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A possible juvenile hypochondroplasia case from the mass grave of Lazzaretto Nuovo Island (Venice)

Matteo BORRINI, Laura RICCADONNA**, Camilla BORRINI****

*Liverpool John Moores University – School of Natural Science and Psychology
M.Borrini@ljmu.ac.uk
**Pontificia Università Teologica "San Bonaventura"
Master in "Antropologia Filosofica e Forense, Criminologia e Tecniche Investigative Avanzate"
***Università degli studi di Firenze, Dipartimento di Storia, Archeologia, Geografia, Arte e Spettacolo

Riassunto
Tra i resti degli individui inumati nel cimitero del Lazzaretto Nuovo (Venezia) durante l'epidemia di peste del 1576, uno scheletro appartenente ad un soggetto sub-adulto presenta una discrasia tra l'età biologica alla morte ricavabile dalla misura delle ossa lunghe dell'arto superiore. Poiché altri indicatori scheletrici concordano con quanto ricavabile dall'omero e dal cinto clavicolo-scapolare, il dato può essere interpretato come una riduzione in senso craniocaudale della lunghezza dei distretti anatomici. In associaizone ad altre modifiche morfologiche leggibili sul campione, al soggetto viene diagnosticata una possibile forma di nanismo ipocondroplasico.

Abstract
Among the remains of individuals buried in the cemetery of the New Lazaretto (Venice) during the plague epidemic of 1576, a juvenile skeleton with a discrepancy between the biological age at death obtained by the diaphyseal length was recovered. Other skeletal indicators from the humerus and the shoulder girdle show a craniocaudal reduction of bone length. Associated with other morphological changes and signs, the individual is diagnosed with hypochondroplasia, a specific form of dwarfism.

Parole chiave: nanismo, ipocondroplasia, bioarcheologia.

Key words: dwafirsm, hypochondroplasia, bioarchaeology.

Introduction

The authors present a preliminary study of the juvenile skeleton ID 24 from the 1576 mass grave of the Lazzaretto Nuovo that was recovered in 2007 during the excavations of Soprintendenza per I Beni Archeologici del Veneto – Nucleo NAUSICAA, Archeoclub d'Italia and Gruppo Archeologico Spezzino (Borrini, 2008). This 8 year-old individual has been selected due to a congenital pathological condition recorder in the skeletal remains.

Dwarfism

The significant reduction of the size of an individual in respect to the population's average is known as dwarfism. The main types are pituitary dwarfism, due to insufficient secretion of growth hormone (GH) or somatotropin (STH) during puberty, and achondroplasia related to decreased cartilage ossification.

In the first case, the lack of STH causes a severe impairment of growth, but the affected individual is well-proportioned; for this reason, in other types such as the Laron dwarfism, the pituitary form is described as harmonic (Berne and Levi, 2009). Achondroplasia, however, is a congenital osteochondrodysplasia that can affect any bone and cartilage and is the most common cause (1/25.000 live births) of disproportionate dwarfism (Marziotti *et al*., 2009). This class of dwarfism is phenotypically characterized by macrocephaly, frontal bossing, craniofacial disproportion, relatively large cranial vault, arched palate, rhizomelic shortness of limbs (which appear short and stubby) with typical disproportion between the epiphysis which is "mushroom-like" and the shaft which is shortened (especially in the lower limb),

brachydactyly, lumbar hyperlordosis, bilateral varus knee and flat foot (*ibidem*, 2009). Although the specific genetic alteration responsible for this pathology is still unknown, in most cases the dominant mutation of the gene located on the short arm of chromosome 4 that encodes the receptor of the type 3 fibroblast growth factor (FGFR3) has been noted. The same genetic alteration is also present in hypochondroplasia (HCH), another short-limbed condition classified in the heterogeneous group of skeletal dysplasias, where the subject has a phenotype comparable to minor stage achondroplasia, with milder marked characteristics (*ibidem*).

In addition, another form of osteodysplastic dwarfism, among the most common with microcephaly, is represented by Seckel's Syndrome also known as bird-headed dwarfism, so called for its typical phenotype: large eyes, beak-like nose, narrow face, receding mandible and dental anomalies (Sauk *et al.*, 1973).

History

Historical documents show how the dwarf has always aroused interest and curiosity as a "freak" or an element discordant with Creation's harmony due to his/her disharmonic aspect. Since the ancient Egyptian civilization a large number of representations depict individuals affected by dwarfism: it is illustrated in an Old Kingdom (2520 BC) sculpture, now in the Cairo Egyptian Museum, where the court official Seneb, an achondroplastic dwarf, is portrayed with his wife and two of their daughters. From the same cultural environment there is the presence of god Bes in the Egyptian pantheon, a minor family and home protector deity depicted as a dwarf with short and stocky limbs.

Deities characterized by the same condition are present also in Ancient Greek, where they are children protectors and dispense fertility as part of the Dionysian cult.

The switch between the divine to the profane symbol of the dwarf happens during the Roman royal and republican ages, when physical deformities and imperfections are not widely accepted, and it continues during the imperial age where dwarfs are used as a court jester or a servant to exhibit. Clue of the changed perception of this disease are the shows and the fights between dwarfs and other "lower categories" - such as wild beasts, women and subjects affected by deformities- that took place in the amphitheaters before the main, more respectable, gladiator fights. A similar treatment was observed in the "freak circus" of the 19th and 20th centuries.

A cultural acceptance of dwarfs will be introduced by the advent of Christianity that encourages a new kind of feeling in human relationships; unfortunately the Medieval age comes back to the concept that the ugliness and physical deformation are a sign of God's punishment, a deserved suffering that must be tolerated as atonement for the sins.

Finally during the Renaissance, trusting they vivacity and intelligence, dwarfs are often appointed as advisers or ladies in waiting as proved by the numerous artistic representations such as the Dwarf Morgante portrait by Bronzino preserved in the Uffizi Gallery in Florence. (Rabino, 1963; Cetorelli Schivo, 2003; 2004).

Analysis of skeleton ID 24

The subject is quite well preserved (Fig. 1) except for the lack of the lower limbs due to the interception of burial by a trench during the Austrian military occupation of the island.

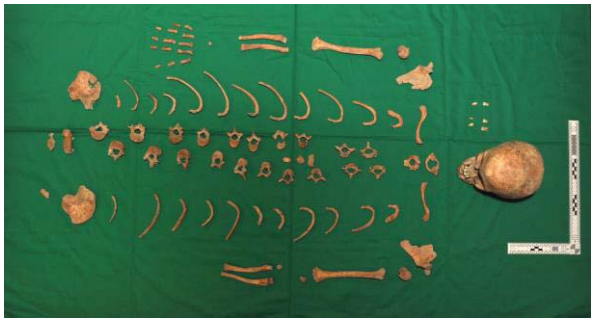

Fig. 1: the skeleton of ID 24 in anatomical position.

According to the juvenile age of the subject, a preliminary morphological sex determination has been conducted using the auricular surface elevation (Mittler and Sheridan, 1992), which allowed a tentative attribution to the female population; further, more reliable genetic analysis could confirm this finding.

The biological age at death estimation, performed by both skeletal and dental developmental methods, showed appreciable discrepancy in the age observed in the different anatomical regions (Tab. 1).

Anthropometric measurements on both forearms delineate a gracile physical structure, and consequently a younger age (5-6 years), compared to the values of the right and left

clavicle, which provided an age range of 8 years +/-9 months, compatible to the value obtained from the teeth (Ubelaker, 1978).

Confirmation of the obtained results comes from the general development of the skeleton, where the complete fusion between *pars basilaris* and *pars lateralis* (Fig. 3) and the lack of fusion of the proximal radial epiphysis, respectively suggest an age older than 5-7 year , but also less than 9 years (Schaefer *et al*. 2009).

In contrast with this estimated age are the lengths of the ulna and radius, which are indicative of an age of 3 years: a cranio-caudal bone length decrease suggests a possible pathological condition.

	Diaphyseal length (mm)	Age (yrs) (Shaefer et al., 2009)	Age (yrs) (Mallegni and Lippi, 2009)
CLAVICLE R	95	8-9	
CLAVICLE L	96	8-9	
SCAPULA R	88	5-6	
SCAPULA L	91	5-6	
HUMERUS R	177	5-6	7-8
HUMERUS L	176	5-6	7-8
RADIUS L	117	3,5-4	5-6
ULNA L	127	3-3,5	4-5

Tab. 1: diaphyseal length and estimated age according to different references.

Osteobiographic data have been used to better figure out the condition responsible for the inconsistency revealed; a moderate curvature of the shaft in both the ulna and radius (Fig. 4), as well as the skull profile characterized by frontal bossing (Fig. 2) and the appreciable rib deformation (Ortner, 2003) could suggest a diagnosis of rickets (Aufderheide and Rodriguez-Martin, 1998), however this is repudiated by the absence of the typical "rachitic rosary" ribs and by the growth delay. On the contrary, the cranio-caudal decrease of the shaft's length with a curvature of the forearm, the frontal bossing and a narrowing of the *foramen magnum* could be compatible with some form of dwarfism.

	Rickets	Achondroplasic dwarfism	Pituitary dwarfism
Shortened cranial base		X	
Deformity of ribs (pigeon breast)	X		
Frontal bossing	X		
Asymmetry of the vertebral bodies	X	X	
Marked narrowing of the vertebral bodies in the left side	X	X	
Cranio-caudal decreasing of age		X	
Normally proportioned body segments, normally shaped long bones			X

Tab. 2: osteobiographic traits as signs of possible pathological condition.

Assuming this last hypothesis as the most probable, the authors tried to collect pieces of evidence to better determine which type of syndrome could be responsible for the changes observed (Tab. 2). The individual can be affiliated with a condition where some anatomical structures are disproportionate, e.g. to a growth defect also called short-limbed dwarfism or disharmonic dwarfism. However it is necessary to reject the possibility of this case being a microcephalic primordial dwarfism (Seckel syndrome or bird-headed dwarfism) due to the absence of microcephaly as well as receding chin and forehead. Also Achondroplasia seems to be excluded by the maintenance of the proportion between epiphysis and diaphysis in long bones, without the classic mushroom shape extremities and thick shaft (*ibidem*).

The present case could not be simply identified as a form of dwarfism characterized by short stature with normally proportioned body segments (harmonic dwarfism). The form of familial low constitution dwarfism and the phenotype related to pituitary growth hormone deficiency, with relatively well-proportioned body (*ibidem*), have to be rejected due to the

internal disproportion in the lengths of the upper limb, despite the epiphysis/diaphysis retained proportion.

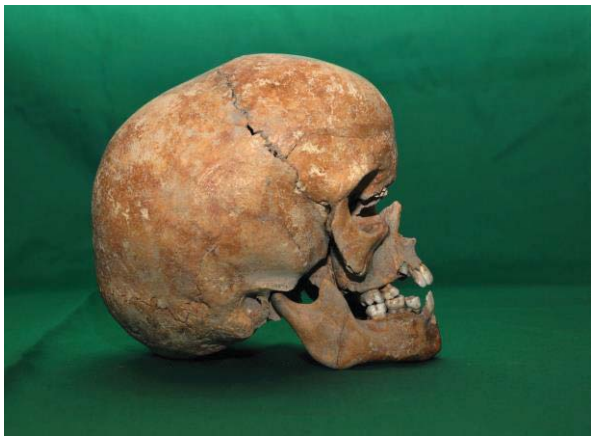

Fig. 2: lateral view of the skull characterized by frontal bossing.

Fig. 3: inferior view of the skull that shows the shape of *foramen magnum* and the complete fusion between *pars basilaris* and *pars lateralis.*

Fig. 4: upper limb with reduction of the forearm's length and curvature of ulna and radio.

Consequently, the hypothetical diagnosis that could fit with the analyzed remains is hypochondroplasia, a disharmonious form of dwarfism less frequent and known than achondroplasia (1 in 180,000/312,000 live births according to Jones, 2006). This form is characterized by a reduced growth which does not alter the morphology of individual bones and the relationships between epiphysis and diaphysis. Possibly due to the juvenile age of the individual, the lack of the lower part of the body and the milder pronunciation of the marks, no other typical signs of this syndrome have been detected, such as short hands and feet, reduced mobility of the elbows, lordosis and bowed legs.

Conclusions

The authors detected on the remains of the 8 year-old skeleton ID 24 a discrepancy in the biological age obtained by measuring the length of each preserved diaphysis, with a cranio-caudal reduction of the estimated age. Related to other morphological signs, such as frontal bossing and a moderate curvature of the forearm, this inconsistency could be realistically attributed to a form of hypochondroplasia. The general appearance of remains, the lack of other deformation in the epiphysis or in the cranium, as well as the lack of a rachitic rosary allow excluding other syndromes (pituitary dwarfism, achondroplasia) or pathological conditions (rickets).

Future analyses are planned to achieve a comprehensive diagnosis and a complete study of the remains from the archeological site of the Lazaretto Nuovo Island.

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