Kroeber Anthropological Society Papers, Nos. 71-72, 1990

Humeral Morphology of Achondroplasia

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Unique humeral morphological features of two prehistoric achondroplastic adult individuals are described. These features are compared to the humerus of a prehistoric non-achondroplastic dwarf and to the humeri of a normal human population sample. A set of unique, derived achondroplastic characteristics is presented. The non-achondroplastic individual is diagnosed as such based on guidelines created by the authors. These guidelines will be useful as an aid in diagnosing achondroplastic individuals from the archaeological record.

INTRODUCTION

For several decades dwarfism has been a prominent topic within the study of paleopathology. It has been represented directly by skeletal evidence and indirectly by artistic representation in the archaeological record (Hoffman and Brunker 1976). Several prehistoric Egyptian and Native American dwarfed skeletons have been recorded, indicating that this pathology is not linked solely with modern society (Brothwell and Sandison 1967; Hoffman and Brunker 1976; Niswander *et al.* 1975; Snow 1943). Artifacts such as paintings, tomb illustrations and statues of dwarfed individuals have been discovered in various parts of the world. However, interpretations of such artifacts are speculative, for it is necessary to allow artistic license for individualistic expression and interpretation. Paintings and sculptures may represent children, for instance. Thus, dwarfism in the prehistoric archaeological record can only be definitively diagnosed through the study of skeletal remains.

Among the several forms of dwarfism, achondroplasia is the most common, both in present and prehistoric times (Brothwell and Sandison 1967; Roberts 1988). Achondroplasia is a specific type of dwarfism classified as rhizomelic micromelia (proximal limb segments are more effected than distal segments). Although all of the limbs are shortened, the humerus and femur are more effected than the distal limb segments. This is due to the fact that in normal individuals the distal limbs grow to a lesser degree and, therefore, appear to be less involved (Rubin 1964). The mechanical effects of normal body mass on shortened limbs leads to a distinct humeral morphology.

The term achondroplasia has often been misused as a general diagnosis for dwarfism (Hall 1986). Some dwarfed specimens have temporarily been diagnosed as achondroplastic, such as in the Hrdlicka Paleopathology Collection in the San Diego Museum of Man (specimens 1915-2-382 and 1915-2-463) (Merbs 1980). The following paper describes a set of humeral morphological characteristics which can be used as a guide to identifying achondroplastic individuals from the archaeological record.

MATERIALS AND METHODS

A comparative population sample, housed by the Lowie Museum of Anthropology (LMA) at the University of California at Berkeley, was derived from a random sample forming a total of sixty adult individuals (thirty males and thirty females) from six different prehistoric archaeological sites within California. Two achondroplastic adult individuals from similar contexts, specimen number 6670 (spc. 6670) and specimen number 9199 (spc. 9199), were analyzed. Spc. 6670 is a complete female skeleton from the Augustine site in Sacramento, California (CA-Sac-127). Spc. 9199 is a left humerus recovered from the Tank site (also called the Topanga or Topanga Canyon site) in Los Angeles, California (CA-LAn-1). A non-achondroplastic dwarf, specimen number 3854 (spc. 3854), was included for comparison in the analysis[1] (Figure 1). Spc. 3854 is from the Mosher Mound site in Sacramento, California (CA-Sac-?). All the individuals are housed by LMA.

Measurements were taken in millimeters using a digital caliper and an osteometric board. Sexing of the individuals was determined by analysis of the pelvis and/or the cranium following the methods outlined by Phenice (1969) and Bass (1987).

DESCRIPTION

The two achondroplastic specimens share several unique features. They both exhibit shortening in the proximal and distal limbs, and both humeral heads exhibit wasting under the inferior border, resulting in distinct mushroom shaped necks. Their humeral lengths range within 155-160 mm, and their deltoid tuberosities are large and rectangular in shape. A tubercle is present on the inferior border of the crest of the lesser tuberosity, at the insertion of teres major muscle (Figure 1). This tubercle is only faintly present in the non-achondroplastic specimen and is absent in the normal humerus (Figure 2).

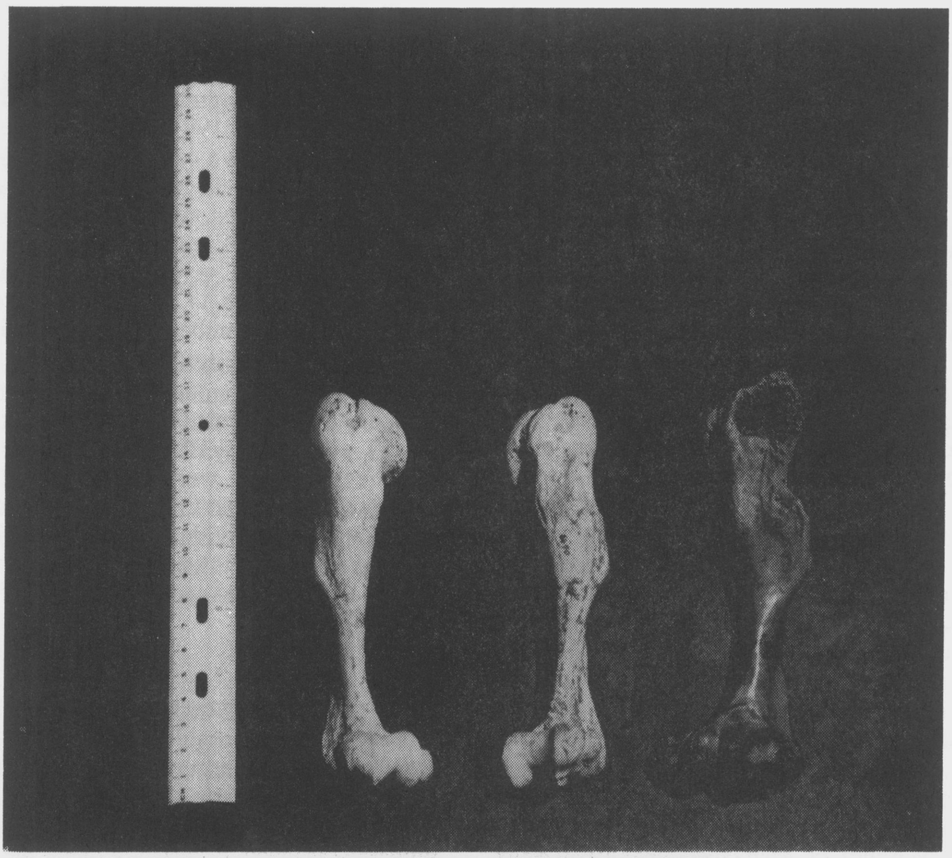
In both achondroplastic specimens the greater and lesser tuberosities are prominent, separated by a broad, shallow intertubercular groove. The shafts are bowed, presenting a medial concavity in anterior view. The epicondylar breadths of the humeri fall within the normal range (Table 1). A groove is present in the midline of the trochlea, located postero-inferiorly. The olecranon fossa is deep and well-developed; however, it is blocked by the medial supracondylar ridge and possesses a narrow, oval opening.

In achondroplastic spc. 6670, humeroradio indices lie within the average of the comparative sample (Table 2). The humeroulnar indices range from 95 to 98. The maximum length of the right humerus of spc. 6670 (158 mm) is nearly equal to the maximum length of the right ulna (155 mm).

Non-achondroplastic spc. 3854 shares some similar morphological features with the achondroplastic individuals. There is a lack of full elbow extension, and, although the proximal limbs are shorter than the distal limbs, the maximum length of the distal limbs falls within the normal range (Table 1). Since the distal limb length exceeds the humeral limb length (Table 2), the humeroradio and humeroulnar indices are greater than 100.

Another similarity spc. 3854 shares with the achondroplastic individuals is that the shafts of the limbs are gracile, particularly on the posterior surface of the semilunar notch of the ulnae and the superior shaft of the radii. Furthermore, arthritis is present in all the limbs. Eburnation is evident on the posterior portion of the left humeral head, while the right humeral head is nearly

Figure 1. Achondroplastic skeletal remains, from left to right: spc. 6670 right humerus; spc. 6670 left humerus; spc. 9199 left humerus.



obliterated by arthritis. Arthritis is also present in the distal metaphysis of the right humerus and on the heads of the distal limbs, especially on the radial heads.

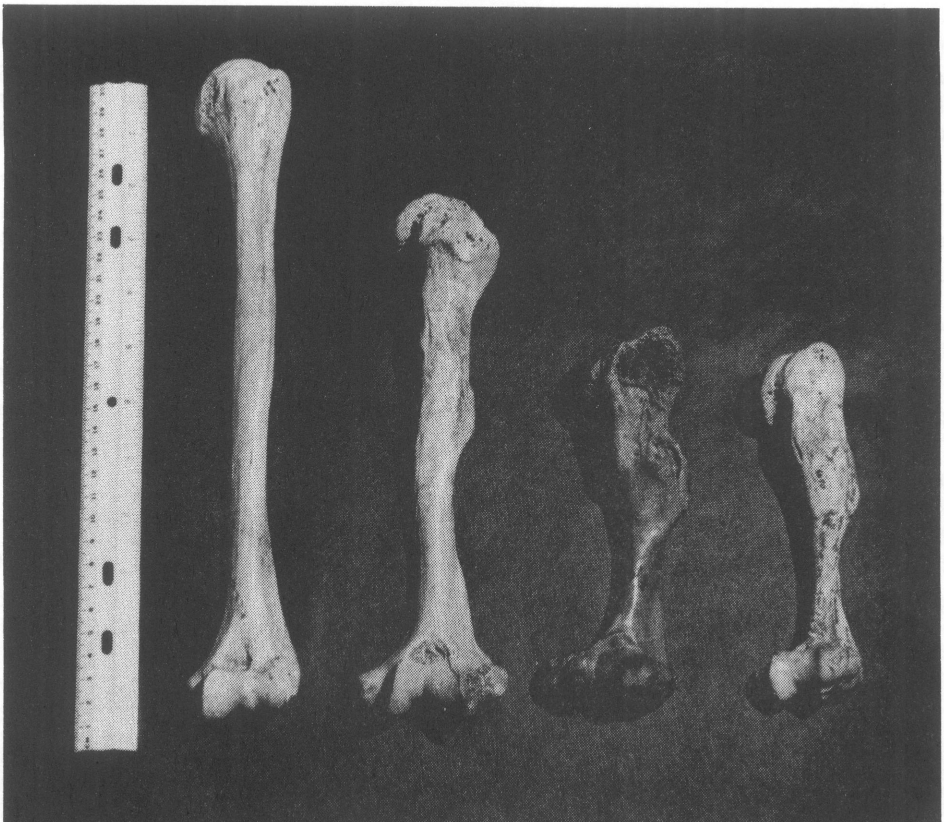
DISCUSSION

Dwarfism is broadly defined as the underdevelopment of the body. There are several types and classifications of dwarfism (Roberts 1988). Classifications are based on the parts of the body which are effected and the types of shortening which occur, such as proportional versus disproportional shortening. The most common form of dwarfism is achondroplasia (Roberts 1988). Achondroplasia is a congenital (autosomal dominant) hereditary disturbance in which cartilage growth rate slows while periostial activity remains normal (Rimoin *et al.* 1970). The result of the decreased growth rate is a quantitative shortening of the humerus (since, in the normal condition, the humerus grows at a greater rate than the distal limbs). The humerus, therefore, appears to be more effected than the distal limbs.

As mentioned earlier, a major distinguishing characteristic of achondroplasia is rhizomelic micromelia. Spc. 6670 reflects this morphology, especially when comparing the humeri with the ulnae. The humeroulnar indices range around 100, indicating that the limbs are of nearly equal length (Table 2). The right humerus exhibits extreme rhizomelic micromelia in that it is only 3 mm longer than the right ulna.

The humeri of achondroplasts present several distinct peculiarities in their morphology. The humeral heads are mushroomed shaped which reflects the developmental pattern of the cartilage growth plate. When the cartilage disk grows appositionally (at the periphery of the disk) instead of interstitially (at the center of the disk), the result is that the humeral head grows laterally (increasing in diameter) instead of superiorly (increasing in height), thereby widening the base of the humeral head (Rubin 1964). This causes the proximal portion of the humeral head

Figure 2. From left to right: normal human humerus; non-achondroplastic humerus (spc. 3854); achondroplastic humerus (spc. 6670); achondroplastic humerus (spc. 9199).



to grow beyond the width of the anatomical neck, producing a lip over the anatomical neck and giving it the mushroom appearance. The altered humeral head morphology does not disturb normal shoulder function (Bailey 1971).

Another peculiarity of achondroplastic morphology is that the humeri have often been described as possessing large rectangular shafts. However, the diaphyses remain normal in width and thickness (Rubin 1964). What gives the achondroplastic humerus its stocky, rectangular appearance is the large, rectangular deltoid tuberosity. When comparing the deltoid tuberosity of the achondroplastic dwarf to the non-achondroplastic dwarf and to normal humeri, it is apparent that, in general, the shorter the limb length the greater the size of the deltoid tuberosity (Figure 2). This is expressed by the width of the deltoid tuberosity (Table 1).[2] Since muscle development among achondroplasts is not effected, muscles grow to their normal size and attach to the smaller bones. The effects of muscles on bones varies proportionally with changing limb lengths. A tubercle usually develops or grows larger at points where there is a greater amount of localized strain. This increases surface area which, in turn, decreases the concentration of stress or strain within the localized area (Hildebrand 1988). Thus, the appearance of additional or larger tubercles on the achondroplastic humerus than on normal humeri may be due to an increase in localized strain, resulting from a larger muscle mass acting on a smaller area of bone.

Achondroplastic humeri are also distinct in that trabeculae are present throughout the shaft (Figure 3). Generally, trabeculae are located at the ends of long bones and are not present past the surgical neck in normal adult humeri; that is, trabeculae are not found within the humeral shaft. Their function is to increase the surface area for

Table 1. Measurements of the comparative sample and the dwarfed specimens.

	HUMERUS			RADIUS	ULNA
	Max. Length	Epicondylar Breadth	Deltoid Tubr. Width	Max. Length	Max. Length
COMPARATIVE SAMPLE					
Total Average	322	59	2	248	269
Male Average	320	63	2	249	270
Female Average	303	56	1	231	252
Total Standard Deviation	18.8	6.3	1.9	17.2	17.0
Male Standard Deviation	18.6	6.5	1.9	15.6	14.9
Female Standard Deviation	15.3	4.0	1.9	13.8	14.1
NON-ACHONDROPLASTIC					
SPECIMEN #3854					
Left	228	71	6	247	250
Right	212	63	2	247	248
ACHONDROPLASTIC INDIVIDUALS					
SPECIMEN #6670					
Left	154	50	12	126	147
Right	158	50	10	121	155
SPECIMEN #9199					
Left	160	59	10	-	-

The epicondylar breadth measurement is the maximum distance between the medial and lateral epicondyle of the humerus. The deltoid tuberosity width measurement is the width of the midshaft subtracted from the maximum medio-lateral measurement of the shaft with the deltoid tuberosity (from the lateral edge of the deltoid tuberosity to the medial edge of the shaft). The maximum lengths of the radii and ulnae were measured from head to styloid process.

contact with bone marrow and vascular tissues (Martin and Burr 1989). Trabeculae may also be indicative of the amount and the direction of stress placed on the skeleton (Hildebrand 1988). Construction of excess trabeculae allows the energy or strain to be maximally absorbed in order to avoid skeletal fatigue or breakage of the bone (Martin and Burr 1989). Thus, the appearance of trabeculae throughout the shaft probably reflects the greater amount of total strain placed on the achondroplastic humerus.

Achondroplastic humeri have often been described as possessing flared metaphyses. However, the epicondylar breadths fall within the normal range (Table 1). Due to the shortened maximum limb length, the metaphyses appear to be flared when in fact their maximum epicondylar breadths fall within the normal range. Furthermore, the medial supracondylar ridge is postero-inferiorly constricted which accentuates the flared appearance of the metaphyses. The downward constriction of the medial supracondylar ridge blocks the opening of the olecranon fossa. This prevents the olecranon process of the ulna from fully entering the olecranon fossa, thus causing the disruption of full elbow extension. In addition, a groove is present in the midline of the trochlea. The trochlear notch of the ulna is divided by a median wedge-like projection which articulates with and rides on the trochlear groove of the humerus.

Spc. 3854 is not classified as an achondroplastic dwarf for several reasons, the primary reason being its distal limb length. Spc. 3854's humerus is shorter than its distal limbs and appears to be the only foreshortened limb, while its ulna and radius fall within the average range of maximum distal limb length (Table 1) and are not shortened. In other words, spc. 3854 does not exhibit rhizomelic micromelia and this is one reason for not classifying it as achondroplastic.

Another reason for not classifying spc. 3854 as achondroplastic is that arthritis is present in most of its metaphyses. The left humeral head is eburnated and the right humeral head is nearly obliterated by arthritis, disrupting normal shoulder function. In contrast, shoulder function in achondroplastics, generally, is uneffected, although the chance of arthritic disruption increases with age (Hall 1986). The dysfunction of the shoulder in spc. 3854 led to atrophy of the shoulder (deltoid) muscle. This is reflected by the small deltoid tuberosity on the right humerus.

A final reason for classifying spc. 3854 as non-achondroplastic is that the superior shaft of the radii is gracile. As stated earlier, the width and cortical thickness of achondroplastic limbs are uneffected.

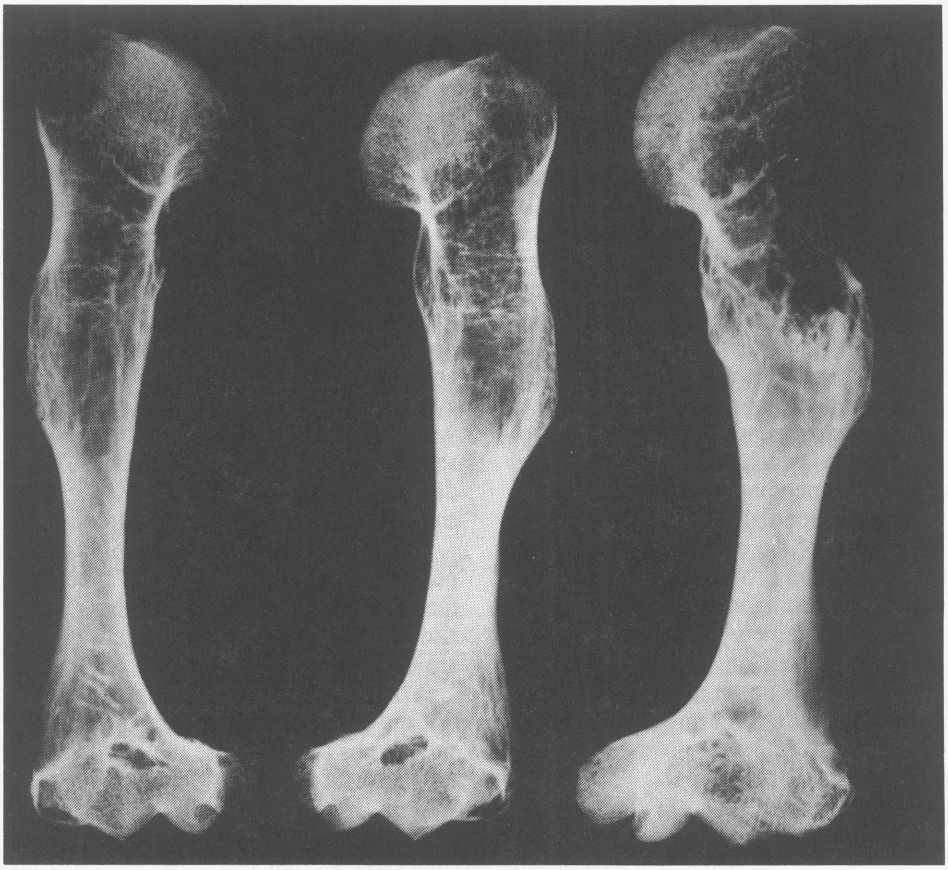
In sum, the overall morphology of spc. 3854 and the occurrence of arthritis in both the left and right limbs indicate that spc. 3854 is not achondroplastic and that its arthritis is not due to

Table 2. Humeroradio and humeroulnar indices.

	Humeroradio Index #	Humeroulnar Index #
SAMPLE POPULATION AVERAGE	77	84
SPECIMEN #3854		
Left	108	110
Right	103	109
SPECIMEN #6670		
Left	82	95
Right	78	101

The average of the sample population's humeroradio indices and humeroulnar indices is calculated. In addition, the individiual indices of spc. 3854 and spc. 6670 are listed. Humeroradio index is the product of radius maximum length times one hundred, divided by humerus maximum length. Humeroulnar index is the product of ulna maximum length times one hundred, divided by humerus maximum length. The humeroradio index reflects the proportional relationship between the maximum length of the radius and the maximum length of the humerus. The greater the number, the longer the distal limb in comparison to the humerus, where a score of one hundred means that the limbs are of equal length.

Figure 3. Radiographs of achondroplastic skeletal remains, from left to right: spc. 6670 right humerus; spc. 6670 left humerus; spc. 9199 left humerus.



trauma but to a congenital defect other than achondroplasia.

SUMMARY

Achondroplastic humeri possess distinct morphological features. The major classification of achondroplasia is rhizomelic micromelia in which all of the limbs are effected and shortened. The achondroplastic humerus possesses a mushroom shaped head, a large, rectangular deltoid tubercle, and a tubercle on the crest of the lesser tuberosity. Generally, there is an inverse relationship between limb length and the width of the deltoid tuberosity. Trabeculae are found throughout the shaft. The distal end is flared and a groove is present in the midline of the trochlea. Blockage of the olecranon fossa by the medial supracondylar ridge limits full elbow extension. These traits are generally found among achondroplastic individuals and are guidelines which can be used to diagnose achondroplasia in the archaeological record.

ACKNOWLEDGMENTS

We would like to give special thanks to Gary Richards and Susan Anton for their inspiration, guidance, patience and invaluable comments. Also to Dr. F. Clark Howell of the Anthropology Department, University of California at Berkeley for his faculty sponsorship, to the Lowie Museum for allowing the authors access to the skeletal material, to Jill Duke of Cowell Hospital for the radiographs and to Christian Lynch for taking and producing the photographs.

NOTES

[1] A case study of specimen number 3854 was done concurrently with this paper by Ross and

Malonzo.

2 It is important to note that the deltoid tuberosity width measurement is extremely variable. This is due to the positive correlation between the mid-shaft width and the deltoid tuberosity width: the wider the midshaft, the larger the deltoid tuberosity. Hence, the deltoid tuberosity width is correlated with two variables (midshaft width and maximum limb length), which results in its high variability.

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