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 (horo, 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

T13
SHORT RADIAL RAY

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

VATER

Holt Oram Syndrome

Trisomy 18 arms

Clenched hand or Syndactyly

- **Trisomy 18** (heart, feet, overlapping fingers)
- **Pena - Shokier** (poor activity, hypertelorism)
- **Arthrogryposis** (contractures, no mvmt)
- **Cerebro-oculo-skeletal** (cataract, microphthalm, ACC, microceph, clubfeet)
- **Smith Lemli Opitz** (toes 2-3, cryptorch, hypospadias, microceph)
- **Triplody** (syndact 3-4th & IUGR)
- **Apert’s** (mitten hand)
- **Amniotic Band syndrome** (asym)
- **EEC** (claw hand & cleft)
Trisomy 18

Arthrogryposis

Cerebro-oculo-skeletal

Syndactyly
Triploidy at 13 wks

Amniotic Bands

EEC 17 wks
HEMIVERTEBRAE

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

Sirenomelia

Short Limbs

- Achondrogenesis (oss spine, shortest limbs)
- Diastrophic Dysplasia (hitch thumb)
- Osteogenesis Imp. (oss skull)
- Thanatophoric (teleph fem, S shape legs, platyspondyly)
- Camptomelic Dysplasia (bowed legs, absent fibula, s'capula)
Thanatophoric Dysplasia
Achondrogenesis

Diastrophic Dysplasia - 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!