Case Studies in High Risk Obstetrics

Charlotte Henningsen, MS,RT(R),RDMS,RVT,FSV,FAIUM
Chair & Professor - Sonography Department
Adventist University of Health Sciences

Objectives

- Describe a variety of obstetrical anomalies and the sonographic findings that may be identified during a sonographic examination
- List pertinent risk factors and laboratory data associated with specific fetal anomalies
- Discuss pregnancy management and possible outcomes related to specific anomalies

Ectopic cordis
- Partial or complete displacement of heart
- Associated with ABS, LBWC, heart defects, abdominal wall defects
- Poor prognosis

Pentalogy of Cantrell
- Ectopia cordis
- Abdominal wall defect
- Diaphragmatic defect
- Pericardial defect
- Heart defect
- Poor prognosis

Amniotic Band Syndrome
- ADAM complex (amniotic deformities, adhesion, mutilation)
- Range of malformations from minor constrictions to complex and bizarre anomalies attributed to bands that stick, entangle and disrupt fetal parts

Trisomy 13
- AKA: Patau syndrome
  - Dr. Klaus Patau
  - 1:5000 births
- Profoundly retarded
- 85% die w/in 1 year

Sonographic Findings

- Holoprosencephaly
  - Facial anomalies
- Limb anomalies
- Heart defects
- Anomalies of other organ systems
Ulnar Ray Defects
- Less common than radial ray defects
- Hypoplasia more common than complete absence
- Associated with syndromes
  - Ulnar-Mammary Syndrome
  - TAR Syndrome

Achondroplasia
- Most common - non-lethal
- 1:66,000 in U.S.
- Autosomal dominant or spontaneous (80%)
- Risk factor: inc. age

Sonographic Findings
- Rhizomelia
- Limb bowing
- Short fingers
- Macrocephaly
- Frontal bossing

findings cont.
- Hydrocephaly
- Saddle nose
- Head/FL discrepancy
- Biometry may be normal <22wks

Anencephaly
- Absence of brain and calvarium
- Most common NTD
- 1:1000 pg in U.S.
- Result of failure of closure of neural tube

Sonographic findings
- “frog-like” appearance
- Absence of cranial vault cephalad to orbits
- Absence of brain
- Vascular malformation
- Polyhydramnios
Acrania
- AKA: exencephaly
- Absence of calvarium
- Abnormal brain
- May progress to anencephaly

Cystic Hygroma
- 1:6000
- Congenital malformation of lymphatics
- Often arise in neck
- Assoc w/ aneuploidy

Sonographic Findings
- Single or multiloculated
- Fluid-filled cavity
- Usually @ post/lat of neck
- Assoc. hydrops

Trisomy 21
- AKA: Down syndrome
- Most common aneuploidy
- Moderately retarded
- 15-20% die in first yr

Sonographic Findings
- Hypoplasia of middle phalanx of fifth digit
- Increased nuchal fold
- Heart defects
- Duodenal atresia
- Short humerus/femur

Dandy-Walker Complex
- Dandy-Walker malformation
- Dandy-Walker variant
- Mega-cisterna magna
- Rare
- Associated with syndromes, abnormal chromosomes, teratogens, intracranial and extracranial anomalies
Sonographic findings
- Enlarged cisterna magna (posterior fossa cyst)
- Splaying of the cerebellar hemispheres
- Absent cerebellar vermis
- Hydrocephalus frequent

Bilat Renal Agenesis
- Most severe GU anomaly
- 1:3000 – 10,000 live births
- Males > Females
- 5% recurrence

Sonographic Findings
- Severe oligo
- Failure to ID kidneys
- Failure to ID bladder

Hydranencephaly
- Result of vascular accident resulting in massive brain destruction
- 1:10,000 births
- Multiple theories including: congenital infections, ischemia, bilateral occlusion of ICA

Sonographic Findings
- Replacement of cerebral hemispheres by cystic cavity
- Intact brain stem
- Normal size, macrocephaly or microcephaly

Trisomy 18
- AKA: Edwards syndrome
  - John H Edwards
- 3:10,000 births
- Profound retardation
- 90% die w/in 1yr
Sonographic Findings

- Clenched hands
- Choroid Plexus cysts
- Anomalies of every organ system

Encephalocele

- Hemiation of meninges or meninges and brain through a defect in the calvarium
- Most defects are midline in occipital region
- Associated with syndromes and abnormal chromosomes
- Prognosis poor when defect includes brain

Limb-Body Wall Complex

- AKA: Short Umbilical Cord Syndrome, Body-Stalk Anomaly
- Result of failure of closure of the ventral body wall, limb reductions, absent umbilical cord

Sonographic Findings

- Short or absent umbilical cord
- Scoliosis
- Thoracoabdominoschisis
- Limb defects
- Facial clefts
- CNS anomalies
- Amniotic Bands

Omphalocele

- Abdominal viscera herniated through a midline defect into base of umbilical cord
- Liver commonly involved
- May contain small or large bowel

US Findings

- Anterior wall defect in midline
- Enclosed by membrane
- Continuous with umb cord
- Ascites
- Polyhydramnios
**Gastroschisis**
- Hemiation of abdominal contents thru an off-midline defect
- Usually to right of cord
- No genetic association
- Defect usually includes small or large bowel, rarely liver
- Not covered by a sac

**US Findings**
- Free-floating bowel loops in amniotic fluid
- Cord insertion to left of defect
- May mimic omphalocele
- Decreased abdominal size
- Polyhydramnios
- Near term, bowel in amniotic fluid may appear matted and dilated

**AMC**
- Arthrogryposis Multiplex Congenita
- AKA: amyoplasia, fetal akinesia deformation sequence
- Multiple congenital contractures
- Possible etiologies: CNS disorder, muscular system disorder, connective tissue/skin disorder
- Associated with numerous syndromes

**Sonographic findings**
- Absent or scant fetal movements
- Persistent contractures
- Postural abnormalities
- Polyhydramnios
- Associated anomalies
- Associated with increased NT in first trimester

**Crossed-fused Ectopia**
- Kidney located opposite to ureteral insertion into bladder
- Fused ≠ not fusion
- Associated with
  - Imperforate anus
  - Skeletal anomalies
  - Heart anomalies

**ARPKD**
- Inherited disorder which causes the development of multiple cysts, enlarging kidneys and reducing function
- 1:20,000 live births
- Associated with mutations of PKHD1 gene
US Findings
• Enlarged, echogenic kidneys
• No CMD
• Unable to ID Bladder
• Severe oligohydramnios

Cystic Renal Dysplasia
• Occurs w/early obstruction of the GU tract
• Decreased renal function
• Usually bilat

Associated with
• VACTERL association
• Chromosomal anomalies
• Syndromes

Spina Bifida
• AKA: Myelomeningocele
• Result of failure of closure of neural tube
• Range of severity

US Findings
• Splaying of post. ossification ctr
• Cleft in soft tissue
• Bulging sac
• Ventriculomegaly
• Chiari II malformation
• Banana sign
• Lemon sign

Holoprosencephaly
• 6-12:10,000 live births
• Result of varying degrees of fusion of forebrain and cerebral structures
• Range of severity: Alobar, Semi-lobar, Lobar
AMC
• Arthrogryposis Multiplex Congenita
• AKA: amyoplasia, fetal akinesia deformation sequence
• Multiple congenital contractures
• Possible etiologies: CNS disorder, muscular system disorder, connective tissue/skin disorder
• Associated with numerous syndromes

Sonographic findings
• Absent or scant fetal movements
• Persistent contractures
• Postural abnormalities
• Polyhydramnios
• Associated anomalies
• Associated with increased NT in first trimester

Prognosis
• Dependent on severity
• Termination may be offered
• Respiratory complications may be lethal