

Anatomy and Standardization

Principles for Representing Biological Structure

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Anatomy and Standardization

Challenges

- Lack of a unifying theory of anatomy;
- Ambiguity about what anatomy is
what does and does not belong in it;
- Lack of universally accepted principles
for representing anatomical reality

Anatomy and Standardization

Questions proposed for discussion

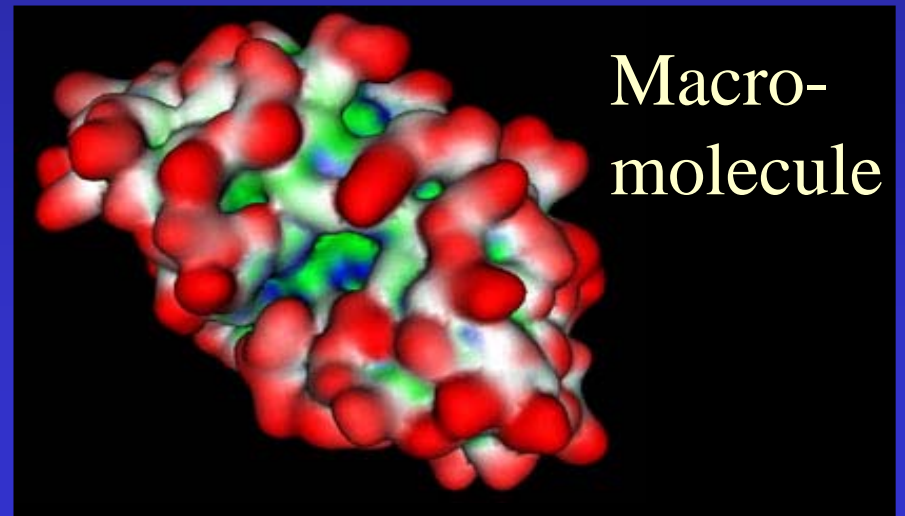
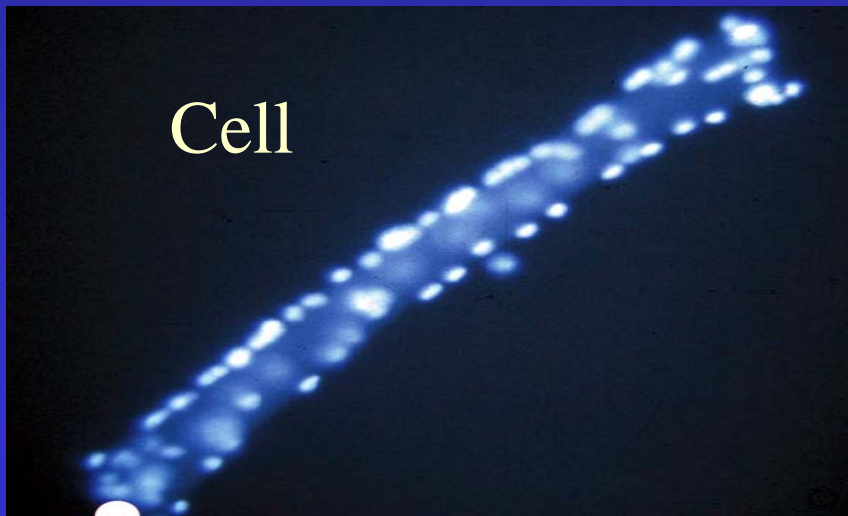
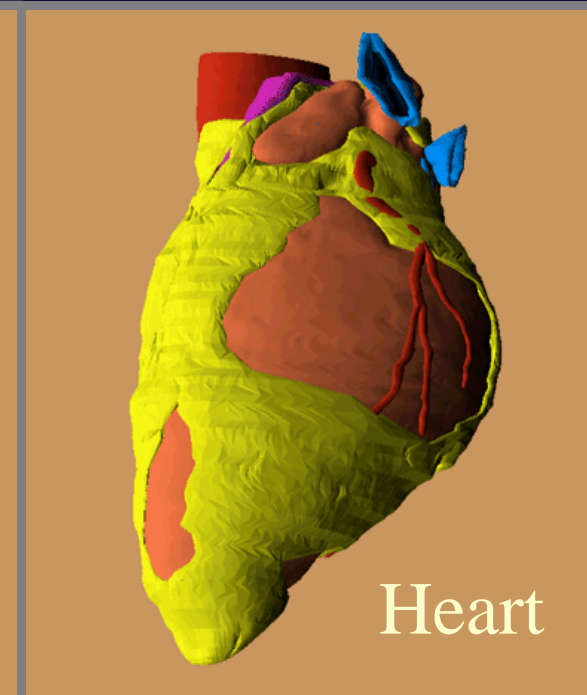
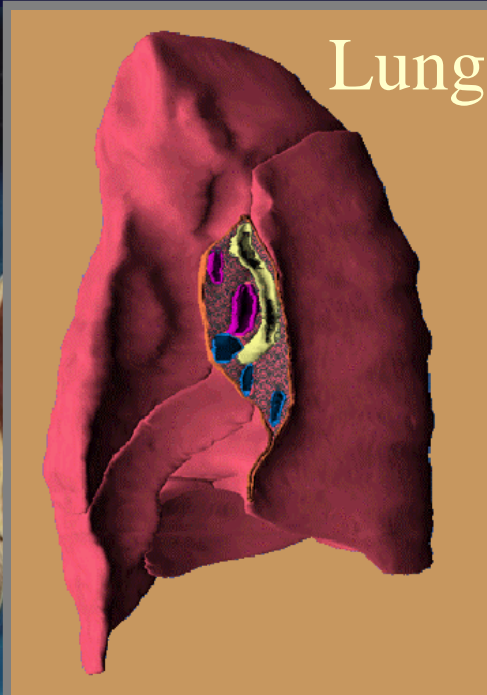
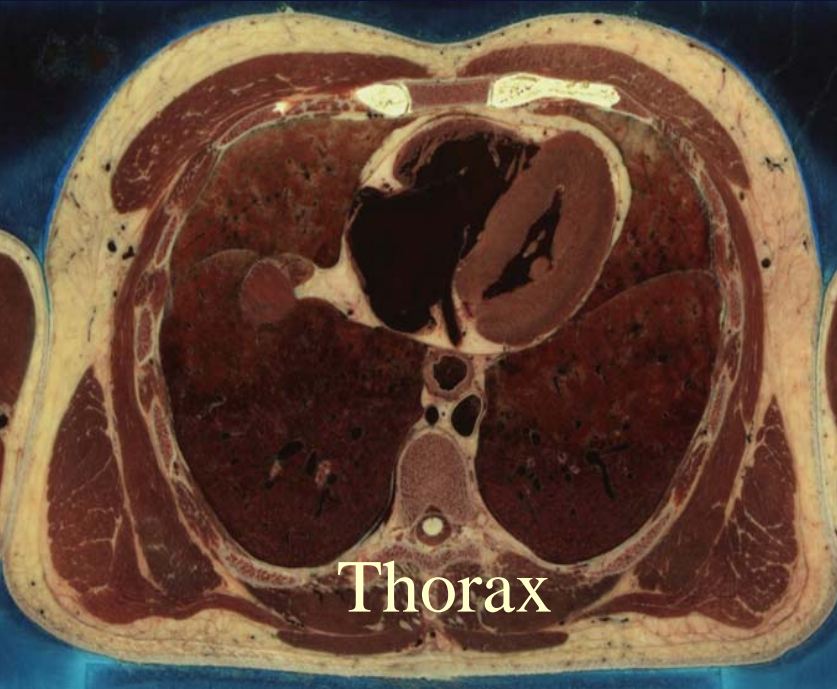
- Is an anatomy standard a matter of standardized terminology, or of something else?
- Do we need a standard for anatomy as a whole, or just for some parts?
- How big should a standard be?
- Should it include for example histology?
- How does the development of an anatomy standard relate to the development of a reference ontology for anatomy?

Anatomy and Standardization

Questions proposed for discussion

- Is an anatomy standard a matter of standardized terminology, or of something else?
- Do we need a standard for anatomy as a whole, or just for some parts?
- How big should a standard be?
- Should it include for example histology?
- How does the development of an anatomy standard relate to the development of a reference ontology for anatomy?

Reality or Views of Reality?



Reality or Views of Reality?



Terminology or Something Else?

NOMINA ANATOMICA

• SIXTH EDITION •

Incisura cardiaca (pulmonis sinistri)
 Margo inferior
 Hilum pulmonis
 Radix (Pediculus) pulmonis
 Lingula pulmonis sinistri
 Culmen pulmonis sinistri
 Lobus superior
 Lobus medius (pulmonis dextri)
 Lobus inferior
 Fissura obliqua
 Fissura horizontalis (pulmonis dextri)

CAVITAS THORACIS [THORACICA]

Regiones pleuropulmonales¹³

Fascia endothoracica
 Membrana suprapleuralis
 Fascia phrenicopleuralis
 Cavitas pleuralis
 Pleura
 Cupula pleurae

Terminologia Anatomica

International Anatomical Terminology

FCAT
Federative Committee on Anatomical Terminology

A02.3.04.002	Cavitas thoracis; Cavitas thoracica	Thoracic cavity; Thorax
A07.1.01.001	Cavitas pleuralis	Pleural cavity
A07.1.02.001	Pleura	Pleura
A07.1.02.002	Pleura visceralis; Pleura pulmonalis	Visceral pleura; Pulmonary pleura
A07.1.02.003	Tunica serosa	Serosa; Serous coat
A07.1.02.004	Tela subserosa	Subserosa; Subserous layer
A07.1.02.005	Pleura parietalis	Parietal pleura
A07.1.02.006	Cupula pleurae	Cervical pleura; Dome of pleura; Pleural cupula
A07.1.02.007	Pars costalis	Costal part
A07.1.02.008	Pars diaphragmatica	Diaphragmatic part
A07.1.02.009	Pars mediastinalis	Mediastinal part
A07.1.02.010	Tunica serosa	Serosa; Serous coat

Terminology or Something Else?

NOMINA
ANATOMICA

Incisura cardiaca (pulmonis sinistri)
Margo inferior
Hilum pulmonis
Radix (Pediculus) pulmonis
Lingula pulmonis sinistri

CAVITAS THORACIS
[THORACICA]

Regiones pleuropulmonales¹³

History of *Terminologia Anatomica*

1850's Discussions of standardizing anatomical terminology

1895 The Basle *Nomina Anatomica*

1989 *Nomina Anatomica* Sixth edition

1998 *Terminologia Anatomica*

Federative Committee on anatomical terminology

Terminologia Anatomica

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A07.1.02.008	Pars diaphragmatica	Diaphragmatic part
A07.1.02.009	Pars mediastinalis	Mediastinal part
A07.1.02.010	Tunica serosa	Serosa; Serous coat

Anatomy Terminology Standard

A03.6.00.001	Juncturae membri inferioris	Joints of lower limb
A03.4.00.001	JUNCTURAE CINGULI PELVICI	JOINTS OF PELVIC GIRDLE
A03.6.01.001	Syndesmoses cinguli pelvici	Syndesmoses of pelvic girdle
A03.6.01.002	Membrana obturatoria	Obturator membrane
A03.6.01.003	Canalis obturatorius	Obturator canal
A03.6.02.001	Symphysis pubica	Pubic symphysis
A03.6.02.002	Discus interpubicus; Fibrocartilago interpubica	Interpubic disc; Interpubic fibrocartilage
A03.6.02.003	Lig. pubicum superius	Superior pubic ligament
A03.6.02.004	Lig. pubicum inferius	Inferior pubic ligament
A03.6.03.001	Articulatio sacroiliaca	Sacro-iliac joint
A03.6.03.002	Lig. sacroiliacum anterius	Anterior sacro-iliac ligament
A03.6.03.003	Lig. sacroiliacum interosseum	Interosseous sacro-iliac ligament
A03.6.03.004	Lig. sacroiliacum posterius	Posterior sacro-iliac ligament
A03.6.03.005	Lig. sacrotuberale	Sacrotuberous ligament
A03.6.03.006	Processus falciformis	Falciform process
A03.6.03.007	Lig. sacrospinale	Sacrospinous ligament
A03.6.03.008	Foramen ischiadicum majus	Greater sciatic foramen
A03.6.03.009	Foramen ischiadicum minus	Lesser sciatic foramen
A03.6.04.001	JUNCTURAE MEMBRI INFERIORIS LIBERI	JOINTS OF FREE LOWER LIMB
A03.6.05.001	Syndesmosis tibiofibularis	Tibiofibular syndesmosis; Inferior tibiofibular joint
A03.6.05.002	Membrana interossea cruris	Interosseous membrane of leg
A03.6.05.003	Lig. tibiofibulare anterius	Anterior tibiofibular ligament
A03.6.05.004	Lig. tibiofibulare posterius	Posterior tibiofibular ligament

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**Rosse C: *Terminologia Anatomica*:
Considered from the Perspective of
Next-Generation Knowledge Sources
Clinical Anatomy; 14:120-133 (2001)**

Terminology or Something Else?

“The oesophagus is a muscular tube ... connecting the pharynx to the stomach. It begins in the neck, level with the lower border of the cricoid cartilage and the sixth cervical vertebra; descending largely anterior to the vertebral column through the superior and posterior mediastina.”

Gray's Anatomy, 38th edition, p. 1751

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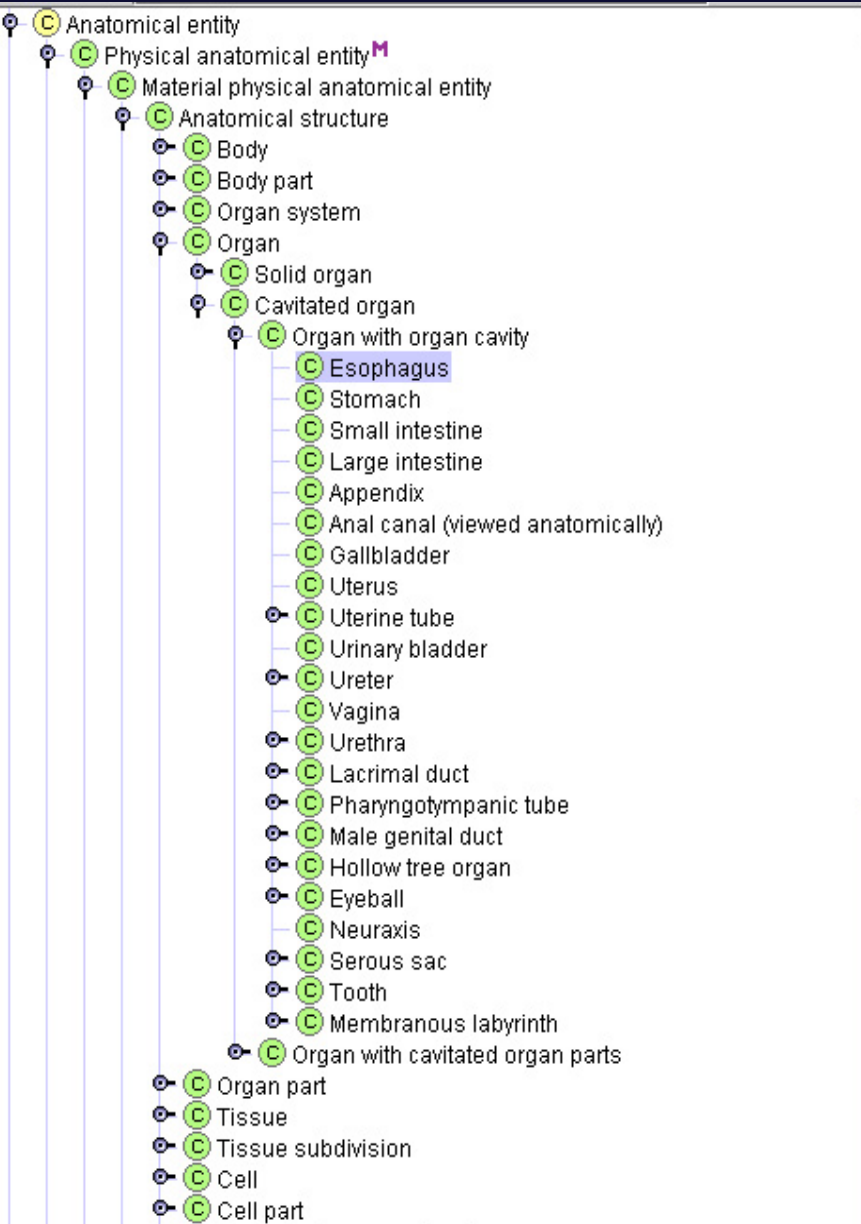
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Something else !!!



Preferred Name **UWDAID**

Synonyms **Non-English Equivalents**

Definition
 Organ with organ cavity which is continuous proximally with the pharynx and distally with the stomach.
 Examples: There is only one esophagus.

Member Of **Part**

- Wall of esophagus
- Lumen of esophagus
- Cervical part of esophagus
- Thoracic part of esophagus
- Abdominal part of esophagus
- Broncho-esophageus

Part Of

Attributed Part

related part	anatomical/arbitrary	shared/unshared	partition
Wall of esophagus	Anatomical	Unshared	Partition 1
Lumen of esophagus	Anatomical	Unshared	Partition 1
Cervical part of esophagus	Arbitrary	Unshared	Partition 2
Thoracic part of esophagus	Arbitrary	Unshared	Partition 2

Attributed Continuous With

related object	coordinate	laterality
Pharynx	Superior	
Stomach	Inferior	

Orientation

related object	coordinate	laterality
Plane of pharyngoesophageal junction	Superior	
Plane of esophagogastric junction	Inferior	

Something Else: Ontology

Ontology

A theory of reality

A conceptualization of what exists

Distinct from VIEWS of reality

e.g., textbooks

Ontology of anatomy

a conceptualization of

component parts of organisms

that exist as natural kinds (universals)

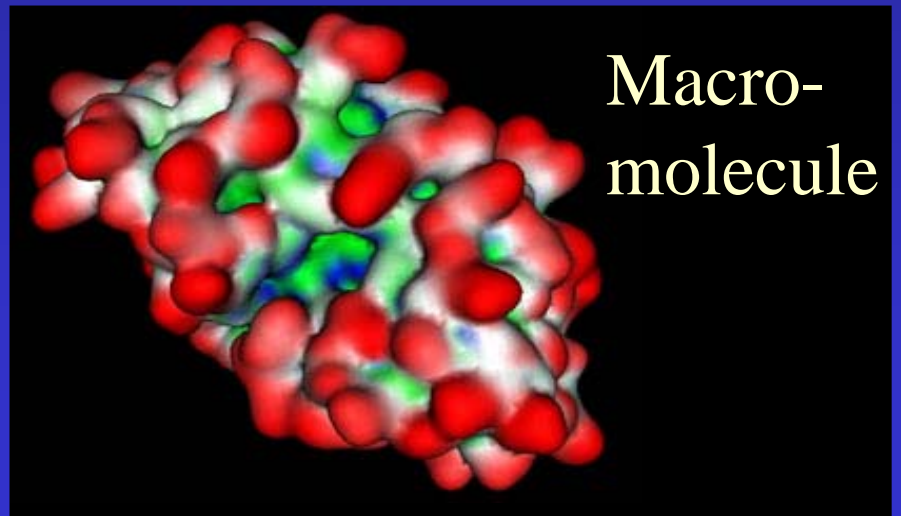
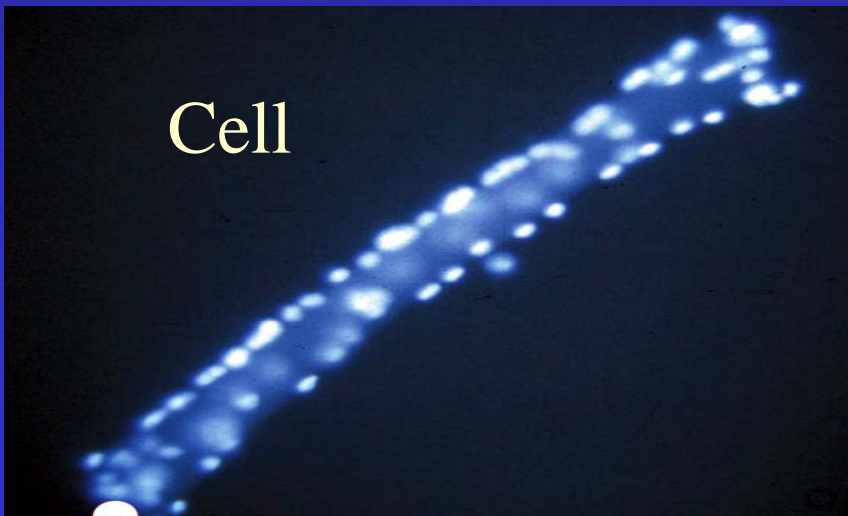
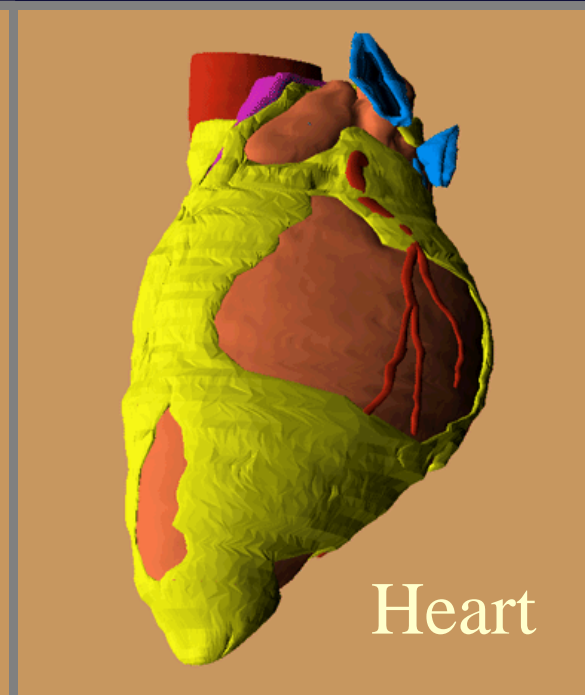
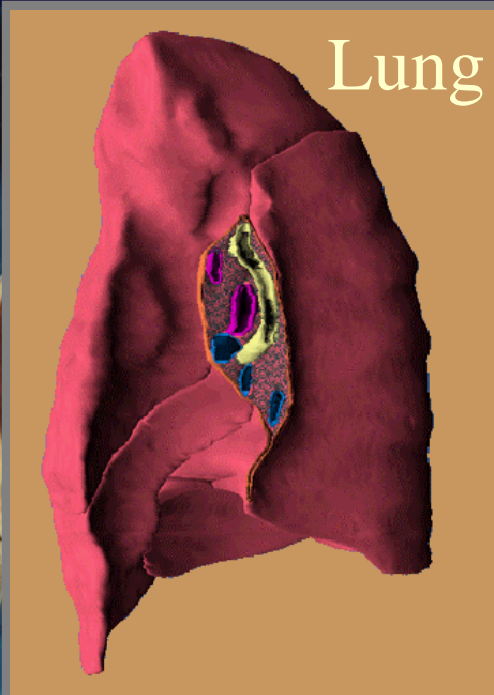
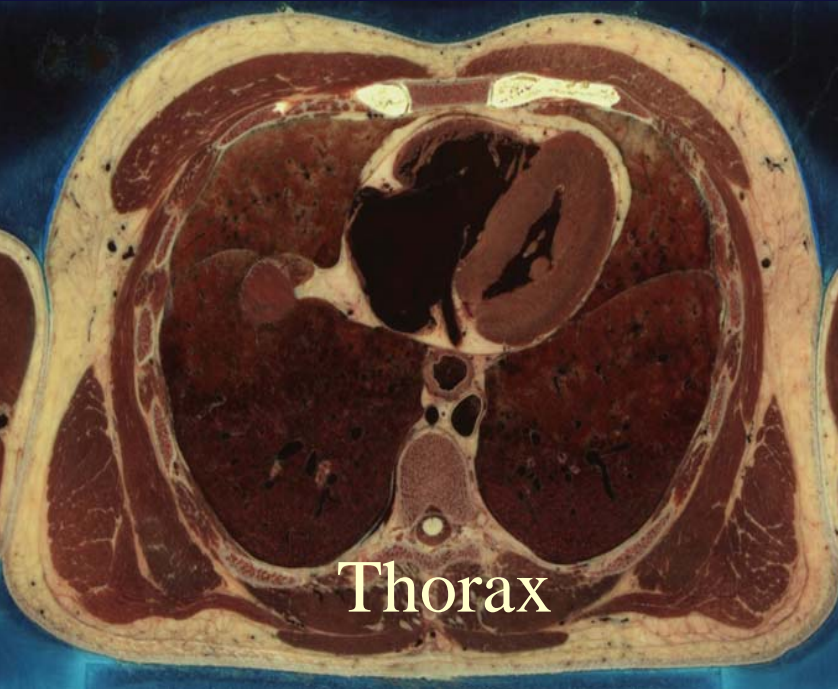
arrangement of component parts

Anatomy and Standardization

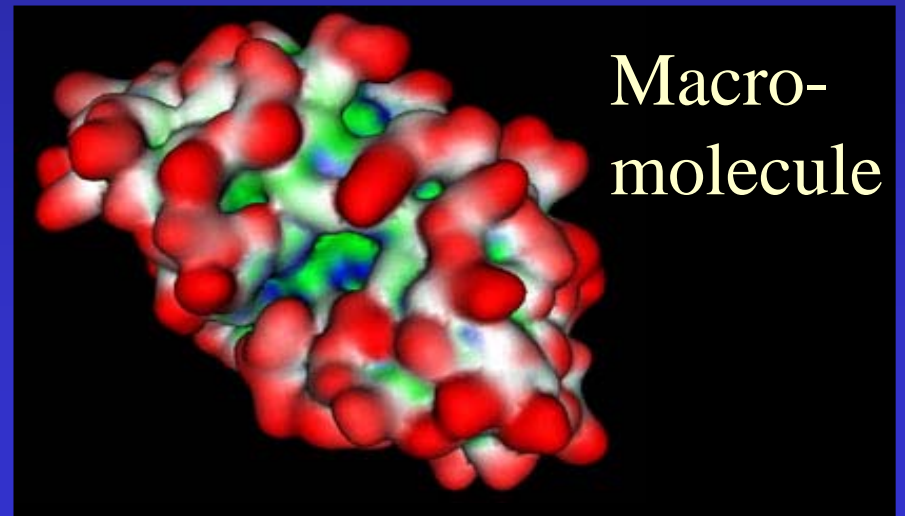
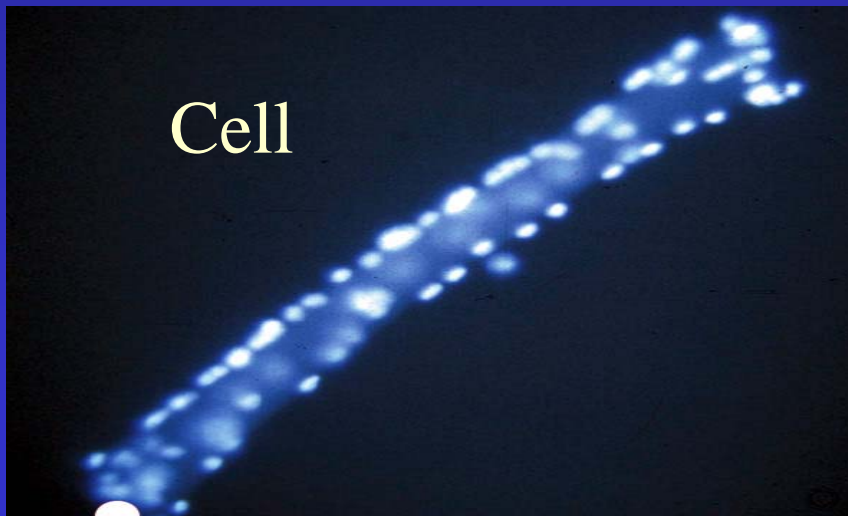
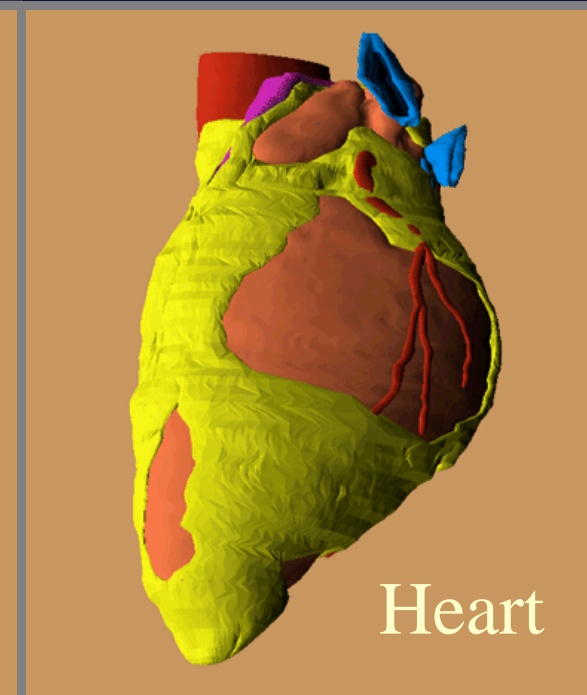
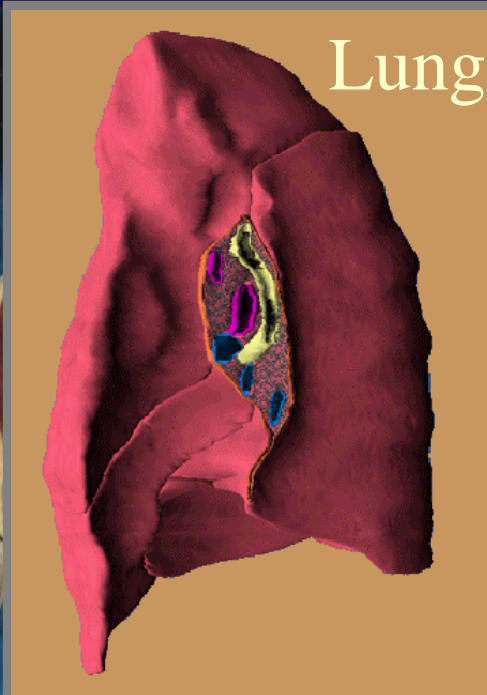
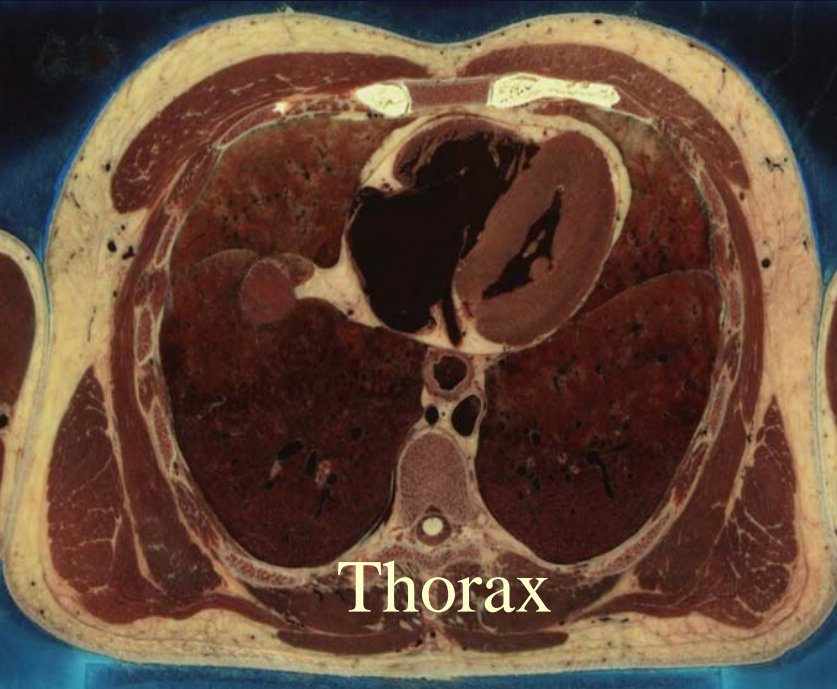
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- Is an anatomy standard a matter of standardized terminology, or of something else?
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Parts or Whole of Anatomy?



Anatomical Structures ?



What is structure?

"Structure" a *homonym* for

1. entity composed of parts;
2. arrangement or interrelation of parts of a whole.

What is structure?

"Structure" a *homonym* for

1. entity composed of parts;

(*e.g., a building, a cell, a plant, brain*)

i.e., a material object

2. arrangement or interrelation of
parts of a whole.

(*e.g., parts of a sentence or a symphony;*

strata of society;

metameric segmentation of earthworm)

i.e., relationships of entities to one another

What is anatomy?

"Anatomy" a *homonym* for

anatomy (structure)

anatomy (science)

What is anatomy?

"Anatomy" a *homonym* for

anatomy (*structure*)

e.g., anatomy of the frog, hand, brain

i.e., structural phenotype of an organism

anatomy (*science*)

systematized branch of knowledge accumulated
about anatomy (structure) or structural
phenotype

Distinction between Anatomical and Non-anatomical structures?

Anatomical structure

Material physical anatomical entity

has inherent 3D shape

generated by *coordinated expression of
organism's own structural genes*

spatially related in patterns

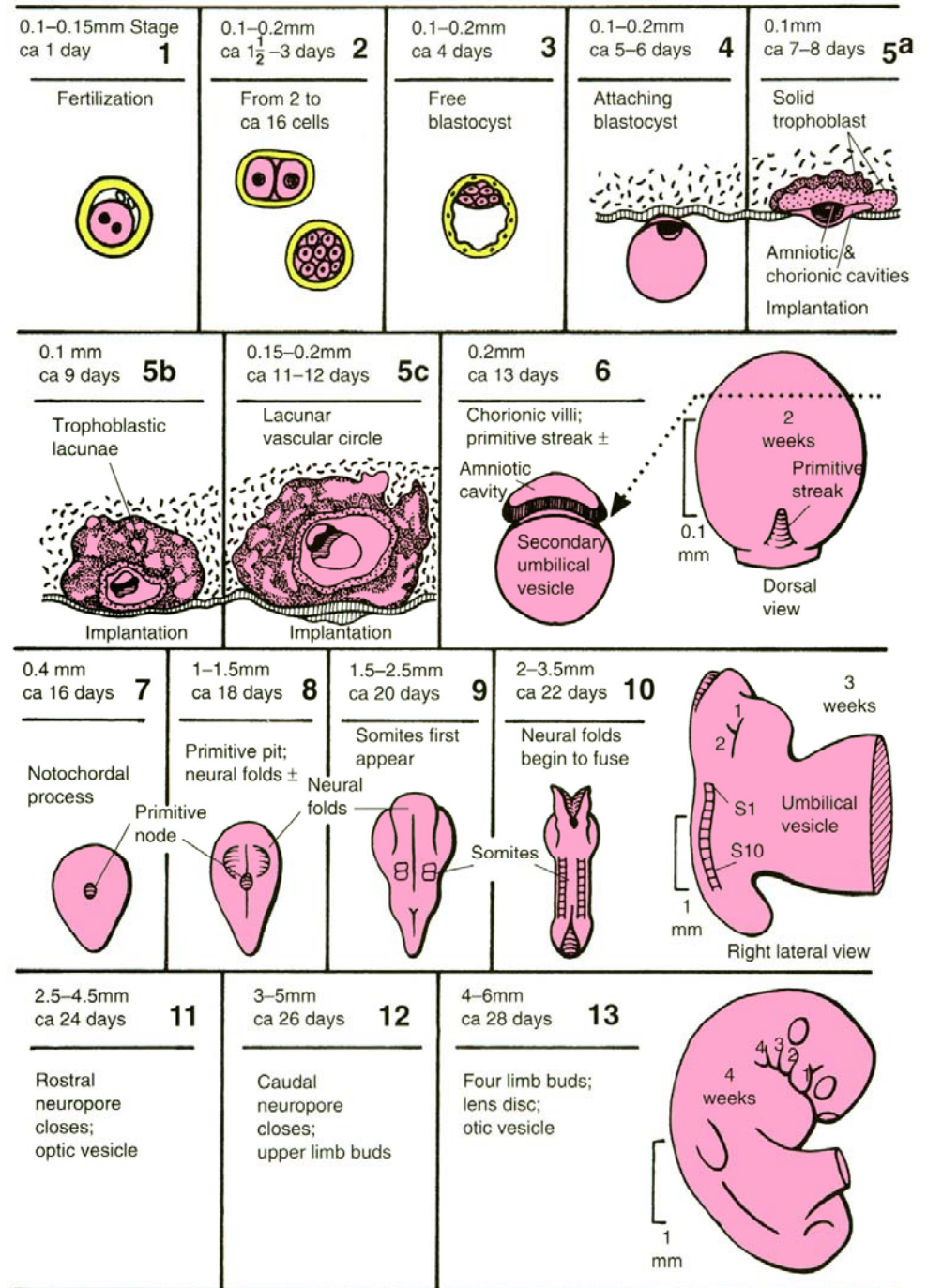
determined by *coordinated gene expression.*

Foundational Model of Anatomy

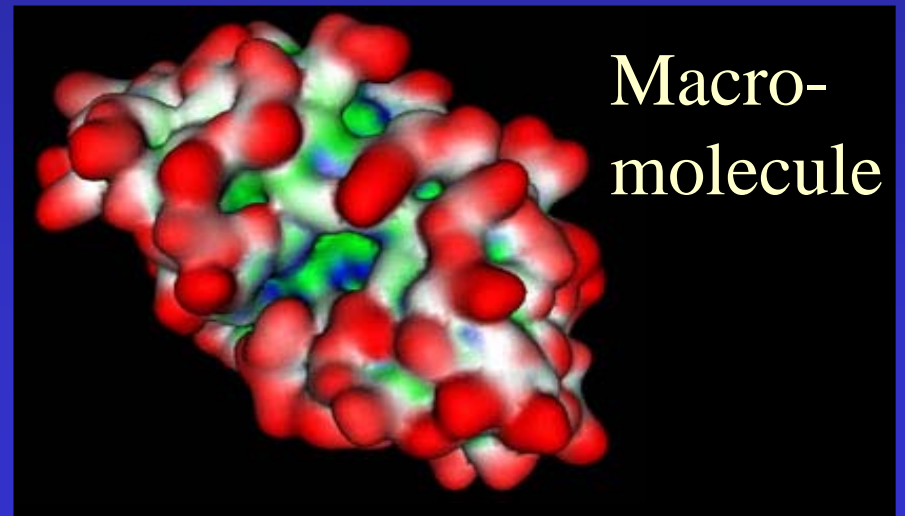
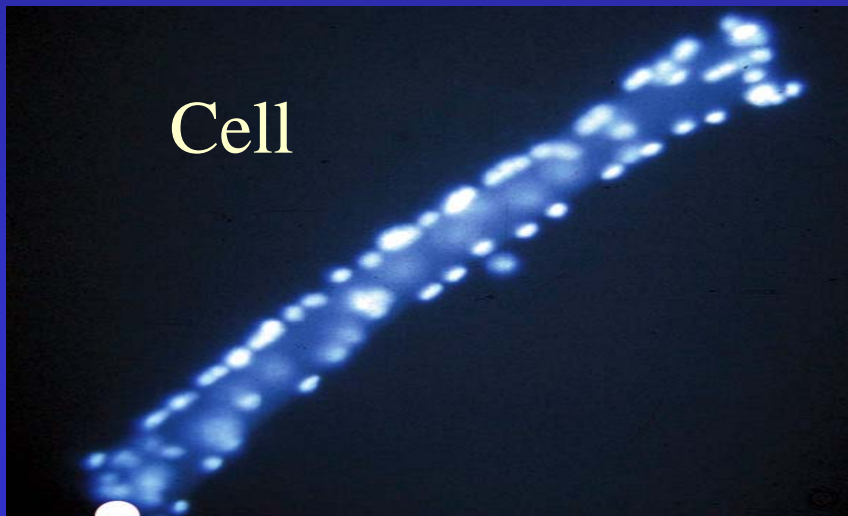
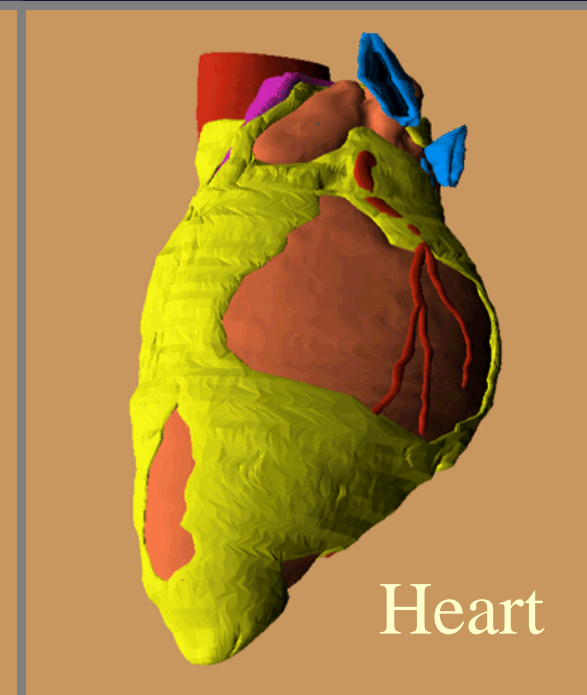
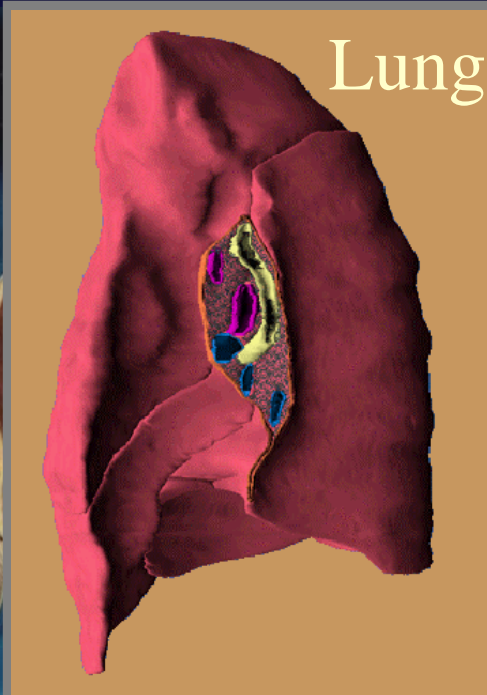
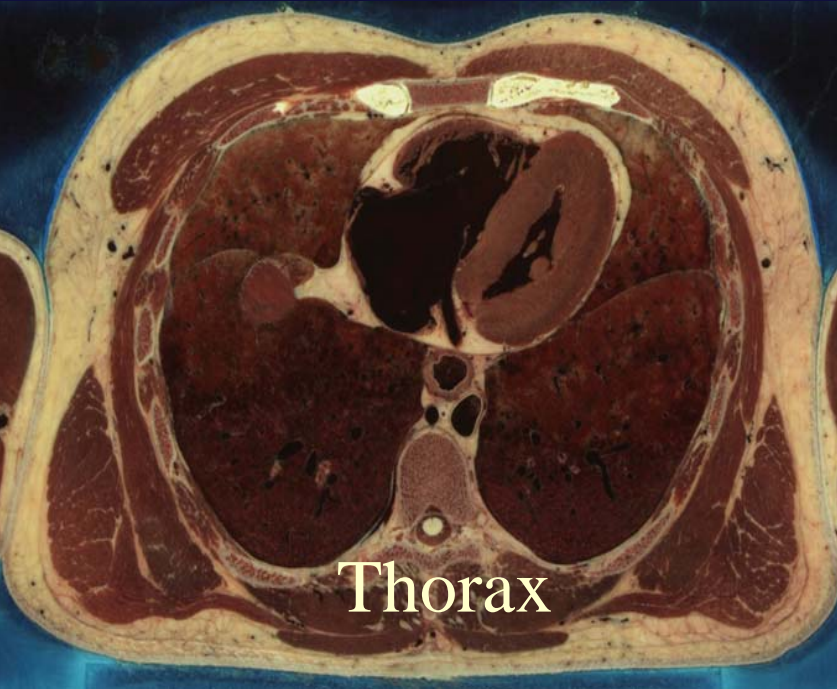
Embryogenesis:

The ontogeny of anatomical structure;
i.e., anatomical structures and their structural arrangement.

Establishment of an organism's structural phenotype



Anatomical Structure?



Anatomy Ontology

Foundational Model of Anatomy

- symbolic representation of

the structural phenotype of the human body (reality)

Declares the principles

for including entities and relationships
that are implicitly assumed
when knowledge of anatomy
is applied in different contexts;

Explicitly defines

entities and relationships
necessary and sufficient for consistently
representing the structure of the
human body.

Implemented as

a reference ontology for anatomy

Foundational Model of Anatomy

Unifying theory of anatomy

High Level Scheme

$FMA = (At, ASA, ATA, Mk)$

where:

At = Anatomy taxonomy

ASA = Anatomical Structural Abstraction

ATA = Anatomical Transformation Abstraction

Mk = Metaknowledge

(principles, rules, axioms)

Anatomy and Standardization

Challenges

- Lack of a unifying theory of anatomy;
- Ambiguity about what anatomy is
what does and does not belong in it;
- Lack of universally accepted principles
for representing anatomical reality

What are principles?

Assertions that guide reasoning and actions
e.g., those required for establishing an
ontology
standard

What kinds of principles?

- Domain-specific principles

Foundational principles for anatomy

(Rosse, Mejino: *J Biomed Inform* 2003)

- Top-level ontology principles

SNAP, SPAN

(Grenon, P and Smith, B., SNAP and SPAN:
Towards Dynamic Spatial Ontology,
Spatial Cognition and Computation 4(1), 69-103, 2004.)

- Implementation principles

(Zhang, Bodenreider: Law and order:
Assessing and enforcing compliance
with ontological modeling principles.
Comp Biol Med 2005; submitted)

Foundational Principles of Anatomy

1. Unified context principle
2. Abstraction level principle
3. Species specificity principle
4. Definition principle
5. Dominant concept principle
6. Organizational unit principle
7. Content constraint principle
8. Relationship constraint principle
9. Coherence principle
10. Representation principle

Foundational Principles of Anatomy

Unified Context Principle:

The abstraction should conform to a strictly structural context.

Only in a structural context is it possible to establish a single inheritance hierarchy (taxonomy) that subsumes all anatomical entities.

Unified Context Principle

Application context: Education, mix structural with functional, clinical concepts



Foundational Principles of Anatomy

Abstraction Level Principle:

The abstraction should represent canonical anatomy;
provide a framework for anatomical variants;
exclude instantiated anatomy.

Foundational Principles of Anatomy

Abstraction Level Principle:

Canonical anatomy

a field of *anatomy (science)*

comprises the synthesis of generalizations

based on anatomical observations that

describe idealized *anatomy (structure)*

implicitly sanctioned by their usage

in anatomical discourse;

deals with *universals* and not individuals.

Foundational Principles of Anatomy

Abstraction Level Principle:

Instantiated anatomy

a field of *anatomy (science)*

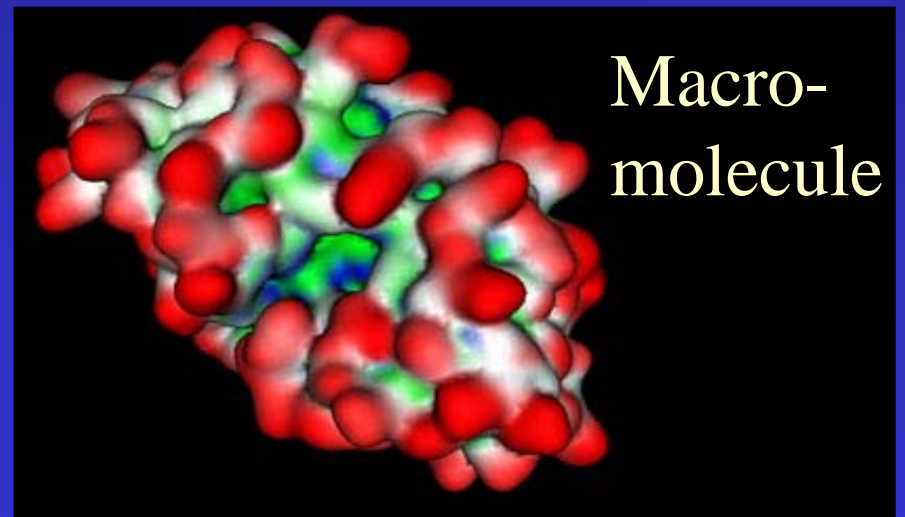
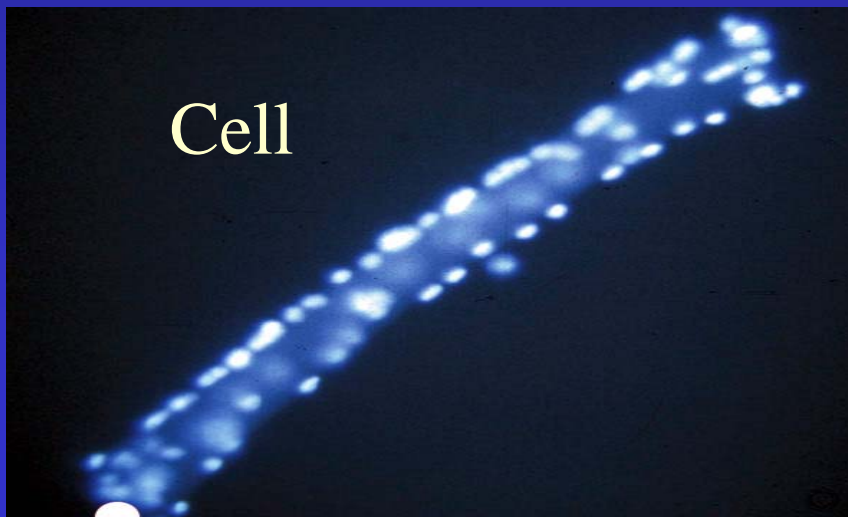
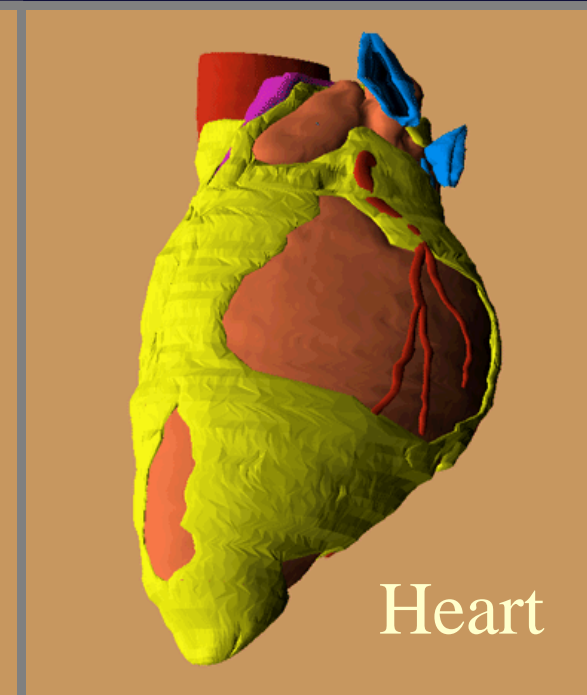
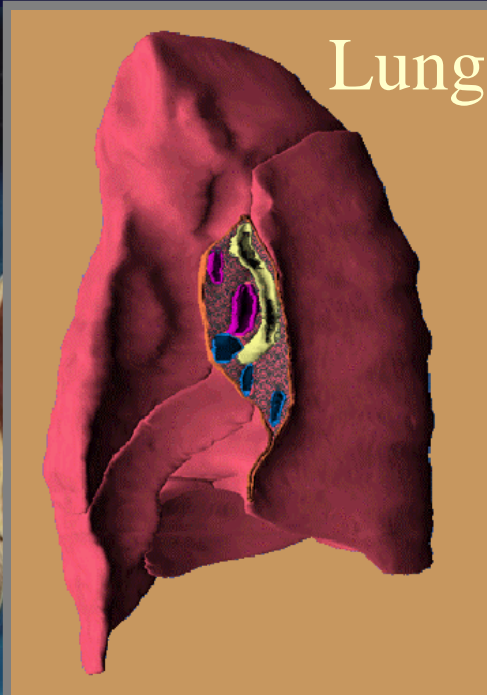
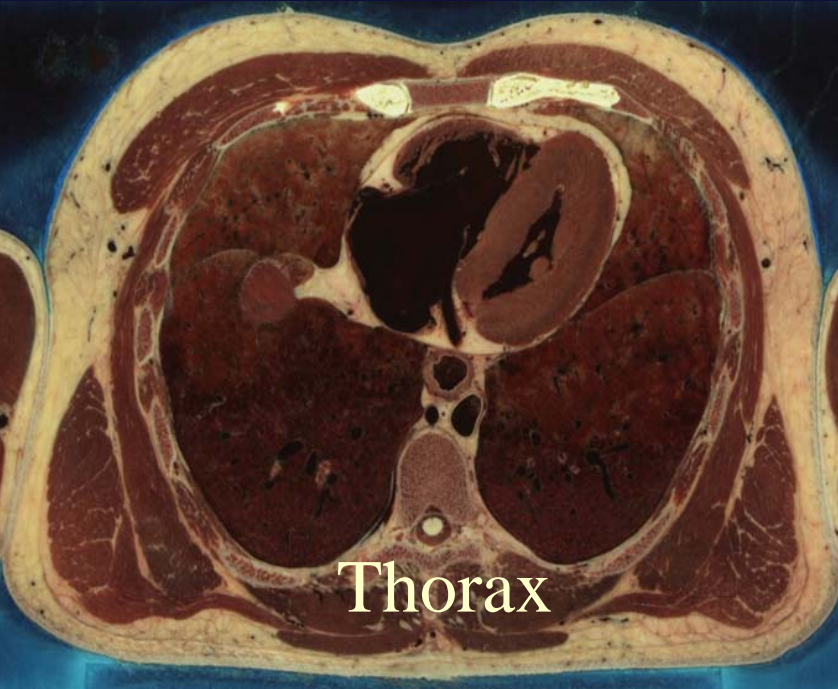
comprises anatomical data

pertaining to *instances*

of organisms and their parts

deals with *individuals* and anatomical data.

Idealized Reality: Universals

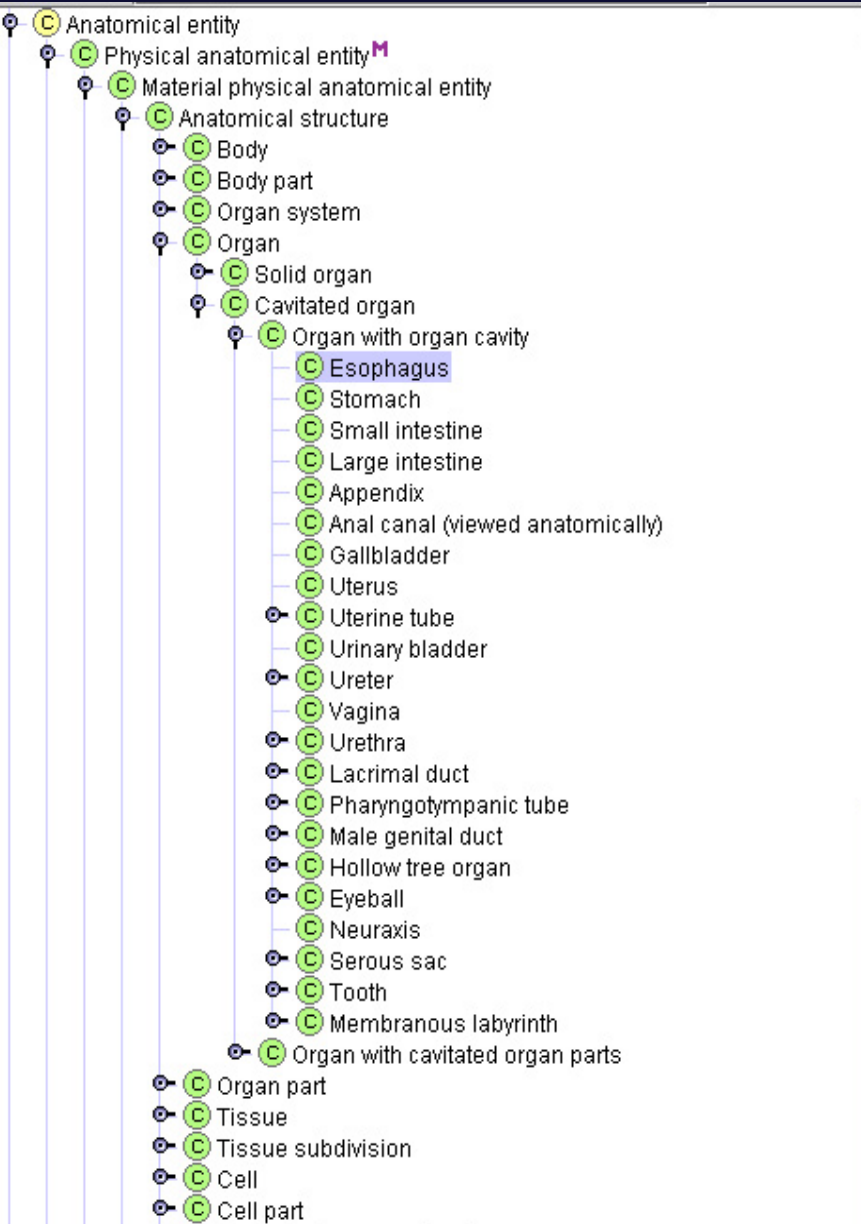


Idealized Reality: Universals

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Gray's Anatomy, 38th edition, p. 1751

Idealized Reality: Classes



Preferred Name **UWDAID**

Synonyms **Non-English Equivalents**

Definition
 Organ with organ cavity which is continuous proximally with the pharynx and distally with the stomach.
 Examples: There is only one esophagus.

Member Of **Part**

- Wall of esophagus
- Lumen of esophagus
- Cervical part of esophagus
- Thoracic part of esophagus
- Abdominal part of esophagus
- Broncho-esophageus

Part Of

Attributed Part

related part	anatomical/arbitrary	shared/unshared	partition
Wall of esophagus	Anatomical	Unshared	Partition 1
Lumen of esophagus	Anatomical	Unshared	Partition 1
Cervical part of esophagus	Arbitrary	Unshared	Partition 2
Thoracic part of esophagus	Arbitrary	Unshared	Partition 2

Attributed Continuous With

related object	coordinate	laterality
Pharynx	Superior	
Stomach	Inferior	

Orientation

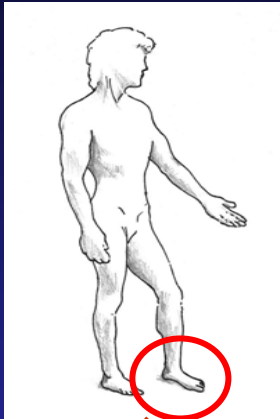
related object	coordinate	laterality
Plane of pharyngoesophageal junction	Superior	
Plane of esophagogastric junction	Inferior	

Foundational Principles of Anatomy

Species Specificity Principle:

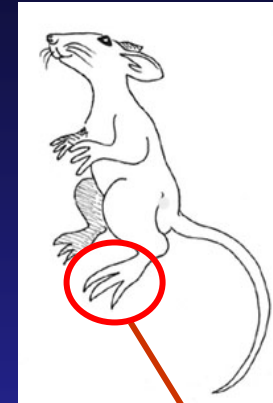
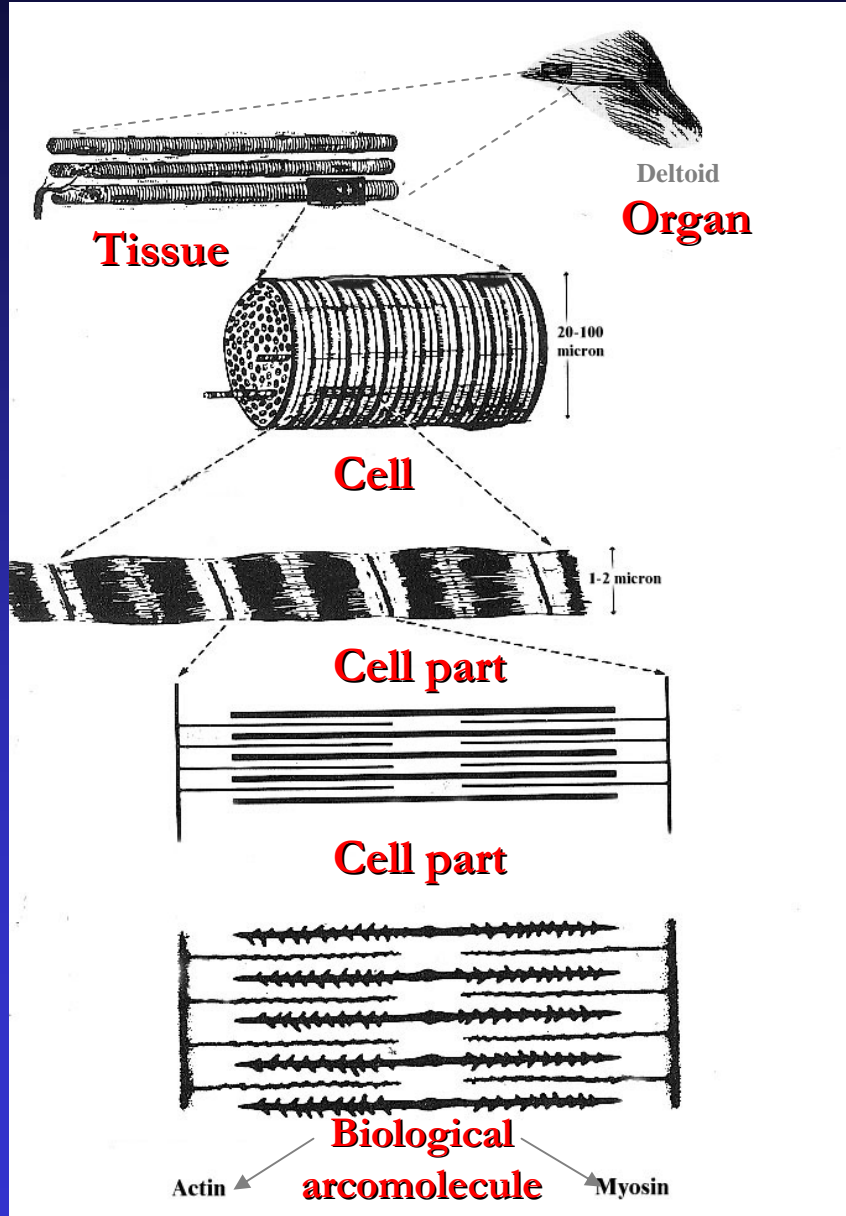
The initial iteration of the abstraction should represent the anatomy of *Homo sapiens*, but at the same time it should serve as a framework for the anatomy of other mammalian and eventually vertebrate species.

Species Specificity Principle



Organism

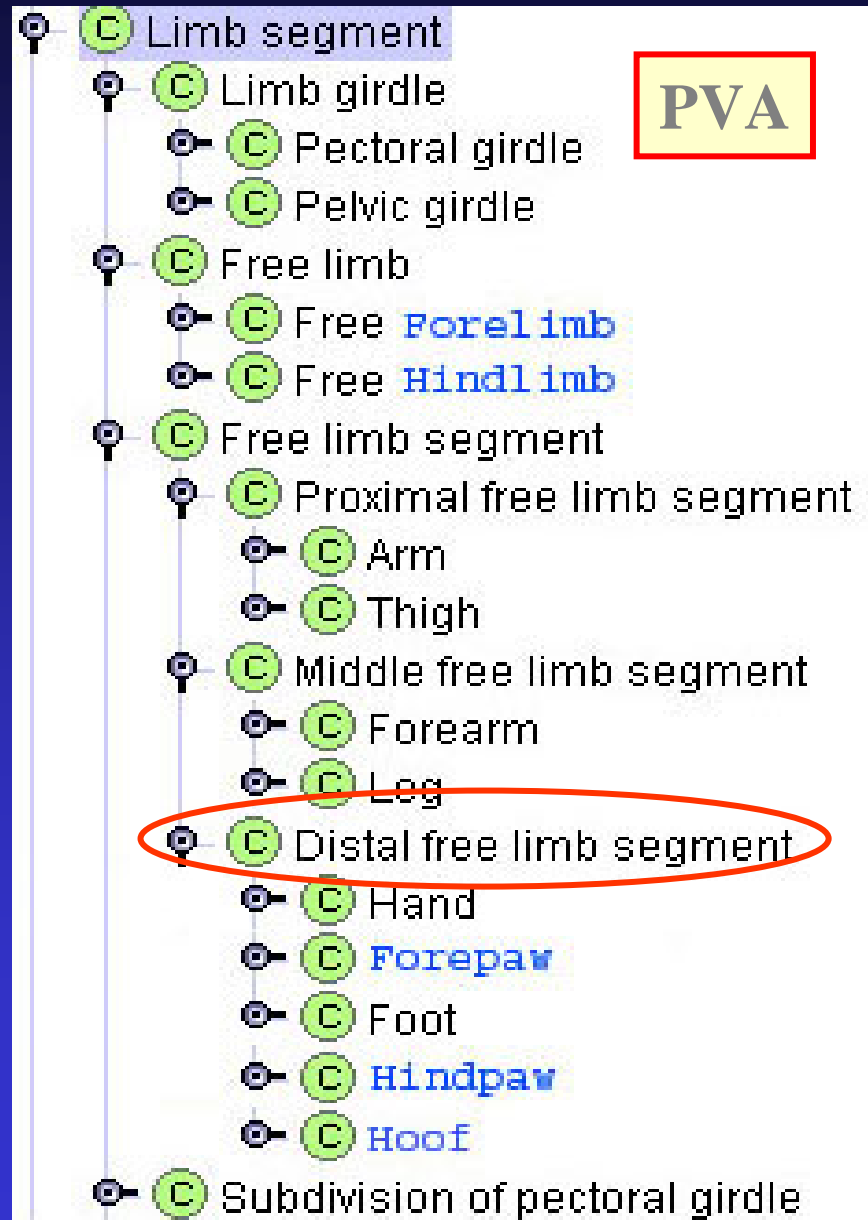
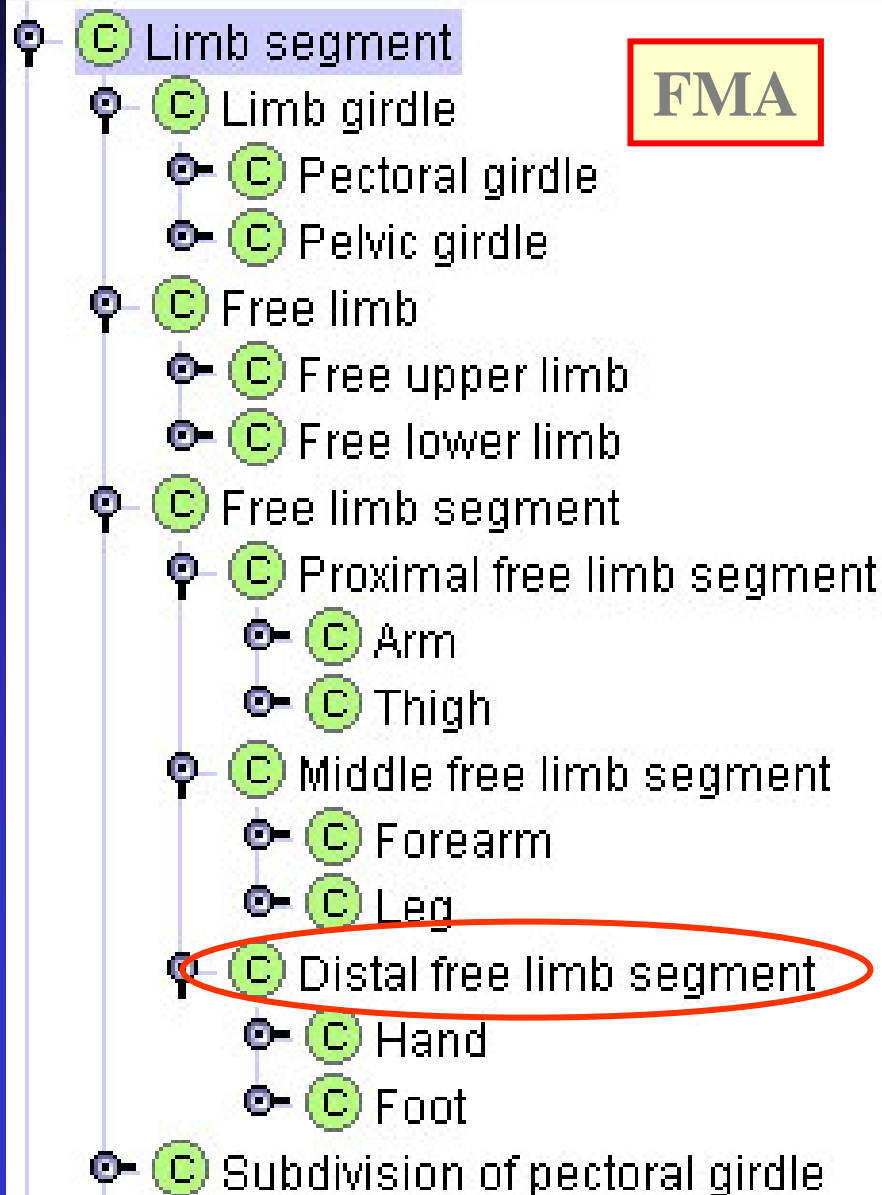
Body part:
Foot



Organism

Body part:
Hindpaw

Species Specificity Principle



Foundational Principles of Anatomy

Definition Principle:

Defining attributes of anatomical entities should be specified in terms of physical attributes and structural relationships of the anatomical entity

Role of Definitions

- To specify a unique taxonomy
- To assure semantic and logical consistency within the ontology
- To assure transitive inheritance of properties through type hierarchy (taxonomy)

Michael J, Mejino JLV, Rosse C.
The role of definitions in biomedical
concept representation.
Procs AMIA Fall Symp 2001; 463-467.

What constitutes Aristotelian definitions?

- An entity is defined by:
 - Class to which it belongs (*genus*)
 - Essential attribute(s) distinguishing it from members of the same class (*differentiae*)
- *Essence* of an entity is the sum of:
 - Properties it inherits transitively from parent classes
 - Attributes assigned to it

Definition Principle

Lung

MSH2001|

Either of the pair of organs occupying the cavity of the thorax that effect the aeration of the blood.

Where should we insert “Lung” in an anatomy taxonomy?

Definition Principle

Lung

MSH2001|

Either of the pair of organs occupying the cavity of the thorax that effect the aeration of the blood.

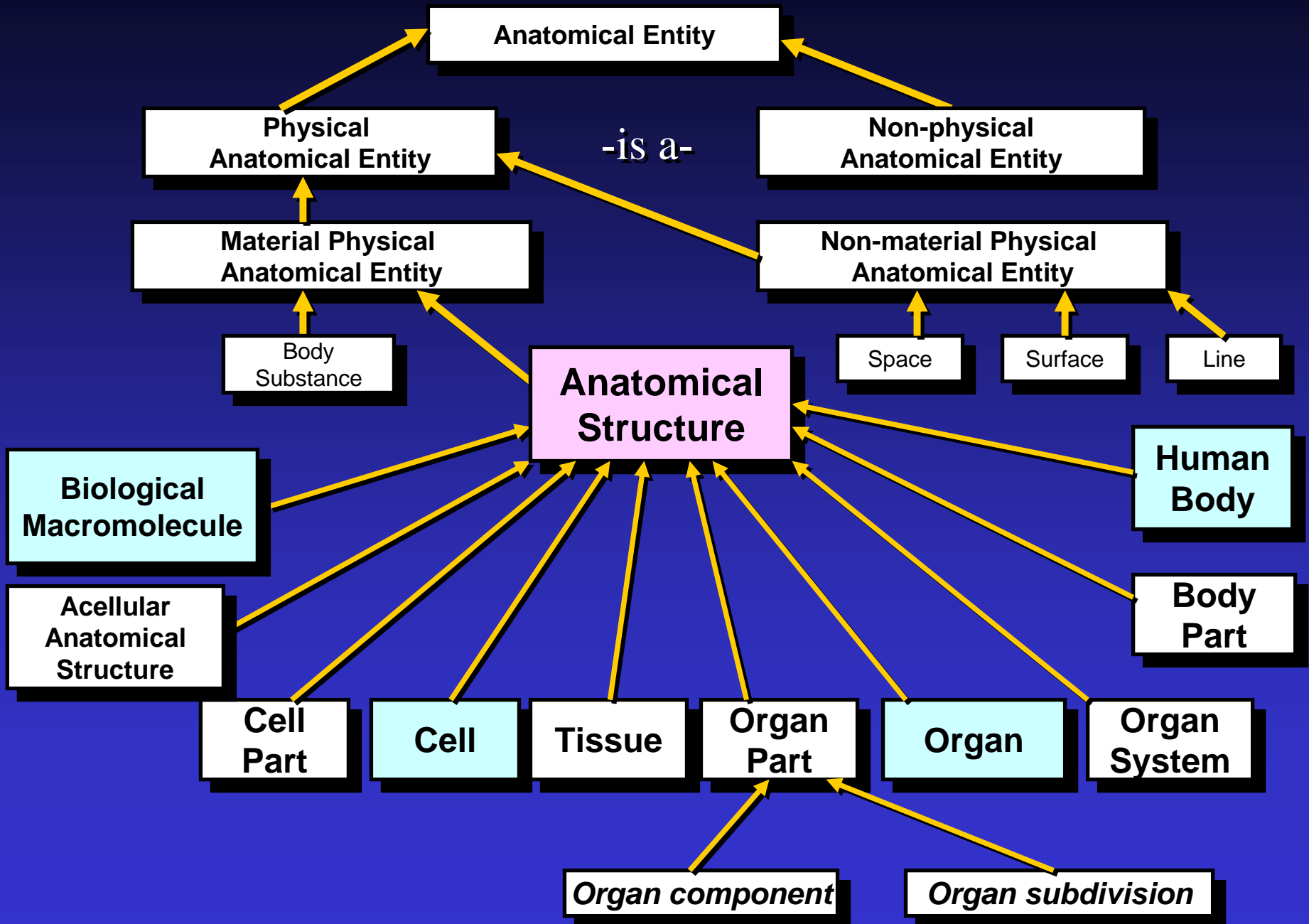
FMA

Lobular organ the parenchyma of which consists of air-filled alveoli which communicate with the tracheobronchial tree.

Foundational Principles of Anatomy

1. Unified context principle
2. Abstraction level principle
3. Species specificity principle
4. Definition principle
5. Dominant concept principle
6. Organizational unit principle
7. Content constraint principle
8. Relationship constraint principle
9. Coherence principle
10. Representation principle

Dominant Concept and Organizational Unit Principles



What kinds of principles?

- Domain-specific principles

Foundational principles for anatomy

(Rosse, Mejino: J Biomed Inform 2003)

- Top-level ontology principles

SNAP, SPAN

(Grenon, P and Smith, B., SNAP and SPAN:
Towards Dynamic Spatial Ontology,
Spatial Cognition and Computation 4(1), 69-103, 2004.)

- Implementation principles

(Zhang, Bodenreider: Law and order:
Assessing and enforcing compliance
with ontological modeling principles.
Comp Biol Med 2005; submitted)

Formal (Top-level) Ontologies

Entities

Continuants

Occurrents

Distinguishing criterion (differentia): Time

Formal (Top-level) Ontologies

Entities

Continuants

Entities which endure *in toto* in every instant of time
at which they exist

Occurrents

Dependent entities which do not endure through time;
unfold themselves in successive temporal phases
(e.g., processes, actions)

Formal (Top-level) Ontologies

Entities

Continuants

Entities which endure *in toto* in every instant of time
at which they exist

Independent continuants

objects, components

Dependent continuants

attributes, roles, qualities, functions

Occurrents

Dependent entities which do not endure through time;
unfold themselves in successive temporal phases
(e.g., processes, actions)

1. Temporal coherence

2. Dependence

3. Univocity

4. Compositionality

5. Objectivity

6. Positivity

7. Explicitness

8. Taxonomic Levels

9. Partonomic Levels

10. Single Inheritance

11. Exhaustiveness

Principles of Formal Ontologies

Principle of Temporal Coherence

An ontology should rigorously distinguish

continuants

occurents

Anatomy is a science of continuants

Anatomical structures, attributes, relationships
and functions are all continuants

FMA complies with principle of temporal coherence

(~70,000 classes of FMA are all continuants)

Excludes functions

Principle of Dependence

If an ontology recognizes a dependent entity
[e.g., surface of lung, parenchyma of lung]
then it should also recognize
the class on which the dependent entity depends [e.g., lung]

FMA is largely compliant:

1,980 classes of “Subdivision of ...” “Component of...”
1,809 the class of ‘whole’ exists (91%)

Principle of Single Inheritance

A class in an ontology's taxonomy should have only one parent on the immediate higher level.

FMA's definition principle enforces compliance with the principle of single inheritance:

an entity can have but one essence

Principle	GO	FMA
Temporal coherence	No	N/A
Dependence	No	N/A
Univocity	No	Yes
Compositionality	No	Yes
Objectivity	No	Yes
Positivity	No	Yes
Explicitness	No	N/A
Taxonomic Levels	No	Yes
Partonomic Levels	No	Yes
Single Inheritance	No	Yes
Exhaustiveness	No	No

What kinds of principles?

- Domain-specific principles

Foundational principles for anatomy

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Principle of Non-duplication

An ontology should represent an entity only once and associate with this single entry all terms that point to the referent entity

- Synonyms
- FMA compliance: treatment of synonyms

Number of terms

Preferred names	69,889
Synonyms	40,683
Total terms	110,572

- ◉ Anatomical structure
 - ◉ Body
 - ◉ Organ
 - ◉ Solid organ
 - ◉ Cavitated organ
 - ◉ Organ with organ cavity
 - ◉ Esophagus
 - ◉ Stomach
 - ◉ Small intestine
 - ◉ Large intestine
 - ◉ Appendix
 - ◉ Anal canal (viewed anatomically)
 - ◉ Gallbladder
 - ◉ Uterus
 - ◉ Uterine tube
 - ◉ Urinary bladder
 - ◉ Ureter
 - ◉ Vagina
 - ◉ Urethra
 - ◉ Lacrimal duct
 - ◉ Pharyngotympanic tube
 - ◉ Male genital duct
 - ◉ Hollow tree organ
 - ◉ Eyeball
 - ◉ Neuraxis
 - ◉ Serous sac
 - ◉ Tooth
 - ◉ Membranous labyrinth
 - ◉ Organ with cavitated organ
 - ◉ Organ part
 - ◉ Tissue

Uterine tube (type=Uterine tube)

Preferred Name UWLDAID

Synonyms

Non-English Equivalents

Definition
 Organ with organ cavity which connects the uterine cavity to the peritoneal cavity.
 Examples: There are only two uterine tubes, the right and the left uterine tubes.

Tuba uterina (type=Concept name, name=su_incus_35616)

Name Authority

Author

Modified By Modification

Date entered/modified Term Status

Language Eponym UMLS ID

Outdated Meaning

FMA Relationship Types

Hierarchical relationships	8
Nonhierarchical	
associative relationships	74
Inverse relationships	38
Atomic properties	121

Instantiation of Relationships

-IS A-	1,105,164
-PART-OF-	972,612
Double assignment	300

Conclusions

Questions proposed for discussion

- Is an anatomy standard a matter of standardized terminology, or of something else?
- Do we need a standard for anatomy as a whole, or just for some (foundational) parts?
- How big should a standard be?
- Should it include for example histology?
- How does a reference ontology for anatomy relate to anatomy standard?

Conclusions

FMA's role for Anatomy Standard

- Proposes a unifying theory of anatomy
- Implements the theory in a rigorous and consistent ontology
conforms to three sets of ontological principles
- Incorporates *Terminologia Anatomica*
international standard for anatomy terminology
- Encompasses in one continuous information space
anatomical entities from the level of
molecules to the whole organism
- Conceptualizes anatomy in a strictly structural context
that generalizes to all views and application domains
- Serves as a template for reference ontologies
in other basic science domains

FMA's Role Anatomy Standard

Should help to answer:

Questions proposed for discussion

- What do we mean by a standard?
- Why do we need a standard?
- What would a CEN standard for anatomy look like?
- Who would use it?
- What sorts of applications should it support?