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Case Study

Madelung's deformity and possible Léri-Weill dyschondrosteosis: Two cases from a Late Intermediate period tomb, Ancash, Peru

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ABSTRACT

Two individuals with bilateral Madelung's deformity were identified in a Late Intermediate period comingled tomb at the northern highland site of Marcajirca, Ancash, Peru (ca. AD 1250). Comparisons of the size and robusticity of the radii and ulnae suggest the individuals represent a male and a female. The difference in the severity of the changes is thought to represent variability in the expression of the deformity seen in males and females in clinical cases. Three comparatively short, thick tibiae were also recovered from this tomb, which may suggest that the individuals demonstrate Léri-Weill dyschondrosteosis, a type of dwarfism characterized by mesomelic shortening. These are the first examples of Madelung's deformity to be described from an archaeological context in South America and offer an insight into the use of tombs (*chullpas*) in Late Intermediate period Ancash.

1. Introduction

This report describes the unusual morphology of two sets of radii and ulnae recovered from a Late Intermediate period (ca. AD 1250) tomb at the northern highland site of Marcajirca in Ancash, Peru (Fig. 1). Situated at the top of a steep-sided, rocky mountain slope on the eastern side of the Cordillera Blanca, the site comprises residential, public, and funerary areas. To date, comingled human skeletal remains have been found in 22 funerary caves and 35 walled tombs (*chullpas)* (Ibarra, 2009).

Chullpa 26 yielded over 1500 disarticulated bones, representing a minimum of 27 adult individuals. Of the 43 adult radii (26 right, 17 left) and 36 adult ulnae (21 right, 15 left) recovered from this tomb, four radii and two ulnae are malformed and comparatively short and thick. The pathological bones appear to be from two individuals, with each individual represented by a right and left radius, and a right ulna. None of the other radii ($n = 158$) or ulnae ($n = 152$) recovered from four additional tombs and one cave demonstrate this anomaly. In addition to the forearm deformities, three comparatively short, thick tibiae were also recovered from *Chullpa* 26. No unusual humeri, femora, metacarpals, metatarsals, or phalanges were observed.

The purpose of this investigation is twofold: first, to perform a differential diagnosis of the deformed bones, and secondly, to understand the bioarchaeological implications of the diagnosis. If the diagnosis is a heritable condition, the presence of two individuals with the same condition in the same tomb can offer insights into the use of Late Intermediate period tombs in the northern highlands of Peru.

2. Materials and methods

A total of 263 adult long bones were available for examination from *Chullpa* 26. Adult bones were identified as having complete fusion of proximal and distal epiphyses. Preservation of the remains was very good, with the exception of the fibulae, which were often too damaged for observation. Measurements of each long bone were taken following the guidelines given in Buikstra and Ubelaker (1994). Radiography was not available.

The presence of malformed and comparatively short but thick radii (2 right, 2 left) and ulnae (2 right) were noted during the initial inventory of the material (Figs. 2–3). Articulation of the bones identified two pairs of radii, each with an associated right ulna. One pair (Individual 1) is shorter and more gracile with a more pronounced deformity, whereas the other pair (Individual 2) is longer and more robust with a less pronounced deformity. Based on size and

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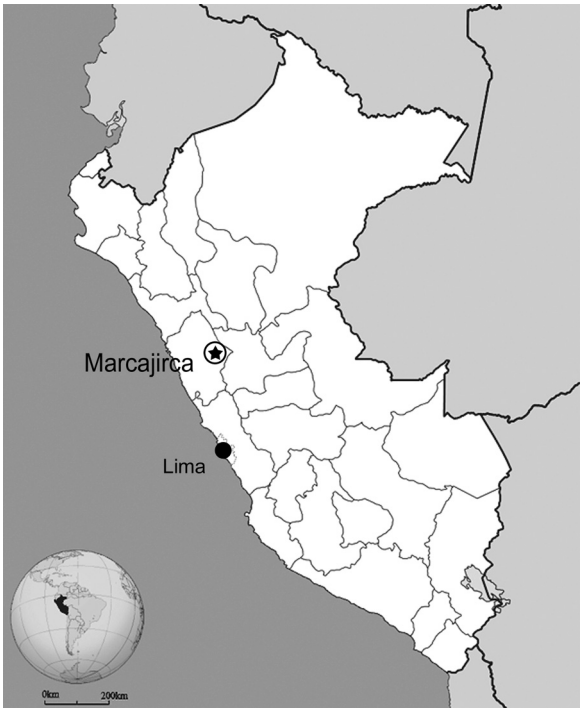

Fig. 1. Location of Marcajirca, Peru.
Modified from Huhsunqu (2009).

robusticity, it is reasonable to suggest that Individual 1 is female and Individual 2 is male.

In addition to the deformed radii and ulnae, one right and two left tibiae from C*hullpa* 26 were found to be shorter yet thicker than the other tibiae from the tomb (Fig. 4). Morphological comparison of the three tibiae determined that they represent two individuals: one smaller and more gracile, and the other larger and more robust. Again, they seem to indicate a male and a female and as these deformities are not found in any of the other samples, it is probable the tibiae and arm bones are from the same two individuals. No other long bones were found to be unusually short or deformed.

Fig. 2. Malformed and short radii compared with normal, average length radii.

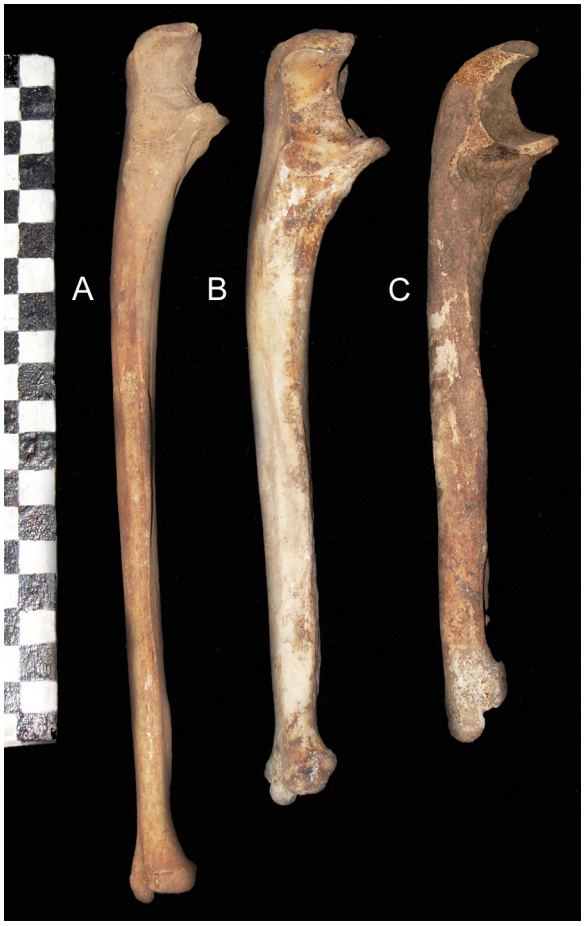

Fig. 3. Malformed and short ulnae compared with normal, average length ulnae. A, Normal; B, Individual 2, C, Individual 1.

3. Results

The affected radii and ulnae have pronounced interosseous crests and strong muscle attachment sites, in particular for brachialis, biceps brachii, pronator quadratus, and pronator teres. The radii bow laterally, and the proximal circumferential articular surface is underdeveloped on all radii, particularly along the lateral margin (Fig. 5). The distal radial articular surfaces have a marked medial deviation, giving them a triangular appearance that likely would have caused the carpals to have a V-shaped formation (Fig. 6). There appears to be posterior dislocation of the ulnar articulation on each radius.

The radii of Individual 1 demonstrate marked lateral bowing. While the proximal end of the left radius is broken postmortem, the right radial head is hypoplastic. The medial side of the articular fovea on the head of the right radius has an osteophyte that articulates with the radial notch of the ulna (Fig. 5A). At the distal end of the interosseous crest of each radius is an elliptical fossa with new bone formation on its floor; the left radial fossa measures a maximum of 11.41 mm long by 6.17 mm wide with a depth of 5.15 mm, and the right radial fossa measures a maximum of 10.25 mm long by 7.51 mm wide with a depth of 5.77 mm (Fig. 6A). The radii lack distinct ulnar notches. The ulna has a deep proximal radial notch and a prominent enthesis on the ulnar tuberosity, at the attachment for brachialis. The distal epiphysis is enlarged with a thickened

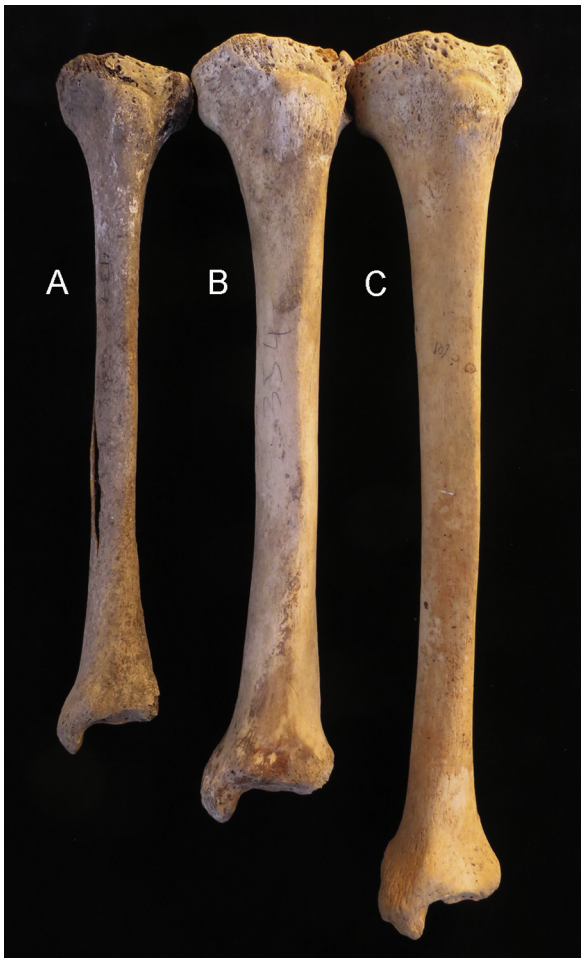

Fig. 4. Short, thick tibiae compared with normal, average length tibia. A, Individual 1; B, Individual 2; C, Normal.

styloid process (10.78 mm in diameter) that is almost equal in size to the ulnar head (12.66 mm in diameter) (Fig. 7C).

The radii of Individual 2 demonstrate similar but less marked features to those of Individual 1(Figs. 2B, 4B). The shafts demonstrate lateral bowing and the heads and circumferential articular surfaces are underdeveloped. Distally the ulnar notches are indistinct and there is an elliptical fossa that measures a maximum of 6.08 mm long by 3.37 mm wide with a depth of 2.94 mm (Fig. 6B). The left radius does not display a fossa.

Like the radii, the ulna demonstrates similar but less pronounced features to that of Individual 1 (Fig. 3B). The distal end is also distorted with a thick styloid process (Fig. 7B). The

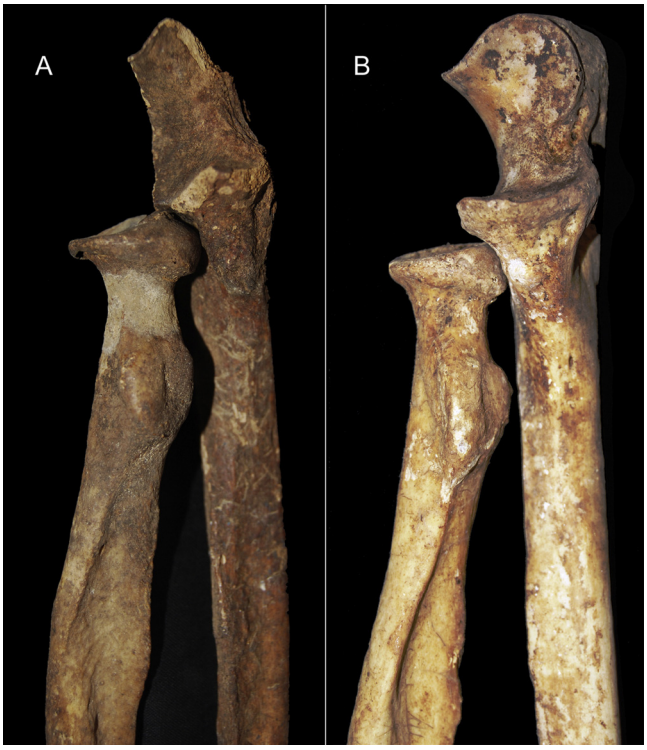

Fig. 5. Proximal radio-ulnar articulation, right arm. A, Individual 1; B, Individual 2.

maximum diameter of the styloid process is 9.01 mm, compared to the maximum diameter of the head at 16.41 mm. The shaft of this ulna is notably robust.

When compared to the average measurements in the sample (Table 1), the affected radii and ulnae are shorter but have greater medio-lateral shaft diameters (ML). While the anterio-posterior (AP) diameters of the affected radii were similar to the sample mean, the AP diameters of the ulnae were slightly greater.

The radius of Individual 1 is 42 mm shorter with a 6 mm greater ML diameter than the mean. The ulna is 36 mm shorter with a 3 mm greater ML diameter and a 4 mm greater AP diameter than the mean.

For Individual 2, the radii were shorter than the mean (right: 21 mm shorter; left: 17 mm shorter), with a greater ML diameter (right, 7 mm wider; left, 5 mm wider). The ulna was 15 mm shorter than the mean, with a 3 mm wider ML diameter and a 5 mm wider AP diameter.

Similar comparisons were carried out for the affected tibiae (Table 2). It is apparent that while the tibiae for both individuals are shorter than the combined average for males and females, they

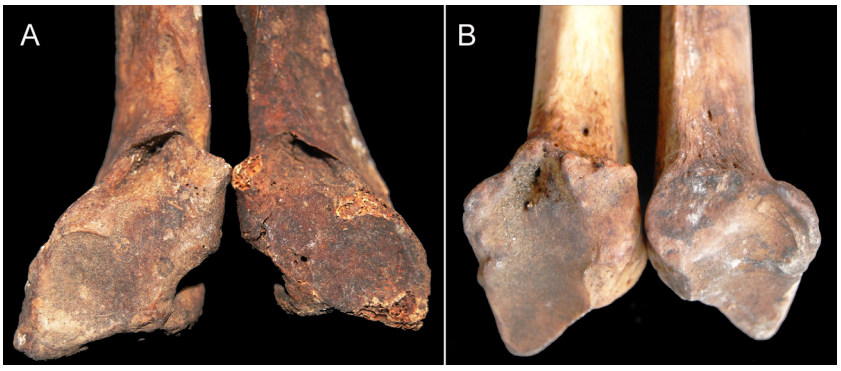

Fig. 6. Distal right and left radii. Medial view. A, Individual 1; B, Individual 2.

Table 1
Measurements of radii and ulnae (mm).

Bone	Maximum length	Range	ML @ mid-shaft	Range	AP @ mid-shaft	Range
Right radius						
Individual 1	176		19		12	
Individual 2	197		20		12	
Adult average[*]	218 (*n* = 51)	183–244	13 (*n* = 58)	10–16	11 (*n* = 58)	9–14
Left radius						
Individual 1	(broken)		–		–	
Individual 2	203		18		12	
Adult average[*]	216 (*n* = 39)	184–260	13 (*n* = 54)	9–18	11.00 (*n* = 54)	8–16
Right ulna						
Individual 1	202		15		17	
Individual 2	223		14		18	
Adult average[*]	238 (*n* = 33)	208–260	12 (*n* = 50)	8–16	13 (*n* = 50)	9–18

ML, medial–lateral diameter; AP, anterior–posterior diameter.
[*] Adult averages based on measurements of sided but unsexed adult radii and ulnae from three comingled tombs (*Cave 3, Chullpa 7, and Chullpa 26*)

Table 2
Measurements of tibiae (mm).

Bone	Condylo-maleolar length	Range	Circumference @ nutrient foramen	Range
Right Tibia				
Individual 1	278		83	
Adult average[*]	329 (*n* = 48)	275–370	83 (*n* = 68)	70–100
Left Tibia				
Individual 1	286		83	
Individual 2	312		111	
Adult average[*]	327 (*n* = 46)	287–381	82 (*n* = 71)	65–99

[*] Comparative mean length based on the averaged measurements of sided but unsexed adult tibiae, from three comingled tombs (*Cave 3, Chullpa 7, and Chullpa 26*).

demonstrate greater circumferences at mid-shaft. In particular, the tibia of Individual 2 is 15 mm shorter than the combined average, but it has a circumference that is 29 mm greater. Although associations between comingled limbs are speculative without DNA analysis, there is a strong possibility that Individual 1 may be associated with the pair of shorter tibiae, whereas Individual 2 may be associated with the relatively short and dense, robust left tibia.

3.1. Differential diagnosis

Various etiologies were considered to account for the malformed radii and ulnae. The observed deformities could have resulted from trauma, or they may be associated with hereditary multiple exostosis, chromosomal abnormalities, and skeletal dysplasia.

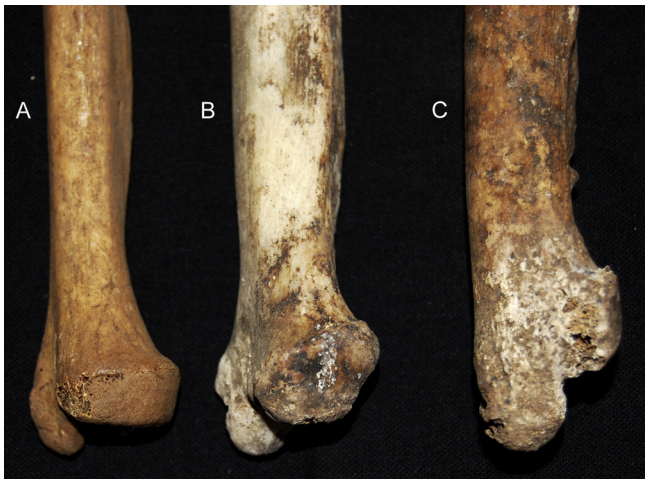

Fig. 7. Distal right ulnae. Anterolateral view. A, Normal; B, Individual 2, C, Individual 1.

Fracture malalignment, may have caused deformities of the shaft and distal epiphysis of the radii (Anderson and Carter, 1995), but the lack of fracture lines or bony calluses rules out this diagnosis. Furthermore, trauma is more likely to occur unilaterally and less likely to occur bilaterally in two individuals with nearly symmetrical presentation.

Forearm and wrist deformities may occur with hereditary multiple exostosis (HME). This condition manifests as numerous projections of bone and cartilage, most often at the knee. Exostoses are typically found on the metaphyses of long bones, but can also occur on the diaphyses. Leg, forearm, wrist, and hand deformities are frequent, and the radius may be longer than the ulna (OMIM, 1986a; Campo et al., 1996). The lack of multiple exostosis on the affected bones, or on any of the bones recovered from the tomb excludes HME as a likely diagnosis.

Wrist deformities can be present with Turner syndrome, a chromosomal abnormality that affects females and caused by the absence or incompleteness of one of the two X-chromosomes (Poznanski and Holt, 1971). Individuals with Turner syndrome have short stature with possible epiphyseal deformities, cubitus valgus, abnormalities of the palate, dysplastic carpals, and shortening of the metacarpals, particularly the fourth. Most cases of Turner syndrome are not inherited, and mesomelic skeletal features are absent or rare (Ogata et al., 2002). As one of the cases involves a probable male, and no abnormal carpals, metacarpals, maxilli, femora or humeri were identified, despite many of these bones being recovered from the tomb, Turner syndrome is an unlikely diagnosis.

Various types of skeletal dysplasia should also be considered, including achondroplasia which is the most common form (Campo et al., 1996). Systemic growth disturbances such as achondroplasia are ruled out as no other dysplastic bone elements were recovered.

Radial longitudinal deficiency ('radial club hand') is a congenital deformity that results from a deficiency of growth in the radial ray, resulting in the abnormal development, underdevelopment, or absence or hypoplasia of the radius, lateral carpal bones, and thumb (Bednar et al., 2009). As a result of this condition, the hand

Fig. 8. Radius and ulna, right arm. A, Individual 1; B, Individual 2.

and wrist demonstrate a characteristic radial deviation. Since the radii in question display a distinct ulnar tilt of the distal epiphyses, radial dysplasia is discounted as a differential diagnosis.

Malformed epiphyses also occur with multiple epiphyseal dysplasia. This condition is characterized by a disorder of cartilage and bone development that results in irregular epiphyses of the limb bones, and is associated with early onset of degenerative joint disease, particularly in the hips and knees (OMIM, 1986b). In a clinical setting, a sloping end of a tibia can lead to a diagnosis of this disorder, due to the deficiency in the ossification of a portion of the distal tibial epiphysis. Wrists and hands are often normal in the mild form of this disorder, whereas they may be short and stubby in the more severe form, and associated with other abnormalities, such as clubfoot, cleft palate, radial ray deficiency, or scoliosis. Since no additional dysplastic epiphyses were observed among the bones from the tomb, it is unlikely that this condition explains the observed forearm deformities. It appears that Individuals 1 and 2 both demonstrate bilateral Madelung's deformity, a rare anomaly of the forearm and wrist (Fig. 8). Described in 1878 by German surgeon Otto W. Madelung, Madelung's deformity (MD) is due to an arrest of growth at the antero-medial aspect of the distal epiphyseal plate of the radius, causing abnormalities of the radius, ulna, and carpal bones (Langer, 1965). Ranging in expression (Zebala et al., 2007), the physical deformity is characterized by shortened and bowed radii and dorsally displaced distal ulnae, resulting in a posterior protuberance of the ulnar head in the wrist of an affected individual (Anton et al., 1938).

The radiographic criteria for a diagnosis of MD include: double bowing of the radius (dorsal and lateral), an enlarged interosseous space between the radius and ulna, shortened radius and ulna, premature fusion of the medial distal radial epiphysis causing a triangulation of the normally rectangular epiphysis, localized decreased bone density on the distal ulnar border of the radius extending for a short distance proximal to the epiphyseal line, osteophyte formation on the ulnar aspect of the distal radius near the epiphyseal line, an antero-medial angulation of the distal radial articular surface, posterior dislocation of the distal radio-ulnar articulation, deformity of the ulnar head and styloid process, and wedging of the carpal bones between the radius and ulna resulting in a V-shape or triangular configuration with the lunate at the apex (Dannenberg et al., 1939). Also described is hypoplasia and flattening of the radial head (Lamy and Maroteaux, 1960), and enlargement and distortion of the distal ulna, with an abnormally dense distal ulnar epiphysis (Henry and Thorburn, 1967).

In their surgical treatment of patients with MD, Vickers and Nielsen (1992) described the presence of an abnormal thickened ligament in the wrist, extending from the anteromedial metaphysis of the radius to the lunate. Now referred to as Vicker's ligament, this fibrous band is approximately 5 mm in diameter. Surgical release of the band in adolescent patients has been found to be an effective treatment for the deformity lending support to the possibility that Vicker's ligament is a contributing factor (Vickers and Nielsen, 1992; Harley et al., 2006). It is possible that the elliptical fossae described in the present cases, and seen in other paleopathological descriptions of MD (e.g., Campo et al., 1996; Canci et al., 2002; Cummings and Rega, 2008), is associated with the presence of the ligament.

Madelung's deformity causes a restriction in wrist movement, and the more severe the deformity, greater the limitations of movement. Patients commonly present with the loss of extension and ulnar deviation, although pronation, supination, and radial deviation may also be affected (Anton et al., 1938; Vickers and Nielsen, 1992; Zebala et al., 2007). While some patients with the deformity may be asymptomatic, some report progressive stiffness and pain, a reduction in grip strength, and concern over physical appearance (Henry and Thorburn, 1967; Plafki et al., 2000; Villeco, 2002; Ghatan and Hanel, 2013). Rupture of the extensor tendons of the fingers has also been reported in some cases of severe deformity (Ducloyer et al., 1991).

Madelung's deformity may be congenital or may result from a traumatic event. Whereas traumatic MD is usually unilateral, congenital MD is typically bilateral and may be either associated with bone dysplasia or be idiopathic in its etiology. Congenital forms of the condition are not apparent during infancy or childhood; rather the deformity slowly develops during growth becoming evident by adolescence. While congenital MD can affect both sexes, it is found more frequently in females (Anton et al., 1938).

While some types of skeletal dysplasia affect the development of an entire limb, other types affect a portion of a limb. In these cases, rhizomelia refers to growth disturbances of the upper segments (humerus and femur), mesomelia describes the relative shortening of the middle segments (bones of the forearm and lower leg), and acromelia refers to the relative shortening of the distal segments (fingers and toes). Following from this, rhizomesomelia refers to growth disturbances of the upper and middle segments, whereas acromesomelia refers to growth disturbances of the middle and distal segments. In the current investigation, acromesomelic and rhizomesomelic forms of dysplasia can be ruled out, since no dysplastic humeri, femora, metacarpals, or metatarsals were recovered from the tomb.

Mesomelic dysplasias associated with MD include Langer mesomelic dyplasia and Léri-Weill dyschondrosteosis. Langer mesomelic dysplasia is due to a homozygous defect or deletion of the short stature homeobox containing gene (SHOX or SHOXY) (Belin et al., 1998; OMIM, 1986c). In addition to MD, individuals with Langer mesomelic dysplasia have severely shortened bones of the forearm and lower leg. The radius and the tibia are short,

Table 3
Previously reported cases of Madelung's deformity and Léri-Weil dyschondrosteosis in the paleopathological literature.

	Location	Chronology	Sex	Age	MD	Diagnosis	Reference
1	England	(Medieval)	Male	Older adult (40–50)	Yes(unilateral)	Post-traumatic	Anderson and Carter (1995)
2	Spain	AD 1000–1200	Male	Middle adult(30–50)	Yes (bilateral)	Idiopathic	Campo et al. (1996)
3	Spain	200–100 BC	Female	Young adult (ca 23)	Yes?(bilateral)	Multiple Epiphyseal Dysplasia? Idiopathic?	Campillo and Malgosa (1991)
4	Italy	800 BC	Male	Older adult (45–50)	Yes (bilateral)	Indeterminate (missing legs)	Canci et al. (2002)
5	Switzerland	AD 500–600	Female	Older adult (ca 60)	Yes (bilateral)	LWD	Lagier et al. (1978)
6	England	AD 100–200	Male	Young adult (20–25)	Yes (bilateral)	LWD	Waldron (2000)
7	England	AD 800–1000	Female	Middle adult(35–44)	Yes (bilateral)	LWD	Cummings and Rega (2008)
8	Netherlands	AD 78–233	Female	Middle adult (ca 35)	No	LWD	Bianucci et al. (2012)
9	Switzerland	Late Neoltihic 5155 ± 45 cal BP	Indeterminate	Sub adult (12)	No	LWD	Milella et al. (2013)

thick, and curved, whereas the ulna and fibula tend to be severely underdeveloped or absent, and mild hypoplasia of the mandible may also be observed (Langer, 1967; OMIM, 1986d). Bearing some similarities, the forearms from *Chullpa* 26 demonstrate a milder expression than what is described for Langer mesomelic dysplasia.

With a similar but less severe phenotype than Langer mesomelic dysplasia, Léri-Weill dyschondrosteosis (LWD) is due to the pseudoautosomal dominant inheritance of a heterozygous defect or deletion of the short stature homeobox containing gene (SHOX or SHOXY), or by a deletion in the SHOX downstream regulatory domain (Belin et al., 1998; Shears et al., 1998; OMIM, 1986d). First described in 1929 by Léri and Weill, LWD is characterized by short stature, mesomelia, and Madelung deformity. The shortening of the forearm tends to be proportionately greater than that of the lower leg (Langer, 1965). Additional findings may include exostoses, particularly on the proximal medial tibia (Waldron, 2000; Léri and Weill, 1929).

Léri-Weill dyschondrosteosis expresses phenotypic variation, even within the same family, likely due the role of sex steroids in skeletal development, background genetic effects, and environmental factors (Shears et al., 1998). Occurring in both sexes, the defect is bilateral and more common and more severely expressed in females, possibly due to sex differences in estrogen levels (OMIM 1986d). Males with LWD may demonstrate muscular hypertrophy (Schiller et al., 2000). The prevalence of this condition is not known.

Like Langer mesomelic dysplasia and LWD, idiopathic MD is also due to a mutation or deletion in the homeobox gene SHOX (Blaschke and Rappold, 2000). However, individuals with the idiopathic form of MD do not demonstrate mesomelia. Nixon (1983) reported MD with dominant familial inheritance that was not associated with bone dysplasia. Plafki et al. (2000) also reported a family with bilateral primary MD within five generations, affecting both females and males, and indicating autosomal dominant inheritance.

It is unfortunate that the tibiae described in our cases are not definitively associated with the forearms, since a diagnosis of LWD cannot be stated with certainty. Nevertheless, LWD deserves strong consideration due to the bilateral nature of the defects, and due to the fact that two individuals in the same tomb present the same deformity, implying a hereditary transmission.

4. Discussion

Madelung's deformity and Léri-Weill dyschondrosteosis are rarely described in the paleopathological literature (Table 3). Six cases of MD have been reported. Of those, one is the result of trauma (Anderson and Carter, 1995), one is idiopathic (Campo et al., 1996), and at least three are associated with LWD (Lagier et al., 1978; Waldron, 2000; Cummings and Rega, 2008). Of the five non-traumatic cases, two are adult females and three are adult males, from England (2), Spain (1), Switzerland (1), and Italy (1), ranging from 800 BC to AD 1000–1200. Another possible idiopathic example of bilateral MD was observed in a young adult female from the Talayotic site of S'Illot de Porros, Mallorca, Spain (200–100 BC) (Campillo and Malgosa, 1991), although the authors suggested that this may be a case of multiple epiphyseal dysplasia. Two additional cases of LWD without MD have been reported from Switzerland (Milella et al., 2013) and the Netherlands (Bianucci et al., 2012), including the earliest example of LWD, dating to the Late Swiss Neolithic (5155 ± 45 cal BP) (Milella et al., 2013).

In the cases described here, Madelung's deformity would have presented as a minor physical deformity with limitations in wrist movement and pain during adolescence. In spite of the deformity, the presence of MD would have not precluded a fully functional and active life, particularly since limitations in wrist movement can be overcome through compensatory movements at the shoulder. Considering the role of the two affected individuals in the community, it is likely that they did demonstrate physical differences from other individuals, particularly with the visible MD and the plausible LWD. Nevertheless, there is no reason to believe that the individuals suffered physical or social disadvantage. Not only did the two individuals live into adulthood, they were given normative burial treatment, testifying to their membership in the community. Importantly, the presence of two individuals with a heritable condition in a single *chullpa* offers insight into tomb use during the Late Intermediate period in the northern highlands of Peru. The use of these above ground mortuary monuments arose during the Early Intermediate period and spread across the Central Andes, becoming the predominant mortuary structure during the Late Intermediate period (Isbell, 1997). It is logical to suggest that tombs were used by kin groups or individuals with a shared ethnic identity; however, such suggestions of prehistoric social structure tend to be based on Colonial records and ethnographic analogy, and less frequently on the human remains themselves. DNA analysis is destructive and often cost prohibitive, although occasional studies have been conducted. One recent investigation was conducted by Baca et al. (2012), to determine if tombs served as family graves at the Late Horizon site Tompullo 2 in Arequipa, Peru. DNA analysis on 41 individuals from six *chullpas* suggested that each tomb contained members of a single patrilineal family group.

The bioarchaeological evidence presented here offers new insight into tomb use. Two cases of heritable MD and possible LWD from one tomb demonstrates that familial relationship was a factor that influenced mortuary practices at Marcajirca. How the individuals were related is not clear, though it is highly likely that they belonged to the same familial group.

5. Conclusions

Macroscopic examination of the bones from *Chullpa* 26 indicates that two individuals exhibit Madelung's deformity, a developmental anomaly of the wrist. These are the first examples of MD to be described from an archaeological context in South America and one of eight cases reported worldwide. Based on size, robusticity, and the expression of the deformity, the individuals were likely a female and a male. Measurements of the comingled tibiae from the tomb identified three that were shorter and thicker than the rest of the tibiae. Although not definitive without DNA analysis, it is probable that the three tibiae demonstrate mesomelic shortening and are associated with the individuals with the forearm deformities, suggesting a diagnosis of Léri-Weill dyschondrosteosis. The likelihood for either inherited idiopathic MD or MD associated with LWD is supported by the fact that two individuals from one tomb have the same condition. The presence of two individuals with the same heritable defect in one tomb indicates that consanguine relations were interred together, offering insight into tomb use at the Late Intermediate period highland site of Marcajirca in Ancash, Peru.

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