FIRST TRIMESTER US
MORE THAN JUST THE NECK

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Objectives

• Review expectations of basic first trimester ultrasound
• Discuss screening for aneuploidy
• Encourage observation of first trimester anatomic findings
Basic First Trimester Scans

- Location
- Viability
- Gestational age
- Number & Chorionicity
- Evaluate adnexae
- Pelvic fluid
- Uterine anatomy
Early First Trimester Scans
VIABILITY?

- **5/10/20 Rule**
  - CRL $\geq 5$
    - Heart Beat
  - MSD $\geq 10$ mm
  - MSD $\geq 20$ mm
    - Fetal pole

- **5.3/10/21 Rule**
  - CRL $\geq 5.3$ mm
    - Heart Beat
  - MSD $\geq 10$ mm
  - MSD $\geq 21$ mm
    - Fetal Pole

Mean Sac Size increases about 1 mm/day

CRL increases about 1 mm/day
Nuchal Translucency

- Include in all 1st trimester scans @11-13⁶ --
  - Even if not doing first trimester serum screening
- Standard measurements with certification
- Difficult measurement to obtain correctly
~ 75% fetuses with Tri 21 have increased NT
~65% absent nasal bone in 1st trimester
Measurement of NT
3.6 mm NT
Effects of NT on Risk for Tri 21 at 12 weeks

11-13\(^6\) Week US. Nicolaides Fetal Med Foundation 2004
<table>
<thead>
<tr>
<th>Nuchal translucency</th>
<th>Chromosomal Defects</th>
<th>Normal karyotype</th>
<th>Major fetal abnormalities</th>
<th>Alive and well</th>
</tr>
</thead>
<tbody>
<tr>
<td>&lt;95th centile</td>
<td>0.2%</td>
<td>1.3%</td>
<td>1.6%</td>
<td>97%</td>
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<tr>
<td>95th–99th centiles</td>
<td>3.7%</td>
<td>1.3%</td>
<td>2.5%</td>
<td>93%</td>
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<tr>
<td>3.5–4.4 mm</td>
<td>21.1%</td>
<td>2.7%</td>
<td>10.0%</td>
<td>70%</td>
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<tr>
<td>4.5–5.4 mm</td>
<td>33.3%</td>
<td>3.4%</td>
<td>18.5%</td>
<td>50%</td>
</tr>
<tr>
<td>5.5–6.4 mm</td>
<td>50.5%</td>
<td>10.1%</td>
<td>24.2%</td>
<td>30%</td>
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<tr>
<td>≥6.5 mm</td>
<td>64.5%</td>
<td>19.0%</td>
<td>46.2%</td>
<td>15%</td>
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</table>
# Abnormalities with Increased NT

<table>
<thead>
<tr>
<th>Central nervous system defect</th>
<th>Gastrointestinal defect</th>
<th>Fetal anemia</th>
</tr>
</thead>
<tbody>
<tr>
<td>Acrania / anencephaly</td>
<td>Crohn’s disease</td>
<td>Blackfan Diamond anaemia</td>
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<tr>
<td>Agenesis of the corpus callosum</td>
<td>Duodenal atresia</td>
<td>Congenital erythropoietic porphyria</td>
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<tr>
<td>Craniosynostosis</td>
<td>Esophageal atresia</td>
<td>Dyserythropoietic anaemia</td>
</tr>
<tr>
<td>Dandy Walker malformation</td>
<td>Small bowel obstruction</td>
<td>Fanconi anemia</td>
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<tr>
<td>Diastematomyelia</td>
<td></td>
<td>Parvovirus B19 infection</td>
</tr>
<tr>
<td>Encephalocele</td>
<td>Genitourinary defect</td>
<td>Thalassaemia-x</td>
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<tr>
<td>Fowler syndrome</td>
<td>Ambiguous genitalia</td>
<td></td>
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<tr>
<td>Holoprosencephaly</td>
<td>Congenital adrenal hyperplasia</td>
<td></td>
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<tr>
<td>Hydrothalus syndrome</td>
<td>Congenital nephrotic syndrome</td>
<td>Fetal akinesia deformation sequence</td>
</tr>
<tr>
<td>Intencephaly</td>
<td>Hydronephrosis</td>
<td>Myotonic dystrophy</td>
</tr>
<tr>
<td>Joubert syndrome</td>
<td>Hypospadias</td>
<td>Spinal muscular atrophy</td>
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<tr>
<td>Macrocephaly</td>
<td>Infantile polycystic kidneys</td>
<td></td>
</tr>
<tr>
<td>Microcephaly</td>
<td>Meckel-Gruber syndrome</td>
<td></td>
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<tr>
<td>Spina bifida</td>
<td>Megacystis</td>
<td>Beckwith-Wiedemann syndrome</td>
</tr>
<tr>
<td>Trigonoocephaly C</td>
<td>Multicystic dysplastic kidneys</td>
<td>GM1 gangliosidosis</td>
</tr>
<tr>
<td>Ventriculomegaly</td>
<td>Renal agenesis</td>
<td>Long-chain 3-hydroxyacyl-coenzyme A dehydrogenase deficiency</td>
</tr>
</tbody>
</table>

### Facial defect

*Facial cleft*

- Acrania/micrognathia: Achondroplasia
- Facial cleft: Achondroplasia
- Micrognathia: Asphyxiating thoracic dystrophy
- Treacher-Collins syndrome: Blomstrand osteochondrodysplasia

### Nuchal defect

- Cleft lip: Cleidocranial dysplasia
- Cystic hygroma: Hypochondroplasia
- Neck lipoma: Hyppophagastasia
- Nuchal defect: Jorcho-Levin syndrome
- Cardiac defect: Kyphoscoliosis
- Di George syndrome: Limb reduction defect
- Pulmonary defect: Osteogenesis imperfecta
- Cystic adenomatoid malformation: Roberts syndrome
- Diaphragmatic hernia: Robinow syndrome
- Fryn syndrome: Short-rib polydactyly syndrome
- Abdominal wall defect: Talipes equinovarus
- Cloacal exstrophy: Thanatophoric dwarfism
- Exomphalos: VACTERL association
- Gastrochisis: Sirenomelia

### Other defect

- Body stalk anomaly: EEC syndrome
- Body stalk anomaly: Body stalk anomaly
- Brachman-de Lange syndrome: Nance-Sweeney syndrome
- Deficiency of the immune system: Noonan syndrome
- Congenital lymphedema: Perlman syndrome
- Stickler syndrome: Unspecified syndrome
- Severe developmental delay: Sirenomelia
Cystic Hygroma and Increased NT

- 5 Medical Centers over 10 years; 944 cases
- Hygroma = Enlarged hypoechoic space at the back of the fetal neck extending the length of the fetal back with obvious septations

Scholl et al: 2012
Karyotype Results
729/944 First Trimester Hygromas

- Normal, 329
- Trisomy 18, 83
- Monosomy X, 88
- Trisomy 13, 26
- Tripolidy, 10
- Mosaic, 10
- Other, 27

Scholl et al. Cystic Hygroma and NT Thickness
Ob GYN Sept 2012
Perinatal Outcomes

• Termination of Pregnancy  447
• Continued pregnancy  295

Outcomes of Continued Pregnancies

Scholl et al; 2012 Hygromas
First Trimester Development

- Ossification of fetal skull unreliable until 11 weeks
  - Acrania, anencephaly
- 4-chamber heart view after 10 weeks
- Normal midgut herniation at 8 – 10 weeks
  - Cannot exclude VWD until then
- Fetal bladder
  - 10 weeks: 50%
  - 11 weeks: 80%
  - 12 weeks: 100%
Early Fetal Anatomical Sonography
Donnelly, GUMed, Malone Best Practice & Research
Clinical Ob GYN 2012

• Excellent review of literature on first trimester embryology and sonography
• 11-14 weeks
• Emphasizes importance of rapidly changing embryonic development at this point with changing “normal” v “abnormal”
5 Week Yolk Sac
Pooh, 2012
4-5 weeks

- Yolk sac located between chorion and amnion
- First seen 5\textsuperscript{th} post menstrual week
- Embryo
  - 2-3 mm linear structure close to uterine wall
  - Heart rate < 100 not predictive of poor outcome

Donnelly: Prenatal Dx 2012
6-9 weeks

• Embryo relatively undifferentiated in appearance
• HR average 130 bpm
• After heart beat, neural tube is next visible structure
  » Hypoechoic linear structure running length of fetus
  » 2 parallel lines
13 weeks pook
• 7 Weeks
  » Rhombencelphalon
    • Single intracranial cystic structure=4th vent
  » Some fetuses with cerebral tissue visible
  » Normal gut herniation

• 8 Weeks
  » Choroid plexus visible
  » Wide 3rd ventricle (Diencephalon)
  » Stomach visible (Visible in 100% by 11 weeks)
  » Atria and ventricles visible
• **9 Weeks**
  » Cerebral hemispheres visible in all
    • Lateral ventricles increase; 3\textsuperscript{rd} ventricle narrows
  » Spine: 2 paralleled echogenic lines
  » Normal mid-gut herniation
  » Esophagus visible as bright streak behind heart
  » Long bones, hands and feet
Systematic Approach 10-14 weeks

- Location of gestation, placenta(s)
- Viability
- Chorionicity
- Anatomy
  - Transverse head: ossified cranial bones, midline echo and choroid plexus
  - Midsaggital face: Nasal bone, orbits, profile
  - Sagittal spine: Intact skin
  - Transverse thorax: 4 chamber heart, axis, situs
  - Transverse/sagittal Abdomen: Stomach, bladder, cord
  - Boney survey
CNS

• Embryologically incomplete in 1st trimester
  » Vermis, corpus callosum, gyri, sulcation

• Acrania
  » Near 100% detection possible by 14 weeks
  » Near 100% detection holoprosencephaly by 14 weeks

• Spina Bifida
  » Brain stem to occipital bone length
  » Aqueduct of sylvius to occiput measurement
Figure 12. Embryo structures visualized by MR image (Carnegie stage 23).

Figure 13. Tomographic sagittal imaging of normal fetus at the beginning of 8 weeks of gestation.
Figure 15. Sagittal section and coronal sections of 10-week-fetus CP; Choroid plexus of the lateral ventricles.
Figure 16. Normal brain development by mid-sagittal 3D US section between 8 and 12 weeks of gestation.
4 Parallel Lines: ONTD Screening

Normal

Loss of 4th line/cisterna magna

1=Upper border of Brain stem
2=Lower border of Brain Stem
3=Lower border of 4th vent w/CP
4=Cisterna Magna

Kavalakis 2012 Prenatal Dx
Intracranial Translucency Measurement

Cursor Placement

Cisterna Magna
Cardiac exam

- Situs
- ¼ of transverse chest
- 4 chambers
- 45 degree
- Displacement of tricuspid valve
Thorax

- **CDH**
  - Diaphragm visible at 10-11 weeks
  - Stomach without fluid in it
  - Low rate of detection
  - Increased NT

- **CPAM**
  - Earliest reported detection at 16 weeks
Gastrointestinal

- **Know your embryology**
  - High detection rate of VWD but also a lot of false positives

- **Abdominal cystic mass**
  - Often resolve—serial scans!
  - All the usual suspects
    - GI, mesenteric cysts, liver cysts, renal
3D/4D sonography moved prenatal diagnosis of fetal anomalies from the second to the first trimester of pregnancy
Ritsuko K. Pooh1 & Asim Kurjak2

Figure 47. Omphalocele at 12 and 13 weeks of gestation 3D reconstructed ultrasound images clearly demonstrates omphalocele. Trisomy 18 was confirmed by chorionic villi sampling in both cases.
Renal System

- Urine production by 10 weeks
- 10 weeks: 50% bladder visualization
- 13 weeks: >95% bladder visualization
- Bladder measurement >10% of CRL is normal
- Oligo of renal origin rare < 14 weeks
- Renal pyelectasis > 1.5 mm AP diameter in 1st trimester
Skeletal abnormalities

• Precise landmarks for femur absent
• Don’t use long bones for EGA at this gestational age
• Assess for presence of limbs in 1st trimester
The Journal of Maternal-Fetal and Neonatal Medicine, 2012; 25(5): 433–455

Figure 43. Upper limb abnormality at 11 weeks of gestation. 3D ultrasound revealed contracted elbow joint abnormality. Right figures show macroscopic appearance of upper limbs of aborted fetus.

Figure 44. Fetal clubfoot at 13 weeks of gestation. Left; 3D image of fetal leg. Right; Legs of aborted fetus. This case was associated with chromosomal aberration.
100% of Dizygous Twins
• Twin intrauterine pregnancy, with two viable embryos. This is a dichorionic, diamniotic pregnancy.
• Embryo A has a posterior right lateral wall placenta while Embryo B has an anterior-fundal placenta mostly on the left side.
Acrania: CRL 22-28 mm

Prenatal Diagnosis
2009: Blaas
Occipital Encephalocele
22 mm CRL
Facial Clefting
Min-Pan: Prenatal Dx 2012

• 1/500 to 1/1000 Live Births
  » Cleft lip  25%
  » Cleft lip and palate  51%
  » Cleft palate  24%

• 2nd Trimester Detection

2 D  Low Risk
  » 33-88% cleft lip
  » 0-22% for isolated
     Lip
  » 0-22% for isolated
     Palate
     cleft palate

3 D  High Risk
  » 100% isolated cleft
     Lip
  » 0-89% isolated
     Cleft palate
Frontomaxillary Facial Angle (FMF)

- Mid-sagittal view of facial profile
- Angle made by a line drawn along the upper surface of the palate and another line which traverses the upper corner of the anterior aspect of the maxilla extending to the external surface of the forehead
  » Objective measure of maxillary growth
  » Abnormal with Tri 21, Cleft Palate
Retrospective Review from Chinese First Trimester Screening Images

- Measured FMF on saved images
- Normal new borns vs those born with cleft lip and/or cleft palate
- Found NO difference in FMF in affected vs control

- FMF may be helpful to screen for mid face hypoplasia, Tri 21 but not helpful for clefting
Cardiac

• Nuchal Translucency >2.5 mm should prompt 2\textsuperscript{nd} trimester echo

• Situs
  » Aorta to left of spine ; IVC to right and anterior
  » Axis 45 degrees from midline
  » ¼ of chest
  » 4 chamber view, offset tricuspid valve
  » Right and left outflow with ductal , aortic arches
### 1st Trimester Detection Cardiac Detects

#### Donnelly Table 2

<table>
<thead>
<tr>
<th>Condition</th>
<th>Syngelaki</th>
<th>Composite of prior studies</th>
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</thead>
<tbody>
<tr>
<td>Coarctation</td>
<td>27%</td>
<td>9.5%</td>
</tr>
<tr>
<td>Tetrology</td>
<td>30%</td>
<td>4.8%</td>
</tr>
<tr>
<td>HLH</td>
<td>40%</td>
<td>21%</td>
</tr>
<tr>
<td>AVSD</td>
<td>33%</td>
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<tr>
<td>DORV</td>
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<tr>
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<td>VSD</td>
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<tr>
<td>TOTAL</td>
<td>29%</td>
<td>7%</td>
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Gender

• Until 14 weeks no appreciable difference in size of genital tubercle

• Angle of tubercle to lumbosacral skin surface
  » >30deg. Male
    • Incorrect assignment male to female
      » 56% at 11; 3% at 12; 0% at 13 weeks
    • Incorrect assignment female to male
      » 5% at 11