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Abnormalities of the first cervical vertebra in a Muslim community from Dobruja (Southeastern Romania): a case study

Andreea Toma¹, Gabriel Vasile^{2*}

Abstract. The individuals buried in the 18th-19th century Muslim cemetery from Constanța *Boreal* show, in addition to numerous dental and bone pathological changes, traumas, and anomalies, some congenital defects identified in two skeletons, after the examination of a third of the individuals. The lack of fusion identified in the middle of the anterior arch of the atlas vertebra is an unusual condition, especially since malformations of the first cervical vertebra are not commonly found in human skeletal material. The two anterior clefts presented in this study are the first anomalies of this type identified in ancient Romania's populations. Nevertheless, defects of the anterior arch of the atlas could represent asymptomatic incidental findings on routine radiographs. Knowledge of this abnormality and its causative factors will contribute to a better understanding of spinal variations and malformations.

Keywords: anterior cleft of the atlas, anomalies, bioarchaeology, Mediaeval period, Dobruja.

Anomalii ale primei vertebre cervicale la o comunitate musulmană din Dobrogea (sud-estul României): un studiu de caz. Indivizii înmormântați în cimitirul musulman din secolele XVIII-XIX de la Constanța *Boreal* prezintă, pe lângă numeroase modificări patologice dentare și osoase, câteva traumatisme și anomalii, dar și defecte congenitale, acestea fiind identificate la doi dintre indivizi în urma examinării a unei treimi din seria scheletică. Lipsa fuziunii din mijlocul arcului anterior al atlasului este o afecțiune neobișnuită, mai ales că astfel de malformații ale primei vertebre cervicale nu sunt frecvent întâlnite în materialele scheletice umane. Cele două fisuri anterioare prezentate în acest studiu sunt primele anomalii de acest tip identificate la populațiile vechi de pe teritoriul României. Cu toate acestea, astfel de defecte ale arcului anterior al atlasului pot fi considerate constatări accidentale asimptomatice la radiografiile de rutină. Cunoașterea acestei anomalii și a factorilor care o determină vor contribui la o mai bună înțelegere a variațiilor și malformațiilor coloanei vertebrale.

Cuvinte cheie: fisura anterioară a atlasului, anomalii, bioarheologie, perioadă medievală, Dobrogea.

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Introduction

In 2019-2020, a preventive archaeological research was organised in *The tumular and flat necropolis from Constanța – Boreal area* archaeological site, which led to the identification of 314 graves belonging to an ancient Muslim cemetery near the former village of Palazu Mare, today a district of Constanța (Dobruja, SE Romania), in addition to graves from the prehistoric and Roman periods. The Muslim cemetery, developed on a dominant hillock with attested ancient flattened mounds, has been provisionally estimated to be within the 18th-19th centuries, until radiocarbon dates become available. Regarding the burial ritual, all the graves (including the investigated burial complexes – M.159 and M.293, discovered at approximately -0.80 m below the current level, **Fig. 1**), are of the same type – rectangular or oval pit with a side niche dug to the south-east (Băjenaru *et alii* 2020; Băjenaru, Petcu-Levei, Lascu 2019a; Băjenaru, Petcu-Levei, Lascu 2019b).

Thus far, almost a third of the total number of individuals has been analysed. Preliminary results for 50 of them have already been published (Toma, Vasile 2022). In addition to several dental and bone pathological manifestations, traumas, and anomalies, two cases of congenital midline clefts of the anterior arch of the atlas caught our attention (the number of cases might change when the rest of the individuals will be analysed).

The atlas is the first cervical vertebra and its structure usually has three primary ossification centres, along with two half-neural and one anterior arch. The anterior arch is cartilaginous until one (Barnes 2012, p. 74) or two years of age, and can sometimes be confused in anthropological investigations with the body of the hyoid bone. The arch consists of a slightly convex anterior surface with a downward-projecting tubercle, and a concave posterior surface with an articular facet for the dens (Schaefer *et alii* 2009, p. 107).

The atlas is one of the main points of head movement (owing to the joint) and acts as an attachment for several muscles. It is not a typical vertebra due to its shape and lack of body and spinous process, but its function and variations in development are important in clinical settings. Most congenital anomalies of the first cervical vertebra are diagnosed incidentally during imaging studies (Van der Velde, Nolet, Cardin 1997, p. 10).

Congenital clefts and other atlas anomalies are rare and can sometimes be confused with neck injuries or trauma (Covantev *et alii* 2021). In contrast to a fracture or trauma, the anterior cleft of the atlas is characterised by smooth, corticated lucency in the midline of the arch, without being associated with widening of the lateral masses (Van der Velde, Nolet, Cardin 1997, p. 14).

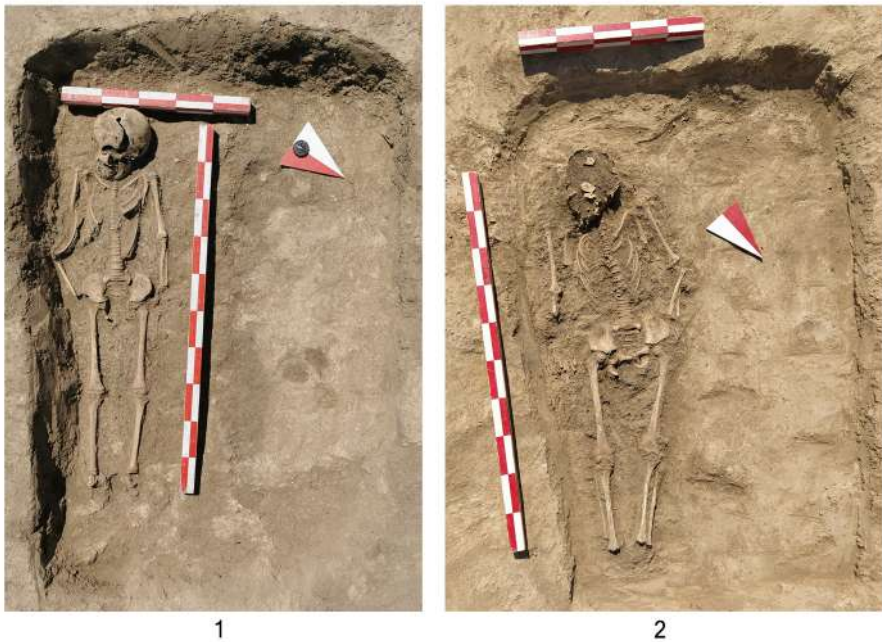


Fig. 1. *In situ* image of individuals M.159 (left) and M.293 (right).
 Fig. 1. Imagine *in situ* a indivizilor M.159 (stânga) și M.293 (dreapta).

A cleft in the anterior arch of the atlas is a rare and inconsequential congenital anomaly that usually remains undetected. Histological examination has shown that this defect, which ranges from 1-5 millimetres in width, is bridged by fibrocartilaginous tissue, resulting in what is generally considered to be a stable atlas. Radiographically, these congenital anterior clefts mimic many types of atlas fractures, including the Jefferson burst fractures and vertical fractures of the anterior arches. The different appearance of congenital clefts and acute atlas fractures helps to distinguish them. However, in most cases, anterior clefts of the atlas remain undetected and have no clinical consequences (Van der Velde, Nolet, Cardin 1997, p. 13).

Material and methods

In the first phase, anthropological analysis of the individuals from M.159 and M.293 followed the compilation of a skeletal inventory of bones and teeth (Nikita, Karligkioti 2019, p. 88-91), as well as the recording of certain postmortem bone alterations (Nikita, Karligkioti 2019, p. 70-73). The age at death of the two individuals was estimated based on the sequence of eruption and development

of the dentition (Ubelaker 1978, p. 47), the fusion of the primary and secondary ossification centres (Schaefer *et alii* 2009), and the maximum dimensions of the diaphyses of the long bones (Facchini, Veschi 2004, p. 93).

With regard to biometrics, a number of measurements were taken, and related indices were calculated both at the cranial (White, Black, Folkens 2012, p. 96-97; Buikstra, Ubelaker 1994, p. 71-78; Bräuer 1988, p. 160-192; Martin 1928, p. 579-678) and postcranial level (White, Black, Folkens 2012, p. 161-263; Martin 1928, p. 992-1068). We also calculated the skeletal stature of the individuals based on the maximum diaphyseal length of the tibia (Visser 1998, p. 415).

Morphoscopic features (Hefner 2009) were recorded together with cranial (Hauser, De Stefano 1989; Mann, Hunt, Lozanoff 2016), postcranial (Finnegan 1978), and dental non-metric characteristics (Scott *et alii* 2018). Finally, the presence of pathological changes in bones and teeth, together with skeletal anomalies (Mann, Hunt 2005; Waldron 2009), were recorded in the analysis.

Results

M.159

In terms of inventory, all components of the neurocranium are complete, with the exception of the sphenoid, which is represented by a few fragments, and the ethmoid, which is completely missing. The viscerocranium is represented by the following osteological components: the maxilla (with numerous destructions), the left palatine, both intact zygomatic bones, and the mandible. 33 teeth were preserved: 12 deciduous and 21 permanent. The postcranial skeleton consists of the sternum (without xiphoid process), ribs (both sides), vertebrae (seven cervical, 12 thoracic, five lumbar), clavicles (diaphyses), scapulae (right acromial process missing), humeri (diaphyses), left radius (diaphysis), ulnae (diaphyses), sacrum, coxae, femora, tibiae, fibulae (diaphysis) and several bones from the hands (one proximal phalanx) and feet (two left tarsals, two right tarsals, two left metatarsals, two right metatarsals and one proximal phalanx).

The skull is mesocranic, with a divergent, metriometopic, and chamemetopic forehead, curved parietals, and dolichostenomandibular mandible. The clavicles are gracile, humeri eurybrachic, and ulnae eurolenic. The left femur has a medium pilaster, is stenomeric, and the right femur has a null pilaster, and is eurimeric. Both tibiae are eurycnemic. The individual has an approximate skeletal stature of 1097.7 mm.

The age at death was estimated to be between 7.0-8.0 years (\pm 24 months) according to the dentition and between 6.2-6.8 years according to the long bone dimensions, placing the individual in the child age category (*infans I/infans II*).

For subadults, the main method used to estimate age at death was based on the sequence of tooth formation, eruption, and development, as the dentition of individuals between 0 and 15 years of age exhibits low sex dimorphism and reduced intra- and interpopulation variability.

At the cranial level, the following anatomical variants were identified: supraorbital groove (bilateral), zygomatico-facial foramen (bilateral), palatine foramen (left), and lambdoid ossicles (right). Allen's fossa (bilateral) and septal aperture (bilateral) were recorded postcranially. Non-metric dental characteristics included: shovel-shaped incisors (I^1 - I^2 bilateral, grade 5), Carabelli's trait (LM^1 , grade 1; RM^1 , grade 3), hypocone (M^1 bilateral, grade 4), and hypoconulid (M_1 bilateral, grade 3).

The following cranial features could be evaluated by morphoscopy: malar tubercle (0) and supranasal suture (0).

Pathologically, we noted the presence of *cribra orbitalia* due to active lesions on both orbital roofs, lingual caries (Lm_2), and linear enamel hypoplasia (I_1 bilateral). At the level of the atlas, the congenital anomaly, known as the midline cleft of the anterior arch, was identified (Fig. 2).

The skeleton shows variable brown and black staining on the skull and at the level of the postcranial bones.



Fig. 2. Anterior atlas arch cleft: first cervical vertebra (M.159) – superior view (left), inferior view (right).

Fig. 2. Defect al arcului anterior al atlasului: prima vertebră cervicală (M.159) – vedere din plan superior (stânga) și inferior (dreapta).

M.293

The skull consisted of small fragments of the parietal and temporal squamae, petrous pyramids, occipital (complete), both maxillae and zygomatic bones, some fragments of the sphenoid bones, and the entire mandible. 31 teeth were preserved: six deciduous and 25 permanent; one tooth is missing (congenital absence). From the postcranial skeleton we recovered ribs (complete), clavicles (diaphyses), scapulae (almost complete, fragmentary), vertebrae (seven cervical, 12 thoracic, five

lumbar), humeri (left proximal epiphysis and diaphyses), right radius (without distal epiphysis), ulnae (diaphyses, better represented on the right), sacrum, coxae, femora, tibiae, fibulae (diaphysis) and several bones of the hands (one left metacarpal, one right metacarpal, and one proximal phalanx) and legs (one left tarsal, one right tarsal, one left metatarsal, one right metatarsal, and one proximal phalanx).

Cranial measurements show a large occipital bone and dolichostenomandibular mandible. Postcranial biometry reveals gracile clavicles, eurybrachic humeri, and platolenic ulnae. The femora have a null pilaster, are platymeric, and both tibiae are mesocnemic. The individual has an approximate skeletal stature of 1198.9 mm.

The skeletal age of the individual was estimated to be between 9.0-10.0 years (\pm 24-30 months) based on dentition and between 7.9-9.0 years based on long bone dimensions, placing the individual in the child age category (*infans* II).

At the cranial level, the following anatomical variants were identified: zygomatico-facial foramen (left) and mastoid foramen (bilateral). Non-metric infracranial features were not observed. Non-metric dental characteristics included: shovel-shaped incisors (I^2 bilateral, grade 6), Carabelli's trait (LM^1 , grade 5; RM^1 , grade 4), hypocone (M^1 bilateral, grade 5), and hypoconulid (M_1 bilateral, grade 3).

The following cranial features could be evaluated by morphoscopy: inferior nasal aperture (3), anterior nasal spine (2), malar tubercle (0), and zygomatic-maxillary suture (0).

Pathological manifestations included congenital absences (LI^1), dental caries (Lm_2 – mesial), dental calculus (LI_1 - LI_2 – lingual; RI_1 – lingual; RI_1 - RI_2 – labial and lingual), linear enamel hypoplasia (LI^2 , RI^1 - RI^2). A midline cleft of the anterior arch was also identified in the anthropological analysis of this individual (**Fig. 3**).

The skeleton shows discolouration of the skull and variable degree of exfoliation of the long bones.



Fig. 3. Anterior atlas arch cleft: first cervical vertebra (M.293) – superior view (left), inferior view (right).

Fig. 3. Defect al arcului anterior al atlasului: prima vertebră cervicală (M.293) – vedere din plan superior (stânga) și inferior (dreapta).

Discussions and conclusions

In this study, we chose to provide a brief description of the pathological conditions found in two children (M.159 and M.293) from the Muslim cemetery of Constanța *Boreal*, and to conclude with an explanation of a rare bone malformation, the anterior cleft of the atlas. The two children are similar in age: 7.0-8.0 years and 9.0-10.0 years, according to the degree of dental development. The maximum lengths of the long bones diaphyses show slightly lower ages at death: 6.2-6.8 years and 7.9-9.0 years respectively.

Pathologies have always been of great interest to scientists; however, more recently, there has been an emphasis on subtle osteological changes and anomalies that reflect the influence of genetic factors, physical stress, and daily activities. By testing hypotheses and incorporating studies of pathology in bioarchaeology, we are able to provide a better perspective and draw valid conclusions regarding the health status of a historical skeletal population (Mann, Hunt 2005).

Within the fields of osteoarchaeology and anthropology, human teeth have been studied to understand the diet, habits, and diseases of past civilisations they may have encountered. Dental caries, dental calculus, and other dental diseases have affected the past populations as long as they existed. Dental caries involve pathological processes in the dental biofilm, which are the result of complex interactions between dietary, genetic, and physiological factors (Bertilsson *et alii* 2022, p. 18-19). Even less is known about the causes that lead to the formation of certain teeth. Heredity plays a role in most cases of congenital absence, but it is not the only causal factor; this process is influenced by trauma, radiation overdose, glandular dysfunction, infection of the developing tooth bud, and systemic conditions (Graber 1978, p. 266-267).

Dental pathologies in adults are related to genetic background and nutritional and hygienic habits, such as dental calculus (microorganisms embedded in the matrix); however, metabolic stress in childhood can be an important factor in the occurrence of linear enamel hypoplasia. In the case of the individuals studied, the horizontal lines that appear on the outer surface of the tooth crown are the result of disturbances in enamel formation during childhood. After these periods of stress, normal enamel formation resumes, leaving thin indentations in the tooth enamel (Limbo 2009, p. 236).

In addition to dental pathology, active porous lesions on the orbital roof have also been observed in individual M.159. The interpretation of these hypertrophic lesions has led to conflicting opinions, as well as various conditions that can produce this pathology in skeletal remains. There is evidence that cancers, infections, and metabolic diseases can produce porous lesions, but this phenomenon is often associated with various types of anaemia (which may be genetic or acquired

in response to various variables, such as diet) (Waldron 2009, p. 136). Genetic anaemias (such as thalassaemia and sickle cell anaemia) are much rarer than acquired anaemias, which are caused by blood loss and nutrient deficiencies. Nutrients needed to maintain red blood cell homeostasis include iron, essential amino acids, and vitamins, such as B12, B6, and folic acid (Walker *et alii* 2009, p. 111).

Genetic information not only influences the incidence of pathologies in populations but can also control the expression of various skeletal anomalies within specific developmental domains in the embryo, as in the case of midline clefts of the anterior arch of the atlas. Malformations of the atlas are rare and include hypoplasia, aplasia, and various clefts (Bonnevillie *et alii* 2004, p. 450-451). The anterior arch of the atlas is usually cartilaginous at birth, and approximately 20% of newborns begin the process of ossification between 6 and 24 months of age (Junewick *et alii* 2011, p. 1231).

This study presents two rare cases of congenital midline clefts of the anterior arch of the atlas, also known as anterior arch rachischisis or anterior spondylschisis of the atlas, which are uncommon developmental anomalies, with only a few cases reported in the literature. What is even more interesting is the fact that this type of anomaly has not yet been detected in anthropological studies conducted in Romania. In a series of cadaver dissections, approximately 2,749 atlases have been examined and it was found that the occurrence of an isolated cleft at the level of the anterior arch was observed in only 0.1% of cadavers (Van der Velde, Nolet, Cardin 1997, p. 12).

Similar percentages of anterior atlas clefts have also been described by Rios *et alii* (2019, p. 3), who state that the frequency of this rare anomaly in modern humans ranges from 0.087% to 0.1%. This is also the case in the research of Kwon *et alii* (2009, p. 525), who obtained similar results, observing the presence of an anterior atlas cleft in approximately 0.09-0.1% of 1,153 patients.

Most studies on congenital anomalies have been conducted using *Homo sapiens* skeletal material. However, according to recent research, among which we mention those carried out by Rios *et alii* (2019) at the site of El Sidrón (Spain) and Palancar *et alii* (2020) at Krapina (Croatia), such anomalies have also been identified in skeletal material belonging to *Homo neanderthalensis*. Rios *et alii*, found this anomaly in two out of 13 individuals (i.e. 15.38%). Palancar *et alii*'s (2020), in their study of three Neanderthal skeletal remains, found that one individual (marked Kr. 99) had a cleft in the anterior arch of the atlas.

In line with the previously described results in both modern humans and Neanderthals, these findings could suggest that the high level of anatomical variation (including the anterior atlas cleft) in Neanderthal atlases may be related

to inbreeding. Over the last few decades, scientists have been studying the genetic diversity that characterises the Neanderthal lineage. Based on genetic evidence, it has been shown that younger Neanderthal sequences show more similarity to modern human DNA than older Neanderthal sequences, suggesting possible inbreeding between Neanderthals and modern humans without ruling out low Neanderthal genetic diversity (Excoffier 2006).

This anomaly, non-fusion of the anterior arch or anterior atlas cleft, can occur in two different ways: absence of the anterior ossification centre, resulting in lateral masses not fusing anteriorly, or non-fusion between the anterior arch ossification centre and the anterior tubercle (Kwon *et alii* 2009).

It is important to distinguish developmental anomalies, such as clefts, from the more common Jefferson burst fractures. In the case of a midline cleft of the atlas, the lateral displacement of the lateral masses is usually 1 to 2 mm, whereas in a Jefferson fracture, it is more than 3 mm. However, anterior midline clefts are much rarer and may be associated with posterior midline clefts, resulting in a bipartite atlas, known as “split atlas” or *atlas bipartite* (Prempeh, Gibson, Bhattacharya, 2002).

Congenital variations in the anatomy of the cervical spine generally have no clinical implications except in the setting of trauma or conditions leading to cervical spine instability. However, congenital cleft of the anterior arch usually results in a weaker atlas (Van der Velde, Nolet, Cardin 1997, p. 14). In most cases, these anomalies are asymptomatic; however, as a consequence of structural instability, individuals may present dizziness, neck pain, headache, and neurological complaints (pins and needles in the arms or loss of strength). The same manifestations may occur in patients with a Jefferson fracture following trauma to the neck or head (De Zoete, Langeveld 2007, p. 62).

Most congenital anomalies of the atlas arch are discovered incidentally during investigations for neck pain or after trauma, and these cases of anterior spondyloschisis of the atlas still require further research, as the causes are not fully understood. In this study, we have chosen to provide a detailed description of the other pathologies present, as well as the parameters recorded in the anthropological study, to have an overall picture of the pathologies discovered and to provide a basis for future studies of the anomaly described.

In conclusion, the reported cases of congenital malformations of the atlas seem to confirm a higher prevalence of variations in the first cervical vertebra in Neanderthals. Finally, studies lead us to hypothesise that these anatomical variations may not be exceptional findings, but rather a consequence of a possible higher prevalence in extinct hominins than in *Homo sapiens* due to population inbreeding. Anthropological analyses will continue on all 314 individuals from

Constanța *Boreal*, including information on diet ($\delta^{13}\text{C}$, $\delta^{15}\text{N}$) and mobility ($\delta^{87}\text{Sr}/\delta^{88}\text{Sr}$), to assess the characteristics and evolution of these communities over time.

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Supplementary information

As part of the anthropological study of the individuals, metric analyses of bones were also recorded. These results are described in Tables 1 and 2, respectively.

Dimensions. Indices	M.159	M.293
1. Maximum skull length (<i>g-op</i>)	172.4	–
7. Length of foramen magnum (<i>ba-o</i>)	–	35.6
11. Horizontal cranial index (8:1) \times 100	78.7	–
8. Maximum skull width (<i>eu-eu</i>)	135.6	–
9. Minimum frontal width (<i>ft-ft</i>)	92.4	–
10. Maximum frontal width (<i>co-co</i>)	117.0	–
112. Transverse frontal index (9:10) \times 100	79.0	–
113. Transverse fronto-parietal index (9:8) \times 100	68.2	–
12. Occipital width (<i>ast-ast</i>)	106.2	105.7
16. Foramen magnum width	26.2	31.1
133. Foramen magnum index (16:7) \times 100	–	87.6
26. Front arch (<i>b-w</i>)	111.0	–
27. Parietal arch (<i>b-l</i>)	118.0	–
29. Front chord (<i>b-b</i>)	100.8	–
30. Parietal cord (<i>b-l</i>)	100.6	–
114. Transverse parieto-occipital index (12:8) \times 100	78.3	–
122. Sagittal frontal index (29:26) \times 100	90.8	–
65. Mandibular bicondylar width (<i>cdl-cdl</i>)	101.7	106.6
66. Mandibular bigonial width (<i>go-go</i>)	81.2	91.6
68. Mandible length (<i>pg-go</i> projection)	81.4	89.1
162. Mandibular index (68:65) \times 100	80.0	83.6

Table 1. The values of the main dimensions (mm), the values of the main cranial indices and the corresponding categories.

Tab. 1. Valorile principalelor dimensiuni (mm), valorile principalilor indici cranieni și categoriile corespunzătoare.

Dimensions. Indices	M.159	M.293
C1	90.6/89.6	102.4/100.5
C6	20.0/20.0	23.0/23.0
(C6:C1) × 100	22.1/22.3	22.5/22.9
H1	176.6/175.9	194.7/197.7
H5	13.0/12.8	14.2/14.4
H6	11.3/12.0	12.9/12.8
(H6:H5) × 100	86.8/93.9	90.6/88.4
R1	127.8/-	-/-
U1	140.5/-	-/156.2
U13	12.5/13.5	13.9/13.9
U14	14.2/14.4	18.2/18.0
(U13:U14) × 100	88.4/94.1	76.7/77.1
F1	243.7/246.3	279.7/282.9
F6	17.0/15.5	18.6/17.7
F7	15.4/16.4	19.5/19.6
F9	18.6/18.8	24.9/25.9
F10	19.0/18.2	19.7/20.3
F21	49.4/48.5	-/-
(F6:F7) × 100	110.4/94.6	95.1/90.3
(F10:F9) × 100	102.6/96.8	79.3/78.5
T1a	199.0/202.0	228.1/230.6
T8a	20.3/20.8	22.5/23.9
T9a	17.7/17.5	15.4/16.1
(T9a:T8a) × 100	87.3/84.3	68.6/67.3
P1	194.7/196.9	224.1/226.6

Table 2. The values of the main postcranial dimensions (left/right, mm), the values of the main postcranial indices (bilateral) and the corresponding categories.

Tab. 2. Valorile principalelor dimensiuni postcraniene (stânga/dreapta, mm), valorile principalelor indici postcranieni (bilateral) și categoriile corespunzătoare.

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