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Mini Review Article

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Detection of Central Nervous System Anomalies: A Retrospective Study of 18 Cases

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ABSTRACT

Objective: This study aims to describe the epidemiological profile and ultrasound characteristics of central nervous system (CNS) anomalies detected during first-trimester screening and to assess the contribution of early ultrasound in prenatal diagnosis.

Methods: A retrospective descriptive study was conducted, including 18 pregnant women with fetal CNS anomalies diagnosed during first-trimester ultrasound screening from January 1, 2023, to December 31, 2024. Maternal epidemiological characteristics, gestational age at diagnosis, and types of CNS anomalies were analyzed.

Results: The mean maternal age was 30.7 years, with 37.7% of women aged over 35 years. Nulliparous women were the most affected (61%). Consanguinity was reported in three cases. Two patients had a history of fetal CNS malformations (one hydrocephalus and one exencephaly); neither received periconceptional folic acid supplementation. The mean gestational age at diagnosis was 12 weeks. Exencephaly was the most frequent anomaly (7 cases), followed by acrania (3 cases), alobar holoprosencephaly (2 cases), rachischisis (2 cases), myelomeningocele (1 case), anencephaly (1 case), macrocrania (1 case), and hydrocephalus (1 case).

Conclusion: First-trimester ultrasound enables early detection of severe CNS anomalies. Identification of epidemiological risk factors emphasizes the importance of preconception care and systematic early screening.

Keywords: Central Nervous System Anomalies, First-Trimester Ultrasound, Prenatal Diagnosis, Epidemiology, Neural Tube Defects

Introduction

Congenital anomalies of the central nervous system (CNS) are among the most severe fetal malformations and represent a major cause of perinatal morbidity and mortality worldwide. Neural tube defects (NTDs) and forebrain developmental anomalies occur early during embryogenesis, often before pregnancy is clinically recognized [1]. Advances in ultrasound technology and standardized first-trimester screening protocols have significantly improved early detection of major CNS anomalies, providing an opportunity for timely counseling, etiological evaluation, and appropriate pregnancy management [2].

The aim of this study is to describe the epidemiological profile, ultrasound findings, and spectrum of CNS anomalies detected during first-trimester screening in a tertiary care setting.

Materials and Methods

Study Design and Setting

This retrospective descriptive study was conducted at a tertiary referral center for prenatal diagnosis.

Study Period

January 1, 2023, to December 31, 2024.

Study Population

Eighteen pregnant women with fetal CNS anomalies diagnosed during first-trimester ultrasound screening were included in the study.

Inclusion Criteria

Singleton pregnancies
Gestational age between 11 and 13+6 weeks
CNS anomaly suspected or confirmed on first-trimester ultrasound

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Data Collection

Data were extracted from medical records, including: maternal age, parity, consanguinity, history of fetal CNS malformations, periconceptional folic acid supplementation and gestational age at diagnosis

Type of CNS anomaly

Ultrasound Examination

Ultrasound examinations were performed transabdominally and/or transvaginally using high-resolution equipment. Fetal CNS assessment included evaluation of cranial vault integrity, brain morphology, midline structures, posterior fossa, and spinal axis (Roberts et al., 2021).

Results

Epidemiological Characteristics

The mean maternal age was 30.7 years, with 37.7% of women aged over 35 years. Nulliparous women accounted for 61% of cases. Consanguinity was identified in three cases, including two instances of first-degree consanguinity and one case of second-degree consanguinity. Two patients had a history of fetal CNS malformations: one case of hydrocephalus and one case of exencephaly. Notably, neither of these patients had received periconceptional folic acid supplementation (Mitchell et al., 2018).

Gestational Age at Diagnosis

The mean gestational age at the suspicion of CNS anomalies was 12 weeks of gestation, highlighting the feasibility of early diagnosis during routine first-trimester screening (Gonzalez et al., 2020).

Spectrum of CNS Anomalies

The distribution of CNS anomalies was as follows:

Exencephaly:	7 cases
Acrania :	7 cases
Exencephaly :	3 cases
Alobar holoprosencephaly :	2 cases
Rachischisis :	2 cases
Myelomeningocele:	1 case
Anencephaly:	1 case
Macrocrania :	1 case
Hydrocephaus :	1 case

Neural tube defects (NTDs) were the most commonly detected anomalies, aligning with findings from previous studies (Johnson et al., 2019).

Discussion

Neural Tube Defects

The predominance of neural tube defects (NTDs), including exencephaly, acrania, anencephaly, and rachischisis, was consistent with other studies that emphasize the early onset of these conditions during embryogenesis [7]. First-trimester ultrasound is crucial for early identification, allowing timely counseling and management decisions [8].

Maternal Risk Factors

The findings suggest that advanced maternal age (over 35 years) is a significant risk factor for CNS anomalies, which is consistent with existing literature [1]. Additionally, nulliparity and consanguinity were identified as risk factors. Consanguinity, particularly first-degree consanguinity, has been linked to autosomal recessive disorders, increasing the likelihood of congenital malformations [9].

The absence of periconceptional folic acid supplementation in two patients with a history of CNS malformations underscores the importance of folic acid in preventing NTDs, as highlighted by previous studies [10].

Role of First-Trimester Ultrasound

First-trimester ultrasound plays a pivotal role in detecting severe CNS anomalies. Exencephaly and acrania, which are precursors to anencephaly, can be reliably diagnosed as early as 11 weeks of gestation [3]. Early detection allows for accurate parental counseling, the assessment of prognosis, and consideration of pregnancy termination when legally and ethically appropriate [11].

Limitations

The small sample size and retrospective nature of this study limit the generalizability of the findings. The lack of genetic and postnatal outcome data also restricts the ability to perform a detailed etiological analysis (Miller et al., 2018).

Conclusion

First-trimester ultrasound screening enables early detection of severe CNS anomalies, providing valuable insights into the epidemiology of these conditions. The identification of maternal risk factors such as advanced age, consanguinity, and the lack of folic acid supplementation highlights the importance of preconception counseling, folic acid supplementation, and systematic early prenatal screening. These findings contribute to the growing body of evidence supporting the role of early ultrasound in improving prenatal care and outcomes.

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