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Received: 3 January 2021 | Revised: 30 January 2021 | Accepted: 19 February 2021

DOI: XXX

RESEARCH ARTICLE

# A case of brachymetacarpia in a skeleton from a Mudejar cemetery from Spain (13th–14th century BC)

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## Abstract

Brachymetacarpia, a form of brachydactyly, is one of the so-called rare diseases because of its low prevalence. Although it is a well-known malformation today, which occasionally requires surgical correction, it is not, or hardly, reported in the palaeopathological literature. The case presented here includes an individual exhumed from the Mudejar cemetery in Uceda (Guadalajara, Spain) dated between the 13th and 14th centuries. It was in an acceptable state of preservation, except for the skull, missing except for the mandible. Its sex was determined as female and the age as a young adult. On examining the hands, the short length of both the 4th and 5th metacarpals and the shortening of the distal phalanx of one of the thumbs were noteworthy. No anomaly was observed in the bones of the feet, which were only partially recovered. Due to the characteristics of the shortening and bones affected, it was considered that the case probably corresponded to type E of brachydactyly in the Bell and Temtamy classifications and to subtype E2 in the Hertzog classification. No data were found in the bones or teeth, suggesting their inclusion in any of the multiple clinical syndromes with this abnormality.

**KEYWORDS**

13th–14th century BC, bioarcheology, brachydactyly, brachymetacarpia, Mudejar, rare diseases

## 1 | INTRODUCTION

Brachydactyly is a term derived from the Greek *brachy* and *dactylos* ("short fingers"). One of the first classifications was proposed in 1932 by McArthur and McCullogh, who differentiated between brachydactyly present at the time of birth and those that developed after it (Gupta & Scheker, 2000). The most widely used classification has been that of Bell, modified by Temtamy and McKusick (1978), in which brachydactyly is the second group of the 10 that comprise malformations of the hand (S.A. Temtamy, 1985).

Brachydactyly can appear in isolation or as part of numerous syndromes and diseases (Flatt, 1994). S.A. Temtamy and Aglan (2008) divide the isolated forms into type A brachydactyly with five subtypes, type B, type C, type D, type E, and brachymetatarsus IV; Sugarman brachydactyly; and Kirner deformity. It is a rare disease, except for types A3 and D, which have a prevalence of around 2% (S.A. Temtamy & Aglan, 2008).

In the present work, a case of brachydactyly is exposed from an individual exhumed in a Medieval cemetery, and its possible etiology is discussed. The importance of the case lies in the infrequent of the disease with low prevalence in the current clinical environment and, above all, after a review of the literature, the low number of cases of

There are no previous work should be referred to specifically and referenced in the new paper. There are no previously published abstracts for meeting presentations that contain partial or similar material to the new paper. There are no disclosures by the authors. The authors give permission for the publication of the manuscript. The authors declare they have no conflict of interest.

 

brachydactyly described in the palaeopathological literature (Cybulski, 1988; Dupras et al., 2010; Kozieradzka-Ogunmakin, 2011; Panzer et al., 2010).

## 2 | MATERIAL AND METHODS

The skeletal remains of 116 nearly complete individuals were exhumed from the Mudejar cemetery of Uceda (Guadalajara, Spain), dated between the 13th and 14th centuries (Figure 1a).

Nearly all of the individuals had, as a common denominator, a burial rite that has its roots in the classical Andalusian model, although it presents (in these last moments and in these territories) significant features of an orthodox relaxation of the rite. Although flexion in the bodies is not common; it is easy to identify bodies in simple fossa (NE–SW) some in the right lateral decubitus position and others in the supine position, with their head turned toward Mecca. One of the bodies, from Tomb (UE) 118, is the object of this study (Ramírez-González & Dorado, 2020; Figure 1b, red square and arrow).

The degree of bone preservation was classified according to the scale proposed by White (2008). This scale includes three grades, with grade one corresponding to the best-preserved bone.

The state of preservation has been calculated, according to Walker et al. (1988) and Safont (2003), through the partial bone preservation index (IP3; includes long bones, pelvic and scapular girdles, cranium, maxilla, and mandible). IP3 = ∑ number of parts conserved/22.

Age was estimated based on the state of epiphyseal fusion (Scheuer & Black, 2000), dental mineralization (Smith, 1991), and dental wear (Brothwell, 1987; Lovejoy, 1985). Sex was established according to the morphological characters of the pelvis (Buikstra & Ubelaker, 1994). Height has been estimated using the equations of Mendonça for the femur (2003), which are based on a contemporary Mediterranean population.

The biometry was collected according to the recommendations of Buikstra and Ubelaker (1994), using a vernier caliper (precision 0.02 mm). The radiological images were obtained with Polyrad SE digital equipment. For the theoretical calculation of the normal length of the metacarpals (MTC), the equations proposed by Aydinlioglu et al. (1998) have been used, calculating the difference between the length obtained and the one that would theoretically correspond to it according to its equations. The length of the 2nd MTC has been used as a reference for the calculation in each hand, because the 3rd right MTC shows signs of an old diaphyseal fracture.

## 3 | RESULTS

The analyzed skeleton corresponds to a female individual, with an estimated age between 25 and 35 years and approximately 156.7 cm height.

The recovered skeleton was quite complete, with an IP3 index of 20/22, although the entire cranium is missing and only part of the mandible remains. The preservation grade is high, White Grade 1.

In the mandible, all teeth were kept in good conditions, including 38 and 48, although there is post-mortem loss of 41–43 and 31–32. The upper molar (26) was recovered. No dental pathology was noted.

The capitate, pisiform, and trapezoid bones have been recovered from the left carpus, whereas the trapezoid and pisiform bones are missing from the right carpus. There are no bone alterations in their size or morphology. All complete metacarpals were well preserved (Figures 2 and 3).

The short length and widening of both 5th metacarpals (MTCs) and the shortening, although to a lesser degree, of both 4th MTCs are shown in Figure 4b.

Osteometric measurements are listed in Table 1.

Applying the mathematical relationship proposed by Aydinlioglu et al. (1998), the result quantifies the shortening of both 5th MTCs at

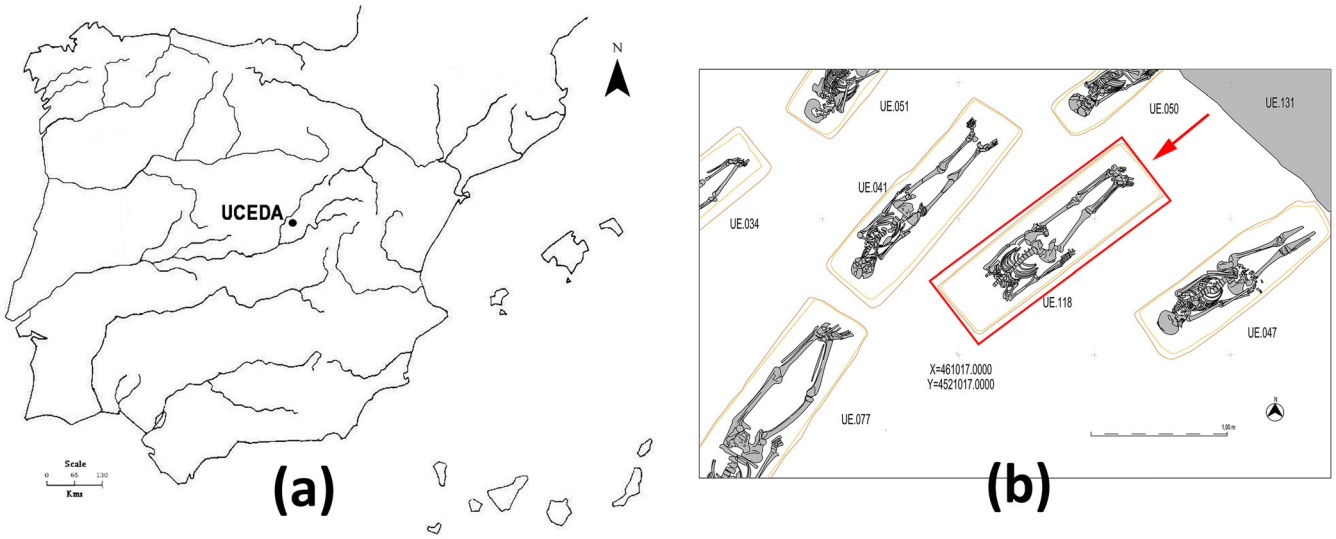


**FIGURE 1** (a) Location of Uceda (Guadalajara, Spain). (b) Planimetry of the grave. UE118 (red arrow)


**FIGURE 2** Bones recovered from the right (R) and left (L) hands. Palmar view

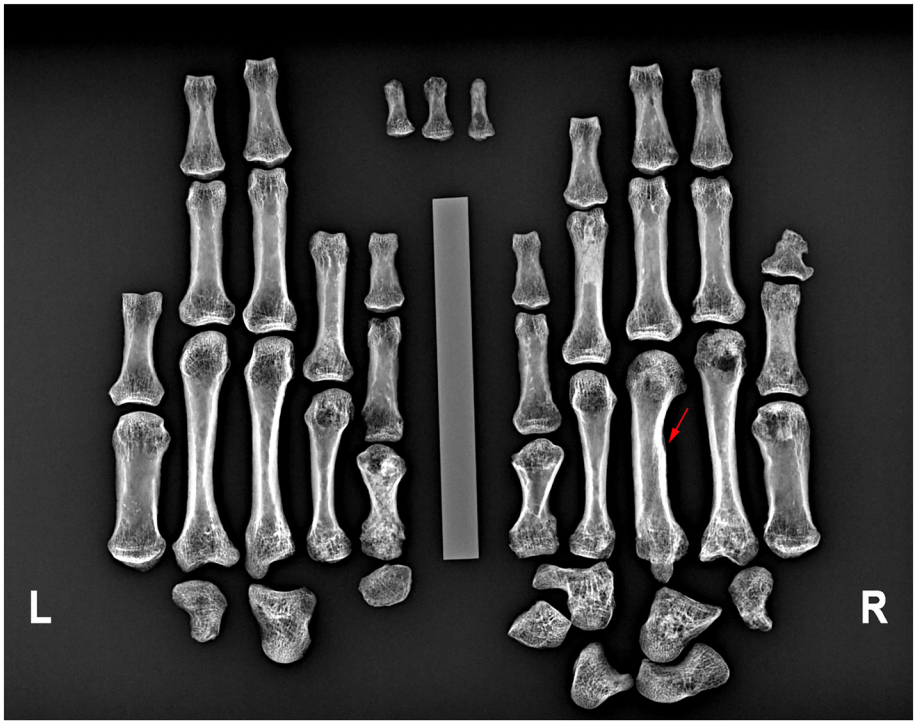


**FIGURE 3** AP X-rays of hands. MTC shortening and 3rd MTC fracture (red arrow)

18.4 and 17.3 mm, whereas in the 4th left and right MTCs, it was 6.9 and 2.1 mm, respectively (Table 2).

All proximal phalanges of both hands and seven middle phalanges were preserved. There were three normal-appearing distal phalanges, measuring 17.1, 16.5, and 15.6 mm, respectively. However, the distal phalanx of the right thumb was recovered, with a reduced length of 12.8 mm, highly deformed and with a widened base (Figure 4a). The contralateral phalanx of the left thumb was not recovered.

The left foot partially conserves the tarsus (calcaneum, cuboid, navicular, and medial cuneiform) with normal appearance, five metatarsals (MTTs) without heads, except the 5th, as well as the proximal phalanx of the great toe. The right foot conserves, equally unaltered,

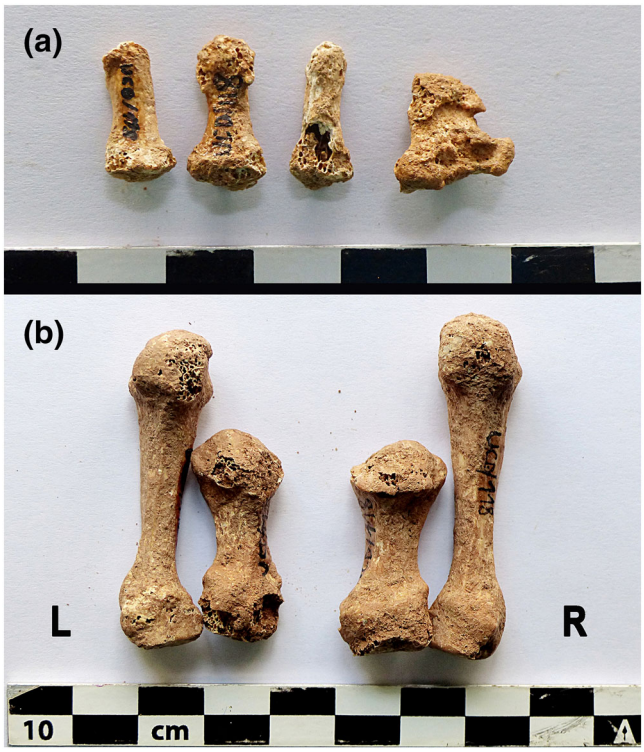


**FIGURE 4** (A) Distal phalanges. Short and deformed distal phalanx of right (R) thumb. (B) 4th and 5th metacarpals of the right (R) and left (L) hands

**TABLE 1** MTC (metacarpal) measurements

| | | R | L |
|---|---|---|---|
| **1st MTC** | **Max. Length** | **43.5** | **43.8** |
| | A-P diameter | 8.1 | 7.8 |
| | M-L diameter | 12.3 | 12.4 |
| **2nd MTC** | **Max. Length** | **63** | **63.7** |
| | A-P diameter | 8.4 | 8.3 |
| | M-L diameter | 7.5 | 7.8 |
| **3rd MTC** | **Max. Length** | **59** | **62** |
| | A-P diameter | 10 | 8.2 |
| | M-L diameter | 9 | 7.5 |
| **4th MTC** | **Max. Length** | **52.2** | **48** |
| | A-P diameter | 7.3 | 7.2 |
| | M-L diameter | 6.5 | 6.9 |
| **5th MTC** | **Max. Length** | **32.7** | **32.1** |
| | A-P diameter | 8.6 | 8.3 |
| | M-L diameter | 7.7 | 7.6 |

*Note*: Measurements in mm.
Abbreviations: A-P, antero-posterior; M-L, mid-lateral.

part of the tarsus (calcaneum, cuboid, and navicular fragment), five MTTs without heads, and the proximal phalanx of the great toe. Likewise, two proximal phalanges with a normal appearance were recovered.

## 4 | DISCUSSION

The skeleton studied corresponds to a young female between 25 and 35 years old. Except for the alterations in the hands, no other bone alterations were observed in the rest of the skeleton, although practically the entire cranium is missing.

The marked shortening of both 5th MTCs (at 18.4 and 17.3 mm) with respect to the expected theoretical length is noteworthy out. Shortening is also observed in both 4th MTCs (at 6.9 mm on the left and at 2.1 mm on the right), slight in the right, although (in both cases) less than that observed in the 5th MTCs.

The right distal phalanx of the thumb appears widened and deformed, with decreased length compared to the rest of the preserved phalanges.

No morphological alterations were observed in the feet.

Bozan et al. (2006) collected, among the current cases treated surgically, the following eight cases of brachymetacarpia in which the lengths of the 4th affected MTCs were between 27 and 37 mm, therefore less than in the case we present; the measurements of the 5th MTC were between 28 and 32 mm, which were similar to those in our case (Table 1). Similarly, Lam et al. (2019) collected four cases in which the lengths of the 4th MTCs, between 32 and 36 mm, were also shorter than those in the case we present, and that of the 5th MTCs, between 34 and 35 mm, were similar to the present case.

The case presented, involving the bilateral metacarpals and the distal phalanx of a thumb, would correspond to type E brachydactyly in the Bell and Temtamy classifications (A. Pereda et al., 2013) and to the E2 subtype in the Hertzog (1968) classification.

Type E brachydactyly is due to shortening of the MTCs and occasionally the MTTs, with normal phalanges, although the distal phalanges may be short (S.A. Temtamy & Aglan, 2008).

In the 10th version of the Nosology and Classification of Genetic Skeletal Disorders, which includes 461 diseases, type E brachydactyly appears as an autosomal dominant hereditary disease linked to the PTHLH and HOXD13 genes (Mortier et al., 2019; A. Pereda et al., 2013). In the database of rare diseases (ORPHA, 2020), it appears with code 93387, as dysostosis disease with anomalies of the extremities, without extraskeletal manifestations.

Hertzog (1968) distinguishes three subtypes of type E brachydactyly. The E1 subtype presents with shortening limited to the 4th MTCs or MTTs, without involvement of the phalanges; in the E2 subtype, shortening affects MTCs in a variable combination of affected MTCs and middle and distal phalanges (distal phalanges of the 1st and 3rd fingers and middle phalanges of the 2nd and 5th fingers); in the E3 subtype, there is variable shortening of the MTCs, without alteration of the phalanges.

Regarding its etiology, in an increasing order of frequency, a possible acquired origin must be considered, its inclusion as part of a syndrome or that it is an isolated malformation.

The least frequent acquired origin has been described in relation to poliomyelitis, trauma, and neurotrophic disorders or surgery (Aski & Patil, 2017). In the case presented, this etiology has been ruled out, because no harmful signs were observed in the shortened MTCs

**TABLE 2** Difference between theoretical and actual lengths (according to Aydinlioglu et al., 1998)

| | | Actual length | Theoretical length | Shortening |
|---|---|---|---|---|
| Right hand | 4th MTC | 52.2 | 54.3 | 2.1 |
| | 5th MTC | 32.7 | 50.0 | 17.3 |
| Left hand | 4th MTC | 48.0 | 54.9 | 6.9 |
| | 5th MTC | 32.1 | 50.5 | 18.4 |

*Note*: Measurements in mm.

(Figure 3) and the bone involvement was multiple and bilateral (Kaziz, 2016; Suresh et al., 2009).

Type E brachydactyly can appear associated with numerous syndromes and diseases. Due to its frequency and the preferential involvement of the 4th and 5th MTCs, brachydactyly must first be ruled out as part of the Aldrich phenotype, which includes, in addition to brachydactyly, pyknic type, rounded face, short stature, and ectopic ossifications, which is associated with pseudohypoparathyroidism and other related disorders (Martos-Moreno et al., 2019; Goswami et al., 2009; S.A. Temtamy & Aglan, 2008; Valizadeh et al., 2013).

However, there are many other syndromes and diseases in which the presence of type E brachydactyly has been described, such as Gorlin-Goltz syndrome (Brechard et al., 2020), Langer-Giedion syndrome (N. Pereda et al., 2012), KBG syndrome (Libianto et al., 2019), brachydactyly mental retardation syndrome (Mahendhar et al., 2018), congenital constriction band syndrome (Satake et al., 2012), Turner syndrome (Page et al., 2017), microcephalic osteodysplastic primordial dwarfism (Bober & Jackson, 2017), and type E brachydactyly with associated hypertension (S.A. Temtamy & Aglan, 2008), lipid disorders (Page et al., 2017), and insulin resistance (Hyari et al., 2006). Likewise, it has been described more frequently in cases of short stature, without other anomalies (Sükür et al., 1997).

In this case, there are obvious limitations in the study, because only the skeleton was available, with most part of the cranium missing, and it was not possible to perform clinical, biochemical, or genetic examinations, which are essential for an adequate etiological diagnosis. However, in the preserved material, no alterations in the skeleton were found that suggest its inclusion as part of a syndrome, such as dental anomalies, bone malformations of the hip, alterations in the length of the radius, cone-shaped epiphyses, and phalanges or divergence between the lengths of the femurs. On the other hand, it is important to note that, in the syndromes that occur with brachydactyly, short stature is common whereas in the analyzed body, the estimated height was approximately 156.7 cm, with the average height in the females excavated from the necropolis being 153.55 cm.

For all these reasons, the possibility that the case may correspond to an isolated malformation of the fingers and toes (i.e., type E brachydactyly without extraskeletal manifestations) must be considered, with the stated limitations.

In relation to the clinical implications of brachymetacarpia, it does not generally affect the functionality of the hand, except for some restrictions in flexion of the metacarpophalangeal joint, not requiring treatment (Saito et al., 2001). The severe shortening that affects the transverse metacarpal arch, however, can lead to weakness in grasping or difficulty in making a hard fist (Lam et al., 2019). In the brachymetacarpia associated with syndromes, the symptomatology will be conditioned by the type of syndrome (S.A. Temtamy & Aglan, 2008).

Currently, surgical treatment is generally carried out for cosmetic reasons (Aski & Patil, 2017; Suresh et al., 2009; Volpi & Fragomen, 2011), because individuals do not show a disability in their daily life, beyond the possible esthetic drawbacks.

Brachydactyly is described more frequently associated with female (Aski & Patil, 2017; Kirkos, 1999; Lam et al., 2019). Cases of type E brachymetacarpia have been described by different authors, but only in the clinical context of living patients (Aski & Patil, 2017; Cohn & Shall, 1986; Kirkos, 1999). However, after reviewing the literature, there are few cases of brachymetacarpia in the palaeopathological literature. Cybulski (1988) reports the presence of brachydactyly in up to eight skeletons from prehistoric burials, Dupras et al. (2010) and Kozieradzka-Ogunmakin (2011) describe such alteration in skeletons from Ancient Egypt, whereas Panzer et al. (2010) describe a case with small stature, mesomelic limb shortening of upper extremities, and brachydactyly of both hands in a mummy in the Capuchin Catacombs of Palermo in Italy.

In our opinion, there are several reasons, other than its rare occurrence, for this lack of palaeopathological references. One reason is the fragility of the bones of the hands and feet before taphonomic agents, together with their small size, make their complete recovery in archeological records infrequent. Another reason is the bone transfers to a secondary burial resulting from a reduction of remains, as it is normal that small bones are not included, which are thus lost for further studies. Also when only a few metacarpals are recovered, what is often, it makes diagnosis difficult because the rest is not available for comparison (Cybulski, 1988). Finally, some affected individuals may have gone unnoticed on palaeopathological examinations.

Although brachydactyly has a low prevalence, with the case presented, we hope that it will alert researchers to its existence when faced with the study of a skeleton of archeological origin.

## AKNOWLEDGMENTS

This work is part of the doctoral thesis that the first author is completing at the University of Granada (Spain). We would like to thank Dr. Miguel Botella, Full Professor, for his collaboration.

## DATA AVAILABILITY STATEMENT

Data sharing not applicable to this article, as no datasets were generated or analyzed during the current study.

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**How to cite this article:** Dorado E, Herrerín J, Ramírez I, Parro L, Carrillo MF, Murillo J. A case of brachymetacarpia in a skeleton from a Mudejar cemetery from Spain (13th–14th century BC). *Int J Osteoarchaeol*. 2021; XX https://doi.orgXX