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PATTERN AND OUTCOMES OF FETAL CONGENITAL ANOMALIES: A PROSPECTIVE STUDY AT A TERTIARY CARE CENTER IN NANDED

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ABSTRACT

Background: Congenital anomalies contribute are a major cause of fetal loss, stillbirth, neonatal mortality, and long-term disability. The causes are multifactorial, involving genetic, chromosomal, nutritional, environmental factors and maternal medical conditions. In resource-limited settings, delayed diagnosis worsens outcomes. Timely identification through screening programs enables better clinical decision-making. **Methods:** This prospective observational study was carried out at a tertiary care center over 18 months (January 2024 to June 2025). A total of 144 antenatal cases diagnosed with congenital anomalies were included. Data on maternal risk factors, gestational age at diagnosis, anomaly type, diagnostic methods, and outcomes were analyzed using descriptive statistics. **Results:** Ultrasonography was the primary diagnostic modality (73.00%). The majority of anomalies (42.36%) were detected during the 20–24-week period. Central nervous system anomalies were most common (31.94%). Nutritional deficiency was the leading modifiable risk factor (19.44%). Pregnancy outcomes included Medical Termination of Pregnancy (MTP) in 71.53% of cases, live births in 18.05%, and stillbirths in 10.42%. Post-intervention survival among live births was low (6.90% of total study population). **Conclusion:** Congenital anomalies were characterized by late diagnosis and poor postnatal survival. The concentration of diagnoses within the 20–24-week window highlights a critical "late diagnosis" period that coincides with the upper legal limit for termination, thereby limiting early intervention. Enhancing first-trimester screening (11–14 weeks), improving maternal nutrition, and expanding neonatal surgical and intensive care services are essential.

KEYWORDS: Congenital anomalies, antenatal screening, fetal outcomes, ultrasonography, maternal risk factors, late diagnosis.

ABBREVIATIONS

ANC: Antenatal Care
CNS: Central Nervous System
CVS: Chorionic Villus Sampling
LSCS: Lower Segment Cesarean Section
MTP: Medical Termination of Pregnancy
NIPT: Non-Invasive Prenatal Testing
NT: Nuchal Translucency
USG: Ultrasonography

INTRODUCTION

Congenital anomalies, commonly referred to as birth defects, include structural, functional, and metabolic abnormalities that develop during intrauterine life. These conditions may be identified during pregnancy, at birth, or later in infancy and contribute significantly to spontaneous abortion, stillbirth, neonatal mortality, and long-term disability in children.[1,2]

Globally, congenital anomalies affect nearly 8 million newborns annually, accounting for approximately 6% of all births and contribute substantially to neonatal morbidity and mortality.[9,12,18] The burden is disproportionately higher in low- and middle-income countries due to limited access to early screening, delayed diagnosis, and inadequate healthcare infrastructure.[3,12]

The etiology of congenital anomalies is complex and often multifactorial, including genetic and chromosomal abnormalities, maternal nutritional deficiencies, infections during pregnancy, consanguinity, exposure to teratogens, and pre-existing maternal medical conditions.[2,6,12] However, in many cases, the exact cause remains unidentified.

Preventive strategies, particularly preconception care and adequate folic acid supplementation, play a crucial role in reducing the incidence of certain anomalies, especially neural tube defects.[5,9] With advancements in prenatal diagnostic techniques, early detection has become increasingly feasible, allowing improved clinical decision-making and better pregnancy outcomes.[6]

Advances in prenatal screening modalities, particularly ultrasonography, along with first-trimester screening methods such as the Nuchal Translucency (NT) scan and double marker test, and newer techniques like Non-Invasive Prenatal Testing (NIPT), have enhanced early detection of chromosomal abnormalities, including Down syndrome. Diagnostic procedures such as chorionic villus sampling (CVS) and amniocentesis provide confirmatory diagnosis in high-risk cases.[4,8]

Early antenatal diagnosis facilitates appropriate parental counseling, prognostication, timely referral to higher centers, and planning of delivery and management, including MTP in severe or lethal anomalies where indicated.[7,10]

The pattern and spectrum of congenital anomalies vary across populations due to genetic, environmental, nutritional, and socioeconomic factors.[1,2,18] Therefore, region-specific data from tertiary care centers are essential for understanding the distribution of anomalies, identifying associated maternal risk factors, and evaluating pregnancy and fetal outcomes.[7]

Despite advancements in prenatal diagnostics, there remains a critical gap in the timing of anomaly detection, particularly in low-resource settings, where many cases are identified only during the second trimester. This delay has significant implications for clinical decision-making and pregnancy outcomes.

Hence, the present study was undertaken to evaluate the pattern of congenital anomalies in fetuses and their outcomes at a tertiary care center. This study also aims to examine the clinical implications of delayed diagnosis within the framework of existing legal and healthcare constraints.

OBJECTIVES

- To study the pattern of congenital anomalies.
- To evaluate fetal outcomes in cases with congenital anomalies.
- To assess methods of pregnancy termination and delivery in affected cases.

METHODOLOGY

Study Design: This prospective observational study was carried out in the Department of Obstetrics and Gynaecology of Dr. Shankarrao Chavan Government Medical College, Nanded, over a period of 18 months from January 2024 to June 2025.

Study Participants: The study included pregnant women aged ≥18 years whose fetuses were diagnosed with congenital anomalies on ultrasonography (USG), as well as cases in which anomalies were detected antenatally or identified during immediate postnatal examination. This also included women referred from peripheral centers for advanced diagnostic evaluation and multidisciplinary management of suspected fetal anomalies. Women aged <18 years and medicolegal cases were excluded.

A total of 144 cases were enrolled using consecutive sampling. This study did not include a control group and was designed as a purely descriptive cohort of pregnancies affected by congenital anomalies.

Statistical Analysis: Data were collected using a predesigned, structured proforma, covering maternal demographics, risk factors, gestational age at diagnosis, type of anomaly, diagnostic methods, and pregnancy and fetal outcomes. Nutritional deficiency was operationally defined based on maternal dietary history, clinical records of irregular antenatal registration, and documented lack of periconceptional folic acid and vitamin B12 supplementation. Biochemical parameters such as hemoglobin levels were available; however, serum vitamin B12 levels were not uniformly assessed. Therefore, nutritional deficiency was primarily defined based on maternal dietary history and supplementation records, which may introduce some degree of misclassification bias.

For the purpose of outcome analysis, neonatal outcomes were assessed only among live-born neonates, excluding stillbirths and terminated pregnancies.

Descriptive statistical methods were used for data analysis and were compiled in Microsoft Excel 2021. No inferential statistical analysis was performed due to the descriptive nature of the study.

Percentages were rounded to two decimal places for improved statistical accuracy.

In cases of termination of pregnancy, multiple methods were used sequentially or in combination in certain patients based on clinical requirements. Therefore, percentages for methods of termination may exceed 100% due to the use of multiple methods in individual patients.

For clarity, “pregnancy outcomes” refer to the mode of pregnancy termination or continuation (e.g., MTP, live

birth, stillbirth), whereas "fetal outcomes" refer to neonatal survival and postnatal clinical status. As a tertiary care referral center, the study is subject to referral bias, with a higher proportion of severe and complex anomalies.

Ethics Approval: The study protocol was reviewed and approved by the Institutional Ethics Committee at Dr. Shankarrao Chavan Government Medical College, Nanded. Written informed consent was obtained from all participants, and strict confidentiality of participant data was maintained throughout the study period. All MTP procedures in this study were performed in accordance with the statutory provisions and gestational limits defined by the **Medical Termination of Pregnancy (MTP)** Act, following appropriate counseling and informed consent from the participants. The findings were presented using tables and charts wherever necessary.

Prenatal Screening and Diagnostic Modalities: A wide range of prenatal screening and diagnostic modalities are available for the early detection of congenital and chromosomal abnormalities at different stages of pregnancy. First-trimester screening plays a crucial role in early risk assessment and includes the **Nuchal Translucency (NT)** scan along with biochemical markers such as the double marker test. These methods enable the identification of pregnancies at increased risk for aneuploidies, including Down syndrome (Trisomy 21), Trisomy 18, and Trisomy 13.

Non-Invasive Prenatal Testing (NIPT), based on the analysis of cell-free fetal DNA circulating in maternal blood, has emerged as a highly sensitive and specific screening tool for common chromosomal abnormalities and is increasingly utilized in clinical practice.[11,12]

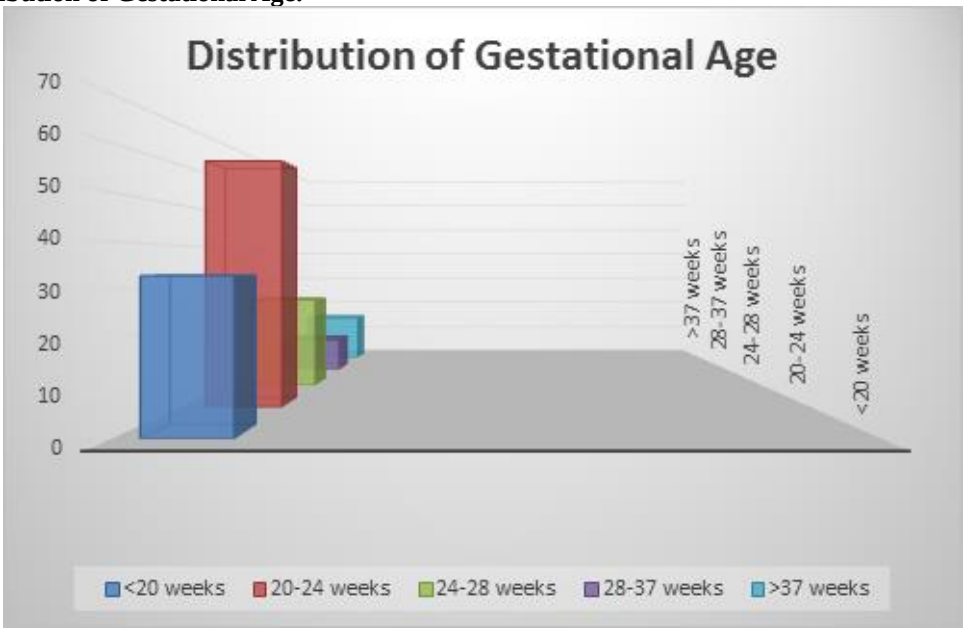
Test	Gestational Age	Specific Anomaly Diagnosed
NT Scan	11–14 weeks	Down syndrome, Trisomy 18, congenital heart defects
Double Marker Test	10–13 weeks	Down syndrome, Trisomy 18, Trisomy 13
NIPT	≥10 weeks	Down syndrome, Trisomy 18, Trisomy 13
Chorionic villus sampling (CVS)	10–13 weeks	β-thalassemia, chromosomal abnormalities
Amniocentesis	15–20 weeks	Down syndrome, neural tube defects, metabolic disorders

For definitive diagnosis, invasive procedures such as CVS and amniocentesis are employed. CVS is typically performed during the first trimester and is useful for detecting chromosomal and genetic disorders, including β-thalassemia. Amniocentesis, usually performed in the second trimester, enables chromosomal analysis and the detection of neural tube defects through biochemical markers such as alpha-fetoprotein.

The combined use of these screening and diagnostic modalities significantly improves early detection, risk stratification, and clinical management of congenital anomalies.[11]

OBSERVATIONS AND RESULTS

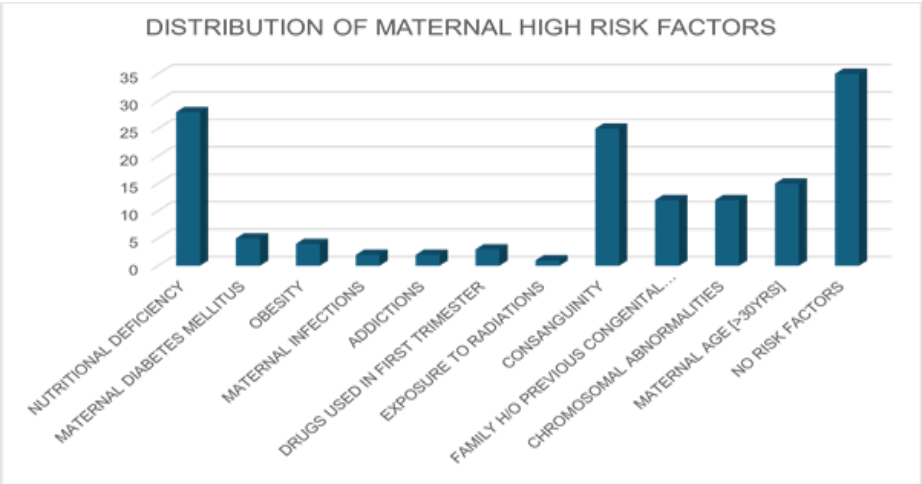
Table 1: Distribution of Gestational Age.



Distribution of Gestational Age (Weeks)	Frequency	Percentage
<20 Weeks	33	22.92%
20–24 Weeks	61	42.36%
24–28 Weeks	25	17.36%
28–37 Weeks	10	6.94%
>37 Weeks	15	10.41%
Total	**144**	**100%**

Table 1 illustrates the distribution of gestational age at diagnosis among cases. The majority of cases (42.36%) were diagnosed during the routine 20–24 weeks anomaly scan, while a significant proportion (22.92%) were identified before 20 weeks, suggesting that earlier screening is feasible but requires further standardization. This was followed by diagnoses between 24–28 weeks (17.36%) and a smaller proportion of cases were diagnosed between 28–37 weeks (6.94%), while (10.41%) were diagnosed after 37 weeks. This distribution suggests a clustering of diagnoses within the second-trimester anomaly scan window, reinforcing concerns regarding delayed detection.

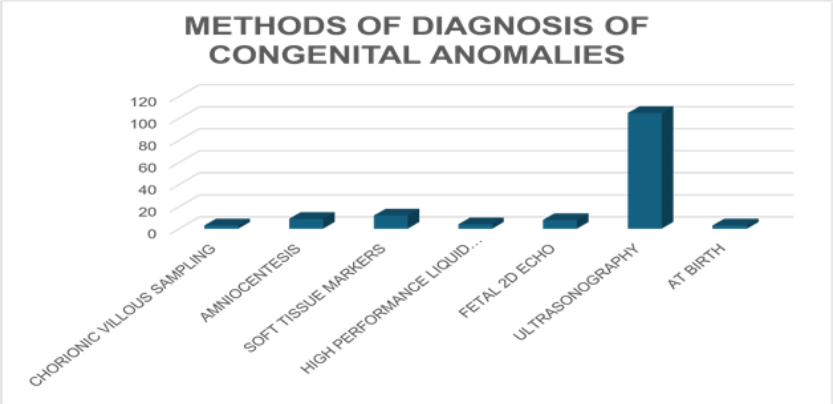
Table 2: Distribution of Maternal Risk Factors.



Distribution of Maternal Risk Factors	Frequency	Percentage
Modifiable Causes		
Nutritional Deficiency	28	19.44%
Maternal Diabetes Mellitus	5	3.47%
Obesity	4	2.78%
Maternal Infections	2	1.39%
Addictions	2	1.39%
Drugs Used in First Trimester	3	2.08%
Exposure to Radiation	1	0.69%
Non-Modifiable Causes		
Consanguinity	25	17.36%
Family History of Previous Congenital Anomaly	12	8.33%
Chromosomal Abnormalities	12	8.33%
Maternal Age [>30 years]	15	10.42%
No Risk Factors	35	24.31%
Total	**144**	**100%**

Table 2 depicts the distribution of maternal risk factors. Nutritional deficiency emerged as the most common modifiable risk factor (28 cases, 19.44%), followed by consanguinity (25 cases, 17.36%) and maternal age >30 years (15 cases, 10.42%). Family history of congenital anomalies and chromosomal abnormalities were each observed in 12 cases (8.33%). Other factors included maternal diabetes mellitus (5 cases, 3.47%), obesity (4 cases, 2.78%), and drug exposure in the first trimester (3 cases, 2.08%). Maternal infections and addictions were each noted in 2 cases (1.39%), while radiation exposure was noted in 1 case (0.69%). No identifiable risk factors were present in 35 cases (24.31%). Nutritional deficiency was identified based on maternal dietary history and clinical records of supplementation status.

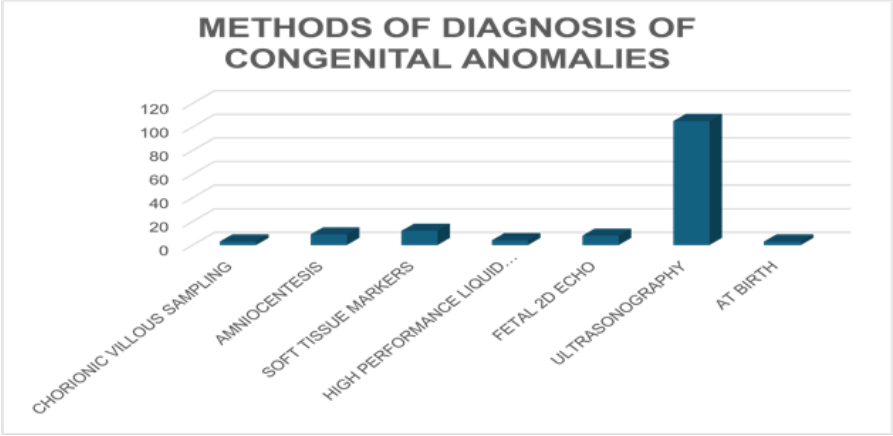
Table 3: Distribution of Methods of Diagnosis of Congenital Anomalies.



Distribution of Methods of Diagnosis of Congenital Anomalies	Frequency	Percentage
CVS	3	2.08%
Amniocentesis	9	6.25%
Ultrasound Soft Markers	12	8.33%
High-Performance Liquid Chromatography (HPLC)	4	2.77%
Fetal 2D Echo	8	5.55%
Ultrasonography	105	73%
At Birth	3	2.08%
Total	144	100%

Table 3 demonstrates the methods used for diagnosis of congenital anomalies. Ultrasonography was the most commonly used modality, accounting for 105 cases (73%). Ultrasound soft markers were identified in 12 cases (8.33%), while amniocentesis was performed in 9 cases (6.25%). Fetal 2D echocardiography was used in 8 cases (5.55%). High-performance liquid chromatography was utilized in 4 cases (2.77%), and chorionic villus sampling was performed in 3 cases (2.08%). In 3 cases (2.08%), anomalies were diagnosed at birth.

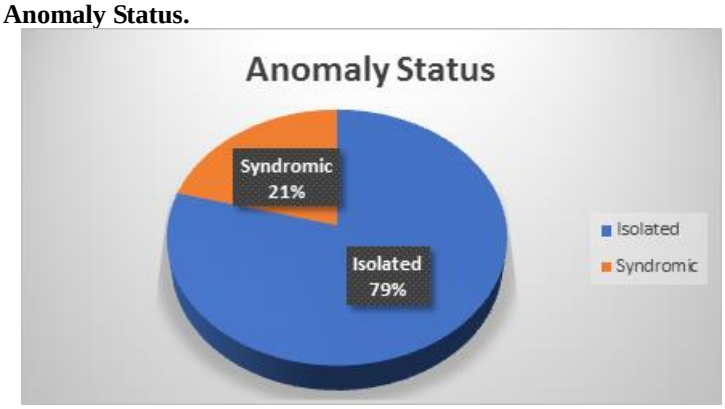
Table 4: Distribution of Birth Defect Categories.



Distribution of Birth Defect Categories	Frequency	Percentage
Central Nervous System	46	31.94%
Cardiovascular System	22	15.27%
Musculoskeletal System	16	11.11%
Gastrointestinal System	15	10.41%
Genitourinary System	15	10.41%
Genetic	12	8.33%
Respiratory System	4	2.77%
Lymphatic System	4	2.77%
Facial	3	2.08%
Hematopoietic	3	2.08%
Total	**144**	**100%**

Table 4 illustrates the distribution of birth defect categories. Central nervous system anomalies were the most common, observed in 46 cases (31.94%), followed by cardiovascular system anomalies in 22 cases (15.27%). Musculoskeletal anomalies were seen in 16 cases (11.11%), while gastrointestinal and genitourinary anomalies were each observed in 15 cases (10.41%). Genetic anomalies accounted for 12 cases (8.33%). Respiratory and lymphatic system anomalies were each observed in 4 cases (2.77%), while facial and hematopoietic anomalies were each noted in 3 cases (2.08%). The predominance of central nervous system anomalies may be attributed to the higher sensitivity of ultrasonography in detecting structural abnormalities such as neural tube defects.

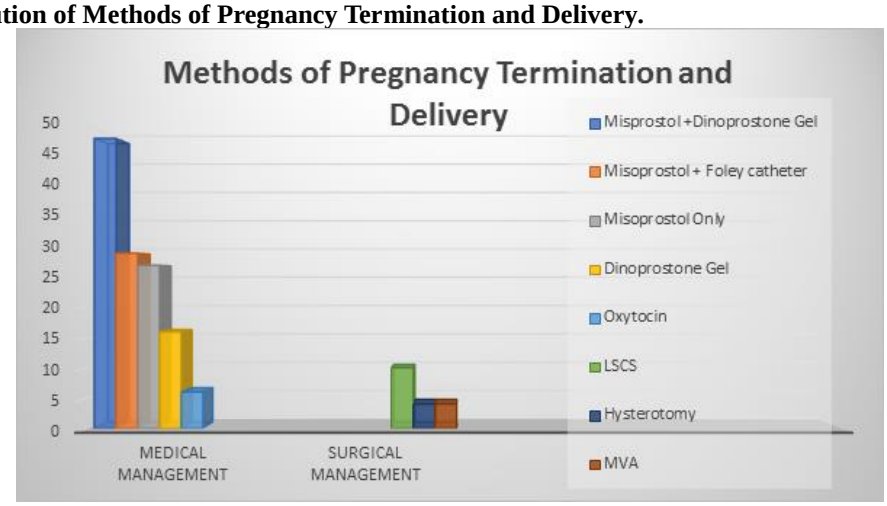
Table 5: Distribution of Anomaly Status.



Distribution of Anomaly Status	Frequency	Percentage
Isolated	114	79.17%
Syndromic	30	20.83%
Total	**144**	**100%**

Table 5 depicts the distribution of anomaly types. The majority of cases were isolated anomalies (114 cases, 79.17%), while syndromic anomalies were observed in 30 cases (20.83%).

Table 6: Distribution of Methods of Pregnancy Termination and Delivery.



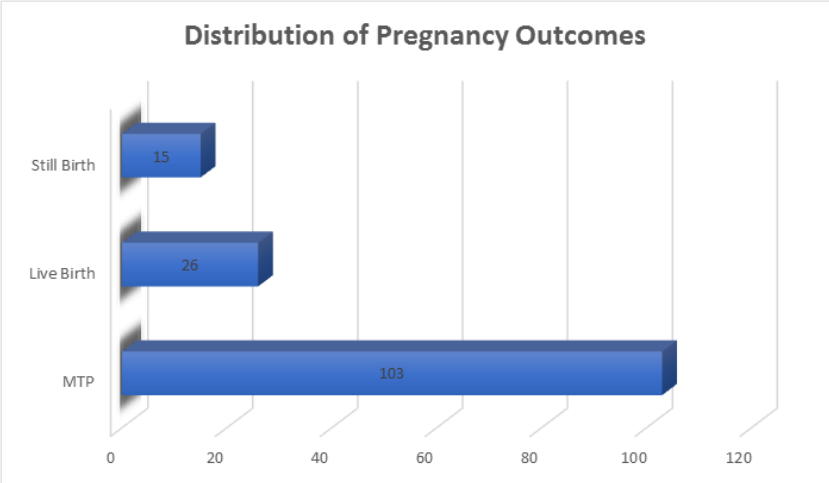
Distribution of Methods of Pregnancy Termination and Delivery	Frequency	Percentage
Medical Methods of Pregnancy Termination		
Misoprostol + Dinoprostone Gel	48	38%
Misoprostol + Foley Catheter	29	23%
Misoprostol Only	27	21.40%
Dinoprostone Gel Only	16	12.70%
Oxytocin	6	4.76%
Surgical Methods		
Lower Segment Cesarean Section (LSCS)	10	6.99%
Hysterotomy	4	2.80%
Manual Vacuum Aspiration (MVA)	4	2.80%

Table 6 demonstrates the methods of termination of pregnancy. Medical methods were most commonly used. The combination of misoprostol and dinoprostone gel was used in 48 cases (38%), followed by misoprostol with Foley catheter in 29 cases (23%) and misoprostol alone in 27 cases (21.4%). Dinoprostone gel alone was used in 16 cases (12.70%), while oxytocin was used in 6 cases (4.76%). Among surgical methods, lower segment cesarean section was performed in 10 cases (6.99%), while hysterotomy and manual vacuum aspiration were each performed in 4 cases (2.80%).

LSCS was performed for obstetric indications in viable pregnancies and represents mode of delivery rather than termination. Therefore, this table includes both termination methods and modes of delivery.

Percentages exceed 100% as multiple methods were used sequentially or in combination in individual cases.

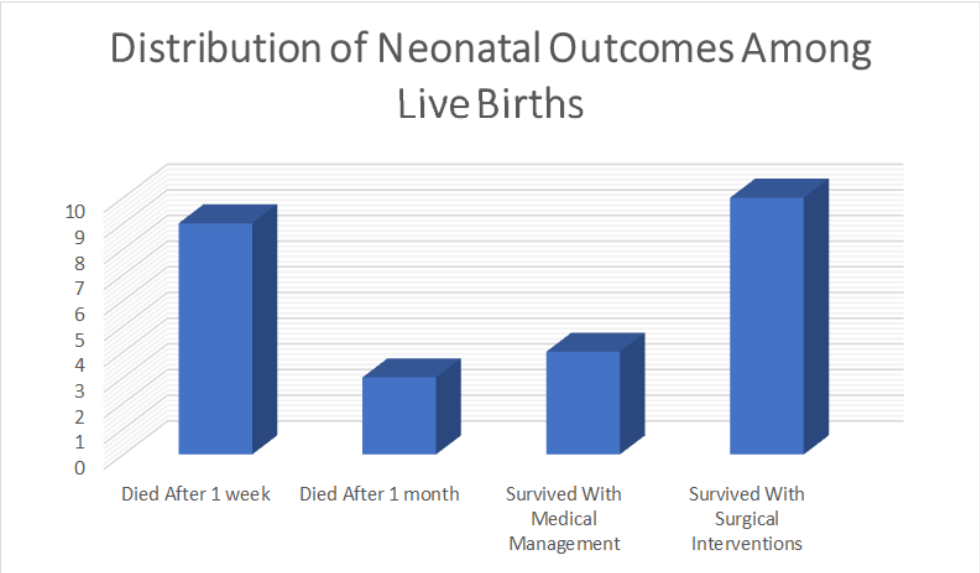
Table 7: Distribution of Pregnancy Outcomes (n=144)



Distribution of Pregnancy Outcomes (n=144)	Frequency	Percentage
MTP	103	71.53%
Live Birth	26	18.05%
Stillbirth	15	10.42%
Total	**144**	**100%**

Table 7 shows the distribution of pregnancy outcomes. MTP was the most common outcome, accounting for 103 cases (71.53%). Live births were recorded in 26 cases (18.05%), while stillbirths occurred in 15 cases (10.42%).

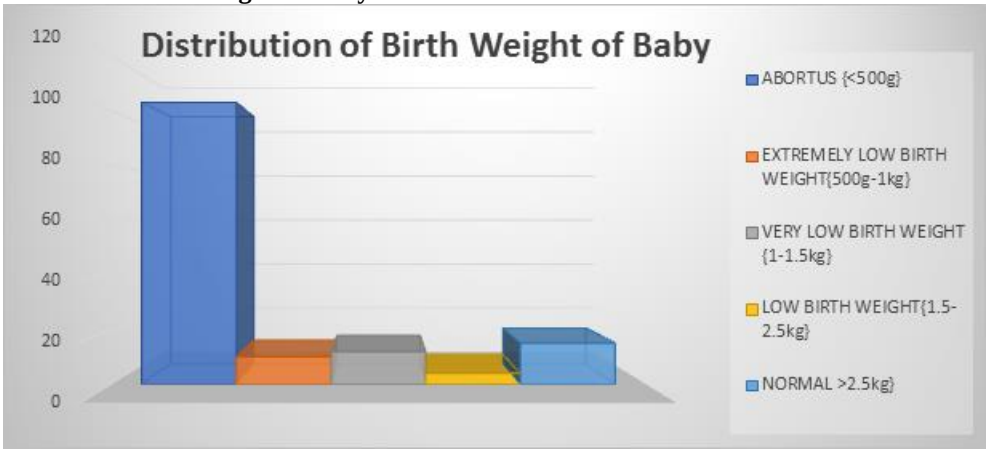
Table 8: Distribution of Neonatal Outcomes Among Live Births (n=26)



Distribution of Neonatal Outcomes Among Live Births (n=26)	Frequency	Percentage
Died After 1 week	9	34.62%
Died After 1 month	3	11.54%
Survived With Medical Management	4	15.38%
Survived With Surgical Interventions	10	38.46%

Fetal outcomes among live-born neonates (n = 26) demonstrated a high rate of early neonatal mortality. Among these, 9 neonates (34.62%) died within the first week of life, while 3 (11.54%) died within one month of life. Survival with conservative (medical) management was observed in 4 cases (15.38%), whereas 10 cases (38.46%) survived following surgical intervention. Despite some survival following intervention, overall outcomes remain suboptimal, reflecting limitations in neonatal care and surgical support. Although 38.46% of live-born neonates survived following surgical intervention, this corresponds to 6.90% of the total study population.

Table 9: Distribution of Birth Weight of Baby.



Distribution of Birth Weight of Baby (kg)	Frequency	Percentage
Abortus [<500 g]	103	71.53%
Extremely Low Birth Weight [500 g–1 kg]	10	6.90%
Very Low Birth Weight [1–1.5 kg]	12	8.3%
Low Birth Weight [1.5–2.5 kg]	4	2.8%
Normal [>2.5 kg]	15	10.40%
Total	**144**	**100%**

Table 9 demonstrates the distribution of birth weight among the study population. The majority of fetuses had a birth weight of less than 500 grams (103 cases, 71.53%). Extremely low birth weight (500 g–1 kg) was noted in 10 cases (6.90%), and very low birth weight (1–1.5 kg) in 12 cases (8.3%). Low birth weight (1.5–2.5 kg) was noted in 4 cases (2.8%), while normal birth weight (>2.5 kg) was noted in 15 cases (10.40%).

DISCUSSION

The present study evaluated the pattern, associated maternal risk factors, and outcomes of congenital anomalies in a tertiary care setting over an 18-month period. Ultrasonography emerged as the primary diagnostic modality (73%), reinforcing its pivotal role in routine antenatal screening, particularly in resource-limited settings.

Although the majority of cases (42.36%) were diagnosed during the standard 20–24 weeks anomaly scan window, this timing represents a clinically significant "late diagnosis" paradox.

The high proportion of second-trimester diagnoses reflects a gap between recommended first-trimester screening and real-world implementation in resource-limited settings.

As per the Medical Termination of Pregnancy (MTP) Act in India (amended 2021), termination for substantial fetal abnormalities is permitted up to 24 weeks under specified conditions.

Therefore, detection within the 20–24 weeks period creates a narrow decision window, often limiting access to safer early termination options.

Recent amendments permit termination up to 24 weeks for substantial fetal abnormalities under specified conditions, making early diagnosis crucial for safe and legally compliant decision-making.

Delayed diagnosis may be attributed to late antenatal registration, referral bias from peripheral centers, and inconsistent implementation of first-trimester screening protocols.

In addition to ultrasonography, first-trimester screening methods such as NT measurement and the double marker test play a crucial role in early risk assessment for chromosomal abnormalities.

Non-invasive prenatal testing (NIPT) has further enhanced detection rates for common aneuploidies. However, confirmatory diagnosis continues to rely on invasive procedures such as chorionic villus sampling (CVS) and amniocentesis. These findings are consistent with previous studies emphasizing the importance of combined screening strategies for early detection of congenital anomalies.[11]

A significant proportion of cases in the present study were associated with modifiable maternal risk factors. Nutritional deficiency was the most common risk factor identified (19.44%) highlighting the importance of micronutrient supplementation.[5,15] This often reflects late antenatal booking, resulting in missed opportunities for periconceptional folic acid and vitamin B12 supplementation, both of which are essential for the prevention of neural tube defects. Similar findings have been reported in other studies highlighting the role of micronutrient deficiencies in the development of congenital anomalies.[5,13,15]

Maternal diabetes and obesity were also observed, supporting the established association between metabolic disorders and adverse fetal outcomes.[16] Other contributing factors included maternal infections, early pregnancy drug exposure, and environmental influences, although these were less frequent.

Among non-modifiable factors, consanguinity (17.36%) and advanced maternal age were notable, reflecting their known association with genetic and chromosomal abnormalities. Consanguinity increases the expression of autosomal recessive disorders, underscoring the need for community-based pre-marital and preconception genetic counseling programs. Simultaneously, the high prevalence of nutritional deficiency highlights the importance of strengthening public health interventions, including food fortification with folic acid and vitamin B12, as well as ensuring adherence to periconceptional supplementation.

A dual-track strategy involving genetic literacy and nutritional advocacy is therefore essential. Integrating culturally sensitive counseling on consanguinity risks into primary healthcare, along with ensuring compliance with micronutrient supplementation, can serve as an effective primary preventive approach.

However, in 24.31% of cases, no identifiable risk factors were present, suggesting a multifactorial or idiopathic etiology.[13,19] This underscores the need for advanced genetic evaluation, such as chromosomal microarray analysis, in future studies.

Most anomalies in this study were isolated (79.17%), while 20.83% were syndromic, findings comparable to other studies conducted in tertiary care settings.[1,3] System-wise analysis revealed that central nervous system anomalies were the most common (31.94%), followed by cardiovascular and musculoskeletal anomalies. This pattern is consistent with previous reports by Singh et al. and Kanhere et al.[1,6,19]

The predominance of central nervous system anomalies in this study may be attributed to nutritional deficiencies and the relative ease of detection of these anomalies on routine ultrasonography. In contrast, studies from developed countries report a higher prevalence of cardiovascular anomalies, likely due to the availability of advanced diagnostic modalities such as fetal echocardiography.[10]

This variation reflects differences in healthcare infrastructure and screening practices influencing the observed pattern of congenital anomalies.

To better contextualize these findings, a comparison with trends observed in developed nations is presented below:

Table: Comparison of Present Study with Developed Countries.

Feature	Present Study (Nanded)	Developed Countries (Reported Trends)
Most Common Anomaly	Central Nervous System (31.94%)	Cardiovascular anomalies
Primary Risk Factors	Nutritional deficiency, Consanguinity	Genetic factors, Advanced maternal age
Peak Time of Diagnosis	20–24 weeks (Second trimester)	11–14 weeks (First trimester screening)
Predominant Outcome	MTP (71.53%)	Surgical correction and long-term survival

Most cases were diagnosed between 20–24 weeks of gestation, consistent with routine anomaly scan timing. The high rate of MTP (71.53%) reflects the detection of severe or lethal anomalies within the legally permissible gestational period. This finding aligns with previous studies conducted in tertiary care settings, where referral bias leads to a higher concentration of complex cases.

While medical methods using Misoprostol and Dinoprostone were most common, surgical methods like LSCS and hysterotomy were utilized when specific

maternal clinical indications or previous uterine scarring precluded medical induction.

Fetal survival was closely associated with the severity of anomalies and the availability of timely neonatal intervention.[8,19] However, the low overall survival rate (6.90% of total study population) observed in the present study is consistent with findings from similar neonatal care settings[17] which underscores the significant gaps in neonatal surgical infrastructure and specialized care services.

Despite antenatal diagnosis, limited access to advanced neonatal intensive care and timely surgical intervention continues to restrict survival in potentially correctable conditions.[14,17] Furthermore, fetal outcomes among non-MTP cases demonstrated high early neonatal mortality, as reported in other neonatal intensive care studies[17] with a substantial proportion of deaths occurring in the immediate postnatal period. This underscores the critical need for strengthening neonatal care facilities and establishing streamlined referral pathways to improve survival outcomes.

To improve outcomes, a streamlined referral pathway is essential. Fetuses diagnosed with surgically correctable anomalies should be delivered at centers equipped with advanced neonatal surgical facilities. Establishing Level III neonatal surgical units integrated with obstetric services can facilitate immediate intervention and reduce neonatal mortality.

Although 18.05% of cases resulted in live births, true survival remained low, with only 6.90% surviving after surgical intervention. In the absence of long-term follow-up, survival in this study primarily reflects the immediate neonatal period or survival until hospital discharge. Future studies incorporating long-term neurodevelopmental outcomes are necessary to assess the quality of survival. These findings emphasize a gap between antenatal detection and neonatal survival in resource-constrained settings.

Despite advancements in prenatal diagnostics, several challenges persist in resource-limited settings. Late antenatal registration, limited awareness of routine screening, and inadequate access to healthcare services contribute to delayed diagnosis and suboptimal outcomes. These barriers are widely reported in developing countries and underscore the need to strengthen antenatal care systems.[14]

For practical implementation, this proposed "Routine implementation of First Trimester Combined Screening" protocol can be integrated into national programs such as the Pradhan Mantri Surakshit Matritva Abhiyan (PMSMA). Incorporating first-trimester screening (11–14 weeks) into routine antenatal visits would enable early risk stratification at the primary care level. Capacity building of healthcare providers, establishment of structured referral pathways, and community-level awareness through frontline workers are essential for successful implementation.

From a public health perspective, improving maternal nutrition, promoting early antenatal registration, and ensuring universal access to prenatal screening are critical strategies to reduce the burden of congenital anomalies. Strengthening primary healthcare systems and expanding access to tertiary care services are essential for improving maternal and neonatal outcomes.

Identified Problem	Research Finding	Proposed Solution
Late diagnosis	42.36% detected at 20–24 weeks	Mandatory 11–14 weeks NT screening
High CNS anomalies	31.94% of total cases	Universal folic acid & vitamin B12 fortification
Low post-surgical survival	Only 6.90% survival	Establish dedicated pediatric surgical ICUs at tertiary centers
Unidentified risk factors	24.31% with no known risk	Universal antenatal USG screening for all registrations

The findings of this study should be interpreted in light of certain limitations, including its single-center design, relatively small sample size, and lack of long-term neonatal follow-up. Despite these limitations, the study provides valuable insights into the pattern and outcomes of congenital anomalies in a resource-limited setting.

Recommendations

- Routine implementation of first-trimester combined screening (11–14 weeks) using nuchal translucency (NT) measurement and biochemical markers to enable early detection of congenital anomalies.
- Ensure universal access to second-trimester anomaly scans (18–20 weeks) to strengthen detection of structural abnormalities.
- Promote early antenatal registration (before 12 weeks gestation) through community-based awareness programs.
- Strengthen periconceptional folic acid and vitamin B12 supplementation, along with consideration of food fortification strategies, to reduce neural tube defects.
- Establishment of dedicated neonatal intensive care units (NICUs) and pediatric surgical facilities at

tertiary care centers to improve survival in correctable anomalies.
- Develop structured referral pathways from primary and secondary care centers to higher-level institutions for timely management of high-risk pregnancies.
- Implementation of genetic counseling services, particularly in populations with a high prevalence of consanguinity.
- Creation of regional and national congenital anomaly registries to improve surveillance, research, and policy planning.
- Integration of prenatal screening protocols into national health programs such as PMSMA to ensure standardized care delivery.

Future Scope
- Conducting multicentric studies with larger sample sizes to enhance generalizability of findings.
- Incorporation of advanced genetic diagnostic techniques, including chromosomal microarray and next-generation sequencing, for better etiological understanding.
- Establishment of long-term neonatal follow-up programs to assess survival, neurodevelopmental outcomes, and quality of life.
- Strengthening preconception care clinics focusing on risk assessment, nutritional optimization, and genetic counseling.
- Evaluating the effectiveness of first-trimester screening implementation within existing national programs.
- Expanding access to fetal medicine services and fetal echocardiography for improved early detection of complex anomalies.

Strengths of the Study
- Prospective study design, allowing systematic data collection and temporal assessment.
- Provides real-world data from a tertiary care setting, reflecting practical clinical scenarios.
- Comprehensive evaluation of maternal risk factors, including both modifiable and non-modifiable determinants.
- Detailed system-wise classification of congenital anomalies, enhancing clinical relevance.
- Inclusion of maternal, fetal, and immediate neonatal outcomes, offering a holistic overview.

Limitations of the Study
- Single-center study, which may limit generalizability to other settings.
- Relatively small sample size (n = 144), potentially affecting statistical power.
- Lack of genetic confirmation (e.g., karyotyping) in the majority of cases limits precise etiological classification.
- Referral bias potentially overestimating anomaly severity, termination rates, and late diagnosis trends, thereby limiting generalizability.
- Absence of long-term neonatal follow-up, restricting assessment of survival beyond hospital discharge and long-term outcomes.
- Lack of a control group, limiting the ability to establish causal associations between maternal risk factors and congenital anomalies.
- Observational design, which precludes definitive causal inference.

Despite these limitations, the study offers meaningful data relevant to similar healthcare settings.

CONCLUSION

Congenital anomalies remain a significant contributor to adverse pregnancy outcomes, with central nervous system anomalies being the most prevalent in this cohort (31.94%). The high rate of MTP (71.53%) reflects both the severity of detected anomalies and the critical influence of the timing of diagnosis. Notably, most anomalies (42.36%) were identified during the routine 20–24-week scan, a period that limits opportunities for early intervention and first-trimester counseling.

The overall survival rate (6.90% of total study population) among live births highlights substantial gaps in neonatal surgical infrastructure and specialized care. This disparity between antenatal detection and neonatal survival outcomes underscores the urgent need to strengthen neonatal intensive care and pediatric surgical services at tertiary care centers.

Additionally, the occurrence of anomalies in 24.31% of cases without identifiable risk factors emphasizes the importance of universal antenatal screening, irrespective of maternal risk profile.

To reduce the burden of congenital anomalies, emphasis should be placed on early first-trimester screening (11–14 weeks), periconceptional folic acid and vitamin B12 supplementation, and community-based counseling regarding consanguinity. A multidisciplinary approach involving obstetricians, radiologists, and pediatric surgeons is essential to improve maternal and neonatal outcomes. Bridging the gap between early detection and timely intervention remains critical to reducing preventable fetal morbidity and improving perinatal outcomes in resource-limited settings.

Summary of key findings
- Central nervous system anomalies were the most common (31.94%), followed by cardiovascular anomalies (15.27%).
- The majority of congenital anomalies (42.36%) were diagnosed during the routine second-trimester scan (20–24 weeks of gestation).
- Nutritional deficiency (19.44%) was the leading modifiable maternal risk factor, highlighting the importance of periconceptional supplementation.

- A high rate of MTP (71.53%) was noted, reflecting the detection of severe anomalies in a tertiary care setting.
- Overall survival rate was 6.90% of the total study population, indicating significant gaps in neonatal surgical and intensive care infrastructure.
- A considerable proportion of cases (24.31%) had no identifiable risk factors, supporting the need for universal antenatal screening.

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