A Helping Hand: Congenital Anomalies of the Upper Extremity

Matthew Winfeld, MD
Perelman School of Medicine
University of Pennsylvania
I have no disclosures
Objectives

- Provide an overview of the spectrum of congenital upper extremity anomalies
- Describe the key imaging findings of these abnormalities
- Discuss the important clinical features of these entities
Overview

- Not enough bones (deficiencies)
- Extra bones
- Fusion/failure of separation
- Malalignment
- Abnormal bone morphology
- Big bones/soft tissues
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Case – 1 month-old boy
**Severe Truncation: Roberts Syndrome**

- **Phocomelia**: deficiency/absence of the proximal-mid extremity
- Autosomal recessive
  - ESCO2 gene involved in chromosome separation
- Rare; 150 cases reported
- All extremities affected
- Multisystem disorder
Case – 6 month-old girl
Thrombocytopenia-Absent Radius

- Absent radii, (near) normal thumbs
- Possibly more extensive upper limb and even lower limb deficiencies
- Autosomal recessive, 1q21.1, 1:1,000,000

Thrombocytopenia
- Severe at birth; bleeding complications
- Transfusion requirements decrease by age 1
- Normalization of platelet count by adulthood
Case – 1 year-old girl
Case – 1 year-old girl
Holt-Oram Syndrome

- Radial anomaly most commonly limited to the thumb
- Autosomal dominant 12q24.1, 1:100,000
- Cardiac manifestations
  - Septal defects (atrial most common)
  - Pulmonic stenosis
Case – 16 year-old boy
VACTERL

Vertebral
Anal
Cardiac
Tracheoesophageal fistula
Esophageal atresia
RADIAL/Renal
Limb
- incidence 16/100,000
Longitudinal Deficiencies: Radial

- Tend to be associated with other non-MSK abnormalities/syndromes
Longitudinal Deficiencies: Radial

- Goldfarb et al. (2006)
  - Reviewed 56 years of surgical data (Wash U)
  - 146 patients with radial deficiencies
  - 55 syndromic

Table 5. Common Syndromes and Associations in the 164 Radial Longitudinal Deficiency Patients

<table>
<thead>
<tr>
<th>Syndrome</th>
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<tbody>
<tr>
<td>TAR</td>
<td>25</td>
</tr>
<tr>
<td>VACTERL</td>
<td>22</td>
</tr>
<tr>
<td>Holt-Oram</td>
<td>7</td>
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<tr>
<td>Fanconi anemia</td>
<td>1</td>
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<tr>
<td>Total</td>
<td>55</td>
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Thumb Hypoplasia

- Goal is to preserve thumb function to be able to perform basic functions (i.e., grab objects, turn doorknobs)
- Reconstruction vs. pollicization
- Key finding: Integrity of carpometacarpal joint
Thumb Hypoplasia: Pollicization

1 year old boy

At 2 years old
Longitudinal Deficiency: Ulnar

- Usually **non-syndromic**
- **Thumb hypoplasia** often coexists
  - Does **NOT** confer a higher risk of associated syndromes/non-MSK malformations
  - If **radius** is hypoplastic/aplastic, classify as radial ray anomaly

1 yo M

24 do F
Case – 11 month-old boy
Central Longitudinal Deficiency: Cleft Hand

- Formerly “typical” cleft hand
- Characteristic “V” configuration
- Autosomal dominant, bilateral
- Also involves the feet
- Associated with cleft lip/palate
Symbrachydactyly

- Short hand with underdeveloped, often webbed digits; “U” shaped
- Formerly “atypical” cleft hand, however no longer believed to be a true central longitudinal deficiency
- Sporadic, unilateral
Case – 7 year-old boy
Poland Syndrome

- Symbrachydactyly + chest wall hypoplasia secondary to in utero vascular disturbance
Overview

- Not enough bones (deficiencies)
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Case – 13 month old girl
Radial (Pre-Axial) Polydactyly

- Sporadic
- 1:3000 live births
- Most common iteration is bifid first metacarpal with duplicated phalanges
- Goal of management is functional thumb
  - Report relative lengths of components and widths/angulation of articular surfaces
More prevalent in people of African descent (1:143)
- Autosomal dominant
- If seen in Caucasian child, higher probability of underlying skeletal dysplasia
  - Ellis van Creveld
  - Chondroectodermal dysplasia

27 day old girl
Ulnar (Post-Axial) Polydactyly

- Management much simpler than radial as function is unlikely to be affected

27 day old girl
Ulnar (Post-Axial) Polydactyly

- Management much simpler than radial as function is unlikely to be affected.
- Rudimentary nubbins can be tied off and will necrose and fall off.

5 month-old girl
Overview

- Not enough bones (deficiencies)
- Extra bones
- **Fusion/failure of separation**
- Malalignment
- Abnormal bone morphology
- Big bones/soft tissues
Case – 9 year-old girl
Case – 9 year-old girl
Apert Syndrome

- Acrocephalosyndactyly syndrome (also Crouzon, Pfeiffer)
- Autosomal dominant
- Syndactyly is characteristic, though severity varies (spade hand, mitten hand, rosebud hand)
Syndactyly

Classifications

- Complete vs incomplete
- Simple (soft tissue only) vs. complex (bone)
- Complicated (syndromic) vs. uncomplicated

Case – 3 year-old girl
Radioulnar Synostosis

- Limits pronation/supination
- Most often sporadic, but can be inherited (AD)
  - Association with sex chromosome duplication
- Type I
  - Proximal fusion 2-6 cm
  - Absence of radial head
Radial head present but dislocated
- Fusion further distal
- Sporadic
- Also limits elbow flexion
Overview

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Case – 4 year-old boy
Clinodactyly

- Fixed interphalangeal malalignment in the radial-ulnar plane
- May be associated with “delta” middle phalanx
- Inherited/isolated or syndromic
COMMON
1. Brachmann-de Lange S. (de Lange S.)
2. Ehlers-Danlos S.
3. Familial brachydactyly A1, A2, A3, A4, C
4. Fanconi anemia (pancytopenia-dysmelia S.)
5. Fetal alcohol S.
6. Fibrodysplasia (myositis) ossificans progressiva
7. Hand-foot-genital S.
8. Holt-Oram S.
9. [Kimer deformity (distal phalanx - seen as isolated anomaly or in Brachmann-de Lange S.)
10. Klinefelter S. (XXY S.)
11. Local disorder (eg, trauma; arthritis; contracture)
12. Marfan S.
13. Metaphyseal chondrodysplasia (Shwachman type)
14. Mitral valve prolapse S. (MVPS)
15. Nail-patella S. (osteo-onychodysplasia)
16. Noonan S.
17. Normal variant; isolated anomaly
18. Oculo-dento-osseous dysplasia
19. Oro-facio-digital syndrome I (Papillon-Leage and Psaume S.) and II (Mohr S.)
20. Otopalatodigital S., type I
21. Poland S. (pectoral muscle aplasia-syndactyly)
22. Silver-Russell S.
23. TAR S. (thrombocytopenia-absent radius S.)
24. Trisomy 21 S. (Down S.)
25. Williams S. (idiopathic hypercalcemia)

+38 uncommon associations

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Camptodactyly

- Fixed interphalangeal joint malalignment in the dorsal-palmar plane
- Estimated prevalence 1%
- Likely isolated if mild
- Associations
  - Aneuploidies (i.e., trisomy 13)
  - Orofacialdigital syndrome
  - Zellweger syndrome
Sprengel Deformity

- Failure of scapular descent
- Usually sporadic
- Wide scapula
- Omovertebral bar
- Limits ROM
- Associated with Klippel-Feil
Overview

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Case – 12 year-old girl
Case -- Compare
Kirner Deformity

- Sporadic or autosomal dominant
- May co-occur with other MSK abnormalities (i.e., pes cavus, kyphosis)
- Fewer syndromic associations than clinodactyly
Case – 12F
Madelung Deformity

- Growth restriction of ulnar/volar distal radius
- Autosomal dominant
- Presents in adolescence with deformity and decreased grip strength
- Vickers ligament (radius to lunate/TFCC)
- Acquired forms are “Madelung-like”
Madelung Deformity

- Management depends on age and degree of deformity
  - Vickers ligament release
  - Distal radial physiolysis
  - Ulnar shortening
Madelung Deformity – Ulnar Shortening
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Case – 9 year-old girl
Multiple Hereditary Exostoses

- Autosomal dominant
- Prevalence 1:50,000 to 1:100,000
- Slightly higher incidence of malignant transformation than isolated osteochondromas
  - Cartilage cap thickness
  - New irregularity/internal lucency
  - Erosion of surrounding bone
Case – 8 month old boy
Anomalous or absent deep venous drainage

Results in superficial venous, capillary, and lymphatic malformations
  - Persistent embryologic venous return???

Local lipomatous and osseous hypertrophy

More common in legs
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