The First Trimester Anatomy Scan

John R. Allbert, MD
Novant Maternal-Fetal Medicine Associates
Anatomic Survey

• Second trimester (18-22 week) scan remains the gold standard
• First trimester evaluation of anatomy introduced in the 1980’s
• NT aneuploid screening has rekindled the interest in the first trimester anatomy scan
1st Trimester Ultrasound

- Viability
- Accurate gestational age
- Aneuploid screening
- Early detection of major anomalies
- Earlier genetic diagnosis
- Critical for the determination of multiple pregnancy chorionicity
- Identify fetuses at high risk of IUGR and PIH
Nuchal Translucency (NT)
Nuchal Translucency (NT)
The Most Critical Component of 1st Trimester Screening

- Fluid collection behind the fetus’ neck
- Measured by U/S between 10+3 and 14+0 wks, (CRL 39-84 mm)
- Excess Fluid accumulation is an indicator of an increased risk of fetal abnormalities
  *Trisomy 21, trisomy 18, & heart defects
- The more fluid indicates a greater risk of an abnormality
Increased NT 5.5 mm
Screening for Down Syndrome

- FIRST TRIMESTER MARKERS
- Advanced maternal age
- Excess Nuchal translucency
- Low PAPP-A
- Elevated hCG
First Trimester Ultrasound
Beyond Aneuploidy

• Abnormalities associated with an increased nuchal translucency
• Screening for other anomalies
  *Direct visualization
  *Indirect visualization
• Risk for perinatal M&M: preeclampsia & IUGR
NT
Pathophysiology

• Cardiac dysfunction
• Altered composition of the extracellular matrix
• Failure of lymphatic drainage
• Fetal anemia
• Hypoproteinemia
• Congenital infection
## Increased NT

<table>
<thead>
<tr>
<th>NT</th>
<th>Aneuploidy</th>
<th>Fetal Death</th>
<th>Anomalies</th>
<th>Alive &amp; Well</th>
</tr>
</thead>
<tbody>
<tr>
<td>&lt;95^{th}%</td>
<td>0.2%</td>
<td>1.3%</td>
<td>1.6%</td>
<td>97%</td>
</tr>
<tr>
<td>95-99%</td>
<td>3.7%</td>
<td>1.3%</td>
<td>2.5%</td>
<td>93%</td>
</tr>
<tr>
<td>3.5-4.4mm</td>
<td>21.2%</td>
<td>2.7%</td>
<td>10.0%</td>
<td>70%</td>
</tr>
<tr>
<td>4.5-5.4mm</td>
<td>33.3%</td>
<td>3.4%</td>
<td>18.5%</td>
<td>50%</td>
</tr>
<tr>
<td>5.5-6.4mm</td>
<td>50.5%</td>
<td>10.1%</td>
<td>24.2%</td>
<td>30%</td>
</tr>
<tr>
<td>≥6.5mm</td>
<td>64.5%</td>
<td>19.0%</td>
<td>46.2%</td>
<td>15%</td>
</tr>
</tbody>
</table>

Soulka AJOG 2005 192:1005
Conditions Associated with Increased NT

- Cardiac defects
- CDH
- Omphalocele
- Achondroplasia
- Asphyxiating thoracic dystrophy
- Blomstrand Osteochondrodysplasia
- Body Stalk Anomaly
- Campomelic Dysplasia
- Fetal Akinesia Deformation Sequence
- Fryn Syndrome
- Hydrolethalus Syndrome
- Jarco Levin Syndrome
Conditions Associated with Increased NT

- Jourbert Syndrome
- Meckel-Gruber Syndrome
- Nance-Sweeny Syndrome
- Noonan Syndrome
- Perlman Syndrome
- Smith Lemli Opitz Syndrome
- Spinal Muscular Atrophy Type I
- Osteogenesis Imperfecta
- Thanatophoric Dysplasia
- Short Rib Polydactyly Syndrome
- Trigonocephaly “C” Syndrome
- VACTERL
- Zellweger Syndrome
13-14 Week Fetal Anatomy Scan

• 5 year prospective study
• N=2876
• Transabdominal and transvaginal if needed
• TV superior with cranium, spine, stomach, kidneys and upper and lower limbs
• TA complete scan in 64%
• TA & TV complete scan in 82%
• Heart not adequately seen in 42%

Ebrashy A, UOG 2010;35:292
<table>
<thead>
<tr>
<th>Anomaly</th>
<th>13-14 weeks</th>
<th>16-20 weeks</th>
</tr>
</thead>
<tbody>
<tr>
<td>Anencephaly</td>
<td>3</td>
<td>0</td>
</tr>
<tr>
<td>Cardiac</td>
<td>1</td>
<td>3</td>
</tr>
<tr>
<td>Omphalocele</td>
<td>1</td>
<td>1</td>
</tr>
<tr>
<td>Gastrochisis</td>
<td>2</td>
<td>2</td>
</tr>
<tr>
<td>Megacystis</td>
<td>2</td>
<td>2</td>
</tr>
<tr>
<td>Skeletal dysplasia</td>
<td>1</td>
<td>2</td>
</tr>
<tr>
<td>Holoprosencephaly</td>
<td>1</td>
<td>0</td>
</tr>
<tr>
<td>Bilateral Talipes</td>
<td>1</td>
<td>1</td>
</tr>
</tbody>
</table>

Ebrashy 2010
## Sensitivity of Major Anomalies
### 13-14 week Ultrasound

<table>
<thead>
<tr>
<th>Author</th>
<th>Year</th>
<th>Number</th>
<th>Sensitivity %</th>
</tr>
</thead>
<tbody>
<tr>
<td>Economides</td>
<td>1998</td>
<td>1632</td>
<td>54</td>
</tr>
<tr>
<td>Carvalho</td>
<td>2002</td>
<td>2853</td>
<td>38</td>
</tr>
<tr>
<td>Taipale</td>
<td>2004</td>
<td>4513</td>
<td>18</td>
</tr>
<tr>
<td>Chen</td>
<td>2004</td>
<td>1609</td>
<td>54</td>
</tr>
<tr>
<td>Soulka</td>
<td>2006</td>
<td>1148</td>
<td>50</td>
</tr>
<tr>
<td>Ebrashy</td>
<td>2010</td>
<td>2876</td>
<td>68</td>
</tr>
</tbody>
</table>
Anomaly Detection Rates

• 100% Acrania, anencephaly, ectopia cordis, encephalocele
• 50-90% DORV, gastroschisis, omphalocele, holoprosencephaly, limb reduction, megacystis
• 1-49% Spina bifida, hydrocephalus, skeletal dysplasia, facial cleft
Suggested Anatomical Assessment
11-13+6 Weeks
Head

- Present
- Cranial bones
- Midline falx
- Choroid-plexus-filled ventricles
Encephalocele
Cephalocele
Cephalocele
Cephalocele
First Trimester
Normal Choroid Plexus

Butterfly
Holoprosencephaly

Sepulva JUM 2004; 23:761
Holoprosencephaly

Sepulva JUM 2004:23:761
Suggested Anatomical Assessment
11-13+6 Weeks
Face

- Eyes with lens
- Nasal bone
- Normal profile/mandible
- Intact lips
Increased NT
Orofacial Clefts

- N=8638
- NT increased in 714 (8.6%),
  513, (5.9%) euploid
- CL and or CL&CP in 18, rate 2.2/1000
- Increased NT risk 19.5/1000
- NT < 95% risk 1/1000, (same as general population)

Timmerman E. Ultrasound Obstet Gynec 2010;36:427
First Trimester Ultrasound
Cleft Palate

• Retronasal triangle: 3 echogenic lines formed by 2 processes of the maxilla and palate, in the coronal section of the fetal face posterior to the nose

• Fetuses with cleft palate will have an abnormally shaped retronasal triangle

Sepulveda W, UOG 2010;25:7
Normal Retronasal Triangle

Sepulveda 2010
Retronasal Triangle
Retronasal Triangle
Unilateral Clefting

Sepulveda 2010
Holoprosencephaly and Cleft Palate

Sepulveda 2010
Facial Profile Dysmorphism

- Agnathia (Otocephaly): rare anomaly complete or severe hypoplasia of the mandible
- Micrognathia: small mandible
- Retrognathia: posterior displacement
- Mandible can be seen as early as 10 weeks
Facial Profile Dysmorphism
Micrognathia/Retrognathia

• Important phenotype of many syndromes
• The mandible requires several elements from different embryonic components to interact and fuse
• Susceptible to a series of molecular and genetic insults
Micrognathia

Orofacial digital syndrome

Fetal akinesia deformation sequence

Acrofacial dysostosis

Paladini D, UOG;2010:35:377
Facial Profile Dysmorphism Syndromes

- Acrofacial dystosis AD
- Treacher Collins AR
- Branchio-oculofacial AD
- Cerebrocostomandibular AD, AR
- Orofaciodigital
- Robin Sequence AD
Facial Profile Dysmorphism
Aneuploidy

- Trisomy 13, 18, 8, 9
- Deletion 3p, 4p, 5p
- Pallister-Killian
- Monosomy X
- Deletion 22q 11.2, 13q, 11q, 9q
- Triploidy
Facial Profile Dysmorphism Diagnosis

- **Subjective:** small or receding chin with prominent upper lip
- **Objective:** small A-P measurement between the two rami (Jaw Index) and decrease in the inferior facial angle (IFA)
- **Midfacial Hypoplasia:** increased frontomaxillary facial angle, (FMF)
- **Retrognathia:** Decrease in the Inferior facial angle, (IFA)
Frontomaxillary Facial Angle (FFA)
Facial Profile Dysmorphia

Mandible

IFA

Borenstein M. UOG 2007;30:928
Micrognathia
First Trimester

Paladini, D UOG 2010;35:377
Facial Profile Dysmorphism
1st Trimester

Borenstein M. UOG 2007;30:928
Facial Profile Dysmorphism

- Check for other anomalies
- Determine karyotype (44-65% aneuploid)
- Consider Robin Sequence
- Risk of recurrence: most are AD and AR
Suggested Anatomical Assessment
11-13+6 Weeks
Spine

- Vertebrae (longitudinal and axial)
- Intact overlying skin
Intracranial Translucency

Chaoui R, OUG 2009;34:249
Intracranial Translucency
Spina Bifida

Compression of the 4th ventricle

Chaoui 2009
First Trimester U/S
Spina Bifida

• Screening with MSAFP
• Ultrasound second trimester
  * Spine
  * Scalloping of the frontal bones
  * Caudal displacement of the cerebellum
Intracranial Translucency

Chaoui R, OUG 2009;34:249
First Trimester Diagnosis
Spina Bifida

• Midsagittal view of the fetal face
• 4\textsuperscript{th} ventricle is between the brainstem and the choroid plexus
• In spina bifida the 4\textsuperscript{th} ventricle is compressed by the caudally displaced hindbrain and cannot be visualized

Chaouï, R UOG 2009;34:249
Intracranial Translucency

Chaoui UOG 2010;35:133
Suggested Anatomical Assessment
11-13+6 Weeks
Chest

• Symmetrical lung fields
• No effusions or masses
Pleural Effusion
8 w 5d
Suggested Anatomical Assessment
11-13+6 Weeks
Heart

- Cardiac regular activity
- Four symmetrical chambers
13+4/7 Week ROFT
13+4/7 weeks LOFT
Increased NT Cardiac Defects Literature Review

- N=3448
- 2.5-3.4 mm, 17/1000
- >3.5 mm, 78.4/1000
- FPR (NT cutoff 2.5-3.0), 4.9%
- Detection Rate 37.5%

Souka AP, AJOG 2005;192:1005
Increased NT >95%
Cardiac Defects

• Left sided defects highest sensitivity
• HLHS 67%
• TGV 50%
• Coarcation 100%
• VSD 50%
• TOF 22%

Hyett JA, BMJ 1999;318:81
Increased NT
Cardiac Defects
Protocol

- Transvaginal ultrasound at 13-15 weeks
- Targeted ultrasound at 18 weeks
- Echocardiogram at 22-26 weeks
Ectopic Cordis
Ectopic Cordis
Suggested Anatomical Assessment
11-13+6 Weeks
Abdomen

- Stomach present in upper left quadrant
- Bladder
- Kidneys
Increased NT
11w3d
13+3/7 Weeks
Case
13+3/7 Weeks
15+3/7 Weeks
Congenital Diaphragmatic Hernia
15=3/7 Weeks
Increased NT
Diaphragmatic Hernia

- 1/4000
- Aneuploidy and other anomalies 50%
- Venous congestion probable cause of nuchal edema
- 37% of all cases of CDH
- 83% of those with NND secondary to pulmonary hypoplasia
- 22% of survivors
13+3/7 Weeks
First Trimester
Diaphragmatic Hernia

Normal

CDH
Omphalocele
Omphalocele
Risk of aneuploidy

• 8-10 weeks all fetuses have midgut herniation
• By 11 5/7 weeks the midgut herniation has retracted
• Neonate with omphalocele risk of aneuploidy 15%
• Second trimester risk is 30%
• 11 4/7-13 6/7 risk is 55-61%
• NT > 2.5, risk is 85%
• NT < 2.5, risk is 4%

Snijders, Ultrasound Obstet Gynec 1995;6:250
First Trimester Bladder
First Trimester Megacystis
First Trimester U/S
Megacystis
Megacystis
Increased NT
Megacystis

- ≥ 7 mm longitudinal diameter
- Trisomy 13 & 18 in 23.6-31.4%
- Euploidy spontaneously resolves in 90%
- >15 mm indicates obstructive uropathy

Kagan KO Ultrasound Obstet Gyn 2010;35:10
Evaluation for Bladder Exstrophy

- Bladder extrophy is a rare and severe malformation, (1/25-50K)
- Prenatal diagnostic signs include absent bladder, low insertion of the umbilical cord with normal kidneys and amniotic fluid volume.
- There may be a lower protruding abdominal mass
- There may be a reduced fetal umbilical cord insertion-to-genital tubercle measurement in early gestation
Evaluation for Bladder Exstrophy

• Prospective cross-sectional study evaluation of 140 patients
• UC insertion-to-genital tubercle length measured in midsagittal section
• Successful measurements were obtained on 134 patients 12-18 weeks
• Two cases with bladder extrophy were <95%
• 12 weeks <11.28 mm, 13 weeks <12.00 mm

Evaluation for Bladder Extrophy
Evaluation for Bladder Extrophy
Suggested Anatomical Assessment
11-13+6 Weeks
Extremities

• Four limbs each with three segments
• Hands and feet with normal orientation
View of Arm @ 12w2d
Fetal Hand 12w2d
Syrinomelia
Anatomy Screen
First Trimester

- Skull
- Cerebral midline echo
- Orbits
- Facial profile
- Spine
- Heart
- Stomach
- Anterior abd wall
- Kidneys
- Bladder
- Upper and lower limbs
- Hands and fingers
- Feet and Toes
Detection Rates 11-14 Weeks
Rossi, AC Obstet Gynecol 2013;122:1160