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A Case of Skeletal Dysplasia in Bone Remains from a Contemporary Male Individual

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The skeletal dysplasias are a large heterogeneous group of disorders characterized by generalized abnormalities of the skeleton. The severity of skeletal dysplasias ranged from individuals with normal stature and survival but early-onset osteoarthrosis to perinatal lethality. This study aimed to investigate bone remains with obvious skeletal abnormalities and to make a differential diagnosis. The remains belonged to a young male individual preserved in the Military Mausoleum with Ossuary, National Museum of Military History (Sofia, Bulgaria). It was established that the abnormalities were due to skeletal dysplasia with increased bone density and *craniometadiaphyseal dysplasia, wormian bone type* was suspected as a probable diagnosis. The individual was fit for service, which suggests a normal stature and mental development without severe malformations.

Key words: skeletal dysplasia, increased bone density, craniometadiaphyseal dysplasia, wormian bone type, Brodie's subacute osteomyelitis.

Introduction

The skeletal dyspasias, or *osteochondrodysplasias*, are a large heterogeneous group of disorders characterized by generalized abnormalities of the skeleton. A chromosomal classification of the skeletal dysplasias is in elaboration, but traditionally they are classified by parts of the skeleton which are involved [15]. Each dysplasia is rare, but the overall birth incidence is estimated to be 1/5000 live births [1]. The most frequent clinical implication is a disproportionate short stature, but it is not uniformly present. The severity of skeletal dysplasias ranged from individuals with normal stature and survival but early-onset osteoarthrosis to perinatal lethality. These disorders can be associated with a variety of orthopedic, neurologic, auditory, visual, pulmonary, cardiac, renal, and psychological complications [8]. Most skeletal dysplasias are associated with normal intellectual development, but there are exceptions to this rule [15].

Differential diagnosis of the skeletal dysplasias required radiographs and complete skeletal survey as well as assessment of the size, structure, and shape of the individual

bones. The region of the skeleton that is affected along with the pattern of skeletal abnormalities and bone density are crucial for a differential diagnosis [15]. Thus, the aim of the present study was to investigate in detail bone remains with obvious skeletal abnormalities and to make a differential diagnosis.

Materials and Methods

The objects of the present study were bone remains of a soldier who served and died in the wars from the beginning of the 20th century. The remains were preserved in the Military Mausoleum with Ossuary, National Museum of Military History (Bulgaria). The remains belonged to a young male individual and consisted of a skull with previously cut out calvaria, right and left humeri, left ulna, right and left tibiae. The mandible and the other postcranial bone were not available.

The physiological age of the individual was determined according to the cranial and postcranial bone development, observation of the cranial sutures and dentition [2, 7, 13, 17].

The skull and the available postcranial bones were macroscopically observed and measured. The characteristics of the angels and indices were given after Martin and Saller scales [9]. The categories "very small", "small", "middle", "large" and "very large" were used after Alekseev and Debets [16]. The stature was calculated based on the lengths of the limb long bones according to the method of Pearson and Lee [11]. A digital radiography was performed on a Nikon XT H 225 system.

Description and discussion of the case

Estimation of the physiological age

The remains belonged to a juvenile individual (16-18 years). It was inferred taking into account that all cranial sutures were open without traces of obliteration and the sphenooccipital synchondrosis was discernible. Furthermore, the third molars were not erupted as well as the second left molar. The radiographs showed an agenesis of these teeth, i.e. missing tooth germs in the maxilla, resulting in hypodontia. The available teeth showed minimal degree of dental attrition without abnormalities except for caries on the mesial surface of the second right molar.

Dental formula:

Right	Left
6 5 X X X X	X X X X 5 6 7

*X – postmortem loss of the tooth

The epiphyses of the long bones had the appearance of separated segments, even though they were fused with the diaphyses except for the distal epiphysis of the ulna, which was detached and missing (**Fig. 1**).

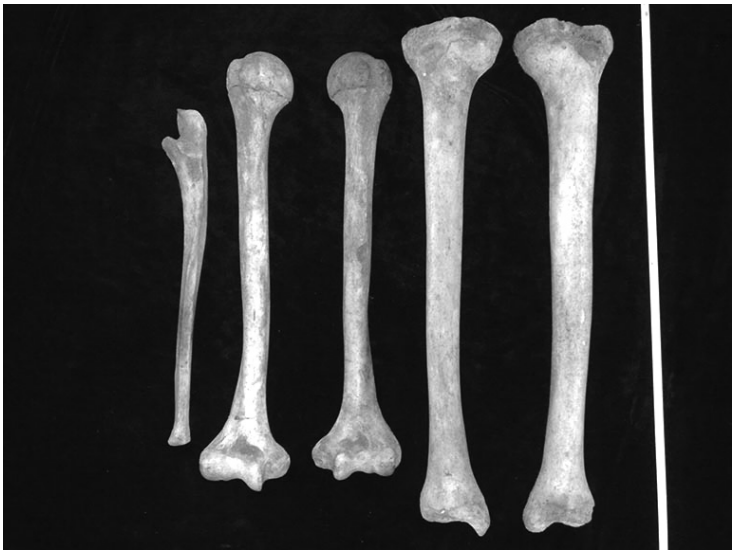

Fig. 1. The available long bones of the skeleton

The skull

The metric, angular and index characterizations of the skull were presented in **Tables 1, 2, 3**. The viscerocranium was broad but short and feebly profiled, i.e. it did not project significantly when the cranium was oriented in the Frankfurt plane (**Fig. 2**). The facial bones were small and underdeveloped. The nasal bones were noticeably wide and depressed, which was precondition for a broad and flat nasal bridge (**Fig. 2a**). The maxillary arch and hard palate were unusually broad and short, which probably was related to the observed hypodontia (**Fig 2c**). In addition, the canine fossae were very deep (**Fig. 2a**).

Table 1. Measurements of the skull after Martin and Saller [9]. The categories were given after Alekseev and Debets [16]

No	Measurements	mm	Category
1	Maximum cranial length	183	Medium
5	Cranial base length	93	Very small
7	Foramen magnum length	47	Very large*
8	Maximum cranial breadth	144	Medium
9	Minimum frontal breadth	108	Very large
10	Maximum frontal breadth	134	Very large
12	Biasterionic breadth	116	Large
16	Foramen magnum breadth	33	Very large
17	Basion-bregma height	132	Medium
20	Porion-bregma height	112	Small
23	Cranial circumference	530	Large

26	Frontal arc	127	Medium
27	Parietal arc	129	Medium
28	Occipital arc	112	Medium
29	Frontal chord	106	Small
30	Parietal chord	114	Medium
31	Occipital chord	92	Small
38	Cranial capacity /by Pearson for male skull/	1449.87 cc	Medium
40	Basion-prosthion length	85	Very small
43	Upper facial breadth	109	Large
45	Bizygomatic breadth	133	Medium
46	Middle facial breadth	101	Large
48	Upper facial height	67	Small
51	Orbital breadth	39	Very small
52	Orbital height	34	Medium
54	Nasal breadth	26	Medium
55	Nasal height	50	Small
56	Nasal bones length	18	–
49a (DC)	Dacrial chord	27	Very large
57 (SC)	Simotic chord	11	Large
DS	Dacrial subtense	7.5	Very small
SS	Simotic subtense	2.5	Small
60	Maxilloalveolar length	45	Very small
61	Maxilloalveolar breadth	67	Large
62	Palatal length	31	Very small*
63	Palatal width	43	Large
64	Palatal height	12	–
FC	Canine fossa depth	10	Very large

* The value of the measuremant is out of the borderlines of the category

Table 2. Angular characterization of the skull after Martin and Saller [9]. The categories were given after Alekseev and Debets [16]

No	Measurements	Degree	Category	Rubrication
72	Total facial angle	85°	Large	Orthognathous
73	Middle facial angle	87°	Large	Orthognathous
74	Alveolar angle	84°	Very large	Mesognathous
75	Nasal bones angle	66°	–	–
75(1)	Angle of nasal projection	21°	Small	–
77	Naso-malar angle	153°	Very large	
< zm	Zygomaxillar angle	148°	Very large	–

Table 3. Index characterization of the skull after Martin and Saller [9]. The categories were given after Alekseev and Debets [16]

Indices		%	Characterisctics	Category
8:1	Cranial index	78.69	Mesocranic	Medium
17:1	Height-length index	72.13	Orthocranic	Small
17:8	Height-breadth index	91.67	Tapeinocranic	Small
9:8	Frontoparietal index	75.00	Eurymetopic	Very large
48:45	Upper facial index	50.38	Mesenic	Small
52:51	Orbital index	87.18	Hypsiconchic	Large
54:55	Nasal index	52.00	Chamaerhinic	Large
61:60	Maxilloalveolar index	148.89	Brachyuranic	Very large*
63:62	Palatal index	138.71	Brachystaphylinic	Very large*
40:5	Alveolar/Gnathic index	91.40	Orthognathic	Small
DS:DC	Dacrial index	40.74	–	Small
SS:SC	Simotic index	33.33	–	Small
16:7	Index of foramen magnum	70.21	–	Very small

* The value of theindex is out of the borderlines of the category

The neurocranium was round in shape (**Fig. 2b**) with medium size and relatively low. The frontal bone was broad and bulging with entirely preserved metopic sutures (**Figs. 2a, b**). The occipital bone was broad and slightly bulging as well. At the posterior part of the sagittal suture a depression was observed (**Fig. 2b**). Foramen magnum was extremely large with notch in its posterior margin (**Fig. 2c**). The basal angle lies at the midpoint between normal and basilar kyphosis, i.e. extensive flection (**Fig. 3**). There was also uncompleted pterygoalar foramen on the left side, formed by the partial ossification of the pterygoalar ligament. The paranasal sinuses were underdeveloped as the frontal sinus was missing. The mastoid air cells were underdeveloped as well. A mild osteosclerosis of the skull was observed.

Multiple Wormian bones (WBs) with mosaic pattern occupied an oval-shaped territory at lambda (**Fig. 4**). They were relatively large in size (**Table 4**). Separate WBs were also placed along the lambdoid suture (**Table 5**). In addition, epipteric bones, postsquamosal bones and WBs in the occipitomastoidal suture were observed bilaterally (**Table 6**). According to Cremin et al. [4], WBs are of significance and could be accepted as a possible indicator of abnormal development when they are 10 or more in number with a diameter exceeding 6 × 4 mm, and arranged in a general mosaic pattern. The association of WBs with definite pathological conditions was widely discussed in our previous work [10].

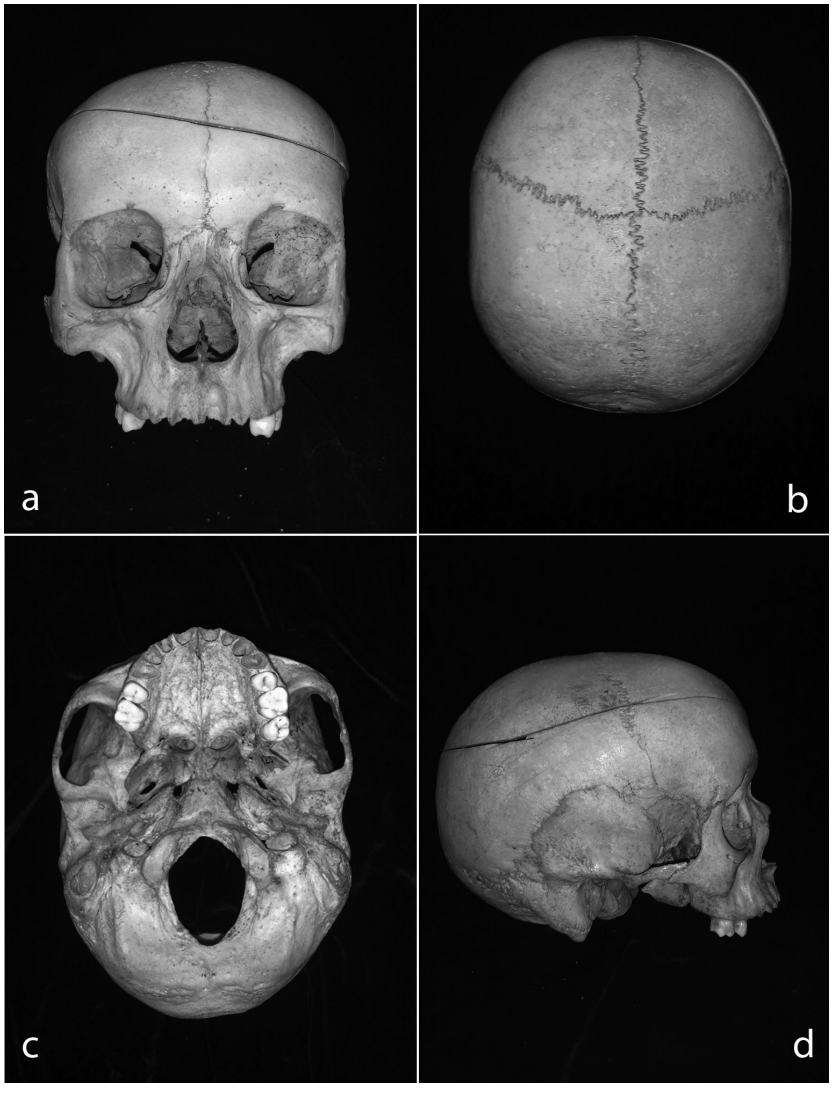

Fig. 2. Views of the skull: a) frontal; b) parietal; c) basilar; d) right lateral

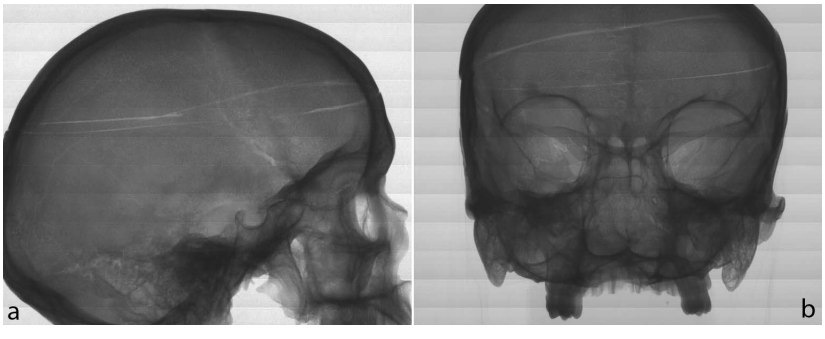

Fig. 3. Digital radiographs of the skull: a) lateral view; b) frontal view

Table. 4. Measurements of the Wormian bones in the region of lambda in mm

No	Wormian bones in lambda	
	length	width
1	26	11
2	24	9
3	15	6
4	21	11
5	25	12
6	17	8
7	16	4
8	15	4
9	19	7
10	15	5
11	18	15
12	24	15
Total	41	47

Table 5. Measurements of the Wormian bones in the lambdoid suture in mm

Wormian bones in lambdoid suture					
Right			Left		
No	length	width	No	length	width
1	14	5	1	19	3
2	18	5	2	16	5
3	21	6	3	17	5
4	16	4	4	9	2
5	22	6	5	13	4
6	–	–	6	17	6
7	–	–	7	16	4
8	–	–	8	8	3
9	–	–	9	12	5

Table 6. Measurements of the other Wormian bones in mm

Bones	Right		Left	
	length	width	length	width
Os epiptericum	22	10	14	4
	–	–	10	10
Os postsquamosum	16	14	14	9
Os Wormi suturae occipitomastoidea	5	6	20	12

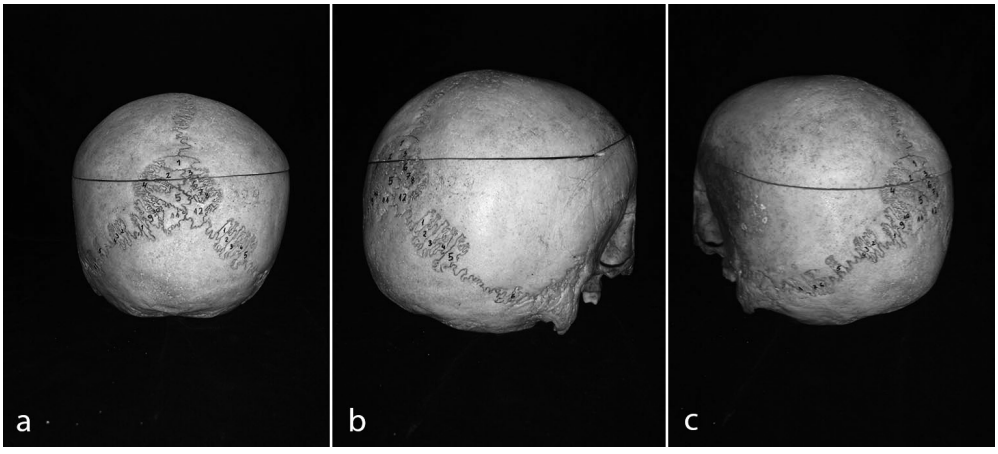

Fig. 4. Wormian bones: a) in lambda region; b) in lambdoid suture on the right side; c) in lambdoid suture on the left side

Postcranial skeleton

The available long bones did not show traces from healed fractures or perimortem inflicts, but the lack of other postcranial bones was an obstacle for complete assessment. The bones displayed thinning of the cortex and expansion of the medullary cavity. Their shape and size were normal except for both humeri whose distal ends were bowed forward (**Fig. 5**).

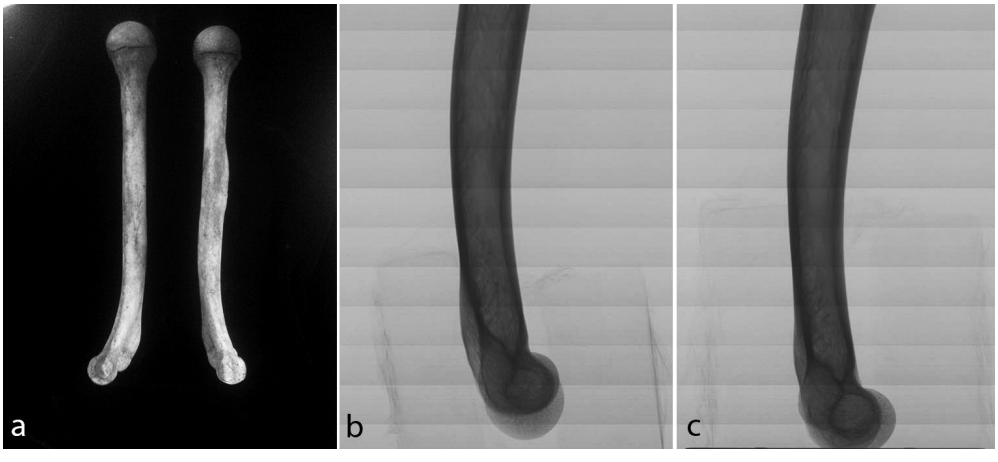

Fig. 5. Bowing of the distal ends of both humeri: a) right and left humeri; b) digital radiograph of the right humerus; c) digital radiograph of the left humerus

The deltoid tuberosity region of the right humerus was swollen and displayed a prolonged lesion – 22 mm long and 9 mm wide (**Fig. 6**). From the proximal end of the lesion started a 12 mm long canal that runs parallel to the bone axis. The aperture of the canal was round (6 × 6 mm). The canal did not communicate directly with the medullary cavity of the bone (**Fig. 6c**). This lesion probably is a consequence of a Brodie's abscess (subacute osteomyelitis) located on the diaphyseal cortex. Brodie's abscesses

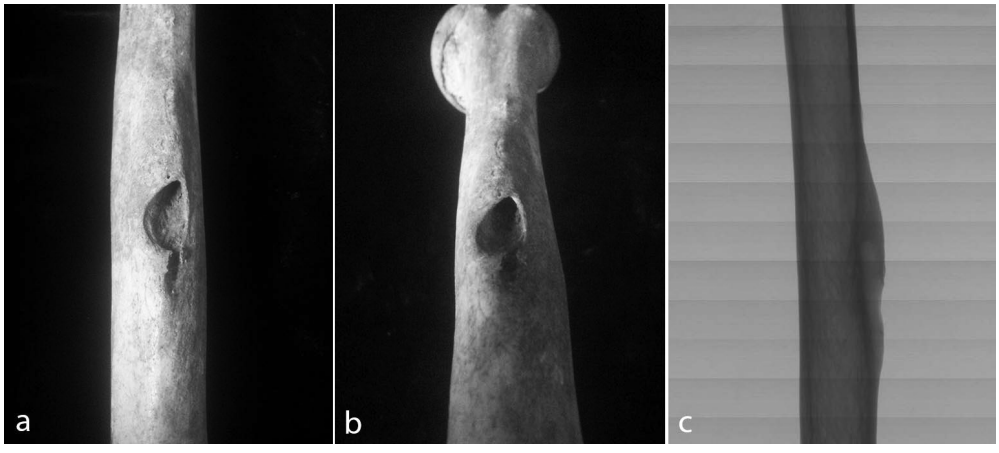

Fig. 6. Bone lesion on the left humerus: a-b) close views of the lesion; c) digital radiograph of the lesion, lateral view

are especially common in the metaphisis in children and occur less frequently in other tubular, flat or irregular bones, including the vertebral bodies, and are diaphyseal in location [12].

The left tibia was twice lighter in weight than the right one. The radiographs revealed excessive cortical thinning with medullary expansion (**Fig. 7**). It has been established that the immobilization causes a bone mass lost by reducing the bone mineral density in the epiphyses and by reducing the cortical wall thickness achieved by advanced endosteal resorption in the diaphyses [5]. Thus, the immobilization of the left leg of the individual due to some kind of trauma could be a reasonable explanation for this localized extra thinning.

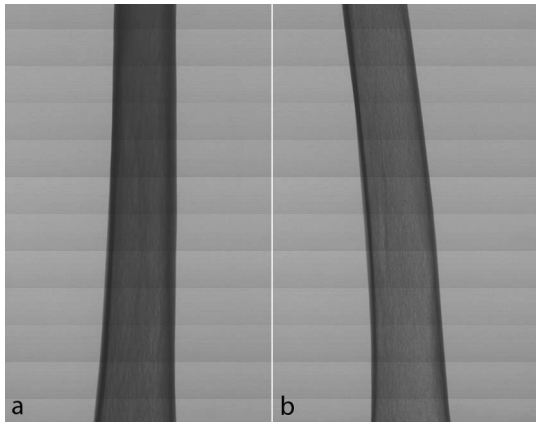

Fig. 7. Digital radiographs showing the cortical thickness of both tibiae: a) right tibia; b) left tibia

The stature of the individual, calculated on the basis of the lengths of the tibiae and the humeri despite the bowing (**Table 7**), was 164.4 cm. The stature was slightly below the mean body height of the recruits in Bulgaria during the period from 1897 to 1920 year which varied around 166 cm [14]. The individuals affected by skeletal dysplasias

most commonly had a disproportionately short stature due to short-trunk or short-limb. Obviously, in our case the individual was with normal length of the limbs, and bearing in mind that it was fit for service, probably was with normal trunk size as well.

Table 6. Lengths of the long bones in mm

Humerus		**Tibia**	
right	**left**	**right**	**left**
321	315	367	370

The differential diagnosis showed that the observed skeletal abnormalities most probably were due to craniotubular dysplasia with increased bone density. *Craniometadiaphyseal dysplasia* (CRMDD)*, wormian bone type* is such a disorder with autosomal recessive inheritance and cranial, metaphyseal and diaphyseal involvement. The generalized manifestation on the cranium includes: a large circumference of the skull; a prominent forehead, frontal bossing and parietal bulging; multiple WBs; a delayed ossification of the cranial vault and wide open anterior fontanelle; a calvarial thickening; a mild sclerosis of the skull base; obliterated paranasal sinuses and basal skull foramina; hypoplastic malar bones; noted maxilla and mandible; high palate; increased caries and dental hypoplasia. The most frequently manifested features on the postcranial skeleton include: an abnormal modeling of tubular bones; lack of normal diaphyseal constriction and poor metaphyseal flaring; osteoporotic bones with thin cortices; bowing of lower limbs; coxa valga; wide long bones, short tubular bones; widen ribs and clavicles; chest deformity; multiple fractures causing severe scoliosis. There were also reported isolated cases of occipital horns, natal teeth, distorted pelvis and short stature [3, 6, 12]. Most of the symptoms of this rare disorder resemble to a great extent those manifested in the investigated bone remains.

Pyknodysostosis is another skeletal dysplasia with increased bone density and similar manifestation on the cranium, but the difference came from the long bones appearance with generalized osteosclerosis, narrowed medullary cavity and short stature, particularly the limbs. By this reason, we suspected CRMDD as more likely diagnosis. However, absence of the other postcranial bones was an obstacle to the complete assessment of the disorder manifestation. Certainly, even though we performed a detailed skeletal survey, an investigation based solely on bone remains has to a great extent an interpretative character, whereas the confirmation of such rare and specific disorder requires more convincing proofs.

Conclusions

The observed abnormalities were due to skeletal dysplasia with increased bone density and CRMDD was suspected as a probable diagnosis. The individual was fit for service, which suggests a normal stature and normal mental development without severe malformations.

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