Mimics of Syndromes

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Congenital Anomalies

10% Newborn
Multiple Congenital Anomalies

Increase Risk of Chromosomal or Genetic Syndrome
High Risk

Major Structural
- CNS
- Face
- Extremities
- Genitourinary
- Heart

Markers
- Echogenic bowel
- Choroid plexus cyst
- Mild renal pelvocalycectasis
- Echogenic intracardiac papillary muscle
- Nuchal Thickening
Multiple Anomalies

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Chromosomal/Syndrome
Case 1 - 25 weeks
Diagnosis?

• Chromosomal - 25-50%
  – Trisomy 13 - 75%
  – Triploidy - 20%
  – Trisomy 18-1-2%

• Single gene mutations - 15-20%
Diabetic Embryopathy

- 10% fetal anomalies diabetic patients
  - Highest in diabetes type 1
  - Can occur in gestational diabetes

- Glucose
  - Interrupts normal cellular metabolism and signaling

- Inversely related gestational age
  - Blastogenic
    - First 4 weeks
Breakdown

- CNS
  - Anencephaly
  - Holoprosencephaly
  - Exencephaly
  - Encephalocele
  - Microcephaly

- Craniofacial
  - Cleft lip
  - Cleft palate
  - Hemifacial
  - Microtia
  - Micrognathia
  - Eye defects
Breakdown

- Cardiac
  - Hypoplastic left heart
  - Transposition
  - Tetralogy of Fallot
  - Septal defects
  - Coarctation
  - Heterotaxia

- Skeletal
  - Sacral agenesis
  - Limb defects
  - Vetebral defects
  - Caudal regression

Heart and Skeletal – VACTERL minus TEF
22 Weeks – Hypoplastic left heart
Case 2 - 29 weeks
Diagnosis?

- Infection
  - CMV

- Genetic?
Congenital Infection

- 2-3% of congenital anomalies

- Nearly 20% of fetal neonatal disease
CNS Infections

- Cytomegalovirus (CMV)-2.2%
- Rubella
- Parovirus
- Toxoplasmosis
- Herpes Simplex
Anomalies with Infection

- Echogenic bowel
- Ascites/hydrops
- Hepatosplenomegaly
- Cardiac anomalies
- CNS
  - Microcephaly
  - Ventriculomegaly
  - Intracranial calcifications/cysts
  - Migrational anomalies
Intracranial Calcifications Infection

- CMV
- Toxoplasmosis
- Rubella
- Herpes Simplex
- Varicella
Differential

- Aicardi Goutieres
- Pseudo TORCH
  - Autosomal recessive inheritance
Case 3 – 22 weeks
Diagnosis

- Thanatophoric Dysplasia
  - Most common .5 in 10,000 births
  - Fibroblast Growth Factor Receptor 3 gene (FGFR3)
    - Temporal lobe dysplasia
      - Hypochondroplasia
      - Achondroplasia
Differential

- Short bones
  - Skeletal dysplasia (< 5<sup>th</sup> percentile or >3 SD below mean)
  - IUGR
  - Chromosomal
    - Trisomy 21 – short femur and humeri
- Normal
  - Asian and Hispanic
Skeletal Dysplasia

- 1 per 5000 live births
- Types
  - Osteodysplasia- bone density/mineralization
  - Chondrodysplasia- cartilage
  - Dysostosis- blastogenesis
Skeletal Dysplasia

- Rizomelia
- Mesomelia
- Micromelia
- Acromelia*
Skeletal Dysplasia

- Achondrogenesis (langer-Saldino)
- Achondroplasia
- Asphyxiating Thoracic Dystrophy
- Campomelic
- Thantophoric Dysplasia
- Cysts liver and pancreas
- Craniofacial anomalies
- Cloverleaf skull
- Frontal bossing
- Hydrops
Case 4 - 26 weeks
Diagnosis?

- Limb Body Wall
- Differential
  - Amniotic band syndrome
  - Pentology of Cantrell
  - Ruptured Omphalocele
  - Large gastroschisis
  - Cloacal extrophy/OEIS
    - Omphalocele
    - Exstrophy
    - Imperforate anus
    - Spinal defects
Limb Body Wall

- Neural tube defect
- Thoraco or Abdominoschisis
- Limb anomalies
- Abnormal short umbilical cord
Limb Body Wall

• 1 in 7500 pregnancies
• Cause
  – Amniotic band
  – Vascular disruption
  – Abnormal embryonic folding
Case 5 – 21 weeks/Monochorionic Twins
Diagnosis?

- Turner Syndrome
- Trisomy 21
- Twin Reversed Arterial Perfusion Syndrome
- Noonan Syndrome
- Congenital Cardiac Disease
Twin Reversed Arterial Perfusion

- 1% of monochorionic twins
- Abnormal placental arterio-arterial and veno-venous anastomosis
  - Diagnosis retrograde flow in the artery of abnormal twin
Twin Reversed Arterial Perfusion

- Acardiac - 75%
- Acephalic
- Absent upper extremities
- Abnormal development of abdomen
- Hydrops
Multiple Anomalies---Differential

- Chromosomal/Genetic Syndrome
- Metabolic
- Infectious
- Dysplasia
- Acquired
- Vascular