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EVALUATION OF CNS ANOMALIES IN FIRST TRIMESTER ULTRASOUND: A PROSPECTIVE STUDY

Radio-Diagnosis

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ABSTRACT

Background: First trimester ultrasound is a critical tool in prenatal care, providing early detection of fetal anomalies. Technological advancements, particularly high-frequency transvaginal ultrasound, have enhanced visualization of fetal structures, including the central nervous system (CNS), at 11-14 weeks gestation. This study aimed to evaluate CNS anomalies in pregnant women during this first trimester of pregnancy by ultrasound. **Methods:** This prospective observational study was conducted from March to September 2022 at GVP Institute of healthcare and medical technology in Vishakhapatnam, India. A total of 250 pregnant women between 11 and 14 weeks gestation undergoing routine ultrasound were enrolled. Inclusion criteria included singleton, viable intrauterine pregnancies with confirmed gestational age. Exclusion criteria were multiple pregnancies, unreliable last menstrual period data, and unwillingness to participate. Ultrasounds were performed using both trans-abdominal and trans-vaginal approaches to assess fetal CNS anatomy, including the cranium, brain, and spinal cord. Key measurements such as crown-rump length (CRL), nuchal translucency (NT), and intracranial translucency (IT) were recorded. Data were analyzed using descriptive statistics. **Results:** The mean maternal age was 24.59 ± 3.41 years, with a mean gestational age of 12.47 ± 0.81 weeks. CNS anomalies were detected in 3.6% of cases, including acrania (1.2%), anencephaly (1.2%), occipital encephalocele (0.8%), and sacral meningocele (0.4%). All affected pregnancies were medically terminated, with fetal autopsies confirming the ultrasound diagnoses. A family history of anomalies and consanguinity were present in 6% of cases, highlighting potential risk factors. **Conclusion:** This study demonstrates that first trimester ultrasonography enables early detection of CNS anomalies, allowing for timely clinical intervention. The relatively low incidence of CNS anomalies underscores the need for routine screening, especially in high-risk populations. Early diagnosis aids in informed decision-making, including consideration for medical termination when appropriate, and promotes improved maternal-fetal care.

KEYWORDS

Central Nervous System Abnormalities, First Trimester Pregnancy, Ultrasonography, Prenatal Diagnosis

INTRODUCTION

Ultrasound (USG) is indispensable in prenatal care, aiding in the early detection of fetal anomalies.1 Introduced in obstetrics by Prof. Ian Donald, USG has advanced significantly with high-frequency transvaginal scans enabling detailed imaging as early as the first trimester.2,3 Initially used for estimating gestational age via crown-rump length (CRL),4 first-trimester ultrasound now identifies chromosomal and structural anomalies. Traditionally performed in the second trimester, anomaly scans are increasingly valuable between 11 and 14 weeks gestation.5,6 Early screening provides reassurance, opportunities for non-invasive prenatal testing (NIPT), and potential early intervention.7 This study aimed to evaluate central nervous system (CNS) anomalies in pregnancies between 11 and 14 weeks gestation.

MATERIALS AND METHODOLOGY

This prospective observational study was conducted from March to September 2022 at GVP Institute of Healthcare and Medical Technology, Vishakhapatnam, India. A total of 250 pregnant women between 11 and 14 weeks gestation were enrolled. Inclusion criteria included singleton, viable intrauterine pregnancies with consistent last menstrual period (LMP) data. Women with multiple pregnancies or unreliable LMPs were excluded.

Participants' obstetric and family histories, including risk factors, were recorded. Ultrasounds were performed using a Samsung HS 70A machine with CA 1-7A and E3-12A transducers. Transabdominal and transvaginal scans were used for comprehensive imaging. Fetal parameters such as CRL, nuchal translucency (NT), and intracranial translucency (IT) were assessed using standard axial and mid-sagittal planes. The fetal spine was evaluated in sagittal, axial, and coronal views to detect anomalies like spina bifida. NT measurements adhered to Fetal Medicine Foundation guidelines for fetuses with CRL between 45 mm and 84 mm.

Statistical Methods

Data were analyzed using IBM SPSS 21.0. Descriptive statistics summarized quantitative variables, while categorical variables were represented by frequencies and proportions. Findings were also visualized using bar diagrams and pie charts.

RESULTS

Of 250 participants, the mean maternal age was 24.59 ± 3.41 years, with most (58.4%) aged between 21 and 25 years. Mean gestational age was 12.47 ± 0.81 weeks. Consanguineous marriages were reported by 6% of participants, while 3.2% had a history of anomalies in previous pregnancies. Family history of anomalies was present in 6%. Medical conditions such as diabetes mellitus (2.8%), hypertension (1.6%), and hypothyroidism (1.6%) were noted, with 2.4% reporting gestational diabetes mellitus (GDM). The demographic characteristics of the study population are summaried in Table 1.

The mean CRL was 5.94 ± 0.83 mm, and mean NT thickness was 1.24 ± 0.95 mm. CNS anomalies were identified in 3.6% of cases: acrania (1.2%), anencephaly (1.2%), occipital encephalocele (0.8%), and sacral meningocele (0.4%). All cases of anomalies underwent medical termination of pregnancy, confirmed by fetal autopsy. The fetal scare parameters and the anomalies detected are summarised in Table 2.

DISCUSSION

Congenital anomalies, defined by Hadzagic et al.8 as morphological abnormalities from the embryonic or fetal period, are increasingly detected early due to advancements in prenatal imaging.9,10 A systematic ultrasonographic approach aids in identifying CNS anomalies, predicting prognosis, and planning interventions.11-13

The mean maternal age in this study was 24.59 years, consistent with findings by Onkar et al.15 and Kunapareddy et al.,14 but slightly lower than in studies by Hassan et al.16 Variations likely reflect geographical differences in reproductive patterns. Gestational age averaged 12.47 weeks, aligning with Kunapareddy et al.14 Timing is crucial for detecting anomalies, as first-trimester scans are often the first detailed assessment for many pregnancies.

Primigravidae constituted 65.2% of participants, similar to Kunapareddy et al.14 but contrasting with the multigravidae majority in Hassan et al.16 These differences may reflect regional reproductive trends. Consanguinity, reported by 6% of participants, is a known risk

factor for congenital anomalies. Family history of anomalies (6%) was comparable to Hassan et al.'s 8%,16 emphasizing the importance of genetic counseling. Pre-existing medical conditions, such as diabetes (2.8%), hypertension (1.6%), and hypothyroidism (1.6%), further underscore the need for comprehensive prenatal care.

The mean CRL and NT thickness in this study were 5.94 mm and 1.24 mm, respectively.

NT measurement is pivotal for detecting chromosomal abnormalities and CNS anomalies, as highlighted by Kunapareddy et al.14 In this study, CNS anomalies were observed in 3.6% of cases, lower than Siddesh et al.'s 31.64%17 but higher than Onkar et al.'s 0.31%.15 Differences in population characteristics and diagnostic criteria likely explain these variations.

Specific anomalies included acrania and anencephaly (1.2% each), occipital encephalocele (0.8%), and sacral meningocele (0.4%). Comparable findings were reported by Onkar et al.15 and Grande et al.18 Early detection enabled timely medical termination, confirmed by fetal autopsies, reinforcing the critical role of first-trimester ultrasonography.

CONCLUSION

This study highlights the effectiveness of first-trimester ultrasonography in detecting CNS anomalies, facilitating timely interventions. With a 3.6% incidence of CNS anomalies, routine early screening is essential, especially in high-risk groups.

Advances in ultrasonography continue to enhance maternal and fetal care, enabling informed decision-making and improved outcomes.

Table 1: Demographic and Clinical Characteristics of the Study Population (n=250)

Parameter	Categories	n (%)
Age (years)	≤ 20	16 (6.4%)
	21 – 25	146 (58.4%)
	26 – 30	78 (31.2%)
	31 – 35	10 (4.0%)
Mean Age ± SD		24.59 ± 3.41
Gestational Age (weeks)	Mean ± SD	12.47 ± 0.81
Parity	Primigravida	163 (65.2%)
	Multigravida	87 (34.8%)
Consanguinity	Yes	15 (6.0%)
	No	235 (94.0%)
Previous Anomalies	Yes	8 (3.2%)
	No	242 (96.8%)
Family History of Anomalies	Yes	15 (6.0%)
	No	235 (94.0%)
History of Drug Intake	Yes	6 (2.4%)
	No	244 (97.6%)
Previous Medical Disorders	Diabetes Mellitus	7 (2.8%)
	Hypertension	4 (1.6%)
	Hypothyroidism	4 (1.6%)
	Cardiac Problems	2 (0.8%)
Pregnancy-related Illness	Gestational Diabetes Mellitus (GDM)	6 (2.4%)

Table 2: Fetal Scan Parameters and CNS Anomalies Detected (n=250)

Parameter	Mean ± SD	Median	Range
Crown-Rump Length (CRL) (mm)	5.94 ± 0.83	5.9	4.1 – 7.6
Nuchal Translucency (NT) (mm)	1.24 ± 0.95	1.1	0.2 – 11.2
Fetal Anomalies Detected	Yes	9 (3.6%)	
	No	241 (96.4%)	
Types of CNS Anomalies	Acrania	3 (1.2%)	
	Anencephaly	3 (1.2%)	
	Occipital Encephalocele	2 (0.8%)	
	Sacral Meningocele	1 (0.4%)	

Figure 3: Sacral meningocele at 12 weeks 3 days gestational age

Figure 4: Occipital encephalocele at 12 weeks 6 days gestational age

Figure 5: Acrania at 12 weeks 3 days gestational age

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