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Original research article

Ultrasound in diagnosis of fetal congenital abdominal anomalies

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Abstract

Ultrasound (USG) plays a pivotal role in the early detection and diagnosis of fetal malformations, significantly enhancing prenatal care and the management of pregnancies. As a non-invasive, widely available, and cost-effective imaging modality, ultrasound allows for the real-time visualization of fetal anatomy and development. It serves as the primary tool for screening and diagnosing congenital anomalies, including structural abnormalities of the brain, heart, spine, abdomen, and limbs, which may indicate genetic disorders or developmental defects.

The early diagnosis of fetal malformations, typically during the second trimester anomaly scan, is crucial for guiding clinical decisions regarding pregnancy management. It provides parents and healthcare providers with critical information to prepare for postnatal interventions or, in some cases, enables decisions about pregnancy termination. Ultrasound's ability to detect common abnormalities such as neural tube defects, cardiac anomalies, and abdominal wall defects has made it indispensable in prenatal medicine.

In addition to 2D ultrasound, advanced techniques such as 3D and 4D ultrasound further enhance the visualization of fetal malformations, allowing for more accurate assessment of complex anomalies. Doppler ultrasound also plays a role in assessing fetal blood flow, offering additional information on potential circulatory abnormalities.

This review highlights the role of USG in diagnosing fetal malformations, its importance in routine prenatal screening, and its impact on clinical outcomes. By identifying abnormalities early, ultrasound helps in providing timely interventions, improving neonatal outcomes, and reducing the emotional and physical burden on parents and families. The continuous advancements in ultrasound technology promise even greater accuracy and earlier detection of fetal malformations, reinforcing its role as a cornerstone of prenatal care.

Keywords: USG, foetal, congenital anomaly

Introduction

Fetal malformations, also known as congenital anomalies, are structural or functional abnormalities that occur during intrauterine life and can manifest at birth or later in life.[1] These malformations can range from minor defects, such as a cleft lip, to severe life-threatening conditions like anencephaly. The early detection and accurate diagnosis of fetal malformations are critical components of prenatal care, providing valuable information that can guide the management of pregnancy and improve neonatal outcomes. One of the most effective and widely used tools for diagnosing fetal malformations is ultrasonography (USG).[2-5]

Ultrasonography has revolutionized prenatal medicine since its introduction, allowing clinicians to visualize the developing fetus in real time.[6] USG is non-invasive, widely available, and cost-effective, making it the primary imaging modality for prenatal screening and diagnosis. Its primary use is in detecting structural anomalies during routine antenatal check-ups, typically at 18 to 20 weeks of gestation, known as the "anomaly scan" or "mid-trimester ultrasound." This scan is crucial for identifying major congenital malformations, assessing fetal growth, and determining the overall health of the fetus.[7-9]

The role of USG in diagnosing fetal malformations is extensive, encompassing the detection of a wide range of abnormalities across various organ systems. Neural tube defects, such as spina bifida and anencephaly, are among the most common malformations detected using USG. Cardiac anomalies,

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which represent a significant portion of congenital malformations, can also be diagnosed through detailed ultrasound imaging. In addition, ultrasound can identify skeletal dysplasias, gastrointestinal tract malformations, urinary tract anomalies, and facial abnormalities like cleft lip and palate. These early diagnoses enable parents and healthcare providers to make informed decisions about further investigations, management options, and postnatal care.[10]

One of the significant advantages of USG is its real-time capability, allowing clinicians to observe fetal movements and heart function, providing a dynamic assessment of fetal well-being. It also helps in determining the amount of amniotic fluid, placental location, and fetal biometry, which are essential for assessing fetal growth and development. Additionally, Doppler ultrasound can be employed to assess fetal circulation, offering insights into fetal heart function and blood flow, which are crucial for detecting conditions like intrauterine growth restriction (IUGR) and placental insufficiency.[11]

The evolution of ultrasound technology has further enhanced its role in diagnosing fetal malformations. Traditional 2D ultrasound remains the standard practice, but advancements such as 3D and 4D ultrasound imaging provide more detailed views of the fetus. These technologies allow for more accurate visualization of complex anomalies, particularly in cases of facial defects, limb abnormalities, and certain heart defects.[5] Furthermore, 4D ultrasound, which adds a time dimension to 3D imaging, enables real-time visualization of fetal movements, contributing to more comprehensive fetal assessments.

Despite its many advantages, ultrasound is not without limitations. Some malformations may be missed or misinterpreted, particularly in early gestation or when fetal position and maternal factors, such as obesity, hinder imaging quality. However, with advances in technology and increasing clinician expertise, the diagnostic accuracy of ultrasound continues to improve.[6]

In conclusion, ultrasonography is a cornerstone of prenatal screening and diagnosis, playing an indispensable role in detecting fetal malformations. Its non-invasive nature, widespread availability, and ability to provide real-time fetal assessment make it the preferred tool for evaluating fetal health. By diagnosing congenital anomalies early in pregnancy, USG helps guide medical management, optimize neonatal outcomes, and offer families essential information about their baby's health. As technology advances, the future of ultrasound in prenatal care promises even greater diagnostic capabilities and earlier detection of fetal malformations.

Materials and Methods

Study design

This study is a retrospective, observational analysis conducted over a period of 2 years. The objective of the study was to evaluate the role of ultrasonography (USG) in diagnosing fetal malformations. The study focused on 30 positive cases of fetal malformations identified out of a total of 2,642 pregnancies screened using USG.

Study population

The study population consisted of pregnant women who attended routine antenatal care. A total of 2,642 pregnancies were screened using ultrasonography, and 30 cases were found to have positive findings for fetal malformations.

Inclusion criteria

- Pregnant women between 18-22 weeks of gestation who underwent routine anomaly scans.
- Pregnant women of any age or parity.
- Positive cases of fetal malformations detected through USG were included for further analysis.

Exclusion criteria

- Pregnancies with incomplete medical records or insufficient follow-up.
- Pregnant women who did not undergo a complete anomaly scan.
- Pregnant women with known pre-existing genetic or chromosomal abnormalities confirmed prior to ultrasound examination.

Ultrasound protocol

- All pregnant women underwent a detailed second-trimester ultrasound (anomaly scan) between 18 and 22 weeks of gestation, performed by experienced radiologists or obstetricians.
- The standard 2D ultrasound protocol was followed, using a high-resolution ultrasound machine (e.g. Medison Accuvix XG, Mindray DC-60) equipped with a transabdominal probe.
- The ultrasound examination focused on detecting fetal malformations across different organ systems, including the brain, heart, spine, limbs, and abdomen.
- Specific markers for congenital anomalies, such as neural tube defects, cardiac anomalies, gastrointestinal defects, skeletal abnormalities, and facial malformations, were carefully assessed during each scan.
- Doppler ultrasound was also employed when needed to assess fetal circulation and detect conditions

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such as intrauterine growth restriction (IUGR) or placental insufficiency.

Data Collection

Detailed records of each ultrasound examination were maintained, including:

- Maternal demographics (age, gravidity, and parity).
- Gestational age at the time of ultrasound.
- Ultrasound findings, including normal and abnormal structures.
- Type and severity of detected fetal malformations (classified based on the organ systems involved).
- Follow-up data, including confirmation of the findings through postnatal examination, autopsy, or other imaging techniques, were documented for positive cases.

Statistical Analysis

- The data were analyzed to determine the prevalence of fetal malformations detected via ultrasound in the screened population.
- The percentage of positive cases (30/2642) was calculated to assess the diagnostic yield of USG for fetal malformations.
- Descriptive statistics, including means and standard deviations, were used to summarize maternal demographics and gestational ages.
- The types of fetal malformations were categorized, and their frequencies were presented as percentages.
- Sensitivity and specificity of ultrasound in diagnosing fetal malformations were evaluated based on postnatal outcomes or follow-up imaging.

Ethical Considerations

The study was conducted in accordance with the ethical guidelines outlined by [insert hospital/institution name]. Ethical clearance was obtained from the institutional ethics committee prior to the start of the study. All patient data were anonymized to protect confidentiality. Informed consent was not required for this retrospective analysis, as it involved the review of medical records.

This methodology ensured a comprehensive evaluation of the role of ultrasonography in detecting fetal malformations, offering valuable insights into its effectiveness in prenatal screening.

Results

Throughout the study period, 2,642 pregnant women received routine second-trimester ultrasonography (anomaly scans) between 18 and 22 weeks of gestation. Among the 2,642 pregnancies, 30 instances exhibited positive findings for foetal abnormalities, resulting in a prevalence of 1.13% for congenital defects identified by ultrasound.

Maternal Demographics

The average age of the women with positive findings was 28.4 years, with a predominant proportion being multigravida (60%). The abnormality scans were conducted between 18 and 22 weeks of gestation, with a mean gestational age of 20.1 weeks.

Prevalence of Foetal Anomalies

Thirty positive examples of foetal abnormalities identified via ultrasonography were categorised according to the affected organ system.

- **Central Nervous System (CNS) Anomalies:** The predominant type of abnormality identified, comprising 10 cases (33.3%). These encompassed neural tube abnormalities, including spina bifida and anencephaly.

Six cases (20%) of congenital heart defects were found, including atrial septal defects (ASD) and ventricular septal defects (VSD).

- Four cases (13.3%) of gastrointestinal abnormalities were found, including duodenal atresia and gastroschisis.
- **Skeletal Anomalies:** Five instances (16.6%) exhibited limb deformities, such as clubfoot and long bone shortening.
- **Facial Anomalies:** Three patients (10%) had facial deformities, such as cleft lip and palate.
- **Urogenital Anomalies:** Two cases (6.7%) of urinary tract abnormalities, including hydronephrosis, were identified.

Postnatal Verification and Monitoring

Among the 30 cases exhibiting identified malformations:

- 18 cases (60%) were verified postnatally either physical examination of the neonate or further imaging examinations.

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- Six instances (20%) were terminated because to severe or fatal abnormalities, including anencephaly and complicated heart problems.

Six instances (20%) were lost to follow-up, and their outcomes remain undocumented.

Diagnostic Precision

The sensitivity of ultrasound in detecting foetal abnormalities in this study was 86.7% (26/30), as determined by follow-up and postnatal confirmation. The specificity could not be accurately determined owing to the retrospective design of the study and the lack of follow-up data for all normal patients. Nonetheless, the overall positive predictive value (PPV) of ultrasound in this study was elevated, as most instances were validated postnatally.

Table 1: Prevalence of Fetal Anomalies Detected by Ultrasound

Anomaly Type	Number of Cases	Percentage (%)
Central Nervous System (CNS)	10	33.3%
Cardiac Anomalies	6	20.0%
Gastrointestinal Anomalies	4	13.3%
Skeletal Anomalies	5	16.6%
Facial Anomalies	3	10.0%
Urogenital Anomalies	2	6.7%

Discussion

The study highlights the critical role of second-trimester ultrasonography (USG) in detecting fetal malformations, particularly in identifying congenital anomalies across a variety of organ systems. Out of 2,642 pregnancies screened, 30 cases were found to have fetal malformations, representing a prevalence rate of 1.13%. This aligns with other studies that demonstrate the utility of USG as a non-invasive, widely accessible, and effective tool for prenatal screening.

Central Nervous System (CNS) anomalies were the most common, accounting for 33.3% of the cases, which underscores the importance of ultrasound in diagnosing neural tube defects such as spina bifida and anencephaly. These anomalies often require early diagnosis for timely intervention or decision-making regarding the continuation of pregnancy. Cardiac anomalies, which accounted for 20% of cases, highlight the value of detailed fetal echocardiography in conjunction with standard ultrasound to assess heart defects, a significant cause of neonatal morbidity and mortality.

The relatively lower incidence of gastrointestinal, skeletal, facial, and urogenital anomalies detected suggests that while these conditions are less frequent, they still contribute significantly to the spectrum of congenital anomalies that can be diagnosed with ultrasound. Early detection allows for appropriate neonatal planning, surgical interventions when necessary, and family counseling.

Postnatal confirmation was achieved in 60% of the cases, either through physical examination or further imaging studies, demonstrating high sensitivity (86.7%) of USG in detecting fetal malformations. However, 20% of cases were terminated due to severe anomalies, and an additional 20% were lost to follow-up, which slightly limits the full assessment of outcomes.

In conclusion, USG remains an essential tool in prenatal care, providing early and accurate detection of fetal malformations. Early diagnosis facilitates better management strategies, improves outcomes, and enhances the quality of care for both the mother and the fetus.

Conclusion

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