CONGENITAL HAND DIFFERENCES

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CONGENITAL DEFORMITIES
Upper Extremity

- Common
- 1 in 626 live births
- Many are single gene disorders

(2008 ASSH SAE qn 68)

The period of time following fertilization during which limb bud development is most rapid and when most congenital anomalies occur is:

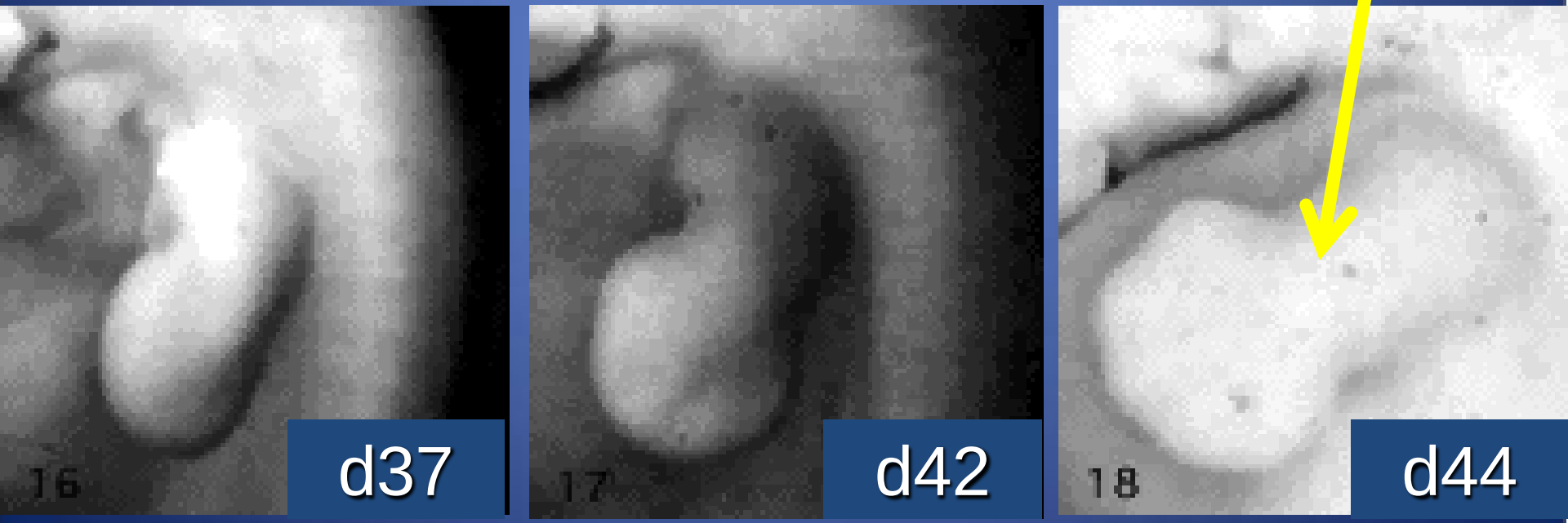
A. 1-4 weeks
B. 4-8 weeks
C. 8-12 weeks
D. 12-14 weeks
E. 14-18 weeks

DIFFERENTIATION OF THE UPPER LIMB

- Limb buds - ventrolateral wall of embryo
- Limb buds appear 4th week (day 26)
- Limb buds develop from day 26-47

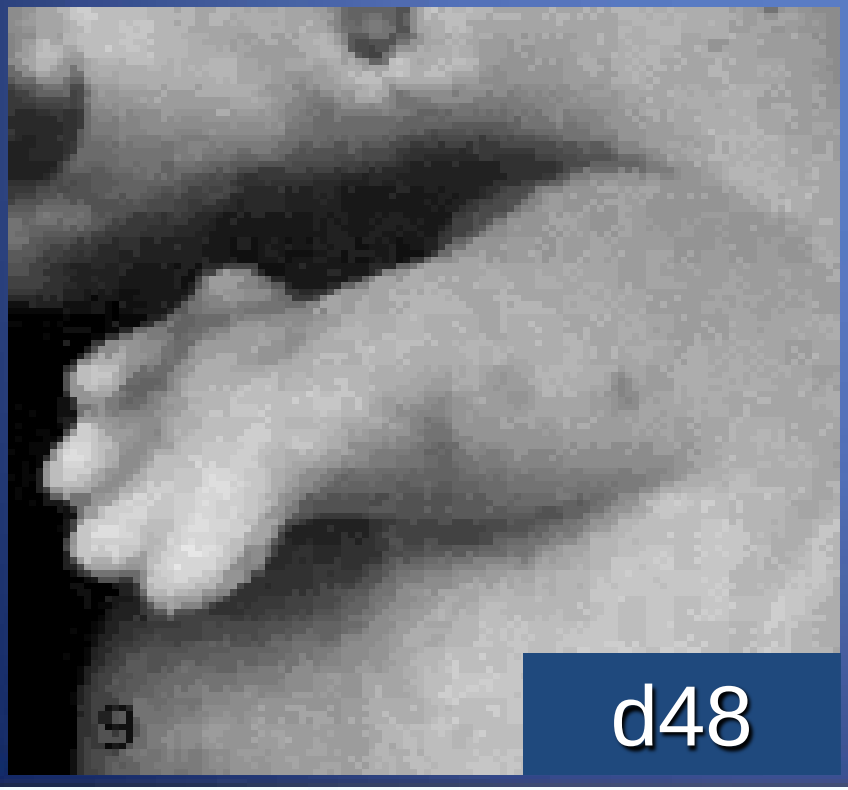
Weeks 5-6

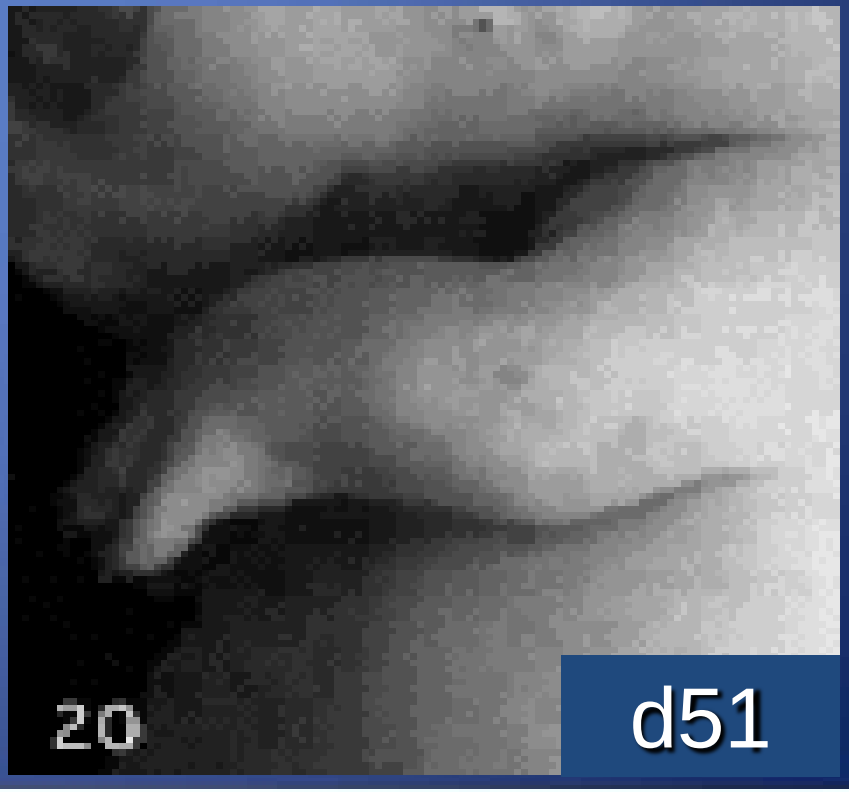
- Hand paddle develops
- Nerve ingrowth from spinal cord rami begins to occur proximally



Weeks 6 - 7

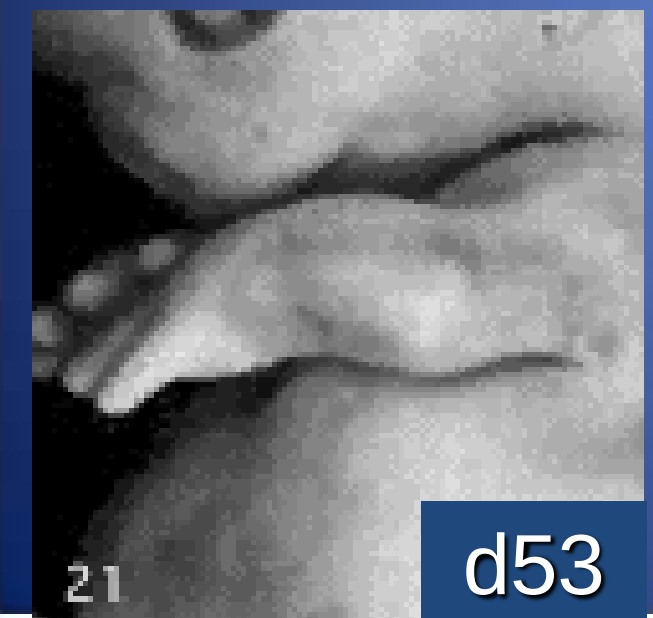
- Fingers begin to separate
- Cartilaginous “bones” form

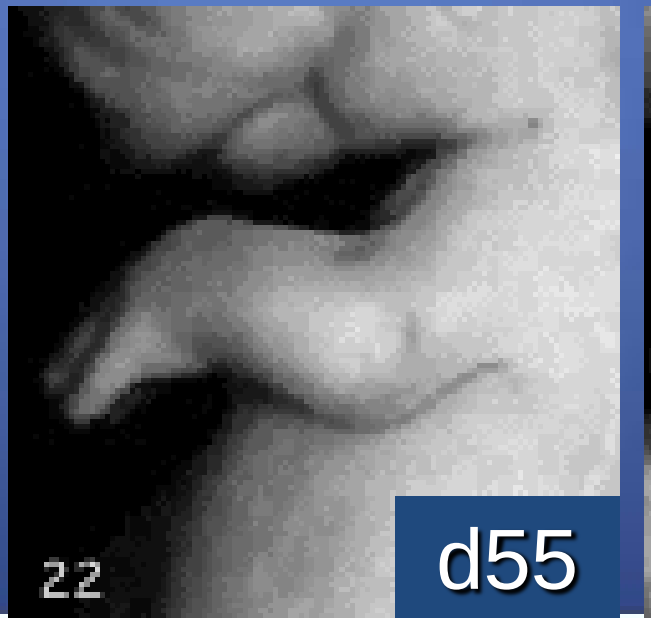


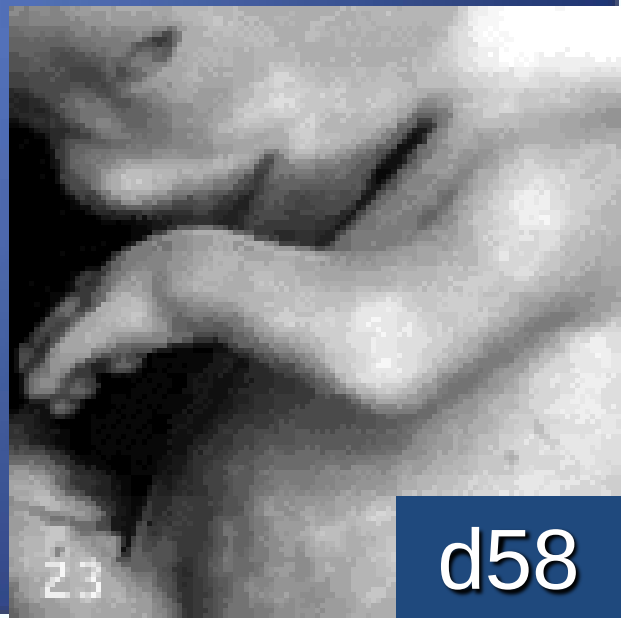


Weeks 7 — 8

- The UE grows and rotates 90°
- Elbows project posteriorly
- Dorsal mesenchymal stem cells – extensors
- Ventral mesenchymal stems cells - flexors







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AIR TRAFFIC CONTROLLERS A.G.M.
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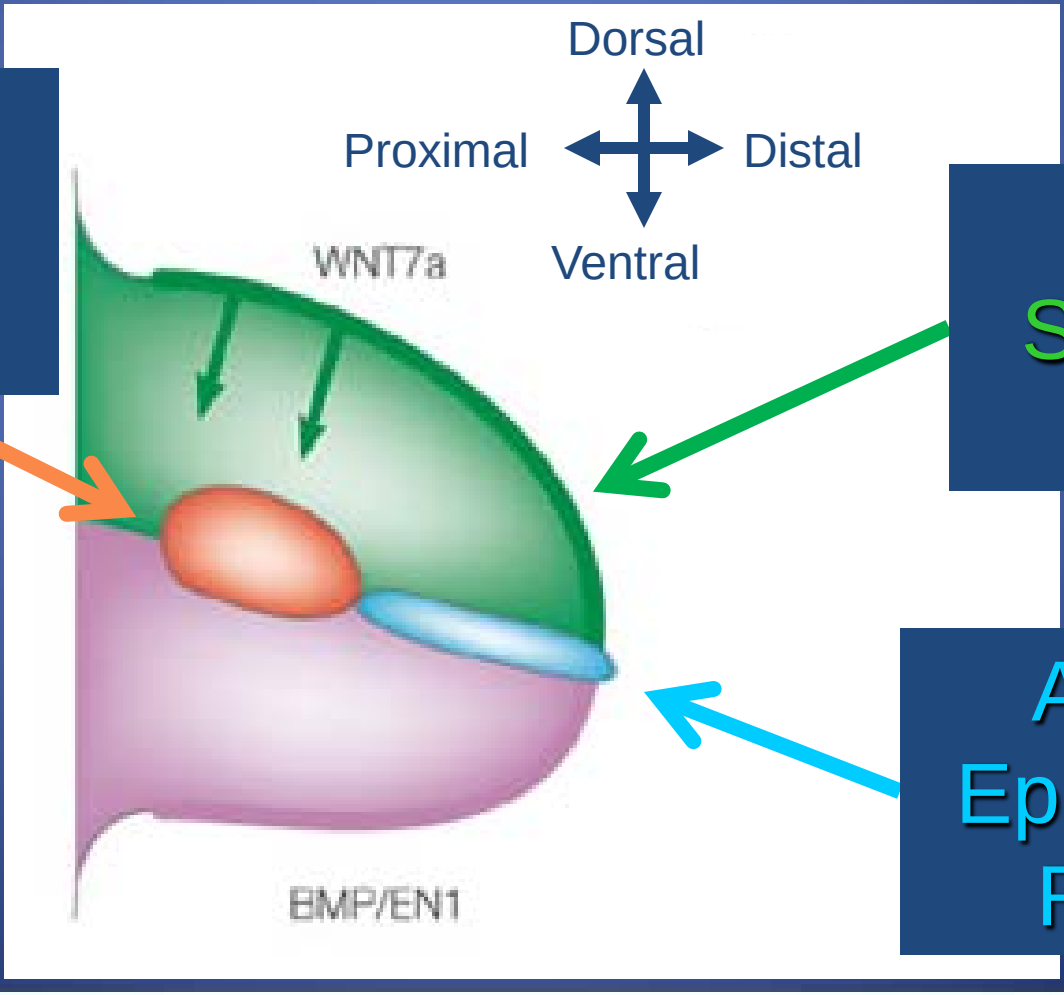
SIGNALING CENTERS IN LIMB DEVELOPMENT

Three centers control limb development

Zone of Polarizing Activity



Wnt Signaling Center

Apical Epidermal Ridge

Transverse Limb Deficit

- Removal of the AER → truncated limb (congenital amputation)
- Placement of the AER elsewhere → ectopic limb
- Can replace AER with Fibroblast growth factor

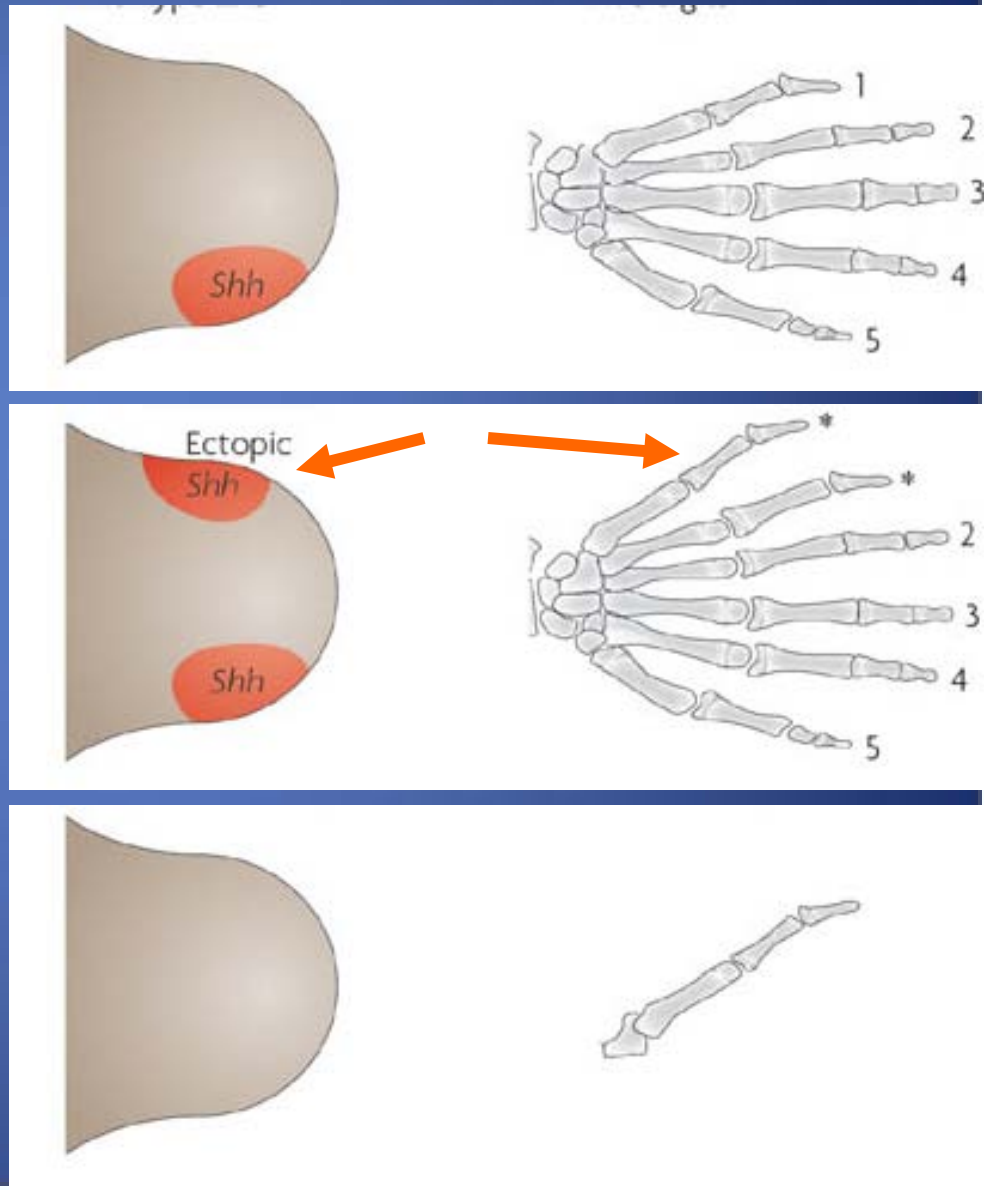
APICAL EPIDERMAL RIDGE

- The AER is also responsible for interdigital apoptosis
- Separates the webbed hand

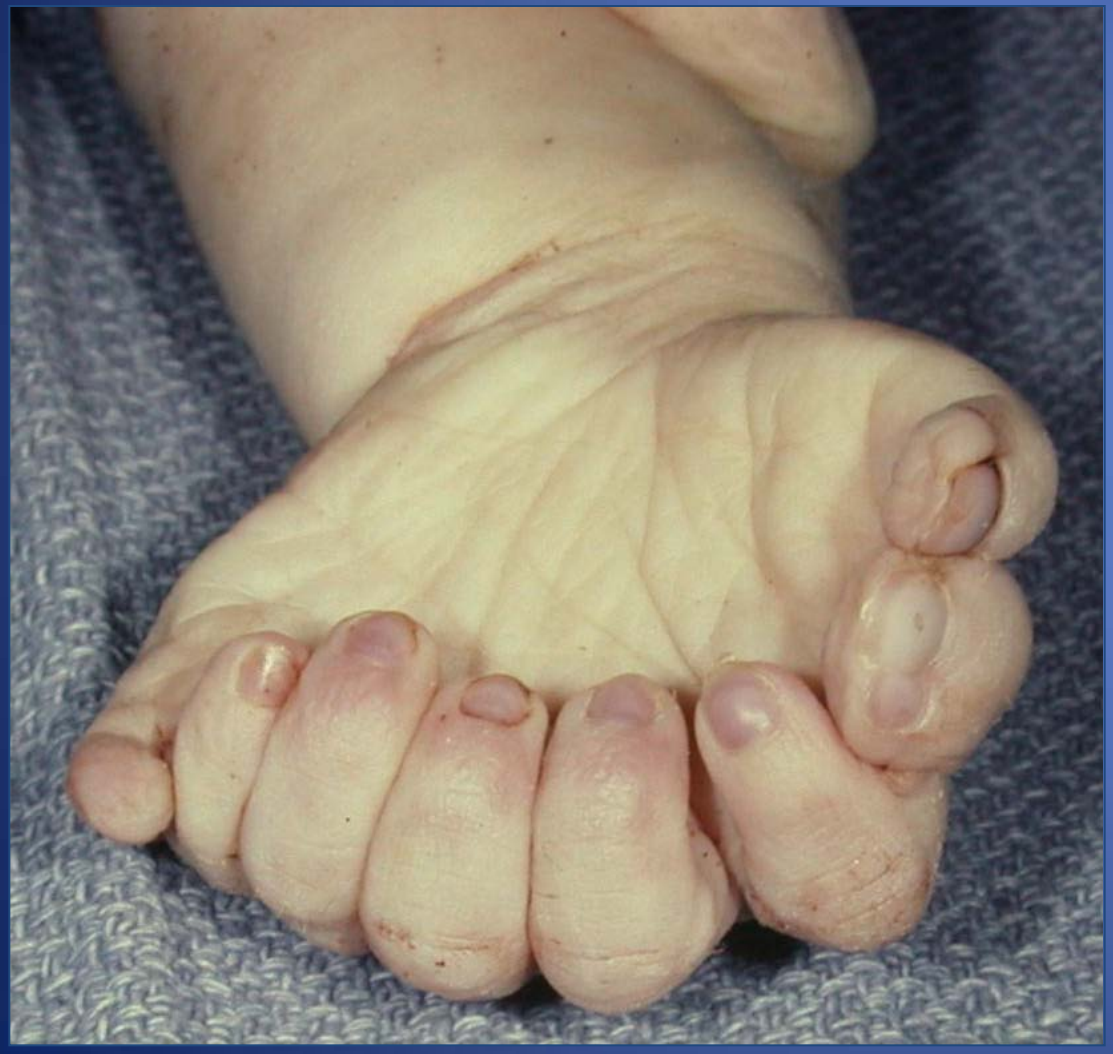
Abnormal ZPA Signaling

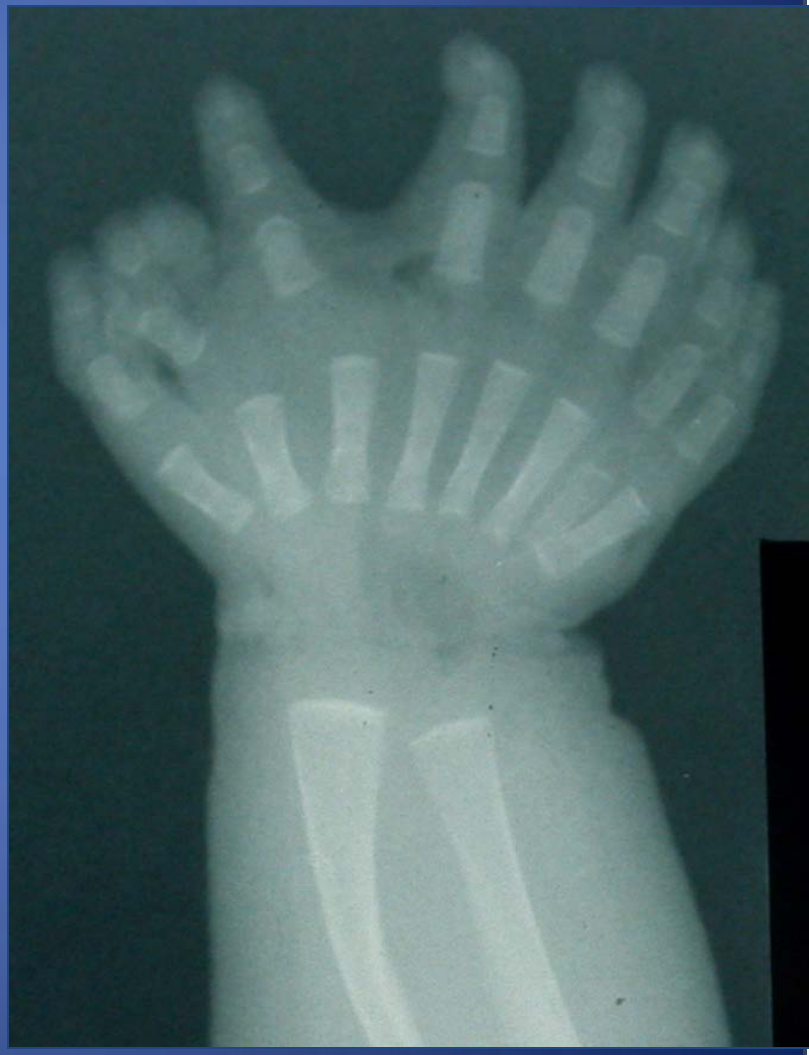
Transplantation of the ZPA or SHH anteriorly → duplication along the radioulnar axis

Remove the ZPA or SHH → fingers fail to form



Mirror Hand Deformity





CONGENITAL HAND ANOMALIES
Classification and Examples

I Failure of formation of parts

II Failure of differentiation

III Duplication

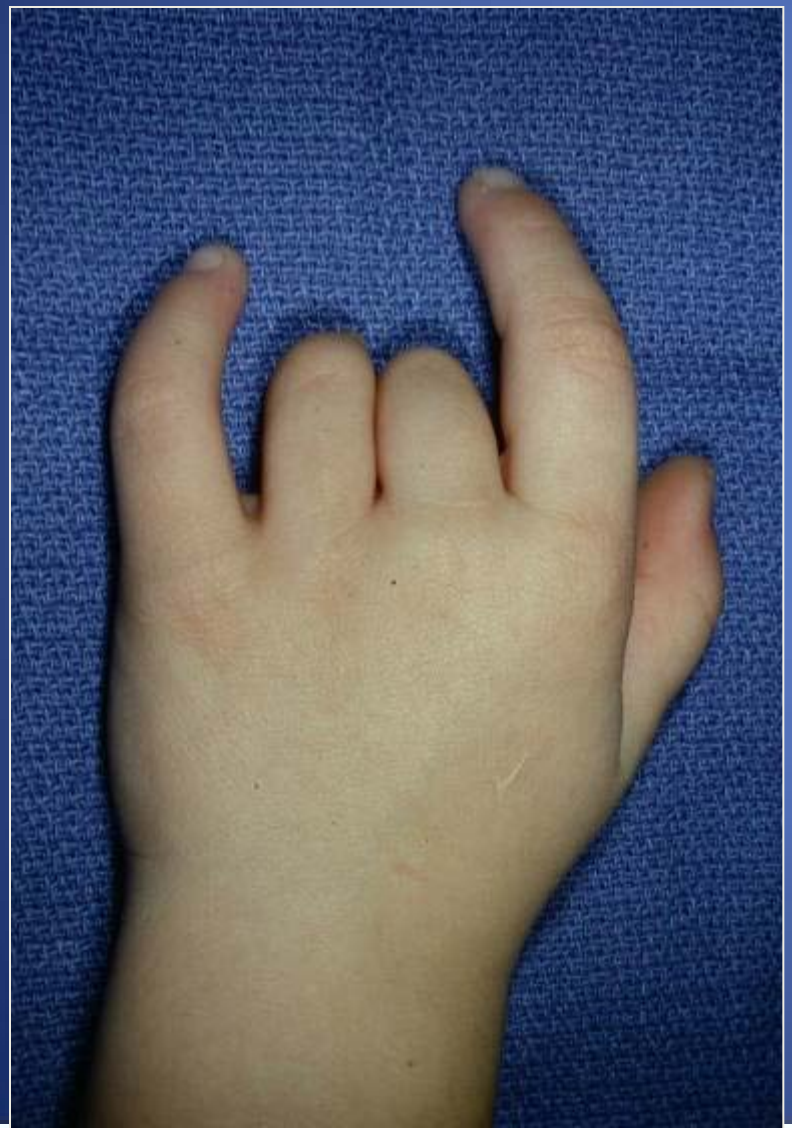
IV Overgrowth

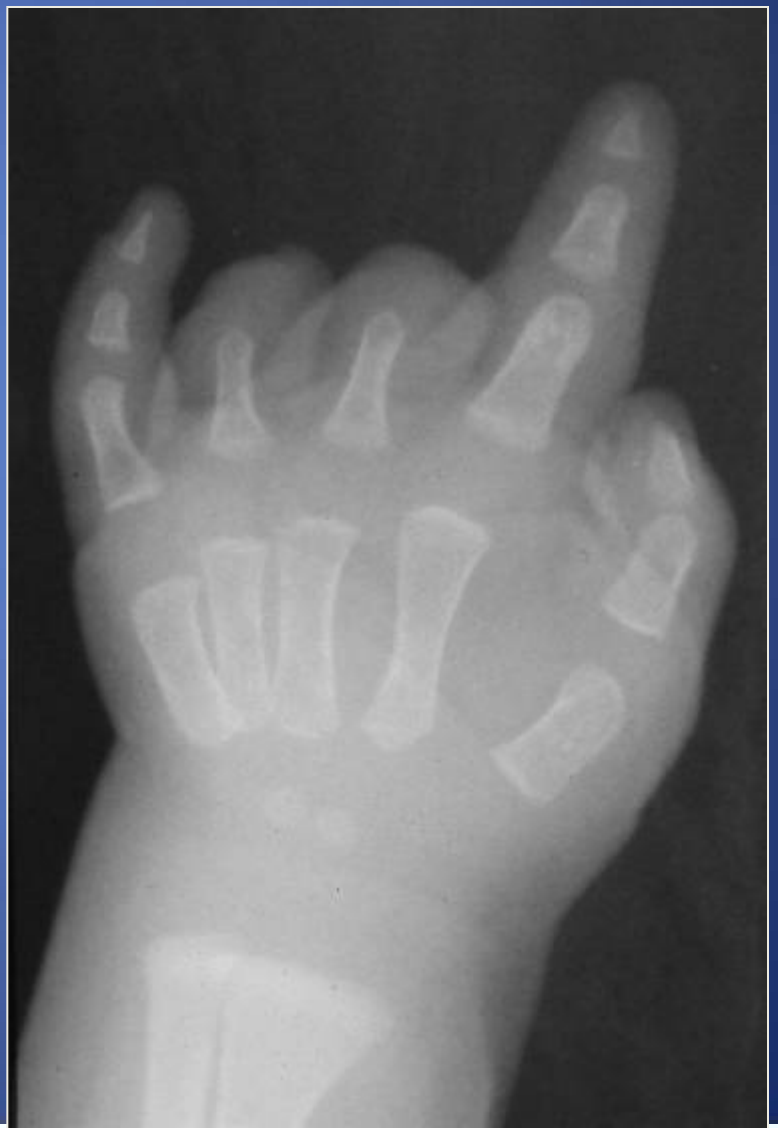
V Undergrowth

VI Constriction band syndrome

VII Generalized skeletal abnormalities

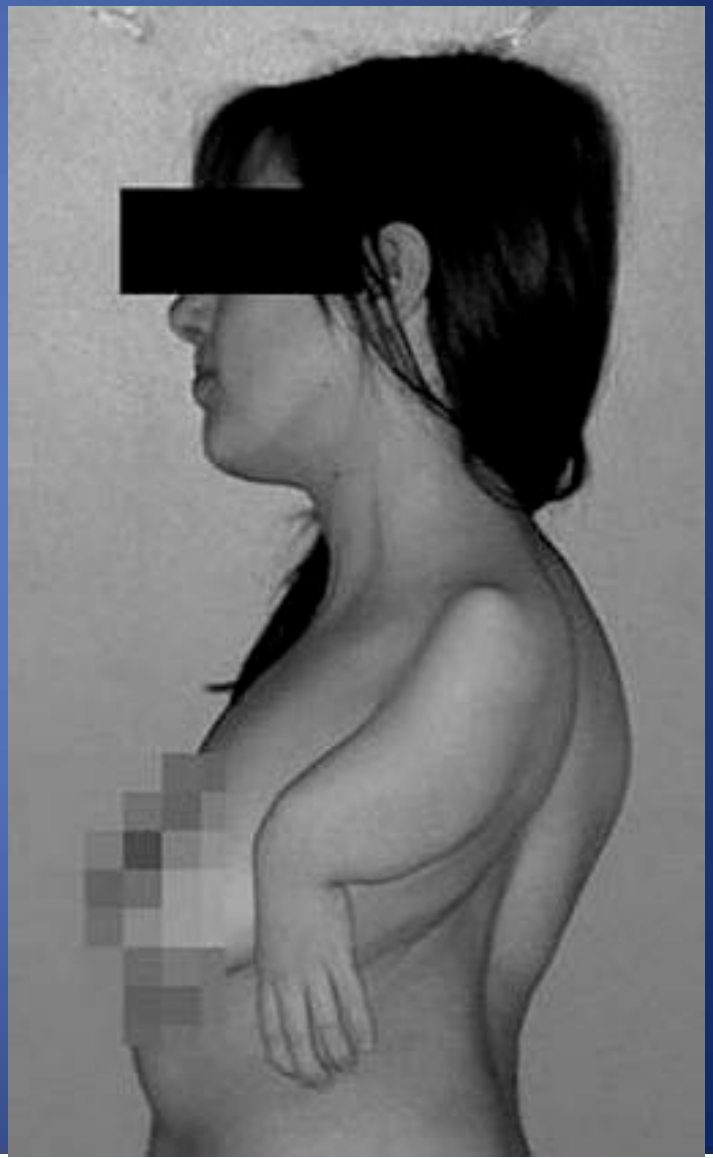
FAILURE OF FORMATION OF PARTS TRANSVERSE DEFICIENCY

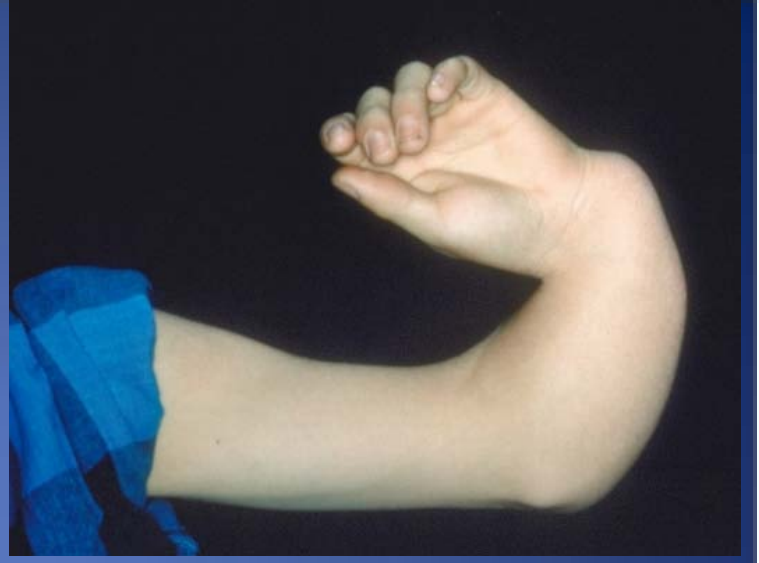




FAILURE OF FORMATION OF PARTS
LONGITUDINAL DEFICIENCY

- Complete
  - longitudinal failure
  - phocomelia
- Partial
  - Radial (preaxial)
  - Central
  - Ulnar (post-axial)

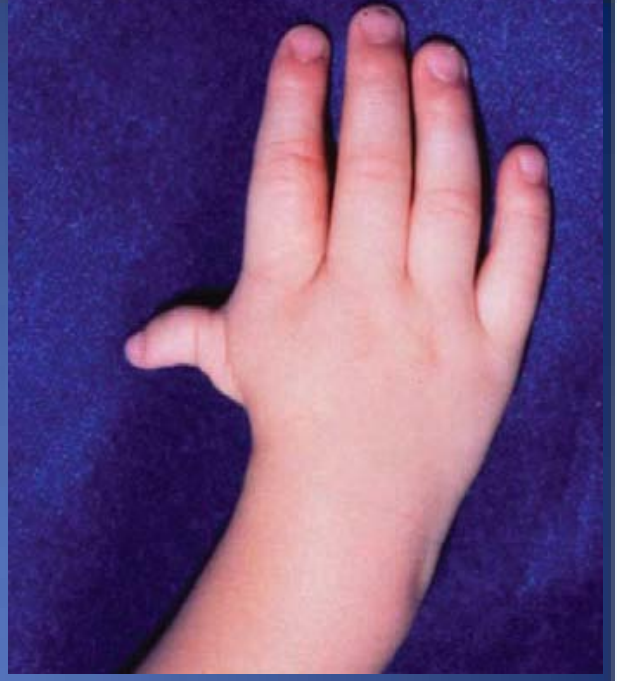


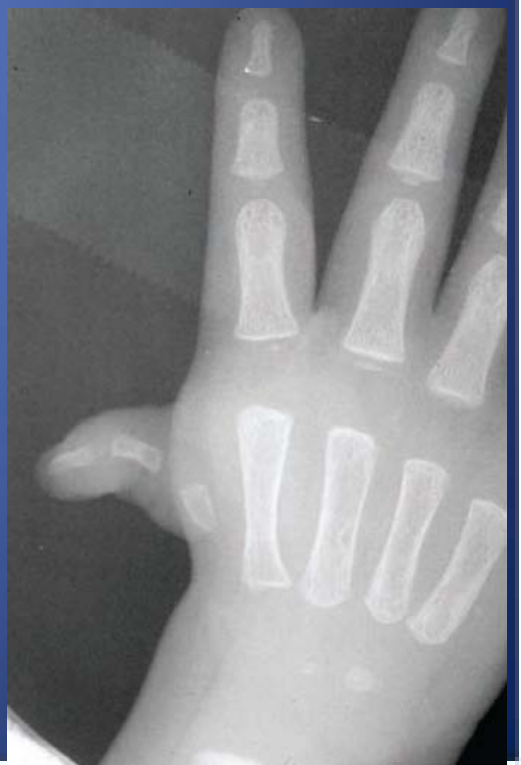


- Radial Longitudinal Deficiency
  - Associated conditions
    - Fanconi anemia
      - Lethal pancytopenia by 5-6 years
      - Need early diagnosis to search for bone marrow donor
    - Thrombocytopenia-absent radius (TAR)
      - Severe thrombocytopenia in infancy
      - Spontaneously improves after 1 year
    - VACTERL
      - Vertebral, anal, cardiac, tracheoesophageal, renal, radial, and lower limb anomalies
    - Holt-Oram
      - Cardiac anomalies, usually septal defects

137. A 10 month-old child presents with the deformity shown in Figures 1 and 2. Treatment should consist of:

- A. Carpometacarpal (CMC) joint stabilization, first web space reconstruction
- B. Metacarpal lengthening, metacarpophalangeal (MCP) joint stabilization, opponensplasty, first web space reconstruction
- C. Thumb ablation and toe to hand transfer
- D. Index finger pollicization
- E. Opponensplasty, MCP joint stabilization, first web space reconstruction





HYPOPLASTIC THUMB

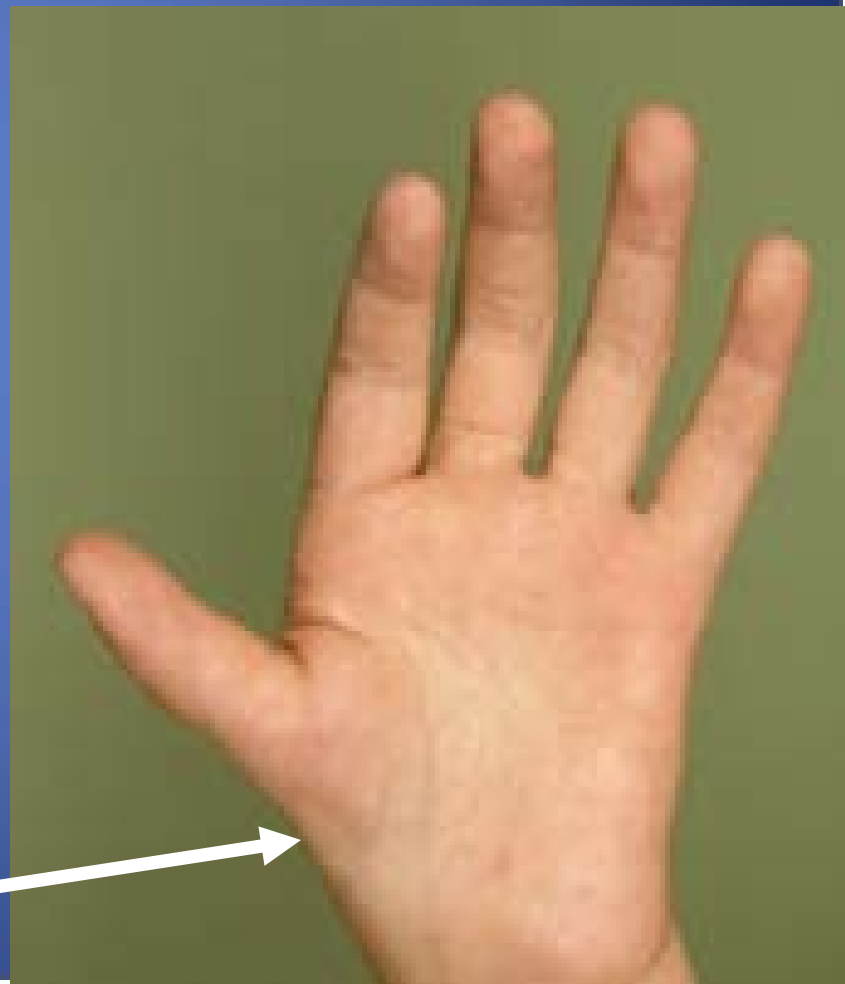
Grade I

Mild shortening

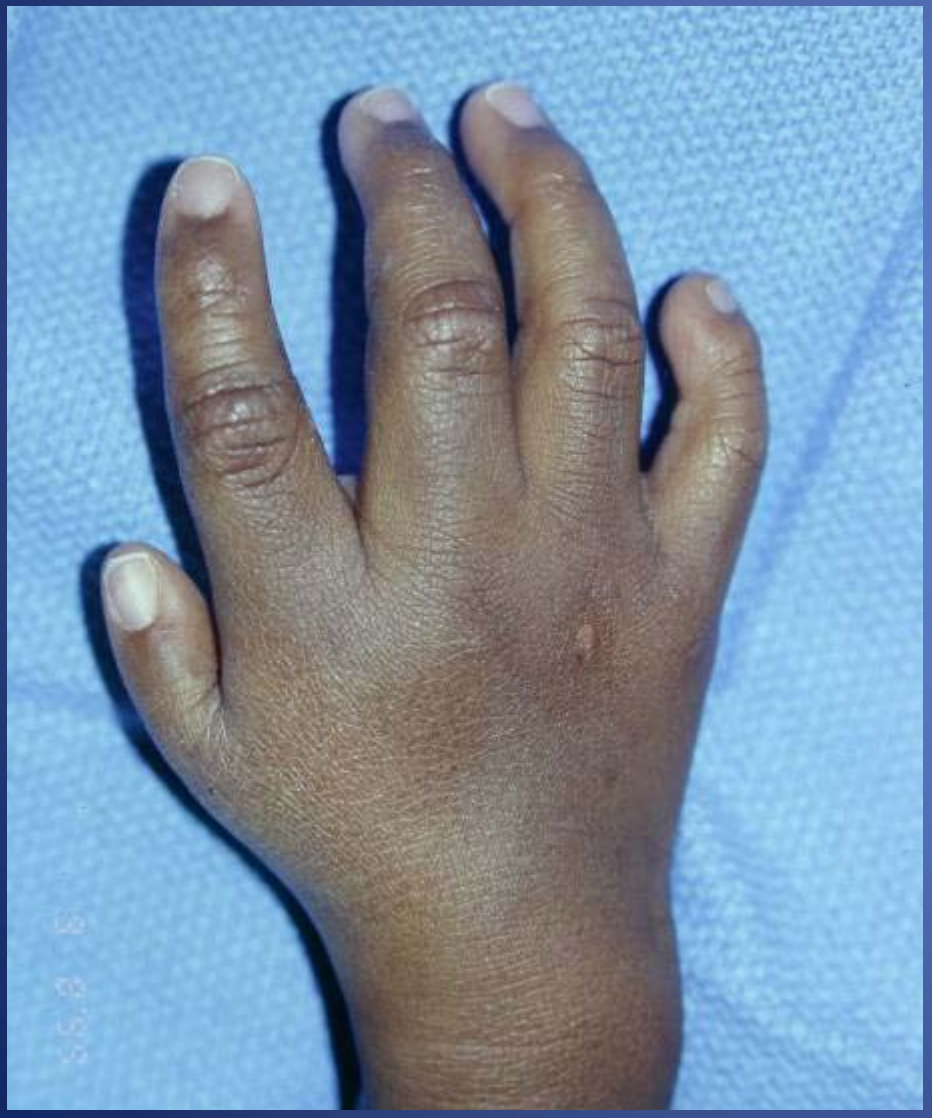
Grade II

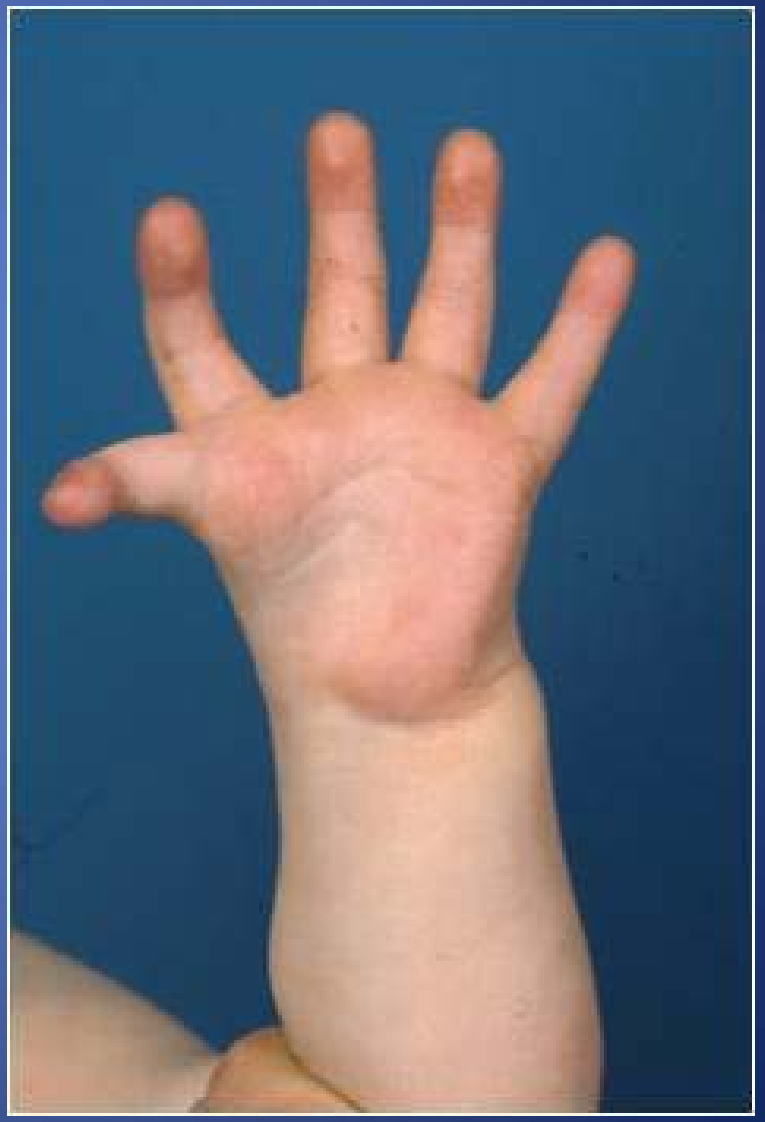
Moderate shortening

Thenar hypoplasia

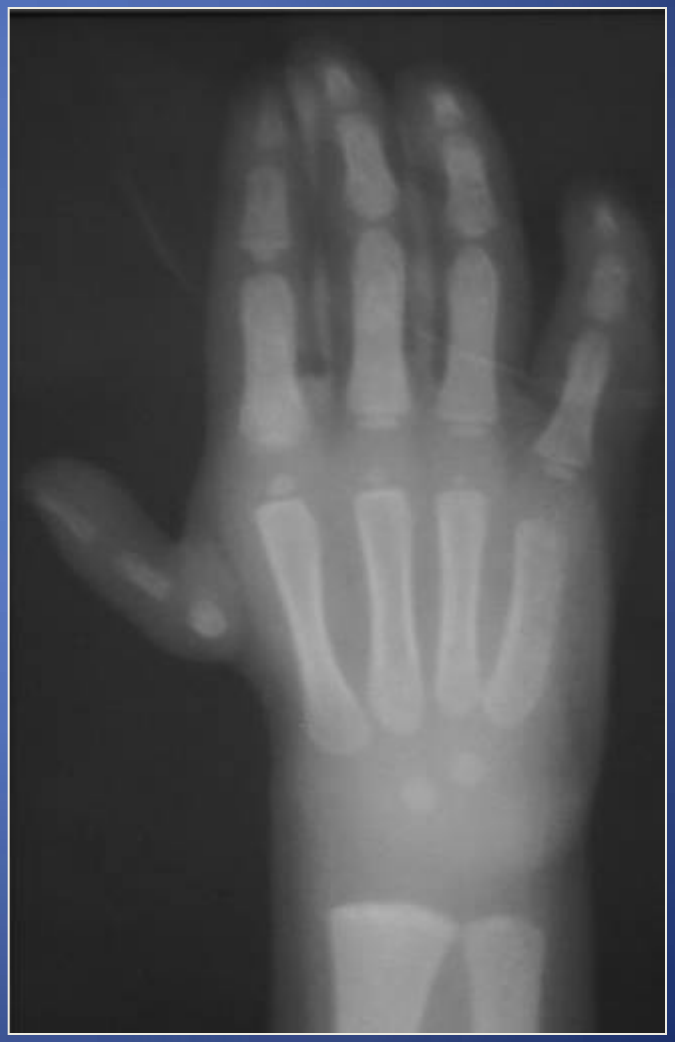


TYPE III HYPOPLASTIC THUMB





TYPE IV - FLOATING THUMB



TYPE V - ABSENT THUMB

- Lateral pinch grip between index and middle fingers
- Secondary rotation of index finger
- Pollicization improves grasp and tip-to-tip pinch

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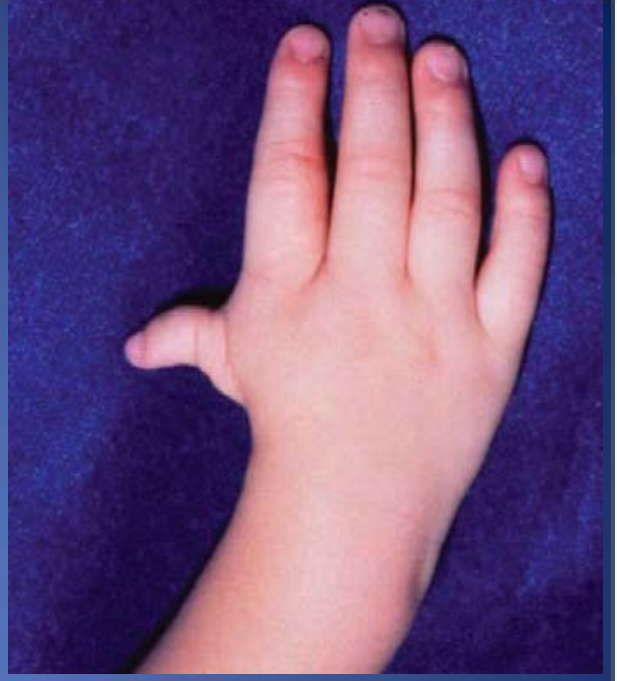
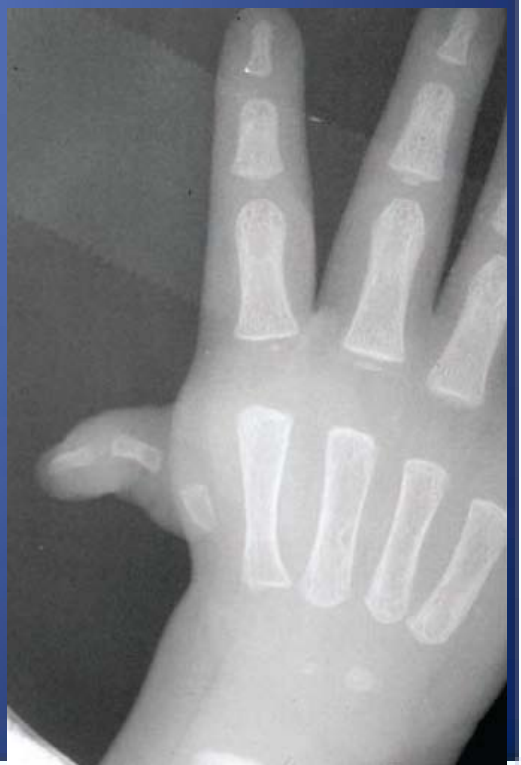
- E. Opponensplasty, MCP joint stabilization, first web space reconstruction

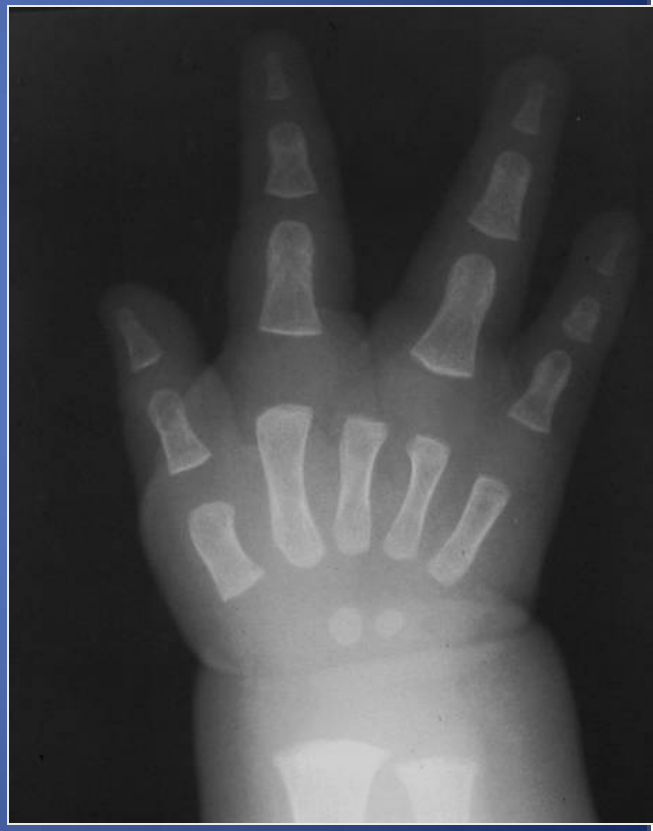




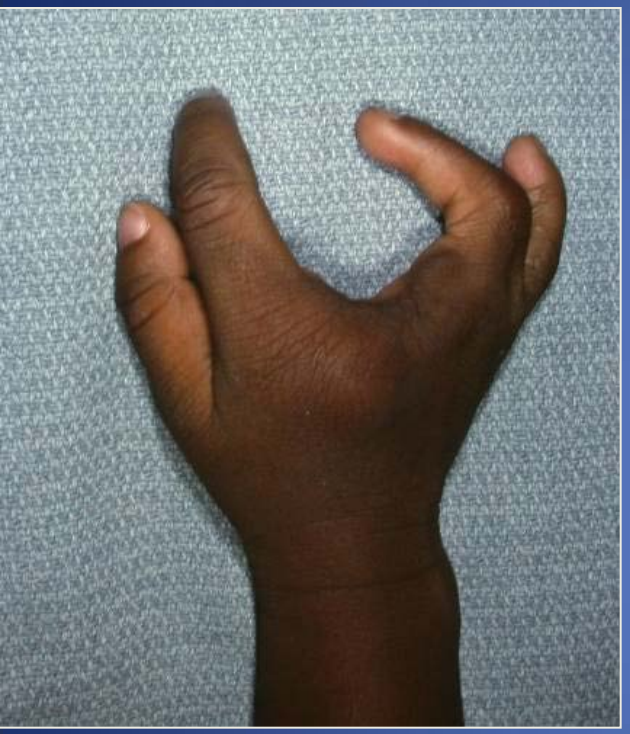
CLEFT HAND

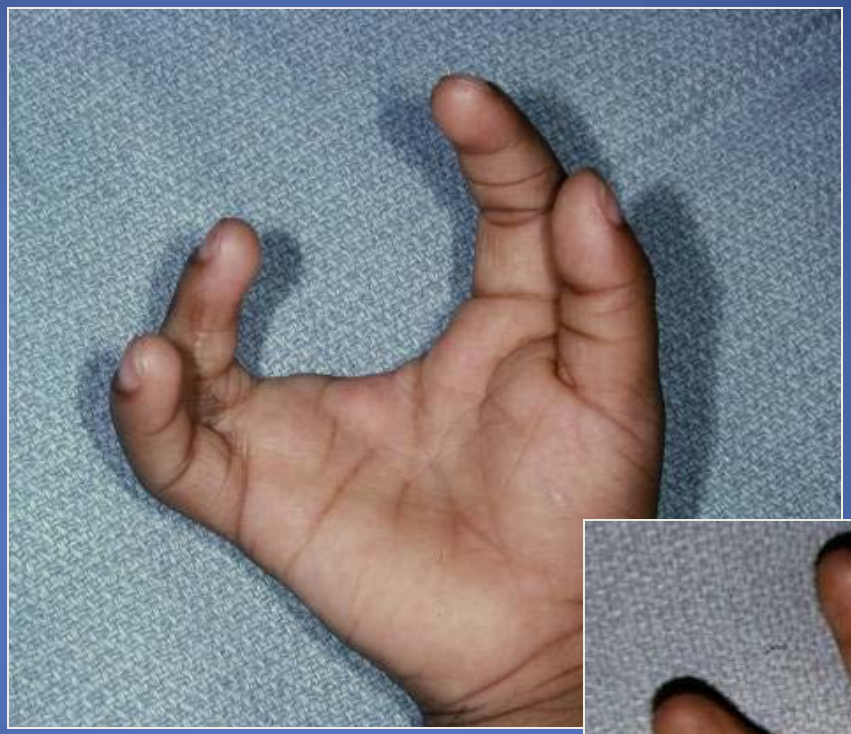
TYPICAL

- Absent middle finger
- Usually bilateral
- Assoc w/ cleft feet (50%)
- Assoc with cleft lip, palate



TYPICAL CLEFT HAND





CONGENITAL HAND ANOMALIES
Classification

I Failure of formation of parts

II Failure of differentiation

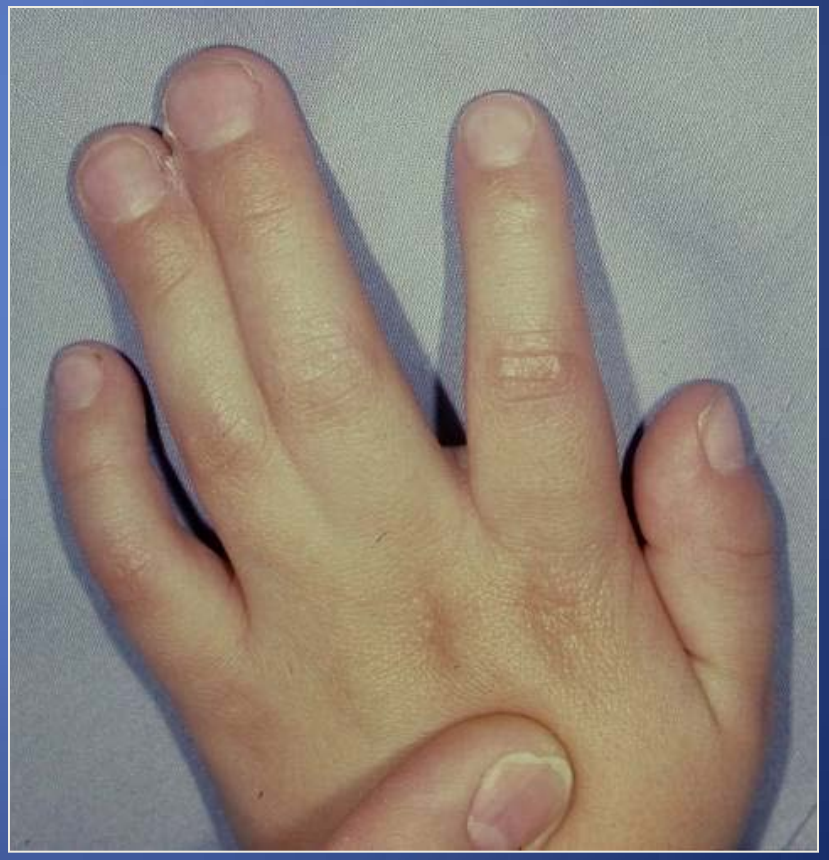
- Syndactyly
- Camptodactyly
- Clinodactyly
- Kirner deformity

SYNDACTYLY

- 1 in 2000 live birth, sporadic vs. familial (10%)
- 50% bilateral
- Middle-ring finger web - most common
- Classification
  - Webbing: “complete” vs “incomplete”
  - Bone: not involved = “simple”
    - involved = “complex”

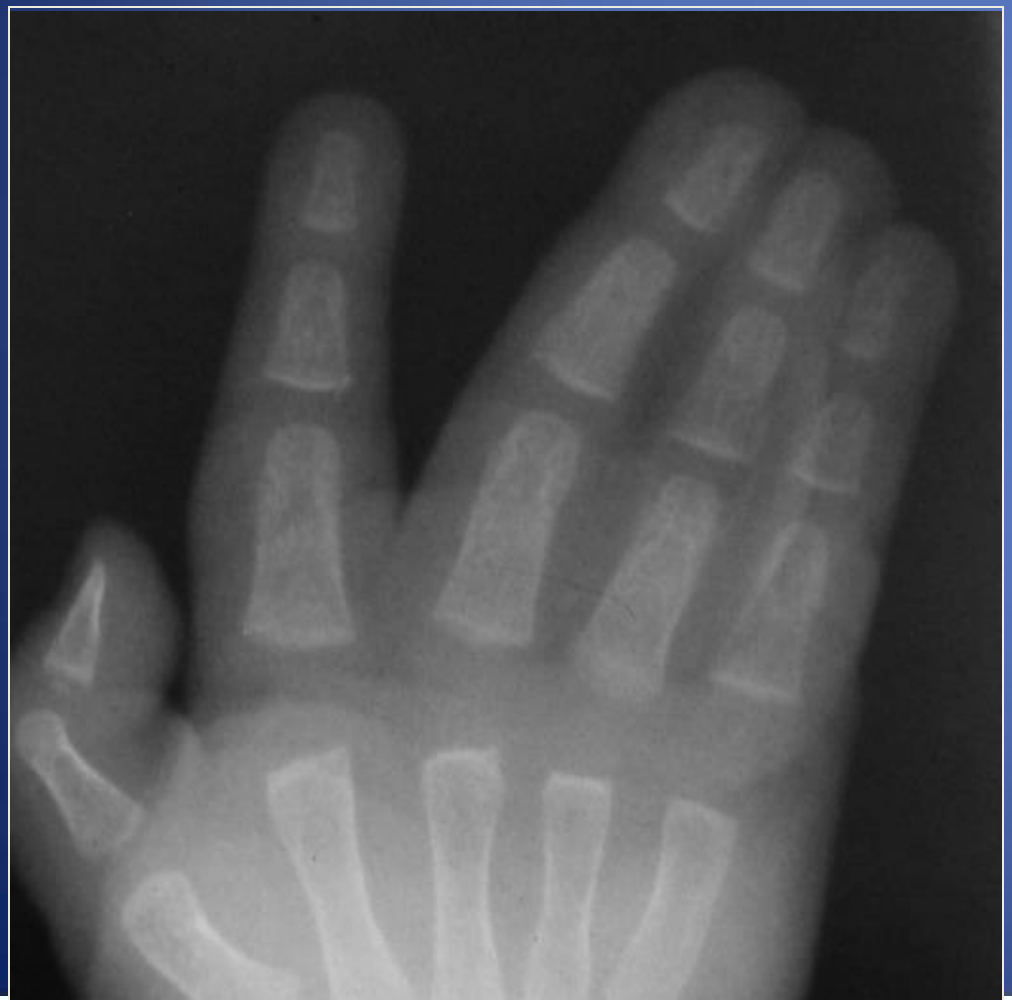
SIMPLE SYNDACTYLY

Incomplete

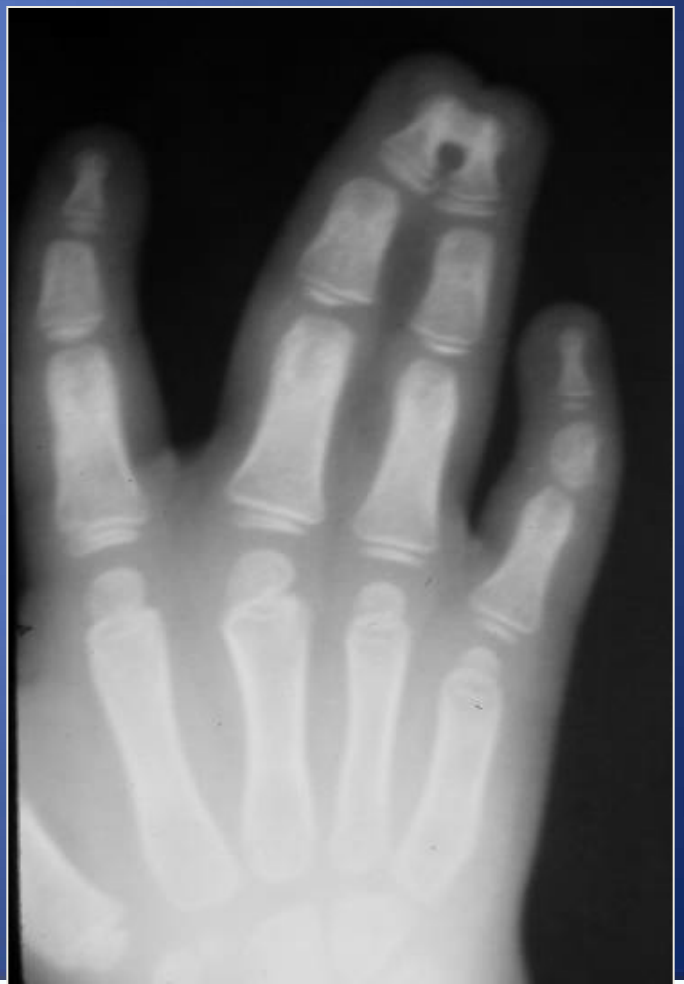


Complete

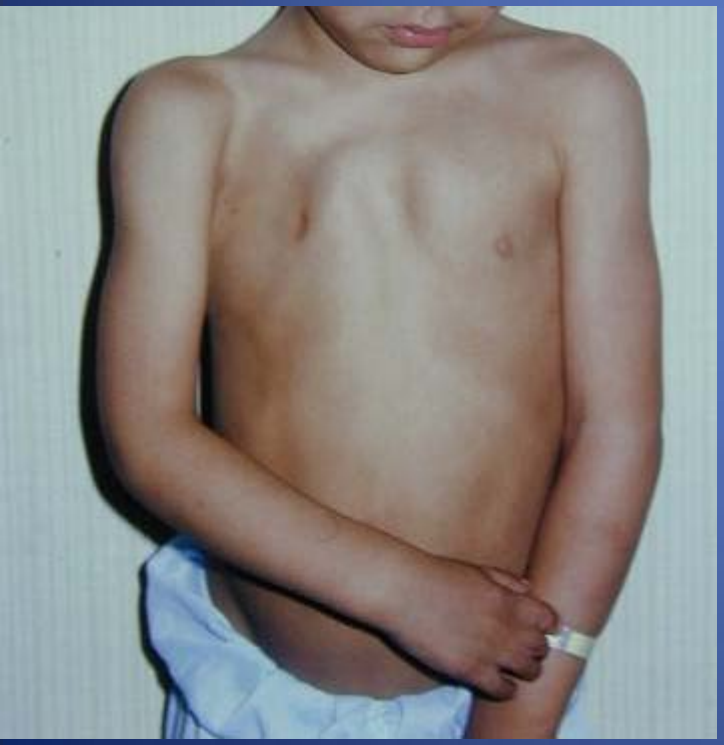
SIMPLE
COMPLETE
SYNDACTYLY
RF tethering MF

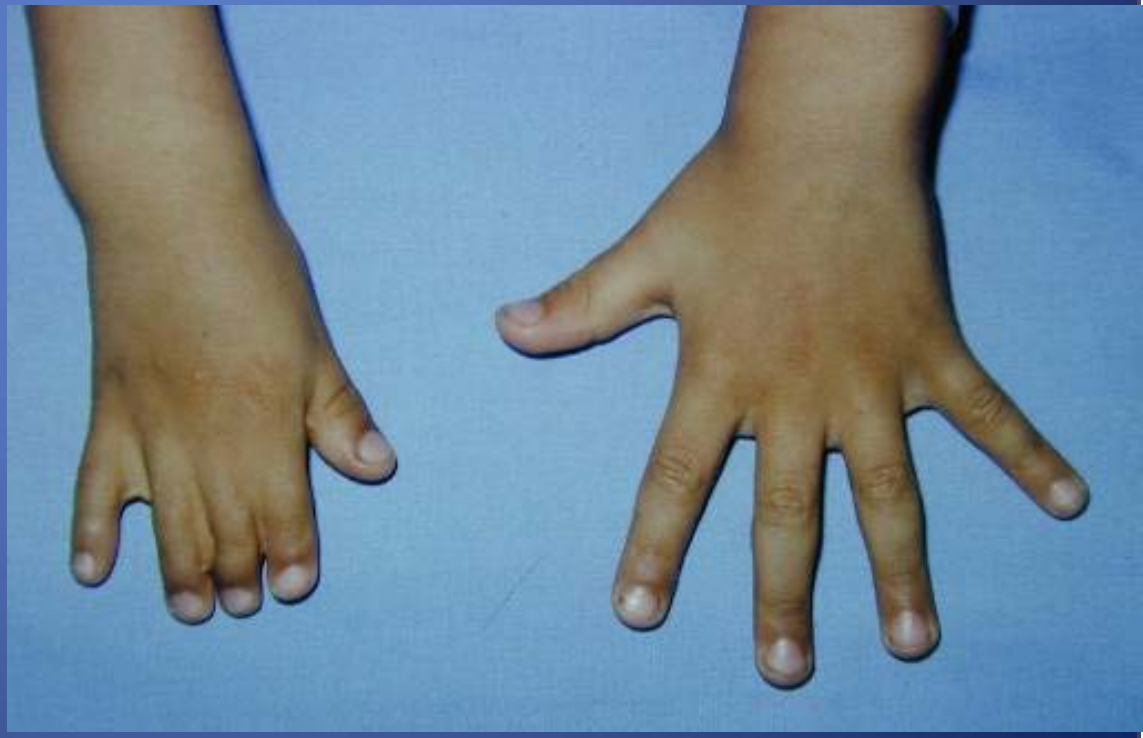


COMPLEX
COMPLETE
SYNDACTYLY



POLAND'S SYNDROME
symbrachydactyly
chest wall deformity
Thought to be vascular anomaly





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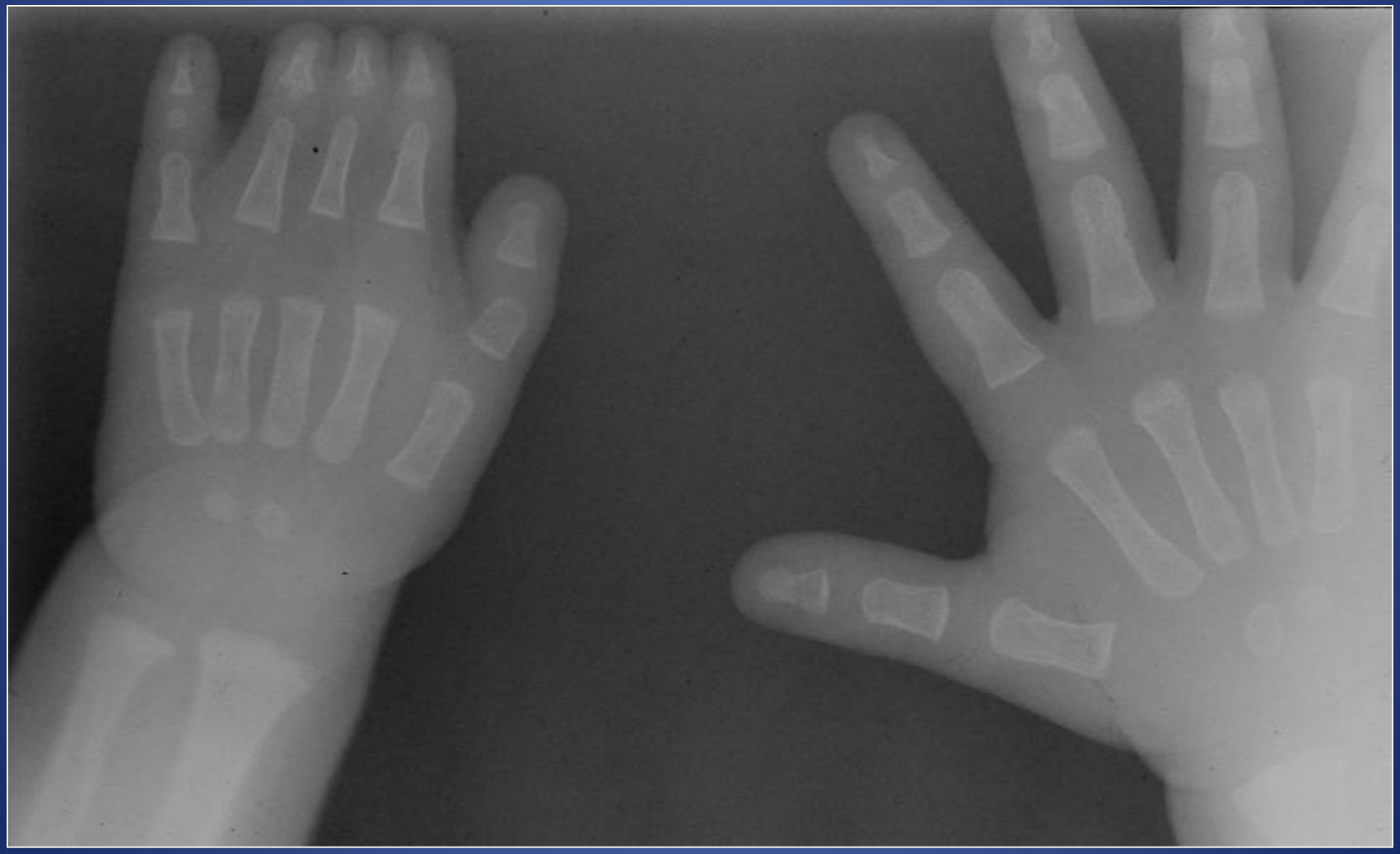
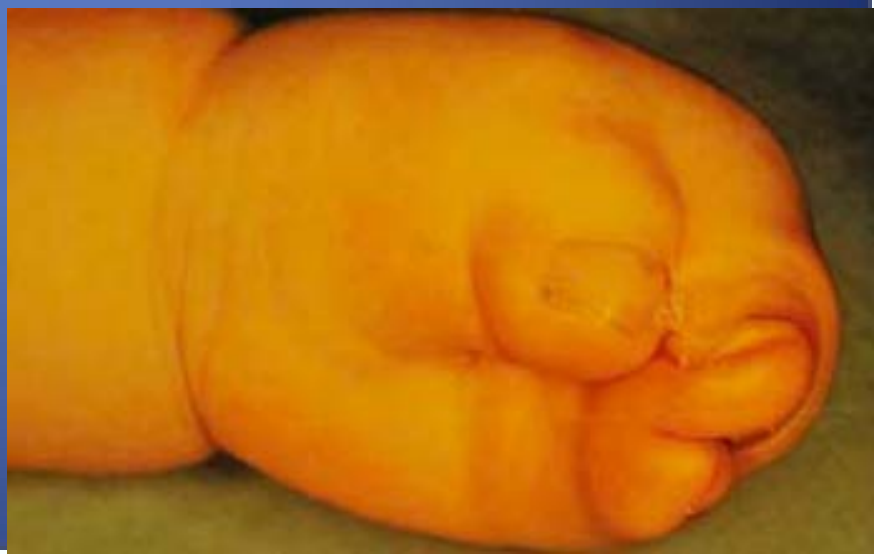


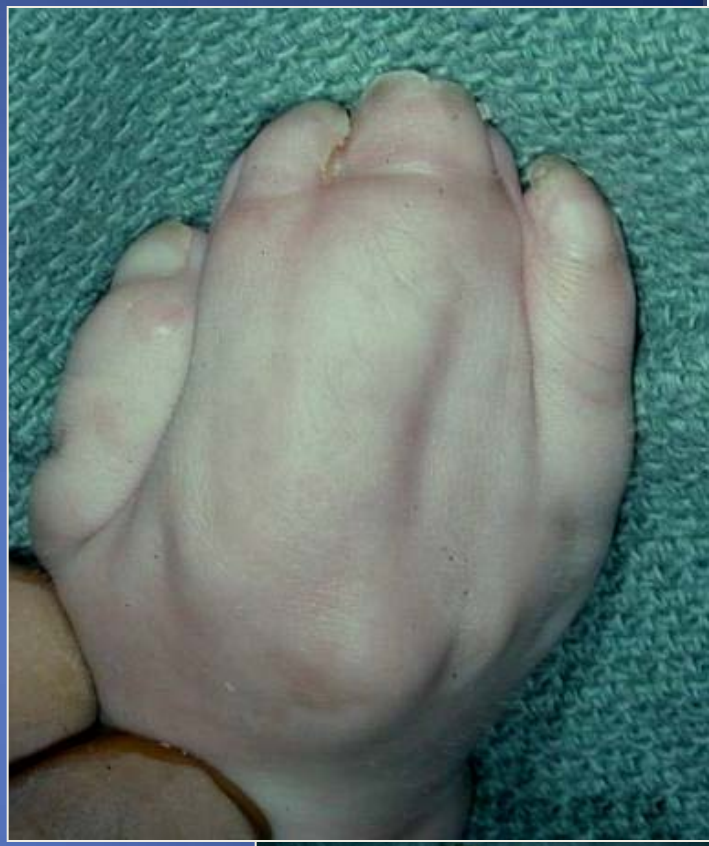
71. The infant whose hands are shown in Figure 1 is expected to have a mutation in which gene?

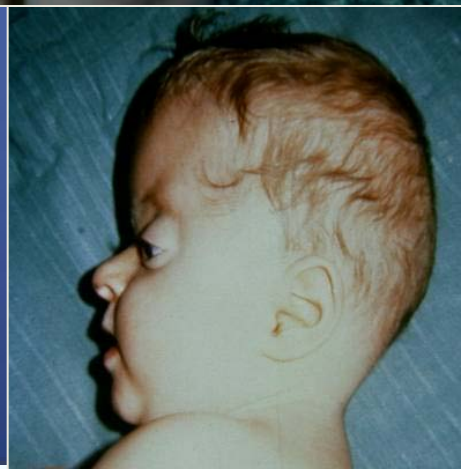
- A. FGF receptor 2 (FGFR2)
- B. Homeobox D13 (HOXD13)
- C. Holt-Oram syndrome 1 (HOS1)
- D. Human zinc factor protein gene 3 (GLI3)
- E. Brachyury T-box 5 (TBX5)

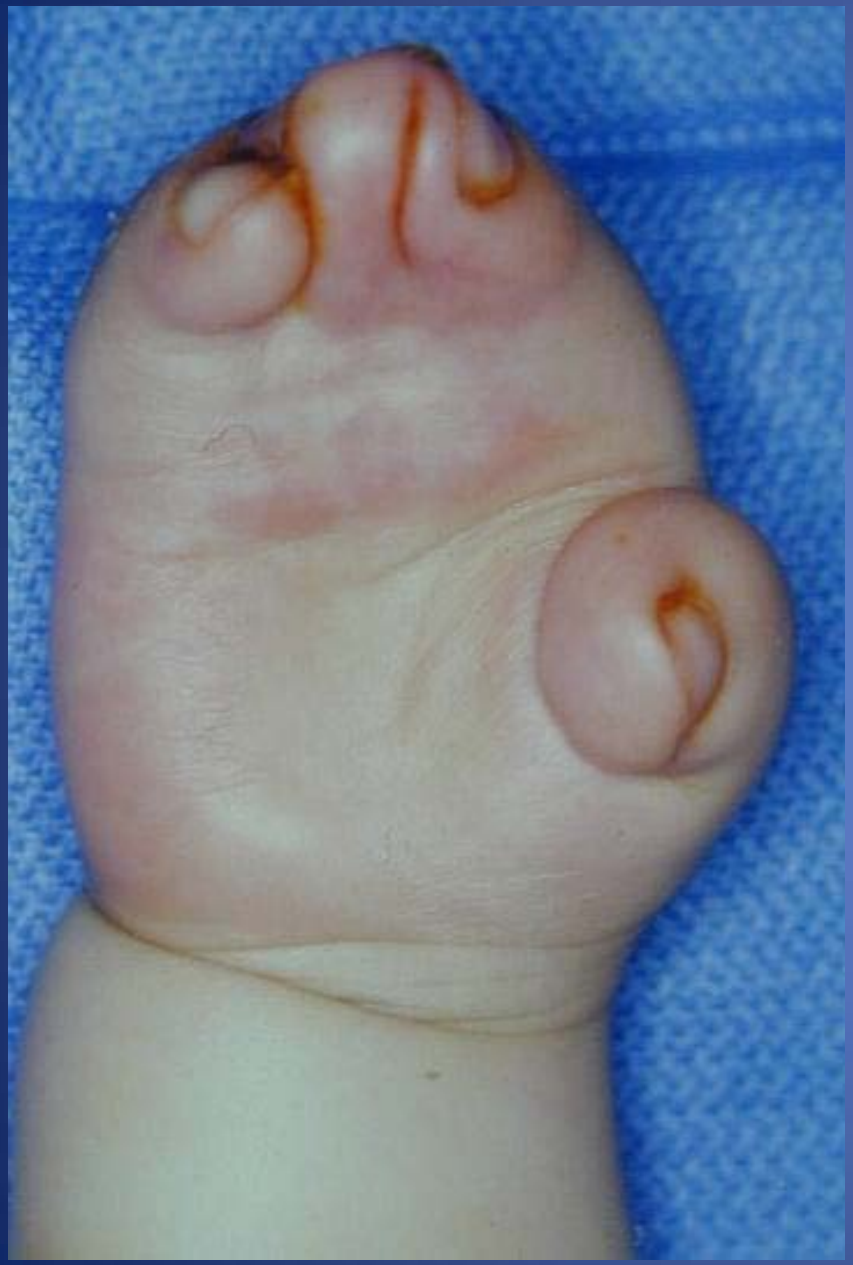


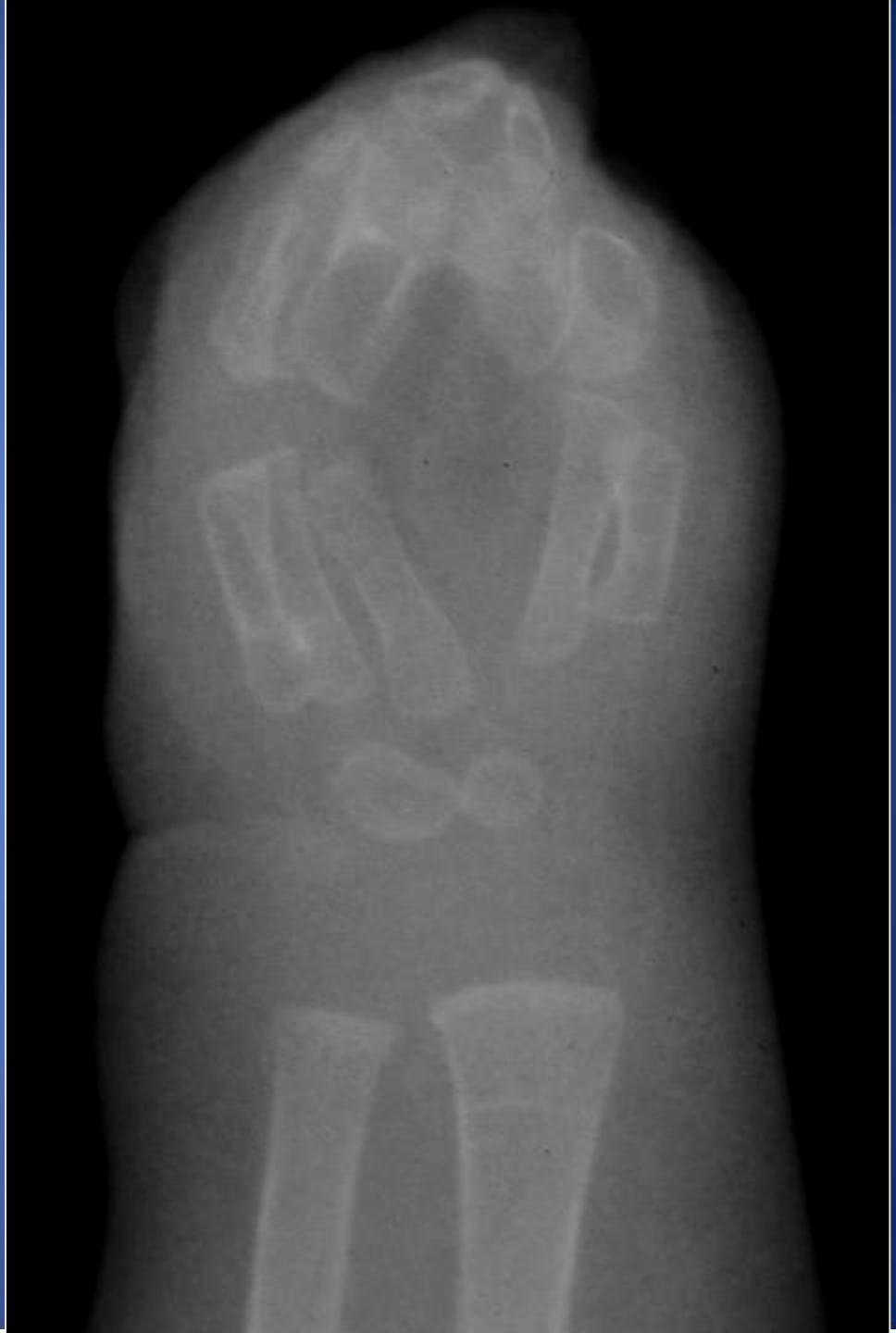
ACROCEPHALOSYNDACTYLY

- APERT SYNDROME
- Mitten or spoon hand
- FGFR 2
- Common nail index, middle & ring fingers
- Complex bony deformities









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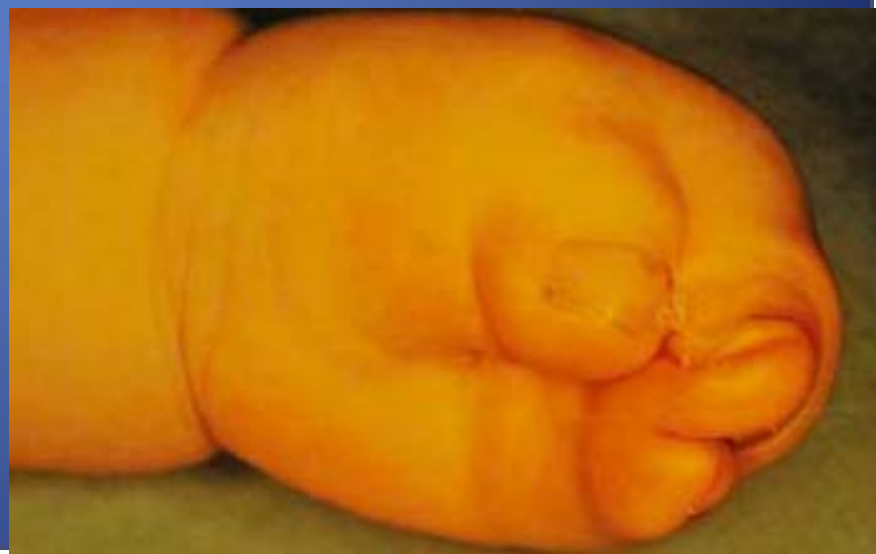
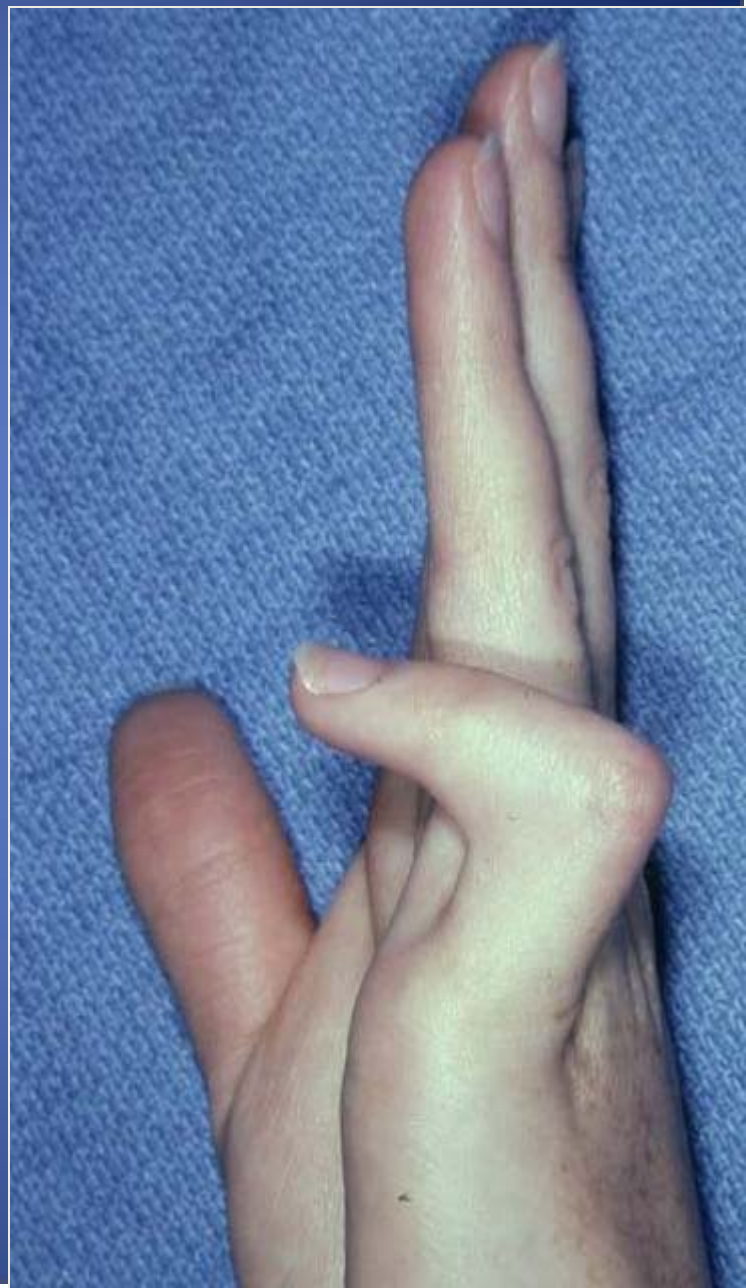


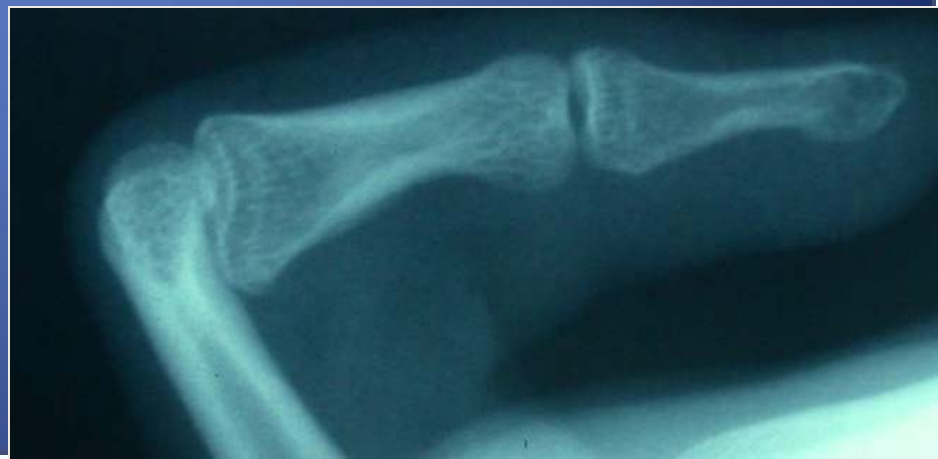
CAMPTODACTYLY

- Flexion contracture PIP joint
- Small finger - 90%
- ? Abnormal insertion of lumbrical muscle
- Bilateral - 66%
- Short FDS
- All structures probably involved



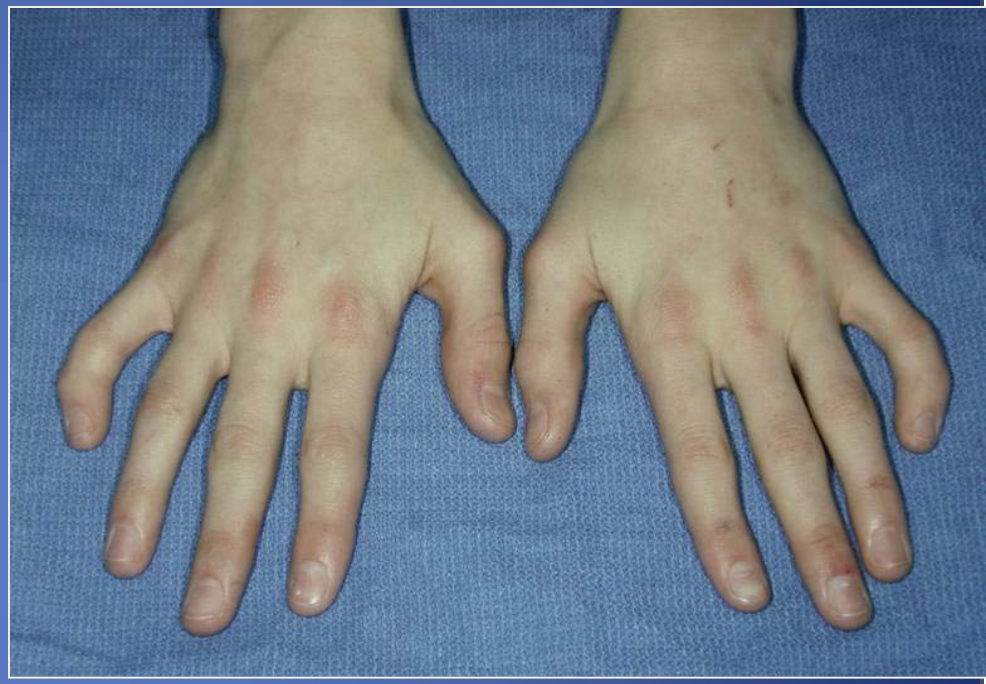
CAMPTODACTYLY Treatment

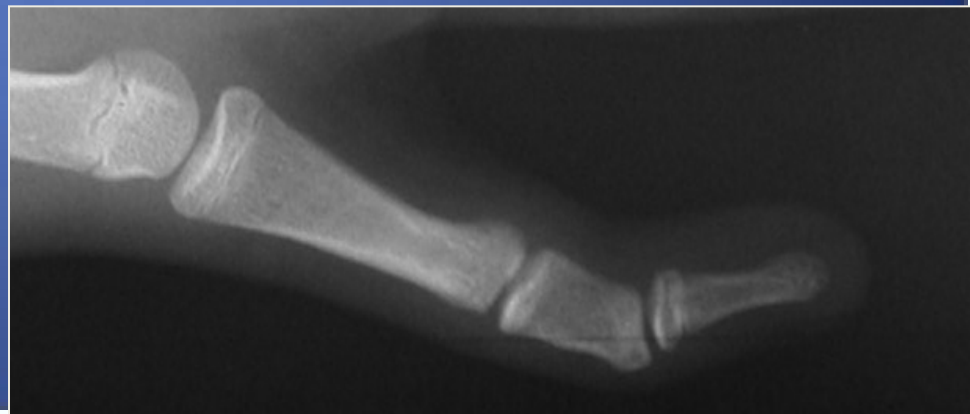
- Advise parents to accept the deformity
- Passive stretching, serial splinting rarely successful
- Consider surgery:
  - Young child with severe contracture
  - Rapid progression
- ???earlier intervention before fixed deformity



CLINODACTYLY

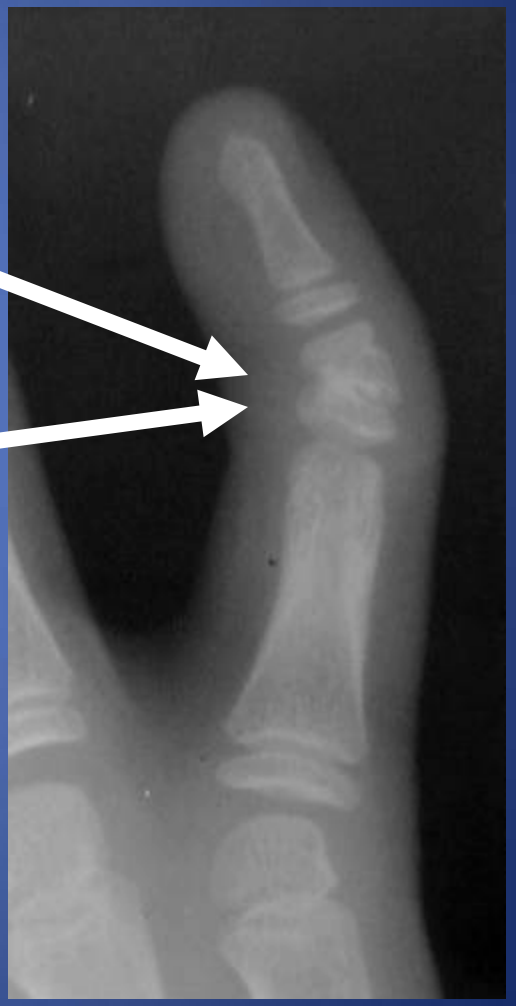
- **Radial-ulnar curvature**
- **Usually small finger**
- **Usually radial deviation**
- **Delta phalanx**





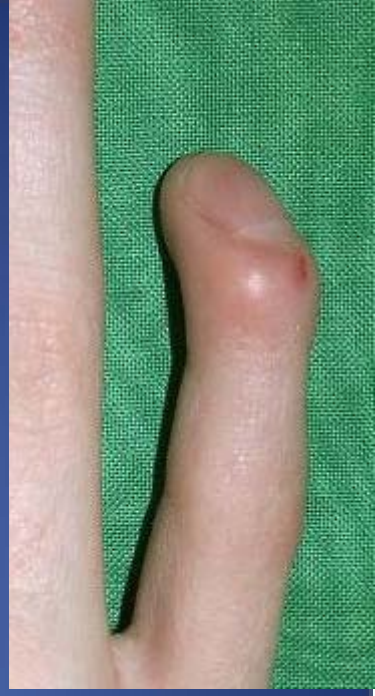
DELTA PHALANX

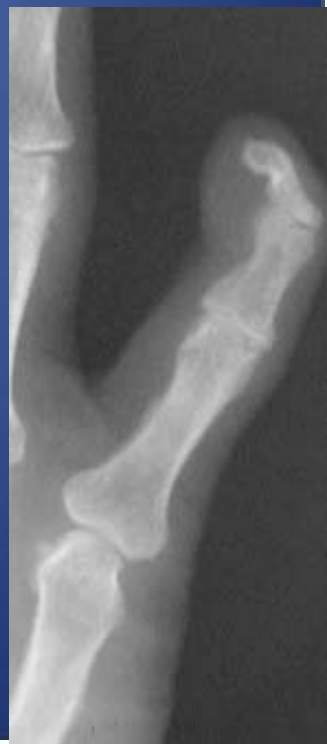
- Trapezoidal-shaped phalanx
- Abnormal C-shaped epiphysis
- Usually middle phalanx of the small finger



KIRNER DEFORMITY

- Palmar-radial curvature, rotation
- Distal phalanx, usually bilateral
- Associated musculoskeletal anomalies
- No functional limitations usually
- Observe
- Splint
- Correctional osteotomy





CONGENITAL HAND ANOMALIES
Classification

I Failure of formation of parts

II Failure of differentiation

III Duplication polydactyly

- Pre-axial = radial Thumb
- Central Index, middle, ring
- Post-axial=ulnar Small finger

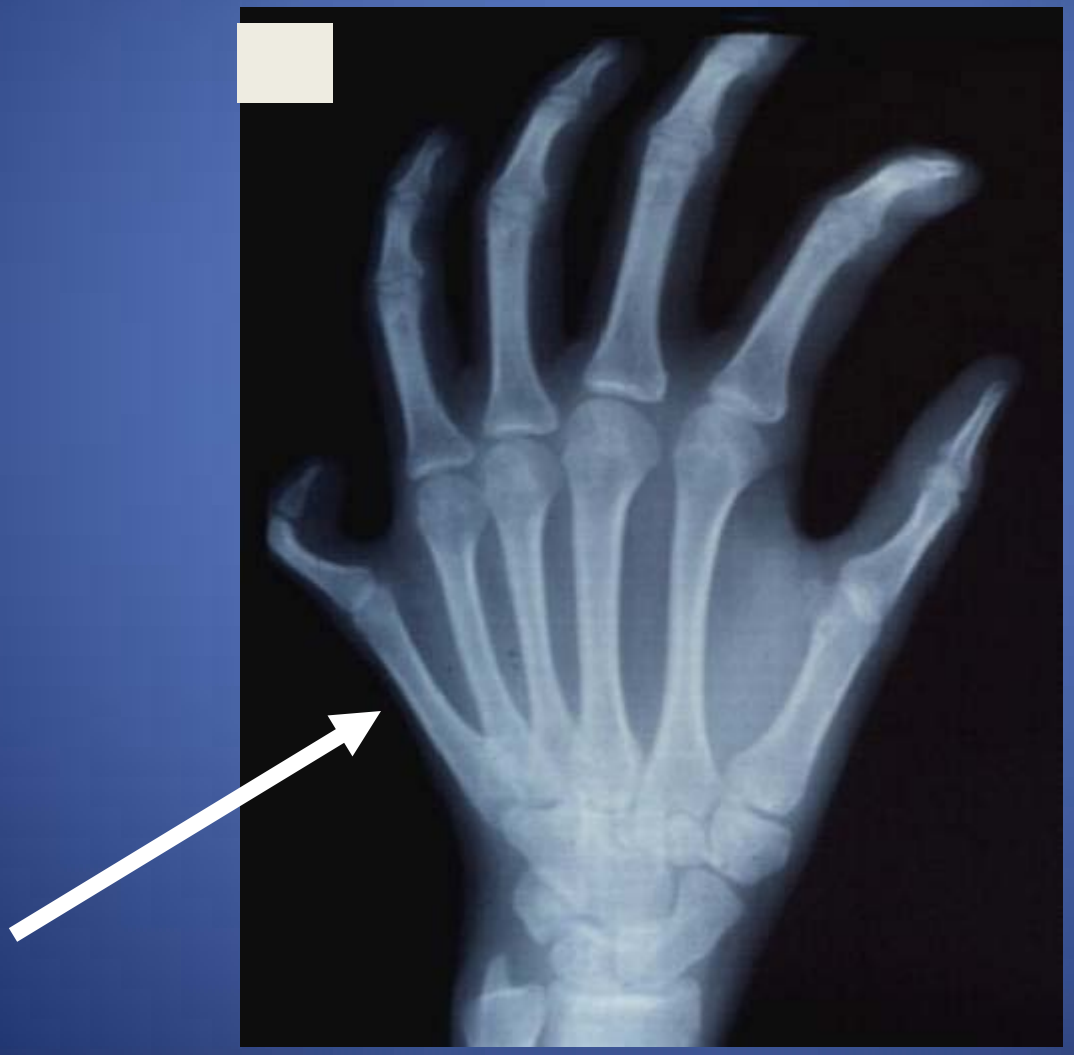
POLYDACTYLY

- Small finger is most commonly involved
- More common in African-American infants (incidence = 1 in 300)
- More common in females

SMALL FINGER POLYDACTYLY
Type 1 – Rudimentary Soft Tissue

SMALL FINGER POLYDACTYLY

Type III – Metacarpal Duplicated

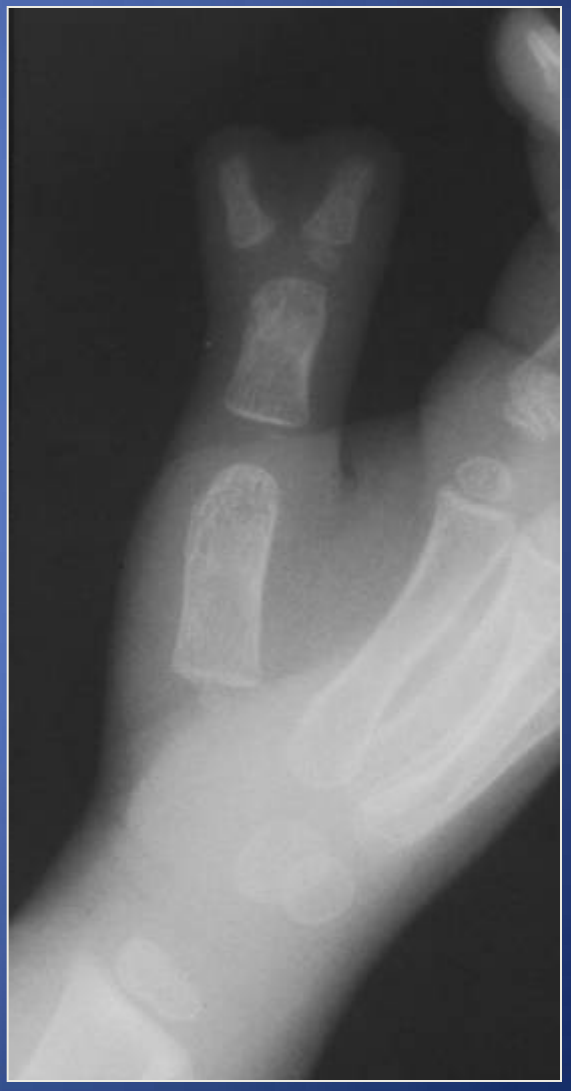


THUMB POLYDACTYLY
“Duplicate Thumb”

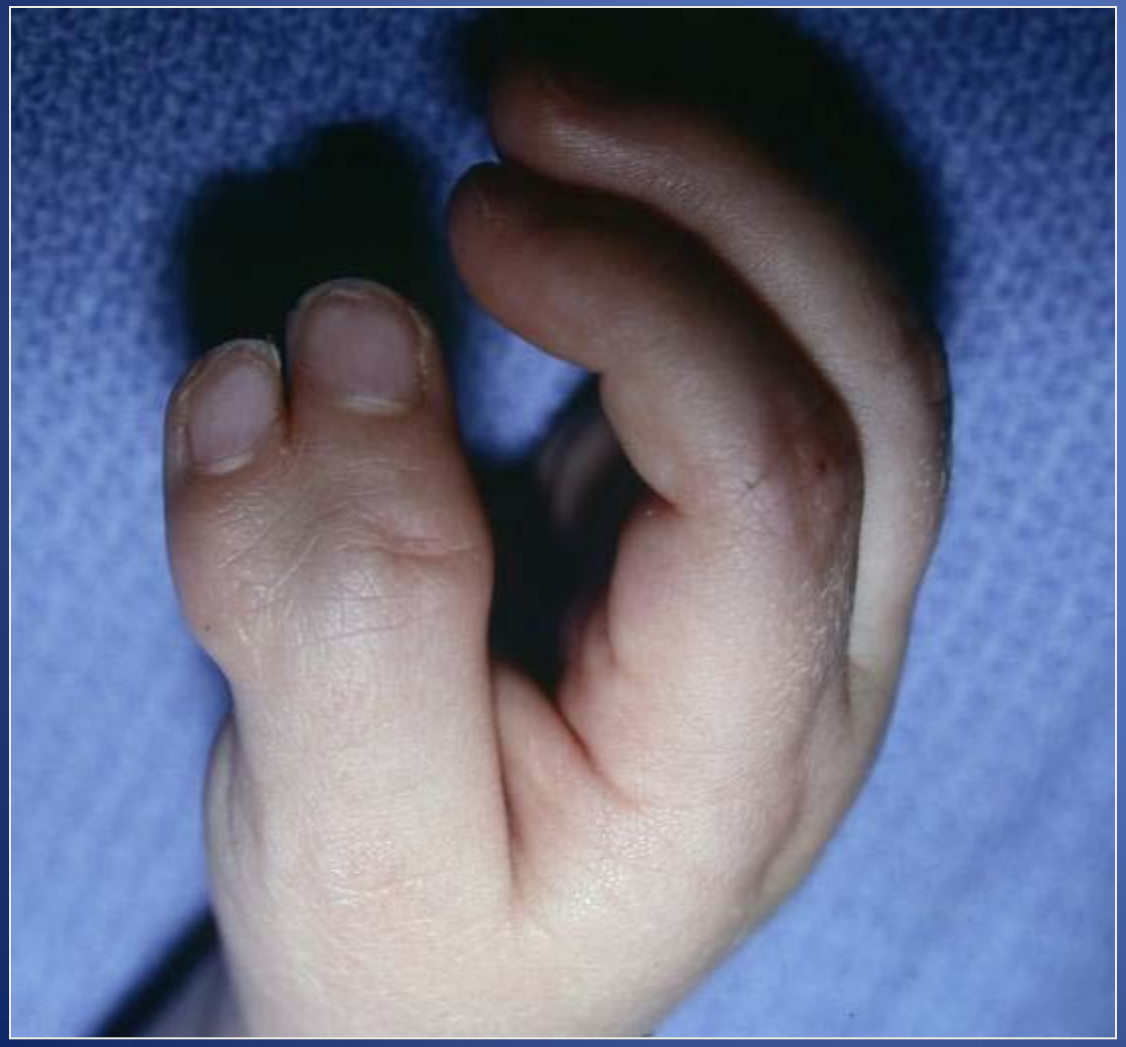
- Most common congenital anomaly----Ikuta
- Second to syndactyly------Cohen

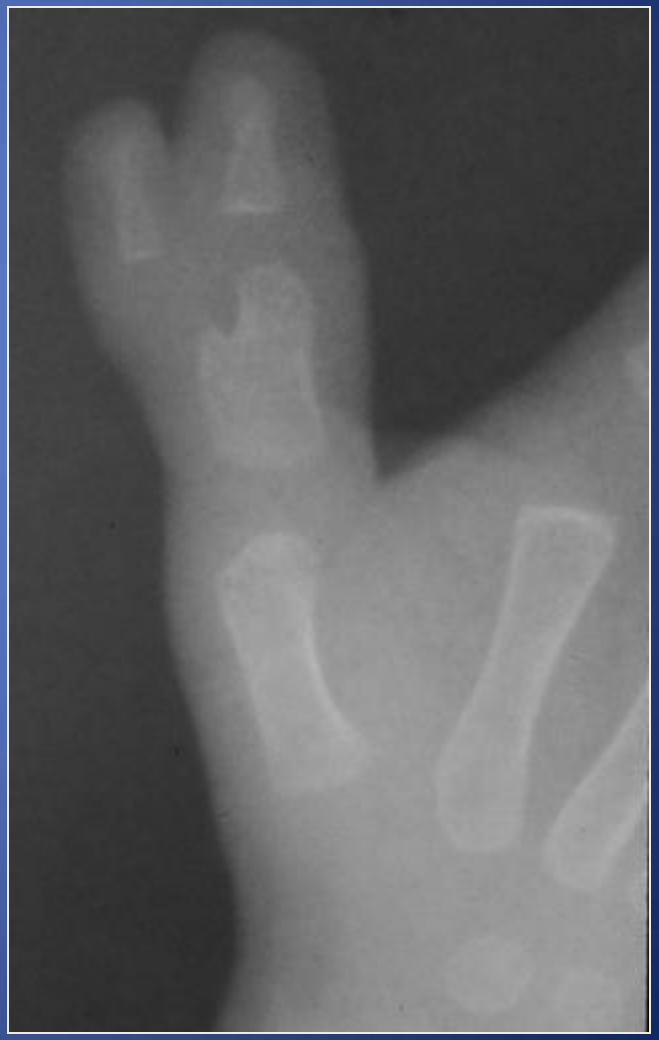
Types	I	II	III	IV	V	VI	VII
							
Bifurcation level	Distal phalanx	IP joint	Proximal phalanx	MP joint	Metacarpus	CM joint	Floating
Cases %	16 5.3%	61 20.1%	14 4.6%	97 31.9%	12 3.9%	25 8.2%	42 13.8%

THUMB DUPLICATION
Type 2

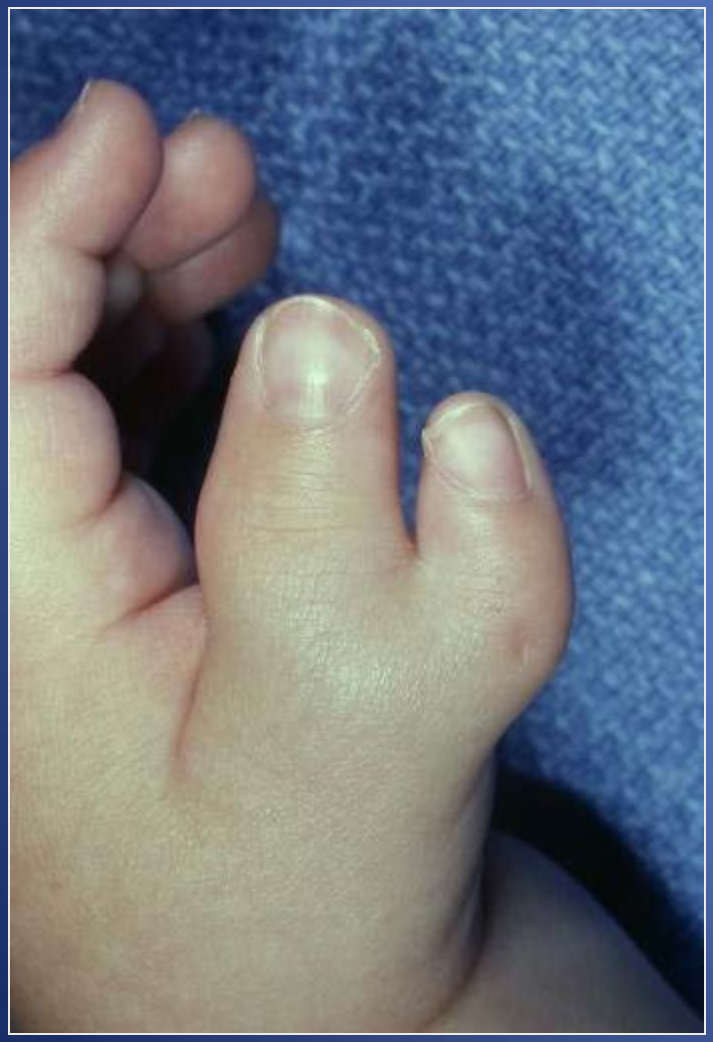


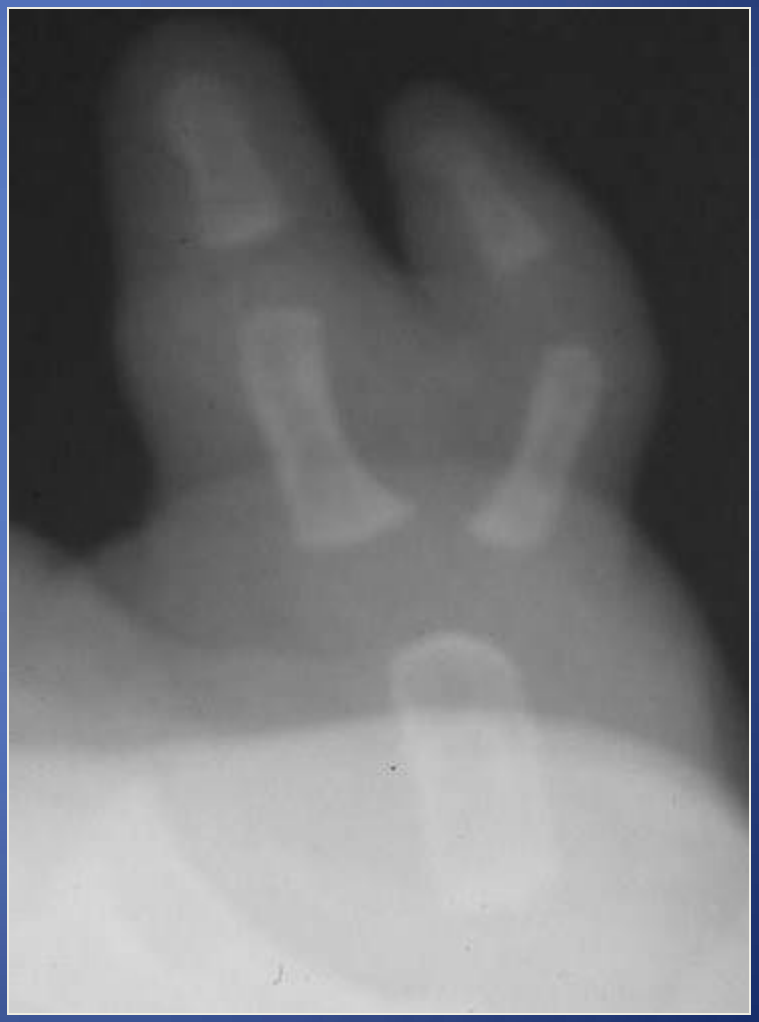
THUMB DUPLICATION
Type 3



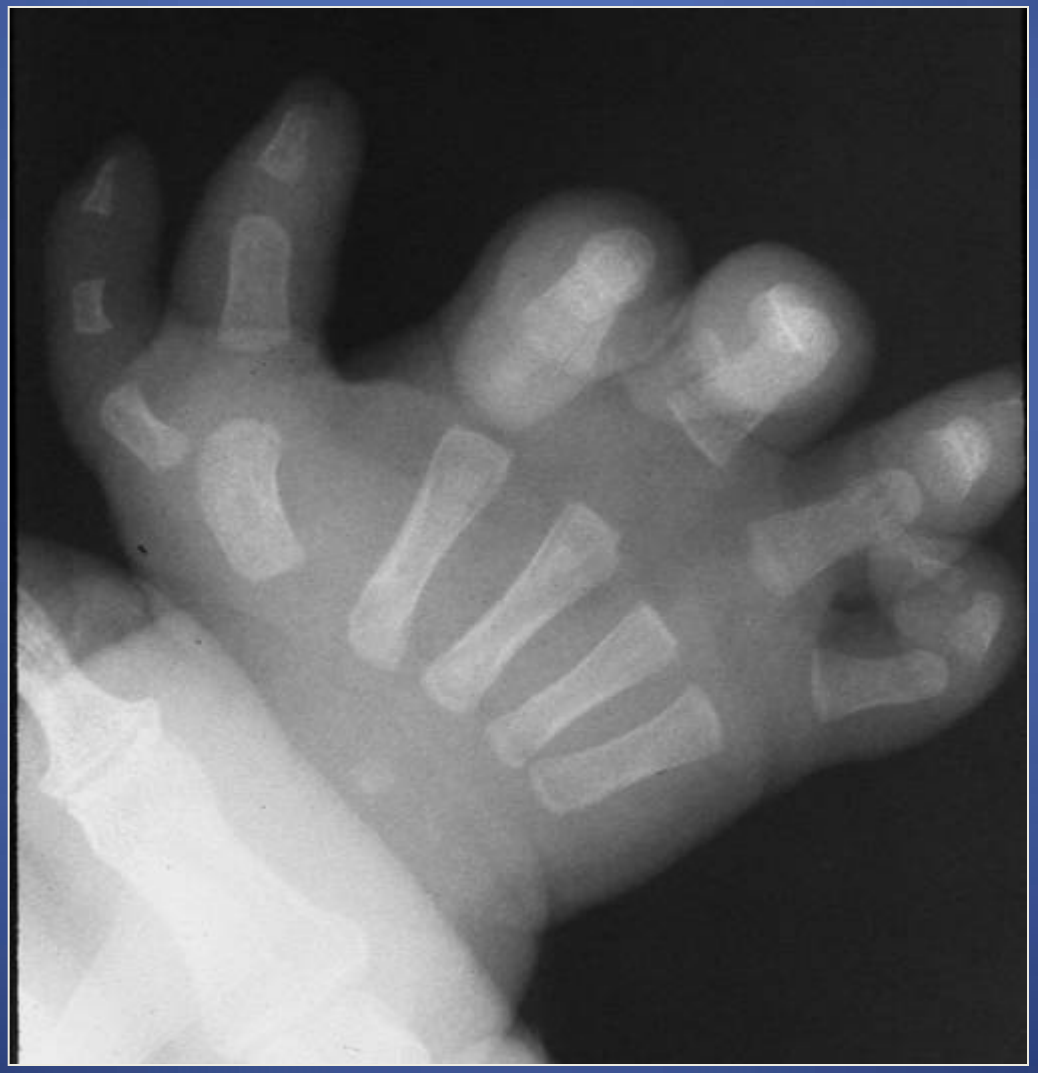


THUMB DUPLICATION
Type 4





TRIPHALANGEAL THUMB
Type 7



CONGENITAL HAND ANOMALIES
Classification

I Failure of formation of parts

II Failure of differentiation

III Duplication

IV Overgrowth

V Undergrowth

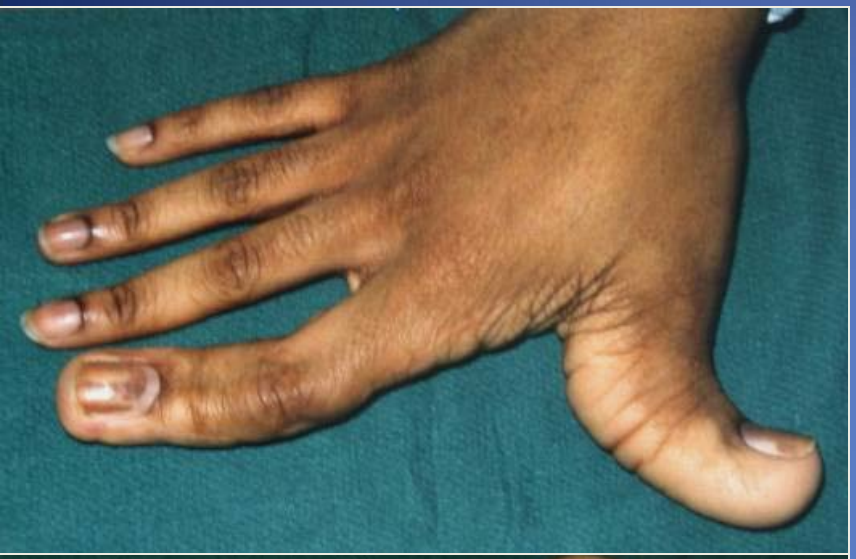
VI Constriction band syndrome

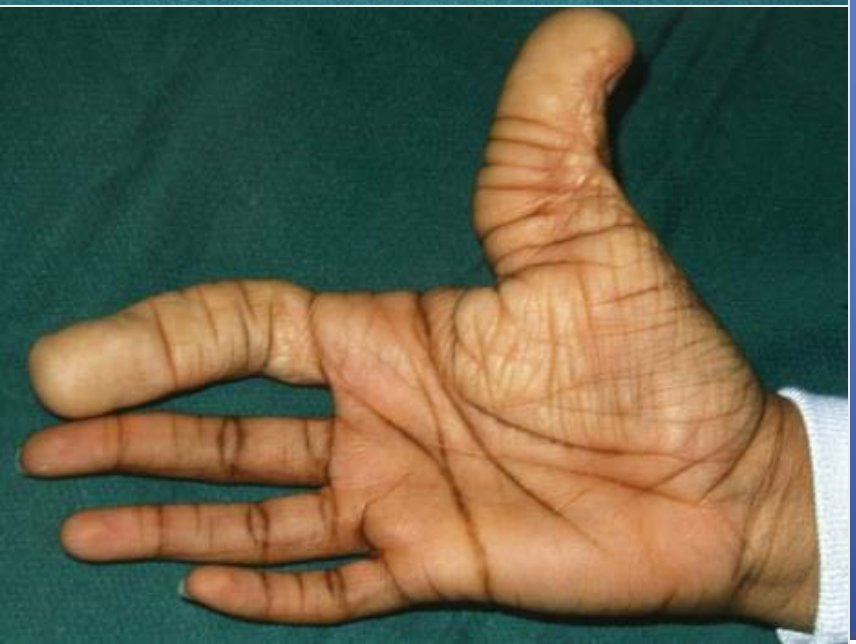
VII Generalized skeletal abnormalities

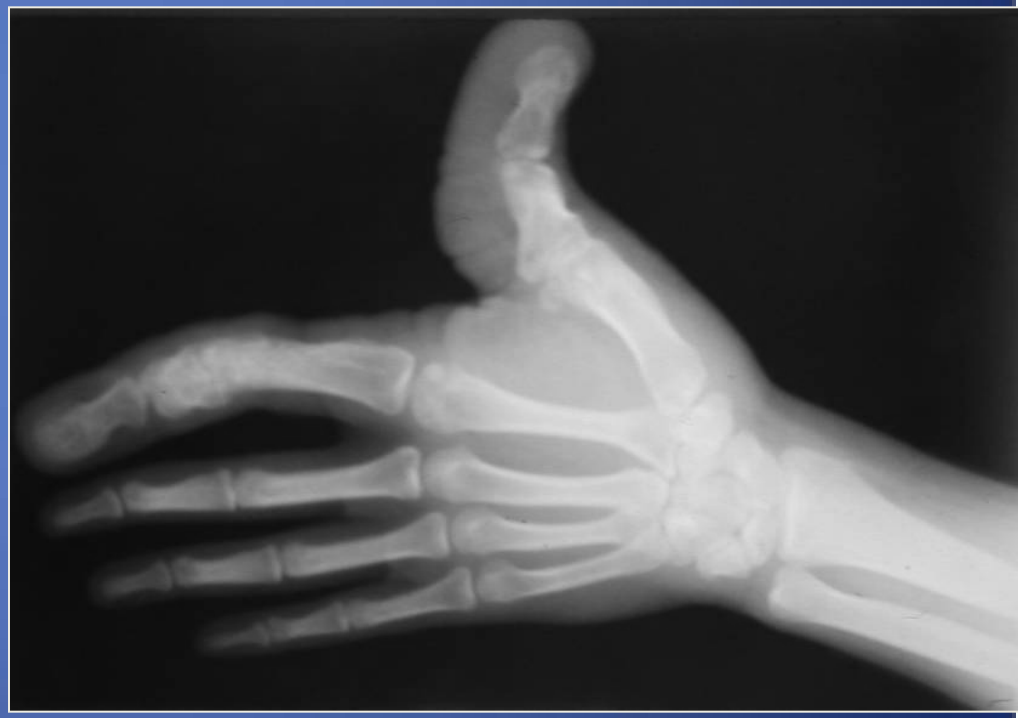
MACRODACTYLY

- 90% unilateral
- Multiple digits vs. single digit = 3 : 1
- Affects radial side of hand
- Index finger most frequently affected
- ? Related to neurofibromatosis

MACRODACTYLY







MACRODACTYLY

- Staged debulking of soft tissue
- Epiphysiodesis
- Wedge osteotomies to correct deviation
- Consider ray amputation

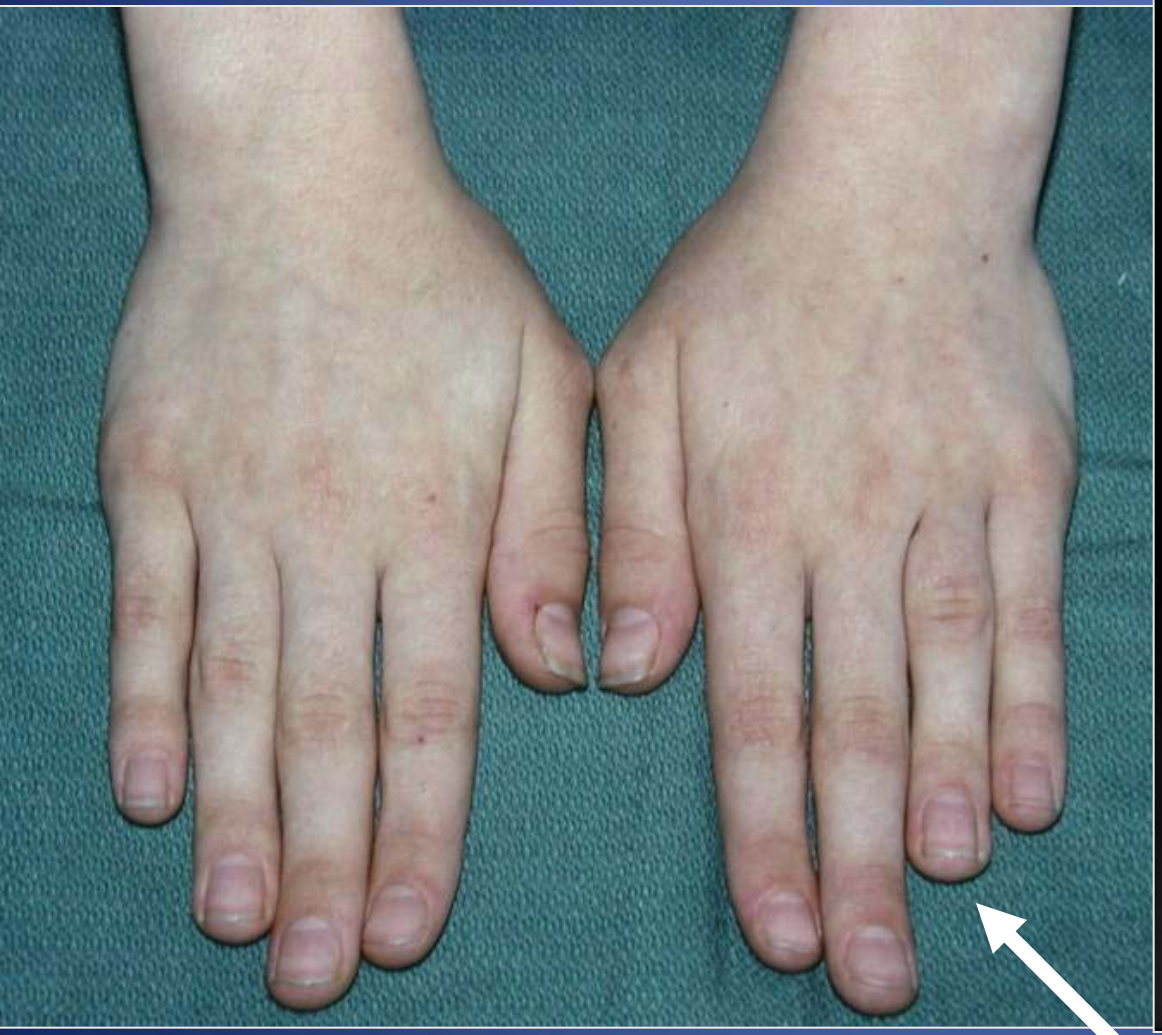
CONGENITAL HAND ANOMALIES
Classification

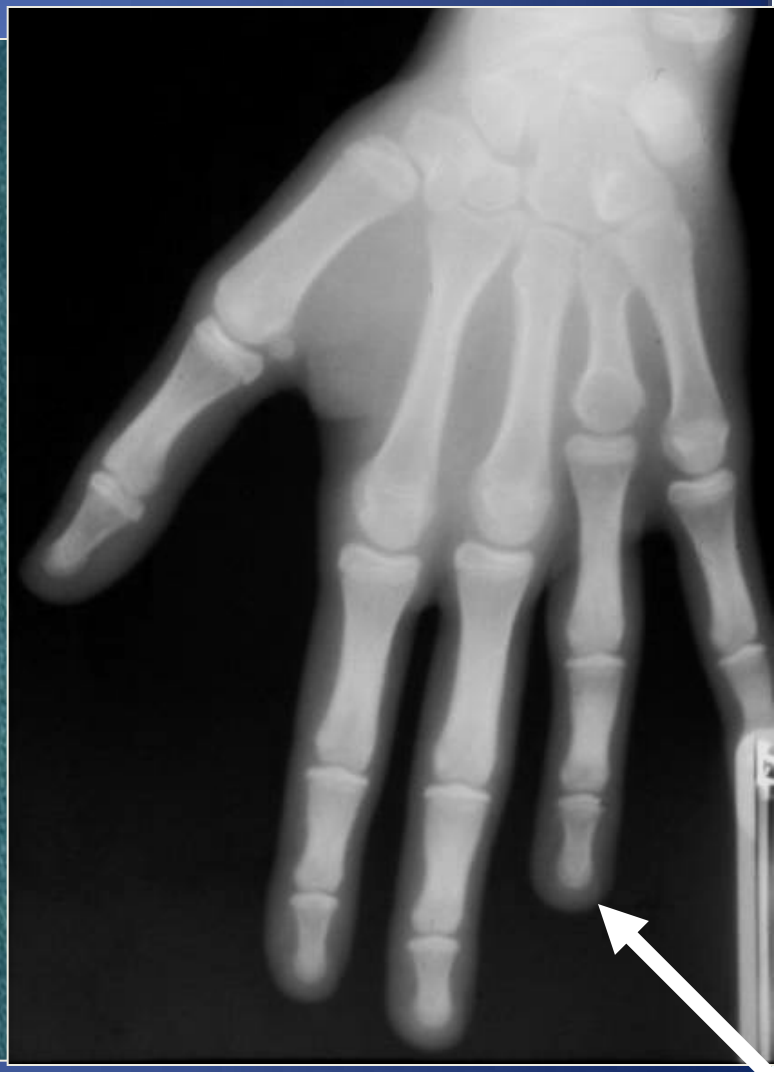
I Failure of formation of parts
II Failure of differentiation
III Duplication
IV Overgrowth

V Undergrowth (brachydactyly)

- Short digit - normal # of bones, 1 is small
- Short metacarpal (brachymetacarpia)
- Short phalanx (brachyphalangia)

BRACHYMETACARPIA





CONGENITAL HAND ANOMALIES
Classification

I Failure of formation of parts

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IV Overgrowth

V Undergrowth

VI Constriction band syndrome

VII Generalized skeletal abnormalities

CONSTRICTION RING SYNDROME

- **Circumferential grooving or transverse amputation**

- **Associated anomalies (40-50%)**
  - ***Club feet; cleft lip and palate***

- **Associated hand anomalies (80%)**
  - ***Syndactyly***

CONGENITAL HAND ANOMALIES
Classification

I Failure of formation of parts

II Failure of differentiation

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IV Overgrowth

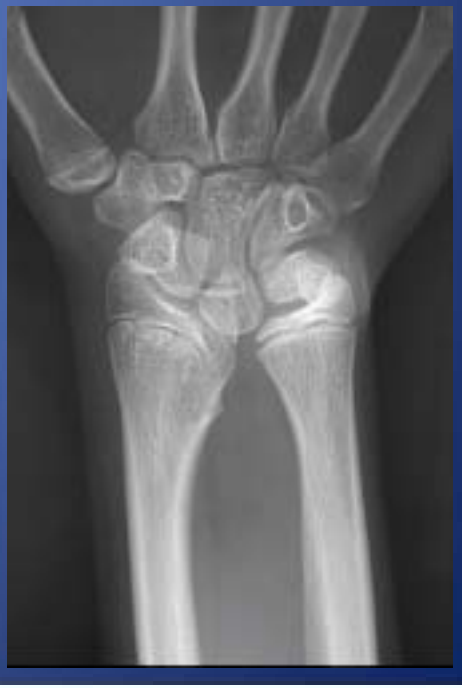
V Undergrowth

VI Constriction band syndrome

VII Generalized skeletal abnormalities

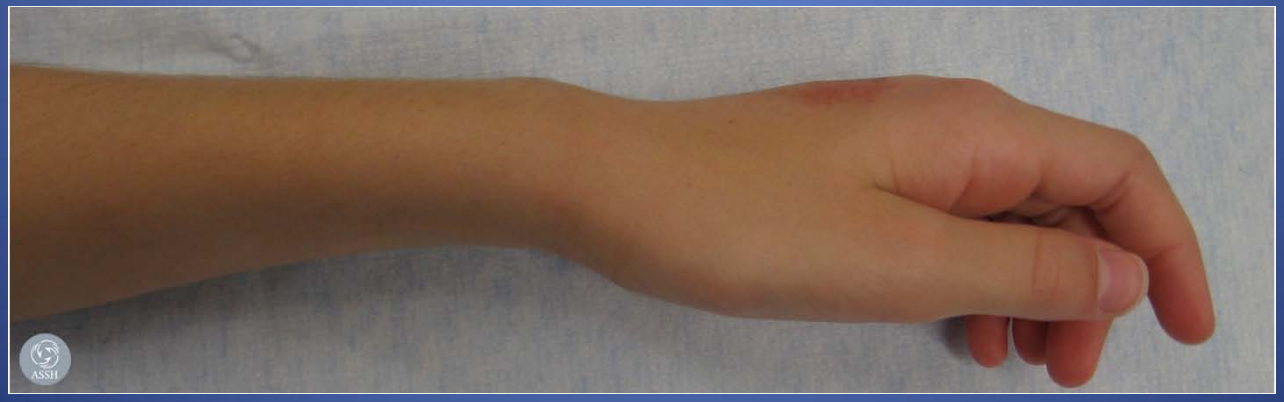
(2008 ASSH SAE qn 68)
Patients with the congenital deformity depicted in Figure 1 often have:

A. Limited elbow range of motion
B. Carpal tunnel syndrome
C. Subluxation of the extensor carpi ulnaris tendon
D. Short stature
E. Mucupolysaccharidase deficiency

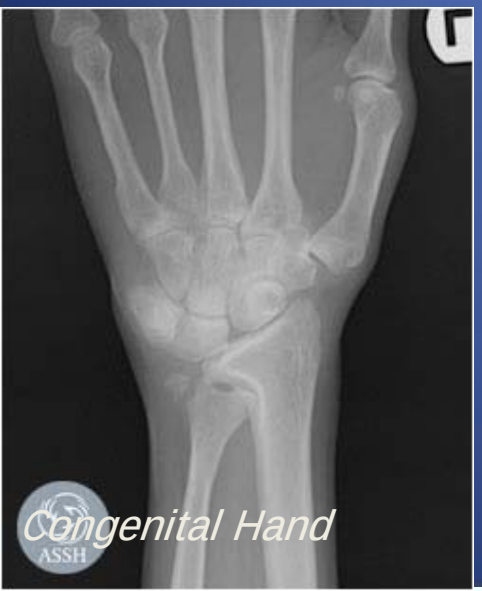


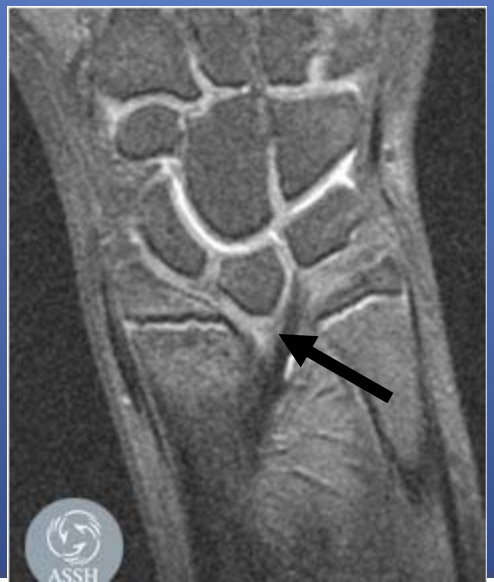
- Madelung Deformity
  - Basics
    - Focal physeal abnormality of distal radius
      - Volar-ulnar corner growth inhibition
    - Abnormal radiolunate ligament from metaphysis of distal radius
      - Vicker's ligament
    - Caused by mutation in SHOX gene
    - Associated with short stature, Turner syndrome, Leri-Weill dyschondrosteosis

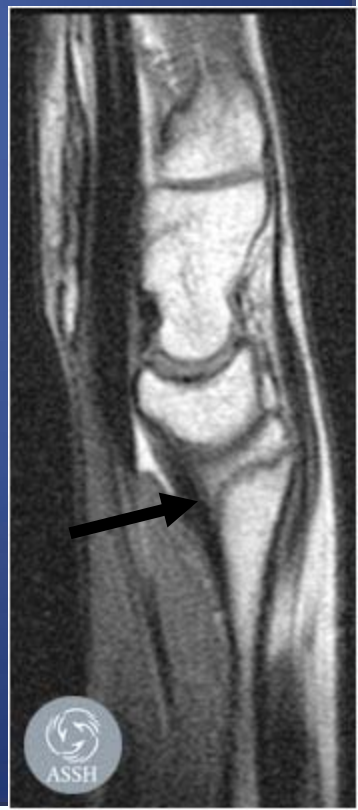
- Madelung Deformity
  - Evaluation
    - Deformity becomes evident in adolescence
    - Volar sag to carpus
    - Dorsally prominent distal ulna
    - Self-limited period of pain in adolescence



- Madelung Deformity
  - Evaluation
    - Radiographs/MRI show typical volar-ulnar physeal fall-off and carpal subluxation
    - MRI reveals Vicker's ligament







- Madelung Deformity
  - Treatment
    - Physiolysis, resection of tethering Vicker's ligament may halt progression if done early enough
  - Dome osteotomy of distal radius to correct established deformity

(2008 ASSH SAE qn 68)

Patients with the congenital deformity depicted in Figure 1 often have:

A. Limited elbow range of motion
B. Carpal tunnel syndrome
C. Subluxation of the extensor carpi ulnaris tendon
D. Short stature
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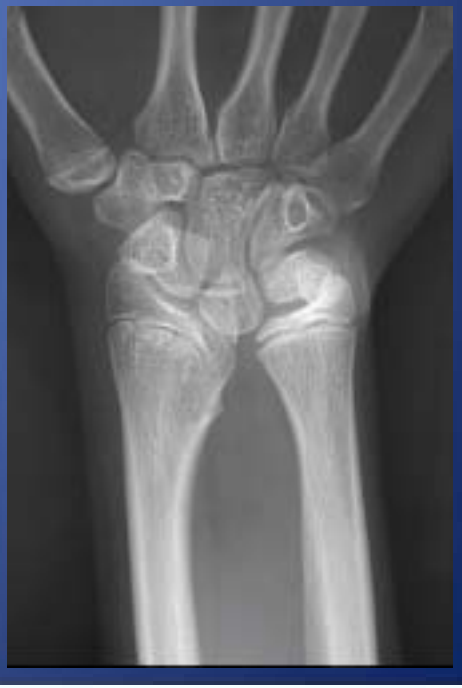
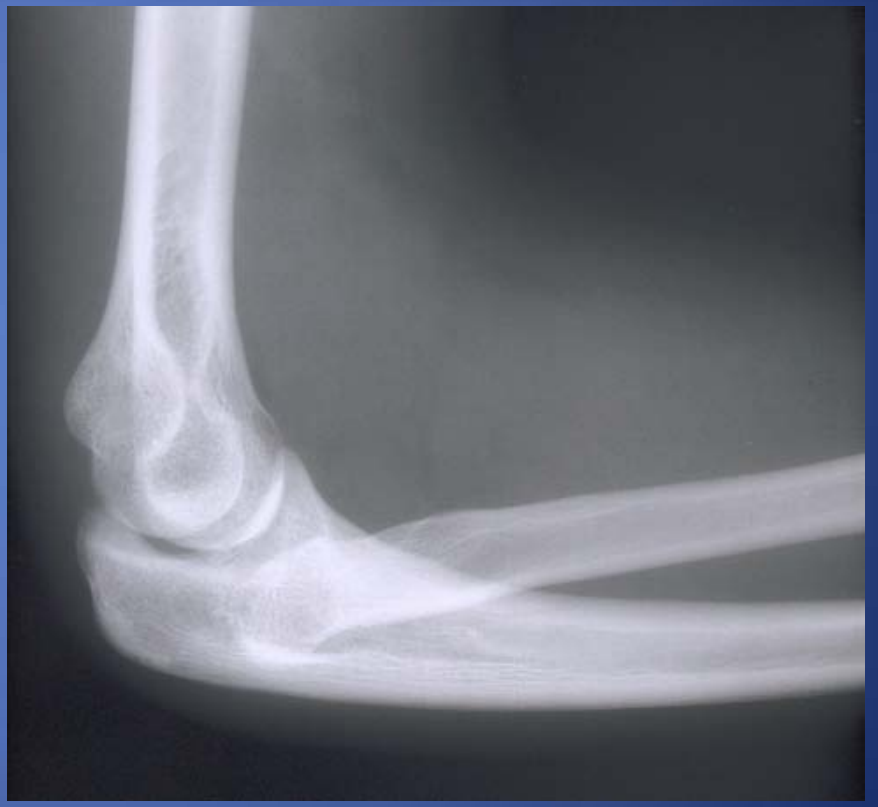


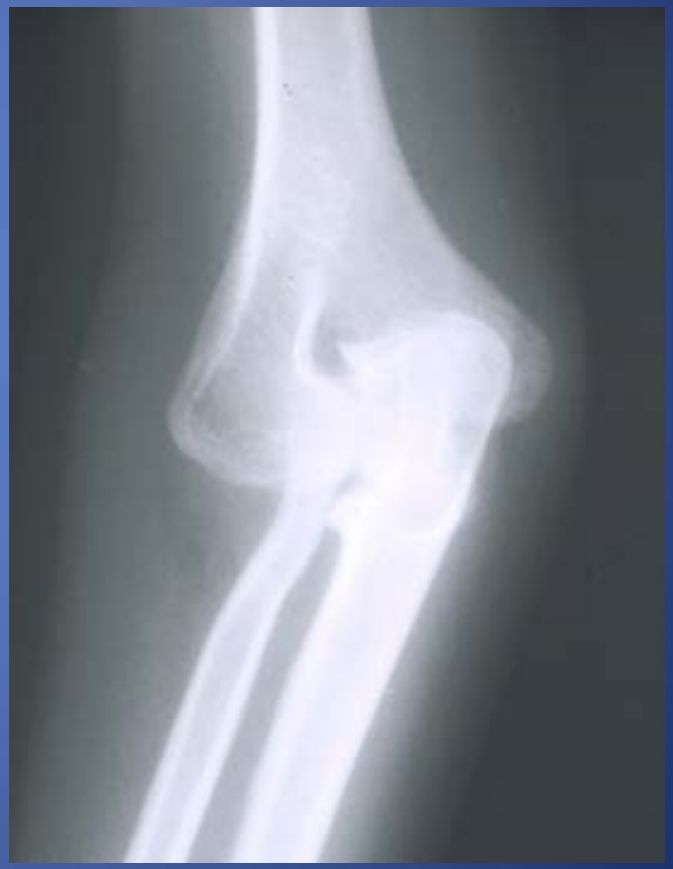
Congenital Dislocation of the Radial Head

- Uncommon
- DDx: traumatic dislocation
- Sporadic or inheritable
- Isolated or syndromic
- 40-60% bilateral



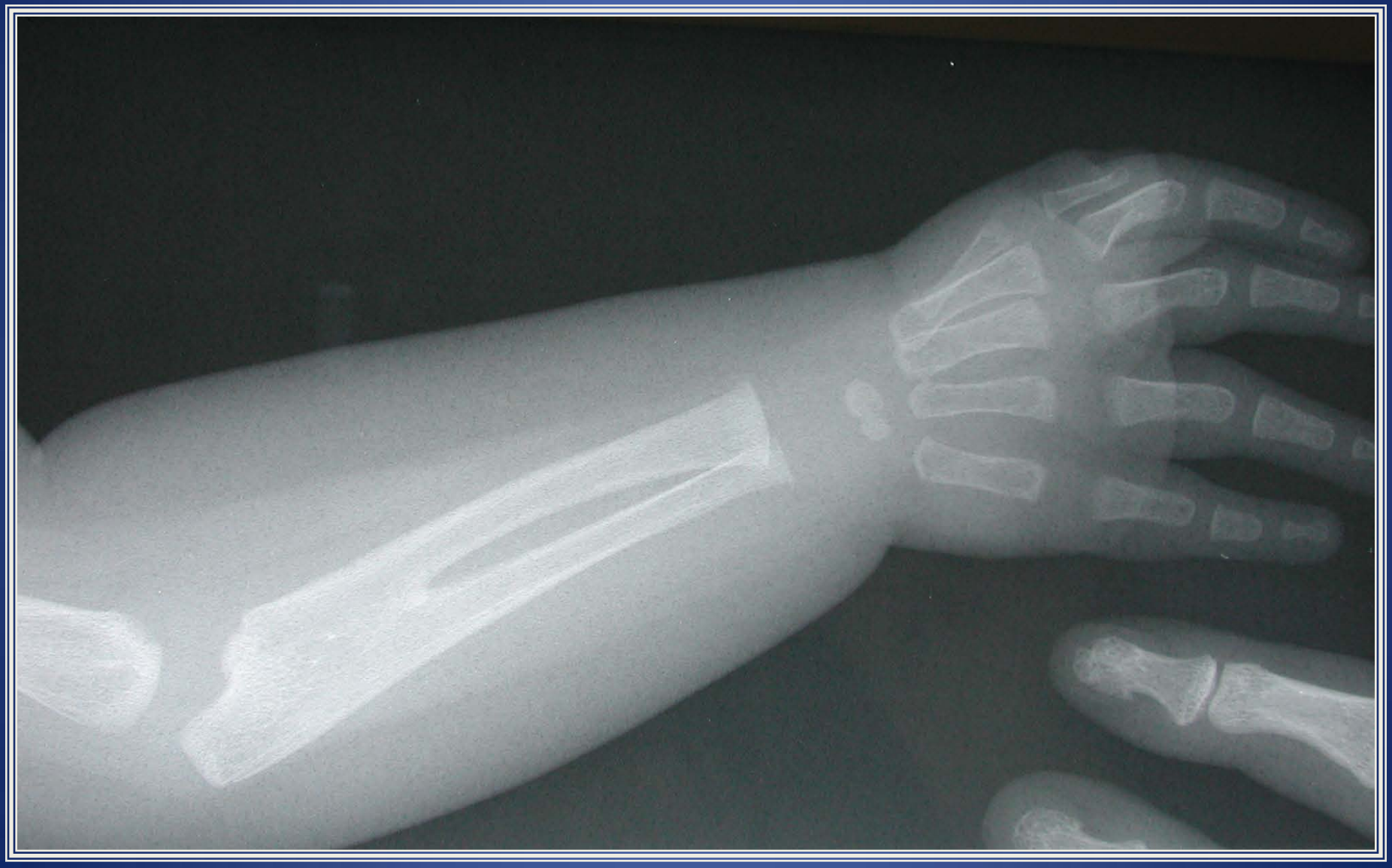
Congenital Dislocation of the Radial Head

- Indications for treatment
  - PAIN
  - Skeletally mature



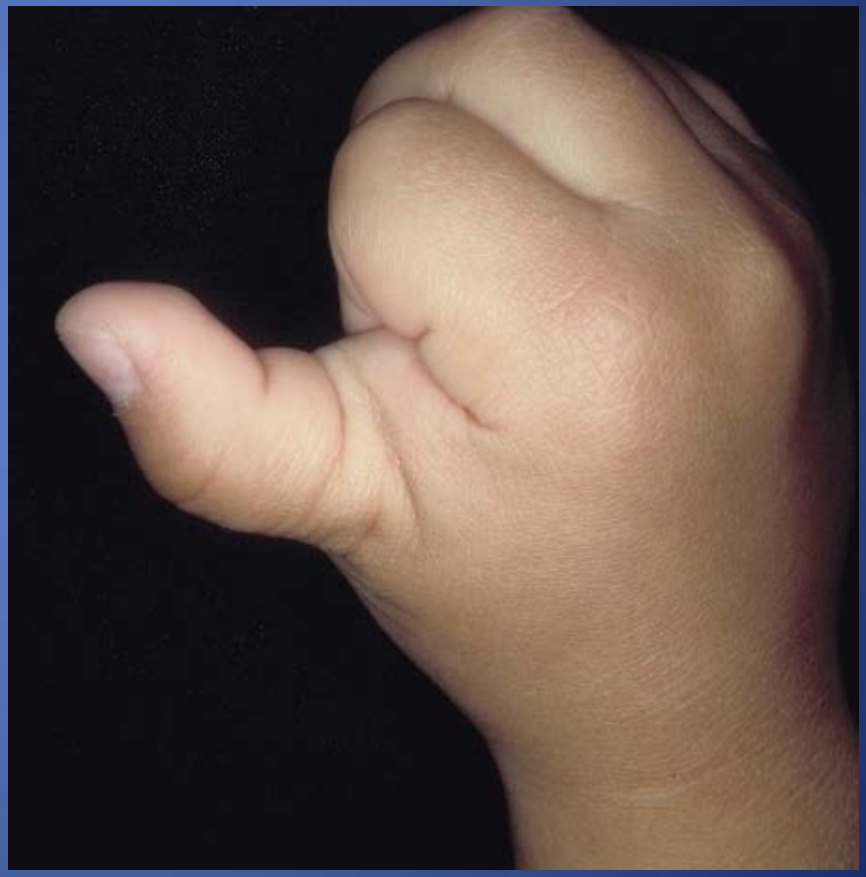
Congenital Proximal Radio-ulnar Synostosis

- Uncommon
- Sporadic, syndromic or other malformations
- 1/3 with fetal alcohol syndrome
- 50-80% bilateral
- No forearm rotation (usually fixed in pronation)
- Indications: function (esp if bilateral)
- Surgical repositioning



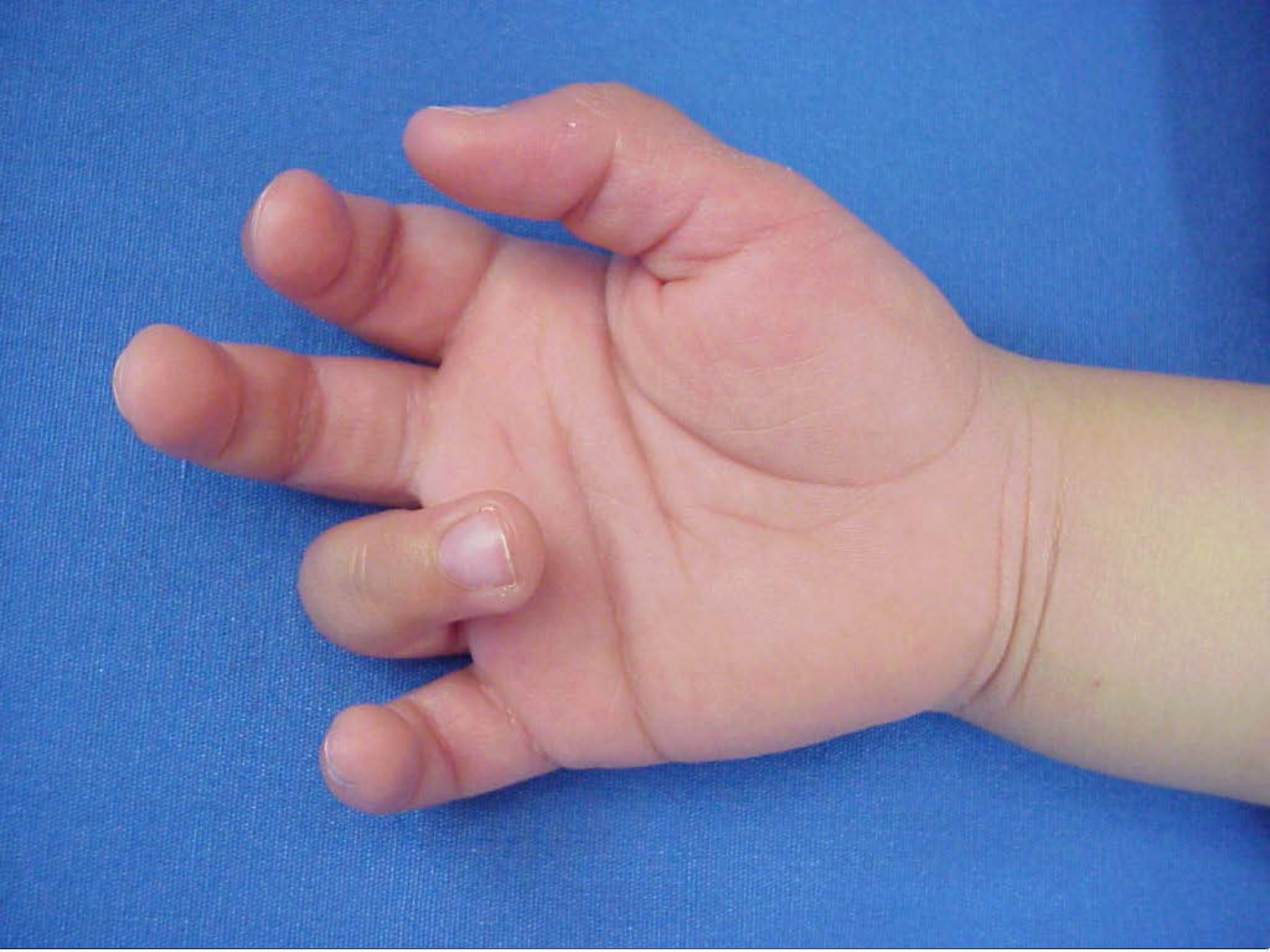
Infantile Trigger Thumb

- Common
- Isolated anomaly
- Developmental, not congenital
- May resolve spontaneously



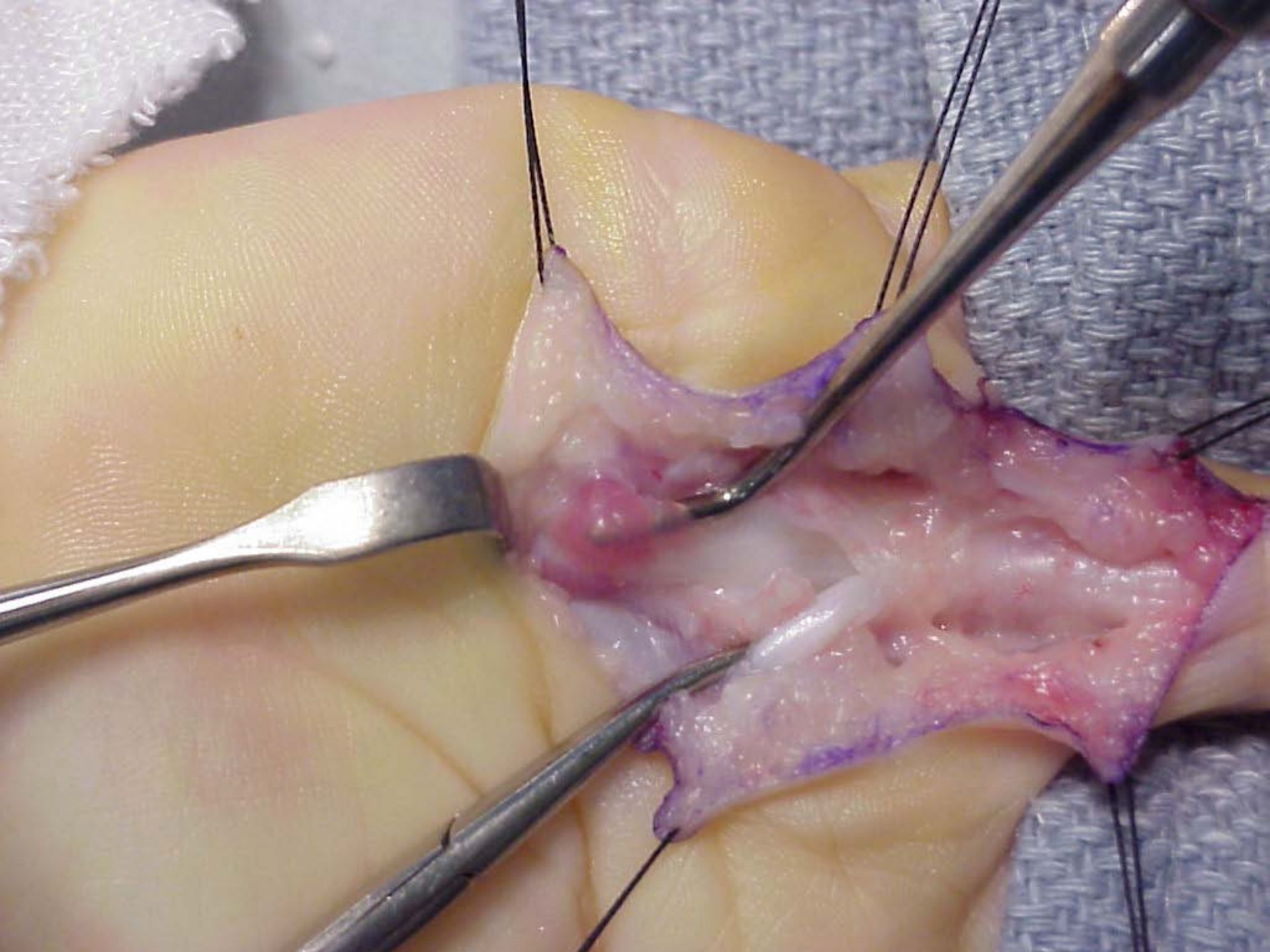
Infantile Trigger Thumb

- Surgical Indications
  - Persists for >1 yr after diagnosis
  - Dx at age > 4 y.o.
- Treatment
  - Surgical release of A1 pulley
  - Excellent results



TRIGGER FINGER

- Different than trigger finger in adult
- Extensile approach
- Look for anomalous muscle/tendon attachment



Thank you