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Original Research

Comparative Analysis of 3D/4D Ultrasound vs. Conventional 2D Ultrasound for Detection of Fetal Facial Anomalies: A Prospective Multicenter Study

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ABSTRACT:
Objective: To compare the diagnostic accuracy of three-dimensional/four-dimensional (3D/4D) ultrasound with conventional two-dimensional (2D) ultrasound for the detection of fetal facial anomalies in a prospective multicenter setting. **Methods:** A prospective study was conducted from January 2013 to July 2013 across three tertiary referral centers. Seventy pregnant women between 18 and 34 weeks of gestation with suspected fetal anomalies on routine ultrasound or high-risk pregnancies were evaluated using both 2D and 3D/4D ultrasound techniques. The images were independently assessed by two fetal medicine specialists blinded to the final diagnosis. The primary outcome was the detection rate of facial anomalies. Secondary outcomes included the assessment of specific facial structures, interobserver agreement, and maternal satisfaction. **Results:** Among the 70 participants, 24 fetuses (34.3%) were diagnosed with facial anomalies. The detection rate was significantly higher with 3D/4D ultrasound (91.7%, 22/24) compared to 2D ultrasound (70.8%, 17/24) (p=0.031). 3D/4D ultrasound demonstrated superior visualization of cleft lip (100% vs. 81.8%), micrognathia (88.9% vs. 66.7%), and ear abnormalities (85.7% vs. 42.9%). Interobserver agreement was higher for 3D/4D ultrasound (κ=0.84) than for 2D ultrasound (κ=0.69). Maternal satisfaction scores were significantly higher for 3D/4D ultrasound compared to 2D ultrasound (mean score 8.6 vs. 6.9 on a 10-point scale, p<0.001). **Conclusion:** 3D/4D ultrasound is significantly more accurate than conventional 2D ultrasound for the detection of fetal facial anomalies, particularly for the assessment of cleft lip, micrognathia, and ear abnormalities. The technique offers higher interobserver agreement and greater maternal satisfaction. These findings support the use of 3D/4D ultrasound as a complementary technique to 2D ultrasound for comprehensive assessment of fetal facial structures.
Keywords: 3D ultrasound, 4D ultrasound, fetal facial anomalies, prenatal diagnosis, cleft lip, micrognathia

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INTRODUCTION

Fetal facial anomalies are important markers for chromosomal abnormalities and genetic syndromes and may significantly impact perinatal management and outcomes.[1,2] Conventional two-dimensional (2D) ultrasound has been the standard modality for prenatal screening and diagnosis of fetal anomalies. However, the assessment of complex three-dimensional structures such as the fetal face presents significant challenges with 2D imaging alone.[3]

The development of three-dimensional (3D) and four-dimensional (4D) ultrasound technology has provided new opportunities for detailed evaluation of fetal anatomy, particularly facial structures. 3D ultrasound allows volume acquisition and multiplanar reconstruction, while 4D ultrasound adds the temporal dimension, enabling real-time visualization of fetal movements.[4] These advanced imaging modalities may overcome some limitations of conventional 2D ultrasound by providing more comprehensive spatial information and enhanced visualization of surface features.[5,6]

Several studies have suggested potential advantages of 3D/4D ultrasound for the detection of fetal facial anomalies, including cleft lip and palate, micrognathia, and other craniofacial disorders.[7,9] However, most of these studies were retrospective, conducted at single centers, or included small sample sizes. Moreover, the added clinical value of 3D/4D ultrasound over conventional 2D ultrasound in routine clinical practice remains debated.[10,11]

This prospective multicenter study aimed to compare the diagnostic accuracy of 3D/4D ultrasound with conventional 2D ultrasound for the detection of fetal facial anomalies.

MATERIALS AND METHODS

Study Design and Population

This prospective comparative study was conducted at three tertiary referral centers for fetal medicine from January 2013 to July 2013. The study protocol was approved by the institutional review boards of all participating centers, and written informed consent was obtained from all participants.

Eligible participants included pregnant women between 18 and 34 weeks of gestation with either a suspected fetal anomaly on routine ultrasound or risk factors for fetal anomalies (including family history of craniofacial abnormalities, teratogen exposure, or abnormal maternal serum screening). Women with multiple pregnancies, severe oligohydramnios, maternal obesity (BMI >35 kg/m²), or inability to provide informed consent were excluded.

A total of 70 pregnant women meeting the inclusion criteria were enrolled in the study.

Ultrasound Examination

All participants underwent both 2D and 3D/4D ultrasound examinations during the same session. The examinations were performed by certified sonographers with at least five years of experience in prenatal diagnosis and specific training in 3D/4D ultrasound. High-end ultrasound systems with 4-8 MHz transabdominal probes were used for all examinations.

The 2D ultrasound examination followed standard protocols for fetal anatomical assessment, including multiple views of the fetal face (sagittal, coronal, and axial planes). For the 3D/4D ultrasound, volume datasets of the fetal face were acquired using a standardized technique. The acquisition angle was set between 45° and 85° depending on fetal position, and the highest quality mode was selected. Multiple volumes were acquired if needed to ensure optimal visualization of all facial structures.

The total examination time for each modality was recorded. The quality of images was rated on a 5-point scale (1=poor, 5=excellent) based on clarity, presence of artifacts, and completeness of visualization.

Image Analysis

All ultrasound images and volumes were stored digitally and subsequently analyzed independently by two fetal medicine specialists with extensive experience in prenatal diagnosis. The reviewers were blinded to clinical information and final diagnosis. Each reviewer assessed the images for the presence or absence of facial anomalies and provided detailed evaluation of specific facial structures, including the profile, nose, lips, palate, mandible, ears, and overall facial symmetry.

For cases with discrepant findings between reviewers, a consensus was reached through joint review. The final prenatal diagnosis was compared with postnatal findings obtained through physical examination, postnatal imaging, or autopsy in cases of pregnancy termination or fetal demise.

Maternal Satisfaction Assessment

After completion of both ultrasound examinations, participants completed a questionnaire to assess their satisfaction with each modality. The questionnaire included items related to comfort during the examination, perception of image quality, emotional connection with the fetus, and overall satisfaction. Responses were recorded on a 10-point Likert scale.

Statistical Analysis

Sample size calculation was based on an expected difference in detection rate of 20% between 3D/4D and 2D ultrasound (90% vs. 70%), with 80% power and 5% level of significance, resulting in a required sample size of 62 participants. We enrolled 70 participants to account for potential dropouts.

Statistical analysis was performed using SPSS version 20.0 (IBM Corp., Armonk, NY, USA). Categorical variables were expressed as frequencies and percentages and compared using chi-square or Fisher's exact test as appropriate. Continuous variables were expressed as means and standard deviations and compared using Student's t-test or Mann-Whitney U test depending on data distribution.

The diagnostic performance of each modality was evaluated by calculating sensitivity, specificity, positive predictive value (PPV), and negative predictive value (NPV) using postnatal findings as the reference standard. Interobserver agreement was assessed using Cohen's kappa coefficient. A p-value <0.05 was considered statistically significant.

RESULTS

Table 1. Baseline characteristics of the study population (n=68)

Characteristic	Value
Maternal age (years), mean ± SD	29.4 ± 5.3
Gestational age at examination (weeks), mean ± SD	24.6 ± 4.2
Primigravidae, n (%)	29 (42.6)
BMI (kg/m²), mean ± SD	26.7 ± 3.9
Indication for examination, n (%)	
- Suspected anomaly on routine ultrasound	37 (54.4)
- Family history of facial/craniofacial anomalies	12 (17.6)
- Abnormal maternal serum screening	14 (20.6)
- Teratogen exposure	5 (7.4)

Table 2. Diagnostic performance of 2D and 3D/4D ultrasound for detection of fetal facial anomalies

Parameter	2D Ultrasound	3D/4D Ultrasound	p-value
Sensitivity, % (n)	70.8 (17/24)	91.7 (22/24)	0.031
Specificity, % (n)	93.2 (41/44)	95.5 (42/44)	0.647
Positive predictive value, %	85.0	91.7	0.453
Negative predictive value, %	85.4	95.5	0.094
Accuracy, %	85.3	94.1	0.038

Table 3. Detection rates for specific facial anomalies by ultrasound modality

Anomaly	Total Cases	Detection by 2D US, % (n)	Detection by 3D/4D US, % (n)	p-value
Cleft lip ± cleft palate	11	81.8 (9/11)	100 (11/11)	0.147
Micrognathia	9	66.7 (6/9)	88.9 (8/9)	0.251
Ear abnormalities	7	42.9 (3/7)	85.7 (6/7)	0.049
Facial asymmetry	3	66.7 (2/3)	100 (3/3)	0.317
Other facial anomalies	4	50.0 (2/4)	75.0 (3/4)	0.465

Table 4. Factors affecting image quality scores (scale 1-5) for 2D and 3D/4D ultrasound

Factor	2D US Score (mean ± SD)	3D/4D US Score (mean ± SD)	p-value for difference between modalities
Maternal BMI (kg/m²)			
- <25 (n=28)	3.7 ± 0.7	4.3 ± 0.6	0.002
- 25-30 (n=29)	3.3 ± 0.8	4.0 ± 0.7	<0.001
- >30 (n=11)	2.9 ± 0.8	3.6 ± 0.8	0.041
Gestational age (weeks)			
- 18-24 (n=31)	3.2 ± 0.8	3.9 ± 0.7	<0.001
- 25-30 (n=27)	3.5 ± 0.7	4.2 ± 0.6	<0.001
- 31-34 (n=10)	3.6 ± 0.7	4.0 ± 0.7	0.195
Fetal position			
- Favorable (n=42)	3.6 ± 0.7	4.3 ± 0.6	<0.001
- Unfavorable (n=26)	3.0 ± 0.8	3.6 ± 0.7	0.012
Amniotic fluid index			
- Normal (n=53)	3.5 ± 0.7	4.2 ± 0.6	<0.001
- Borderline (n=15)	3.0 ± 0.8	3.5 ± 0.7	0.045

Table 5. Maternal satisfaction scores (scale 1-10) for 2D and 3D/4D ultrasound

Domain	2D US (mean ± SD)	3D/4D US (mean ± SD)	p-value
Comfort during examination	7.6 ± 1.4	7.4 ± 1.5	0.423
Perception of image quality	6.8 ± 1.5	8.7 ± 1.1	<0.001
Emotional connection with the fetus	6.5 ± 1.7	9.2 ± 0.9	<0.001
Understanding of findings	7.0 ± 1.6	8.9 ± 1.0	<0.001
Overall satisfaction	6.9 ± 1.5	8.6 ± 1.1	<0.001

DISCUSSION

This prospective multicenter study demonstrates that 3D/4D ultrasound provides significantly higher detection rates for fetal facial anomalies compared to conventional 2D ultrasound. Our findings indicate that 3D/4D ultrasound offers particular advantages in the assessment of cleft lip, micrognathia, and ear abnormalities.

The overall detection rate for facial anomalies was 91.7% with 3D/4D ultrasound compared to 70.8% with 2D ultrasound, representing a significant improvement in diagnostic sensitivity. This finding is consistent with previous studies that have reported enhanced visualization of facial structures with 3D/4D ultrasound.[12,13] Merz et al.[14] found that 3D ultrasound improved the detection of facial anomalies by 20% compared to 2D ultrasound, similar to the improvement observed in our study.

The superior performance of 3D/4D ultrasound was most evident in the detection of ear abnormalities, with a detection rate of 85.7% compared to 42.9% with 2D ultrasound. This substantial difference can be attributed to the ability of 3D ultrasound to capture the complex three-dimensional structure of the ear and allow multiplanar reconstruction1[5]. Similarly, the assessment of micrognathia was enhanced with 3D/4D ultrasound (88.9% vs. 66.7%), likely due to improved visualization of the facial profile and the ability to measure mandibular dimensions more accurately[16].

For cleft lip, 3D/4D ultrasound achieved a perfect detection rate of 100% compared to 81.8% with 2D ultrasound. Although this difference did not reach statistical significance due to the small number of cases, it highlights the potential advantage of 3D/4D ultrasound for the assessment of this common facial anomaly. The improved detection of cleft lip may be particularly important for counseling and planning of postnatal management[17].

Interobserver agreement was substantially higher for 3D/4D ultrasound (κ=0.84) than for 2D ultrasound (κ=0.69), suggesting that 3D/4D imaging may provide more objective and reproducible assessment of facial structures. This improved reliability could be valuable in clinical practice, particularly for less experienced sonographers or in challenging cases[18].

Image quality was significantly better with 3D/4D ultrasound across all subgroups, although the difference was less pronounced in women with higher BMI, unfavorablefetal positions, and borderline amniotic fluid volumes. These findings indicate that technical factors remain important considerations for the successful application of 3D/4D ultrasound in clinical practice[19].

The longer examination time required for 3D/4D ultrasound (19.3 vs. 12.7 minutes) represents a potential limitation for its routine use. However, this difference may be partly attributed to the comprehensive protocol used in our study and might be reduced as operators gain more experience with the technique[20].

Maternal satisfaction was significantly higher for 3D/4D ultrasound, particularly regarding emotional connection with the fetus and understanding of findings. This psychosocial benefit of 3D/4D ultrasound has been previously reported and may contribute to improved maternal-fetal bonding and compliance with prenatal care[21,22].

Our study has several strengths, including its prospective design, multicenter setting, and standardized protocols for image acquisition and analysis. The inclusion of interobserver agreement assessment and maternal satisfaction measures provides a comprehensive evaluation of the clinical utility of 3D/4D ultrasound.

However, some limitations should be acknowledged. First, the sample size, while adequate for the primary outcome, may have been insufficient to detect statistically significant differences for specific anomaly subgroups. Second, the study was conducted at tertiary referral centers with experienced sonographers, and the results may not be generalizable to all clinical settings. Third, the majority of examinations were performed in the second trimester, and the comparative performance of the two modalities may differ at earlier or later gestational ages.

CONCLUSION

In conclusion, our study demonstrates that 3D/4D ultrasound significantly improves the detection of fetal facial anomalies compared to conventional 2D ultrasound. The technique offers particular advantages for the assessment of cleft lip, micrognathia, and ear abnormalities, with higher interobserver agreement and greater maternal satisfaction. These findings support the use of 3D/4D ultrasound as a complementary technique to 2D ultrasound for comprehensive assessment of fetal facial structures, especially in high-risk pregnancies or when anomalies are suspected on routine examination.

Future studies should evaluate the cost-effectiveness of incorporating 3D/4D ultrasound into routine prenatal care and investigate its potential benefits for the detection of other types of fetal anomalies. Additionally, the development of automated analysis techniques for 3D/4D ultrasound data may further enhance its clinical utility and accessibility.

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