The ANthropological Notation Ontology (ANNO): A core ontology for annotating human bones and deriving phenotypes

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Abstract. The Anthropological Notation Ontology (ANNO) allows the systematic and standardized classification of recovered bone finds into the skeletal system, the description of the skeletal pieces, and the definition of functions for the derivation of different phenotypes of humans in forensic and historical anthropology. ANNO consists of two components: ANNOdc, a domain-core ontology providing core entities such as basic anatomical categories, and ANNOds, a domain-specific ontology used for annotating structures of the human skeleton. ANNO is integrated into AnthroWorks3D, a photogrammetry pipeline and application for the creation and analysis of 3D-models of human skeletal remains. The integration is based on the three-ontology method with the General Formal Ontology as the top-level ontology, ANNOdc as the task ontology and ANNOds as the domain ontology. Thus, AnthroWorks3D only needs to implement access to the entities (classes and properties) of the task ontology, whereas the entities of the corresponding domain ontology are imported dynamically. ANNO supports the analysis of skeletal and bone finds in forensic and historical anthropology, facilitating the standardization of data annotation and ensuring accurate preservation of information for posterity.

Keywords: Ontology, Ontology development, Anthropology

1. Introduction

Data from historical and prehistoric anthropology [1], as well as modern forensic science, provides a comprehensive understanding of deceased individuals from different time periods. This understanding encompasses their identity, health status, aspects of their behavior, lifestyle, culture and even the circumstances of their death [2]. For example, anthropological methods can be used to determine individual properties (phenotypes) such as their sex, age, height and ancestry and other individualizing traits.

Digitization and digitalization offer several advantages to anthropology, including remote work, without the effects of wear and tear on the physical skeletal specimens. Current problems of access can be resolved, as examinations can be conducted even if the skeletal material from individuals or collectives are not present at the institution or may have already been reburied, which facilitates interdisciplinary collaborative research. Moreover it allows for

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linking data of different study samples across geographical boundaries. Data sets consolidated this way and formalized through the Anthropological Notation Ontology (ANNO) presented here allow for efficient data analysis techniques, such as data and text mining that promote deeper insights.

Apart from contextual information, most informative clues can be found directly on the bone. Anthropologists use human anatomy, specifically the skeletal system and further tissues of the musculoskeletal system, to classify and describe and thus document bones, anatomical structures and diagnostic features within the overall framework of the skeleton in a comprehensive and transparent fashion.

By applying the same principles used for mapping the Earth's surface "terrain", a detailed map of the human body can be created. This map contains visible anatomical surface structures, as well as artificial objects such as measurement points or content-related or methodologically based classifications and boundaries [3]. ANNO represents such a map by providing an ontology for accurate and exhaustive definitions that allow to unequivocally locate these, facilitate their retrieval and furthermore serve as a basis for the objective examination, including measurements.

ANNO consists of two components: ANNOdc, a domain-core ontology providing core entities such as basic anatomical concepts and classifications, and ANNOds, a domain-specific ontology employed for annotating human skeletal structures. In its current version, the ontology refers to standardized normal adult anatomy, excluding developmental aspects, variations, pathologies and other related factors. ANNO is integrated into AnthroWorks3D (AW3D), a photogrammetry pipeline and application for generating and analyzing 3D-models of human skeletal remains, and published over multiple channels, see table 1.

2. Related Work

There are various reference materials for the description of anatomical structures, ranging from anatomical atlases to nomenclatures. The latter aim for an established standardized naming and systematization. There are currently two prominent ontologies in the field of general anatomy:

2.1. Terminologia anatomica (TA)

The TA [6] is a hierarchy of anatomical structure concepts for the entire human body. For historical reasons, it uses a terminology consisting of Latin and originally Greek, which was later latinized. For each anatomical structure, the TA provides the "preferred", i.e. standard, Latin term and its English equivalent as well as an individual identification number. In some cases, Latin and English synonyms are also included, e.g. *malar bone* is an English synonym of the Latin preferred term *Os zygomaticum* and its English equivalent *zygomatic bone*. The identification number is crucial, as certain landmarks are mostly listed without bone affiliation, resulting in multiple occurrences. For instance, the *processus zygomaticus* can be found both at the *os frontale* and the *os temporale*. Eponyms are terms derived from proper nouns such as the name of a person, place or thing, and they are considered a type of synonyms since they stand for the same anatomical structure. For example, *ossa digitorum manus / pedis* (engl. *phalanges of the hand / foot*) are the small bones of the fingers and toes and are also denoted as *phalanges manus / pedis* which are named after the Greek word *phalanx* for a dense rectangular infantry formation. In addition to the outdated print

Table 1
Structure, properties and publication of ANNO.

ANNOdc (domain-core) , ANNOds (domain-specific)			
General Formal Ontology [4]			
https://annosaxfdm.de/ontology/			
RickView [5] at https://annosaxfdm.de/ontology/			
https://github.com/annosaxfdm/ontology			
https://ols.imise.uni-leipzig.de/ontologies/anno			
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edition [7], an extended second edition (TA2) is available online [6]. Furthermore, there is a commercially available independent companion print publication with German [8], English [9] and Spanish [10] editions of which at least the German edition is regularly updated. This book contains textual and visual descriptions, whereby anatomical structures are indicated through lines on the illustrations.

2.2. Foundational Model of Anatomy (FMA)

The FMA [11] ontology represents the physical organization of human anatomy by mapping relations to one another. It allows the knowledge it contains to be represented in a way that is humanly comprehensible and machine-interpretable. The FMA uses the Basic Formal Ontology (BFO) as its top-level ontology. Corresponding to its relational nature, new higher level concepts are introduced and used for reorganizing the actual anatomical structures diverging from the TA's hierarchical structure. Thus, in 90 % of the cases this leads to new creations of anatomical concepts [12] whereas only 1 % contain textual definitions [13].

We create manual links between ANNO and the FMA, which we use to connect ANNO with the TA2 by using unofficial links between FMA and TA2 provided by Wikidata.

2.3. Current Research Gaps

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There are several issues and research gaps when it comes to naming and defining anatomical concepts and instances that neither current ontologies nor reference literature—general or subject-specific—can address adequately.

The primary issue lies in the absence of standardization. While both the TA and FMA have been proposed as a means of standardizing terminology, they haven't garnered sufficient acceptance to facilitate the adoption of a unified and consistent body of work to establish standardization in actual practice [14–16]. Instead, the usage of terminology is as fluent as any other language, depending on its socio-cultural environment, such as schools or language areas [14, 15, 17–19]. Thus, the usage of English designations is preferred in the Anglo-American sphere while Latin terms are commonly used in German-speaking areas [8, 20, 21]. As a consequence there are at times various translations for the same anatomical terms [22]. Yet, knowledge of Latin is generally declining. With the terms hence becoming more abstract to the people using them, there is also a common prevalence for spelling divergences or grammar mistakes [8, 16, 23, 24]. Moreover, in textual descriptions Latin and the native language are commonly used interchangeably for stylistic purposes. For instance, [8] uses the German terms of the bones in the explanation of sutures. Meanwhile, the existing terminology is not flexible enough for language-like usage as, for example, there is an inconsistent use of singular and plural forms or gaps in lateralized landmarks.

Anatomical terms, unless historically evolved, typically describe the location, affiliation or function of a concept or structure [21]. However, this naming convention is often inconsistent and non-intuitive. An example, showing the variability in illustrative descriptions are the Tuberculum articulare of the Processus zygomaticus, which in [25] is described as saddle-shaped although it is not called *sella*, the Latin name for saddle. It is moreover worth pointing out that not every anatomical structure that contributes to an articulation in some way automatically carries the term articular with it, such as the *caput mandibulae*, which connects the Mandibula to the rest of the Cranium. The Tuber frontale, which serves as an indicator for sex in anthropology, is listed as an eminentia in the TA [6]. Likewise, the Protuberantia mentale is referred to as Eminentia mentale in a widely-used anthropological method that is also used for sex determination. As can be seen, the incentive for synonyms is high. However, the current resources fail to provide comprehensive lists of synonyms in use. This in turn may lead to misattribution: For example, the FMA entry with the preferred name *External acoustic aperture* and non-English equivalent *Porus acusticus externus* (fma: 61301) lists *external acoustic meatus* (*Meatus acusticus externus* in Latin) as a synonym. However, it is not the same, rather the Porus is—as its English name indicates—the opening to the meatus [8]. Occasionally, there are discrepancies and inconsistencies found in the definitions of individual anatomical structures. For instance, the position of the *Corpus ossis pubis* varies across different sources [8, 26–29].

Another issue is that neither FMA nor TA are tailored to fit the requirements of anthropology or any other specialized field [11]. For example, articular surfaces of the individual bones are relevant anthropologically, yet not all articular surfaces (e.g. on the *Ossa carpi* or *Ossa metatarsi*) are defined in the TA or FMA, resulting in further

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gaps. A similar situation exists with respect to osteometric measurements. Established and to some extent standardized osteometric measurement points are only found on the cranium and mandible [30]. However, since there are measurement distances on all bones, the respective start and end points must also be defined. Last but not least, both ontologies suffer from a lack of consideration given to anatomical variants [20]. Moreover, different disciplines require a different level of detail, which is why nose surgery works with anatomical terms of very detailed anatomical structures [31], which to the greater part are not found in the FMA or the TA that primarily aim for the standardization of general (clinical) anatomy [11]. A work of dental anthropology even split the aforementioned Tuberculum articulare (Articular tubercle) in two structures, naming the other "articular eminence" [32]. However, there is limited interdisciplinary coordination between the individual disciplines and general anatomy.

Furthermore, anatomical resources often lack adequate visual representation of anatomical structures. The labeling is often selective, varies by source and mostly relies on arrows for local designation. However, since they are multi-dimensional structures, it is essential to provide a marker over the entire landmark and delineate it accurately. The latter also requires a clear textual definition. Unfortunately, there are currently no equivalent standardized definitions for skeletal anatomy, meanwhile anatomical resources only provide sparse information on this topic.

To address these issues, it is necessary to develop unified, textual, visual, and detailed descriptive definitions for anatomical concepts and instances. These definitions should be presented within an easily understandable ontology, with ANNO contributing to filling this specific niche.

3. Ontological Architecture

The ontological architecture of ANNO consists of three interrelated layers representing by a top-level ontology, a domain-core ontology and a domain-specific ontology:

- 1. We use the General Formal Ontology (GFO) [4] as top-level ontology of ANNO. We refer specifically to the categories "Material object", "Attributive", "Relator" as well as the spatial entities of GFO, see fig. 1.
- 2. ANNOdc (section 4, fig. 2) is a domain-core (dc) ontology that defines the core entities of the domain. This includes general anatomical categories (Bone and Tooth), a category for describing their characteristics (Anatomical property), anatomical spatial entities (Anatomical space, surface, line and point) as well as the category *Phenotype* to model the rules for determining human phenotypes. ANNOdc is embedded in the GFO, i.e. the ANNOdc classes are subclasses of GFO classes.
- 3. ANNOds (section 5) is a domain-specific (ds) ontology for describing domain-specific entities to be used for annotating the parts of the human skeleton. These are bones, teeth, their parts and compounds, such as *mandible*, *Mental protuberance* or the facial skeleton. It is also used for modeling their properties and relations, such as the distance between *Mentale dexterum* and *Mentale sinistrum*, needed to derive human phenotypes like sex or height. ANNOds is embedded in ANNOdc.

4. Description and Foundation of ANNOdc

The core ontology development occurs in close collaboration between ontologists and anthropologists. The objective is twofold: to adequately represent the most basic anatomical and anthropological entities and to keep the ontology relatively simple and compact. This approach ensures that domain experts can easily handle the ontology and efficiently integrate it into the AnthroWorks3D software, see section 6.

4.1. Terminology for descriptive anatomy

The position of all anatomical entities is described in a standardized fashion using conceptual axes (a kind of anatomical line), planes (a kind of anatomical surface) as well as directions (relative anatomical locations). Directions are relative to the body based on the standard anatomical position (positio anatomica ta2:72, fma:23132). It refers to standing upright, facing forward, palms and toes pointing forward, thumbs to the side, and observed from

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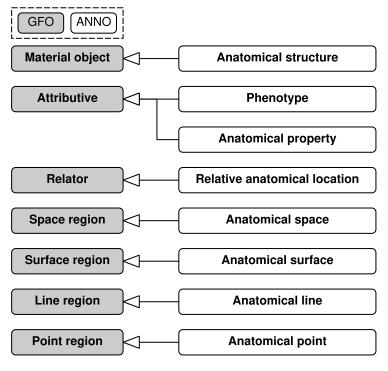


Fig. 1. Integration of ANNO with the top-level GFO ontology.

the perspective of the respective individual. Although an infinite number of axes and planes can be defined within the human body, generally three principal axes and planes are used [26]. They are mutually perpendicular and provide the three spatial coordinates. The three principle axes, shown in fig. 3 are:

- The longitudinal or vertical axis runs in the superior-inferior direction in an upright position, perpendicular to the ground. It intersects with the frontal and sagittal planes.
- The sagittal axis runs in the ventral-dorsal direction, from the front to the back surface of the body and vice versa. It intersects with the sagittal and transverse planes.
- The transverse or horizontal axis extends from left to right, intersecting with the frontal and transverse planes.

The three principal planes, depicted in fig. 3, are:

- The sagittal plane, i.e all vertical planes parallel to the sagittal suture of the skull and running from anterior to posterior in the upright position. The median (sagittal) plane divides the body into two symmetrical halves.
- The frontal plane (= coronal plane) comprising all planes parallel to the forehead (frons) or the coronal suture of the skull, running vertically from one side of the body to the other in the upright position.
- The transverse plane including all horizontal cross-sectional planes, relative to the upright position, dividing the body into cranial and caudal sections. They run perpendicular to the longitudinal axis of the body.

Each one has two anatomical axes as boundaries in the sense of GFO, which allows boundaries to occur inside a structure and not necessarily at the extremities. For example, the coronal (frontal) plane has the transversal (horizontal) and longitudinal (vertical) axes as boundaries.

An anatomical direction or relative anatomical location, as shown in fig. 3, is the position of an anatomical entity (e.g., a bone structure) in relation to another anatomical entity (e.g., another bone, an axis, a plane or a point such as the center of the body). Relative anatomical locations are classified according to the anatomical terms of location (e.g., cranial or superior = "towards the end of the skull", dexter = "right" or distal = "in direction towards the end of a limb"). For example, the Glabella is located laterally to both Arcus superciliaris (Arcus superciliaris sinister und Arcus superciliaris dexter). A relative anatomical location can be considered as an individual relation between two

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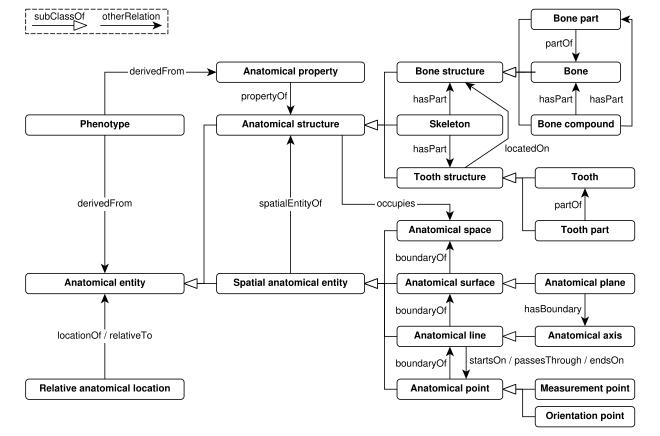


Fig. 2. The ANNOdc domain-core ontology.

anatomical entities (target entity and reference entity) and can therefore be modeled using a GFO relator. Relators are composed of roles (in our case target and reference) and have the power to relate arbitrary entities (in our case, anatomical entities) [35].

4.2. Anatomical entity

An anatomical entity is either an anatomical (material) structure or a spatial anatomical entity, such as Cranium or FrontalPlane.

4.3. Anatomical structure

An anatomical structure refers to any anatomical division or material anatomical entity. ANNO describes three kinds of anatomical structures: tooth structures (teeth and teeth parts), bone structures (single bones, bone parts and bone compounds), as well as the complete skeleton. The GFO defines a material object gfo: MaterialObject as a solid concrete entity that belongs to the material region of the world, has mass, consists of matter and occupies space [36]. Accordingly, we define anno: Anatomical Structure as a gfo: Material Object in the anthropological context. The closest equivalent in the FMA is the Material anatomical entity fma: 67165, the subclass of Physical anatomical entity fma: 61775 that has mass.

4.4. Skeleton

The largest or most comprehensive skeletal anatomical structure, the human skeleton, is the framework composed of all the bones and teeth of a human being. The TA [6] equates the skeleton with the "Systema skeletale (skeletal

system)", which refers to the passive part of the musculoskeletal system, including the articulated bony skeleton with teeth, as well as cartilage, ligaments, and other joints. ANNO does not adopt this equivalence, and "Skeleton" here refers to the entirety of a human's bones and teeth. However, since teeth constitute their own tissue, they are treated separately, allowing all other components of the skeletal system, such as cartilage or ligaments (as tissues) or joints (e.g. as specific aspects), to be integrated and referenced separately in ANNO. The anthropologically meaningful distinction between "Skeleton" and "Systema skeletale" can be maintained, while still referencing both within the TA concept.

4.5. Tooth structure

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Tooth structures comprise teeth and their parts. A human tooth is an individual unit of the human dentition (synonymous for the teeth as a whole). Teeth are part of the skeleton, yet they are characterized by their own distinctive tissue and thus treated separately from bones. Teeth, such as the Dens caninus (canine tooth) are located on bone structures. The FMA has the equivalent class "Tooth" fma: 12516. A tooth part is any portion of a tooth. They are not included in the initial scope of the ontology, but they are one of the aspects by which it can be expanded.

4.6. Bone structure

Bone structures comprise all possible parts of the human skeleton, which are individual bones, bone parts, bone compounds as well as the complete skeleton excluding the teeth. The FMA does not have a common superclass for those parts but instead mounts them in different parts of its hierarchy.

Table 2

Classes of ANNOdc along with exemplary ANNOds subclasses as well as interlinks to the FMA and the GFO top-level ontology.

* LO is the line between Lambda and Opisthion, the occipital saggittal arc.

ANNOdc class	ANNOds Examples	FMA equivalent / ID	GFO superclass
AnatomicalEntity		Physical anatomical entity 61775	
AnatomicalStructure	Skeleton	Material anatomical entity 67165	MaterialObject
SpatialAnatomicalEntity		immaterial anatomical entity 67112	SpaceEntity
AnatomicalSpace			SpaceRegion
AnatomicalSurface		Anatomical surface 24137	SurfaceRegion
AnatomicalLine	LO*	Anatomical line 9657	LineRegion
AnatomicalPoint		Anatomical point 9658	PointRegion
MeasurementPoint	Lambda		
OrientationPoint	Orbitale		
AnatomicalPlane	FrontalPlane	Anatomical plane 242982	
AnatomicalAxis	SagittalAxis		
BoneStructure			
Bone	Mandibula	Bone organ 5018	
BonePart	ArcusAlveolaris	Related to Segment of bone organ 281808, Zone of bone organ 10483	
BoneCompound	Cranium	Comparable to union of Skeletal system 23881 and subdivision of skeletal system 85544	
ToothStructure			
Tooth	DensCaninus	Tooth 12516	
ToothPart			
Phenotype	SexGiles19671		Attributive
AnatomicalProperty			Attributive
RelativeAnatomicalLocation	Sinister	Anatomical qualitative coordinate 30346	Relator

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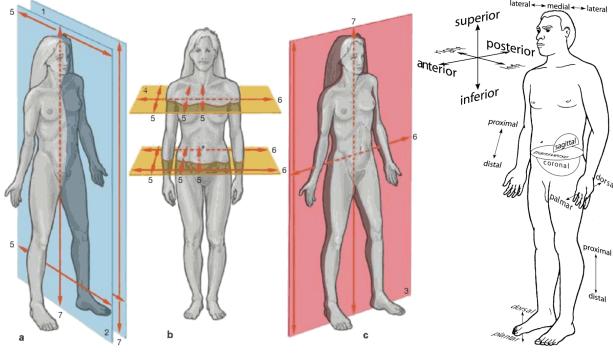


Fig. 3. Left: Anatomical Planes and Axes: 1 saggital plane, 2 midsaggital plane, 3 frontal plane, 4 transverse or horizontal plane, 5 saggital axis, 6 transverse axis, 7 longitudinal or vertical axis. a: Planum sagittale (sagittal plane), includes sagittal and longitudinal axes; the midsagittal plane passes through the midline of the body, b: Planum transversale (Transversal plane), includes transverse and sagittal axes, c: Planum frontale (frontal plane): includes longitudinal and transverse axes. Right: Anatomical relative location: directional terms used in anatomy and anthropology. Modified figure based on [33, 34].

A bone or bone element is a single self-contained bony skeletal entity. Bones, such as Mandibula (Mandible) and Os occipitale (Occipital Bone) are individual bone organs. FMA does have a "Bone" class (fma:30317) but that only has a single subclass "Skull bone" (fma:30317). Instead, "Bone organ" (fma:5018) is the equivalent class.

A bone compound is a section of the skeleton combining multiple bones or bone parts together, depending on the classification system chosen. This allows for the representation of any partonomy that need not necessarily be compatible with one another. Bone compounds, such as the Cranium (skull bone) consist of further bone compounds, individual bones (which in turn consist of bone parts) and bone parts. The FMA does not have a single equivalent class, but it is similar to the union of "Skeletal system" (fma:23881) and "Subdivision of skeletal system" (fma:85544). On the skeletal level, bone parts comprise any piece or portion of a bone. An anatomical landmark is any distinct structure on a bone. In ANNO, it is also referred to as a bone part.

4.7. Spatial anatomical entity

An anatomical entity without mass is classified as a spatial anatomical entity, which is either an anatomical space (three-dimensional), an anatomical surface (two-dimensional), an anatomical line (one-dimensional) or an anatomical point (zero-dimensional). It is equivalent to the FMA "Immaterial anatomical entity" (fma: 67112).

An anatomical space is a space region (three-dimensional) occupied by an anatomical structure. An anatomical surface is a boundary (two-dimensional) of an anatomical space.

An anatomical point (or osteometric landmark) is any immaterial, conceptual point that marks a location on a bone, either to create measurements (anatomical lines) or locate other anatomical points, e.g. by aligning the bone in specific planes for a measurement. The former serve as measurement points, the latter - relevant to the measurement procedure as orientation points. An anatomical point is thus a boundary (zero-dimensional) of an anatomical line.

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An anatomical line is a conceptual, immaterial line on a bone passing between at least two measurement points that, in the form of distances, circumferences or angles is used to collect measurements representing aspects of a bone's dimensions. An anatomical line is a boundary (one-dimensional) of an anatomical surface. Anatomical lines can connect or pass through anatomical entities (e.g., an edge between two or an angle between three anatomical entities). The length of the line or the angle degree can be measured and used in functions to infer individual phenotypes. For example, the anatomical line ZyDexterumZySinistrum between the Zygion Dexterum and the Zygion Sinistrum is used in the sex determination function for the phenotype SexGilesElliot196319 [37].

4.8. Phenotype

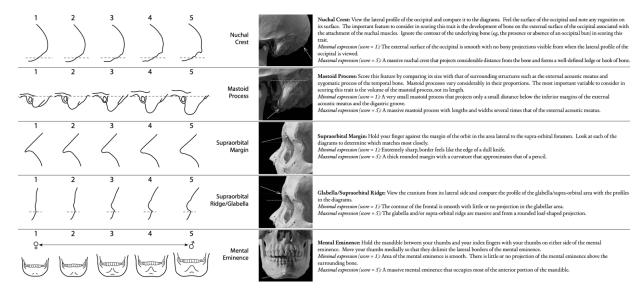


Fig. 4. Exemplary assessment of anatomical properties for the derivation of the phenotype sex, where five cranial bone parts are evaluated for their level of morphological expression. Method after Walker [38]. Image taken unmodified from [38].

Usually, a phenotype is considered as a (combination of) bodily feature(s) or observable characteristic(s) of an organism, such as sex, body height or body weight [39-41]. Since phenotypes are individual properties, they can be considered as attributives in the GFO sense. The phenotype notion has also been analyzed in detail within the framework of the Core Ontology of Phenotypes [42].

In the case of ANNO, the phenotype can be derived using functions comprising obtained measurements. Discriminant functions, e.g. based on the Cranium, allow for the assignment to sex while estimation of stature is attained by using linear regressions, e.g. based on Femur or Tibia length measurements. RDF is not optimized for mathematical formulas so we model those as literals.

As an example, the discriminant function no. 6 developed by Vodanovic uses the measurement of the angus mandibulae to assign sex, thus deriving a phenotype. For the left side it is given as:

$0.17 \cdot [ppam sinistrum-go sinistrum-paim sinistrum] - 20.43$

The right side is calculated analogously using (ppam dexterum-go dexterum-paim dexterum). The threshold or sectioning point is given at 0.2, meaning that if the result is greater than this value, then sex is assigned to male, if it is lower it is assigned to female.

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4.9. Anatomical property

An anatomical property is a characteristic (e.g., shape) of an anatomical structure. Anatomical properties are considered as attributes or qualities in GFO [43]. These are dependent individuals that characterise other individuals (in our case, anatomical structures).

Other than by means of osteometry using measurements and functions, an anatomical landmark's morphology can be used to examine anthropological aspects or phenotypes such as age or sex. For instance, assessment of the degree of an anatomical landmark's morphological expression (the anatomical property in this case) offers information about the sex of the remains of the individual being examined. While the morphological examination was not within the scope of the project, it was nonetheless ensured that future extension in this respect is possible.

5. Development of ANNOds

			label:en	label:de	label:la {form: singular}	label:la {form: plural}
	Os hamatum		Hamate bone	Hakenbein	Os hamatum	Ossa hamata
		Os hamatum dexter	Hamate bone right	Hakenbein rechts	Os hamatum dextrum	Ossa hamata dextra
		Os hamatum sinister	Hamate bone left	Hakenbein links	Os hamatum sinistrum	Ossa hamata sinistra
Os metacarpi			Metacarpal bones	Mittelhandknochen	Os metacarpi	Ossa metacarpalia
	Metacarpal bo	one 1	Metacarpal bone I	Mittelhandknochen 1	Os metacarpi 1	Ossa metacarpalia 1
		Metacarpal bone 1 right	Metacarpal bone 1 right	Mittelhandknochen 1 rechts	Os metacarpi 1 dextrum	Ossa metacarpalia 1 dextri

Fig. 5. Excerpt of the spreadsheet-based input template used by the anthropologists.

While ANNOdc is created by the ontologists in consultation with the domain experts, ANNOds is developed by the domain experts themselves. For this purpose they were provided with a spreadsheet-based SMOG [44] template by the ontologists, see fig. 5, eliminating the requirement of having a background in RDF and ontologies. The template is based on the structure of ANNOdc, so that the entered data is compliant with it: The ANNOds classes are subclasses of the ANNOdc classes (see table 2) and properties (see fig. 2) from ANNOdc are used. The spreadsheet is transformed to an OWL 2 ontology consisting of a taxonomy, annotations and some simple axioms on the basis of property restrictions. This approach ensures intuitive and unimpeded data input and a valid end result. Additionally, ANNOds is validated using SHACL shapes¹, which requires metaclasses. For example, all directly specified and transitive subclasses of Bone are also explicitly individuals of the metaclass BoneClass because of the limitations of SHACL. In addition, the objective is to initiate the process of data entry, encompassing selected bones of the skeleton. Given the overwhelming number of bone parts present in the cranium, the initial selection was narrowed down to a selection of representative and relevant parts. Overall, however, the aim is to include those that are of anthropological relevance, i.e. that contribute to navigation, localization, and identification on the bone. Those structures included in the definitions of others were also to be defined. For bones that lie on the median sagittal plane (e.g., mandible or sternum), bilateral landmarks are defined, each with a side reference. For osteometric landmarks, all those already established in the core literature are to be used. Furthermore, those that are relevant for meaningful measurement distances and can be annotated in AnthroWorks3D should be used. For the measurement distances, those should be selected that are established in the majority of the core literature as well as necessary for discriminant functions and functions for estimating height and weight. The functions chosen were those with diagnostic value. These were discriminant functions for sex determination and regression functions for body height and body weight estimation.

¹Contained in dist/shacl.ttl in https://zenodo.org/record/8380382.

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5.1. Process for creating definitions and measurements

For the definitions and measurements, a representative minimum amount of anthropological and anatomical English- and mostly German-language literature was compared in order to develop the definitions from their information. Notably, Latin or latinized ancient Greek terms often missing in the English literature and the FMA were included. Overall, the name of the structure, measurement or point is noted in Latin in singular and plural forms, English, German, synonyms in all three languages, the FMA and TA ID, and any information on function and delineation. This requires a positional description, such as the Punctum superioris capitis femoris as the superior located point of the Caput femoris. For the measurements, in addition to the name of the measurement, the type (e.g., distance measurement) and the measurement instrument were also recorded. The subsequent visual definitions were made in the different anatomical views marking the area of the anatomical structures and the position of the anatomical points.

5.2. Functions

The functions are divided into discriminant functions for sex determination and regress functions for body height. The sex of a specific individual within a population may be estimated using a function on skeletal measurements that is specific or similar to this population. Based on a threshold value, skeletons are classified into male, probably male, indifferent, probably female, and female. The exact number of categories may vary depending on the particular method used. ANNOds covers functions for at least one European, African, American, and Asian ethnicity or population.

5.3. Resolving incongruities

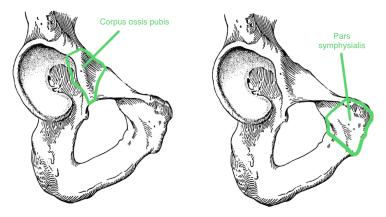


Fig. 6. The *Corpus ossis pubis* (pubic body) forms the proximal part of the *Os pubis* and includes the anterior part of the *Acetabulum* as well as the *Eminentia iliopubica*, which is a small elevation on the *Corpus ossis pubis*. According to the *TA* and others [6, 8, 25, 34, 45–47], this landmark is located in the region of the *Facies symphysialis*, which is defined here as the *Pars symphysialis*.

The domain experts reconciled conflicting literature to arrive at a consistent and logical result, for example for the Corpus ossis pubis, see fig. 6: Since the Corpi of the Os ischii and Os ilii are located in the area of the Acetabulum, it is logical to place the Corpus ossis pubis in this region as well [27, 31, 48–50], thereby forming the Acetabulum from all Corpi (Corpus ossis ilii, Corpus ossis ischii, and Corpus ossis pubis). The Pars symphysialis ossis pubis has ventral-medially the Facies symphysialis, which connects the paired Ossa pubes of the pelvic bones. In the Pars symphysialis, the Ramus inferior ossis pubis and Ramus superior ossis pubis merge into each other. This redefinition closes the resulting gap in the definition of the area between the Ramus inferior / superior ossis pubis. ANNO also corrects the commonly missing differentiation between Cranium and Mandibula in English literature, with no Latin equivalent for the overall term "skull". Instead, Cranium is established as a synonym for the skull [33, 47].

Further contributions of ANNOds include:

- Consistent Latin declension (including in plural).
- Transparency through detailed source references; for measurement distances, origin works and reference works are indicated by dividing into primary and core sources.

- Dissected group structures.
- Redefinitions such as joint surfaces, which are often not named in anatomy literature but are anthropologically relevant, and osteometric measurement points outside the skull, which are not established in the literature.

6. Use Case: Integration into AnthroWorks3D

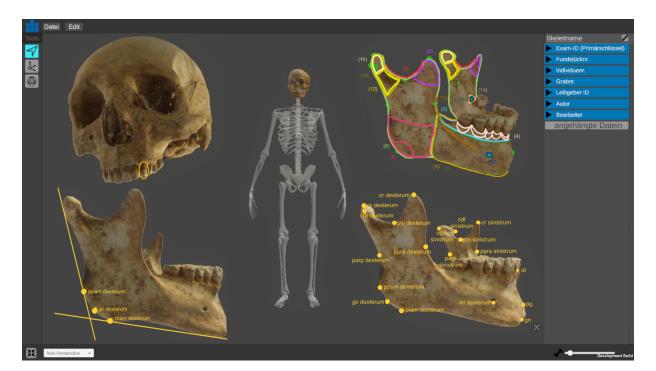


Fig. 7. Collage of different views in AnthroWorks3D of a 3D model of a *cranium* (skull) inserted into a placeholder skeleton (source: [51]). The cranium is a bone compound consisting of 29 bone elements. One of them, the *mandibula* (mandible), is flexibly connected to the rest of the skull by the mandibular joint. In the top right, various bone parts on the Mandibula are visually emphasized, with part 14 representing the angulus mandibulae (mandibular angle). In the top left, the *dens caninus maxillaris dexter* (right maxillary secondary canine tooth), a tooth structure, is highlighted within a circle. The tooth cusp is a part of the tooth. It refers to the projection that divides the occlusal surface of most of the teeth, with the exception of the incisors. At the bottom right, anatomical points (osteometric landmarks) of the right lateral side of the *mandibula* are depicted. The *gonion dextrum* (right gonion; *gonion dextrum*, abbreviated as *go dextrum*, depicted alternative spelling of dextrum: dexterum) forms part of an anatomical line measuring the *angulus mandibulae*, as shown in the bottom left. Skeletal material courtesy of Dr. Birgit Grosskopf, Department of Historical Anthropology and Human Ecology, University of Göttingen.

AnthroWorks3D, see fig. 7, is a German-language tool that combines user-friendly techniques of photogrammetry with insights from user experience research and knowledge from game development and offers a procedure to create 3D skeletal models serving as digital twins which can subsequently be examined virtually. This facilitates anthropological work to be location-independent and parallel without exercising wear and tear on the skeletal material. The examination can be performed as often as desired, even if the skeletal individuals or collections are not available at the institute or have already been reburied. We use our ontological architecture (top-level ontology, domain core ontology, domain specific ontology) to integrate ANNO into AnthroWorks3D based on the three-ontology method [52].

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Lower Limb
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  @language"
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                                                                             Femur right
 "@language" : "la",
"@value" : "Incus"
                                                                            Foot
                                                                             Metatarsal bones
                                                                              Metatarsal bones 1
     //www.w3.org/2000/01/rdf-schema#subClassOf":
                                                                              Metatarsal bones 2
         "https://ols.imise.uni-leipzig.de/ontologies/anno#Ossicles
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 "@value" : "52752
                                                                              Metatarsal bones 5
                                                                             Phalanges
"https://ols.imise.uni-leipzig.de/ontologies/anno#id" : [ {
 "@value" : "B0037'
```

Fig. 8. Left: ANNO converted to the AnthroWorks3D JSON input format. Right: Bone selection in the imported hierarchy.

One of the advantages of the three-ontology method is that the software only needs to implement access to the entities (classes and properties) of ANNOdc ontology, whereas the classes of ANNOds are processed dynamically, as shown in fig. 8. In addition, it is now possible to perform sex determinations using discriminant functions, as shown in fig. 9.

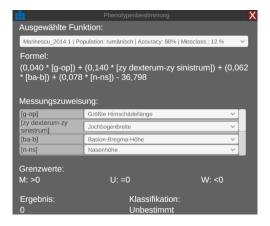


Fig. 9. Sex determination in AnthroWorks3D using the imported ANNO discriminant functions.

7. Conclusion and Future Work

By contributing systematic, standardized and clear definitions for (material and spatial) anatomical entities regarding the human skeleton as well as anthropological aspects such as the derivation of phenotypes, ANNO formalizes knowledge in the fields of anatomy and anthropology. It encompasses all the essential elements for the routine use of anatomical terms in daily anthropological practice. Integrating the ontology into AW3D enables its immediate application in anthropological analysis.

The ontology is interlinked with the TA and FMA and provides transparency by including all sources used to generate its content. Moreover, it provides a method for conceptualizing the ontology and generating its content. The ontology replaces an old, hard-coded format in AnthroWorks3D, which saves development time, separates annotation file compatibility from software versions, eases annotation through hierarchical browsing and improves interoperability and customization. Also, ANNO comes with extensive documentation of the process and available resources. Hence, foundations have been laid for its use and further development and management.

Navigating through plenty of options for designing an ontology makes consistency challenging to achieve. As documented in the TA [6, part II], the choices frequently reflect the preference of a party having something included

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or named or structured in a certain way because it is considered relevant by a particular party. ANNO represents a specialized ontology that builds upon existing ontologies such as FMA and TA. However, meeting the diverse needs of the field and interdisciplinary requirements can only be accomplished through an ongoing, gradual process over time.

While ANNO serves as a foundation for describing anatomical terms, additional work is required to comprehensively cover the anthropological domain. For instance, it only covers standardized normal adult anatomy. Moreover, in order to remain permanently suitable for practical usage, the ontology must be continuously reviewed, updated, enriched or adapted to further needs. Moreover, ANNO presents all necessary prerequisites to being extended to the other aspects of anthropological work with human skeletal remains.

Apart from continuing data entry, future work may involve the inclusion of other anthropologically relevant properties such as those that capture the morphology of the human skeleton and contribute to deriving phenotypes such as sex, age and developmental aspects, pathologies or ancestry. In addition, tooth parts, deciduous teeth as well as anatomical variants could be added in the future.

Regarding skeletal anatomy, the representation of preservation status through the ontology would be of great use for anthropological work. Furthermore, anatomical terms describing aspects of bone morphology or function, such as the Latin word "processus" which appear numerous times as part of anatomical designations indicating a projection, could be reviewed and flexibly systematized through ANNO, making them analyzable as well.

ANNO's potential applications include forensic, historical and prehistoric anthropology, as well as pathology and medicine, and the field of computer science, especially medical informatics.

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References

- [1] G. Grupe, M. Harbeck and G. McGlynn, Prähistorische Anthropologie, Springer Spektrum, 2015, pp. 217–221.
- [2] M. Heuschkel, S. Becker, S. Jeraufke, C. Lucas, M. Mohaupt, D. Labudde, T. Kirsten and B. Grosskopf, Zur Spurensuche an menschlichen Skeletten, Humboldt-Universität zu Berlin, 2021, pp. 127–136. doi:10.18452/22415.
- [3] S. Standring, A brief history of topographical anatomy, Journal of anatomy 229(1) (2016), 32-62.
- $[4]\ F.\ Loebe,\ P.\ Burek\ and\ H.\ Herre,\ GFO:\ The\ General\ Formal\ Ontology, \\ Applied\ Ontology\ (2022),\ 1-36.\ doi:10.5281/zenodo.5205420.$
- [5] K. Höffner, RickView: Lightweight Standalone Knowledge Graph Browsing Powered by Rust., arXiv e-prints (2023).
- [6] Federative International Programme for Anatomical Terminology, *Terminologia Anatomica*, 2nd edn, FIPAT.library.dal.ca, 2019, errata version 2.07 from 16 August 2021. https://fipat.library.dal.ca/ta2/.
- [7] Federative Committee on Anatomical Terminology, Terminologia Anatomica, Thieme, 1998. ISBN 9783131143617.
- [8] W. Dauber and H. Feneis, Feneis' Bild-Lexikon der Anatomie, 11 edn, Thieme, 2019.
- [9] H. Feneis, Pocket atlas of human anatomy, Thieme, 2007.
- [10] W. Dauber, Feneis. Nomenclatura Anatómica Ilustrada, Elsevier, 2021. ISBN 9788413820033.
- [11] C. Rosse and J.L.V. Mejino, *The Foundational Model of Anatomy Ontology*, in: *Anatomy Ontologies for Bioinformatics: Principles and Practice*, A. Burger, D. Davidson and R. Baldock, eds, Springer, London, 2008, pp. 59–117. doi:10.1007/978-1-84628-885-2_4.
- [12] O.P. Gobée, D. Jansma and M.C. DeRuiter, AnatomicalTerms.info: Heading for an online solution to the anatomical synonym problem hurdles in data-reuse from the Terminologia Anatomica and the foundational model of anatomy and potentials for future development, *Clinical anatomy (New York, N.Y.)* 24(7) (2011), 817–830. doi:10.1002/ca.21185.
- [13] C.J. Mungall, C. Torniai, G.V. Gkoutos, S.E. Lewis and M.A. Haendel, Uberon, an integrative multi-species anatomy ontology, *Genome Biology* 13(1) (2012), R5. doi:10.1186/gb-2012-13-1-r5.
- [14] B.D. Martin, D. Thorpe, V. DeLuna, T. Howard, J. Hagemeyer and N. Wilkins, Frequency in Usage of Terminologia Anatomica Terms by Clinical Anatomists, *Journal of Biomedical Education* 2014(2) (2014), 1–9. doi:10.1155/2014/950898.
- [15] B.E. Hirsch, Does the Terminologia Anatomica really matter?, Clinical anatomy (New York, N.Y.) 24(4) (2011), 503–504.

1.0

2.7

- [16] D. Kachlik, V. Baca, I. Bozdechova, P. Cech and V. Musil, Anatomical terminology and nomenclature: past, present and highlights, *Surgical and radiologic anatomy: SRA* **30**(6) (2008), 459–466. doi:10.1007/s00276-008-0357-y.
 - [17] R. O'Rahilly, Anatomical Terminology, Then and Now, Cells Tissues Organs 134(4) (1989), 291–300. doi:10.1159/000146705.
 - [18] O.P. Gobée, D. Jansma and M.C. DeRuiter, Anatomical Terms.info: Heading for an online solution to the anatomical synonym problem hurdles in data-reuse from the Terminologia Anatomica and the foundational model of anatomy and potentials for future development, *Clinical Anatomy* **24**(7) (2011), 817–830.
 - [19] M. Ocak, H.A. Aktaş, M.B. Uzuner, F. Geneci, Ç. Aşkit and M.F. Sargon, A comparison of the anatomical terminology in the last 25 years, Journal of the Anatomical Society of India 66 (2017), S31–S33. doi:10.1016/j.jasi.2017.09.002.
 - [20] D. Kachlik, V. Musil and V. Baca, Contribution to the anatomical nomenclature concerning general anatomy and anatomical variations, Surgical and Radiologic Anatomy 38 (2016), 757–765.
 - [21] T. Buklijas, The Science and Politics of Naming: Reforming Anatomical Nomenclature, ca. 1886–1955, Journal of the History of Medicine and Allied Sciences 72(2) (2017), 193–218.
 - [22] M. Loukas, I. Aly, R.S. Tubbs and R.H. Anderson, The naming game: A discrepancy among the medical community, *Clinical Anatomy* **29**(3) (2016), 285–289.
 - [23] D. Kachlik, V. Musil and V. Baca, Terminologia Anatomica after 17 years: inconsistencies, mistakes and new proposals, Annals of Anatomy-Anatomischer Anzeiger 201 (2015), 8–16.
 - [24] P.E. Neumann, One vowel or two? Diphthongs, digraphs, ligatures, and diaereses, oh my!, Clinical Anatomy 30(8) (2017), 1013–1016.
 - [25] K. Zilles and B. Tillmann, Anatomie, Springer, 2010.

2.7

- [26] M. Schünke, E. Schulte and U. Schumacher, *Prometheus LernAtlas-Allgemeine Anatomie und Bewegungssystem*, Vol. 1, 5th edn, Thieme, 2018.
- 18 [27] F. Paulsen and J. Waschke, Sobotta Allgemeine Anatomie und Bewegungsapparat, Elsevier GmbH, Urban & Fischer, München, 2017.
 - [28] G. Aumüller, G. Aust, A. Conrad, J. Engele, J. Kirsch and G. Maio, Duale Reihe Anatomie, 5th edn, Duale Reihe, Thieme, 2020.
 - [29] A. Rauber and F. Kopsch, Anatomie des Menschen. Lehrbuch und Atlas, Thieme, 1987.
 - [30] H.L. Barlett, Wesen und Methoden der Anthropologie 1 Wissenschaftstheorie, Geschichte, morphologische Methoden / mit Beitr. von H. L. Barlett, in: Anthropologie Handbuch d. vergleichenden Biologie d. Menschen; zugl. 4. Aufl. d. Lehrbuchs d. Anthropologie, begr. von Rudolf Martin, R. Knußmann, ed., Fischer, Stuttgart [u.a.], 1988. http://slubdd.de/katalog?TN_libero_mab2635674.
 - [31] S. Standring and N. Anand (eds), Gray's anatomy: The anatomical basis of clinical practice, 41 edn, Elsevier Health Sciences, 2016.
 - [32] K.W. Alt and Rösing, Friedrich W., Teschler-Nicola, Maria, Dental anthropology: Fundamentals, limits and prospects, Springer, Wien, 1998. ISBN 978-3-7091-7498-2.
 - [33] F. Paulsen, J. Waschke, T. Klonisch and S. Hombach-Klonisch, Sobotta: Atlas of human anatomy: General Anatomy and Muscoskeletal System, 15 edn, Sobotta, Vol. 1, Elsevier/Urban & Fischer, M"unchen, 2011. ISBN 978-0-7234-3639-3.
 - [34] T.D. White, M.T. Black and P.A. Folkens, *Human osteology*, Third edition edn, Elsevier Academic Press, Amsterdam and Boston and Heidelberg, 2012. ISBN 978-0-12-374134-9.
 - [35] F. Loebe, Towards an Ontology of Categories and Relations, in: Proceedings of the Joint Ontology Workshops 2018 Episode IV: The South African Spring, Cape Town, South Africa, September 17-18, 2018, L. Jansen, D.P. Radicioni and D. Gromann, eds, CEUR Workshop Proceedings, Vol. 2205, CEUR-WS.org, 2018. https://ceur-ws.org/Vol-2205/paper29_ontocom3.pdf.
 - [36] F. Loebe, P. Burek and H. Herre, Developing GFO 2.0 Further—Initiating the Modules of Space and Material Objects, in: Proceedings of the Joint Ontology Workshops 2021 Episode VII: The Bolzano Summer of Knowledge, Bolzano, Italy, September 11-18, 2021, E.M. Sanfilippo, O. Kutz et al., eds, CEUR Workshop Proceedings, Vol. 2969, CEUR-WS.org, 2021. https://ceur-ws.org/Vol-2969/paper69-FOUST.pdf.
 - [37] E. Giles and O. Elliot, Sex determination by discriminant function analysis of crania, *American journal of physical anthropology* **21**(1) (1963), 53–68.
 - [38] J.E. Buikstra and D.H. Ubelaker, Standards for data collection from human skeletal remains. Arkansas Archaeological Survey Research Series No. 44, Fayetteville: Arkansas Archaeological Survey (1994).
 - [39] R.H. Scheuermann, W. Ceusters and B. Smith, Toward an ontological treatment of disease and diagnosis, in: Proceedings of the 2009 AMIA Summit on Translational Bioinformatics, 2009, pp. 116–20. https://philarchive.org/archive/SCHTAO-29.
 - [40] M. Mahner and M. Kary, What exactly are genomes, genotypes and phenotypes? And what about phenomes?, *Journal of theoretical biology* **186**(1) (1997), 55–63. doi:10.1006/jtbi.1996.0335.
 - [41] R. Hoehndorf, A. Oellrich and D. Rebholz-Schuhmann, Interoperability between phenotype and anatomy ontologies, *Bioinformatics* **26**(24) (2010), 3112–3118.
 - [42] A. Uciteli, C. Beger, T. Kirsten, F.A. Meineke and H. Herre, Ontological representation, classification and data-driven computing of phenotypes, *Journal of Biomedical Semantics* 11(1) (2020), 1–17.
 - [43] P. Burek, F. Loebe and H. Herre, Towards GFO 2.0: Architecture, modules and applications, in: *Formal Ontology in Information Systems*, IOS Press, 2020, pp. 32–45.
 - [44] A. Uciteli, C. Beger, S. Kropf and H. Herre, Spreadsheet Model Generator (SMOG): A Lightweight Tool for Object-Spreadsheet Mapping., Studies in health technology and informatics 267 (2019), 110–117.
 - [45] W. Dauber and H. Feneis, Feneis' Bild-Lexikon der Anatomie, 5th edn, Thieme Flexibooks, Thieme, 2007.
 - [46] R.L. Drake, A.W. Vogl and A.W. Mitchell, Gray's anatomy for students, 4th edn, Elsevier Health Sciences, 2019.
- [47] M. Schünke, E. Schulte and U. Schumacher, *Prometheus: LernAtlas der Anatomie: Kopf, Hals und Neuroanatomie*, Vol. 3, Georg Thieme Verlag, 2009.
 - [48] F. Anderhuber, F. Pera and J. Streicher, Waldeyer-Anatomie des Menschen: Lehrbuch und Atlas in einem Band, 2012.

- [49] N.R. Langley, L.M. Jantz, S.D. Ousley, R.L. Jantz and G. Milner, Data collection procedures for forensic skeletal material 2.0, *University of Tennessee and Lincoln Memorial University* (2016).
- [50] M. Marinescu, V. Panaitescu and M. Rosu, Sex determination in Romanian mandible using discriminant function analysis: Comparative results of a time-efficient method, *Rom J Leg Med* **21**(4) (2013), 305–8.
- [51] M.-L. Heuschkel, H. Fritzsch, F. Schmiedel, S. Meier, P. van der Burgt, B. Grosskopf, A. Flux, M. Mohaupt, D. Labudde and T. Kirsten, AnthroWorks3D—A Digital Tool for Anthropological Analyses in Cultural Heritage Institutions and Beyond, CIDOC Conference 2020: Digital Transformation in Cultural Heritage Institutions, Geneva. Workshop. https://www.youtube.com/watch?v=yeGwR80UpUI.
- [52] R. Hoehndorf, A.-C.N. Ngomo and H. Herre, Developing consistent and modular software models with ontologies, in: *New Trends in Software Methodologies, Tools and Techniques*, IOS Press, 2009, pp. 399–412.