The 1st trimester scan

- Routine ultrasound examination is an established component of prenatal care
  - If there are adequate resources
  - Access
- Current advancements in technology have resulted in improvements in resolution
  - Early fetal development can now be assessed & monitored in detail

The 1st trimester scan

studies up to 13w 6d
- Embryo until 10 weeks
- Fetus after 10 weeks
The 1st trimester scan

• Provide information which will optimize prenatal care and lead to the best possible outcomes for both mother and fetus.

The 1st trimester scan

• Confirm viability
• Establish gestational age
• Evaluate for possible ectopic gestation

The 1st trimester scan

• Identify multiple pregnancies
  – Determine amnionicity & chorionicity

• Nuchal translucency measurement
  – As a component of Ultrascreen

• Potential to identify some major anomalies

Multiple gestations

• Incidence has increased significantly mostly in women over 30

• This is due to both
  – Increasing maternal age
  – Assisted reproductive techniques

Multiple gestations

• Determine number of embryos

• Significance
  • Increased risk for both mother and fetus
  • Up to 12% of perinatal deaths occur in multiple gestations
  • Perinatal mortality for twins is 5-10% greater than for singletons
All multiple pregnancies are high-risk pregnancies requiring close antepartum monitoring.

Zygosity
- Dizygotic (fraternal) twins
  - Fertilization of two separate ova
  - Account for approximately 70% of twins
  - Increased incidence with
    - Increasing maternal age
    - Ethnicity (highest in Blacks, lowest in Asians)
    - Family history
    - Assisted reproductive techniques

Monozygotic twins
- Arise from the division of a single zygote
- Account for approximately 30% of twin pregnancies
  - Relative frequency is approximately 1 per 250 live births worldwide
  - Slightly increased incidence in patients receiving ovulation induction agents

Chorionicity
- # of placentas

Amnionicity
- # of sacs

Dizygotic twins
- Two separate zygotes
- Two separate blastocysts
  - Each implant independently
  - Dichorionic-diamniotic
    - Two placentas unless blastocyst implantation is close enough to result in the formation of one fused placenta

Monozygotic twins
- Dichorionic-diamniotic (~1/3)
  - Division during first 3 days
- Monochorionic-diamniotic (~2/3)
  - Division between day 4 & day 8
- Monochorionic-monoamniotic
  - Division between day 8 & 11
  - Conjoined
Multiple gestations

- **Dichorionic-diamniotic twins**
  - Perinatal mortality ~ 10%
  - Preterm birth
  - IUGR
  - Slightly increased risk of anomalies

- **Monochorionic-diamniotic**
  - Perinatal morbidity & mortality 3-5 times > dichorionic twins
  - Preterm delivery
  - IUGR
  - Anomalies
  - Placental
    - Twin-twin transfusion
    - TRAP
    - Twin embolization syndrome

- **Monochorionic-monoamniotic**
  - Highest mortality because of risk of cord entanglement

Sonographic determination of amnionicity & chorionicity

- Accurate diagnosis has very important prognostic implications
- Prognosis varies with *chorionicity* rather than zygosity

- Transvaginal approach is preferred
  - Better resolution
- *Chorionicity* significantly impacts prognosis
  - The sooner it is established, the earlier a management plan can be determined

Role of sonography

- Determination of *chorionicity* is very reliable in the first trimester
- Always go to the *initial exam* when evaluating multiple pregnancies
- From 6-9 weeks, a thick septum is present separating the chorionic sacs
chorionicity

Dichorionic-diamniotic twins
Trichorionic-diamniotic triplets

Dichorionic twins
• Twin “peak”; lambda sign

Monochorionic-diamniotic twins

Monoamniotic twins

Monochorionic-monoamniotic twins

Conjoined twins
8 weeks 3 days
Conjoined twins

Thoracopagus conjoined twins

Ischiopagus conjoined twins
Conjoined twins

The 1st trimester scan

- **Nuchal translucency measurement**
  - Component of the Ultrascreen
- Potential to identify some major anomalies

Early evaluation of fetal anatomy

- **Nuchal translucency**
  - In conjunction with PAPP-A and free beta hCG

Early evaluation of fetal anatomy

- **Thickened NT**
  - Aneuploidy
    - Trisomies (21 and 18)
    - Turner syndrome
  - Cardiac anomalies
  - Chest masses
    - Diaphragmatic hernia
  - Omphalocele
  - Skeletal anomalies
  - Unexplained fetal demise

NT measurement
Adverse outcome (death, major anomaly) in euploid fetuses with increased nuchal lucency

Evolution

Ultrason; 11 weeks 3 days

Trisomy 18

Trisomy 18

Thickened NT

Evolution

Trisomy 18

Thickened NT
Thickened NT

Hypoplastic right heart

euploid

Monochorionic-diamniotic twins

10 weeks

Fetus B

Fetus A

Polyalveolar lung

Thickened NT

• euploid

• How has the increased use of NIPT impacted the role of ultrasound in the 1st trimester?
NIPT

- The use of free fetal DNA (cfDNA) from maternal plasma is now increasingly offered as a screening test for fetal aneuploidy
  - Circulating cell-free fetal DNA comprises ~ 3-13% of total cell free maternal DNA
  - Detection of trisomy 21, 18 & 13
  - Offers parental reassurance
  - Decreases the risk of pregnancy loss from invasive procedures

Fetal anomaly detection

- Which anomalies can we reasonably expect to detect prior to 14 weeks?

sonoembryology

Is this normal?

Normal 12 week

- The choroid filled lateral ventricles dominate the intracranial image at 11-14 weeks
  - Thin brain mantle
- Hemispheres should appear symmetric
  - Separated by a clearly visible falx

Exencephaly-anencephaly

- No calvarium
- Disorganized angiomatous stroma above the orbits
  - Progressively destroyed by chemical & mechanical forces
- Lethal
  - Multifactorial etiologies
encephalocele

- Defect in bony skull & dura
  - Protrusion of intracranial structures
  - Diverse appearance depending on herniated content
  - Associated with multiple syndromes
    - Meckel-Gruber most common (encephalocele, polycystic kidneys, polydactyly)
**holoprosencephaly**

- Complex malformation
  - Single ventricle
  - Fused thalami
  - Facial anomalies in ~ 70%

Continuum
- Alobar --- semilobar --- lobar

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**Early evaluation of the chest**

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**Early evaluation of fetal anatomy**

- 12 weeks 6 days

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**Early evaluation of fetal anatomy**
Congenital diaphragmatic hernia

CDH: sonography

- Cystic mass in the chest
- No stomach “bubble” in the abdomen
- Deviated heart
  - Abnormal cardiac axis may be the only clue

CDH: associated anomalies

- Present in about 50% of cases
  - Central nervous system: 30%
  - Cardiac: 20%
  - Renal
  - Spinal
- Aneuploidy
  - Up to 1/3 of cases

Congenital diaphragmatic hernia

- Pulmonary hypoplasia is worse than from other chest masses of comparable size
  - Hypoplasia is always present
  - Small histologically immature lungs
  - Pulmonary hypertension
    - Muscular hypertrophy of arterial walls
- If isolated without liver herniation, survival is ~80%
Sonoembryology: Mid-gut herniation, 9w 1d

Mid-gut herniation
- Normal embryologic process
- Due to rapid growth of midgut during the 1st trimester
- Bowel returns to abdomen by 11-12 weeks
- Liver never herniates

Physiologic mid gut herniation

omphalocele
10 weeks

Gastroschisis
12 weeks 4 days

Gastroschisis
17 weeks 4 days
gastroschisis

Early evaluation of fetal anatomy

Cloacal extrophy

• Low abdominal wall defect
• Absent bladder

Early evaluation of fetal anatomy

Omphalocele with thickened NT

Early evaluation of fetal anatomy

Single ventricle
Trisomy 18
omphalocele
14 weeks 2 days

Beckwith-Wiedemann

Beckwith-Wiedemann Syndrome
• Best diagnostic imaging clues
  – Large for dates fetus
  – Enlarged kidneys
  – omphalocele
  – Macroglossia
    • Protruding tongue

Beckwith-Wiedemann
• In utero
  – Polyhydramnios
  – Maternal risk of preeclampsia
  – Preterm delivery
• Neonatal period
  – Neonatal airway obstruction if severe
  – hypoglycemia

Early evaluation of GU tract
• Megacystis at 10-14 weeks
  – ~ 50% resolve
  – Association with aneuploidy
  – Increased incidence of obstructive uropathy
• Normal bladder length < 6 mm
  – Mild megacystis 7-11 mm (Grade 1)
  – Moderate megacystis 12-15 mm (Grade 2)
  – Severe megacystis > 15 mm (Grade 3)

megacystis
• Increased incidence of aneuploidy
  – Often increased Nuchal Translucency
  – Usually mild-moderate bladder size
• Early manifestation of lower urinary tract obstruction
  – Often severe increase in bladder length
megacystis

marked megacystis

Thickened NT

Megacystis + thickened NT

34 year old patient

11 weeks 3 days

Anterior abdominal wall defect
extremities

Body stalk anomaly
- Lethal malformation characterized by attachment of visceral organs to the placenta
  - Short/absent cord
- Thoraco-abdominal wall defect
- Scoliosis
- Limb defects

Amniotic band syndrome
- Thought to result from early rupture of the amnion with subsequent entrapment of fetal structures by the bands originating from the chorionic side of the amnion which are “sticky”
- Ischemia resulting from the constriction may lead to amputation
- Bizarre asymmetric defects

Early evaluation of Musculoskeletal System

Normal extremities
11 weeks 2 days
Short-rib polydactyly syndrome

- Group of rare lethal osteochondrodysplasias
- Autosomal recessive
  - Transvaginal imaging in high-risk families
  - Postnatal confirmation of diagnosis important for recurrence risk counselling if new diagnosis

Early evaluation of Musculoskeletal System

12 week 2 days

VACTERL

- Vertebral
- Anal atresia
- Cardiac
- Tracheo-esophageal fistula
- Esophageal atresia
- Renal
- Limb
VACTERL
• Nonrandom association of 7 core abnormalities

• Defective differentiation of mesoderm prior to day 35
  – Mechanism unknown
  – Euploid

triploidy
• 69 chromosomes
  – Entire extra haploid set
  – 75% maternal
  – 25% paternal (partial mole)
• Early severe IUGR
• Multiple anomalies
  – Ventriculomegaly
  – Cardiac anomalies
  – Cystic hygroma
conclusions

• A comprehensive sonogram in the first trimester provides an evaluation of both the pregnancy and the maternal pelvis
  – Consequent improvement in obstetric care
• An awareness of normal anatomy in the 1st trimester is necessary
  – Even if the study is performed for other reasons

conclusions

• Anomalies evident in the 1st trimester are often
  – More complex
  – Higher association with aneuploidy
  – Increased likelihood of hydrops
  – May be transient

conclusions

• In the uncomplicated pregnancy without clinical concerns, the 1st trimester scan should be scheduled at ~11-14 weeks
  – This provides the opportunity to evaluate fetal anatomy as well as confirming gestational age, viability and determining fetal number

conclusions

• Earlier diagnosis offers many advantages
  – Patient learns of the fetal problem with sufficient time to consider management options
  – Terminations earlier in pregnancy are safer & cheaper
  – Less traumatic for the mother both psychologically & physically with a greater likelihood of preserving patient privacy
  – Availability of termination in the latter half of the 2nd trimester is soon to be less available

conclusions

• Cannot overemphasize the value of parental reassurance in patients with a previous history of a fetus with a major anomaly or genetic abnormality

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