# Congenital hand malformations in an anatomical donor: a potential case of brachydactyly type B

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## SUMMARY

This study investigates the anatomy of a cadaveric hand presenting with a rare malformation that we identified as a form of brachydactyly. We performed a layer-by-layer dissection of the affected cadaveric hand and documented both the normal and variant anatomical compositions that were present. The osteology revealed complete aplasia of the proximal and middle phalanges of the second, third, fourth, and fifth digits along with truncated distal phalanges of the fourth and fifth digits. The radius, ulna, carpal bones, and the first digit were unremarkable. Neurovasculature followed standard anatomical courses. Normal development of surrounding musculature was observed.

Congenital hand malformations are infrequently observed in the anatomy lab and provide a unique learning opportunity for students and faculty alike. While hand deformities are frequently imaged through noninvasive techniques such as X-rays and CT imaging, they are infrequently dissected. Through dissection, clinicians and anatomists may be able to adopt a more tangible appreciation for the underlying anatomy of this condition.

**Key words**: Brachydactyly – Anonychia – Clinodactyly – Hand malformations

## INTRODUCTION

The upper limb begins developing between weeks four to eight beginning with the formation of two paired limb buds (Guéro, 2018; Simet and Cassidy, 2021). There are several factors that can lead to limb dysmorphologies including but not limited to alternations in signaling biology, environment, and individual genotype (Simet and Cassidy, 2021). Congenital hand malformations occur in approximately 27.2 per 10,000 births (Goldfarb et al., 2017). One of these hand malformations is called brachydactyly. Brachydactyly is a general term that refers to disproportionately short digits in both the upper and lower extremities, resulting from abnormal development of either the metacarpals, phalanges, or both (Tentamy and Aglan, 2008). In this report, we investigated the anatomy

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of a cadaveric hand afflicted with brachydactyly by performing a layer-by-layer dissection to document any unusual findings present. Anatomists have been documenting variations in hand morphology for many centuries, but there are very few reports on the gross anatomy of congenital hand conditions such as brachydactyly (Henle, 1871; Macalister, 1866; Wood, 1866).

Surgical separation of syndactyly, osteotomy for deviated digits or bone grafting for short digits may be indicated in treatment of brachydactyly to enhance function (Stevenson and Hall, 2005). Therefore, there are clinical advantages in studying these limb dysmorphologies through dissection. The knowledge gained from dissecting these limb dysmorphologies provides crucial information of anatomical variation as well as confidence regarding treatment decisions such as surgical separation.

## CASE REPORT

We describe a case of a 71-year-old female human cadaver that presented with brachydactyly that is rarely observed within the anatomy lab. The specimen was fixed with an embalming solution of formaldehyde and phenol. The cause of death was documented by the medical examiner as upper gastrointestinal bleeding. A layer-by-layer dissection was performed of the afflicted cadaveric hand and we potentially identified rare anatomical findings. The contralateral hand, and lower extremities presented with normal anatomy. The unskinned hand presented with truncation of second, third, fourth, and fifth digits, clinodactyly (curved finger) of the second and third digits, and anonychia (missing nails) of all digits with the exception of the thumb (Fig. 1). Clinodactyly, although present on the second and third digits, typically occurred in the fifth digit (Goldfarb and Wall, 2015). The inferior portion of the hand had been partially skinned by students for the medical anatomy course before we began this study. The next figure shows the completely skinned hand (Fig. 2). Thenar and hypothenar musculature had standard proximal and distal attachments. The superficial palmar arch and its branches were normal other than being truncated due to the shortness of the fingers. We next removed the thenar musculature and isolated

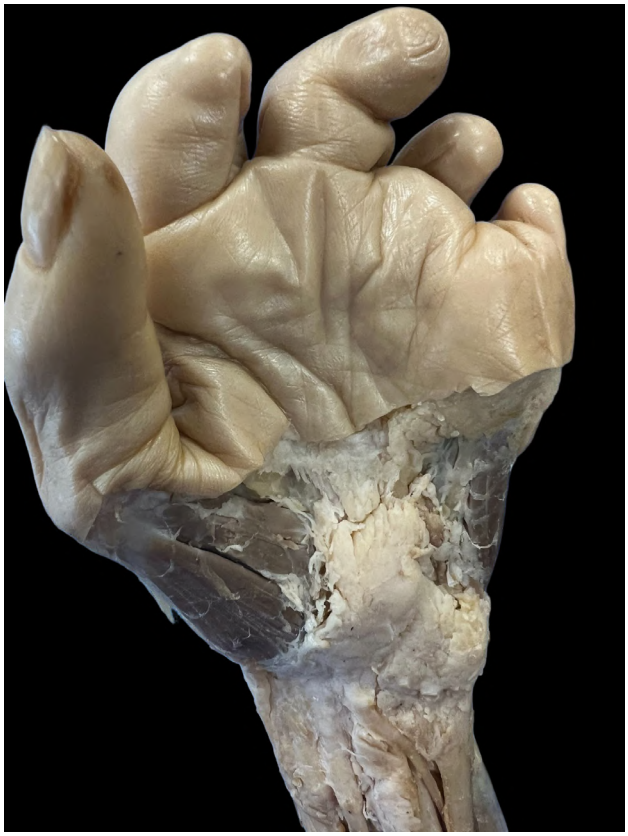

**Fig. 1.-** Left handed brachydactyly: The specimen presented with shortening of the second to the fifth digits along with anonychia of the second to fifth digits.

the lumbrical muscles and the tendons of the superficial flexor, flexor digitorum profundus (FDP) and flexor digitorum superficialis (FDS) muscles. All four lumbrical muscles were present but appeared slightly hypertrophic and longer in length than normally observed. Following standard anatomical description, the FDP tendons passed through splits in the FDS tendons, but FDS had a distal attachment on the distal phalanges as opposed to its regular attachment on the sides of the middle phalanges (Fig. 3). FDPs attachments were unremarkable. The lumbricals, FDS and FDP were removed in order to reveal the palmar interossei and the adductor pollicis muscle (Fig. 4). Both transverse and oblique heads of adductor pollicis were present. Finally, all remaining muscles were removed from their attachments in order to isolate the carpals, metacarpals, and phalanges. The osteology revealed complete aplasia of the proximal and middle phalanges of the second, third, fourth, and fifth digits along with truncated distal phalanges of the fourth and fifth digits (Fig. 5). The radius, ulna, carpal bones, and the first digit were unremarkable.

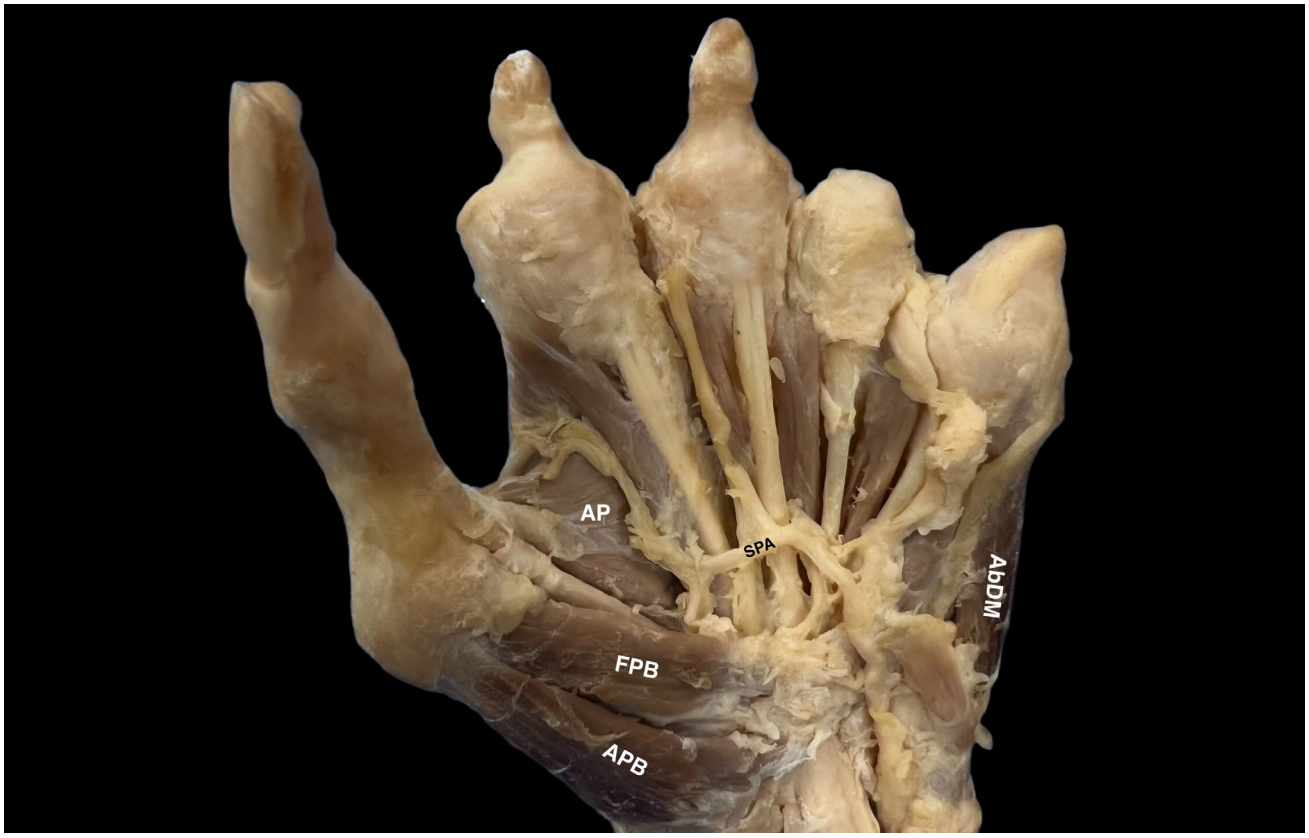


**Fig. 2.-** Skinned hand: The thenar and hypothenar musculature is shown along with the superficial palmar arch. (FPB; flexor pollicis brevis; AbPB; abductor pollicis brevis; AdP; adductor pollicis; AbDB; abductor digiti minimi; SPA; superficial palmar arch).

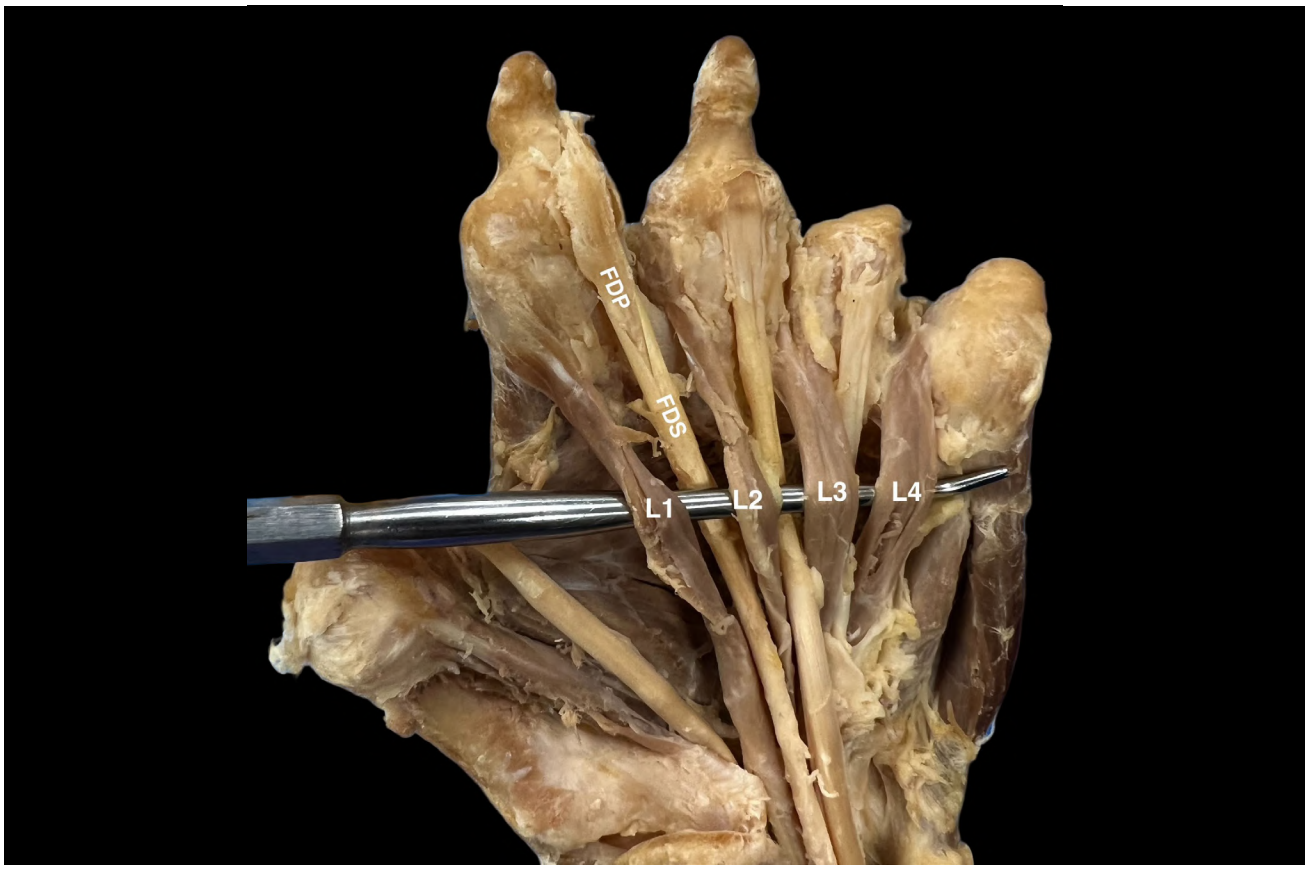


**Fig. 3.-** Lumbricals and FDS and FDP muscles: The distal attachments of the four lumbricals and FDS and FDP muscles are shown. (L1-L4; lumbricals 1-4; FDS; flexor digitorum superficialis; FDP; flexor digitorum profundus).

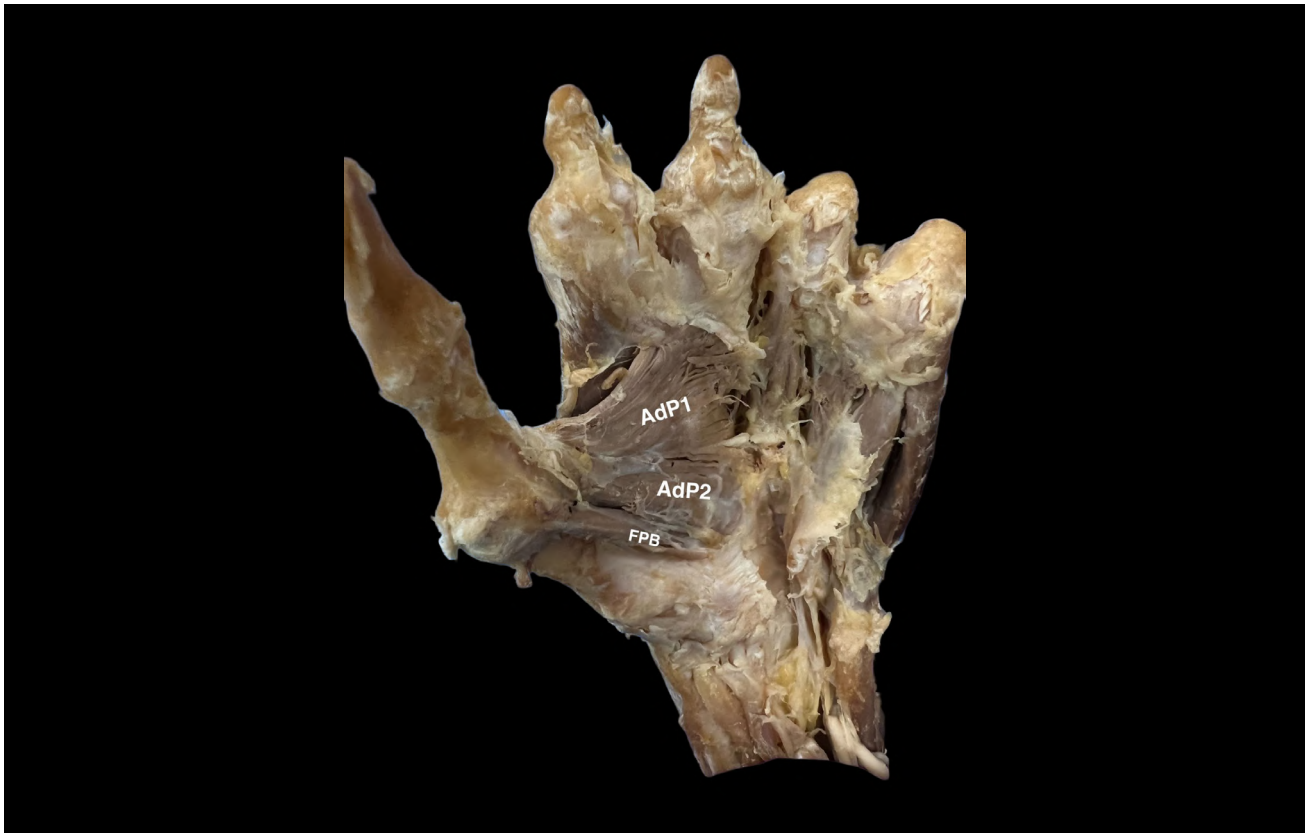


**Fig. 4.-** Adductor pollicis and palmar interossei muscles: The two heads of the adductor pollicis and the deep head of flexor pollicis brevis. (AdP1; transverse head; AdP2; oblique head; FPB; deep head of FPB).

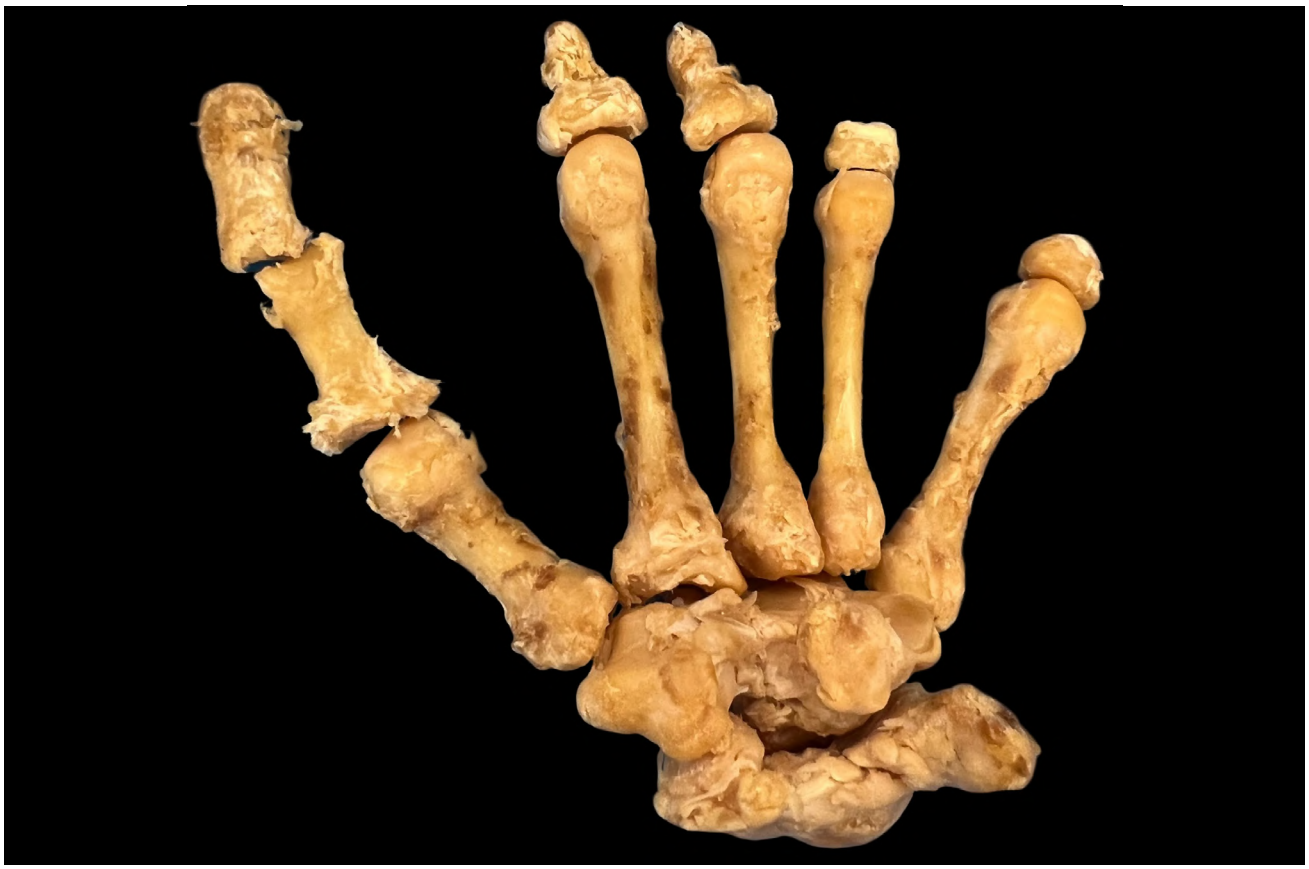

**Fig. 5.-** Osteology: All proximal and middle phalanges of the second to fifth digits are absent. The distal phalanges of the fourth and fifth digits are shortened.

## DISCUSSION

Bell and Temtamy developed a system that is commonly used to classify brachydactyly based on anatomical grounds (Bell, 1951; Temtamy and Aglan, 2008). There are eleven main types of isolated brachydactyly, each characterized by aplasia or hypoplasia of a part of the digit (Temtamy and Aglan, 2008). Fitch's classification provides a comprehensive analysis of each of the main types and also further subdivides Type E (Fitch, 1979). Our findings did not precisely align with any of the categories described in either of the aforementioned systems. Our donor presented with shortening of the 2nd to 5th digits, where all proximal and middle phalanges were absent. Additionally, there was clinodactyly in the 2nd and 3rd digits and anonychia in all digits except the thumb. Our case most closely resembles type B brachydactyly, which is characterized by absence or hypoplasia of the terminal parts of the index to little fingers with complete absence of fingernails (Temtamy and Aglan, 2008). However, the thumb frequently shows variation of the distal phalanges, which was not present in our donor. On the other hand, the case does not appear to match any type of symbrachydactyly either. We hypothesize the congenital variation in the donor to be closest to type IIIA of symbrachydactyly, however a symbrachydactyly is typically associated with ectodermal elements such as nails. Our case lacks this kind of tissue (Goodell et al., 2016). Symbrachydactyly typically results from a lack of blood flow to the tissue during embryological development. In our donor, the digital vessels, and distal median and ulnar nerve branches were unaffected other than being truncated due to the overall shortness of the digits. Overall, based on the osteology findings, we hypothesize that the congenital hand malformation observed in this donor was type B brachydactyly.

## CONCLUSION

Brachydactyly is infrequently observed in the anatomy lab as they have an extremely low prevalence, except for the A3 and D classification being roughly 2% (Temtamy and Aglan, 2008). X-rays or CT imaging would provide stronger evidence of the type of brachydactyly present in the patient. Future iterations of similar studies should include acquisition of clinical images prior to anatomical dissection. Even when they are observed, these hand deformities are frequently imaged through noninvasive techniques such as x-rays and CT imaging rather than dissection. Therefore, this case provides a unique learning opportunity of the various patterns of malformation in brachydactyly with the tangible appreciation for the important underlying anatomy of this condition.

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## AUTHOR CONTRIBUTIONS

*Lin*: project development, manuscript writing. *In*: manuscript editing. *Baribeau*: manuscript editing. *Wisco*: manuscript editing, PI.

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