Congenital Hand Anomalies

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Introduction:

Hi everyone. My name is Harold Zhu, and I am a first-year medical student at the University of Alberta. This PedsCases podcast provides a clinical approach to recognizing and managing Congenital Hand Anomalies. This episode was developed in collaboration with Dr. Regan Guilfoyle, a Pediatric Plastic Surgeon and Associate Professor at the University of Alberta.

The human hand represents the apex of evolutionary mechanical complexity, serving as our primary tool for interacting with the world. When a child is born with a hand "difference", a term we prefer over "deformity" to focus on a patient-centered approach, it carries profound implications for their developmental trajectory and self-concept. [1.]

Learning Objectives:

By the end of this podcast, listeners will be able to:

1. Describe the key stages and molecular signaling centers of embryologic hand development.
2. Categorize major malformations using the Oberg, Manske, and Tonkin (OMT) classification.
3. Recognize clinical "red flags" and systemic associations, specifically for radial ray defects.
4. Outline the clinical rationale for surgical timing in common anomalies like syndactyly and polydactyly.

Clinical Case:

Let's start with a scenario. You are a medical student on your neonatal rotation. You are asked to examine "Aiden," a 2-day-old baby boy. His parents are healthy and the pregnancy was uncomplicated, but they are understandably distressed about differences in Aiden's hands.

On your exam, you notice that on Aiden's right hand, the middle and ring fingers are fused together up to the level of the fingernail. His left hand appears typical at first, but you notice a small, rudimentary "nubbin" of skin, about the size of a pea, hanging off the ulnar side of his left little finger.

Clinical Pearl: When you identify a hand difference, always perform a "head-to-toe" exam. A hand anomaly is often the first visible sign of a systemic syndrome involving the heart, kidneys,

or bone marrow, that could be life-threatening. [2,3]

Why does the topic matter?:

Congenital upper limb anomalies occur in approximately 21.5 per 10,000 live births. While many are isolated, others serve as essential clinical markers for conditions involving the heart, kidneys, or hematopoietic system. [4.]

Beyond the medical, there is a significant psychosocial component. Primary research indicates that 58% of children with congenital hand differences experience significant stress, and a striking 43% of their parents may be unaware of this stress. [1] Early recognition allows a multidisciplinary team, including surgeons, therapists, and psychologists, to support the family from day one through external support, education ,and other specialized support programs. [1]

Pathophysiology and Embryology:

To understand what went "different," we must look at weeks 4 through 8 of gestation. [5] The hand develops from a mesenchymal limb bud along three primary axes:

1. Proximodistal Axis (Shoulder to Fingertip): Regulated by the Apical Ectodermal Ridge (AER), which secretes Fibroblast Growth Factors (FGF) to maintain outgrowth. [5] Disruption here leads to limb truncations or transverse deficiencies. [5]
2. Anteroposterior Axis (Thumb (D1) to Little Finger (D5)): Specified by the Zone of Polarizing Activity (ZPA) through a gradient of the protein Sonic hedgehog (Shh). [5] High Shh specifies the ulnar side, while low Shh allows the thumb to form. [5] Ectopic signaling here causes duplications, like polydactyly. [5]
3. Dorsoventral Axis (Back of hand to Palm): Regulated by molecules like Wnt which ensure you have nails on the back, and flexural creases on the palm. [5]

Key processes in the development of the hand include the initial hand plate forming as a solid paddle. [5] Between weeks 6 and 8, the tissue between the fingers must undergo apoptosis, or programmed cell death. [7,8] A failure of this process results in syndactyly, or webbed fingers. [7,8]

Classification, OMT vs Swanson System

Historically, surgeons used the Swanson system based on "clinical appearance." However, the international community has transitioned to the Oberg, Manske, and Tonkin (OMT) system, which focuses on the developmental mechanism. [9]

- Malformations: Primary failures of the developmental axes, like polydactyly or radial deficiency. [9]

- Deformations: Occur when a normally formed limb is restricted by external forces, such as amniotic band syndrome. [9]
- Dysplasias: Abnormal tissue organization, such as macrodactyly, where all tissues in a finger are overgrown. [9]

Clinical Presentation and Physical Exam:

Your history should include prenatal exposures to medications like valproate (or other known teratogenic drugs) and a detailed family history, as conditions like postaxial polydactyly often follow an autosomal dominant pattern. [10,11] A thorough evaluation begins with a comprehensive history. You should also gather general info like age, sex, and hand dominance.[12] Start with open-ended questions to let the patient or parents tell their story, then home in on specifics. [12] For chronic or congenital conditions, ask broader questions: When did the problem begin and what is the course like? Is pain constant or intermittent, and can it be localized? Are there associated paresthesias, stiffness, or weakness? Crucially, ask how the condition affects daily activities, vocation, and hobbies. [12]

The Physical Exam:

-Position yourself across from the patient with their hands resting on a hand table, ensuring arms are exposed from the elbow distally. Always evaluate both extremities to provide a baseline for comparison. [12]

-Inspection and Palpation: Observe hand positioning and use, specifically looking at the digital cascade or resting posture. Note skin color, localized or generalized swelling, and any scars, which can be clues to prior trauma. Inspect the nail complex for pitting or brittle plates, as these may indicate systemic disease or nutritional deficiencies. Have the patient flex their fingers and oppose the thumb, observing for normal range of motion or rotational deformities like scissoring. [12]

-Vascular Assessment: Assess digits for color, warmth, and capillary refill in the nailbed (normal is < 3 seconds). Palpable peripheral pulses should be noted; if absent, a Doppler exam for triphasic flow is necessary. [12]

-Sensory Testing: This consists of light touch, which can be performed with a fingertip of cotton swab; and 2-point discrimination which can be performed with a paper clip. Fashion the paper clip so that the two ends can be held against the skin simultaneously and their relative distance can be changed by the examiner. Normal ranges are less than 6 mm for static and 3 mm for dynamic discrimination. [12]

-Motor Function: Differentiate musculotendinous from neurological conditions using the tenodesis effect: wrist extension should produce finger flexion, and wrist flexion should produce

finger extension. A lack of this pattern indicates a disruption of the musculotendinous units. This is a good test to use in patients who are unable to participate in a physical exam. Use the Bunnell test to evaluate intrinsic tightness. [12]

-The Thumb: Assess motion in multiple planes: radial and palmar abduction, opposition, and circumduction. [12]

-Imaging: Standard radiographs should include 3 views: anteroposterior (AP), lateral (with fingers fanned), and oblique. Special views, like a true AP and lateral of the thumb ray, may be required to visualize the trapeziometacarpal joint.[12]

Differential Diagnosis of Radial Ray Deficiency:

Radial club hand is a spectrum of complex malformations of the radial forearm, ranging from thumb hypoplasia to a completely absent radius.[13] It is the most common longitudinal deficiency, occurring more in boys (3:2 ratio) with an incidence between 1 in 5,000 and 1 in 55,000 births.[13] The etiology is thought to involve Sonic hedgehog (Shh) mutations affecting FGF function.[13]

Presentation and Evaluation:

Children often present with a radially directed angulation of the hand, frequently perpendicular to the forearm.[13] Bony abnormalities are apparent, but muscles, tendons, nerves, and joints are also affected. [13]The ulna is typically short and bowed.[13]

Bayne and Klug Classification:[13]

- Type I: Short distal radius (growth plate present).
- Type II: Hypoplastic radius (with growth plate deficiency).
- Type III: Partial absence of the radius.
- Type IV: Total absence of the radius (most common).

Systemic Associations & Workup:[13]

Only one-third of cases are isolated. Differential diagnoses include TAR syndrome, Fanconi anemia, VACTERL/VATER, Holt-Oram, and trisomies 13 and 18. Screening requires:

1. CBC: To screen for bone marrow issues.
2. Renal Ultrasound: To evaluate for VACTERL anomalies.
3. Echocardiogram: For septal defects (Holt-Oram).
4. Spine X-rays: To check for vertebral defects.
5. Chromosomal Breakage Studies: Mandatory for all children with radial or thumb deficiencies to rule out life-threatening Fanconi anemia.

Management:

Treatment starts soon after birth with stretching and splinting. [14]Surgical treatment is staged: Stage 1 involves aligning the hand onto the distal ulna (centralization or radialization) before age one, followed 6 months later by Stage 2 (thumb reconstruction or index pollicization). [14]

Management of Syndactyly, Polydactyly, and the Hypoplastic thumb

Surgical indications aim to improve hand function and cosmesis while limiting long-term complications. [14]

Syndactyly:

This occurs in 2-3 per 10,000 live births, often associated with toe webbing. [14]

- Timing: Release should be done at 3–6 months only if finger size is mismatched and growth is causing deviation. Otherwise, delay until 1–2 years to decrease the risks of general anesthetic. [14]
- Technique: Zigzag flaps are used to break up longitudinal scar lines. Supple dorsal skin flaps create the commissure; grafts are almost always necessary in the radial and ulnar base of the webspace due to overall deficiency of skin in this area. Webspace creep, or loss webspace depth is a possible complication of surgery that can be corrected with 4-flap z-plasty in the future if needed. [14]

Polydactyly:

- Preaxial (Radial): Often sporadic. triphalangeal thumbs follow an autosomal dominant pattern and associated with syndromes like Holt-Oram or Blackfan-Diamond anemia. [14] Neither duplicated thumb is normal.[14]Treatment involves preserving the most functional digit (typically ulnar) and reattaching the collateral ligaments. [14]
- Postaxial (Ulnar): Incidence is 1 in 143 in infants of African descent.[14] Type B (rudimentary) digits can be suture ligated at birth, though this may cause tender neuromas in 16% of cases.[14] Elliptical excision with neurovascular ligation is often recommended. [14]

Thumb Hypoplasia:

This follows the Blauth classification (Type I to V). [14]

- Type II/IIIA: Present with a stable MCP joint with instability of the UCL and benefit

from opponensplasty (using FDS tendon transfer) and web space deepening. [14]

- Type IIIB/IV/V: These involve instability at the MCP joint (Type IIIB) or complete absence of the thumb (Type V). The procedure of choice is index finger pollicization, performed between 1.5 and 2 years of age.[14]

Summary and take home messages

To wrap up, here are the three most high-yield points from this podcast:

1. The Hand is a Window: Hand differences such as a radial ray defect is an urgent indicator to screen the heart, kidneys, and bone marrow. [2,3]
2. Timing is Growth-Dependent: While simple syndactyly release is often delayed until 1–2 years, border digit syndactyly (thumb/index or ring/pinky) needs referral and release by 3-6 months to prevent the shorter digit from permanently tethering and curving the longer one.[14]
3. Address the Psychosocial Gap: 58% of children experience stress related to their hand difference, yet 43% of parents are unaware.[1] A multidisciplinary approach involving psychological screening is essential for the child's development.[1]

Thanks for listening to this PedsCases podcast. For more information, visit PedsCases.com.

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