Syndactyly-Polydactyly: A Patterned Approach to Diagnosis and Categorization with Genetic Correlation

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Learning objectives

This electronic exhibit will review radiographic patterns and categorizations of syndactyly-polydactyly with emphasis on a structured approach to imaging findings and genetic significance. The embryology of limb development, findings of non-syndromic and syndromic syndactyly, and polydactyly will be discussed.

Background

Embryology

Limb buds are formed between the 4th and 8th weeks of gestational age with signaling centers in the limb bud controlling the presence of absence of asymmetry. There is an apical ectodermal ridge (AER) determining the proximal-distal axis and the zone of polarizing activity (ZPA) for the anterior-posterior axis (i.e. thumb to little finger). ZPA positioning is influenced by several encoding proteins including the hedgehog (Shh) pathway. Mutations in genes associated with Shh, and pathways regulating apoptosis and ossification are involved in the pathogenesis of syndactyly and polydactyly.

Syndactyly

Syndactyly is defined as the function of the soft tissues between digits with or without bony fusion. This fusion can be divided into simple (only soft-tissue involvement, complex (side-to-side bony fusion). Additionally, fusions can be divided into complete (fusion extending to finger tips) or incomplete (partial fusion).

Epidemiology

Syndactyly/polydactyly is one of the most common congenital hand deformities (approximately 1 in 2000). There is a male predominance occurring in a ratio of 2:1. Family history is usually contributory in 10-40% of cases, most common due to an autosomal dominant inheritance with variable penetrance and expressivity. However, the occurrence may be an isolated finding or in a syndrome.

Findings and procedure details
Non-Syndromic Syndactyly

Non-syndromic syndactyly only involves the digit and results in appendage malformation. There are nine sub-groups (SD1 to SD9).

SD1
- Zygodactyly
- Varied phenotype - can affect upper and/or lower limbs
- In the hand, cutaneous or bony involvement of the 3rd or 4th web space
- Only autosomal dominant inheritance
- 4 sub-types (zygodactyly 1-4)

SD2
- Synpolydactyly
- Most heterogeneous malformations of the non syndromic syndactylies
- Autosomal dominant inheritance
- 3rd and 4th finger syndactyly with polydactyly within the web space

SD3
- Complete syndactyly of the 4th and 5th digits
- Bilateral
- Most commonly a soft-tissue syndactyly with absent or rudimentary 5th middle phalanx
- Disease spectrum with oculodengodigital dysplasia (ODDD)

SD4
- Haas Type
- Very rare (4 cases reported in literature)
- Complete bilateral soft-tissue syndactyly
- Polydactyly with extra digit and metacarpal
- Autosomal dominant inheritance

SD5
- Most common affects the 3rd and 4th digits or the 4th and 5th digits of the hand
- Associated bony fusion of metacarpals
- Autosomal dominant as well as X-linked recessive inheritance have been described

SD6
• "Mitten hand" deformity
• Unilateral syndactyly of the 2nd to 5th digits
• Limited research into genetic cause

SD7

• Very rare phenotype characterized by Drs. Cenani and Lenz
• Autosomal recessive inheritance reported
• Fusion of metacarpals
• "Disorganization" of phalangeal development
• Severe shortening and fusion of ulna and radius

SD8

• Characterized by bony fusion of the 4th and 5th metacarpals with syndactyly
• X-linked recessive trait

SD9

• Mesoaxial Synostotic Syndactyly (MSSD)
• Complete syndactyly and synostosis of 3rd and 4th digits
• Bony reduction in the proximal phalanges
• Aplasia/hypoplasia of the middle phalanges of the 2nd and 5th digits
• Soft-tissue syndactyly of the toes

Other Syndactylies

Acrosyndactyly

• Syndactyly associated with congenital construction bands
• No genetic basis

Syndromic Syndactylies

• Extensive and growing list of syndromes
• Most common include Poland syndrome, and Acroencephalosyndactyly (Types I-V)

Poland Syndrome

Ipsilateral chest wall and upper extremity involvement with some of the following features:

• Hypoplasia/aplasia of the pectoralis muscle and other chest wall muscles
• Hypoplastic skin/subcutaneous tissue/breast
• Absent ribs and costal cartilage
• Shortened upper arm and forearm
• Simple, complete, or incomplete syndactyly

No definite cause has been identified with an estimated incidence of 1:30,000

**Acroencephalosyndactyly**

Craniosynostosis and syndactyly

• Type I: Apert syndrome (Mid face hypoplasia, foot and hand syndactyly)
• Type II: Carpenter syndrome (Acrocephaly, foot and hand syndactyly)
• Type III: Saethre-Chotzen syndrome (eyelid and cranial abnormalities, syndactyly of the 2nd and 3rd fingers)
• Type IV: Goodman syndrome (now classified with type II)
• Type V: Pfeiffer syndrome

**Management**

• Surgery is the mainstay of management
• Intervention is non-urgent, but age related targets exist for aesthetic and functional considerations
• Multiple digit involvement is surgically released in stages, with border digits released first
• Full thickness skin grafts and flaps used for soft tissue coverage

**Polydactyly**

• Presence of supernumerary digits
• Isolated or a syndromic feature
• Currently, over 221 syndromes associated with polydactyly
• Can be classified as ulnar, radial, and central polydactyly

**Radial (preaxial) polydactyly**

• Up to 1 in 3000 births
• Wassel classification (Type 1 to 7) most widely used
• Highest incidence of Type 4
• Usually isolated but Type 7 is associated with autosomal dominant inheritance
• Surgical management requires complicated reconstruction

**Ulnar (postaxial) polydactyly**

• Presentation varies from a well developed digit (type A) to a skin tag (type B) according to Temtamy and McKusick classification
• Up to 1 in 143 live births in African Americans, compared to 1 in 1339 live births in Caucasians
• Autosomal dominant inheritance for isolated presentation
• Autosomal recessive inheritance and syndromic association seen in Caucasian populations
• Can be treated with simple excision, or reconstruction in cases of joint involvement

**Central Polydactyly**

• Duplication of a non-border digit
• Rare form of polydactyly
• Most commonly seen in the ring finger and middle finger
• Most often presents as a part of synpolydactyly (SD2)

**Images for this section:**
Fig. 2: SD1
Fig. 3: SD2
Fig. 4: SD2
Fig. 5: SD3

Different patients

Fig. 6: SD3
Fig. 8: SD4 (Sato D et al. [3])
Fig. 9: SD5
**Fig. 10:** SD5 (Zhao et al.[4])
Fig. 11: SD6

Fig. 12: SD6
Fig. 14: SD7 (Elliott et al.[5])
Fig. 15: SD8

From Lonardo et al. [6]

Fig. 16: SD8 (Lonardo et al. [6])
Fig. 18: SD9 (Malik et al.[7])
**Fig. 19:** Ulnar (Postaxial) Polydactyly
Conclusion

This pictorial review illustrates the phenotypes of non-syndromic and syndromic syndactyly/polydactyly with emphasis on a structured findings approach to description and classification. Prenatal and perinatal imaging can help contribute to the clinical pattern and genetic prognosis.

Personal information

References


