Approach and Goals

- Basic approach: speak the plastic surgeon’s language
- Immediate goal: accurate diagnosis and classification of craniofacial anomalies
- Ultimate goal: improved parental counseling and patient outcome

Overview

- The normal face
- Cleft lip and palate
- Abnormal profile
- Micrognathia
- Abnormal head shape
- Ear anomalies

The Normal Face

Anatomy of the Lip

- Vermillion border
- White roll
Anatomy of the Nose

- Tip
- Nostril
- Alar base
- Philtrum

Anatomy of the Ears

- Top of helix should be at level of inner canthal line

The Normal Profile

- Forehead and chin on same plane
- Nasal bone should be present
- Nose should project beyond plane of forehead and chin
- Top of ear at level of orbit

Abnormal head shape

- Brachycephaly
- Dolichocephaly
- Turibrachycephaly
- Microcephaly
- Macrocephaly
- Trigonocephaly
Abnormal head shape: etiologies

• Craniosynostosis
• Microcephaly/Macrocephaly
• Open neural tube defect
• Hemifacial microsomia
• Local deformation (oligo, fibroids)
• Syndromes

Cleft lip and palate

Cleft lip

Description of Cleft Lip

• Sidedness: unilateral, bilateral
• Symmetry: symmetric, asymmetric
• Extent: complete, incomplete
• Severity: mild, moderate, severe

Subtypes of Cleft Lip

• Mini-microform
• Microform
• Minor-form
• Incomplete
• Complete
Examples of minor form cleft lip

• Cleft lip and palate often go undetected on screening and targeted fetal sonography
• Detection rate low for cleft lip, lower for cleft palate
• It is important to know if the palate is cleft
  • accurate prenatal counseling of parents
  • to prepare parents for postnatal repair
  • if the palate is not cleft, the child will not have abnormal speech, recurrent ear infections or diminished midfacial growth

Problem

2D and 3D US of Cleft Palate

• Ultrasound accuracy in detecting cleft palate in the setting of cleft lip remains low, despite best 2D and 3D imaging
• Sensitivity 33-63%
• Specificity 84-95%


Cleft Palate: Limitations of US

• Why is accuracy of US so low?
• Unfavorable fetal position
• Shadowing by adjacent bony structures


Cleft Palate: Fetal MRI

• Preliminary work suggests sensitivity and specificity of fetal MR for detection of cleft palate is high (>90%)

Estroff JA, et al. IFMS 2011: Accuracy of Fetal MR in detection of cleft secondary palate in the setting of cleft lip

Cleft Lip and Palate: our approach

• Ultrasound for lip and alveolus=primary palate
• MRI for secondary palate, both hard and soft
Primary and Secondary Palate

• Primary palate= lip and alveolus; all structures anterior to incisive foramen

• Secondary palate= all structures posterior to incisive foramen. Includes part of the hard palate and all of the soft palate

Types of Cleft Palate: Veau Classification

The “Y Diagram”

Sonography

• Lips
• Nostrils
• Vomer
• Maxilla and Mandible
• Orbits
• Profile
• Position of tongue


MR

- Midline sagittal: primary bony palate and secondary soft palate; fetal profile

- Coronal: Vomer intersecting bony palate; inner and outer orbital distances

- Axial: Tooth-bearing alveolus

Associated Signs of Cleft Palate in Presence of Cleft Lip

<table>
<thead>
<tr>
<th>Axial/Coronal views</th>
<th>Sagittal view</th>
</tr>
</thead>
<tbody>
<tr>
<td>Lips – cleft</td>
<td>Profile – midface retrusion</td>
</tr>
<tr>
<td>Nares – flattened or deformed</td>
<td>Position of tongue – high</td>
</tr>
<tr>
<td>Vomer – deviated away from side of cleft; often midline if bilateral cleft lip and palate</td>
<td></td>
</tr>
<tr>
<td>Maxilla – interrupted alveolus, wide gap</td>
<td></td>
</tr>
<tr>
<td>Orbits – hypertelorism</td>
<td></td>
</tr>
</tbody>
</table>

Sonographic Protocol for Evaluation of Face and Neck: 2D & 3D

1. Skull and Brain
   - Axial, Coronal, and Sagittal planes
     - Skull shape (normal, dysmorphology, teatrocephaly; unusual)
     - Sinuses (open, air filled)
     - Normal indenting bone
     - Measurements: Normal brain size compared to long bone and abdominal biometry for gestational age
     - IPO/OFV: vs. GA, AD, FL, HL
     - Sign normal head, normal, thickened, micro

2. Face
   - Axial, Coronal, and Sagittal planes
     - Maxilla: normal bony anatomy
     - Nares: symmetric, not flattened or cleft
     - Vomer: symmetric
     - OD/OD: 2.0, orbit size, symmetry: 2.0 with normal size globes and normal interocular distance
     - Mandible: normal or severe cleft alveolus
     - Mandible: normal or severe cleft alveolus
     - Tongue position, size
     - Nasal – nose shape positions
     - Footprint normal, cleft, hypoplasia
     - Mandible normal, retrotrochoid
     - Chin: normal vs. small (macroglossia), retracted (prognathism) or not

3. Neck
   - Axial, Coronal, and Sagittal planes
     - Head position
     - Length of spine, curvature
     - Length of skull, skull thickening
     - Length of vertebral, humerus, and spine processes
     - Presence of branchial, tracheal, and parathyroid
     - Evaluate vallecula and pyriform sinuses, glottis, subglottic, and supraglottic regions
     - Cervical spine

MR

- Midline sagittal: primary bony palate and secondary soft palate; fetal profile

- Coronal: Vomer intersecting bony palate; inner and outer orbital distances

- Axial: Tooth-bearing alveolus
Always evaluate:
- Corpus callosum
- Cerebellar vermis
- Rest of fetal body for associated anomalies

Importance of accurate classification
- If the secondary palate is NOT cleft, the child will NOT have hearing, speech and feeding difficulties
- If the secondary palate IS cleft, all of these problems are expected AND the surgery is more extensive

Abnormal fetal profile
- Sloping forehead
- Bulging forehead
- Midface hypoplasia
- Micrognathia
- Retrognathia
- Arhinia
- Agnathia

Cleft examples
Unilateral complete cleft lip and palate
Bilateral cleft lip and palate

- Vomer usually midline
- 2 tooth buds in intermaxillary segment
- Hypertelorism
Twins: Referred at 35 weeks for a facial abnormality in one twin.

Final diagnosis: Apert Syndrome

Atypical clefts

DOL 2: bilateral coronal craniosynostosis
Tessier Cleft


Tessier I4 facial cleft

- Frontal encephalocele
- Cleft palate
- Hypertelorism
- Airway obstruction

Abnormal Fetal Profile

- Sloping forehead
- Midface hypoplasia
- Absent nose
- Absent mouth
- Micrognathia
- Protuberant tongue
- Masses; hydrops

Sloping forehead
**Wolf-Hirschhorn Syndrome (4p-)**

- Broad bridge of the nose continuing to forehead
- Microcephaly
- High forehead with prominent glabella
- Ocular hypertelorism, epicanthus, highly arched eyebrows, short philtrum, downturned mouth
- Micrognathia
- Poorly formed ears with pits/tags
- IUGR, hypotonia

**History:**

- 26 week fetus with sloping forehead

---

**Mouth and jaw anomalies**

- Micrognathia: small mandible
- Agnathia: absence of mandible
- Otocephaly: hypoplasia or absence of mandible
- Microstomia or aglossia: small or absent mouth
- Macrostomia: large or wide mouth
- Macroglossia: enlarged tongue

---

**Micrognathia**

- Micrognathia: small mandible
- Retrognathia: posteriorly displaced receding chin
- Glossoptosis: abnormal posterior position of tongue
Robin Sequence: Triad

- Cleft palate
- Micrognathia
- Glossoptosis

Robin Sequence: Etiology

- Primary abnormality: ? small mouth
- Tongue falls back= glossoptosis
- Inhibits fusion of palate leading to cleft

Robin Sequence: Prenatal Diagnosis

- Polyhydramnios
- Micrognathia
- High arched cleft palate

Robin Sequence: Prognosis and Management

- Concern for upper airway obstruction
- Neonatal respiratory distress
- Feeding problems
- If survive infancy, jaw variably grows, and child often does well
- Autosomal recessive recurrence risk
- Differential dx includes: T13, T18
Syndromes Associated with Micrognathia

- Etiology: monogenic, chromosomal, teratogenic, disruptive
- Nager syndrome
- Stickler Syndrome
- CHARGE syndrome
- Miller-Dieker syndrome
- Goldenhar syndrome
- Treacher-Collins syndrome
- Etc


Craniosynostosis

- Premature fusion of one or more cranial sutures
- Abnormal calvarial shape
- Exorbitism/exophthalmos
- FGFR (fibroblast growth factor related)
- Often syndromic (Apert, Crouzon, Pfeiffer)

Referral History

- 21 weeks: scoliosis and possible meningocele

21w6d
Summary of imaging findings at 21 weeks, 6 days

- Brachycephaly
- Hypertelorism
- Exorbitism
- Midface hypoplasia
- Broad short mandible and maxilla
- Macroglossia
- Scoliosis
- "Tail"

Outcome: demise at 27 weeks
Presumed Pfeiffer syndrome

Nose anomalies

- Arhinia: absent nose
- Midface hypoplasia
- Nasal glioma
- Encephalocele
- Accessory nostril

Arhinia: absent nose
Agnathia: congenital absence of the lower jaw

Ear anomalies
- Anotia
- Microtia
- Low set ears
- Protruding ears
- Auricular tags
Goldenhar Syndrome: hemifacial microsomia

Midface hypoplasia, hypotelorism, lowset ears, ACC, clenched hands:

Profile looks normal, but no nasal bone: T21

History

- 25 w: Multiple anomalies:
  - agnathia, microstomia, pelvic kidney and polyhydramnios
- 29 2/7w: admitted for PPROM
- 29 3/7w: STAT C-section- found fully dilated and fetus in breech position

26w5d poly, micrognathia, microstomia, low large ears
Outcome

- Delivery of a 2 lb 10 oz infant
- Cyanotic, floppy, no heart rate
- Chest compressions
- Emergency tracheostomy
- During procedure infant became cyanotic and lost HR. CPR was initiated.
- Transillumination: right pneumothorax
- Unresponsive; died.

Face Masses

Presumed facial hemangioma

Thank you for your attention.