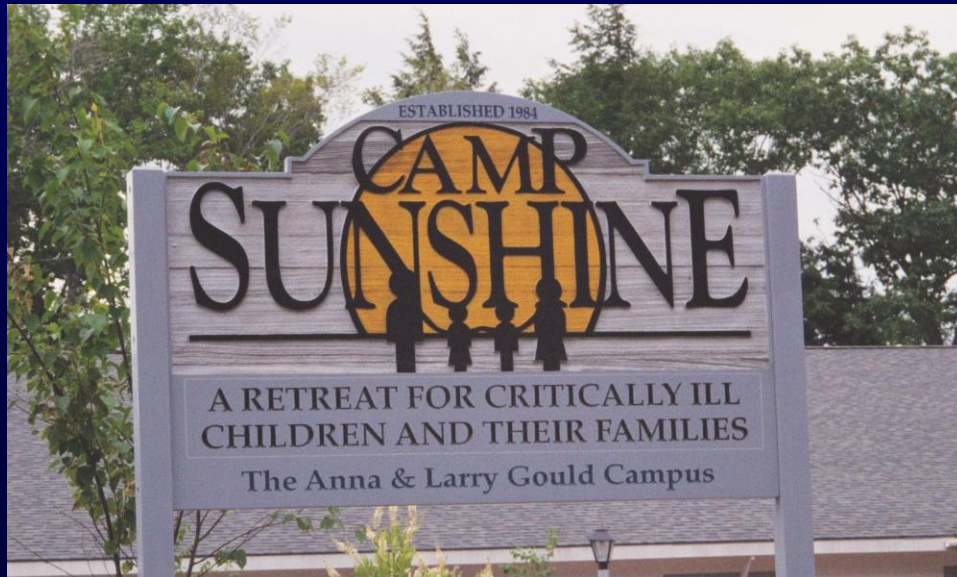


Hand Differences in Fanconi Anemia

Scott H. Kozin, MD

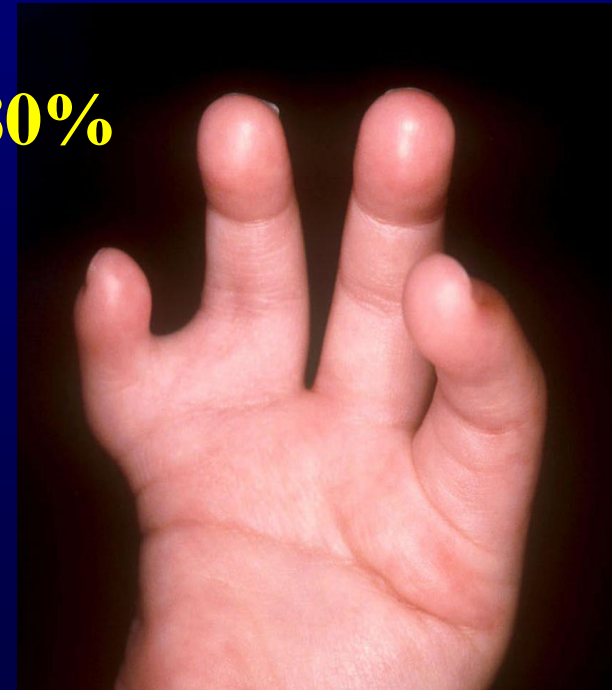
Shriners Hospital for Children

Philadelphia, PA



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



Type	X-ray Findings	Clinical Features
I Short radius	Distal radial epiphysis delayed in appearance Normal proximal radial epiphysis Mild shortening of radius without bowing	Minor radial deviation of the hand Thumb hypoplasia is the prominent clinical feature requiring treatment
II Hypoplastic	Distal & proximal epiphysis present Abnormal growth in both epiphyses Ulna thickened, shortened, & bowed	Miniature radius Moderate radial deviation of the hand
III Partial absence	Partial absence (distal, middle, proximal) of radius Distal 1/3-2/3 absence most common Ulna thickened, shortened, & bowed	Severe radial deviation of the hand
IV Total absence	No radius present Ulna thickened, shortened, & bowed	Most common type Severe radial deviation of the hand



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

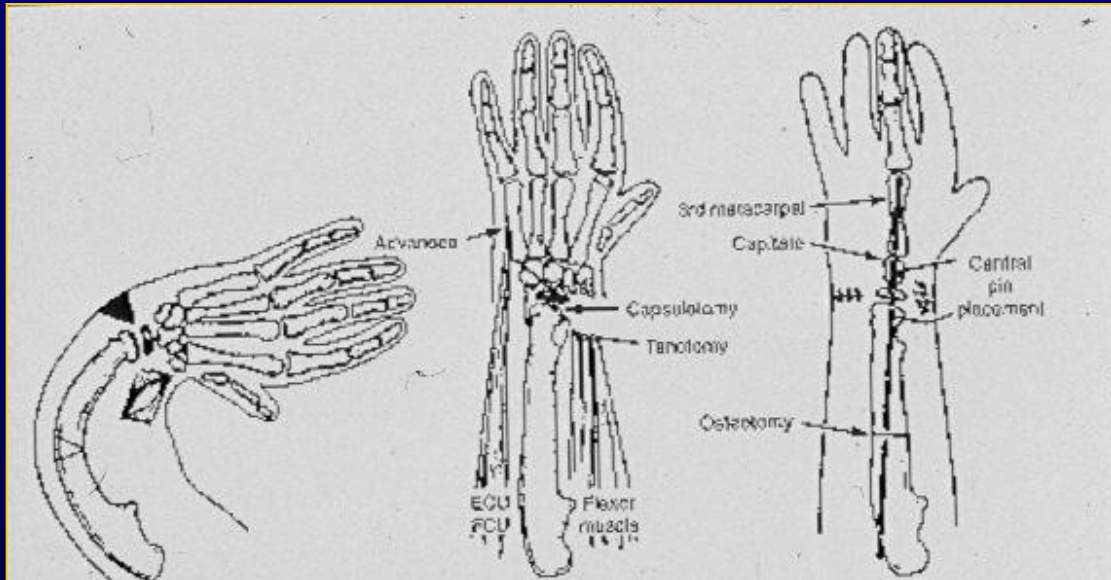
- Centralization indications/ contraindications
 - Elbow motion
 - Systemic conditions
 - Age



Centralization Principles

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

Centralization



Centralization

- \pm ulnar osteotomy (greater than 30 degrees)



Results & Complications

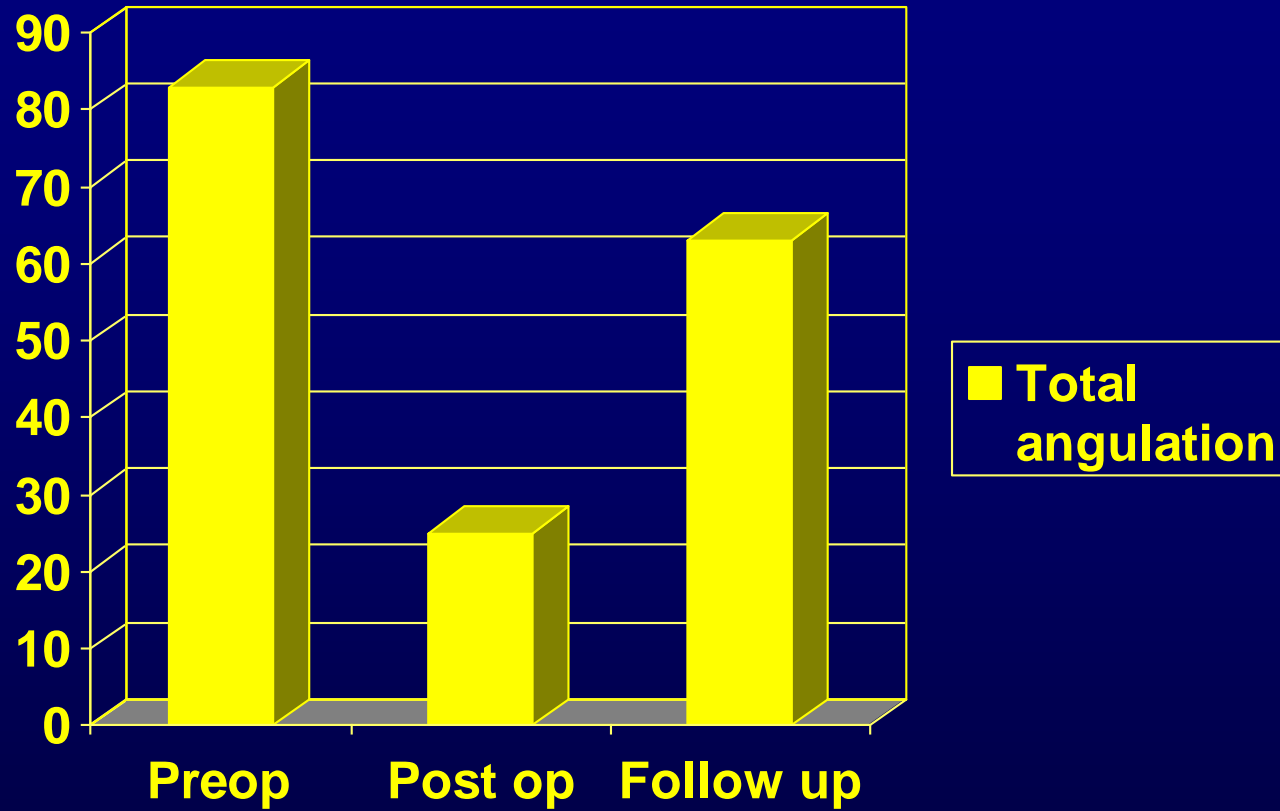
- Correction
- Recurrence
- Stiffness
- Functional impact



Results & Complications



Damore E, Kozin SH, Thoder JJ, Porter S:
J Hand Surg 2000;25A:745-751.



Damore, Kozin, Thoder, Porter. J Hand Surg 2000

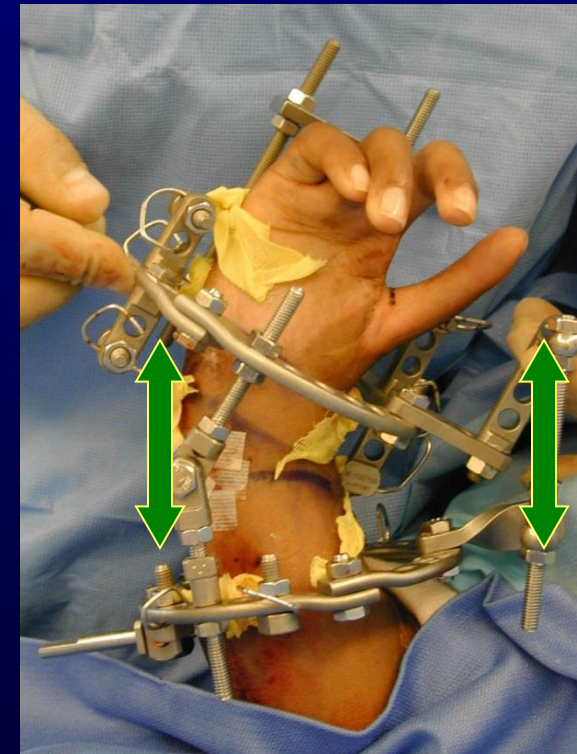
Does it Help Function?

- 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

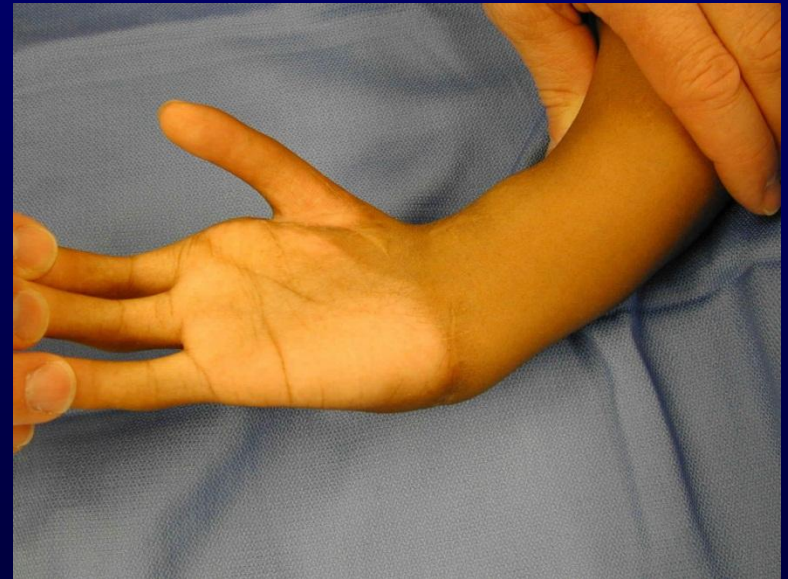
Treatment Advances

- Soft tissue lengthening
- Bone lengthening

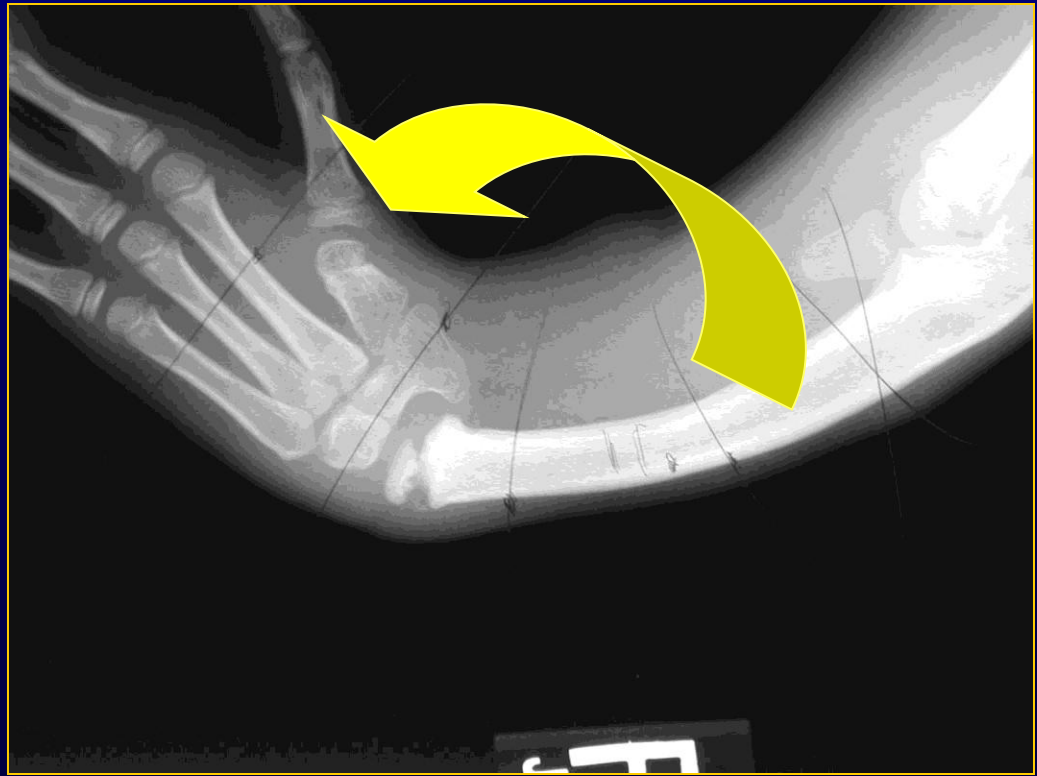
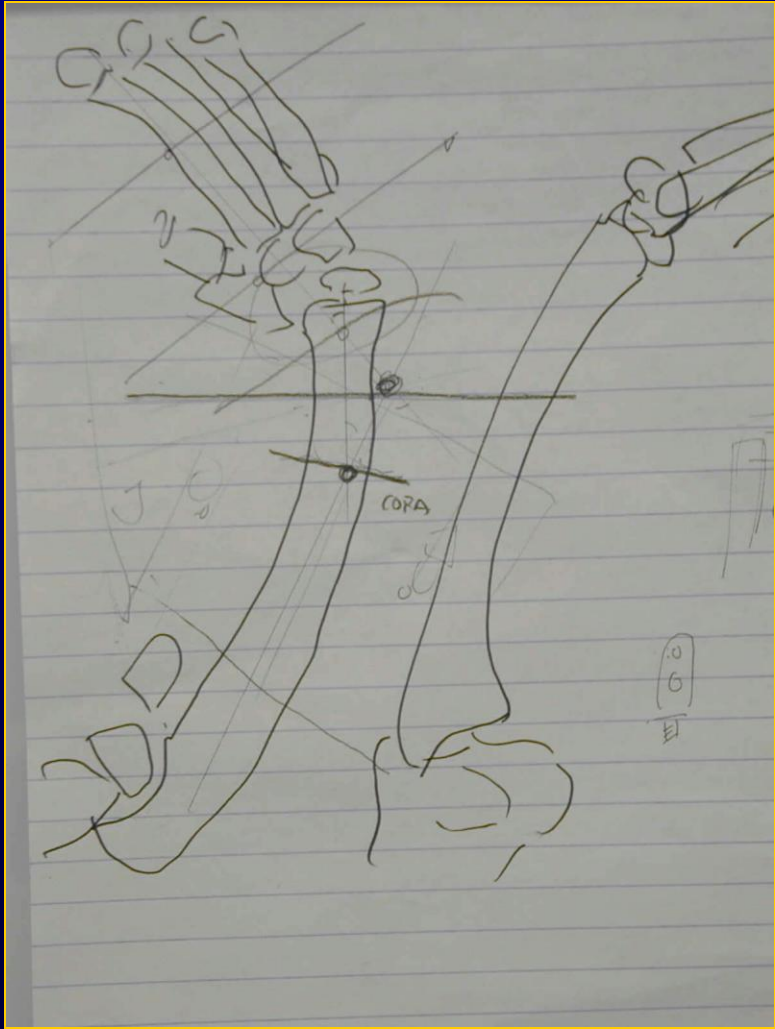


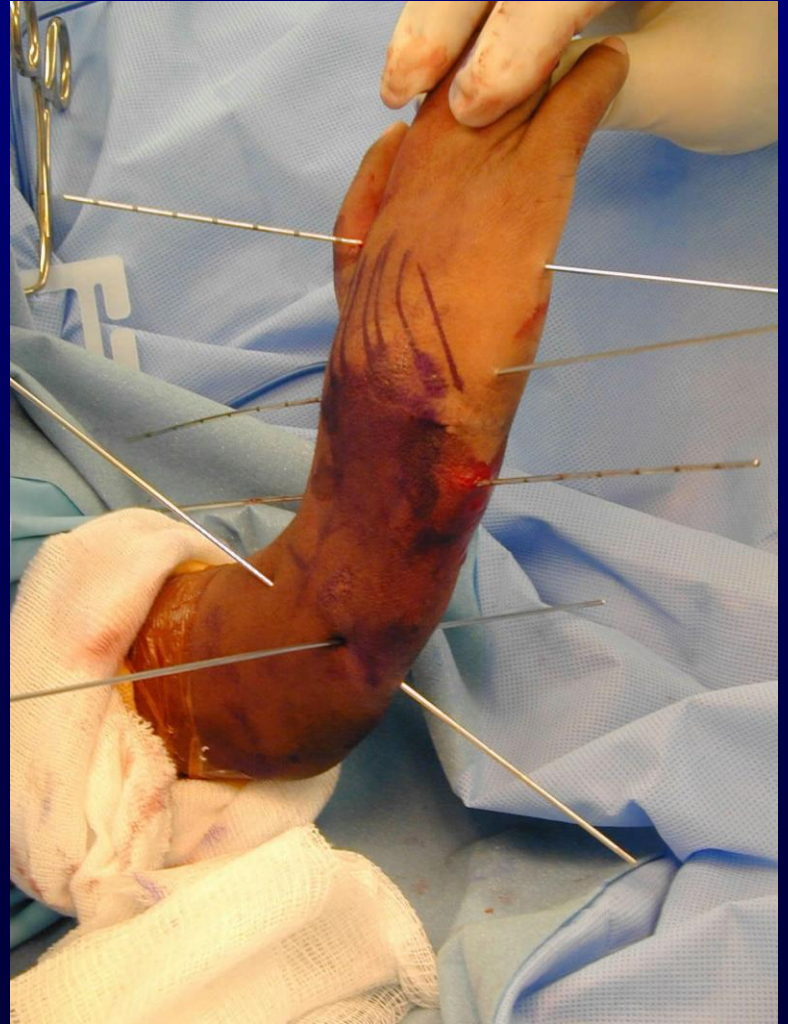
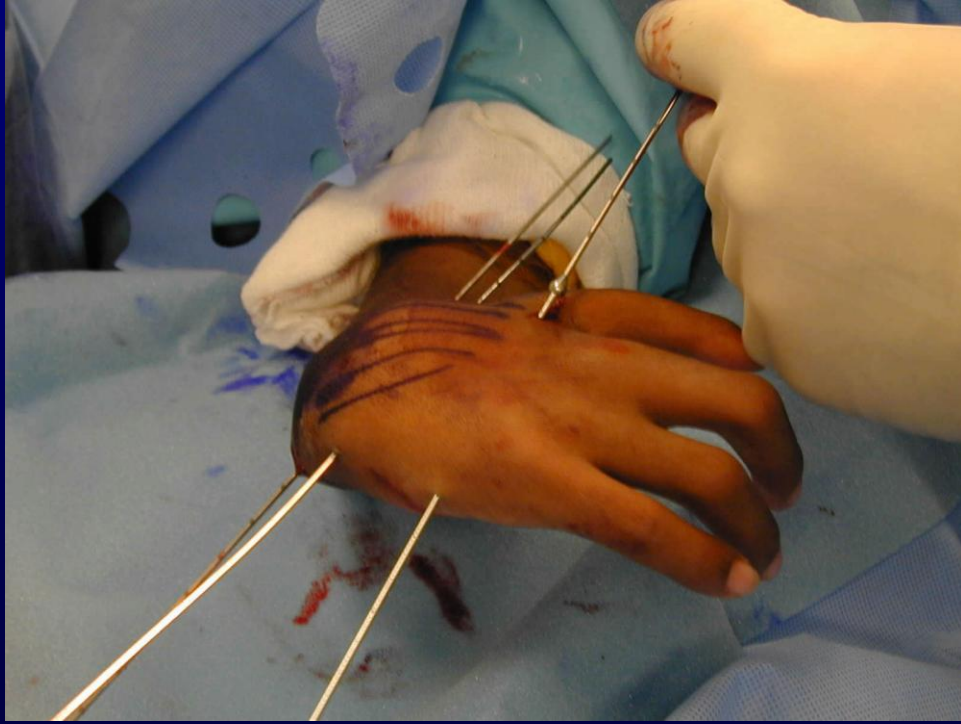
Case Example

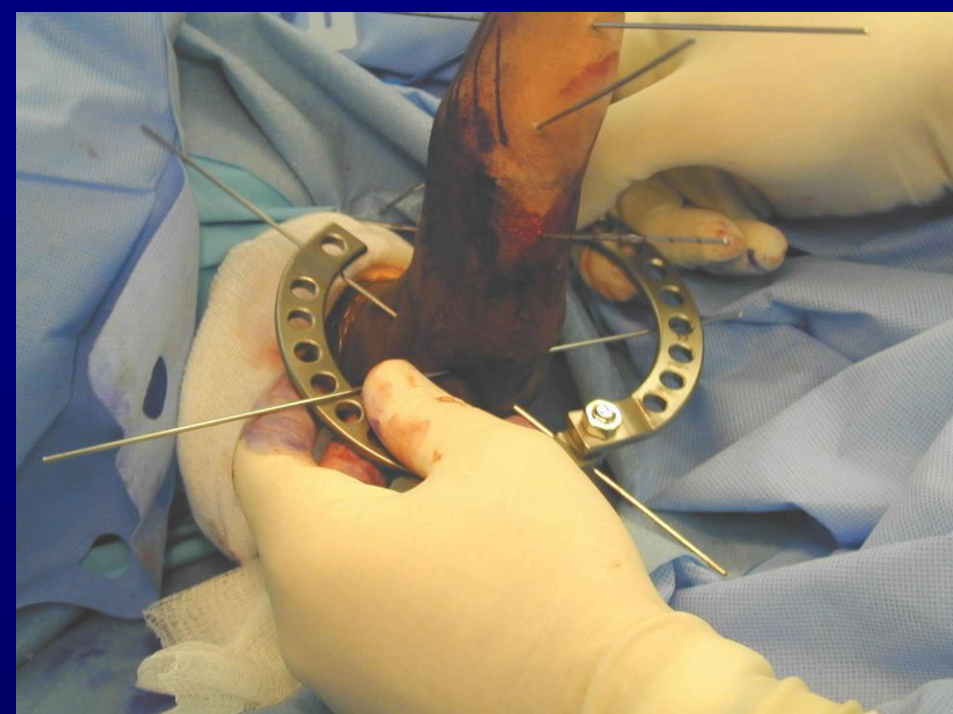
- MJ- 9 year-old previous centralization at age 2 with recurrence







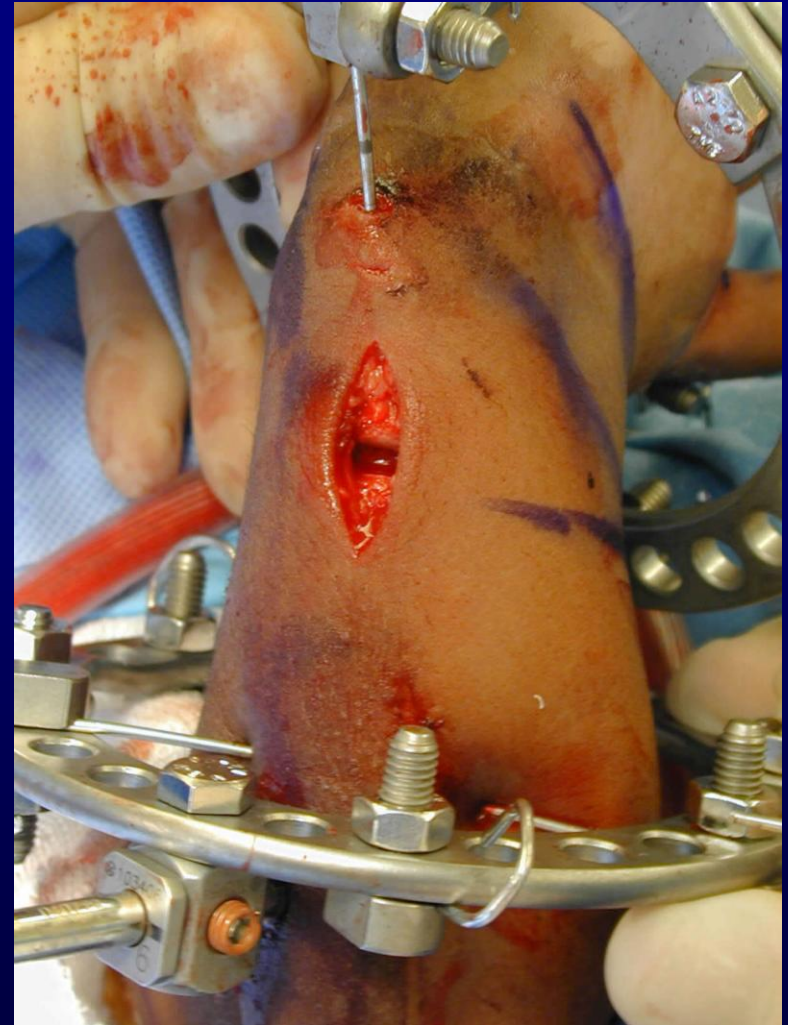
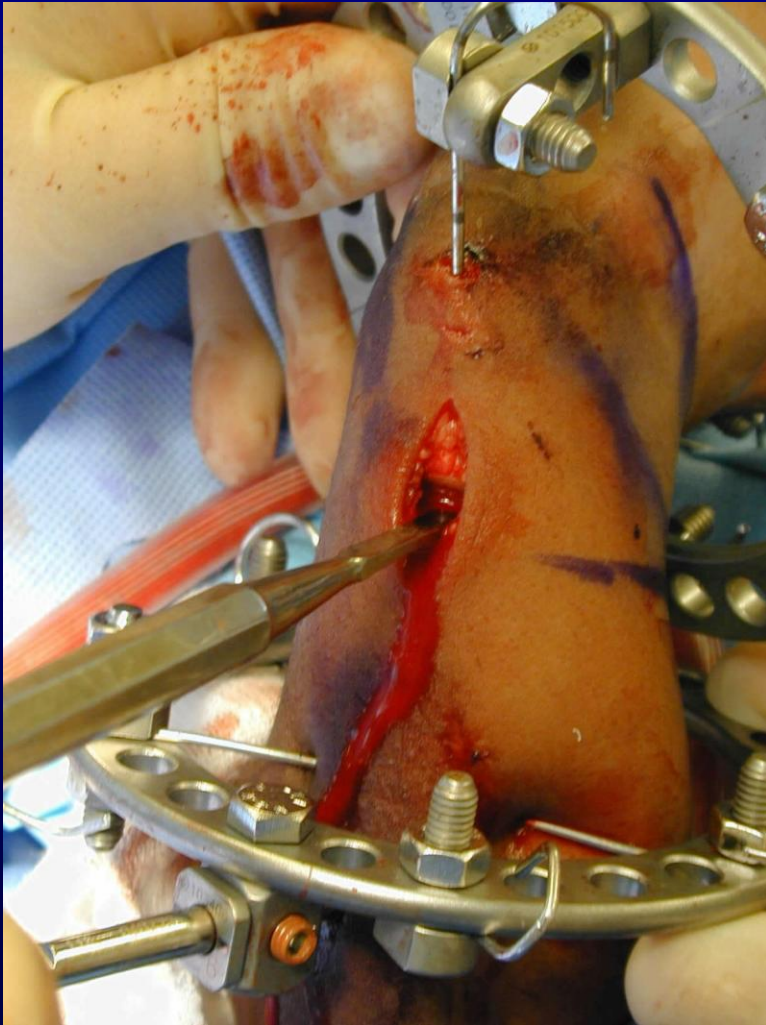




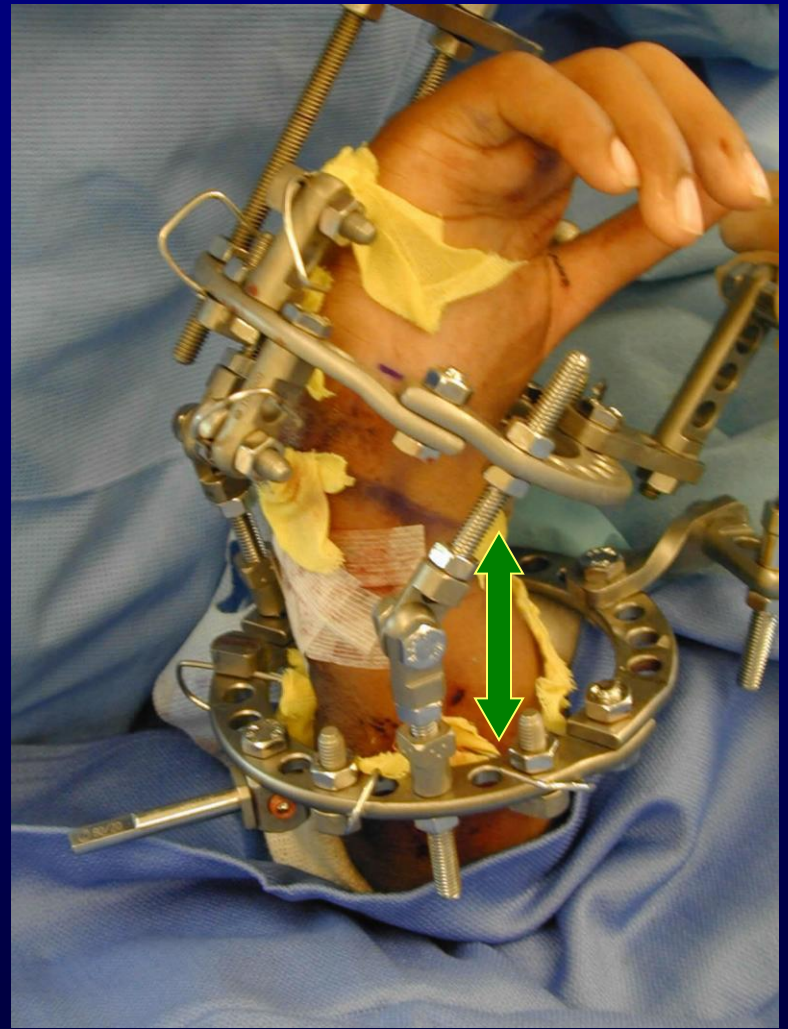
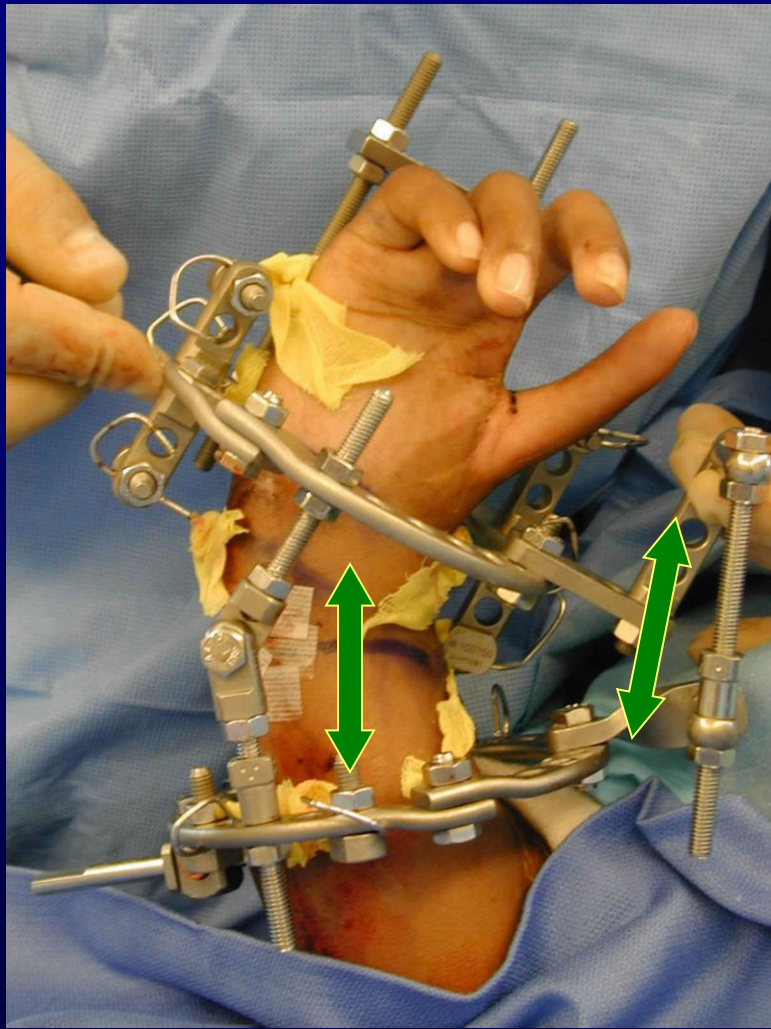
Assemble rings

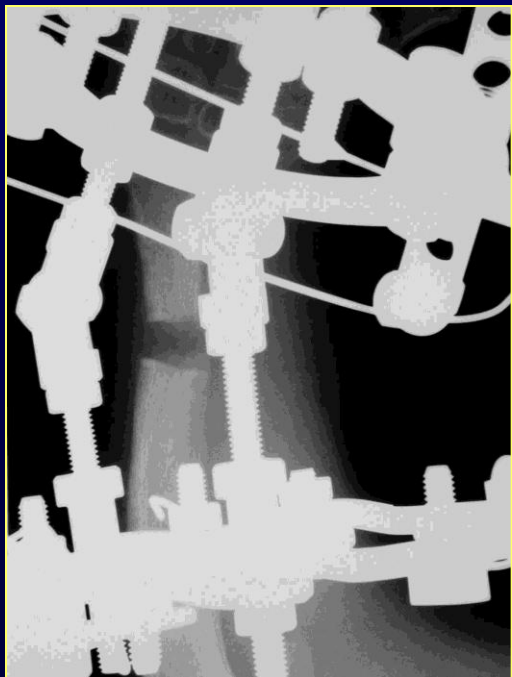
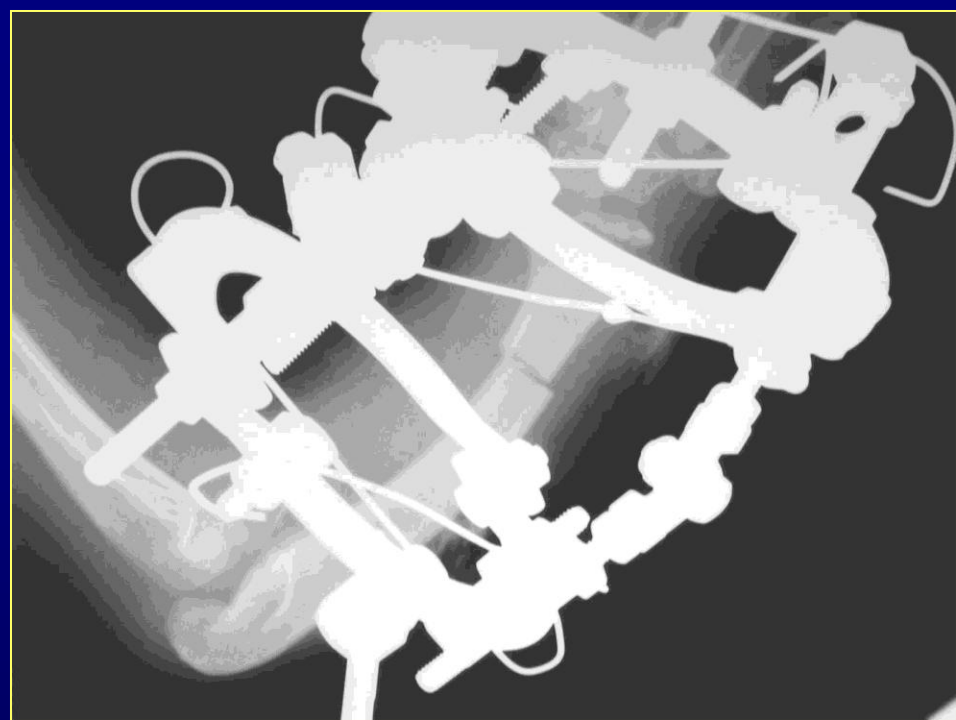
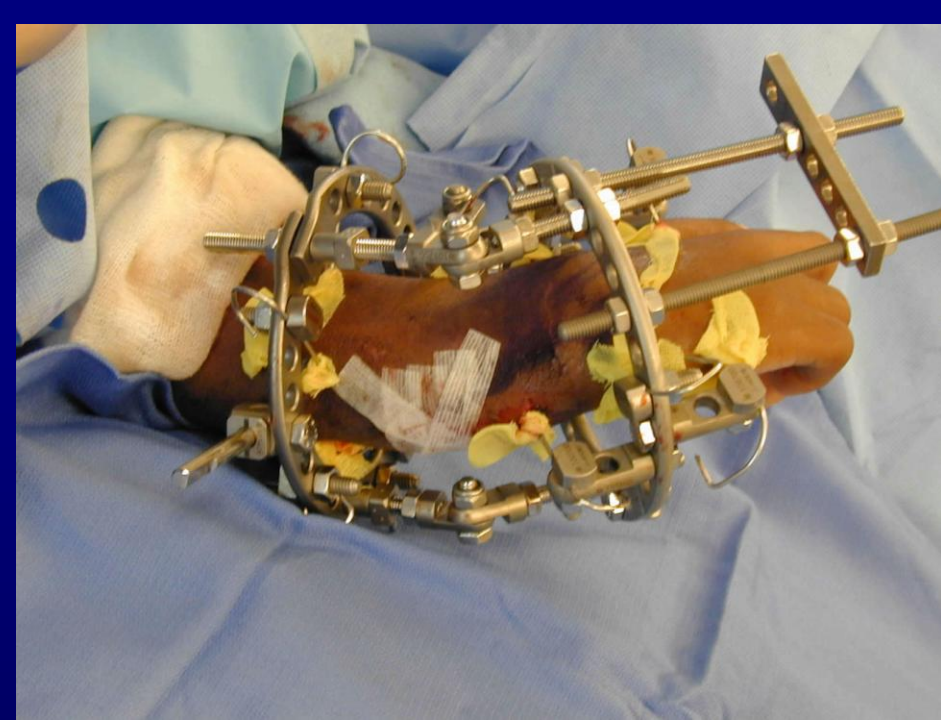
Tighten wires





Corticotomy

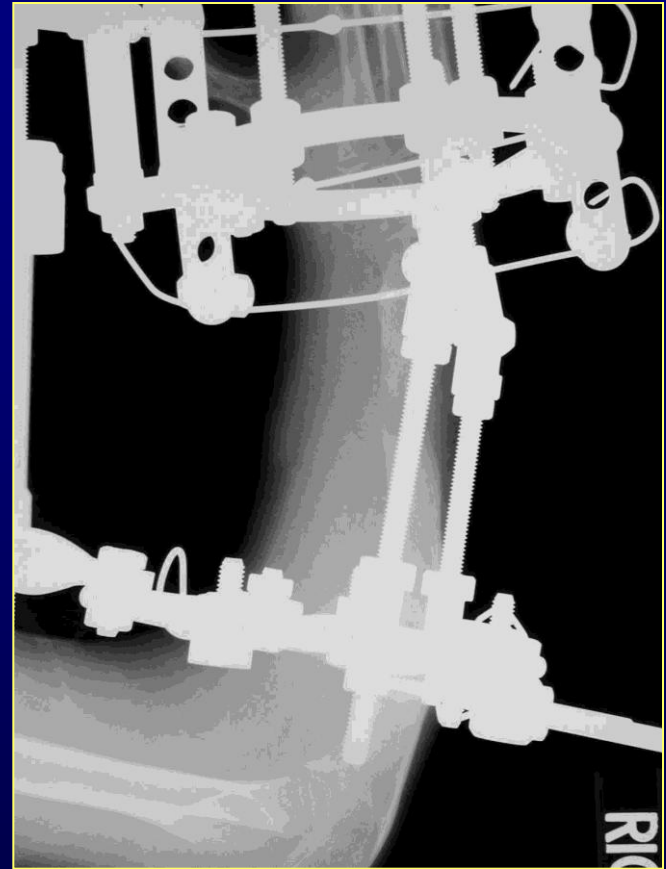




Latency period
Lengthening



Angular correction
& lengthening



Regenerate
bone



WOW



BUT



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



	Findings	Treatment
I	Minor generalized hypoplasia	Augmentation
II	Absence of intrinsic thenar muscles First web space narrowing UCL insufficiency	Opponensplasty 1st-web release UCL reconstruction
III	Similar findings as type II plus: Extrinsic abnormalities Skeletal deficiency A: Stable carpometacarpal joint B: Unstable carpometacarpal joint	A: Reconstruction B: Pollicization
IV	Pouce flottant or floating thumb	Pollicization
V	Absence	Pollicization

Treatment Paradigm

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



Treatment Paradigm

- Type IIIB → Pollicization
- Type IV → Pollicization
- Type V → Pollicization

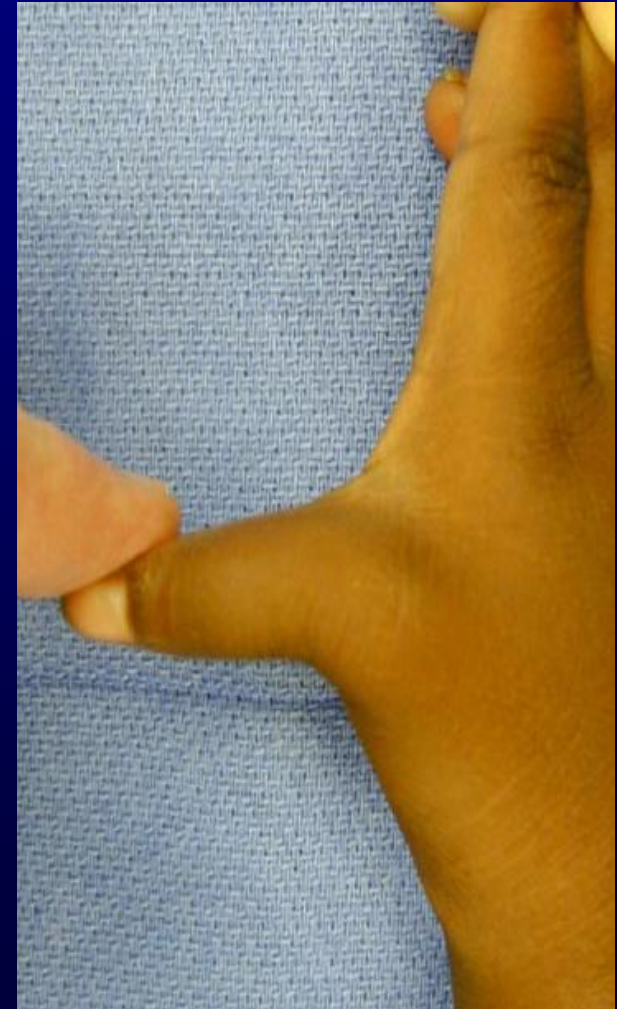


Mild Hypoplasia (Blauth I)

No Treatment Needed



Thenar Hypoplasia (Blauth II)



Opponensplasty (Blauth II)



Pollicization

- Timing
- Usually 1 year
 - Developmental
 - Ease



Skin Incision

- Multiple options

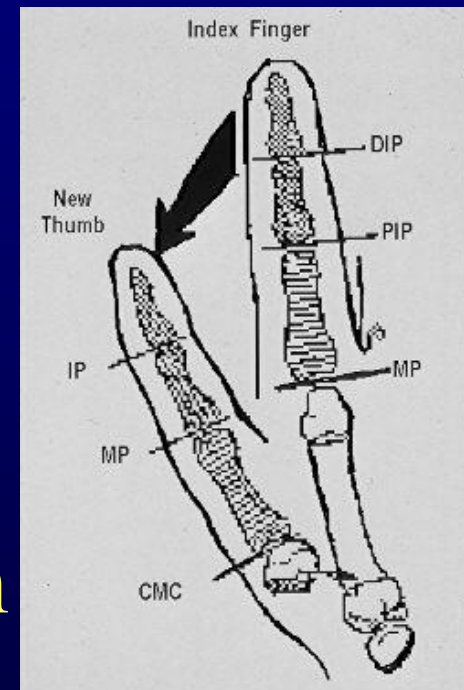


Skin Incision



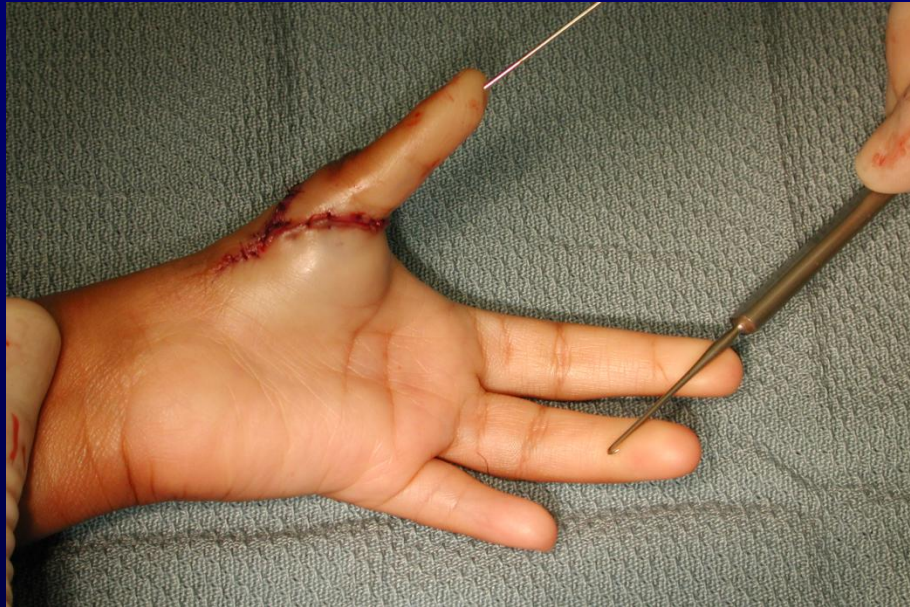
Bone & Joint Reorganization

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



Outcome

- Related to degree of index mobility



Kozin SH et al. Functional Results After Index Finger Pollicization for Congenital Aplasia or Hypoplasia of the Thumb. J Hand Surg 17A:880-884, 1992.

Results

- Enhance function
- Grasp > pinch





Complications

- Stiffness
- Decreased motors → tendon transfer

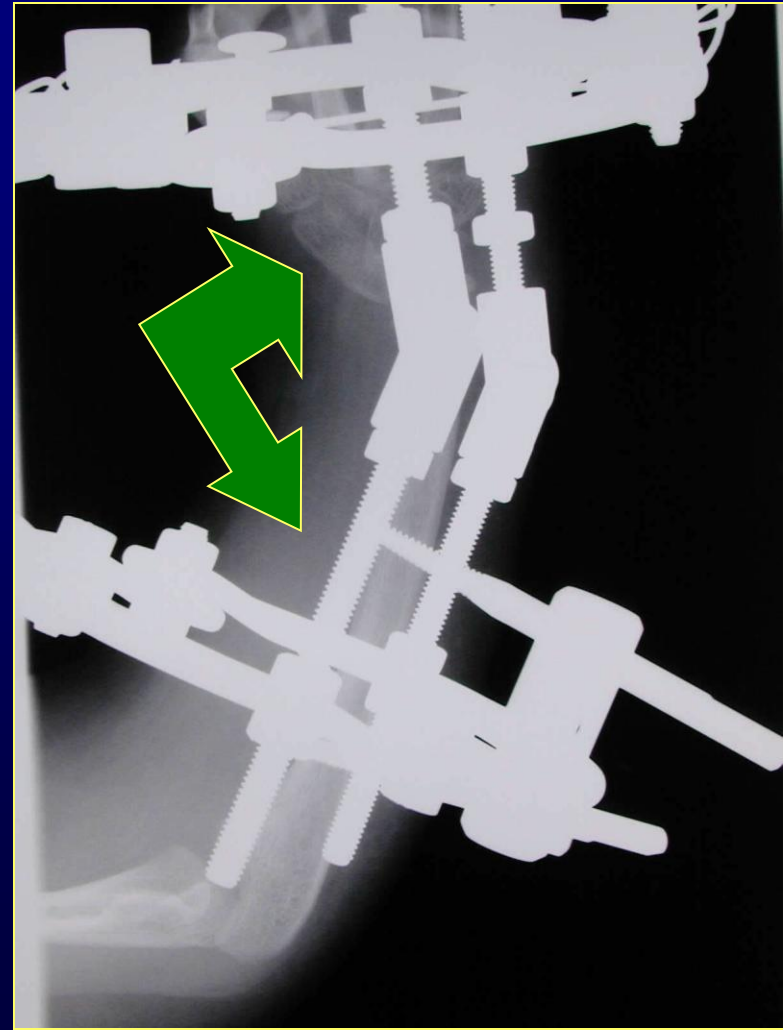
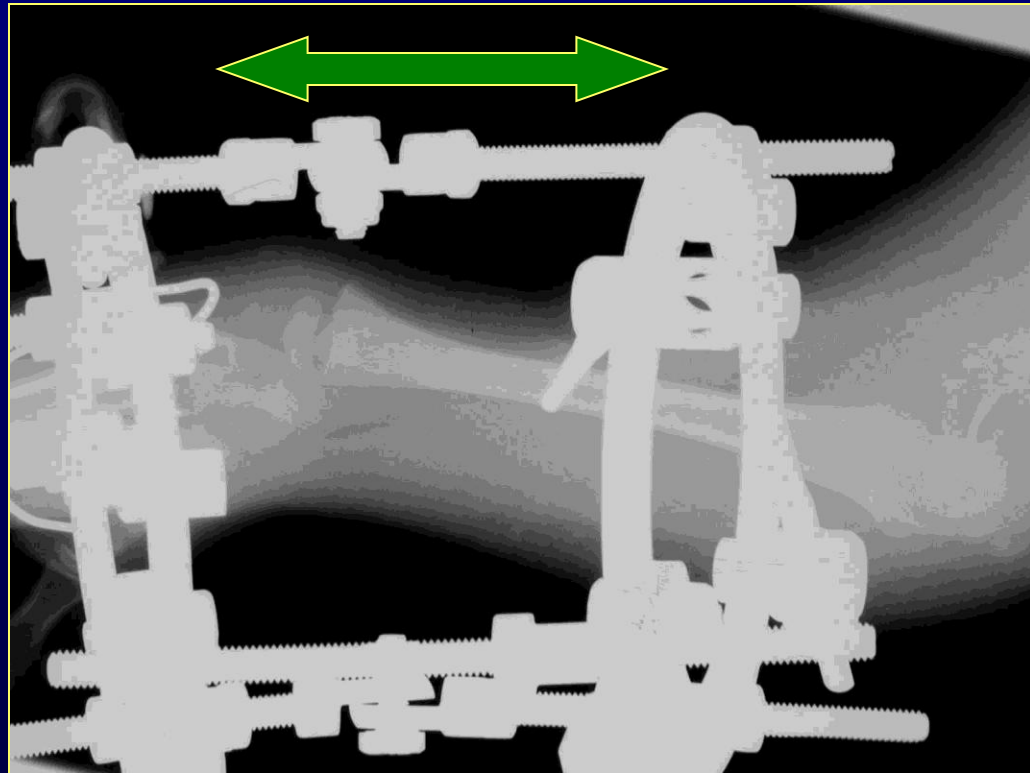


Thank You



9 year-old with recurrent deformity

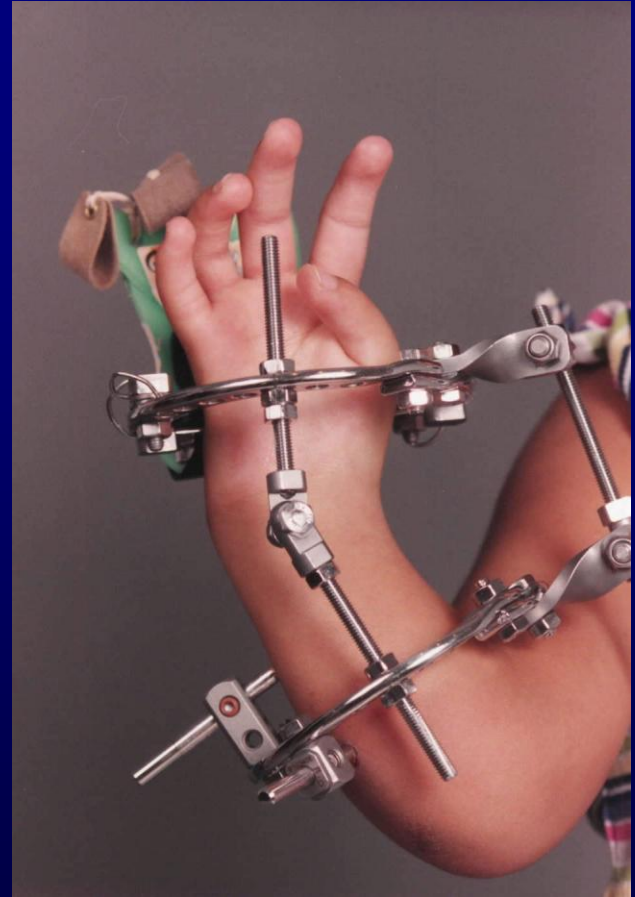




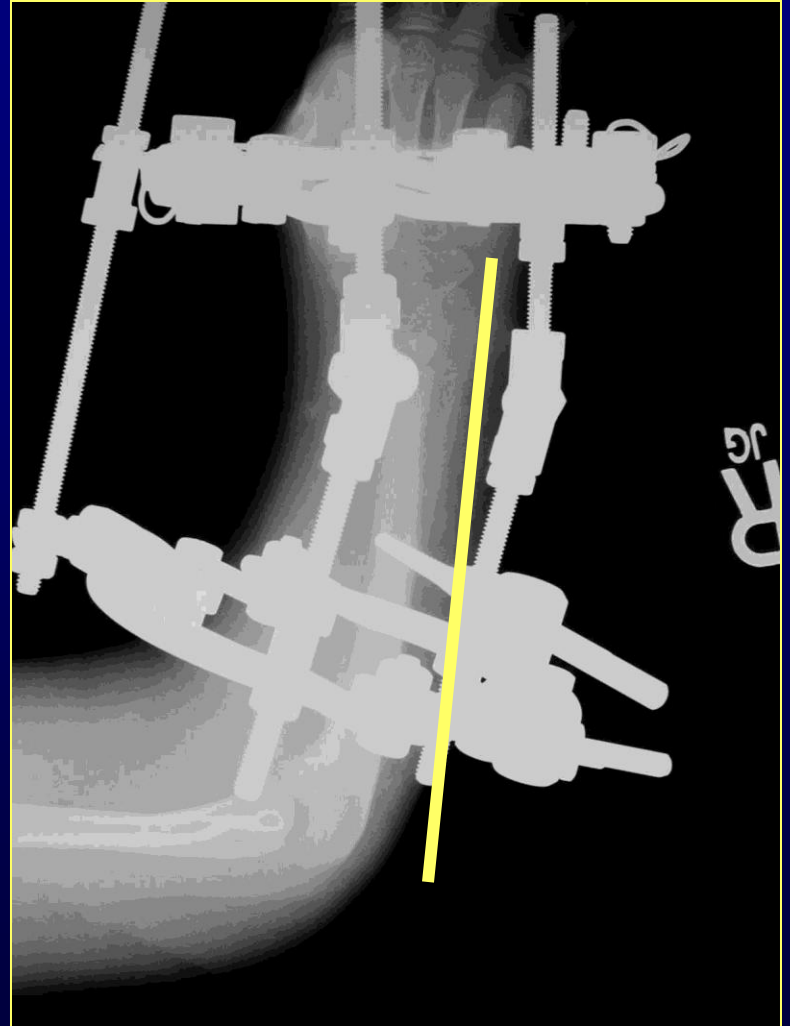
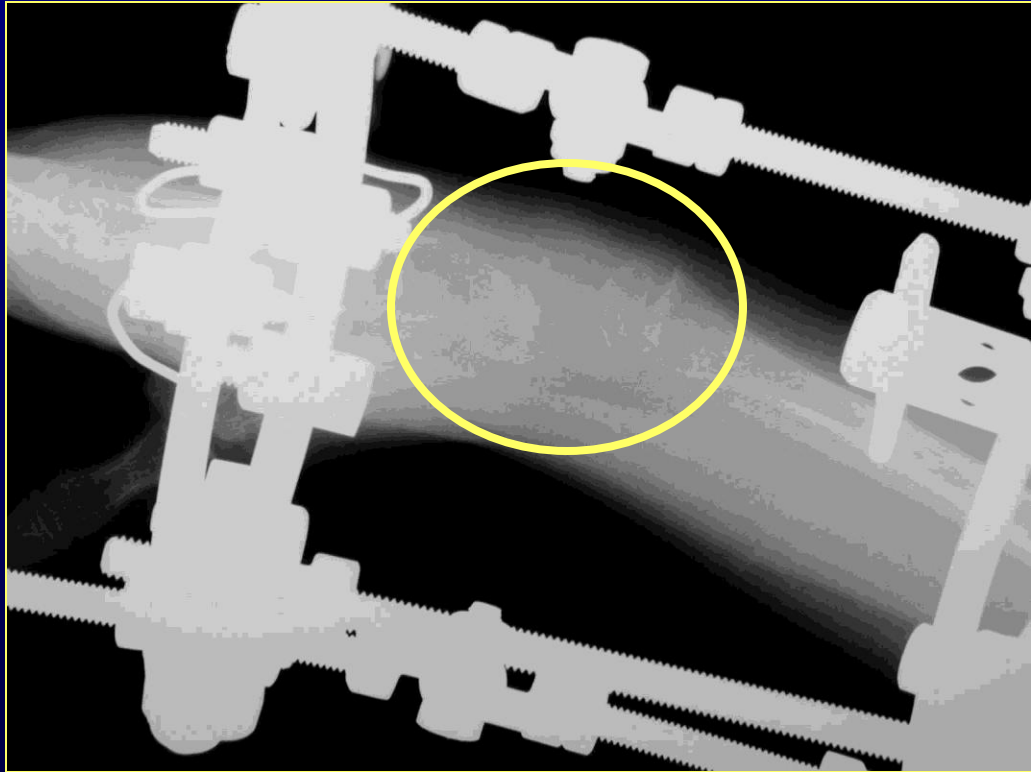


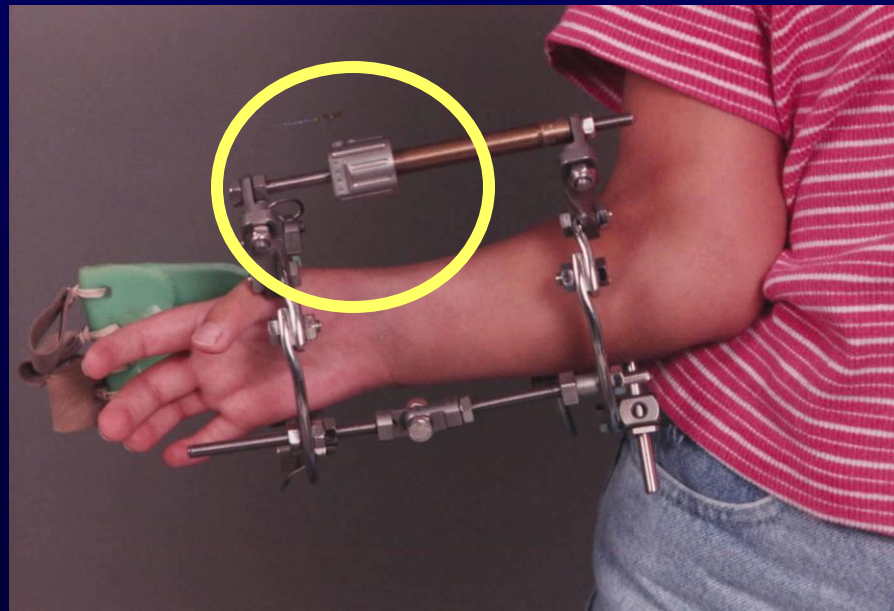


Flexion

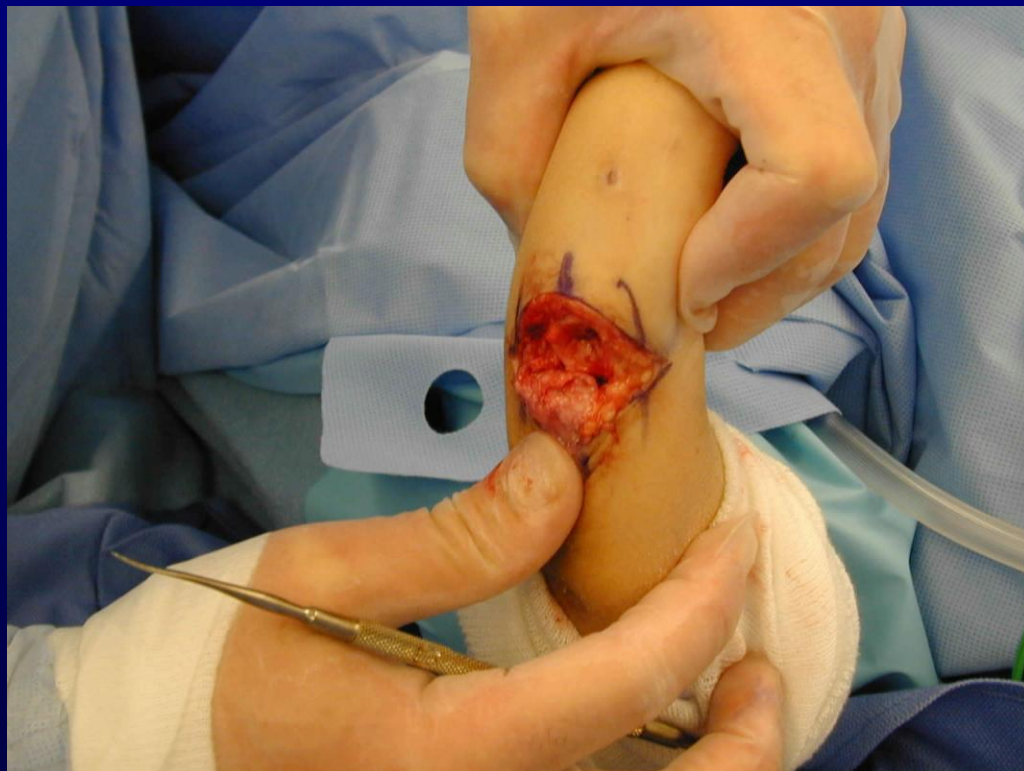


Extension





Formal centralization





Ulnar approach

K-wire fixation







Extension

Flexion

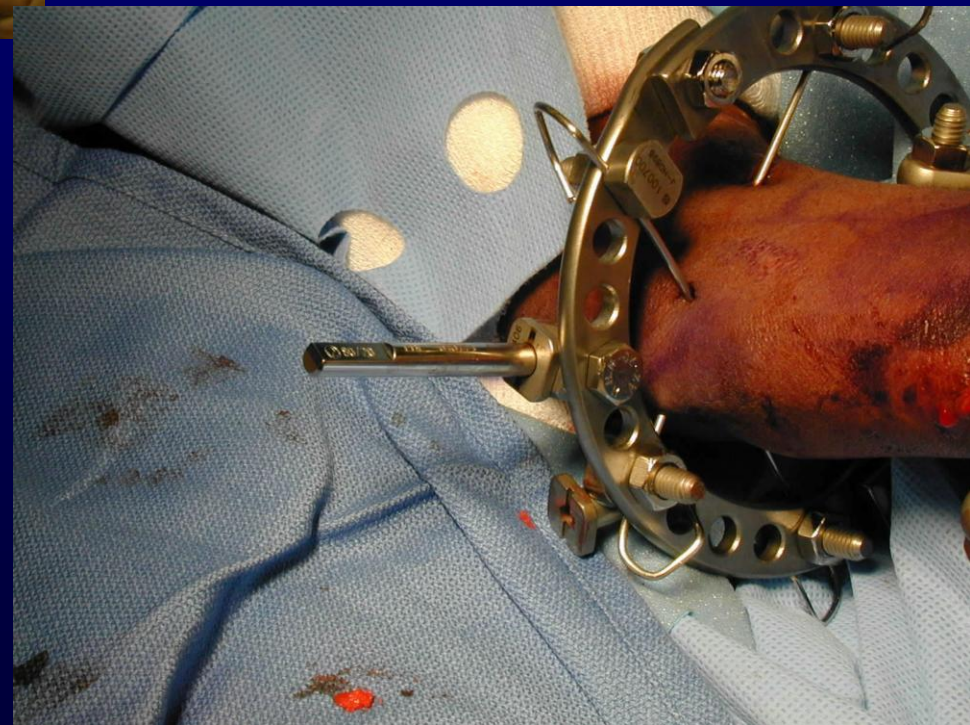


Results

- Related to degree of index mobility



Additional half-pins
as needed





Hand Differences in Fanconi Anemia

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



Goldfarb et al. Functional Outcome after Centralization for Radial Dysplasia. J Hand Surg 2002;27A:118-124.

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

**Law et al. The Canadian Occupational Performance Measure.
CJOT 57: 1990;87.**

Hand Differences in Fanconi Anemia

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

