Hand Differences in Fanconi Anemia
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Physical Anomalies with FA

- Abnormal pigmentation 75%
- **Skeletal (Alter 1993)** 60-80%
- Growth retardation 56%
- Microcephaly 43%
- Renal anomalies 28%
- Strabismus 26%
- Hypogonadism 22%
Skeletal Anomalies

- Upper Extremity 70%
  - 1° radial deficiency
- Lower Extremity 4%
- Spine 2%
Radial Deficiencies

• Total or partial absence of preaxial border
• Variable phenotype
• May be clue to early diagnosis
Radial Deficiencies

- Bilateral 50%
- Incidence within the same family is small (5-10% of reported cases)
Associated Syndromes

- Holt-Oram syndrome (cardiac defects)
- TAR (thrombocytopenia, absent radius)
- Fanconi anemia (aplastic anemia)
Associated Syndromes

- VACTERRL (vertebral, anal atresia, cardiac, tracheoesophageal fistula, esophageal atresia, renal defects, radial dysplasia, lower limb abnormalities)
Anomalies

• Bone
  – Scapula is often reduced in size
  – Deficient capitellum and trochlea common
Anomalies

• Bone
  – Ulna is approximately 60% normal length
  – Ulna thickened and frequently bowed toward the absent radius
Anomalies

• Nerve
  – Ulnar nerve is normal
  – Radial nerve usually terminates elbow
Anomalies

- **Nerve**
  - Enlarged median nerve substitutes sensation to the radial aspect of the hand
  - Positioned in the fold between the wrist and forearm
Anomalies

- Tendon
  - Muscles that originate or attach to the radius abnormal or absent
  - FCU, ECU, interossei, and hypothenar muscles are often normal
Anomalies

- Artery
  - Normal brachial and ulnar artery
  - Radial artery is often absent
  - Interosseous arteries well developed
<table>
<thead>
<tr>
<th>Type</th>
<th>X-ray Findings</th>
<th>Clinical Features</th>
</tr>
</thead>
<tbody>
<tr>
<td>I Short radius</td>
<td>Distal radial epiphysis delayed in appearance Normal proximal radial epiphysis Mild shortening of radius without bowing</td>
<td>Minor radial deviation of the hand Thumb hypoplasia is the prominent clinical feature requiring treatment</td>
</tr>
<tr>
<td>II Hypoplastic</td>
<td>Distal &amp; proximal epiphysis present Abnormal growth in both epiphyses Ulna thickened, shortened, &amp; bowed</td>
<td>Miniature radius Moderate radial deviation of the hand</td>
</tr>
<tr>
<td>III Partial absence</td>
<td>Partial absence (distal, middle, proximal) of radius Distal 1/3-2/3 absence most common Ulna thickened, shortened, &amp; bowed</td>
<td>Severe radial deviation of the hand</td>
</tr>
<tr>
<td>IV Total absence</td>
<td>No radius present Ulna thickened, shortened, &amp; bowed</td>
<td>Most common type Severe radial deviation of the hand</td>
</tr>
</tbody>
</table>
Radial Deficiency
Treatment Goals

• Maximize length
• Improve appearance
• Improve function
• Create stable wrist
Natural History

• 90 to 100  radial deviation
Treatment Paradigm

- Stretching
- Splinting
- Surgery
Radial Deficiency - Stretching

- Start early & frequent splint changes
Surgery

- Centralization indications/ contraindications
  - Elbow motion
  - Systemic conditions
  - Age
Centralization Principles

- Release tight radial structures to allow centralization
- Carpal realignment over ulna
- Tendon balance
Centralization
Centralization

- ± ulnar osteotomy (greater than 30 degrees)
Results & Complications

- Correction
- Recurrence
- Stiffness
- Functional impact
Results & Complications

Does it Help Function?

• Effect on function unclear
• Individualized per patient
• Self-esteem?
• ??

Goldfarb, Manske, and colleagues. Functional outcome after centralization for radius dysplasia
Hand Surg 2002;27A:118-124
Treatment Advances

- Soft tissue lengthening
- Bone lengthening
Case Example

• MJ- 9 year-old previous centralization at age 2 with recurrence
Assemble rings

Tighten wires
Corticotomy
Latency period Lengthening
Angular correction & lengthening

Regenerate bone
4 years later
Now What?

• Considerations
  – Form
  – Function
  – ??
Questions

• Recurrence is common, if not universal
• Centralization improves appearance, but may not significantly enhance function
• What is the role of centralization in FA kids??
Hypoplastic thumb

- Subgroup of radial deficiency
- Classification of thumb deficiency
  - Important for treatment paradigm
<table>
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<th>Findings</th>
<th>Treatment</th>
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</thead>
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<tr>
<td>I Minor generalized hypoplasia</td>
<td>Augmentation</td>
</tr>
<tr>
<td>II Absence of intrinsic thenar</td>
<td>Opponensplasty 1st-web release</td>
</tr>
<tr>
<td>muscles</td>
<td>UCL reconstruction</td>
</tr>
<tr>
<td>First web space narrowing</td>
<td></td>
</tr>
<tr>
<td>UCL insufficiency</td>
<td></td>
</tr>
<tr>
<td>III Similar findings as type II</td>
<td>A: Reconstruction B: Pollicization</td>
</tr>
<tr>
<td>plus:</td>
<td></td>
</tr>
<tr>
<td>Extrinsic abnormalities</td>
<td></td>
</tr>
<tr>
<td>Skeletal deficiency</td>
<td></td>
</tr>
<tr>
<td>A: Stable carpometacarpal joint</td>
<td></td>
</tr>
<tr>
<td>B: Unstable carpometacarpal joint</td>
<td></td>
</tr>
<tr>
<td>IV Pouce flottant or floating</td>
<td>Pollicization</td>
</tr>
<tr>
<td>thumb</td>
<td></td>
</tr>
<tr>
<td>V Absence</td>
<td>Pollicization</td>
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</table>
Treatment Paradigm

- Type I  →  No treatment
- Type II  →  Reconstruction
- Type IIIA  →  Reconstruction
Treatment Paradigm

- Type IIIB → Pollicization
- Type IV → Pollicization
- Type V → Pollicization
Mild Hypoplasia (Blauth I)
No Treatment Needed
Thenar Hypoplasia (Blauth II)
Opponensplasty (Blauth II)
Pollicization

- Timing
- Usually 1 year
  - Developmental
  - Ease
Skin Incision

• Multiple options
Skin Incision
Bone & Joint Reorganization

- DIP joint → IP joint
- PIP joint → MP joint
- MP joint → CMC joint
- Metacarpal → Trapezium
Outcome

• Related to degree of index mobility

Results

- Enhance function
- Grasp > pinch
Complications

- Stiffness
- Decreased motors → tendon transfer
Thank You
9 year-old with recurrent deformity
Formal centralization
Ulnar approach

K-wire fixation
Extension

Flexion
Results

- Related to degree of index mobility
Additional half-pins as needed
Hand Differences in Fanconi Anemia

• Outcome
• Measurement tool
  – DASH
  – Jebsen-Taylor
• Reliable assessment

Canadian Occupational Performance Measure

- Client-identified goals
- Client rated scores on performance of and satisfaction with FES
- Likert-type scale between 1-10
  - 1 cannot perform & extremely dissatisfied
  - 10 performs very well & extremely satisfied

Hand Differences in Fanconi Anemia

- Failure of formation - hypoplastic thumb
- Complications after pollicization
  - Malposition