



Building a Core Ontology in Medicine

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who

where

why



acknowledgements:

Ivano APOLLONI

Massimo BATTAGLIA

Aldo GANGEMI

Nicola GUARINO

Claudio MASOLO

Geri STEVE

... and all the LOAkers



who

where

why



Laboratory of Applied Ontology of CNR-ISTC

www.loa-cnr.it

Ontology Summer School July 2004



who

where

why



some questions

- **is ontology useful in medicine?**
 - ◆ **only in medical research?**
 - ◆ **what about health-care practice?**

- **how would one persuade an “ontoskeptical”?**



**“Have over 35 years of health informatics
made **Europe healthier?** ” (Bryden 2003)**

ontology → informatics

informatics → health informatics

health informatics → Europe healthier



do we need ontology in medicine?

- ◆ **medical terminology**
- ◆ **clinical guideline representation**
- ◆ **Evidence-Based Medicine**



Semantic Mining WP 21

- ◆ **To contribute to a consensus on a bio-medical "upper ontology".**
- ◆ **To contribute to the convergence of clinical and bio-ontologies.**



ROME

Reference Ontology for MEdicine



ROME wasn't built in a day

...



ROME wasn't built in a day

...

1.7.2004 - 31.12.2006



sources for ROME:

- ◆ **ON 9**
- ◆ **GALEN-CORE**
- ◆ **UMLS**
- ◆ **UMLS-SN**
- ◆ **HL7 reference model**



sources for ROME:

- ◆ ON 9
- ◆ GALEN-CORE
- ◆ UMLS
- ◆ UMLS-SN
- ◆ HL7 reference model







proceedings AMIA 1998

**“An Ontological Analysis
of the UMLS MetathesaurusTM”**

Domenico M. Pisanelli, Aldo Gangemi, Geri Steve

Istituto di Tecnologie Biomediche, CNR, Rome, Italy.



Tool for representation

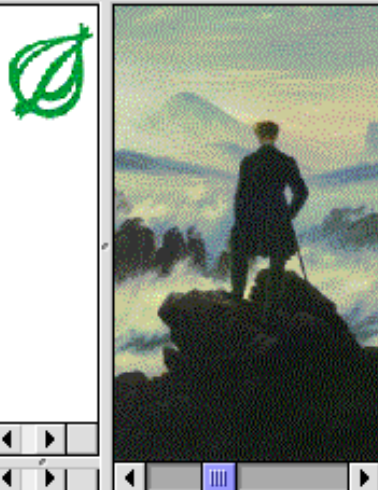
ONTOLINGUA

Tool for representation and classification

LOOM

Tool for browsing and editing

ONTOSAURUS

Location: <http://onto62.irmkant.rm.cnr.it/loom/shuttle.html>

Concept ALGA

Theory: [NATURAL-KINDS](#) Package: LoomSeen from:

[New Concept](#)
[New Relation](#)
[Make Instance](#)
[Edit Concept](#)
[Export Concept](#)
[Delete Concept](#)
[Find Matching Instances](#)
[Bookmark](#)

Documentation

A chiefly aquatic plant that contains chlorophyll, but does not form embryos during development and lacks vascular tissue.

Definition

```
(defconcept ALGA
  :ANNOTATIONS ((DOCUMENTATION "A chiefly aquatic plant that contains chlorophyll, but does not
embryos during development and lacks vascular tissue."))
  :ANNOTATIONS ((PROBLEM "(1) needs a special axiomatization: it involves the localization in a
region-type, the biological necessity, the notion of life, and an interactional notion.
(2) with substances should be interpreted as a mass-componency; see theory:meronymy"))
  :CONTEXT :NATURAL-KINDS
  :IS-PRIMITIVE (:AND PLANT
    (:SOME LIVES-IN
      WATER)
    (:SOME HAS-COMPONENT
      CHLOROPHYLL)
    (:SOME HAS-DEVELOPMENTAL-FORM
      (:AND BIOLOGIC-OBJECT
        (:NOT EMBRYONIC-STRUCTURE)))
    (:SOME HAS-COMPONENT
      (:AND BIOLOGIC-OBJECT
```

Update

Reload





The ontologization of the Metathesaurus

what we represented:

- ◆ the preferred names of the CUIs (e.g. "Fibromyalgia");
- ◆ the instances of PAR (=“has_parent”) relations between different CUIs that UMLS mutated from its sources (e.g. "Fibromyalgia" PAR "Muscular-diseases");
- ◆ the relations between CUIs that UMLS imported from sources (e.g. " COPAD protocol" USES "Asparaginase");
- ◆ the instances of PAR relations between a CUI and its "semantic types" (e.g. "Fibromyalgia" PAR "Disease-or-Syndrome");
- ◆ the definition of the CUIs in plain text, as reported in authoritative sources such as medical dictionaries.



what should not be given for granted:

**all instances of PAR relation
are equivalent to genuine IS_A**



some instances of **PAR** relation

Ocular Orbit	C0029180	PAR	Eye	C0015392
Cranial Cavity	C0230041	PAR	Head	C0018670
Hip Joint	C0019558	PAR	Hip	C0019552
Dental Pulp Cavity	C0011403	PAR	Tooth	C0040426
2-Aminopurine	C0000263	PAR	Adenine	C0001407
Adenine	C0001407	PAR	2-Aminopurine	C0000263



some instances of **PAR** relation

Ocular Orbit	C0029180	PAR	Eye	C0015392
Cranial Cavity	C0230041	PAR	Head	C0018670
Hip Joint	C0019558	PAR	Hip	C0019552
Dental Pulp Cavity	C0011403	PAR	Tooth	C0040426

2-Aminopurine	C0000263	PAR	Adenine	C0001407
Adenine	C0001407	PAR	2-Aminopurine	C0000263

MeSH



some instances of **PAR** relation

Ocular Orbit	C0029180	PAR	Eye	C0015392
Cranial Cavity	C0230041	PAR	Head	C0018670
Hip Joint	C0019558	PAR	Hip	C0019552
Dental Pulp Cavity	C0011403	PAR	Tooth	C0040426

2-Aminopurine C0000263 **PAR** Adenine C0001407
Adenine C0001407 **PAR** 2-Aminopurine C0000263

MeSH

CRISP



the true story:



the true story:

Adenine = 6-Aminopurine



2-Aminopurine



Reference Ontology for MEdicine

version 0.1

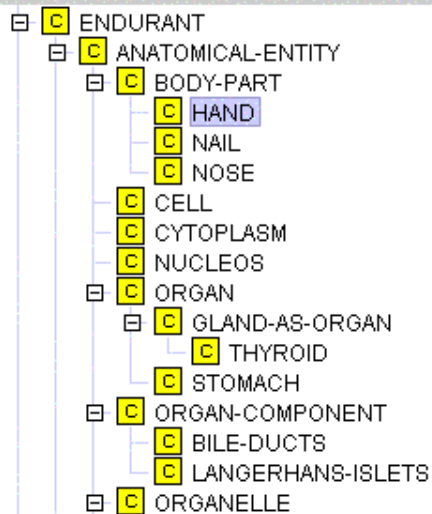
about 100 concepts

- developed in RACER v. 1-17-20
- can be exported in DAML+OIL, OWL
- can be browsed by Rice, OilEd, Protégé



Class Hierarchy

Hierarchy



Supers

C BODY-PART

Done

Name

HAND

Properties

- SubclassOf
- SameClassAs

Documentation

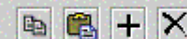
Classes

BODY-PART



Restrictions

type	property	filler



C:\RACER\oiled\U-15-oiled.daml



Relationship Superclass

- :THING A
- :SYSTEM-CLASS A
- XMLSchema:anyType
- Ontology
- daml_oil:Thing
- BODY-SYSTEM
 - ENTITY
 - BODY-JUNCTION
 - BODY-LOCATION-OR-REGION
 - BODY-SPACE
 - BODY-SPACE-AND-JUNCTION
 - BODY-SUBSTANCE
 - ENDURANT
 - ANATOMICAL-ENTITY
 - BODY-PART
 - HAND
 - NAIL
 - NOSE
 - CELL
 - CYTOPLASM
 - NUCLEOS
 - ORGAN
 - GLAND-AS-ORGAN
 - THYROID
 - STOMACH
 - ORGAN-COMPONENT
 - BILE-DUCTS
 - LANGERHANS-ISLETS
 - ORGANELLE
 - TISSUE

- Superclasses
- ANATOMICAL-ENTITY

ORGAN-COMPONENT (type=Class)

Name: ORGAN-COMPONENT

comment: []

damlProperties

Name	Type	Cardinality	Other Facets
S :NAME	String	single	
S :DOCUMENTATION	String	multiple	
S 1	Instance	multiple	classes={:THING}
S 2	Instance	multiple	classes={:THING}
S 3	Instance	multiple	classes={:THING}
S 4	Instance	multiple	classes={:THING}
S 5	Instance	multiple	classes={:THING}
S 6	Instance	multiple	classes={:THING}

Restriction

onProperty	type	value
5	hasClass	ORGAN

QualifiedRestriction

onProperty	hasClassQ	type	value

Logicaldefinition

type	value



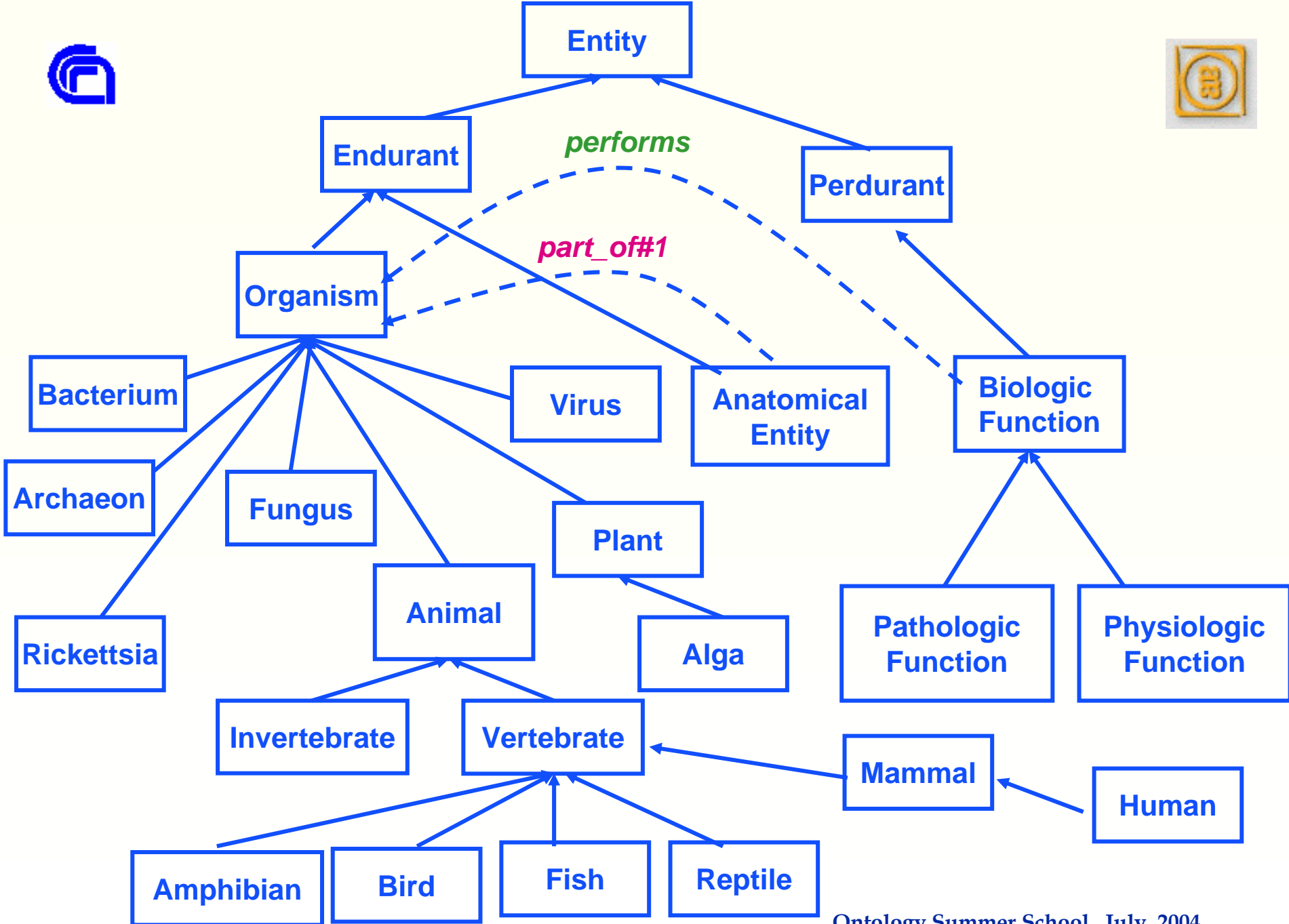
issues

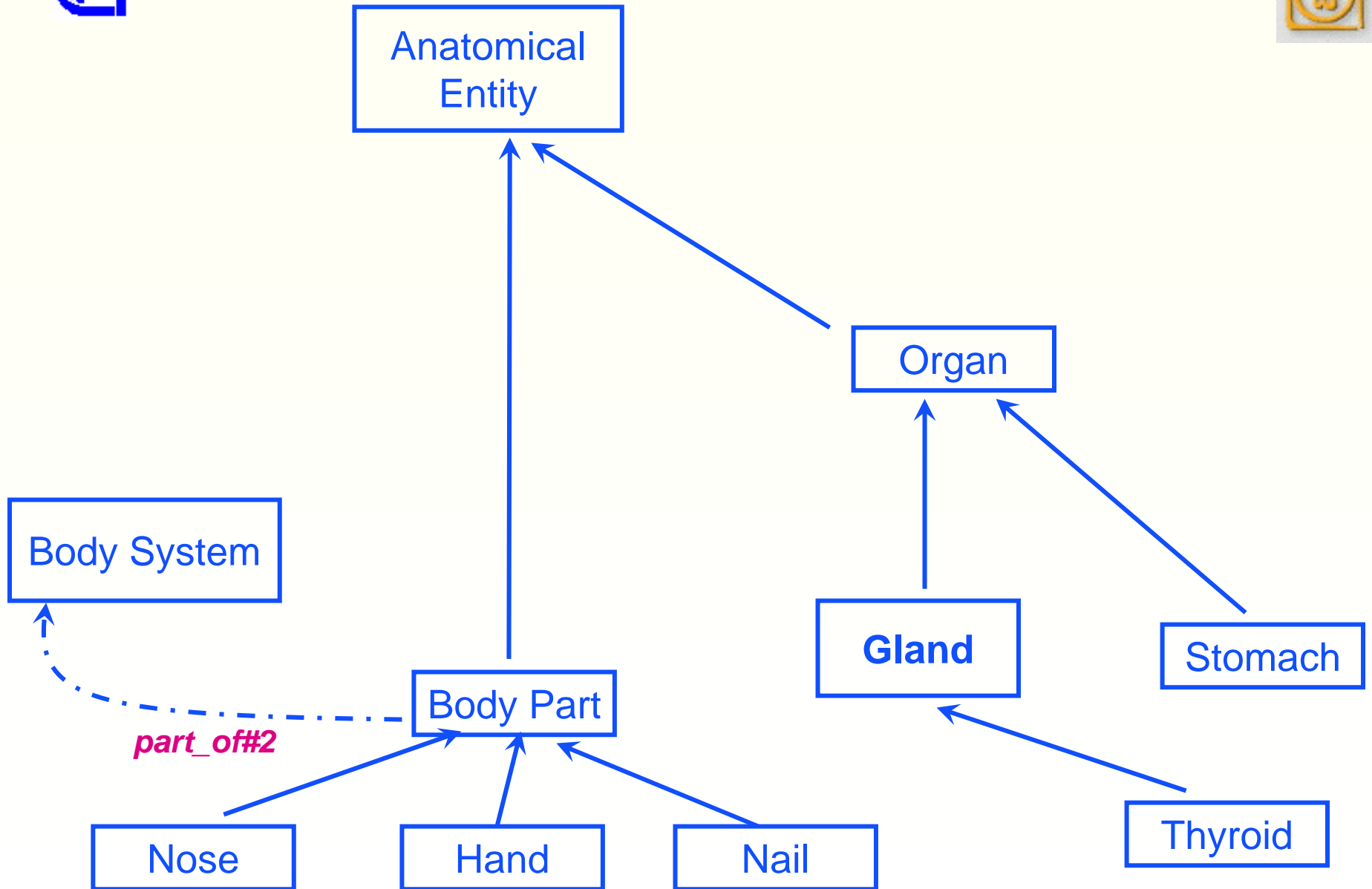
- ◆ mereology
- ◆ “holes and other superficialities”
- ◆ are there such things as “instances of models”?

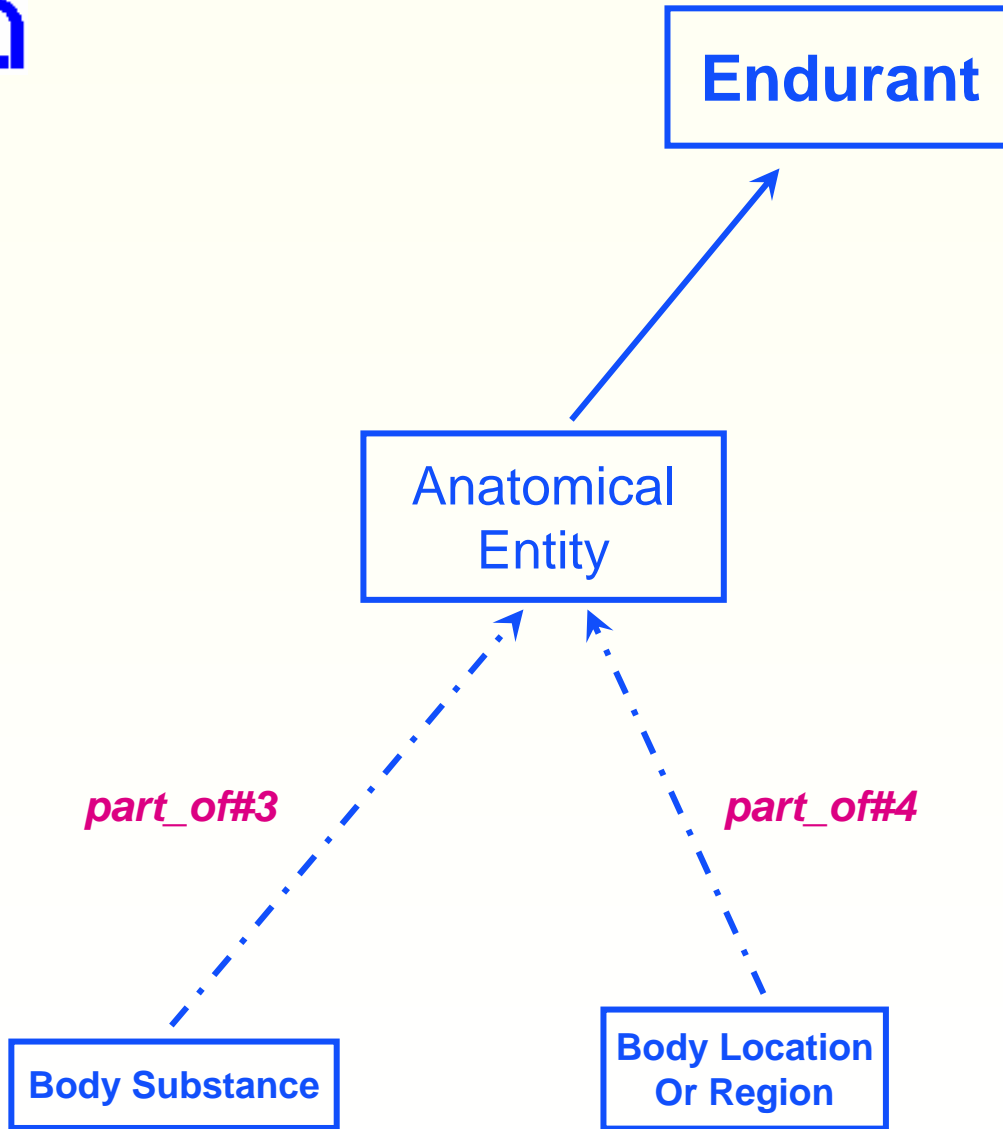


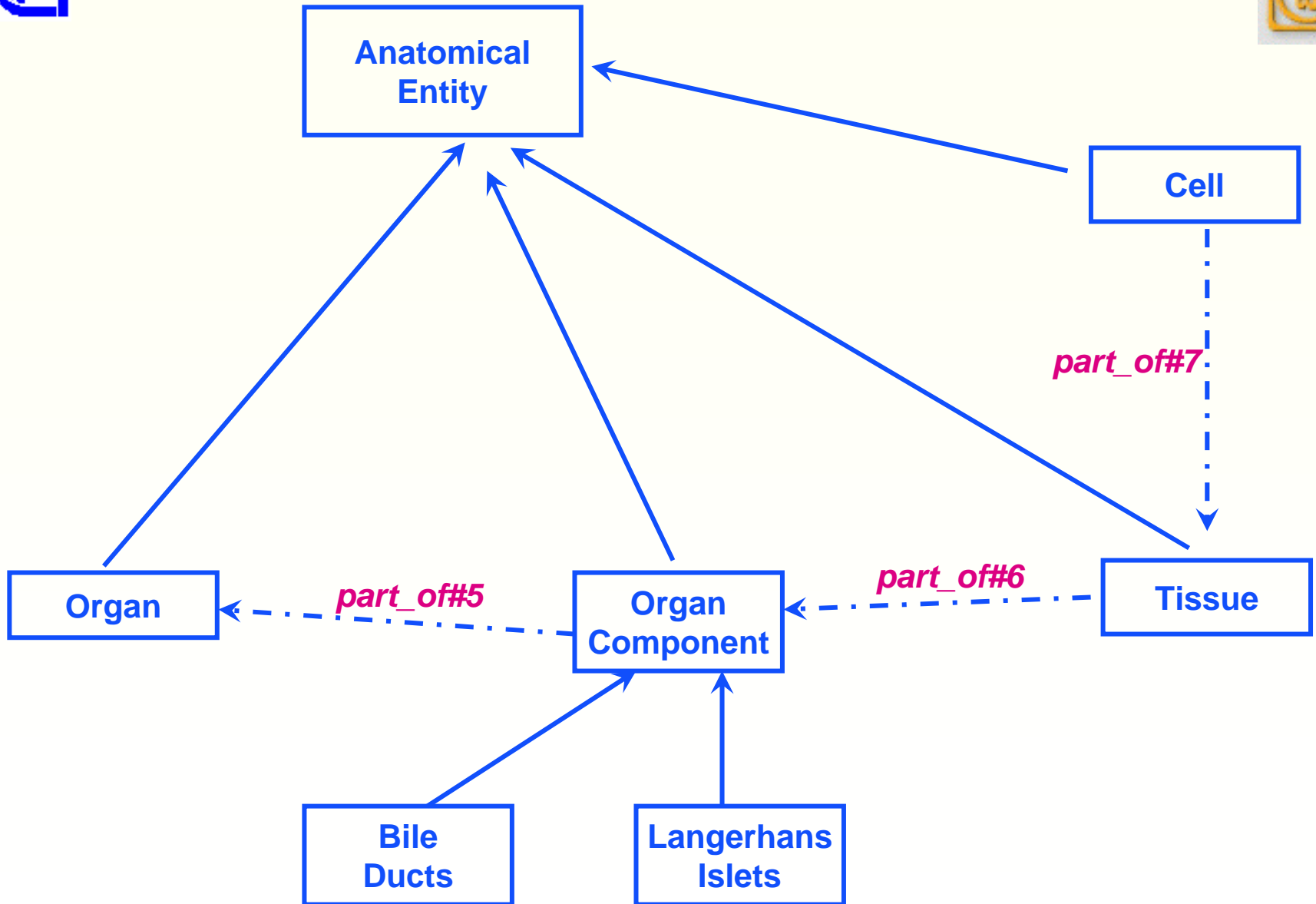
issues

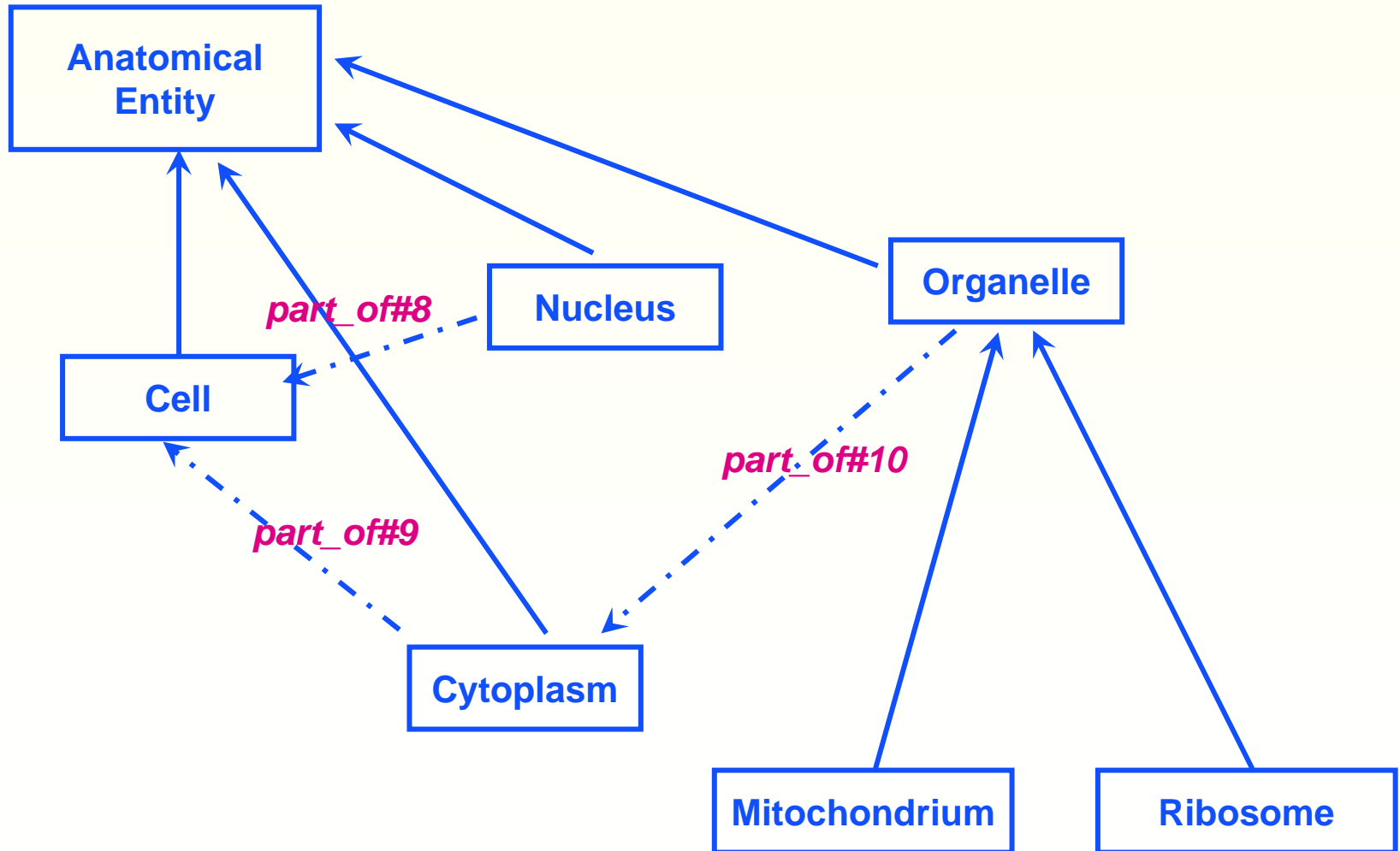
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issues

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issues

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**Body Space
or Junction**

Body Space

Body Junction

**Body Space
and Junction**

MS Sans Serif 10 G C S % . 100%

E6467 =

bodySpJncPadriFigli.xls

	A	B	C	D	E
6431	C1184665	Functional joint type	C1184669	Saddle joint	
6432	C1184665	Functional joint type	C1184670	Ellipsoid joint	
6433	C1184665	Functional joint type	C1184671	Cotyloid joint	
6434	C1184709	Set of canals	C0231099	Set of incisive canals	
6435	C1184709	Set of canals	C1184710	Set of diploic canals	
6436	C1184709	Set of canals	C1184711	Set of alveolar canals of maxilla	
6437	C1184709	Set of canals	C1184712	Set of lesser palatine canals	
6438	C1184709	Set of canals	C1184713	Set of longitudinal canals of modiolus cochleae	
6439	C1184839	Set of anatomical features	C1184840	Set of recesses, fossae and folds	
6440	C1184842	Osseous cochlear canal	C1184843	Right osseous cochlear canal	
6441	C1184842	Osseous cochlear canal	C1184844	Left osseous cochlear canal	
6442	C1184890	Subdivision of surface of occipital bone	C1184892	Clivus of occipital bone	
6443	C1185636	Set of recesses of tympanic membrane	C0229360	Anterior recess of tympanic membrane	
6444	C1185636	Set of recesses of tympanic membrane	C0229361	Superior recess of tympanic membrane	
6445	C1185636	Set of recesses of tympanic membrane	C0229362	Posterior recess of tympanic membrane	
6446	C1185742	Commissure	C0225942	Commissure of pulmonary valve	
6447	C1185742	Commissure	C0225954	Commissure of mitral valve	
6448	C1185742	Commissure	C0225964	Commissure of aortic valve	
6449	C1185742	Commissure	C0935053	Commissure of labium majus	
6450	C1185742	Commissure	C1186000	Labial commissure of mouth	
6451	C1186200	Subdivision of epitympanic recess	C1186201	Epitympanic recess proper	
6452	C1186200	Subdivision of epitympanic recess	C1186223	Cupular part of epitympanic recess	
6453	C1186257	Spiral sulcus	C1186300	Inner spiral sulcus	
6454	C1186257	Spiral sulcus	C1186301	Outer spiral sulcus	
6455	C1186729	Cavity of endoplasmic reticulum vesicle	C1186730	Cavity of smooth endoplasmic reticulum vesicle	
6456	C1186729	Cavity of endoplasmic reticulum vesicle	C1186731	Cavity of rough endoplasmic reticulum vesicle	
6457	C1187007	Anatomical line of skin	C1180284	Wrinkle line of skin	
6458	C1187007	Anatomical line of skin	C1180285	Flexure line of skin	
6459	C1254133	Lumen of main bronchus	C0225609	Lumen of right main bronchus	
6460	C1254133	Lumen of main bronchus	C0225631	Lumen of left main bronchus	
6461	C1256475	Inferior division bronchus	C1183962	Lumen of inferior division bronchus	
6462					
6463					
6464					

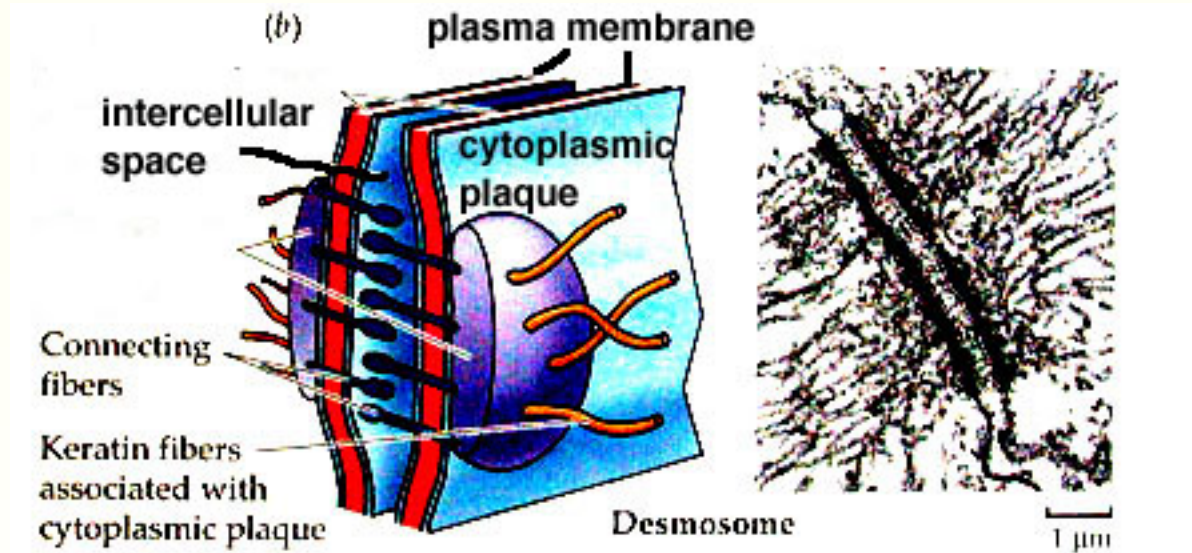


some examples of microscopic junctions

Desmosomes	C0011703
Synapses	C0039062



Desmosomes



Desmosomes tightly link adjacent cells but permit materials to move around them in the intercellular space. Anchored in dense plaques, cell adhesion proteins cross the intercellular space, binding adjacent cells together. Keratin fibers extend through the cytoplasm from one plaque to another.

<http://academic.brooklyn.cuny.edu>

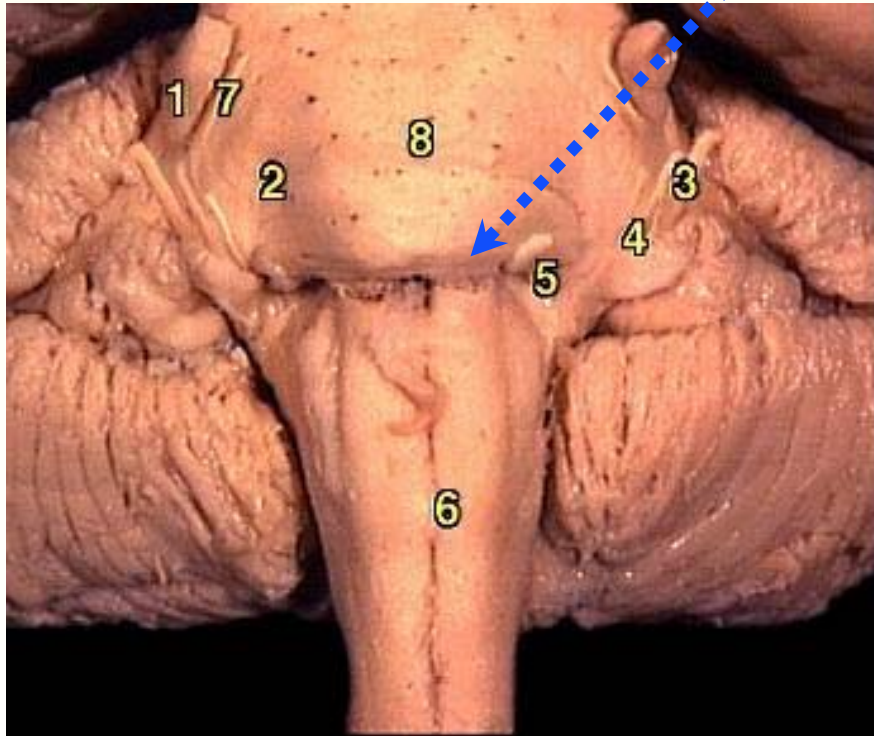


some examples of macroscopic junctions

Cerebellopontine Angle	C0007764
Hip Joint	C0195582
Elbow Joint	C0013770



Cerebellopontine Angle



Anatomy of the cerebellopontine angle showing the relationship of the various cranial nerves.

1. Sensory root of trigeminal nerve
2. Pons
3. Vestibulocochlear nerve
4. Facial nerve
5. Abducent nerve
6. Medulla oblongata
7. Motor root of trigeminal nerve
8. Basilar sulcus

<http://www.mrcophth.com>



some examples of microscopic spaces

Cavity of endoplasmic reticulum vesicle
Cavity of lysosome

C1186729
C1179902

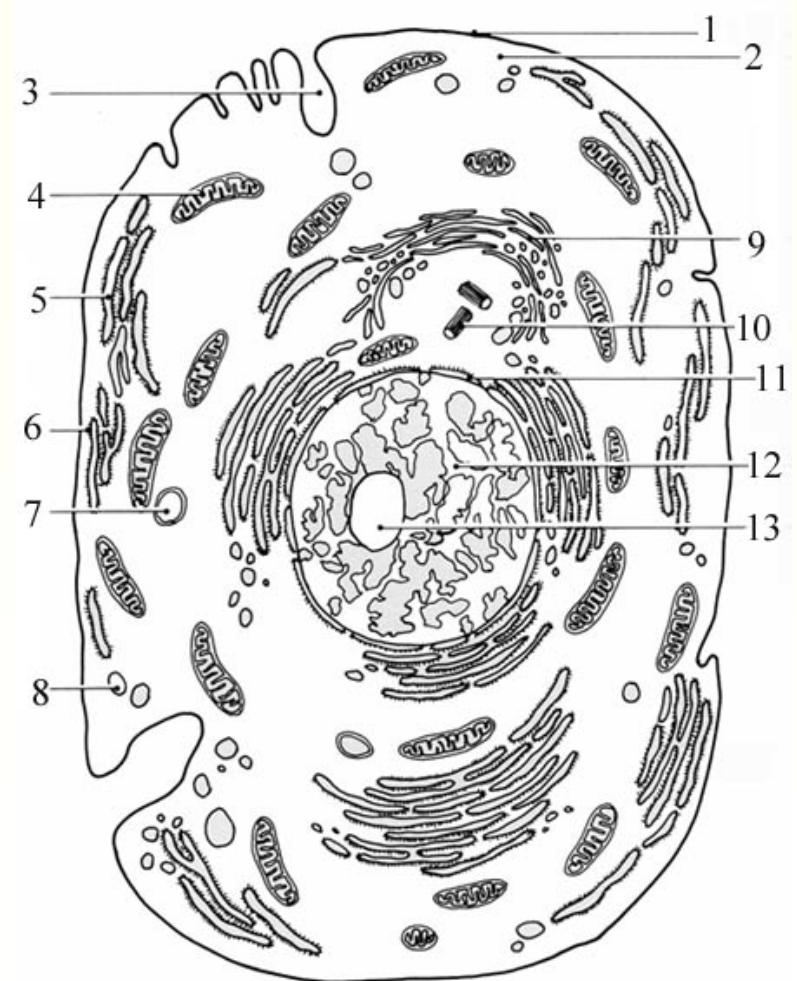


**Cavity of endoplasmic
reticulum vesicle**

5

Cavity of lysosome

7



<http://www.sci.port.ac.uk>

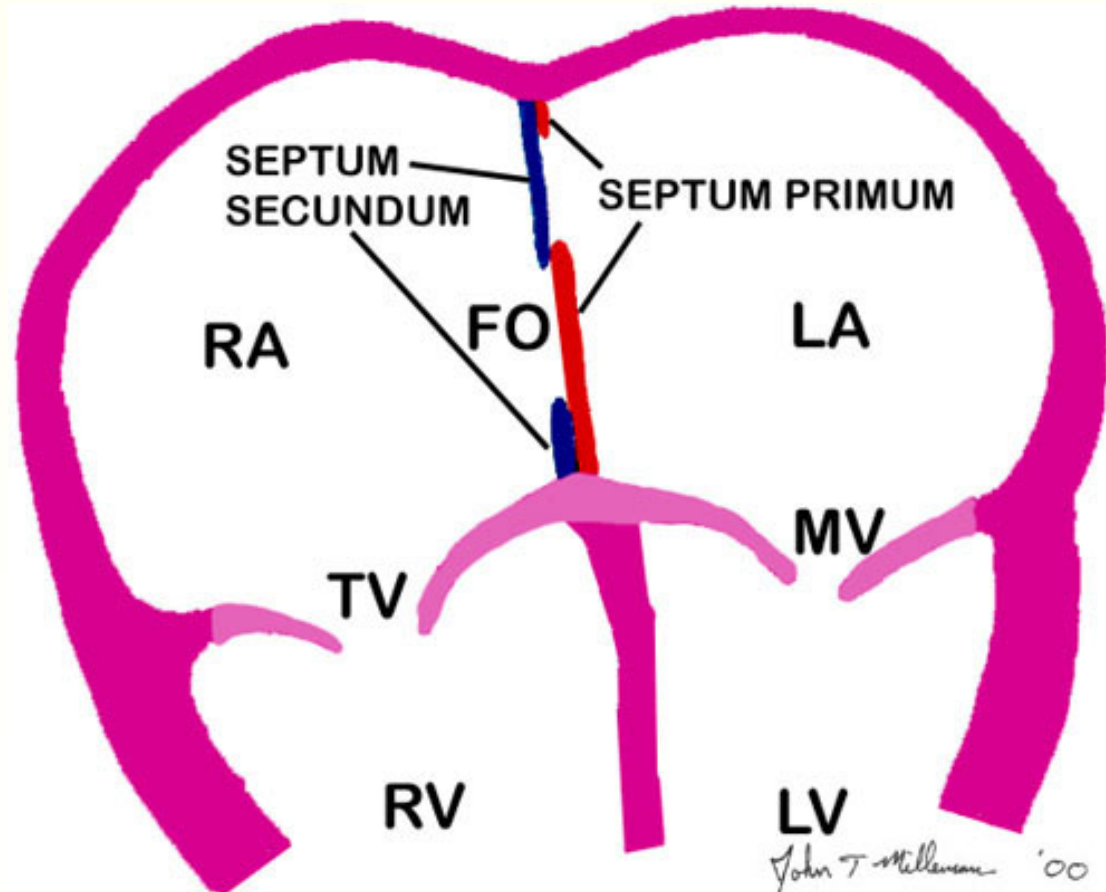


some examples of macroscopic spaces

Cavity of left atrium	C0503989
Cavity of incisor tooth	C0927607



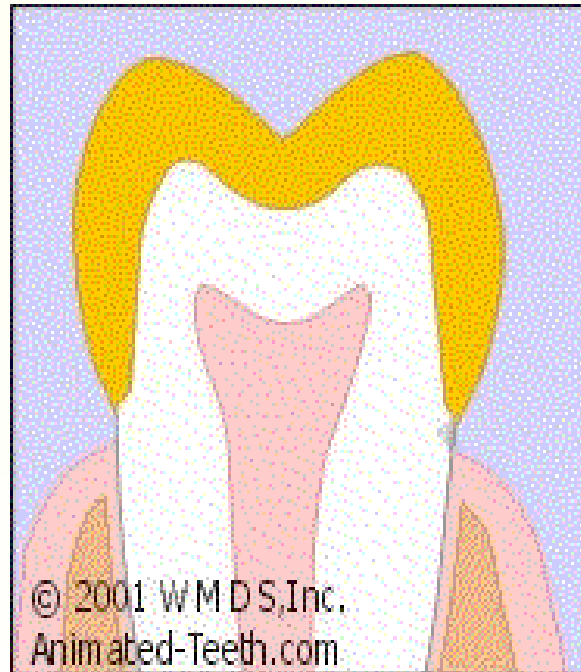
Cavity of left atrium



<http://www.vh.org>



Cavity of incisor tooth



<http://animated-teeth.com>

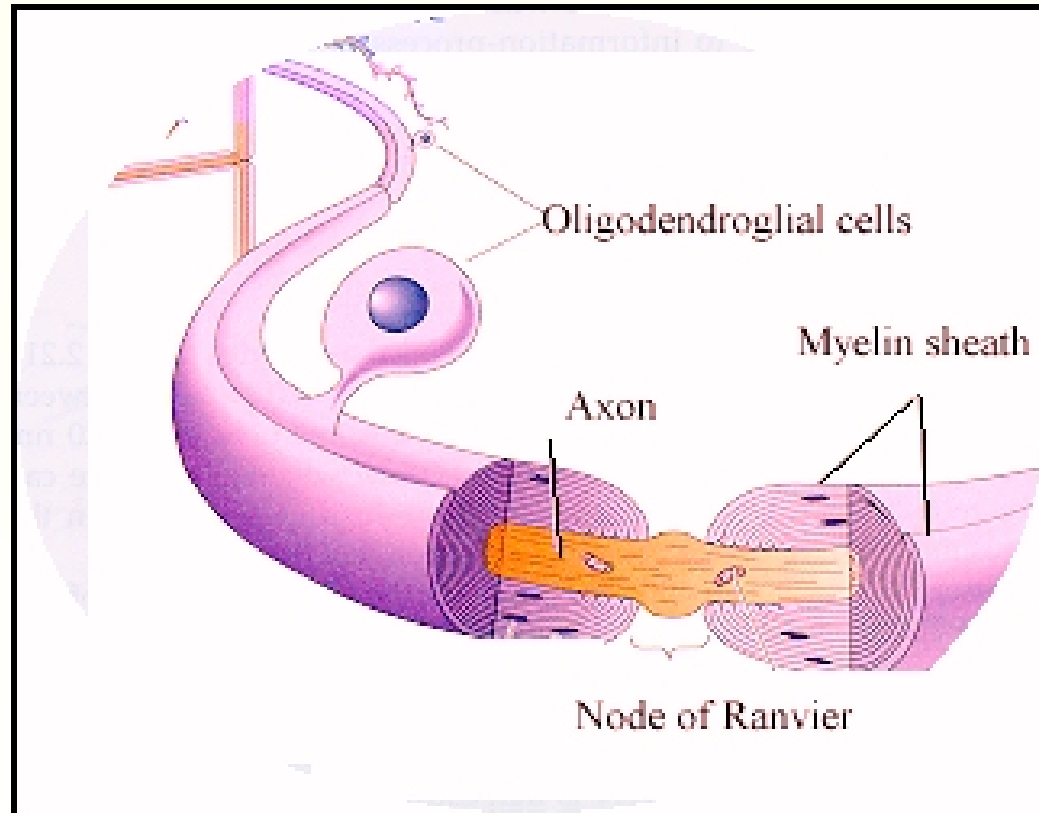


some examples of “space and junctions”

Ranvier's Nodes	C0034667
Lumen of esophagus	C0227194



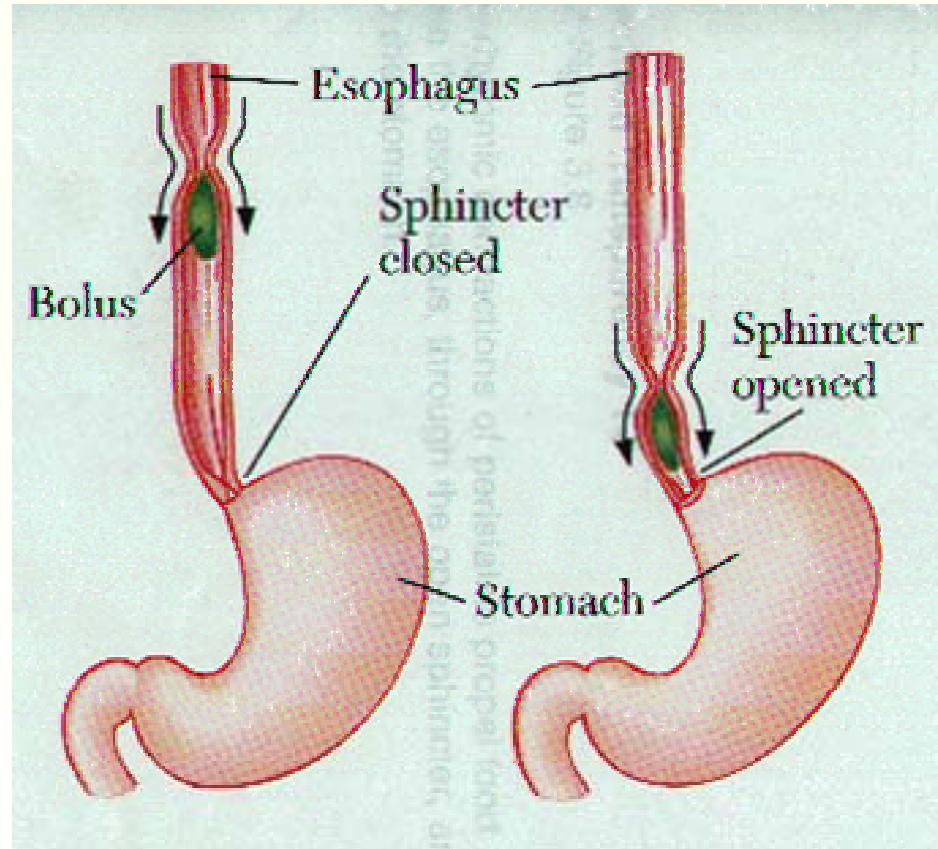
Ranvier's Nodes



<http://aids.hallym.ac.kr>



Lumen of esophagus



<http://www.health.utah.edu>



issues

- ◆ mereology
- ◆ “holes and other superficialities”
- ◆ are there such things as “instances of models”?



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*This is not a pipe
try to smoke it !*

© René Magritte



the ambiguity of everyday language:

“We have the same car”

“We went to the same school”

“We read the same book”



the ambiguity of everyday language:

“We have the **same** car”

“We went to the **same** school”

“We read the **same** book”



**ontology must eliminate
the ambiguity of everyday language:**

“We have the **same car”**

“We went to the **same school”**

“We read the **same book”**



Medical Dictionary

The latest word on medical terms

4 Words Found.

To view an entry in the list, highlight it and click on GO TO.

diagnosis
diagnosis related group
differential diagnosis
DRG

Go To

Main Entry: di·ag·no·sis

Pronunciation: "dI-i g- 'nO-s&s, -&g-

Function: *noun*

Inflected Form(s): *plural di·ag·no·ses /- "sEz/*

1 a : the art or act of identifying a disease from its signs and symptoms

b : the decision reached by diagnosis <a *diagnosis* of pneumonia>

2 : a concise technical description of a taxon



diagnosis

judgment

**e.g. “my presenile dementia as
diagnosed by dr.Dobbs on
July 8th 2003”**

taxon

**e.g. “presenile dementia”
(290.1 ICD-9-CM)**



diagnosis

judgment

e.g. “my presenile dementia as diagnosed by dr.Dobbs on July 8th 2003”

taxon

e.g. “presenile dementia”
(290.1 ICD-9-CM)

process

e.g. the investigation process carried out by dr.Dobbs



diagnosis

judgment

non-physical endurant

taxon

non-physical endurant

process

perdurant



conclusions

**need to converge towards a bio-medical
reference ontology**

**need to address untrivial issues with formal
approaches**

