FIRST TRIMESTER ULTRASOUND
WHAT CAN WE SEE TODAY?
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First Trimester Ultrasound

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LEcTure Objectives

By the end of this lecture the viewer should:

• Understand the reasons behind an 11-14 week anatomic assessment
• Understand how to carry out a systematic anatomic assessment at 11-14 weeks
• Understand the various types of anomalies amenable to detection at 11-14 weeks: what should be detected, what may be detected, and what cannot be detected
• Understand the limitations of fetal assessment at 11-14 weeks
• Understand the safety concerns at 11-14 weeks
• Understand future direction of fetal evaluation in the first trimester

OBJECTIVES

Background

Why Early?

Systemic Evaluation

Future Direction

Anomaly Detection

Limitations

PEARLS & CONCLUSIONS

• Affect 3-5% of all pregnancies
• Most common cause of infant mortality
• Suboptimal detection 16-77%

CONGENITAL MALFORMATIONS

Reem S. Abu-Rustum has no disclosures.

Abu-Rustum

Rosano et al. J Epidemiol Community Health 2000; 54:660

NTD at 12w6d

Majority have no risk factors

MUST SCREEN THE ENTIRE POPULATION

Why Early?

Background

Systemic Evaluation

Future Direction

Limitations

Anomaly Detection

Why DETECT EARLY?
MAIN CONSIDERATIONS

- Workup
  - Options
  - TDF Limitations
  - Safety
  - Explain sudden IUFD
  - Natural progression
  - Psychological
  - Obstetric care
  - Early Reassurance

Deaths / 100,000 abortions

Abortions in the USA 1988 - 1997

Bartlett et al 2004

NT

NIPT

Technical Advances

GLOBAL IMPLEMENTATION
AS A RESULT OF…

Today we have access to more than 70% of pregnancies who are undergoing NT assessment at 11-14 weeks

Pearls & Conclusions

Safety

Options

TOP Limitations

Workup

Explain sudden IUFD

Natural progression

Psychological

Obstetric care

Early Reassurance

Clinical Advances

3.6 per 1000

23.9 per 1000

2000 lb

Keep in Mind…

Reiff et al. Prenatal Diagnosis 2016; 36:260

Maiz et al. Prenatal Diagnosis 2016; 36:449 – 455

Barlett et al. Obstet Gynecol 2004; 103:729

Bartlett et al. Obstet Gynecol 2004; 103:729

Salveson et al. UOG 2011; 37:625

Today we have access to more than 70% of pregnancies who are undergoing NT assessment at 11-14 weeks

But…

Today we have access to more than 70% of pregnancies who are undergoing NT assessment at 11-14 weeks
... A SHIFT IN THE ROLE OF NT WITH NIPT…

IT IS NOW POSSIBLE TO RULE OUT

FIRST TRIMESTER DETECTION OF STRUCTURAL ANOMALIES

In the most ideal situations, FTS can detect
• Up to 82% of anomalies and
• 2/3 of cardiac lesions

The overall detection rate of early ultrasound was 591 of 996 (59%). With regard to location of fetal malformation, the highest detection rate was observed for neck anomalies (24/65 [36%]) followed by anomalies of the abdomen (39/149 [26%]), brain and spine (81/358 [23%]), heart (201/418 [48%]), limbs (36/105 [34%]), genitourinary tract (40/116 [34%]), and face (8/23 [34%]). Other types of malformations were associated with a detection rate of 15 of 41 (30%). Box 1 groups the types of malformation according to their detection rates.
Background

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Future Direction

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Anomaly Detection

**Pearls & Conclusions**

- **Limit**s are 11w0d-13w6d
- Ideally 12-13 weeks
- Skull ossifies at 10.5-11 weeks
- Fetuses have **exomphalos** at 9-10 weeks that resolves by 12 weeks

**MUST KEEP IN MIND TIMING...**

- Just as in the second trimester
  - Fetal position and dexterity
  - Placental localization
  - Full anatomic evaluation

**PLACENTA**

**SYSTEMATIC APPROACH**
**MG Characteristic**

<table>
<thead>
<tr>
<th>Normal Controls (n=86)</th>
<th>Facial Clefts</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Isolated</td>
</tr>
<tr>
<td>In Gap</td>
<td>34 (39.5%)</td>
</tr>
<tr>
<td>Partial Gap</td>
<td>2 (2.4%)</td>
</tr>
<tr>
<td>Complete Gap</td>
<td>2 (2.4%)</td>
</tr>
<tr>
<td>Gap &lt; 1.5 mm</td>
<td>6 (100%)</td>
</tr>
<tr>
<td></td>
<td>13/47 (27.7%)</td>
</tr>
</tbody>
</table>

**THE MID-SAGITTAL VIEW**

**SYSTEMATIC APPROACH**
SYSTEMATIC APPROACH

ABDOMEN

Image adapted from Bault et al. UOG 2010; 36:11

EVANASCENT PELVIC LUCENCY IN ANAL ATRESIA

SYSTEMATIC APPROACH

PELVIS

Image adapted from Bault et al. UOG 2010; 36:11

SPINE
**INTRACRANIAL TANSLUCENCY**

**12W1D FETUS**

**AND AT 14W2D**

**RATIO OF BPD TO TAD IN NTD**

- BPD/TAD ~ 1

**EXTREMITIES**

**ABSTRACT**

Objective. To evaluate the diagnostic accuracy of intracranial translucency (IT) in the detection of spina bifida (SB) in the first trimester of pregnancy.

Methods. We included studies assessing the accuracy of sonoanatomic measurements of IT in a trans-sagittal view of the baby due to position of SB in the first trimester of pregnancy. The primary outcome was the accuracy of IT in the prediction of spina bifida. Sensitivity, specificity, positive and negative likelihood ratios (LR+) and diagnostic odds ratio for the overall predictive accuracy of IT were computed.

Results. Nine studies (12978 fetuses) were included in the meta-analysis. IT was successfully measured in the majority of fetuses. The meta-analysis demonstrated that the sensitivity was 0.80 (95% CI 0.78-0.88), specificity 0.72 (95% CI 0.63-0.83), positive LR 2.07 (95% CI 1.77-2.50), negative LR 0.30 (95% CI 0.22-0.40), and diagnostic odds ratio 13.87 (95% CI 5.87-32.97).

Conclusions. The use of IT as a screening method in the detection of ventral wall defects in the first trimester of gestation remains controversial. Based on the available evidence, it appears that the potential for IT to be a reliable predictor can be further examined in a high-risk cohort. Positive results may provide leading to unnecessary parental anxiety. © 2015 Wiley Periodicals, Inc.
THE FIRST TRIMESTER SWEEP

CONGENITAL HEART DISEASE

• Most common major abnormality
• Incidence: 8.8/1000 live births
• 30% with associated defects
• Contributes to >50% of congenital anomaly-related deaths in childhood

NON-SELECTED POPULATION IN NORWAY
• 30,149 fetuses
• Detection Rate at 57%

HOW GOOD ARE WE?

Friedberg et al. J. Pediatr. 2009; 155:26
• Prospective 1 year study
• Northern California
• Fetuses and infants with CHD < 6 months
• 98/309 diagnosed (36%)

Prenatal recognition of CHD rose from 17% in 1994 to 30% in 1995 and 50% in 1996.

Conclusions—A simple training program for obstetric ultrasonographers increased their ability to detect serious congenital heart disease at a routine 18- to 20-week anomaly scan.
**THE FIRST TRIMESTER HEART**

- Rice Grain
- Coin: 1 Euro

**KEEPING IN MIND…**

**MUST ACQUIRE SKILL IN**

**THE SECOND TRIMESTER…**

**IN ORDER TO OBTAIN THESE VIEWS**

**SYSTEMATIC APPROACH**

18-22 WEEKS

**RISK FACTORS**

- Cardiac Abnormalities 5/1000 (0.5%)
- Diabetic Mum 10-15/1000 (1-1.5%)
- Previous Affected Child 20/1000 (2%)
- NT > 3.5 mm 50-70/1000 (5-7%)
• Pooled Sensitivity NT >95th Centile is 46%
• Pooled Sensitivity NT >99th Centile is 21%

<table>
<thead>
<tr>
<th>Type of CHD</th>
<th>NT &gt; 95th Centile</th>
<th>Abnormal</th>
</tr>
</thead>
<tbody>
<tr>
<td>Conotruncal</td>
<td>30.6%</td>
<td>81.6%</td>
</tr>
<tr>
<td>Univentricular Hearts</td>
<td>37.9%</td>
<td>96.6%</td>
</tr>
<tr>
<td>Combined CHD</td>
<td>57.9%</td>
<td>94.7%</td>
</tr>
<tr>
<td>Total</td>
<td>51.7%</td>
<td>74.1%</td>
</tr>
</tbody>
</table>

WHAT CAN WE SEE?

EARLIEST REPORTS
MORE RECENTLY...

- Haak et al
  UOG 2002; 20:9
  Transvaginal 92%

- Huggon et al
  UOG 2002; 20:22
  Transabdominally 84%

NEW TREND

Utrasound Obstet Gynecol 2003; 22:6-8

Opinion

First-trimester fetal echocardiography: is the future now?

CARDIAC IMAGING AT 11-14 WEEKS

Key Points
- Heart Develops GA 5-8 Weeks
- Chest AP diameter is about 2.5 cm at 12-13 weeks

Can Assess
- Position
- Connections
- Symmetry of 4 Chambers
- 2 AV valves/Septum (Doppler)
- Septal Continuity
- 2 Semilunar Valves (Doppler)
- Normal Cross Over of Arteries

ANATOMIC LANDMARKS

- Left atrium and aorta occupy the center of the chest
- Aorta points to the right shoulder as it exits then heads posteriorly towards the spine
- Pulmonary artery (PA) points to the left shoulder as it exits
- Outflow tracts cross over, with the PA being more anterior than the left ventricular outflow tract
- Postulation of the PA, the aorta and PA are almost parallel

ANATOMIC LANDMARKS

- Right ventricle is the most anterior, below the sternum
- Left atrium is connected to the spine near central structure in the chest
- Aorta is just anterior to the left of the spine
- Tricuspid valve is more lateral than aortic valve
- Aorta exits near exit of left atrium
- Moderator band is in the right ventricle
- Atria traversed by the left ventricle

SYSTEMATIC APPROACH

TRANSVERSE VIEWS
SYSTEMATIC APPROACH

SAGITTAL VIEWS

Abd Circ
Apex
LVOT - Ao
RVOT - PA
3VV
4CV

ESTABLISHING SITUS

4 CHAMBER VIEW

Sinkovskaya et al. UOG 2010; 36:676
Sinkovskaya et al. UOG 2014; 44:10

CARDIAC AXIS 30-60
TRICUSPID REGURGITATION

PULMONARY VEINS

TA at 13w1d Using Linear 9MHz Probe

R PULMONARY VEINS

Diagram Courtesy of L. Daou, MD

SYSTEMATIC APPROACH TRANSVERSE VIEWS

OUTFLOW TRACTS

OUTFLOW TRACTS
OUTFLOW TRACTS

SYSTEMATIC APPROACH
TRANSVERSE VIEWS

CROSS OVER

3 VESSEL VIEW

3 VESSEL VIEW
TA at 13w2d Using RM6C/OB

SYSTEMATIC APPROACH
TRANSVERSE VIEWS

AORTIC ARCH & DESCENDING AORTA

RIGHT ATRIAL INFLOW

IVC  SVC  RA  DAo

RIGHT ATRIAL INFLOW

TV at 13w1d Using RIC 1-12

RIGHT ATRIAL INFLOW

IVC  SVC  RA  DAo

RIGHT ATRIAL INFLOW
Use of spatiotemporal image correlation at 11–14 weeks' gestation

Use of spatiotemporal image correlation at 11–14 weeks' gestation

C. VOFINO, T. CO, R. AM. RUTTIMI, S. DARIAN SARD, V. GALLO, A. DORRESCU, H. DAVY and J. JAMU

Departments of Ob/Gyn and Gynecology, University Hospital, Brussels, Belgium. Centre for Advanced Fetal Care, Hospital, Department of Radiology, Brussels, Belgium.
**RESULTS**

<table>
<thead>
<tr>
<th>1st period</th>
<th>2nd period</th>
<th>3rd period</th>
<th>p-value</th>
</tr>
</thead>
<tbody>
<tr>
<td>% Views out of 8</td>
<td>4.76 (59.5%)</td>
<td>6.0 (75%)</td>
<td>7.89 (98.6%)</td>
</tr>
<tr>
<td>Average Time (sec)</td>
<td>262.4 (4.87m)</td>
<td>429.3 (7.13m)</td>
<td>560.1 (9.3m)</td>
</tr>
<tr>
<td>BMI</td>
<td>24.08</td>
<td>24.0</td>
<td>23.5</td>
</tr>
<tr>
<td>CRL</td>
<td>72.2</td>
<td>72.7</td>
<td>72.1</td>
</tr>
</tbody>
</table>

**CONCLUSION**

- Fetal cardiac evaluation is feasible in the first trimester
- At least 52 exams and an average time of 10 minutes needed
- Time allocation and gained sonographer experience are the most significant factors
Various Developmental Limitations with Structural Defects

- Always detectable
- Somewhat detectable
- Never detectable

First Trimester Detection of Structural Abnormalities

<table>
<thead>
<tr>
<th>Always Detectable</th>
</tr>
</thead>
<tbody>
<tr>
<td>Acrania</td>
</tr>
<tr>
<td>Cystic hygroma</td>
</tr>
<tr>
<td>Exompholos/Gastroschisis</td>
</tr>
<tr>
<td>Ectopia Cordis</td>
</tr>
<tr>
<td>Megacystis</td>
</tr>
<tr>
<td>Sirenomelia/Limb Abnormalities</td>
</tr>
</tbody>
</table>

Acrania

- Always Detectable
- Acrania
- Cystic hygroma
- Exompholos/Gastroschisis
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- Megacystis
- Sirenomelia/Limb Abnormalities
FIRST TRIMESTER DETECTION OF STRUCTURAL ABNORMALITIES

Always Detectable
- Acrania
- Cystic hygroma
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- Sirenomelia/Limb Abnormalities

Pentalogy of Cantrell

ECTOPIA CORDIS AT 11W6D

Always Detectable
- Acrania
- Cystic hygroma
- Exompholos/Gastroschisis
- Ectopia Cordis
- Megacystis
- Sirenomelia/Limb Abnormalities
MEGACYSTIS

- Acrania
- Cystic hygroma
- Exomphalos/Gastroschisis
- Ectopia Cordis
- Megacystis
- Sirenomelia/Limb Anomalies

FIRST TRIMESTER DETECTION OF STRUCTURAL ABNORMALITIES

LIMB AMPUTATION

- Holoprosencephaly
- Cephalocele
- Choroid plexus cysts
- Facial clefts
- NTD
- Univentricle/HLHS/HRHS
- Congenital diaphragmatic hernia
- Skeletal dysplasias
- Renal agenesis/hydronephrosis
- Single umbilical artery
- Anal atresia

FIRST TRIMESTER DETECTION OF STRUCTURAL ABNORMALITIES

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Various Developmental Limitations with Structural Defects

First trimester detection of structural abnormalities:

- Always detectable
- Somewhat detectable
- Never detectable
HOLOPROSENCEPHALY

FIRST TRIMESTER DETECTION OF STRUCTURAL ABNORMALITIES

Somewhat Detectable
- Holoprosencephaly
- Cephalocele
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PROBOSCIS AT 12W6D

CEPHALOCELE

FIRST TRIMESTER DETECTION OF STRUCTURAL ABNORMALITIES

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CHOROID PLEXUS CYST
FIRST TRIMESTER DETECTION OF STRUCTURAL ABNORMALITIES

Somewhat Detectable
- Holoprosencephaly
- Cephalocele
- Choroid plexus cysts
- Facial clefts
- NTD
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FACIAL CLEFTS

NTD AT 12W6D

NTD AT 12W6D

NTD AT 12W6D
FIRST TRIMESTER DETECTION OF STRUCTURAL ABNORMALITIES

- Somewhat Detectable
  - Holoprosencephaly
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  - Skeletal dysplasias
  - Renal agenesis/hydronephrosis
  - Single umbilical artery
  - Anal atresia

HYPOPLASTIC RIGHT HEART

12 WEEKS 6 DAYS

UNIVENTRICLE

DEXTROCARDIA AT 13W3D

AV CANAL

TETRALOGY OF FALLOT AT 12W6D
FIRST TRIMESTER DETECTION OF STRUCTURAL ABNORMALITIES

Somewhat Detectable
- Holoprosencephaly
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- Skeletal dysplasias
- Renal agenesis/hydronephrosis
- Single umbilical artery
- Anal atresia

Abu-Rustum, et al. UOG 2011; 38:190

Left CDH at 12W5D

FIRST TRIMESTER DETECTION OF STRUCTURAL ABNORMALITIES

Somewhat Detectable
- Holoprosencephaly
- Cephalocele
- Choroid plexus cysts
- Facial clefts
- NTD
- Univentricle/HLHS/HRHS
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- Skeletal dysplasias
- Renal agenesis/hydronephrosis
- Single umbilical artery
- Anal atresia

Abu-Rustum et al. UOG 2010; 36 (supplement 1) 168-305

RHIZOMELIA AT 12W6D

First and second trimester anomalies.

FIRST TRIMESTER DETECTION OF STRUCTURAL ABNORMALITIES

Somewhat Detectable
- Holoprosencephaly
- Cephalocele
- Choroid plexus cysts
- Facial clefts
- NTD
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Abu-Rustum et al. 2010; 36 (supplement 1) 168-305

TALIPES AT 12W6D

First and second trimester anomalies.

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RENAI PELVISES

FIRST TRIMESTER DETECTION OF STRUCTURAL ABNORMALITIES

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SINGLE UMBILICAL ARTERY

FIRST TRIMESTER DETECTION OF STRUCTURAL ABNORMALITIES

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  - Cephalocele
  - Choroid plexus cysts
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  - Anal atresia

ANAL ATRESIA

FIRST TRIMESTER DETECTION OF STRUCTURAL ABNORMALITIES

- Various Developmental Limitations with Structural Defects
  - Always detectable
  - Somewhat detectable
  - Never detectable
FIRST TRIMESTER DETECTION OF STRUCTURAL ABNORMALITIES

Never Detectable?
- Dandy-Walker malformation
- Ventriculomegaly
- Agenesis of the corpus callosum
- Vermian agenesis
- Mild valvular heart abnormalities
- Late appearing coarctation of the aorta
- Pulmonary abnormalities
- Duodenal atresia
- Bowel obstruction
- UPJ obstruction and other mild renal abnormalities

FIRST TRIMESTER DETECTION OF STRUCTURAL ABNORMALITIES

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DANDY WALKER MALFORMATION AT 13W3D

FIRST TRIMESTER DETECTION OF STRUCTURAL ABNORMALITIES

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- Bowel obstruction
- UPJ obstruction and other mild renal abnormalities

ROLE OF CARDIAC AXIS

FIRST TRIMESTER DETECTION OF STRUCTURAL ABNORMALITIES

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- Ventriculomegaly
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- Vermian agenesis
- Mild valvular heart abnormalities
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- Pulmonary abnormalities
- Duodenal atresia
- Bowel obstruction
- UPJ obstruction and other mild renal abnormalities
PLEURAL EFFUSION AT 12W1D

Background

Pearls & Conclusions

Why Early?

Future Direction

Systematic Evaluation

Limitations

Anomaly Detection

Training

Machinery

Maternal Body Habitus

Developmental Stage

Undue Anxiety

Time Consuming

Greatest challenge is the LOW RISK PATIENT!

TECHNICAL/PERSONAL LIMITATIONS

SAFETY IN THE FIRST TRIMESTER

SAFETY IN THE FIRST TRIMESTER

Bromley et al. JUM 2014; 33:1209

UFHealth University of Florida Health

UFHealth University of Florida Health

UFHealth University of Florida Health

UFHealth University of Florida Health
Background

Why Early?

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Pearls & Conclusions

ANATOMY AT 8-10 WEEKS?
VOTINO ET AL UOG 2014; 44: 10

9W4D FETUS CRL 28.7 MM

UNIQUE TO THE FIRST TRIMESTER

9W4D FETUS CRL 28.7 MM
RESULTS

ultimately

Background

Why Early?

Systemic Evaluation

Future Direction

Limitations

Anomaly Detection

PRACTICAL PEARLS

Conclusions

Detailed first trimester fetal assessment is feasible
Critical role in the era of NIP
Powerful tool for early reassurance
May diagnose over 70% of major anomalies/CHD
Does not replace the second trimester scan
Its incorporation into clinical practice is inevitable

IT IS TIME TO LOOK BEYOND THE NT

CONCLUSION

Learn the Past and Research the Present to Predict the Future…

HIPPOCRATES
The Future is so incredibly bright...