“Thenar Dysplasia in Radial Polydactyly Depends on the Level of Bifurcation.”

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Financial Disclosure Statement: No funding was received for this article.

This study is registered in UMIN Clinical Trials Registry
Trial registry name: “カスタムメイド超音波３D画像による体表解剖に関する研究および臨床診断における有効性に関する研究” (Studies on superficial anatomies of human body by 3D ultrasound)
Registration identification number: UMIN000014396
URL: https://upload.umin.ac.jp/cgi-open-bin/ctr/ctr_view.cgi?recptno=R000016753

This study was accepted to be orally presented at 4th European Symposium on Pediatric Hand Surgery and Rehabilitation, which will be held in Paris, France in June 15-16, 2017.

Short Running Head

Thumb Dysplasia in Radial Polydactyly

Thenar Dysplasia in Radial Polydactyly Depends on the Level of Bifurcation

Abstract

Background

Little is known about thenar dysplasia in radial polydactyly, other than that thenar hypoplasia occasionally occurs in radial polydactyly with triphalangism. In particular, the phenotype and level of duplication associated with thenar dysplasia remains unclear.

Methods

The abductor pollicis brevis (APB) and flexor pollicis brevis (FPB) muscles were visualized using three-dimensional ultrasound and their horizontal geometry was assessed using a biaxial level classification system. Subjects were categorized into three phenotypes according to the developmental condition of the radial thumb. The relationship between the level of distribution of the muscles and the level of the bifurcation of the radial thumb was investigated.

Results

Nineteen patients with radial polydactyly without triphalangism were included. There were ten patients with the non-floating type, three with the floating type, and six with the rudimentary type. All patients with bifurcation at or more distal to the metacarpophalangeal (MCP) joint had normal thenar muscle distribution, but the muscles in patients with bifurcation at or more proximal to the level of the metacarpals were confined, regardless of phenotype. The level of muscle distribution was strongly correlated with the level of the bifurcation of the radial thumb.

Conclusions

These findings suggest that formation of the thenar muscles in the longitudinal direction in radial polydactyly might depend on the level of bifurcation of the radial thumb. The presence of thenar dysplasia even in floating-type or rudimentary-type duplications is of clinical and etiological importance.

Key Words

Radial polydactyly, thenar muscle, dysplasia, ultrasound

Introduction

For radial polydactyly, reconstructive surgery has been widely accepted as a result of increased knowledge about associated skeletal and soft tissue abnormalities[1-4]. However, even though all these anomalies are addressed intraoperatively, complete functional restoration of the thumb remains challenging[5, 6]. In 1982, Miura stated issues remaining after reconstruction for radial polydactyly are adduction contracture of the thumb and thenar dysplasia[7]. Although more than 30 years have passed since Miura's statement, little is known about thenar dysplasia in radial polydactyly apart from the fact that thenar hypoplasia occurs in special cases of radial polydactyly with triphalangism[8].

The objective of this study was to investigate developmental conditions of the thenar muscles in radial polydactyly without triphalangism using three-dimensional (3D) ultrasonography. In particular, the relationship between thenar dysplasia and the level of bifurcation of the radial thumb was evaluated.

Materials and methods

This study began in 2014 after it was approved by the ethics committee of our institution. The patients' families received an explanation about the 3D ultrasound examination and surgery, and written informed consent was obtained. Triphalangeal-thumb polydactyly or polysyndactyly, and a six-fingered hand were not included because these types were known to have distinct pathogenic backgrounds[9, 10].

The 3D ultrasound method was described previously[11]. Briefly, the thenar region was scanned using a linear array transducer (Preirus; Hitachi Aloka Medical Co., Ltd., Tokyo, Japan) in the transverse direction under precise speed regulation at 3 mm/s. During scan, the thumb was placed in the 45-degree radially-abducted position. Consecutive images were acquired at 0.2 mm intervals and processed to reconstruct 3D data using imaging software (ImageJ, U. S. National Institutes of Health (NIH), Bethesda, Maryland, USA). Anatomical identifications were accomplished on the basis of multi-planar observation, initially using coronal sections, followed by sagittal and axial sections. Especially for coronal sections, the outlines of the first metacarpal were memorized to be shown in more superficial sections as anatomical references. (Supplementary figure. 1).

To systematically investigate developmental conditions of the thenar muscles in patients with radial polydactyly of various degrees of severity and phenotypes, subjects were categorized according to the level of bifurcation and the developmental condition of the radial thumb. For bifurcation, nine levels were defined along the phalanges and metacarpal (Fig. 1). For developmental condition of the radial thumb, three phenotypes were defined. The non-floating type had a true articulation or an osseous continuity between the radial and ulnar thumbs. The floating type had a radial thumb connected to the ulnar thumb by fibrous tissues and at least one pair of articulated phalanges in the radial thumb. The rudimentary type had a more underdeveloped radial thumb that is comprised of soft tissues only or contains no more than one osseous component. To analyze horizontal geometry of the abductor pollicis brevis (APB) and flexor pollicis brevis (FPB), a biaxial level classification system was further defined (Fig. 1). Distributions of the insertion and origin of the FPB were classified by levels along the metacarpal axis and levels along the longitudinal axis, respectively. The APB distribution was assessed only on the insertion side by levels along the metacarpal axis. The relationship between the level of muscle distribution and the level of the bifurcation of the radial thumb was analyzed using Pearson's correlation coefficients for samples bifurcating at Levels 1–7 because the levels from Level 1 to Level 7 were aligned regularly.

Results

Consecutive nineteen patients were included in this study. There were no bilateral cases. Additional examinations using nine unaffected hands of infants verified that the APB on the insertion side, FPB on the insertion side, and FPB on the origin side located at Levels 6, 6, and 5, respectively. Scatter plots of the distribution of the thenar muscles are shown in Fig. 2. In all hands with Level 6 or more distal bifurcation, muscle distribution was within the normal range for both APB and FPB regardless of phenotypic differences in the radial thumb (Supplementary figure 2). In contrast, in hands with Level 5 or more proximal bifurcation, the level of muscle distribution was strongly related to the level of the bifurcation, especially for APB and FPB on the insertion side. Notably, even the origin side of the FPB was distributed proximally according to the proximity of the bifurcation of the radial thumb, especially in cases with Level 3 or more proximal bifurcation. Representative cases were presented in Supplementary figure 3-6.

Discussion

In this study, a strong correlation between the level of APB and FPB distribution and the level of bifurcation of the radial thumb was found. Herein, we propose that etiologically, muscle formation in the longitudinal direction is limited by the level of bifurcation. Surprisingly, proximal confinement of the thenar muscles was found even in patients with a metacarpal duplication of the floating or rudimentary type, suggesting that the mechanism we proposed may apply to different phenotypes of radial polydactyly, although the presence of pathogenic factors in the transverse direction is speculated (Fig. 3). These results provide two-fold clinical implications. One is that aberrant APB and FPB insertion can exist even in floating and rudimentary types. The other is that longitudinal underdevelopment of the thenar muscles can influence surgical outcomes. Lourie et al. reported unusual cases showing a zigzag deformity after ligation treatment for an extra pedunculated thumb with at the radial aspect of the metacarpal[12]. Similarly, we experienced APB underdevelopment in cases of a rudimentary extra thumb with metacarpal bifurcation (Supplementary figure 5 and 6). Although APB atrophy observed in the postoperative case (Supplementary figure 5) indicates the possibility that detachment of the APB insertion might have occurred during surgery, APB underdevelopment shown in the non-treated case (Supplementary figure 6) rather suggests that APB hypoplasia originally existed. It is known that APB insufficiency causes joint deformities, thus potential APB hypoplasia might be associated with secondary joint deformities in Wassel type V and VI[13, 14]. Further studies on surgical outcomes for radial polydactyly using assessment systems involving palmar abduction[15, 16] would clarify the clinical importance of the thenar dysplasia observed in this study.

Authors' role/participation in the authorship of the manuscript

SS conducted the project of this study and performed all examinations. SS, MM and MU analyzed the data.

All the authors contributed to the writing of the paper.

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Figure legends

Fig. 1

The biaxial level classification system and its application

Seven levels are defined for the thenar region along two anatomical axes. In addition to the seven levels, two levels are defined distally in order to classify the level of bifurcation of the radial thumb (top). An ultrasound coronal image of a non-floating-type duplication with Level 5 bifurcation (bottom). In this case, the APB on the insertion side, FPB on the insertion side, and FPB on the origin side all distribute at Levels 5. APB, abductor pollicis brevis; FPB, flexor pollicis brevis; CMC, carpometacarpal; MCP, metacarpophalangeal; IP, interphalangeal.

Fig. 2

Scatter plots and lines of best fit showing relationships between distributions of APB and FPB versus the level of bifurcation. Pearson's correlation coefficients for patients with bifurcation at the level of the metacarpals are also shown. Yellow bars indicate normal muscle distributions. APB, abductor pollicis brevis; FPB, flexor pollicis brevis.

Fig. 3

Schematic illustration showing a proposed mechanism of thenar dysplasia associated with radial polydactyly.

Supplementary figure 1

Ultrasound images showing normal thenar anatomy in a 1-year-old infant. **A**, coronal; **B**, sagittal; **C**, axial sections. Lines with numbers indicate the location of the images. Green lines indicate the outline of the first metacarpal. Yellow lines show the outline of the thenar muscles. FPL is used as a landmark that separates the superficial and deep part of the muscles (Red circles). APB, abductor pollicis brevis; AD, adductor pollicis; FPB, flexor pollicis brevis; FPL, flexor pollicis longus; OP, opponens pollicis; CT, carpal tunnel; CMC, carpometacarpal joint; MCP, metacarpophalangeal joint; 1st MC, first metacarpal; d, depth from the skin.

Supplementary figure 2

Photos, radiographs, and coronal ultrasound images of patients with radial polydactyly bifurcating at Level 7 or more distally. Yellow lines show the outline of the superficial thenar muscles. Note that all the muscles distribute distally to the level of the metacarpophalangeal joint (red arrowheads). Abbreviations are as indicated in Fig. 2.

Supplementary figure 3

A 6-month-old patient with a non-floating-type duplication with Level 1 bifurcation. **A**, The thenar eminence seems hypoplastic (right). Note that the proximal thenar crease is lacking (arrow). **B**, Radiograph. **C**, Coronal; **D**, axial; and **E**, sagittal ultrasound sections. The thenar muscles are confined proximally to Level 2 (red arrowheads). The distal portion of the muscles is located in the direction of the MCP joint of the radial thumb (yellow arrowheads). Yellow lines show the outline of the thenar muscles. Red circles indicate the FPL in a cross-sectional view. Abbreviations are as indicated in Fig. 2. L and R indicate left and right, respectively.

Supplementary figure 4

A 1-year-old patient with a floating-type extra thumb bifurcating at Level 3. **A**, The thenar eminence seems slightly hypoplastic (right, arrow). **B**, Radiograph. **C**, Coronal; **D**, axial; and **E**, sagittal ultrasound sections. The thenar muscles are confined proximally to Level 3 (red arrowheads). Note that there is no muscle distribution in the direction of the MCP joint of the ulnar thumb (yellow arrowheads). Yellow lines show the outline the thenar muscles. Red circles indicate the FPL in a cross-sectional view. Abbreviations used are indicated in Fig. 2. L and R indicate left and right, respectively.

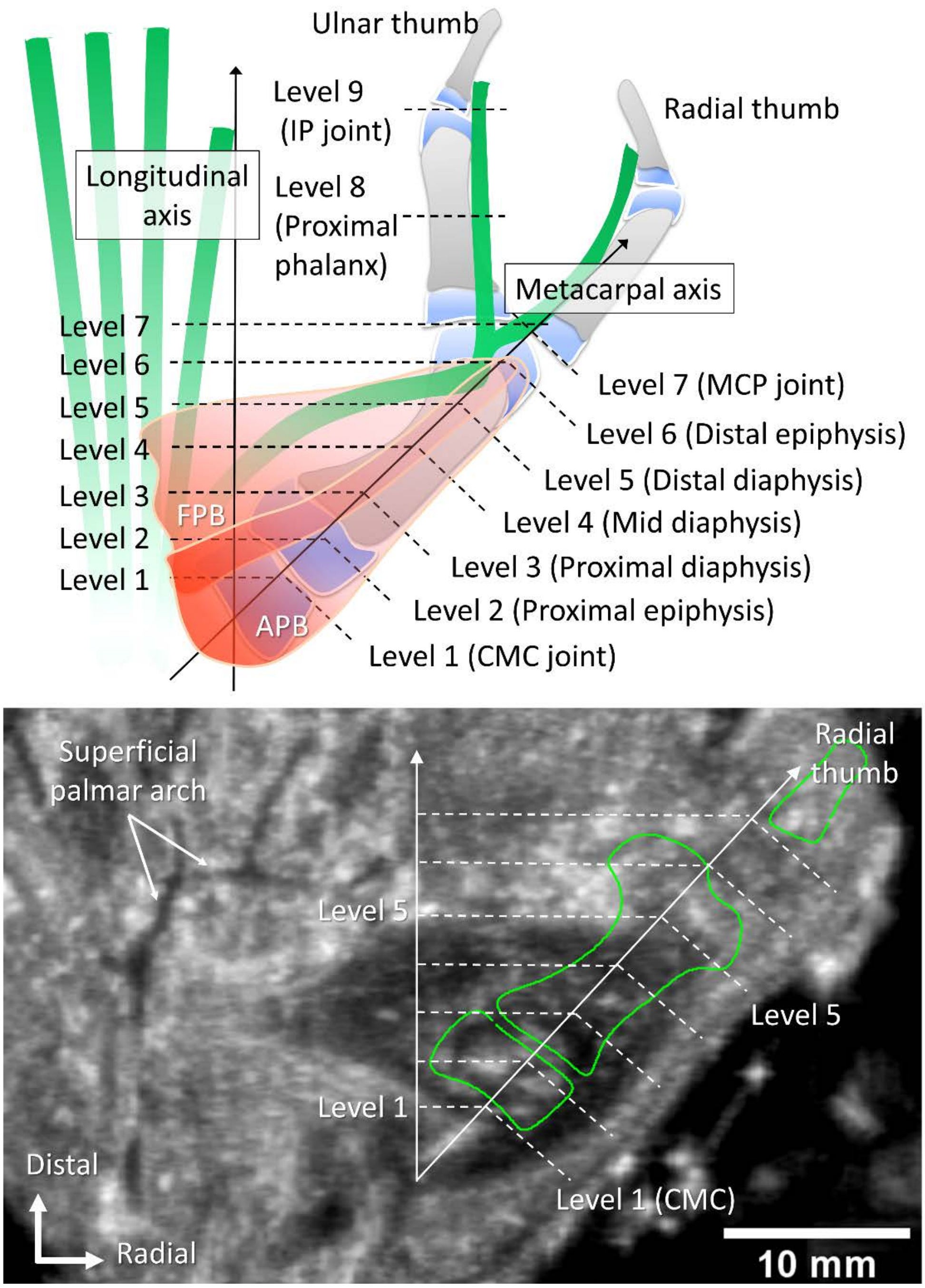
Supplementary figure 5

A 4-year-old patient who underwent ablative surgery for rudimentary-type duplication with Level 4 bifurcation four years previously. **A**, The thenar eminence seems underdeveloped radially (arrows). Palmar abduction of the affected thumb is insufficient (bottom, right). **B**, Preoperative appearance (left) and radiograph (right). **C**, Coronal; **D**, sagittal; and **E**, axial ultrasound sections. Axial sections at the level of the epiphyseal line demonstrate APB atrophy (red arrowheads) although FPB is shown is less underdeveloped (yellow arrowheads). Red circles indicate the FPL in a cross-sectional view. Abbreviations used are indicated in Fig. 2. L and R indicate left and right, respectively.

Supplementary figure 6

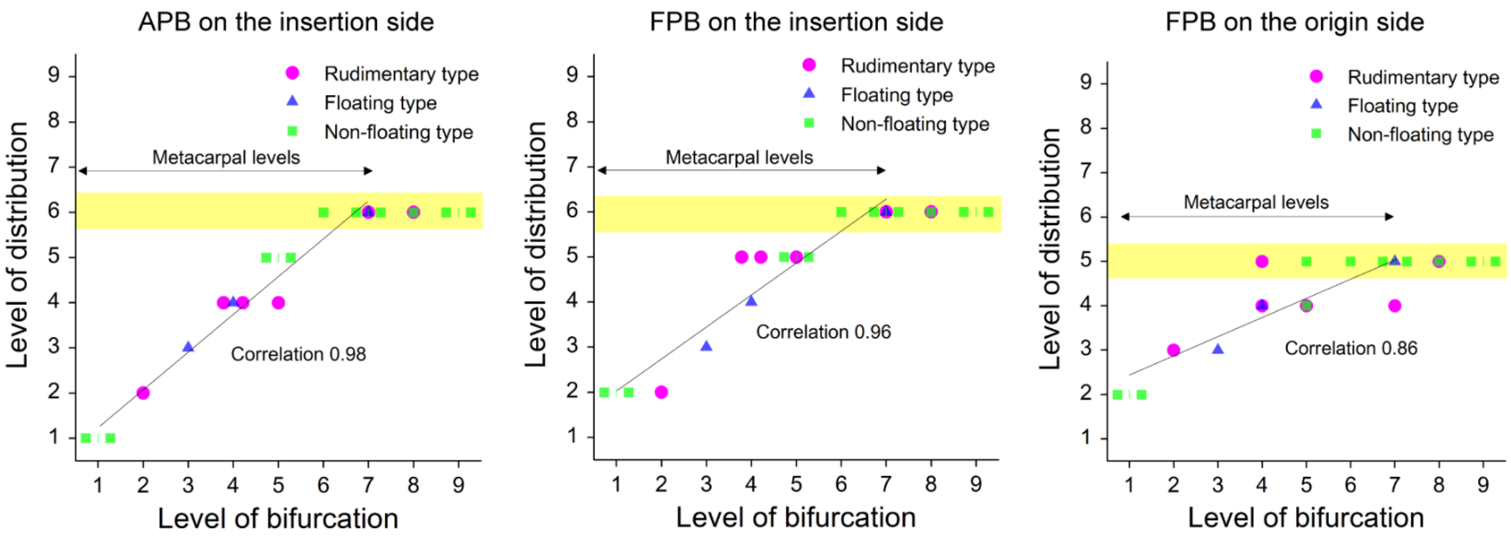
A 14-year-old patient with untreated rudimentary-type duplication with Level 2 bifurcation**.** **A**, The thenar eminence seems underdeveloped (top, black arrow). The involved thumb shows a deformity in the flexion position (bottom, white arrow). **B**, Radiograph. **C**, Coronal; **D**, sagittal; and **E**, axial ultrasound sections. The thenar muscles are confined proximally to Level 2 (red arrowheads). Axial sections at the level of the epiphysis demonstrate muscle atrophy (yellow arrowheads). Yellow lines show the outline of the thenar muscles. Red circles indicate the FPL in a cross-sectional view. Abbreviations used are indicated in Fig. 2. L and R indicate left and right, respectively.

Fig. 1



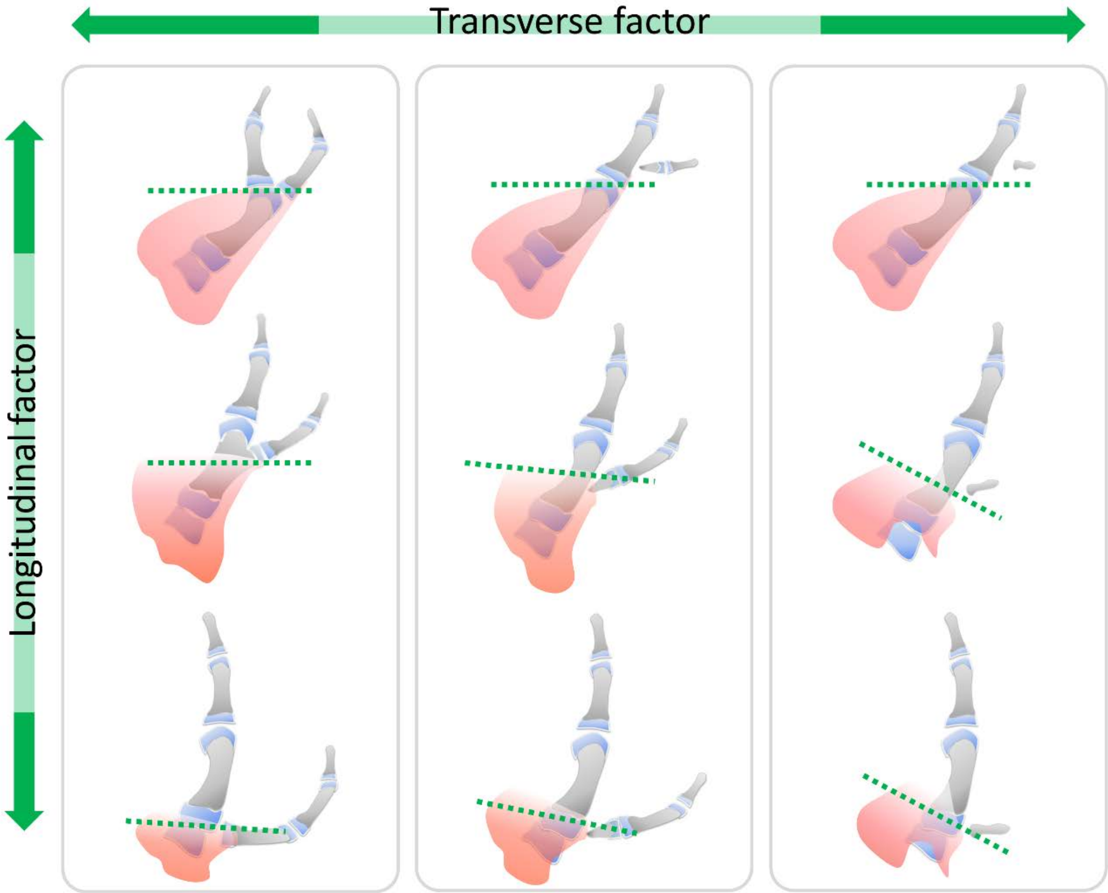
Ulnar thumb
Radial thumb
Level 9
(IP joint)
Level 8
(Proximal
phalanx)
Longitudinal
axis
Metacarpal axis
Level 7
Level 6
Level 5
Level 4
Level 3
Level 2
Level 1
FPB
APB
Level 7 (MCP joint)
Level 6 (Distal epiphysis)
Level 5 (Distal diaphysis)
Level 4 (Mid diaphysis)
Level 3 (Proximal diaphysis)
Level 2 (Proximal epiphysis)
Level 1 (CMC joint)
Superficial
palmar arch
Radial
thumb
Level 5
Level 5
Level 1
Level 1 (CMC)
Distal
Radial
10 mm

Fig. 2

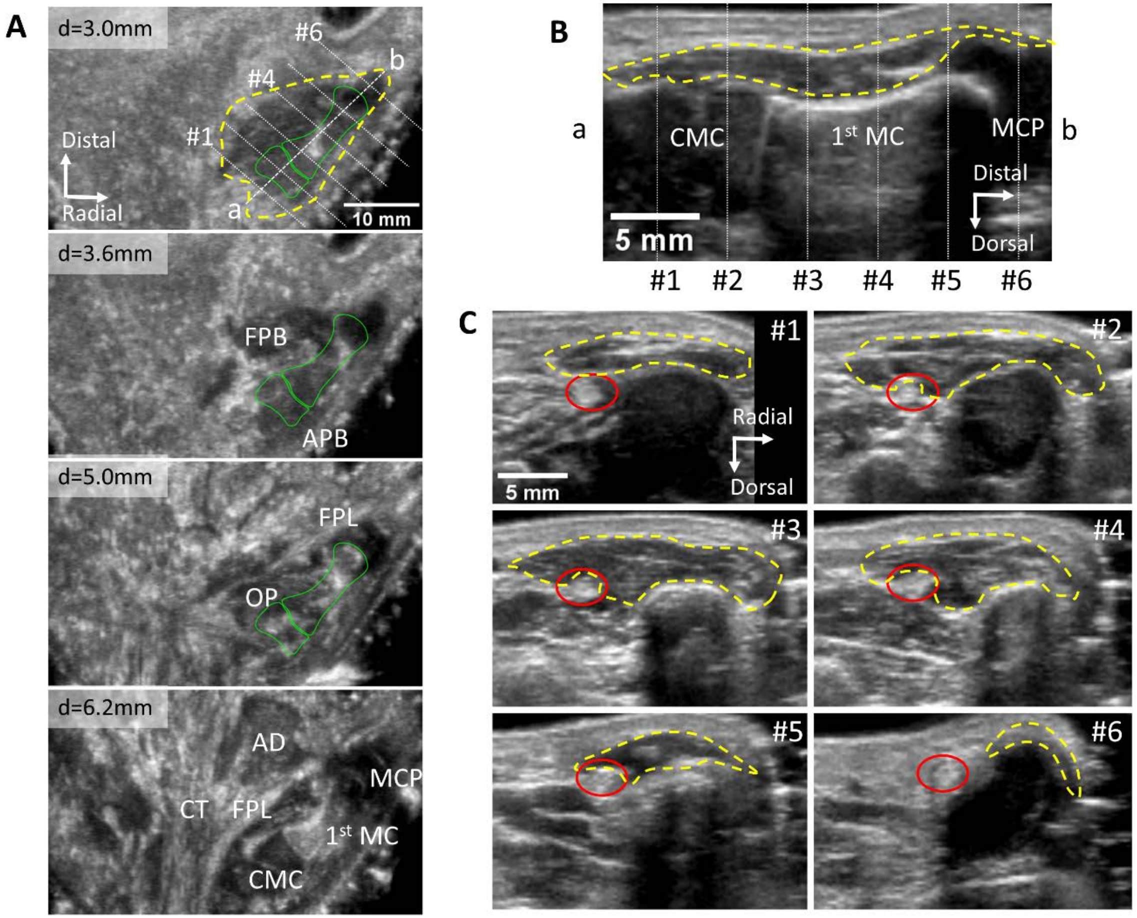


APB on the insertion side
FPB on the insertion side
FPB on the origin side
Rudimentary type
Floating type
Non-floating type
Metacarpal levels
Correlation 0.98
Correlation 0.96
Correlation 0.86
Level of distribution
Level of bifurcation

Fig. 3



Supplementary figure 1



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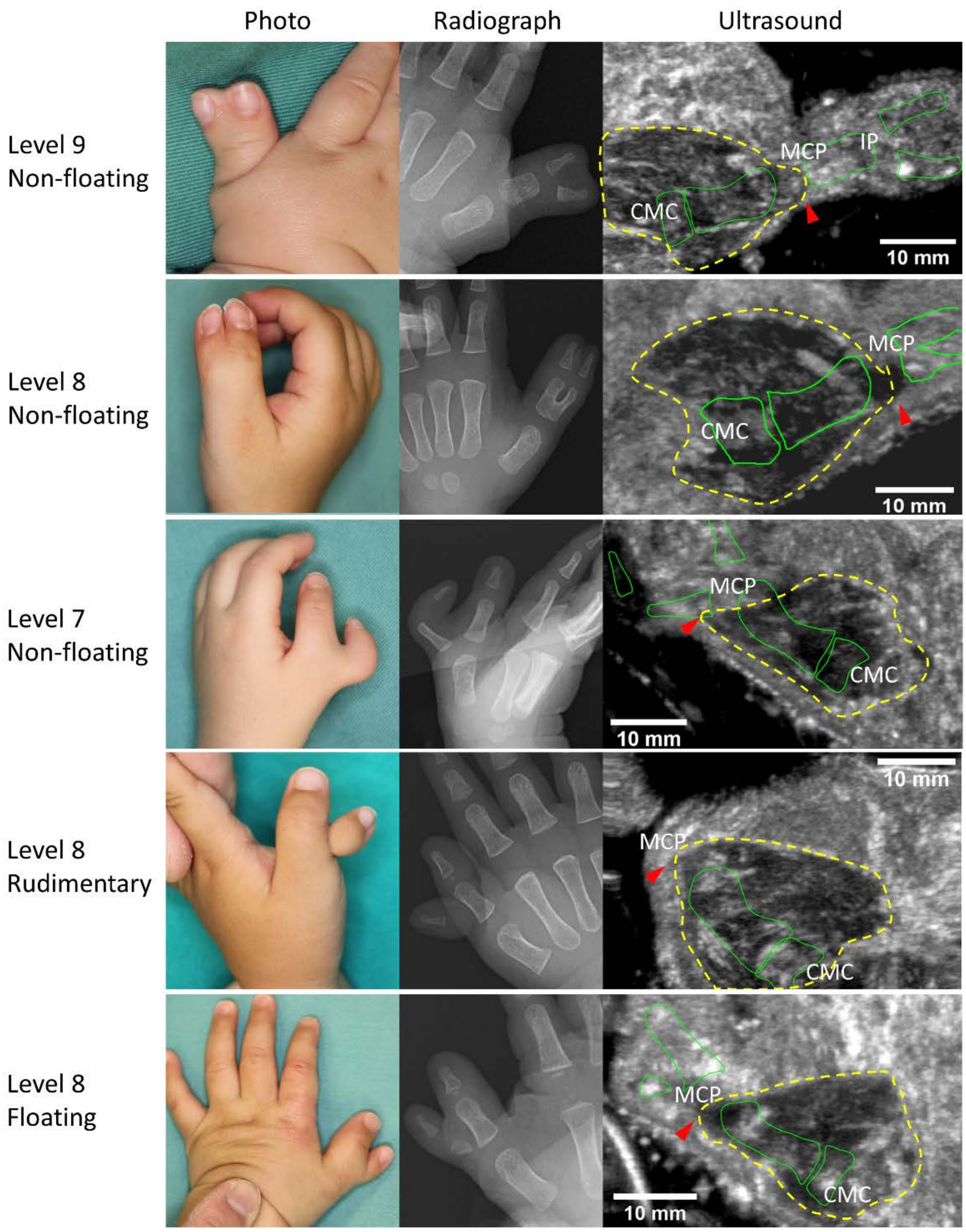

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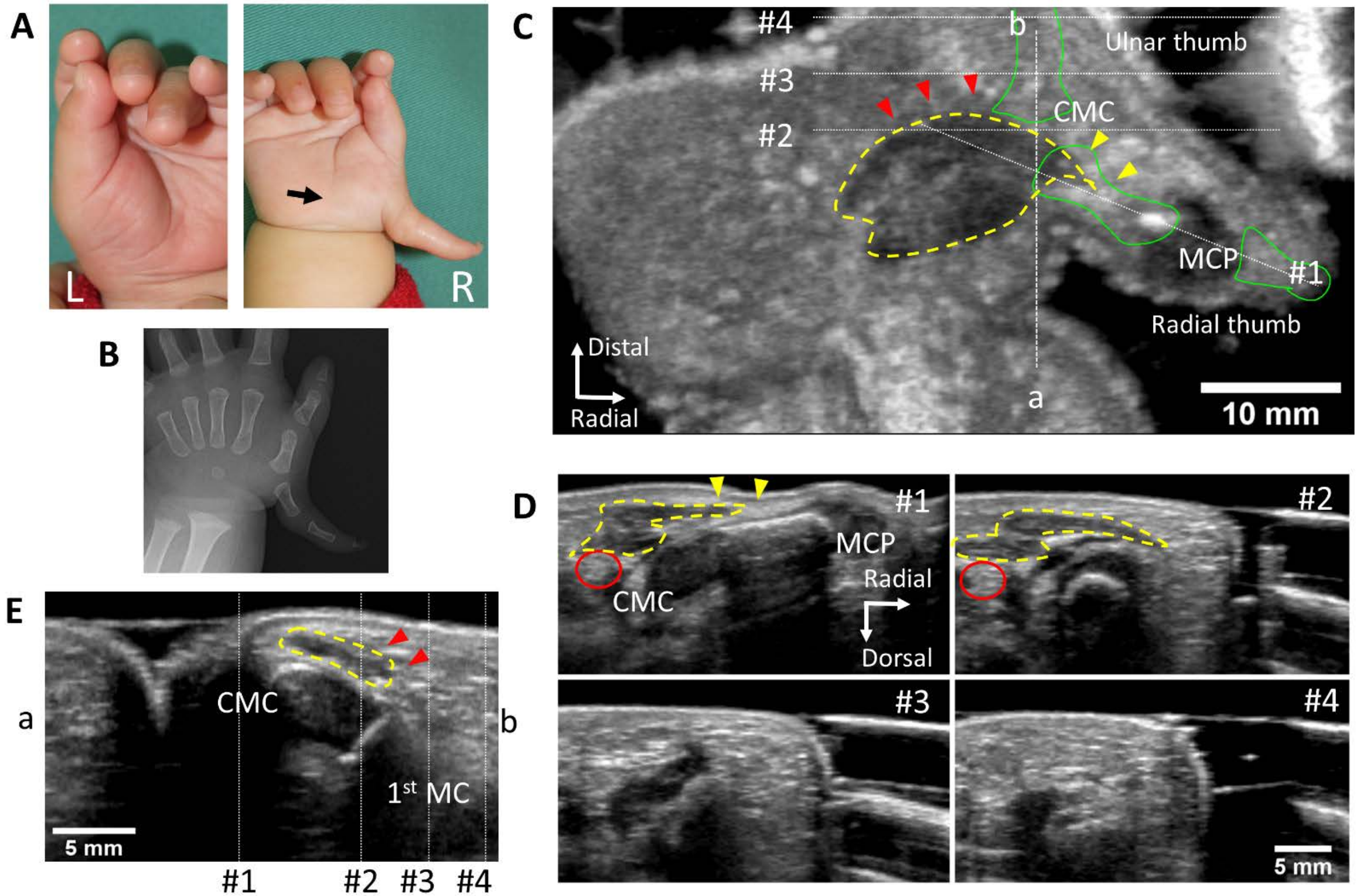

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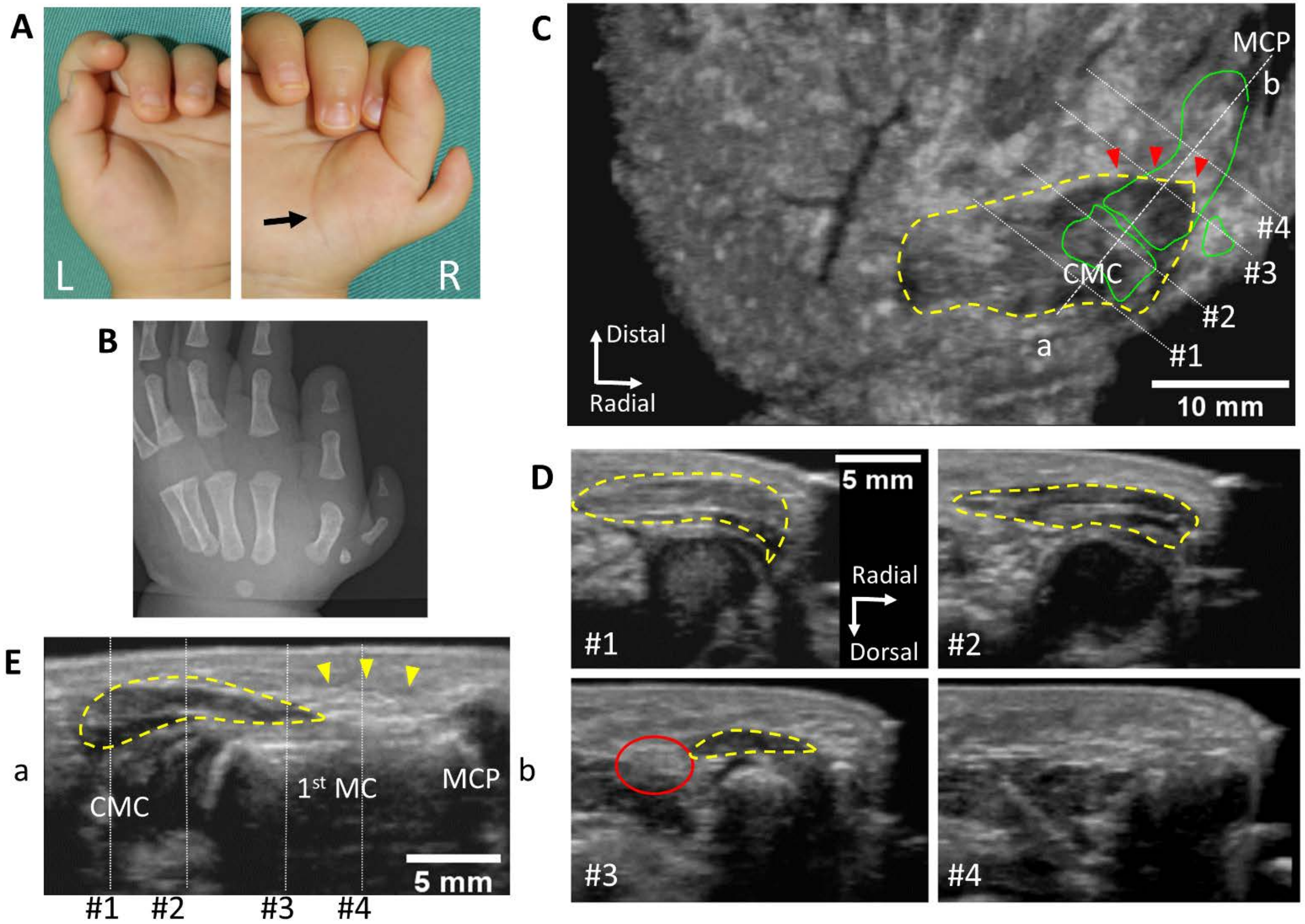
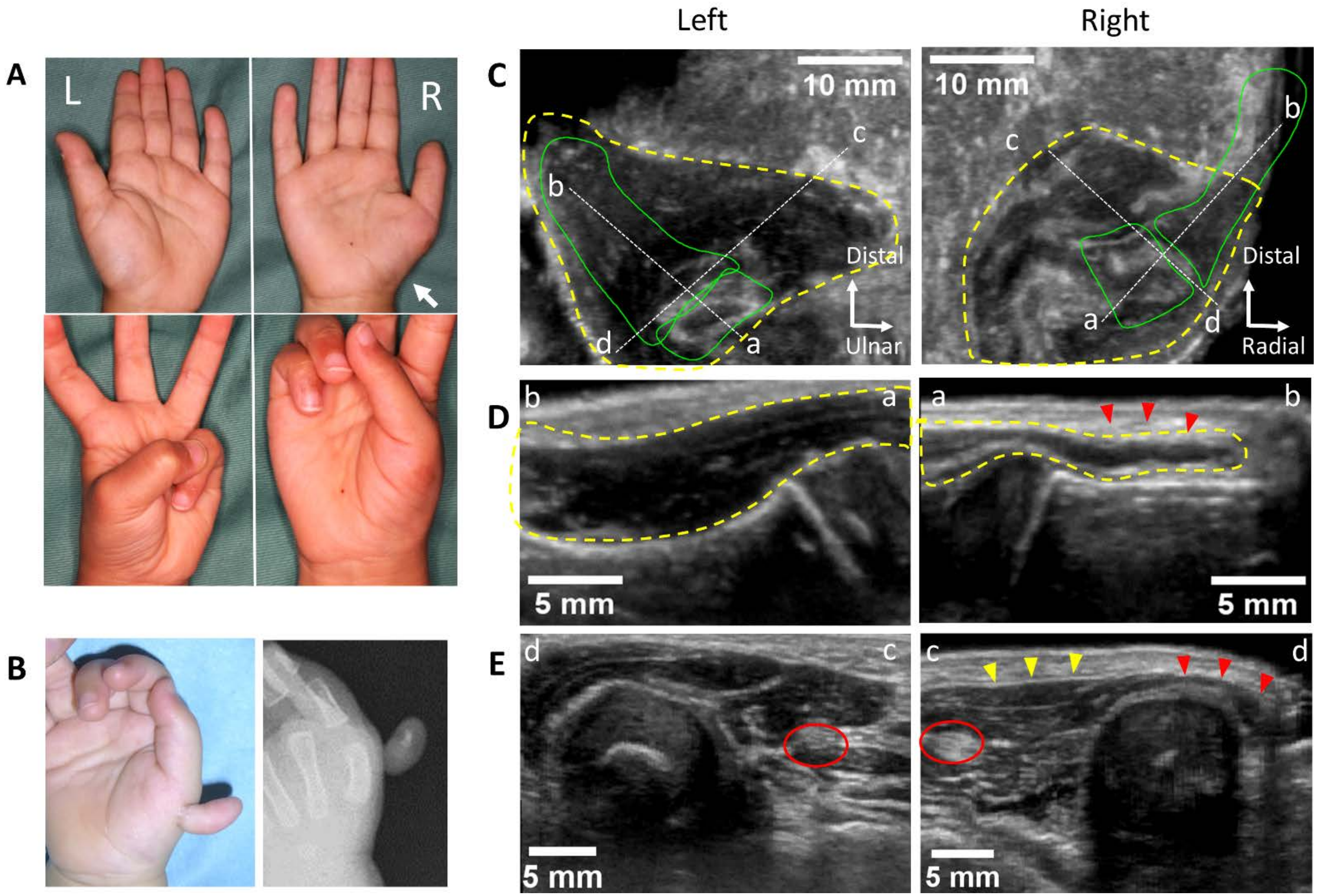


Supplementary figure 5



Supplementary figure 6