INTRODUCTION

• Causes extreme parental distress because of it being so visible.
• Examination of the fetal face is extremely important to demonstrate whether the fetus is normal or is it a part of aneuploidies or syndromes.\(^1,2\)
• Chromosomal abnormalities are present in 32 to 77\% of fetuses with facial defects depending on the type of defect.\(^3\)
• Imaging at 16 to 18 weeks of gestation can delineate the detailed facial anatomy\(^4\) (Figs 1 and 2).
• Primarily three views are taken to evaluate the fetal face that include coronal view, transverse view and sagittal view.
• Normal liquor amnii with outlining of the fetal face is important for demonstration by ultrasound.
• Soft tissues of the upper and lower lip and the nostrils are delineated by the modified coronal plane.
• Palate can be delineated by a modified coronal and modified longitudinal view.
• Fetal orbits and lens can be delineated by the coronal views.
• Nasal bone and mandible are delineated by the longitudinal view.
• Ears are delineated by the modified longitudinal plane (Tables 1 and 2).
• Oligohydramnios, maternal obesity and fetal position restrict visualization of the detailed fetal facial anatomy.
• 3D ultrasound now offers improved delineation of fetal facial anatomy in normal and malformed fetuses (Figs 3 to 8).
• Various fetal activities and fetal moods can be demonstrated beautifully on 3D ultrasound (Figs 9 to 13).
• Among facial activities observed by 4D-US, simultaneous eyelid and mouthing movements dominate between 30 and 33 weeks of gestation. Pure mouth movements, such as mouth opening, tongue expulsion, yawning and pouting are present, but at a significantly lower incidence.\(^5\)
• 4D sonography depicts different facial expressions and movements, which might represents fetal awareness. As of now one can only study the quality and not the quantity of facial movement patterns.\(^6\)
• Fetal behavioral patterns directly reflect the developmental and maturational processes of the central nervous system. 4D observation of fetal and early neonatal period may add to better understanding of the neurological development of the fetus.\(^7\)
Table 1: Imaging facial anatomy

1. Upper, lower lip and nostrils – Modified coronal plane
2. Palate – Modified coronal and modified longitudinal views
3. Orbits and the lens – Coronal view
4. Nasal bones and mandible – Longitudinal view
5. Ears – Modified longitudinal planes

Table 2: Fetal face

- Orbits
  - Globe
  - Lens
  - Ocular diameter
  - Intercocular distance
  - Binocular distance
- Lips
  - Continuity
- Nostrils
  - Number
  - Continuity
- Maxilla
  - Continuity of alveolar ridge
- Mandible
  - Size and position of chin
- Ear
  - Number
  - Structure
  - Position
• 4D ultrasonography is a powerful tool in the assessment of fetal behavior, and studies show that there is a continuity from fetal to neonatal behavior, especially in terms of isolated eye blinking movements, mouth and eyelid opening, yawning, tongue expulsion, smiling, scowling and hand movements directed to other parts of the face.

**ORBITS AND LENS**

• Axial coronal view of the orbits can visualize the globe and the lens (Fig. 14).
• Ocular diameter: Measured from medial inner to medial lateral wall of the orbit.
• Interocular distance: Measured from medial inner wall of one orbit to medial inner wall of the other orbit.
• Binocular distance: Measured from lateral inner wall of one orbit to lateral inner wall of the other orbit.
• Normal values for ocular diameters, interocular distances and binocular distances for different gestational ages are available (Figs 15 to 17).
Lips, Maxilla, Mandible
Nasal Bone and Tongue

- Modified coronal view for lips and maxilla\textsuperscript{10} (Figs 18 to 21).
- 3D ultrasound is more beneficial for evaluating facial malformations (Figs 22 to 26).
- Surface display rendering and multiplanar reconstruction in three planes define facial malformations much better.\textsuperscript{11}
• Axial plane view of the maxilla and soft tissue overlying it and the tooth-bearing alveolar ridge (Fig. 27).
• Color Doppler can sometimes be used to assess a normal palate. This can be seen with color flow imaging with respiratory activity showing movement in the nasopharynx superior to an intact palate (Fig. 28).
• For nasal bone mid-sagittal view of the fetal profile should be obtained. Charts of nasal bone length for various periods of gestation are available.12
• Longitudinal (facial profile) view and transverse view of the mandible, tongue, hard palate and nasal bridge (Figs 29 to 32).
Fig. 29: Fetal mandible and base of oral cavity as seen on transverse section

Fig. 30: Normal palate seen with color flow imaging with respiratory activity showing movement in the nasopharynx superior to an intact palate

Fig. 31: Sagittal view showing the maxilla (solid line) and mandible (dashed line)

Fig. 32: Sagittal view of a normal fetal profile showing the osseous and soft tissue components. With the fetal mouth open the normal positioning of the tongue can also be seen (solid line)

Ears
- Usually the ear in the near field if outlined by amniotic fluid is clearly demonstrated.
- Fetal ear in the far field with the fetus in lateral position is difficult to visualize.
- Rescan after few minutes for change of fetal position and try and delineate with a coronal approach of the cranium.
- Coronal and parasagittal (modified longitudinal planes) views are taken (Figs 33A and B).
- 3D ultrasound also clearly demonstrates the pinna (Figs 34A to D).

CLEFT LIP AND CLEFT PALATE

Definition
- Cleft lip and cleft palate are defects involving in the upper lip and in the upper jaw and palate.
- Most defects are lateral.
- This is the most common congenital facial anomaly.\(^ {13}\)
- Clefts are present in 0.15% of births (representing 7.5% of all anomalies).\(^ {14}\)

Ultrasound Appearance
- The cleft can be incomplete/complete, unilateral/bilateral, lateral/midline and symmetric/asymmetric (Figs 35A to 49).
- Clefts can involve:
  - Only the upper lip.
  - Upper lip and the anterior portion of maxilla.

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Fig. 33A and B: Parasagittal view showing the fetal ear
• Upper lip, the anterior portion of maxilla and the posterior portion of palate.
• Only the posterior portion of palate.

- Cases of isolated cleft palate with no cleft lip are very difficult to identify on routine 2D views and are usually picked up on 3-dimensional sonography.
- Cleft palate alone involves the soft palate commonly rather than the hard palate. Color Doppler identifies the flow of fluid in the oral and nasal cavities simultaneously.15,16
- The bilateral cleft lip and cleft palate variety are the easiest to be diagnosed as they are easily recognizable sonographically on all routine 2D views for the face.

• Unilateral cleft lip and cleft palate can be diagnosed by a coronal and transverse axial view.17
• Median cleft lip is also seen by the modified coronal view.

What else to Check?
• Can be associated with chromosomal abnormalities like trisomies 13, 18, 21 and triploidy so look for other sonographic stigmata of these abnormalities.18
• The severe cleft chances of chromosomal abnormalities increase.19
Fig. 36: 2D reconstruction of the cleft upper lip as shown on 3D in Figure 44. This helps the parents to understand better.

Fig. 37: Midline cleft seen in the upper lip in an end on view.

Fig. 38: Unilateral cleft in the upper lip involving the nose as well.

Fig. 39: Unilateral cleft lip.

Fig. 40: Bilateral cleft lip.

Fig. 41: Bilateral cleft lip with inner extension seen on transverse view.

Fig. 42: Bilateral cleft lip.

Fig. 43: Bilateral cleft lip as seen on 3D reconstruction with all three views and 3D mode.
Fig. 44: 3D reconstruction of the bilateral cleft lip as seen from the side

Fig. 45: Bilateral cleft lip and palate

Fig. 46: End on view showing the cleft lip and palate

Fig. 47: Bilateral cleft lip as seen on the profile view to show upward extension and end on view to show inner extension

Fig. 48: Bilateral cleft lip as seen on 3D reconstruction

Fig. 49: Bilateral cleft lip on 3D

Fig. 50: Amniotic bands (arrow) can be associated with a cleft lip or palate

- Can also be associated with anencephaly, holoprosencephaly and amniotic band syndrome (Fig. 50).
- The prognosis depends on the severity of the cleft and associated malformations.

Prognosis
- If the finding is isolated, the prognosis is very good.
- It depends on the accompanying anomalies.
- Midline defects have a poor prognosis.

Be Careful
1. The facial profile view, however adequately visualized, does not confirm normal anatomy of the lips, palate and nostrils.
2. Comment on normal anatomy only with favorable fetal position otherwise rescan afterwards.
3. Do a detailed search for anomalies whenever you detect a cleft.
4. Latest technologies assist us with software and sections for visualizing not only the hard palate but also the soft palate as well. Accurate visualization of the soft palate requires an excellent initially acquired volume, fluid between the fetal tongue and palate, and curving of the plane to follow the structure of the palate. The oblique-face or flipped-face views make it possible to visualize the soft palate well in selected cases. 20

**HYPOTELORISM**

**Ultrasound Appearance**

The interocular distance and binocular distance (Fig. 51) are below 2 standard deviations of the mean (Fig. 52). 21

**What else to Check?**

- Most commonly associated with holoprosencephaly (Fig. 53).
- Associated facial abnormalities like cyclopia, ethmocephaly, cebocephaly (Fig. 54) and cleft lip and cleft palate should also be looked for. 22-24

**Fig. 51:** Fetal orbits seen in the coronal view to assess for hypo/hypertelorism

**Fig. 52:** Hypotelorism seen in a case of semilobar holoprosencephaly. The ocular diameter in this case was 12 mm, the interocular distance was 8 mm and the binocular distance was 29 mm

**Prognosis**

- Also associated with trisomies 13 and 21 so look in for associated abnormalities of these syndromes.
- Prognosis depends on the severity of holoprosencephaly, which is most commonly associated with trisomy 13. 25

**HYPERTELORISM**

**Ultrasound Appearance**

The interocular distance and binocular distance (Fig. 51) are above 2 standard deviations from the mean. 21

**What else to Check?**

- Usually associated with anterior (frontonasal) cephaloceles.
- Also associated with median cleft face syndrome and triploidy. 26

**Prognosis**

Prognosis depends on the size of cephalocele and other associated malformations.

**MICROGNATHIA**

**Ultrasound Appearance**

- Seen subjectively as a small mandible with a receding chin with the lower lip strikingly posterior to the upper lip (Fig. 55). 27
Midline sagittal view or profile view (Fig. 56) is most helpful to evaluate the size of mandible in respect to the face. Charts for measurements of the mandible are available for objective evaluation of micrognathia. The use of inferior facial angle and mandible width/maxilla width ratio helps sonographic recognition and characterization of fetal micrognathic mandibles in utero.

What else to Check?
- Often accompanied with congenital heart defects and skeletal dysplasias.
- Frequently associated with trisomies 18 and 13.
- Results in polyhydramnios because of difficulty in swallowing.
- At birth, micrognathia results in respiratory distress and difficult intubation.

Prognosis
- Depends on associated anomalies.
- Poor prognosis on antenatal detection.

MICROPHTHALMIA

Definition
- It is seen as a small globe.
- This is confirmed by orbital measurements.

Ultrasound Appearance
- Can be detected in the early second trimester when orbital measurements are taken (Figs 57 and 58).
- Can be unilateral or bilateral.

What else to Check?
- Look for sonographic stigmata of chromosomal abnormalities.
- Look especially for other facial and extremity malformations.

Prognosis
- It varies with etiology and associated malformations.

REFERENCES


