%), orofacial (5.8%), and abdominal wall/gastrointestinal anomalies (3.1%). Neural tube defects, including myelomeningocele and anencephaly, predominated among CNS malformations. Live births accounted for 62% of cases, stillbirths 30%, and 8% were medically terminated.

Conclusion: Congenital anomalies remain a significant contributor to perinatal morbidity and mortality in northern India. Musculoskeletal and neural tube defects were the leading types. Strengthening prenatal screening, folic acid supplementation, and establishing regional anomaly registries are crucial to prevention and early management.

Keywords: Congenital Anomalies, Neural Tube Defects, Musculoskeletal Malformations, Perinatal Outcome.

Introduction

Congenital malformations, or birth defects, are structural or functional abnormalities that arise during fetal development and are present at birth. These conditions remain a significant public health concern because they contribute substantially to perinatal and infant deaths worldwide. According to the WHO, congenital anomalies occur in about 2–3% of all live births and account for hundreds of thousands of neonatal deaths every year. In developing countries, including India, the impact is particularly severe due to limited antenatal screening, late diagnosis, and inadequate neonatal care facilities. These defects range in severity from minor cosmetic deformities to major malformations that can lead to stillbirth or lifelong disability. Beyond the medical consequences, congenital anomalies have deep psychological, social, and economic implications for families and place a considerable burden on healthcare systems.

The causes of congenital anomalies are complex and often multifactorial. They may result from genetic or chromosomal abnormalities, environmental exposures, maternal illnesses, nutritional deficiencies, or a combination of these factors. Maternal infections such as rubella or cytomegalovirus, uncontrolled diabetes, use of teratogenic drugs, alcohol intake, and poor maternal nutrition are recognized risk factors. One of the most important preventable causes is folic acid deficiency, which is closely associated with neural tube defects (NTDs) such as anencephaly, spina bifida, and encephalocele. In many developed nations, mandatory folic acid supplementation has led to a marked decline in the incidence of NTDs.

However, in India, irregular antenatal care and lack of awareness about preconceptional supplementation continue to result in a higher burden. Other frequently encountered congenital anomalies involve the musculoskeletal, cardiovascular, genitourinary, gastrointestinal, and respiratory systems, as well as orofacial clefts. The distribution and type of these defects vary across different geographic regions, reflecting differences in genetic makeup, environmental influences, and healthcare access.

Several hospital-based studies from India have attempted to estimate the prevalence and pattern of congenital anomalies, but their findings are inconsistent. A recent study by Chikkagowdra et al. (2023) from Karnataka observed an incidence of 7.3 per 1,000 live births, with central nervous system anomalies forming the largest group, followed by cardiovascular and musculoskeletal malformationsJSAFOG-15-725. Other studies have reported musculoskeletal and genitourinary defects as more common, emphasizing that the pattern differs by region and population characteristics. The national pooled estimate stands at around 4.8 per 1,000 births, though tertiary centers often report higher rates because they receive complicated and high-risk pregnancies. Differences in reporting methods, diagnostic capabilities, and study design also contribute to the variation. Reliable multicentric data are still scarce, especially from northern regions such as Jammu and Kashmir, where no large-scale retrospective studies have been undertaken to date. Understanding these regional variations is essential to guide preventive strategies and strengthen antenatal diagnostic programs.

Recognizing this gap, the present retrospective multicentric study was carried out in two major tertiary care hospitals in Srinagar and New Delhi—Lal Ded Hospital, Government Medical College Srinagar, and SSKH, Lady Hardinge Medical College, New Delhi. Together, these institutions handle more than 17,000 deliveries over a six-month period, offering a representative sample for assessing the regional burden of congenital anomalies. The main objectives were to determine the incidence of congenital malformations among total deliveries, analyze their distribution by organ system, and identify common patterns that could inform preventive and management strategies. By providing contemporary data from a high-volume tertiary setting, this study aims to enhance understanding of the current trends in congenital anomalies, encourage community-level preventive measures such as folic acid supplementation, and promote routine antenatal anomaly screening. The findings are also intended to support the establishment of a regional registry for systematic surveillance and multidisciplinary management of affected newborns and their families.

Materials and Methods

Study design: Retrospective multicentric descriptive study.

Study sites:

1. Lal Ded Hospital, Government Medical College, Srinagar
2. SSKH, Lady Hardinge Medical College, New Delhi

Study duration: January – June 2024 (6 months)

Sample size: 17,200 total deliveries (>24 weeks gestation).

Inclusion criteria: All live births, stillbirths, and terminated fetuses beyond 24 weeks of gestation with diagnosed structural congenital anomalies.

Data collection: Medical records and neonatal files were retrospectively reviewed for demographic details, anomaly type, gestational age at diagnosis, and neonatal outcome. Anomalies were classified according to EUSCAT (European Surveillance of Congenital Anomalies) guidelines.

Data analysis: Results were compiled and analyzed using descriptive statistics (percentages and proportions).

Results

A total of 17,200 deliveries were recorded across both tertiary centers — Lal Ded Hospital (LDH) and SSKH, Lady Hardinge Medical College New Delhi — during the six-month study period (January–June 2024). Among these, 412 neonates and fetuses were identified with structural or functional congenital anomalies. The overall incidence was calculated to be 2.4% (24 per 1,000 live births). The distribution of anomalies was analyzed by organ system, pattern, and frequency.

1. Distribution by System Involvement

Musculoskeletal anomalies constituted the largest group, accounting for 31.0% of all cases. Within this category, clubfoot (talipes equinovarus) and polydactyly were the predominant defects. Central nervous system (CNS) anomalies formed the second most common category (23.8%), with neural tube defects (NTDs) such as myelomeningocele, anencephaly, and spina bifida being the most frequent findings. Cardiovascular system (CVS) anomalies represented 15.0% of total defects, primarily septal defects and transposition of the great vessels (TGV).

Anomalies of the genitourinary system accounted for 12.1%, with hypospadias and chordee being the commonest findings. Respiratory system anomalies comprised 7.0%, with congenital diaphragmatic

hernia (CDH) and tracheoesophageal fistula (TEF) identified most frequently. Orofacial clefts contributed 5.8%, where isolated cleft lip was more prevalent than combined cleft lip and palate. A smaller proportion (3.1%) involved gastrointestinal and abdominal wall defects, including anorectal malformations, duodenal atresia, omphalocele, and gastroschisis.

Table 1: Distribution of Congenital Anomalies by Organ System

System Involved	Number of Cases (n=412)	Percentage (%)	Most Common Anomalies Observed
Musculoskeletal	128	31.0	Clubfoot, Polydactyly
Central Nervous System	98	23.8	Myelomeningocele, Anencephaly, Spina bifida
Cardiovascular System	62	15.0	Septal defects, Transposition of great vessels
Genitourinary System	50	12.1	Hypospadias, Chordee
Respiratory System	29	7.0	Congenital diaphragmatic hernia, Tracheoesophageal fistula
Orofacial Region	24	5.8	Isolated cleft lip, Isolated cleft palate
Gastrointestinal & Abdominal Wall	13	3.1	Anorectal malformation, Duodenal atresia, Omphalocele, Gastroschisis
Total	**412**	**100.0**	—

2. Monthly Distribution and Institutional Contribution

Of the total cases, Lal Ded Hospital accounted for 67% (n=276), while SSKH contributed 33% (n=136), corresponding to their relative delivery volumes. On average, 2,000–2,200 deliveries per month occurred at Lal Ded Hospital and 900–1,100 deliveries per month at SSKH. The trend in anomaly detection remained relatively stable across the study period, with slight peaks in March and May 2024, possibly reflecting increased antenatal screening activity during these months.

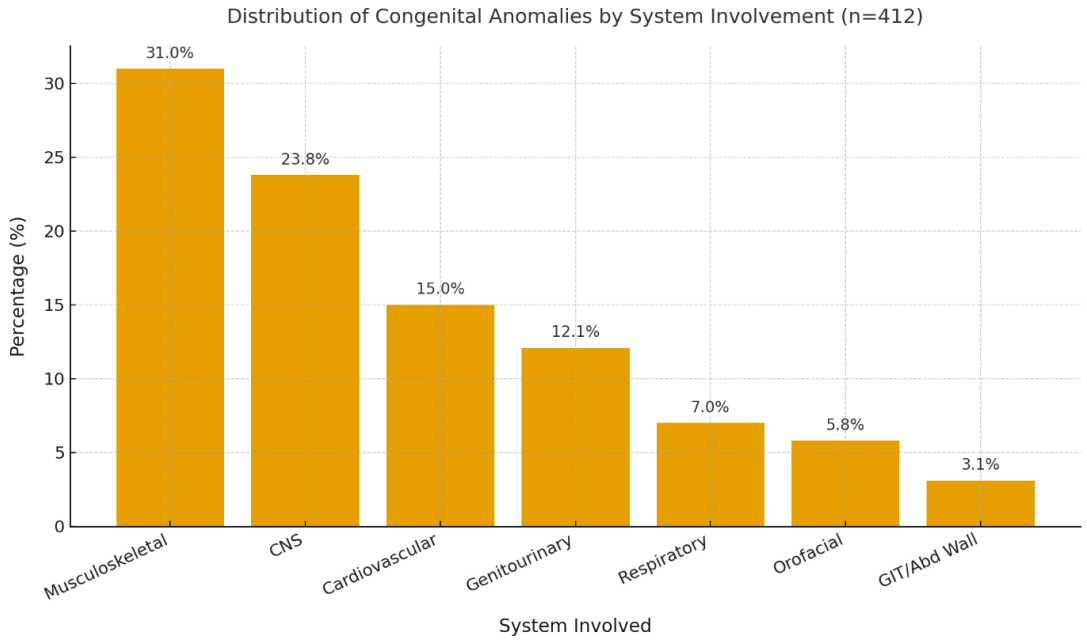

Figure 1: Proportion of Congenital Anomalies by System

3. Pattern of Neural Tube Defects (NTDs)

Among the 98 CNS anomalies, neural tube defects constituted the majority (71.4%). Myelomeningocele (MMC) was the leading lesion, observed in 38 cases (38.8%), followed by anencephaly (22 cases, 22.4%), and spina bifida (10 cases, 10.2%). Other rarer CNS anomalies included hydrocephalus, encephalocele, and agenesis of the corpus callosum.

Table 2: Distribution of Central Nervous System Anomalies

Type of CNS Anomaly	Number of Cases (n=98)	Percentage (%)
Myelomeningocele	38	38.8
Anencephaly	22	22.4
Spina bifida	10	10.2
Hydrocephalus	9	9.2
Encephalocele	7	7.1
Agenesis of corpus callosum / others	12	12.3

4. Associated and Multiple Anomalies

In approximately 8% (n=33) of the affected neonates, more than one system was involved. The most frequent combinations were CNS with musculoskeletal defects and CNS with orofacial clefts. These multisystem anomalies were associated with higher rates of stillbirth and early neonatal death compared to isolated defects.

5. Outcome and Observations

Among the affected cases, 62% (n=255) were live births, 30% (n=124) were stillbirths, and 8% (n=33) represented medically terminated pregnancies following antenatal diagnosis of severe or incompatible anomalies (such as anencephaly or multiple system involvement). The perinatal mortality rate was significantly higher among cases with CNS and cardiovascular defects.

Discussion

Congenital malformations are a persistent cause of perinatal and neonatal morbidity and mortality, particularly in developing countries where infectious causes of infant deaths have declined but structural birth defects remain under-addressed. In this multicentric retrospective study involving 17,200 deliveries across two tertiary hospitals in Srinagar, 412 neonates were found to have congenital anomalies, giving an incidence of 2.4% (24 per 1,000 live births). This rate is slightly higher than the national pooled estimate of 4.8 per 1,000 births but comparable with other hospital-based studies in India. Variations in reported incidence often arise due to differences in diagnostic methods, regional factors, and referral bias. Tertiary hospitals, which receive complicated and high-risk pregnancies, generally report higher rates than community surveys. The findings reaffirm that congenital anomalies continue to contribute significantly to neonatal morbidity in regions with high delivery loads and limited periconceptional preventive measures.

The spectrum of malformations observed in this study showed that musculoskeletal anomalies were most common, followed by central nervous system (CNS), cardiovascular, and genitourinary defects. Musculoskeletal defects accounted for nearly one-third of all cases, with clubfoot and polydactyly being the leading types. This pattern differs from the findings of Chikkagowdra et al., who reported CNS defects as the predominant group in their study from Karnataka. Regional differences in genetic makeup, dietary habits, and maternal health likely explain these variations. In Kashmir, long winters and limited sun exposure, and possible nutritional deficiencies may affect fetal growth and development. Musculoskeletal anomalies are also easier to detect at birth, unlike cardiovascular or internal defects that require specialized imaging. Despite being generally correctable, these anomalies can lead to lifelong disability if untreated, emphasizing the importance of early diagnosis and intervention.

Central nervous system anomalies were the second most common group, with neural tube defects (NTDs) such as myelomeningocele, anencephaly, and spina bifida forming the majority. These results mirror trends from other Indian studies, which continue to report a high frequency of NTDs despite their preventable nature. The persistence of these defects reflects ongoing gaps in folic acid supplementation and preconceptional counseling. In countries with mandatory folate fortification, the prevalence of NTDs has markedly declined. However, in India, many pregnancies remain unplanned, and nutritional supplementation is often initiated late. Early antenatal registration, widespread education about folic acid intake, and better implementation of maternal nutrition programs are needed to address this gap. Additionally, socioeconomic factors and limited awareness contribute to delayed diagnosis and management of affected pregnancies.

Cardiovascular anomalies ranked third in frequency, with septal defects and transposition of great vessels being most common. The proportion likely underrepresents the actual burden since many congenital heart diseases remain undetected without echocardiography. In high-resource settings, the use of fetal echocardiography and neonatal screening has increased early detection and survival rates. In contrast, many cardiac defects in low-resource regions are diagnosed only after clinical symptoms develop. Genitourinary anomalies, primarily hypospadias and chordee, comprised 12.1% of cases, showing a clear male predominance. Environmental, hormonal, and genetic factors may contribute to these malformations, but consistent epidemiological data from India are limited. These anomalies, while

rarely fatal, carry important implications for future sexual and reproductive health and thus require timely surgical correction and counseling.

Less common anomalies included those of the respiratory tract, gastrointestinal system, orofacial region, and abdominal wall. Congenital diaphragmatic hernia (CDH) and tracheoesophageal fistula (TEF) were notable respiratory anomalies, often presenting as neonatal surgical emergencies. Orofacial clefts accounted for nearly 6% of total defects, with isolated cleft lip being more common than combined cleft lip and palate. These anomalies have both functional and psychosocial consequences and are often associated with maternal nutritional deficiencies or teratogenic exposures. Abdominal wall defects, such as omphalocele and gastroschisis, though rare, contribute to significant neonatal morbidity and require advanced surgical care. About 8% of affected neonates in this study had multiple system involvement, most often CNS and musculoskeletal combinations, suggesting possible chromosomal or syndromic associations. These complex cases underscore the need for cytogenetic studies and genetic counseling in tertiary centers.

Outcomes among affected pregnancies varied by anomaly type and severity. In this series, 62% of affected babies were live births, 30% were stillbirths, and 8% were medically terminated following prenatal diagnosis of lethal or multiple anomalies. Perinatal mortality was highest among cases involving CNS and cardiovascular defects. These figures align with other Indian hospital-based studies, reaffirming that congenital anomalies remain a major cause of preventable perinatal loss. Improved prenatal screening—particularly targeted anomaly scans between 18 and 22 weeks—could help identify severe defects earlier. The availability of fetal medicine units at tertiary centers like those in this study demonstrates progress, but timely referrals and early pregnancy detection remain critical challenges, especially in rural populations. Strengthening antenatal ultrasonography and linking peripheral centers with referral hospitals can substantially improve detection and outcomes.

The findings from this multicentric study highlight the ongoing need for preventive and diagnostic measures to address congenital anomalies. Primary prevention should focus on ensuring adequate periconceptional folic acid supplementation, maternal immunization, control of chronic illnesses, and avoidance of teratogens. Secondary prevention requires universal access to quality prenatal screening and ultrasonography, enabling early detection and informed decision-making. Tertiary prevention, including neonatal surgical interventions, rehabilitation, and parental counseling, is essential to improve quality of life for affected children. Establishing regional and national registries for congenital anomalies would aid in monitoring trends, evaluating interventions, and guiding policy decisions. A coordinated effort among obstetricians, pediatricians, neonatologists, and public health professionals is needed to reduce the burden of congenital malformations in India. The present study contributes valuable data from a high-delivery region and reinforces that sustained preventive action can meaningfully reduce perinatal morbidity and mortality associated with congenital anomalies.

Conclusion

In this multicentric retrospective study from two tertiary care hospitals in Srinagar and New Delhi, congenital anomalies were identified in 2.4% of total births, reflecting a substantial burden of preventable neonatal disorders. Musculoskeletal and central nervous system malformations were most frequent, followed by cardiovascular and genitourinary defects. Neural tube defects, despite being largely avoidable through periconceptional folic acid supplementation, remained prominent, underscoring gaps in preventive maternal care. Early diagnosis through routine prenatal screening and improved access to fetal medicine services are essential for reducing morbidity and mortality. Establishing regional surveillance and congenital anomaly registries can facilitate systematic data collection and guide public health interventions. Focused health education, nutritional counseling, and multidisciplinary management are critical to minimizing disability and improving neonatal outcomes. Strengthening preventive strategies remains central to lowering the burden of congenital malformations in India's tertiary healthcare settings.

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