ABSTRACTS

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ENLARGED PARIETAL FORAMINA (EPF) IN THE OSTEOARCHAEOLOGICAL RECORDS OF THE CARPATHIAN BASIN

Z. Bereczki [1] , E. Molnár [1], G. Tóth [2], A. Marcsik [1], A. Hegyi [3]

Enlarged Parietal Foramina (EPF) is a unique developmental condition that has been present in osteoarchaeological materials since the Pleistocene. Contemporary imaging techniques provide solid evidence of this condition in modern populations and cases also show familial aggregation. However, EPF is rarely reported in the osteoarchaeological literature. We have gathered three possible cases of EPF in the Carpathian Basin so far, the remains of 30-35 years old female from the 8th - 10th century CE from the Avar cemetery of Debrece-Bellegeló, Bordás-Tanya (Gr. no. 339). The individual shows unequivocal signs of EPF, just like the skull remains of a 3-5 years old child from the 10th - 11th century CE early Hungarian cemetery of Balatonudvari - Fóvenyes (Gr. no. 286). The third possible case derives from the 1st - 4th century CE roman cemetery of Sárvár (Gr. no. 214). The 47-53 years old male also shows signs compatible with EPF, however, the first investigators of the material describe the lesions as a double trepanation. In our paper we give a detailed account of these cases and also discuss the differential diagnosis of EPF and other skull openings.
EPF is not a simple epigenetic trait. It may be linked to syndromes and epilepsy and may create a weakened site in the cranium prone to injuries. Hence, people with EPF in historical periods might have been mildly disabled compared to members of the non affected population. We may obtain a very interesting insight into the health standards of these past periods if we meticulously examine bone remains for these signs.

1: Department of Biological Anthropology, University of Szeged, Hungary
2: Institute of Biology, University of Western Hungary, Szombathely, Hungary
3: Biology Teaching Unit, Department of Physiology, Anatomy and Neurology, University of Szeged, Hungary
Corresponding author: Z. Bereczki, bereczki.zsolt@bio.u-szeged.hu

APPROACH TO THE FORMATION OF PALEOPATHOLOGICAL COLLECTIONS IN RUSSIA YESTERDAY AND TODAY

N. Berezina [1], A. Buzhilova [1]

The anthropological and ethnographical collection of the Moscow State University started as an educational exhibition for Moscovites in 1879, and the pathological collection was a part of the exhibition from the early beginning.
The purpose of the collection was to organize a database to work with paleoanthropological pathological findings, as well as medical research of clinical cases. For example, much attention was paid to changes in the shape of the skull and brain during different variants of early fusion of the sutures of the skull and microcephaly. There was assembled a significant collection of Russian materials, and casts of busts, brains and skulls of microcephalics from France, Germany, Estonia, Italy, USA and South America for the exhibition.
The collection of pathological cases was expanded and supplemented at the beginning of the 20th century by medical and paleoanthropological cases. Another significant Russian pathological collection was collected in the first half of the 20th century by the radiologist and one of the first paleopathologists D.G. Rokhlin in St. Petersburg. It was intended for medical students, and partly preserved till now.
At present time, paleopathological collections in Russia are replenished only by archaeological cases. The old and new parts of collections serve as comparative material and for the training of students of anthropology. With the development of new diagnostic methods, it is important to return to the old parts of the collections to clarify the diagnosis. For ethical reasons, we cannot add new cases of rare diseases to the collection, so it is important to preserve and study the heritage of our predecessors. This research was partly supported by Grant RFBR 17-29-04125.

1: Research Institute and Museum of Anthropology, Lomonosov Moscow State University
Corresponding author: N. Berezina, berezina.natalia@gmail.com

THE ANTHROPOLOGICAL COLLECTION OF THE NATURAL HISTORY MUSEUM VIENNA, A POTENTIAL FOR STUDYING RARE DISEASE

M. Berner [1] , S. Eggers [1]

The osteological collection of the Natural History Museum in Vienna with over 40,000 skeletons is a well known and widely used collection for anthropological studies. Despite the long collection history and large number of skeletal remains, so far no systematic palaeopathological data collections or compilations have been carried out. A closer look into the inventory book reveals that some skulls or skeletons were collected specifically because of their pathological or abnormal appearance, but some cases with apparent visible pathological conditions are also noted for archaeological series. These observations include cases with congenital abnormalities, predominantly descriptions of premature closure of cranial sutures or deformations/malformations of postcranial elements. Nevertheless, these observations have rarely been published, as in general publications including descriptions of such cases are difficult to trace. In our presentation, we want to address the potential of the museum's collection for a systematic inspection and data collection for studying rare diseases in the past. Given that the majority of the collection stems from neighbouring geographical regions from the Neolithic to the Early Medieval, such an endeavour could take into account a diachronic perspective, investigating changes in the frequency of rare diseases over time.

1: Natural History Museum-Vienna, Anthropology Department
Corresponding author: M. Berner, margit.berner@nhm-wien.ac.at

"THE GIANT AMONG THEM": A POSSIBLE CASE OF PITUITARY GIGANTISM FROM THE ROMAN CEMETERY OF EN CHAPLIX, AVENTICUM, SWITZERLAND (1ST-3RD C. AD)

C. Bourbou [1], M. Milella [1]

Skeletal growth and development is controlled by the growth hormone (hGH) and thyroxin from the pituitary and thyroid glands, respectively. Over- or insufficient hormone production can influence the size, shape and density of skeletal elements during growth and development, as well as later in adulthood. Disorders of the pituitary gland leading to growth hormone hypersecretion and gigantism (PG) are extremely rare today and scarcely documented in the paleopathological literature. This results not only in a fragmentary knowledge about their incidence in the past, but also about the social correlates of these conditions in ancient cultures. Here we present an adolescent male individual from the Roman necropolis of en Chaplix at Avenches (Western Switzerland, 1st-3rd century AD), buried prone and showing a suite of skeletal features (e.g. excessive long bone length, alteration of bone morphology) consistent pituitary gigantism. Besides being the second known possible case of pituitary gigantism from Roman times, the specific burial modality of this individual (the customary ritual for this time and geographic contexts is represented by supine inhumations) allows unprecedented insights about the social perception of gigantism in the past.

1: University of Fribourg/University of Zurich
Corresponding author: C. Bourbou, chryssab@gmail.com

THINKING ABOUT RARE DISEASES

J. Buikstra [1 2] , A. Cormier [1 2], A. Wissler [1 2]

This presentation considers the manner in which the identification and interpretation of Rare Diseases best inform paleopathology and bioarchaeology. We also contemplate how our study of past Rare Diseases may usefully advance contemporary perspectives on impairment and disability. We begin by considering the manner in which Rare Diseases are defined in the US and Europe, followed by a discussion of how best to gain knowledge of Rare Diseases, as expressed in archaeological materials. Standard texts in (paleo)pathology, Rare Disease databases, and clinical search engines are critically reviewed. We also explore the history of study for Rare Diseases, a field that is rapidly changing through insights gained from molecular biology. It is argued that for us to fully appreciate Rare Diseases in the past, relevant fields of study should include Anatomy (skeletal, dental, and soft tissue), Cell Biology, Human Embryology, Human Growth and Development, Human Genetics, Molecular Genetics, Palliative Care, Archaeology, and (Paleo)pathology. We also assert that the precise identification of conditions is less important than defining life course experiences of those affected by rare disease and the community that supported them. Finally, the malleability of individual identities of those suffering from Rare Diseases and their communities of concern are illustrated.

1: Center for Bioarchaeological Research, Arizona State University
2: Repatriation Office, National Museum of Natural History
Corresponding author: J. Buikstra: buikstra@asu.edu

UPPER PALEOLITHIC CASE OF DEFORMED FEMORA OF SUNGHIR 3 IN CONTEXT OF PALEOGENETIC DATA

A. Buzhilova[1 2]

The double child burial (Sunghir 2 and 3), one of the most spectacular and elaborate Upper Paleolithic funerary examples, was found in 1969 near the town of Vladimir, about 200 km northwest of Moscow. The Sunghir 3 skeleton is in a good state of preservation. Based on dental maturation, the age of it can be estimated at around 9–10 years. The Sunghir 3 femora are short and exhibit marked symmetrical antero-posterior bowing. Bowing affects the whole diaphysis and shows a regularly incurved profile, with the highest point at midshaft. Pathology is confined only to the femora. The isolated nature of the Sunghir 3 anomalies points to cases reported in the medical literature as ‘congenital bowing of long bones’ (CBLB). These are a group of rare conditions exhibiting localized, sometimes bilateral, bowing and shortening which are nonspecific and may result from different causes, including abnormalities of the primary cartilaginous anlage. Localized ossification disturbances might explain the shortening and the coincidence of maximum midshaft curvature with the position of the primary ossification center, as well as the lack of involvement of other skeletal parts. Other possibilities, like early bilateral midshaft fracture, acute plastic bowing deformities, or faulty fetal posture, are unlikely. The analysis of the European Upper Paleolithic record reveals a higher frequency of pathological deformed individuals, like the Sunghir children, Kostenki 14 (Russia) and individuals of Dolní Věstonice (Moravia). The genetic affinities between the Sunghir individuals, Kostenki, and the ‘Věstonice cluster’ are discussed in the context of developmental abnormalities.

1: Research Institute and Museum of Anthropology, Lomonosov Moscow State University
2: Institute of Archaeology, Russian Academy of Sciences
Corresponding author: A. Buzhilova: albu_pa@mail.ru

BEYOND THE PHENOTYPE: DETECTING DISEASE WITH ANCIENT DNA

L. M. Cassidy [1] , D. G. Bradley [1]

The growing field of ancient genomics has created a paradigm shift in our approach to disease detection in prehistory.
We apply these new avenues of discovery to three distinct prehistoric Irish populations. The incidence and distribution of known pathological mutations in each group is assessed, ranging from single point mutations, such as the C282Y mutation of the haemochromatosis gene, to large chromosomal scale abnormalities. Optimal methods for detection are discussed as well as potential pitfalls.
Finally, the anthropological implications of our results are considered, ranging from the inference of environmental stressors acting upon a population to the treatment of vulnerable members of a society.

1: Smurfit Institute of Genetics, Trinity College Dublin
Corresponding author: D. Bradley, dbradley@tcd.ie

MRI: A NEW POWERFUL TOOL IN PALEOPATHOLOGICAL DIFFERENTIAL DIAGNOSIS

M. Čavka [1]

Magnetic Resonance Imaging (MRI) is one of the most used imaging methods in modern medicine; for example in 2016 United States had population rate of annual MRI scans of 118 per 1000 people. MRI in clinical settings is still undeveloped and underused in paleopatology; it has been used only several times (in scientific study of mummies) as there aren't enough unbound protons in dehydrated bodies. Although some efforts have been made during the '80s with rehydration of mummified samples, the first successful MRI of a whole dry mummy was done by Ruehli et al. in Zurich in 2007 using the ultra short echo time sequence (UTE). From 2008-2012 mummified remains from the Egyptian Collection of the Archeological Museum in Zagreb, Croatia have been scanned on X-ray, CT and MRI in the University Hospital Dubrava, Zagreb, Croatia, and a case of ancient rare disease has been published in 2012 by our group. We have suggested that a young male, from the period between 950–790 B.C. was suffering from Langerhans cell histiocytosis (LCH), a rare group of disorders without well-understood aetiology. Furthermore we classified it as Hand-Schueller-Christian disease, one of three syndromes associated with LCH, which has the incidence up to 0.5 per 100000 children per year. As this was the first case of MRI use in differential diagnosis in paleopathology, we want to reassess the use of MRI in paleopathology in the context of rare diseases.

1: Dept. of diagnostic and interventional radiology, University Hospital Centre, Zagreb
Corresponding author: M. Čavka, mislav.cavka@yahoo.com

A FETUS WITH MULTIPLE ABNORMALITIES BORN IN 1735

D. Müller [1], H. Tönnies [2 3], H. Neitzel [3], K. Sperling [3], S. Clayton [4]

First published in 1737 as “Monstrum humanum rarissimum” by the physician Gottlieb Friderici, the striking appearance of the fetus continues to attract scientific enquiry. The fetus is well preserved under spirit in a closed glass vessel allowing continuing discoveries with the application of new technologies. We report new findings from Next Generation Sequencing (NGS) and review earlier array-Comparative Genomic Hybridisation (aCGH) results. Variant calling and filtering for common variation and variant consequences recovered ~20,000 Single Nucleotide Variants (SNV). A custom Copy Number Variation (CNV) calling pipeline and filtering recovered rare CNVs.

1: Klinikum Chemnitz, Säuglingsklinik, Chemnitz
2: Robert Koch-Institut, Geschäftsstelle Gendiagnostik-Kommission
3: Institut für Medizinische Genetik und Humangenetik, Charité, Campus Virchow-Klinikum, Humboldt Universität
4: Max-Planck-Institut für Menschheitsgeschichte
Corresponding author: S. Clayton, clayton@shh.mpg.de

“MEDICINALIUM OBSERVATIONUM EXEMPLA RARA” BY REMBERT DODOENS (1581)

M. Dooms [1]

Rembert Dodoens was a Flemish physician also known as Rembertus Dodonaeus, who was born in Mechelen (now Belgium) on the 29th of June 1517. His most famous book is *Cruydenboeck* (“herb book”) and in 1581, he wrote *Medicinalium observationum exempla rara. Medicinalium observationum exempla rara, recognita et aucta. Accessere et alia quaedam, quorum elenchum pagina post praefationem* exhibit by Rembert Dodoens medici Caesarei along with Valesco de Tarenta, Alexander Benedictus, Antonio Benivieni, Maternus Cholinus, Mathias Cornax, Achilles Pirmin Gasser and Gilles de Hertoghe. After a general introduction and a list of cited authors, Rembert Dodoens gives extensive list of some 200 rare diseases in the 16th century such as *Aneurisma, Calculus in vesica* (stone in the bladder), *Catalepsis* (seizure), *Diabetes, Dysenteria, Gemini pueri* (twins), *Lapides in vessica fellis* (gall stones), *Mania cum Melancholia affinitatem habet* (mania with melancholia) *Scorbutus, Tetanos, Vermis in vesica* (worms in the bladder) *and Vomitus sanguinis* (vomiting blood). Throughout the Middle Ages, to be diagnosed with a (rare) disease had major social (“stigmatization”) and medicinal implications for the individual. Some communities, knowing the importance of an accurate diagnosis, established multidisciplinary groups (“expert centers”) to review suspected cases. Cousin marriage was common at that time in Europe, which resulted in multiple genetic diseases. Children were born at home from teenage house-wives sometimes with the help of a midwife following the guidelines of Eucharius Rhodion’s Der Rosengarten (1513). Long-time breastfeeding by the mother or by a wet nurse was general practice. Infanticide became exceptional in the 16th century, and unwanted children were left at the door of church or abbey, and the clergy was assumed to take care of their upbringing.

1: University Hospitals Leuven, Belgium
Corresponding author: M. Dooms, marc.dooms@uzleuven.be

SPECIMEN COLLECTIONS AND MUSEUMS: USING THE PAST TO INFORM THE PRESENT AND THE FUTURE

W. Edwards [1]

For many years specimen collections have largely been seen as at best, quaint and obsolete and at worst worthless and rather unpleasant. However recently there have been some signs of a rediscovery of such collections and their true potential. This presentation aims to give an overview of the purposes of Museums and Curation; comparing different Museums, collection strategies and the application of new technologies. The aim is to demonstrate the vital role of such collections in research, teaching, public engagement and fund raising. Some skeletal specimens will be used as examples.

1: Gordon Museum, Kings College London
Corresponding author: W. Edwards, william.edwards@kcl.ac.uk

CAN DUCHENNE MUSCULAR DYSTROPHY BE A MARKER FOR THE CARE GIVEN TO DISABLED CHILDREN IN THE PAST?

S. Eggers [1], M. Berner [1]

The treatment of persons with physical disabilities is extremely variable across time and space. Affected persons may be pampered, given respected status and special social functions, but they may also be treated as economic liabilities, reluctantly kept alive or totally rejected. Historical accounts show that often neglect, ignorance and fear contributed to social isolation of persons with disabilities.
Physical disabilities include paralysis. Year-long limb paralysis causes bone atrophy due to trauma, poliomyelitis, cerebral palsy, multiple sclerosis or even neural or muscular degeneration.
Today we recognise more than 30 different types of progressive muscular dystrophies. Together they account for a prevalence of circa 22 affected/ 100,000 persons/year. This is only four times smaller than the prevalence of tuberculosis or trisomy 21. Since muscular dystrophies (MD) are genetic diseases, they must also have afflicted people in the past as often as today. This is especially so for Duchenne muscular dystrophy (DMD). Apart from being one of the most frequent genetic diseases (affecting one in every 3500 live-born boys), DMD is X-chromosomal recessive (providing a 50% recurrence risk in male children of usually unaffected carrier mothers). Since DMD progressively weakens limbs to the incapacity of ambulation at age 10 and leads to complete paralysis and death at about age 20, DMD should be detectable as severe bilateral limb atrophy in the archaeological record.
Depending on the age at death and the accompanying archaeological contexts, DMD candidate cases can shed light on the care and significance of children incapable of ambulation in ancient communities.

1: Natural History Museum-Vienna, Anthropology Department
Corresponding author: S. Eggers, Sabine.Eggers@nhm-wien.ac.at

SELECTED CASES OF CONGENITAL MALFORMATIONS AND PATHOLOGIES IN A HISTORICAL WET-SPECIMEN COLLECTION

P. Eppenberger [1]

Paleopathological research is highly dependent on well-documented pathological specimens from individuals who have not received modern advanced pharmacological and/or surgical treatment. The aim of this presentation is, therefore, to highlight the historical wet specimen collection of the Institute for Evolutionary Medicine of the University of Zurich (IEM), which is as yet a relatively little known Swiss reference series of high potential value for paleopathological research. Our collection consists of over 2000 pathological specimens from the late 19th and early 20th centuries preserved in formalin solution, and clinical background information as well as autopsy reports are available for a large number of specimens. Since these specimens do not only contain skeletal material but also soft tissues, they constitute a valuable supplementary reference series to the many well-documented historical human skeletal reference series such as the collections of the Mütter Museum at the College of Physicians of Philadelphia, the Warren Anatomical Museum at Harvard Medical School in Boston, the National Museum of Health and Medicine at the Armed Forces Institute of Pathology in Washington DC, or the IEM's Galler Collection in Zurich. In this talk several examples of rarely documented congenital malformations and pathologies from the IEM's wet specimen collection, such as conjoined twins, holo-acranial anencephaly, polydactyly, Klippel-Feil syndrome or congenital generalized osteosclerosis shall be presented.

1: Institute of Evolutionary Medicine, University of Zurich
Corresponding author: P. Eppenberger, patrick.eppenberger@iem.uzh.ch

A PROBABLE CASE OF OSTEOPETROSIS TARDA IN AN ADULT NEOLITHIC SKELETON FROM PALATA 2-OFANTO RIVER VALLEY (CANOSA-SOUTH ITALY): RADIOLOGICAL, HISTOLOGICAL AND CONFOCAL LASER MICROSCOPY STUDY

M. Favia[1], D. Ferorelli[1], V. Scattarella [2], S. Sublimi Saponetti [2], F. Radina [3], E. Dellù [3], S. Sivilli [3], A. Dell'Erba [1], G. Favia [1]

In Canosa di Puglia - S Italy (loc. Palata 2) a stratigraphic deposit connected to a Neolithic settlement was brought to light in 2008. The burial referred to the individual US 21 made in a quadrangular pit and dated to the middle of 6th millennium B.C. (radiocarbon dating: LTL 5188A 6.561 ± 50 BP, calibrated 2 sigma 5.630-5.460 BC). The anthropological analysis has shown that it is a male subject, with an age between 21 and 40 years with macroscopical evidence of generalized hyperplastic osteosclerosis with increase of bone thickness. Skull and long and flat bones have been studied with X-rays and biopsied. Serial histological section were made and observed with a Nikon Eclipse confocal laser microscopy. The bone showed an increase in density, and in thickness of the compact cortical bone with very large osteons; the medullary bone is constituted by large and massive trabeculae, with decrease of medullary spaces, and with a compact-bone like appearance. The confocal laser histological pictures showed the co-existing presence of areas with different degree of calcification and negative fluorescence of the hyper-calcified atypical bone. The radiological and the histological findings are typical of a hyperplastic sclerosing bone disorder, with high grade of calcification, with very few remodelling areas, and with the pathognomonic aspects of the Osteopetrosis tarda, a rare hereditary bone disorder that presents in adulthood in a benign clinicopathological form.

1: Interdisciplinary Department of Medicine, University of Bari (Italy)
2: Department of Biology, University of Bari (Italy)
3: Superintendence Archeology, Fine Arts and Landscape, for the metropolitan city of Bari (Italy)
Corresponding author: M. Favia, matteofavia@me.com

A PARENTAL RELATIONSHIP BETWEEN THREE NEOLITHIC SKELETAL REMAINS FROM AN APULIAN TOWN (ITALY): A CASE OF FAMILY-CELIAC DISEASE WITH HIGH-GRADE OSTEOPOROSIS.

D. Ferorelli [1], M. Favia [1], F. Radina [2], E. Dellù [2], S. Lonero Baldassarra [1], V. Scattarella [3], G. Favia [1], A. Dell'Erba [1]

In 1997 in Rutigliano (Bari) a little town in South Italy - was accidentally discovered a monumental grave. A large funerary structure with quadrangular plant. It has been dated to the Final Neolithic Age (Diana culture, final 5th - beginning of 4th millennium B.C.) and contained three skeletal remains in crouched position: one male of 30-35 years old, one male and one female of 21-25 years old. The aim of the study was to define any possible parental relationship between individuals and their physical and health conditions. Anthropological, dental, histological and genetic comparisons were carried out. Teeth and bones were analyzed with optical and confocal laser microscopy; DNA extraction was performed with a modified protocol of NucleoSpin DNA Trace kit (Macherey Nagel). Radiological examinations have shown widespread osteoporosis in all the examined bone districts; histopaleopathological studies revealed high-grade osteoporosis and many areas of hypoplasia of enamel. Paleonutritional analysis showed values connected to calcium-related elements of more than 50% lower in two subjects compared to the third. Genetic studies revealed a parental relationship between these skeletal remains. If one could imagine extremely different nutritional habits among the three individuals, the plausible kinship, the radiological findings and especially the histopaleopathological ones agree with the presence of family-celiac disease. This multidisciplinary study in this specific sample has found a case of family-celiac disease with high-grade osteoporosis.

1: Interdisciplinary Department of Medicine, University of Bari (Italy)
2: Superintendence Archeology, Fine Arts and Landscape, for the metropolitan city of Bari (Italy)
3: Department of Biology, University of Bari (Italy)
Corresponding author: D. Ferorelli, davideferorelli@gmail.com

A CASE OF SCLEROSING BONE DYSPLASIA FROM 16TH CENTURY SARDINIA: CAMURATI-ENGELMANN DISEASE?

V. Giuffra [1], D. Caramella [2], M. Milanese [3]

The skeletal remains of a male aged 45-55 years displaying several bone anomalies were unearthed from the Alghero plague cemetery "loc. Quarter", a burial site dating back to 1582-1583 AD outbreak. The skeleton is characterised by marked thickening of the cranial vault and a symmetrical enlargement of the diaphyses of the long bones of the upper and lower limbs. The metaphyses appear to be involved, while the epiphyses are spared. X-rays and CT showed increased irregular cortical thickness and narrowing of the medullary cavity. The individual was affected by a sclerosing bone dysplasia, a genetic disease characterised by increased bone density. Sclerosing bone dysplasias are rare genetic diseases whose variety makes the clinical diagnosis challenging; in our case the mature age of the individual, with evident signs of late stage of the disease, helped us identifying the disorder. Differential diagnosis allowed to suggest a possible case of Camurati-Engelmann disease, also known as progressive diaphyseal dysplasia. It is an autosomal-dominant condition caused by mutations within the transforming growth factor-β1 (TGFβ1) gene on chromosome 19q13, characterised by anomalies in intramembranous bone formation. It is a very rare disorder, and approximately 200 cases have been reported. The individual from Alghero should be symptomatic, since he is likely to have experienced the most common clinical symptoms, such as pain in the limbs and fatigability, and to have had an unusual gait. This case represents the unique paleopathological evidence of Camurati-Engelmann disease so far.

1: Division of Paleopathology, Dept. of Translational Research and New Technologies in Medicine and Surgery, University of Pisa (Italy)
2: Division of Diagnostic and Interventional Radiology, Dept. of Translational Research and New Technologies in Medicine and Surgery, University of Pisa (Italy)
3: Dept. of History, Human Sciences and Education, University of Sassari, (Italy)
Corresponding author: V. Giuffra, valentina.giuffra@unipi.it

RARE PATHOLOGIES IN DEEP TIME - A FRESH LOOK AT BONES OF FOSSIL ANIMALS

Y. Haridy [1], F. Witzmann [1]

Paleontologists have long been interested in paleopathology as an 'anomaly science' often only documenting the presence of an abnormality without diagnoses. More recently due to improved technology diagnosing these early pathologies has become possible, and paleontologists at the Museum für Naturkunde Berlin have made many new discoveries. Here we present an overview of some of the rare diseases that have been diagnosed at the Museum für Naturkunde Berlin: (1) Congenital vertebral malformations like hemi- and wedge vertebra and congenital block vertebra in early amphibians (up to 300 million years old) and in dinosaurs (up to 150 million years old), as well as spina bifida in a 250 million years old dicynodont, a mammalian ancestor. (2) Paget's disease of bone, although not rare in humans today, has only been reported two times in fossils: in a dinosaur (150 million years old) and an early reptile (300 million years old). (3) Schmorl's nodes are not rare in humans, but similar structures on the vertebral bodies are exceptional in fossil non-mammalian vertebrates. However, these cases do not represent true Schmorl's nodes because reptiles do not have intervertebral discs like mammals, but rather have synovial joints between the vertebral bodies. Thus we designated this pathology as synovial cysts as reptilian analog of Schmorl's nodes. (4) Bone cancer is very rare in the fossil record and we will present one of its earliest occurrences in a fossil turtle (240 million years old). (5) Polydactyly and malformed extremities in ancient amphibians (300 million years old) as consequences of failed limb regeneration, analogous to modern salamanders that are the only tetrapods today that can fully regenerate their limbs. We discuss the impacts of each of these findings, as well as future directions in better recognising rare diseases in the fossil record, and finally how these diseases aid in understanding the evolution of healing.

1: Museum für Naturkunde Berlin
Corresponding author: Y. Haridy, Yara.Haridy@mfn.berlin

NGS-BASED ANALYSIS OF RARE SKELETAL PHENOTYPES

U. Kornak [1 2 3]

More than 400 of the approximately 5000 rare hereditary disorders display significant skeletal involvement. While specific skeletal phenotypes like achondroplasia are easily diagnosed clinically many other skeletal disorders show strong overlap. Since the advent of next generation sequencing (NGS) genetic profiling has therefore become a main tool for differential diagnosis of skeletal disorders besides radiological assessment. The approaches range from disease-specific gene panels to whole exome and whole genome sequencing. The unprecedented flood of genetic information generated by these approaches can only be dealt with through powerful bioinformatics. A recent and important innovation was the prioritization of genetic variants according to phenotype relevance, which relies on a computable phenotype description using the human phenotype ontology. Several examples will be given for phenotypes solved by this strategy. But NGS goes beyond the identification of genomic alterations. RNA-sequencing can improve the diagnostic yield by revealing change of splicing and expression levels. Chromatin profiling as done in HiC or Capture C can elucidate how copy number variants like microdeletions perturb the architecture of the genome and lead to aberrant gene expression during skeletal development. While amazing progress has been achieved we are still far from understanding the 98% of the genome that are not harboring genes. A joint evolution of sequencing technology, databases, and software tools is needed to fully appreciate the information contained in the genome.

1:Institut für Medizinische Genetik und Humangenetik, Charité-Universitätsmedizin Berlin (Germany)
2: BIH Center for Regenerative Therapies, Charité-Universitätsmedizin Berlin (Germany)
3: Max-Planck-Institut für Molekulare Genetik, Berlin (Germany)
Corresponding author: U. Kornak, uwe.kornak@charite.de

SCLEROSING BONE DYSPLASIAS AND THE OSSIFICATION PATHWAY: DIFFERENTIAL DIAGNOSIS OF A DIFFUSE SCLEROSIS IN A SKULL OF AN IDENTIFIED MALE (EARLY 20TH CENTURY COIMBRA, PORTUGAL)

B. M. Magalhães [1 2], L. Catarino [3], I. Carreiro [4], R. Gomes [1 2], R. C. Ramos [1 2 4], V. Matos [1 2], A. L. Santos [1 2]

This work aims to discuss the differential diagnosis of a sclerosing dysplasia. The skull of a 61 year-old male from the International Exchange Skull Collection who died in 1928 (registered cause of death: albuminous nephritis [Bright disease]/uraemia) was macroscopically and radiologically observed and subjected to X-ray fluorescence (XRF). The cranium and mandible were weighed on a digital scale and fifteen measurements and five indexes were compared with reference data. Although the skull presents normal morphology confirmed by the values of the measurements and indexes, the weight is about three times (cranium: 1.731kgs, mandible: 0.151kgs) heavier than the average (Silva et al., 2009). Radiologically, the skull shows diffuse thickening, with the trabecular bone assuming a cortical appearance. The diffuse aspect is enhanced in the diploe of the calvaria and mandible, and less in the remaining facial bones. Elemental analysis (XRF) revealed higher levels of Cu, Zn and Pb when compared with other identified skulls. Accumulation of these elements is usually associated with diagenetic effects; however, higher levels of Cu may be associated with albumin. Osteopetrosis, Voorhoeve disease, Camurati-Engelmann disease, Erdheim-Chester disease (which is clinically associated with chronic renal failure), or several asymptomatic sclerosing bone dysplasias are possible diagnosis. This work underlines the difficulties of a definitive diagnosis, especially in the absence of the postcranial skeleton, and shows that a broader diagnosis is the best choice when pathognomonic characteristics are absent.

1: Department of Life Sciences, University of Coimbra (Portugal)
2: Research Centre for Anthropology and Health (CIAS), University of Coimbra (Portugal)
3: Geosciences Center, University of Coimbra (Portugal)
4: Coimbra Hospital and University Centre (CHUC) (Portugal)
Corresponding author: B. Magalhães, bruno.miguel.silva.magalhaes@gmail.com

IN SILICO PALEOPATHOLOGY: VIRTUAL AND QUANTITATIVE APPROACHES FOR THE ANALYSIS OF DEVELOPMENTAL ANOMALIES OF THE SKELETON

M. Milella [1]

Genetic disorders affecting skeletal growth and development include a large range of conditions characterised by a marked clinical diversity and genetic heterogeneity. Their rarity and their phenotypic variability, spanning from mild developmental deviations to gross anomalies, presents a challenge when trying to assess and discuss them in osteoarchaeological specimens. These issues are further complicated by the fragmentation typically characterising most skeletal remains from archaeological contexts.
The increasing availability of quantitative three-dimensional analytical tools represents a solid ally to the paleopathologist, especially when dealing with complex skeletal presentations and fragmentary remains. This contribution illustrates the application of a range of approaches (e.g. virtual reconstruction, tomographic analysis, and geometric morphometrics) in the analysis and discussion of two paleopathological cases from Switzerland involving rare developmental anomalies: a Neolithic child affected by Léri Weill dyschondrosteosis, and a medieval male presenting the bilateral lack of fusion of the triradiate cartilage. The results of the qualitative and quantitative analyses are in both cases discussed from a developmental, functional, and behavioural perspective.

1: Department of Anthropology, University of Zurich
Corresponding author: M. Milella, marco.milella@aim.uzh.ch

ISOLATED OR RESPECTED? - SEVERE CRANIOFACIAL CLEFT FROM THE 10TH CENTURY AD IN HUNGARY

E. Molnár [1], A. Hegyi [1 2], G. Pálfi[1], Z. Bereczki [1], L. Kis [1], E. Pap [3], A. Marcsik [1]

Orofacial clefts (OFC) comprise cleft lips, with or without cleft palate, and cleft palate alone. OFCs result from arrested development during embryogenesis and are amongst the most common congenital anomalies in modern populations. There is a well-known worldwide variation in the prevalence of OFCs at birth, ranging between 0.45–3.62/1000. Their complex aetiology has been postulated to involve chromosome rearrangements, gene mutations and environmental factors.
Despite its high frequency in recent populations, these conditions are rarely reported in archaeological context. This discrepancy is likely to be the result of a number of factors, including high mortality in infants born with OFCs, which reduces the possible recovery of the condition in skeletal remains from past populations. The paucity of recovered OFC cases encourages close attention to each case in order to document the development of the pathology and to investigate how past cultures addressed this condition. This study presents the results of the macromorphological analysis of a juvenile male individual coming from an isolated burial dated to the 10th century AD (Hungarian Conquest Period) presenting osteological signs of a severe craniofacial cleft. The right and left maxillae did not close and a funnel-shaped gap can be seen between them. All incisors and the left canine are missing, and the right upper canine is abnormally located. According to the special grave goods (e.g., a saddle with bone plating, a sabretache plate, silver mountings of a belt and a horse skull showing abnormalities) it can be supposed that this juvenile male played a particular role in the community.

1: Department of Biological Anthropology, University of Szeged (Hungary)
2: Biology Teaching Unit, Department of Physiology, Anatomy and Neurology, University of Szeged (Hungary)
3: István Türr Museum, Baja (Hungary)
Corresponding author: E. Molnár, molnar.era@hotmail.com

MULTIPLE OSTEOCHONDROMAS: CURRENT UNDERSTANDING AND IDEAS FOR THE FUTURE

E. Murphy [1], C. McKenzie [2]

Multiple osteochondromas, confusingly also known by a myriad of other names, is a rare inherited autosomal dominant condition of enchondral bone growth which primarily involves the development of multiple osteochondromas – or exostoses capped with cartilage – at the metaphyseal regions of the long bones. The condition has been most thoroughly studied among White populations which have prevalences of 0.9 to 2 individuals per 100,000. Strikingly higher rates have been identified for more isolated modern populations, however, and the palaeopathological evidence suggests this may also have been true in the past. The formation of multiple osteochondromas is the best known characteristic but the disease also involves the development of a suite of orthopaedic deformities, including disproportionate short stature; limb-length discrepancies; forearm deformities; coxa vara of the femora; valgus deformities of the knees and ankles; asymmetry of the pectoral and pelvic girdles; shortening of the foot and hand bones and scoliosis. While it is relatively straightforward to identify the osteochondromas, synonymous with the condition, it is more difficult to precisely describe some of the orthopaedic complications using conventional analysis. Clinical studies have indicated that the size and number of lesions can vary significantly so a further complication may be that only individuals with more severe manifestations of the condition will be recognized archaeologically.
To date, some 18 palaeopathological cases of multiple osteochondromas have been identified. The presentation will provide an overview of the condition with recourse to these cases, particularly those from Ballyhanna, Co. Donegal, Ireland. Suggestions for analysis beyond morphology will also be advanced.

1: Archaeology and Palaeoecology, School of Natural and Built Environment, Queen's University Belfast (N. Ireland)
2: Department of Archaeology, University of Exeter (England)
Corresponding author: E. Murphy, eileen.murphy@qub.ac.uk

DWARFISM IN PREDYNASTIC AND EARLY DYNASTIC EGYPT: NEW EVIDENCES FROM THE ELITE CEMETERY HK 6, HIERAKONPOLIS

A. Pieri [1 2]

Several types of growth disorders are known today and they can be divided between disproportionate and proportionate dwarfism. By far the higher number of cases come from Ancient Egypt, either in the form of tomb illustrations, statues or as human remains.
The skeletal evidences are of very high value, being the most objective evidence of the existence of this genetic condition.
In this view the recent discoveries in the Elite cemetery HK 6, in Hierakonpolis, are incredibly important. The most surprising discovery of the last few seasons was in fact the skeletons of two individuals affected by a form of dwarfism. Both these dwarfs presented almost all of the features typical of achondroplasia, however a careful observation revealed some differences in the morphology of the bones, which might suggest, a peculiar variation of this disorder.
The presence of two individuals, possibly affected by a variation of the common dwarfism, from the same cemetery and about the same time, open several questions about their relationship, provenance and provide several new information about the role that these individuals had in the predynastic society.

1: Hierakonpolis Expedition
2: Museo Civico Archeologico di Cecina (Italy)
Corresponding author: A. Pieri, anna.pieri@gmail.com

TWO WOMEN (18TH -20TH CENTURY) WITH SHORT STATURE: THE CHALLENGE OF THE DIFFERENTIAL DIAGNOSIS

A. L. Santos [1], M. Arrieta [2], M. do Rosario Martins [3], V. Martins [4]

Short stature may result from familial genetic, chondrodysplasias, endocrine, malnutrition, and/or chronic illnesses. This work discusses the possible aetiologies for the short stature of two females.
Individual 1: 26-years-old, from the Coimbra Identified Skeletal Collection, who died of measles in 1920. Her estimated height is 138.91-144.3cm, approximately 3 standard deviations below the average female stature for early 20th century Portugal. The crural, brachial, humero-femoral and intermembral indexes show a proportionate body, uncommon in dwarfism. Skeleton examination revealed a small skull, with prominent frontal, prognathism, obliterated sagittal suture, long/oval *foramen magnum*, small mandible with *masculine* features. Other lesions are multi-enamel hypoplasias, cribra orbitalia, porotic hyperostosis and proliferative reaction in the petrous portion of the temporal. Ribs sternal ends are flattened/ wider and vertebrae present developmental defects (e.g. atlas with both left transverse foramen and posterior tubercle open, absence of the right transverse foramen in axis, sacrum with six vertebrae). The differential diagnoses take into account clinical and paleopathological knowledge. Among the possible aetiologies are Turner syndrome and idiopathic short stature.
Individual 2: glove (similar size of the index and ring fingers) and shoe (12cm) worn by a woman at age 55, who lived at end of 17th/early 18th century. According to the inventory description at the Museum of Science, she had a proportionate body and the height of a 6 year-old-girl. The stature (less than 1m) points to achondroplasia or hypopituitarism.
These individuals clearly show the difficulties of retrospective diagnosis because there are more than 200 conditions which cause serious growth problems.

1: Department of Life Sciences, University of Coimbra (Portugal)
2: Departamento de Ciencias Naturales, Universidad Nacional de Río Cuarto (Argentina)

3: Museum of Science, University of Coimbra (Portugal)
4: Maternity Bissaia Barreto, Coimbra Hospital and Universitary Centre (CHUC) / Portugal)
Corresponding author: A. L. Santos, alsantos@antrop.uc.pt

RARE DISEASES AND WHERE TO FIND THEM. TOWARDS A DIGITAL ATLAS OF ANCIENT RARE DISEASES

W. Schmidle [1], E. Petiti [2], J. Haelm [2], J. Gresky [2]

Reports on rare diseases from archaeological contexts are most often published as case studies in specialised literature. The data is not standardised, and especially older publications are missing a lot of information that would be of interest when comparing with other cases. The current situation heavily affects the visibility of the available data on rare diseases in archaeology. We are developing a digital atlas on ancient rare diseases as an open access online platform for a more systematic approach to rare diseases. Apart from the possibility to collect standardised information for better comparison of cases, the atlas will provide tools to search for rare diseases in geographic areas or in chronological periods. Pictures and descriptions of the most important skeletal changes will help in diagnosing rare diseases in archaeological specimens. We have created a data model and implemented a prototype using data about cases of dwarfism. The digital atlas of rare diseases is conceived as a project for the E-RIHS DIGILAB for standardised, interoperable and if possible multilingual heritage data and will become part of the iDAI.world, the catalogue of online services of the DAI. The atlas makes use of available norm data for diseases (ICD-11), places (iDAI.gazetteer), periods (iDAI.chronontology) and literature (iDAI.bibliography Zenon). Additional literature references, place and periods can be added back to these systems as new norm data.

1: IT Department, DAI Berlin (Germany)
2: Department of Natural Sciences, DAI Berlin (Germany)
Corresponding author: W. Schmidle, wolfgang.schmidle@dainst.de

A POSSIBLE CASE OF HISTIOCYTOSIS-X AND THE DIFFERENTIAL DIAGNOSES IN MACERATED BONE SPECIMENS

M. Schultz [1]

Histiocytosis, also known as Langerhans cell histiocytosis (LCH), was formerly known as histiocytosis X. It is not a uniform disease, but represents a rare disease group that involves specific cells of the immune system. The cause of the cell changes leading to the individual diseases of this group is still unknown. In fact, LCH often behaves like cancer.
In this article, a late-antiquity case from Neuburg / Danube in Bavaria (Germany) is presented again, which has already been examined macroscopically and radiologically (Zink and Nerlich 2001). The differential diagnoses resulting from the microscopic examination are discussed.

1: Zentrum Anatomie, AG Paläopathologie, Georg-August-Universität, Göttingen, (Germany)
Corresponding author: M. Schultz, mschult1@gwdg.de

OSTEOARCHAEOLOGICAL IDENTIFICATION OF RARE DISEASES FROM THE OSTEOLOGICAL COLLECTION OF THE CROATIAN ACADEMY OF SCIENCES AND ARTS

M. Šlaus [1]

This talk focuses on two diseases from the Osteological collection of the Croatian Academy of Sciences and Arts. The first is a case of proportionate dwarfism identified in an adult female from the Zvekovice site near Dubrovnik. This type of dwarfism is characterized by proportionate shortening of long bones that exhibit marked medullary stenosis and thin, sclerotic skulls lacking diploic space and is known as Kenny-Caffey syndrome. It is most likely caused by a deletion in the TBCE gene and is extremely rare. The first case was published in 1967 and so far fewer than 60 cases have been reported worldwide with, obviously, this case representing the oldest example of the disease.

The second disease is present in the Kranj skeletal series from Slovenia and is a joint disease characterized by polyarticular, bilateral lesions exhibiting no new bone formation, no peri-articular erosions, no osteopenia, periosteal reactions or bony ankylosis ruling out the more recognizable joint diseases such as rheumatoid arthritis, juvenile idiopathic arthritis, psoriatic arthritis, Reiter's syndrome, osteochondritis dissecans, or gout. The disease follows a regular progression in severity beginning with small lytic lesions leading to greater areas of destruction with irregular geographical shapes, finishing with complete destruction of the joint surface. The lesions are more frequent in females than males and affect individuals from all age groups. A potential diagnosis may be multicentric reticulohistiocytosis also known as lipoid dermato-arthritis, a rare systemic histiocytic polyarhritis. If so, this would be the first example of this disease in an archaeological series.

1: Croatian Academy of Sciences and Arts
Corresponding author: M. Šlaus, mslaus@hazu.hr

A PROBABLE CASE OF AMELOGENESIS IMPERFECTA FROM A NORTHERN ITALY MEDIEVAL CEMETERY

C. Tesi [1], O. Larentis [1], R. Fusco [1], M. Licata [1]

A case of severe dental enamel defects was discovered during the archaeological excavations of the Medieval cemetery of San Biagio in Cittiglio (Varese, Northern Italy). The remains of an infant individual, aged 18 months - 2 years, show intensive pathological alterations at the enamel of the deciduous dentition. In particular, the crowns of the maxillary teeth exhibit circumferential bands of missing enamel and extended erosion of the underlying dentine. Areas of defects are mostly detected at the labial surfaces of the anterior teeth and at the buccal and occlusal surfaces of molars. These lesions are localised approximately at the middle of the dental crown, without affecting the cervical area, and display sharp and undercut margins, showing evidence of chipped-off enamel. Moreover, almost all the teeth exhibit an opaque yellow-brownish discolouration of the enamel that spares the cervical region.

A detailed differential diagnosis that comprises plane-form enamel hypoplasia, congenital syphilis, fluorosis, molar-incisor-hypomineralisation, early childhood caries, dentinogenesis imperfecta and amelogenesis imperfecta (AI), allowed us to assume the AI as the most probable diagnosis. Since in osteoarchaeological literature only a few cases of AI are recorded, we think that this occurrence related to an infant subject can be interesting for the paleopathological field, representing a new evidence of a severe genetic dental condition.

1: Centre of Research in Osteoarchaeology and Paleopathology, Department of Biotechnology and Life Sciences, University of Insubria, Varese (Italy)
Corresponding author: C. Tesi, ctesi@uninsubria.it

A GLOBAL PERSPECTIVE ON DWARFISM – LITERATURE REVIEW

B. Teßmann [1]

Dwarfism is a rare disease and belongs to the group of skeletal dysplasias. This is a congenital disorder of the bone and cartilage tissue (osteodysplasia or osteochondrodysplasia). In total, there are over 200 different types of dwarfism. The most common type of dwarfism is achondroplasia, which occurs at a frequency of 1:10 000 live births. Achondroplasia is inherited by a chromosomal abnormality or triggered by gene mutations. The dwarfism can therefore be hereditary indexed, but also occur suddenly. In addition, there are also quite different causes, such as metabolic diseases or malnutrition. In archaeological cases the precise diagnosis of their causes is often impossible. With dwarfism no intellectual restrictions go hand in hand, which is why dwarfs took in the Egyptian and Greek antiquity social special positions. The skeletons usually show no signs of violence. This is certainly testimony to the fact that these people were treated with sympathy with their handicap and had their place in society. The article gives an overview of archaeological finds and explores how prehistoric and historical societies dealt with this phenomenon. The oldest evidence of dwarfism comes from the late Paleolithic in southern Italy. Here, too, a short-stature man in a pure hunting and gathering community had his firm place in the group. Due to his small height and massive restriction in the mobility of his arms, he was probably dependent on the help of other group members, for example in hunting. For indigenous communities in North America, on the other hand, there seems to be historical evidence showing that infants with disabilities have been exposed to the prairie.

1: Museum für Vor- und Frühgeschichte, Berlin SMPK
Corresponding author: B. Teßmann, barbaratessmann@gmx.de

DEEP IN THE MARROW AND BLOOD: A PROBABLE CASE OF ANCIENT LEUKEMIA IN THE SOUTH AMERICAN ANDES

J. M. Toyne [1], C. Schow [2]

While cancer today seems prevalent and highly insidious, evidence for cancer in the past is limited to few case studies and its differential diagnosis from skeletons remains challenging. Yet, various manifestations of probable cancers have been identified from bone lesion across the globe. It remains essential that these examples are situated within the broader cultural context to provide a richer understanding of the uniqueness, evolution, and impact of these different types of diseases. In this case study, we construct an osteobiography from the partially mummified remains of an individual discovered in an archaeological burial complex from Chachapoyas, Peru (~1350-1450 AD) using osteological, paleopathological, radiographic, and isotopic analyses. The adult male (30 to 44 years) is incomplete, but demonstrates diffuse small round and oval resorptive lesions across the cortical surfaces of the ribs, vertebrae, and basilar portion of the occipital bone. Lesions are not localised, and do not have sclerotic or lytic margins, thus appearing to be derived from a systemic process.We consider Cushing's Syndrome, hyperparathyroidism, and multiple myeloma as possible diagnoses, but feel that based on the pattern, distribution, and size of the lesions as well as the epidemiological considerations of age and sex that adult T-cell leukaemia is most likely. The advanced stage of this condition resulted in extensive osteoporosis and collapse of the upper thoracic vertebrae, and it is likely that this individual survived with this debilitating condition for some time. Yet, his burial within an elaborate cliff side tomb with other individuals with evidence of advanced kyphosis due to infectious disease suggests that the expression of his disease either did not affect his social standing or perhaps enhanced it. Thus, rare diseases were recognized in this ancient society.

1: University of Central Florida, College of Sciences
2: University of Central Florida, College of Medicine
Corresponding author: M. Toyne, J.Marla.Toyne@ucf.edu

SKELETAL MANIFESTATIONS OF LANGERHANS CELL HISTIOCYTOSIS ON A 1964 DOCUMENTED CASE

M. Voulgari[1], M. Piagkou[2], K. Moraitis[1]

The reported manifestation of Langerhans cell histiocytosis (LCH) in skeletal remains from both bioarchaeological and forensic anthropological contexts is singular. LCH is a condition that could according to researchers show similarities with metabolic diseases, inflammations as well as malignant neoplasms. It is maybe a disease that could not be classified by morphology. Formerly known under a variety of different names such as histiocytosis X, eosinophilic granuloma, Hand-Schüller-Christian disease or Letterer-Siwe disease, LCH is a rare disease of the family of histiocytosis characterized by the accumulation of histiocytic cells in various tissues.
The objective of this study is to report a case of LCH in the remains of a 3-year-old girl, who, according to the available medical records, was suffering from a disease appointed as Hand-Schüller-Christian. The lesions were examined both macroscopically and radiographically.
A detailed description of the lesions as well as a differential diagnosis were undertaken in order to provide a clearer idea about the disease and how it could be identified in skeletal material. The lesions were large showing beveled edges and found predominantly in the skull, pelvis, femur, vertebrae, and ribs while the metacarpals and metatarsals were spared. Little or no bone formation was found. The X-rays showed osteolytic lesions with beveled edges and multilocular lesions with scalloped borders as well as lesions with a hole-within-hole appearance.

1: Dept. of Forensic Medicine and Toxicology, School of Medicine, National and Kapodistrian University of Athens (Greece)
2: Dept. of Anatomy, School of Medicine, National and Kapodistrian University of Athens (Greece)
Corresponding author: M. Voulgari, myrsinivoulg@gmail.com

EVIDENCE FOR RARE DISEASES IN ANCIENT EGYPTIAN MUMMIES AND SKELETONS

A. Zink [1], E. Hower-Tillmann [2], S. Lösch [2]

For more than 20 years the Institute for Mummies and the Iceman in Bolzano, Italy and the Department of Anthropology in Bern, Switzerland have been involved in the study of mummies and skeletal findings at various archaeological sites in Egypt. The excavations that are headed and coordinated by the German Archaeological Institute in Cairo included the important necropolis of Abydos,Thebes-West, Dashur and Buto and ranging from the Pre and Early Dynastic Period to the New and Middle Kingdom until the Roman and Coptic times. Up to now, data have been collected from approximately 1900 mummies and skeletons, which are covering about 4000 years of the ancient Egyptian history. The aim of this ongoing project is to perform a comparative analysis of the various sites and time periods in order to obtain profound insights into the health and living conditions of the old Egyptian populations. The study includes a detailed analysis of pathological conditions, such as dental diseases, trauma, degenerative disease, signs of malnutrition and infectious diseases. Moreover, rare pathological conditions were detected in a few skeletal and mummified remains from different sites and time periods. These comprised the presence of cranial dysostosis, achondroplasia and cerebral palsy. In addition, in a radiological study of the mummy of king Tutankhamun an aseptic bone necrosis (Kohler disease) was observed in his left foot. The different findings of rare diseases in the ancient Egyptian human remains will be presented and the implications regarding diagnosis and the social and cultural impact will be discussed.

1: EURAC Research, Institute for Mummy Studies, Bolzano (Italy)
2: Department of Physical Anthropology, Institute of Forensic Medicine, University of Bern (Switzerland)
Corresponding author: A. Zink, Albert.Zink@eurac.edu

ACHONDROPLASIA IN THE ANCIENT WORLD

J. Bąk [1], M. Popowska [1], S. Sołowczuk [1]

The approach to dwarves in the ancient world varied. On one hand, they played a special part - treated as supernatural beings, who were messengers of the gods. On the other hand, their illness was perceived as ugliness, for which there was no place in the contemporary canon of beauty. What's more, they were often understood as "living toys" to cheer up others with their disabilities. In this work, the presentation of dwarfs in the art of selected civilisations of the ancient world will be thoroughly reviewed. Particularly noteworthy are the monuments belonging to the Egyptian, Roman, Greek and Mayan culture. We can find the image of dwarfs on numerous artefacts, including sculptures or ceramic vessels, where their anatomical features were presented with exceptional accuracy. How can one explain the role of dwarves in contemporary societies in the light of what we know from archaeological sources? It turns out that an anthropologist's help is necessary because bone material provides valuable information about the health status of individuals and their socio-economic status. Dwarfism is a manifestation of achondroplasia - a genetically conditioned disease caused by mutations of the gene found on chromosome 4. In the bone material, the disease is manifested in the characteristic symptoms of shortening the stems of the bone roots, thus disturbing the proportion of the entire skeleton. In addition to anthropological analyses, the study of the short stature in humans gives a large field for analysis using molecular biology techniques. Undoubtedly, this is an interesting topic that is not given much attention. The above work will be a preliminary outline of new directions of research and its aim is to trigger a broader discussion of the issues raised.

1: Wroclaw University of Environmental and Life Sciences
Corresponding author: J. Bąk, judytabak@onet.eu

KLIPPEL-FEIL SYNDROME, DESCRIPTION OF THE RARE DISEASE BY THE EXAMPLE OF THE CASE REPORT OF THE SKELETON FROM WĄGROWIEC (POLAND, 14TH–17TH CENTURIES)

B. Drupka [1], J. Wysocka [1], W. Nowaczewska [1]

Klippel-Feil syndrome is a rare genetic disorder that can be diagnosed by the presence of two or more non-segmented cervical vertebrae with an occurrence estimated at one on 40,000 to 50,000 live births. In clinical manifestations, KFS demonstrates significant diversity, in which fusion of C2 with C3 and C5 with C6 vertebrae is most frequently observed. The clinical studies present a wide range of reports concerning the occurrence of this syndrome. Nevertheless, relatively few cases have been presented in the literature about archaeological material, especially from Poland.
Klippel-Feil syndrome was diagnosed on the skeleton of an individual from the cemetery adjacent to the Church of St Jacob the Apostle in Wągrowiec, in north-western Poland. The grave number 25 was excavated within an archaeological layer dated to the 14th–17th centuries. The presented case describes the co-occurrence of a rare type of fusion of two cervical vertebrae (C6 with C7) with a supernumerary, well-developed rib, fused to the shaft of the first thoracic rib and articulated via synovial joints with the seventh cervical vertebra. A cervical rib is also an uncommon abnormality with the range of incidence, depending on the considered population, established at 0.05% to 3.0%. The presented poster is based on the case described in the article published in International Journal of Osteoarcheology by authors of the abstract in 2018 (doi: 10.1002/oa.2726).

1: University of Wroclaw
Corresponding author: B. Drupka drupka.beata@gmail.com

A PROBABLE CASE OF KLIPPEL-FEIL SYNDROME IN A NEOLITHIC SKELETON FROM APULIA: RADIOLOGICAL AND HISTOLOGICAL ANALYSIS WITH CONFOCAL LASER MICROSCOPY

M. Favia[1], D. Ferorelli[1]

Klippel-Feil syndrome (KFS) is a vertebral genetical-based developmental pathology with segmental fusion of two or more vertebrae (more frequently cervical) into a block with a single spinous process, neural arch and vertebral body. In the Neolithic archeological site of Cala Colombo, Torre a Mare (Bari, Italy) multiple skeletal remains have been recovered during archaeological excavation, and the following osteological analysis showed a case with a vertebral block consisting in fusion of two cervical vertebrae (probably C4-C5). This specimen has been studied with standard X-rays and a 3D model was obtained by using TC scan. The fusion involves the inferior face of the upper vertebra with the superior face of the lower one, the pedicles and the articular apophyses. The anterior face of the vertebrae showed an osteophyte formation. Bone biopsies of the vertebral fragments were obtained and serial histological sections were observed with a Nikon Eclipse confocal laser microscopy, with double laser scanning with green and red wave length. The fusion zone showed a cartilage layer with a laminar structure in direct contact with the bone tissue superiorly and inferiorly. Sporadic presence of chondroid tissue in the surrounding bone tissue was also observed. These features allow us to evaluate a pathogenesis of alteration during differentiation and maturation of chondroid tissues, with atypical development of intervertebral disc. The morphological, radiological and histological features suggest that the most likely diagnosis is type II KFS.

1: Interdisciplinary Department of Medicine, University of Bari (Italy)
Corresponding author: M. Favia, matteofavia@me.com

A POSSIBLE CASE OF CROUZON SYNDROME FROM MODERN AGE SIENA (ITALY)

V. Giuffra [1], F. Sbrana [2], D. Caramella [3], D. Giustini [3], B. Tixier [2], G. Fornaciari [1]

The skeleton of a female adult found in archaeological excavations carried out in Siena (central Italy) and dated back to the Modern Age showed a severe skull malformation. The deformity was due to premature bilateral closure of the coronal suture, left squamous suture and bilateral occipitomastoid suture; the synostosis caused a turribrachycephalic shape, characterised by reduction of the antero-posterior diameter, increase in the biparietal diameter and high and flat frontal bone. Radiological examination evidenced increased digital markings in the endocranial surface of the frontal bone, demonstrating probable brain compression. The craniosynostosis resulted to be associated with other malformations, such as shallow orbits, likely to have determined ocular proptosis, hypertelorism, slight underdevelopment and flattening of the facial bones, mandibular prognathism and consequent malocclusion, but there was absence of anomalies in the remaining hand and foot bones. These features appeared to be related not to an isolated condition, but to a more complex genetic syndrome, suggesting a possible case of Crouzon syndrome, a genetic disorder caused by mutations in the *FGFR2* gene, mapped to chromosome locus 10q25-10q26. Nowadays, craniofacial disorders associated with craniosynostosis syndromes are surgically corrected in the infantile age. Therefore, the interest of this case consists not only in its rarity in paleopathology, but also in the opportunity of observing the lesions typical of this congenital disorder in an adult subject.

1: Division of Paleopathology, Dept. of Translational Research and New Technologies in Medicine and Surgery, University of Pisa (Italy)
2: Department of History and Cultural Heritage, University of Siena, Italy (Italy)
3: Division of Diagnostic and Interventional Radiology, Department of Translational Research and New Technologies in Medicine and Surgery, University of Pisa, (Italy)
Corresponding author: V. Giuffra, valentina.giuffra@unipi.it

RECONSIDERING OSTEOPETROSIS: A CASE FROM NEOLITHIC ALBANIA

J. Gresky [1], E. Petiti [1],

Osteopetrosis, a heritable disorder, is rare in recent times and even scarcer is its presence in the paleopathological record.
We present here one of two oldest cases of osteopetrosis (as for the other case, refer to Favia *et al.*, p. 8 in this volume) of a skeleton from 4600 BC from Albania, proving the presence of this disorder even in Neolithic times. Only both humeri, the left radius and left femur of a young adult individual of unknown sex were present for examination. Plain radiography, computed tomography, digital microscopy, as well as plain and polarizing microscopy aided in diagnosis. All bones show a marked sclerosis with obliteration of the medullary cavity, particularly in the metaphyses and in the middle of the diaphyses. Radiodense areas, bone-within-bone, are found in the epi- and metaphyses of all bones, bands of alternating lucency are also present in all bones. Two very well healed fractures of both the proximal shaft of the right humerus and the distal shaft of the left radius are present. Furthermore, an "Erlenmeyer flask" deformity of the distal metaphysis of the left femur can be observed.
The adult age of the individual, the specific radiographic signs, the fractures and the "Erlenmeyer flask" deformity of the left femur point to the diagnosis of an autosomal dominant osteopetrosis type II (ADO II). Its occurrence within the period of giant-settlements might have been provoked by interactions between genetic predispositions and changed external environmental factors.

1: Department of Natural Sciences, DAI Berlin (Germany)
Corresponding author: J. Gresky, julia.gresky@dainst.de

NEOLITHIC INDIVIDUAL WITH POSSIBLE KLIPPEL-FEIL SYNDROME AND HIS PLACE IN COMMUNITY

Z. Hukeľová [1], M. Krošláková [1]

In this paper, a possible case of Klippel-Feil syndrome (KFS) from Vráble, Slovakia, is presented and discussed as regards the individual's life within community. KFS is a rare congenital disease, most frequently manifesting as synostosis of the cervical spine. In a Neolithic male from Slovakia aged 15 - 25, fusion of two cervical vertebrae was recorded. Synthesis of only a few vertebral segments is typical for KFS type II, usually associated with autosomal dominant inheritance. In addition to the unification of vertebrae, a notochord defect (linear cleft) on the lower thoracic vertebra was visible. Organ malformations and other health problems often accompany KFS, potentially affecting individual's life within a community. Despite that the bones of the man with possible KFS were moved after death, probably by small animals, and the position of this individual could not be firmly estimated, no skull has been associated with the skeleton. Two more individuals at the site were interred without their skulls, moreover in positions different from the rest of the Vráble skeletons. Moreover, both these skeletons manifest congenital defects of the spine (spina bifida, spondylolysis, and butterfly vertebra). All considered, different treatment of individuals with anomalies and/or of abnormal health/appearance is suggested for this Neolithic community.

1: Institute of Archaeology (Slovak Academy of Sciences)
Corresponding author: Z. Hukeľová, zuzana.hukelova@savba.sk

A CASE OF ACHONDROPLASIA? THE CHILD FROM DIEPENSEE

B. Jungklaus [1], L. Zamstein [1]

During excavations that took place 2005 - 2007 a disturbed burial was found in a village from the 13th-15th century AD located South of Berlin. The lower extremities and a displaced right humerus of a child (burial 372) were retrieved. The long bones appeared strikingly short and compact. Further, they showed some distinctive changes that are typical for those observed in skeletal dysplasias like achondroplasia. Caused by a genetic mutation resulting in a defective endochondral ossification, achondroplasia mainly affects the limbs, the skull, vertebrae as well as vertebral bodies. The skull was missing as were most of the postcranial bones. The pathological changes of the long bones suggest that the child suffered from achondroplasia, the most common skeletal dysplasia. But given the early death of the child and the lack of most of the bones other dysplasias have to be taken into consideration. Archaeological cases of skeletal dysplasias are rare and so far, there are no known cases of archaeological human remains from Brandenburg exhibiting dwarfism. This is the first case of a child with possible achondroplasia from Brandenburg.

1: Anthropologie-Büro Jungklaus
Corresponding author: B. Jungklaus, B.Jungklaus@t-online.de

TWO CASE STUDIES OF RARE GENETIC DISORDERS IN CHILDREN OF MEDIEVAL AND EARLY MODERN KIEV

A. Kozak [1]

80 non-adult skeletons from regular burials and mass graves of 10th-13th century and 110 skeletons of subadult individuals from the monastic cemetery and town necropolis of 16th-18th century of Kiev, Ukraine, were examined for morphology and diseases by macroscopic methods. In the mass grave dated to 1240 AD, the remains of 9 adults and 8 children, from new-born to 13 years old, were unearthed. The skull of a 2 to 4 years old child showed signs of craniosynostosis. The sagittal and the central part of the coronal sutures were completely fused. On this skull the traces of metabolic disorders, possibly severe anaemia and scurvy, otitis media and some meningeal reactions have been observed. The two main differential diagnoses discussed here are: genetic disorders and those diseases which lead to a premature fusion of the cranial sutures, due to bone remodelling processes on the skull vault.
In the grave 92 on the cemetery of Desjatinna Church dated to the 16th - 18th century, a stillborn child at 30-32 gestational weeks with bilateral radio-ulnar synostosis has been discovered. Both, radius and ulna, were fused at their proximal ends. Congenital disorders and the consequences of foetal trauma are discussed as differential diagnoses.

1: Institute of Archaeology Kiev
Corresponding author: A. Kozak, akozak26@ukr.net

BILATERAL DYSMELIA OF HUMERUS, ULNA AND RADIUS AND UNILATERAL HYPOPLASIA OF THE MANDIBULA IN AN EARLY MEDIEVAL SKELETON

C. Meyer [1], K. W. Alt [2]

We describe a case of a multi-focal skeletal dysplasia in an adult male from an Early Medieval cemetery from Mannheim, Germany. The left side of the mandible is dysplastic, the condyle sits rather low on the mandibular ramus and appears stunted, the mandibular body is posteriorly waisted. Although the right side of the mandible is not completely preserved it appears to have a normal morphology. The overall appearance of the lower jaw is clearly asymmetric.
Further dysplasia (dysmelia) can be found in the bones forming the elbow joints. The distal parts of both humeri and the proximal parts of both ulnae and radii appear malformed. Although the bone surfaces show postmortem taphonomic damage the abnormal joint morphology is clearly evident and suggestive of an absence of the respective epiphyses. Both humeral diaphyses appear rather slender for a male individual of this series suggesting at least a partial atrophy of the upper limbs. Unfortunately, most of the bones of the hands were not recovered but a few phalanges are present. These appear normal, like the rest of the preserved skeleton. As there are at least three dysplastic regions in this skeleton and the malformations of the elbow joints are bilateral and symmetric, a congenital condition of a skeletal dysplasia currently appears as the most reasonable explanation. Other pathological conditions fail to explain all recognized changes. So far, the dysplasia of this individual could not be matched to a previously described syndrome from the medical literature.

1: OsteoARC – OsteoArchaeological Research Centre (Germany)
2: Centre of Natural and Cultural Human History, Danube Private University (Austria)
Corresponding author: C. Meyer, chr.meyer@email.de

DYSPLASIA OF THE LOWER ARM IN A FEMALE FROM THE MEROVINGIAN PERIOD IN CENTRAL GERMANY

J. Nováček[1 2]

During a rescue excavation in Gotha-Boilstädt (Central Germany), a Merovingian burial ground from the 6th and 7th century CE was found. Among 53 individuals from this burial ground, one individual, a female with an age-at-death of 40-50 years, showed an unusual case of dysmelia of the right arm. The shoulder (clavicle and shoulder blade) and upper arm were shortened and displayed weak muscle attachment marks and a lower robustness compared to the left side. The main difference, however, was observed in the bones of the lower arm. The ulna was highly shortened and distorted; the radius was deformed almost beyond recognition. Unfortunately, no hand bones were preserved, but it seems likely, that the hand was also highly deformed, judging from the morphology of the wrist joint facet of the radius. Probably, the weak muscle attachment marks of the upper arm and shoulder resulted from an inactivity atrophy due to the deformation of the lower arm (and possibly hand). Therefore, a diagnosis of erb-duchenne palsy (obstetric brachial plexus palsy) does not seem plausible. As the deformity was most pronounced in the hand and wrist, poliomyelitis was not the presumable reason, but it cannot be excluded as a differential diagnosis. The most plausible differential diagnosis, however, seems to be Madelung's deformity, a congenital malformation of the wrist, occurring in about 1-3% of all hand and wrist deformities. The differential diagnoses will be discussed.

1: Thüringisches Landesamt für Denkmalpflege und Archäologie Weimar
2: Institut für Anatomie und Zellbiologie, Universitätsmedizin Göttingen
Corresponding author: J. Nováček, jan.novacek@tlda.thueringen.de

HEREDITARY HAEMORRHAGIC TELANGIECTASIA (HHT) – A RARE GENETIC DISORDER AS A POSSIBLE CAUSE OF CHRONIC IRON DEFICIENCY ANAEMIA

J. Nováček[1 2], K. Scheelen-Nováček [3 4], I. Chao [2], M. Schultz [3]

In the palaeopathological record, except for haemoglobinopathies (e.g. thalassemia, sickle-cell disease), genetic defects are not commonly considered as possible causes of anaemia. In our contribution, we focus on one of these commonly unknown, but possible causes. Hereditary haemorrhagic telangiectasia (HHT), also known as Osler-Weber-Rendu disease, is a genetic disorder which occurs in one in 5000 people. It is transmitted through an autosomal dominant inheritance pattern and leads to abnormal blood vessel formation. Arteriovenous malformations commonly affect larger organs, such as the lungs, the liver, and the brain. Telangiectasias characteristically occur in the skin and mucous membranes. Especially lesions in the gastrointestinal tract may bleed periodically, which usually remains undetected, but results in chronic iron deficiency anaemia in about 20% of the patients. With regard to this frequency, also in an archaeological context, HHT can be considered as a possible reason for iron deficiency anaemia. In a female body donor (age-at-death 68 years) who reportedly suffered from HHT, coupled with severe anaemia, possible skeletal changes commonly related to chronic anaemia were observed and documented. The characteristic filling of long bone diaphyses with spongy bone was frequently present and pronounced. On the other hand, the surprisingly thin skull bones hardly showed any vestiges of possible (healed) external cranial cribra. The individual had no orbital cribra, quite the contrary, the orbital roofs were extremely thin and translucent. The results will be presented and discussed.

1: Thüringisches Landesamt für Denkmalpflege und Archäologie Weimar
2: Institut für Anatomie und Zellbiologie, Universitätsmedizin Göttingen
3: Institut für Anatomie und Embryologie, Universitätsmedizin Göttingen
4: Institut für Biologie und Chemie, Universität Hildesheim
Corresponding author: J. Nováček, jan.novacek@tlda.thueringen.de

A RARE CASE OF ANGELMAN SYNDROME FROM THE 17TH CENTURY CEMETERY OF RAVENNA (ITALY)

A. Pasini[1], N. Rinaldo[1], B. Bramanti[1 2]

Archaeological excavations at the cemetery of the St. Biagio Church in Ravenna (Emilia Romagna, Italy) led to the discovery of several hundred graves dated from the 17th to the 19th century. Among the numerous skeletal remains, which are currently under analysis at the Laboratory of Archaeo-anthropology and Forensic Anthropology of the Ferrara University, some uncommon cases are sometimes recovered and offer the opportunity to analyse the presence of rare diseases during the past.
Skeletal remains from Individual 207 belonged to an adult male presenting a particular set of disorders, which led us into hypothesising a shared aetiology more than a casual appearance of several unrelated features. Cranial traits included reduced and disproportioned size of the neurocranium and marked occipital flattening, strong maxillary hypoplasia and diffused oral disorders such as an almost complete ante-mortem tooth loss and severe mandibular ablution and prominence, auditory exostosis and osteophytic reactions on the occipital condyles; post-cranial anomalies included ossification defects, asymmetry of the lower limbs and features related to postural defects. All these characteristics seem to be compatible with a diagnosis of Angelman syndrome (i.e. “Happy Puppet” Syndrome), a rare genetic disease occurring in about 1 case over 12/20.000 and presenting typical traits such as severe developmental delay and malformations of the skull and facial bones (including microcephaly and disproportioned growth of the skull, posterior flattening of the head, maxillary hypoplasia and prominent mandible, frequent oral disorders connected to excessive chewing behaviours), developmental delay, and movement or balance disorders. Individual affected by this syndrome often reach adult life. This unique finding could help in a better understanding of rare diseases in the past.

1: University of Ferrara
2: University of Oslo
Corresponding author: A. Pasini, alba.pasini@unife.it

SYNDROMIC BILATERAL LAMBDOID AND SAGITTAL SYNOSTOSIS (MERCEDEZ BENZ PATTERN CRANIOSYNOSTOSIS) IN A MODERN-AGE SKELETON FROM RAVENNA, ITALY: A RARE CASE OF CROUZON SYNDROME?

N. Rinaldo[1], A. Pasini[1], F. Sciané[1], E. Gualdi Russo[1]

Bilateral lambdoid and sagittal synostosis, also known as Mercedez Benz pattern craniosynostosis, is a complex and extremely rare disorder characterized by the premature fusion of the lamboid suture and the posterior part of the sagittal suture. This particular pattern of craniosynostosis was found in a subadult, aged about 10-15 years, discovered during the archaeological excavations carried out in the cemetery of San Biagio Church (Ravenna, Northern Italy) and dated back between the 17th and the 19th century. The juvenile subject presented a characteristic head malformation with a posterior plagiocephaly associated with turricephaly and mild brachycephaly. Moreover, besides the craniosynostosis, other skeletal malformations or anatomical anomalies were found, such as mid-face hypoplasia and crowding of teeth with consequent malocclusion, torus palatinus, spondylolysis with vertebral deformities and bifid rib. All these features seem to be related to a more complex genetic syndrome and suggest a probable case of Crouzon syndrome, that might be associated with a Chiari I malformation, that commonly occurs in case of lambdoid craniosynostosis. This diagnosis is supported by the fact that the malformation did not seem to have affected the bones of the extremities. This finding represents a unique case in the archaeological context and this case report provides the opportunity to detect, even in the past, the presence of a rare disturbance that is nowadays surgically corrected during the first years of life.

1: University of Ferrara
Corresponding author: N. Rinaldo, natascia.rinaldo@unife.it

GENETIC DETECTION OF ACHONDROPLASIA IN HISTORICAL SKELETAL MATERIAL

M. Rusinko [1], K. Brzobohatá [1], E. Drozdová [1], D. Fialová [1]

Achondroplasia is an inherited bone disorder that occurs in one in 20,000 to 30,000 live recent births. It is the most prevalent form of genetically determined human dwarfism. People suffering from achondroplasia are characterized by disproportionately short proximal segments of limbs and distinctive craniofacial appearance. 95% of cases is caused by a point mutation p.Gly380Arg in the FGFR3 gene which regulates enchondral ossification of long bones (FGFR3 codes for fibroblast growth factor receptor). The collection of small skeletons is normally studied by traditional anthropometric and morphological approach. Low preservation status of osteological material and chemical inhibitors often prevent such samples from being confirmed genetically. To this day, there is no universal method for genetic diagnosis of achondroplasia from skeletal remains. The aim of the study was to establish a laboratory protocol suitable for genetic confirmation of achondroplasia in suspected bones. More methods of DNA isolation and amplification were adopted prior to Sanger sequencing of the critical locus. The isolation method with magnetic particles (PrepFiler™ BTA Forensic DNA Extraction Kit) and KAPA2G Robust PCR Kit proved to be efficient. The individual from 18th-19th century suspected of having achondroplasia was confirmed to be negative. This opens the discussion, what other health conditions could led to the specific phenotype. The established protocol can be used for genetic testing of further individuals from museum collections, which were, up to now, studied only by anthropological methods.

1: Department of Experimental Biology, Faculty of Science, Masaryk University, Brno (Czech Republic)
Corresponding author: M. Rusinko, rusinko.martin.94@gmail.com

UNUSUAL CONGENITAL PATHOLOGICAL DISORDER FROM THE CHURCH EL SALVADOR (XVI–XVIII CENTURY, TOLEDO, SPAIN)

N. Sarkic[1], J. Herrerin, R. Dinares

Individual SALV-009, excavated at the necropolis of El Salvador (Toledo, Spain), was found incomplete and with obvious signs of pathology. Namely, the right leg was noticeably shorter than the left one. The individual was determined to be female, with an estimated age of death between 25 and 39 years old. Apart from the leg, her right forearm was also a bit shorter and curved, and her stature was short (145±1.67 cm). Osteoarthritis on right femur, tibia and patella, as well as Charles' facets and very marked linea aspera of the femur, are strong proofs that she was using the right leg in everyday activities, despite the deformation. After detailed morphological investigation, scan images and X-ray analyses, we discarded traumatic or infectious (poliomyelitis) origin of the pathology, and estimated aetiology as congenital.
Among the diseases that can lead to such an important congenital shortening of a limb we determinate Congenital Short Femur (CSF) as the more plausible diagnosis. CSF is a rare and complex congenital deformity, in which a patient has a smaller femur, sometimes accompanied by serious alterations and even in the absence in the iliofemoral joint.

1: Independent
Corresponding author: N. Sarkic, nsarkic@gmail.com

COULD SOME OSSEOUS LESIONS BE PREDICTIVE OF GENETIC ANEMIA? DISCUSSION ABOUT SKELETAL MARKERS OF BETA THALASSEMIA

F. Scianò[1], M. Schultz, E. Pacciani, E. Gualdi-Russo, B. Bramanti

The study of thalassemia syndrome in archaeological human remains is of growing interest in the field of paleopathology. However, there is still uncertainty in the diagnosis of this disease in human skeletal remains. Depending on the globin strain damaged, the thalassemic syndromes are classified as α-thalassemia or β-thalassemia. Each of these two categories includes different forms of the disease depending on the homozygous, heterozygous or compound heterozygous status of the person affected (Galanello and Origa, 2010; Perisano et al., 2012).
The bone lesions found on ancient human remains are difficult and uncertain to interpret. In archaeological samples, several non-specific lesions were mostly suggested as probable evidences of β-thalassemia syndrome in sub-adults affected and adult carriers. These not-specific skeletal lesions suggested the skull as the skeletal region with the larger number of evidences predictive of thalassemic syndrome. Some postcranial lesions were also proposed as important factors for the identification of β-thalassemia carriers.
This preliminary work aims to clarify which skeletal lesions are “diagnostic” and which only “indicative/not-diagnostic” of β-thalassemia syndrome. The goal is to provide bio-archaeologist and paleo-pathologists with new knowledge gained from the observation of clinical cases, and radiological and histologic analysis. This information may help in identifying with more confidence putative β-thalassemia carriers and select samples to submit to further genetic confirmation.

1: University of Ferrara
Corresponding author: F. Scianò, scnfpp@unife.it

DEVELOPMENT ANOMALIES OF THE VERTEBRAL COLUMN IN PORTUGUESE PREHISTORIC SAMPLES

A. M. Silva [1], S. Tereso [1]

The spine can provide a large amount of information about an individual's physical condition, lifestyle, epidemiology and familial links of birth defects. The aim of this research was the analysis of developmental anomalies of the vertebral column of Portuguese prehistoric skeletal collections. Thus far, the analysed samples included more than 200 individuals. Still, one of the main limitation of this study is the nature of the samples, recovered from collective tombs, were the bones were recovered very fragmented, disturbed with few or complete absence of anatomical connection. Despite these limitations, until now, the observed anomalies included atlas *bipartite*, clefting of the posterior arch of the atlas and several block vertebra involving cervical and thoracic vertebrae, including a possible case of Klippel-Feil syndrome.

1: Laboratory of Prehistory, CIAS, University of Coimbra
Corresponding author: A. M. Silva, amgsilva@antrop.uc.pt

BILATERAL PROTRUSIO ACETABULI IN A MEDIEVAL SKELETON FROM TRANSYLVANIA: A CASE FOR MARFAN SYNDROME

K. L. Filipek [1], K. Tucker [1], I. Stanciu [1], M. Oliverson [1], M. Crowther [1], J. Snyder [1]

The Jucu de Sus Necropolis is a multi-period cemetery site located in the Transylvanian region of modern-day Romania. The site is associated with diachronic settlements from the Roman period through the early Medieval Period (4th - 12th centuries AD), yielding hundreds, possibly thousands, of burials. Previous sample excavations undertaken in 2007 by archaeologists from the Institute of Archaeology and Art History (Cluj-Napoca) yielded numerous burials from the early medieval period (n=80), and with Transylvania Bioarchaeology, resumed excavations there in 2014 as part of a bioarchaeology training program. This case study examines the remains of Burial 26, an adult female (aged 25-45) from the medieval period (radiocarbon dates TBC) with a rare presentation of bilateral protrusio acetabuli. The acetabular bone changes coupled with skeletal hypergracility and increased stature indicate that the bilateral protrusio acetabuli is most likely associated with Marfan Syndrome, a genetic disorder of connective tissue that affects approximately 1:10,000 (Lundby et al. 2011). The degree of pelvic intrusion shows femoral neck impingement, which would have caused substantial joint restriction. Previous to medical intervention, people with Marfan syndrome had significantly shortened lifespans, not living much past their teens (Keane and Pyeritz 2008). Burial 26's adult age indicates a system of a support in place from a time period that isn't often associated with palliative care and longevity in the face of disease. Further, this is the first instance of Marfan Syndrome, and its associated skeletal changes, appearing in the archaeological record.

1: Transylvania Bioarchaeology
Corresponding author: K. Tucker, Katie.Tucker@winchester.ac.uk

SYNDROMIC CRANIOSYNOSTOSIS AND FIBROBLAST GROWTH FACTOR RECEPTORS (FGFRS) MUTATIONS: TOWARDS DIFFERENTIAL DIAGNOSIS IN PAST POPULATIONS THROUGH SKELETAL AND GENETIC ALTERATIONS.

S. Zdral[1 2], María José Trujillo-Tiebas[1 2]

Craniosynostosis is the premature fusion of one, several or all calvarial sutures, with a consequential alteration in cranial volume and shape. When archaeological populations are studied, it is not uncommon to find cases of premature craniosynostosis and unintentional alterations of the cranial morphology. Regarding its classification, there are craniosynostoses produced by environmental or genetic causes, and within the latter exist the syndromic craniosynostosis (approximately 15%). Syndromic craniosynostosis involvement goes beyond cranial alterations, including facial dysmorphisms, syndactyly and other modifications in the phalanges, fusion of several vertebrae or dental malocclusion. Retardation in growth and/or alterations in skeletal maturation have also been described. The most common are the Apert (MIM 101200), Pfeiffer (MIM 101600), Crouzon (MIM 123500) and Muenke Syndrome (MIM 602849), caused principally by alterations in the FGFR2 and FGR3 genes, with an autosomal dominant inheritance. These syndromes differ in their frequency but they are all classified as rare diseases. The aims of this study are two: 1) To describe the skeletal alterations that occur in each one of the syndromic craniosynostoses produced by mutations in the FGFR2 and FGFR3 genes making use of the available clinical information; and 2) To perform a differential diagnosis as updated and accurate as possible between isolated and syndromic craniosynostosis as well as between genetic and environmental craniosynostoses. With all this, it is intended to guide the researchers in the study of archaeological individuals who present these skull alterations.

1: Universidad Autónoma de Madrid
2: Fundación Jiménez Díaz, Madrid
Corresponding author: S. Zdral, sofia_zn22@hotmail.com

SPOTTED BONES IN BUSCHKE-OLLENDORF SYNDROME: A RARE AND BENIGN DISEASE WITH CLINICAL SIGNIFICANCE

S. Zdral[1 2], I. Lorda [1 2], M. J. Trujillo-Tiebas[1 2]

The Buschke Ollendorf Syndrome (BOS) is a rare, benign and asymptomatic syndrome that consists of the joint appearance of two lesions: osteopoikilosis on the bone and connective tissue nevus on the skin. Focusing on bone alterations, osteopoikilosis consist of multiple sclerotic lesions with circular shape that are observed as radiodense areas through an X-Ray. They are usually found in the epiphyses and metaphyses of long bones and presents bilateral distribution. Their discovery is often accidental when radiographs are taken for different purposes. BOS is caused by heterozygous mutations of the LEMD3 gene, with an autosomal dominant pattern of inheritance and its prevalence is estimated at 1:20,000 individuals, affecting both sexes equally. Bone changes usually occur within the first decade of life. In paleopathology, the published works regarding BOS are practically non-existent. However, in 2003, Ortner was one of the first authors to talk about this radiological sign. The differential diagnosis includes analysis from bone metastases to lesions of taphonomic origin. In this study, a current clinical case is presented, in order to show how to make a more suitable differential diagnosis of this syndrome.

1: Universidad Autónoma de Madrid
2: Fundación Jiménez Díaz, Madrid
Corresponding author: S. Zdral, sofia_zn22@hotmail.com

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rare-disease-workshop@dainst.de

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