An Approach To Fetal Skeletal Syndromes using 2D and 3D

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Be A detective

Identifying syndromes correctly requires becoming a detective:

The Sentinel Feature Method

- Once you find a malformation, form a list of possible syndromes.
- Then, look for the one additional feature that is most "sentinel" to each syndrome on your list, to identify the correct one.
- Then, look at the pattern of malformations to make sure you have arrived at the most likely Dx.

POLYDACTYLY

- Meckel Gruber (post axial cystic kidneys & encephalocele)
- Trisomy 13 (holo, face, heart, mult.)
- Short rib polydactyly (cleft type 2)
- Carpenter (craniosyn.-preaxial)
- Mohr (oro/facial/dig)- pre & post axial & face abn - cleft)
- Familial (√fam hist)

Pre and Post-axial polydactyly
Meckel Gruber

• 3D in the first trimester
• Imaging the fetal digits at 9 weeks

10 weeks Polydactyly and acrania

Carpenter’s Syndrome

Majewski Syndrome - SRPDS type 2
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**SHORT RADIAL RAY**

- VATER (vert, anal, TE, renal, radial)
- TAR (Thrombocytopenia Absent Radius)
- Holt-Oram (heart defect + dominant)
- Amniotic Bands (asymmetry)
- Fanconi's Anemia (no thumb)
- Trisomy 18 (mult anom, heart etc…)
- Nager (severe micrognath & ear abn.)

VATER

Nager syndrome 28 wk – prior affected child

NAGER

One volume

Holt Oram Syndrome
Clenched hand or Syndactyly

- Trisomy 18 (heart, feet, overlapping fingers)
- Pena-Shokier (poor activity, hypertelorism)
- Arthrogryposis (contractures, no movement)
- Cerebro-oculo-skeletal (cataract, microphthalmia, ACC, microcephaly, clubfoot)
- Smith-Lemli-Opitz (toes 2-3, cryptorchidism, hypospadias, microcephaly)
- Triploidy (syndactyly 3-4 and IUGR)
- Apert's (mitten hand)
- Amniotic Band syndrome (asym)
- EEC (claw hand & cleft)
HEMIVERTEBRAE

• Jarco-Levin (+ syndactyly, short spine)
• Sirenomelia sequence/caudal regression (kidneys, legs, mvmnt)
• Klippel-Feil (neck & face)
• MURCS (mullerian, renal aplasia, C5-T1)
• VATER (vert, anal, TE, renal, radial)
• Poland S. (absent pectoralis, upper spine)
• Isolated vertebral body defect (most common)
Sacral Agenesis

Sirenomelia

Short Limbs

• Achondrogenesis (loss spine, shortest limbs)
• Diastrophic Dysplasia (hitch thumb)
• Osteogenesis Imp. (loss skull)
• Thanatophoric (teleph fem, S shape legs, platyspondyly)
• Camptomelic Dysplasia (bowed legs, absent fibula, vestigial scapula)
Diastrophic Dyspl. - low thumb

Osteogenesis Imperfecta Congenita

OI type 1 - autosomal dominant - affected mother

The Sentinel Feature

- Fetal ultrasound specialists are fetal dysmorphologists and detectives.
- Once you see an anomaly, make a list of possible syndromes. Then for each syndrome, look for the sentinel or distinctive feature that is characteristic enough to help narrow your list to the most likely syndrome.
- Then, use the pattern of all the malformations you see to arrive at a specific diagnosis - most of the time, you will be right!!.

How to Detect syndromes

- Assume that each fetus is abnormal until proven otherwise.
- Check every anatomical landmark on all fetuses during each scan.
- Look for patterns, understand the timing of when malformations appear, and when uncertain, get a follow-up.

Thank you for your attention!