5. FETAL DIAGNOSIS AND IMAGING: ULTRASOUND

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ULTRASONOGRAPHY

1. Definition of ultrasound
   a. Frequency
   b. Resolution vs. penetration

2. Historical perspective

3. How ultrasound works
   a. The components of the machine
      i. Transducer
         1. sector, linear, curvilinear
      ii. Display
      iii. Signal transduction and information processing
   b. Generating an ultrasound “ping”: the piezoelectric effect
      i. Physics
      ii. Steering the beam
   c. Biomechanical effects and safety
      i. Tissue effects
      ii. Measuring sound intensity

4. Indications for ultrasound in pregnancy:
   a. NICHD and ACOG vs. the rest of the world
      i. NICHD indications: 28 of them!
      ii. Cost-benefit
      iii. The real world: 70% of pregnant women in the U.S. get scanned
b. The RADIUS trial
   i. Randomized trial of automatic ultrasound in pregnancy vs ultrasound only if indicated.

   ii. Results

   iii. Lessons learned
       1. training, training, training!

5. What do we look for: Ultrasonogram content
   a. First trimester ultrasound
      i. Number, location and viability

      ii. adnexae

   b. Second and third trimester
      i. Number, location, viability

      ii. Placental location

      iii. Fluid volume

      iv. Anatomic examination

   c. Structure vs. function

6. How Sensitive and How Accurate is Ultrasound at Detecting Structural Fetal Anomalies? Training, training, training redux

INVASIVE DIAGNOSTIC TECHNIQUES
1. Amniocentesis
   a. Purpose: screening vs diagnostic testing

   b. Indications
      i. Abnormal screening tests, advanced maternal age, family history of heritable disease, abnormal anatomy

   c. Technique
      i. Timing: 15-16 weeks
         1. risks are timing-associated

      ii. guidance: ultrasound

      iii. Equipment / tools
d. What do we get from it?
   i. Amniocytes
      1. genetic testing
      2. genotyping
   
   ii. Amniotic fluid
      1. AFP, enzyme

  e. Limitations / Risks
     i. Time limitations

2. Chorionic Villus Sampling
   a. Purpose: purely diagnostic testing
   
   b. Indications
      i. Abnormal screening tests, advanced maternal age, family history of heritable disease,
   
   c. Technique
      i. Timing: 10-12 weeks
      
      ii. Transvaginal vs transabdominal approach
      
      iii. Guidance: ultrasound
      
      iv. Equipment / tools
   
   d. What do we get from it?
      i. Chorionic villi
   
   e. Limitations / Risks
      i. Limb reduction defects
      
      ii. Placental mosaicism

3. Percutaneous Umbilical Blood Sampling
   a. Purpose: determine fetal acid-base status, Hgb concentration, karyotype
   
   b. Indications: non-immune hydrops fetalis; increased risk for significant fetal anemia
   
   c. Technique
      i. Timing
ii. Guidance: ultrasound

iii. Equipment / tools

d. What do we get from it?
   i. Fetal red cells, lymphocytes, serum

e. Limitations / Risks
   i. Risks associated with accessibility of cord insertion and underlying fetal condition.

**CAN NON-INVASIVE TESTING REPLACE OR SUPPLEMENT INVASIVE TESTING?**

1. Ultrasound to refine the risk for aneuploidy
   a. First trimester: Nuchal Translucency

   b. Second trimester: the Genetic Sonogram

   c. Screening vs. diagnosis

2. Fetal Cells and Fetal DNA in Maternal Circulation