CONGENITAL HAND DIFFERENCES

Lisa Lattanza, MD
Chief Division of Hand and Upper Extremity Surgery
UCSF Medical Center

CONGENITAL DEFORMITIES
Upper Extremity

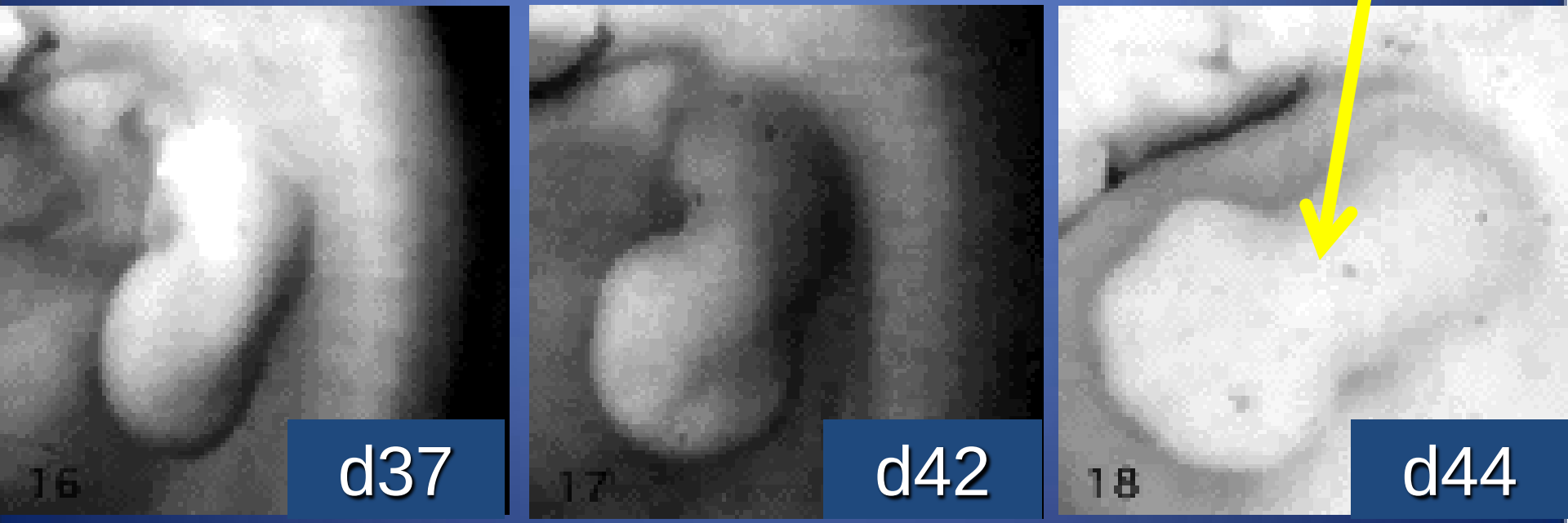
- Common
- 1 in 626 live births
- Many are single gene disorders

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

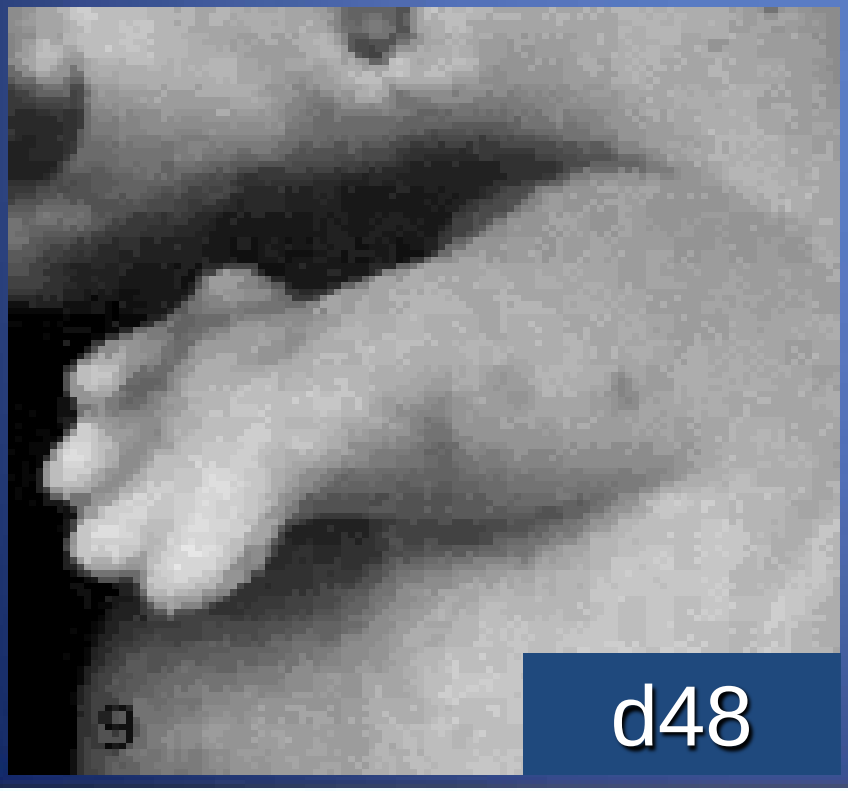
Weeks 5-6

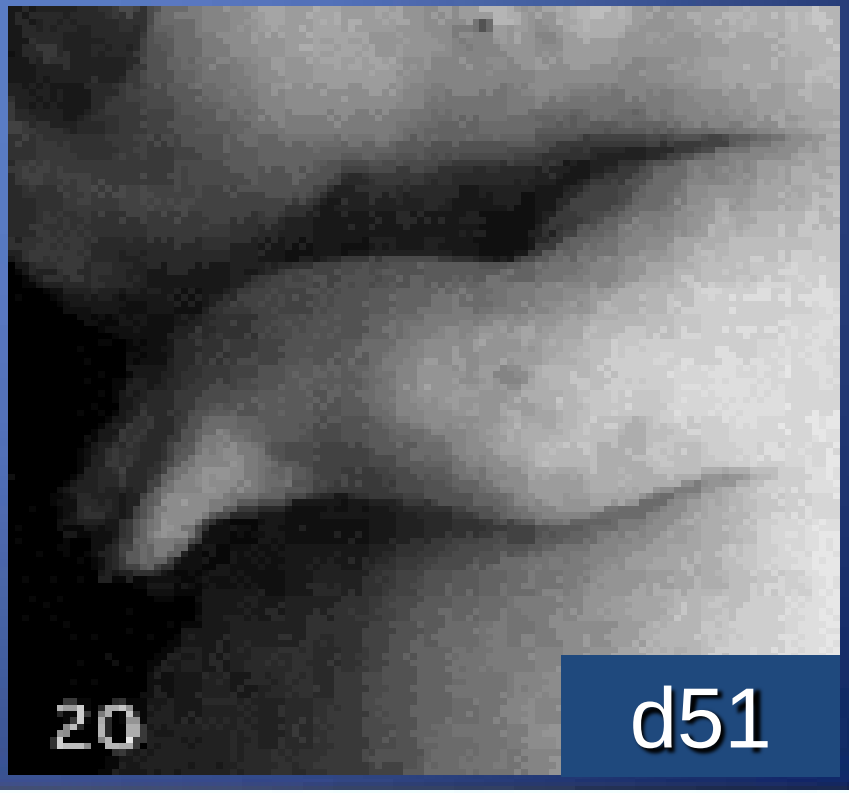
- Hand paddle develops
- Nerve ingrowth from spinal cord rami begins to occur proximally



Weeks 6 - 7

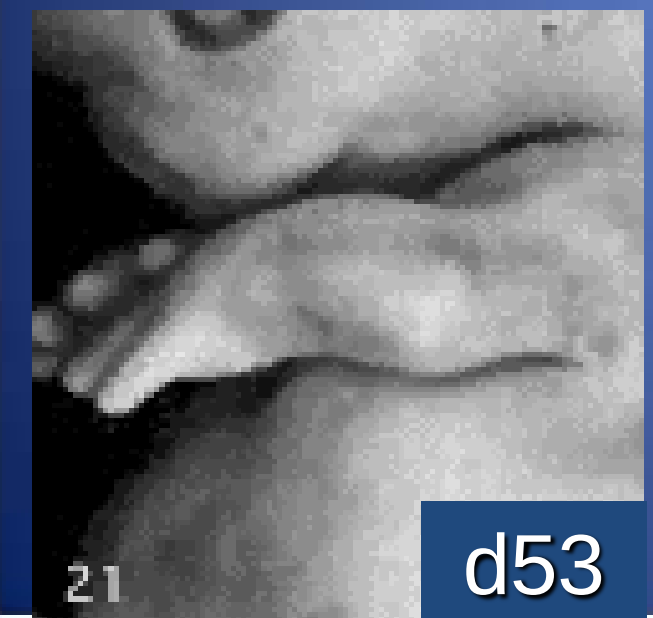
- Fingers begin to separate
- Cartilaginous “bones” form

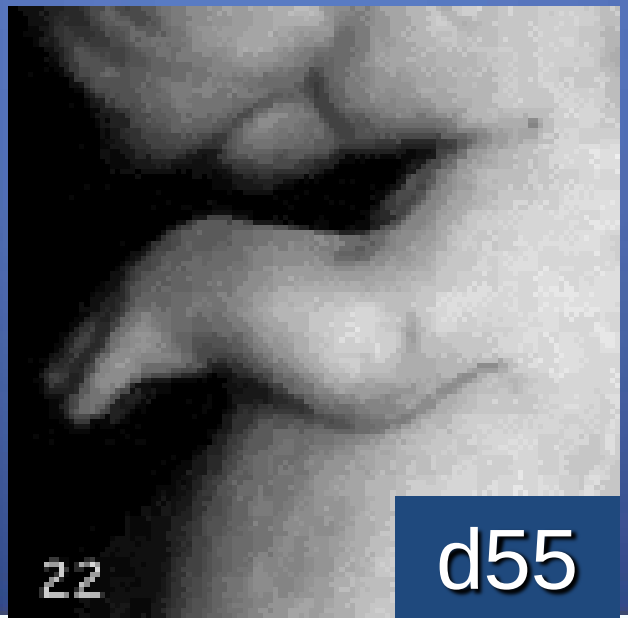


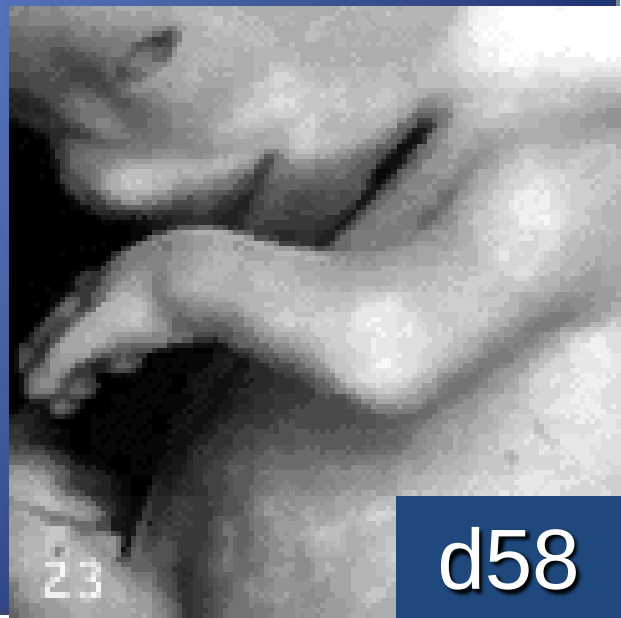


Weeks 7 — 8

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







(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

Preferred Response: B

Discussion: The most rapid period of limb development is between 4 and 8 weeks after fertilization. The majority of congenital anomalies occur during this period of time. At 8 weeks gestation all limb structures are present and further development is characterized by maturation and differentiation of existing structures

Limb Development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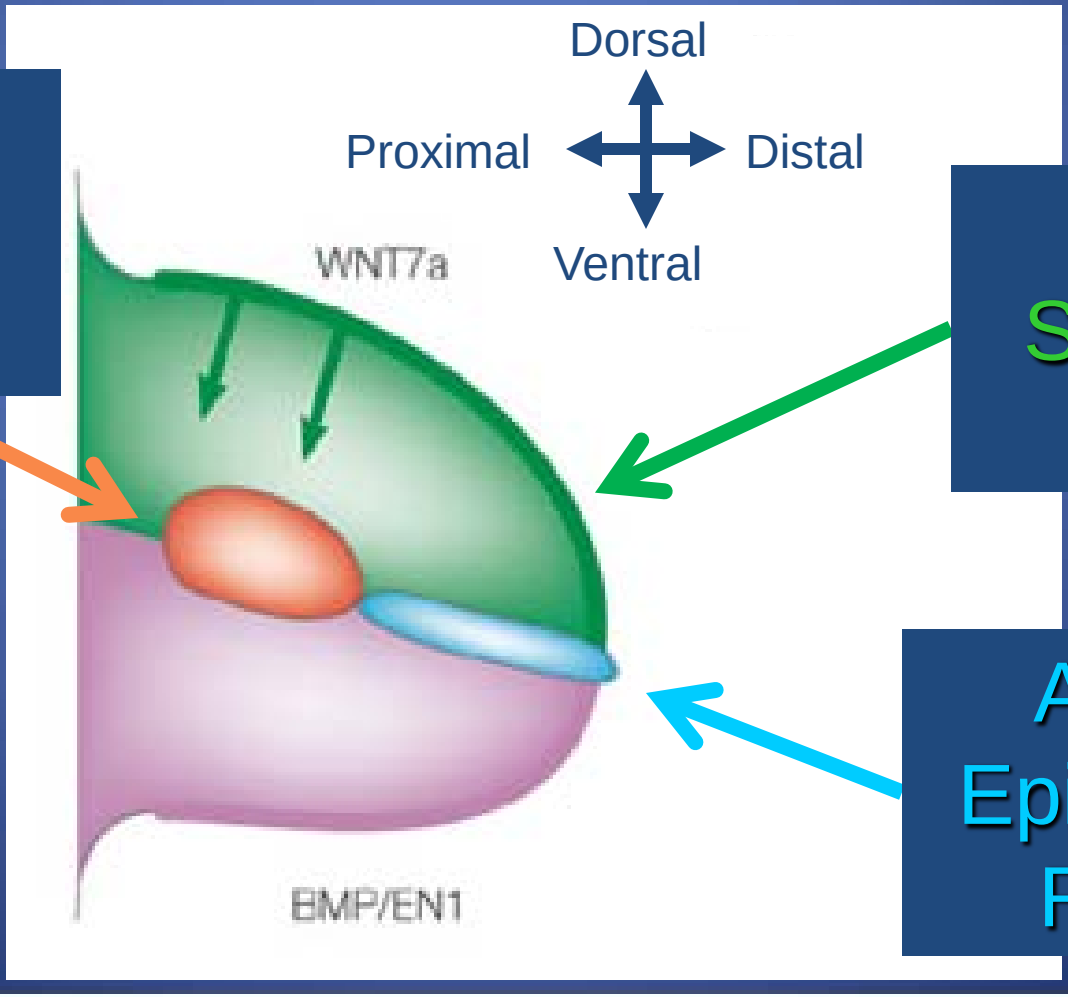
SIGNALING CENTERS IN LIMB DEVELOPMENT

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

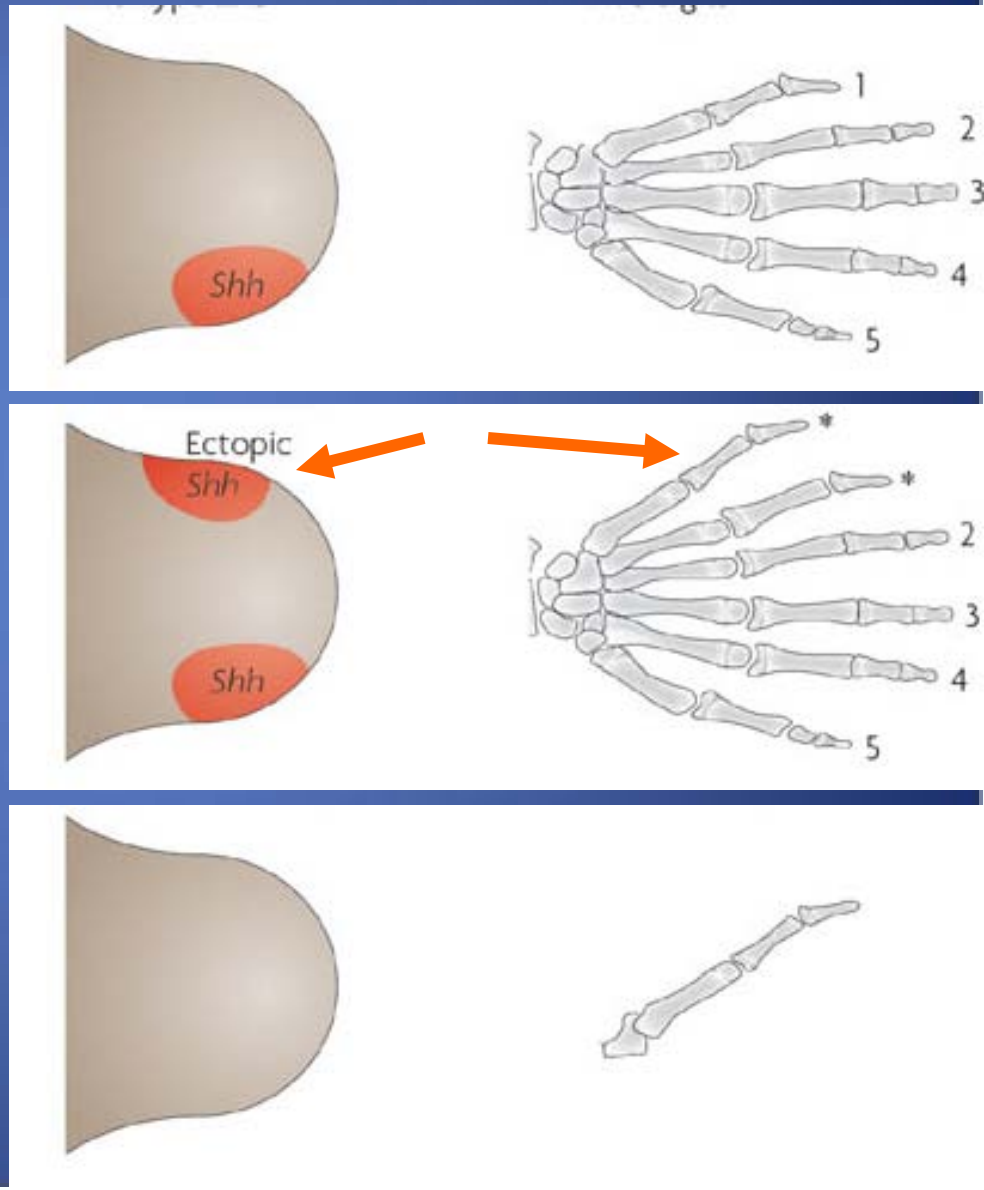
APICAL EPIDERMAL RIDGE

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

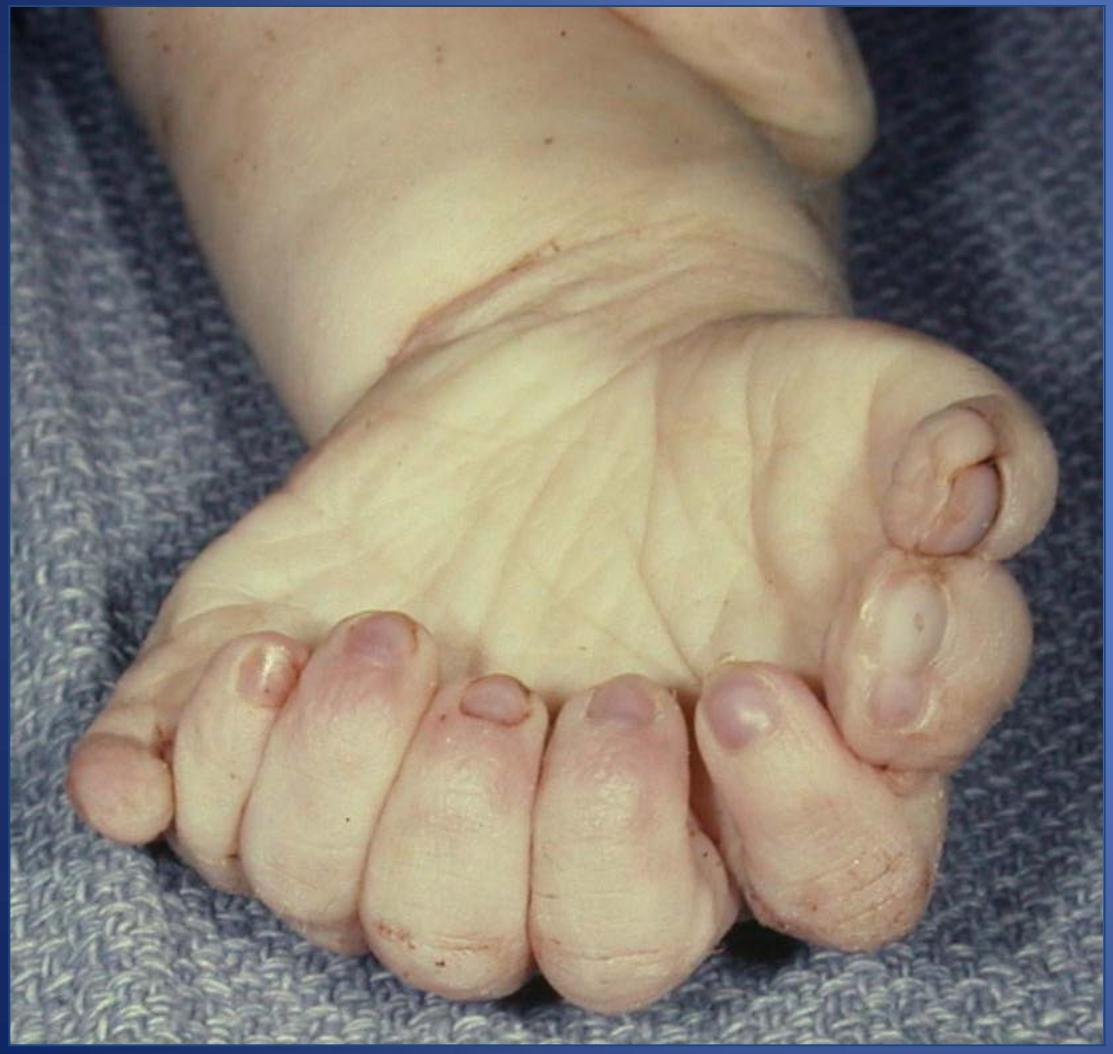
Abnormal ZPA Signaling

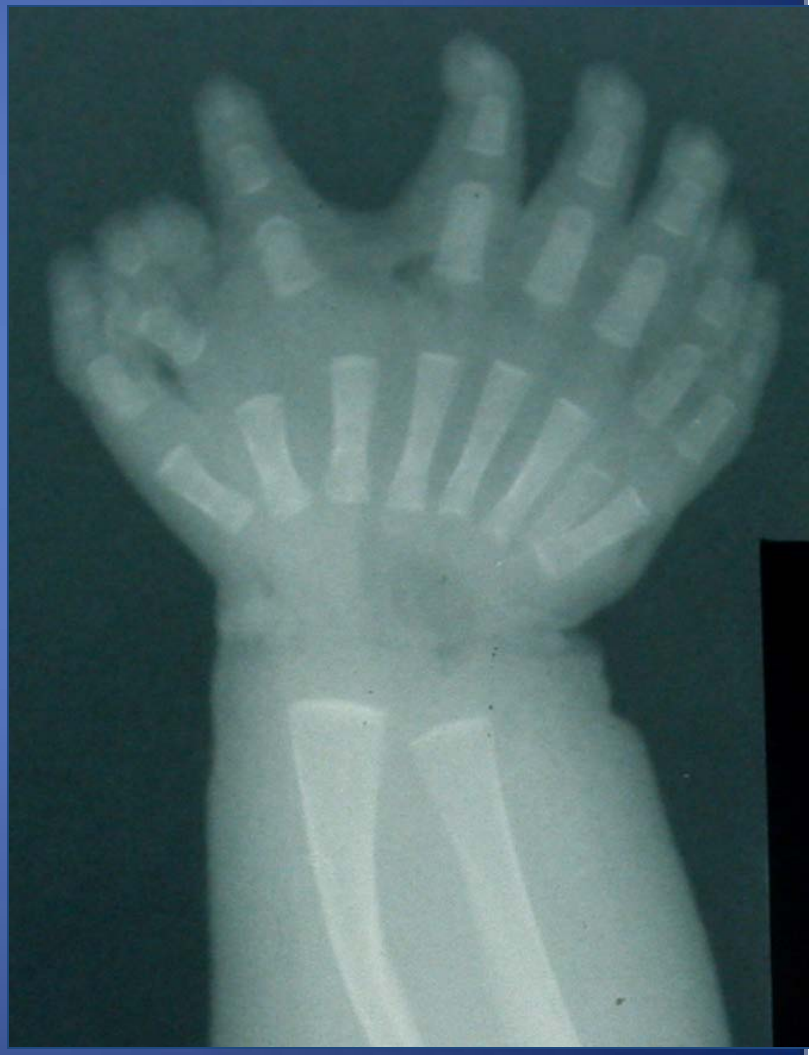
Transplantation of the ZPA or SHH anteriorly → duplication along the radioulnar axis

Remove the ZPA or SHH → fingers fail to form



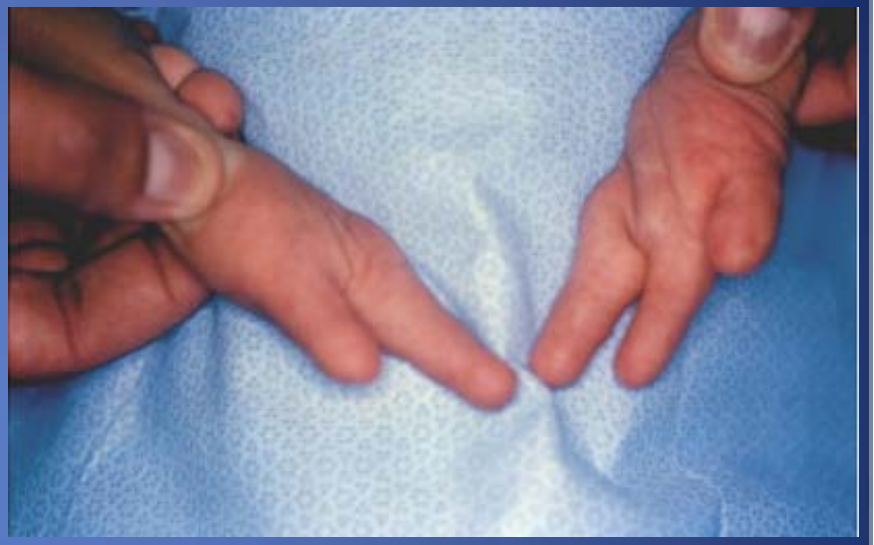
Mirror Hand Deformity

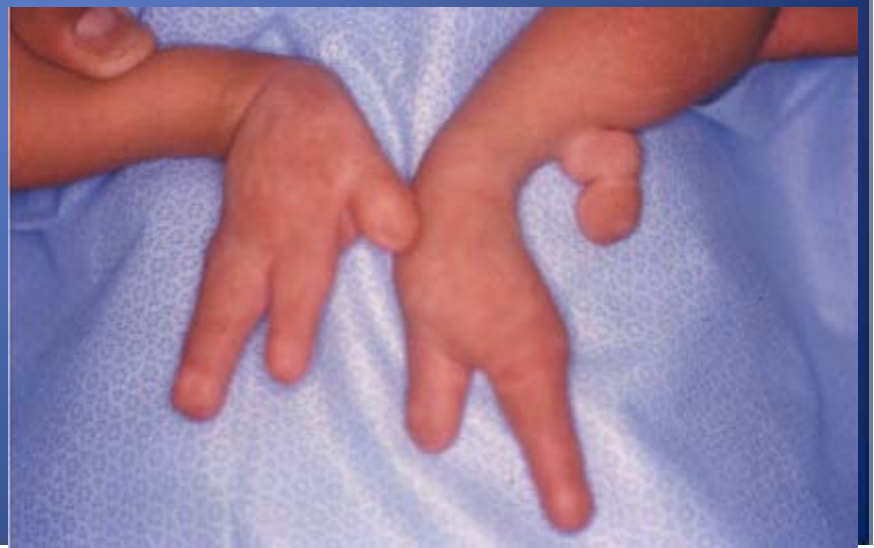




PALMAR DUPLICATION
Wnt7a Gene Mutation

Both hands have thick palmar skin and no hair or nails DORSALLY





Summary

Signaling Center	*Responsible Substance*	*Action*	*Anomaly*
Apical ectodermal ridge	Fibroblast growth factors	Proximal to distal development, interdigital apoptosis	Transverse deficiency, syndactyly
Zone of Polarizing Activity	Sonic Hedgehog protein	Radioulnar limb formation	Mirror Hand
Wnt Pathway	Wnt7a	Ventral and dorsal axis	Nail patella syndrome

CONGENITAL HAND ANOMALIES
Classification and Examples

I Failure of formation of parts

II Failure of differentiation

III Duplication

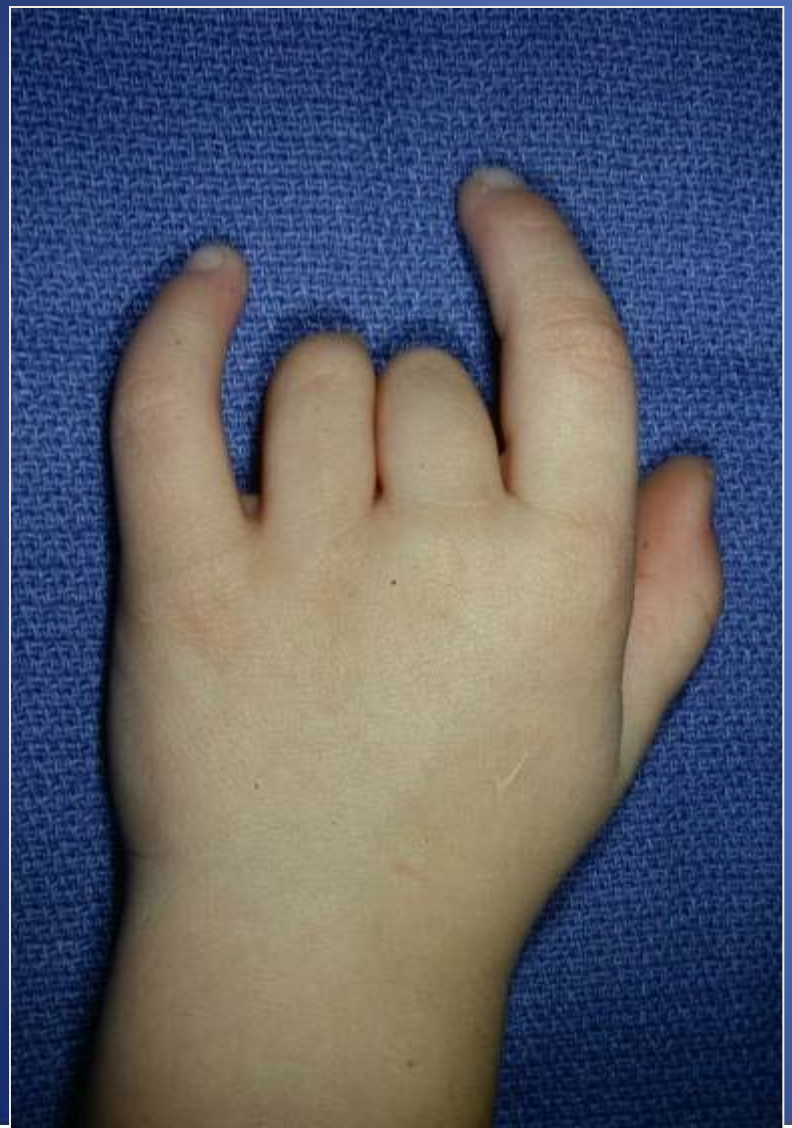
IV Overgrowth

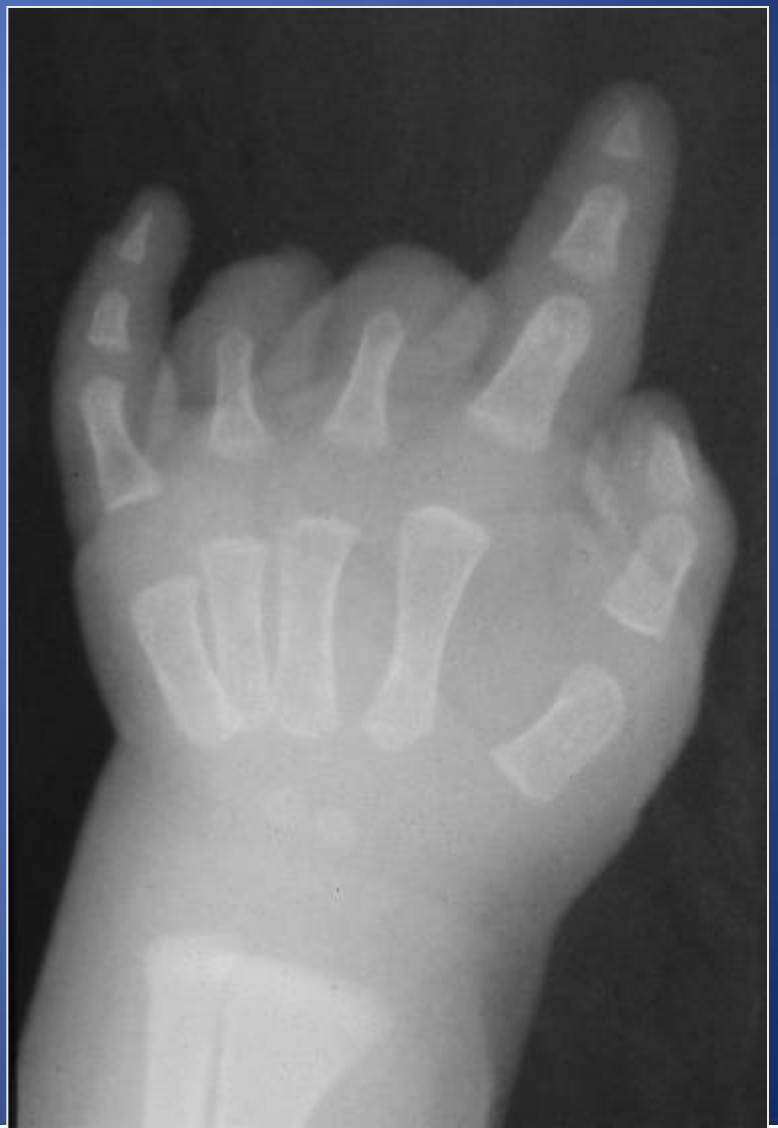
V Undergrowth

VI Constriction band syndrome

VII Generalized skeletal abnormalities

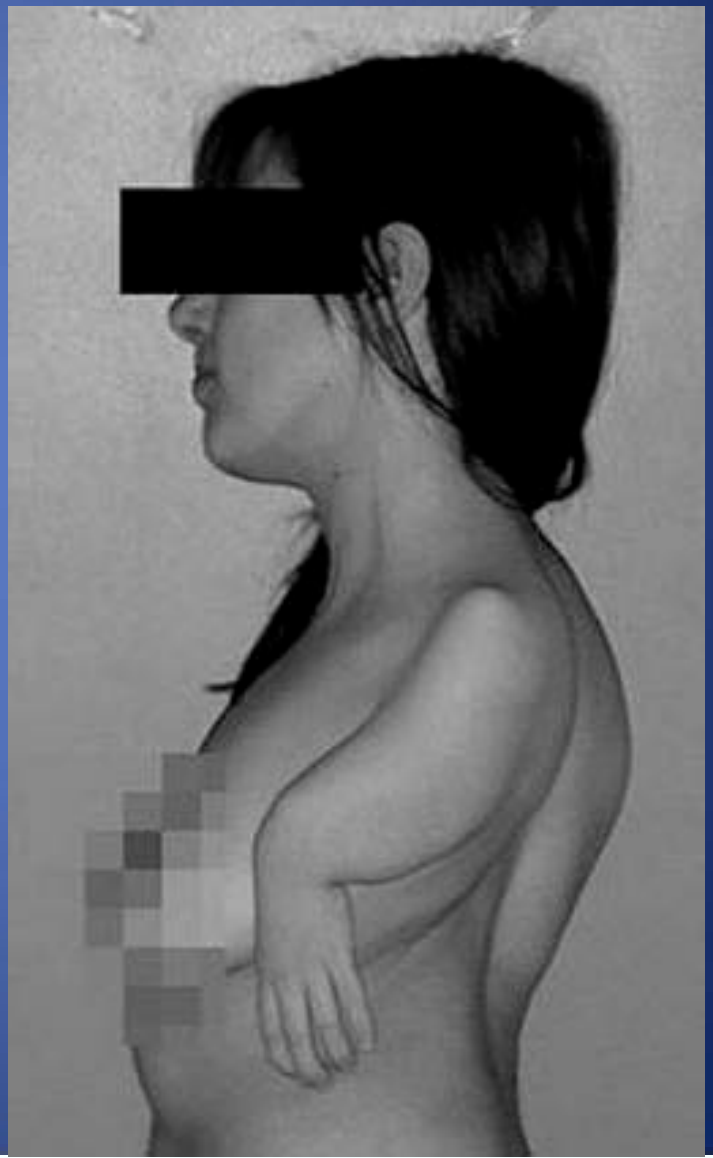
FAILURE OF FORMATION OF PARTS TRANSVERSE DEFICIENCY

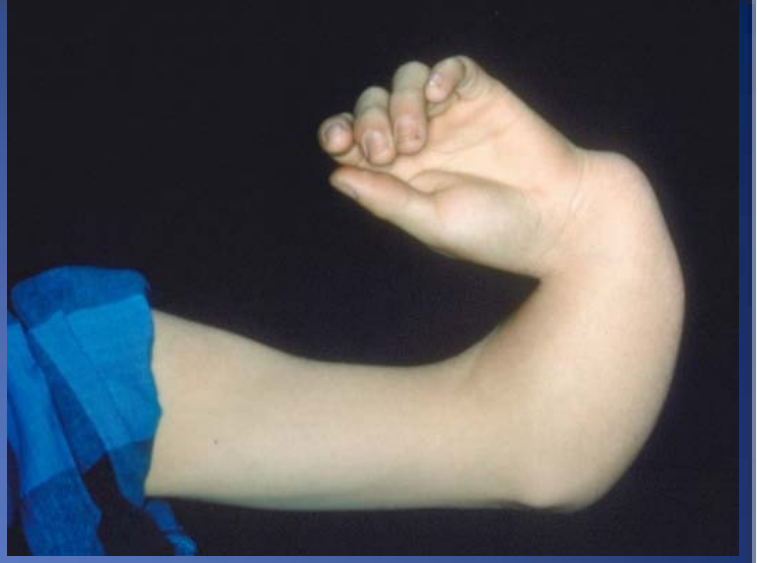




FAILURE OF FORMATION OF PARTS
LONGITUDINAL DEFICIENCY

- 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

HYPOPLASTIC THUMB

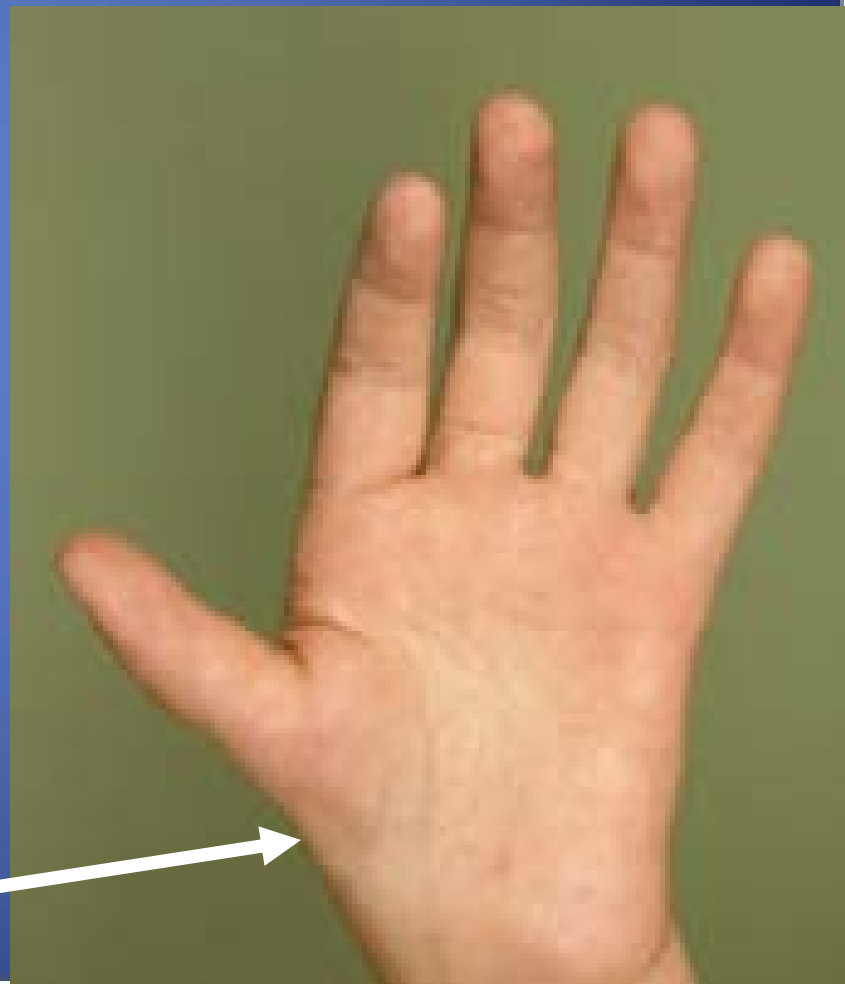
Grade I

Mild shortening

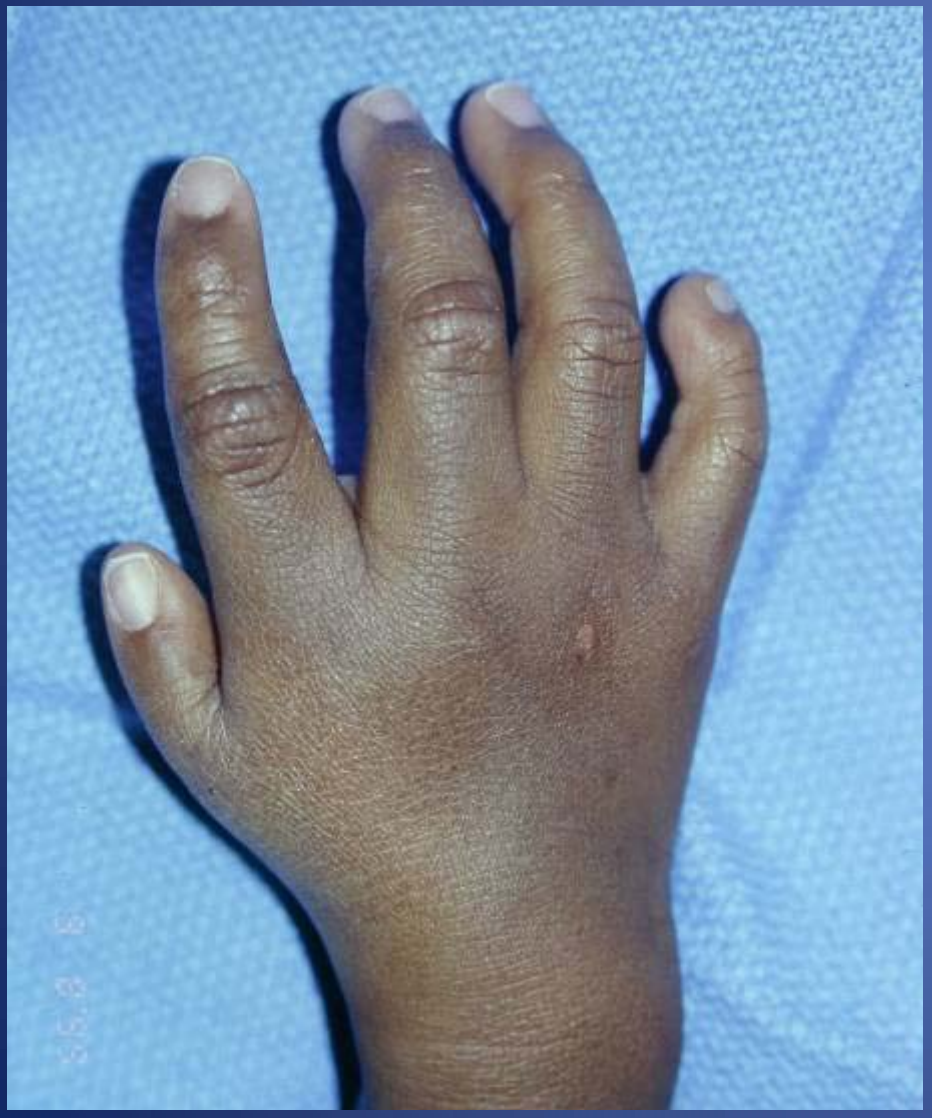
Grade II

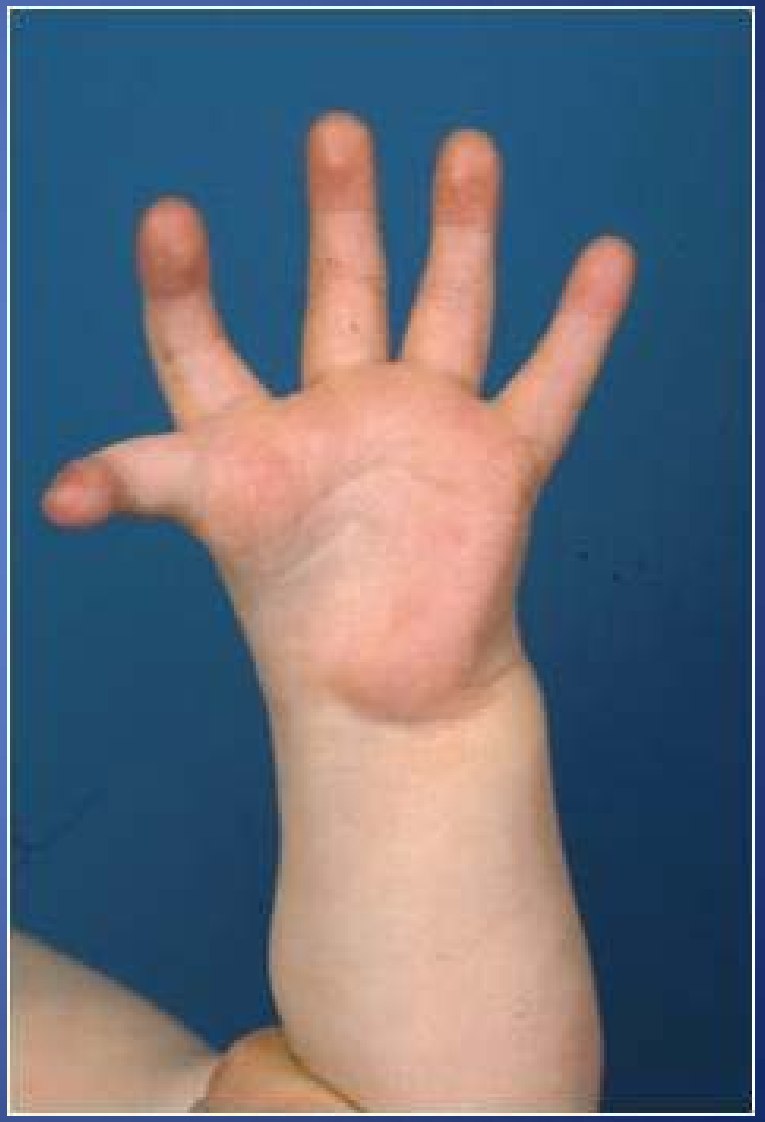
Moderate shortening

Thenar hypoplasia

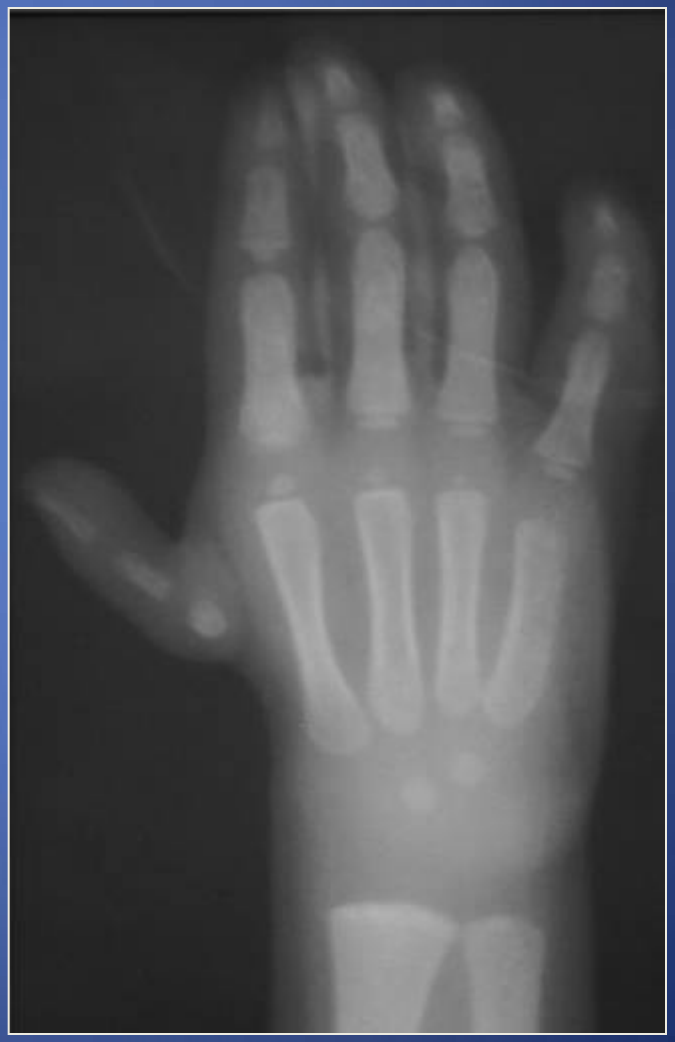


TYPE III HYPOPLASTIC THUMB





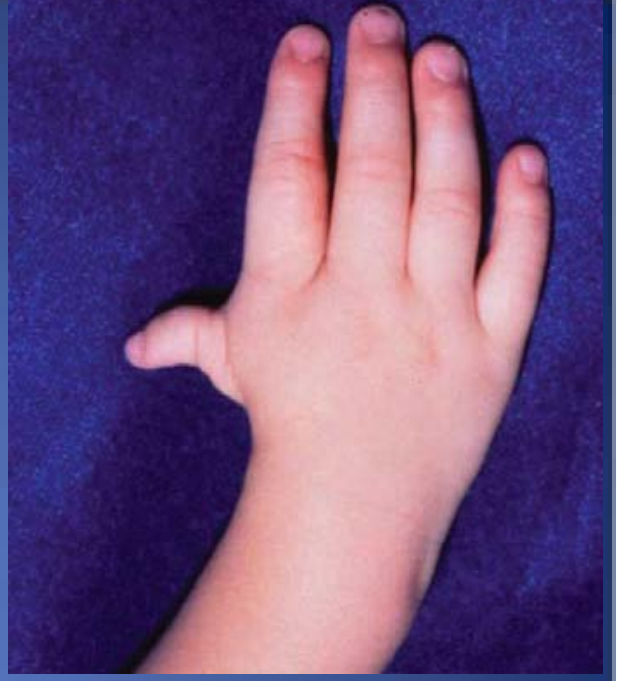
TYPE IV - FLOATING THUMB

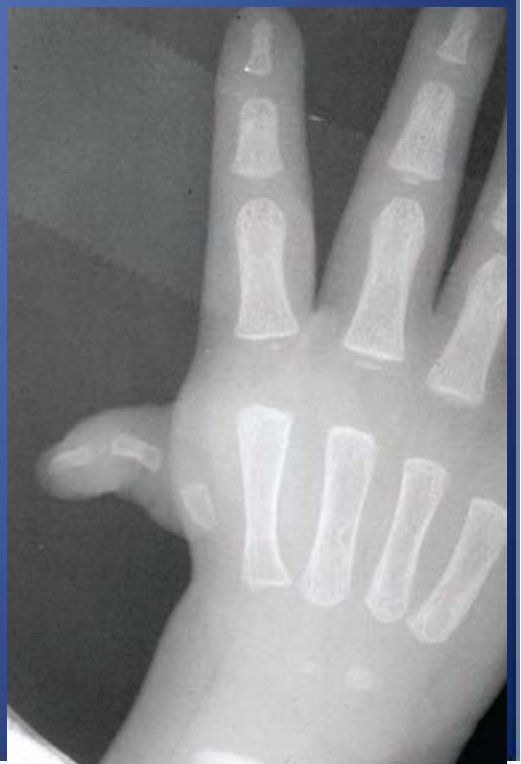


TYPE V - ABSENT THUMB

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

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



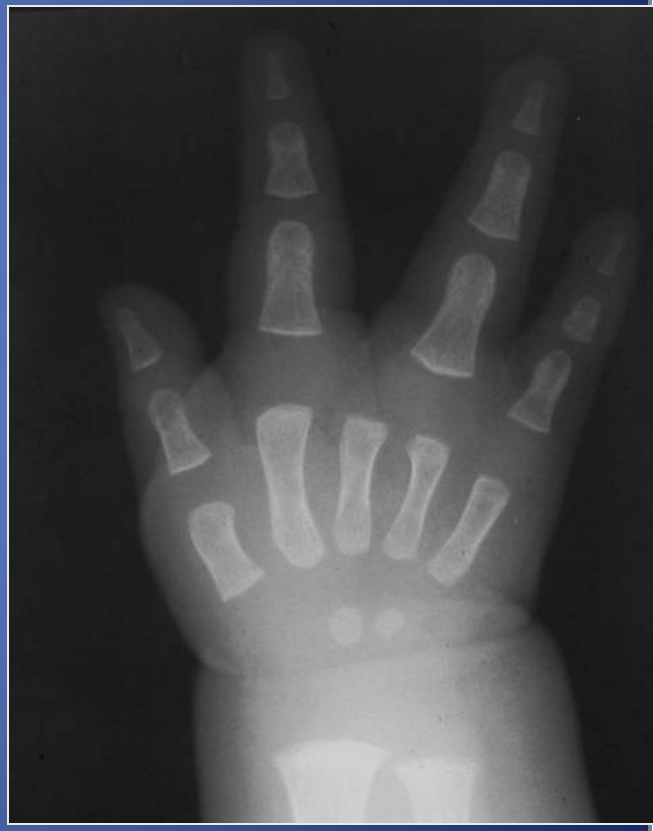


- A. Carpometacarpal (CMC) joint stabilization, first web space reconstruction
- B. Metacarpal lengthening, metacarpophalangeal (MCP) joint stabilization, opponensplasty, first web space reconstruction
- C. Thumb ablation and toe to hand transfer
- D. Index finger pollicization
- E. Opponensplasty, MCP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

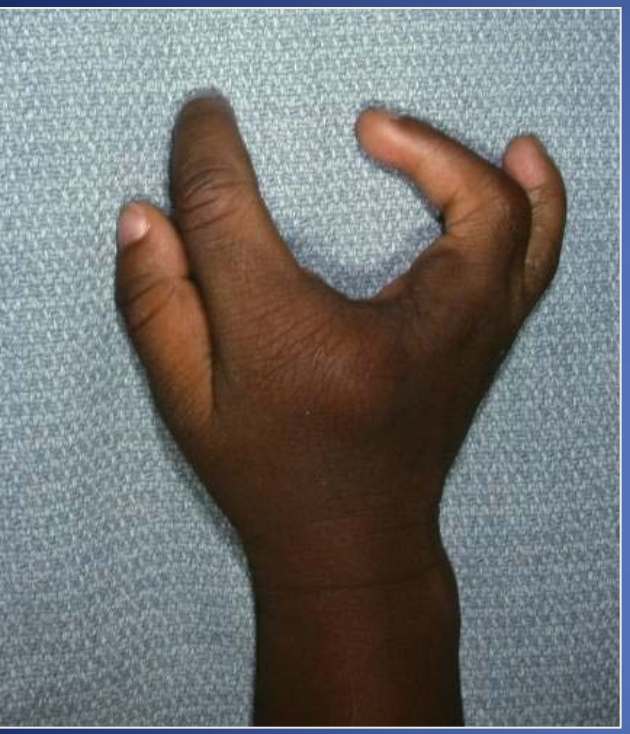
CLEFT HAND

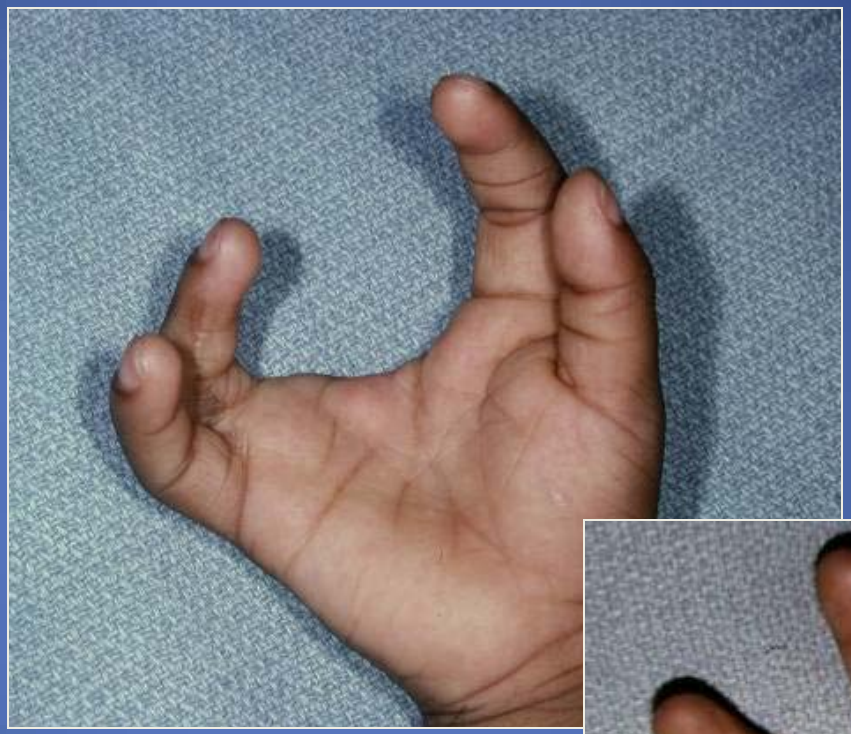
TYPICAL

- 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

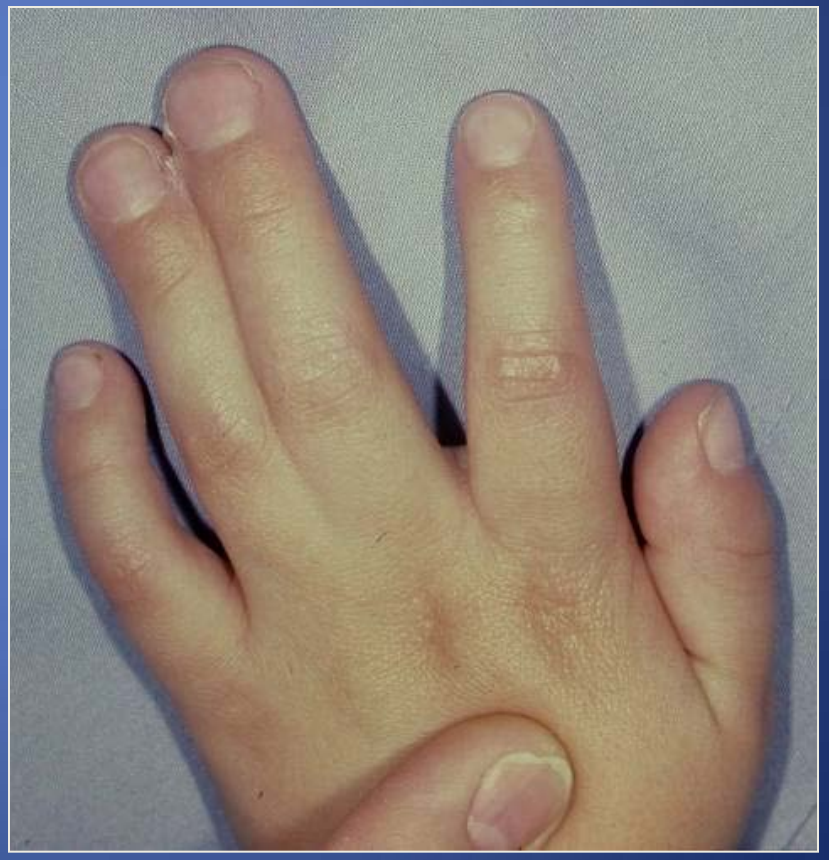
- Syndactyly
- Camptodactyly
- Clinodactyly
- Kirner deformity

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”

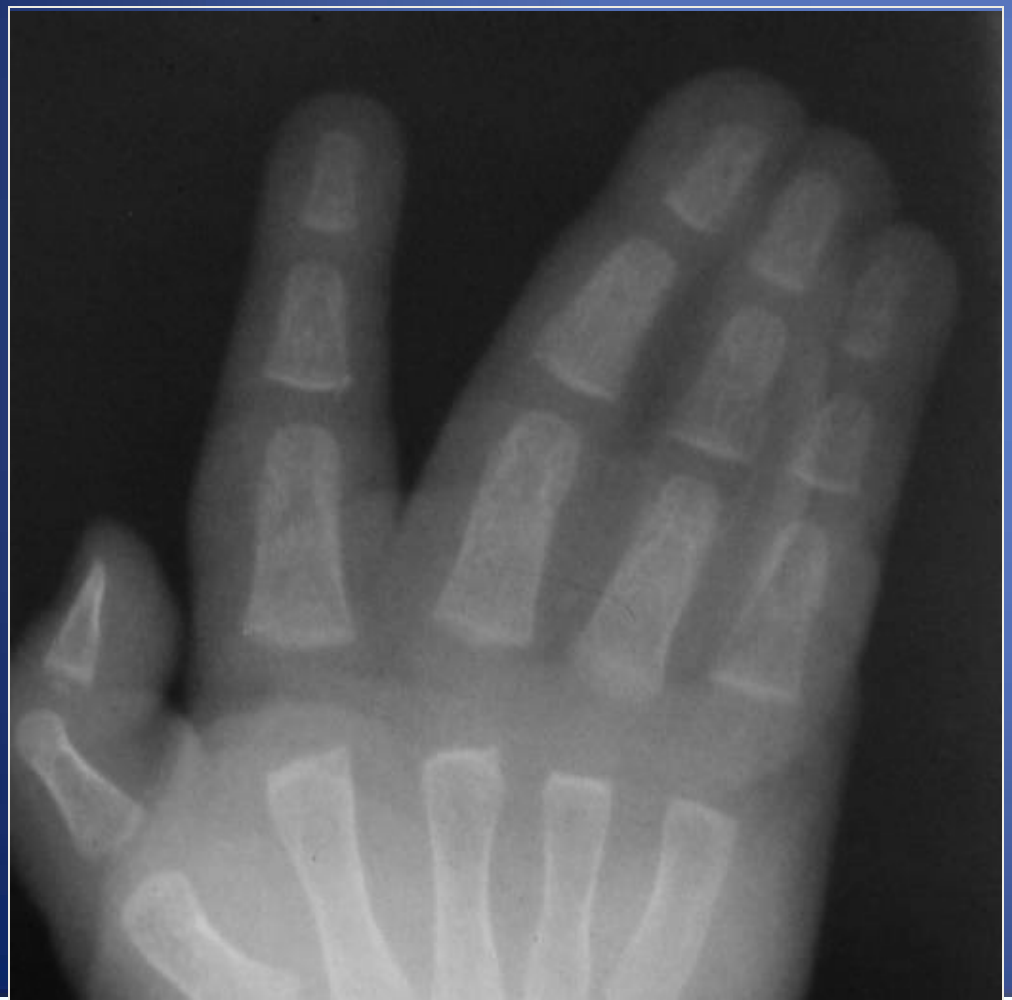
SIMPLE SYNDACTYLY

Incomplete

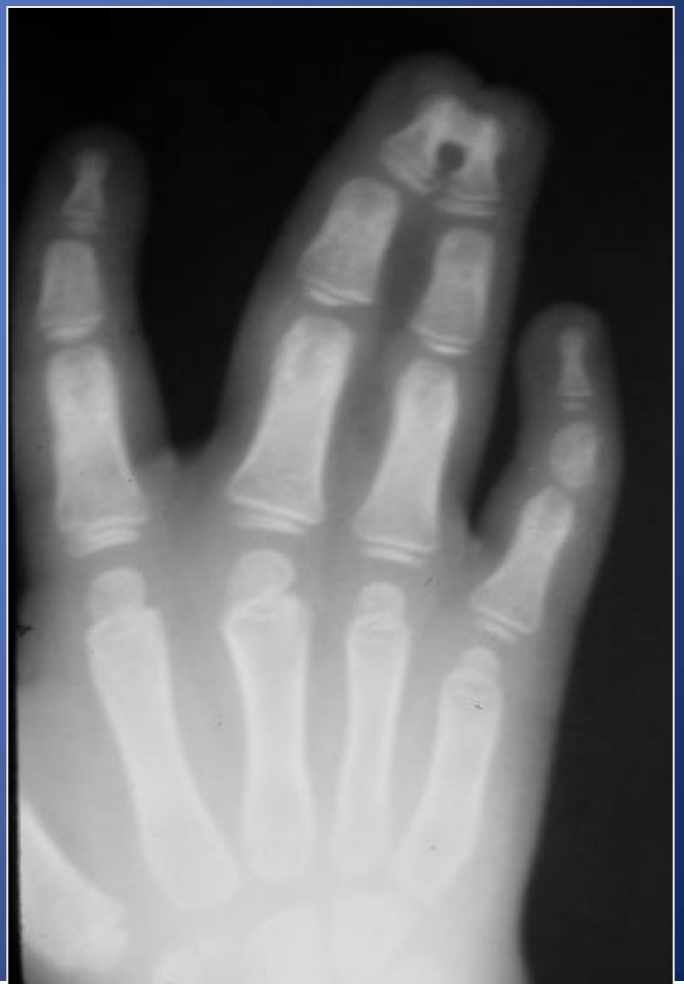


Complete

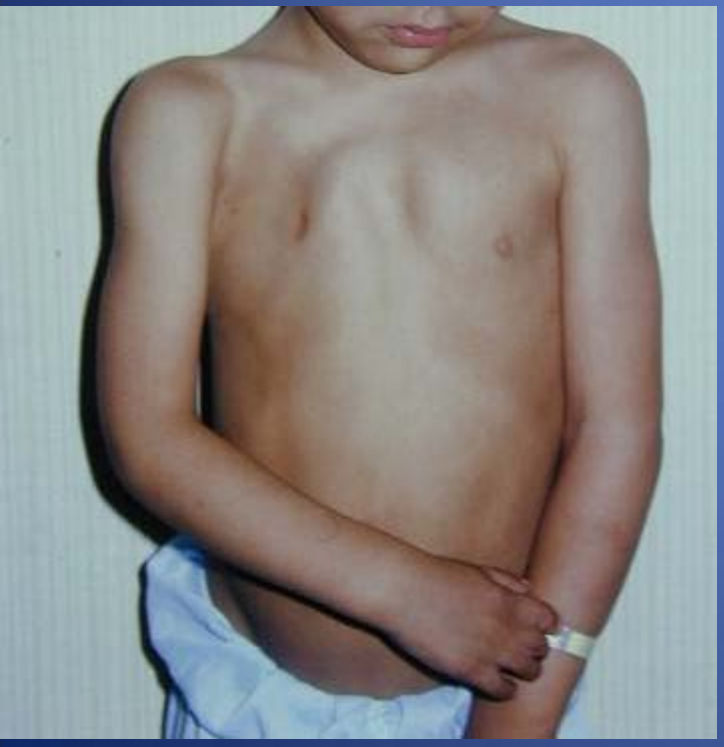
SIMPLE
COMPLETE
SYNDACTYLY
RF tethering MF

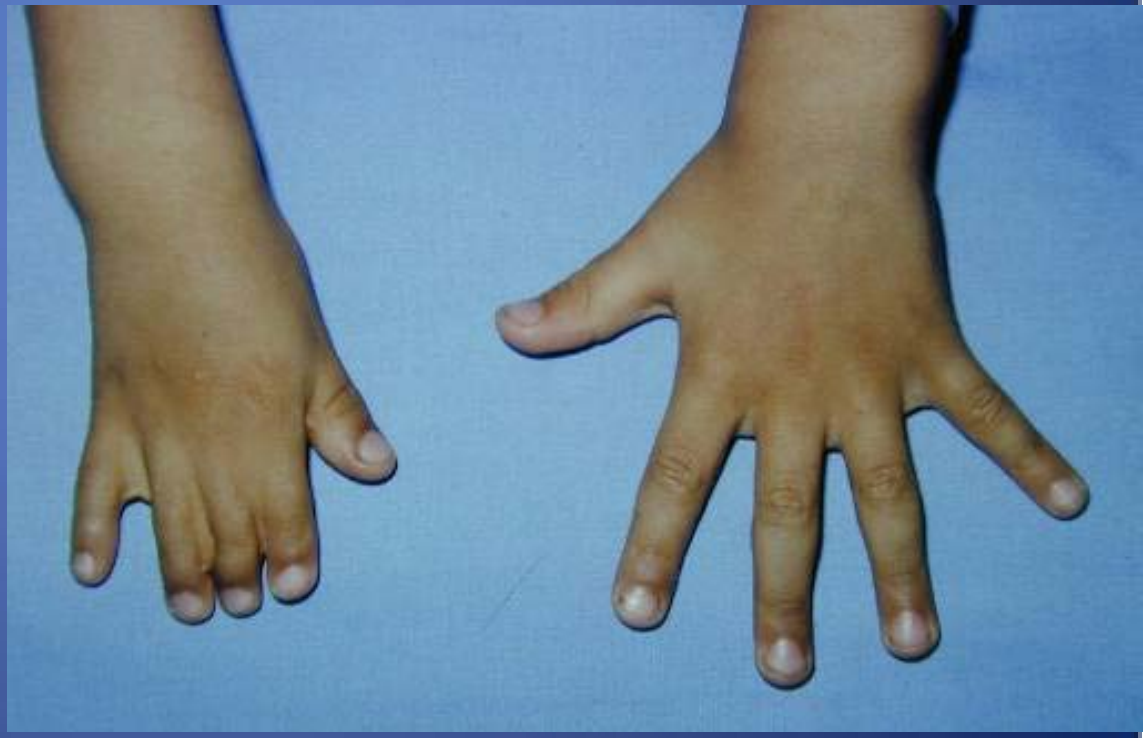


COMPLEX
COMPLETE
SYNDACTYLY

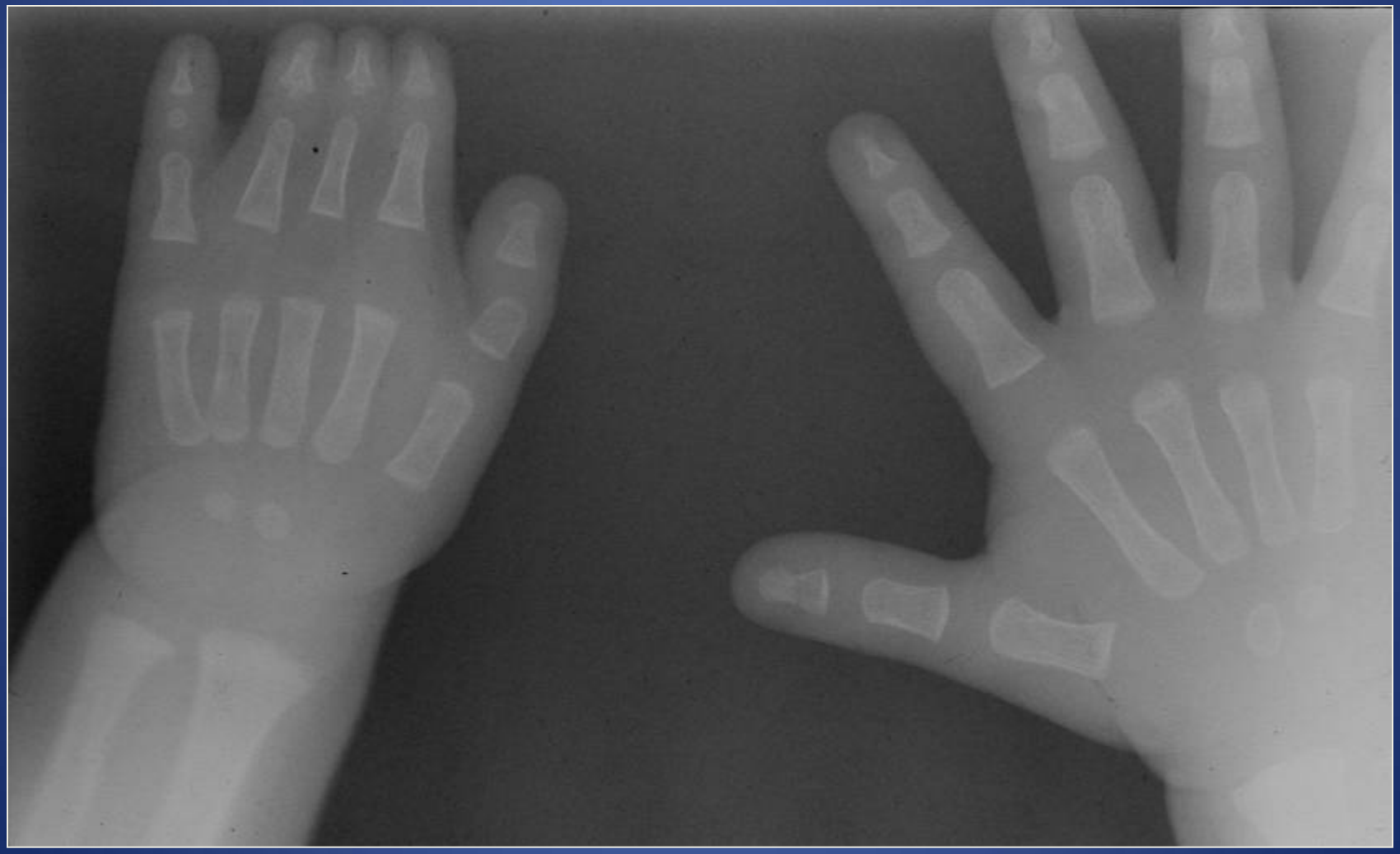


POLAND’S SYNDROME
symbrachydactyly
chest wall deformity
Thought to be vascular anomaly



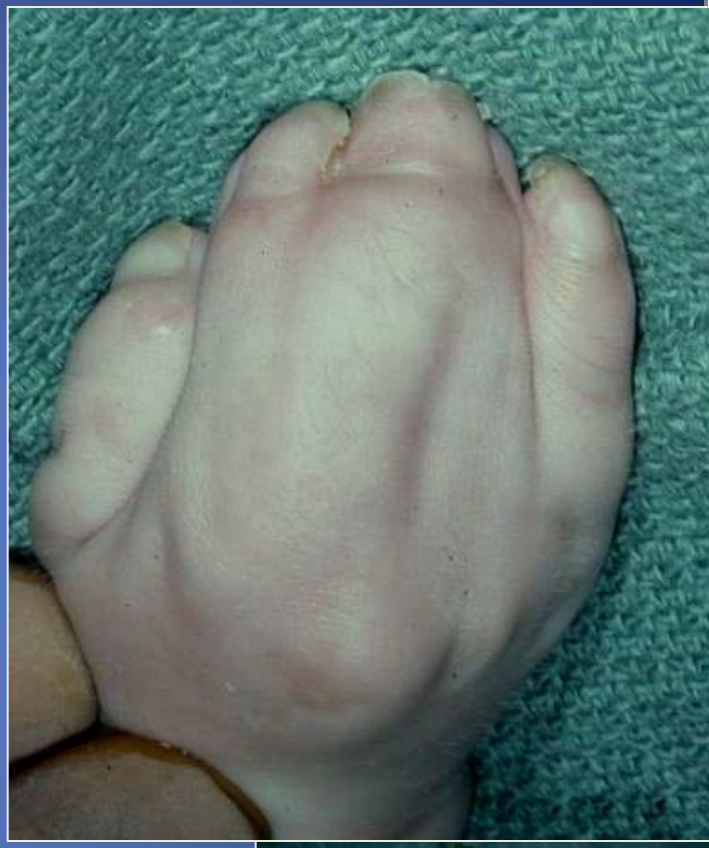


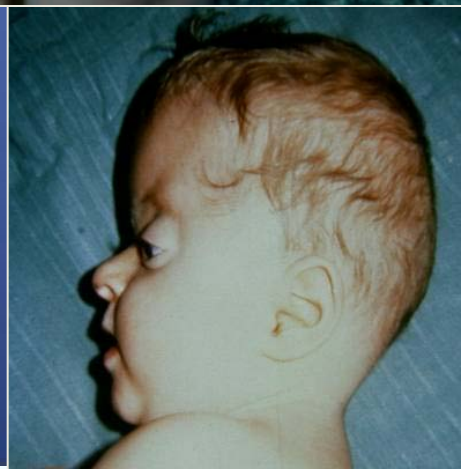
POLAND'S SYNDROME
absent PIP joints

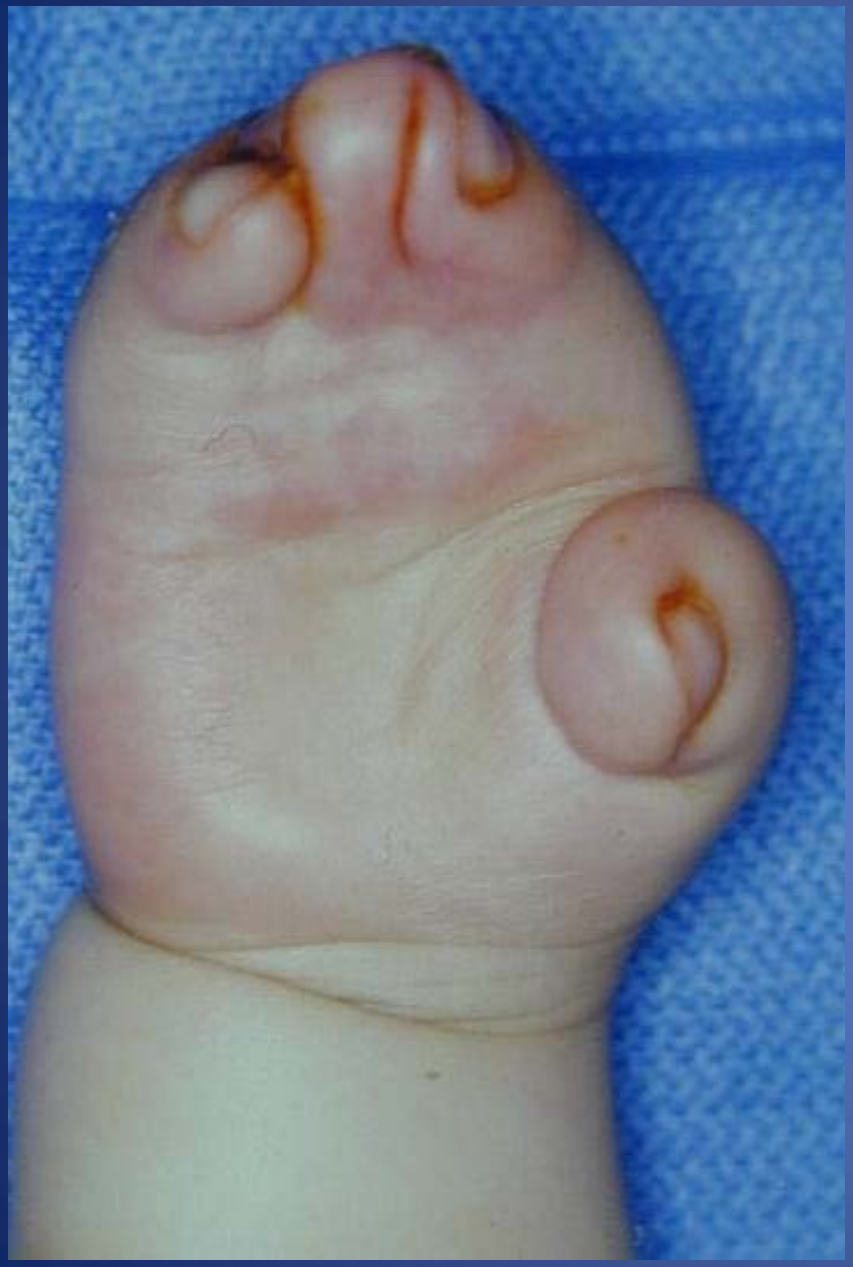


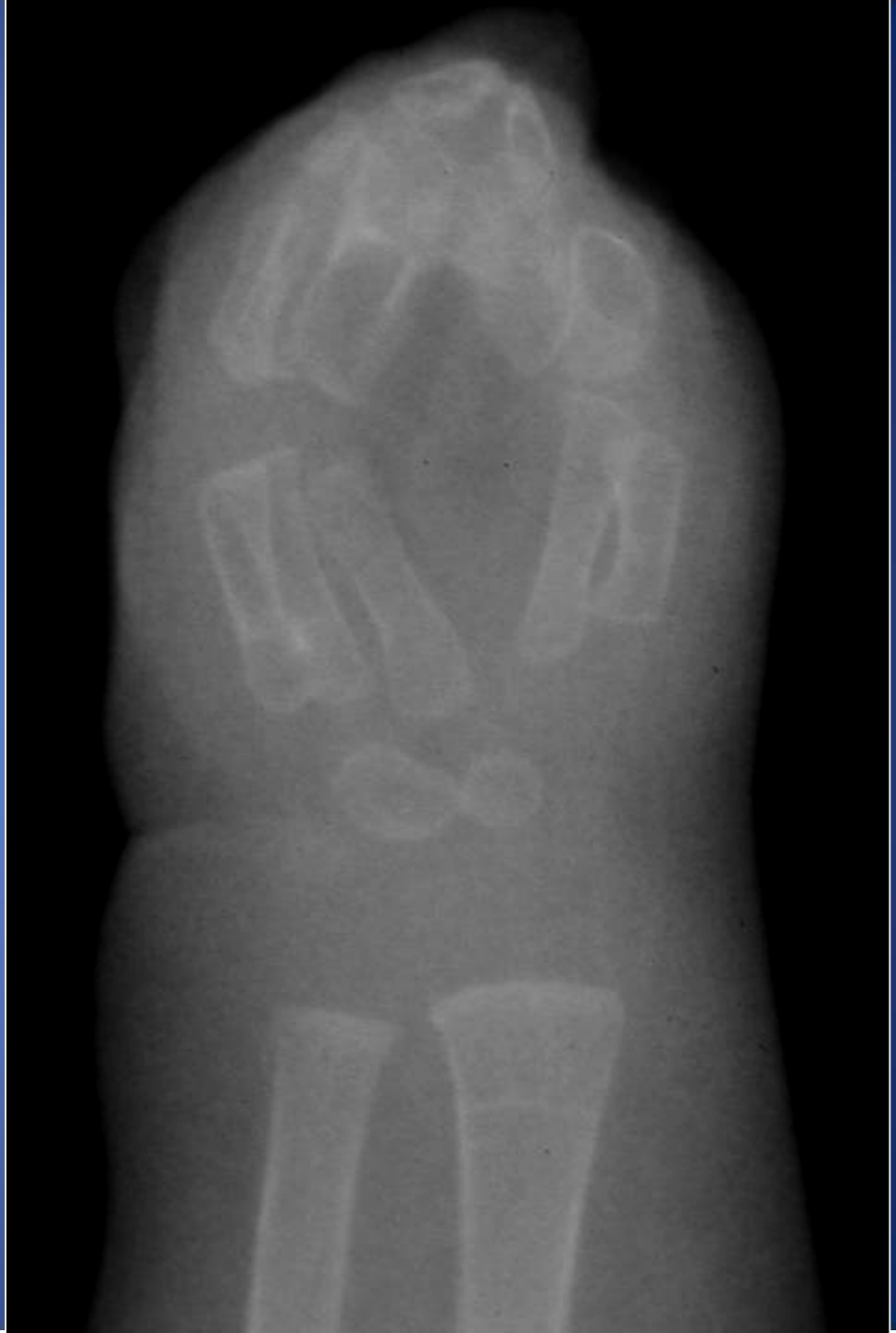
ACROCEPHALOSYNDACTYLY

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

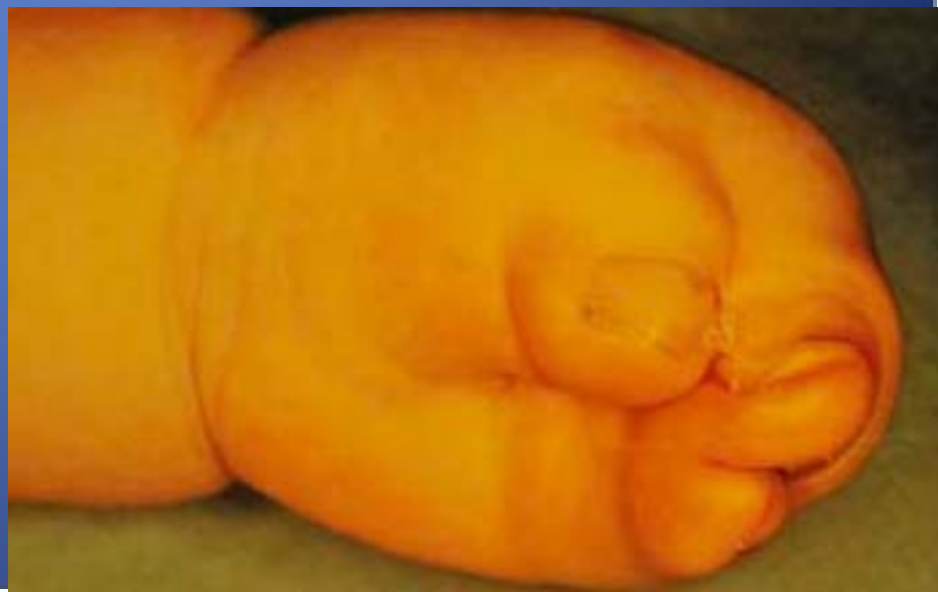






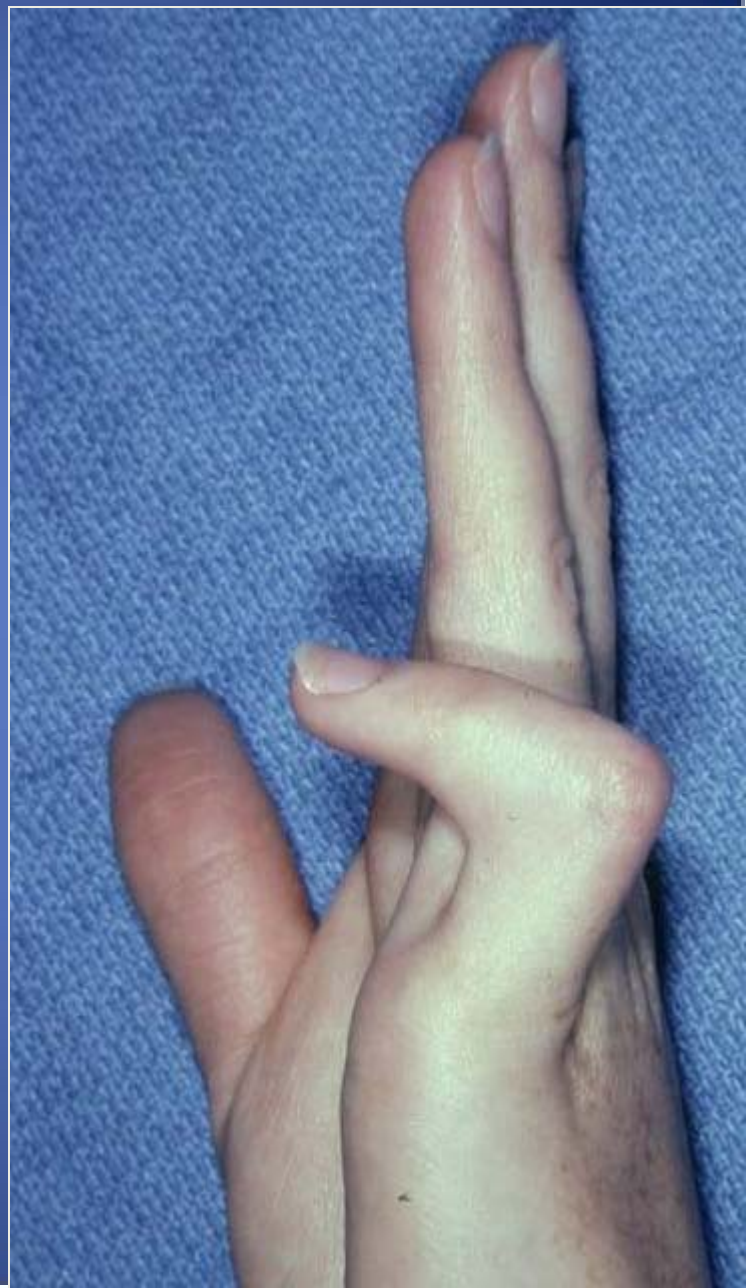


- 71. The infant whose hands are shown in Figure 1
- is expected to have a mutation in which gene?
- A. FGF receptor 2 (FGFR2)
- B. Homeobox D13 (HOXD13)
- C. Holt-Oram syndrome 1 (HOS1)
- D. Human zinc factor protein gene 3 (GLI3)
- E. Brachyury T-box 5 (TBX5)



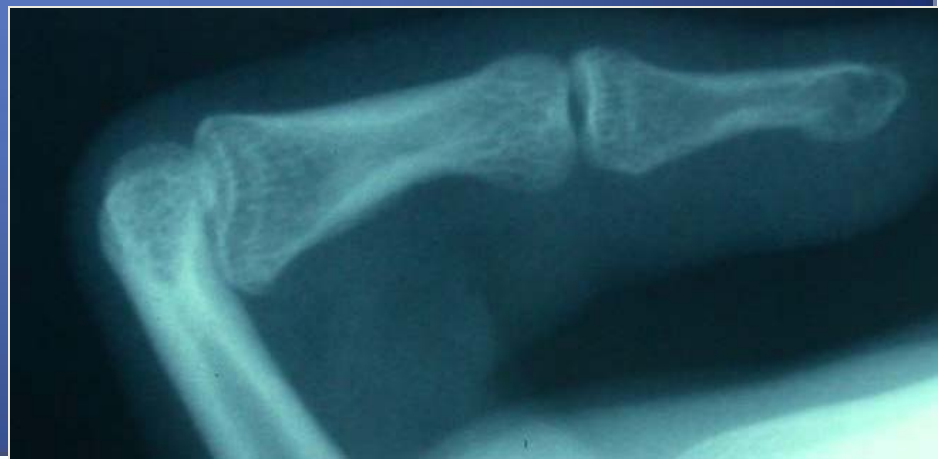
CAMPTODACTYLY

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



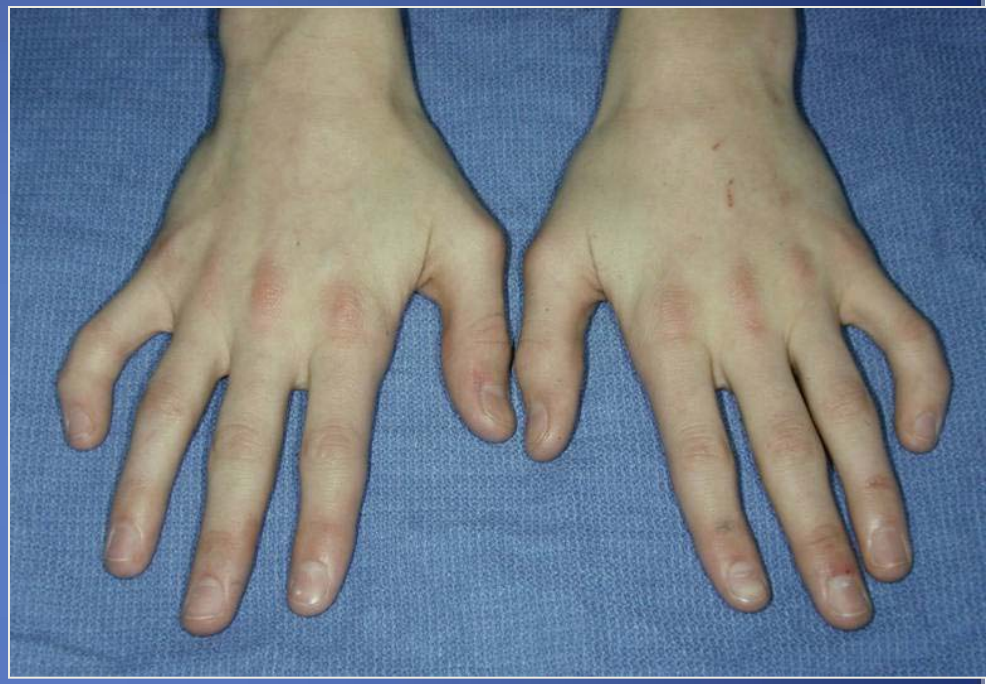
CAMPTODACTYLY Treatment

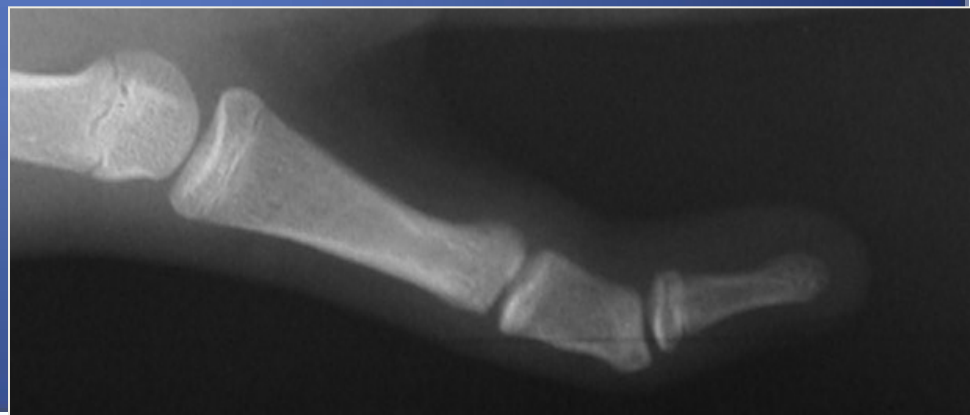
- Advise parents to accept the deformity
- Passive stretching, serial splinting rarely successful
- Consider surgery:
  - Young child with severe contracture
  - Rapid progression
- ???earlier intervention before fixed deformity



CLINODACTYLY

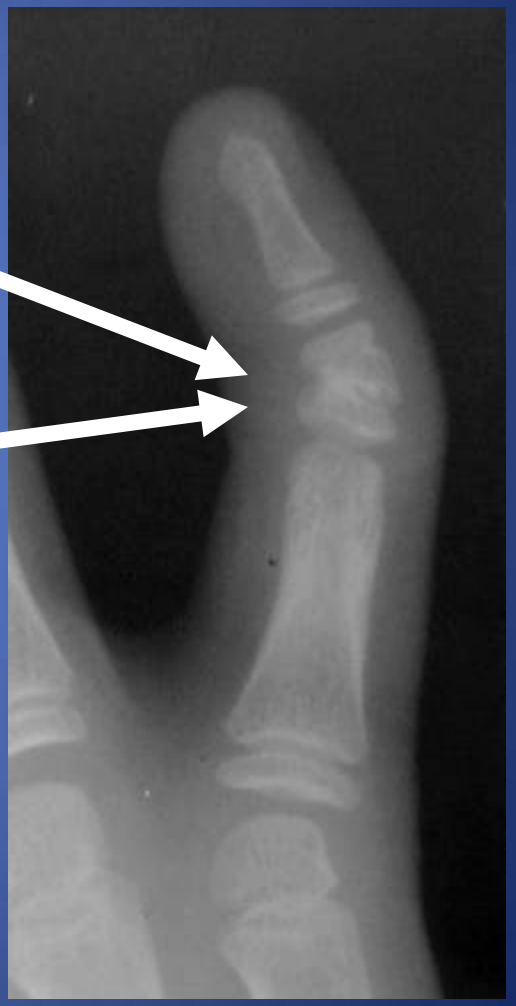
- **Radial-ulnar curvature**
- **Usually small finger**
- **Usually radial deviation**
- **Delta phalanx**





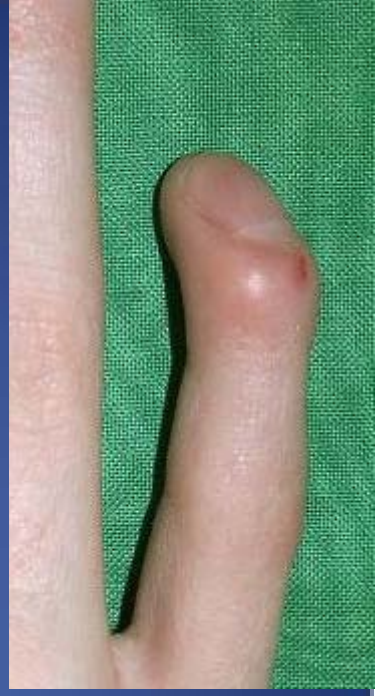
DELTA PHALANX

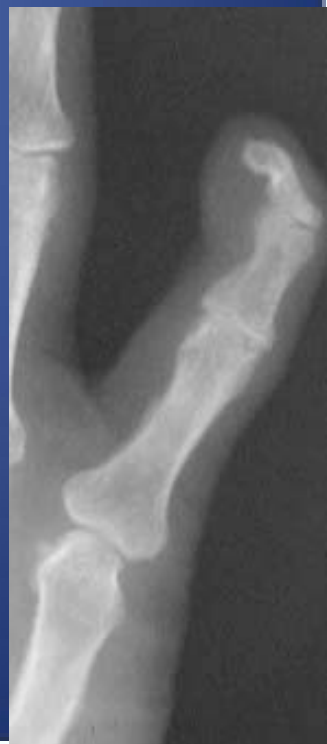
- Trapezoidal-shaped phalanx
- Abnormal C-shaped epiphysis
- Usually middle phalanx of the small finger



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

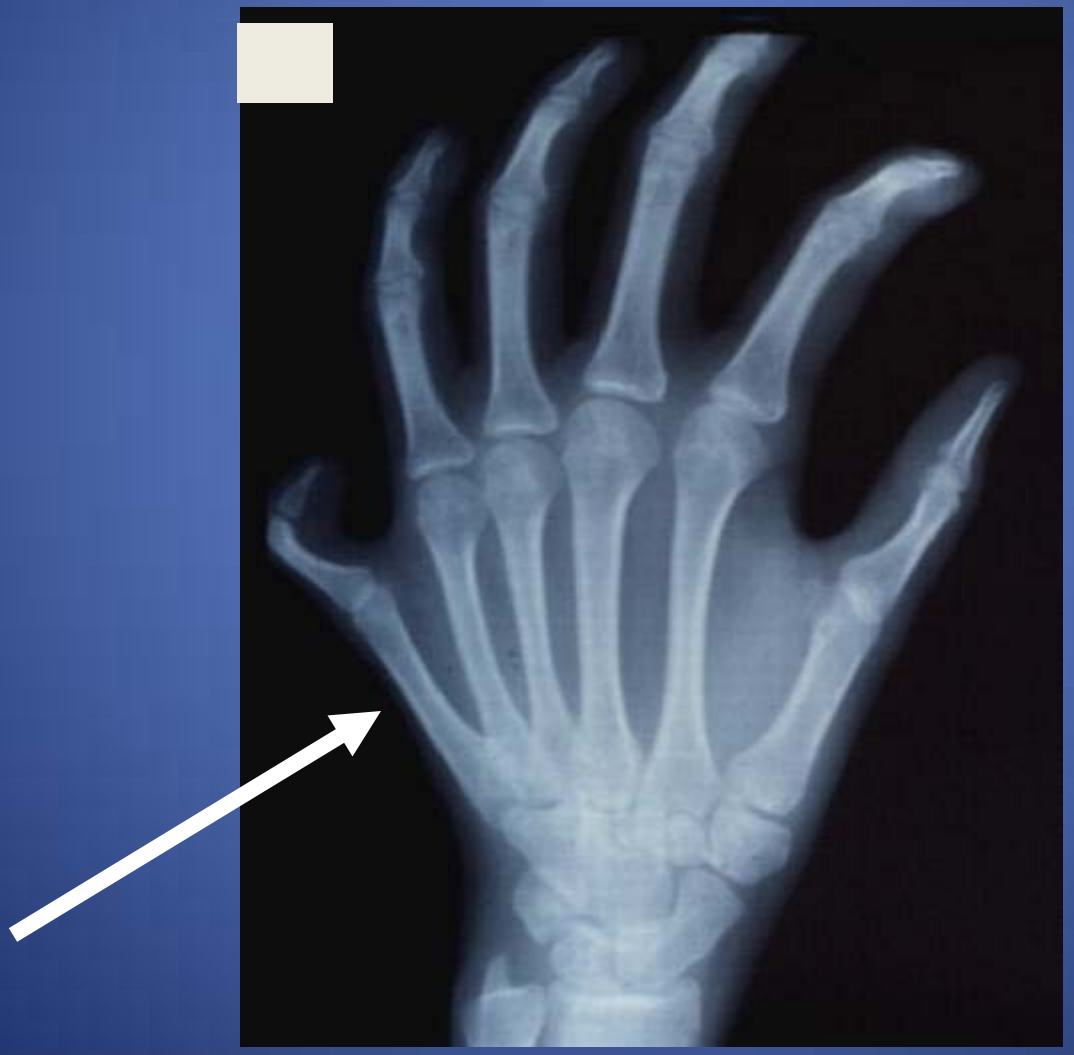
POLYDACTYLY

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

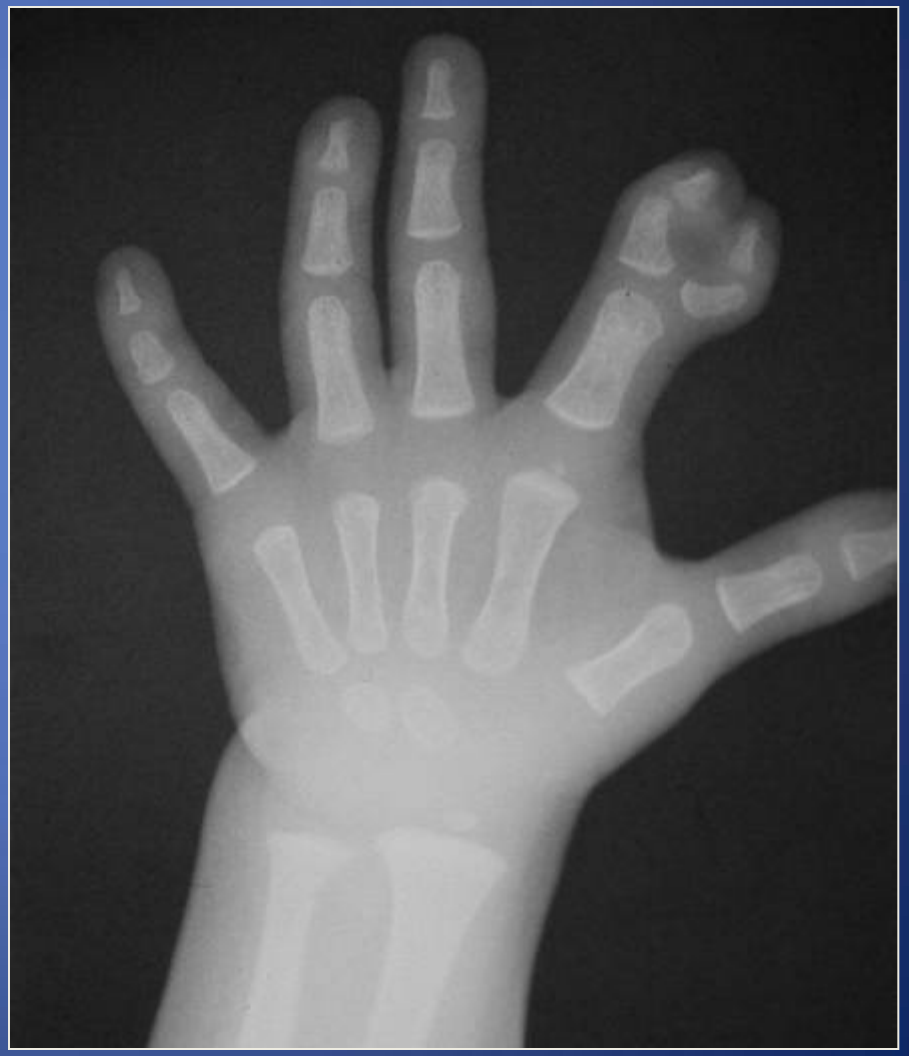
SMALL FINGER POLYDACTYLY
Type 1 – Rudimentary Soft Tissue

SMALL FINGER POLYDACTYLY

Type III – Metacarpal Duplicated



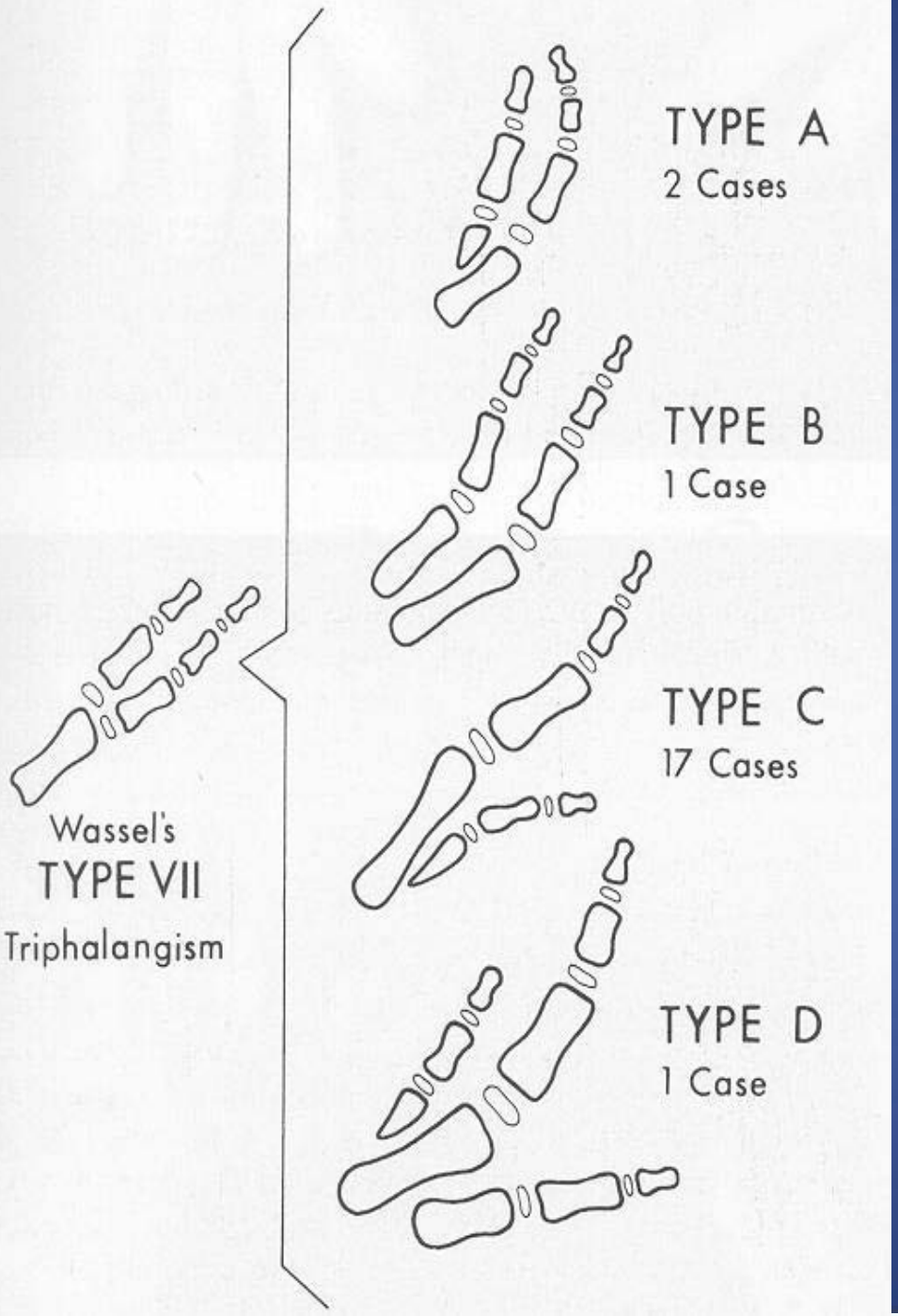
INDEX FINGER DUPLICATION



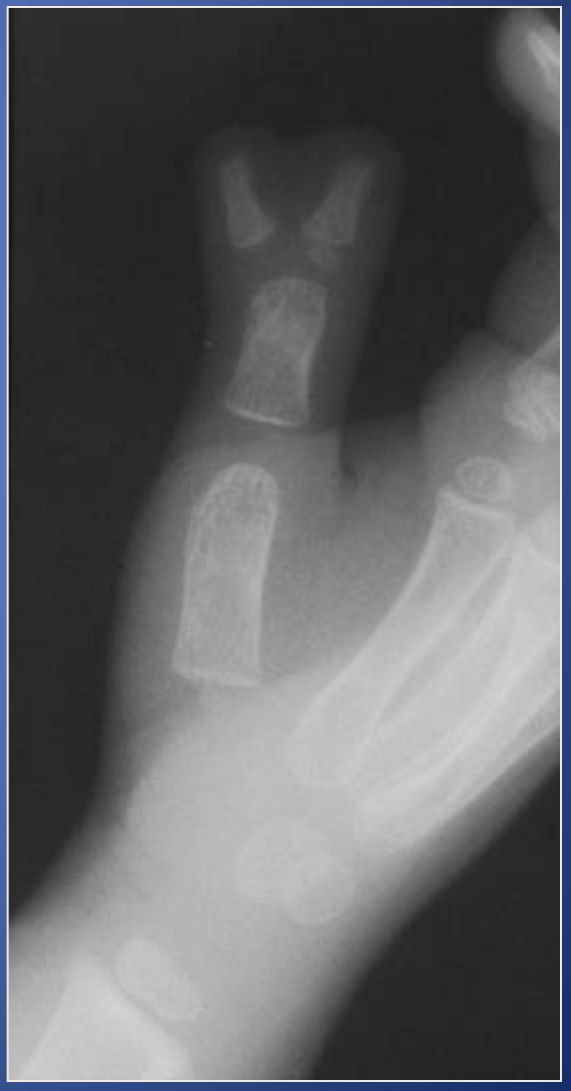
THUMB POLYDACTYLY
“Duplicate Thumb”

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

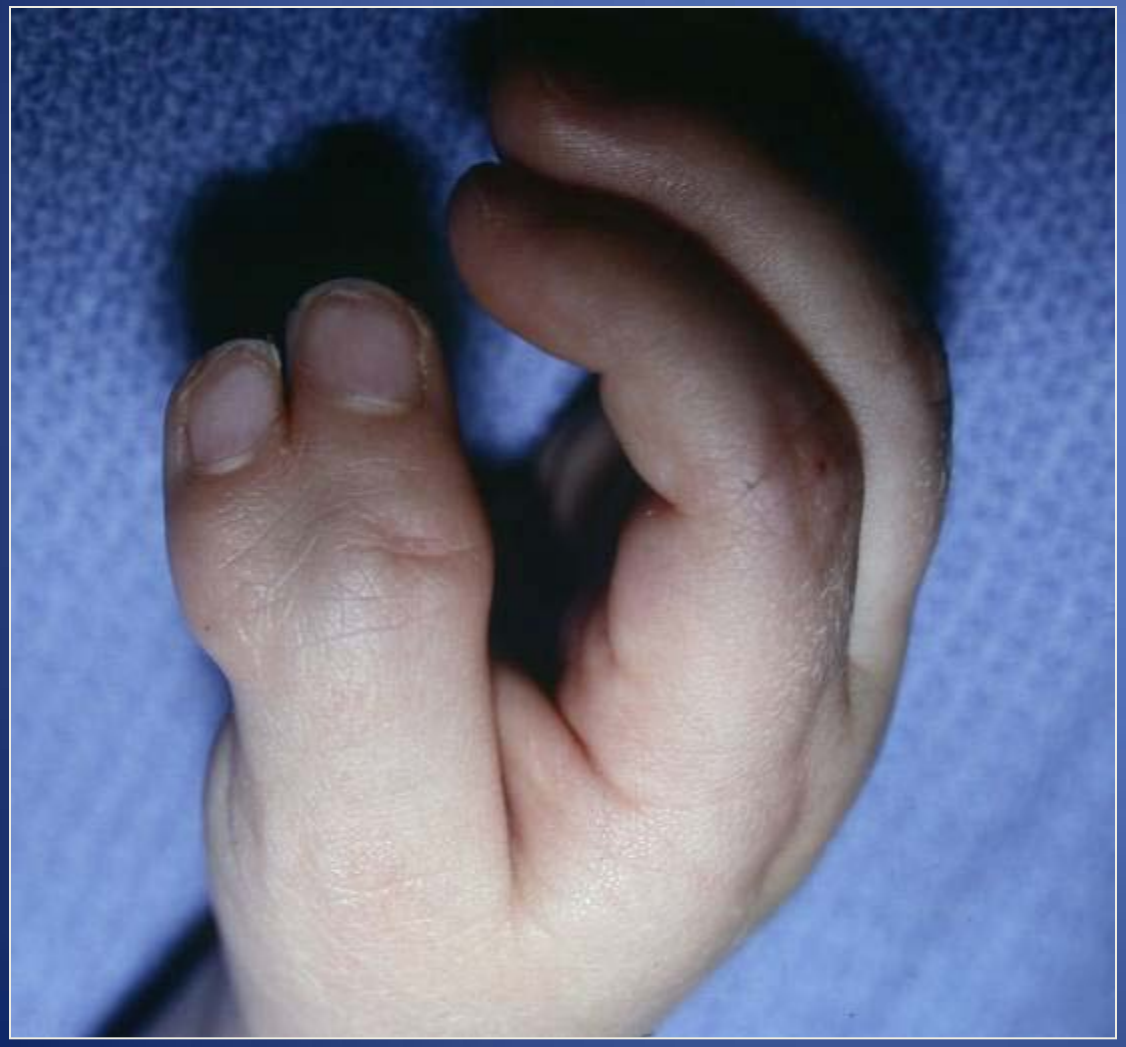
Types	I	II	III	IV	V	VI	VII
							
Bifurcation level	Distal phalanx	IP joint	Proximal phalanx	MP joint	Metacarpus	CM joint	Floating
Cases %	16 5.3%	61 20.1%	14 4.6%	97 31.9%	12 3.9%	25 8.2%	42 13.8%

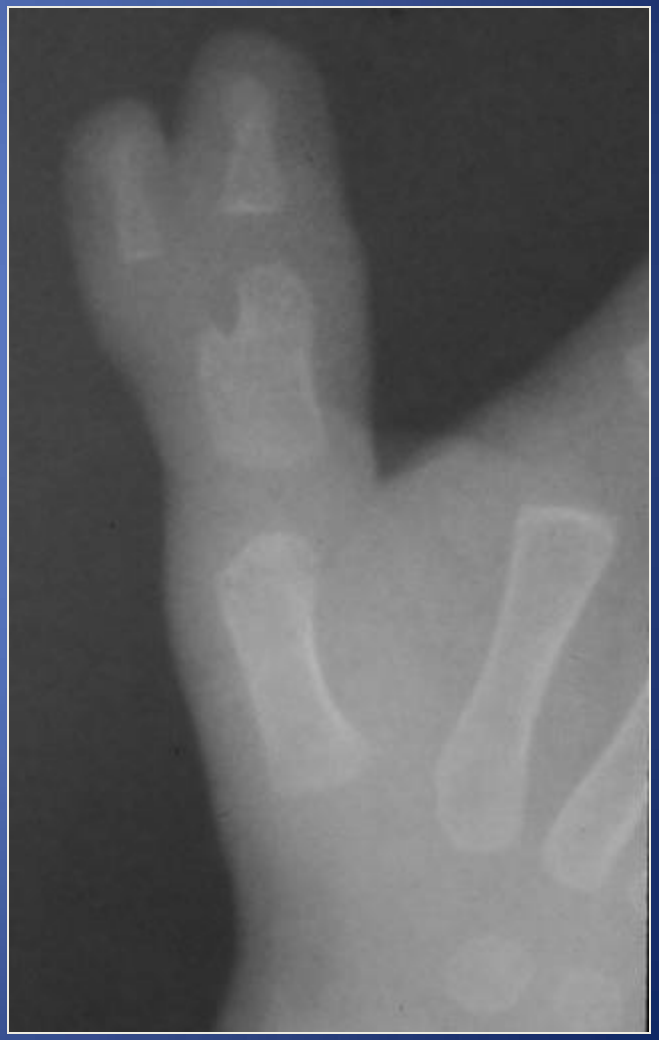

TYPE A
2 Cases
TYPE B
1 Case
TYPE C
17 Cases
TYPE D
1 Case
Wassel's
TYPE VII
Triphalangism

THUMB DUPLICATION
Type 2

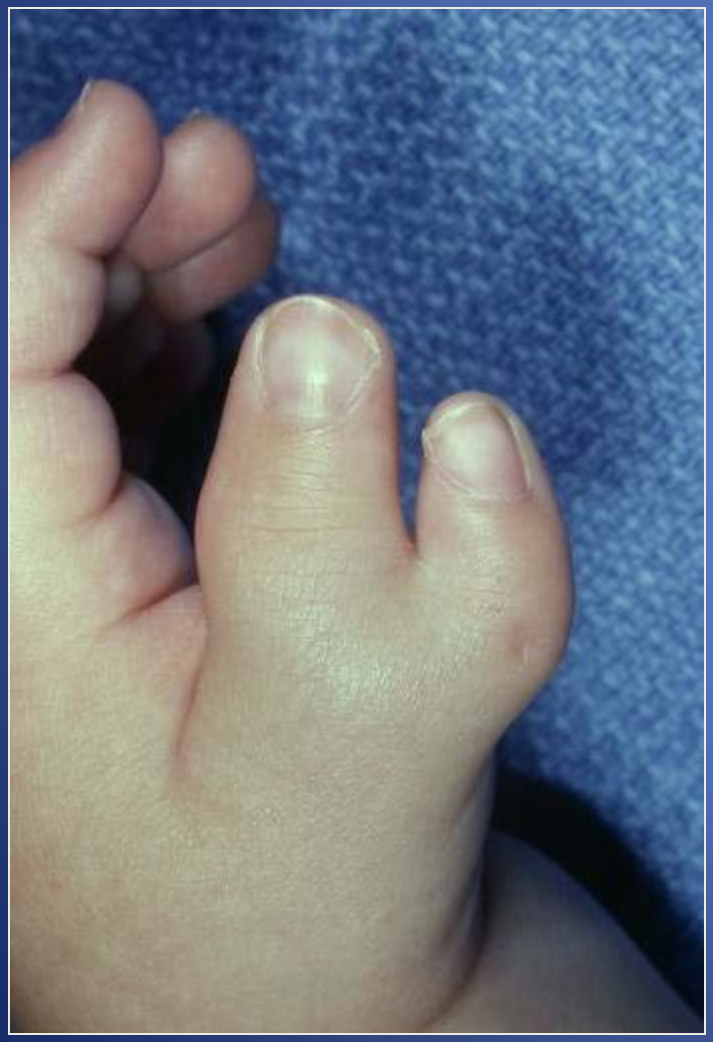


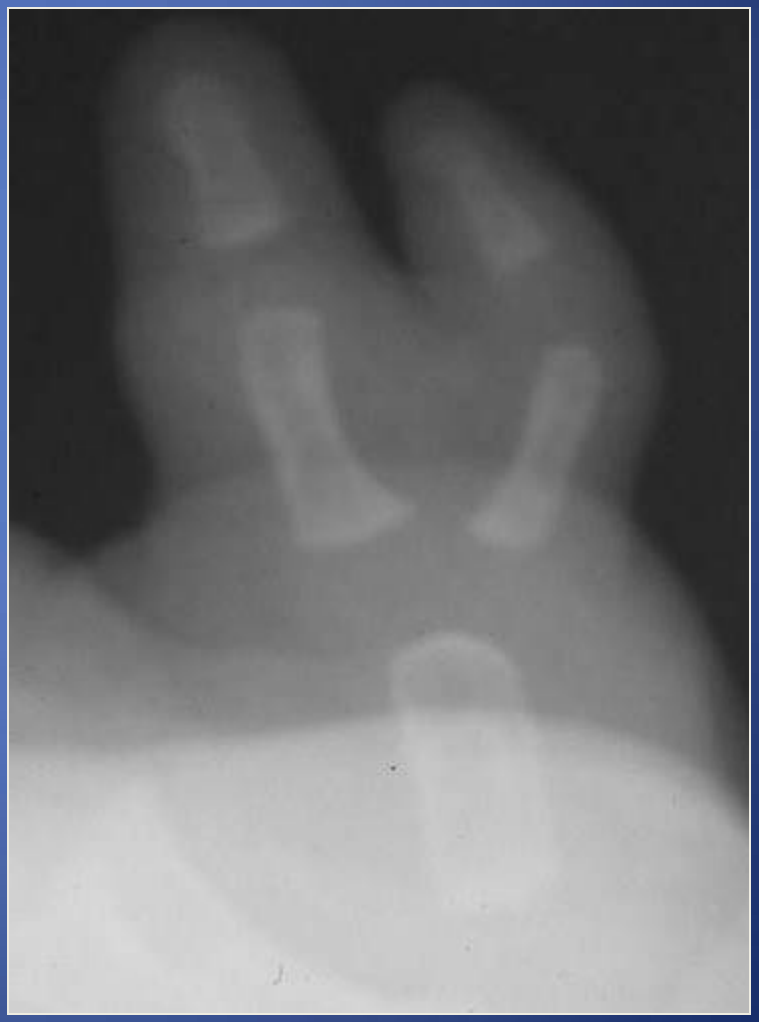
THUMB DUPLICATION
Type 3



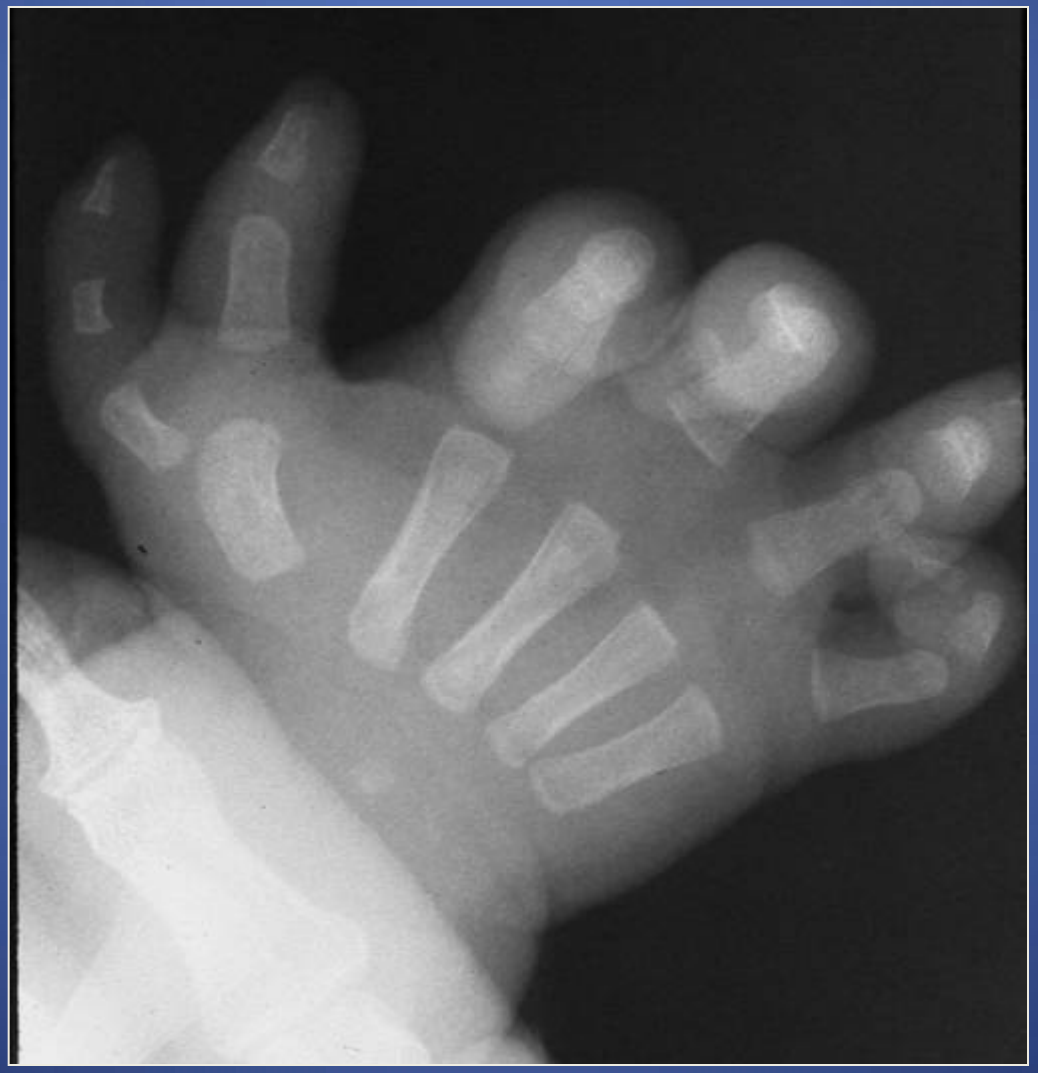


THUMB DUPLICATION
Type 4





TRIPHALANGEAL THUMB
Type 7



CONGENITAL HAND ANOMALIES
Classification

I Failure of formation of parts

II Failure of differentiation

III Duplication

IV Overgrowth

V Undergrowth

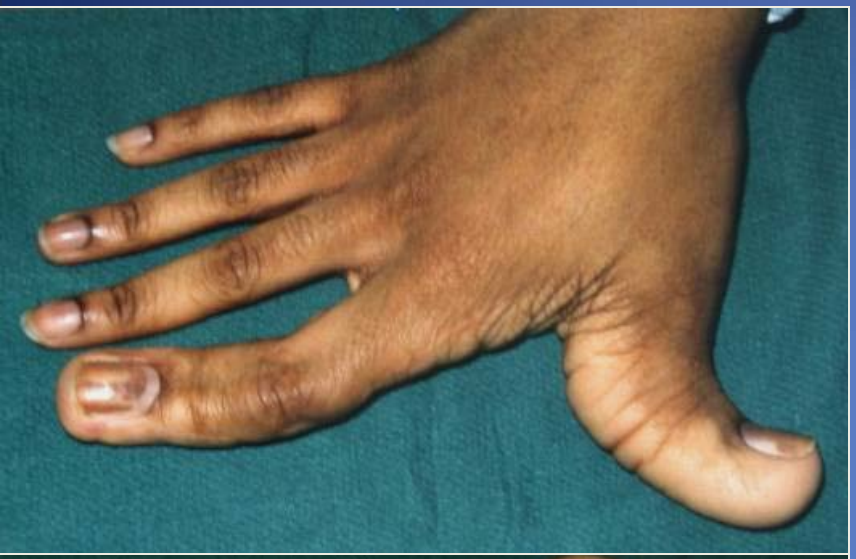
VI Constriction band syndrome

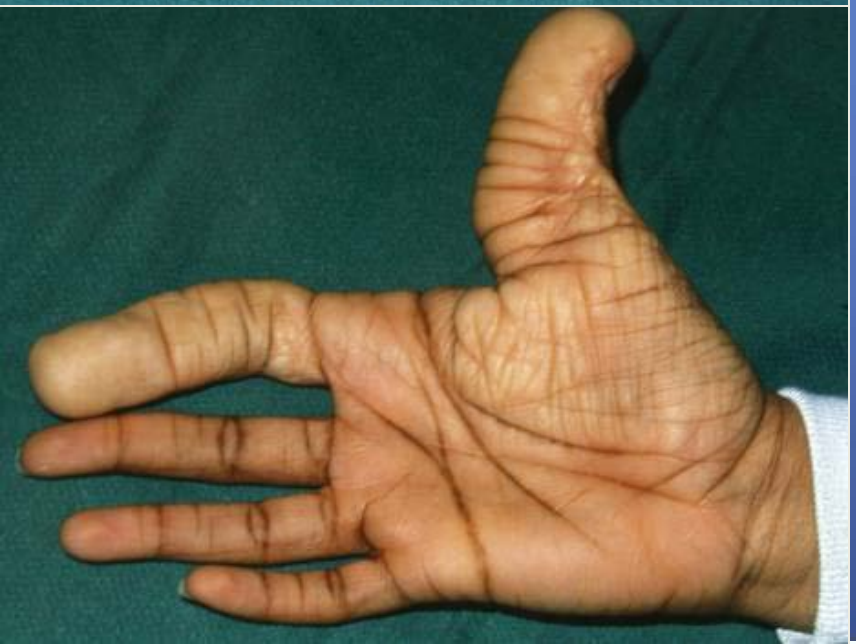
VII Generalized skeletal abnormalities

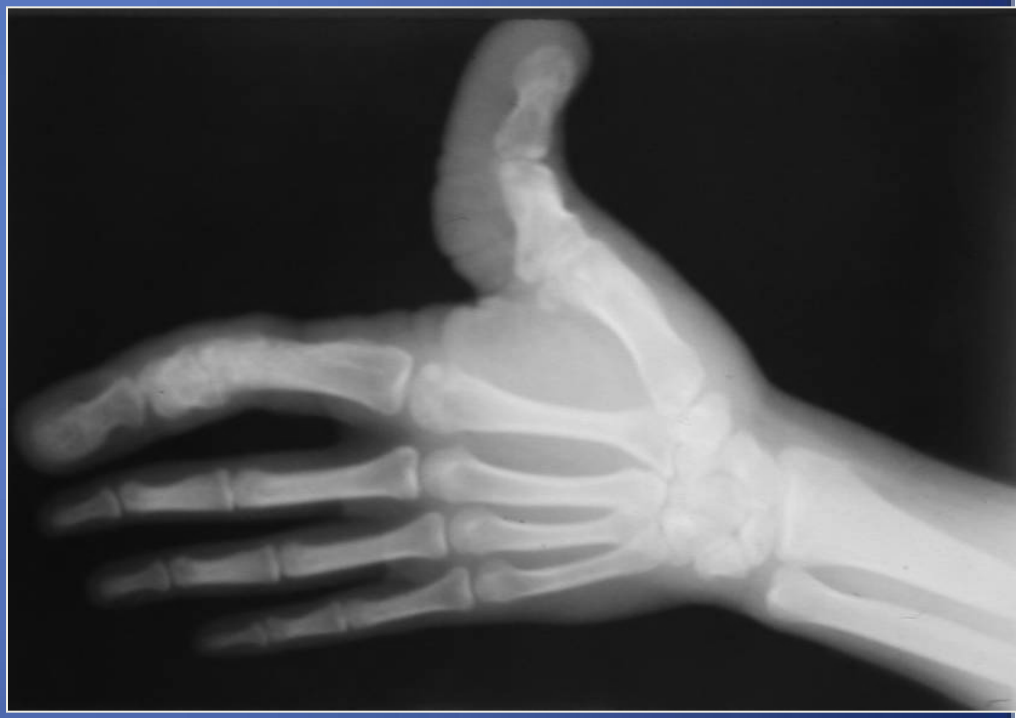
MACRODACTYLY

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

MACRODACTYLY





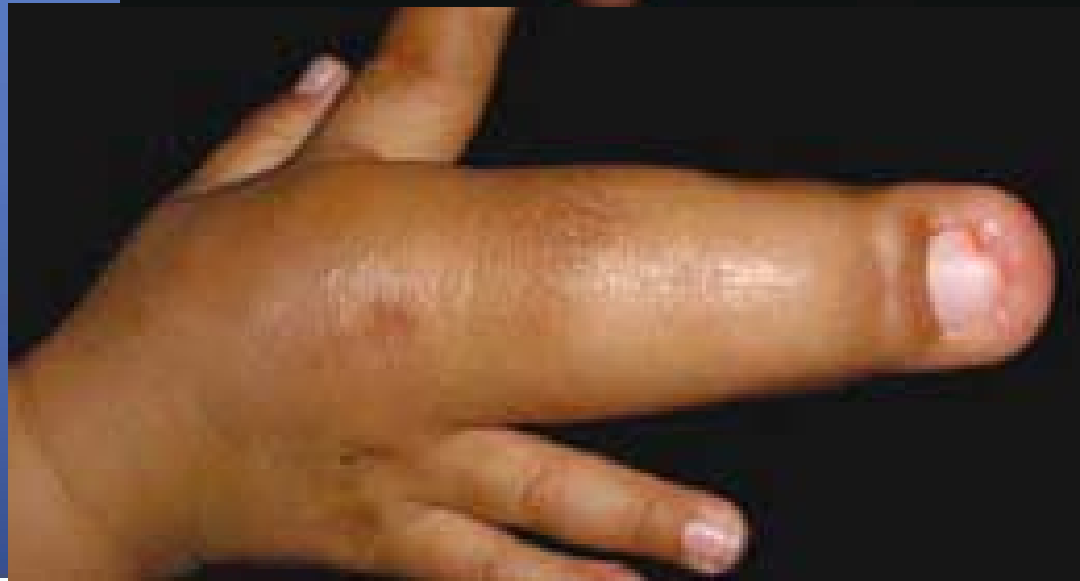


MACRODACTYLY

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

101. Which of the following treatment options is not effective in arresting the longitudinal overgrowth in the 9-month-old child with the condition depicted in Figures 1 and 2?

A. Epiphysiodesis
B. Epiphysectomy
C. Digital neurectomy
D. Digital shortening
E. Debulking



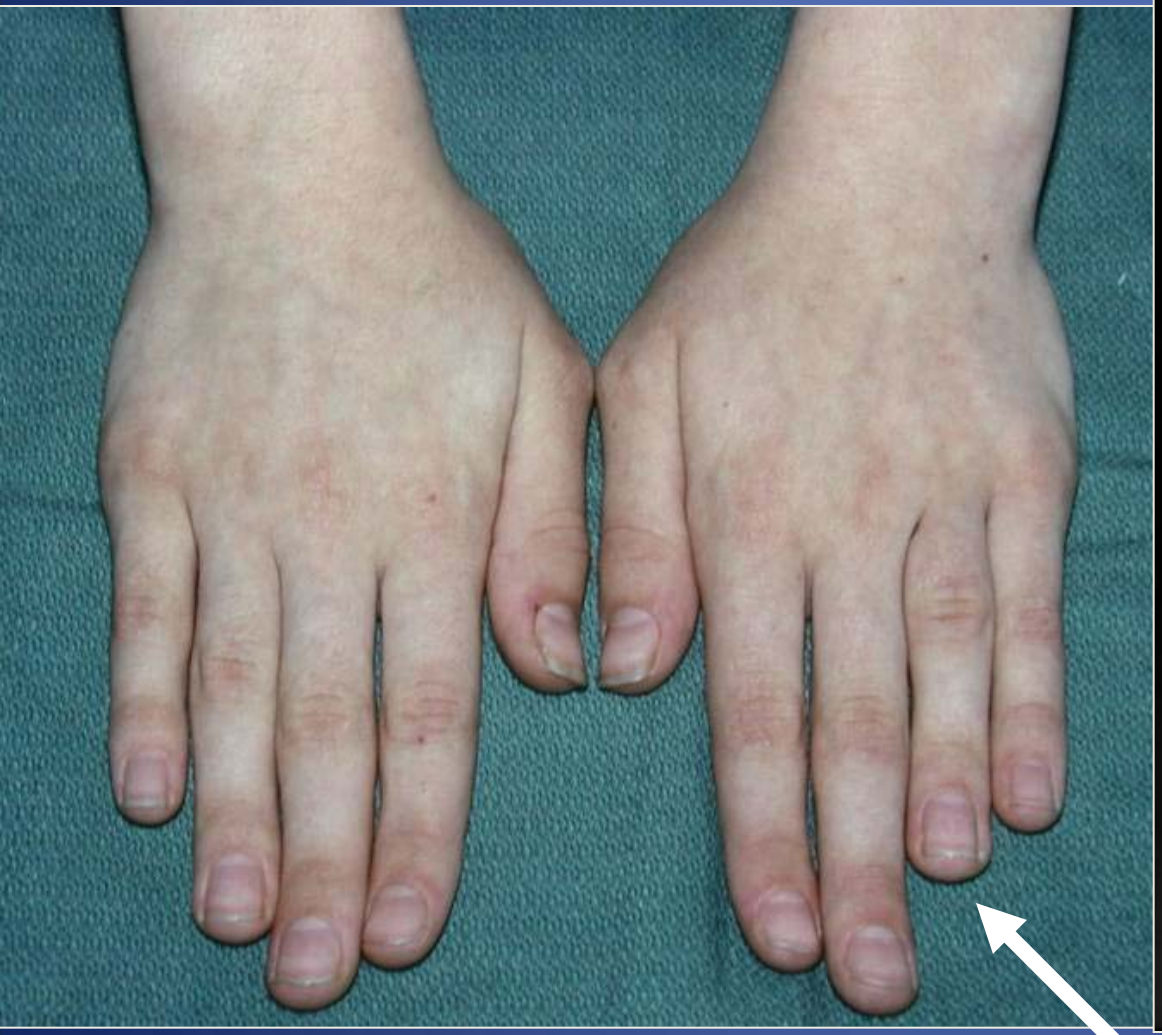
CONGENITAL HAND ANOMALIES
Classification

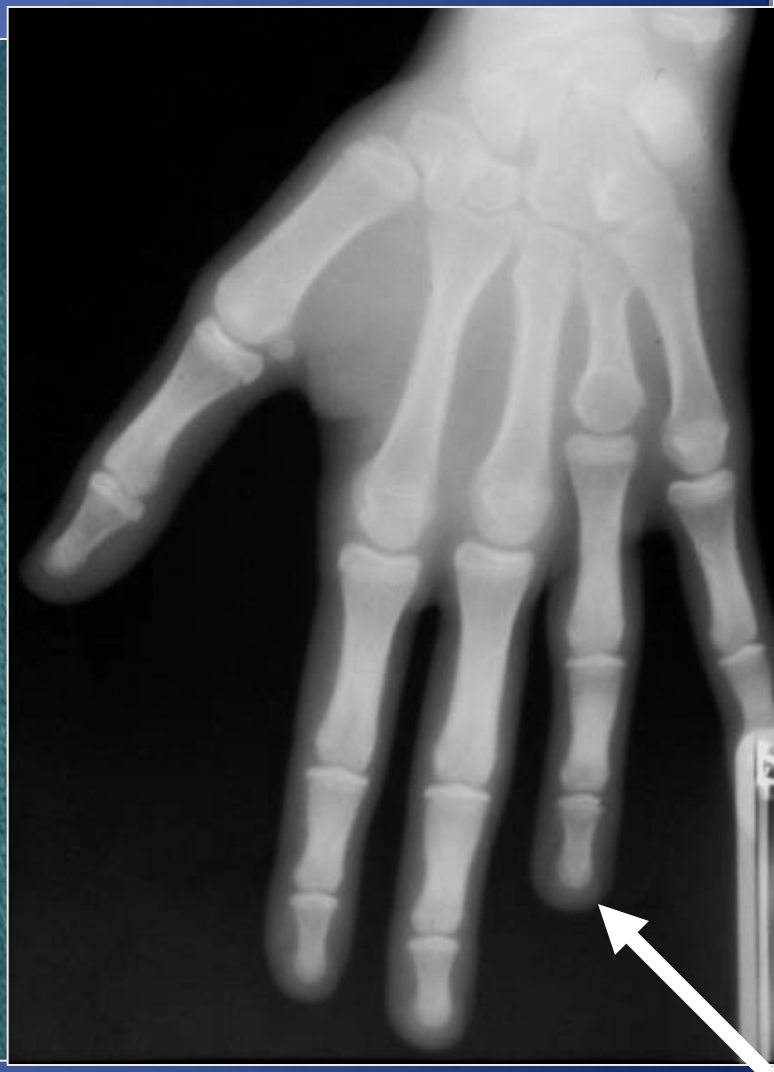
I Failure of formation of parts
II Failure of differentiation
III Duplication
IV Overgrowth

V Undergrowth (brachydactyly)

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

BRACHYMETACARPIA





CONGENITAL HAND ANOMALIES
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VI Constriction band syndrome

VII Generalized skeletal abnormalities

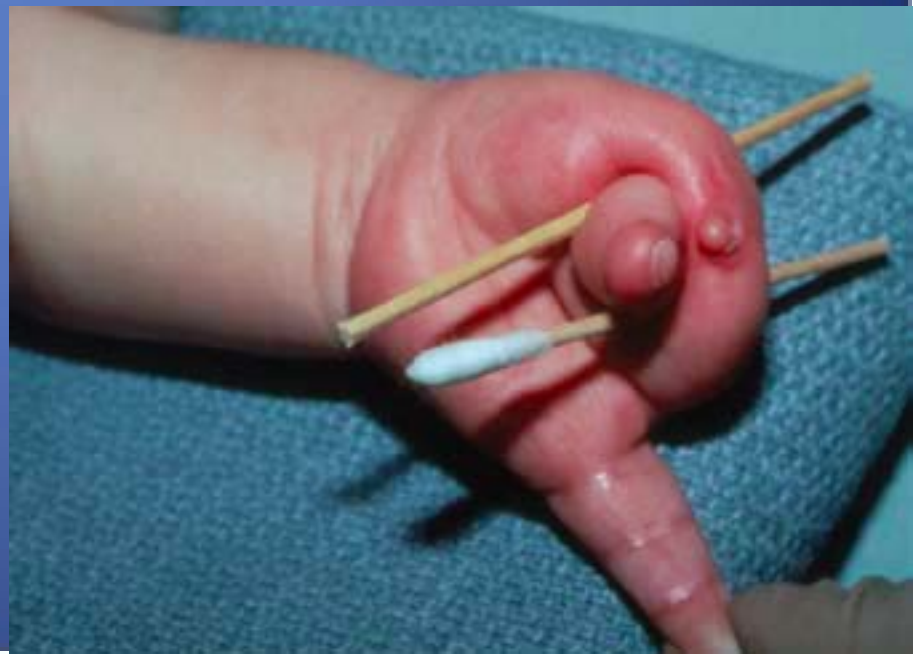
CONSTRICTION RING SYNDROME

- **Circumferential grooving or transverse amputation**

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

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

- 21. Acrosyndactyly (Figure 1), or fusion of the terminal
- portion of two or more digits with proximal sinuses
- between the digits, is seen in:
- A. Constriction band syndrome
- B. Poland's syndrome
- C. Cleft hand
- D. Simple, incomplete syndactyly
- E. Simple, complete syndactyly



CONGENITAL HAND ANOMALIES
Classification

I Failure of formation of parts

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IV Overgrowth

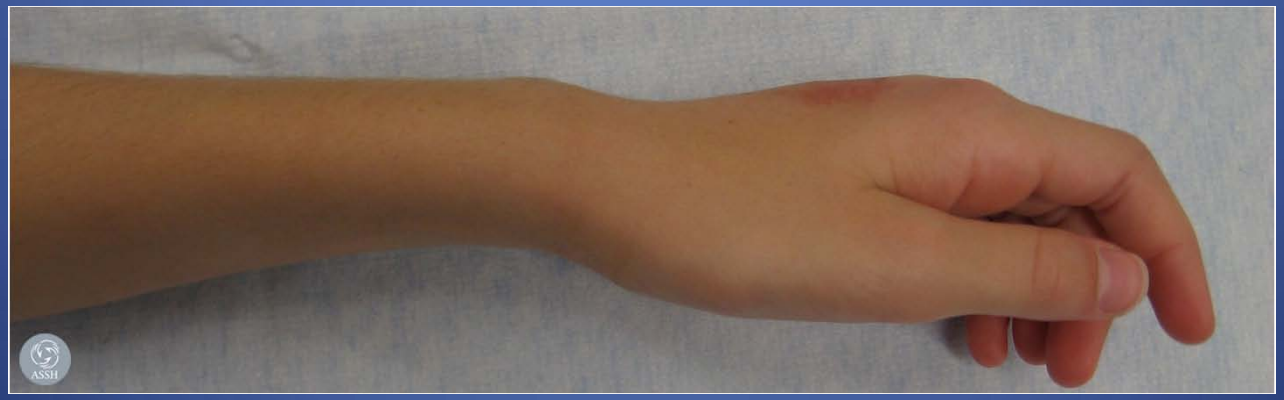
V Undergrowth

VI Constriction band syndrome

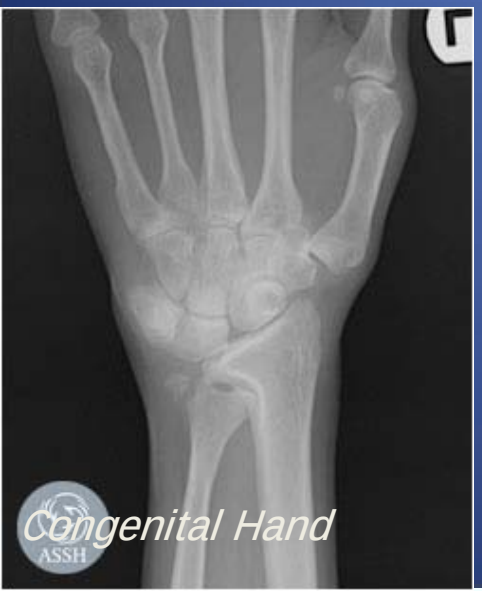
VII Generalized skeletal abnormalities

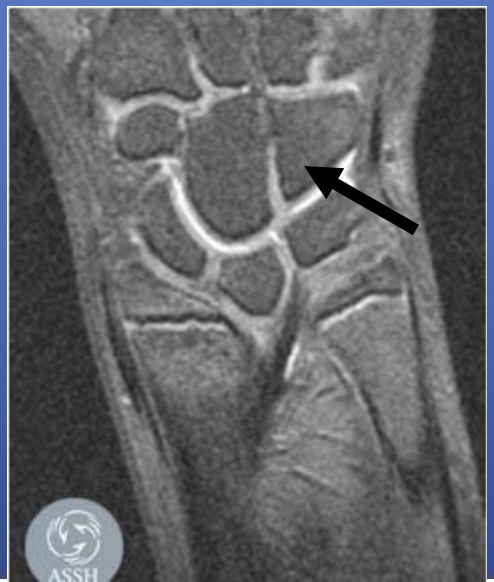
- Madelung Deformity
  - Basics
    - Focal physeal abnormality of distal radius
      - Volar-ulnar corner growth inhibition
    - Abnormal radiolunate ligament from metaphysis of distal radius
      - Vicker's ligament
    - Caused by mutation in SHOX gene
    - Associated with short stature, Turner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

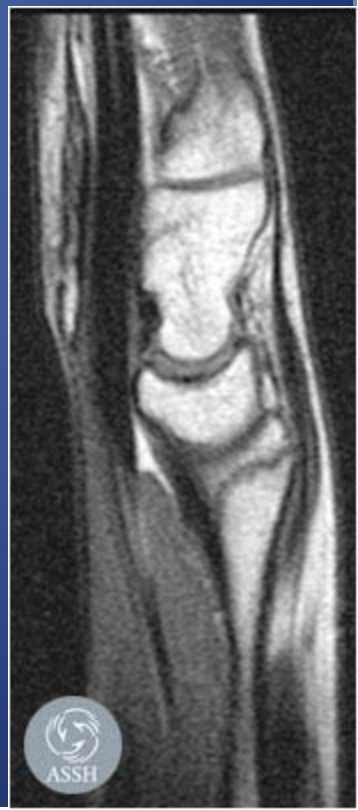
- Madelung Deformity
  - Evaluation
    - Deformity becomes evident in adolescence
    - Volar sag to carpus
    - Dorsally prominent distal ulna
    - Self-limited period of pain in adolescence



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



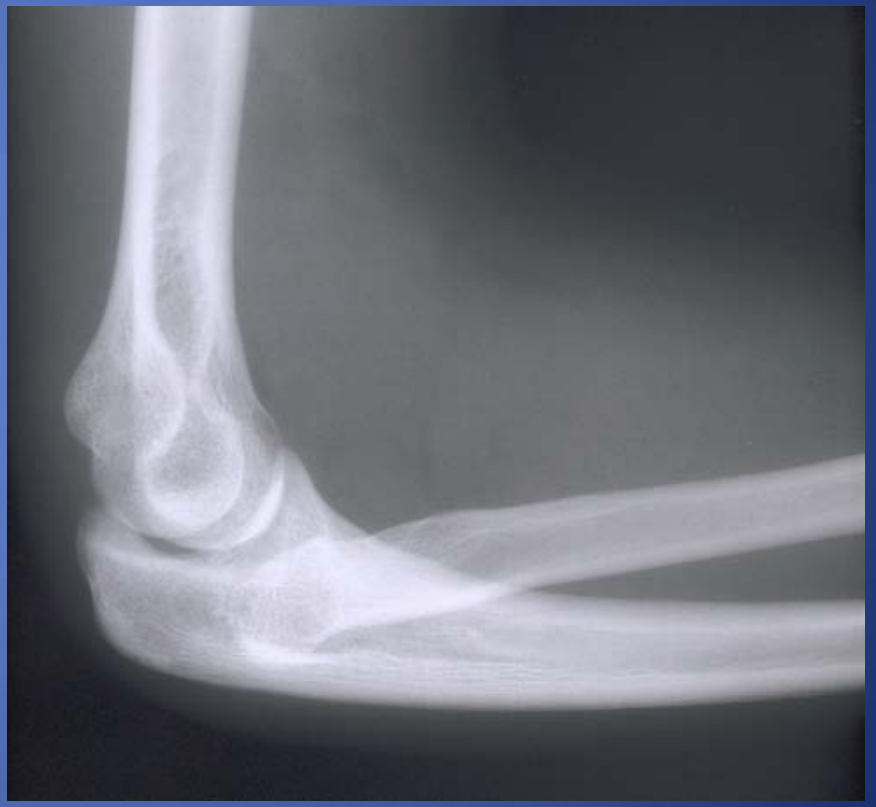




- Madelung Deformity
  - Treatment
    - Physiolysis, resection of tethering Vicker's ligament may halt progression if done early enough
  - Dome osteotomy of distal radius to correct established deformity

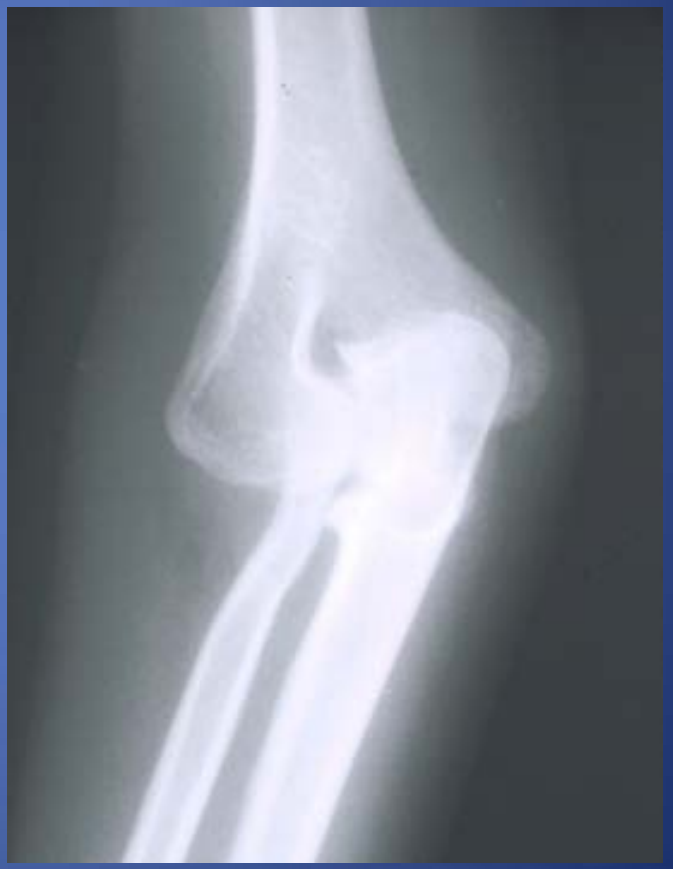
Congenital Dislocation of the Radial Head

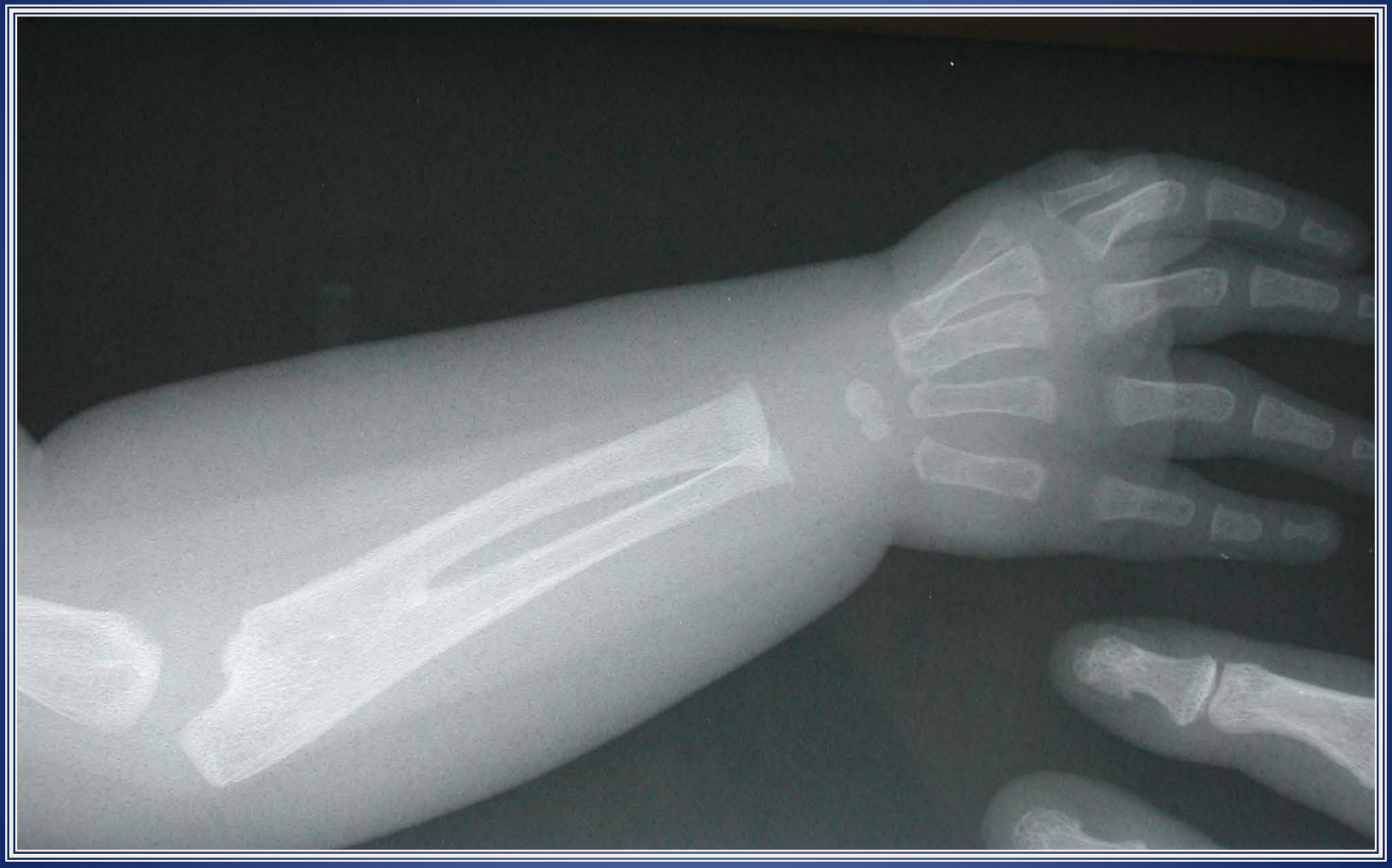
- Uncommon
- DDx: traumatic dislocation
- Sporadic or inheritable
- Isolated or syndromic
- 40-60% bilateral



Congenital Dislocation of the Radial Head

- Indications for treatment
  - PAIN
  - Skeletally mature



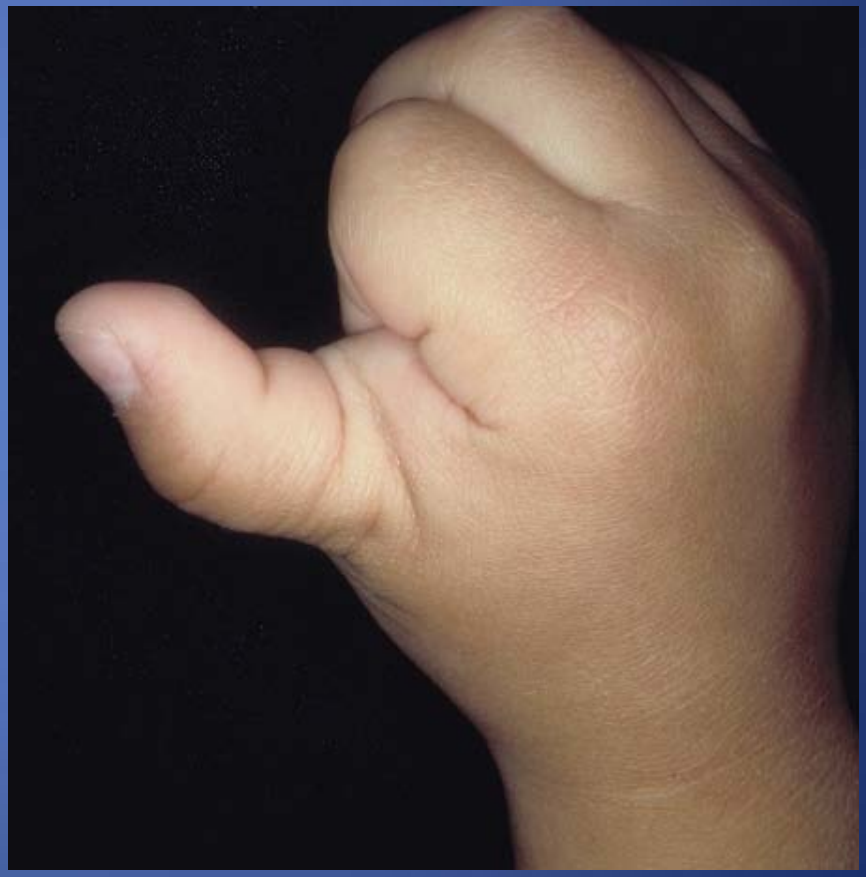


Congenital Proximal Radio-ulnar Synostosis

- 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

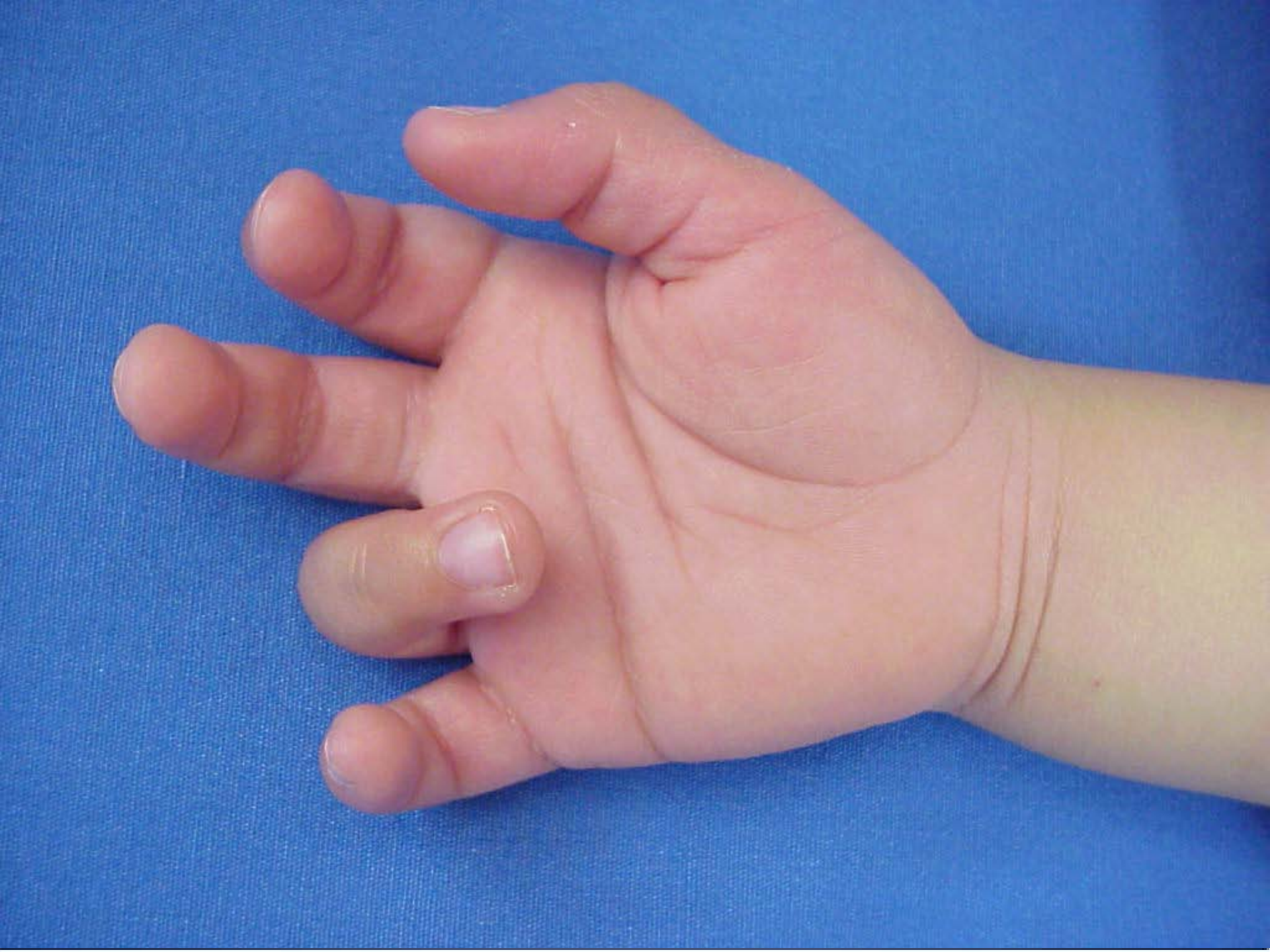
Infantile Trigger Thumb

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



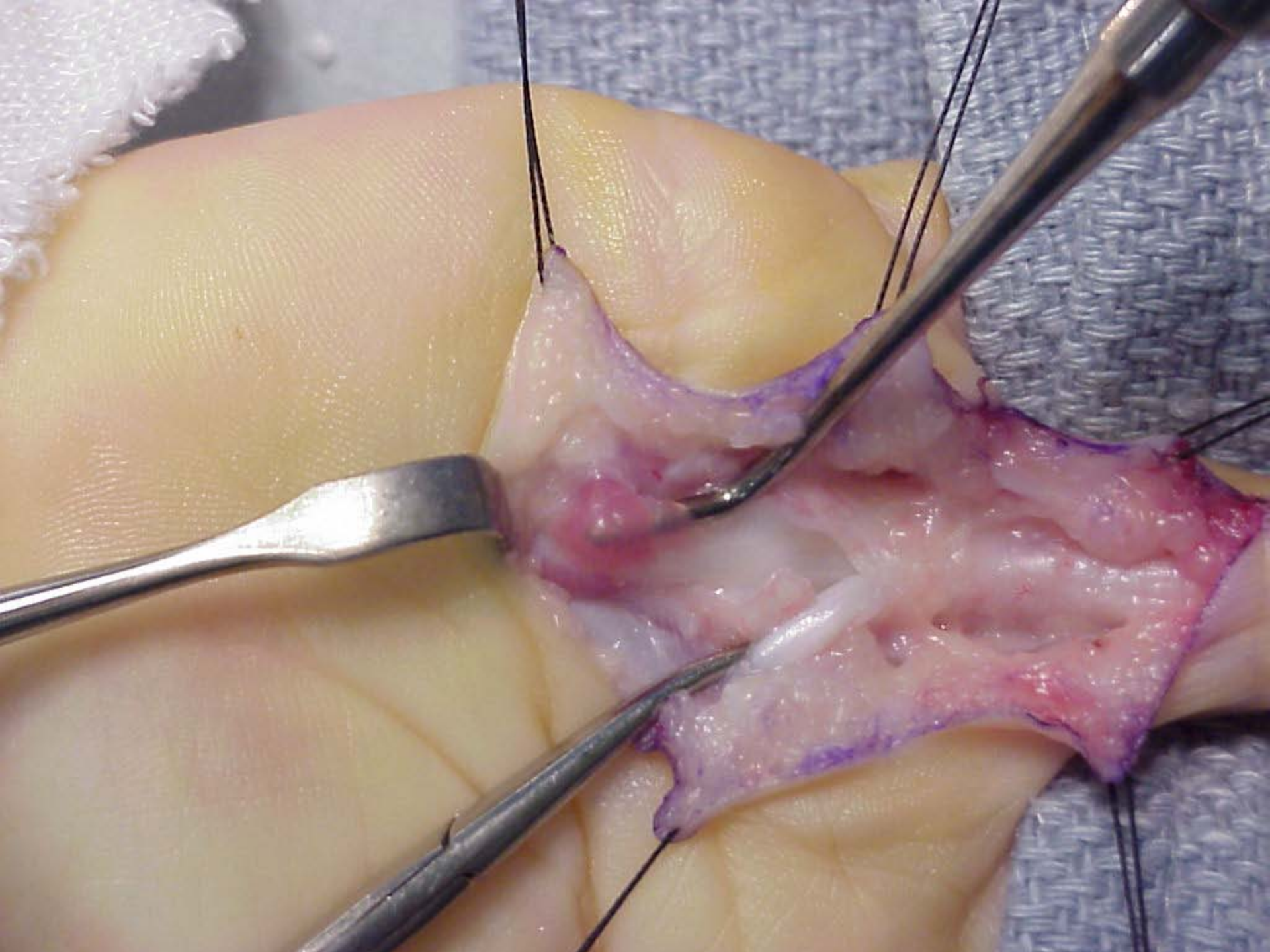
Infantile Trigger Thumb

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



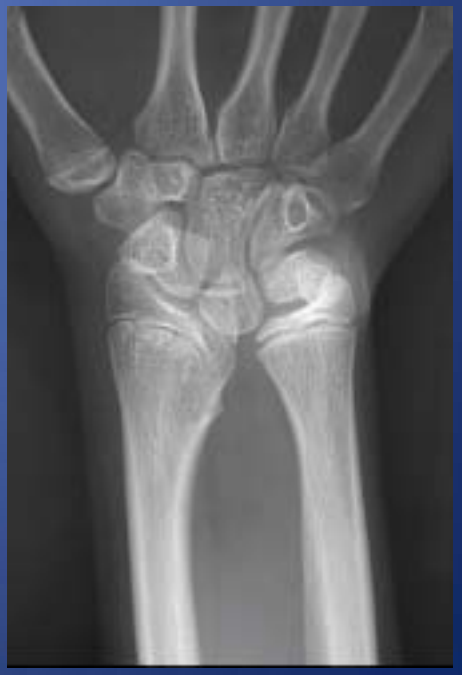
TRIGGER FINGER

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



(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

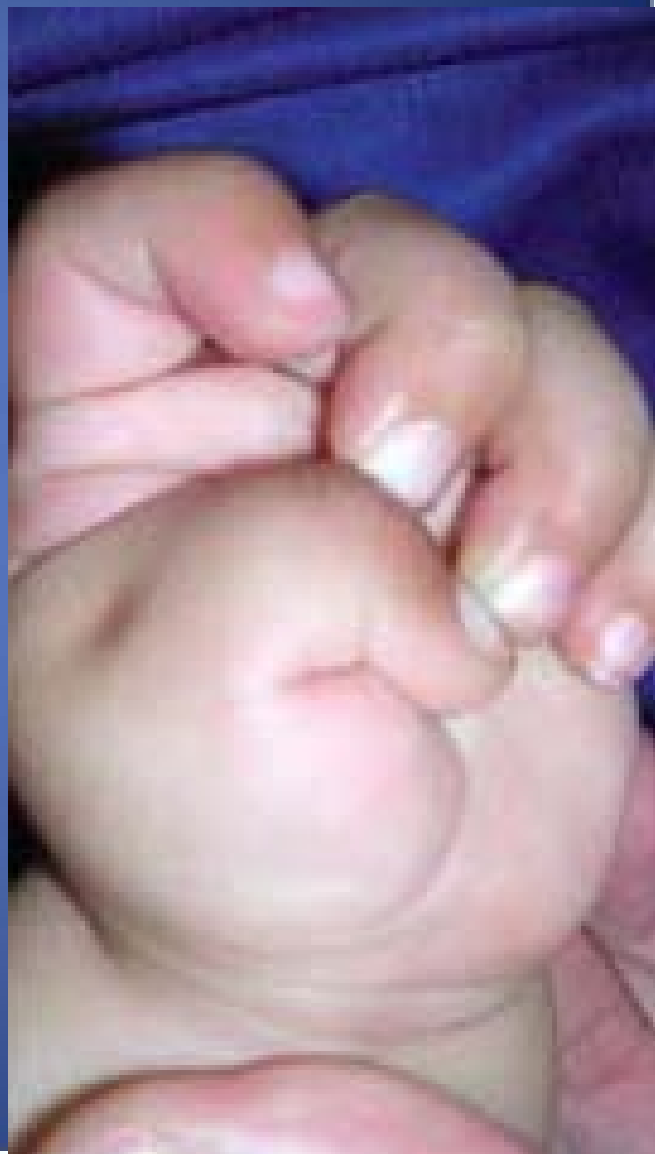


Preferred Response: D

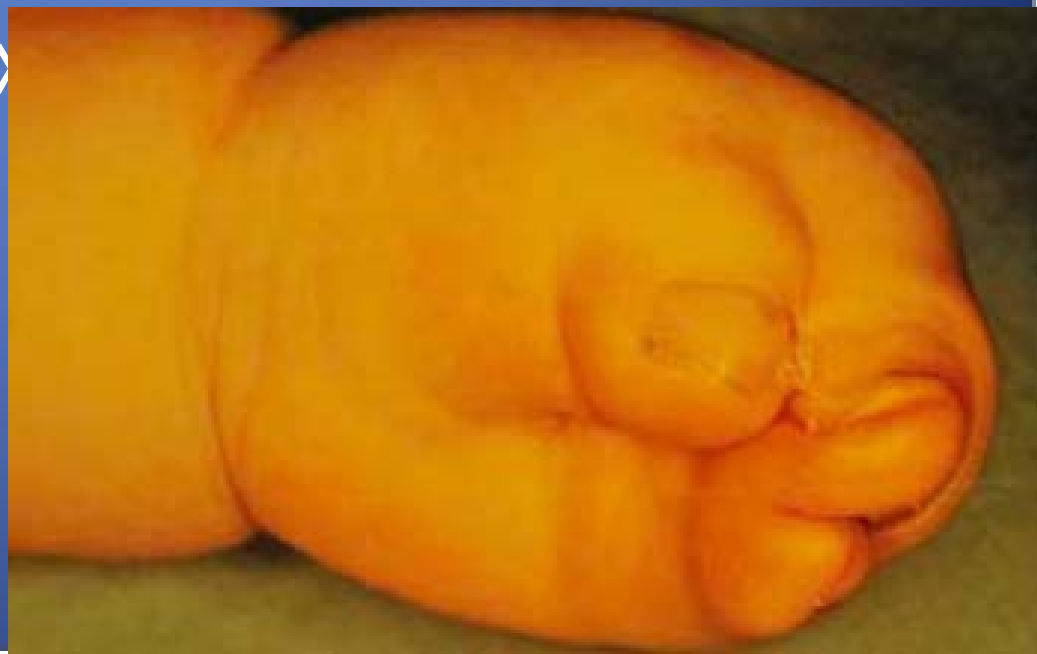
Discussion: Most patients with Madelungs have a dyschondrosteosis, or a combination of the deformity of the distal radius, short forearms, and short stature. Elbow range of motion is rarely affected and carpal tunnel syndrome and extensor tendon abnormalities are uncommon as well.

An 8-month-old child is seen with the thumb deformity in Figure 1. The MCP joint is flexed, but supple, and easily passively extended. The child cannot actively extend the MCP or the IP joint of the thumb. There are no other abnormalities seen in this child. In considering surgery for this deformity, what common abnormality is found?

A. Thickening in the flexor pollicis longus tendon
B. Stenotic A1 pulley of the thumb
C. Absence of the flexor pollicis longus
D. Hypoplastic extensor pollicis brevis
E. Absence of the brachioradialis

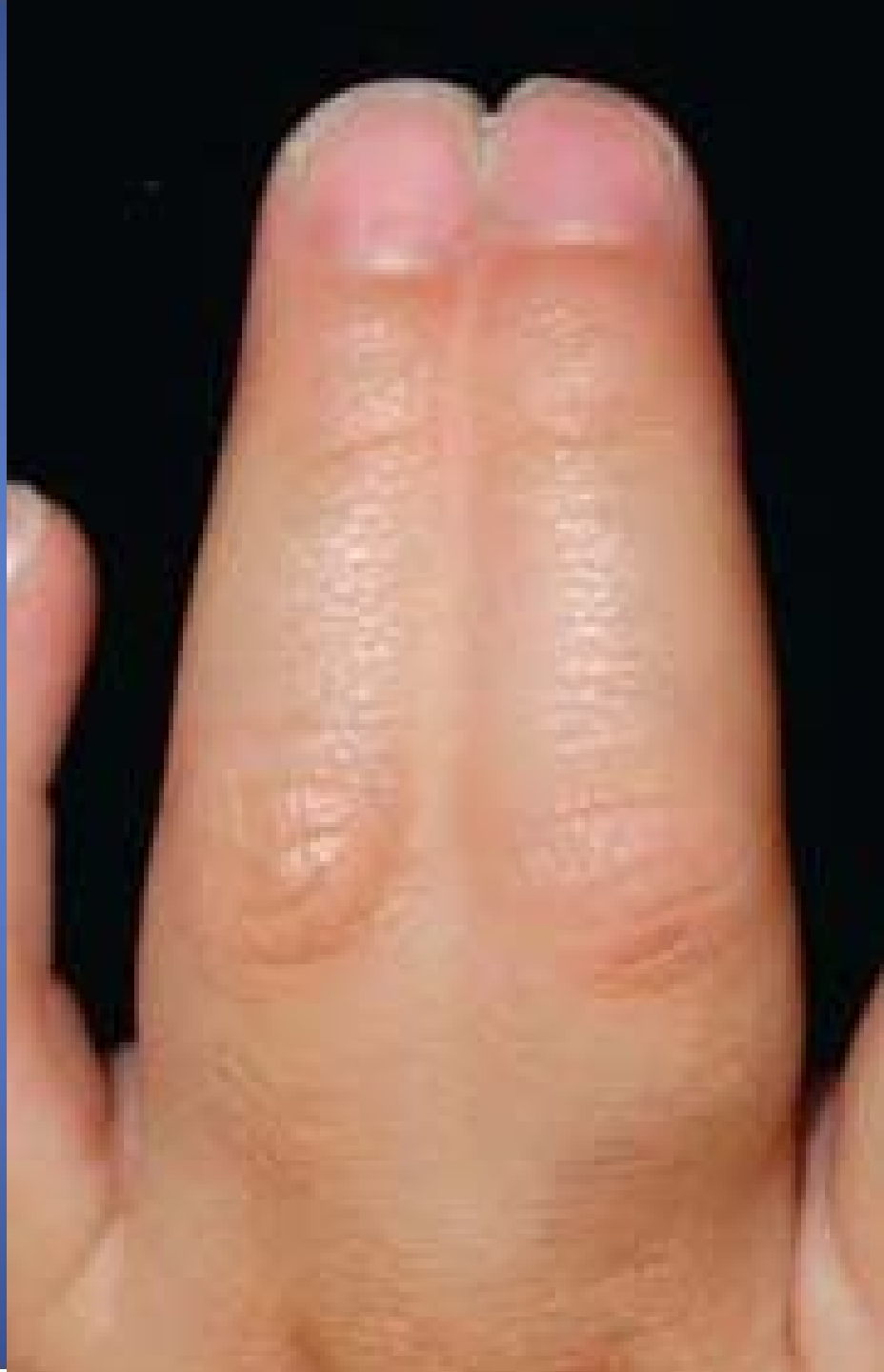


- 71. The infant whose hands are shown in Figure 1
- is expected to have a mutation in which gene?
- A. FGF receptor 2 (FGFR2)
- B. Homeobox D13 (HOXD13)
- C. Holt-Oram syndrome 1 (HOS1)
- D. Human zinc factor protein gene 3 (GLI3)
- E. Brachyury T-box 5 (TBX



74. The abnormality depicted in Figure 1
occurred during embryogenesis between weeks:

A. 1 and 2
B. 3 and 4
C. 5 and 7
D. 8 and 10
E. 11 and 13



100. The hand finding most commonly associated with the condition depicted in Figure 1 is:

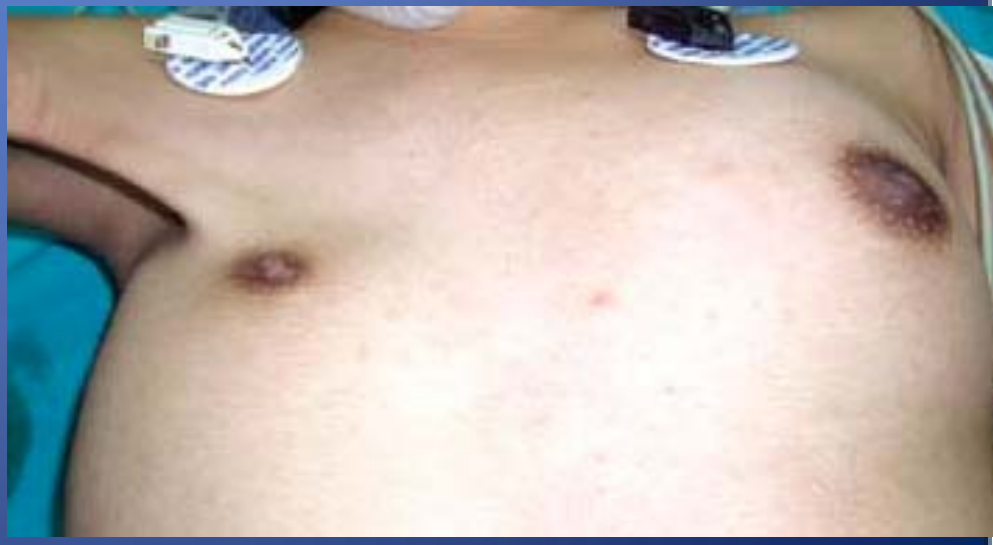
A. Normal hand

B. Hypoplastic hand

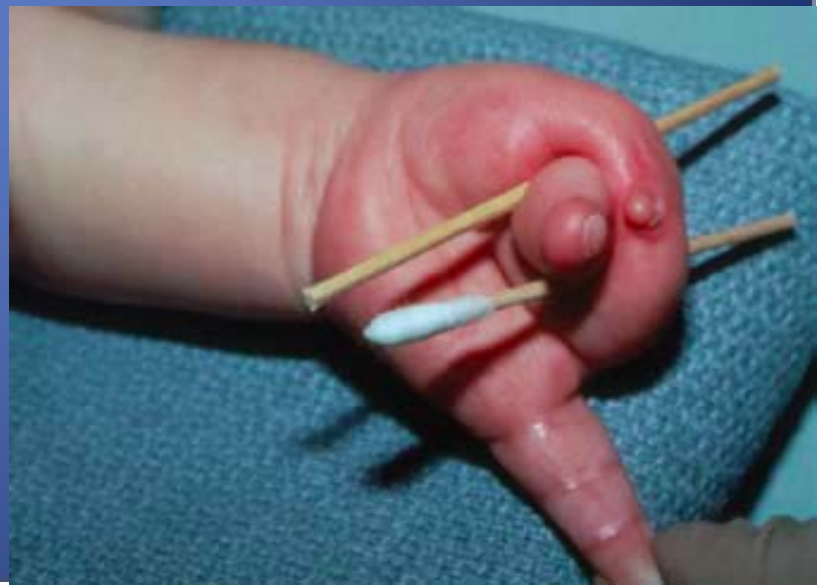
C. Radial club hand

D. Brachysyndactyly

E. Transverse deficiency proximal to the MCP joints

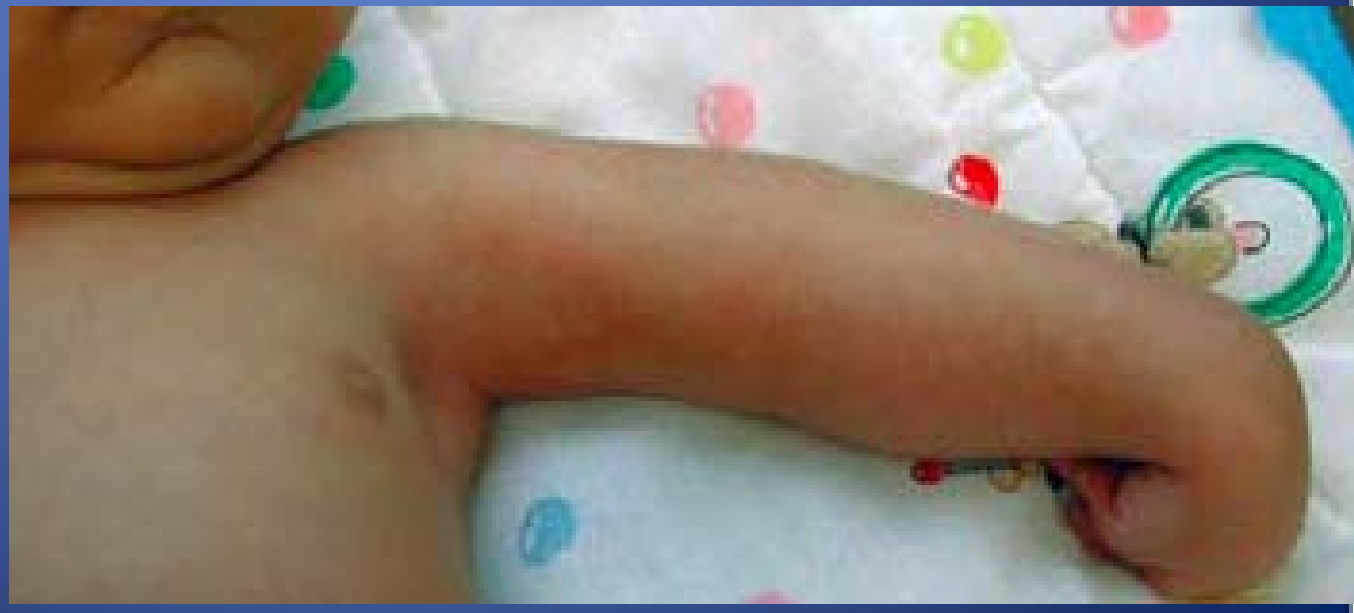


- 21. Acrosyndactyly (Figure 1), or fusion of the terminal portion of two or more digits with proximal sinuses
- between the digits, is seen in:
- A. Constriction band syndrome
- B. Poland's syndrome
- C. Cleft hand
- D. Simple, incomplete syndactyly
- E. Simple, complete syndactyly



66. What is the diagnosis of the infant seen in Figure 1?

A. Congenital radioulnar synostosis
B. Ulnar dimelia
C. Arthrogryposis multiplex congenita
D. Aperts syndrome
E. Congenital contractural arachnodactyly of the elbow



- 84. In hypoplasia of the thumb, what factor would
- favor pollicization?
- A. Absence of thenar muscles
- B. Narrowing of the first web space
- C. Insufficiency of the ulnar collateral ligament of the
- MP joint
- D. Extrinsic tendon abnormalities (flexor and extensor)
- E. Unstable carpometacarpal joint

The patient with the depicted hand deformity (Figures 1, 2 and 3), is more likely to have:

A a craniofacial deformity
B amniotic disruption syndrome
C a cleft hand deformity
D synpolydactyly
E renal abnormalities