International Journal of Paleopathology 33 (2021) 146–157

Contents lists available at ScienceDirect

International Journal of Paleopathology

journal homepage: www.elsevier.com/locate/ijpp

Three cases of brachydactyly type E from two commingled tombs at the Late Intermediate period - Late Horizon site of Marcajirca, Ancash, Peru

Anne R. Titelbaum [a,*], Samantha Fresh [b], Bronwyn E. McNeil [c], Bebel Ibarra Asencios [d]

[a] Basic Medical Sciences, University of Arizona College of Medicine – Phoenix, Phoenix, AZ, 85004, USA
[b] Department of Sociology and Anthropology, College of Charleston, Charleston, SC, 29424, USA
[c] Science World British Columbia, BC, V6A 3Z7, Canada
[d] Department of Anthropology, Tulane University, New Orleans, LA, 70118, USA

ARTICLE INFO

Keywords:
Rare diseases
Developmental anomalies
Brachymetacarpia
Brachymetatarsia
Albright's hereditary osteodystrophy
South America

ABSTRACT

Objective: Here we present the first known examples of brachydactyly from an Andean archaeological context by describing the affected bones, presenting a differential diagnosis, and discussing the cultural implications of there being shortened metapodials in multiple tombs.
Materials: 3232 well-preserved tubular bones representing an MNI of 250 human adults.
Methods: Each bone was visually inspected. Measurements were taken with an osteometric board, sliding calipers, and a flexible tape measure.
Results: Of 1210 metapodials excavated from eight burial contexts, ten were atypically short.
Conclusions: The ten shortened elements represent an MNI of three individuals with brachydactyly from two tombs. The presence of at least two individuals with brachydactyly in one tomb adds support to a previous suggestion that tombs were used for familial interment. It is plausible that the third individual from a different tomb was related to the other two, and the different burial contexts may reflect postmarital practices.
Significance: These cases offer insight into tomb use and underscore the importance of identifying rare developmental anomalies in the archaeological record as their presence may indicate genetic relationships within or among archaeological cemeteries.
Limitations: With commingled contexts and incomplete recovery of skeletal remains, individualizing the brachydactylous elements was not possible. There is also a lack of comparative data from other Andean sites.
Suggestions for Further Research: Identify more Andean cases of brachydactyly to learn if the relatively frequent involvement of the first digit is more common among Andean skeletal samples than North American, or if it is unique to Marcajirca.

1. Introduction

Brachydactyly is a general term referring to the shortening of a digital ray due to the hypoplasia or aplasia of one or more segments (Stevenson, 2006). Single or multiple digits may be affected in an individual, and expression may be unilateral or bilateral, symmetric (affecting the same digits on both sides) or asymmetric (affecting different digits on each side). The condition may be acquired, arise as an isolated trait, or be part of a syndrome. In the hands, the middle phalanges are most commonly affected (Bell, 1951). In an archaeological sample however, it would be difficult to assess hypoplasia or aplasia of phalanges due to incomplete recovery of these bones, and due to the variability in size of the various phalanges (especially in commingled contexts). Therefore, it is less likely that brachydactyly would be identified through the phalanges, and more likely that hypoplasia of the metacarpals (brachymetacarpia) and/or metatarsals (brachymetatarsia) will be the form of brachydactyly that will be identified in paleopathological contexts (Case, 1996; Cybulski, 1988). While any of the metapodials may be affected, it is most often the fourth metacarpal and metatarsal that are affected (Stevenson, 2006).

Clinically, brachymetacarpia and brachymetatarsia are considered rare, in that fewer than 1/2000 individuals are affected (Temtamy and Aglan, 2008; Temtamy and McKusick, 1978). Brachymetatarsia is less common than brachymetacarpia, with a clinical incidence of 0.02 % to

* Corresponding author.
E-mail address: atitelb@email.arizona.edu (A.R. Titelbaum).

https://doi.org/10.1016/j.ijpp.2021.04.006
Received 30 December 2020; Received in revised form 17 March 2021; Accepted 10 April 2021
Available online 29 April 2021

0.05 % (Mah et al., 1983; Urano and Kobayashi, 1978). Brachymetatarsia is often bilateral (72 % of cases) with a strong prevalence for females (Schimizzi and Brage, 2004; Munuera Martínez et al., 2004; Urano and Kobayashi, 1978). Since isolated brachymetacarpia and/or brachymetatarsia is a heritable, rare, and easily observed developmental anomaly, its presence may offer insight into genetic relationships within or among burial populations (Case et al., 2017; Stojanowski and Schillaci, 2006; Alt and Vach, 1998). Further, since isolated brachydactyly is conveyed by autosomal dominant inheritance, it should be observable in multiple generations (Case, 1996).

Previously, two cases of Madelung's Deformity and possible mesomelic dwarfism (Léri-Weill dyschondrosteosis) from one Late Intermediate Period (ca. AD 1250) commingled tomb were reported from Marcajirca, a highland site in Ancash, Peru (Fig. 1) (Titelbaum et al., 2015). Since then, ten additional atypically short skeletal elements representing a minimum number of three individuals (MNI) from two additional commingled tombs have been discovered, all of which are metapodials. Here we present the first examples of brachydactyly to be described from an archaeological context in South America. In addition to brachydactyly being rarely reported in the paleopathological literature, the presence of two individuals with heritable brachydactyly in one tomb corroborates the previous suggestion that tombs were used for familial interment (Titelbaum et al., 2015). It is possible the third individual from a different tomb may have been related to the other two, and the different burial contexts may reflect postmarital practices. These cases offer insight into tomb use, suggest the continued use of the site over time by an extended kin group, and underscore the importance of identifying rare developmental anomalies in the archaeological record.

2. Materials and methods

Human remains at Marcajirca have been found in accessible aboveground walled tombs (*chullpas*) (n = 35), and subterranean burials discovered beneath structure floors (n = 2), some of which have been tested archaeologically. While human remains in *chullpas* were commingled, the subterranean burials yielded a combination of individual interments and commingled remains. In nearly all contexts, the human remains were in overall excellent condition.

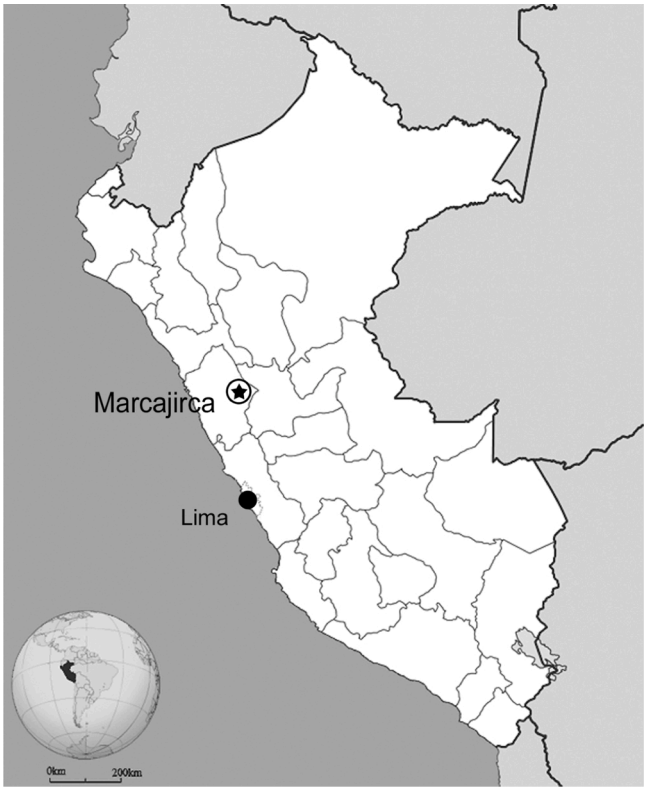

Fig. 1. Location of Marcajirca, Peru. Modified from Huhsunqu (2009).

Radiocarbon dates place the occupation of Marcajirca between AD 1132–1648, a period that corresponds to the Late Intermediate Period (AD 1000–1450) and the Late Horizon-to-Early Colonial transition (AD 1450−1650) (Ibarra Asencios, 2021). *Chullpas* tend to date within the Late Intermediate Period (e.g., 865 ± 20 BP, cal AD 1184–1267, YU-4523, wood, *Chullpa* 7), whereas human remains from the subterranean burials have yielded somewhat later dates that mark the Late Intermediate Period to Late Horizon transition (e.g., 14C date of 485 ± 15 BP, cal AD 1433–1459, UCI-185297, human phalanx, Structure 7) (Ibarra Asencios, 2021).

Archaeological testing of six *chullpas* and the two subterranean burials produced an MNI of 250 adults and 110 subadults, based on counts of long bones and mandibles. Adult bones were identified as those with complete epiphyseal fusion. Each excavated bone was visually inspected for bone formation or loss, abnormalities of shape and size, trauma, developmental anomalies, and degenerative changes. Observations and measurements of long bones followed standard guidelines (Buikstra and Ubelaker, 1994). Radiography was not available at the time of observation.

During the analysis of the skeletal remains, atypically short adult metapodials were observed in two separate burial contexts: *Chullpa* 7 and Structure 7, a subterranean burial. No other adult tubular bones from these two contexts and no other adult metapodials from the other six burial contexts demonstrated unusual length (Table 1). Since the affected elements were limited in distribution to *Chullpa* 7 and Structure 7, subsequent analysis focused on the remains from these contexts. Because metapodial brachydactyly is typically the result of premature fusion of an epiphysis, bones without complete epiphyseal closure were excluded from subsequent analysis. Also omitted from further analysis were the phalanges: since the majority of the remains were commingled and incomplete, siding and numerically assigning the present phalanges would have been a challenging endeavor (Cybulski, 1988).

***Chullpa* 7** yielded an MNI of 53 adults, based on the number of recovered femora. Adult bones included 473 long bones, 107 metacarpals, and 108 metatarsals (Tables 1, 2). Long bones included 79 humeri (42 right, 37 left), 55 ulnae (29 right, 26 left), 65 radii (30 right, 35 left), 105 femora (53 right, 52 left), 89 tibiae (42 right, 47 left), and 80 fibulae (40 right, 40 left).

Structure 7 yielded an MNI of 22 adults, based on the number of recovered mandibles. Adult remains included 172 long bones, 59 metacarpals, and 73 metatarsals (Tables 1,2). Long bones included 28 humeri (15 right, 13 left), 26 ulnae (13 right, 13 left), 29 radii (17 right, 12 left), 33 femora (18 right, 15 left), 27 tibiae (15 right, 12 left), and 29 fibulae (16 right, 13 left).

Measurements of the metapodials were taken with an osteometric board, sliding calipers, and a flexible tape measure. Measured dimensions included maximum length, dorsal-palmar/plantar diameter of the head and base, medial-lateral diameter of the head and base, and the circumference at midshaft.

3. Results

Of the 1210 metapodials excavated from eight burial contexts at Marcajirca, ten (0.83 %) were atypically short (Table 3). These ten shortened elements were observed among the 347 metapodials recovered from *Chullpa* 7 and Structure 7, and included one right first metacarpal, one left fifth metacarpal, four first metatarsals (three right, one left), three fourth metatarsals (two right, one left), and one right fifth metatarsal. These metapodials represent an MNI of three affected individuals (1.20 %) of the estimated MNI of 250 from the eight burial contexts (Table 4). While these elements demonstrated shortness in length, the measurements of their heads, bases, and midshaft

Table 1
Adult tubular bones recovered from eight burial contexts at Marcajirca.

Burial Context	Long Bones		Metacarpals			Metatarsals			Adult MNI
	R	L	R	L	?	R	L	?	
Chullpa 6	104	107	56	45	0	46	54	1	20
Chullpa 7	236	237	60	40	7	60	40	8	53
Chullpa 8	99	101	12	12	0	21	13	0	30
Chullpa 13	137	116	31	26	5	39	42	1	39
Chullpa 14	125	106	20	27	13	26	22	1	25
Chullpa 26	147	137	43	33	0	65	70	0	27
Structure 7	94	78	26	31	2	30	29	14	22
Structure 10	99	99	46	30	9	19	31	4	34
Totals	*1041*	*981*	*294*	*244*	*36*	*306*	*301*	*29*	*250*

Key: *R* = right, *L* = left, *?* = indeterminate.

Table 2
Adult metacarpals and metatarsals recovered from *Chullpa* 7 and Structure 7.

Burial Context	MC1			MC2			MC3			MC4			MC5			MC?		
	R	L	?	R	L	?	R	L	?	R	L	?	R	L	?	R	L	?
Chullpa 7	7	8	0	14	7	0	12	8	0	5	2	0	17	7	0	5	8	7
Structure 7	9	5	0	4	5	0	4	9	0	3	5	0	6	7	0	0	0	2
Burial Context	MT1			MT2			MT3			MT4			MT5			MT?		
	R	L	?	R	L	?	R	L	?	R	L	?	R	L	?	R	L	?
Chullpa 7	9	7	2	11	7	0	12	4	0	11	7	1	7	10	1	1	5	4
Structure 7	5	6	0	4	6	0	6	6	0	9	8	0	6	3	0	0	0	14

Key: *MC* = metacarpal, *MT* = metatarsal, *?* = indeterminate, *R* = right, *L* = left.

Table 3
Percentage of affected adult metapodials.

Burial Context	Affected Metapodials	Total Metapodials	% Affected
All eight burial contexts	10	1210	0.83 %
Chullpa 7	4	215	1.86%
Structure 7	6	132	4.54%

Table 4
Percentage of the minimum number of affected adult individuals.

Burial Context	Affected MNI	Total MNI	% Affected
All eight burial contexts	3	250	1.20 %
Chullpa 7	1	53	1.89 %
Structure 7	2	22	9.09 %

MNI = minimum number of individuals.

circumferences were similar to those of the other adult metapodials (Table 5).

From *Chullpa* 7, four shortened metapodials were observed: one left fifth metacarpal (Fig. 2), one right and one left first metatarsal (Fig. 3), and one right fifth metatarsal (Fig. 4), representing an MNI of one individual with brachydactyly (1.89 %) from this tomb. No shortened long bones of the upper or lower extremities were observed.

From Structure 7, six shortened metapodials were observed among the commingled remains: one right first metacarpal (Fig. 5), two right first metatarsals (Figs. 6,7), and one left and two right fourth metatarsals (Figs. 8,9), representing an MNI of two brachydactylous individuals (9.09 %) from this burial context. Although the head of the left fourth metatarsal (MT4) was damaged on the dorsal aspect, the remaining plantar portion demonstrated morphology similar to the heads of the other shortened MT4s, with plantar elongation of the articular surface and diffuse porosity (Fig. 9). While no shortened long bones of the extremities were observed among the remains of Structure 7, symphalangism of distal and intermediate foot phalanges was present, though the side, position, and association with an individual was indeterminate.

Table 5 presents measurements of the shortened metapodials compared with the mean measurements of sided but unsexed non-brachydactylous adult metacarpals and metatarsals. Due to the low number of complete metapodials, measurements of metapodials from multiple sepulchers were combined to achieve the mean averages. As shown, all measurements of the shortened bones fall within the ranges for the rest of the population's metapodials with the exception of length: for nearly all of the shortened bones, the lengths ranged 5–20 mm shorter than the averaged population measurements. The shortened metacarpals measured 5–16 mm (12.3–34 %) shorter than average, and the shortened metatarsals measured 9–20 mm (9.8–33.12 %) shorter. The one exception was a right first metatarsal from Structure 7, that measured 50 mm in length, while the range was 48–67 mm. Although this bone's length was found to measure 2 mm longer than the low end of the range, it was considered to be shortened due to its overall morphology, including the proportionately and absolutely large dimensions of the head, base, and midshaft circumference (Table 5, Fig. 7).

Independent-samples two tailed t-tests were conducted, to A) compare the measurements of the grouped short first metatarsals with the measurements of the average length first metatarsals, and B) compare the lengths of the grouped short fourth metatarsals with those of the average fourth metatarsals. The results found:

A) There was a statistically significant difference in the length of the short first metatarsals (M = 44.5, SD = 3.87) and average first metatarsals (M = 55.39, SD = 4.3); t (43) = 4.87, $p = 0.000016$. These results suggest that the short first metatarsals are significantly shorter than those of average length.
B) There was a statistically significant difference in the length of the short fourth metatarsals (M = 49.67, SD = 3.51) and average fourth metatarsals (M = 61.84, SD = 4.66); t (33) = 4.39, $p = 0.00011$. These results suggest that the short fourth metatarsals are significantly shorter than those of average length.

Table 5
Measurements of the short metacarpals and metatarsals recovered from *Chullpa* 7 and Structure 7, compared with the mean measurements of sided but unsexed non-brachydactylous elements. Measurements in millimeters.

Bone	Max Length	Head DP	Head ML	Base DP	Base ML	MS Circumf
1st Metacarpal						
Structure 7, Right	35*	13.5	14	15	18	34
Adult average	39.9 ($n = 14$)	11.4 ($n = 14$)	13.9 ($n = 14$)	13.2 ($n = 14$)	13.7 ($n = 13$)	28.3 ($n = 14$)
Adult Range	37–46	10–13.5	12.5–16	12–15	12–15	22–35
5th Metacarpal						
Chullpa 7, Left	31*	12.5	12.5	10.5	13.5	22.5
Adult average	47 ($n = 11$)	10.8 ($n = 10$)	10.7 ($n = 11$)	10.6 ($n = 10$)	11.8 ($n = 11$)	20.4 ($n = 11$)
Adult Range	43–52	10–12	9–12.5	9–15	10–14	18–23
1st Metatarsal						
Chullpa 7, Left	44*	17	20	23	21	40
Chullpa 7, Right	41*	15	19	25	20	45
Structure 7, Right	50	22	21	29	22	47
Structure 7, Right	43*	18	n/a	24	16	41
Adult average	55.4 ($n = 41$)	19.2 ($n = 40$)	20.4 ($n = 39$)	25.1 ($n = 40$)	18.9 ($n = 38$)	41.9 ($n = 41$)
Adult Range	48–67	16–24	16.5–25	17–30	15–22	33–50
4th Metatarsal						
Structure 7, Left	50*	14	9	16	12	26
Structure 7, Right	46*	16	10	15	14	30
Structure 7, Right	53*	17	11	17	12	26
Adult average	61.8 ($n = 32$)	13.2 ($n = 30$)	9.5 ($n = 30$)	16.7 ($n = 31$)	11 ($n = 32$)	26.1 ($n = 32$)
Adult Range	55–73	11–16	8–12	14–21	9–14	21–37
5th Metatarsal						
Chullpa 7, Right	41*	11	11	13	18	24
Adult average	61.3 ($n = 16$)	12.7 ($n = 15$)	10.3 ($n = 15$)	13.6 ($n = 16$)	18.9 ($n = 16$)	27.3 ($n = 16$)
Adult Range	54–68	10–15	8–13	11–17	16–23	24–30

Key: *Max* = maximum; *DP* = dorsal palmar/plantar; *ML* = medial-lateral; *MS Circumf* = midshaft circumference.
* = measurement is below that of the adult average and range.

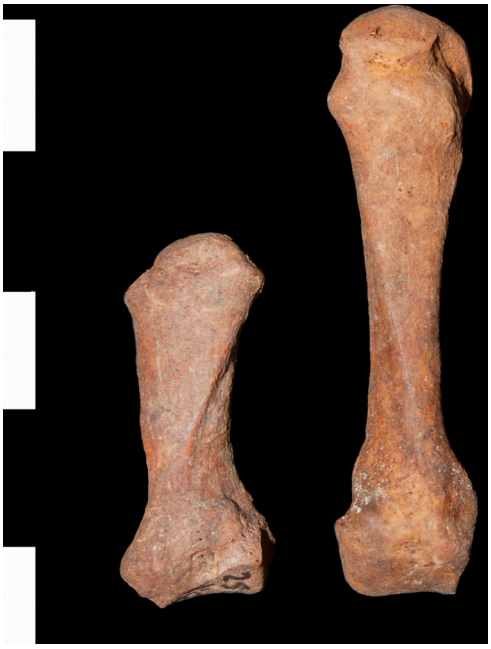

Fig. 2. *Chullpa* 7, shortened left fifth metacarpal pictured with a non-brachydactylous adult right fifth metacarpal for comparison. Dorsal view.

4. Differential diagnosis

Brachydactyly is the shortening of the digital rays of the hands and/or feet due to hypoplasia or aplasia of metacarpals, metatarsals, and/or phalanges. Clinically, there is a wide range of presentation, and the condition may be classified into various types, based on what combination of bones are affected. Most cases appear along genetic lines, occurring as isolated cases or associated with a variety of syndromes (Poznanski et al., 1977; Temtamy and McKusick, 1978), though it may arise following trauma. Even when brachydactyly appears along genetic lines, there is interfamilial variability in the digits affected (Bell, 1951; Brailsford, 1945). In the present differential diagnosis, systemic bone growth disorders such as achondroplasia and multiple epiphyseal dysplasia are considered unlikely, since shortened long bones of the extremities were not observed among the skeletal remains of the two sepulchers.

4.1. Brachydactyly typology

Brachydactyly has been categorized into types based on the main digital ray or the portion of a ray that is affected. The earliest and most commonly used classification of the nonsyndromic brachydactylies was devised by Bell (1951), after examining 124 pedigrees consisting of 1336 individuals. Bell (1951) discerned five types of brachydactyly (A–E), and this typology was later modified by Temtamy and McKusick (1978) and Fitch (1979) (Table 6). It should be noted that many of these typologies present challenges in an archaeological context, since the full complement of hand and foot bones are not consistently recovered. Overall brachydactyly is clinically rare, with the exception of Bell's Type A3 and D (see below), which have a prevalence of around 2% (Temtamy and Aglan, 2008; Temtamy and McKusick, 1978). Fitch (1979) noted that symphalangism may be found with brachydactyly types A, B, C, and E.

The hallmark of Bell's Type A1 is hypoplasia of the middle phalanges in digits two through five of the hands or feet, though the proximal phalanges of the hallux and pollex may also be affected (Bell, 1951). This presentation may be accompanied by shortening of the metacarpals or metatarsals of digits four and five, and findings are typically bilateral (Stevenson, 2006). Individuals with Type A1 tend to demonstrate short stature (Bell, 1951; Stevenson, 2006). Type A2 differs from A1 in that shortening is confined to the middle phalanx of the second digits, and in Type A3, it is the middle phalanx of only the fifth digits that are shortened.

Bell's Type B is often an isolated finding, characterized by hypoplastic or aplastic distal phalanges and nails, in addition to shortened middle phalanges (Bell, 1951). The number of digits affected is variable, but exclude the pollex and hallux.

Fig. 3. *Chullpa* 7, shortened right (on left) and left (in middle) first metatarsals pictured with a non-brachydactylous adult left first metatarsal. Dorsal view.

Fig. 4. *Chullpa* 7, shortened right fifth metatarsal pictured with a non-brachydactylous adult right fifth metatarsal for comparison. Dorsal view.

Fig. 5. Structure 7, shortened right first metacarpal. Dorsal view.

With Bell's Type C, the fourth digit is either normal or relatively long, whereas digits two and three are usually short. The most characteristic shortening seen with this type is that of the second and third middle phalanges, and the proximal phalanges may be affected by hypersegmentation and fusion (Bell, 1951). Fitch (1979) added to this description, noting that the middle phalanx of digit five and metacarpals may also be affected. Reports indicate that individuals with Type C may demonstrate additional abnormalities, including short stature, hip disease, short and abnormally shaped ulnae, radial hypoplasia, and Madelung's deformity among others (Fitch, 1979:41).

The typical presentation of Bell's Type D is a short broad distal phalanx of the thumb, while the rest of the hand is normal (Bell, 1951). This presentation has also been referred to as "Stub Thumb" or "Murderer's Thumb", the latter name bequeathed by practitioners of palmistry at the turn of the 20th century (Poznanski et al., 1977; Stevenson, 2006; Benham, 1901:171). The shortening may be unilateral or bilateral, and the hallux may also be affected.

Bell's Type E (BDE) can also be referred to as brachymetacarpia or brachymetatarsia, as this form of brachydactyly consists of shortened metacarpals and/or metatarsals (Bell, 1951). Findings are often asymmetric, and there may also be involvement of the phalanges (Stevenson, 2006; Pareda et al., 2013; Herzog 1968). BDE can arise secondary to trauma that occurs prior to normal epiphyseal fusion, be isolated as an autosomal dominant trait with variable expression, or be associated with a number of more complex syndromes, including Albright hereditary osteodystrophy, acrodysostosis, and Turner syndrome. While any of the digits may be affected, the fourth and fifth metacarpals and metatarsals are most commonly affected, and individuals may demonstrate short stature (Stevenson, 2006). The population frequency of BDE is unknown, but it is rare as an isolated anomaly (Temtamy and Aglan,

Fig. 6. Structure 7, shortened right first metatarsals pictured with a non-brachydactylous adult right first metatarsal for comparison. Dorsal view.

Fig. 7. Structure 7, shortened right first metatarsal pictured with a non-brachydactylous adult right first metatarsal for comparison. Lateral view.

2008).

Unfortunately, without complete sets of phalanges, it is impossible to know if there are multiple types of brachydactyly represented by the remains at Marcajirca. However, given that the observed shortened bones consisted of metacarpals and metatarsals, and given the overall rarity of brachydactyly, the parsimonious suggestion is that rather than there being multiple types of brachydactyly, the shortened elements can be categorized as BDE. The question becomes whether the BDE was acquired, isolated, or associated with a syndrome.

4.2. Acquired BDE

Trauma, infection, or infarction may affect an individual's digit during growth and development. For trauma or infection to affect a metapodial such that it achieves adult proportions in dimensions other than length, the injury would have had to affect the epiphyseal cartilage, leading to premature fusion (Arslan, 2001; Cybulski, 1988:368). Trauma to the epiphyseal cartilage would likely displace the epiphysis relative to the metaphysis, which should be observable in skeletal remains (Case, 1996). This type of injury tends to be unilateral and isolated. While it is plausible that traumatic brachydactyly may occur in a single individual in a series, it seems less likely for it to occur among multiple individuals in two burial contexts, especially if there is a possibility of bilateral presentation. Further, other than their apparent size difference, the shortened elements at Marcajirca demonstrate no other macroscopic evidence of a healed fracture or infection, such as abnormalities of shape, remodeling, traumatic arthritis, or joint fusion (Buikstra and Ubelaker, 1994).

Brachydactyly may also arise due to metaphyseal infarction that leads to osteonecrosis in the metadiaphyseal region and premature fusion of the epiphyses (Bosch et al., 2011). However, metaphyseal infarction is most commonly associated with sickle cell disease, which did not affect prehistoric Andean populations (Greenfield, 1980).

4.3. Isolated BDE

BDE may arise as a heritable isolated condition, not associated with a syndrome. While isolated BDE with normal stature has been attributed to a heterozygous mutation in the homeobox D13 gene (*HOXD13*), isolated BDE with short stature has been attributed to a mutation in the parathyroid hormone-like hormone gene (*PTHLH*) (OMIM, 2020; Genetic and Rare Diseases Information Center (GARD, 2018a; Pareda et al., 2013). Both forms are autosomal dominant with variable expressivity. Though phenotypic expression is variable, with wide intrafamilial variation, shortened fourth metatarsals are most frequently seen (Pareda et al., 2013; Johnson et al., 2003; Holt, 1975; Bell, 1951; Brailsford, 1945).

4.4. Syndromic BDE

Brachydactyly may occur with numerous familial multiple malformation syndromes. The syndromes most commonly associated with brachydactyly include Albright's hereditary osteodystrophy, acrodysostosis, and Turner syndrome. However, there are challenges involved in identifying and differentiating among these syndromes both in clinical contexts and in paleopathology: the degree of involvement of brachydactyly in each syndrome is variable with variable presentation, the syndromes often involve other nonspecific skeletal changes (e.g. dental crowding), the syndromes usually involve soft tissue/organ changes that are not preserved archaeologically, and some of the syndromes involve variations in hormone levels that cannot be assessed from skeletal remains.

4.4.1. Albright's hereditary osteodystrophy (AHO)

AHO is a rare heritable disorder characterized by a set of clinical features that include short stature, obesity, round face, subcutaneous ossifications, cone shaped epiphyses, and skeletal anomalies (Genetic and Rare Diseases Information Center (GARD, 2018b; OMIM, 2016a; Pareda et al., 2013; Poznanski et al., 1977). Brachydactyly is a typical feature of AHO, with involvement of the first distal phalanx and the third, fourth, and fifth metacarpals, and often with asymmetrical presentation (Poznanski et al., 1977).

AHO is the phenotype for pseudohypoparathyroidism (PHP), a group of several related genetic disorders where there is end-organ resistance to parathyroid hormone and multiple hormone resistance (Poznanski et al., 1977). PHP results from mutations in several genes, including the *GNAS* locus on the maternal allele (OMIM, 2016a; Genetic and Rare Diseases Information Center (GARD, 2016a). Among individuals with PHP, 70–78 % demonstrate brachydactyly, though presentation is variable (Pareda et al., 2013). When the mutation is inherited on the paternal allele, an individual will have the clinical features of AHO, but without hormone resistance, which is referred to as

Fig. 8. Structure 7, shortened left (second from left) and two right fourth metatarsals pictured with a non-brachydactylous adult left fourth metatarsal for comparison. Dorsal view.

Fig. 9. Structure 7, shortened right and left fourth metatarsals. Lateral views.

pseudopseudohypoparathyroidism (PPHP) (OMIM, 2016b; Genetic and Rare Diseases Information Center (GARD, 2016b; Pareda et al., 2013). With PPHP the fourth metacarpal is frequently shortened, though all metacarpals and metatarsals may be affected (OMIM, 2016b). Both PHP and PPHP are autosomal dominant.

Poznanski et al. (1977) examined radiographs of the hands of individuals with PHP, PPHP and BDE to determine if there were differentiable morphological features among the conditions. They found that the presentation of individuals with PHP and PHPP were nearly identical, and radiographically indistinguishable from individuals with BDE. Since the skeletal pattern of BDE is not informative on its own, to differentiate among the various conditions it would be necessary to perform biochemical analyses (Pareda et al., 2013; Poznanski et al., 1977).

Table 6
Bell (1951) classification of brachydactyly with the Fitch (1979) classification. After Stevenson (2006:968).

Type A: Brachymesophalangy: shortening is confined mainly to the middle phalanges
Type A-1: Brachymesophalangy II-V: brachybasophalangy I (Fitch type 9)
Type A-2: Brachymesophalangy II (Fitch type 2)
Type A-3: Brachymesophalangy V (Fitch type 3)
Type B: the middle and distal phalanges are short or absent
Aplasia distal phalanges, II-V
Hypoplasia middle phalanges, II-V
Broad, bifid distal phalanges, I (Fitch type 8)
Type C: the middle and proximal phalanges of digits 2 and 3 are affected
Brachymesophalangy II, III
Hypersegmentation, proximal phalanges II, III (Fitch type 11)
Type D: the distal phalanx of digit 1 is affected
Short, broad distal phalanx (Fitch type 1)
Type E: one or more of the metacarpals or metatarsals are affected
Brachymetacarpia
Brachymetatarsia
Fitch type 4: Brachymetacarpia II
Fitch type 5: Brachymetacarpia IV
Fitch type 6: Brachymetatarsia IV
Fitch type 7: Brachymetacarpia IV; brachymetatarsia IV
Fitch type 10: Short stature, brachymetacarpia, brachytelephalangy

4.4.2. Acrodysostosis (with or without multihormonal resistance)

Acrodysostosis is a rare skeletal dysplasia caused by heterozygous mutation in the *PRKAR1A* gene (OMIM, 2016c). Phenotypically, it is characterized by severe generalized shortening of the metacarpals and metatarsals of digits 2–5, cone shaped epiphyses, short stature, spinal stenosis, craniofacial anomalies such as maxillary hypoplasia and nasal hypoplasia, and individuals with acrodysostosis often have intellectual disabilities (OMIM, 2016c; Genetic and Rare Diseases Information Center (GARD, 2016c; Pareda et al., 2013; Poznanski et al., 1977). Digit 1 tends to be less frequently affected (Pareda et al., 2013). While AHO and acrodysostosis demonstrate some similarities, overall hand size with acrodysostosis is smaller with more marked shortening of the

metacarpals, and the two can be distinguished at the genetic level (Poznanski et al., 1977). Given the overall rarity of acrodysostosis, combined with the relatively high frequency of digit 1 being affected at Marcajirca (5/10), it is less likely that this syndrome accounts for BDE at Marcajirca.

4.4.3. Turner syndrome (TS)

TS is a chromosomal disorder affecting females, where one sex chromosome is normal, and the other is missing or structurally altered. While patient presentation depends on the portion of the chromosome that is missing, individuals with TS tend to have short stature, gonadal dysgenesis, primary amenorrhea, and skeletal changes such as scoliosis, micrognathia, and brachydactyly (Genetic and Rare Diseases Information Center (GARD, 2016d; Pareda et al., 2013). When brachydactyly occurs, it most commonly involves digit four, though the third and fifth metacarpals may also be affected (Poznanski et al., 1977). Since most females affected by TS are infertile, most cases of TS are not inherited (Genetic and Rare Diseases Information Center (GARD, 2016d). At Marcajirca, the shortened metapodials represent at least 3 cases from two burial contexts, which suggests the brachydactyly was inherited. Therefore, TS can likely be removed from the differential diagnosis.

4.5. BDE at Marcajirca

The shortened metapodials recovered at Marcajirca represent at least 3 cases of brachydactyly from two burial contexts, which suggests the condition was inherited. Therefore, acquired brachydactyly and TS can likely be removed from the differential diagnosis. Acrodysostosis and isolated BDE with short stature can also probably be ruled out, as brachydactyly involved digit 1 in 5/10 instances, and none of the long bones were appreciably short or stocky. Possible diagnoses could be isolated BDE with normal stature, or PHP/PPHP; however, without biochemical analysis, it would be challenging to differentiate between these conditions from skeletal findings. Therefore, while a syndromic cause cannot be ruled out, the more conservative suggestion is that the cases be considered isolated BDE, conveyed through autosomal dominant inheritance with variable expressivity.

5. Discussion

Brachydactyly was the first human developmental anomaly recognized as an example of Mendelian inheritance, by anthropologist William Curtis Farabee in his dissertation from Harvard (Farabee, 1903). Nevertheless, paleopathological reports about brachydactyly have been infrequent. In some instances, they are embedded within the descriptions of syndromes. For example, Kozieradzka-Ogunmakin (2011) presented a case of multiple ephiphyseal dysplasia in a possible male Old Kingdom Egyptian skeleton. Among the numerous skeletal findings were a short and angled fourth left metacarpal and fourth left metatarsal (the right sided elements were absent).

Other reports briefly mention sporadic or isolated cases as part of general paleopathological analyses. In the New World, brachydactyly has been reported from Canada, the United States, and Mexico (see Table 7). While the majority of examples (58.8 %) involve the fourth metatarsal (Barnes, 1994; Loveland and Gregg, 1988; Pokotylo et al., 1987; Merbs and Vestergaard, 1985; Reed, 1981), a brachydactylous first metacarpal was seen at Fairty, an Iroquois ossuary in Ontario, Canada (Anderson, 1963), a shortened fourth metacarpal was pictured from La Playa, Mexico (Barnes, 2012), a shortened first metatarsal was noted in an individual from Pueblo de Las Humanas in Gran Quivira National Monument, New Mexico (Reed, 1981), and unilateral shortening of the right fifth metatarsal was observed in a female from Pueblo Bonito, New Mexico (NMNH 327071) (Barnes, 2012). This latter individual also demonstrated additional developmental anomalies, including bilateral os intermetatarseum.

Several paleopathological reports discuss brachydactyly as the primary focus. Loveland and Gregg (1988) presented a single case of brachymetatarsia from the Kaufman-Williams site in Texas. The affected bone was the right fourth metatarsal of a young adult male that measured 11 mm shorter than the contralateral bone, though both bones demonstrated the same circumference. The authors suggested that while this was the only example of brachydactyly observed, it reflected autosomal dominant inheritance, and either there were other examples that were not yet recovered, or the individual did not pass the trait.

Cybulski (1988) described an MNI of ten individuals with brachydactyly from four sites on the north mainland coast of British Columbia. These cases were observed in eight skeletons and isolated remains. Metatarsals were more commonly affected than metacarpals, right-sided elements were more often affected than the left, and bilateral involvement was observed in at least three individuals. While the most frequently affected bone was the fourth metatarsal ($n = 8$, MNI = 6), also shortened were first metatarsals ($n = 3$), and first ($n = 2$), fourth ($n = 2$), and fifth ($n = 1$) metacarpals (Cybulski, 1988:366). For the bones where an unaffected contralateral side was present in the same individual, the shortened metacarpals were 6−13 mm shorter and the shortened metatarsals were 10−11 mm shorter. No additional skeletal changes or pathology suggested syndromic conditions. Cybulski (1988) therefore suggested that the incidents reflect an isolated inherited trait likely involving autosomal dominance with incomplete penetrance, and raised the possibility of genetic connections between the sites.

Case (1996) examined the protohistoric Mobridge and Sully skeletal collections from South Dakota for developmental anomalies of the hands and feet and identified six individuals with brachydactyly. Similar to Cybulski's (1988) findings, Case (1996) found that metatarsals were more commonly affected than the metacarpals, and the fourth metatarsal was the most commonly affected bone.

More recently, Giuffra et al. (2014) described a case of brachymetatarsia from a Medieval context in Sardinia. The affected individual was a middle adult female with bilateral shortening of the fourth metatarsal and symphalangism of the distal interphalangeal joint of the left fifth toe. The affected metatarsals measured approximately 20 mm shorter than the rest of the population. Since the condition was present bilaterally, the authors ruled out an acquired etiology, but were unable to determine if it was an isolated or syndromic condition.

Compared with the previous reports, the examples from Marcajirca show some similarities. Metatarsals were more commonly affected than metacarpals (8:4), right-sided elements were more often affected than the left (7:3), and the affected bones measured 5−20 mm (9.8–33.1 %) shorter than the average bone lengths. Additionally, there were no examples of brachymetacarpia or brachymetatarsia of digits two and three. However, while the fourth metatarsal was most commonly affected in other investigations, at Marcajirca the most commonly affected bone was the first metatarsal ($n = 4$; MNI = 3) followed by the fourth ($n = 3$; MNI = 2). Unfortunately, the remains were from commingled contexts, so it was not possible to see the full pattern of brachydactyly, determine the sex of the individuals, or investigate if brachydactyly co-occurred with other developmental anomalies.

In the cases described above and at Marcajirca, the affected individuals may have cosmetically presented with brachydactyly. In most cases, the individuals would have experienced no functional deficits (Suresh et al., 2009; Harris and Beath, 1949). It is possible however, that hand function could have been affected with either a shortened pollex, with deficits in pinch function during grasping, or shortened digits four and five, with loss in grip strength (Arslan, 2001; Suresh et al., 2009). It is also possible that brachymetatarsia could have been associated with pain (e.g., from transfer lesions or callus formation) and deformity, including claw toe or cock-up toe where there is dorsiflexion of the metatarsophalangeal joint and flexion of the interphalangeal joints of the affected digit (Schimizzi and Brage, 2004; Munuera Martínez et al., 2004; Arslan, 2001). While the individuals at Marcajirca may have demonstrated minor physical differences, particularly with brachymetatarsia of digit four, there is no reason to believe they suffered social

Table 7
New World cases of brachydactyly.

Case	Site	Location	Time Period	Sex	Side	MC1	MC2	MC3	MC4	MC5	MT1	MT2	MT3	MT4	MT5	Reference
Unknown	Fairty Ossuary	Ontario, Canada	(protohistoric)	?	?	1										Anderson, 1963
Burial 6	Cache Creek	British Columbia, Canada	1330 +/- 260 BP	F	L/R									2		Pokotylo et al., 1987
Burial 303	Parizeau	British Columbia, Canada	1500 BC-AD 500	F	R	1										Cybulski, 1988
Burial 345	Boardwalk	British Columbia, Canada	1500 BC-AD 500	F	R									1		Cybulski, 1988
Burial 372	Boardwalk	British Columbia, Canada	1500 BC-AD 500	M	L						1					Cybulski, 1988
Burial 397	Boardwalk	British Columbia, Canada	1500 BC-AD 500	M	L/R						1			2		Cybulski, 1988
Burial 455	Lachane	British Columbia, Canada	1500 BC-AD 500	M	L/R									2		Cybulski, 1988
Burial 457	Lachane	British Columbia, Canada	1500 BC-AD 500	M?	R									1		Cybulski, 1988
Burial 505	Baldwin	British Columbia, Canada	1500 BC-AD 500	M	R						1					Cybulski, 1988
Burial 516	Baldwin	British Columbia, Canada	1500 BC-AD 500	F	R/L				2	1						Cybulski, 1988
Isolated 353	Boardwalk	British Columbia, Canada	1500 BC-AD 500	?	R									1		Cybulski, 1988
Isolated 812	Baldwin	British Columbia, Canada	1500 BC-AD 500	?	L	1										Cybulski, 1988
Isolated 853	Baldwin	British Columbia, Canada	1500 BC-AD 500	?	R									1		Cybulski, 1988
Burial 6	Inscription House	Arizona, USA	AD 1050–1300	?	L									1		Reed, 1981
1514–19	Sundown	Arizona, USA	AD 1100–1200	F	L/R									2		Merbs and Vestergaard, 1985
FS-4518	Las Humanas	New Mexico, USA	AD 1545–1672	F?	L?						1					Reed, 1981
Room 329	Pueblo Bonito	New Mexico, USA	AD 850–1150	?	L									1		Barnes, 1994
NMNH 327,071	Pueblo Bonito	New Mexico, USA	AD 850–1150	F	R										1	Barnes, 2012
NMNH 383,150	Mobridge	South Dakota, USA	AD 1700–1830	F	R						1					Case, 1996
NMNH 383,040	Mobridge	South Dakota, USA	AD 1700–1830	M	R									1		Case, 1996
NMNH 382,896	Mobridge	South Dakota, USA	AD 1700–1830	?	L/R									2		Case, 1996
NMNH 383,097	Mobridge	South Dakota, USA	AD 1700–1830	?	L					1						Case, 1996
NMNH 381,400	Sully	South Dakota, USA	AD 1600–1750	M	L								1	1		Case, 1996
NMNH 381,428	Sully	South Dakota, USA	AD 1600–1750	F	L									1		Case, 1996
Burial 30	Kaufman-Williams	Texas, USA	AD 1650–1700	M	R									1		Loveland and Gregg, 1988
TOTAL				M = 8 F = 9 ? = 8	L = 16 R = 17 ? = 1	3	0	0	2	2	5	0	1	20	1	
Chullpa 7	Marcajirca	Ancash, Peru	AD 1075–1450	?	R						1				1	
Chullpa 7	Marcajirca	Ancash, Peru	AD 1075–1450	?	L						1					
Structure 7	Marcajirca	Ancash, Peru	AD 1450–1650	?	R	1					2			2		
Structure 7	Marcajirca	Ancash, Peru	AD 1450–1650	?	L					1				1		
TOTAL				? = 10	R = 7 L = 3	1	0	0	0	1	4	0	0	3	1	

Key: *MC* = metacarpal, *MT* = metatarsal, *M* = male, *F* = female, *L* = left, *R* = right, *?* = indeterminate.

disadvantage, as they were given normative burial treatment, testifying to their membership in the community.

Importantly, the presence of a minimum of three individuals with an uncommon heritable condition in two sepulchers offers insight into tomb use during the Late Intermediate Period-to-Late Horizon transition in the northern highlands of Peru. While it may seem logical to suggest that tombs were used by kin groups or individuals with shared ethnic identity, such suggestions tend to be based on Colonial records and ethnographic analogy, and less frequently on the human remains. In one study however, Baca et al. (2012), analyzed the DNA of 41 individuals from six *chullpas* at the Late Horizon site Tompullo 2 in Arequipa, Peru to determine if tombs served as family graves. The results suggested that each tomb contained members of a single patrilineal family group. In another study, Velasco (Velasco, 2018) found that Late Intermediate period *chullpas* in Colca Valley, Peru contained phenotypically similar individuals through biodistance analysis of cranial nonmetric traits.

While consanguinity at Marcajirca cannot be determined without DNA assessments, it is likely the individuals with brachydactyly were related. Two cases of an uncommon heritable developmental anomaly from one tomb is highly suggestive that genetic relationship was a factor that influenced mortuary practices at Marcajirca. How the individuals were related is not clear, though it is likely that they belonged to the same familial group. That brachydactyly was also observed in at least one other individual from *Chullpa* 7 suggests that this individual had genetic ties to the others, and the difference in interment may be reflective of postmarital residence practices. And since *Chullpa* 7′s radiocarbon date precedes those of the human remains from Structure 7, the cases of brachydactyly may also suggest that there was continued use of the site over time by an extended kin group.

5.1. The clinical and paleopathological rarity of brachydactyly

As a final consideration, how does the observed frequency of brachydactyly contribute to an understanding of rare conditions? While the clinical literature considers BDE to be a rare condition in that it affects fewer than 0.05 %, with brachymetatarsia being less common than brachymetacarpia (Temtamy and Aglan, 2008; Urano and Kobayashi, 1978; Mah et al., 1983), the minimum incidence of brachydactyly at Marcajirca is 1.2 % based on the MNI of eight burial contexts, with brachymetatarsia (1.2 %) slightly more frequent than brachymetacarpia (0.80 %). If specific burial contexts are considered, the frequency of brachydactyly rises to 1.89 % for *Chullpa* 7 and 9.09 % for Structure 7. On the one hand, these frequencies support the suggestion that the brachydactylous individuals were related, especially those from Structure 7. On the other hand, they raise the possibility that brachydactyly, though uncommon, may not be as rare paleopathologically as it seems to be in the clinical literature.

As described, paleopathological reports concerning isolated brachydactyly are few, which may underscore the overall rarity of the condition. However, the infrequent reports may also be associated with underreporting due to sampling bias, resulting from incomplete recovery, commingling, or taphonomic damage that make brachydactyly more challenging to identify (Burnett, 2005). That said, when brachydactyly is reported, archaeological frequencies tend to be higher than those reported in the clinical literature (e.g., Cybulski, 1988; Case et al., 2017). This higher frequency may be due to the genetic relatedness among the paleopathological sample; indeed, small population sizes and factors of endogamy may lead to higher frequencies of rare heritable traits, especially if it is a kin group that is represented in a burial context (Case et al., 2017; Cybulski, 1988).

Among modern populations, it is possible that brachydactyly is more common than reported in the clinical literature. Part of the reason it may be underreported may also have to do with sampling bias in that a clinician is generally limited to observing individuals that seek medical attention. Secondly, most brachydactyly research has focused on the hand rather than the foot (Cybulski, 1988). Third, brachydactyly may be subtle, and may not be identified during visual inspection. Indeed, it is not uncommon for brachydactyly to be found incidentally during clinical consult for a different reason (e.g., Suresh et al., 2009).

Further, how brachydactyly is diagnosed may affect clinical frequencies. For feet, brachymetatarsia is diagnosed radiographically when one metatarsal head is ≥ 5 mm proximal to the parabolic arc of the other metatarsal heads (Bartolomei, 1990). For hands, brachymetacarpia of the fourth metacarpal may be diagnosed through a positive ‘metacarpal sign’, originally identified by placing a pencil on the heads of the fourth and fifth metacarpals on the back of a closed fist: in affected patients, the pencil would intersect the head of the third metacarpal, indicating that the fourth was short (Archibald et al., 1959). While this method was improved with radiography, it was found to give false positives, it was unable to detect shortening when all metacarpals were affected, and assessment of the other bones was subjective (Poznanski et al., 1972, 1977). Another method of diagnosis is metacarpophalangeal pattern profile analysis, where bone lengths are measured on radiographs and used to create plots to generate a pattern that can be compared to length standards for age and sex (Poznanski et al., 1972; Garn et al., 1972). While this method is more objective, population-specific standards need to be used for comparison since growth rates and bone size vary by population (Laurencikas and Rosenborg, 2000). Further, adult bones may overlap, precluding measurements, and degenerative changes may cause difficulties in defining the ends of bones (Poznanski et al., 1972). It is therefore possible that visual inspection and radiography may miss some clinical cases (Case, 1996; Poznanski et al., 1972; Burnett, 2005).

Anatomical samples from cadaver populations offer another perspective on frequency, since it is less probable that the individuals are related, and likely represent a larger gene pool (Case et al., 1998; Burnett, 2005). Burnett (2005) examined skeletons from the Raymond A. Dart Collection, primarily composed of adults of South African descent from the 1920s-present, and found that brachydactyly was present in 1.22 % of the sample, with brachymetatarsia (0.83 %) more frequent than brachymetacarpia (0.22 %). Among the Terry Collection, comprised of individuals from the late 19th-early 20th centuries in the United States, Case et al. (2017) examined feet and observed that 0.29 % had shortened first metatarsals, while 0.88 % had short fourth metatarsals. The frequency data from these anatomical samples further suggest that while brachydactyly is uncommon, it may not be as rare as reported by the clinical frequencies. These data also suggest that brachymetatarsia is more frequently observed than brachymetacarpia.

Considering the paleopathological, clinical, and anatomical findings, it seems that clinical and paleopathological frequencies may underrepresent the prevalence of brachydactyly. That said, it is apparent that frequencies of brachydactyly vary by the techniques of diagnosis and the sample population, which in turn varies by location, time period, sample size, overall size of the gene pool, and relatedness among the individuals.

In light of this, when applying the terminology of “rare” to prehistoric conditions, it is probably best to avoid strict frequency-based definitions, as frequency may change over time and space: what is rare today may not have been rare in the past and what is rare in one region, may be more common in another. For conditions like BDE that are not maladaptive, once they arise they will continue to be expressed in subsequent generations, barring genetic drift. And if they arise in smaller, more genetically isolated populations, there will be relatively higher frequencies than in larger populations with greater gene flow. Rare conditions then, can refer to conditions that generally affect a small percentage of the overall archaeological population and tend to be infrequently encountered, identified, and reported in paleopathological literature.

6. Conclusions

Identifying rare conditions in prehistory is important for understanding the antiquity of disease, and their presence may help us

understand how individuals endured their conditions and how they were treated by their communities. Further, heritable, low frequency traits may offer insight into relationships within or among archaeological cemeteries. As such, they may indicate which individuals are genetically related, and provide information concerning the social organization of a cemetery. This is particularly true when considering a trait that is conveyed by autosomal dominant inheritance, since the trait may be observable in multiple generations.

The present investigation described ten shortened metapodials recovered from Marcajirca, a Late Intermediate to Late Horizon highland side in Ancash, Peru. These elements were excavated from two commingled burial contexts, and represent an MNI of three (1.20 %) of an estimated 250 individuals from eight burial contexts. Measurements of the shortened bones demonstrated that while the base, heads, and midshaft circumferences were comparable to the average size metapodials in the population, the difference in length was statistically significant ($p < 0.001$).

Since there were no atypically short long bones recovered from these contexts, the shortened elements can be categorized as BDE, which describes digits that are short due to the metacarpals/metatarsals. BDE may be acquired, isolated, or associated with syndromes, such as PHP and PPHP. Since multiple individuals at Marcajirca were affected, acquired BDE was ruled out, and a heritable form of BDE is suggested. Unfortunately, the skeletal pattern of brachydactyly does not differ appreciably between isolated BDE and PHP/PPHP, and differentiating among them would require biochemical analysis. The conservative suggestion then, is that the cases of brachydactyly from Marcajirca be considered isolated BDE, a clinically rare condition conveyed through autosomal dominant inheritance with variable expressivity.

The observations at Marcajirca confirm previous archaeological findings that suggest the fourth metatarsal is a commonly affected bone in BDE, and that metatarsals are more affected than the metacarpals. However, differing from previous studies, the number of instances of preaxial (digit 1) brachydactyly was the same as postaxial (digits 4 and 5). This investigation therefore adds to regional and site-oriented population studies on the frequency of brachydactyly and provides information concerning its variant expressions.

There are limitations of this study. First, a general challenge in identifying a rare disease in paleopathology is that not only are soft tissue indications not preserved, skeletal recovery is not always complete, especially in commingled contexts. Second, since the remains at Marcajirca were recovered from commingled contexts, it was not possible to associate the brachymetacarpia/brachymetatarsia with individuals to see the full pattern of brachydactyly. It is therefore unclear if multiple digits were affected in an individual, if there was bilateral expression, and if phalanges were also shortened. Also unknown is how many individuals were actually affected, their sex, and if they demonstrated additional developmental anomalies. Further limitations come from the lack of comparative data from other Andean sites. It would be interesting to know where else brachydactyly is observed in the Andes, and to learn if the relatively frequent involvement of the first digit is more common among Andean skeletal samples than North American, or if it is unique to Marcajirca.

In spite of the limitations, the presence of at least two individuals with BDE in one tomb adds support to the previous suggestion that tombs were used for familial interment. The four additional brachydactylous elements from a different tomb represent at least one more individual that may have been related to the other two, and the different burial contexts may reflect postmarital practices and the continued use of the site over time by an extended kin group. These cases therefore offer insight into tomb use, and underscore the importance of identifying rare developmental anomalies in the archaeological record as their presence may indicate genetic relationships within or among archaeological cemeteries.

Acknowledgements

This investigation was supported by the Huari-Ancash Bioarchaeological Field School and the University of Arizona College of Medicine - Phoenix. Deep appreciation is extended to the field school students and staff of the 2013-2017 seasons. We would like to acknowledge the hard work of the editors of the IJPP and the organizers and editors of this special issue, Drs. Julia Gresky and Emmanuelle Petiti, and thank them for putting this volume together. We are very grateful for the thoughtful and constructive feedback from the editors and two anonymous reviewers who offered excellent suggestions to improve this manuscript. The human skeletal remains were excavated and analyzed by the bioarchaeological field school, and reinterred at Marcajirca.

References

Alt, K.W., Vach, W., 1998. Kinship studies in skeletal remains-concepts and examples. In: Alt, K.W., Rosing, F.W., Teschler-Nicola, M. (Eds.), Dental Anthropology: Fundamentals, Limits, and Prospects. Springer, Wien, pp. 537–554.

Anderson, J.E., 1963. The People of Fairty: an Osteological Analysis of an Iroquois Ossuary, 193. National Museum of Canada Bulletin, Canada Department of Northern Affairs and National Resources, Ottawa, pp. 28–129.

Archibald, R.M., Finby, N., De Vito, F., 1959. Endocrine significance of short metacarpals. J. Clin. Endocrinol. Metabol. 19, 1312–1322. https://doi.org/10.1210/jcem-19-10-1312.

Arslan, H., 2001. Metacarpal lengthening by distraction osteogenesis in childhood brachydactyly. Acta Orthop. Belg. 67, 242–247. PMID: 11486686.

Baca, M., Doan, K., Sobczyk, M., Stankovic, A., Węgleński, P., 2012. Ancient DNA reveals kinship burial patterns of a pre-Columbian Andean community. BMC Genet. 13, 30.

Barnes, E., 1994. Polydactyly in the Southwest. Kiva 59, 419–431. https://doi.org/10.1080/00231940.1994.11758248.

Barnes, E., 2012. Developmental Field Anomalies of the Human Skeleton: a Paleopathology Perspective. Wiley-Blackwell, Hoboken, New Jersey.

Bartolomei, F.J., 1990. Surgical correction of brachymetatarsia. J. Am. Podiatr. Med. Assoc. 80, 76–82. https://doi.org/10.7547/87507315-80-2-76.

Bell, J., 1951. On brachydactyly and symphalangism. In: Penrose, L.S. (Ed.), Treasury of Human Inheritance. University Press, Cambridge, pp. 1–17.

Benham, W.G., 1901. The Laws of Scientific Hand Reading. The Knickerbocker Press, New York.

Bosch, D.G.M., van Nieuwenhoven, C.A., Hoogeboom, A.J.M., 2011. Brachydactyly: a rate complication of sickle cell anemia. Clin. Dysmorphol. 20, 172–173. https://doi.org/10.1097/MCD.0b013e328345895f.

Brailsford, J.F., 1945. Familial brachydactyly. Br. J. Radiol. XVIII, 167–172.

Buikstra, J.E., Ubelaker, D.H., 1994. Standards for data collection from human skeletal remains. Arkansas Archeological Survey Research Series No. 44. Fayetteville, Arkansas.

Burnett, S.E., 2005. Developmental Variation in South African Bantu: Variant Co-occurrence and Skeletal Asymmetry. PhD Dissertation. Arizona State University, Tempe.

Case, D.T., 1996. Developmental Defects of the Hands and Feet in Paleopathology. MA Thesis. Arizona State University, Tempe.

Case, D.T., Ossenberg, N.S., Burnett, S.E., 1998. Os intermetatarseum: a heritable accessory bone of the human foot. Am. J. Phys. Anthropol. 107, 199–209. https://doi.org/10.1002/(SICI)1096-8644(199810)107, 2<199::AID-AJPA6>3.0.CO;2-Q.

Case, D.T., Jones, L.B., Offenbecker, A.M., 2017. Skeletal kinship analysis using developmental anomalies of the foot. Int. J. Osteoarchaeol. 27, 192–205. https://doi.org/10.1002/oa.2529.

Cybulski, J.S., 1988. Brachydactyly, a possible inherited anomaly at prehistoric Prince Rupert Harbour. Am. J. Phys. Anthropol. 76, 363–376. https://doi.org/10.1002/ajpa.1330760309.

Farabee, W.C., 1903. Hereditary and Sexual Influences in Meristic Variation: a Study of Digital Malformations in Man. Doctoral Dissertation. Harvard University, Cambridge.

Fitch, N., 1979. Classification and identification of inherited brachydactylies. J. Med. Genet. 16, 36–44. https://doi.org/10.1136/jmg.16.1.36.

Garn, S.M., Hertzog, K.P., Poznanski, A.K., Nagy, J.M., 1972. Metacarpophalangeal length in the evaluation of skeletal malformation. Radiology 105, 375–381. https://doi.org/10.1148/105.2.375.

Genetic and Rare Diseases Information Center (GARD), 2016a. Reference for Pseudohypoparathyroidism, #10758 [Last Update: 07/13/2016]. (accessed 03/06/2019). https://rarediseases.info.nih.gov/diseases/10758/pseudohypoparathyroidism.

Genetic and Rare Diseases Information Center (GARD), 2016b. Reference for Pseudopseudohypoparathyroidism, #7860 [Last Update: 03/09/2016]. (accessed 03/06/2019). https://rarediseases.info.nih.gov/diseases/7860/pseudopseudohypoparathyroidism.

Genetic and Rare Diseases Information Center (GARD), 2016c. Reference for Acrodysostosis, #5724 [Last Update: 11/01/2016]. (accessed 12/16/2020). https://rarediseases.info.nih.gov/diseases/5724/acrodysostosis.

Genetic and Rare Diseases Information Center (GARD), 2016d. Reference for Turner Syndrome, #7831 [Last Update: 07/13/2016]. (accessed 03/06/2019). https://rarediseases.info.nih.gov/diseases/7831/turner-syndrome.
Genetic and Rare Diseases Information Center (GARD), 2018a. Reference for Brachydactyly Type E, #987 [Last Update: 04/22/2018] (accessed 03/06/2019). https://rarediseases.info.nih.gov/diseases/987/brachydactyly-type-e.
Genetic and Rare Diseases Information Center (GARD), 2018b. Reference for Pseudohypoparathyroidism, #5770 [Last Update: 01/03/2018] (accessed 12/16/2020). https://rarediseases.info.nih.gov/diseases/5770/albrights-hereditary-osteodystrophy.
Giuffra, V., Bianucci, R., Milanese, M., Tognotti, E., Montella, A., Caramella, D., Fornaciari, G., Bandiera, P., 2014. A case of brachymetatarsia from Medieval Sardinia (Italy). Anat. Rec. 297, 650–652. https://doi.org/10.1002/ar.22883.
Greenfield, G.B., 1980. In: Lippincott, J.B. (Ed.), Radiology of Bone Diseases, third ed. Philadelphia.
Harris, R., Beath, T., 1949. The short first metatarsal: its incidence and clinical significance. J. Bone Jt. Surg. 31, 553–565.
Holt, S.B., 1975. Polydactyly and brachymetapody in two English families. J. Med. Genet. 12, 355–366.
Huhsunqu 2009. http://upload.wikimedia.org/wikipedia/commons/7/7b/Perulocationmap.svg.
Ibarra Asencios, B., 2021. A Diachronic Study of Mortuary Variability in the North Highlands of Peru (AD 200-1600): Implications for Understanding Funeral Practices and Ancestor Veneration. PhD Dissertation. Department of Anthropology, Tulane University, New Orleans.
Johnson, D., S-h, Kan, Oldridge, M., Tembath, R.C., Roche, P., Esnouf, R.M., Giele, H., Wilkie, A.O.M., 2003. Missense mutations in the homeodomain of HOXD13 are associated with brachydactyly types D and E. Am. J. Hum. Genet. 72, 984–997. https://doi.org/10.1086/374721.
Kozieradzka-Ogunmakin, I., 2011. Multiple epiphyseal dysplasia in an Old Kingdom Egyptian skeleton: a case report. Int. J. Paleopathol. 1, 200–206. https://doi.org/10.1016/j.ijpp.2011.10.002.
Laurencikas, E., Rosenborg, M., 2000. Swedish metacarpophalangeal standards compared with previously published norms. Acta radiol. 41, 498–502. https://doi.org/10.1080/028418500127345785.
Loveland, C.J., Gregg, J.B., 1988. Brachydactyly in a prehistoric Texas skeleton. Plains Anthropol. 33, 399–404. https://doi.org/10.1080/2052546.1988.11909404.
Mah, K.K., Beegle, T.R., Falknor, D.W., 1983. A correction for short fourth metatarsal. J. Am. Podiatry Assoc. 73, 196–200. https://doi.org/10.7547/87507315-73-4-196.
Merbs, C.F., Vestergaard, E.M., 1985. The paleopathology of sundown, a prehistoric site near Prescott, Arizona. In: Merbs, C.F., Miller, R.J. (Eds.), Health and Disease in the Prehistoric Southwest. Anthropological Research Papers No. 34. Arizona State University, Tempe, pp. 85–103.
Munuera Martínez, P.V., Lafuente Sotillos, G., Dominguez Maldonado, G., Salcini Macias, J.L., Martionez Camuna, L., 2004. Morphofunctional study of brachymetatarsia of the fourth metatarsal. J. Am. Podiatr. Med. Assoc. 94, 347–352.
Online Mendelian Inheritance in Man, OMIM®, 2016a. Johns Hopkins University, Baltimore, MD. MIM Number: 103580 (Albright Hereditary Osteodystrophy with Multiple Hormone Resistance): 04/06/2016 (accessed 12.06.2020). https://omim.org/entry/101800.
Online Mendelian Inheritance in Man, OMIM®, 2016b. Johns Hopkins University, Baltimore, MD. MIM Number: 612463 (Pseudopseudohypoparathyroidism (PPHP)): 04/06/2016 (accessed 12.06.2020). https://omim.org/entry/612463.
Online Mendelian Inheritance in Man, OMIM®, 2016c. Johns Hopkins University, Baltimore, MD. MIM Number: 101800 (Acrodysostosis 1 with or Without Hormone Resistance): 07/09/2016. https://omim.org/entry/101800.
Online Mendelian Inheritance in Man, OMIM®, 2020. Johns Hopkins University, Baltimore, MD. MIM Number: 113300 (Brachydactyly, Type E1): 02/06/2020 (accessed 11.27.2020). http://omim.org/entry/113300.
Pareda, A., Garin, I., Garcia-Barcina, M., Gener, B., Beristain, E., Miren Ibañez, A., Perez de Nanclares, G., 2013. Brachydactyly E: isolated or as a feature of a syndrome. Orphanet J. Rare Dis. 8, 141. https://doi.org/10.1186/1750-1172-8-141.
Pokotylo, D.L., Binkley, M.E., Curtin, A.J., 1987. The Cache Creek Burial Site (EeRh 1), British Columbia. Contributions to Human History. British Columbia Provincial Museum, Victoria, B.C. Canada.
Poznanski, A.K., Garn, S.M., Nagy, J.M., Gall, J.C., 1972. Metacarpophalangeal pattern profiles in the evaluation of skeletal malformations. Radiology 104, 1–11. https://doi.org/10.1148/104.1.1.
Poznanski, A.K., Werder, E.A., Giedion, A., Martin, A., Shaw, H., 1977. The pattern of shortening of the bones of the hand in PHP and PPHP-a comparison with brachydactyly E, turner syndrome, and acrodysostosis. Radiology 123, 707–718. https://doi.org/10.1148/123.3.707.
Reed, E.K., 1981. Human skeletal material. In: Hays, A.C. (Ed.), Contributions to Gran Quivira Archaeology. National Park Service, U.S. Department of the Interior, Washington D.C, pp. 75–118.
Schimizzi, A., Brage, M., 2004. Brachymetatarsia. Foot Ankle Clin. N. Am. 9, 555–570. https://doi.org/10.1016/j.fcl.2004.05.002.
Stevenson, R.E. (Ed.), 2006. Human Malformations and Related Anomalies. Oxford University Press, Oxford.
Stojanowski, C.M., Schillaci, M.A., 2006. Phenotypic approaches for understanding patterns of intracemetery biological variation. Yearb. Phys. Anthropol. 49, 49–88. https://doi.org/10.1002/ajpa.20517.
Suresh, S.S., Abraham, R., Ravi, P., 2009. Isolated symmetrical brachymetacarpia of the thumb-case report. Hand 4, 424–426. https://doi.org/10.1007/s11552-009-9185-8.
Temtamy, S.A., Aglan, M.S., 2008. Brachydactyly. Orphanet. J. Rare Dis. 3, 15. https://doi.org/10.1186/1750-1172-3-15.
Temtamy, S.A., McKusick, V.A., 1978. The genetics of hand malformations. BDOAS XIV, 1.
Titelbaum, A.R., Ibarra, B., Naji, S., 2015. Madelung's deformity and possible Leri-Weill dyschondrosteosis: two cases from a Late Intermediate period tomb, Ancash, Peru. Int. J. Paleopathol. 9, 8–14. https://doi.org/10.1016/j.ijpp.2014.11.004).
Urano, Y., Kobayashi, A., 1978. Bone-lengthening for shortness of the fourth toe. J. Bone Joint Surg. Am. 60, 90–93. PMID: 624764.
Velasco, M.C., 2018. Open sepulchers and closed boundaries? Biodistance analysis of cemetery structure and postmarital residence in the late prehispanic Andes. Am. J. Phys. Anthropol. 166, 906–920. https://doi.org/10.1002/ajpa.23594.