Fetal anomalies of the hands and feet

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Introduction

• Limb malformations:
  0.6 % fetal US
  0.49 – 3.5/10000 live births
  Upper > lower 2:1
  Uni > bilateral 4:1

• Difficult to recognize, often overlooked
Introduction

- Spectrum:
  - simple / complex
  - isolated / associated

- Importance of detection:
  - functional & psychosocial impact
  - diagnostic & pronostic significance
May be the clue for a specific Δ

Acrocephalosyndactyly, Pfeiffer type
Objectives

• To emphasize the necessity of a systematic examination

• To review the normal appearance

• To discuss major hands & feet anomalies:
  significance
  prenatal work-up
  differential diagnosis
US examination: when?

- Best timing: late 1<sup>st</sup> trim – early 2<sup>nd</sup> trimester
- 3<sup>rd</sup> trim:
  - closed hands
  - less relative amniotic fluid
  - hidden parts
US examination: how?

• Systematically proximal – distal
4 limbs
presence, morphology
position, motion
proportions

• 2D abdo /transvag, 3D complementary, (MRI)
Normal appearance

- 8-9 wga: limb buds
- 10-11 wga: appendicular long bones, phalanges
- 12-13 wga: metacarpals, metatarsals
- 20-24 wga: calcaneum, talus

- Other ossification centers after birth

(Mahony, J Ultrasound Med 1984)
Premature appearance of ossification centers

• **Diffuse:**
  - chondrodysplasia punctata
  - Warfarin

• **Tarsal location:**
  - N (artifact)
  - aneuploidies
  - IUGR

19 wga, chondrodysplasia punctata
Condradi-Hunermann
Hands & feet anomalies

- aN morphology
- aN number
- aN size
- aN motion
- aN alignment

(Rypens, Radiographics 2006)
Absence: e.g.: amniotic constriction band

- 1/56 fetuses – 1/1200 live births
- Unknown etiology:
  - Intrinsic germline aN
- ? Vascular disruption event
  - Early amnion rupture
- Disruptions, deformations & malformations:
  - Simple groove → amputation
  - acrosyndactyly
  - cranial defects, body wall defects…

(Goldfarb J Bones & Joint Surg 2009)
Amniotic constriction band

- ASYMMETRICAL defects
- > 1 limb
- Rare loose adhesive bands
- Usually sporadic
- ΔΔ:
  Teratogens (thalidomide, warfarin, phenytoin, cocaine, misoprostol, CVS, ...)
  Developmental symbrachydactyly
  Syndromes: Adams-Oliver, EEC, clefts with bands...
Alignment anomalies

- Club foot
- Rockerbottom foot
- Club hand
- Clenched hand
- Camptodactyly
- Clinodactyly
- Phocomelia
- ....
Club foot: equinovarus talipes

- Hind foot equinus & varus
  Forefoot adduction +/- cavus
- 1/1000 live births (N Am, Europe)
  (Asia: 0.4 – Hawaï: 7)
- M/F 2:1
- Bilat +/- 50%

Club foot: diagnosis

- Tibia, fibula, foot in frontal view
- CONSTANT
- Ankle angulation
- Abnormal foot print
- +/- calf thinning
Club foot: multifactorial etiology

- Heredity (familial 16 - 25 %)
- Neuromuscular (CNS, arthrogryposis)
- Mechanical (crowding)
- Teratogens (DM, fever, early amniocentesis)

- Often unknown

Club foot: management

- Prognosis: associated anomalies
- Detailed fetal US
- Karyotype:
  YES if associated / complex
  ? If isolated
  (0 – 3.6 % aneuploïdies: T18, sex chrom)

- If isolated: no correlation with postnatal severity
Club foot: differential

- Normal (transient, early 1\textsuperscript{st} T, late 3\textsuperscript{rd} T)
- Crowding (up to 10 \% false +)
- Metarsus adductus
- More complex leg anomaly
- Rockerbottom foot

\textbf{→} look at hindfoot, plantar sole, leg, dynamics
Rockerbottom foot

- Prominent heel
- Rounded bottom of foot
- +/- dorsiflexed 1st toe
- Best seen in lateral view
- Frequently associated:
  - T 13, T 18
  - neural tube defects
  - dysplasia, Potter
Clubhand

= permanent wrist deviation

Akinesia

Abnormal radius > ulna

(ΔΔ: direction, forearm)

12 wga
Radial clubhand

- Rarely isolated and sporadic
- Usually associated with
  - Aneuploidy: T 18, T 21, 13 del, ring chrom 4
  - VACTERL
  - Radial hypoplasia sequence (thumb, metacarpal, trapezium, scaphoid, humerus)
  - Hematologic disorders: Fanconi, TAR, Aase
  - Syndromes: Holt-Oram, Cornelia de Lange
  - Teratogens: DM, valproate...

( Kennelly Prenat Diagn 2007)
Radial club hand: management

- Detailed fetal US
  Uni / bilateral*
  Isolated / associated*

- Thumb!
  Fanconi: ⚫
  TAR: +
  Aase: triphalangeal
Radial club hand: management

- Cardiac US
  (TAR, Aase, Holt-Oram, Cornelia de Lange)
- Genetic counsel
- Fetal blood sampling
- Chromosomal breakage study
  (Fanconi, Roberts)
- Specialized center
Clenched hand

- Index (and 5th digit) overlaps a clenched fist formed by the other digits
- Index IPP flexed & ulnarly deviated
- Thumb adducted
- « constant »

![Image of clenched hand and ultrasound images]
Clenched hand

- Strongly suggestive of T 18 (40 %)
- When T 18 suspected: look at hands & feet, heart radial ray sequence, clenched hand, club foot, rockerbottom foot
- T 18: great phenotypic variability
  1\textsuperscript{st} T low serum markers
  3\textsuperscript{rd} T: IUGR, polyhydramnios, hand aN
Clenched hand: differential

- T 18, T 13, T 9
- Akinesia / hypokinesia
- Normal (transient!)
- Adducted thumbs (X-linked aqueductal stenosis)
Camptodactyly

- = flexion contracture of the PIP
- frequently asymmetrical
- may be isolated (late progression)
- ! when severe, multiple digits
  → Karyotypic aN (T18, 13, 15)
  → Contracture S
Clinodactyly

= fixed abnormal radial deviation of the DIP
Small 2nd phalanges
- Up to 60 % of T21
- Familial and usually isolated (AD)
- 18 % of normal population

Not significant if isolated
→ search for associated aN

Trisomy 21 - Hand Features
Fetal akinesia deformation sequence

- = arthrogryposis, Pena Shokeir, amyoplasia...
- 3/10000 live births

- Secondary to \( \downarrow \) motion (7 – 8 wga)
- Not a specific disease but an end-result
  - neurologic, muscular, connective tissue, skeletal, crowding...

- Causes: infection, drugs, toxics, FAS, heredity…
FADS: diagnosis

- Absence of fetal movement
- Severe flexion / extension deformities
- Usually generalized, symmetric
- More severe distally
- Lower > upper limbs
- +/- polyhydramnios, lung hypoplasia

ΔΔ: T 18
Number anomalies

- Polydactyly
- Oligodactyly
- Syndactyly
- Ectrodactyly
- Polysyndactyly
- ....
Polydactyly

- The most common hand anomaly
- 1/683 pregnancies

Pre-axial (radial / tibial)

Central

Post-axial (ulnar / fibular)

(Zimmer Am J Obstet Gynecol 2000)
Polydactyly

- **Incomplete:**
  - soft tissue
  - often isolated
  - often overlooked
  - autoamputation

- **Complete (bone):** more often associated
Post-axial polydactyly

- More frequent (afro-american)
- Usually isolated (AD, favorable)
- Can be associated with:
  - T13
  - Meckel-Gruber
  - Bardet-Biedl
  - Smith-Lemli-Optiz
  - SRPS…

Meckel-Gruber Syndrome
Pre-axial polydactyly

- Less frequent
- Possibly syndromic:
  - SRPS
  - Carpenter S
  - T21
  - VACTERL
  - Diabetes mellitus
- ! « thick » thumb, great toe
Syndactyly

- = abN connection between adjacent digits (moving together)
- Simple (soft tissue) / complex (bone)
- Complete / incomplete
- Isolated / complicated
- Often 2nd – 3rd digits
- Toes: very difficult to Δ
Syndactyly

2-3 / 10000 live births
Familial occurrence (AD, isolated, incomplete penetrance, variable expressivity)

! Complex:
acrocephalosyndactylies (Apert)
Poland
Ectrodactyly (lobster-claw deformity)

- long deficiency of the central digit(s) & meta
- V / U shaped cleft
- +/- syndactyly, aplasia, hypoplasia
- Isolated
- Associated:
  - EEC S
  - Roberts S
Circumstances of discovery

- Targeted scan:
  family history, teratogens (DM, drugs)

- Other fetal anomalies:
  IUGR, lg bone shortening
  CNS anomalies, craniostenosis…

- Fortuitous

24 wga, long bone shortening, ambiguous genitalia, feet!
→ Smith Lemli Opitz
Management: multidisciplinary

- Thorough fetal examination (! Thumb)
- Cardiac US
- Repeated focused US / 3D US / MRI (CNS)
- Genetic counseling (familial inquiry, karyotype,..)
- Specific examinations (FBS, chromosome breakage studies)
- Orthopedic, plastic surgeons, psychologist,..
Please, look at the hands & feet!

Thank You!