

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