Fetal Facial Abnormalities: From 2D Sonography to HDlive and HDlive Silhouette Mode

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ABSTRACT

Fetal face examination is essential to complete a full anatomical scan. Two-dimensional (2D) sonography is the basic standardized tool used. The anatomical nature of the fetal face should be accurately judged along with the exclusion of all possible anomalies, but this is sometimes difficult to achieve with 2D sonography alone. Three-dimensional (3D) ultrasound provides detailed data on the fetal face. Many studies showed its superiority for detecting anomalies that were missed by 2D sonography. HDlive provides life-like images of the fetal face, and the HDlive silhouette mode provides hologram-like images of structures of interest. In this article, the development of ultrasound from 2D to conventional 3D and new rendering modes, such as HDlive and the HDlive silhouette mode will be presented, highlighting these new techniques over conventional 2D sonography for detecting fetal facial anomalies.

Keywords: 2D sonography, 3D ultrasound, Facial abnormality, Fetus, HDlive, HDlive silhouette mode.


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INTRODUCTION

Fetal face scanning represents a challenging task for obstetricians. Two-dimensional (2D) sonography is the standard tool described in universally approved guidelines for fetal scanning including fetal face examination.1,2 Although new technology has provided higher-quality 2D images, there are still limitations in fetal face examination, especially when the fetal position is suboptimal. This is due to the fact that the fetal face is a three-dimensional (3D) structure with complex curvatures. This renders its comprehensive evaluation using 2D sonography alone insufficient. The importance of fetal face examination is that the detection of fetal anomalies can offer diagnostic clues for different chromosomal and genetic disorders.3-13

This review considers fetal facial evaluation and related studies in line with the development of ultrasound from 2D to 3D, with its different rendering modes.

TWO-DIMENSIONAL SONOGRAPHY

The use of 2D sonography in obstetrics and gynecology began in 1958 by Donald et al.14 The 2D sonography was used for the detection of congenital anomalies in 1972, when Campbell15 and his group reported the diagnosis of a case of anencephaly at 17 weeks of gestation. It became widely used because it is a safe, noninvasive tool with real-time imaging. However, for this technique, the examiner must have experience to interpret the images for diagnosis because in some cases 2D sonography has limitations for evaluation.16,17 Conventional 2D sonography has a sensitivity of 40.4% for the detection of fetal anomalies.18 Because of the curvature of the fetal face, the physician has to evaluate it in many planes as axial, sagittal, coronal, and oblique views in each fetus. Also, the examiner has to create a 3D view of the image in his/her mind because the image is displayed in two dimensions only.

Despite its limitations and difficulties to evaluate the fetal face, many articles have described the visualization of various facial structures and detection of numerous facial anomalies using 2D sonography at different gestational ages.19-26

CONVENTIONAL 3D ULTRASOUND

The 3D ultrasound provides a three-orthogonal plane image capable of facilitating thorough assessment of fetal facial characteristics and the detection of anomalies. Since its introduction, the adjunct use of 3D ultrasound has been shown to be superior to 2D sonography alone owing to its ability to demonstrate planes that could not be visualized using conventional 2D sonography (Fig. 1). Four-dimensional (4D) ultrasound is a real-time visualization of the 3D image. This technique represents the same application as 3D ultrasound but it has the benefit of being applicable even with a moving fetus, while 3D ultrasound...
Ais suitable only for a fetus at rest. Kurjak et al.27 stated that adding a temporal factor, which is characteristic of 4D ultrasound, facilitates the visualization of facial expressions. This is due to its ability to detect fetal facial movements, which might reflect fetal brain and central nervous system development.

**HDlive**

HDlive is a new surface-rendering mode that is characterized by an adjustable light source that enables depth perception through lighting and shadowing effects.28 This results in real skin-like color images of the fetus. This technique has been shown to be advantageous for the prenatal diagnosis of various fetal anomalies.29-39 Pooh and Kurjak40 reported that HDlive can depict fetal facial structures as early as in the first trimester. Because of its adjustable light source and shadowing effect, HDlive demonstrates abnormal positions of fetal facial structures in the first trimester, such as low-set ears, more clearly than 2D sonography and 3D ultrasound. Improvements in the physician’s confidence for diagnosis and the counseling of parents can be achieved using this technique. Details of a normal fetal face can be clearly demonstrated using this technique early in gestation (Fig. 2). The adjustable light source can create images with fine details of the face (Figs 3A and B). This enables precise identification of the location and relation between different fetal facial structures.

**HDlive Silhouette Mode**

The HDlive silhouette mode provides vitreous-like clarity of fetal structures.41 The main principle of this technique is that it delineates the outline of a structure of interest, but keeps the core transparent. So, the cystic fetal facial structures, such as eyeballs can be revealed through the fetal surface.42 It was shown to be beneficial for assessment of the fetal heart.43 Also, it has the ability to demonstrate structures that lie behind the structure of interest.44 Therefore, it may be a good alternative overcoming the
problem of the fetal face obscured behind a fetal arm, which can be encountered when any other display or rendering modes are used.

FETAL FACIAL PART

Eyes

A routine mid-trimester scan performed using 2D sonography is limited to detection of the orbit and lens, as 2D sonography has a limited ability to accurately detect the fetal eyes (Figs 4A and B). Some charts were developed to identify anomalies as hypo- and hypertelorism using 2D sonography, but these methods remain difficult to apply. With the appearance of 3D ultrasound, an improved evaluation of the fetal eye was achieved. The advantages of 3D ultrasound over 2D sonography in eye anomaly detection have been unclear due to variable results. The 3D ultrasound showed the same detection rates as 2D sonography for identifying anomalies. However, more image planes and multiple features in the same image can be viewed using 3D ultrasound, especially in cases of hypertelorism (Fig. 5).

Dyson et al reported that 3D ultrasound could also demonstrate a case of hypertelorism missed by 2D sonography.

HDlive has the additional advantage of an adjustable light source; therefore, depth perception can be achieved. This point is valuable as in the palpebral fissure visualization, which can be demonstrated with fine details using this technique (Fig. 6). This fact was confirmed by Hata et al where HDlive was able to demonstrate hypotelorism and hypertelorism with a more realistic appearance compared with conventional 3D ultrasound. HDlive with its adjustable light source was also superior to 3D ultrasound for detecting upward slanting eyes and narrow eyelid folds, which were missed by 3D ultrasound (Figs 7A to C). Moreover, HDlive showed images identical to the postpartum ones on the simultaneous display of cyclopia in addition to a clearer demonstration of the midline-situated proboscis and generated more accurate images compared with conventional 3D ultrasound (Figs 8A to F). Experienced operators familiar with this technique can adjust the light source in the fetal profile view to easily identify an exophthalmic-protruded eye.

The unique characteristic of the HDlive silhouette mode to show the delineated outlines of structures of interest while simultaneously showing its inner core makes it more beneficial for identifying the fetal face and associated anomalies. The ability to view the lenses with the HDlive silhouette mode is a marked advantage over conventional 3D ultrasound and HDlive, which cannot demonstrate them (Figs 9A and B).

Ear

Ear examination has markedly advanced due to the development of ultrasound technology. The 2D sonography can
Figs 7A to C: The fetal face reconstructed by HDlive in a case of trisomy 21 at 29 weeks and 5 days of gestation. Flattened nose, angle of the mouth turned down, and upward slanting eyes with a left oblique view (A) frontal view; (B) right oblique view; and (C) are evident (Courtesy: Reprinted with permission from Hanaoka et al (A and B) and Hata et al (C)).

Figs 8A to F: Conventional 3D sonographic (A to C) HDlive; and (D to F) images of cyclopia and a proboscis at 32 weeks of gestation (Courtesy: Reprinted with permission from Hata et al (A to C) and (D to F)).

Figs 9A and B: HDlive (A) HDlive silhouette mode; and (B) images of a normal fetus at 24 weeks and 4 days of gestation that clearly show the eyes, nose, and mouth. The HDlive silhouette mode can precisely depict the right lens (L). This is superior to conventional 3D ultrasound and HDlive.
demonstrate the fetal ear length, which was described in some reports as an indicator of chromosomal abnormalities. However, the correlation between the fetal ear length and chromosomal abnormalities is not as strong as the fetal ear position as marker of chromosomal abnormalities, such as trisomy 18 and trisomy 21. However, it is always difficult to detect the fetal ear position using 2D sonography. Three-dimensional ultrasound can demonstrate a profile view of the fetal face to judge the position of the ear (Fig. 10). A line drawn from the outer canthus of the eye to the occiput is used as an indicator of the fetal ear. As we previously mentioned, the HDlive technique with its adjustable light source can demonstrate the fetal eye better than 3D ultrasound, and, therefore, the ability to accurately assess the fetal ear position can be achieved using HDlive compared with 3D ultrasound. This can be easily understood when comparing 3D with HDlive images.

Lip and Mouth

The detection of anomalies of the lip and mouth is mandatory for obstetricians and sonographers during pregnancy. This is due to difficulties that can be encountered at birth during resuscitation when cleft lip and palate are present.

Since the introduction of 3D ultrasound, the adjunct use of this tool has been shown to be superior to 2D sonography alone (Figs 11A and B). Pretorius and Nelson demonstrated four cases in which normal lips were seen by 3D ultrasound alone, while they were undetected by 2D sonography. This can be explained by the inability to obtain a correct midsagittal plane in some cases, which might result in diagnostic inaccuracies. Merz et al performed one of the earliest studies involving 618 pregnant women to address the value of 3D ultrasound in fetal facial examinations. Abdominal 3D ultrasound was able to demonstrate all facial anomalies in 25 cases with abnormal faces, while 2D sonography missed 5 cases (2 cases were cleft lip/palate). Even when transvaginal 3D ultrasound was used at an earlier gestational age in this study, clear images of the fetal face could be obtained as early as 9 weeks using the surface rendering mode. Moreover, the proper correction of deviation of the exact plane of the fetal profile, which might be necessary during 2D sonographic examination, can be achieved using this technique. This confirms the benefit of 3D ultrasound for obtaining the midsagittal profile. Data from Dyson et al were consistent with the previously mentioned study by Merz et al, confirming the superiority of 3D ultrasound over 2D sonography for detecting cases of cleft lip and palate. The 3D ultrasound provides the ability to evaluate orthogonal and axial planes at the same time. Therefore, simultaneous
assessment of the alveolar ridge and upper lip can be achieved. This is of marked importance because an isolated cleft palate can be missed in the diagnosis because of acoustic shadows caused by facial bones.\textsuperscript{11} Moreover, 3D ultrasound has the ability to provide an image of the fetal face/profile when the position is unfavorable to give a clear image with 2D sonography. This can allow an earlier and a more accurate diagnosis of cleft lip/palate compared with the use of 2D sonography alone.\textsuperscript{50} Previous studies showed a significantly shorter time to examine the fetal face using 3D ultrasound relative to 2D sonography alone (5.33 vs 6.2 seconds respectively, \(p < 0.001\)), resulting in 100% for all of the sensitivity, specificity, positive predictive value, and negative predictive value in the diagnosis of fetal cleft lip/palate. Also, in cases that can be diagnosed by 2D sonography, the use of 3D ultrasound enables accurate visualization of the cleft and increases diagnostic confidence compared with 2D sonography alone.\textsuperscript{51} HDlive can provide clearer and more discernible images of the fetal cleft lip compared with conventional 3D ultrasound. Accurate localization of the lesion as well as providing realistic images that can be useful in counseling of the parents and plastic surgeons represents the main benefit of this rendering mode over conventional 3D ultrasound. Moreover, HDlive was shown to be superior to 3D ultrasound for demonstrating details of the fetal cleft lip.\textsuperscript{33}

**Jaw**

Jaw anomalies, especially micrognathia, have attracted the interest of fetal sonography specialists due to the long list of chromosomal abnormalities that have been found to be associated with this anomaly. Micrognathia can be diagnosed based on a subjective impression, provided that a clear midsagittal fetal profile view is obtained. This is an easy method. An objective diagnosis of micrognathia depending upon the jaw index and inferior facial angles has different limitations, and sometimes it is impossible to use.\textsuperscript{52} Paladini\textsuperscript{52} demonstrated that 3D ultrasound with its surface rendering mode and maximum mode is very advantageous when used as an adjunct tool in diagnosing micrognathia. Dyson et al\textsuperscript{47} reported that all cases of micrognathia were seen only by 3D ultrasound and were missed by 2D sonography. Three-dimensional ultrasound has a role in quantitative and objective assessment of the jaw for micrognathia.

Hanaoka et al\textsuperscript{34} stated that HDlive could detect a case of micrognathia that was missed by conventional 3D ultrasound, probably due to its ability to clearly demonstrate the fetal profile and evaluate the chin accurately (Fig. 12). The HDlive silhouette mode might also be beneficial due to its ability to show curvatures of the fetal face; therefore, chin anomalies can be judged. However, there has been no report on its use in the antenatal diagnosis of micrognathia.

**Nose**

With an appropriate view of the fetal face, the nasal bone is visible using 2D sonography in more than 99% of normal karyotype fetuses.\textsuperscript{19} There have been many studies on the detection of fetal chromosomal abnormalities through fetal nasal bone screening during the second and third trimesters.\textsuperscript{21} However, an upturned nose or a flat nose that is associated with chromosomal abnormalities cannot be demonstrated well by 2D sonography. Hanaoka et al\textsuperscript{34} reported that an upturned nose was detected by conventional 3D ultrasound as well as the HDlive mode. However, in the same study, HDlive could demonstrate a flattened nose, which was missed by 3D ultrasound. Clear identification of the proboscis with HDlive images is possible compared with conventional 3D ultrasound images (Fig. 8).

**LIMITATIONS**

Conventional 3D ultrasound, HDlive, and the HDlive silhouette mode share some of the same limitations. Inadequate amniotic fluid in front of the fetal face impairs the image quality. A moving fetus may represent a problem during data acquisition while using conventional 3D ultrasound but not 4D ultrasound. The obstruction of the region of interest by the placenta, umbilical cord, or other body parts affects the image quality in all modes used but, as we mentioned, the HDlive silhouette mode is superior in this point.

\[\text{Fig. 12: HDlive clearly reveals microcephaly and micrognathia in a case of trisomy 18 at 27 weeks and 6 days of gestation (Courtesy: Reprinted with permission from Hanaoka et al\textsuperscript{34})}\]
CONCLUSION

The fetal face is a complex structure, and the diagnosis of its anomalies requires the use of new ultrasound techniques, such as conventional 3D ultrasound, HDlive, and the HDlive silhouette mode. The 3D ultrasound is superior to 2D sonography in detecting fetal facial anomalies. HDlive can provide life-like images of the fetal face and help to better identify and describe different fetal anomalies. The HDlive silhouette mode is superior when the fetal face is obscured by structures, such as fetal limbs or the umbilical cord when all other modes cannot properly display the face. These new techniques shorten the time needed for examination, and facilitate diagnosis. However, to master them, a learning curve is needed to accurately interpret the reconstructed images. HDlive and the HDlive silhouette mode can be beneficial adjunct diagnostic tools together with conventional 2D/3D ultrasound, with relative advantages over conventional modes in some cases of facial anomalies. Further studies involving a large sample size are needed to accurately determine the sensitivity and specificity of these new techniques in the diagnosis of various fetal facial abnormalities.

REFERENCES