Cranial Evaluation and the Fetal Face

Lawrence D. Platt, MD
Professor of Obstetrics and Gynecology
David Geffen School of Medicine at UCLA
Los Angeles, CA

Learning Objectives

• At the end of the presentation the attendee will
  1. Be able to recognize the fetal palate and appropriate landmarks
  2. Understand the importance of 3 D Rendering in the evaluation of the fetal lip, palate and ears
  3. Understand the value of 3 D rendered imaging in seeking consultations for patients with malformations
  4. Will Understand the role of Volume contrast imaging in assessing the cranial sutures

42 weeks and 31 weeks

Faces First Trimester
Getting the fetal profile

Surface mode of fetus with severe micrognathia

Various faces of normal fetuses

Surface mode of fetal face -- achondroplasia with frontal bossing and depression of the nasal bridge
Abnormal Inferior Facial Angle is abnormal

Predictability of Fetal Mandible Length on Ultrasound

The fetal mandible measurement: an objective determination of fetal jaw size

C. Otto and L. D. Platt

Fetal Mandible v. GA

Fetal Mandible vs. BPD

Retrognathia/micrognathia

Abnormal fetal faces: Down syndrome, Micrognathia, Apert syndrome
**Profile**

**Evaluation of Fetal Profile**

125 Cases

- Orthogonal display
- Abdominal transducer
- True profile in 70%
- Off profile by up to 3-20 degrees in 30%

Merz et al, UOG 9:237, 1997

**Facial Profiles**

- Aiding Genetic Diagnoses

**Landmarks of the Ear**

1. Helix
2. Crus of helix
3. Auricular tubercle
4. Antihelix
5. Crura of antihelix
6. Triangular fossa
7. Scaphoid fossa
8. Concha of auricle
9. External acoustic meatus
10. Tragus
11. Antitragus
12. Intertragal notch
13. Lobule of auricle

**Ears**
Infant with cat-cry syndrome

Mandibulofacial dysostosis

(L) Trisomy 13-15. Note the sharp angulation of the crus helicis, and hypoplasia of the antihelix and lobule

Normal for Comparison
Fetal Ear

Fetal Ear Length: A predictor of aneuploidy?

- 1,848 pts with singleton pregnancies with genetic amnio in 2nd or 3rd trimester
  - 1,311 fetuses evaluated with complete data for ear length, biometry, and anatomic survey
  - 48 (3.7%) had abnormal karyotype
  - 34/48 abnl karyotypes, considered significant; 11/34 (32.4%) abnl karyotypes had short EL
  - Incr BPD/EL ratio of ≥4.0 in fetuses with abnl karyotype

- Conclusion:
  - Short fetal EL noted on U/S, either alone or in combination with other U/S detected structural abnormalities, may be a useful parameter in predicting fetal aneuploidy


Normal Anatomy of the Oral Cavity

- The lip and palate form in the first trimester
- The lip closes at 5-6 weeks (conceptual age)
- The palate closes at 10 weeks (conceptual age)
- Clefts could be detected as early as the 11-12th postmenstrual weeks of pregnancy
- However most clefts are detected at the time of the initial 18-20 week ultrasound

Lips & Palate

Associated Anomalies

- A child with CL/P has a 30% chance of having the cleft in association with a syndrome
- A child with CP has a 50% chance of having the cleft in association with a syndrome

Lips

90210 vs 07631

Incomplete and Complete Cleft Lips (Pre-repair)

Unilateral Bilateral Unilateral Bilateral

Photo credit: Mary Spano, New York University Institute of Reconstructive Plastic Surgery. Used with permission.
Diagnostic accuracy of ultrasound in predicting CL/P

- CP can hardly be diagnosed c. standard 2D
- Low uptake of CL/P in low risk pregnancies (15%)
- Fetuses with multiple anomalies more frequently identified than isolated clefts
- Once a defects is identified ultrasound diagnosis of laterality and palate involvement is very precise (> 95%)
- Soft palate is not be reliably identified

Cleft Lip and Palate
31 Cases

- 28 Cleft lip + or - palate
  - 22 Cleft palate
    » 2D - 10/22 and 3D - 19/22
- 3 suspected clefts were normal
- 7 changed management
  - 3 for termination of pregnancy
  - 1 for continuation of pregnancy
  - 3 had no further workup (amnio)

Johnson et al, Radiology 217:236, 2000

Sonographic Diagnosis

- Depends on
  - Type of cleft
  - Severity of the cleft
  - Gestational age of the fetus
  - Presence of associated anomalies
  - Fetal position
  - Maternal body habitus
  - Expertise of sonographer/sonologist

Detection Rates of Cleft Lip/Palate Using Transabdominal Sonography

- 2D ultrasound in low-risk populations
- GA varied among studies; average was 24 weeks
- Demonstrates a range of diagnostic accuracy
  - Most studies report detection between 9-50%
  - However, reported rates ranged from:
    - 9-100% for cleft lip ± cleft palate
    - 0-22% for cleft palate only
    - 0-73% for all types of cleft
- False-positive diagnosis was low

Ultrasound Obstet Gynecol 2017

Detection Rates of Cleft Lip/Palate Using Transabdominal Sonography

- 3D US in high-risk populations
- GA varied among studies; average was 24 weeks
- High range of diagnostic accuracy by experts
  - Most studies reported detection rates of 100%
  - However, reported rates ranged from:
    - 100% for cleft lip only
    - 86-90% for cleft lip and palate
    - 0-89% for cleft palate only
- 3D US has high detection rates, but not for cleft palate only.

2D Ultrasound: Unilateral Left Cleft Lip

• 3 scanning planes result in the following views:
  – Profile
  – View of the nose and lips
  – Transverse section through the upper lip

Sagittal Coronal Axial

Courtesy of Ana Monteagudo, Used with permission.

Evaluation of the Upper Lip

• Ultrasound evaluation of the face and upper lip is part of the anatomical survey¹
• Reported detection rates of CL/P c. 2D US in low-risk population are low, ranging from 23 to 58%²
• To improve the dx. of CLP 2D & 3D/4D US need to be systematically employed. Using this protocol the detection rate for CLP can increases to as much as 87.5%³
• Recently, with the increased use of US during the 1st trimester as part of the “First Trimester Screen” reports of the diagnosis of CLP are emerging.


Profile is sometime misleading

CL/P unilateral CL/P bilateral

2D Ultrasound: Bilateral Left Cleft Lip

Maxillary pseudomass

2D Evaluation: Upper Lip and Palate

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How about an “early” diagnosis?

12 week Fetus
Median view of the fetal face that includes the nasal bone, then rotating the transducer 90° and slightly tilting its orientation to bring the frontal processes of the maxilla and primary palate into the same plane.

Face: Retronasal Triangle

2D vs. 3D Ultrasound
For the Detection of Cleft Lip and Palate

2D vs. 3D: Advantages of 3D
- Realistic images of the fetal face
- Facial expressions can be observed
- Hard-to-see structures can be imaged
- The family can see the true appearance of the fetus
- Counseling by other non-imaging physicians is facilitated

Cleft Palate
Normal for comparison
Cleft Lip and Palate

Cleft Lip and Palate: Pre and Post

2D vs. 3D: Advantages of 3D (Continued)

• Bedside or off-line analysis
• Off-site expert consult
• Data is easily revisited for additional evaluation
• Short processing time
• Effective teaching tool (simulating real time scanning)

2D vs. 3D: Limitations of 3D

• Inherent limitations of 3D sonography include:
  – Acoustic shadowing
  – Fetal movements
  – A fluid-tissue interface
  – Fetal position and maternal obesity

Cleft Palate/Cleft Lip

• 4th most common birth defect in U.S.A.
• Incidence: 1 in 700
• Clefts of the lip and primary palate
  – Occur independently from clefts of the secondary palate or vice versa
  – Caused by failure of the palatine to close between 5-8 days of embryogenesis
  – Readily seen on 2D ultrasound; however, due to limitations of 2D, majority are seen in neonates
  – 3D more optimal for antenatal diagnosis
Looking at the Fetal Palate:
Reverse Face: The “Campbell Technique”

Reversed facial view technique as described by Campbell, et al. When the 3D volume is rendered, the transverse hard palate can be identified.


Campbell S, Lees C, Moscoso G, Hall P. Ultrasound antenatal diagnosis

3D Reverse Face (3D RF)

- Technique:
  - To evaluate the lips and alveolar ridge
  - The face is rotated 180° on the vertical axis to assess the secondary palate by the 3-D RF view (the cut plane is directed from the back of the skull towards the front)
  - 3-D volume is rendered


Looking at the Fetal Palate:
Reverse Face: The “Platt Technique”

Start with this picture and acquire the volume


Panel A = Fetal profile (acquired image)

Acquired multiplanar image

Reference point placed at the level just below the philtrum

Acquired multiplanar image

Rendered image

Sequence of rendered images

The mandible and corresponding alveolar ridge are seen in Panel A.

The tongue can be seen in Panel B.
Panel A illustrates the acquisition. The rendered image in this fetus demonstrates a right-sided cleft lip.

The image is rotated so that the green cut plane is directed at the level of the chin (Panel B).

The cut plane is now in the proper location to begin the sequential analysis of the mandible maxilla.

“Flipped Face” Technique

- Once images obtained
  - Different filters can be selected to highlight soft tissues, bone and cartilage

Gradient Light

X-Ray

Surface Smooth

Surface

The rendered image at the level of the cleft of the alveolar ridge near the maxillary bone using different filters.

The surface smooth and the surface rendering filters provide the greatest detailed images of the tissue.

“Flipped Face” Technique

- First 50 cases examined achieved 100% confirmation following birth
- Technique improves upon capabilities to identify
  - Mandible
  - Maxillary bones
  - Respective alveolar ridges
  - Hard and soft palates

“Flipped Face” Technique

- Limitations
  - Predominantly related to acquisition of an adequate facial volume sans shadowing of the palate
    - Shadowing can be misinterpreted as a defect
  - Further study needed of the soft palate using this technique

Conclusion

- “Flipped face” technique
  - Rapid, highly effective method for identifying the fetal lips, alveolar ridges, hard and soft palates
  - As early as the 2nd trimester
  - After volume acquisitions, <2 minutes required to render, display and analyze
Looking at the Fetal Palate: 
Angled Insonation: The “Pilu Technique”

- Transducer is angled at an oblique angle of 45° to insonate the secondary palate
- A 3D volume is acquired
- 3D reconstruction of axial and coronal planes

Hard palate

Maxilla (Alveolar ridge)

Angle of insonation (45°)

Comparison of “Reverse face,” “Flipped Face” and “Oblique Face”

- Which method is best?

Table 2 Percentage of fetuses without cleft lip or palate (n = 50) in which different structures were well visualized using each technique

<table>
<thead>
<tr>
<th>Feature</th>
<th>Reverse-face view (% n)</th>
<th>Flipped-face view (% n)</th>
<th>Oblique-face view (% n)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Lip</td>
<td>100 (50/50)</td>
<td>100 (50/50)</td>
<td>100 (50/50)</td>
</tr>
<tr>
<td>Alveolar ridge</td>
<td>100 (50/50)</td>
<td>100 (50/50)</td>
<td>100 (50/50)</td>
</tr>
<tr>
<td>Hard palate</td>
<td>78 (39/50)</td>
<td>84 (42/50)</td>
<td>86 (43/50)</td>
</tr>
<tr>
<td>Soft palate</td>
<td>0 (0/50)</td>
<td>16 (8/50)</td>
<td>26 (13/50)</td>
</tr>
</tbody>
</table>

Comparison of “Reverse face,” “Flipped Face” and “Oblique Face”

- Which method is best?

Table 1 Percentage of fetuses with cleft lip and palate (n = 10) in which abnormal findings were well visualized using each technique

<table>
<thead>
<tr>
<th>Feature</th>
<th>Reverse-face view (% n)</th>
<th>Flipped-face view (% n)</th>
<th>Oblique-face view (% n)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Lip</td>
<td>100 (10/10)</td>
<td>100 (10/10)</td>
<td>100 (10/10)</td>
</tr>
<tr>
<td>Alveolar ridge</td>
<td>100 (9/9)</td>
<td>100 (9/9)</td>
<td>100 (9/9)</td>
</tr>
<tr>
<td>Hard palate</td>
<td>94 (77/82)</td>
<td>85 (77/85)</td>
<td>100 (95/100)</td>
</tr>
<tr>
<td>Soft palate</td>
<td>0 (0/7)</td>
<td>14 (1/7)</td>
<td>14 (1/7)</td>
</tr>
</tbody>
</table>

Technique:
- A 2D profile is obtained
- Transducer is angled at an oblique angle of 45° to insonate the secondary palate
- A 3D volume is acquired
- 3D reconstruction of axial and coronal planes
3D Imaging of the Fetal Face – Recommendations from the International 3D Focus Group

- **Purpose of the recommendations**
  - To demonstrate the different possibilities of assessing the normal and abnormal fetal face with 3D/4D ultrasound
  - To give the operator an overview of the benefit resulting from the application of that technology


### Display Modes

- **Three-orthogonal-plan display**
  - All conventional scan planes can be viewed
  - Simultaneous display of all three perpendicular sectional planes provides an ideal basis for a detailed survey of the fetus and allow the demonstration of a specific 3D plane to be precisely controlled.
  - To optimize the position of the face, the cursor dot should be placed between the orbits in the coronal plane and the face rotated in all planes until it is symmetrical.

### Parallel-plane (= tomographic) display

- Region of interest can be displayed in several parallel slices
- This enables a detailed tomographic survey of the fetal face in sagittal, coronal, and transverse slices at different distances:
  - Sagittal plane helpful to demonstrate the profile and orbits
  - Coronal – to assess the orbits and the palate
  - Transverse – to evaluate the orbits, palate, maxilla, and mandible

### 3D Surface Display

- 3D rendering algorithms depict the fetal face: surface mode, soft surface mode, light mode and soft light mode
- Pre-requisites for high-quality 3D US images:
  - Sufficient amniotic fluid in front of the structure
  - Absence of overlying or adjacent structures (or removed with the electronic scalpel). Electronic scalpel cannot be used to overcome acoustic shadows/artifacts

### Transparent Display

- Different transparent modes: maximum mode, x-ray mode
- Maximum mode used to visualize hyperechogenic structures
- Provides a complete survey of the fetal skill and facial bones
Sutures, cranial bones

3D Imaging of the Fetal Face – Recommendations from the International 3D Focus Group

- Display Modes
  - 4D Ultrasound (Real-Time 3D US)
    - Allows real-time visualization in any of the modes
    - Enables the physician to study fetal facial movements, i.e., yawning, sucking, swallowing, mimicking
    - Offers parents the opportunity to observe fetal movements
    - Diagnostically, 4D US is always helpful in the moving fetus, and 3D US in the fetus at rest

Assessment of the Fetal Face by 3D US in the 1st Trimester

| 1. Anatomy (3D orthogonal plane display) | sagittal view | profile with forehead, nose, lips and chin
| (3D surface display, transparency = maximum intensity display) | coronal view | forehead, orbits, nasal bone, maxilla and mandible
| transverse view | frontal, orbits, both nasal bones, maxilla and mandible
| 2. Biometry | sagittal view | frontonasal facial angle (fronto-nasal angle = 45°)
| (maximum intensity display) | coronal view | orbits with maxilla (symmetry)
| (maximum intensity display) | transverse view | orbits with maxilla (symmetry)
| 3. Documentation | coronal view | frontal view

Assessment of the Fetal Face by 3D US in the 2nd and 3rd Trimester

| 1. Anatomy (3D orthogonal plane display) | sagittal view | profile with forehead, nose, lips and chin
| (3D surface display, transparency = maximum intensity display) | coronal view | forehead, orbits, nasal bone, maxilla and mandible
| transverse view | frontal, orbits, both nasal bones, maxilla and mandible
| 2. Biometry | sagittal view | frontonasal facial angle (fronto-nasal angle = 45°)
| (maximum intensity display) | coronal view | orbits with maxilla (symmetry)
| (maximum intensity display) | transverse view | orbits with maxilla (symmetry)
| 3. Documentation | coronal view | frontal view

Assessment of the Fetal Face by 3D US in the 2nd and 3rd Trimester

| 2. Biometry | sagittal view | nasal bone
| (3D surface radiography) | coronal view | orbital diameter
| transverse view | orbital diameter
| 3. Documentation | nasal bone

Limitations and Pitfalls

- Fetal or probe movements during data acquisition lead to motion artifacts
- Surface rendering cannot be done in cases of severe oligohydramnios
- Overlying or adjacent structures interfere with surface rendering and must first be removed with the electronic scalpel
- Artifact defects caused by long bones of arms or legs in front of the fetal face
- Setting the threshold too high or faculty manipulation of the electronic scalpel leads to iatrogenic structural defects
- Surface display of the fetal face in the early embryologic development (1T or early 2T) does not show the typical human facial appearance seen in the last 2T or in the 3T
  - Should not be misinterpreted as a malformation of the fetal face
Craniofacial Cleft Classifications

**Cleft 0**, or median craniofacial dysraphia, its course is outlined from the anterior fontanelle through the frontal bone, crista galli, midline of the nose, columella, lip, and maxilla, and may actually involve the tongue, lower lip, and mandible. Its cranial extension is cleft 14.

**Cleft 1**, or paramedian craniofacial dysraphia, courses through the frontal bone and the olfactory groove of the cribiform plate, between the nasal bone and the frontal process of the maxilla, and through the maxilla between the central and lateral incisors. Its cranial extension is cleft 13.

**Cleft 2**, or paranasal cleft, is similar to cleft 1, but it is slightly more lateral. Its cranial extension is cleft 12.

Tessier Cleft

Tessier Cleft Ear Tag
Craniofacial Clefting

Postnatal Confirmation

Fetal Orbits

Orbital and ocular anomalies

Rosati, et al (Italy, 2002)
- Evaluated 2,717 fetuses
- 11-16 wks GA, high-resolution TVU
- Interocular distance (IOD), binocular distance (BOD) and orbital diameter (OD) obtained

- Results
  - Orbital measurements increased linearly throughout early pregnancy with good correlation to GA
- TVU is able to visualize and measure OD accurately in early pregnancy
  - Reference ranges developed can be used to evaluate normal development and aid in prenatal diagnosis of syndromes with orbital growth defects and other associated anomalies

Normal

Hypotelorism (holoprosencephaly or severe craniostenosis)

Hypertelorism (normal variant, cleft lip/palate, craniostenosis)

Ocular anomalies

Anophtalmia/microphthalmia

Cataract

Unilateral

Bilateral

Congenital cataracts

12 weeks 5 days

15 weeks 4 days

Intrauterine evolution of microphthalmia

- Well documented cases of infants with microphthalmia with normal appearing eyes in early gestation*
- Microphthalmia can be a progressive intrauterine disease and prenatal diagnosis with sonographic is not certain


Fetal Head

- Symmetry of pathology
  - Hemorrhage
  - Schizencephaly
  - Ventriculomegaly

- Sutures
  - Craniosynostosis
  - Abnormal appearance

- Mild ventriculomegaly
  - Dandy Walker variant
  - Identify corpus callosum

Sutures


**Syndromic craniosynostosis**

Cloverleaf skull (Pfeiffer lethal)

Normal and abnormal fetal skulls

Cloverleaf

Trigonocephaly

**Conclusion**

- The face reveals anatomic and chromosomal anomalies
- No sonographic evaluation of the fetus is complete without examining the face
- No fetal neuroscan is complete without scrutinizing the fetal face
- In addition to using 2D become familiar with utilizing 3D US for the task

**Thank You**