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OSTEOLOGICAL REPRODUCTIONS

Human Female Achondroplasia Dwarf Skull BCD-279



Osteological Evaluation Report

Prepared by

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Human Female Dwarf Skull, Achondroplasia

<u>Product Number:</u>	BCD-279
<u>Specimen Evaluated:</u>	Original Specimen
<u>Skeletal Inventory:</u>	One intact cranium One intact mandible

General Osteological Observations:

Skull:

The neurocranium is large in relation to the face. The forehead bulges and double bossing is visible on the frontal bone (**See Figure 1**). The nasal bridge is depressed, the nasal spine is large and the nasal bones appear constricted, bending sharply and jutting outward (**See Figure 2**).



Figure 1: Frontal View of Cranium



Figure 2: Lateral View of Cranium



The foramen magnum is extremely small, a condition known as *foramen magnum stenosis* (See Figure 3). The condition carries a number of clinical consequences, including serious nerve compression.

Figure 3: Foramen Magnum Stenosis

Dentition:

The individual was completely edentulous at the time of death (See Figure 4). All dental alveoli are healed and the surrounding bone is greatly resorbed. On both the maxilla and mandible, the labial portion of the alveolar ridge is sharp, and the buccal portion is thick and rounded. The mandibular height is less than 1 centimeter, and the mandible is so reduced that the superior surface of the alveolar ridge encroaches upon the mental foramina.

Dental crowding is reported as a condition of achondroplasia. The complete loss of teeth in this individual may have been secondary to dental crowding and malalignment.

The shape of the mandibular condyles is modified. This is a typical result of total tooth loss and the subsequent realignment of the maxilla and mandible.



Figure 4: Edentulous Jaw:

The alveolar ridges are healed and severely reduced following long-term tooth loss. The mandibular height is less than 1 cm.

Features of Race:

It is not possible to scientifically analyze race in this specimen due to lack of known-race comparative material for dwarfs. (For general interest, it is noted here that achondroplasia is not race-specific. The condition is said to occur in 1 in 15,000 to 40,000 newborns, regardless of race.)

Features of Sex:

Methods for sex determination from the skull are based on populations of normal individuals of specific ancestry. Achondroplasia greatly affects the skull, so standard methods are not applicable. (However, see entire skeleton SCD-279-A. In spite of its small size, the pelvis demonstrates the classic characteristics of a female genotype. The pubis is elongated and the sub-pubic angle is wide. The ventral arc is obvious.)

Features of Age:

The age at death is known to be around 60, and the overall skeletal age is consistent with that of a person beyond middle age (based on analysis of postcranial skeleton in addition to the skull). The diseases of the elderly are all present: the synovial joints are arthritic (e.g., shoulders, elbows, knees); the cartilaginous joints have degenerated (e.g., pubic symphysis, auricular surface of the ilium, vertebral body surfaces); the costal cartilage has begun to ossify; the vertebral bodies are fringed with osteophytes and compression fractures are present; the maxilla and mandible are edentulous.

Evidence of Disease (Pathology):

The skeletal modifications of achondroplasia are all present as well as severe arthritis and other age-related diseases. Please see the report for SCD-279 for the modifications to the postcranial skeleton. The conditions affecting the skull are 1) foramen magnum stenosis (small opening for passage of spinal cord into skull), and 2) complete tooth loss (edentulous).

SUMMARY

(Note: Determinations are based upon examination of postcranial skeleton in addition to skull (see report for SCD-279) :

Race:	Undetermined, although the nasal form is consistent with European origin.
Sex:	Reported to be female. The pelvic form, particularly the pubis, supports that information.
Age:	Reported to be around 60. The skeletal evidence does not contradict that information. The remains appear to be middle aged or older, based on generalized degenerative changes in all joints.
Pathology:	Achondroplasia and associated skeletal modifications
Trauma:	No perimortem trauma is visible. Antemortem trauma can be attributed to aging and achondroplasia.

Educational Resource Suggestions:

1. The skull alone is diagnostic of achondroplasia. The forehead bulges and double bossing is visible on the frontal bone. The bridge of the nose is abnormally depressed, and the nasal bones jut outward at a more-than-normal angle. The foramen magnum is extremely small, a condition known as *foramen magnum stenosis*
2. The face is small in relation to the size of the brain case. This is typical of achondroplasia, but it is accentuated in this individual by the total loss of teeth during life. The alveolar ridges that would have held the tooth sockets resorbed following tooth extraction, thus further reducing the height of the face. Alveolar ridge absorption is a normal occurrence, not related to achondroplasia.
3. Educators may want to use the skeleton (SCD-279) as a launching point for the discussion of (1) endochondral bone formation, and the mechanism for calcification of cartilaginous models in long bone growth; (2) the plasticity of bone under stress. For example, note the effects of wheelchair use on the density and configuration of upper limbs in comparison to lower limbs.

References:

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Hecht, J.T., Horton, W.A., Reid, C.S., Pyeritz, R.E., Chakraborty, R., (1995). Growth of the foramen magnum in achondroplasia. *American Journal of Medical Genetics*, **32**(4):528 – 535

Nehme AM, Riseborough EJ, Tredwell SJ., (May 1976) Skeletal growth and development of the achondroplastic dwarf. *Clin Orthop Relat Res.*,116:8-23.

Richette, P, Bardin, T, Stheneur, C. (2008) Achondroplasia: from genotype to phenotype *Joint Bone Spine*, **75**(2): 125-30.

Disclaimers:

This report is meant only as a teaching tool for introductory level students of the anatomical, anthropology or forensic sciences who might be using this specimen to learn human and forensic osteology. My opinions are based solely upon the material presented to me. This is somewhat artificial as in real forensic investigations additional studies would be undertaken prior to the formulation of diagnoses and the production of a report. These studies might include plain film radiography, computed tomography (CT) studies, histological analysis, etc. My opinions regarding race and sex are based only upon non-metric analyses.

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Bone Clones Disclaimer on Ancestry Assessment

The assessment of ancestry from human skeletal remains, particularly the skull, is a component historically included in the creation of a biological profile for forensic purposes. This practice involves the analysis of morphoscopic traits and metric variables that may exhibit population-specific patterns of variation. However, it is important to recognize the significant scientific and ethical limitations of this practice.

Race is not a biologically valid concept. Contemporary biological anthropology holds that race is a social construct with no discrete biological basis. Human variation exists on a continuum, shaped by complex interactions between genetics, environment, and culture—not distinct “racial” categories. Therefore, the identification of “race” or “ancestry” based solely on skeletal features is scientifically problematic and cannot be performed with high accuracy or precision.

Although some morphological traits of the cranium may reflect broad population-level patterns due to shared evolutionary history, these traits do not map neatly onto socially defined racial categories. Furthermore, categories such as “Asian,” “European,” or “African” are socially constructed labels that do not fully capture genetic or phenotypic diversity, and they should not be interpreted as exact or absolute identifiers. As such, ancestry estimation based on skeletal features should not be interpreted as the identification of race, and results should be presented with appropriate caution and clear communication of limitations.

Historically, law enforcement agencies have requested ancestry estimations as part of forensic reports. However, many biological anthropologists today are increasingly hesitant to include this component, as doing so may inadvertently reinforce outdated and harmful typological thinking—the idea that humans can be classified into discrete biological “types” based on physical features. Such typologies have a long and problematic history and are not supported by modern science.

In cases where ancestry estimation is included, it is done with the understanding that it is a probabilistic assessment—not a definitive classification—and it must be contextualized within a broader ethical framework that prioritizes scientific integrity, individual dignity, and the avoidance of reinforcing racial stereotypes.