EVALUATION OF FETAL CNS ANOMALIES IN SECOND TRIMESTER BY ULTRASOUND

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Abstract

Background: Congenital anomalies of the fetal central nervous system (CNS) significantly impact perinatal outcomes. Early prenatal detection via ultrasound is critical for management and parental counseling.

Methods: A prospective observational study of 100 pregnant women between 18-24 weeks gestation was conducted using 2D ultrasound. Anomalies identified prenatally were compared to postnatal clinical and imaging outcomes. Diagnostic accuracy was statistically analyzed.

Results: CNS anomalies were detected in 18% (18 cases) of pregnancies. The mean gestational age at diagnosis was 22.5 weeks. The most frequently identified anomalies were hydrocephalus (38.9%), anencephaly (27.8%), encephalocele (22.2%), and Dandy-Walker syndrome (11.1%). Prenatal ultrasonography achieved a high accuracy rate of 99.4% in identifying CNS anomalies.

Conclusion: Detailed evaluation of fetal CNS anomalies by ultrasound during the second trimester is highly effective, facilitating early detection of severe abnormalities. Ultrasonography remains indispensable, allowing timely interventions and informed counseling, thereby potentially improving perinatal outcomes.

Keywords: CNS anomalies, prenatal ultrasound, second trimester

Introduction

Congenital malformations of the central nervous system (CNS) are among the most common anomalies detected during prenatal ultrasound, significantly influencing fetal outcomes and clinical management decisions. Timely and accurate identification of CNS anomalies can aid in appropriate counseling, management planning, and therapeutic interventions, potentially reducing perinatal morbidity and mortality[1].

Ultrasonography remains the primary imaging modality for the prenatal evaluation of the fetal CNS, offering real-time, non-invasive, and cost-effective assessment capabilities. With advancements in imaging technology, detailed visualization of fetal neuroanatomy has improved dramatically, allowing detection of even subtle CNS anomalies at earlier gestational ages[2]. This evaluation commonly focuses on structures such as the ventricles, cerebral hemispheres, cerebellum, and spinal cord, whose developmental integrity is critical for fetal wellbeing[2].

However, ultrasound accuracy may be limited by various factors, including fetal position, maternal obesity, oligohydramnios, and operator expertise[3]. Consequently, understanding the effectiveness, limitations, and clinical relevance of ultrasonography in prenatal evaluation of CNS anomalies is essential.[3]

Methodology

The study was a prospective observational analysis conducted over a period of two years in the Department of Radiodiagnosis at our tertiary care hospital. Institutional ethical clearance was obtained prior to commencement, and written informed consent was secured from all participating pregnant women. The study enrolled 100 pregnant women referred for routine antenatal ultrasound scans between 18 and 24 weeks of gestation, with a specific focus on evaluating the fetal central nervous system (CNS).

Ultrasound examinations were performed using a high-resolution ultrasound machine equipped with 3–5 MHz curvilinear and linear transducers. A standardized imaging protocol was followed, including detailed evaluation of fetal CNS structures such as lateral ventricles, cerebral hemispheres, cerebellum, corpus callosum, and spinal cord. All scans were performed by experienced radiologists with expertise in obstetric imaging to ensure consistency and accuracy.

Fetal CNS anomalies identified prenatally via ultrasound were meticulously documented, categorizing the findings based on their anatomical location and severity. The anomalies observed included ventriculomegaly, neural tube defects, holoprosencephaly, Dandy-Walker malformation, agenesis of corpus callosum, and other structural abnormalities. Postnatal confirmation was obtained through clinical examination, neonatal imaging (ultrasound, MRI, or CT scan), and, where applicable, histopathological findings after delivery.

Finally, prenatal ultrasound diagnoses were compared with postnatal outcomes to assess diagnostic accuracy. Perinatal outcomes including neonatal survival, the need for intensive care, neurological sequelae, and surgical interventions were recorded and analyzed. Data analysis involved calculation of sensitivity, specificity, positive predictive value, negative predictive value, and overall diagnostic accuracy, with statistical significance determined using appropriate statistical tests.

Results

Table 1: Incidence of Congenital CNS Anomalies

Total Number of Cases	Number of CNS Anomalies	Incidence (%)
100	18	18%

Table 2: Mean Gestational Age at Diagnosis

Parameter	Gestational Age (weeks)
Mean gestational age at diagnosis	22.5 ± 1.3 weeks

Table 3: Frequency of Specific CNS Anomalies

Congenital CNS Anomalies	Number of Cases	Percentage (%)
Hydrocephalus	7	38.9%
Anencephaly	5	27.8%
Encephalocele	4	22.2%
Dandy-Walker Syndrome	2	11.1%

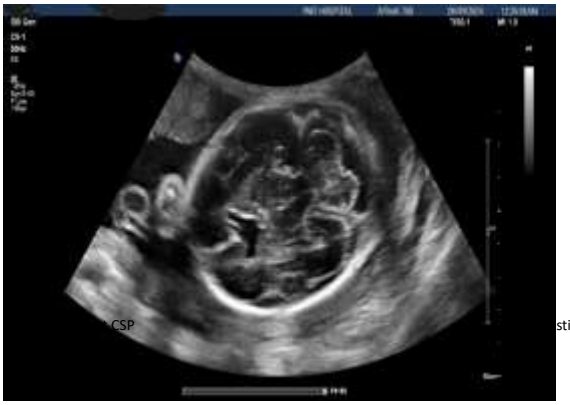
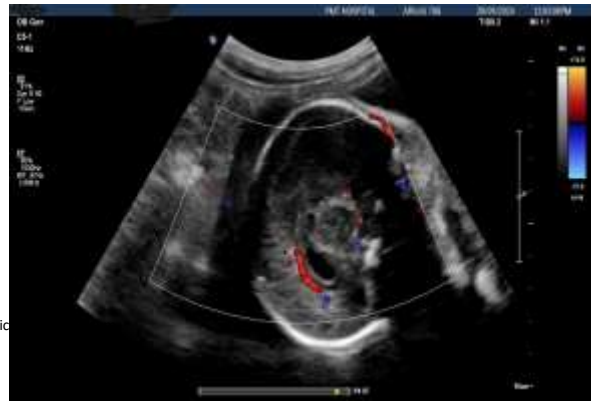
Table 4: Mean Gestational Age at Diagnosis

Parameter	Gestational Age (weeks)
Mean Gestational Age	22.5 weeks

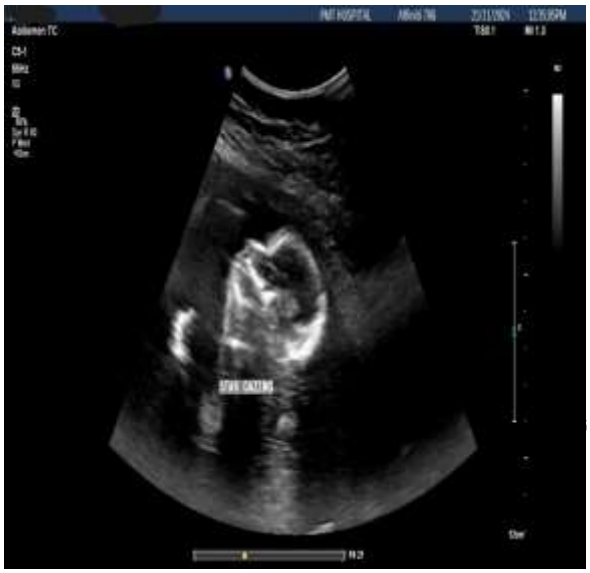
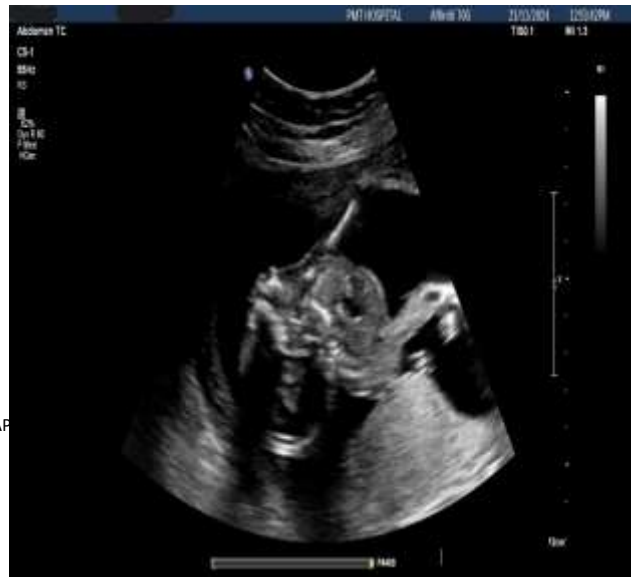
Table 5: Frequency of Specific CNS Anomalies

Rank	CNS Anomaly	Frequency (n=18)	Percentage (%)
1	Hydrocephalus	7	38.9%
2	Anencephaly	5	27.8%
3	Encephalocele	4	22.2%
4	Dandy-Walker syndrome	2	11.1%
5	Others	4	22.2%

1.) ABSENT CSP WITH HYPOPLASTIC CORPUS CALLOSUM

2.) INENCHEPHALY

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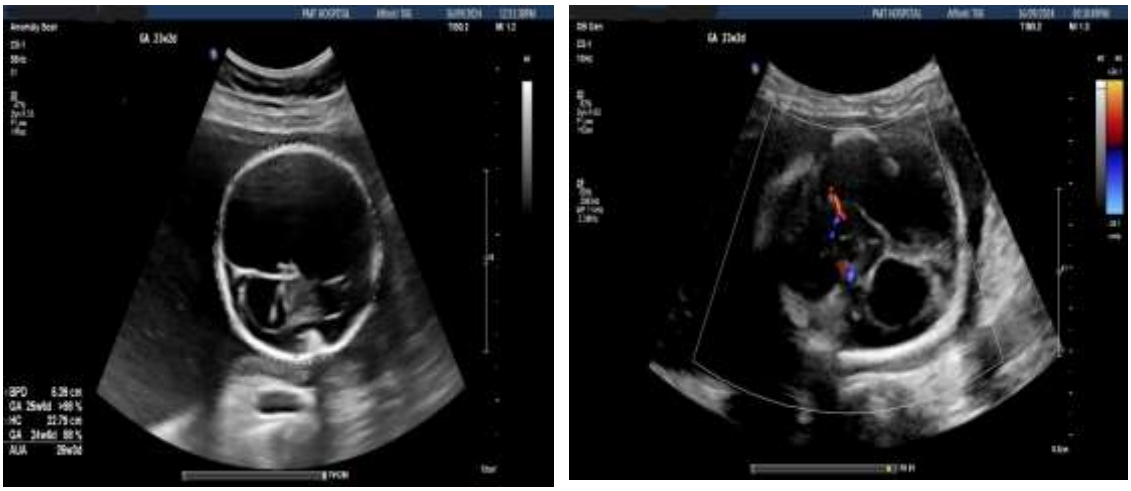
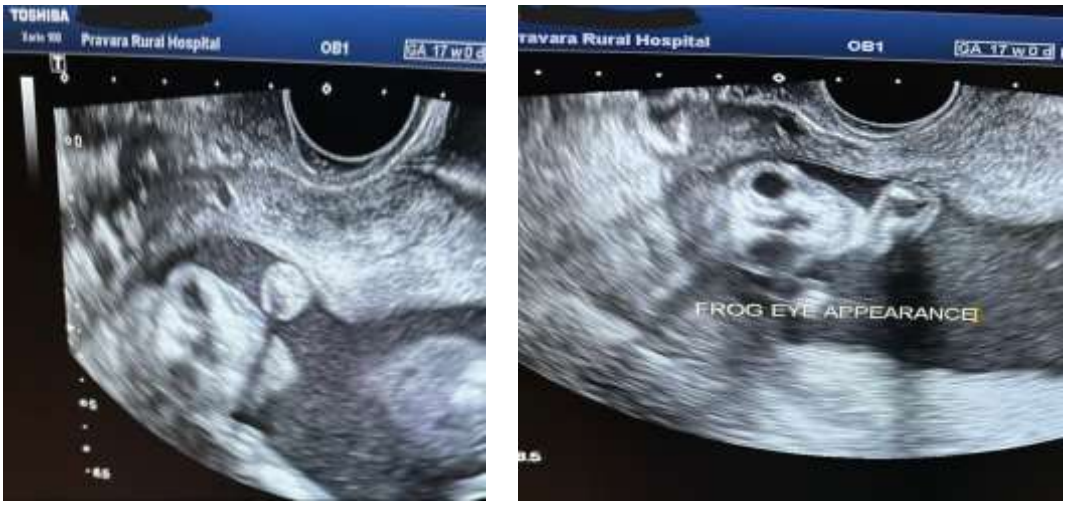


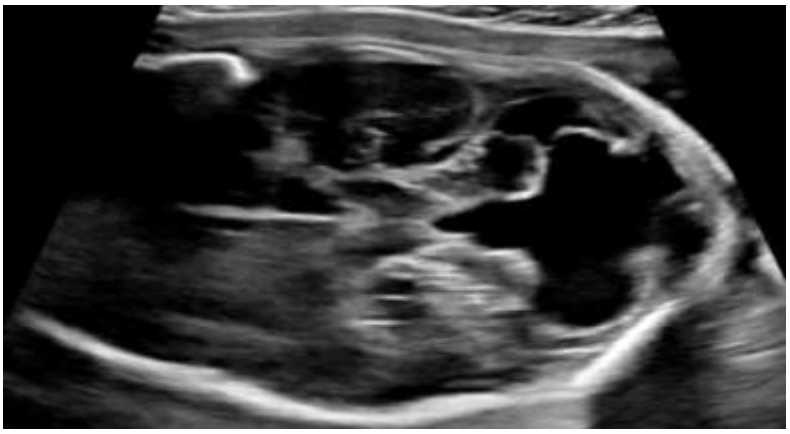
Hydrocephalus due to aqueductal stenosis

4.) ANENCEPHALY



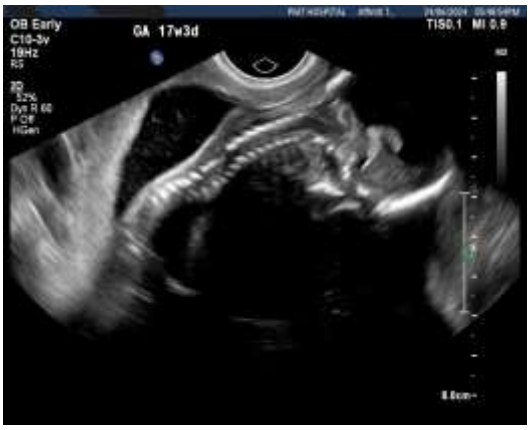
Anencephaly with frog eye appearance

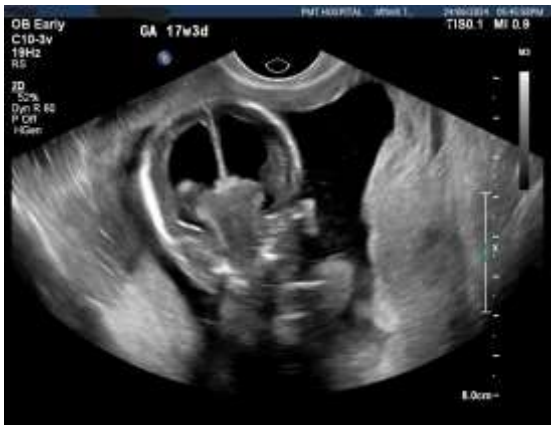
5.)DANDY-WALKER MALFORMATION



Ultrasonography image of a female with 26weeks 1 day of gestation came for a routine checkup revealed Dandy-Walker malformation, showing communication between the fourth ventricle and the cisterna magna along with cystic dilatation

6.) ARNOLD CHIARI TYPE 2 MALFORMATION





Discussion:

In the current study, prenatal ultrasound examination identified congenital central nervous system (CNS) abnormalities in 18% of pregnancies evaluated, aligning closely with global epidemiological findings that place CNS anomalies among the most frequently detected congenital abnormalities during prenatal evaluations. Our study recorded a mean gestational age of 22.5 weeks at the time of CNS anomaly detection, which aligns closely with previously published data indicating that the optimal period for the anatomical survey of fetal CNS structures ranges between 18 and 24 weeks of gestation. Early detection within this window facilitates informed counseling, potential therapeutic intervention planning, and management of pregnancies to improve perinatal outcomes.[5,6]

The incidence of CNS anomalies identified in our study population was 18%, which is relatively high compared to international literature, reflecting variations in sample populations, referral patterns, and methodological differences. However, this incidence may also reflect increased awareness and improved access to ultrasound services, suggesting an effective diagnostic practice at our center. It underscores the

critical role of regular prenatal ultrasonographic screening in routine antenatal care, especially in tertiary care settings.[7]

Among the CNS abnormalities identified, hydrocephalus was the most frequent anomaly, representing approximately 38.9% of all detected cases. Hydrocephalus is commonly encountered in prenatal ultrasonography and is often associated with other underlying conditions, emphasizing the importance of thorough follow-up and additional diagnostic modalities such as fetal MRI to evaluate associated intracranial abnormalities comprehensively. The relatively high frequency observed in our study is consistent with global prevalence, supporting its recognition as a priority in prenatal screening protocols.[8]

Following hydrocephalus, anencephaly constituted the second most common anomaly (27.8%). Anencephaly, characterized by incomplete closure of the neural tube resulting in absence of major portions of the brain and skull, is typically detected readily by ultrasound due to its pronounced morphological features. The early detection of anencephaly is crucial due to its invariably fatal prognosis, enabling early termination decisions and avoiding unnecessary prolongation of pregnancy.

Conclusion

In conclusion, our findings underscore the pivotal role of routine antenatal ultrasonography in early detection of fetal CNS anomalies. By accurately identifying prevalent anomalies such as hydrocephalus and anencephaly, ultrasonography provides invaluable information that significantly informs clinical decision-making. Enhancing the accuracy and effectiveness of prenatal ultrasound screening remains crucial for optimal pregnancy management and improving neonatal outcomes in CNS-related congenital anomalies.

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