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DIAGNOSTIC UTILITY OF FIRST-TRIMESTER ULTRASOUND IN THE EARLY DETECTION OF MAJOR FETAL ANOMALIES: A CROSS-SECTIONAL EVALUATION

Obstetrics & Gynaecology

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ABSTRACT

Background: First-trimester ultrasound is increasingly used for early detection of major fetal anomalies, offering clinical advantages over traditional second-trimester scans. **Objectives:** To assess the diagnostic utility of first-trimester ultrasound in identifying major structural fetal anomalies. **Methods:** This hospital-based cross-sectional observational study was conducted over one year in the Department of Obstetrics and Gynaecology, Rohilkhand Medical College and Hospital, Bareilly. A total of 250 pregnant women between 9+0 and 13+6 weeks of gestation were included. All participants underwent detailed first-trimester ultrasonography using high-resolution transabdominal and transvaginal probes. NT measurement and biochemical markers were assessed in selected cases. Anomalies were documented and analyzed. **Results:** Out of 250 participants, 20% were found to have major fetal anomalies in the first trimester. The most commonly detected anomalies were anencephaly (10%), acrania (6.4%), and abnormal nuchal translucency (6.4%). Significant associations were observed between anomaly detection and abnormal CRL and MGS measurements ($p<0.05$). A history of congenital anomalies also showed a strong correlation with first-trimester anomaly detection. **Conclusion:** First-trimester ultrasound is a reliable and effective modality for early detection of major fetal anomalies. Its integration into routine antenatal care—particularly for high-risk populations—may significantly enhance prenatal screening protocols and improve perinatal outcomes.

KEYWORDS

First-trimester ultrasound; fetal anomalies; nuchal translucency; prenatal screening; early diagnosis; congenital malformations.

INTRODUCTION

First-trimester ultrasound has emerged as a vital tool in early fetal evaluation, extending its role beyond aneuploidy screening to the detection of major structural anomalies.[1]

Traditionally reserved for the second trimester, anomaly scans can now be performed between 11 to 13+6 weeks[2], thanks to advancements in high-resolution imaging and skilled sonography. Conditions such as acrania, anencephaly, cystic hygroma, and omphalocele can often be identified during this early window, offering crucial time for parental counseling and management decisions.[3]

While second-trimester scans remain the standard, studies by experts like Timor-Tritsch[4] and Salomon[5] have shown that early ultrasound, when performed with appropriate training and protocols, can provide comparable diagnostic accuracy. Factors such as fetal position, maternal obesity, and amniotic fluid volume may affect image quality, but the use of transvaginal and 3D modalities has helped overcome many of these challenges.[3,6]

This study aims to evaluate the diagnostic utility of first-trimester ultrasound in detecting major fetal anomalies, assessing its reliability and potential incorporation into routine prenatal care.

MATERIALS AND METHODS

This hospital-based cross-sectional observational study was conducted in the Department of Obstetrics and Gynaecology at Rohilkhand Medical College and Hospital, Bareilly, Uttar Pradesh, over a one-year period (November 2022 to October 2023).

A total of 250 pregnant women attending the OPD/IPD in the first trimester (gestational age 9+0 to 13+6 weeks; CRL 45–84 mm) were included after obtaining informed written consent. The sample size was calculated with assumptions of 3.7% prevalence, 5% precision, and a 95% confidence level (Z = 1.96), resulting in a final sample of 250.

Women with gestational age outside the defined limits, poor fetal visualization (due to obesity or surgical scars), or who declined consent were excluded. All participants underwent first-trimester ultrasound using high-resolution systems (EPIQ-7, IU22, Aloka SSD 3500, Philips HD series), employing transabdominal or transvaginal probes based on image quality. The scans aimed to confirm viability, determine fetal number, establish gestational age, and assess biometric measurements. For those undergoing first-trimester screening, NT was measured and maternal serum markers (PAPP-A, β-hCG) were evaluated using the FMF algorithm. NT scans were performed only by FMF-certified sonographers, while dating scans were conducted by both certified and experienced sonographers.

A general fetal anatomical survey was performed during all scans, though not standardized. Anomalies detected were classified according to the affected system—CNS, cardiac, gastrointestinal, genitourinary, musculoskeletal, and others.

Fetuses with multiple anomalies were categorized by the most severe defect. Cases with chromosomal abnormalities confirmed by invasive testing or non-pathological anatomical variants were excluded. No distinction was made between cystic hygroma and NT ≥3.5 mm due to overlapping ultrasound appearances.

Statistical analysis was performed using SPSS version 23.0. Quantitative data were presented as mean ± standard deviation and analyzed using paired t-tests. Categorical data were assessed using Chi-square or Fisher's exact test. A p-value of less than 0.05 was considered statistically significant.

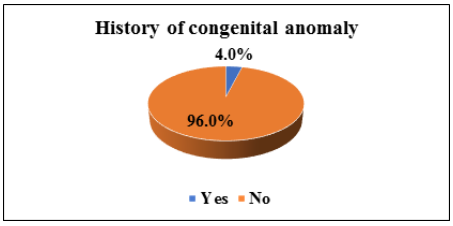
RESULTS

In our study, the majority of pregnant women were between 20–30 years (60.8%), with a higher proportion residing in rural areas (57.2%). Most were nulliparous (44%), and 16% had diabetes as a comorbidity. These baseline demographics provide insight into the population studied and its potential impact on anomaly detection — as shown in Table 1.

Table 1: Demographic and Clinical Characteristics of Pregnant Women Undergoing First-Trimester Ultrasound

	Variable	Frequency (n=250)	Percentage (%)
Maternal Age	**20–30**	152	60.8
	31-40	98	39.2
Residence	**Urban**	107	42.8
	Rural	143	57.2
Parity	**0**	110	44.0
	1	83	33.2
	2	35	14.0
	>2	22	8.8
Comorbidity	**Diabetes**	40	16.0
	None	210	84.0

In figure 1: The History of Congenital anomaly has been shown



Fígure 1: Hístory Of Congenítal Anomaly

Among the fetal anomalies detected in the first trimester, anencephaly was most prevalent (10%), followed by acrania (6.4%) and abnormal nuchal translucency (6.4%). A significant majority (72%) of pregnancies were anomaly-free in the first-trimester scan — as shown in Table 2.

Table 2: The Types Of Congenítal Anomalíes Díagnosed Ín 1st Trímester Dístríbutíon

Congenítal anomalíes	Frequency (n=250)	Percentage
Acranía	16	6.4
Anencephaly	25	10
Cystíc Hygroma	3	1.2
Abnormal N.T.	16	6.4
Hydrocephalus	6	2.4
Mícrocephaly	3	1.2
Spína bífída	1	0.4
No	180	72

A significant association was found between a history of congenital anomalies and the presence of certain anomalies in the current pregnancy. Notably, hydrocephalus (40%) and cystic hygroma (15%) were more common in women with a positive history, indicating a predictive value of past obstetric history in early anomaly detection — as shown in Table 3.

Table 3: The Types Of Congenítal Anomalíes Díagnosed Ín 1st Trímester And Hístory Of Anomaly

Congenítal anomalies	Hístory of anomaly		P value
	No (n=220)	Yes (n=30)	
Acranía	1 (0.6%)	2 (10.0%)	<0.001
Anencephaly	5 (2.8%)	0 (0.0%)	
Cystíc Hygroma	0 (0.0%)	3 (15.0%)	
Abnormal N.T.	11 (6.1%)	0 (0.0%)	
Hydrocephalus	5 (2.8%)	8 (40.0%)	
Mícrocephaly	4 (2.2%)	0 (0.0%)	
Spína bífída	1 (0.6%)	0 (0.0%)	
Normal	153 (85.0%)	7 (35.0%)	

Biometric abnormalities in crown-rump length (CRL) and mean gestational sac (MGS) diameter were significantly more frequent in fetuses with anomalies. Abnormal CRL was seen in 45% of anomaly cases (p=0.026), and abnormal MGS in 67.5% (p<0.001), suggesting these measurements are valuable early indicators of structural defects — as shown in Table 4.

Table 4: Mean Crl And Gs Measures Ín The Fírst Trímester To The Congenítal Anomalíes Present Ín The First Trimester

		Anomaly found (1st trímester)		P value
		No (n=210)	Yes (n=40)	
MCRL	Abnormal	43 (20.4%)	18 (45.0%)	0.026
	Normal	117 (55.7%)	22 (55.0%)	
MGS	Abnormal	37 (17.6 %)	27 (67.5%)	<0.001
	Normal	123 (58.5%)	13 (32.5%)	

DISCUSSION

First-trimester ultrasonography has evolved into a key tool for early detection of major fetal anomalies. Earlier, mid-gestation was the standard for anomaly screening, but with improved imaging and transvaginal approaches, detection is now possible between 11 to 13+6 weeks. This early identification allows for timely decision-making and better outcomes, especially in high-risk pregnancies.

In our study, 20% of cases had detectable anomalies in the first trimester, most commonly hydrocephalus, abnormal NT, and anencephaly. This aligns with **Fouad et al. (2018)** who reported a 9.4% anomaly detection rate, with hydrocephalus being most frequent.[2] **Iliescu et al. (2013)** found a 40.6% detection rate at 12–13+6 weeks, including 76.3% of major structural anomalies, especially CNS and cardiac defects.[7] **Colosi et al. (2015)** and **Liu et al. (2017)** also noted increasing maternal age and rural residence as associated factors, similar to our findings.[8,9]

Bardi et al. reported high first-trimester detection rates for acrania and encephalocele, while **Singh et al.** observed good early detection of lethal anomalies, though some conditions like renal anomalies were missed.[10,11] **Grande et al.** demonstrated unexpectedly high detection rates for skeletal (69%) and cardiac (57%) anomalies with advanced imaging.[12] **Edwards and Hui** showed that nearly half of all major anomalies can now be identified in the first trimester.[13]

Additionally, our study found a significant correlation between increased CRL and GS abnormalities and anomaly presence, consistent with **Fouad et al.**. **Baer et al.** reported that increased NT was strongly associated with major birth defects (RR: 1.6–2.1), reinforcing the predictive value of NT measurements.[2,14]

While early scanning offers clinical advantages, challenges remain—cost, training, and access to advanced imaging. As suggested by prior studies, early ultrasound is most effective in high-risk populations.

Strengths

The strengths of the study are the utilization of standardized ultrasound protocols and FMF-certified sonographers, ensuring reliable assessments. It focused specifically on early gestational age, offering valuable insight into the diagnostic potential of first-trimester scans for structural anomalies within a real-world, hospital-based Indian clinical setting.

Limitations

The limitations include a single-center design and the absence of follow-up scans to confirm missed anomalies. Operator dependency and image quality variations due to maternal factors may have influenced detection rates. The findings may not be generalizable to all healthcare settings or populations.

CONCLUSION

First-trimester ultrasound demonstrates promising diagnostic accuracy for early detection of major fetal anomalies. Early identification supports timely clinical decisions and better outcomes. Incorporating such scans into routine prenatal care can significantly enhance antenatal screening, especially in high-risk pregnancies.

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Consent: Written consent secured.

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