PLEASE CLICK ON THE FOLLOWING LINK TO WATCH THE LECTURE ONLINE:-

https://www.youtube.com/watch?v=7rheURiG6Qk&list=PLuBRb5B7fa_fRRpcuUO-I1JFGuAGVF9Qy&index=7

Congenital Anomalies of the hand

Dr. Motaz Qasaimeh

Agenda

- Embryology and Classification of Congenital Upper Limb Anomalies
- Radial Longitudinal Deficiency: Radius Hypoplasia
- Radial Longitudinal Deficiency: Thumb Hypoplasia
- Ulnar Longitudinal Deficiency (ULD)
- Symbrachydactyly
- Cleft Hand
- Camptodactyly
- Clinodactyly
- Congenital Clasped Thumb
  - Radial Polydactyly
  - Ulnar Polydactyly
  - Macrodactyly
  - Amniotic Band Syndrome (ABS)

Embryology and Classification of Congenital Upper Limb Anomalies

•**Developmental Timeline**:

•The upper limb begins to form around the fourth week of human development from the lateral plate mesoderm.

•By the sixth week, the basic shape of the limb is recognisable, and by the eighth week, finer details like fingers start to differentiate.

•**Significant Signaling Centers**:

•**Apical Ectodermal Ridge (AER)**: **Proximal-Distal (P-D) Axis**:

• through the release of growth factors like FGFs.

•**Zone of Polarizing Activity (ZPA)**: **Anterior-Posterior (A-P) Axis**: Known as the radial-ulnar axis,

• through the secretion of Sonic Hedgehog (Shh).

•**Wnt7a and En1**: These molecules regulate **Dorsal-Ventral (D-V) Axis**:

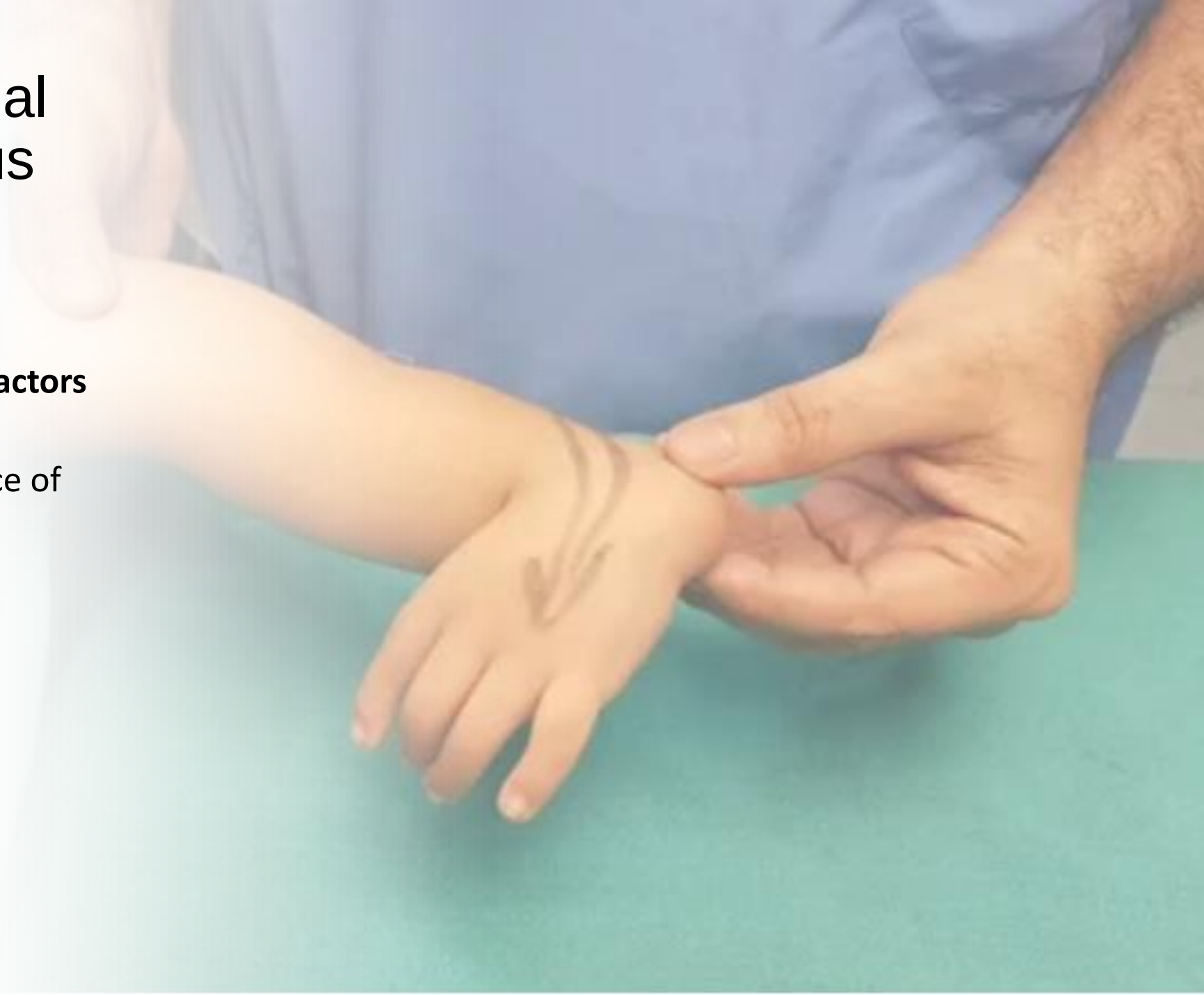
, with Wnt7a promoting dorsal identities and En1 ensuring ventral differentiation.

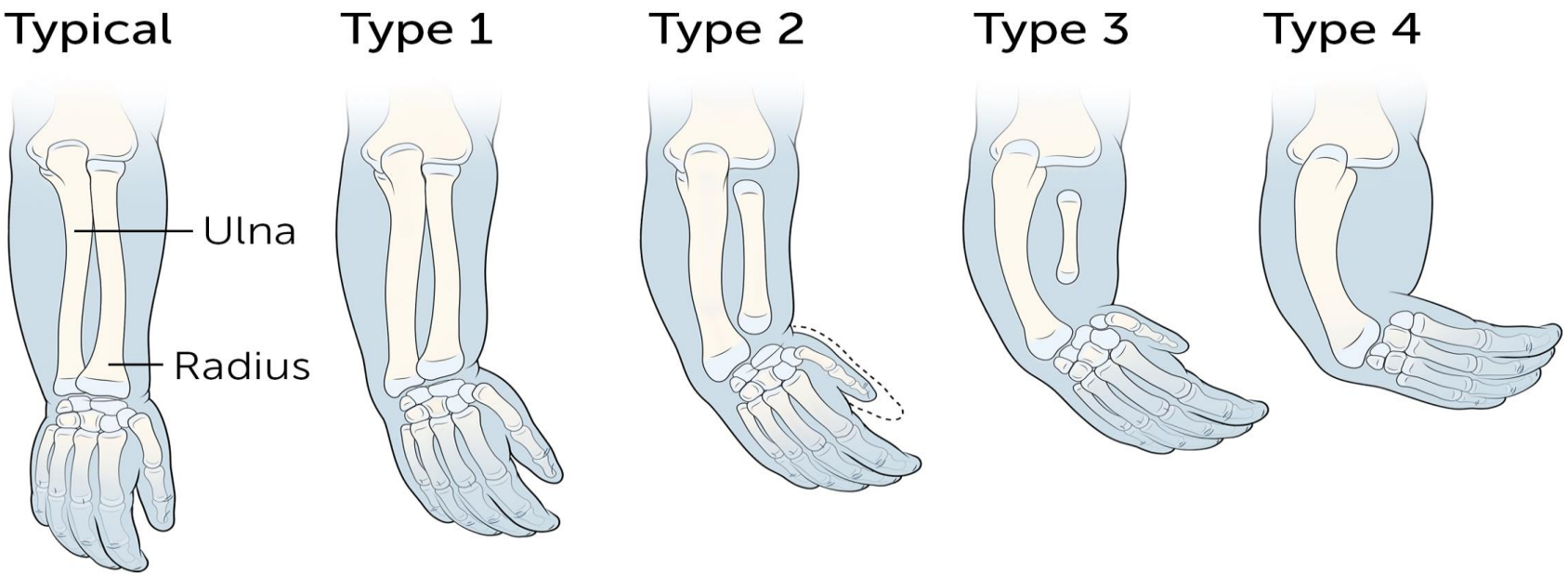
- **Classification of Congenital Upper Limb Anomalies**
- **Malformations**:
  - Result from intrinsic developmental disruptions leading to structural abnormalities (e.g., radial longitudinal deficiency, syndactyly).
  - These are typically not caused by external factors and may involve mutations in developmental genes.
- **Deformations**:
  - Occur due to extrinsic factors affecting a normally developing limb, such as compression or vascular disruption (e.g., constriction by amniotic bands).
  - These can sometimes be corrected after birth if addressed early.
- **Dysplasias**:
  - Involve abnormal tissue development and organization, affecting the growth and functional integrity of the limb (e.g., macrodactyly, which involves excessive growth of the fingers or toes).
- **Overgrowth and Undergrowth**:
  - **Overgrowth Syndromes**: Conditions like Proteus syndrome, where parts of the body grow disproportionately.
  - **Undergrowth**: Conditions like ulnar hypoplasia, where a part of the limb is underdeveloped.

Radial Longitudinal Deficiency: Radius Hypoplasia

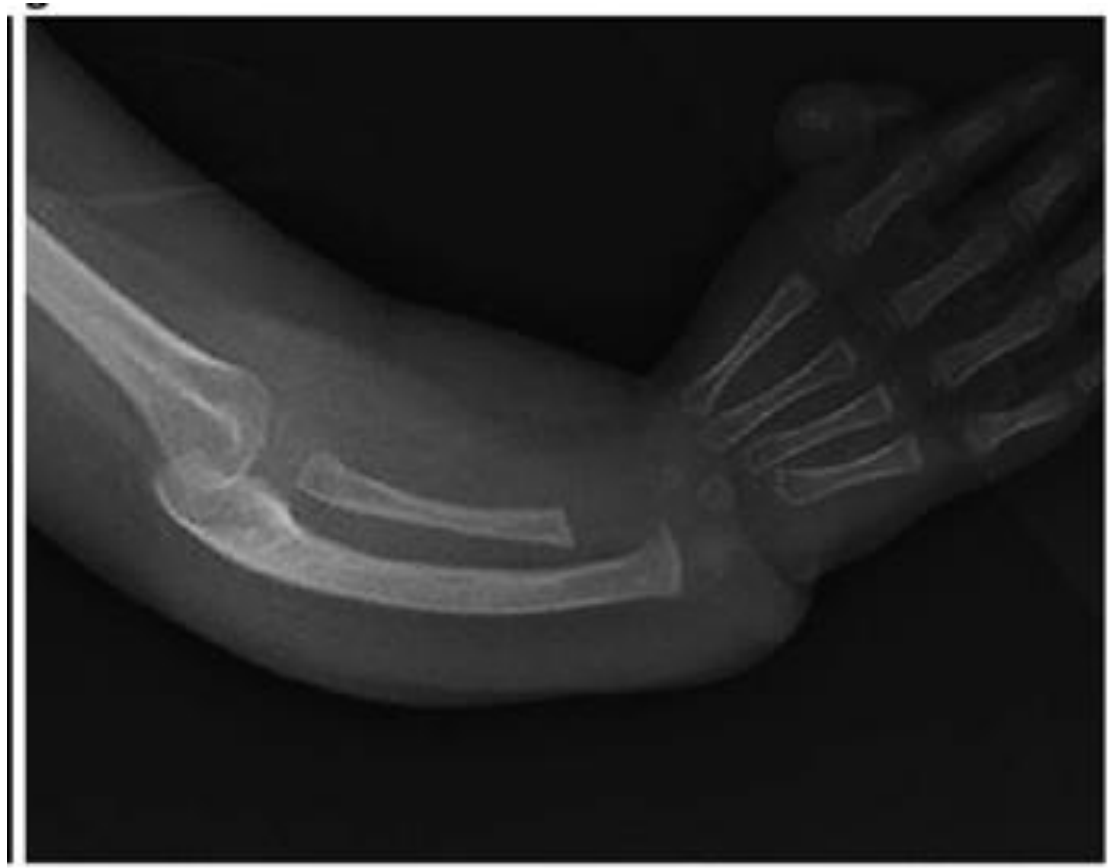
- **Embryology and Genetic Factors**

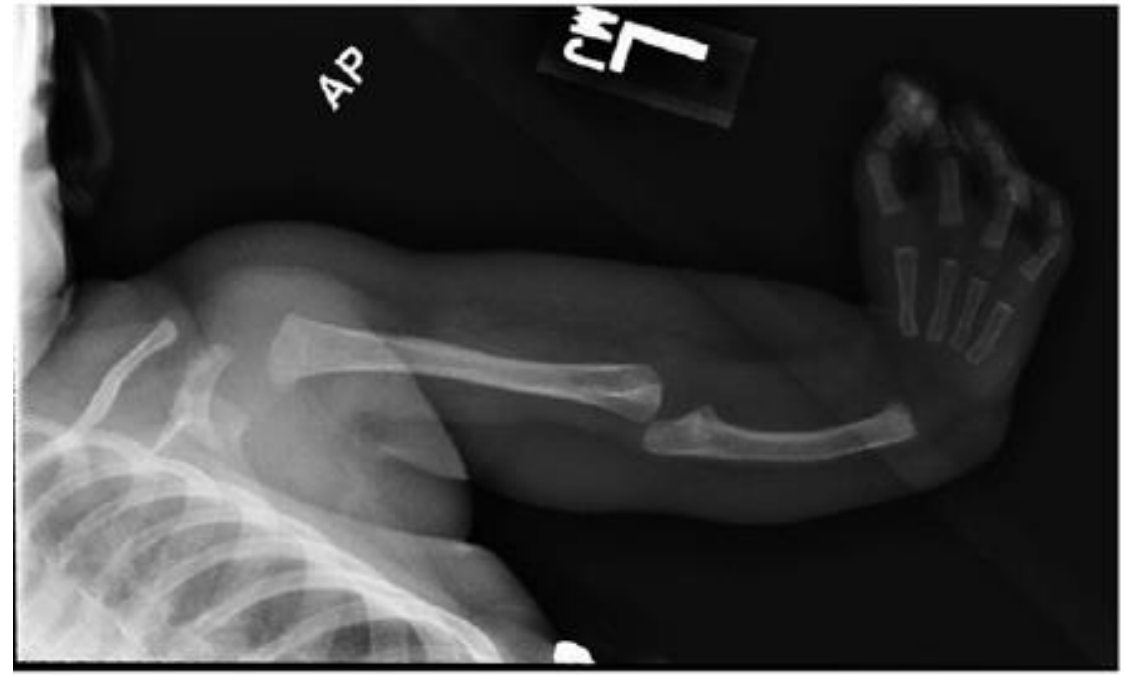
Defined As congenital underdevelopment or absence of the radius





- **Detailed Classification**
- **Bayne and Klug Classification**:
  - **Type I**: Shortening of the radius, preserving wrist and hand function with minimal intervention required.
  - **Type II**: Pronounced bowing of the radius, often needing surgical intervention to prevent further deformity and improve function.
  - **Type III**: Substantial part of the radius is missing, leading to significant functional limitations.
  - **Type IV**: Total absence of the radius; the most severe form, typically requiring multiple surgical procedures for functional correction and aesthetic improvement.

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- **Associated Anomalies and Syndromes**
  - Holt-Oram Syndrome (cardiac and upper limb anomalies)
  - Fanconi Anemia
  - VACTERL Association (vertebral, anal, cardiac, tracheal, esophageal, renal, and limb anomalies)
  - Thrombocytopenia-absent radius (TAR) syndrome

- **Comprehensive Management Strategies**
- **Conservative Treatments**:
- **observation**
- indicated if absent elbow motion or biceps deficiency
- hand deformity allows for extra reach to mouth in presence of a stiff elbow

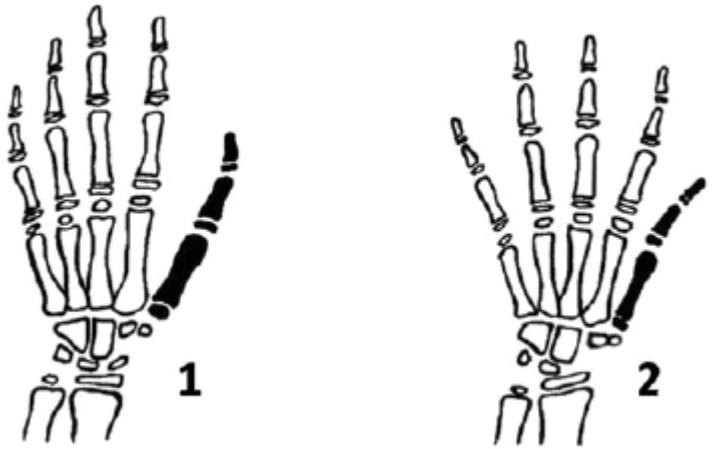
  - **Physical Therapy**: Focuses on maintaining the range of motion and stretching, strengthening the limb.
  - **Orthotic Devices**: Custom splints or braces to support proper limb positioning and prevent worsening of the deformity.

- splints or braces to support proper limb positioning and prevent worsening of the deformity.
- **Surgical Approaches**:
  - **Centralization or Radialization**: Procedures to realign the hand over the ulna, improving functional use of the hand.
  - **Ulnar Osteotomy**: Corrects deformities of the ulna when significant bowing is present.
  - **Reconstructive Surgeries**: Aim to improve the functionality and appearance of the hand and thumb, including thumb reconstruction or pollicization when necessary.
- **Follow-Up and Prognosis**:
  - **Regular Monitoring**: Essential to adjust treatments as the child grows and develops.
  - **Prognosis**: Varies based on the type of RLD and the timeliness and effectiveness of the intervention. Early and tailored treatments can significantly enhance outcomes.

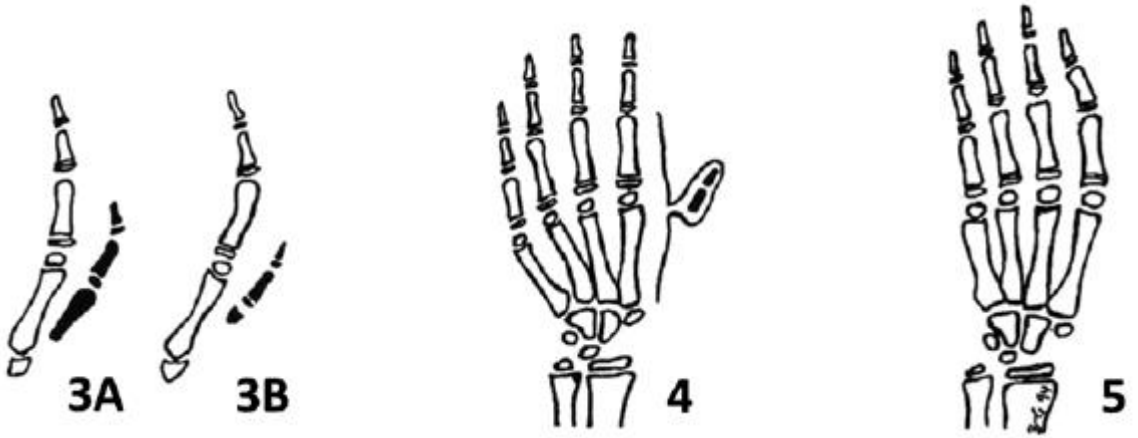
Radial Longitudinal Deficiency: Thumb Hypoplasia

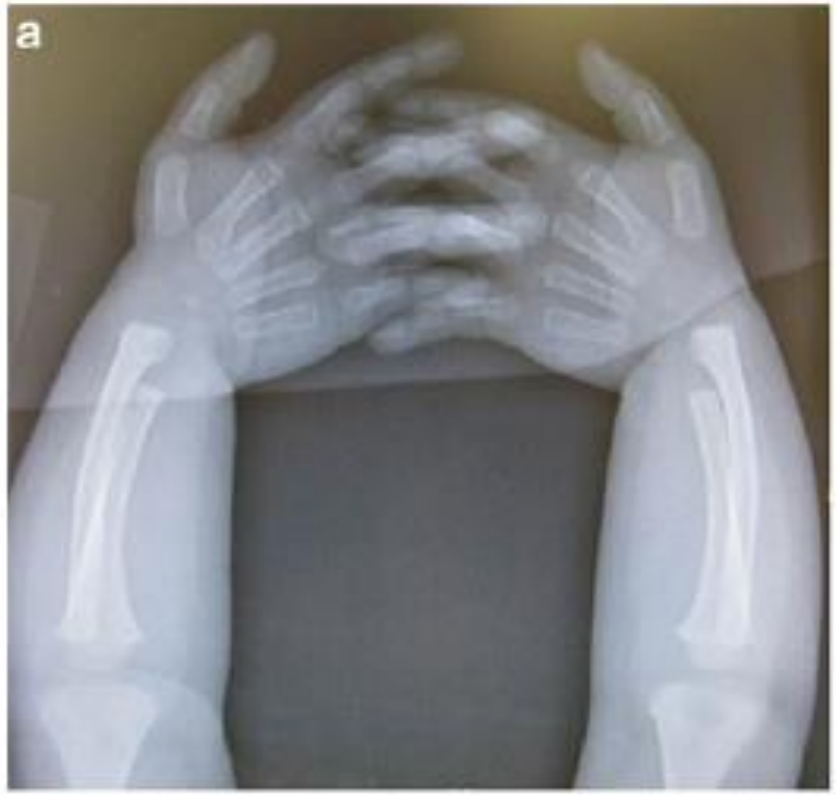
- **Embryology**
- **Development**: Thumb Hypoplasia is commonly associated with Radial Longitudinal Deficiency (RLD), manifesting early in limb development during the 4th to 7th weeks of gestation.
- **Genetic Influence**: It often involves disruptions in critical signaling pathways that regulate limb outgrowth, notably the Sonic Hedgehog (Shh) pathway and Fibroblast Growth Factors (Fgfs).

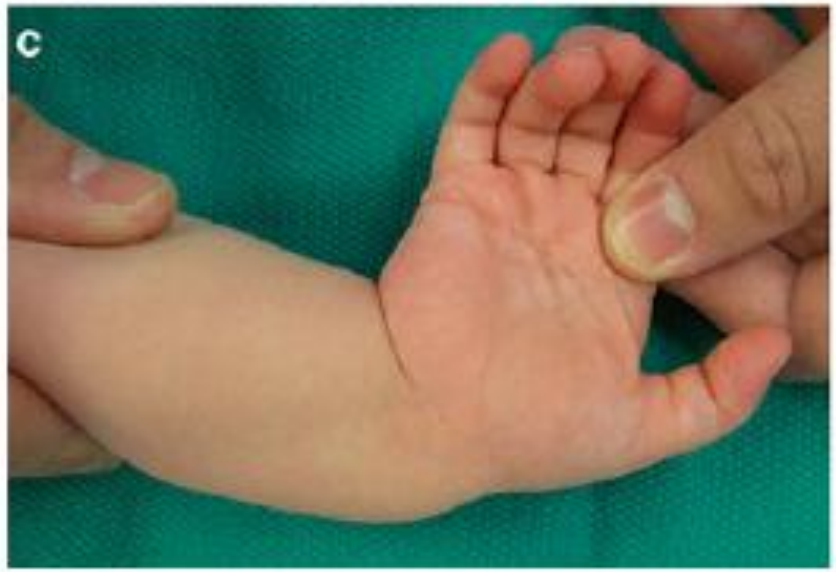
- **Classification (Blauth Classification modified by Manske)**
- **Grade 1**: Slight hypoplasia of the thenar musculature; stable and functional joints; typically, no surgery required.
- **Grade 2**:
  - **2**: instability of the MCP joint; atrophied thenar muscles, may require minor surgical interventions such as first web release or MCP joint stabilization and opponensplasty



- **Grade 3**:
  - **3A**: absence of instability of the CMC joint (stable CMC joint)
  - **3B**: Absence of the CMC joint (unstable CMC joint); typically addressed through pollicization (index finger converted to function as a thumb).
- **Grade 4**: Absence of the thumb metacarpal; pollicization is recommended.
- **Grade 5**: Complete absence of the thumb; options include pollicization or toe-to-hand transfer for creating a functional thumb.



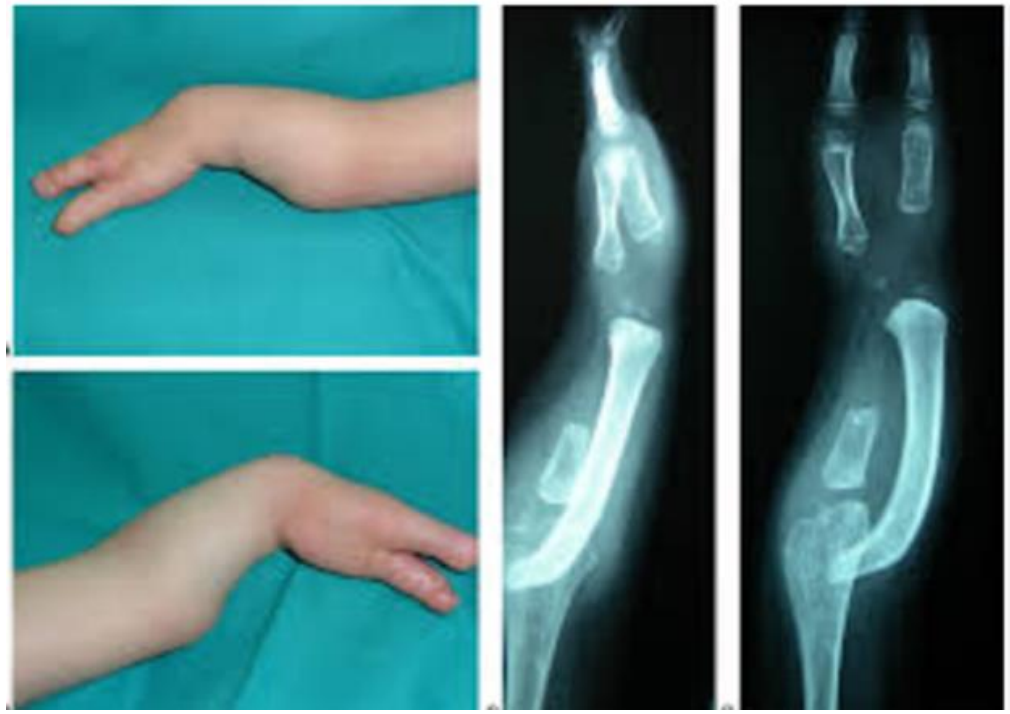




Ulnar Longitudinal Deficiency (ULD)

- **Definition and Incidence**
- **Ulnar Longitudinal Deficiency (ULD)**, also known as ulnar dysplasia, is a rare congenital disorder primarily affecting the ulnar side of the forearm and hand. It involves underdevelopment or absence of ulnar-sided structures, impacting both skeletal and soft tissues.
- **Incidence**: Occurs in approximately 1 in 25,000 live births, usually presenting unilaterally and sporadically without a clear inheritance patternbryology

- **Development**: ULD arises from disruptions in the signaling pathways responsible for the development of ulnar-sided forearm structures, notably the Sonic Hedgehog (Shh) signaling which plays a key role in the differentiation of ulnar-sided digits and bones .

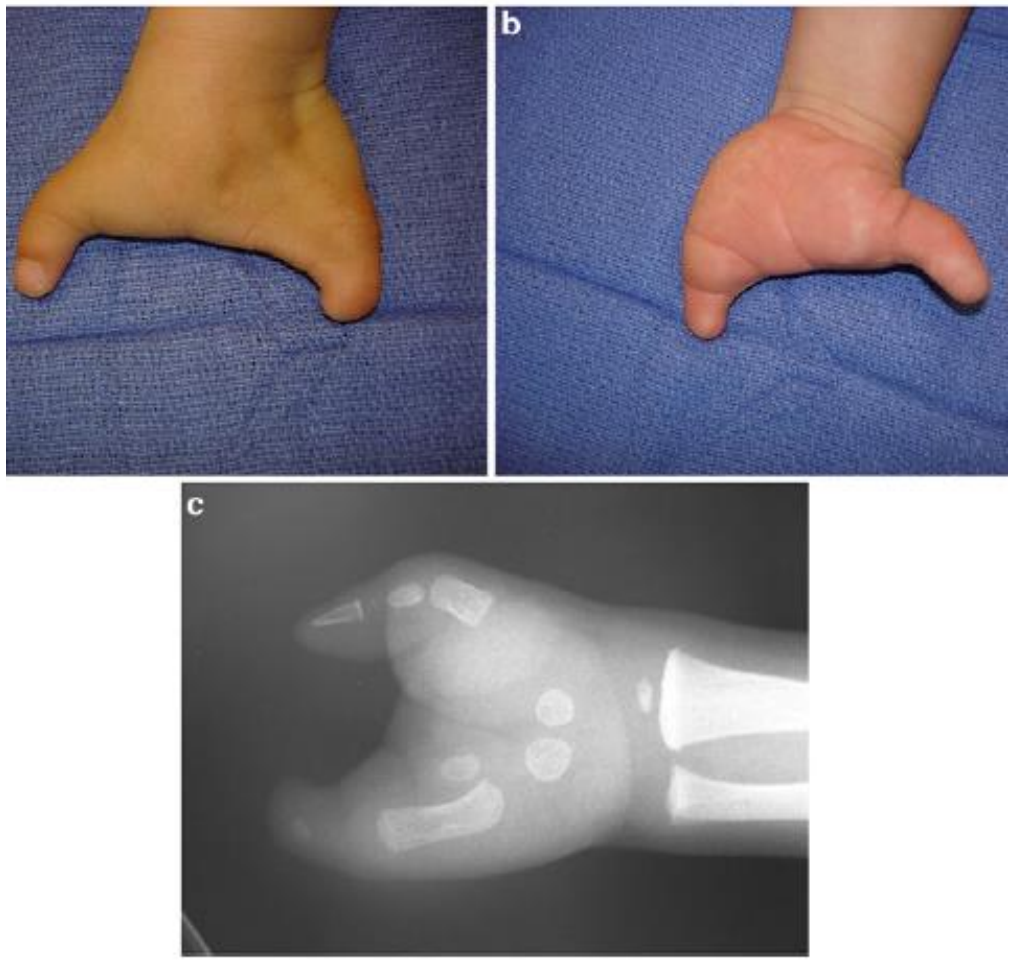


- **Classification**
- **Bayne Classification Modified**:
  - **Type I**: Normal length ulna with ulnar-sided hand anomalies.
  - **Type II**: Hypoplasia of the ulna with an intact proximal and distal ulnar epiphysis.
  - **Type III**: Partial aplasia of the ulna, missing distal or middle third.
  - **Type IV**: Total aplasia of the ulna.
  - **Type V**: Complete absence of the ulna with radiohumeral synostosis, indicating severe deformities .

- **Management Strategies**
- **Non-operative Management**: Includes early intervention with stretching and splinting to maintain joint mobility. Splints may be used to align the wrist and improve hand function in cases of mild deformity.
- **Surgical Procedures**: Focus on hand reconstruction and correction of associated ulnar deficiencies, including:
  - **Syndactyly Release**: Separation of fused fingers.
  - **Thumb Reconstruction**: Enhancing thumb functionality through procedures like opponensplasty and pollicization, especially in cases of thumb absence.
  - **First Web Space Deepening**: Critical for enhancing thumb movement and opposition.
  - **Forearm and Wrist Surgery**: May involve excision of the ulnar anlage, radial osteotomy, or creation of a one-bone forearm to improve stability and alignment .

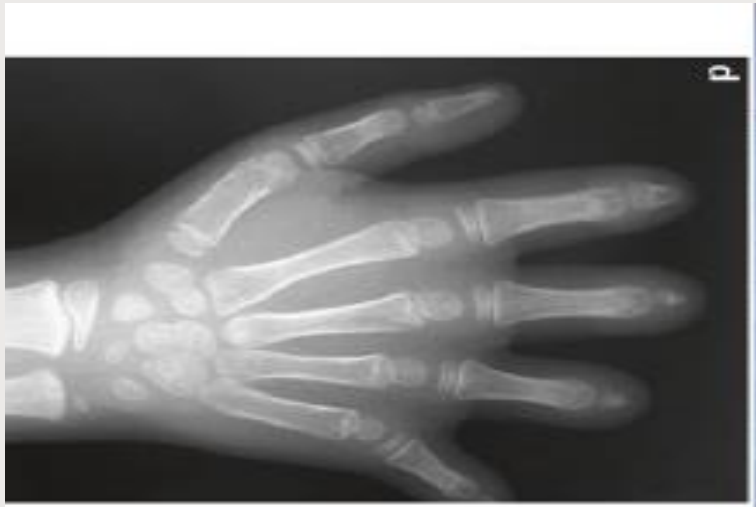
Symbrachydactyly

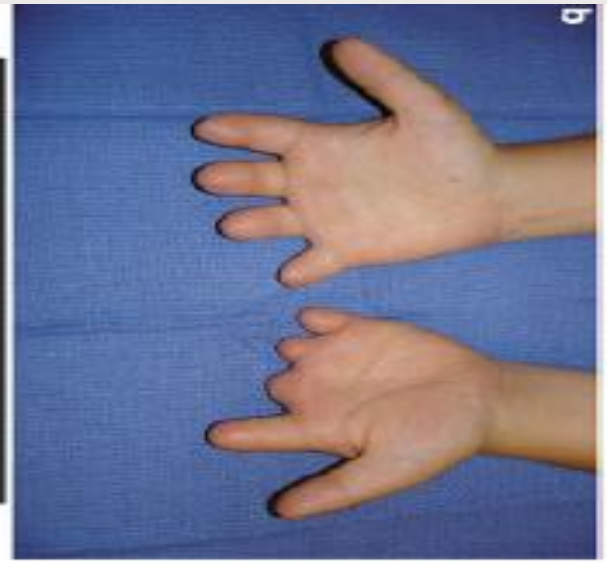
- **Symbrachydactyly** is a congenital anomaly affecting the hands, characterized by shortened or absent fingers and sometimes the fusion of digits (syndactyly).
- This condition results from transverse deficiencies in the development of the hand, often presenting unilaterally.



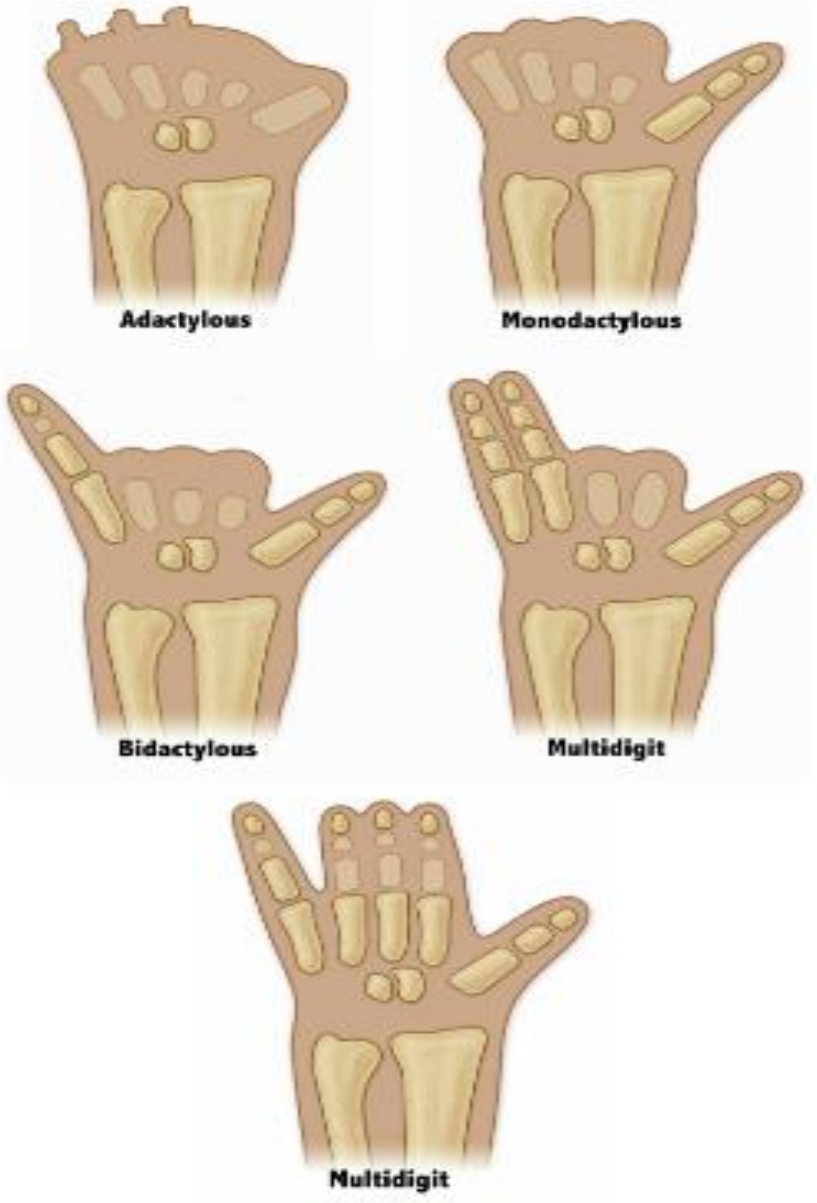
- **Etiology**
- The exact cause of symbrachydactyly is not fully understood. It is believed to result from defects in mesodermal development during limb formation.
- **Vascular disruptions**, such as those affecting the subclavian artery, have been linked to this condition, like other syndromes such as Poland’s syndrome and Klippel-Feil syndrome.
- There is no clear hereditary pattern for symbrachydactyly, and no known animal model exists.

- **Clinical Features**
- **Shortened digits**: Fingers may be partially formed, often with the middle phalanges missing.
- **Syndactyly**: Fusion of digits is a common feature.
- **Nubbins**: In severe cases, fingers may be replaced with small soft tissue projections called "nubbins."
- **Hypoplasia**: The hand may be underdeveloped, with defects sometimes extending into the forearm.
- **Tendon defects**: Extensor tendons are often normal, but flexor tendons may form amorphous masses in the carpal tunnel.



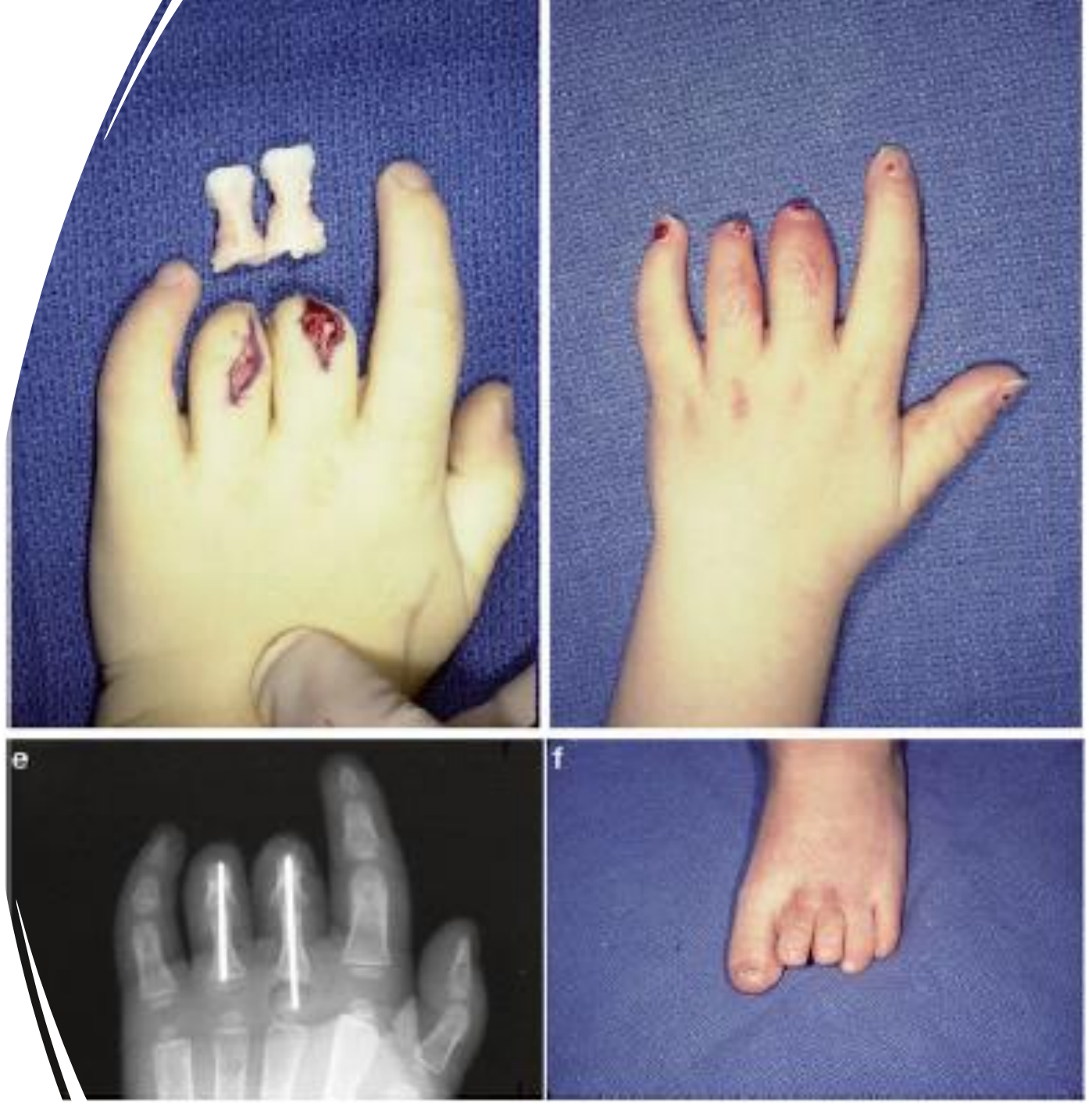


- **Classification**
- **Blauth and Gekeler Classification** (1971):
  - **Type I** (Brachymesophalangia): Shortened fingers with missing middle phalanges, often associated with incomplete syndactyly.
  - **Type II** (Oligodactyly): Absence of some or all of the central three digits, sometimes referred to as an “atypical cleft hand.”
  - **Type III** (Monodactyly): Presence of only the thumb with all four other fingers absent.
  - **Type IV** (Peromelia/Adactyly): Complete absence of all digits, sometimes with small nubbins representing finger remnants.

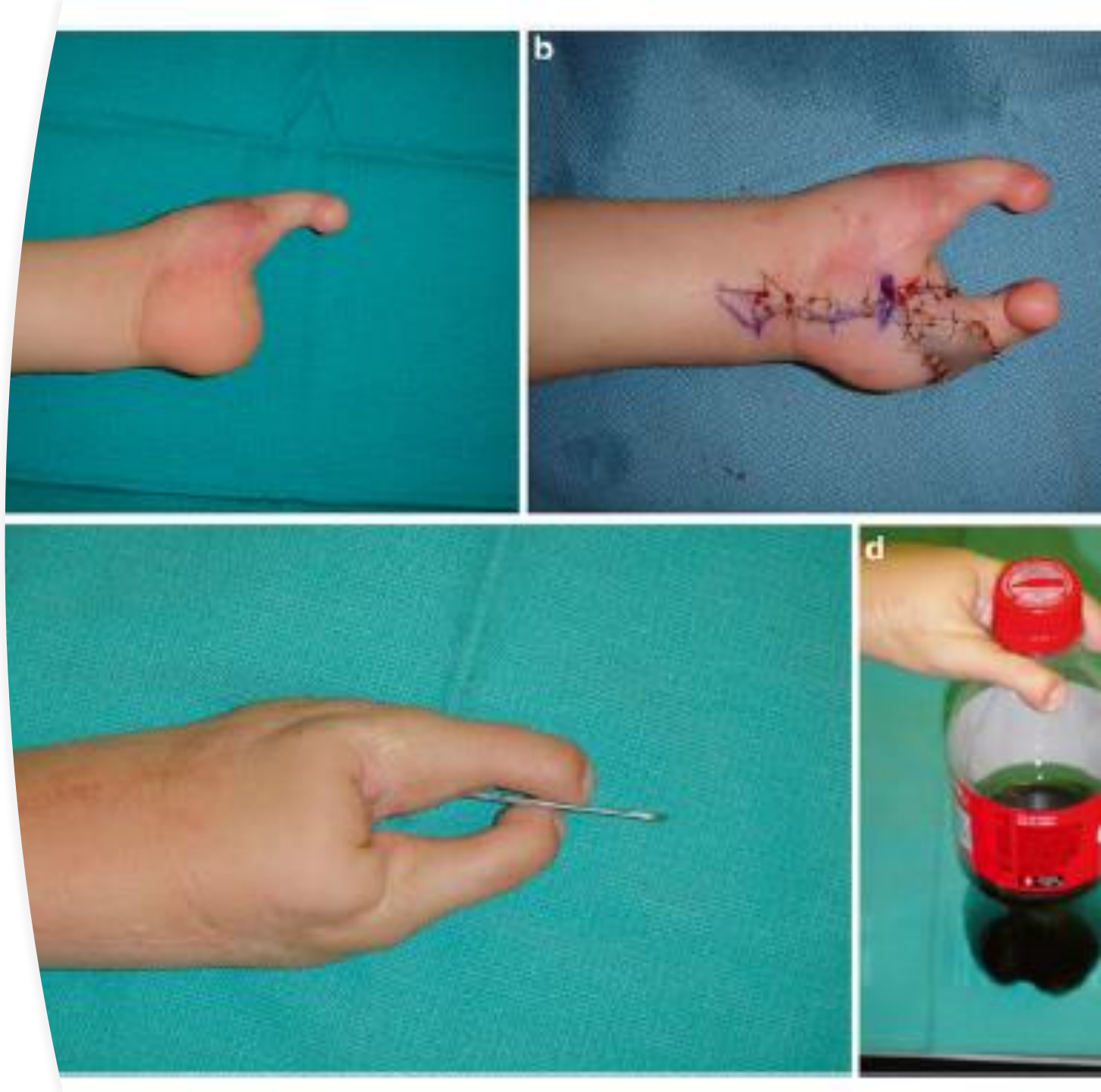


Surgical Treatment

- **Non-vascularized Toe Phalangeal Bone Grafts**: These are used to treat shortened digits by transferring phalanges from the toes.



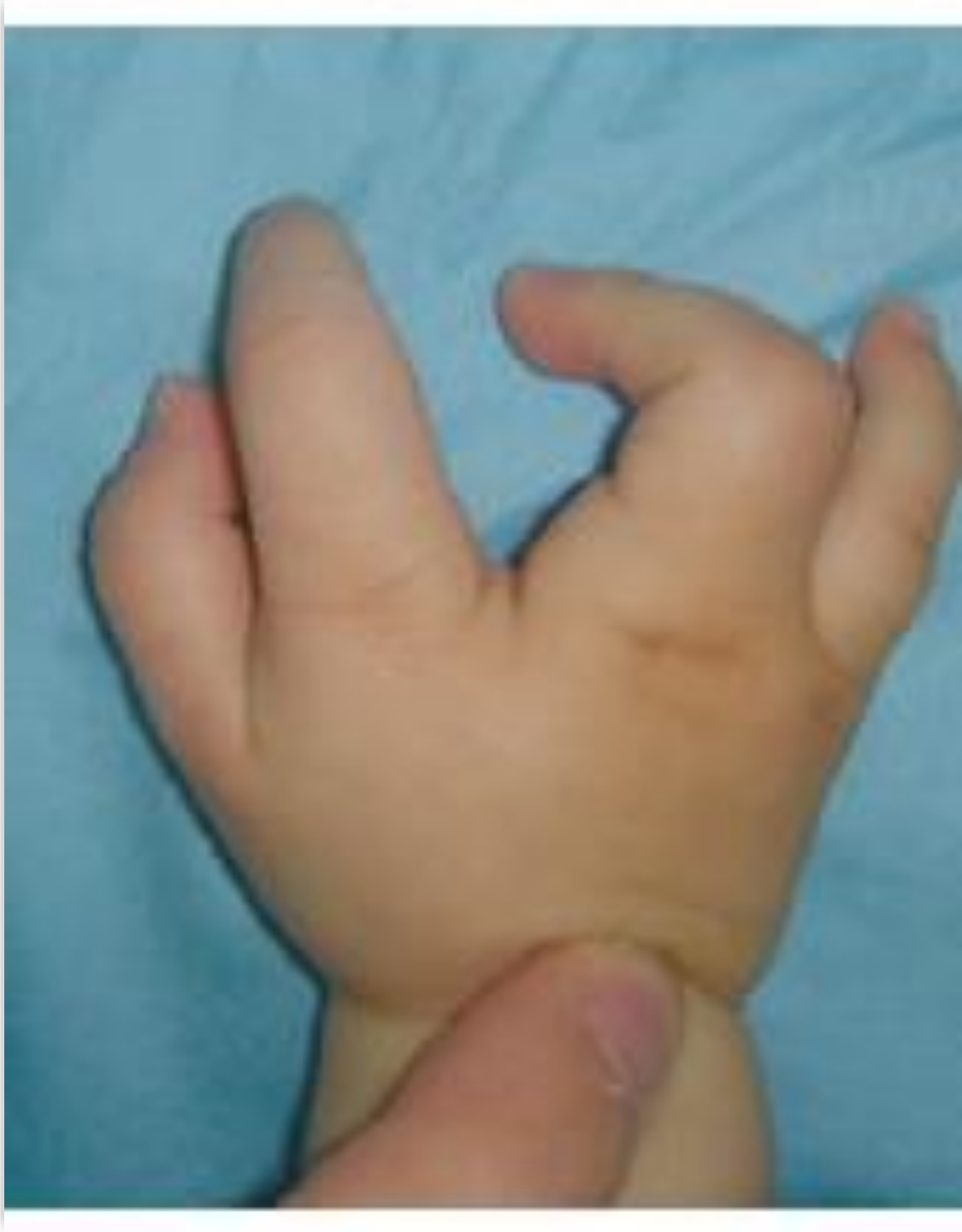
- **Microsurgical Toe-to-Hand Transfers**: Can be used for more severe cases where multiple digits are absent. This procedure involves transferring one or more toes to the hand to reconstruct missing fingers.



- **Distraction Osteogenesis**: A surgical procedure used to lengthen bones in cases of severe brachydactyly, where the digits are abnormally short.

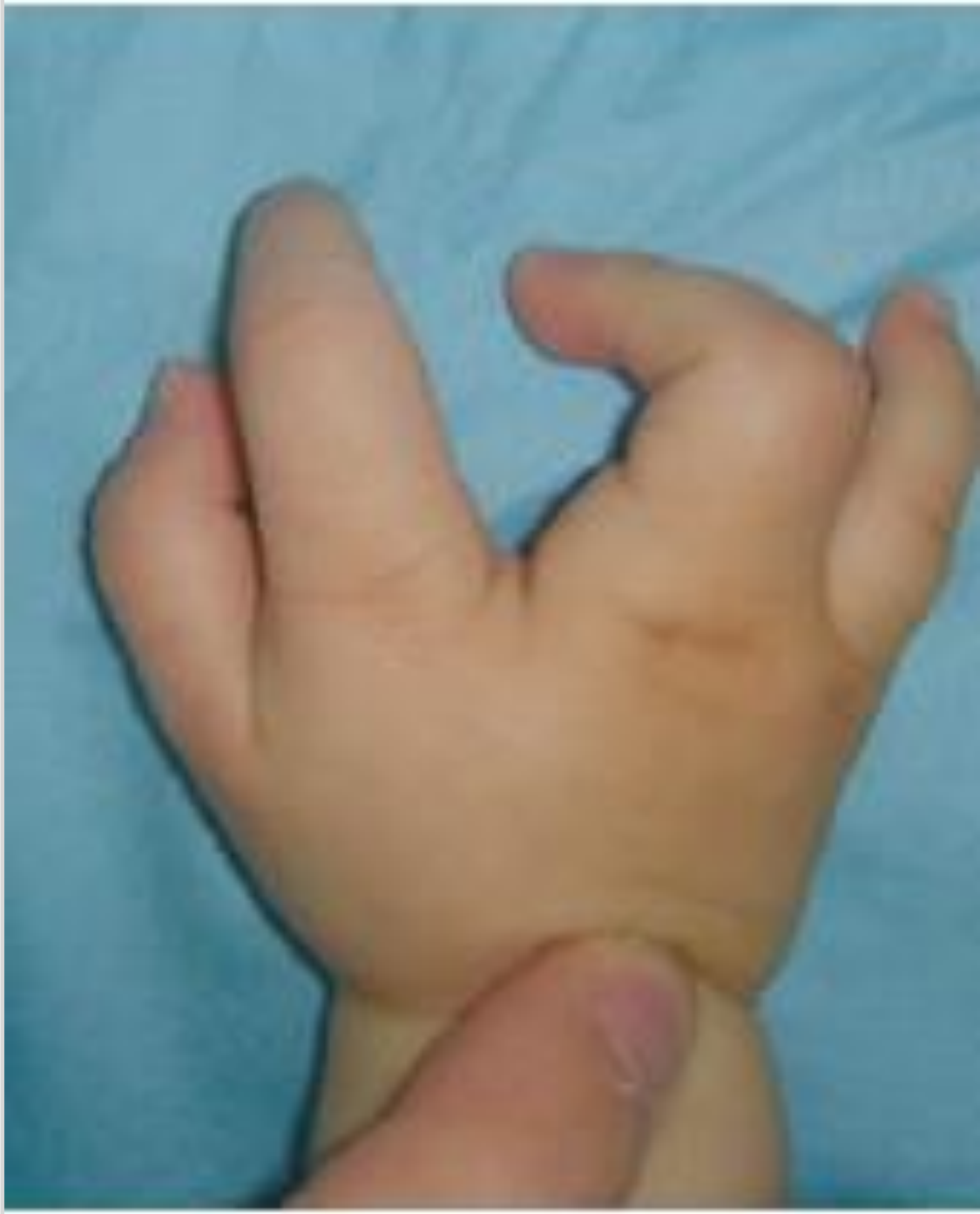
Cleft Hand

- **Presentation**: Cleft hand is characterized by the absence of one or more central digits, often leading to a V-shaped cleft.
- The condition is most commonly missing the middle fingers and may include syndactyly of the remaining digits, contributing to significant functional and cosmetic concerns.

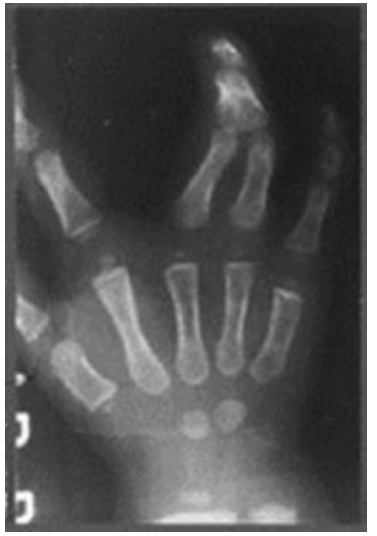


Cleft Hand

- **Etiology**
- **Genetic Factors**: The development of cleft hand can occur sporadically or be inherited in an autosomal dominant pattern. This congenital anomaly shows variable expressivity and incomplete penetrance, highlighting the complexity of its genetic basis.

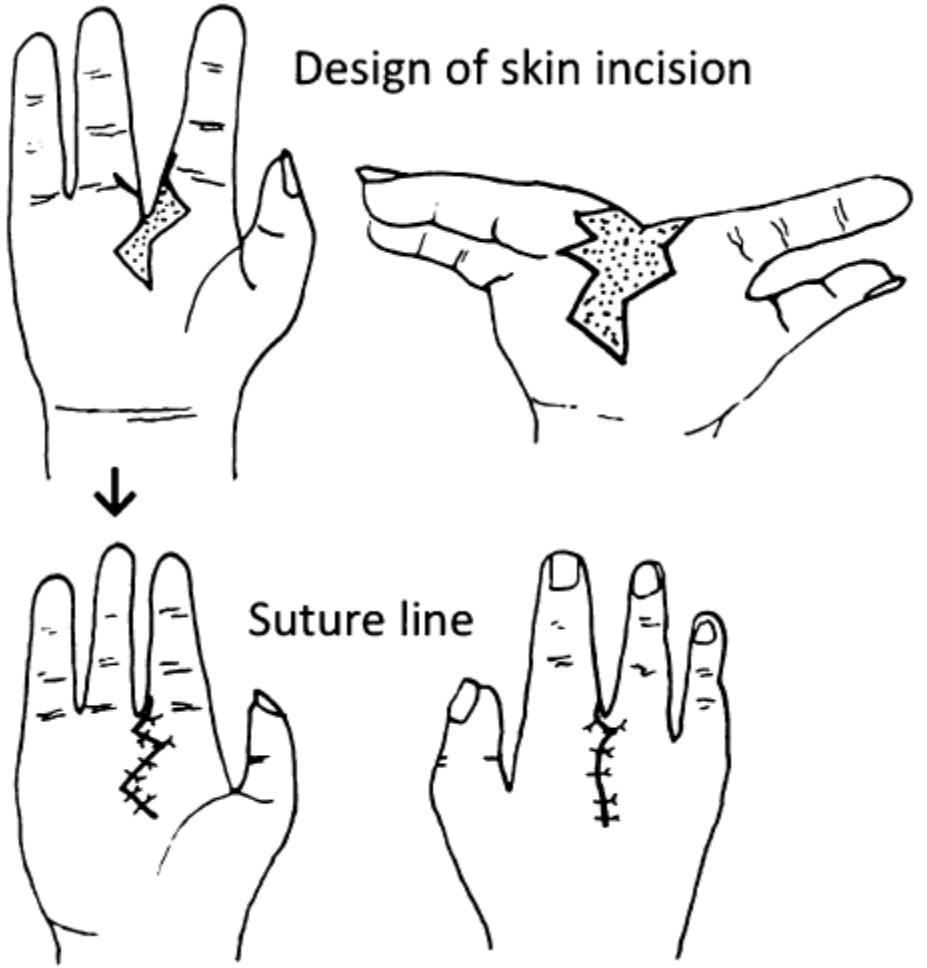


- Atypical cleft hand is best classified as transverse deficiency or symbrachydactyly.
- In longitudinal deficiency, the congenital absence of digits is confined to the long axis of the upper limb and is classed as ulnar deficiency, radial deficiency, or central deficiency (cleft
- hand)
- There is a theory that supports the concept that a common etiological mechanism of cleft hand
- is the combination of the development of central polydactyly and syndactyly.



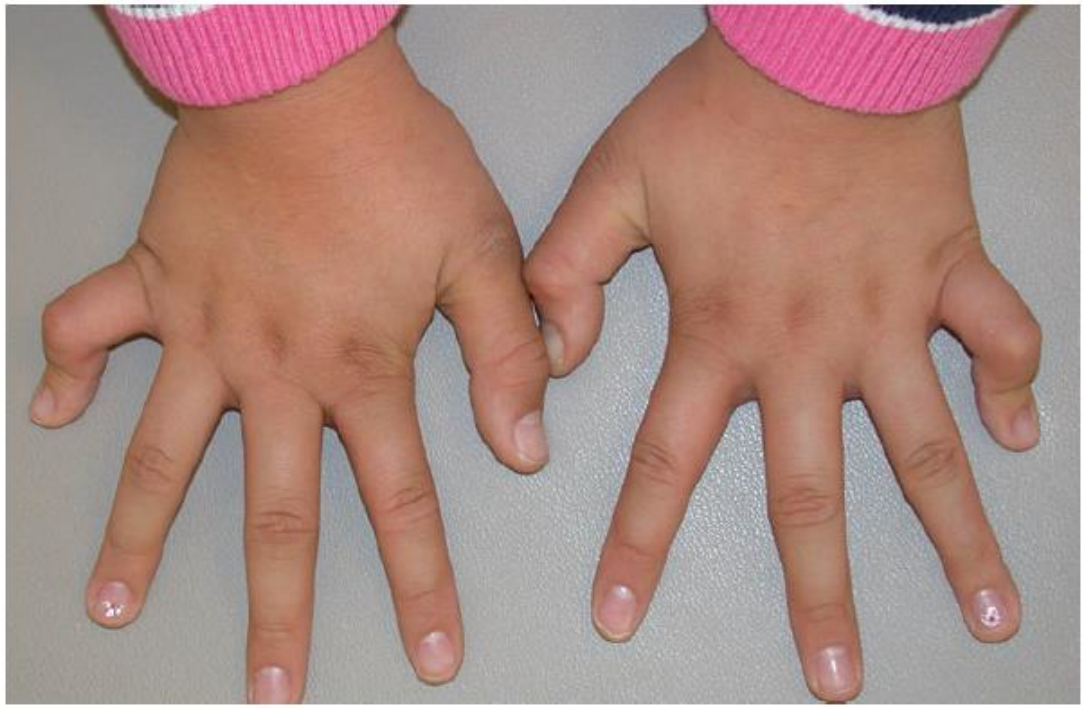
- **Surgical Management**
- **Reconstructive Techniques**: Aim to close the cleft and restore a more normal appearance and functionality. Procedures may include:
  - **Local Flaps**
  - **Z-Plasty**
  - **Direct Closure Techniques**

  - These surgeries are tailored to each patient, taking into account the unique anatomical challenges presented by the cleft.



Camptodactyly

- **Camptodactyly** is characterized by a progressive flexion deformity of the proximal interphalangeal (PIP) joint, primarily affecting the ulnar digits, particularly the little finger.



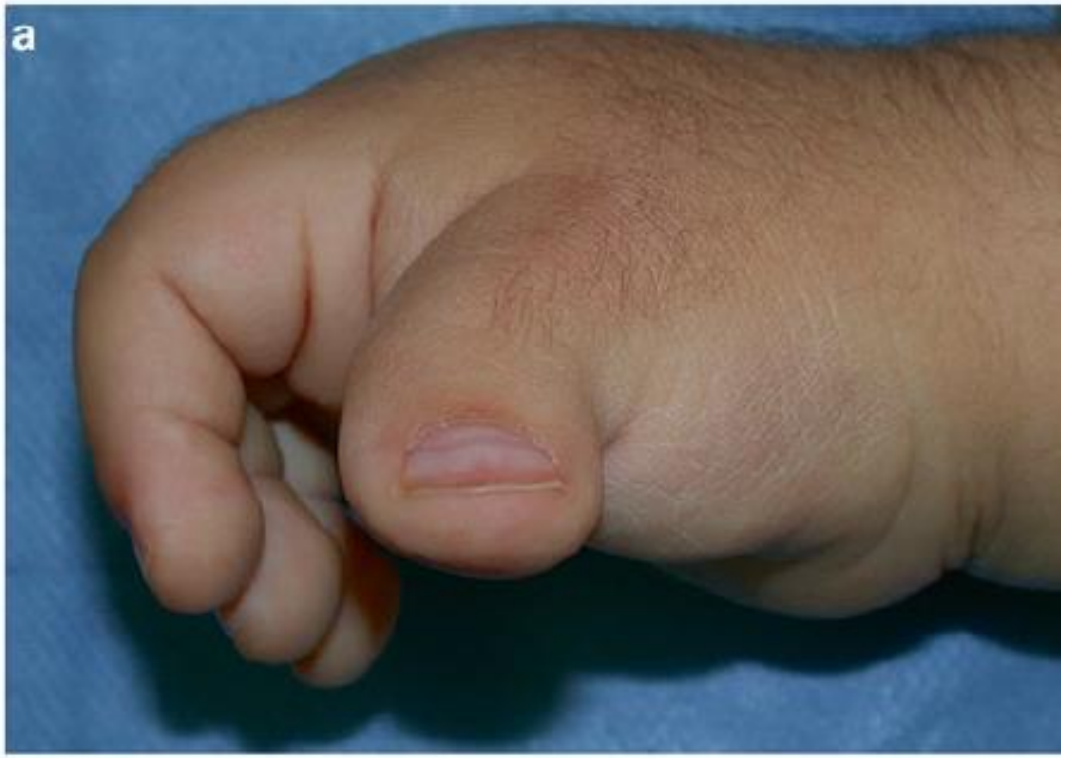
- **Etiology**
- The exact cause of camptodactyly is unknown, but it is often associated with abnormalities in the flexor digitorum superficialis (FDS) tendon and lumbrical muscle abnormalities, leading to an imbalance between flexor and extensor forces.
- **Pathology**
- Abnormalities around the PIP joint, including a contracted or underdeveloped FDS tendon, lead to a flexion deformity which can result in secondary bony changes over time.

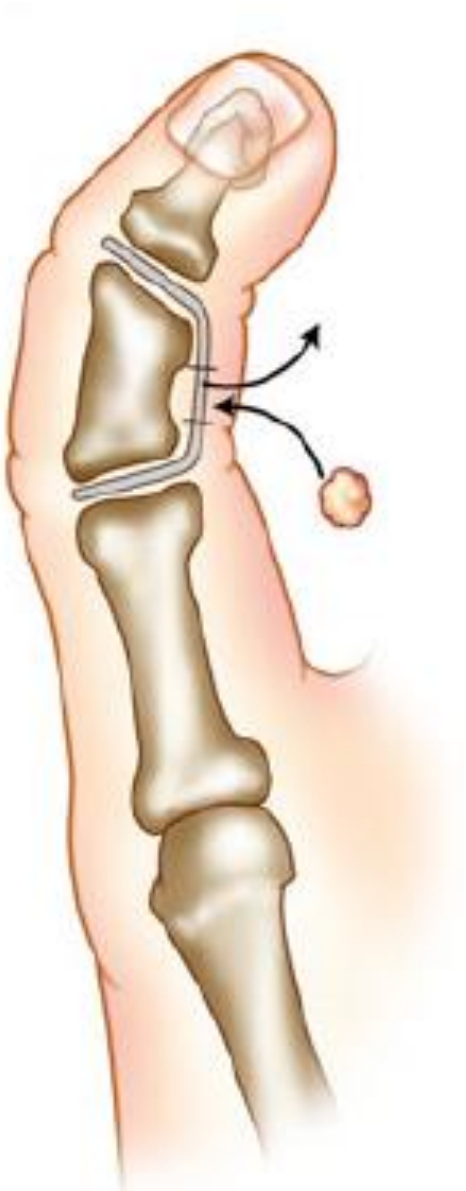
Camptodactyly

- Camptodactyly is classified based on the age of onset and severity:
  - **Type I**: Presents in infancy, affecting one or two fingers.
  - **Type II**: Presents during adolescence, affecting multiple digits.
  - **Type III**: Severe form associated with other congenital abnormalities, often involving multiple digits bilaterally.
- **Treatment**
- Non-surgical management is preferred, particularly in infants and young children, using static or dynamic splinting.
- Surgical interventions may be necessary for severe cases where functional impairment occurs, focusing on releasing contracted soft tissues and correcting any bony deformities.

Clinodactyly

- **Clinodactyly** refers to a congenital curvature of a digit, usually the little finger, involving radial or ulnar deviation at the level of the proximal interphalangeal joint.

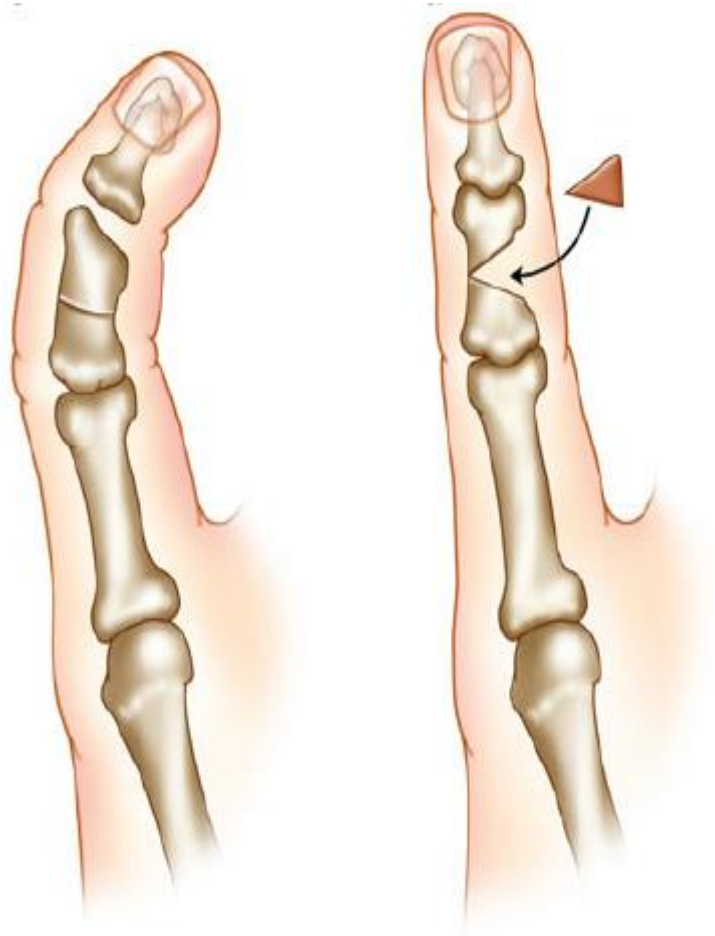




Clinodactyly

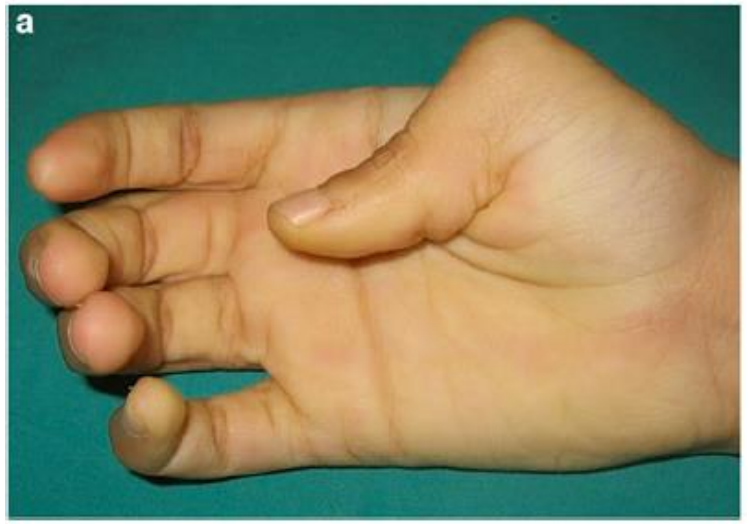
- **Etiology**
- Often caused by an abnormally shaped middle phalanx, resulting in progressive angulation as the child grows.
- **Pathology**
- The middle phalanx may be triangular (delta phalanx) due to an abnormal longitudinally oriented epiphysis, leading to angular deformities.

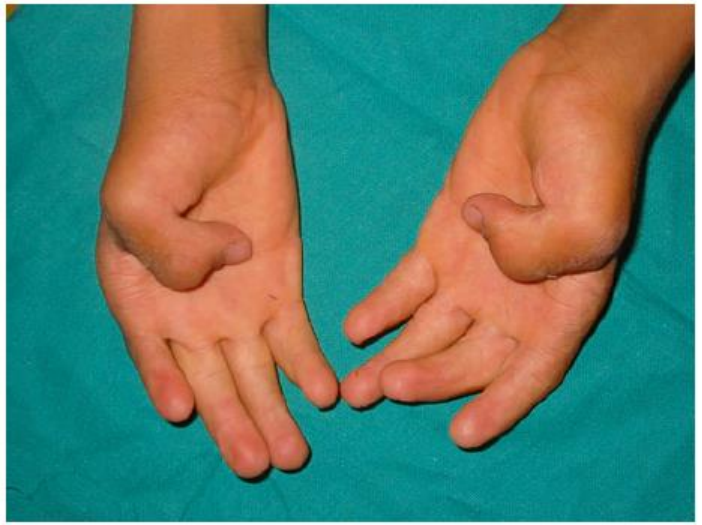
- **Classification**
- Based on the degree of deviation:
  - **Mild**: Less than 10 degrees.
  - **Moderate**: Between 15-30 degrees.
  - **Severe**: More than 30 degrees, often requiring surgical intervention.
- **Treatment**
- Nonsurgical management is recommended for mild forms, primarily for cosmetic concerns.
- Surgical correction, such as osteotomies, may be required for severe angulation that interferes with hand function.



Congenital Clasped Thumb

- **Congenital Clasped Thumb** is characterized by the inability to extend the thumb at the metacarpophalangeal (MCP) joint. It may also affect the interphalangeal (IP) joint, resulting in the thumb remaining flexed and adducted into the palm. This condition is distinct from spastic thumb deformities such as those seen in cerebral palsy, which are not congenital





- Clinical Presentation
- - The primary feature is the lack of active extension at the MCP joint of the thumb. Associated signs can include narrowing of the web space, wrist extension contracture, and camptodactyly of other fingers. The thenar muscles might show mild to severe hypoplasia, indicating more complex thumb hypoplasia rather than isolated clasped thumb
- Pathoanatomy:
- - The deformity typically stems from a **flexor-extensor imbalance**, where the extensor tendons, especially the **extensor pollicis brevis (EPB)** and **extensor pollicis longus (EPL)**, are hypoplastic or present only as vestigial strands. The flexor tendons and related structures contribute to the flexion contracture

- **Classification**
- The condition can be classified into several types based on severity and associated conditions:
  - **Type I**: Isolated clasped thumb.
  - **Type II**: Clasped thumb with finger contractures.
  - **Type III**: Associated with radial ray hypoplasia.
  - **Type IV**: Miscellaneous group, including cases with polydactyly

- **Associated Syndromes**
- Frequently associated with **arthrogryposis**, a syndrome characterized by multiple joint contractures, indicating a more widespread musculoskeletal issue

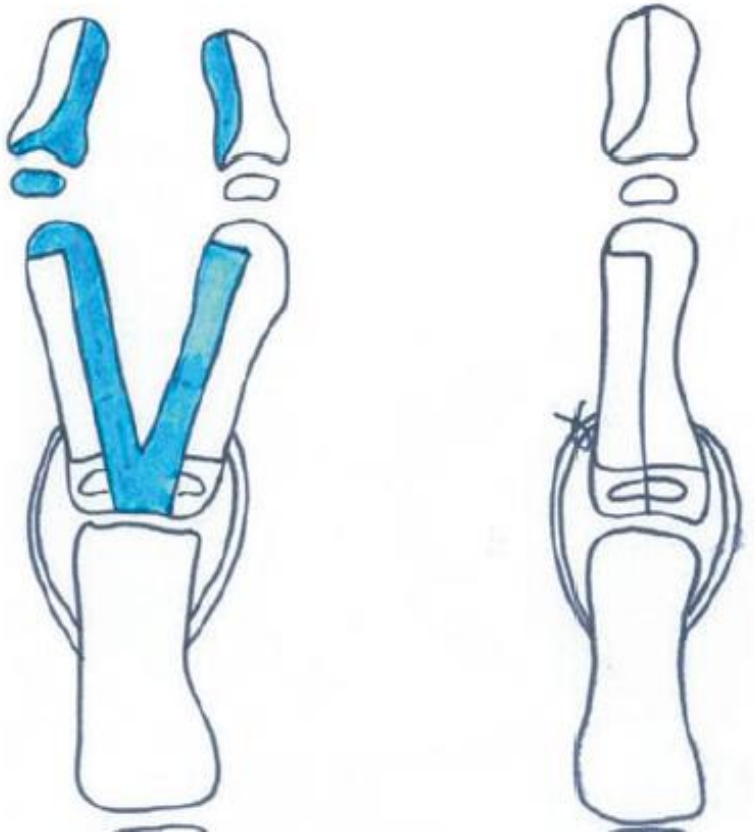
- **Treatment**
- **Non-surgical**: Includes splinting and passive stretching, particularly effective in infants and children for mild cases.
- **Surgical Management**: Tailored to the severity, involving tendon transfers to restore thumb extension, web space widening, and MCP joint stabilization. Surgical interventions are generally recommended after the age of 1.5 years to maximize outcome effectiveness

Radial Polydactyly

- **Definition and Epidemiology**
- **Radial Polydactyly**, also known as thumb duplication or preaxial polydactyly, involves the presence of an extra thumb or parts of an extra thumb on the radial side of the hand. It's one of the most common congenital hand anomalies.
- The condition occurs sporadically, though there's a higher hereditary predisposition when associated with a triphalangeal thumb, particularly noted in some familial cases
- **Genetics and Development**
- The **SHH (Sonic Hedgehog) pathway**, specifically the zone of polarizing activity regulatory sequence (ZRS), plays a crucial role in the anteroposterior axis of limb development. Mutations in ZRS and other related genes like **GLI3** are often associated with variations of radial polydactyly

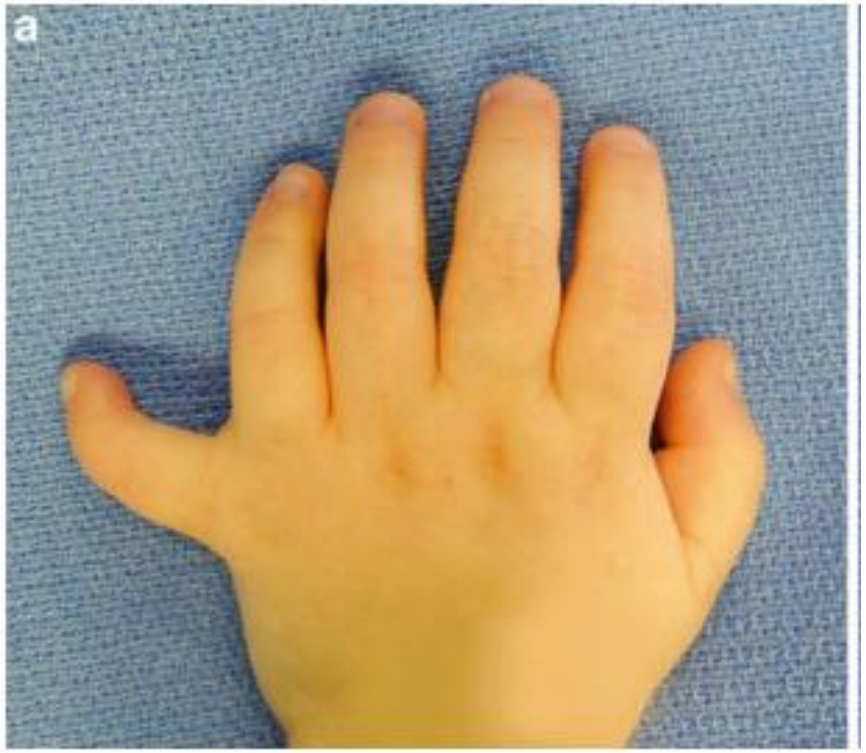
- **Classification**
- **Wassel Classification** is the most widely used system, categorizing the anomaly based on the level of duplication:
  - **Type I**: Bifid distal phalanx with a common epiphysis.
  - **Type II**: Complete duplication of the distal phalanx.
  - **Type III**: Duplicated distal phalanx with bifurcated proximal phalanx.
  - **Type IV**: Complete duplication of the proximal phalanx (most common).
  - **Type V**: Bifurcation of the first metacarpal.
  - **Type VI**: Complete duplication of the entire first digit.
  - **Type VII**: A triphalangeal thumb accompanied by a normal thumb

- **Surgical Management**
- The primary goal of surgery is to create a stable, functional, and aesthetically pleasing thumb.
- Techniques vary based on the type of duplication and may include excision, osteotomy, tendon realignment, and collateral ligament reconstruction
- Simple Excision: For rudimentary duplicated thumbs with no bony connections.
- Excision and Reconstruction: For more complex forms, involving arthroplasty and corrective osteotomy, particularly indicated for Wassel types I-IV.
  Bilhaut-Cloquet Procedure: Used in cases where both thumbs are underdeveloped; involves resecting the central portion of the duplicated segment and fusing the remaining parts
- On-Top Plasty



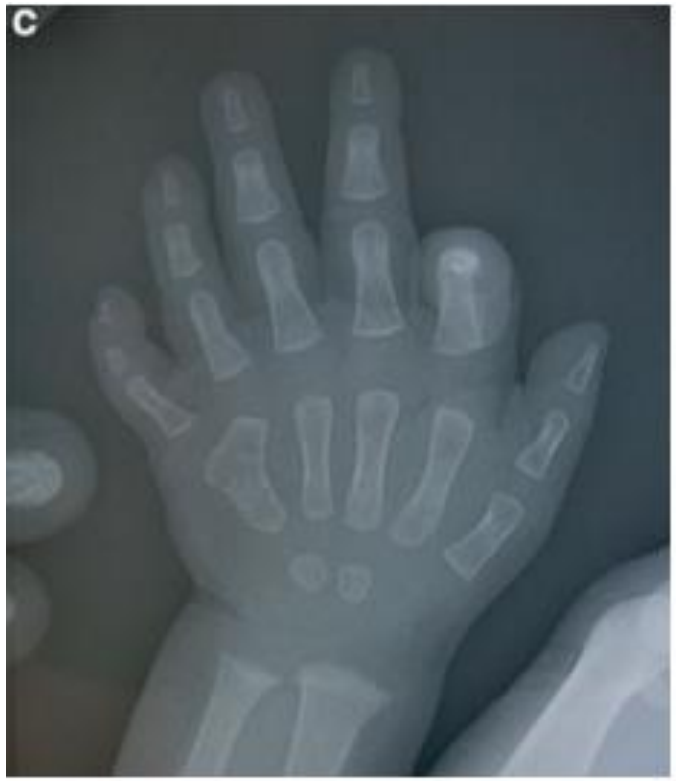
Ulnar Polydactyly

- **Ulnar Polydactyly**, also known as postaxial polydactyly, is the duplication of digits along the ulnar side of the hand.
- Classified into two main types:
  - **Type A**: Well-developed digits that are functional and may contain bone, joint, and tendon structures.
  - **Type B**: Rudimentary digits, often just soft tissue nubbins, which lack skeletal structures
- **Epidemiology**
- More common in certain populations, particularly African Americans where it occurs in about 1 in 150 newborns, compared to Caucasians at 1 in 1400

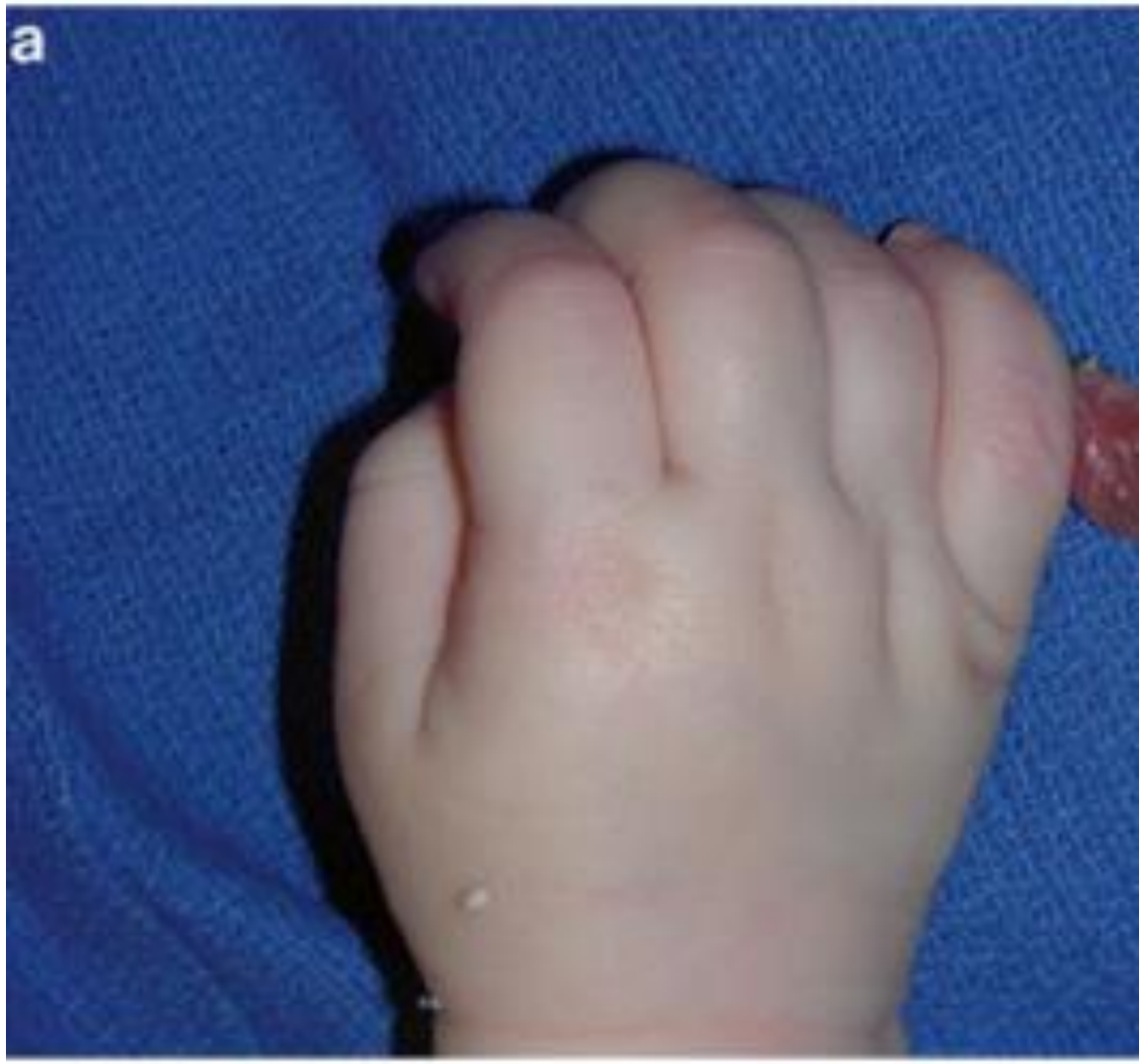


Clinical Presentation and Diagnosis

- Often diagnosed prenatally or through early postnatal physical exams.
- Type A requires detailed imaging for surgical planning, whereas Type B may just present as soft tissue on the ulnar side without the need for complex imaging

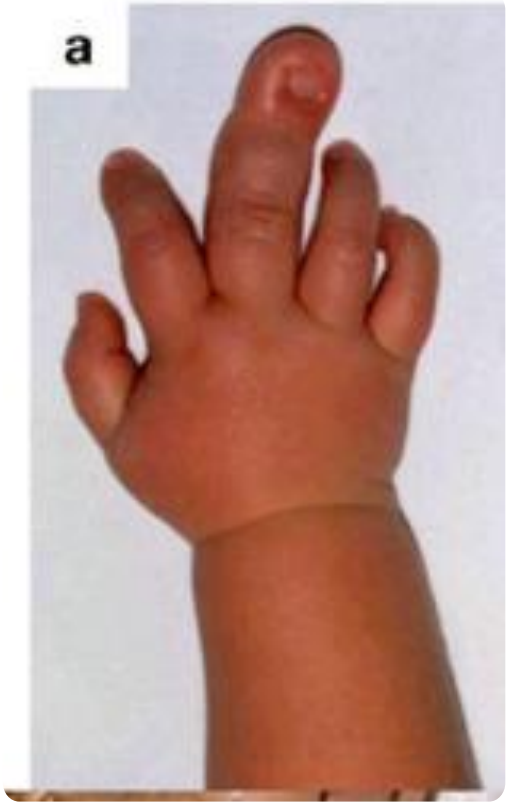


- **Treatment**
- Type A: Requires surgical removal of the extra digit and reconstruction of the hand to preserve and optimize function.
- Type B: Can often be treated non-surgically with suture ligation, leading to auto-amputation, but may require surgical excision if complicated by neuromas or other issue



Macrodactyly

- **Macrodactyly** is a congenital condition characterized by the overgrowth of the bones and tissues of the digits. It can affect one or more fingers or toes and is noticeable at birth or becomes more apparent as the child grows.
- **Epidemiology**
- Macrodactyly often does not follow a pattern of inheritance nor is it typically associated with other malformations.
- **Associated Conditions**
- Frequently associated with neurofibromatosis and other complex syndromes like Proteus syndrome or CLOVES syndrome, where the overgrowth is part of a broader pattern of anomalies.



- **Classification**
- Static Macrodactyly: The size of the digit is large at birth and remains proportionate as the child grows.
- Progressive Macrodactyly: The digit continues to grow disproportionately, becoming more prominent with age.

- Types

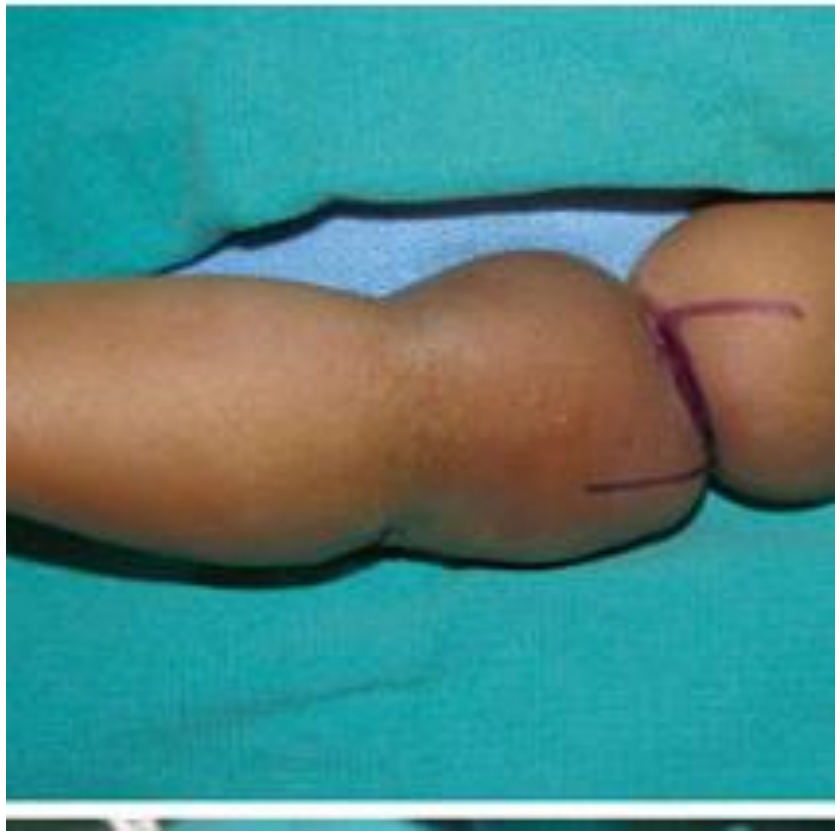
- Lipomatous Macrodactyly: The most common type, involving an overgrowth of all digital structures but without nerve infiltration.
- Neurofibromatosis-associated Macrodactyly: Characterized by nerve involvement along with the enlargement of bones and soft tissues, primarily affecting the median nerve territory.
- Digital Hyperostosis: Rare, involving bilateral symmetric or asymmetric enlargement of the digits without significant nerve or fatty tissue overgrowth .

- **Treatment**
- Conservative Treatment: Includes monitoring and physical therapy to manage symptoms and maintain functionality.
- Surgical Treatment: This may involve debulking of soft tissues, bone reduction surgery, or even amputation in severe cases. The aim is to improve functionality and appearance while managing the size and proportion of the affected digits.

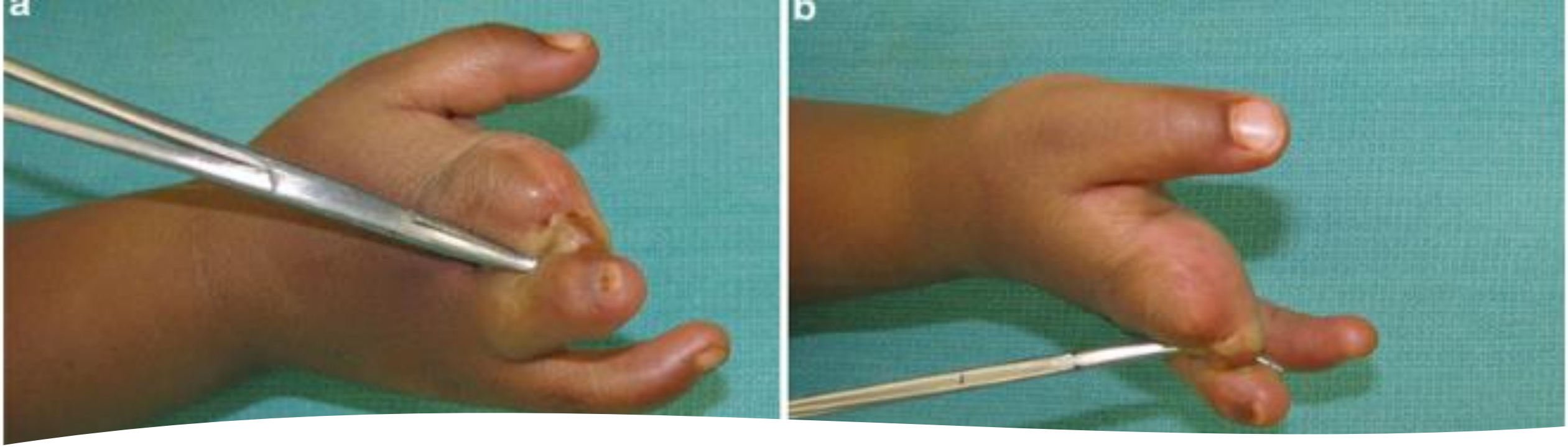
- **Prognosis**
- The outcome depends on the type and progression of the condition. Static forms typically have fewer complications and require less intervention, whereas progressive forms may need multiple surgeries and have a more guarded prognosis.

Amniotic Band Syndrome (ABS)

Amniotic Band Syndrome (ABS) is a congenital disorder characterized by fibrous bands that can wrap around limbs, fingers, or other parts of the fetus. These bands may constrict parts of the body, potentially leading to deformations or amputations.

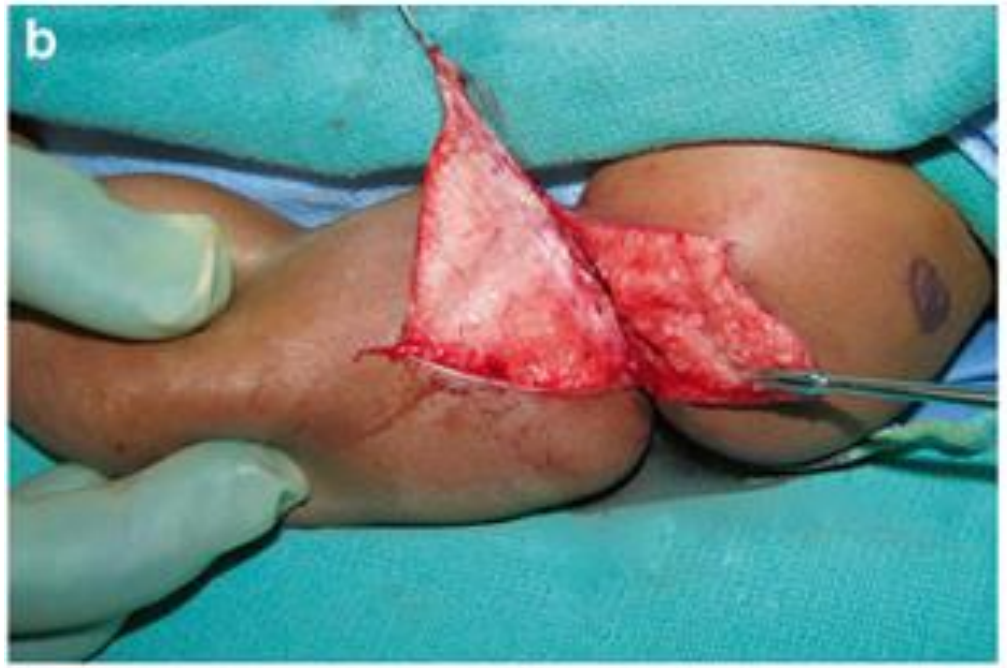


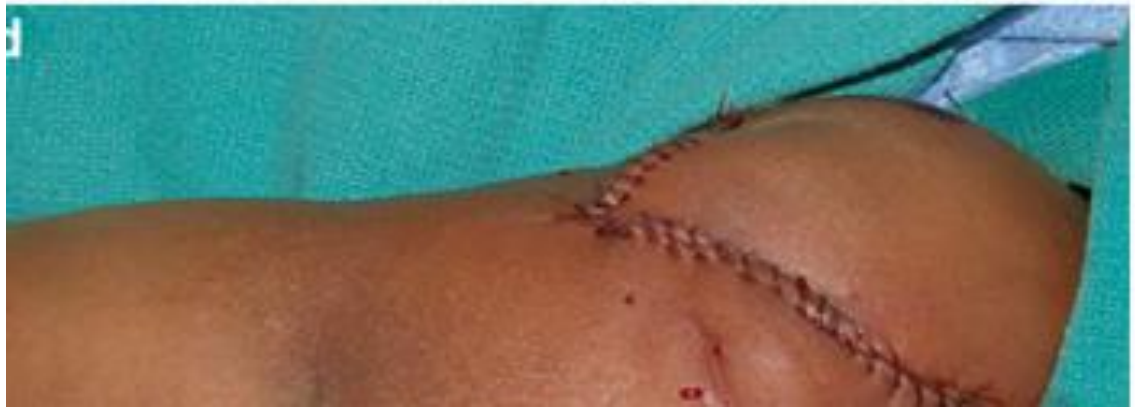
- **Etiology**
- Thought to result from early rupture of parts of the amnion, leading to fibrous strands that entangle fetal parts.



- **Clinical Presentation**
- Variability: The presentation of ABS can range from simple indentations on limbs to severe limb amputation.
- Common Manifestations: Include limb and digital amputations, constriction rings around limbs, acrosyndactyly, and in severe cases, craniofacial, thoracic, and visceral anomalies
- **Diagnostic Tools**
- Ultrasound: Utilized for prenatal detection of amniotic bands and associated deformities.
- MRI: Offers detailed images of anatomical structures and can reveal the extent of band involvement, especially useful for planning surgical interventions.

- **Management Strategies**
- Surgical Intervention: Necessary to correct deformities and release constriction bands.
- Procedures may include z-plasty or other reconstructive surgeries to alleviate band constriction and preserve affected limbs.
- Postoperative Care: Focuses on managing scarring, preserving limb function, and monitoring for developmental progress





- **Complications**
- Physical Disabilities: Depending on the severity and location of the bands, ABS can lead to significant functional impairments.
- Psychological Impact: Emotional and psychological support for families is crucial, as the visual and functional impacts of ABS can be considerable.

THE END

Any question?