The Normal Heart

Atrioventricular Septal Defect

The Normal Abdomen

Duodenal Atresia

Ventral Wall Defects

Gastroschisis
Ventral Wall Defects

Omphalocele

11 week fetus

Ventral Wall Defects

- Gastrochisis
  - 1 in 5000 live births
  - Paraumbilical defect involving all layers of the abdominal wall
  - Associated GI tract abnormalities
  - 10% other fetal anomalies

- Omphalocele
  - Extrusion of the abdominal contents into the base of the umbilical cord
  - Associated anomalies - 50 to 80 percent
  - Associated chromosomal abnormalities – 40 60%
  - Beckwith-Wiedeman

Holoprosencephaly

- Trisomy13
Trisomy 13
- Cardiac defects
- CNS abnormalities
- Cystic hygroma
- Facial abnormalities - cleft lip and palate
- Echogenic kidneys (polycystic)
- Intrauterine growth restriction
- Holoprosencephaly
- Microcephaly
- Neural tube defects
- Ocular abnormalities
- Omphalocele
- Polydactyly
- Rocky bottom feet
- Minor markers
  - Echogenic intracardiac focus
  - Mild ventriculomegaly
  - Pyelectasis
  - Single umbilical artery

Clover Leaf Skull
- Skeletal Dysplasia
- Thanatophoric dwarfism
- Craniosynostosis

Strawberry Shaped Skull

Specific Minor Markers
- Thick Nuchal Fold
  - First sonographic marker associated with DS
  - Measured 15 - 20 wks
  - >/= 6 mm
  - Identifies 40% of fetuses with DS
  - Single most specific and sensitive marker for DS
  - Confers highest risk of aneuploidy

Specific Minor Markers
- Renal Pyelectasis
  - 0.3 to 4.5%
  - >/= 4 mm AP diameter
  - PPV 1 in 340
  - AP diameter varies over time
  - Association greatest when other anomalies seen
  - Clinical relevance may lie in post-delivery concerns

Specific Minor Markers
- Echogenic Bowel
  - 0.2 to 1.4% scans
  - Normal variant
  - Fetal aneuploidy
  - IUGR
  - Cystic fibrosis
  - Bleeding
  - Congenital infection

Specific Minor Markers

Specific Minor Markers

Specific Minor Markers

Specific Minor Markers
Specific Minor Markers

- **Echogenic Intracardiac Focus**
  - 4% of scans
  - Highest in Asians; lowest in Blacks
  - Calcifications of papillary muscle of unknown etiology
  - Can be single or multiple
  - Most are in the left ventricle
  - Original studies in high risk populations
  - In low risk population does not increase risk significantly
  - Carries lowest risk for aneuploidy

- **Calcifications of papillary muscle of unknown etiology**
  - Can be single or multiple
  - Most are in the left ventricle
  - Original studies in high risk populations
  - In low risk population does not increase risk significantly
  - Carries lowest risk for aneuploidy

Specific Minor Markers

- **Choroid Plexus Cyst**
  - 1% incidence
  - Can be single or multiple
  - Associated with trisomy 18 (Not trisomy 21)
  - High risk group – 1 in 128
  - Low risk group – 1 in 189
  - Recent studies show risk low when heart and hands appear normal >/= to 18 weeks

Specific Minor Markers

- **Nasal Bone**
  - Most recent addition
  - 1st trimester
    - 73% of fetuses with trisomy 21 vs 0.5% of normal fetuses
    - Unable to obtain in 6% of cases
  - 2nd trimester controversies
    - Absence vs Presence vs Hypoplastic
    - Nasal bone length ratios
    - Interpretation varies by ethnic group

ACOG Practice Bulletin
May 2016 Number 163

- Minor markers
  - Nonspecific findings more common in fetuses with Down syndrome than those without
    - Common in unaffected fetuses as well
    - Increased nuchal skinfold
    - Highest risk of aneuploidy
    - Isolated echogenic intracardiac focus
    - Lowest risk of fetal aneuploidy
    - Isolated presence of minor marker
      - Analyte screening or cell free DNA should be offered
    - Major limitations of 2nd trimester ultrasonographic markers
      - Lack of standardization in characteristics that define a positive test result

ACOG Practice Bulletin
May 2016 Number 163

- **Level C Recommendations**
  - Isolated minor marker
    - Offer aneuploidy screening if not yet completed
  - Positive result from traditional screening
    - Cell-free DNA versus definitive testing
      - May delay definitive diagnosis/fail to identify aneuploidy
  - Parallel/simultaneous testing with multiple screening options
    - Not cost-effective and should not be performed
  - Multifetal gestations, fetal demise
    - Serum-based aneuploidy screening should be discouraged
    - Increased risk for inaccurate test result in these circumstances
Specific Minor Markers

- Other markers
  - Clubfoot
  - Single umbilical artery
  - Dangling of the choroid plexus with normal atrial dimension
  - Iliac angle widening
  - Hypoplasia middle phalanx 5th digit
  - Clinodactyly
  - Delayed fusion of chorion/amnion
  - Sandal-gap toe

Statistics Review

- Positive predictive value
  - Chance that a positive test will end up in an affected fetus
- Negative predictive value
  - Chance that a fetus will be normal and is not affected
- Likelihood ratio (positive)
  - How much more likely is a fetus with a specific finding to have a chromosomal abnormality than one without the finding
- Likelihood ratio (negative)
  - How much more likely is absence of a finding in a normal fetus vs an affected fetus

Statistics and Prenatal Diagnosis

- Sensitivity - true positives
  - How well the test identifies fetuses with aneuploidy
- Specificity - true negatives
  - How well the test identifies fetuses with nl chromosomes
- Positive Predictive Value
  - Chance that a positive test will result in an affected fetus
- Negative Predictive Value
  - Chance that a negative test will result in a normal fetus
- Likelihood Ratio (positive)
  - How much more likely a fetus with a specific finding will have a chromosomal abnormality than one without the finding
- Likelihood Ratio (negative)
  - How much more likely is absence of a finding in a normal fetus vs an affected fetus

Risk Modification (Example)

- Maternal age 35 years = 1/270 risk for trisomy 21
- 1st trimester screen places risk at 1 in 450
- Ultrasound shows bilateral pyelectasis
  - Likelihood ratio of 1.6
- 1/450 x 1.6 = 1 in 281
  - Still a low risk result

Likelihood Ratios-Trisomy 21

<table>
<thead>
<tr>
<th>Finding</th>
<th>Likelihood ratio</th>
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<tbody>
<tr>
<td>Structural Defect</td>
<td>25</td>
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<tr>
<td>Nuchal Thickening</td>
<td>18.6</td>
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<tr>
<td>Echogenic Bowel</td>
<td>5.5</td>
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<tr>
<td>Short Humerus</td>
<td>2.5</td>
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<tr>
<td>Short Femur</td>
<td>2.2</td>
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<tr>
<td>Echogenic Intracardiac Focus</td>
<td>2</td>
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<tr>
<td>Renal Pyelectasis</td>
<td>1.6</td>
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<tr>
<td>Normal Ultrasound</td>
<td>0.4</td>
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Minor Marker Follow-up

- Third trimester follow-up
  - Renal pyelectasis
  - Echogenic bowel
  - Short femur and/or humerus
- No follow-up necessary
  - Choroid plexus cyst
  - Echogenic bowel
- Detailed counseling
  - Hypoplastic or absent nasal bone
  - Echogenic bowel
  - Thickened nuchal skinfold