First Trimester Ultrasound
Diagnosis of Fetal Abnormalities:
What You Need to Know

Alfred Abuhamad, MD.
Eastern Virginia Medical School

My Prediction

• The first trimester ultrasound will play a major role in clinical management of pregnancy in the next 3-5 years
  • (up to 13 6/7 weeks)

Current Role of First Trimester Ultrasound

• Accurate pregnancy dating
• Early diagnosis of multiples
• Pregnancy risk assessment
• Evaluation of the adnexae

Estimation of Gestational Age

• If a patient reports no menstrual dates, ultrasound in the first or second trimester should date the pregnancy and establish the EDD
• Ultrasound dating of pregnancy is most accurate in the first trimester

Criteria for CRL Measurement

• Focal zone at appropriate level
• Image magnified
• Embryo/fetus imaged in neutral position
• Maximal length of embryo/fetus shown
• Maximal length of embryo/fetus measured in a straight line from cranial to caudal


Estimation of Gestational Age

• CRL are most accurate in fetal neutral position and between 11+0 and 13+6 weeks of gestation
• Use the mean of three discrete CRL measurements.
• Assign EDD by CRL if > 5 days discrepancy at less than 9 weeks of gestation
• Assign EDD by CRL if > 7 days discrepancy at 9 - 13 6/7 weeks of gestation
Evidence of early first-trimester growth restriction in pregnancies that subsequently end in miscarriage

**Multiple Pregnancy**

- **Chorionicity:**
  - 100% in 1st Trimester
  - 90% in 2nd and 3rd Trimesters

**Pregnancy Risk Assessment**

- Hemorrhagic Cyst
- Cystic Teratoma
- Pedunculated Leiomyoma
- Endometrioma

**Evaluation of the Adnexae**

- **Evolving Role of First Trimester Ultrasound**
  - women
  - Screening for fetal malformations

**Biometry in First Trimester**
The Role of Early Gestation Ultrasound in the Assessment of Fetal Anatomy in Maternal Obesity

Letty Romary MD, Elna Skidmore MD, PhD, Salerno AS MFR, Tina D. Cunningham PhD, Shannon Markello, Simon Hanco, Lindsey Steward, Las Perlas MD, Jennifer Phillips MD, Alfred Alhabash MD

Table 4. Completion Rates of Combined Morphologic Examinations by US.

<table>
<thead>
<tr>
<th>US (2-3 SD)</th>
<th>US (4-5 SD)</th>
<th>US (6+ SD)</th>
<th>Combined Morphologic Examinations (CM)</th>
</tr>
</thead>
<tbody>
<tr>
<td>92.0% (n = 120)</td>
<td>74.0% (n = 120)</td>
<td>56.0% (n = 120)</td>
<td>83.0% (n = 120)</td>
</tr>
</tbody>
</table>

Table 5. Completion Rates of Morphologic Examination by Hill Without Upper Lip.

<table>
<thead>
<tr>
<th>Overall</th>
<th>US (1-2 SD)</th>
<th>US (3-4 SD)</th>
<th>US (4-5 SD)</th>
<th>US (5-6 SD)</th>
<th>US (6+ SD)</th>
</tr>
</thead>
<tbody>
<tr>
<td>75.0% (n = 120)</td>
<td>74.0% (n = 120)</td>
<td>52.0% (n = 120)</td>
<td>56.0% (n = 120)</td>
<td>56.0% (n = 120)</td>
<td></td>
</tr>
</tbody>
</table>

Evolving Role of First Trimester Ultrasound

- malformations
- pregnancy termination

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### Factors Impacting Detection Rates

- Use of an anatomical protocol during first trimester anomaly screening ($P<0.0001$)

### Why the Midsagittal Plane?

- Same as the Nuchal Translucency (NT) plane
  - Standardized criteria for NT plane
  - Established mechanism for certification
  - Large number of physicians and sonographers already certified

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### Implementation of a National Nuchal Translucency Education and Quality Monitoring Program

- In range
- Out of range: low
- Out of range: high

Midsagittal Plane

![Midsagittal Plane Image]

Nasal Bone

- Abnormal if:
  - Absent
  - Short
  - Hypoplastic

Aneuploidy Screening

<table>
<thead>
<tr>
<th>NT Measurement</th>
<th>Rapid</th>
<th>Trimester 21</th>
<th>Trimester 22</th>
<th>Trimester 23</th>
</tr>
</thead>
<tbody>
<tr>
<td>NT thickness</td>
<td>5.0</td>
<td>2.0</td>
<td>0.3</td>
<td>0.3</td>
</tr>
<tr>
<td>Nuchal translucency</td>
<td>0.0</td>
<td>0.0</td>
<td>0.1</td>
<td>0.1</td>
</tr>
<tr>
<td>Ductus venosus</td>
<td>1.0</td>
<td>0.5</td>
<td>0.3</td>
<td>0.3</td>
</tr>
</tbody>
</table>

Nasal Bone – Trisomy 21

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Posterior Fossa – Axial View

13 weeks

Spina Bifida

Dandy Walker

Other CNS Anomalies

Ventriculomegaly
**Ventriculomegaly**

- Thinning of CP
- CP filling less than ½ of LV
- CP borders do not touch lateral ventricular wall
- Separation of CP from superior borders of LV
- Enlarged 3rd Ventricle

**Normal Ventrices**

**Ventriculomegaly**

**Anencephaly / Exencephaly**

**Axial Planes**

- Normal
- Holoprosencephaly
Cord Insertion

Physiologic Midgut Herniation

Omphalocele

Omphalocele (Pentalogy)
Gastroschisis

- Smooth Borders
- Omphalocele
- Irregular Borders
- Gastrochisis

Bladder

- Seen in 88% at 12 weeks
- Seen in 92–100% at 13 weeks
- Renal source of AF is > 16 weeks

Fetal Bladder

- Chromosomal anomalies in 24%
- With normal chromosomes:
  - Resolution in 90%
  - Renal anomalies in 10%

Megacystis (7-15 mm)

Ultrasound Obstet Gynecol 1996; 7: 95–100
Megacystis (> 15 mm)

- Chromosomal anomalies in 11%
- With normal chromosomes:
  - Renal anomalies in 100%

Fetal Gender

- Inaccurate before 12 weeks
- Accuracy increases with advancing gestational age
- Greater than 95% at > 13 weeks

Ductus Venosus

3D - Extremities

Aneuploidy Screening
Trisomy 21

Table 6.2 First Trimester Features of Trisomy 21

- Thickened nuchal translucency (NT)
- High human chorionic gonadotropin (HCG), ß-HCG
- Low pregnancy-associated plasma protein A (PAPP-A)
- Absent or hypoplastic nasal bone
- Reversal of flow in ductile or high impedance flow in ductus venous
- Tricuspid regurgitation
- Increased femoral-medial facial angle (FMF), short maxilla reflecting midface hypoplasia
- Abnormal right subclavian artery
- Ethrogic focus
- Ethrogic bowel
- Renal tract dilation
- Increased peak velocity in the hepatic artery
- Ductus venous directly draining into the inferior vena cava
- Structural anomalies such as atrioventricular septal defect, tetralogy of Fallot, and others

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Trisomy 18

Marked thickened nuchal translucency (NT), cystic hygroma, early hydronephrosis
High human chorionic gonadotropin (HCG), ß-HCG
Low pregnancy-associated plasma protein A (PAPP-A)
Normal nasal bone
Cardiac anomalies: tachycardia, left ventricular outflow obstruction, hypoplastic left heart syndrome, coarctation of the aorta, absent left subclavian artery, tricuspid regurgitation
Renal anomalies: horseshoe kidney, renal pelvis dilation
Ductus venous abnormalities: directly draining into inferior vena cava, reversal flow of A-wave or high impedance
Female gender

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Trisomy 13

Monosomy X

Table 6.4 First Trimester Features of Monosomy X

- Marked thickened nuchal translucency (NT), cystic hygroma, early hydronephrosis
- High human chorionic gonadotropin (HCG), ß-HCG
- Low pregnancy-associated plasma protein A (PAPP-A)
- Normal nasal bone
- Cardiac abnormalities: tachycardia, left ventricular outflow obstruction, hypoplastic left heart syndrome, coarctation of the aorta, absent left subclavian artery, tricuspid regurgitation
- Renal anomalies: horseshoe kidney, renal pelvis dilation
- Ductus venous abnormalities: directly draining into inferior vena cava, reversal flow of A-wave or high impedance
- Female gender

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Monosomy X

Triploidy

Table 6.5 First Trimester Features of Triploidy

- Nuchal translucency: Normal
- p53 (paternal) 0.18 Mmol
- p53 (maternal) 0.08 Mmol
- Growth: Short crown-rump length, severe growth restriction with diaphragmatic hernia
- Head: Proportional large head, dilated fourth ventricle, compressed posterior fossa as a clue for spine bifida, holoprosencephaly
- Heart: Cardiac anomalies, abnormal right subclavian artery, ethrogic focus, tricuspid regurgitation
- Abdomen: Polyhydramnios, single umbilical artery, absent gall bladder, ethrogic kidneys
- Limbs: Shortened femur, amelia, club feet, joint laxity
- Two ß-HCG, ß-human chorionic gonadotropin, PAPP-A, pregnancy-associated plasma protein A

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Triploidy

Midsagittal Plane in First Trimester

- Good screening modality for fetal abnormalities
- Rather than doing away with NT, we need to enhance training and focus on detection of fetal abnormalities
- The role of first trimester ultrasound will expand over the next years