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Original Research Article

Prevalence of various types of congenital malformations: a hospital based descriptive cross-sectional study

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ABSTRACT

Background: World Health Organization (WHO) defines birth defect as structural or functional anomalies during intrauterine life which are identified during prenatal, at birth and sometime later in the infancy. The international statistical classification of diseases includes birth defects in chapter XVII as congenital malformations, deformations and chromosomal abnormalities.
Methods: This was a descriptive cross sectional observational hospital-based study conducted on prevalence of various types of congenital malformations at Department of Obstetrics and Gynecology, RNT Medical College, Udaipur. Study period from February 2024 to February 2025.
Results: Among the 250 mothers, the majority were primigravida (G-1) at 39.6%, followed by G-2 at 27.9%, G-3 at 20.1%, G-4 at 8.5%, and multigravida beyond four pregnancies (>G-4) at 4%. 211 mothers (86.2%) had no previous history of abortion, while 39 (13.8%) reported prior abortions. Among these, 19 cases (7.6%) were induced and 20 cases (8%) were spontaneous, indicating a slightly higher prevalence of spontaneous abortions. majority of congenital anomalies were seen in mothers aged 20-30 years (58.8%), with central nervous system (CNS) defects being the most common across all age groups.
Conclusions: This study highlights that congenital malformations, CNS defects were the leading anomalies, followed by cardiovascular and genito-urinary (GU) malformations. Most cases occurred among mothers aged 20-30 years, and first and second pregnancy, with a higher prevalence in rural populations.

Keywords: Congenital malformations, WHO, Central nervous system

INTRODUCTION

Human development is an intricate and elaborate process, from a single-cell zygote to a complex multicellular organism. Fortunate are those fetuses that navigate this journey without any obstacles. The birth of a malformed baby is a tragic event for any family and society as a whole. The influence of teratogens, such as pathogens, extensive chemical exposure, environmental pollution, and the indiscriminate use of drugs by mothers in their daily lives, has led to an increased incidence of congenital abnormalities in newborn children. Congenital anomalies are significant causes of prenatal mortality and morbidity. Consequently, antenatal diagnosis and fetal therapy have become crucial in the field of human embryology, as noted by Dolk. WHO defines birth defect as structural or functional anomalies during intrauterine life which are identified during prenatal, at birth and sometime later in the infancy.[1]

According to march of dimes (MOD) global report on birth defects, 7.9 million births occur annually worldwide with serious birth defect. 94% of these births defects occur in low-and middle-income countries (LMIC). In India, out of every 100 babies born annually, 6 to 7 have a child birth defect (CBD), translating to 1.7

million (MOD, 2006).[2] The recent studies from India show the incidence as 0.66% to 4.08%. In developed world, CBDs affect one in every 33 babies born each year. National health and family survey (NHFS)-5 report (2019-21) India, reported overall 3.3% prevalence of foetus with congenital anomalies, out of which 3.9% was observed in urban females and 3.0% in rural population.

According to Rashtriya Bal Swasthya Karyakram, India with a birth cohort of 26 million annually accounts for highest share of CBD globally. Here, CBD mortality (8.6%) ranked as the fourth leading cause of neonatal death, after neonatal preterm birth (27.7%), neonatal encephalopathy due to birth asphyxia and trauma (14.5%), and lower respiratory infections (11.0%).[3]According to joint WHO and MOD meeting report, 7% of all neonatal mortality and 3.3 million under five deaths have been attributed to CBDs.[4]

The birth defects spectrum involves different organ systems, the most common being congenital heart diseases (CHD), neural tube defects (NTDs), and Down syndrome followed by hemoglobinopathies, musculoskeletal (MSK) disorders and others. The prevalence and mortality of neonate due to birth defects varies among different culture and communities and also different regions depending on the presence or absence of these risk factors. Babies with birth defect require significant medical, surgical interventions. Examples include spina bifida, anencephaly, heart defects and orofacial clefts.[5]

Major congenital anomalies result in mortality, morbidity, and disability and minor anomalies have minor or no significant health problem and tend to have limited social or cosmetic consequences. Examples of minor anomalies include single palmar crease and clinodactyly (mild curvature of a finger). Once the hospital-based surveillance is well established, one can then begin to identify neonates with congenital anomalies born at home who are taken to hospitals or local clinics for services, to assess the possibility of implementing a population-based surveillance programme. Although genetic factors contribute to birth defects risk, environmental (i.e., non-genetic) factors influence the risk for birth defects as well.[6] Every year more than 8.14 million children are born with a serious birth defect, due to genetic or environmental causes.[7] This study has been conducted to find out the prevalence and pattern of congenital malformations (magnitude and distribution of birth defects), identify high- risk populations or identify clusters (aggregation of cases), to monitor trends in the prevalence of different types of birth defects in different categories of neonates and pregnant mother diagnosed to have birth defect child admitted to our hospital. It is of paramount importance from preventive strategic point of view to know about the characteristics of neonates and malformations. Hopefully information available from this study will help the health managers to evolve preventive strategy to decrease the prevalence of the congenital malformations in the study area.[8]

Aims and objectives

Aim and objectives were to determine the incidence of congenital malformations at a tertiary care center and to study the various types of malformations.

METHODS

Study design

This was a descriptive cross sectional observational hospital-based study

Study place

Study conducted on prevalence of various types of congenital malformations at department of obstetrics and gynecology, RNT Medical College, Udaipur.

Study size

Sample size is calculated using the formula

$N=Z^2PQ/L^2$

Z=1.96, P=Prevalence, Q=1-P and L is precision.

According to a study by Singh et al where overall prevalence of congenital malformations was 59/10,000 live births.[16] Keeping a confidence interval of 95% and margin of error 5%, sample size calculated is 226. Assuming 10% drop out rate sample size came to be 249 after roundoff we will take sample of 250.

Study period

Study carried out from February 2024 to February 2025.

Inclusion criteria

All women coming during 1st, 2nd and 3rd trimester for termination of pregnancy due to congenital anomalies in foetus (confirmed by USG), all women delivering an anomalous fetus and all the live born, still born, intrauterine deaths and neonatal deaths with congenital anomalies will be included in the study.

Exclusion criteria

Mothers not willing to participate in the study were excluded from study.

Methodology

Study conducted in department of obstetrics and gynecology at RNT Medical College Udaipur, Rajasthan from February 2024 to February 2025 with all women coming to a scan showing any type of congenital anomaly in the fetus admitted to the labor room. Patients may be in the 1st, 2nd or 3rd trimester. After filling out appropriate

forms (Form I-for upto 20 weeks, form E-for between 20-24 weeks of gestation (after filling form D) in cases of termination of pregnancies with anomalies beyond 24 weeks and obtaining required permission from medical board. Meanwhile patient's details and relevant history (ante-natal visit record, family history, consanguinity etc) will be recorded in a suitable proforma. Details of investigations like ultrasonography, fetal echocardiography and laboratory studies will be noted and entered into the record. After the patient aborts or delivers the fetus, an examination of the fetus will be carried out. The fetus' weight, gender (if it can be determined), period of gestation and the type of anomaly will be noted down.

RESULTS

Gender wise distribution in our study

Among 250 babies with congenital malformations, 180 (72%) male and 70 (28%) were female, indicating a higher prevalence of congenital anomalies among male infants.

Outcome of birth

Out of 250 cases of congenital malformations, 215 (86.22%) resulted in live births, 6 (3.53%) were stillbirths, and 29 (10.25%) involved medical termination of pregnancy (MTP) up to 24 weeks, indicating that the majority of affected pregnancies led to live births.

Gestational age

Among 250 cases of congenital malformations, majority were detected in later stages of gestation, with 93 cases (37.2%) occurring after 34 weeks and 101 cases (40.4%) between >24-34 weeks. Earlier gestational detections were less frequent, with 45 cases (18%) between >14–24 weeks, and only 11 cases (4.4%) before 14 weeks.

Residence

Out of 250 mothers, 179 (74.9%) resided in rural areas, while 71 (25.1%) were from urban areas, indicating a predominantly rural population in the study.

Maternal age

Among the 250 mothers studied, the majority were aged 20-25 years (39.46%), followed by those aged 25-30 years (21.77%). Women aged 30-35 years accounted for 18.03%, and those above 35 years made up 15.31%. The least represented group was mothers below 20 years of age, comprising only 1.7% of the total.

Mothers' education

Among mothers, majority had education up to the primary level (26.5%) and high school (25.44%). Middle school (17.67) and graduate-level education were equally common (24.38%), while only 2.12% mother's illiterate.

Obstetric history

Among the 250 mothers, the majority were primigravida (G-1) at 39.6%, followed by G-2 at 27.9%, G-3 at 20.1%, G-4 at 8.5%, and multigravida beyond four pregnancies (>G-4) at 4%. This indicates that most congenital malformations occurred in the first and second pregnancies.

Previous history of abortion

Out of 250 cases, 211 mothers (86.2%) had no previous history of abortion, while 39 (13.8%) reported prior abortions. Among these, 19 cases (7.6%) were induced and 20 cases (8%) were spontaneous, indicating a slightly higher prevalence of spontaneous abortions.

Previous history of contraceptive use

Among the 250 mothers, 130 (48%) had not used any contraceptive method. Oral contraceptive pills (OCPs) were the most commonly used method among users, reported by 67 mothers (26.8%), followed by Cu-T (9.2%), Antara injections (6%), barrier methods (4%), and implants (2%).

History of malformation in family

Out of 250 cases, 215 mothers (87.63%) reported no family history of the congenital malformations, while 35 cases (12.37%) had a positive family history and indicating a smaller but notable proportion of the hereditary influence.

Use folic acid (>3 months)

Out of 250 cases, 243 mothers (97.5%) reported using folic acid for more than 3 months during pregnancy, while only 7 mothers (2.5%) did not, highlighting a high rate of compliance with folic acid supplementation.

Maternal chronic diseases and use of regular drugs

Out of 250 mothers, 207 (82.8%) had no history of chronic diseases or regular drug use during pregnancy, while 43 (17.2%) reported the presence of chronic conditions and/or ongoing medication use.

Attend regular antenatal clinic

Out of 250 mothers, 205 (82%) attended regular antenatal clinics during their pregnancy, while 45 (18%) did not receive consistent antenatal care.

Use of tobacco

Out of 250 mothers, 234 (94.3%) reported no use of tobacco during pregnancy, while 16 mothers (5.7%) admitted to tobacco use.

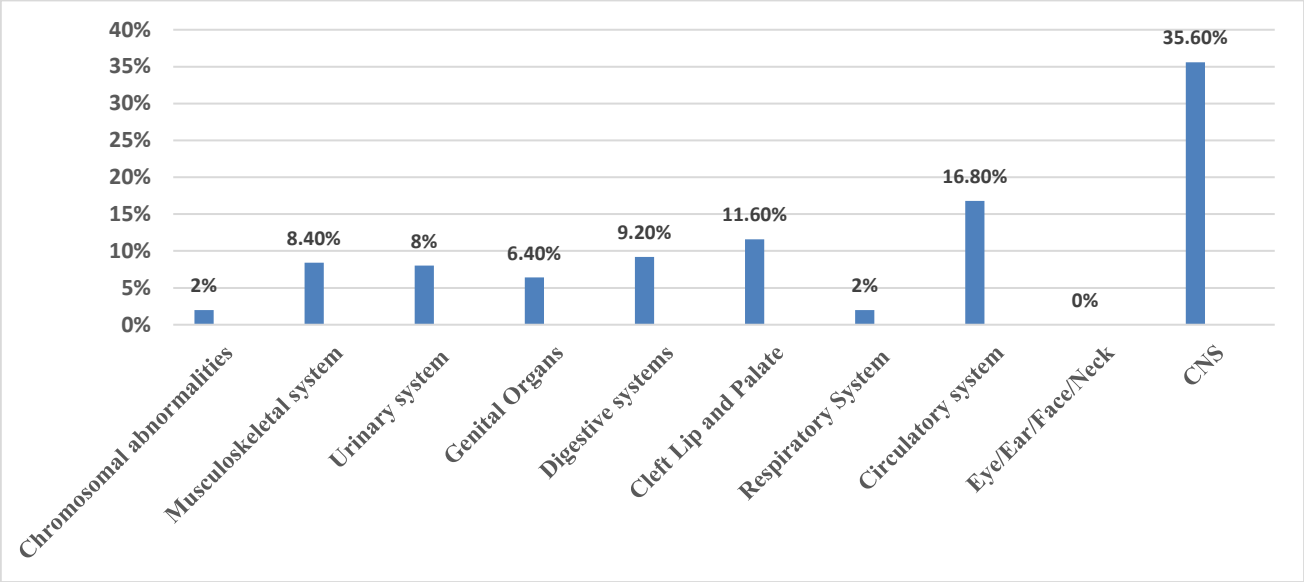

Figure 1: System wise classification of congenital malformations.

Where among the 250 observed congenital anomalies, the CNS malformations were the most common, accounting for 89 cases (35.6%). This was followed by circulatory system defects with 42 cases (16.8%), and GU system malformations with 36 cases (14.4%).

Orofacial defects were observed in 29 cases (11.6%), and digestive system anomalies in 23 cases (9.2%). Musculo-skeletal (MSK) system anomalies were seen in 21 cases (8.4%). Less frequent were chromosomal abnormalities, with 5 cases (2.0%), and respiratory system anomalies, also 5 cases (2.0%). Notably, there were no reported cases of malformations in eye, ear, face and neck category, nor under other congenital malformations. This distribution highlights that neural and cardiovascular anomalies constitute majority of congenital malformations in these babies.

Classification of congenital malformation with gender of baby

The distribution of congenital malformations among 250 cases shows a clear predominance in males (72.0%) compared to females (28.0%). The most common system involved was the CNS, accounting for 35.6% of total cases (25.6% in males, 10.0% in females). This was followed by the circulatory system with 16.8% (12.0% males, 4.8% females), GU system with 14.4% (10.4% males, 4.0% females), and orofacial defects contributing 11.6% (8.4% males, 3.2% females). Other notable categories included the digestive system (9.2%) and MSK system (8.4%), both showing higher incidence in males. Chromosomal abnormalities were seen only in males (2.0%), while respiratory system anomalies were rare (2.0% overall). No malformations were observed in the eye, ear, face and neck or other congenital malformations categories. Overall, all systems showed a male preponderance across almost all malformation types.

Classification of congenital malformation with age of the mothers

Among the 250 congenital malformation cases, the highest number occurred in mothers aged 20-25 years (83 cases, 33.2%), followed by 25-30 years (64 cases, 25.6%), 30-35 years (53 cases, 21.2%), and >35 years (45 cases, 18.0%). Only 5 cases (2.0%) were reported in mothers under 20.

The most affected system was the CNS (89 cases, 35.6%), with the peak incidence in the 20-25 years group (12.4%), followed by decreasing frequency in older age groups. Circulatory system defects (16.8%) followed a similar pattern, most commonly seen in mothers aged 20-25 years. Other prevalent anomalies included GU system (14.4%), orofacial defects (11.6%), and digestive system malformations (9.2%), again most frequently observed in the 20-25 age group.

Overall, the majority of congenital anomalies were seen in mothers aged 20–30 years (58.8%), with CNS defects being the most common across all age groups.

Congenital malformation with obstetric history

Among the 250 congenital malformation cases, the highest number were seen in primigravida (G-1) mothers-99 cases (39.6%), followed by G-2 (28.0%), G-3 (20.0%), G-4 (8.4%), and >G-4 (4.0%).

The CNS was the most commonly affected system (89 cases, 35.6%), most frequently seen in G-1 (12.0%) and G-2 (11.2%) pregnancies.

Similarly, circulatory system malformations (16.8%) and GU anomalies (14.4%) were also predominantly observed in lower gravidity (G-1 and G-2).

Other significant anomalies included orofacial defects (11.6%), digestive system (9.2%), and MSK defects (8.4%), again with higher incidence in early pregnancies. Chromosomal abnormalities were rare (2.0%) and mostly associated with first-time pregnancies. Overall, the majority of congenital anomalies occurred in G-1 and G-2 pregnancies (67.6%), with CNS defects being the most common across all gravidity levels. The incidence of anomalies declined progressively with increasing gravidity (Table 1).

Among the 250 cases of congenital malformations, 207 mothers (82.8%) had no history of chronic disease or regular drug use, while 43 mothers (17.2%) had existing chronic illnesses and/or were on regular medication.

The most common malformations across both groups were CNS anomalies (35.6%), with 30.4% in mothers without chronic conditions and 5.2% in those with such conditions. Similarly, circulatory system defects (16.8%) were more frequent in the no-disease group (13.6%) compared to 3.2% in the affected group.

Other malformations such as GU (14.4%), orofacial (11.6%), and digestive system anomalies (9.2%) were also more prevalent in mothers without chronic diseases, though a notable number were still present in those with comorbidities. Chromosomal abnormalities (2.0%) and MSK defects (8.4%) showed modest presence in both groups. Overall, while the majority of congenital anomalies occurred in mothers without chronic diseases or regular drug use, 17.2% of cases were associated with maternal comorbidities, indicating that pre-existing maternal health conditions may contribute to a subset of congenital malformations-especially in the central nervous and circulatory systems.

Classification of congenital malformation with contraceptive used

Among 250 congenital malformation cases, 52.0% of mothers had not used any contraceptives, while the remaining 48.0% had used various forms: OCPs (26.8%), Cu-T (9.2%), Antara (6.0%), implants (2.0%), and barrier methods (4.0%).

The CNS was the most affected system (35.6%), with the highest occurrence in non-contraceptive users (19.2%), followed by OCP users (9.2%) and lower frequencies across other methods. Circulatory system defects (16.8%) followed a similar trend, again most common in non-users (8.0%) and OCP users (4.4%).

Other malformations-orofacial (11.6%), GU (14.4%), and digestive system anomalies (9.2%)-were also more frequent among non-contraceptive users. MSK (8.4%) and chromosomal abnormalities (2.0%) were seen in all groups but remained most common in non-users and OCP users. No anomalies were reported in the eye, ear, face and neck or other congenital categories.

Overall, a higher proportion of congenital malformations was observed in mothers who had not used any contraceptives, especially affecting the central nervous and circulatory systems. However, malformations were also present, though less frequently, across all contraceptive user groups.

Among the 250 cases of congenital malformations, 234 cases (93.6%) were observed in mothers without a history of tobacco use, while 16 cases (6.4%) were associated with maternal tobacco usage during pregnancy.

The most frequent malformations in both groups included: CNS anomalies (35.6%), with 33.2% in non-users and 2.4% in tobacco users. Circulatory system defects (16.8%)-16.0% in non-users vs. 0.8% in users. Other commonly affected systems were orofacial (11.6%), GU (14.4%), and digestive system (9.2%) defects, all showing lower but notable contributions from the tobacco-exposed group.

No malformations were reported in the eye, ear, face and neck, or other unspecified congenital categories.

Overall, while the majority of congenital malformations occurred in pregnancies without maternal tobacco exposure, a small but significant 6.4% were linked to tobacco use, with most anomalies seen in the central nervous and GU systems, suggesting potential teratogenic effects of tobacco during pregnancy.

Table 1: Maternal chronic diseases and use of regular drugs.

System of malformation	No	Yes	Total
CNS	76 (30.4%)	13 (5.2%)	89 (35.6%)
Eye, ear	0 (0.0%)	0 (0.0%)	0 (0.0%)
Circulatory system	34 (13.6%)	8 (3.2%)	42 (16.8%)
Respiratory system	4 (1.6%)	1 (0.4%)	5 (2.0%)
Orofacial defect	24 (9.6%)	5 (2.0%)	29 (11.6%)
Digestive system	19 (7.6%)	4 (1.6%)	23 (9.2%)
GU system	30 (12.0%)	6 (2.4%)	36 (14.4%)
MSK system	17 (6.8%)	4 (1.6%)	21 (8.4%)
Other congenital malformations	0 (0.0%)	0 (0.0%)	0 (0.0%)
Chromosomal abnormalities, not elsewhere classified	3 (1.2%)	2 (0.8%)	5 (2.0%)
Column totals	207 (82.8%)	43 (17.2%)	250 (100%)

Table 2: Classification of malformation with history of tobacco usage.

System of malformation	No (%)	Yes (%)	Row total (%)
CNS	83 (33.2)	6 (2.4)	89 (35.6)
Eye, ear,	0 (0.0)	0 (0.0)	0 (0.0)
Circulatory system	40 (16.0)	2 (0.8)	42 (16.8)
Respiratory system	4 (1.6)	1 (0.4)	5 (2.0)
Orofacial defect	27 (10.8)	2 (0.8)	29 (11.6)
Digestive system	22 (8.8)	1 (0.4)	23 (9.2)
GU system	34 (13.6)	2 (0.8)	36 (14.4)
MSK system	20 (8.0)	1 (0.4)	21 (8.4)
Other congenital malformations	0 (0.0)	0 (0.0)	0 (0.0)
Chromosomal abnormalities, not elsewhere classified	4 (1.6)	1 (0.4)	5 (2.0)
Column totals	234 (93.6)	16 (6.4)	250 (100)

DISCUSSION

Congenital anomalies represent a significant cause of neonatal morbidity and mortality worldwide, with a particularly high burden in LMICs such as India. Despite advances in perinatal screening, antenatal care, and neonatal intensive care, many malformations remain undiagnosed until birth or later, contributing to preventable deaths and lifelong disabilities. Udaipur, had varied urban and rural populations, where factors such as poor antenatal booking, maternal malnutrition, consanguinity, and lack of access to diagnostic facilities may contribute to a higher prevalence of congenital anomalies. Therefore, understanding the pattern and system-wise distribution of anomalies in this setting is essential for developing preventive strategies, strengthening antenatal surveillance, and guiding resource allocation for pediatric surgical and medical services.

CNS anomalies

CNS anomalies consistently emerge as the most common category across multiple studies. The present study from Udaipur found CNS malformations in 35.6% of cases, dominated by anencephaly, myelomeningocele, hydrocephalus, encephalocele, meningocele, and spina bifida. These findings align with reports from Khan et al (Karachi), where hydrocephalus (25.2%) and anencephaly (16.8%) were leading anomalies, and from Sharma et al (Ranchi), who reported 56% CNS anomalies, with anencephaly being the most common. Similarly, Sowmiya et al (Coimbatore) highlighted CNS as the leading group, and Nishupriya et al (Jammu and Kashmir) reported 54.6% CNS defects. NTDs remain a major contributor, often linked with folic acid deficiency, consanguinity, and maternal hypertension.[9-12]

Cardiovascular system anomalies

The second most frequent category in present study was CVS anomalies (16.8%), with patent ductus arteriosus, atrial septal defect, duct-dependent cyanotic heart disease, ventricular septal defect, and complex lesions identified. This aligns with findings from Laxmanan (Coimbatore), where CVS anomalies contributed 26%, and Verma et al (Northern India), who reported 33.4% cardiovascular malformations, often requiring surgical intervention. Padmanabhan et al also noted 35.6% CVS prevalence, while Singh et al found circulatory malformations as the leading group (20%), with Down syndrome being common. Higher mortality associated with CVS malformations has been highlighted in Padmanabhan et al study, where they accounted for max neonatal deaths.[13-16]

Gastrointestinal anomalies

In the present study, 9.2% of anomalies involved the gastrointestinal tract (GIT), including malformations such as diaphragmatic hernia, gastroschisis, exomphalos, and cloacal exstrophy. Comparable results were reported by Padmanabhan et al where 9.2% of malformations were GIT, and Verma et al also observed a considerable burden (19.8%). Although GIT anomalies are less frequent than CNS and CVS, they often require urgent surgical correction.[14,15]

Table 3: CNS anomalies across studies.

Authors	Location	CNS prevalence/key findings
Present study	Udaipur, India	35.6%-anencephaly, myelomeningocele, hydrocephalus, spina bifida
Khan et al[9]	Karachi, Pakistan	Hydrocephalus (25.2%), anencephaly (16.8%), spina bifida (14%)
Sowmiya et al[10]	Coimbatore, India	CNS most common, anencephaly predominant
Sharma et al[11]	Ranchi, India	56% CNS defects, anencephaly most frequent
Nishupriya et al[12]	Kathua, J and K	54.6% CNS anomalies, mainly NTDs

Table 4: CVS anomalies across studies.

Authors	Location	CVS prevalence/ key findings
Present study	Udaipur, India	16.8%-PDA, ASD, duct-dependent CHD, VSD, complex lesions
Laxmanan[13]	Coimbatore, India	26% CVS anomalies, leading group
Verma et al[14]	Northern India	33.4% CVS anomalies, high surgical need
Padmanabhan et al[15]	India	35.6% CVS anomalies, mortality highest in this group
Singh et al[16]	Queen Elizabeth Hospital	Circulatory 20%, Down syndrome most common single defect

Table 5: GIT anomalies across studies

Authors	Location	GIT prevalence / key findings
Present study	Udaipur, India	9.2% diaphragmatic hernia, gastroschisis, exomphalos
Verma et al[14]	Northern India	19.8% GIT anomalies
Padmanabhan et al[15]	India	9.2% GIT anomalies

Musculoskeletal anomalies

MSK malformations accounted for 8.4% in the present study, with cases such as achondroplasia, arthrogryposis, and talipes. This is lower compared to several studies where MSK anomalies predominated, such as Sinha et al reporting 52.2%, and Vinodh et al (Kerala) who found 24% MSK prevalence.[17,18] Sharma et al (Ranchi) also noted 23% MSK anomalies, with cleft lip and palate being common.[11] The variability may be due to regional genetic factors, consanguinity rates, and diagnostic practices.

Table 6: MSK anomalies across studies,

Authors	Location	MSK prevalence/key findings
Present study	Udaipur, India	8.4% achondroplasia, arthrogryposis, talipes
Sinha et al[17]	India	52.2% MSK anomalies, CTEV most common
Vinodh et al[18]	Kerala, India	24% MSK anomalies
Sharma et al[11]	Ranchi, India	23% MSK anomalies, cleft lip-palate common

Genitourinary anomalies

In the present study, 14.4% of anomalies were genitourinary (GU)-related, with polycystic kidney disease, posterior urethral valves, hydronephrosis, multicystic dysplastic kidney, and ectopia vesicae being identified. This is consistent with Padmanabhan et al who reported 13.8% GU malformations, and Verma et al where 19.8% GU anomalies were noted. Sinha et al also reported a high prevalence (26.1%).[14-16]

Table 7: GU anomalies across studies.

Authors	Location	GU prevalence/key findings
Present study	Udaipur, India	14.4%-polycystic kidney, posterior urethral valves, hydronephrosis
Verma et al[14]	Northern India	19.8% GU anomalies
Padmanabhan et al[15]	India	13.8% GU anomalies
Sinha et al[17]	India	26.1% GU anomalies

Orofacial anomalies

The present study recorded 11.6% orofacial defects, mainly cleft lip and palate. Khan et al (Karachi) similarly observed cleft lip in 8.4%, while Sharma et al (Ranchi) reported cleft lip-palate as the commonest MSK anomaly. Sinha et al reported 6.4% cleft palate.[9,11,17]

Table 8: Orofacial anomalies across studies.

Authors	Location	Orofacial prevalence/key findings
Present study	Udaipur, India	11.6%-cleft lip and palate
Khan et al[9]	Karachi, Pakistan	8.4% cleft lip
Sharma et al[11]	Ranchi, India	Cleft lip-palate commonest MSK anomaly
Sinha et al[17]	India	6.4% cleft palate

Chromosomal and other anomalies

The present study reported 2% chromosomal anomalies (Down syndrome), which is in line with Singh et al who highlighted Down syndrome as the commonest chromosomal defect (4.1/10,000 live births). Respiratory anomalies, though rare (2%), were represented in Udaipur exclusively by choanal atresia.

Table 9: Chromosomal/ other anomalies across studies.

Authors	Location	Key Findings
Present study	Udaipur, India	2% Down syndrome; 2% respiratory (choanal atresia)
Singh et al[16]	Queen Elizabeth Hospital	Down syndrome (4.1/10,000 live births)

Limitations

Single-center study design

The study was conducted at a single tertiary care center (R. N. T. Medical College and associated M. B. Hospital, Udaipur), which may limit the generalizability of the findings to other regions or healthcare settings.

Hospital-based sample

As the study included only hospital-attending cases, it may not accurately represent the true prevalence or pattern of congenital malformations in the general community, particularly those not seeking institutional care.

Lack of a comparison/control group

The absence of a control group of pregnancies without congenital malformations limits the ability to identify or establish associations with potential risk factors.

Late gestational detection bias

A majority of congenital malformations were detected in later gestational ages (>24 weeks), which may have led to underrepresentation of early-detected or early-terminated cases, thereby affecting the overall pattern of findings.

Reliance on self-reported maternal history

Information regarding folic acid intake, drug use, environmental exposure, tobacco use, and family history was based on maternal recall, which is subject to recall bias and reporting inaccuracies.

CONCLUSION

This study highlights that congenital malformations, Central nervous system defects were the leading anomalies, followed by cardiovascular and GU malformations. Most cases occurred among mothers aged 20–30 years, and first and second pregnancy, with a higher prevalence in rural populations.

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