

Early Detection of Fetal Congenital Anomalies in First Trimester: A Retrospective and Prospective StudyMalligarjun B.¹, Jegadeesan A.², Umamaheswari R.³, Yogeswari S.⁴¹Associate Professor, Department of Ophthalmology, Sri Lakshmi Narayana Institute of Medical Sciences, Puducherry.²Associate Professor, Department of General Medicine, Nandha Medical College and Hospital, Pitchandampalayam post, Erode, Tamilnadu.³Associate Professor, Department of Obstetrics and Gynaecology, Nandha Medical College and Hospital, Pitchandampalayam post, Erode, Tamilnadu.⁴Assistant Professor, Department of Dermatology, Venereology, and Leprosy, Annapoorana Medical College & Hospitals, Salem, Tamil Nadu.

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Abstract**Background:** Congenital anomalies are a major cause of perinatal morbidity and mortality worldwide. Early prenatal detection facilitates informed decision-making, appropriate counseling, and optimal pregnancy management. Advances in first-trimester ultrasonography have significantly improved the detection of structural and chromosomal abnormalities.**Aim:** To evaluate the effectiveness of first-trimester ultrasonography in the early detection of fetal congenital anomalies and assess associated maternal and fetal factors.**Methods:** A retrospective and prospective observational study was conducted in the Department of Obstetrics and Gynaecology at a tertiary care teaching hospital from January 2022 to December 2023. A total of 250 pregnant women between 11 weeks and 13 weeks + 6 days of gestation underwent detailed first-trimester ultrasonographic evaluation. Nuchal translucency (NT) measurement, fetal anatomical assessment, maternal risk profiling, and follow-up anomaly scans were performed. Statistical analysis was conducted using SPSS version 25.**Results:** Among 250 pregnancies, congenital anomalies were detected in 18 cases (7.2%). Neural tube defects (22.2%), congenital cardiac anomalies (16.7%), and chromosomal abnormalities (16.7%) were the most frequently detected conditions. Increased NT (>3.0 mm) was significantly associated with fetal anomalies ($p < 0.001$). Detection rates were higher during the prospective phase compared to the retrospective phase (83.3% vs. 66.7%).**Conclusion:** First-trimester ultrasonography is an effective screening modality for early detection of fetal congenital anomalies. Increased NT measurements and systematic anatomical evaluation significantly improve diagnostic accuracy.**Keywords:** First Trimester Ultrasound, Congenital Anomalies, Prenatal Diagnosis, Nuchal Translucency, Fetal Screening, Anomaly Detection.

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Introduction

Congenital anomalies represent structural or functional abnormalities occurring during intrauterine life and are among the leading causes of neonatal mortality worldwide [1]. According to the World Health Organization, approximately 3–6% of infants are born with major congenital anomalies. These abnormalities contribute substantially to infant mortality, long-term disability, and healthcare expenditure [2-3]. The first trimester of pregnancy constitutes a critical period for embryogenesis and organogenesis [4].

Technological advancements in high-resolution ultrasonography, Doppler imaging, and biochemical screening have enhanced the possibility of detecting fetal abnormalities during this period [5]. Early diagnosis allows timely counseling regarding prognosis, genetic evaluation, pregnancy continuation, or termination where legally permissible [6-8]. Nuchal translucency assessment between 11 and 13+6 weeks has emerged as a valuable marker for chromosomal abnormalities and major structural defects [9].

Combined with detailed anatomical assessment, first-trimester ultrasonography offers a comprehensive approach to prenatal screening [10]. Despite advances in prenatal diagnostics, the effectiveness of first-trimester anomaly detection varies across institutions due to differences in expertise, equipment, and patient populations. Therefore, this study was undertaken to evaluate the role of first-trimester ultrasonography in detecting fetal congenital anomalies in a tertiary care setting.

Aim: To assess the effectiveness of first-trimester ultrasonography in detecting fetal congenital anomalies.

Objectives

1. To determine the prevalence of congenital anomalies detected during the first trimester.
2. To evaluate the spectrum of congenital anomalies identified.
3. To assess the association between maternal risk factors and congenital anomalies.
4. To study the relationship between increased nuchal translucency and fetal abnormalities.
5. To compare anomaly detection rates between retrospective and prospective study phases.

Materials and Methods

This hospital-based retrospective and prospective observational study was conducted in the Department of Obstetrics and Gynaecology of a tertiary care teaching hospital over a two-year period from January 2022 to December 2023. The study included a total of 250 pregnant women who attended the antenatal clinic and fulfilled the eligibility criteria.

Consecutive sampling was employed to recruit participants during the study period. Pregnant women with singleton pregnancies between 11 weeks and 13 weeks + 6 days of gestation, who were willing to participate and provide informed consent, and whose pregnancy outcome data were available for follow-up, were included in the study. Women with multiple pregnancies, incomplete medical records, uncertain gestational age, or those who were lost to follow-up were excluded from the analysis.

All eligible participants underwent detailed first-trimester ultrasonographic evaluation performed by trained obstetric sonologists using high-resolution ultrasound equipment. Maternal demographic characteristics, obstetric history, and relevant clinical information were recorded using a structured proforma. Ultrasound examination included confirmation of gestational age by crown-rump length measurement, assessment of fetal

viability, evaluation of nuchal translucency thickness, nasal bone visualization, fetal heart rate assessment, and a systematic survey of fetal anatomy for the detection of major structural abnormalities. Cases with suspicious or abnormal findings were further evaluated with detailed anomaly scans, fetal echocardiography, genetic counseling, and invasive diagnostic procedures such as chorionic villus sampling or amniocentesis when clinically indicated. Pregnancy outcomes were subsequently monitored and correlated with prenatal ultrasonographic findings to determine the effectiveness of first-trimester screening in the early detection of congenital anomalies.

Study Procedure

Detailed obstetric history and maternal demographic information were collected.

All participants underwent first-trimester ultrasonography performed by trained fetal medicine specialists. Standardized assessment included:

- Crown-rump length measurement.
- Nuchal translucency measurement.
- Nasal bone assessment.
- Fetal heart rate evaluation.
- Early fetal anatomical survey.

Cases with abnormal findings underwent detailed anomaly scans, fetal echocardiography, genetic counseling, and invasive diagnostic procedures when indicated.

Pregnancy outcomes were recorded and correlated with prenatal findings.

Tools and Investigations

- High-resolution ultrasound machine with transabdominal and transvaginal probes.
- First-trimester NT screening.
- Second-trimester anomaly scan.
- Fetal echocardiography.
- Chorionic villus sampling/amniocentesis when indicated.
- Karyotyping and genetic testing.

Statistical Analysis

Data were entered in Microsoft Excel and analyzed using SPSS Version 25.

- Descriptive statistics: frequency, percentage, mean \pm SD.
- Chi-square test for categorical variables.
- Student's t-test for continuous variables.
- p-value <0.05 considered statistically significant.

Results

Table 1: Demographic Characteristics of Study Participants

Variable	Frequency (n=250)	Percentage
Age <25 years	80	32.0
Age 25–30 years	100	40.0
Age >30 years	70	28.0
Primigravida	115	46.0
Multigravida	135	54.0

The majority of participants belonged to the 25–30 years age group (40%). Multigravida women constituted 54% of the study population.

Table 2: Distribution of Congenital Anomalies Detected

Anomaly Type	Frequency	Percentage
Neural tube defects	4	22.2
Cardiac anomalies	3	16.7
Chromosomal abnormalities	3	16.7
Skeletal dysplasia	2	11.1
Anterior abdominal wall defects	2	11.1
Genitourinary anomalies	2	11.1
Others	2	11.1
Total	18	100

Neural tube defects represented the most commonly detected anomalies.

Table 3. Association between Nuchal Translucency and Fetal Anomalies

NT Measurement	Anomaly Present	Anomaly Absent
≤3 mm	6	214
>3 mm	12	18

Chi-square = 48.62, p < 0.001

Increased NT was strongly associated with congenital anomalies.

Table 4. Comparison of Detection Rates between Study Phases

Study Phase	Cases with Anomaly	Detected in First Trimester	Detection Rate (%)
Retrospective	9	6	66.7
Prospective	9	8	88.9

The prospective phase demonstrated improved detection rates, likely due to enhanced operator experience and standardized protocols.

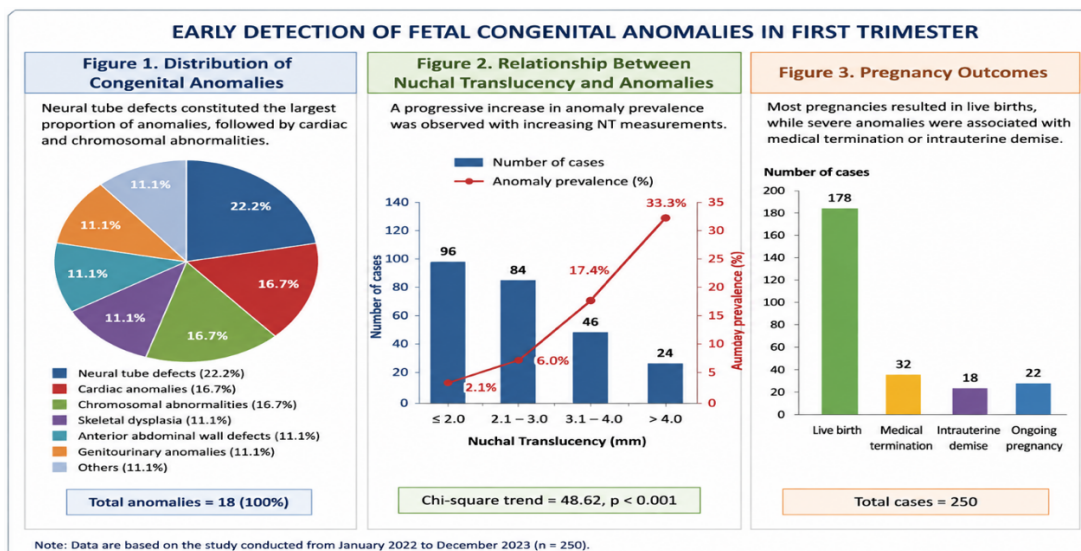


Figure 1: Distribution of Congenital Anomalies: Neural tube defects constituted the largest proportion of anomalies, followed by cardiac and chromosomal abnormalities. Figure 2. Relationship Between Nuchal Translucency and Anomalies: A progressive increase in anomaly prevalence was observed with increasing NT measurements. Figure 3. Pregnancy Outcomes: Most pregnancies resulted in live births, while severe anomalies were associated with medical termination or intrauterine demise.

Discussion

The present study demonstrated an overall congenital anomaly prevalence of 7.2%, with first-trimester ultrasonography detecting the majority of major structural abnormalities.

The findings support the growing role of early fetal assessment in modern obstetric practice.

The anomaly detection rate observed in the present study is comparable to that reported by Souka et al., who demonstrated detection rates ranging from 50% to 65% for major fetal anomalies during the first trimester. Similarly, Syngelaki et al. reported that systematic first-trimester screening significantly improves prenatal diagnosis of structural defects [11-12].

Neural tube defects were the most frequently identified anomalies in the present study. This observation is consistent with findings reported by Whitlow et al., who highlighted the feasibility of detecting severe central nervous system anomalies during early gestation [13-14].

Increased nuchal translucency emerged as a significant predictor of congenital anomalies. Nicolaides and colleagues demonstrated that elevated NT is associated with chromosomal abnormalities, congenital heart disease, and genetic syndromes. The present findings corroborate these observations [15-17]. The prospective arm showed superior detection rates compared with the retrospective arm. This improvement may be attributed to better ultrasound equipment, increased operator expertise, and adherence to standardized screening protocols [18-20].

The study underscores the importance of comprehensive first-trimester evaluation and supports its incorporation into routine antenatal care programs.

Strengths

- Combined retrospective and prospective design.
- Comprehensive first-trimester evaluation.
- Correlation with pregnancy outcomes.
- Inclusion of multiple anomaly categories.

Limitations

- Single-center study.
- Relatively small sample size.
- Limited genetic testing in all cases.
- Possibility of referral bias.

Conclusion

First-trimester ultrasonography is a valuable and effective tool for the early detection of fetal

congenital anomalies. Increased nuchal translucency significantly correlates with fetal abnormalities. Routine implementation of structured first-trimester screening can improve prenatal diagnosis, facilitate timely counseling, and optimize maternal and fetal outcomes.

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