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-INVASIVETESTING 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