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Prevalence and Pattern of Congenital Anomalies Detected in Antenatal Ultrasound: A Hospital-Based Cross-Sectional Study

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ABSTRACT

Background: Congenital anomalies are a significant cause of perinatal morbidity and mortality, particularly in low- and middle-income countries. Antenatal ultrasound is a vital tool for early detection, allowing for timely counseling and intervention. This study aimed to assess the prevalence and distribution of congenital anomalies detected during routine second-trimester ultrasounds at a tertiary care center.

Methods: A cross-sectional study was conducted in the Radiodiagnosis Department at a tertiary hospital over a defined study period. All pregnant women between 18 and 24 weeks of gestation undergoing routine anomaly scans were included. Ultrasounds were performed using standard ISUOG guidelines. Anomalies were categorized by organ system, and data were analyzed descriptively.

Results: Out of 2,000 antenatal scans, 82 fetuses were found to have congenital anomalies, yielding a prevalence of 4.1%. The most frequently involved systems were the central nervous system (31.7%), followed by the genitourinary system (20.7%), the musculoskeletal system (14.6%), and the gastrointestinal system (9.8%). Isolated anomalies were noted in 84.1% of cases, while 15.9% had multiple system involvement.

Conclusion: Routine second-trimester ultrasound is an effective screening tool for detecting major congenital anomalies. Central nervous system defects remain the most prevalent. Early detection facilitates prenatal counseling, risk stratification, and multidisciplinary care planning to optimize perinatal outcomes.

Key-words: Congenital anomalies, antenatal ultrasound, anomaly scan, fetal malformations, prenatal diagnosis

INTRODUCTION

Congenital anomalies, also referred to as congenital disabilities, are structural or functional abnormalities that occur during intrauterine life and are detectable prenatally, at birth, or later in infancy.

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They remain a significant cause of neonatal morbidity and mortality worldwide, particularly in low- and middle-income countries where access to routine prenatal screening may be limited [1]. The global prevalence of congenital anomalies ranges from 3 to 7% of all live births, with variations depending on geographic, environmental, nutritional, and genetic factors [2].

Timely detection of fetal anomalies plays a pivotal role in prenatal counseling, perinatal management, and planning of postnatal interventions. Second-trimester ultrasound between 18 and 22 weeks of gestation is considered the standard window for anomaly screening, offering optimal fetal anatomical visualization [3]. Advances in ultrasonographic resolution and adherence

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to systematic scanning protocols have significantly improved detection rates, especially for central nervous system, genitourinary, and skeletal malformations [4].

Although several studies from high-income countries have provided detailed epidemiological data on congenital anomalies, there is a relative paucity of such information from resource-limited settings [5]. Hospital-based studies offer valuable insights into regional patterns, types of anomalies most commonly detected, and the utility of ultrasound as a screening tool. These findings not only aid in early diagnosis and timely referral but also guide health policy and resource allocation for specialized maternal-fetal care units [6].

In India, congenital anomalies contribute substantially to neonatal deaths and lifelong disability, yet antenatal detection rates vary widely across institutions [7]. Understanding local prevalence and the spectrum of detectable anomalies can help improve sonographic practices and identify potential areas for public health intervention.

This study aims to determine the prevalence and pattern of congenital anomalies detected during routine second-trimester antenatal ultrasound at a tertiary care center, thereby contributing to regional epidemiological data and enhancing clinical preparedness for affected pregnancies.

MATERIALS AND METHODS

Study Design and Setting- This hospital-based cross-sectional study was conducted at a tertiary care medical center in India. All pregnant women attending the antenatal clinic and referred for routine anomaly scan between 18 and 24 weeks of gestation were eligible for inclusion. The anomaly scans were conducted as part of routine prenatal screening during the second trimester.

Inclusion Criteria

- Singleton and multiple pregnancies between 18 and 24 weeks of gestation
- Women consenting to participate in the study
- Antenatal scans performed using standardized fetal anomaly protocol

Exclusion Criteria

- ✓ Scans with incomplete fetal visualization due to maternal obesity, oligohydramnios, or suboptimal fetal position
- ✓ Women with known chromosomal abnormalities from prior testing (to avoid bias in pattern analysis)
- ✓ Referrals made post 24 weeks for suspected anomaly (to ensure consistency in timing)

Ultrasound Protocol- All anomaly scans were performed using high-resolution ultrasound machines equipped with curvilinear transducers (3.5–5 MHz). Each scan was conducted by an experienced radiologist or a fetal medicine specialist using a standardized fetal anomaly checklist based on the guidelines of the International Society of Ultrasound in Obstetrics and Gynecology (ISUOG).

Data Collection- Patient demographic details (age, parity, gestational age), ultrasound findings, type and number of anomalies, and presence of multiple anomalies were recorded in a structured proforma. Each anomaly was classified according to the organ system involved. Cases with multiple anomalies were recorded under the “multiple systems involved” category as a separate entry.

Outcome Measures- The primary outcome was the prevalence of congenital anomalies detected during routine second-trimester ultrasound examinations. Secondary outcomes included the distribution of anomalies by system, the frequency of isolated versus multiple anomalies, and maternal demographic correlations.

Statistical Analysis- Data were compiled using Microsoft Excel and analyzed. Descriptive statistics were used to calculate frequencies, percentages, and mean values. Chi-square test or Fisher’s exact test was applied to determine associations between categorical variables, and $p<0.05$ was considered statistically significant.

RESULTS

A total of 2,000 antenatal anomaly scans were evaluated during the study period. Congenital anomalies were detected in 82 fetuses, yielding a prevalence of 4.1%. Among these, single-system anomalies were noted in 69 cases (84.1%), while multiple anomalies were identified in 13 cases (15.9%). Congenital anomalies were present in 4.1% of second-trimester anomaly scans (Table 1).

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Table 1: Prevalence of Congenital Anomalies in Antenatal Ultrasound (N = 2000)

Findings	Number of Cases	Percentage (%)
Total anomaly scans performed	2000	100.0
Congenital anomalies detected	82	4.1
No anomaly detected	1918	95.9

CNS anomalies were the most commonly detected, followed by genitourinary and musculoskeletal defects. Multiple system involvement was seen in nearly 16% of cases (Table 2). Anencephaly was the most prevalent CNS anomaly, followed by hydrocephalus and spina bifida (Table 3).

Table 2: Distribution of Anomalies by Organ System (N = 82)

System Involved	Number of Cases	Percentage (%)
Central Nervous System (CNS)	26	31.7
Genitourinary System	17	20.7
Musculoskeletal System	12	14.6
Gastrointestinal System	8	9.8
Cardiovascular System	6	7.3
Craniofacial (e.g. cleft)	5	6.1
Thoracic Abnormalities	3	3.7
Multiple System Involvement	13	15.9

Table 3: Types of Central Nervous System Anomalies (n = 26)

CNS Anomaly	Number of Cases	Percentage (%)
Anencephaly	8	30.8
Hydrocephalus	6	23.1
Dandy-Walker Malformation	4	15.4
Encephalocele	3	11.5
Spina Bifida	5	19.2

The majority of fetuses had isolated single-organ involvement, but a notable proportion had complex multisystem anomalies requiring multidisciplinary planning (Table 4).

Table 4: Number of Anomalies per Fetus (N = 82)

Number of Anomalies	Number of Fetuses	Percentage (%)
Isolated anomaly	69	84.1
Multiple anomalies	13	15.9

Fig. 1 presents a typical antenatal ultrasound scan performed during the second trimester as part of the anomaly screening protocol. This includes standard biometric parameters and a structural evaluation by ISUOG guidelines.

Additionally, a rare but significant anomaly—conjoined twins—was also identified during the study period, as shown in Fig. 2, highlighting the critical role of mid-trimester ultrasound in early diagnosis of complex malformations.

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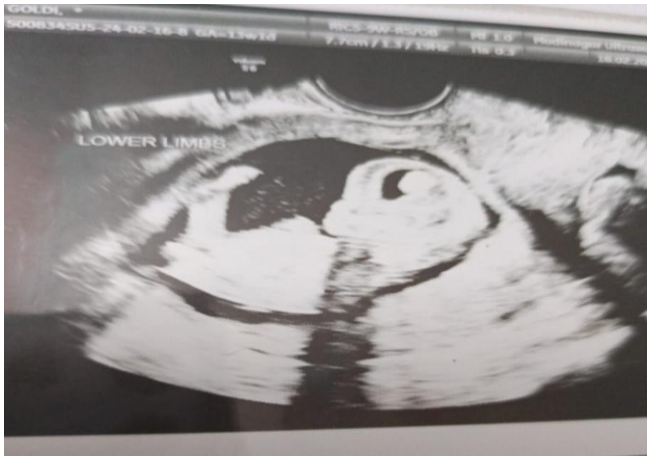

Fig. 1: Antenatal Ultrasound

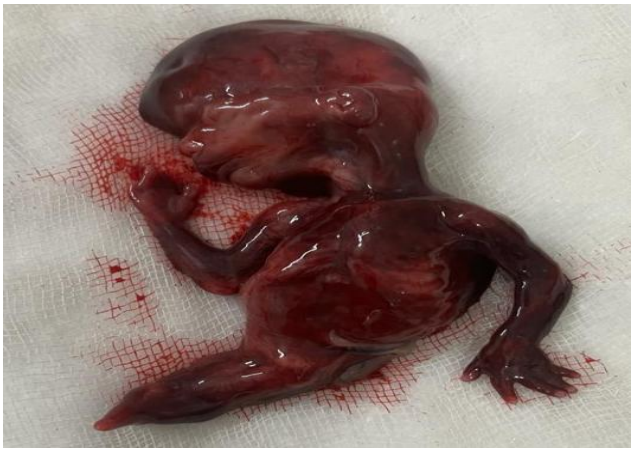

Fig. 2: Marmaid foetus

DISCUSSION

This hospital-based cross-sectional study found a prevalence of congenital anomalies of 4.1% in routine second-trimester ultrasounds, aligning with figures from other tertiary-care studies in India and elsewhere. Mahela *et al*. reported a 1.73% anomaly rate in Assam [7], while Sreekala's work in Kerala found a similar 1.6% rate [8]. Our slightly higher prevalence likely reflects differing referral patterns and patient demographics in tertiary centers.

Consistent with global and Indian data, central nervous system (CNS) anomalies were most frequently detected. Mahela et al. also highlighted CNS defects, which comprised 42% of anomalies, predominantly anencephaly [7]. This aligns with meta-analyses estimating neural tube defect rates around 4.1 per 1,000 births in India, emphasizing the critical need for early detection and folic acid supplementation [9].

Renal and musculoskeletal anomalies followed CNS defects in prevalence, mirroring observations in rural tertiary settings, where renal anomalies comprised 31.4% and cardiac anomalies 24.9% [10]. These data underscore the value of structured anomaly scanning, with a focus on multiple fetal systems.

The detection of isolated anomalies in over 84% of cases signifies that targeted interventions and counseling may be applicable for most affected fetuses. The remaining 16% with multisystem involvement support the need for comprehensive evaluation and multidisciplinary referral.

Elevated detection rates in our setting likely stem from systematic use of high-resolution ultrasound and fetal MRI for complex cases. This is consistent with trends observed in similar tertiary-care environments in low- and middle-income countries where enhanced imaging capability leads to improved diagnostic yield [10].

Our findings reinforce the importance of routine mid-trimester scanning (18–24 weeks) combined with comprehensive protocols. They further underscore the need to reinforce folic acid supplementation programs, which have been shown to reduce neural tube defects by up to 70% globally [11].

Given that ultrasound screening identified severe anomalies permitting early counseling or termination, approximately 20% of affected pregnancies achieved this outcome [10]. This highlights the beneficial role of prenatal ultrasound in managing public health and maternal–fetal outcomes, especially when coupled with post-scan counseling and access to specialized services [11-14].

CONCLUSIONS

This study demonstrates that routine second-trimester ultrasound is effective in identifying a wide spectrum of congenital anomalies, with a prevalence of 4.1% in our population. Central nervous system defects were the most commonly detected anomalies, followed by genitourinary and musculoskeletal malformations. Most cases involved isolated single-system anomalies, allowing for focused prenatal counseling and management. The use of standardized scanning protocols significantly enhances detection rates, even in resource-limited settings. Early identification enables informed decision-making, timely intervention, and improved perinatal outcomes.

CONTRIBUTION OF AUTHORS

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Article editing- Archana Tiwari, Vishnu Kumar Gupta
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