Assessment of the Fetal Face and Neck

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Objectives

- Illustrate abnormalities of head shape
- Discuss facial abnormalities
  - Isolated malformations
  - Chromosomal anomalies
  - Genetic and non-genetic syndromes
  - Environmental insults
  - CNS abnormalities
- Discuss neck masses
  - Cystic hygromas
  - Lesions that would interfere with airway at delivery

Skull Shape

- Macrocrania
  - Tumors or hydrocephalus
- Abnormal shape
  - Trisomy 18
  - NTD
  - Craniosynostosis
- Microcephaly
  - Sloping forehead
  - Overlapping sutures/collapsed skull - Zika

Macrocrania

- Tumors or Hydrocephalus
- If head size is too large, vaginal delivery may not be possible

Skull Shape

- Macrocrania – tumors or hydrocephalus

Strawberry Skull

- Trisomy 18
Lemon Skull

- NTD

Sloping Forehead of Microcephaly

- Microcephaly
- Sloping forehead
- Parieto-occipital step
- Torcular enlargement

Scalp mass

- Hemangioma
- Encephalocele
- Lymphangioma
- Teratoma
- Cystic hygroma

Hemangioma

- Important to determine that the mass is extracranial
Encephalocele

Facial Anomalies and Chromosomal Anomalies
- 146 fetuses with facial defects
  - Micrognathia 37/56 (66%)
  - Macroglossia 10/13 (77%)
  - Cleft lip/palate 31/64 (48%)
  - Hypotelorism or cyclopia 5/11 (45%)
  - Nasal hypoplasia/proboscis 6/19 (32%)

Nicolaides Fetal Diagn Ther 1993;8:9

Protruding Tongue
- Think: Beckwith Weidemann
  - Macrosomia
  - Omphalocele
  - Macroglossia
  - Childhood tumors
  - Neonatal hypoglycemia

Assessment of the Lips and Palate
- Coronal View
  - Lips
  - Nose
  - Nostrils

Coronal view
- Orbits/eyelids

Midline sagittal view
- Profile
  - Nose
  - Upper lip
  - Lower lip
  - Chin
  - MR with soft palate
Retrognathia

- The line orthogonal to the vertical part of the forehead at the level of the synostosis of the nasal bones and the line joining the tip of the mentum and the anterior part of the most protruding lip is less than 50 degrees on a sagittal midline view.


Micrognathia

- Micrognathia is when the mandible width/maxilla width ratio (obtained at the alveolar level 1 cm behind the anterior osseous border) is less than 0.8.


Micrognathia

- Treacher Collins
- Goldenhar
- Roberts
- Chromosomal (Trisomy 13, 18)
- Orofacial digital

Pierre Robin Sequence

- Micrognathia
- Cleft plate with high arch palate
- Glossoptosis (tongue falls back)
- Polyhydramnios
Agnathia Microstomia Syndrome (Otocephaly)

3D Normal Faces

Images courtesy of Dolores Pretorius

Toothbuds

- Axial images of mandible to assess tooth buds
- Smooth continuous arc
- 10 symmetric toothbuds
- 4 anterior toothbuds should be seen after 19 weeks gestation

Ulm Prenat Diagn 1995:15:368-372

Orbitals Measurements

- Symmetric
- Normative data for measurements
  - Ocular diameter
  - Interocular diameter
  - Binocular diameter

Holoprosencephaly

- The face predicts the brain*
  - Normal
  - Cyclopia – single eye blind ending proboscis above the median eye
  - Ethmocephaly (extreme hypotelorism, arrhinia, blind-ended probosis)
  - Cebocephaly – hypotelorism, single-nostril nose
  - Median cleft lip and palate (with hypotelorism)
  - Bilateral cleft lip and palate

DeMeyer Pediatrics 1964:34:256-263
Holoprosencephaly

Images courtesy of Dolores Pretorius

Hypotelorism and Hypotelorism

Orbital abnormalities

- Hypotelorism
- Hypertelorism
- Anophthalmia
- Microphthalmia
- Cataracts
- Masses

Hypertelorism

- Abnormally wide-set eyes
  - Isolated abnormality
  - Part of numerous syndromes
    - Craniosynostosis
    - Apert
    - Crouzon
    - Pfeiffer
    - Median facial cleft
    - Agenesis of the corpus callosum
  - Mechanical disruption, anterior encephalocele
  - Chromosomal anomalies

Hypotelorism

- Decrease in the normal interocular distance, typically below the 5th percentile for gestational age
  - Holoprosencephaly
  - Chromosomal
  - Microcephaly
  - PKU
  - Meckel-Gruber
  - Cleft lip/palate
  - Crouzon

Microphthalmia/Anophthalmia

- Small or absent orbit
  - Chromosomal
    - Trisomy 13, 18, 9
  - Syndromes
    - Goldenhar-Gorlin syndrome (hemifacial microsomia)
    - Lenz syndrome (X-linked microphthalmia)
Midface Retrusion/Hypoplasia
- Chromosomal
- Syndromes
  - Median facial cleft
  - Teratogen exposure

Midface Hypoplasia

Cleft Lip and Palate
- CL/P distinct from isolated CP
- 0.15% of births (7.5% of all anomalies)
  - 50% CL/P
  - 20% CL
  - 30% CP

Patient took Tegretol during early pregnancy

Moore, the Developing Human, W.B. Saunders
Cleft Lip and Plate

- 1.5/1000 live births
- Higher in utero
  - American Indians 1/300
  - Asians 1/600
  - Blacks 1/2500

Isolated Cleft Palate

- No ethnic differences

Classification Cleft Lip/Palate

- 1 – unilateral lip
- 2 – unilateral lip/palate
- 3 – bilateral lip/palate
- 4 – median
- 5 – random – amniotic band
Cleft Alveolus

Cleft Lip and Alveolus

Cleft Lip and Palate

Cleft Lip – slightly lateral to midline
+ Cleft adjacent alveolar ridge

Bilateral Cleft

Cleft Lip and Palate

Images courtesy of Dolores Pretorius
Bilateral CL/P
Premaxillary Protuberance

Trisomy 13

Median Cleft
Holoprosencephaly

Median cleft with holoprosencephaly

Cleft Lip and Palate

Image courtesy of John Mulliken, M.D.
### Teratogens for CL/P
- Rubella
- Thalidomide
- Retinoic acid
- Valproic acid
- Phenytoin
- Hydantoin

### Syndromes for CL/P
- Too many to list
- CP – 46.7% associated malformation
- CL/P – 36.8%
- CL – 3.6%
  - Stoll, Craniofac J, 2000;37:41-7

### Ears
- Normal ear size and length
  - Small in Trisomy 21
- Low-set ears
  - Syndromes
- Preauricular tags
  - Syndromes
- Abnormal lobulations
  - Syndromes

### Syndromes Associated with Abnormal Ears
- Chromosomal abnormalities
- Pharyngeal Arch syndromes
- Holoprosencephaly
- Anencephaly
- Crouzon's dysostosis
- Treacher Collins syndrome
- CHARGE association
- VACTERL association

### The Neck
- Nuchal cord
- Cystic hygroma
- Goiter
- Tumors
Important definitions for Neck Soft Tissues

- Cystic hygroma – discrete fluid, loculations
- Nuchal thickening – no discrete fluid
- Nuchal translucency - 10-14 weeks
- Lymphangioma – unilateral

Nuchal Translucency

11-14 weeks Sliding scale for abnormal thickness with respect to CRL

Nuchal Thickening

- ≥ 6 mm is abnormal
- Suboccipital bregmatic plane
  - Cavum septi pellucidi
  - Cerebral peduncles
  - Cerebellar hemispheres
- 40% sensitive for Trisomy 21
- Even if it resolves, it still carries a risk of chromosomal abnormality

Cystic Hygroma

- Anomalies of lymphatic system
- Single or multiple cysts within the soft tissue
- Usually in the neck
- Most common neck mass in utero

1/200 SAB at CRL >30 mm
1/1000 live births
- Bryne Hum Pathol 1984 15:61-67
Large Cystic Hygroma
- 40-50% chromosomal abnormalities
- Especially Turner syndrome

Turner Syndrome

Lymphangioma
**Associated Anomalies in Early Second Trimester**

<table>
<thead>
<tr>
<th>Cystic Hygroma</th>
<th>Non-septated mass</th>
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<tbody>
<tr>
<td>Persist</td>
<td>56%</td>
</tr>
<tr>
<td>Aneuploidy</td>
<td>72%</td>
</tr>
<tr>
<td>Hydrops</td>
<td>50%</td>
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<tr>
<td>Other anomalies</td>
<td>52%</td>
</tr>
<tr>
<td>Pregnancy loss</td>
<td>88%</td>
</tr>
<tr>
<td>Non-septated</td>
<td>2%</td>
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<tr>
<td>Mass</td>
<td>5.6%</td>
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<tr>
<td></td>
<td>1.7%</td>
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<tr>
<td></td>
<td>15%</td>
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<tr>
<td></td>
<td>6%</td>
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Bronshitein, Obstet Gynecol 993;81:683-687

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**Goiter**

- Congenital hypothyroidism 1/3700 births
- Maternal Graves disease leads to abnormal fetal thyroid function ±12%
  - Transplacental transfer of maternal thyroid stimulating autoantibodies in Graves
- **DRUGS**
  - PTU for Graves disease
  - Carbimazole for Graves disease
  - Amiodarone
  - Lithium
  - 131I
- Fetal goiter
  - Solid
  - Hypoechogenic
  - + polyhydramnios with hyperextended neck

- Solid neck mass in region of thyroid
- Hyperextension of neck
- Polyhydramnios
Epignathus
- Oral teratoma
- Nasopharyngeal teratoma
- Facial teratoma
- 1/35,000 live births
- Can grow intracranially
- Polyhydramnios

Cervical Teratoma
- Anterior neck
- Solid of mixed cystic/solid
- May have calcifications
- Hyperextension of neck if large
- Polyhydramnios
- Preterm delivery

Oropharyngeal teratoma 28 weeks
Thyroid Teratoma

- Ex utero intrapartum treatment
- Fetus partially delivered, maintained on placental support until an airway is established

EXIT

Oropharyngeal Teratoma 17 weeks

Oropharyngeal Teratoma 28 weeks
Summary

- The face predicts the brain…and many other syndromes and chromosomal abnormalities
- Neck masses need careful assessment, at times with MRI, to appropriately triage for airway management at delivery