Ultrasound Screening for Congenital Anomalies

Amelia Sutton MD, PhD
Maternal Fetal Medicine
University of Alabama at Birmingham

Disclosures

- None

Objectives

- Review epidemiology of congenital anomalies and purposes of screening
- Discuss the components of first and second trimester ultrasounds
- Explore the role of ultrasound in the era of cell-free DNA screening
Epidemiology of Congenital Anomalies

• The incidence of is 2-4%
• Most are sporadic or multifactorial
• Aneuploidy affects 1/750 livebirths

Most Common Types of Defect

• Congenital heart defects
  – 0.5-0.8% of live births
  – VSD is the most common

Most Common Types of Defects

• Neural tube defects
  – 2/1000 live births
• Cleft lip/palate
  – 1/750 live births
Sensitivity of Ultrasound

- Detection rates of 16-44% for all anomalies prior to 24 weeks
- Overall detection rate for lethal anomalies up to 84%

Screening for Congenital Malformations in Low Risk Patients

- RADIUS Trial—AJOG, 1994, 171:392
  - 2.3% major anomaly rate
  - 35% detection of major anomalies
  - No change in perinatal outcome

Recommendations

- At least one ultrasound should be offered to all women between 18-20 weeks
- Endorsed by ACOG, SMFM, AIUM, RCOG, SOGC, ACR, SPR, SRU...
Purposes of Screening

• Pregnancy management options
• Antenatal interventions
• Delivery planning
• Postnatal interventions
• *Family preparation*

In addition to above:

– Assess the role of heredity and the recurrence risk for specific disorders
– Make options for dealing with recurrence risk available to at risk individuals
– Enhance reproductive options for at risk individuals

Indications for first trimester sonography

• Confirmation of IUP
• Evaluation of suspected ectopic pregnancy
• Evaluation of bleeding or pain
• Evaluation of uterine abnormalities of pelvic masses
• Diagnosis of multiple gestations
• Aneuploidy screening or CVS
• Assessing for fetal anomalies in high-risk patients
Standard first trimester ultrasound: essential components

- Uterus, adnexa, cul-de-sac
- Gestational sac(s)
- Yolk sac
- Fetal pole with CRL
- Fetal heart rate
  - M-mode

Indications for second trimester sonography

- Estimation of gestational age
- Screening for fetal anomalies
- Evaluation of fetal growth
- Evaluation of bleeding
- Assessment of fetal presentation
- Assessment of amniotic fluid volume
- Amniocentesis or other procedure

Selected components of standard second trimester sonography

- Maternal anatomy
- Placenta
- Placental cord insertion
- Amniotic fluid volume
- Fetal heart rate
- Biometry
- Multiple gestation
Selected components of standard second trimester sonography

- Head, face, and neck:
  - Lateral cerebral ventricles
  - Choroid plexus
  - Midline falx
  - Cavum septi pellucidi
  - Cerebellum
  - Cisterna magna
  - Upper lip
  - Nuchal fold

Selected components of standard second trimester sonography

- Chest
  - Four chamber heart
  - Left ventricular outflow tract
  - Right ventricular outflow tract

Selected components of standard second trimester sonography

- Abdomen
  - Stomach
    - Presence, size, location and situs
  - Kidneys
  - Bladder
  - Cord insertion site
  - Cord vessel number
Selected components of standard second trimester sonography
- Spine
  - Cervical, thoracic, lumbar, sacral
- Extremities
- Gender
  - In multiples and when “medically-indicated”

Selected indications for targeted sonography
- Suspected anomaly
- Abnormal aneuploidy screen
- Advanced maternal age*
  - If desires option for diagnostic testing
- Family history (1st degree) of congenital abnormality
- Family history of birth defect

Selected indications for targeted sonography
- Pre-gestational DM
- Teratogenic exposure
- TORCH infection
- Monochorionic twins
- Growth restriction
- Oligo- or polyhydramnios
- Fetal arrhythmia
Selected indications for fetal echocardiography

- Suspected cardiac anomaly
- Pre-gestational diabetes
- First-degree relative with congenital cardiac anomaly
- Monochorionic gestation
- Teratogen exposure
- Maternal PKU

Selected indications for fetal echocardiography

- Fetal arrhythmia
- Fetal aneuploidy
- Hydrops
- Increased nuchal translucency
- Early-onset growth restriction

Soft Markers

<table>
<thead>
<tr>
<th>Marker</th>
<th>LR for T21</th>
</tr>
</thead>
<tbody>
<tr>
<td>Nuchal fold</td>
<td>9.8</td>
</tr>
<tr>
<td>Short humerus</td>
<td>4.1</td>
</tr>
<tr>
<td>Echogenic bowel</td>
<td>3.0</td>
</tr>
<tr>
<td>Echogenic focus</td>
<td>1.1</td>
</tr>
<tr>
<td>Pelvic fistula</td>
<td>1.0</td>
</tr>
<tr>
<td>Choroid plexus cysts</td>
<td>7.1 (for T18)</td>
</tr>
</tbody>
</table>
SMFM Guideline

• In patients with negative serum screening or negative cell-free DNA screening, an isolated soft marker (EIF or CPC) should be described as not clinically significant or a normal variant.

Norton et al 2017 AJOG

SMFM Guideline

• Do not report or “normal variant:”
  – EIF
  – CPC
  – Sandal toe gap
  – Clinodactyly

• Per routine:
  – Pelviectasis
  – Ventriculomegaly
  – Echogenic bowel
  – Thick nuchal fold
  – Single umbilical artery
  – Hypoplastic nasal bone
  – Short humerus/femur

Norton et al 2017 AJOG

SMFM Guideline

• Women with fetal anomalies should be offered diagnostic testing with chromosomal microarray.

Norton et al 2017 AJOG
Resources

- AIUM Practice parameter for the performance of obstetric ultrasound examinations. 2013
- Norton et al. The role of ultrasound in women who undergo cell-free DNA screening. 201