Foundations of Ontology 1

Barry Smith

http://ifomis.org

The problem

About 30,000 genes in a human

Probably 100-200,000 proteins

Individual variation in most genes

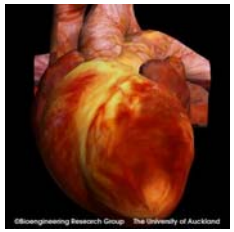
100s of cell types

100,000s of disease types

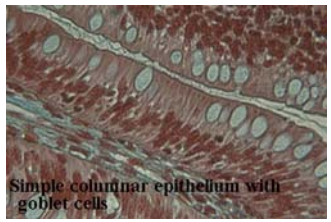
Organism

- Musculo-skeletal system
- Circulatory system
- Respiratory system
- Digestive system
- Nervous system
- Urinary system
- Reproductive system
- Endocrine system
- Lymphoidal system

Organ

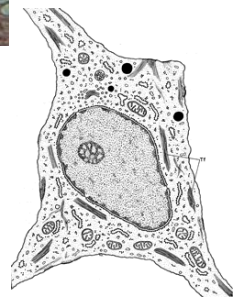


Tissue

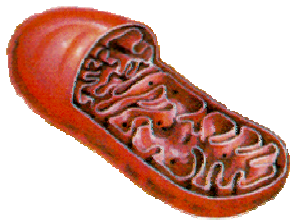


- Muscle tissue
- Nerve tissue
- Connective tissue
- Epithelial tissue
- Blood

Cell

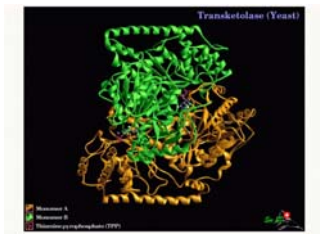


Organelle

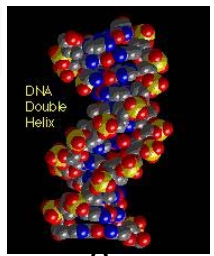


- Mitochondria
- Nucleus
- Endoplasmic reticulum
- Cell membrane

Protein



DNA



The Challenge

Each (clinical, pathological, genetic, proteomic, pharmacological …) information system uses its own terminology and category system

biomedical research demands the ability to navigate through all such information systems

How can we overcome the incompatibilities which become apparent when data from distinct sources is combined?

Answer:

“Ontology”

Three senses of ontology

1. Philosophical sense: an inventory of the types of entities and relations in reality
2. Knowledge engineering sense: an ontology as a consensus representation of the concepts used in a given domain
3. GO/OBO sense: a controlled vocabulary

Ontology as a branch of philosophy

seeks to establish

the basic *formal-ontological structures*

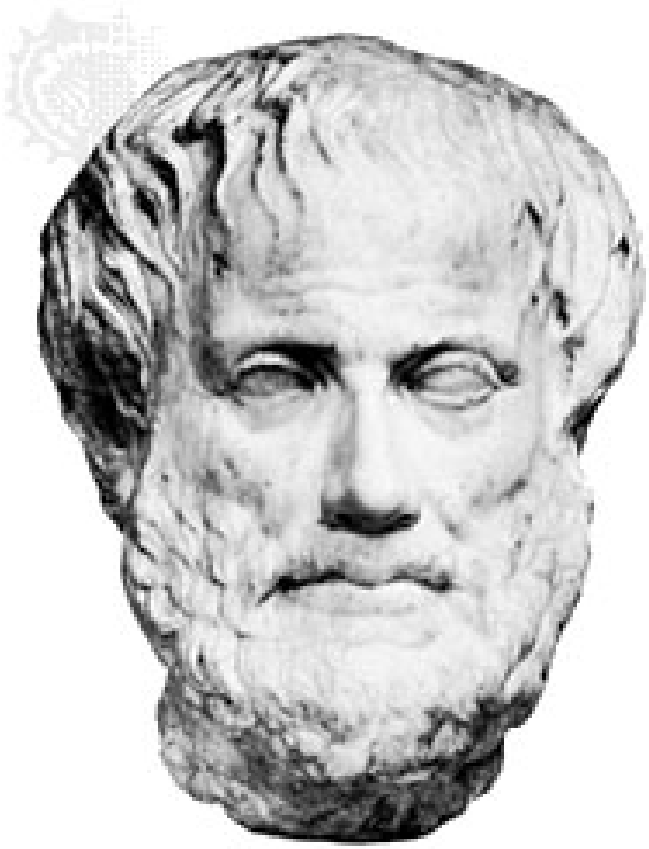
the kinds and structures of objects, properties, events, processes and relations in each *material* domain of reality

Formal ontology an analogue of pure mathematics

Can be *applied* to different domains

Material ontology a kind of generalized chemistry or zoology

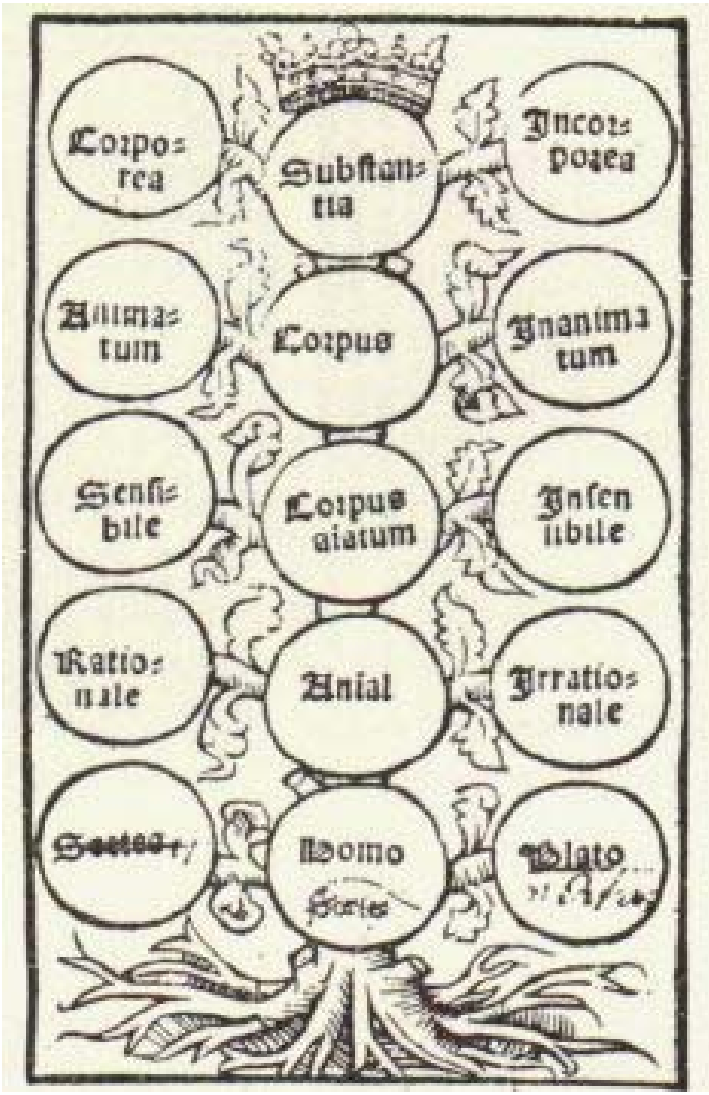
(Aristotle's ontology grew out of biological classification)



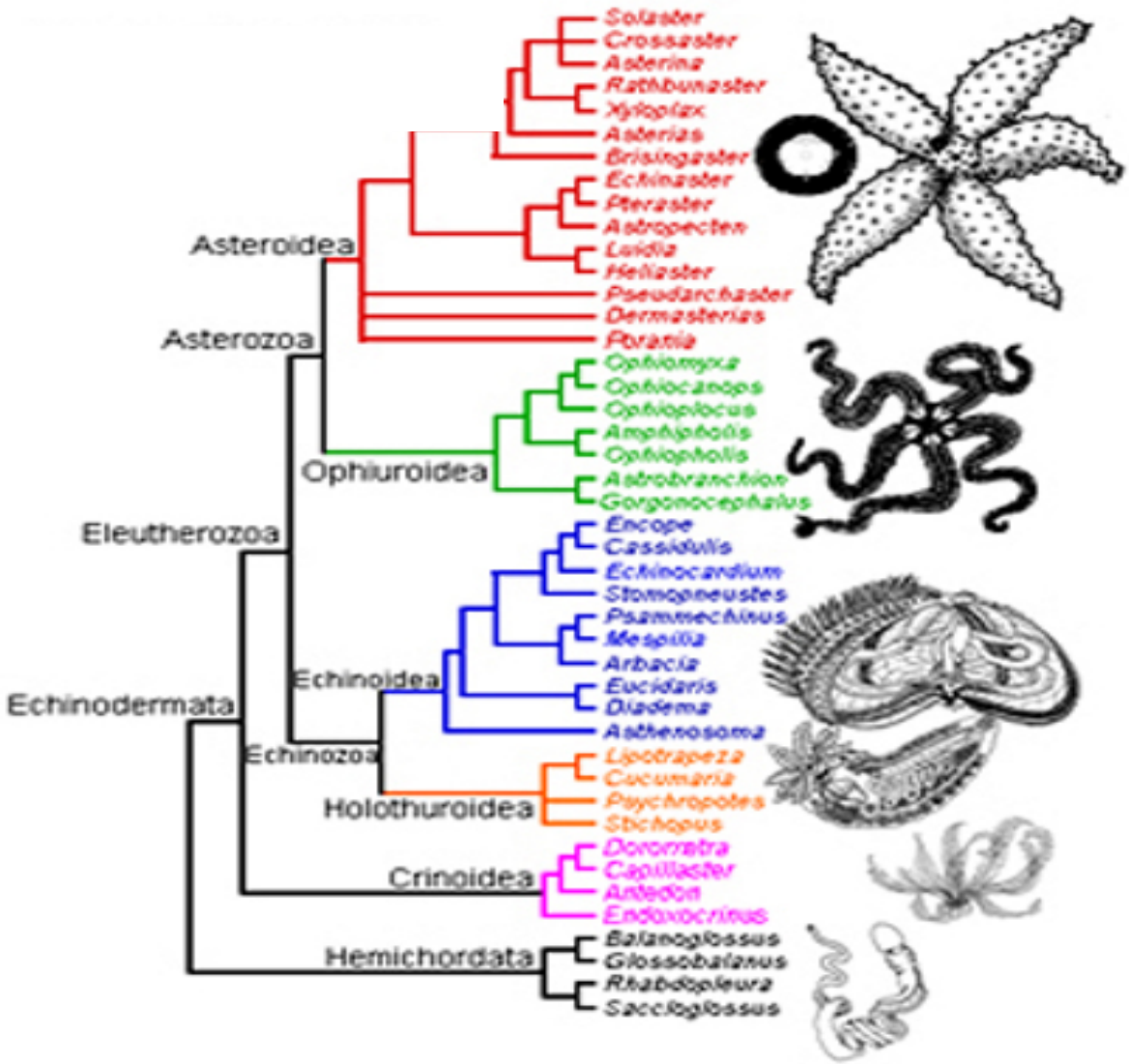
world’s first ontologist

World‘s first ontology

(from Porphyry’s *Commentary* on Aristotle’s *Categories*)



Linnaean Ontology



Formal Ontology

– theory of part and whole
– theory of dependence / unity
– theory of boundary, continuity and contact
– theory of universals and instances
– theory of continuants and occurrents (objects and processes)
– theory of functions and functioning
– theory of granularity

Formal Ontology

the theory of those ontological structures (such as part-whole, universal-particular) which apply to all domains whatsoever

Formal Ontology vs. Formal Logic

Formal ontology deals with the interconnections of *things*

with *objects* and *properties*, *parts* and *wholes*, *relations* and *collectives*

Formal logic deals with the interconnections of *truths*

with *consistency* and *validity, or* and *not*

Formal Ontology vs. Formal Logic

Formal ontology deals with formal ontological structures

Formal logic deals with formal logical structures

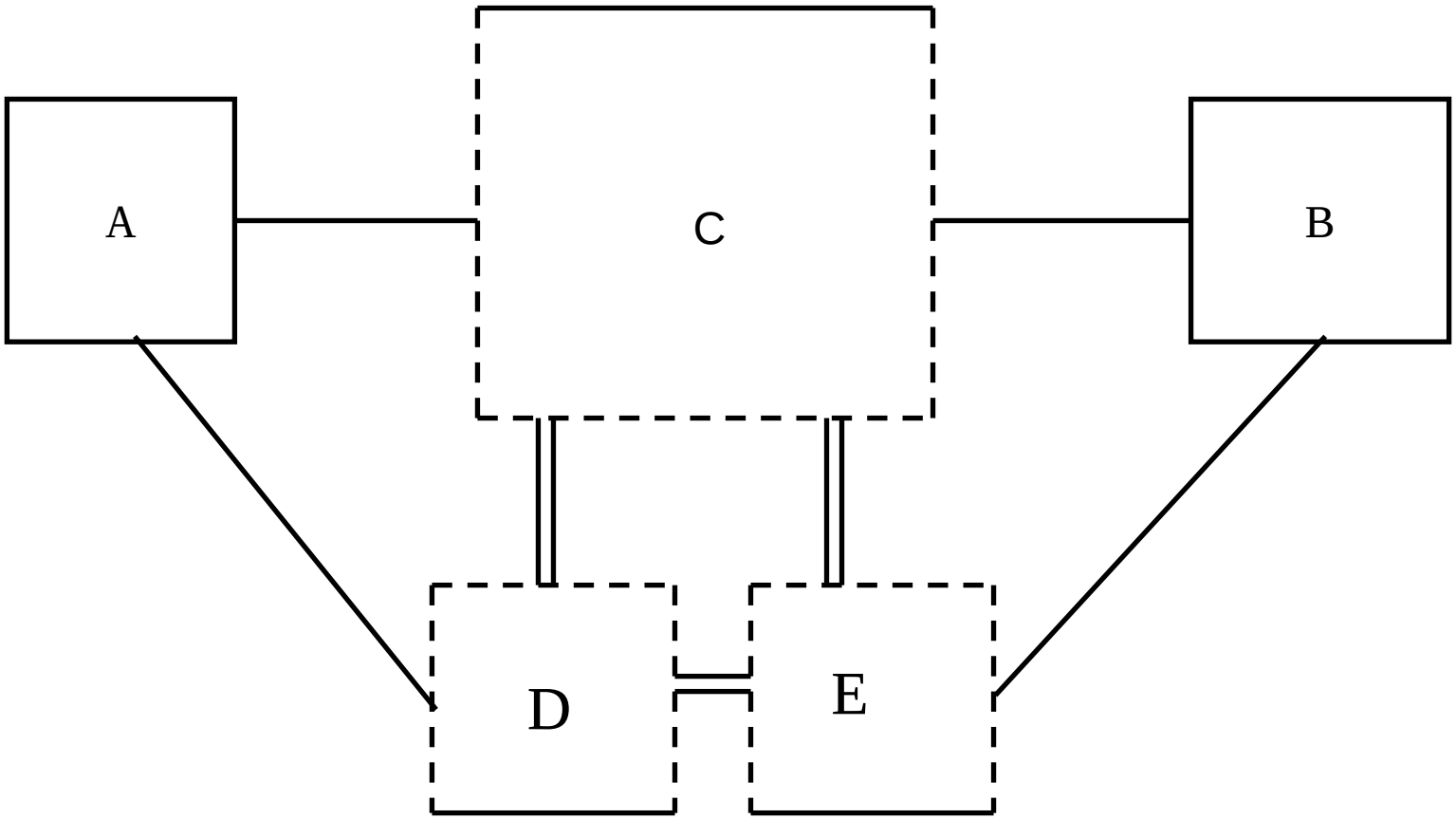
(Epistemology deals with ways of gaining knowledge)

Formal-Ontological Categories

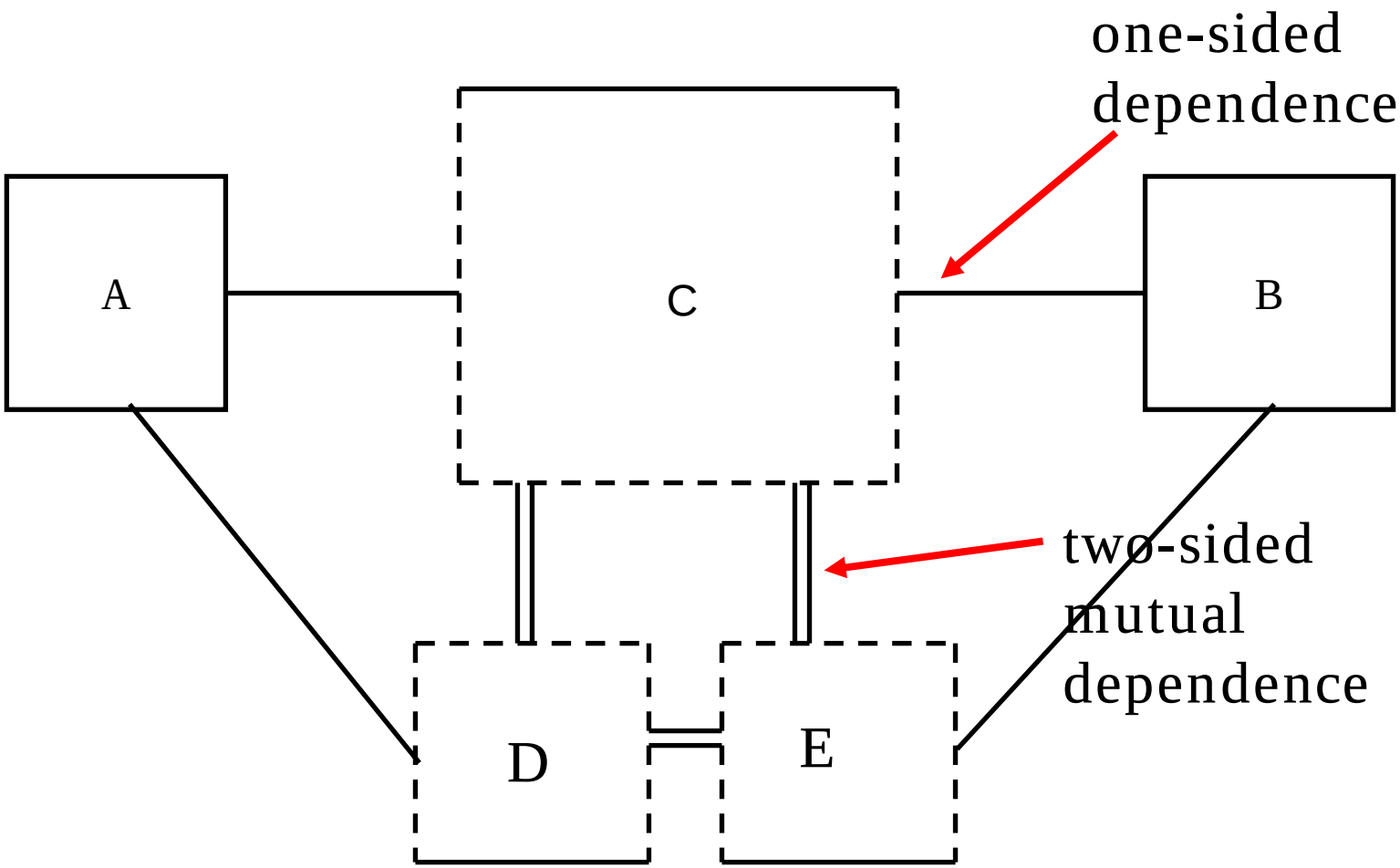
substance
process
function
unity
plurality
site
dependent part
independent part

are able to form complex structures in non-arbitrary ways joined by relations such as ***part***, ***dependence***, ***location***.

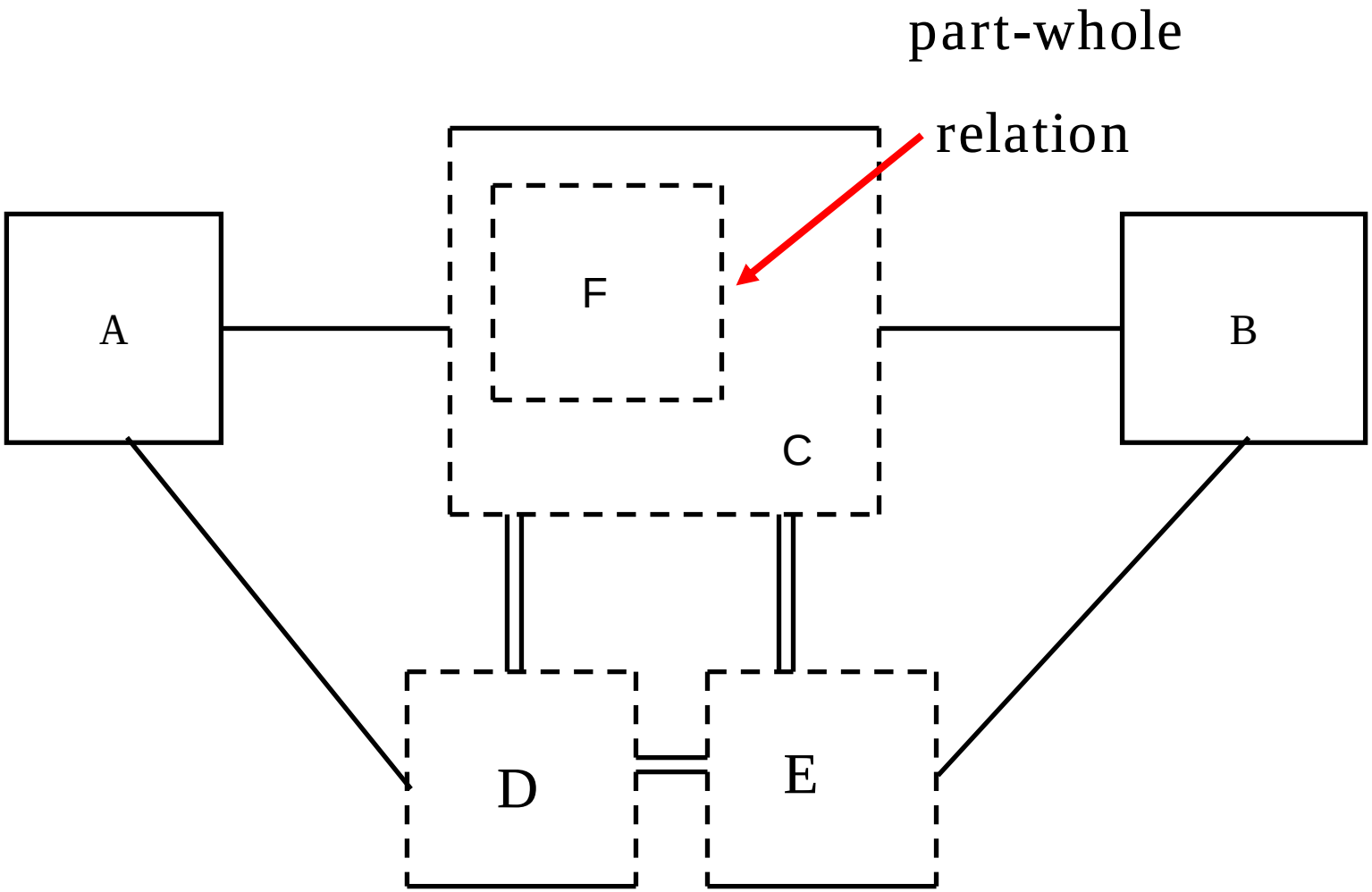
Example of a Formal-Ontological Structure



Ontological Structure



Ontological Structure



A Network of Domain Ontologies

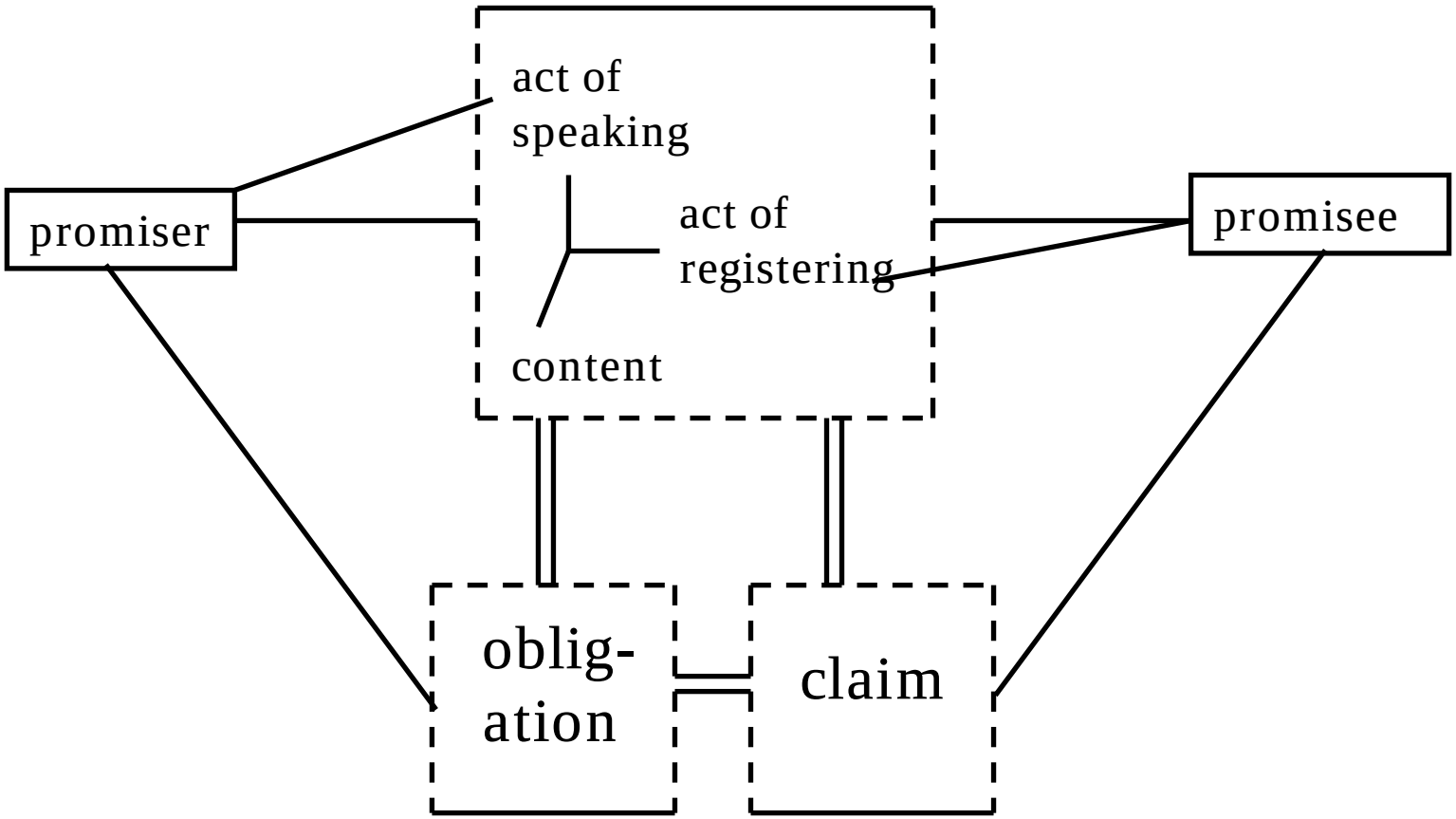
Basic Formal Ontology

Material (Regional) Ontologies

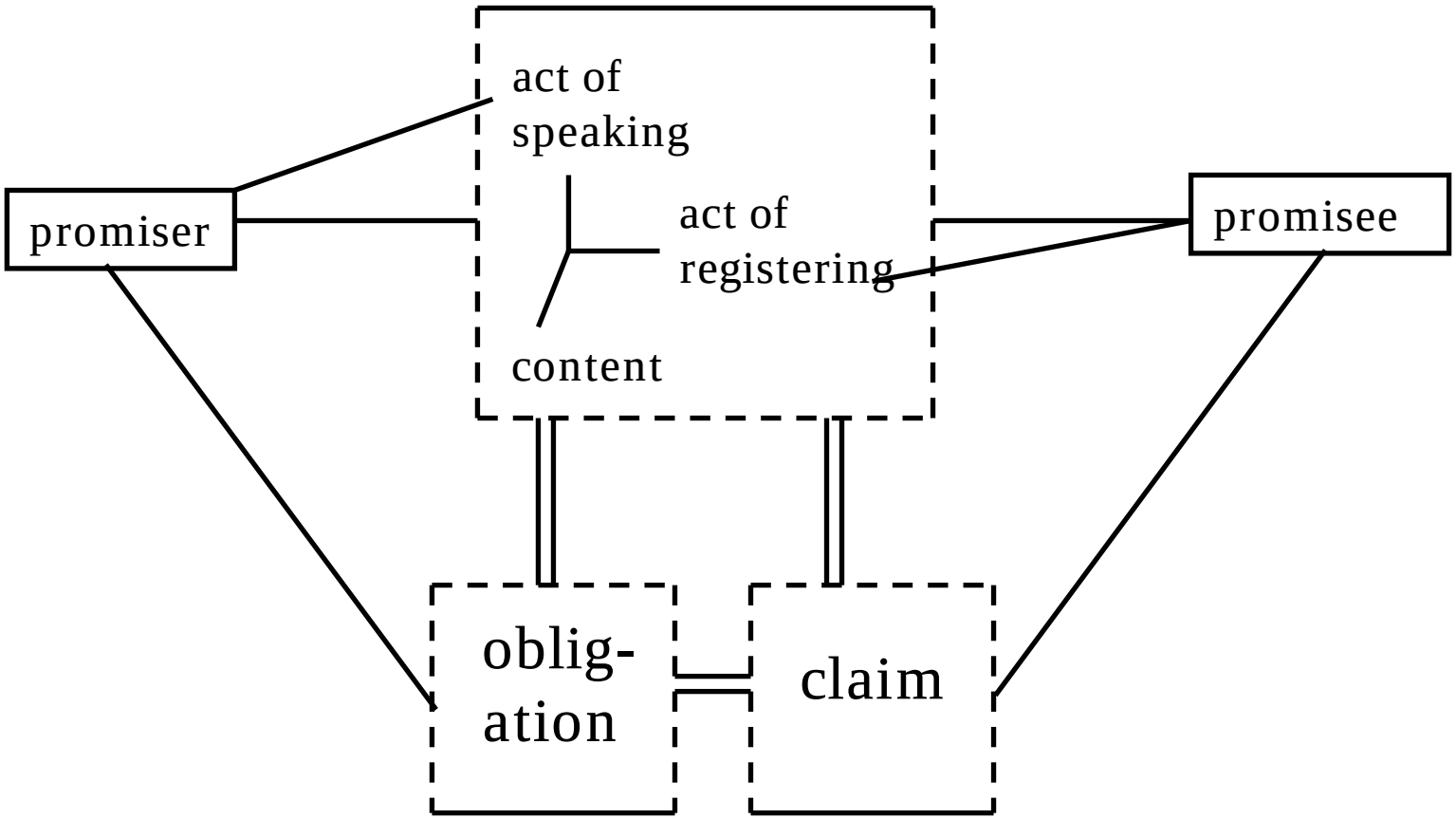
In formal ontology

as in formal logic, we can grasp the properties of given structures in such a way as to establish *in one go* the properties of all formally similar structures

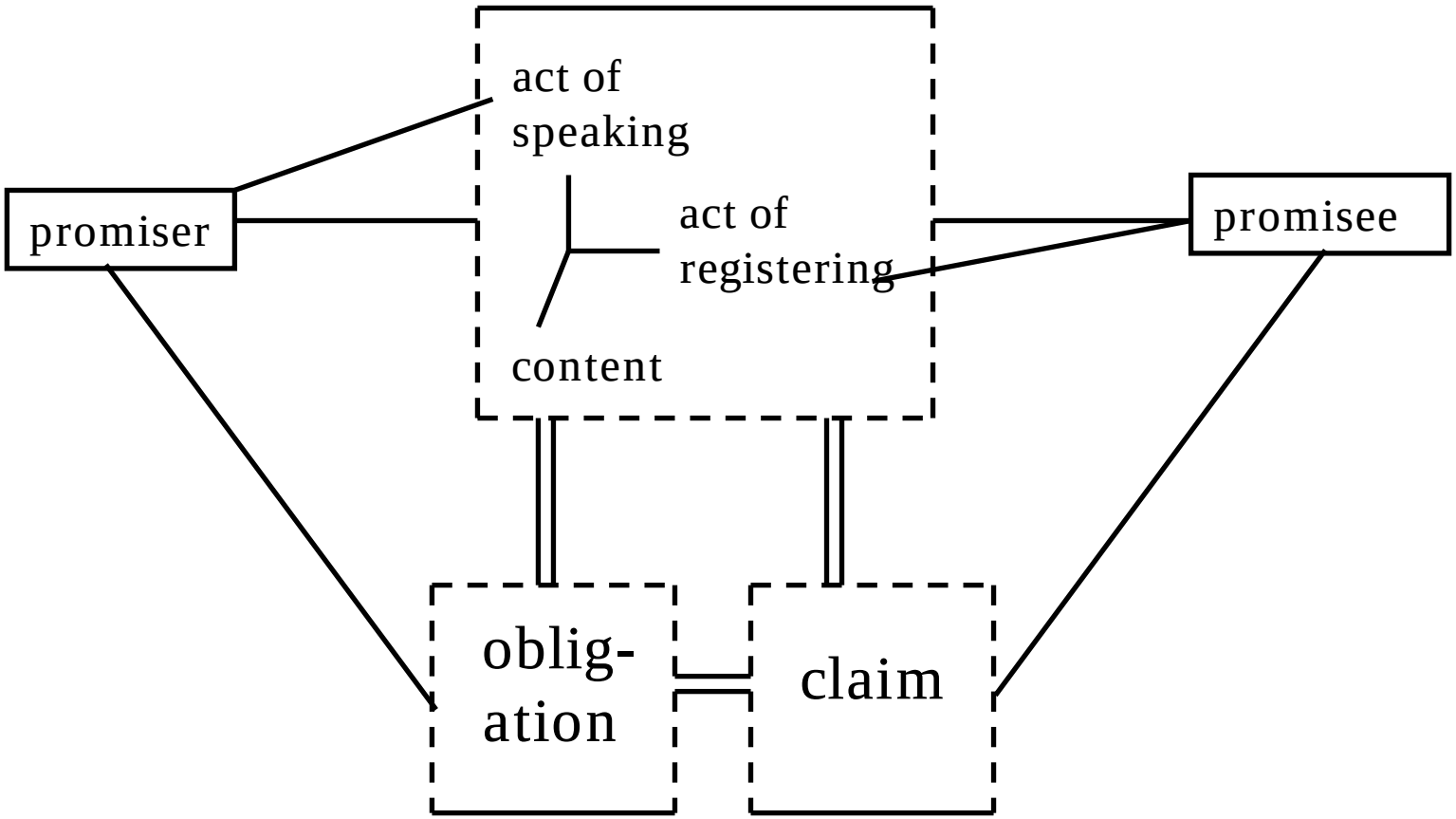
Material Ontology of Social Interaction



A Window on Reality



Universals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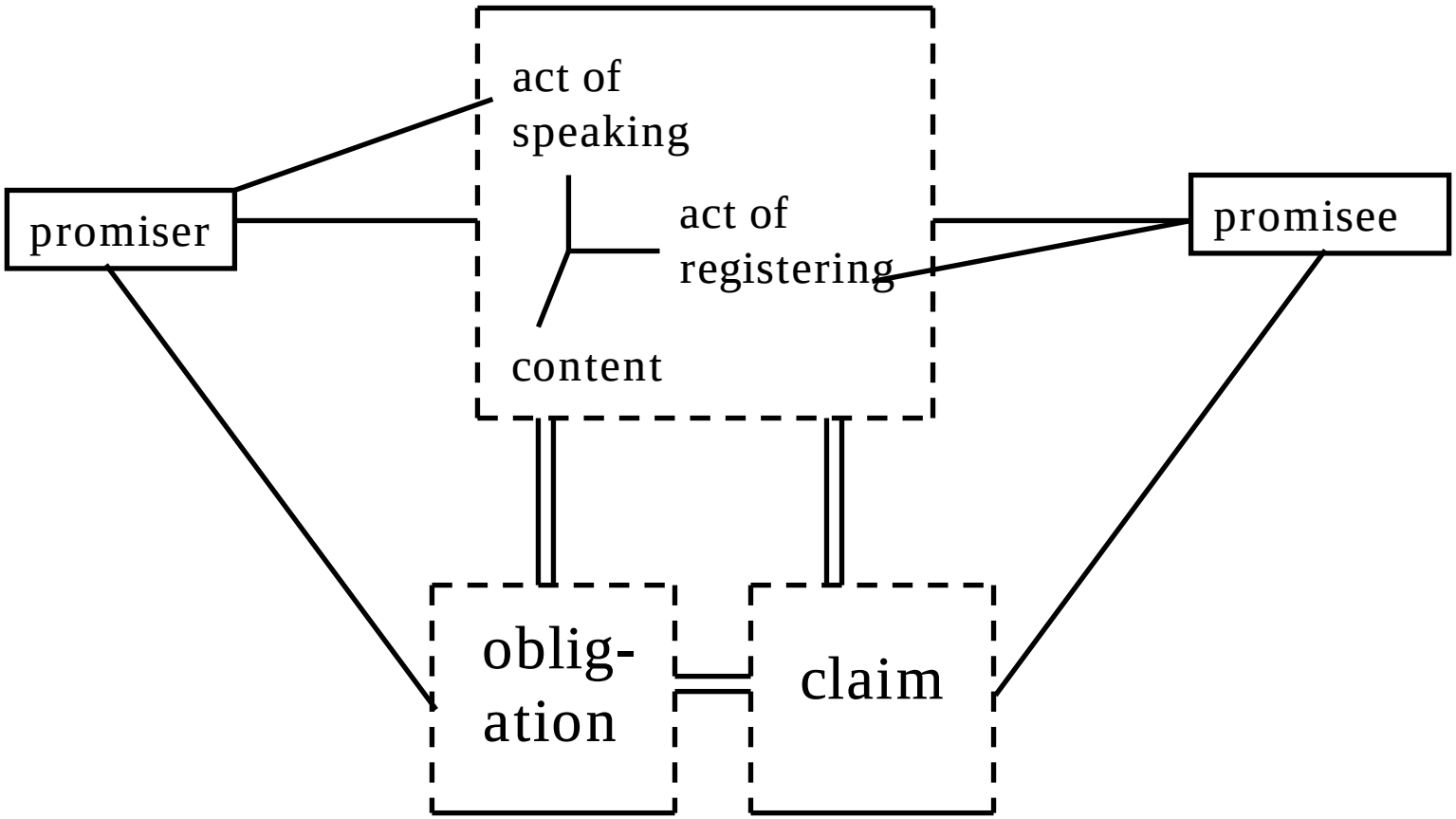
Instances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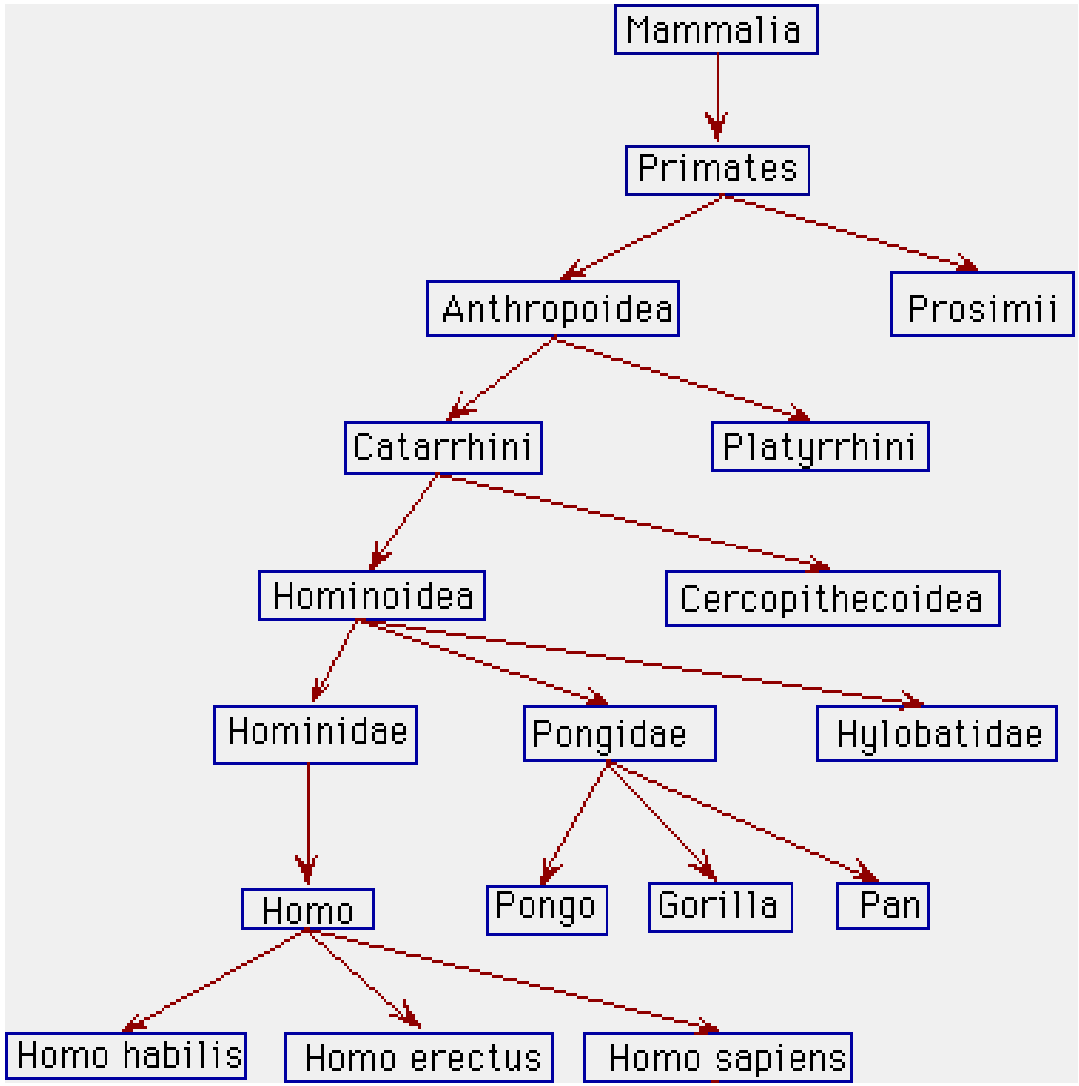
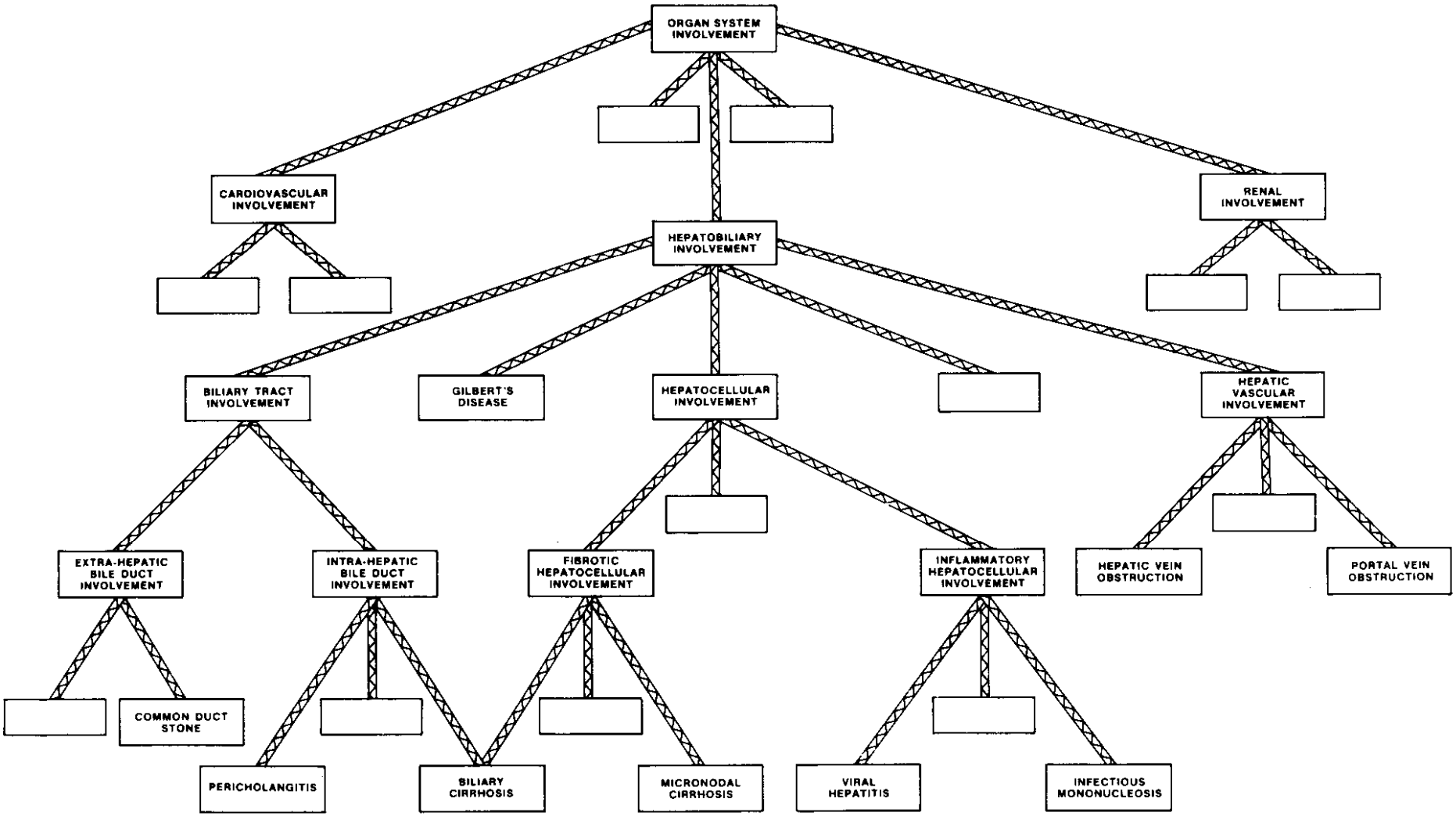
A Window on Reality

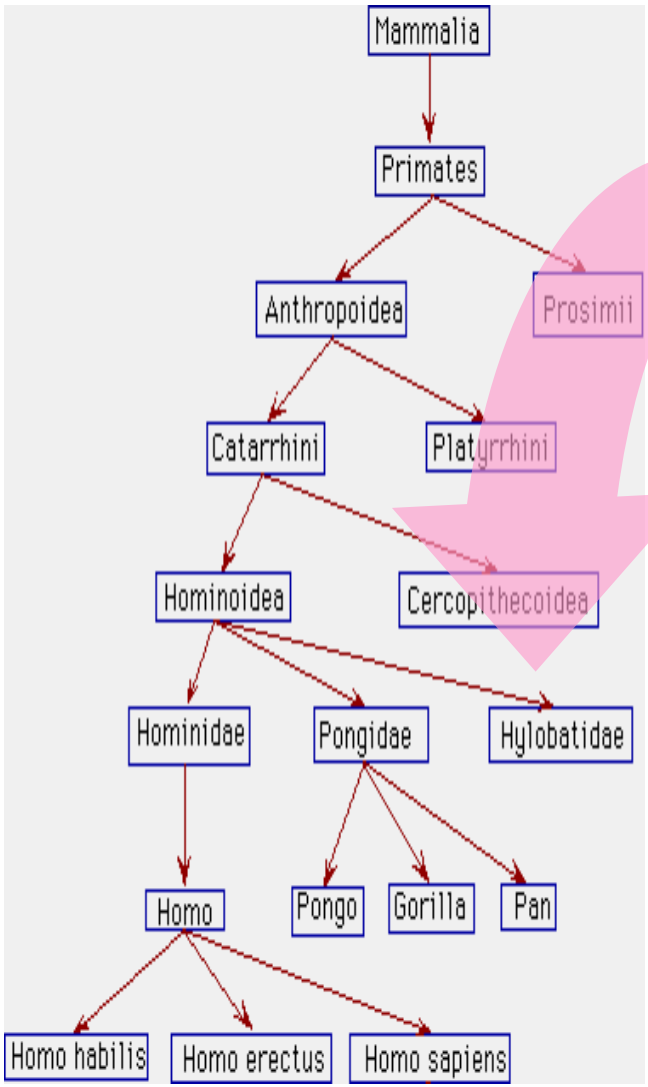


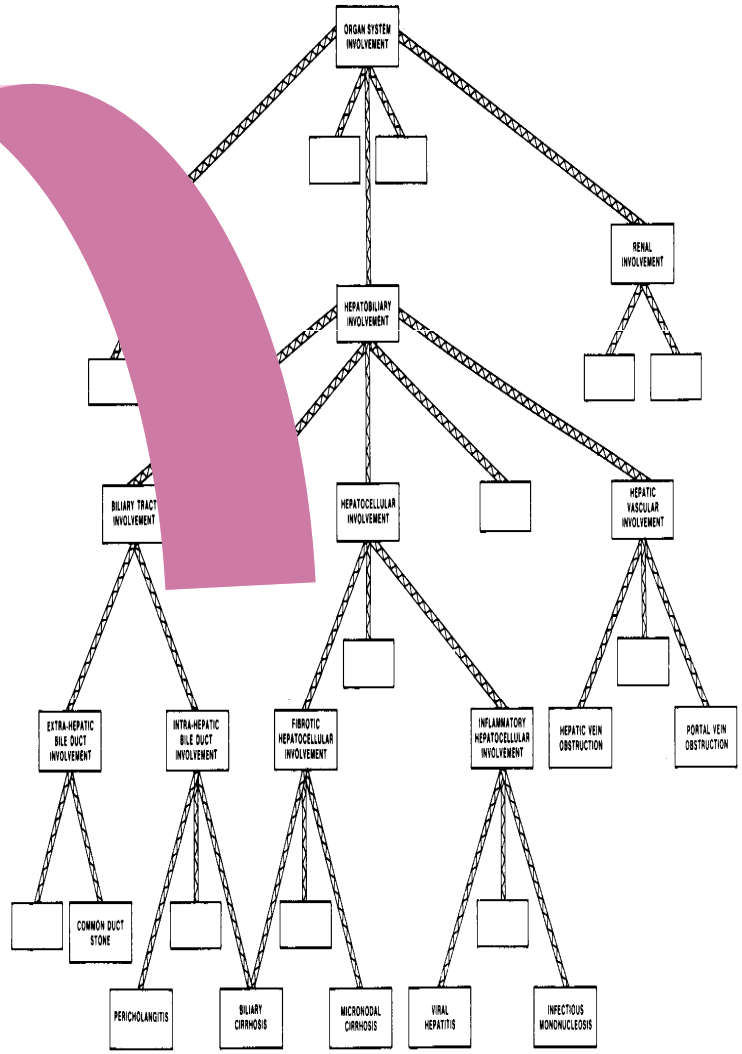
Medical Diagnostic Hierarchy



a hierarchy in the realm of diseases

Dependence Relations

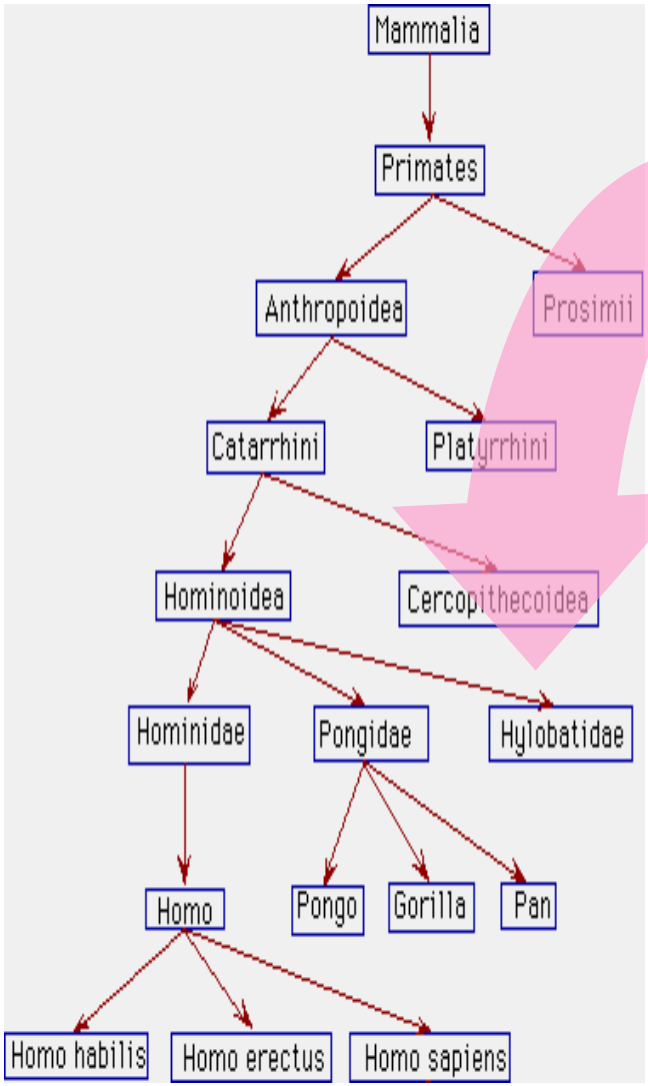


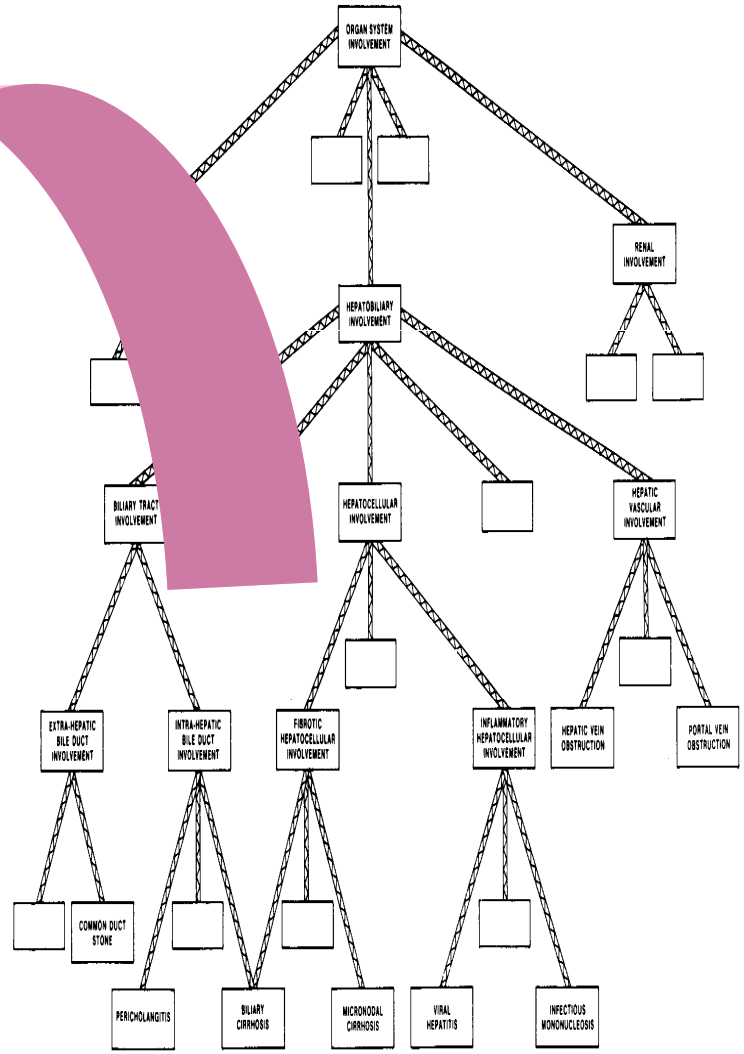


Organisms

Diseases

A Window on Reality

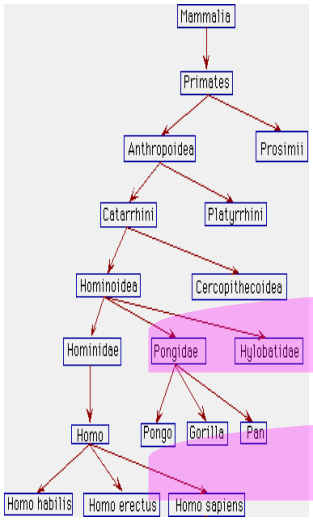


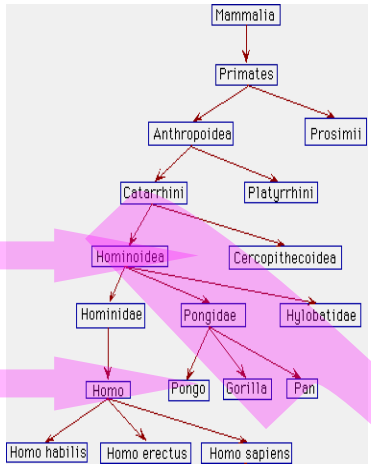


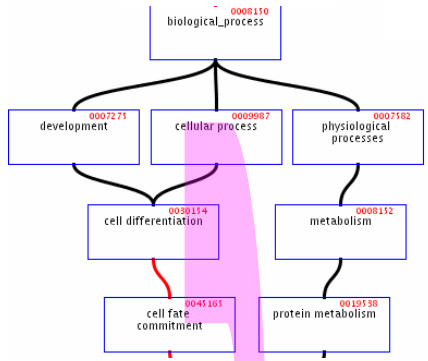
Organisms

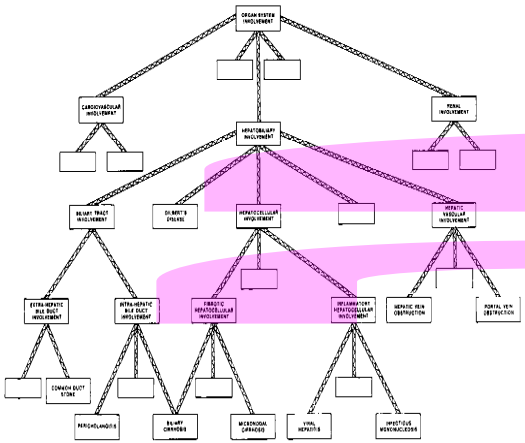
Diseases

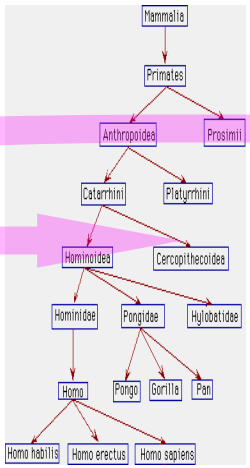
A Window on Reality

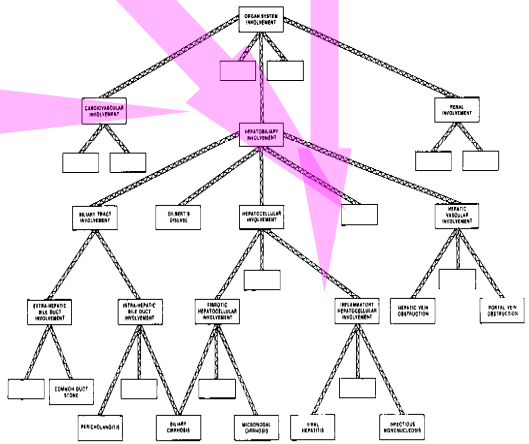




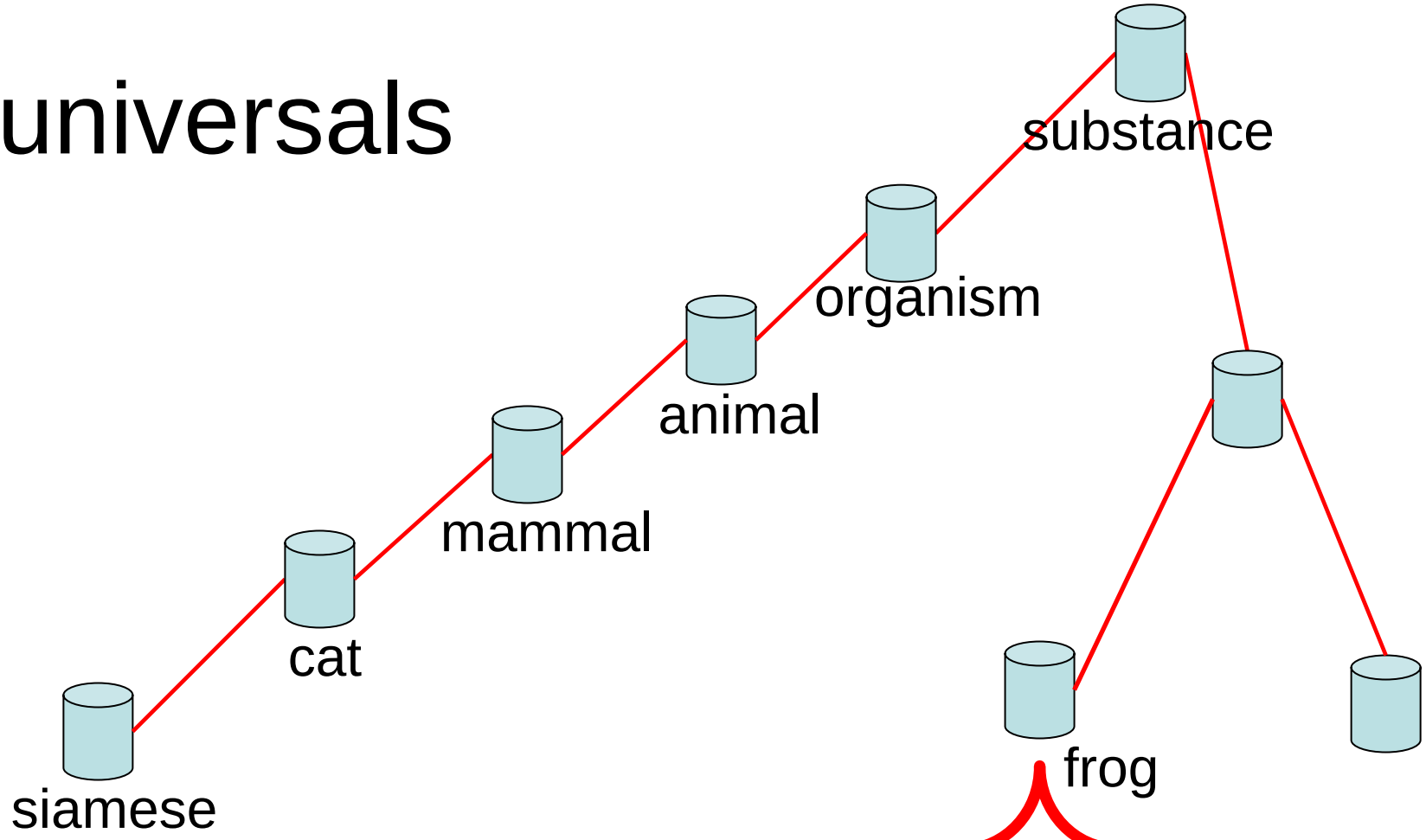




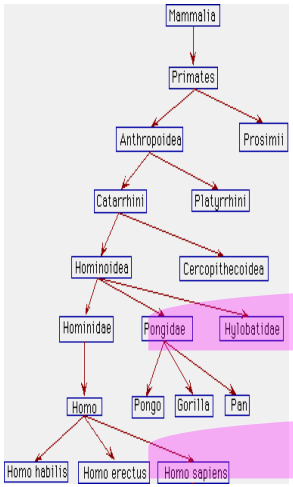


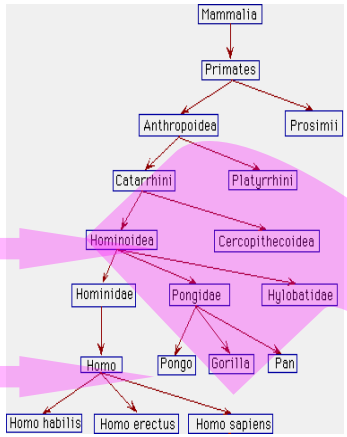


universals



instances


Mammalia
Primates
Anthropoidea
Prosimii
Catarrhini
Platyrrhini
Hominoidea
Cercopithecoidea
Hominidae
Pongidae
Hylobatidae
Homo
Pongo
Gorilla
Pan
Homo habilis
Homo erectus
Homo sapiens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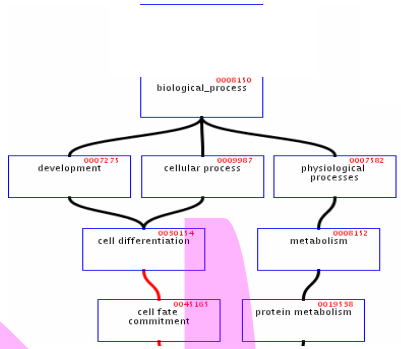
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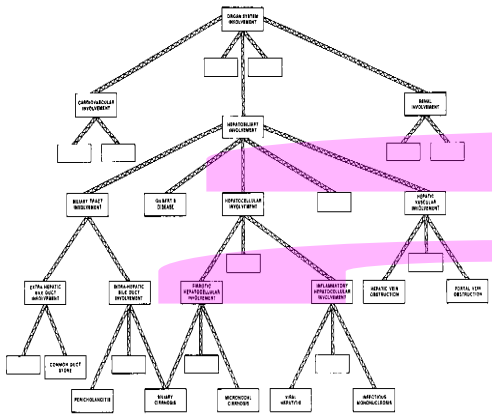
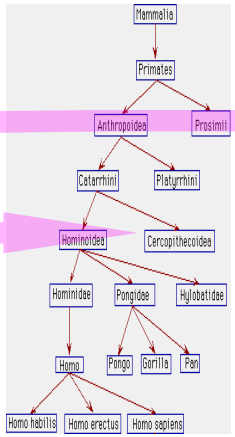
development
cellular process
physiological processes
cell differentiation
metabolism
cell fate commitment
protein metabolism

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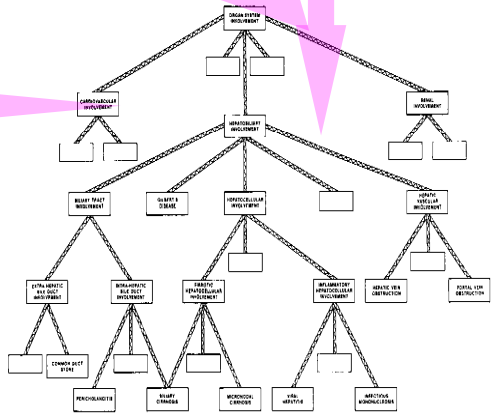
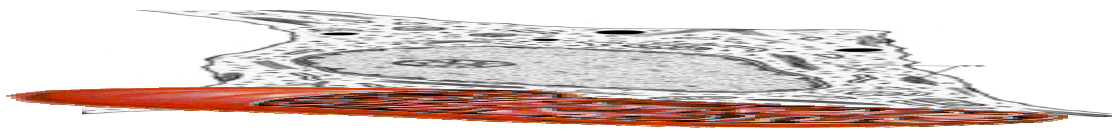






Many current standard ‘ontologies’ ramshackle because they have no counterpart of formal ontology

The Universal Medical Language System (UMLS)

a compendium of source vocabularies including:

- HL7 RIM
- SNOMED
- International Classification of Diseases
- MeSH – Medical Subject Headings
- Gene Ontology

Problem: The different source vocabularies are incompatible with each other

Problem: They contain bad coding

which often derives from failure to pay attention to simple logical or ontological principles or from principles of good definitions

Bad Coding

Plant roots ***is-a*** Plant

Plant leaves ***is-a*** Plant

Pollen ***is-a*** Plant

Both testes ***is a*** testis

Both uterii ***is a*** uterus

Bad definitions

Heptolysis $=_{def}$ the cause of heptolysis

Biological process $=_{def}$ a biological goal that requires more than one function

UMLS Source Vocabularies

HL7 RIM

SNOMED

International Classification of Diseases

MeSH – Medical Subject Headings

Gene Ontology

To reap the benefits of standardization

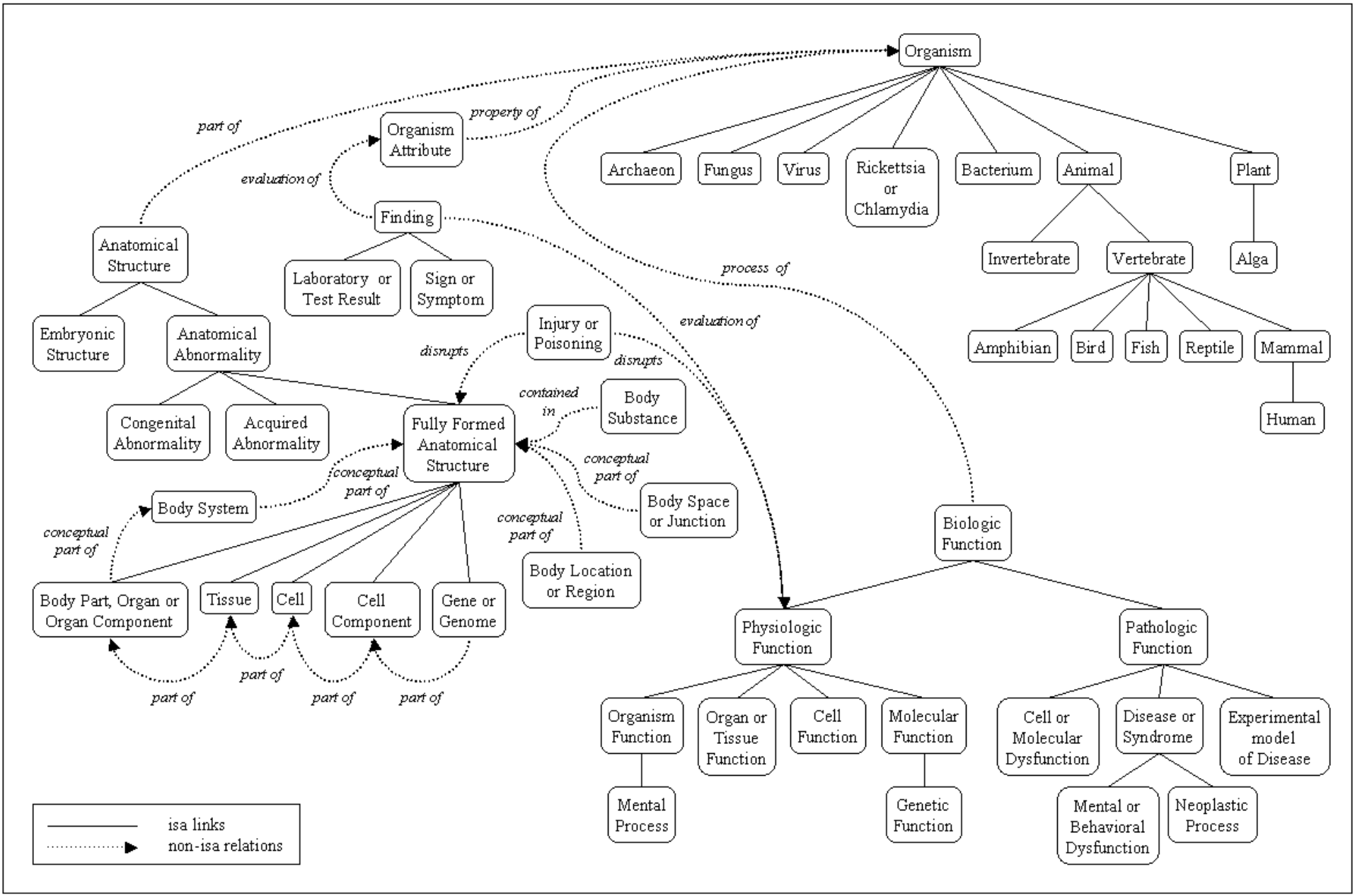
we need to make ONE SYSTEM out of many different terminologies

→ UMLS Semantic Network

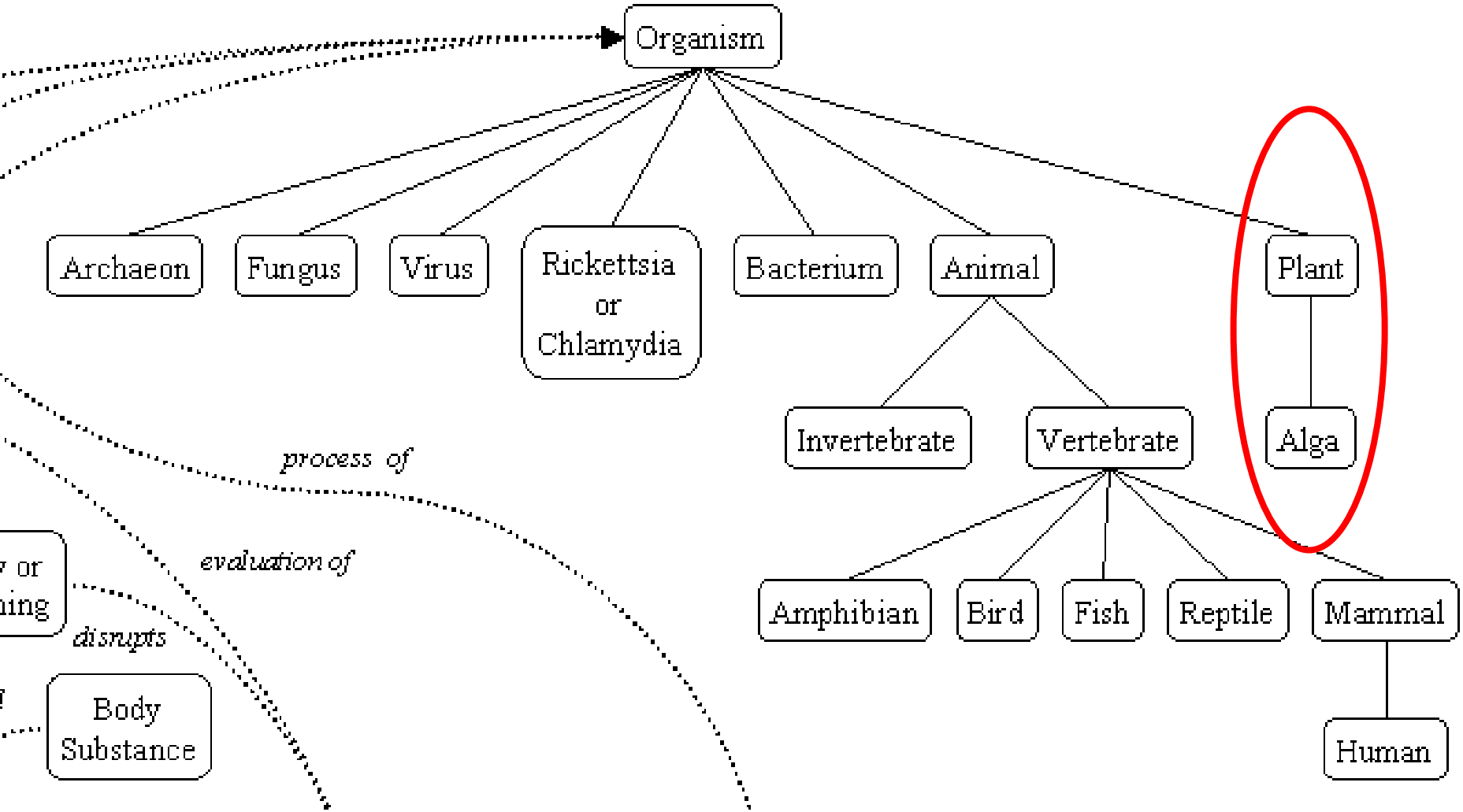
nearest thing to an “ontology” of the UMLS

134 Nodes, 54 Relationship-Types between these Nodes, forming a graph with 6000 Edges

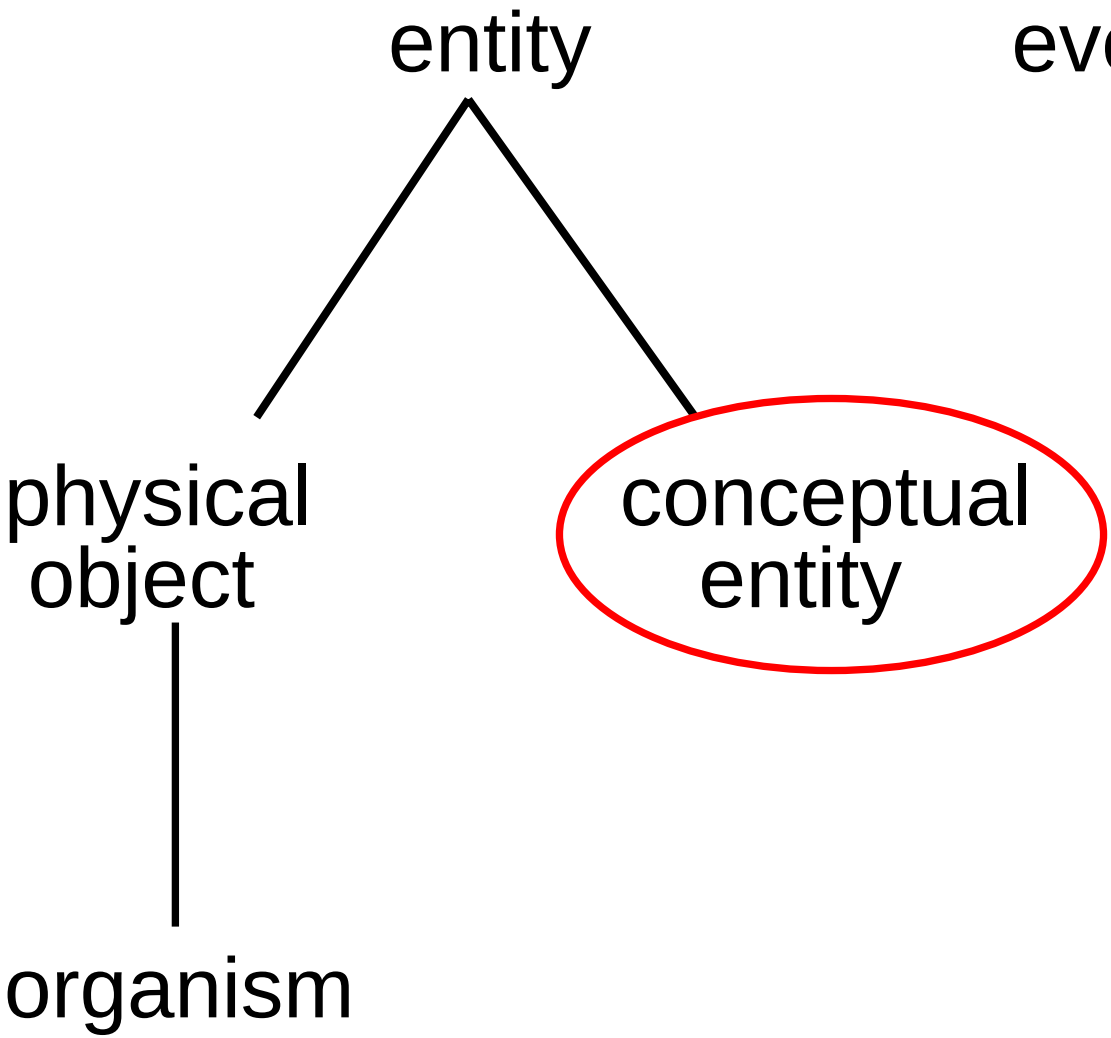
(built by linguists …)



Fragment of the UMLSemantic Network


Organism
Archaeon
Fungus
Virus
Rickettsia or Chlamydia
Bacterium
Animal
Plant
Invertebrate
Vertebrate
Alga
Amphibian
Bird
Fish
Reptile
Mammal
Human
process of
evaluation of
disrupts
Body Substance

UMLS Semantic Network



conceptual entity

Organism Attribute

Finding

Idea or Concept

Occupation or Discipline

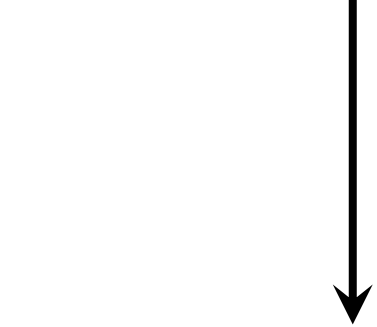
Organization

Group

Group Attribute

Intellectual Product

Language



Idea or Concept

- **Functional Concept**
- **Qualitative Concept**
- **Quantitative Concept**
- **Spatial Concept**
  - **Body Location or Region**
  - **Body Space or Junction**
  - **Geographic Area**
  - **Molecular Sequence**
    - **Amino Acid Sequence**
    - **Carbohydrate Sequence**
    - **Nucleotide Sequence**

Budapest

is an *Idea or Concept*

- Idea or Concept
  - **Functional Concept**
  - **Qualitative Concept**
  - **Quantitative Concept**
  - **Spatial Concept**
    - **Body Location or Region**
    - **Body Space or Junction**
    - **Geographic Area**
    - **Molecular Sequence**
      - Amino Acid Sequence
        - **Carbohydrate Sequence**
        - **Nucleotide Sequence**

Problem: Confusion of concepts and entities in reality

Blood Pressure Ontology

The hydraulic equation:

BP = CO*PVR

arterial blood pressure (BP) is directly proportional to the product of blood flow (cardiac output, CO) and peripheral vascular resistance (PVR).

UMLS-Semantic Types:

blood pressure is an *Organism Function*,

cardiac output is a *Laboratory or Test Result* or *Diagnostic Procedure*

BP = CO*PVR thus asserts that

blood pressure is proportional either to a laboratory or test result or to a diagnostic procedure

Problem: Confusion of reality with our (ways of gaining) knowledge about reality

What are the terms of ontologies

Concepts?

The Concept Orientation

Work on biomedical ontologies grew out of work on medical dictionaries and nomenclatures

Has focused almost exclusively on ‘concepts’ conceived (sometimes called ‘classes’, sometimes confused with terms/descriptions).

Concept-orientation also common in KR,

has led to the entrenchment of an assumption according to which all that need be said about classes can be said without appeal to time or instances.

This, however, has fostered an impoverished regime of definitions in which the use of identical terms (like ‘part’) in different systems has been allowed to mask underlying incompatibilities.

is-a

Standard definition:

A ***is-a*** *B* =def every instance of *A* is an instance of *B*

= standard definition of computer science

adult **is-a** *child*

animal owned by the Emperor ***is-a*** *animal*

mammal ***is-a*** *object weighing less than 200 kg*

correct reading of ***is-a***

1. *A* and *B* are natural kinds,
2. there are times at which instances of *A* exist,
3. at all such times these instances are **necessarily (of their very nature)** also instances of *B*

1. *eukaryotic cell* **is-a** *cell*
2. *mammal* **is-a** *animal*
3. *death* **is-a** *biological process*

Ontologies

Here *A* and *B* are *universals*

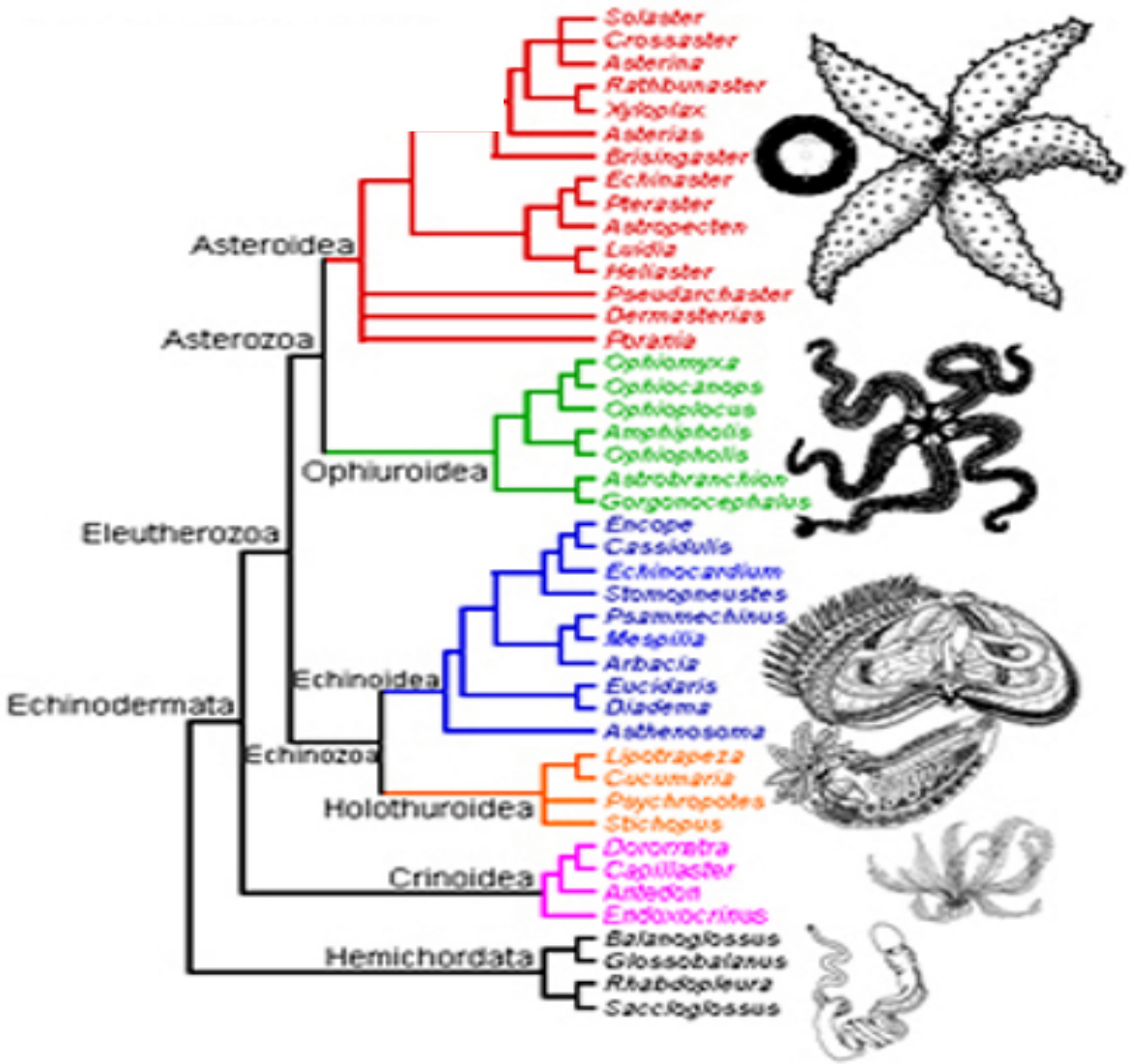
(= natural kinds, types …, roughly analogous to biological species)

Universals have *instances* (you and me, your headache, my coughing)

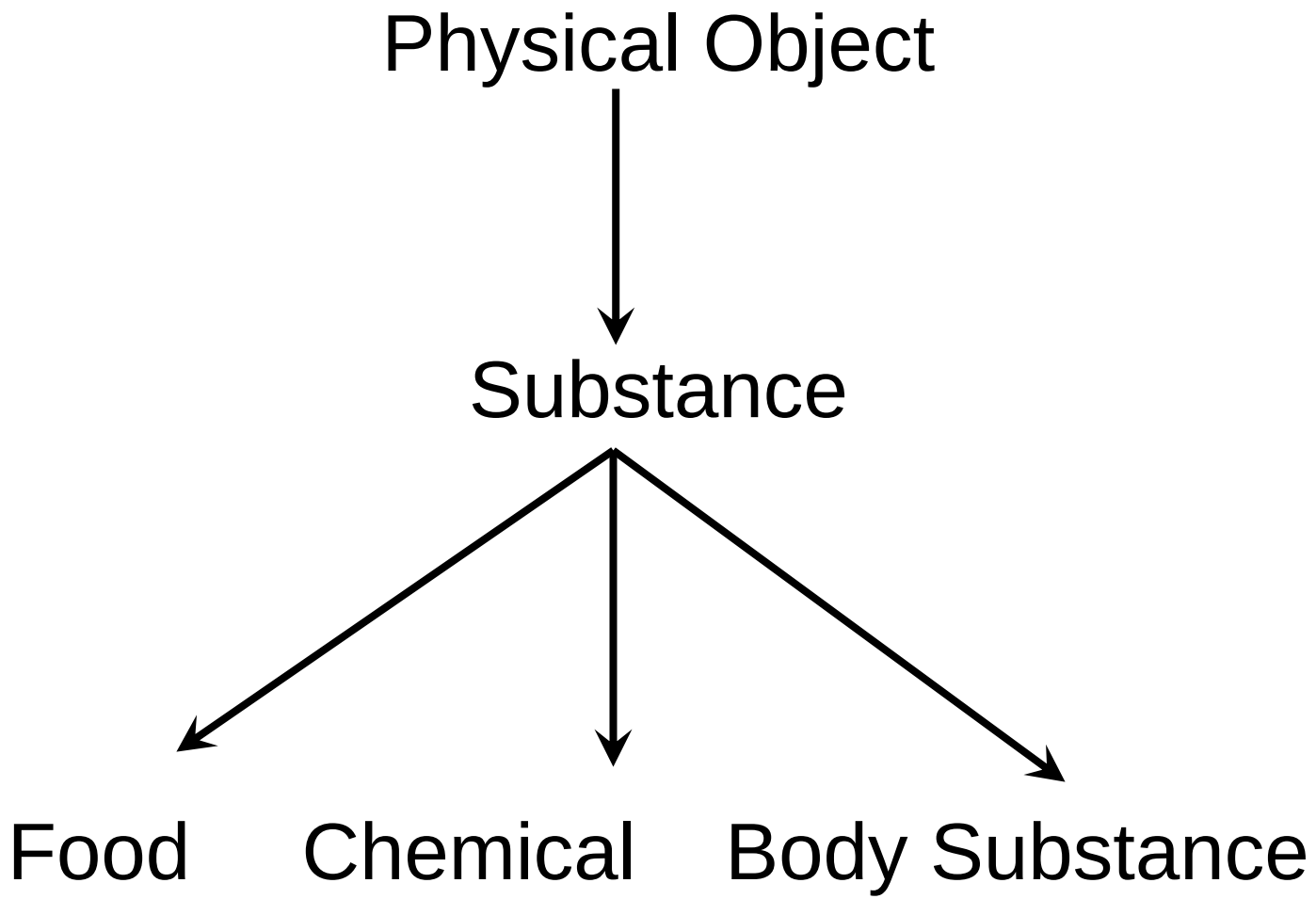
Instances are elite individuals

they are those which instantiate universals (entering into biological laws)

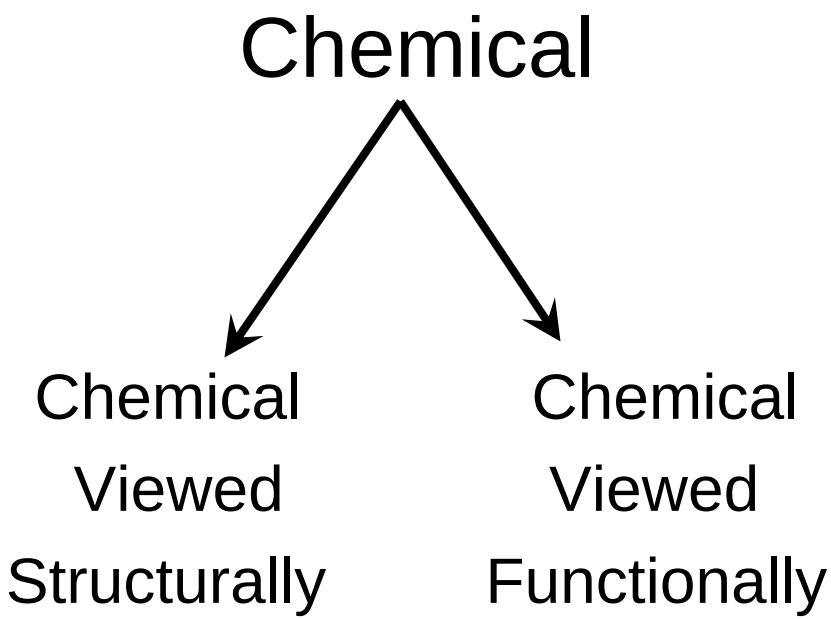
Linnaean Ontology

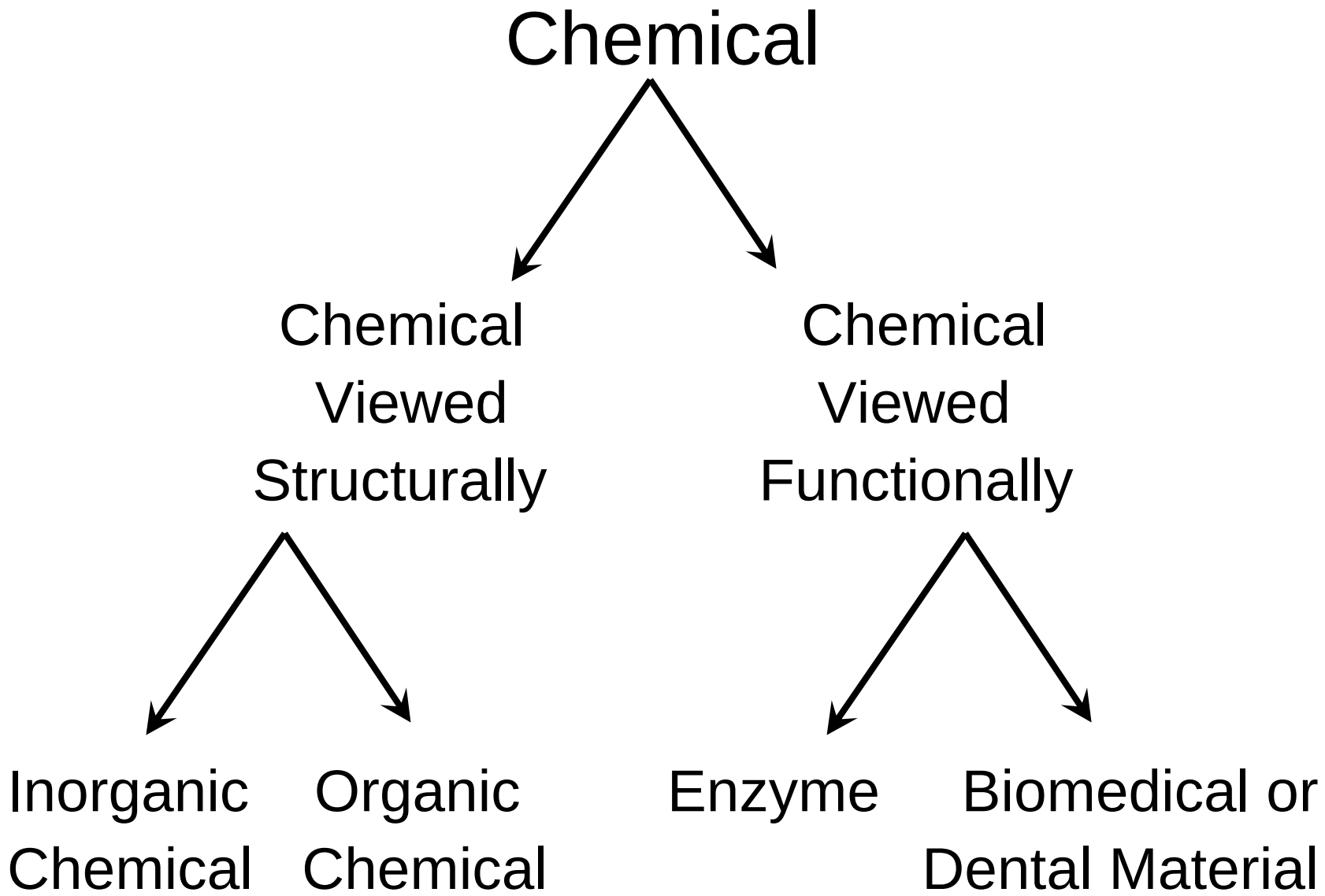


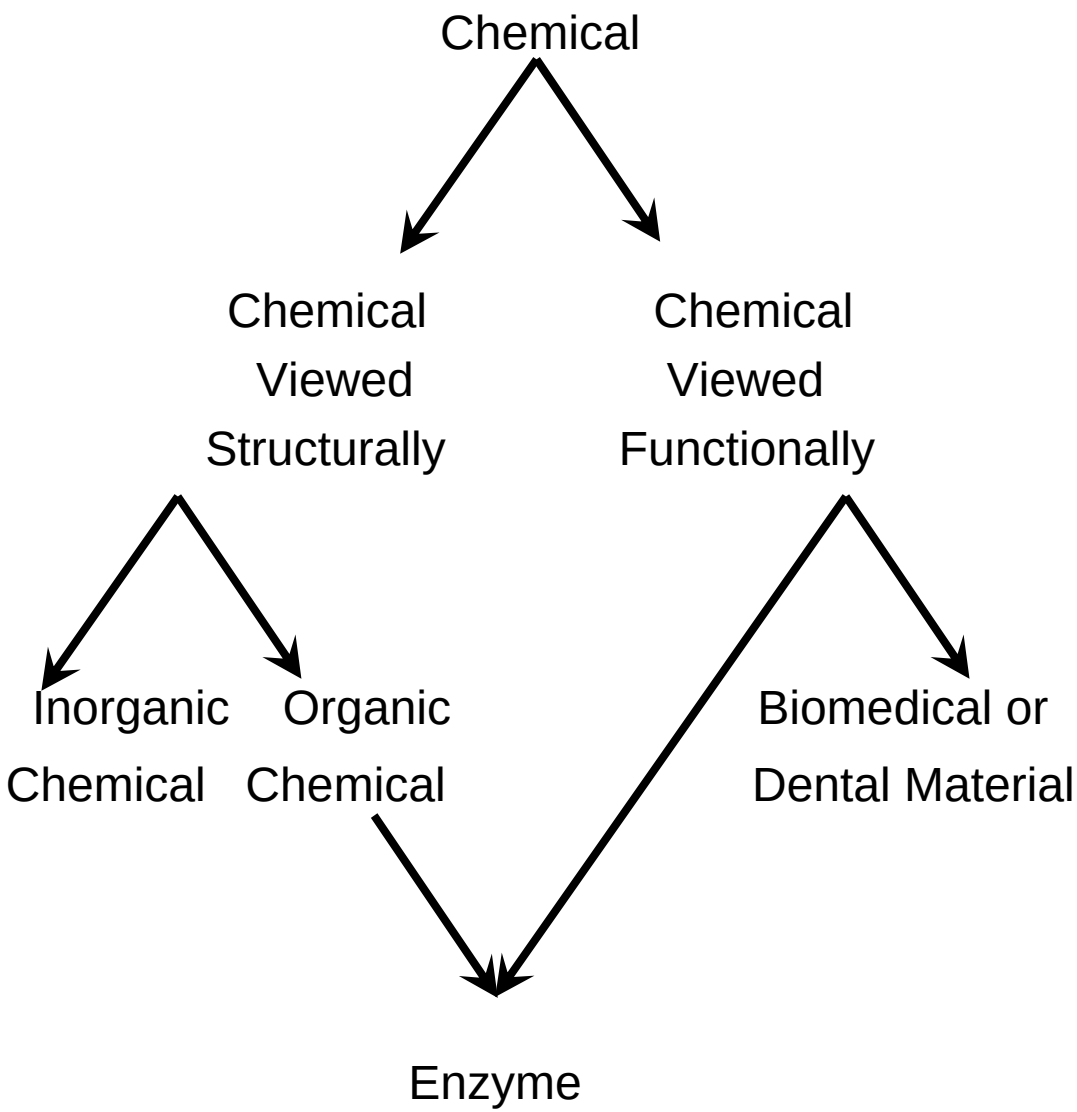
Confusion of Ontology and Epistemology



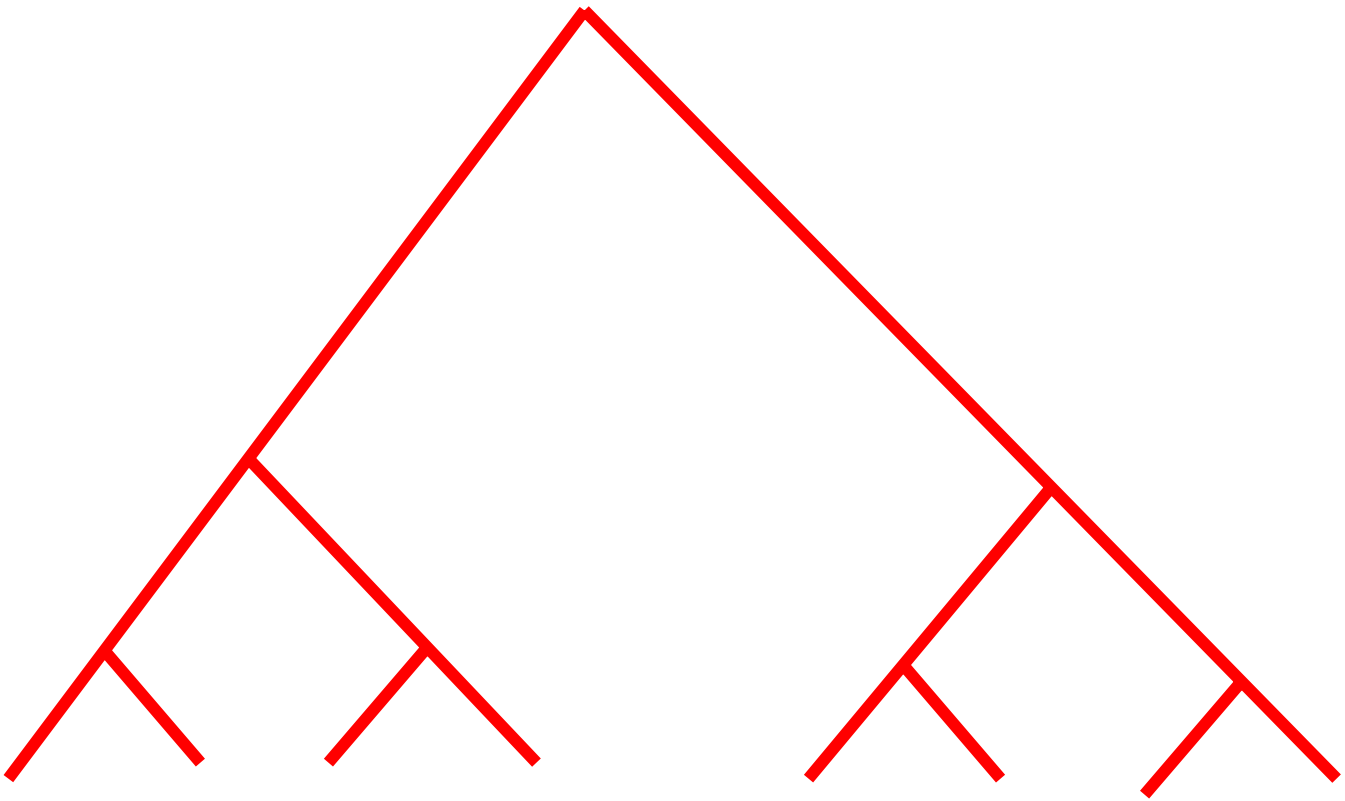
Confusion of Ontology and Epistemology




Chemical
Chemical Viewed Structurally
Chemical Viewed Functionally
Inorganic Chemical
Organic Chemical
Enzyme
Biomedical or Dental Material

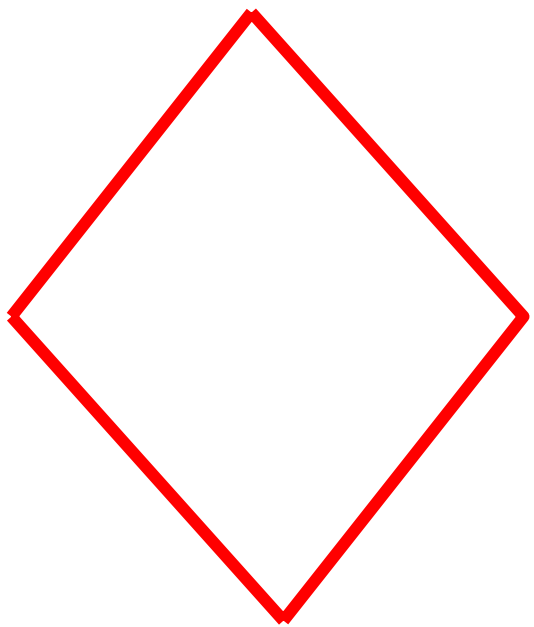

Chemical
Chemical Viewed Structurally
Chemical Viewed Functionally
Inorganic Chemical
Organic Chemical
Biomedical or Dental Material
Enzyme

Is biological classification Linnaean?

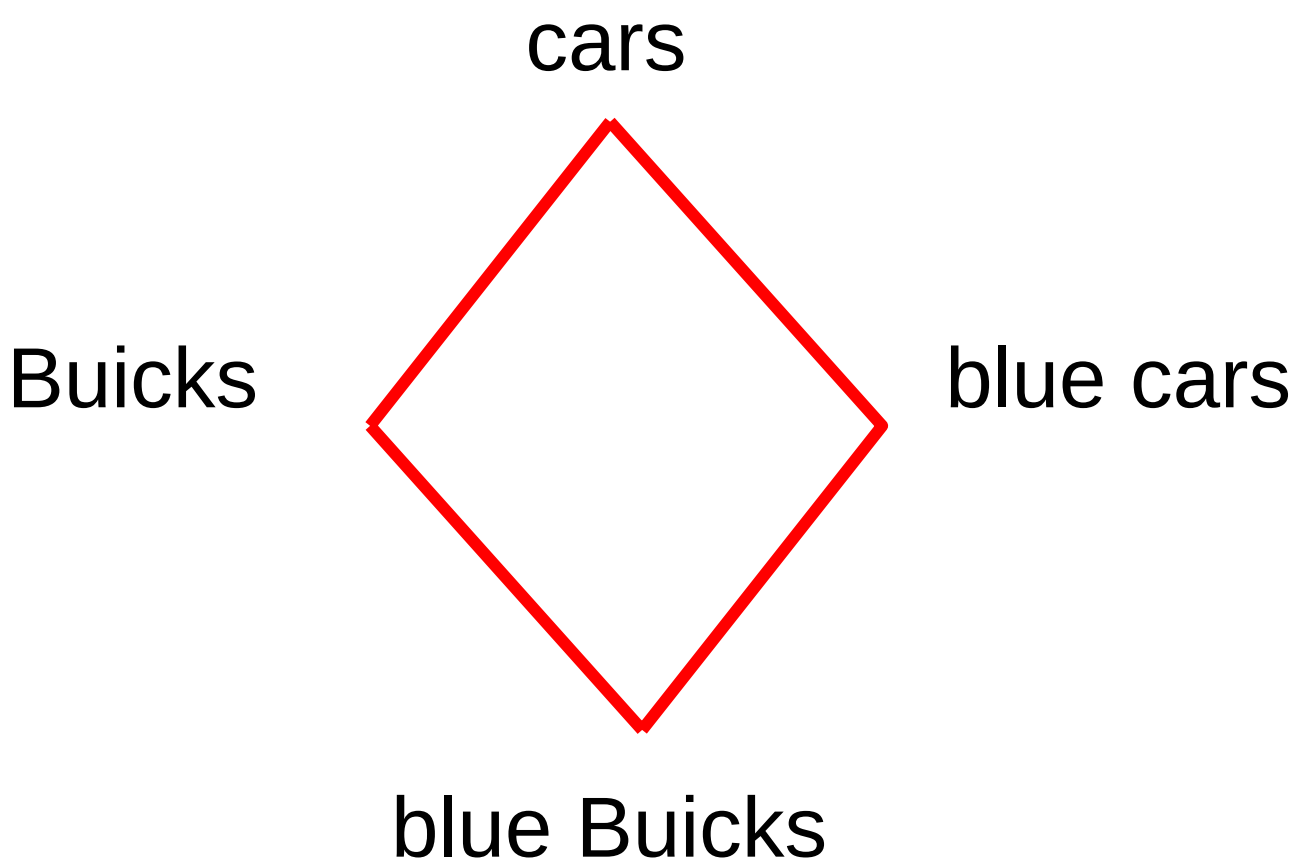


Principle of Single Inheritance

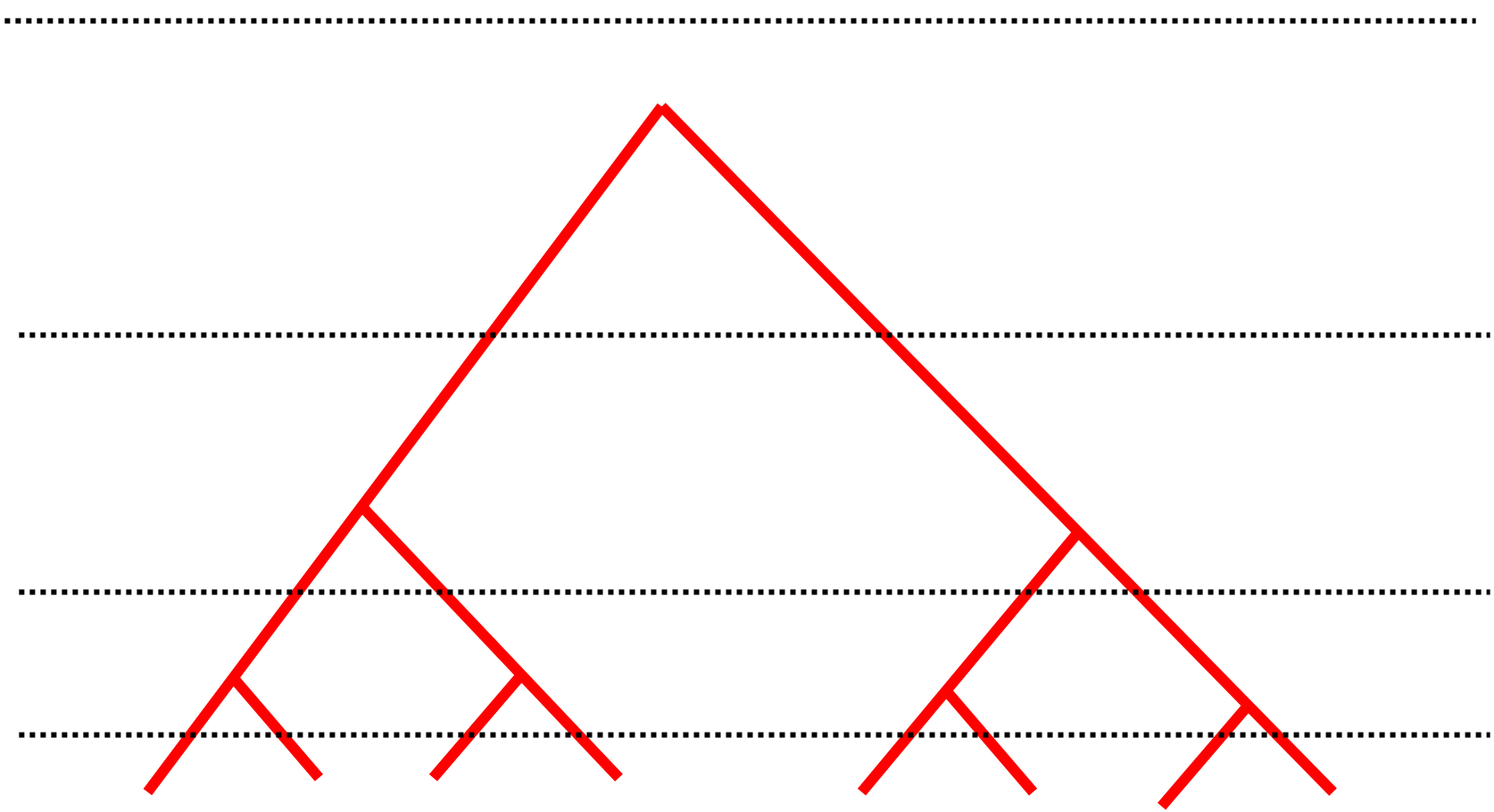
(rule of thumb) no class in a classificatory hierarchy should have more than one parent



The Problem of Multiple Inheritance



Principle of Taxonomic Levels



Principle of Taxonomic Levels

the terms in a classificatory hierarchy should be divided into predetermined levels (analogous to the levels of *kingdom*, *phylum*, *class*, *order*, etc., in traditional biology).

‘depth’ in GO’s hierarchies not determinate because of multiple inheritance

Principle of Exhaustiveness

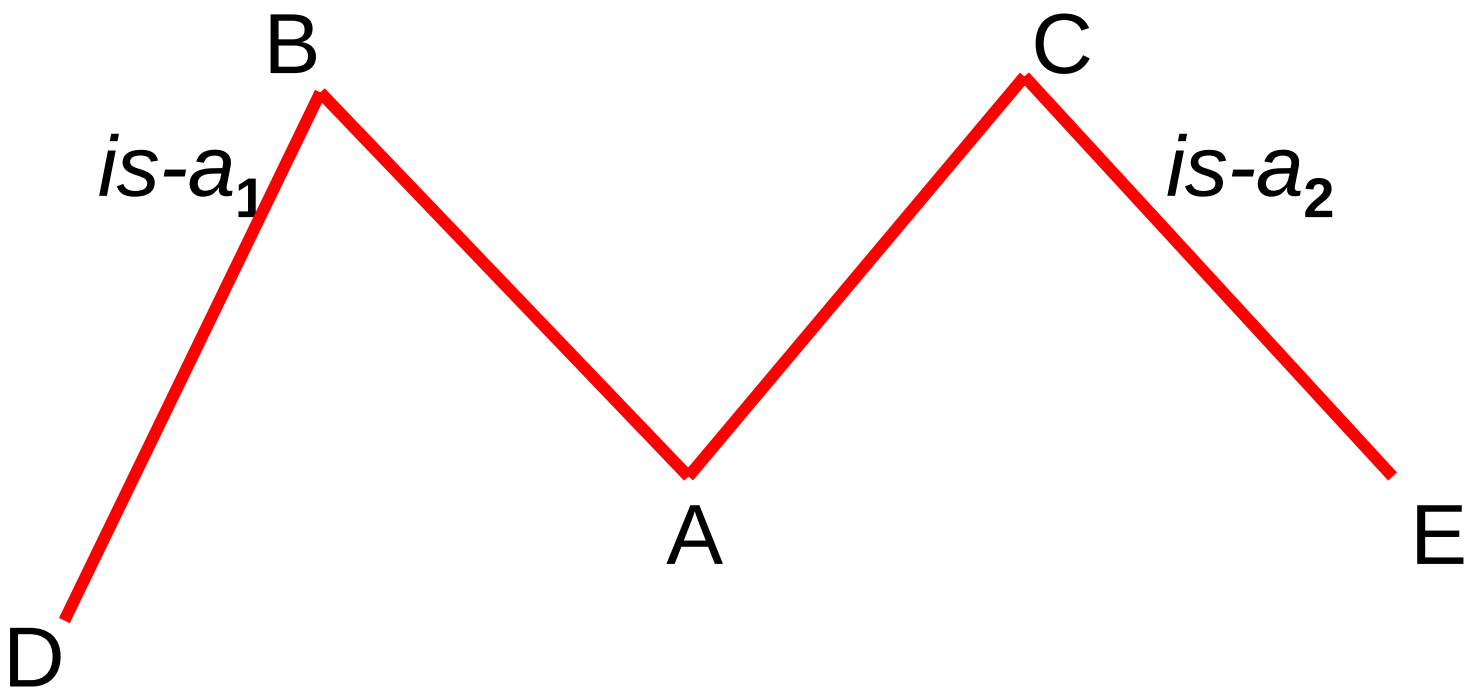
the classes on any given level should exhaust the domain of the classificatory hierarchy.

Single Inheritance + Exhaustiveness = JEPD

Exhaustiveness often difficult to satisfy in the realm of biological phenomena; but its acceptance as an ideal is presupposed as a goal by every scientist.

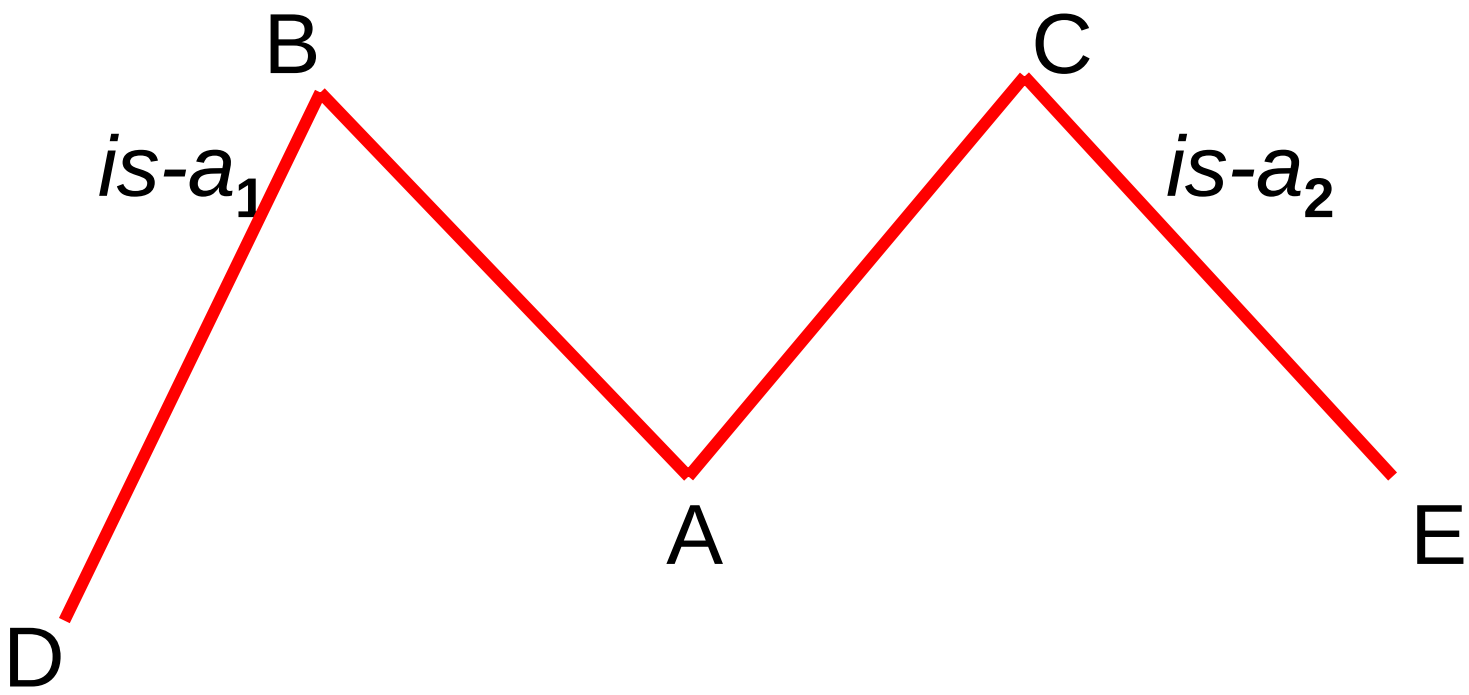
Single inheritance accepted in all traditional (species-genus) classifications

Problems with multiple inheritance



‘sibling’ is no longer determinate

Problems with multiple inheritance



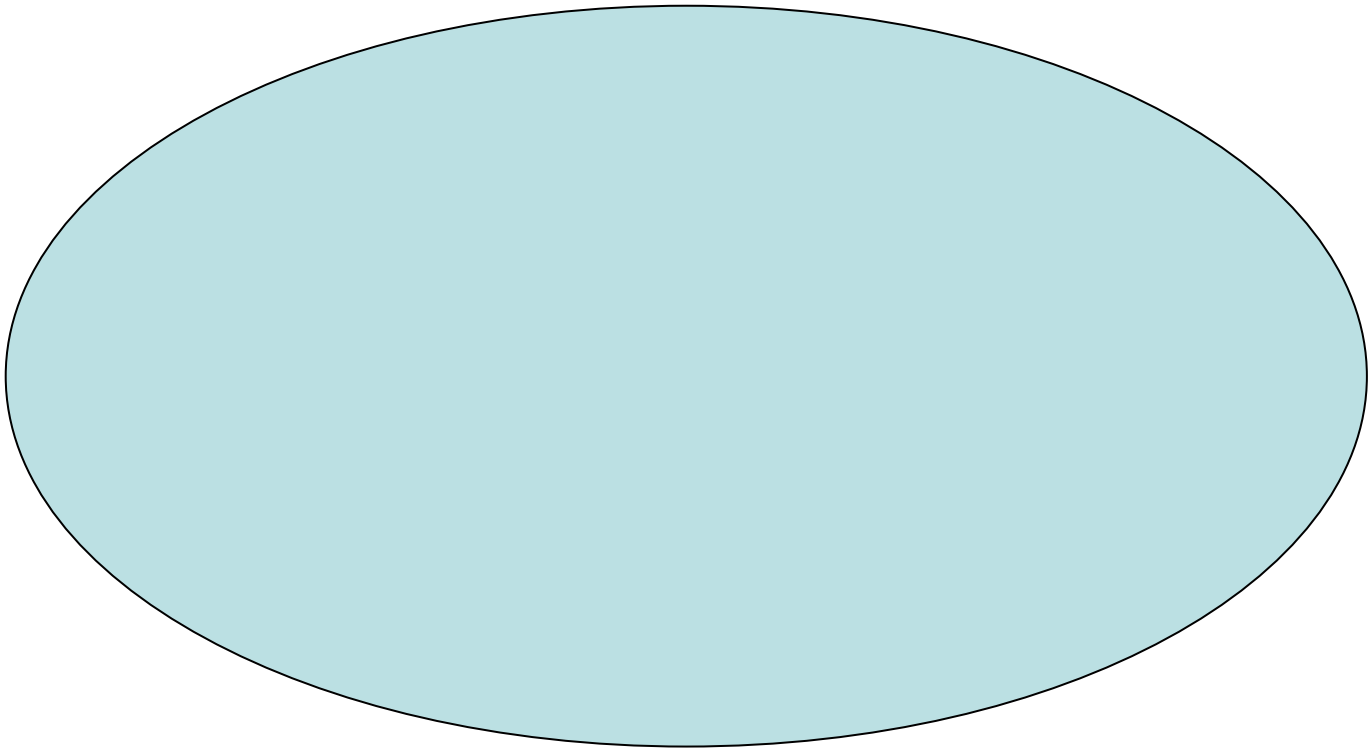
‘*is*_*a*’ is no longer univocal

when '***is-a***' is pressed into service to mean a variety of different things

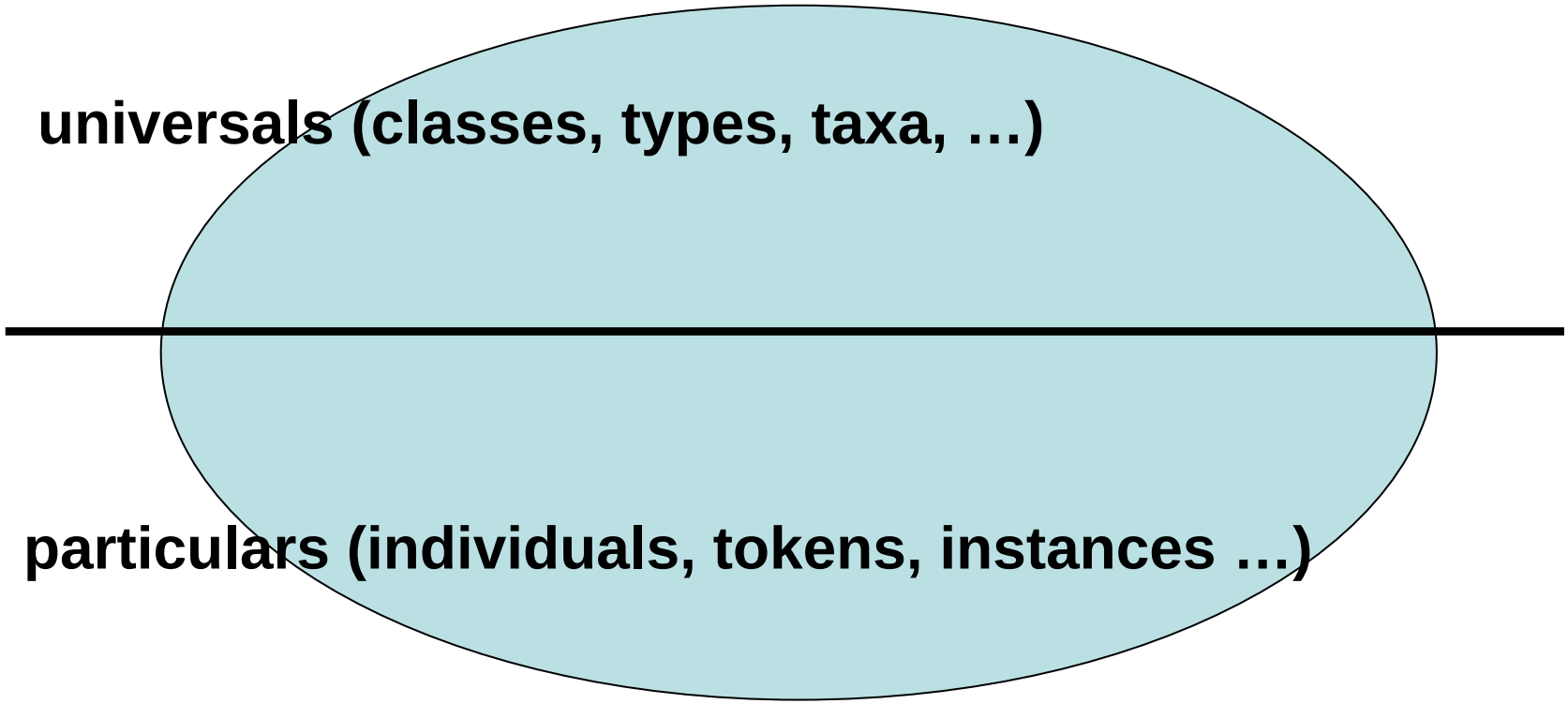
- the resulting ambiguities make the rules for correct coding difficult to communicate to human curators
- they also serve as obstacles to integration with neighboring ontologies

How are universals and instances related together?

Entities



Entities



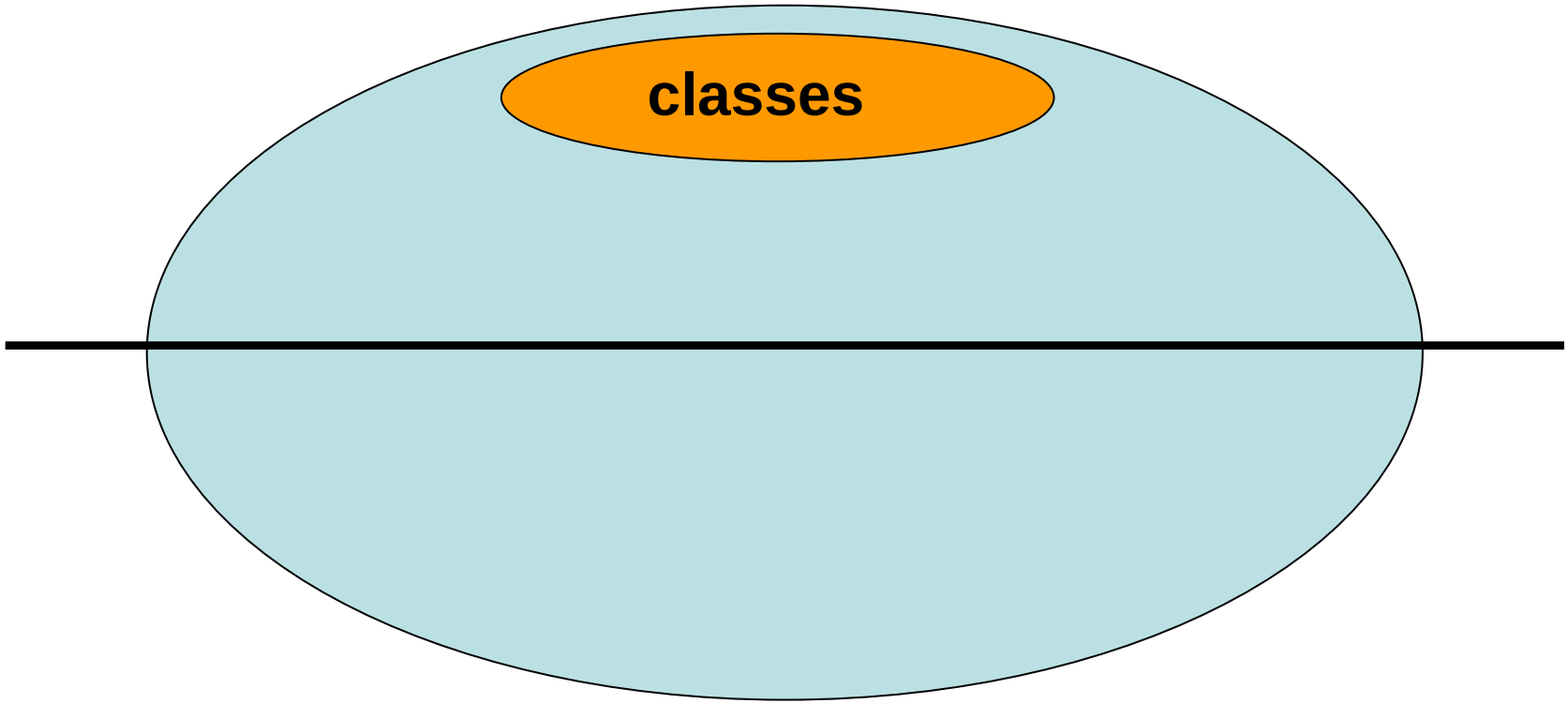
Axiom: Nothing is both a universal and a particular

Two Kinds of Elite Entities

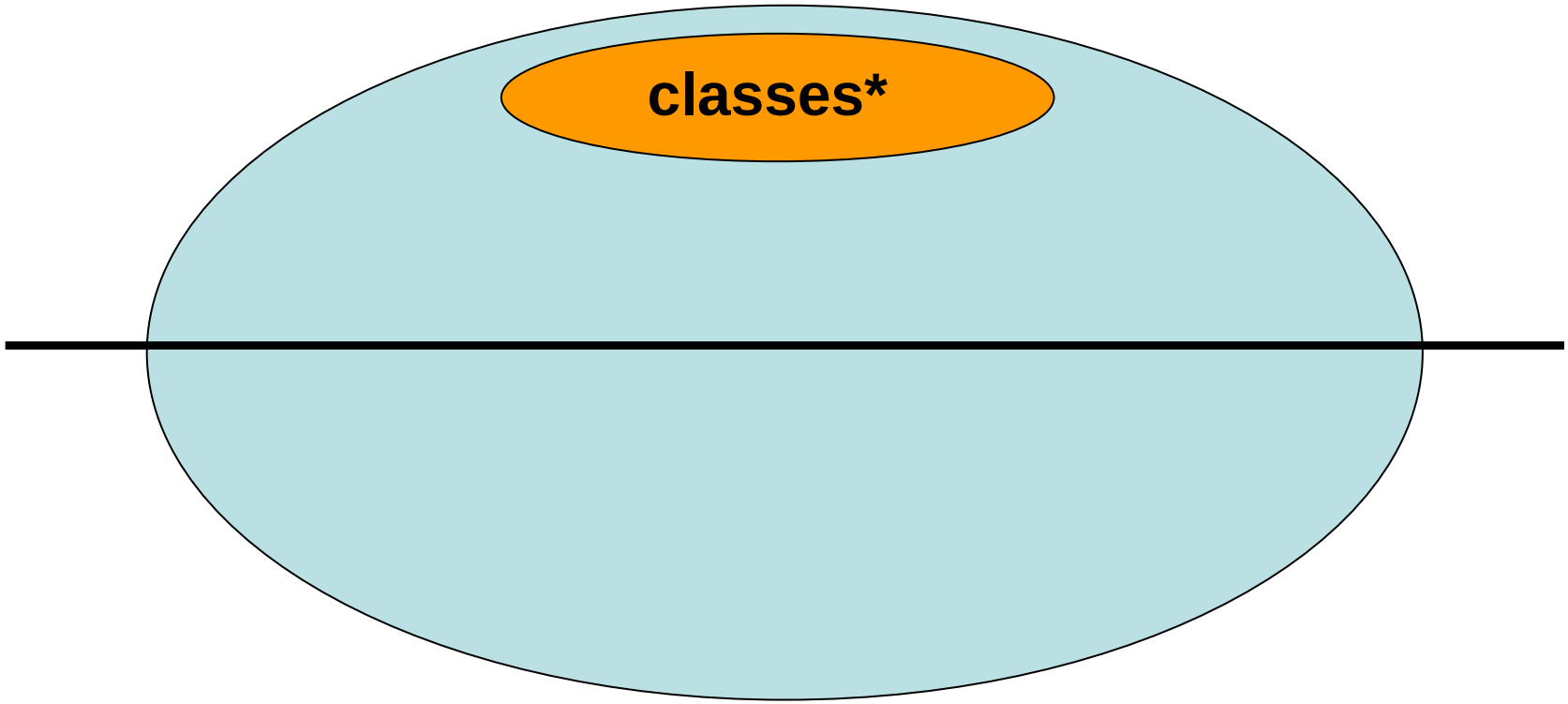
classes, within the realm of universals

instances within the realm of particulars

Entities

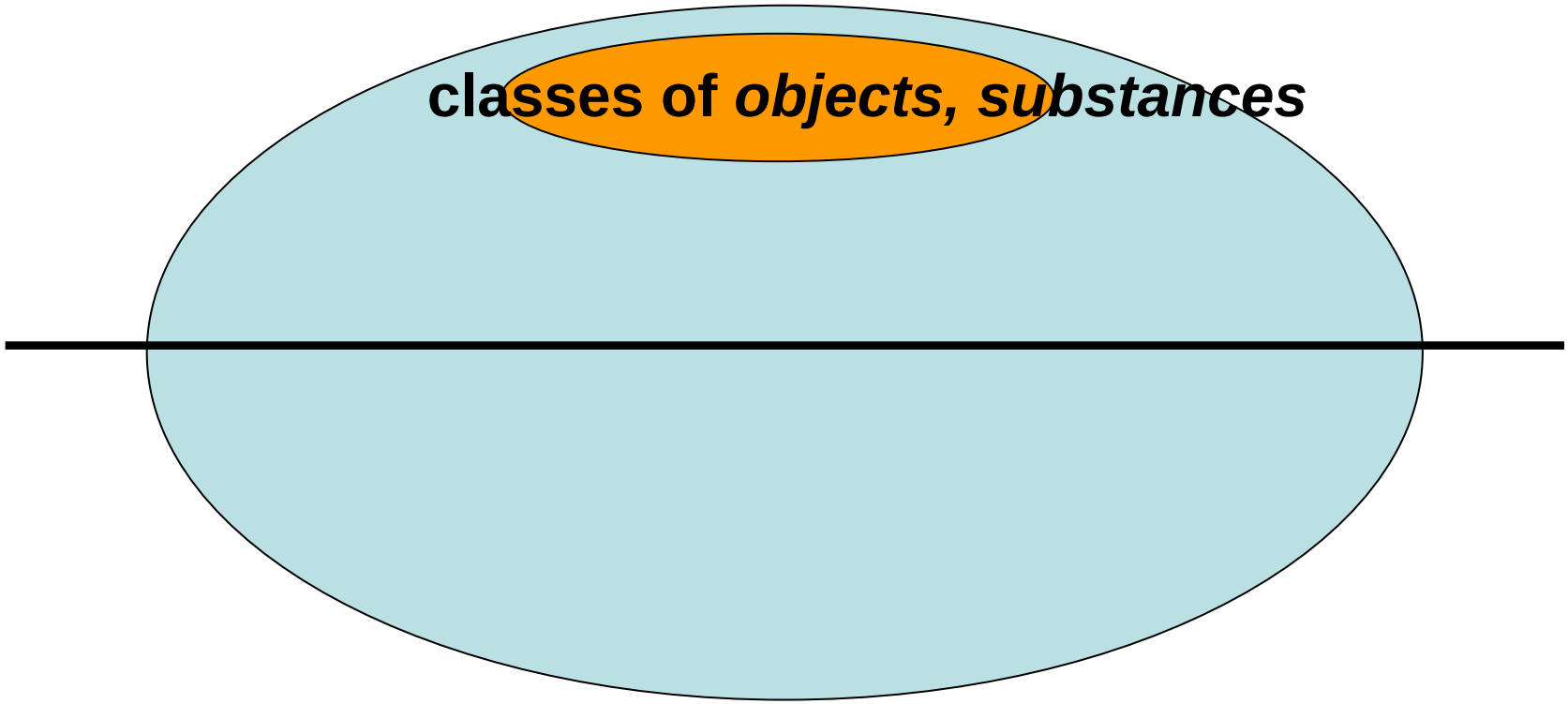


Entities



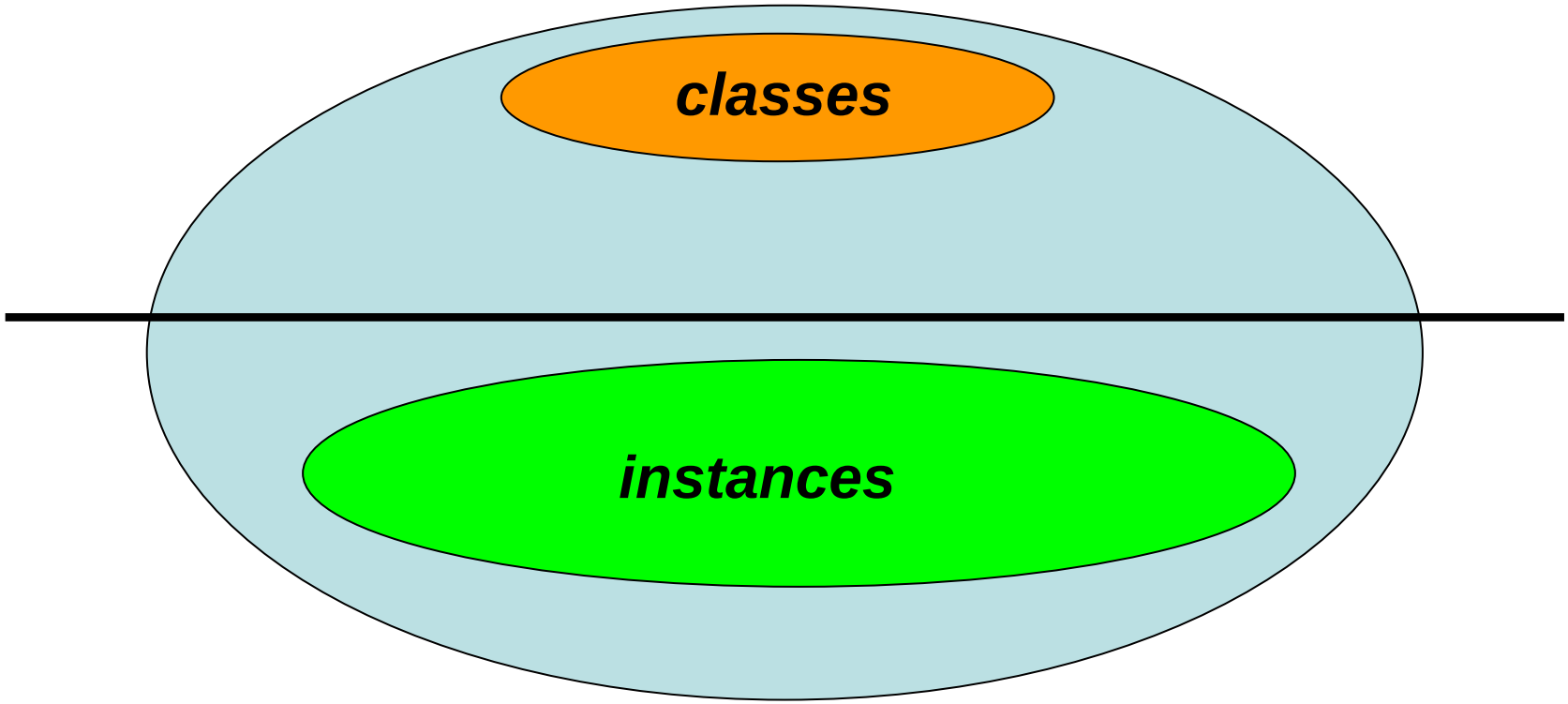
***natural, biological**

Entities



need modified axioms for classes of functions, processes, pathways, reactions, etc.

Entities



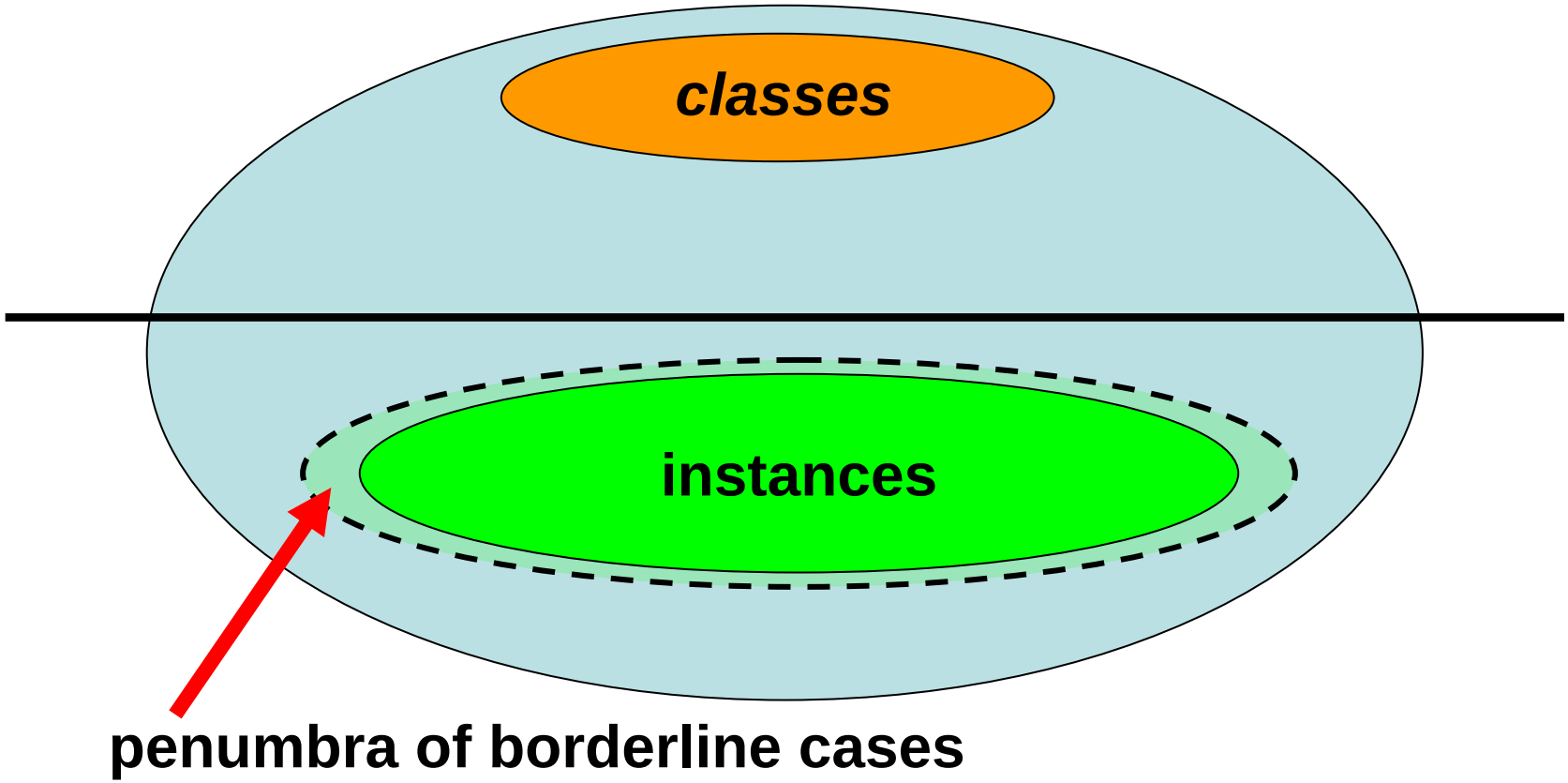
Classes are *natural* kinds

Instances are *natural* exemplars of *natural* kinds

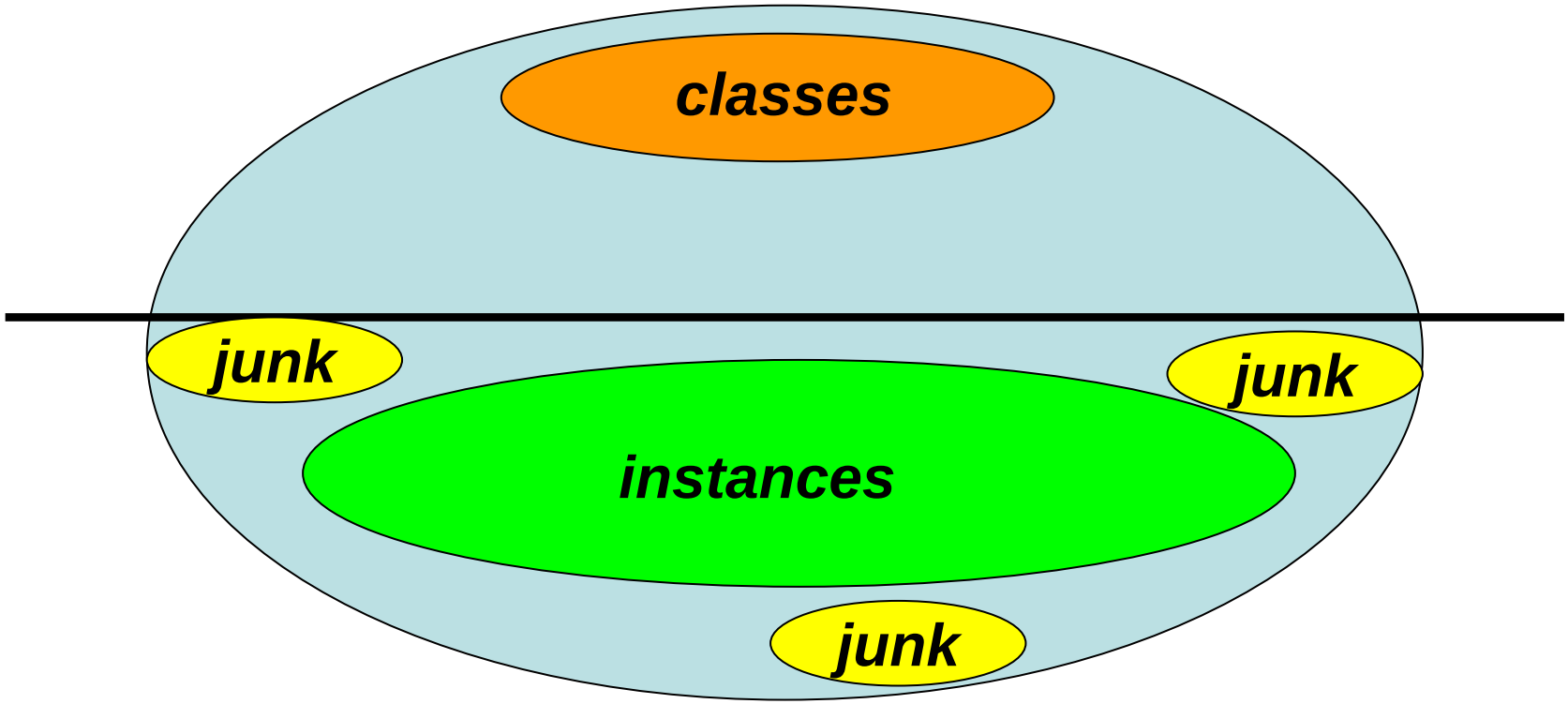
(problem of non-standard instances)

Not all individuals are instances of classes

Entities



Entities



example of junk: beachball-desk

Primitive relations: ***inst*** and ***part***

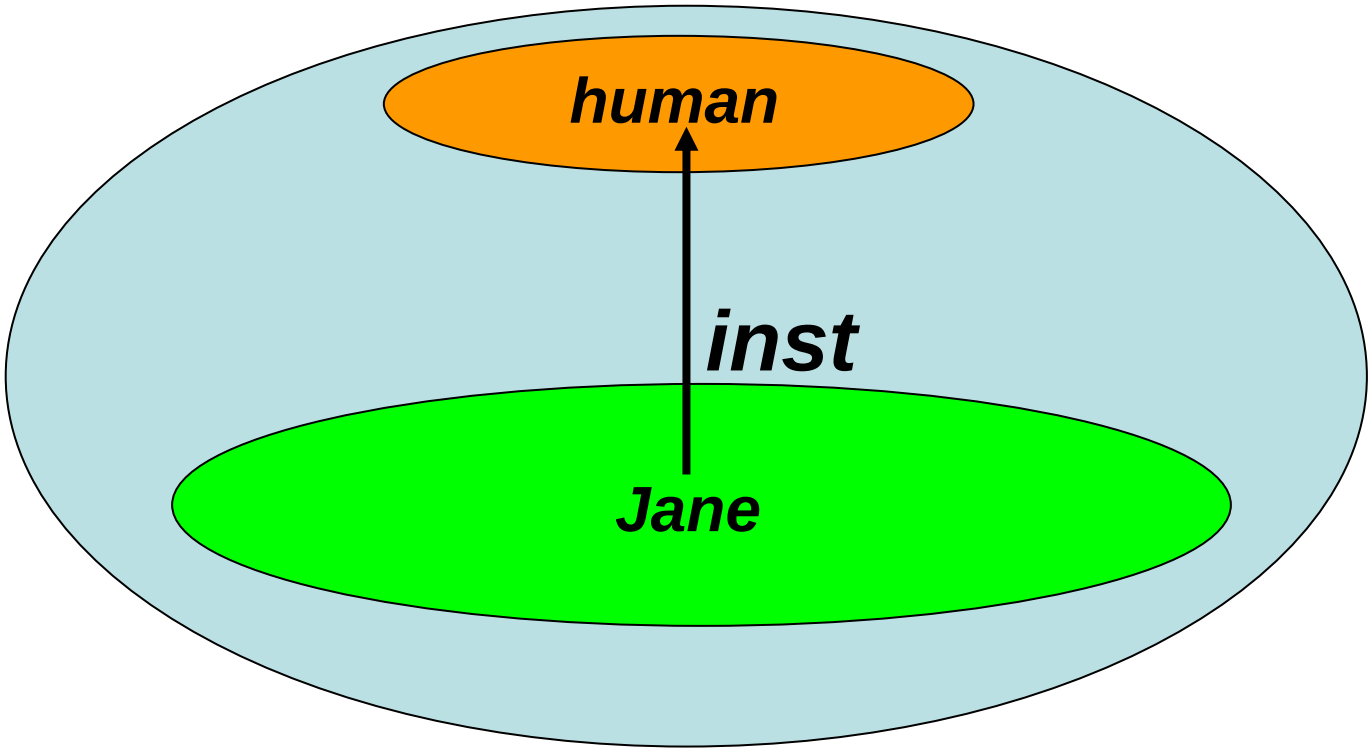
inst(Jane, human being)

part(Jane's heart, Jane's body)

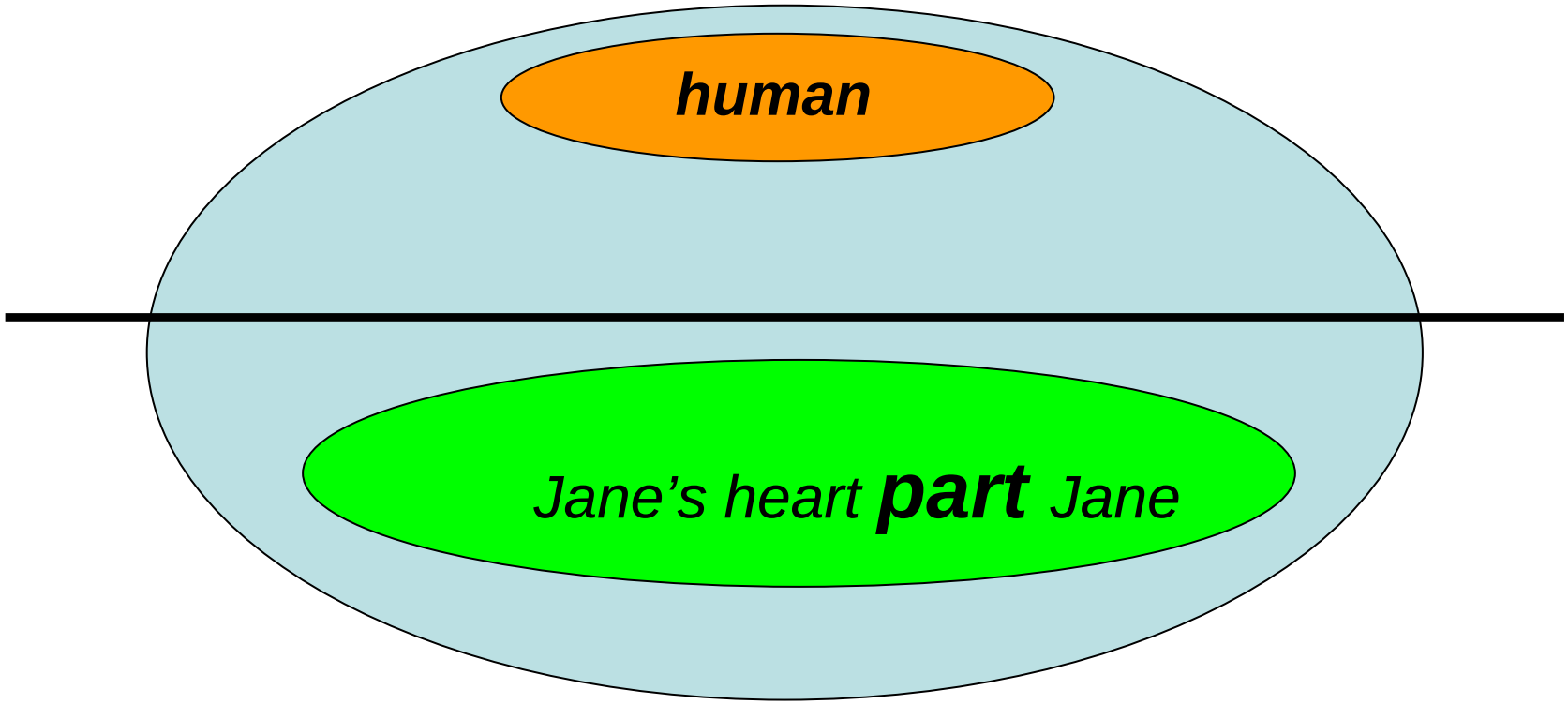
A class is anything that is instantiated

An instance as anything (any individual) that instantiates some class

Entities



Entities



part *as a relation between individuals*

subject to the **usual axioms of mereology**

Two primitive relations: ***inst*** and ***part***

inst(Jane, *human being*)

part(Jane's heart, Jane's body)

A universal is anything that is instantiated

An instance is anything (any individual) that instantiates some class

Two primitive relations: ***inst*** and ***part***

Axioms governing ***inst*** :

(1) it holds in every case between an instance and a class, in that order;

(2) that nothing can be both an instance and a class.

Axioms governing ***part*** (= ‘proper part’)

(1) it is *irreflexive*

(2) it is *asymmetric*

(3) it is *transitive*

(4) it holds only between individuals

(usual mereological axioms)

Part_for and Has_Part

A part_for B $=_{\text{def}}$
given any *x,* if ***inst***(*x*, *A*) then there is some *y* such that ***inst***(*y*, *B*) and ***part***(*x*, *y*)

B has_part A $=_{\text{def}}$
given any y, if ***inst***(y, *B*) then there is some x such that ***inst***(x, *A*) and ***part***(*x*, *y*)

human testis part_for human being,
But not: *human being has_part human testis*.
human being has_part heart,
But not: *heart part_for human being.*

The usual part_of relation as a relation between universals

A part_of B $=_{\text{def}}$ *A part_for B & B has_part A*

*A*s exist only as parts of *B*s and *B*s are structurally organized in such a way that *A*s must appear in them as parts.

Analogous problems for nearly all foundational relations of ontologies and semantic networks:

A causes B

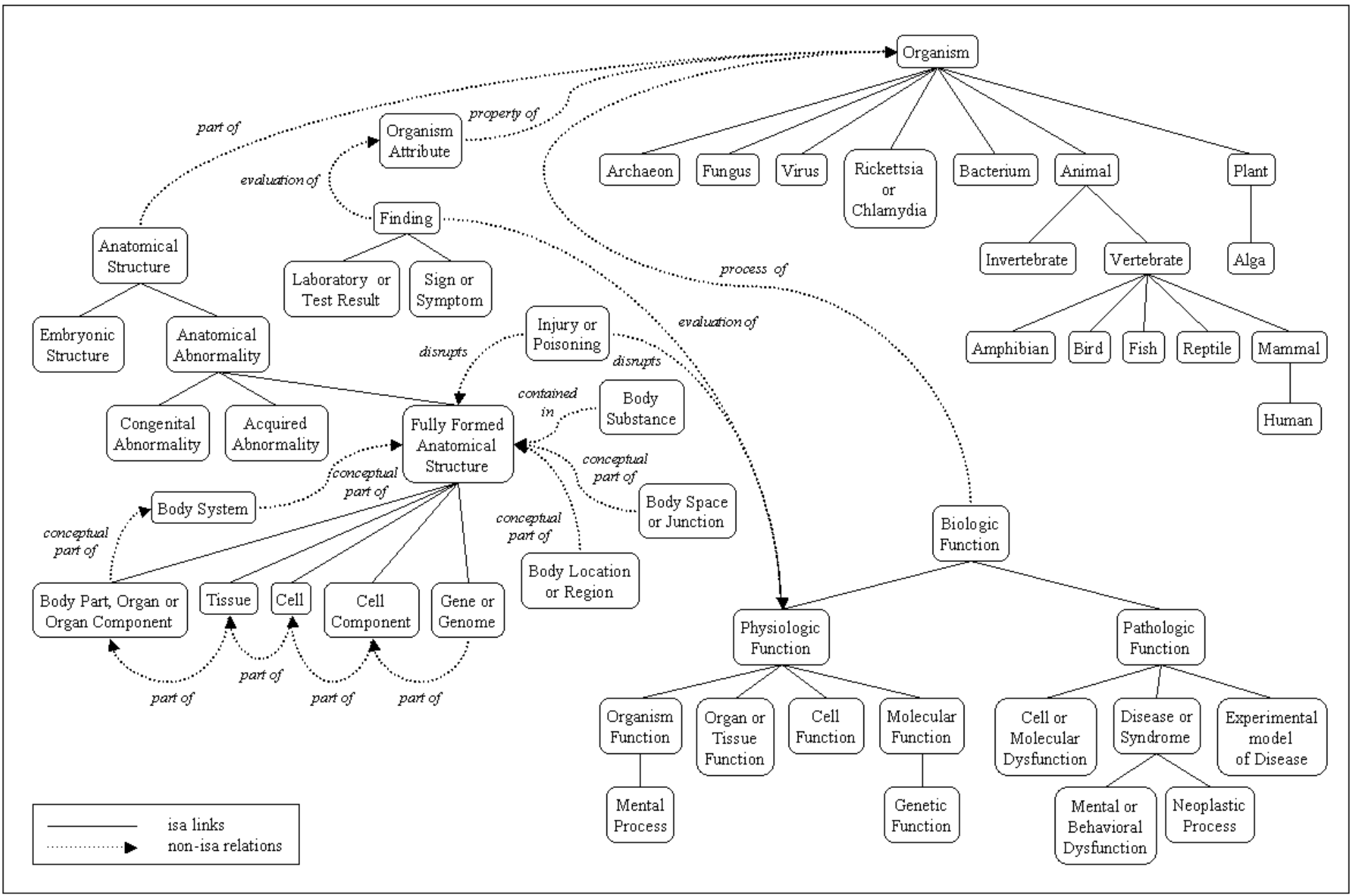
A is associated with B

A is located in B

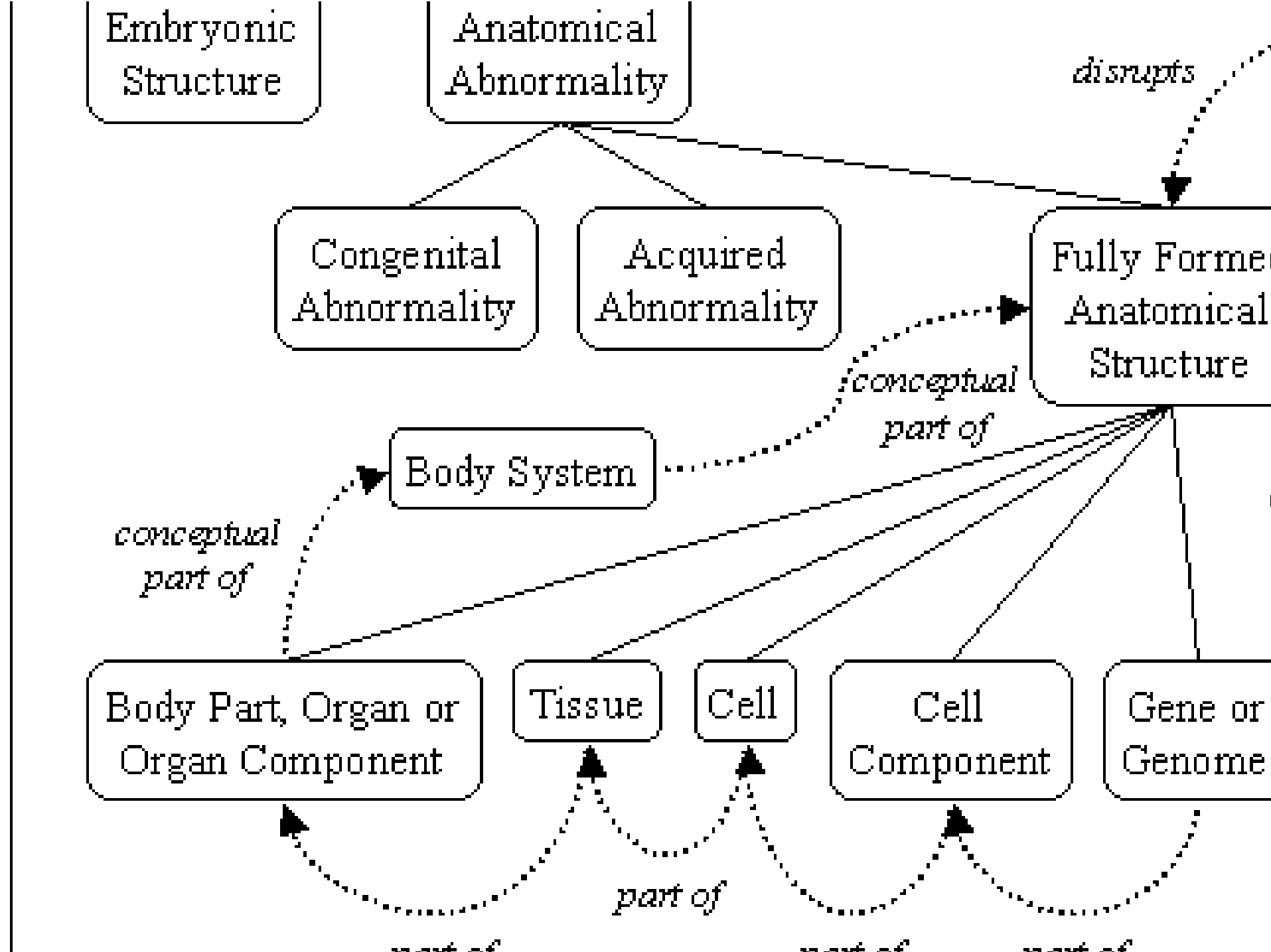
etc.

Reference to instances is necessary to clear up these problems

if they can be cleared up at all …



Fragment of the UMLSemantic Network


Embryonic Structure
Anatomical Abnormality
disrupts
Congenital Abnormality
Acquired Abnormality
Fully Formed Anatomical Structure
conceptual part of
Body System
conceptual part of
Body Part, Organ or Organ Component
Tissue
Cell
Cell Component
Gene or Genome
part of

Mental Process ***precedes*** Molecular Function

Mental Process ***precedes*** Genetic Function

Experimental Model of Disease ***precedes*** Cell or Molecular Dysfunction

Acquired Abnormality ***affects*** Bird

Experimental Model of Disease ***affects*** Fungus

Physiologic Function ***affects*** Reptile

Antibiotic ***causes*** Experimental Model of Disease

Biomedical or Dental Material ***causes*** Mental or Behavioral Dysfunction

Manufactured Object ***causes*** Disease or Syndrome

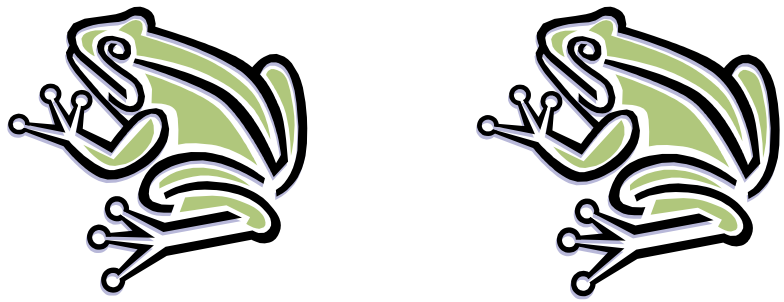
Vitamin ***causes*** Injury or Poisoning

Fungus ***location_of*** Vitamin

Organization ***location_of*** Diagnostic Procedure

What are universals?

invariants in reality



satisfying biological laws

(there are truths about universals in biological textbooks)

Universals are Not Sums

Universals are distinguished by granularity: they divide up the corresponding domain into whole units or members, whose interior parts and structure are traced over. The *universal* human being is instantiated only by human beings as single, whole units.

A mereological sum is not granular in this sense (molecules are parts of the mereological sum of human beings)

Universals are Not Sets

Both universals and sets are marked by granularity – but universals are timeless

Both a universal and a set is laid across reality like a grid consisting (1) of a number of slots or pigeonholes each (2) occupied by some member.

But a set is *determined by its members*. This means that it is (1) associated with a specific number of slots, each of which (2) must be occupied by some specific member.

A universal survives the turnover in its instances: it is specified neither (1) what the number of associated slots should be nor (2) what individuals should occupy these slots. *Both may vary with time*.

A universal is not determined by its instances as a state is not determined by its citizens

A universal may vary with time as an organism may vary with time (by gaining and losing molecules)

Universals are Not Sets

A set is an abstract structure, existing outside time and space. The set of Romans timelessly has Julius Caesar as a member.

Universals *exist in time*.



Two Questions

1. What does “Functional” mean in expressions like “Functional Genomics” ?

2. How can we use the answer to this question to help us understand notions fundamental to medicine such as “health” and “disease” ?

Towards an Tri-Categorial Ontology

of Structures, Functions and Processes

Definition of Function in UMLS Semantic Network

Functional Concept $=_{df}$ A concept which is of interest because it pertains to the carrying out of a process or activity.

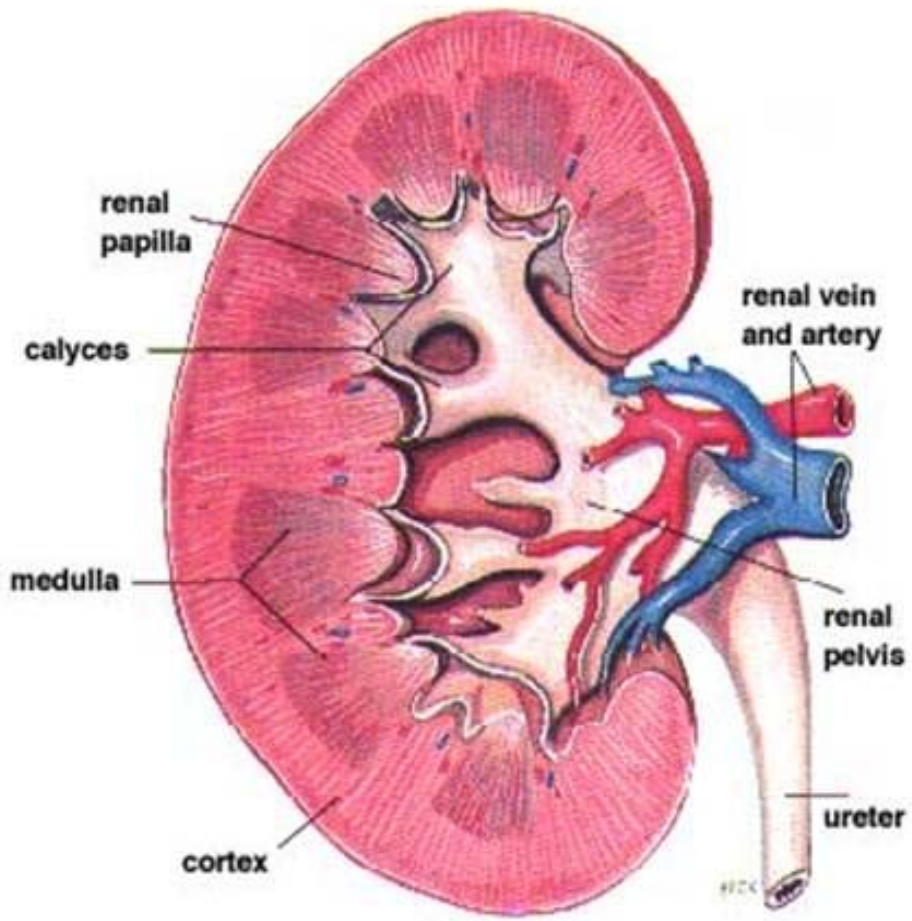
Function ≠ Functional *Concept*

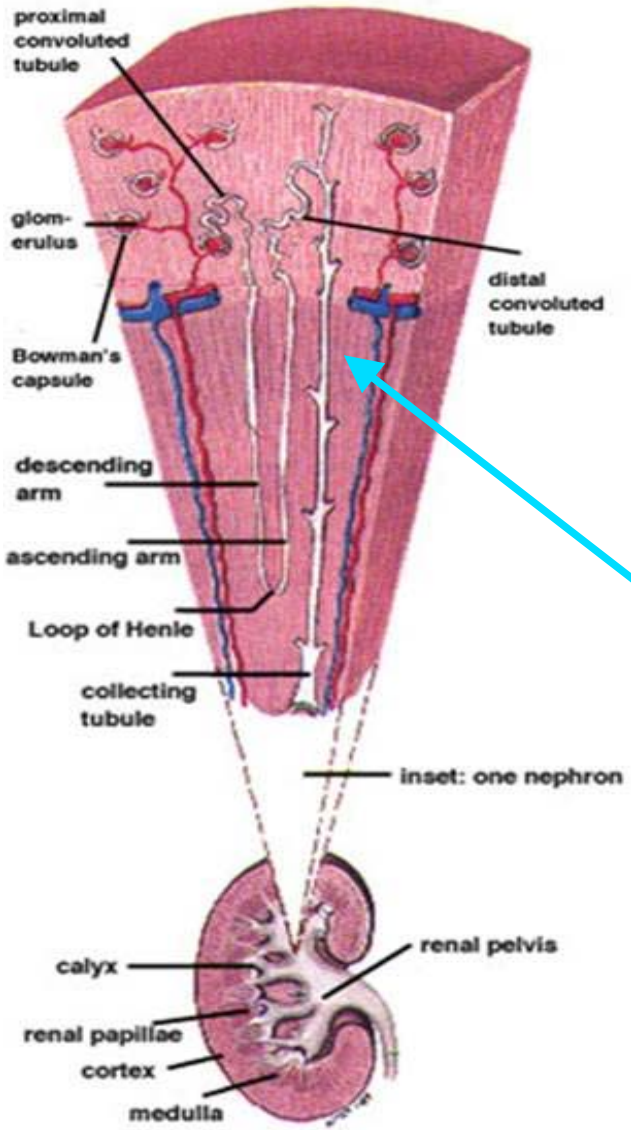
Function ≠ *Realization* of a Function

The Kidney

From Andrew Lonie, University of Melbourne

Your entire blood volume flows through your kidneys every few minutes, leaving behind excess water, solutes and waste materials



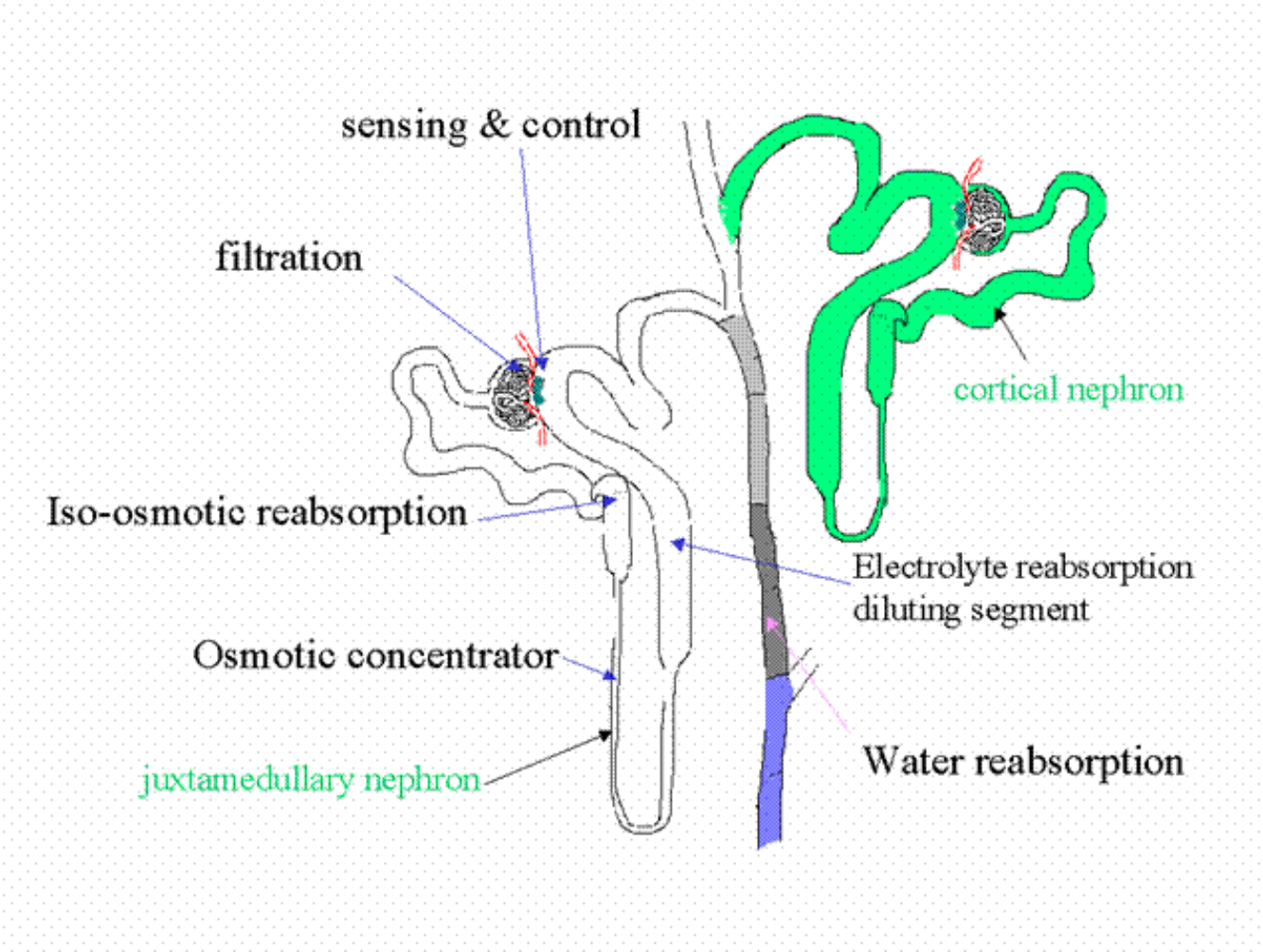


Essentially a massively parallel filter composed of 10^5 to 10^6 nephrons

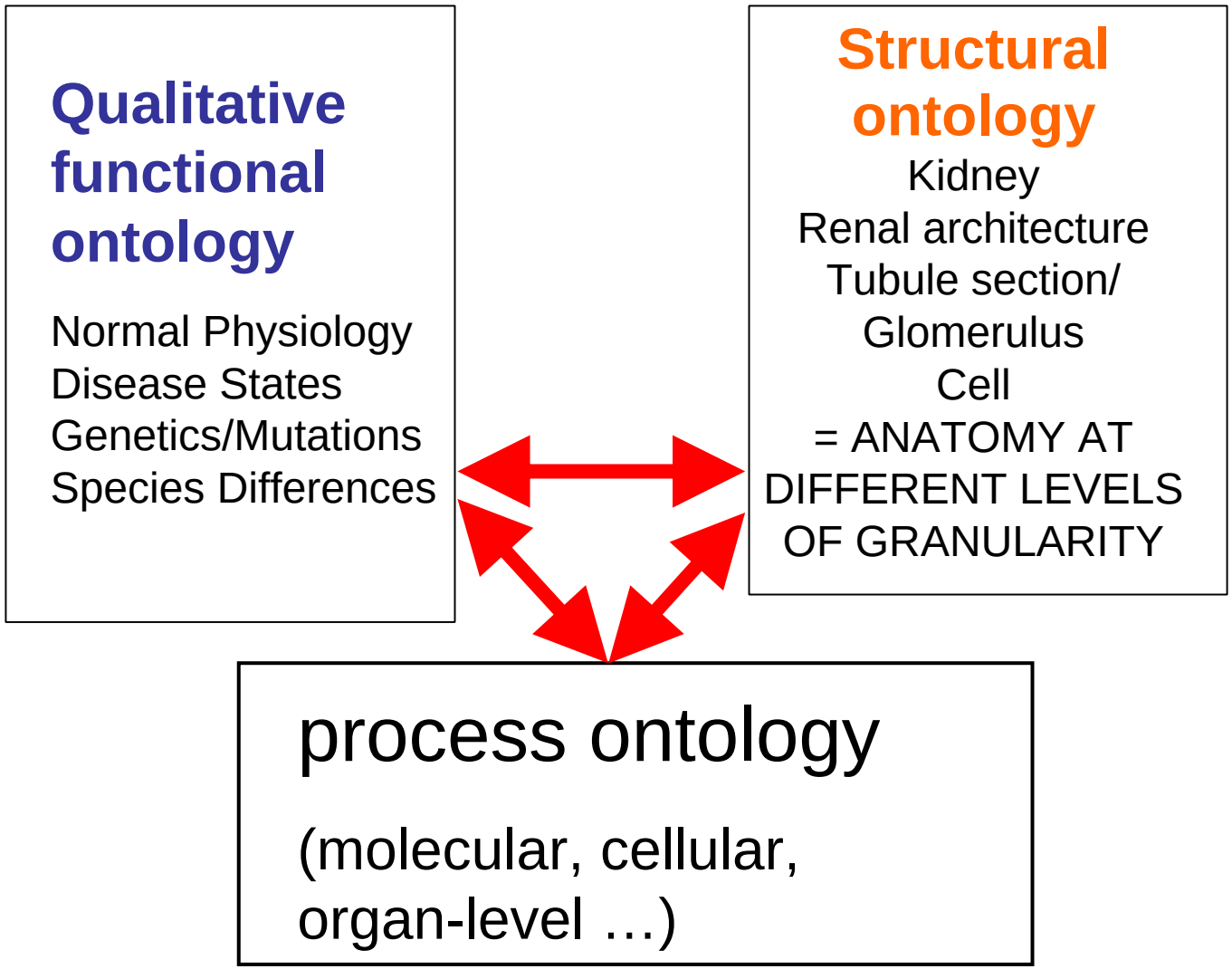
The *nephron* is the functional unit of the kidney

Each *nephron* is a very convoluted, long, thin tube lined with biochemical pumps

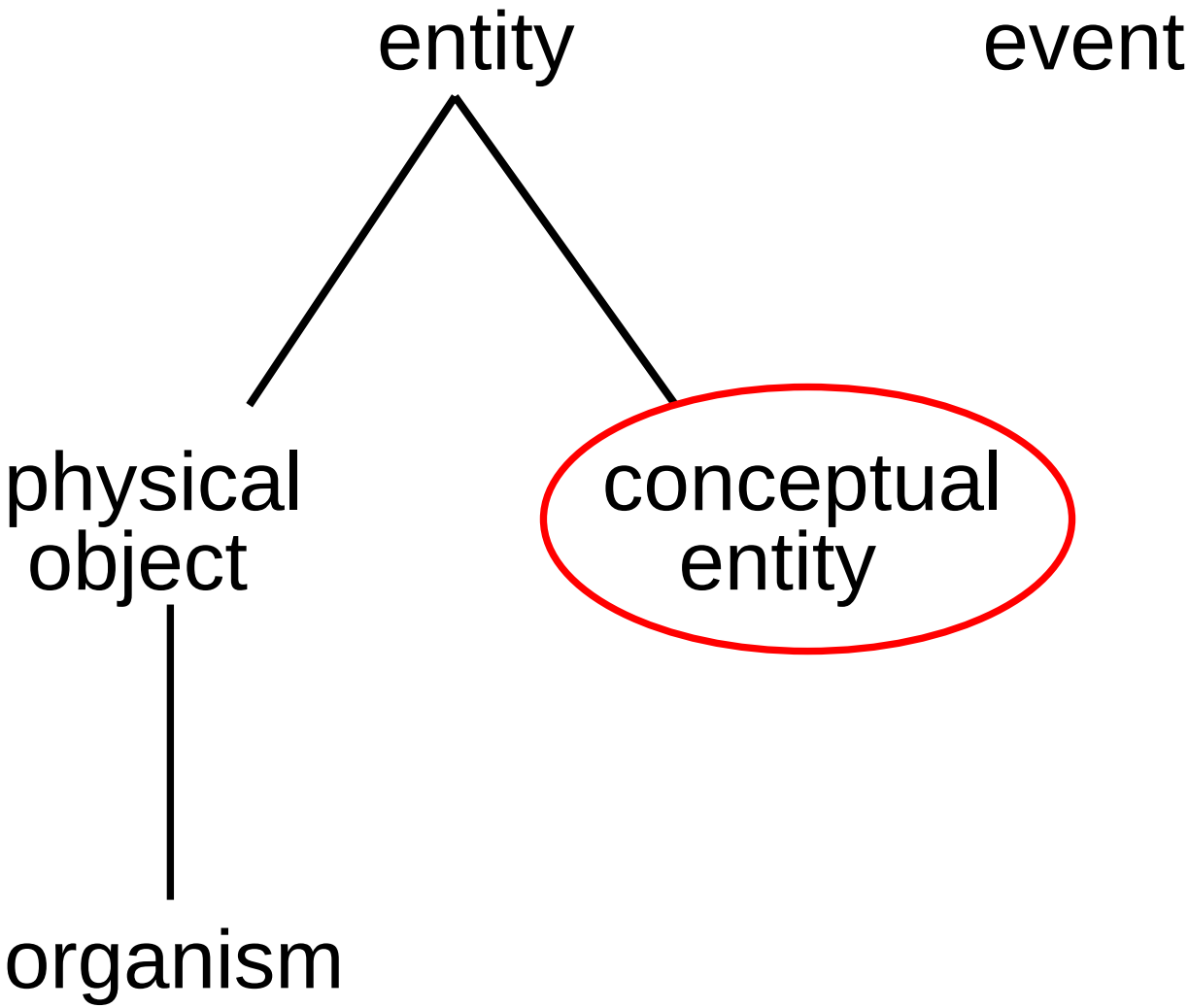
- 10 functional segments
- 15 different cell types



Structural and functional representation



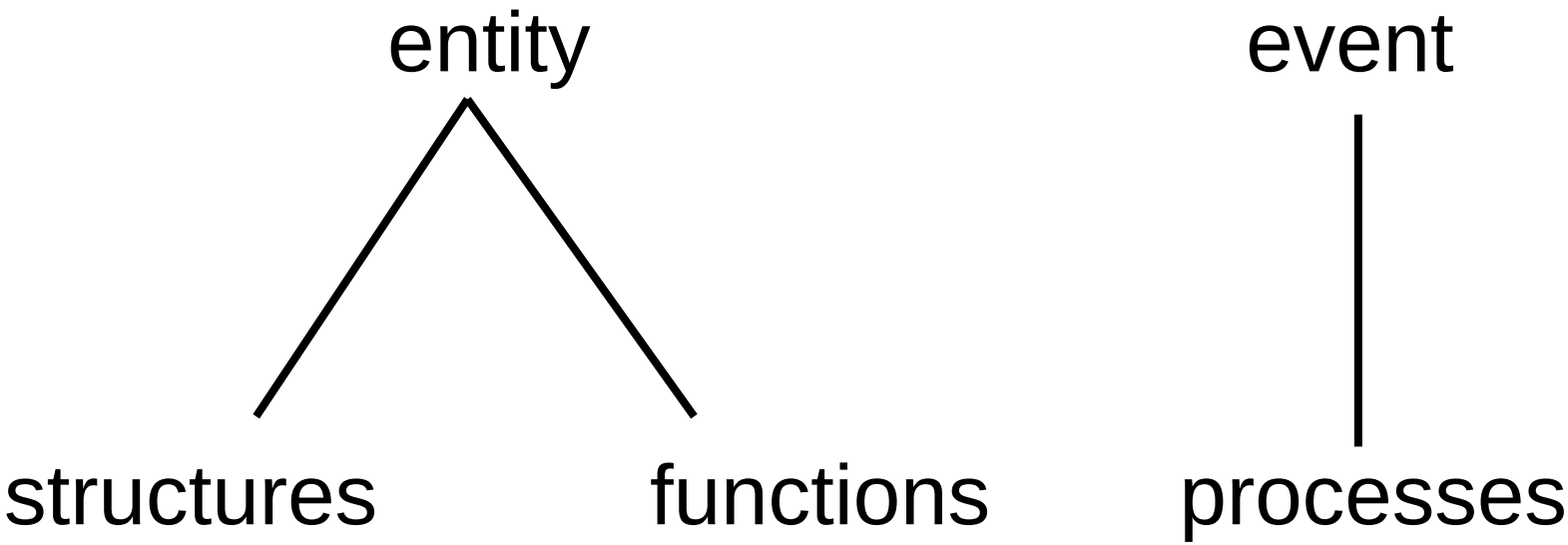
UMLS Semantic Network



Tri-Categorial Ontology present also in GO: The Gene Ontology

3 ‘ontologies’ (large telephone directories) of standardized designations for gene functions and products

RUMLS Semantic Network



GO's three disjoint term hierarchies

the *cellular component* (***structure***) *ontology*,

e.g. flagellum, chromosome, cell

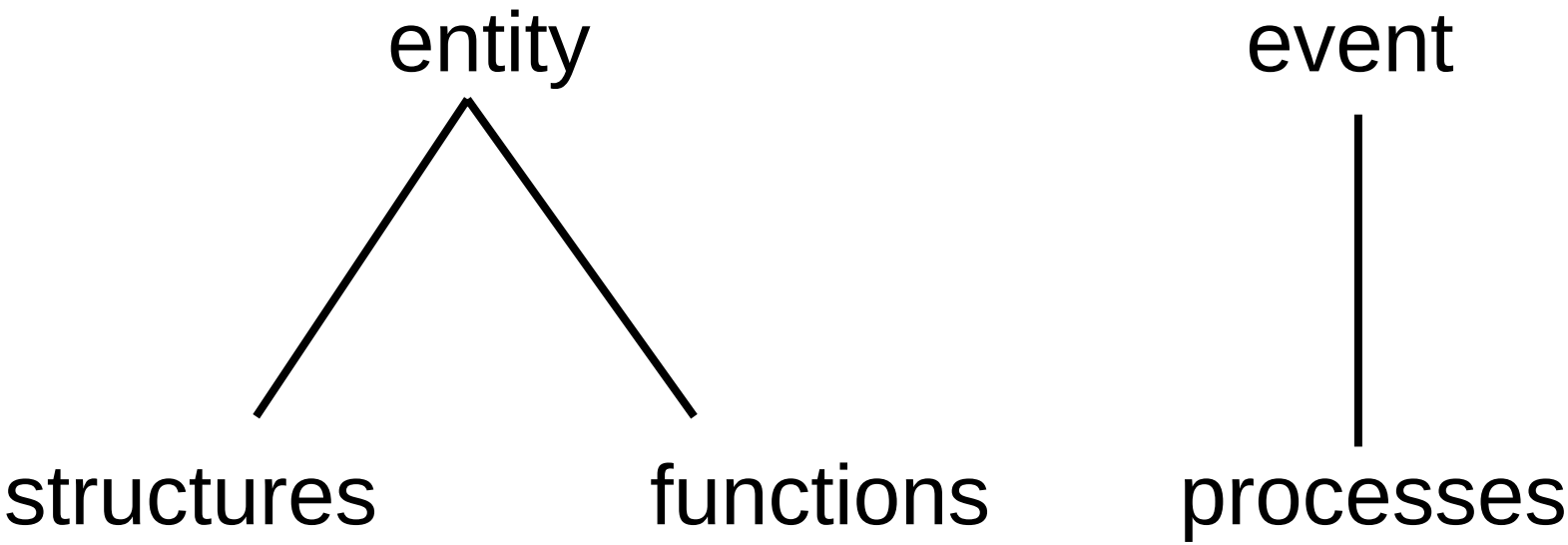
the *biological **process** ontology,*

e.g. glycolysis, death

the *molecular **function** ontology,*

e.g. ice nucleation, binding, protein stabilization

RUMLS Semantic Network



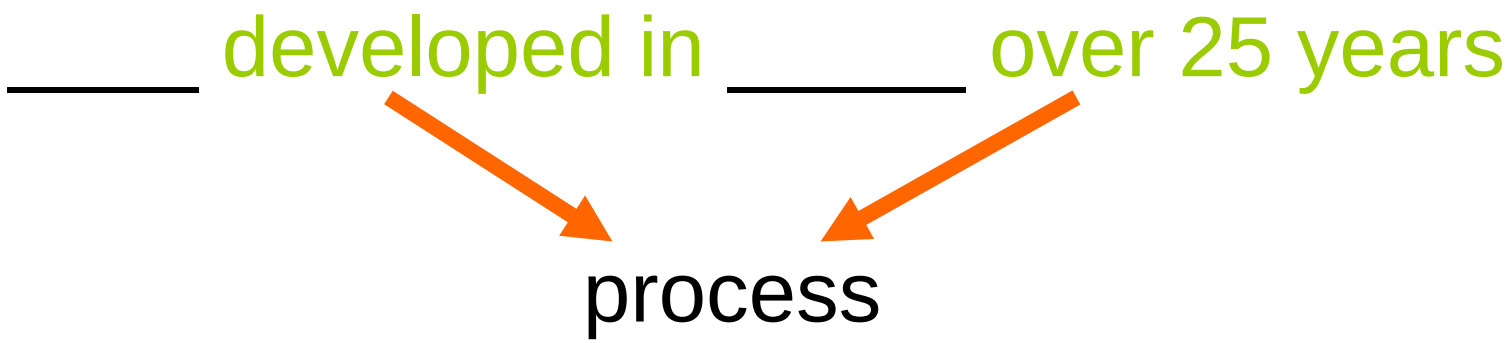
Functional Genomics

What does “Functional“ mean?

The Problem

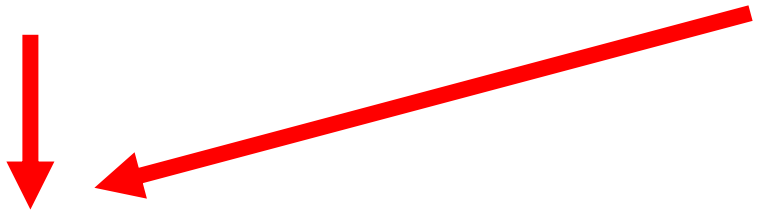
The tumor developed in John’s lung over 25 years

The Problem



The Problem

The tumor developed in the lung over 25 years



substances

things

objects

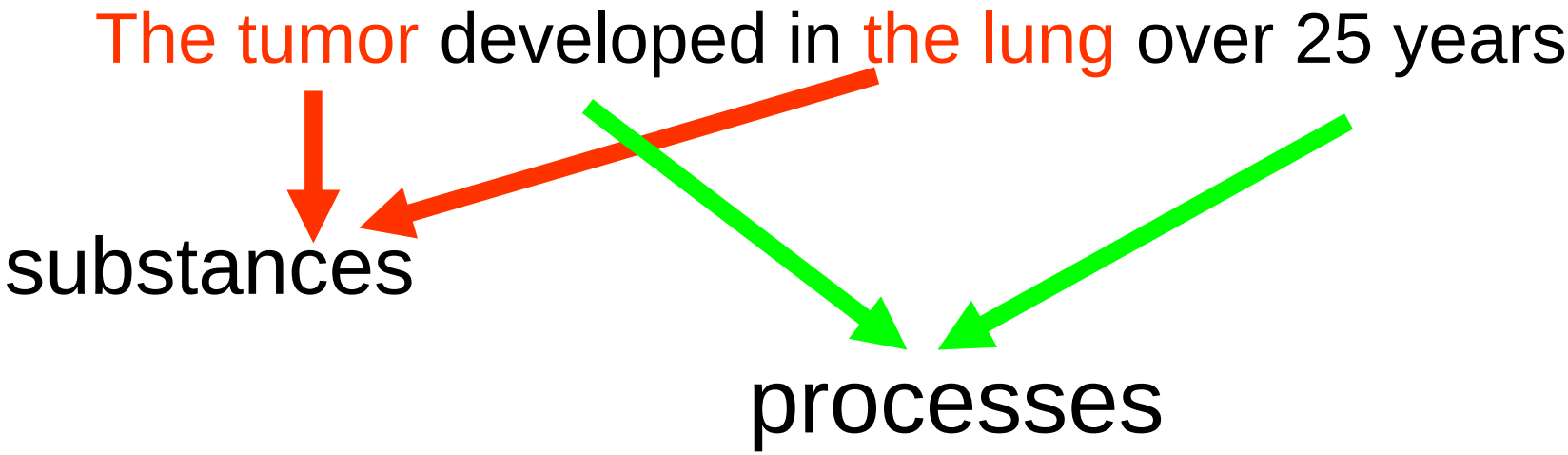
continuants

The Problem

The tumor developed in John's lung over 25 years

PARTHOOD NOT DETERMINATE

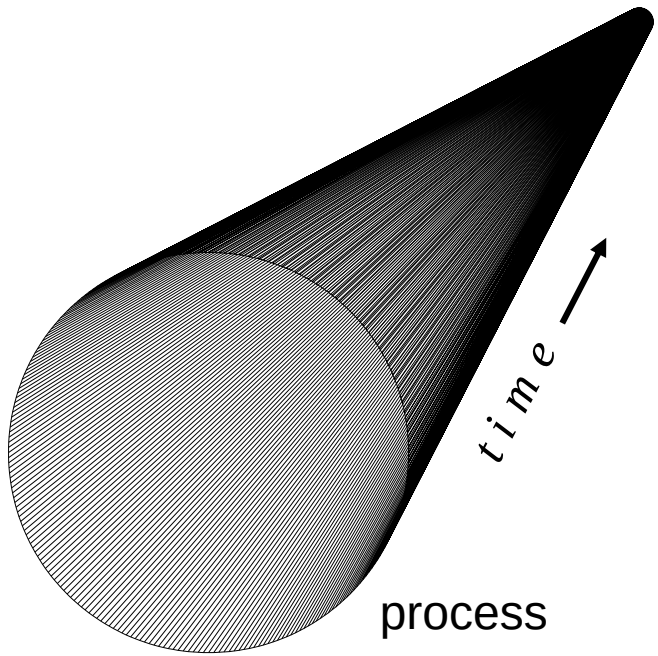
The Problem



GLUING THESE TOGETHER YIELDS ONTOLOGICAL MONSTERS

Substances and processes exist in time in different ways

substance



process

SNAP vs SPAN

Endurants vs perdurants

Continuants vs occurrents

In preparing an inventory of reality

we keep track of these two different kinds of entities in two different ways

Fourdimensionalism

- only processes exist
- time is just another dimension, analogous to the three spatial dimensions
- substances are analyzed away as worms/fibers within the four-dimensional plenum

There are no substances

Bill Clinton does not exist

Rather: there exists within the four-dimensional plenum a continuous succession of processes which are similar in a Billclintonizing way

Fourdimensionalism (the SPAN perspective) is right in everything it says

But incomplete

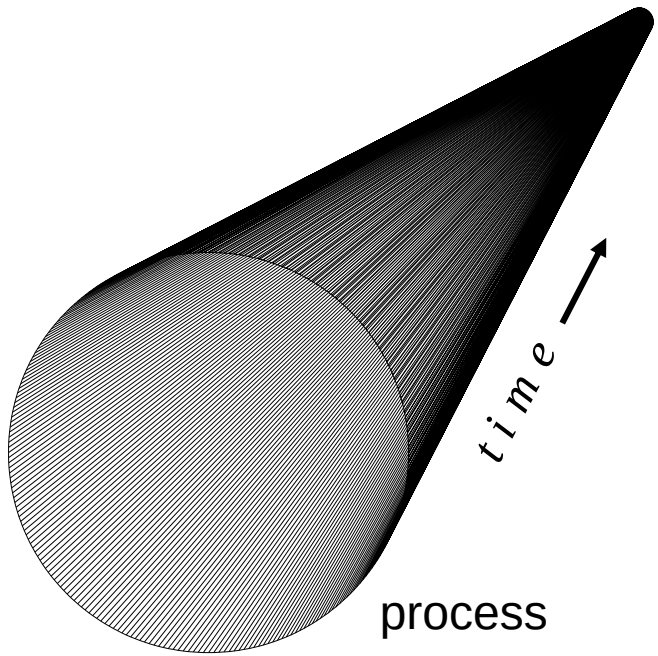
Need for Two Orthogonal, Complementary Perspectives

SNAP and SPAN

Snapshot ontology

substance

Video ontology



process

SNAP and SPAN

stocks and flows

commodities and services

product and process

anatomy and physiology

SNAP and SPAN

SNAP entities

- have continuous existence in time
- preserve their identity through change
- exist *in toto* if they exist at all

SPAN entities

- have temporal parts
- unfold themselves phase by phase
- exist only in their phases/stages

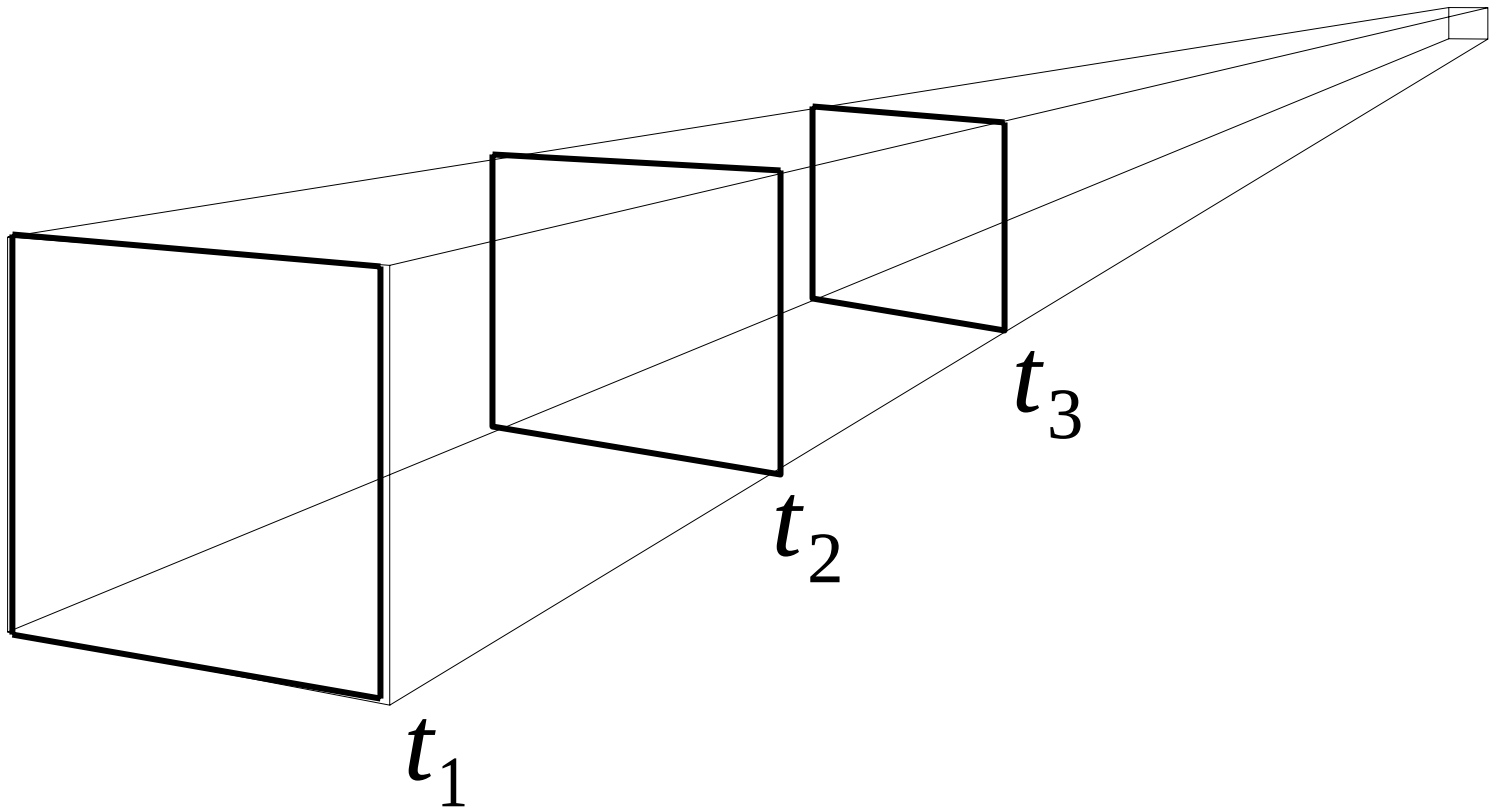
You are a substance

Your _life_ is a process

You are 3-dimensional

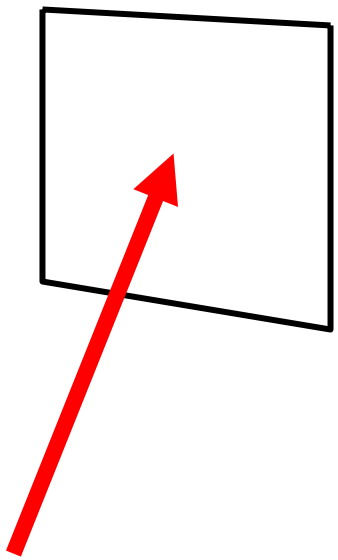
Your _life_ is 4-dimensional

Many SNAP Ontologies



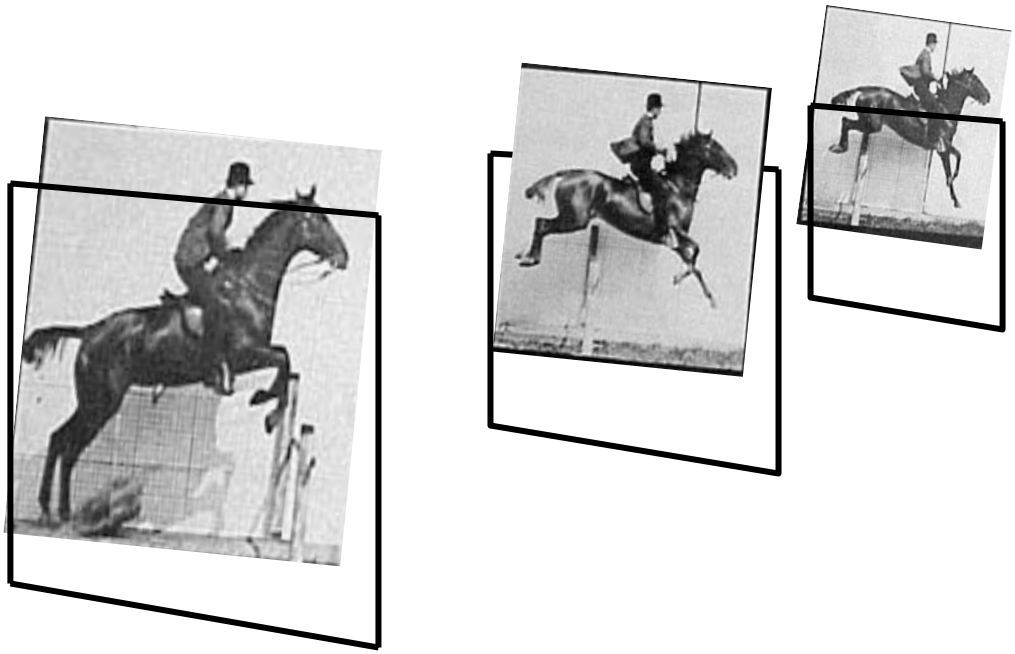
here time exists outside the ontology,
as an *index* or *time-stamp*

each $SNAP_i$ section through reality



includes everything which *exists*
(present tense)

mereology works without restriction (parthood is everywhere determinate) in every $SNAP_i$ ontology



Three kinds of SNAP entities

1. SNAP Independent Entities (you and me)
2. SNAP Dependent Entities
3. Spatial regions

SNAP dependent entities

States, powers, qualities, functions, dispositions, plans, shapes, liabilities, propensities…

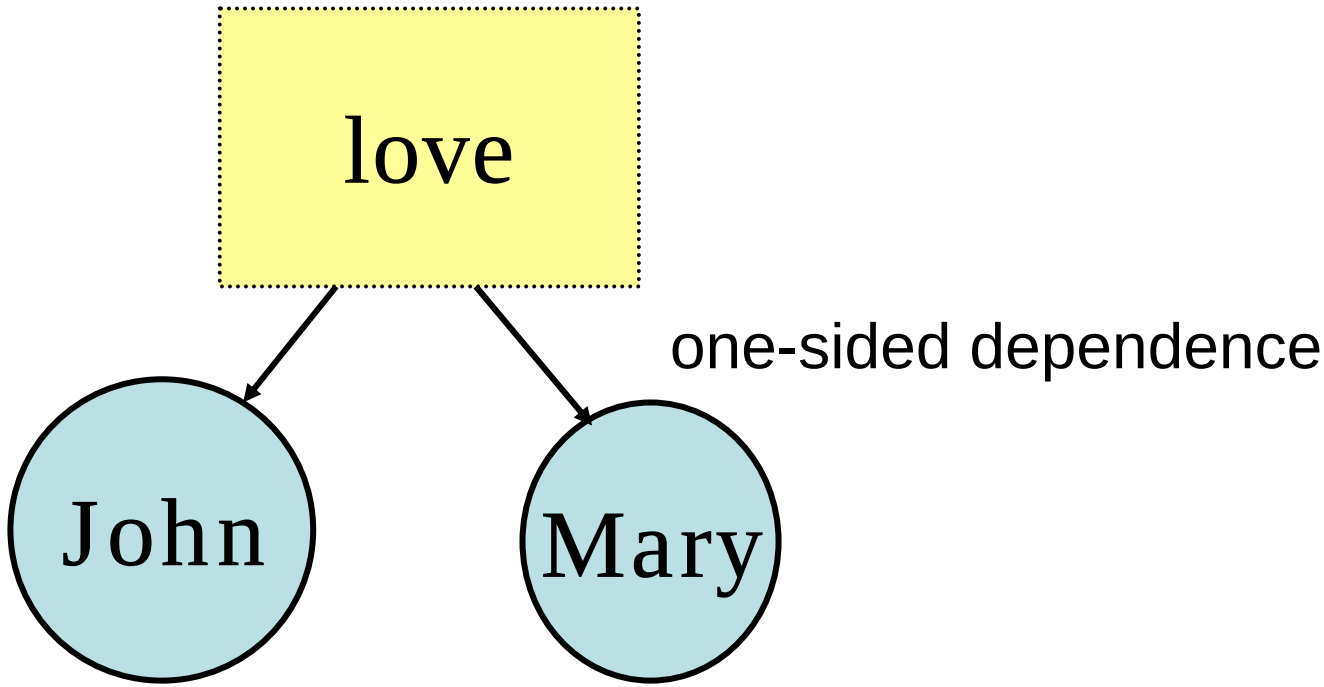
SNAP dependent entities:

one-place:

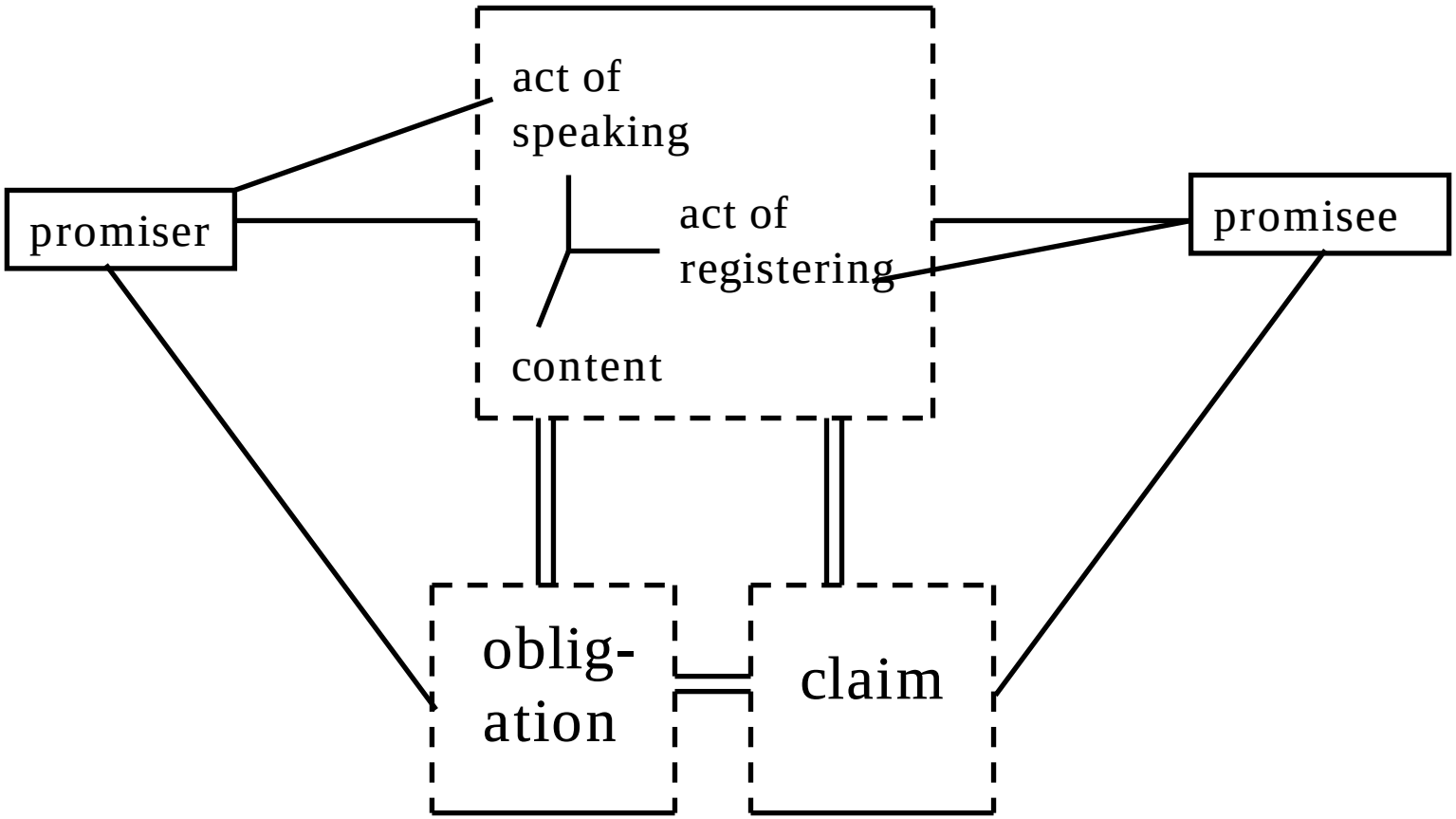
your temperature, color, height
my knowledge of French
the whiteness of this cheese
the warmth of this stone
the fragility of this glass

relational SNAP dependent entities

stand in relations of one-sided dependence to a plurality of substances simultaneously



A Window on Reality



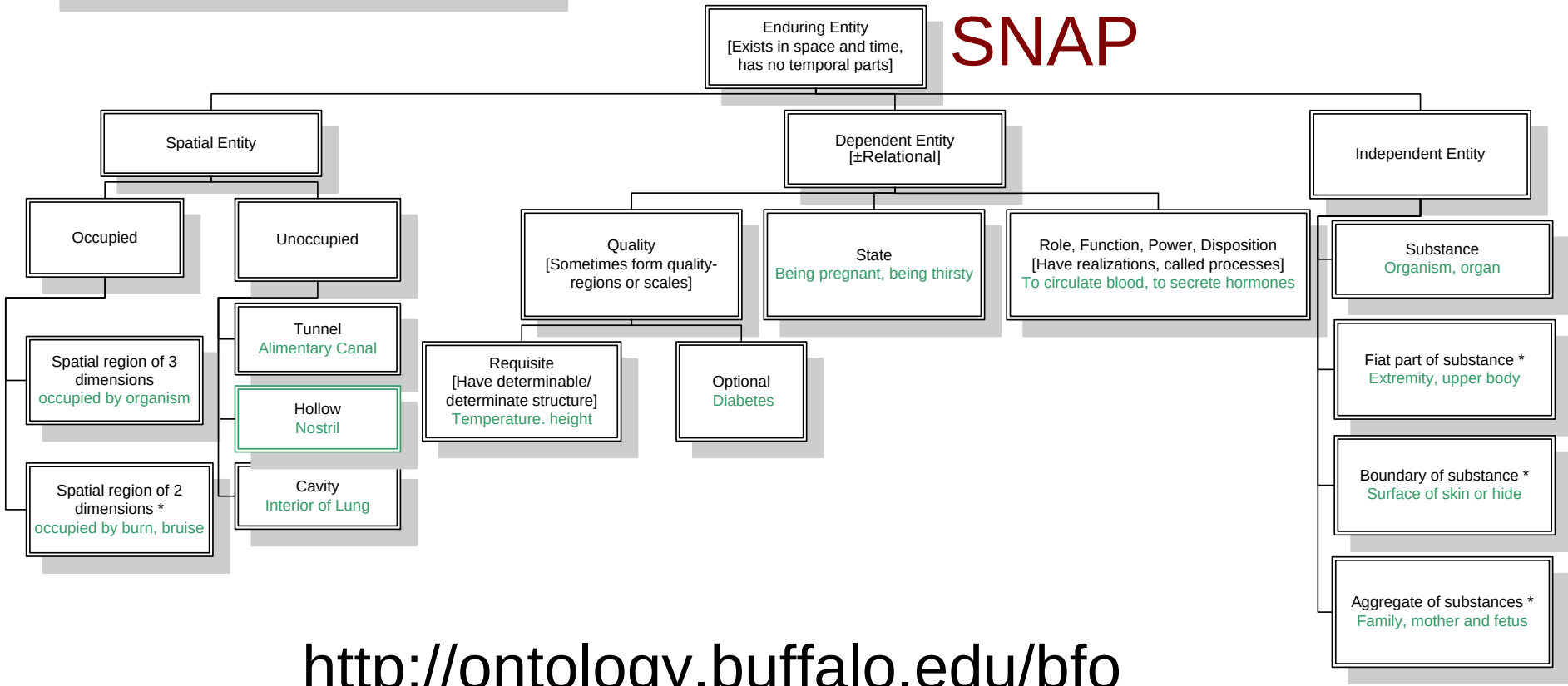
Spatial regions + sites (contexts, niches, environments)

Organism species *evolve into* environments

Domesticated spatial regions: rooms, nostrils, your alimentary tract

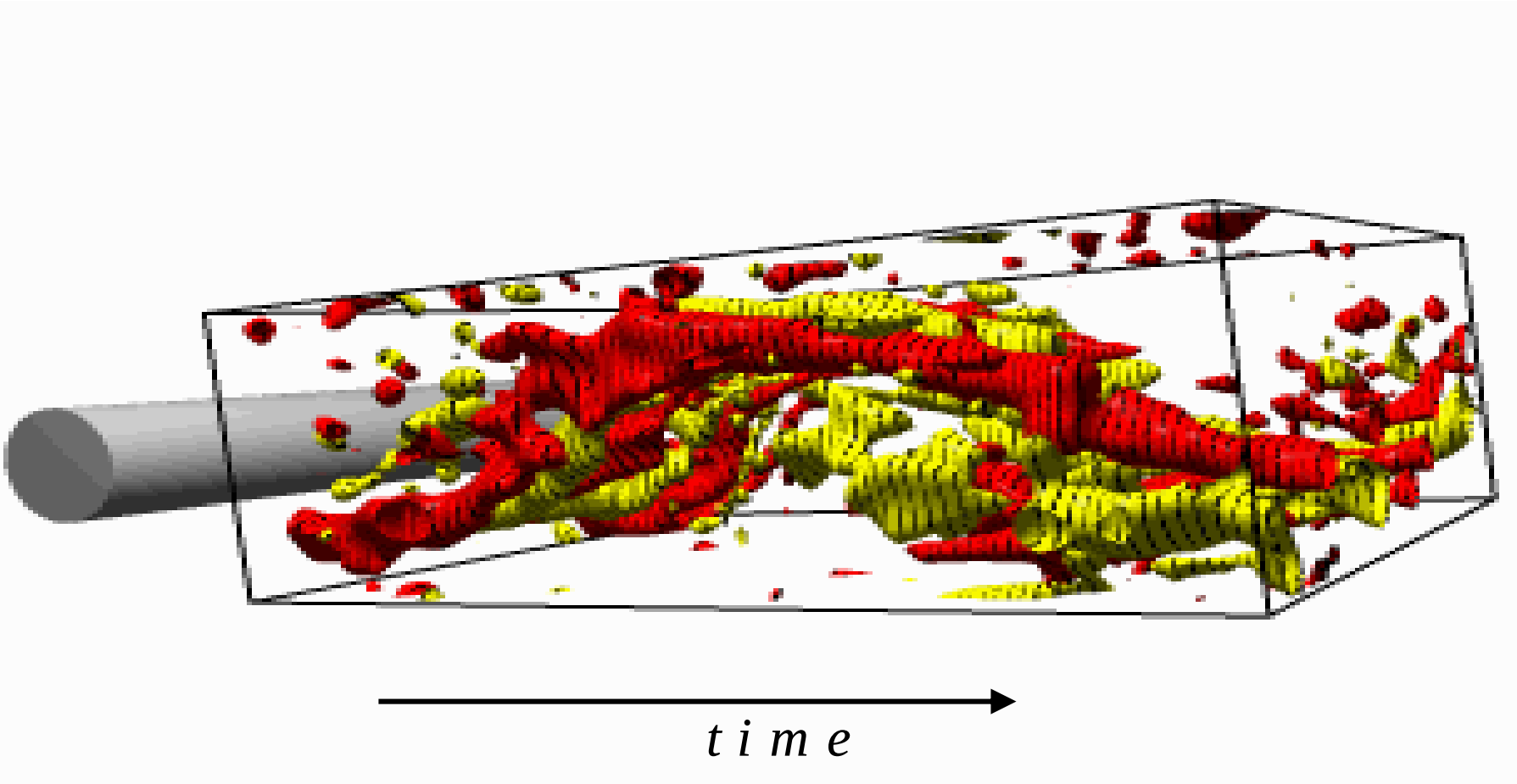
Fiat spatial regions: JFK designated airspace

SNAP: Entities existing *in toto* at a time

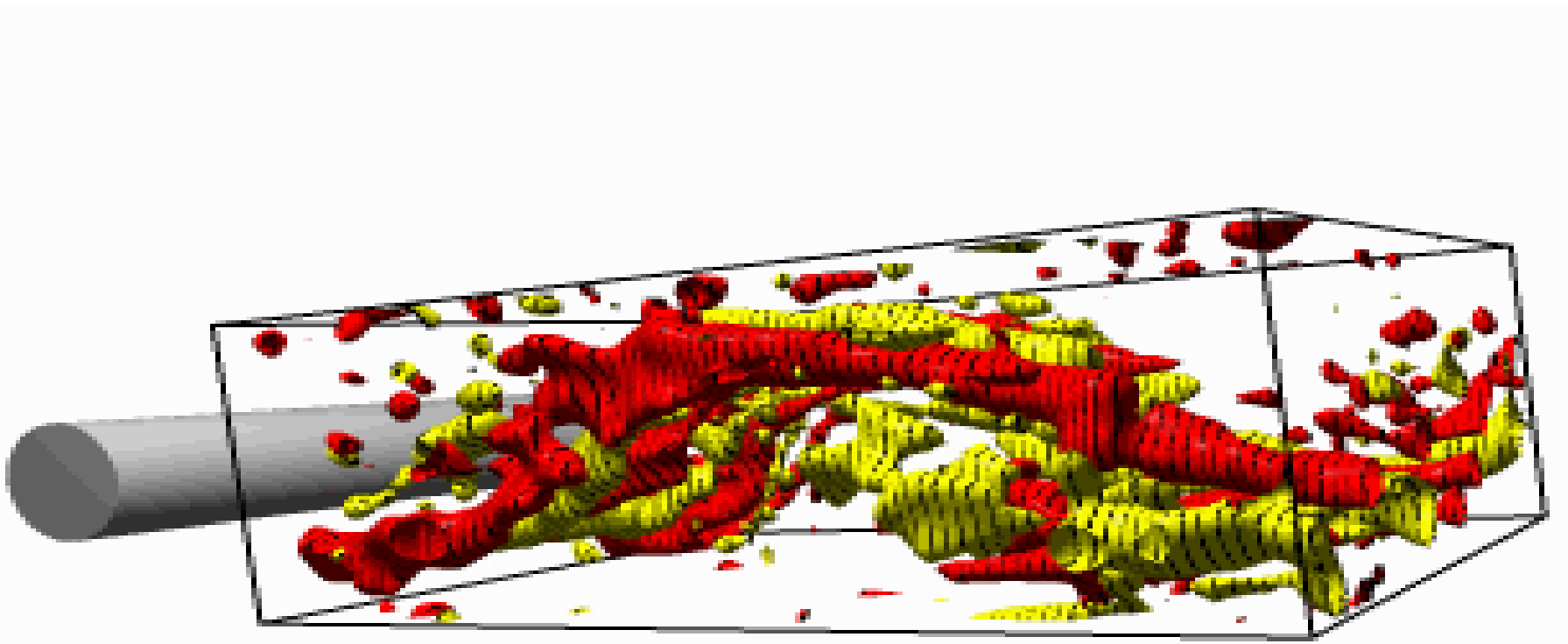


http://ontology.buffalo.edu/bfo

The SPAN Ontology

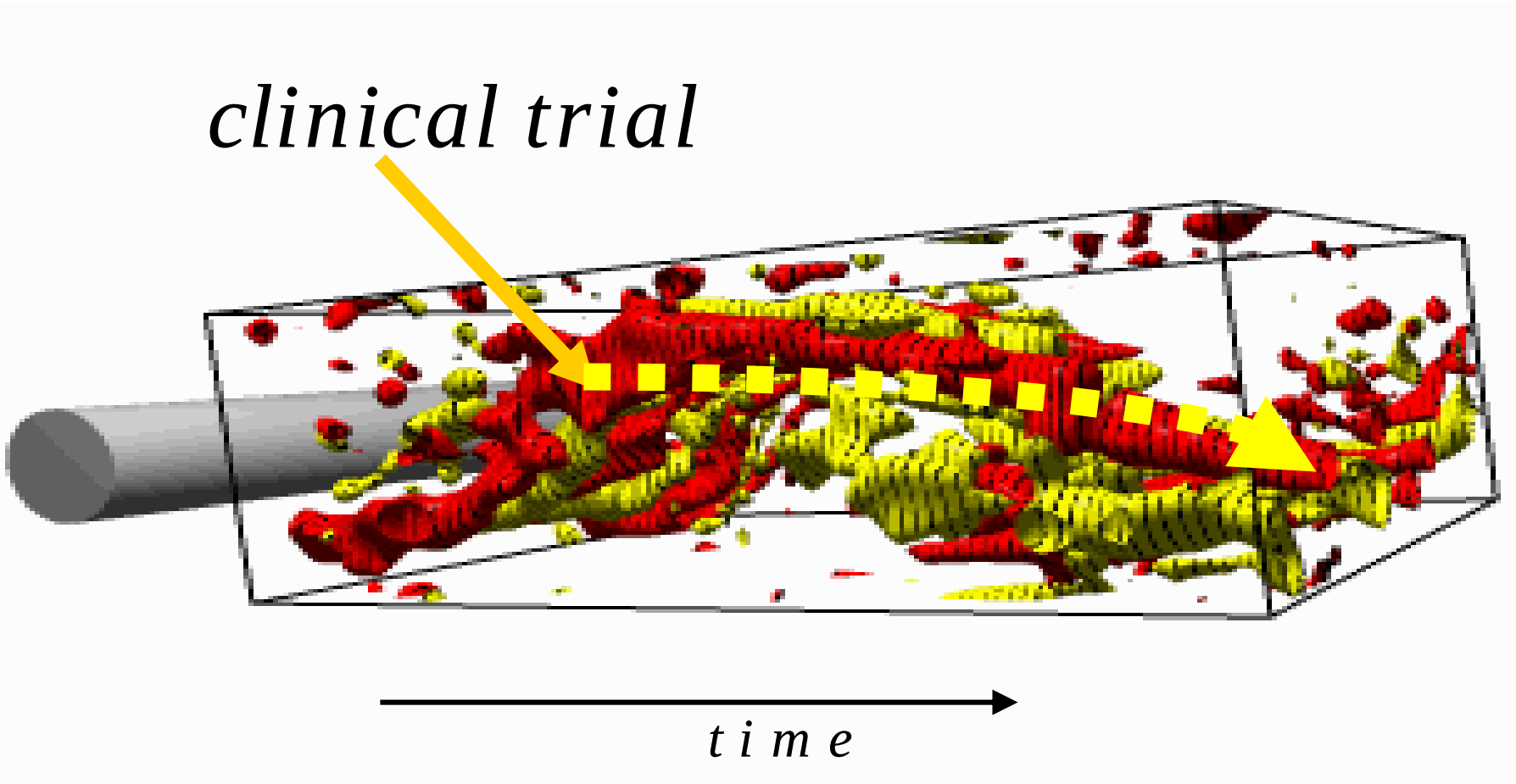


The SPAN ontology

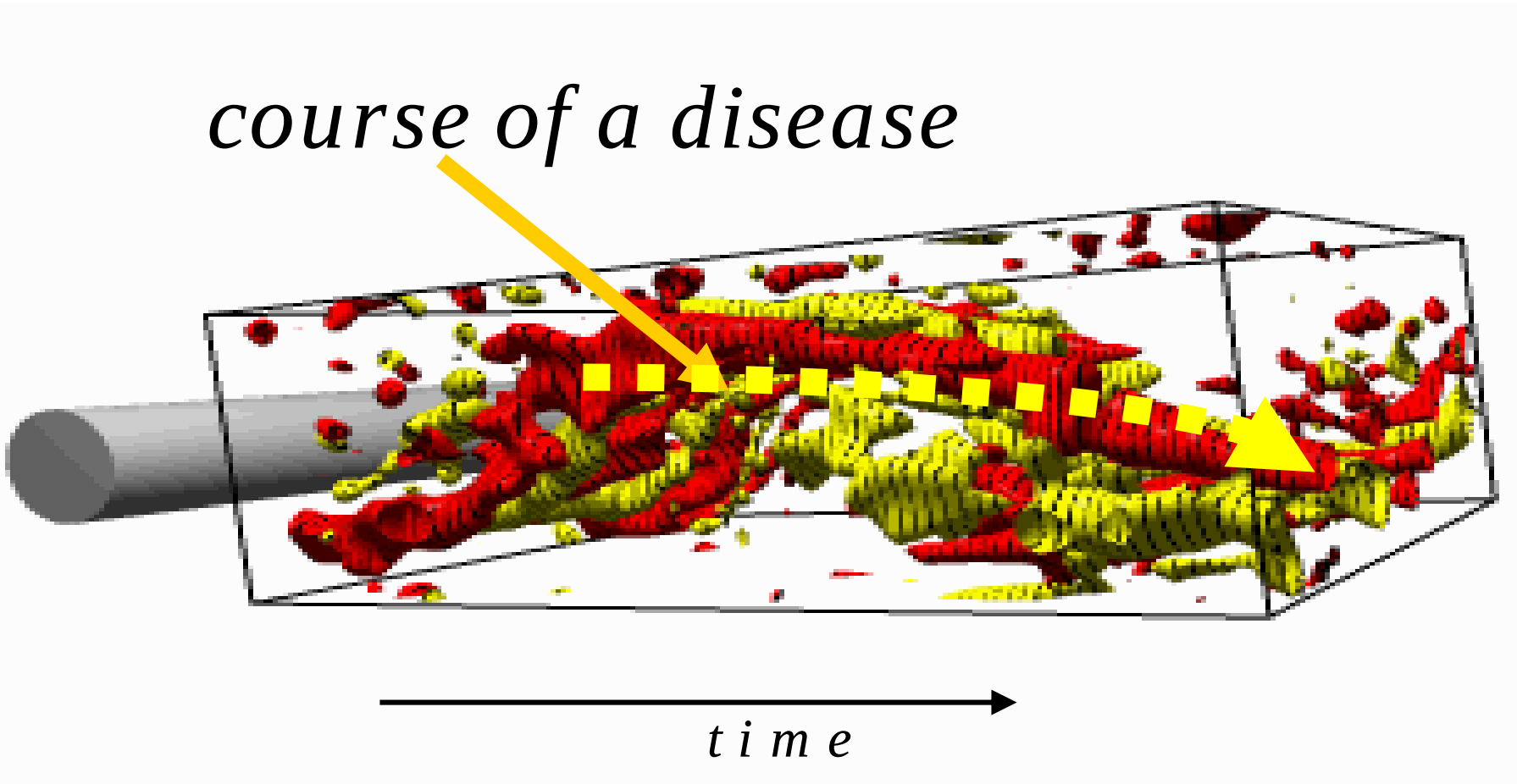


here time exists as part of the domain of the ontology

mereology works without restriction everywhere here



mereology works without restriction everywhere here



Processes, too, are dependent on substances

One-place vs. relational processes

One-place processes:

your getting warmer

your getting hungrier

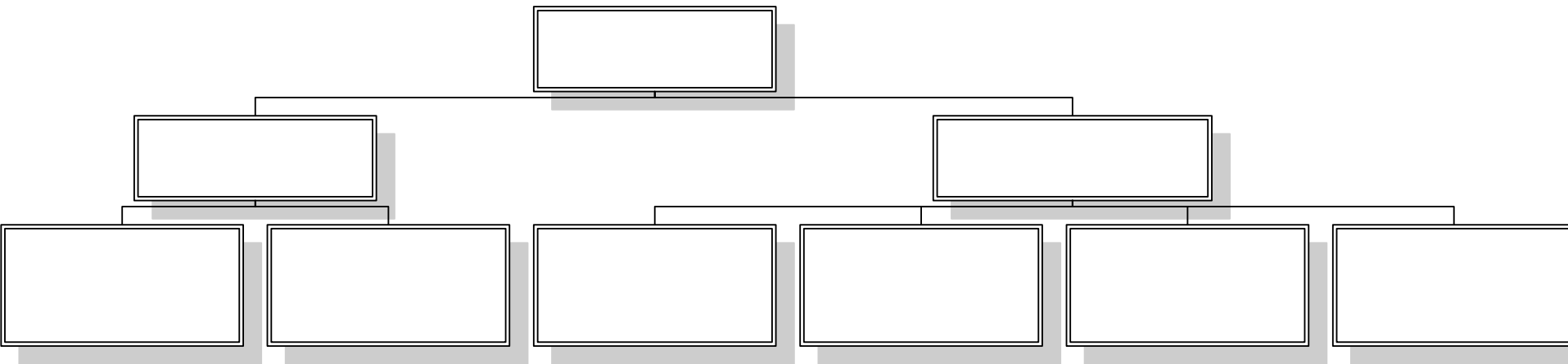
Relational processes

kissings, thumpings, conversations,

dancings, promisings, infectings, bindings

join their carriers together into *collectives* of greater or lesser duration

SPAN: Entities extended in time



http://ontology.buffalo.edu/bfo

Two kinds of SPAN entities

1. Processes (including events: process-boundaries, settings)
2. Spatio-temporal regions

How do you know whether an entity is SNAP or SPAN?



problem cases

forest fire

hurricane Maria

traffic jam

ocean wave

disease

anthrax epidemic

forest fire:

a process

a pack of monkeys jumping from tree to tree and eating up the trees as they go

the Olympic flame:

a process or a thing?

(anthrax spores are little monkeys)

A disease

The course/history of a disease

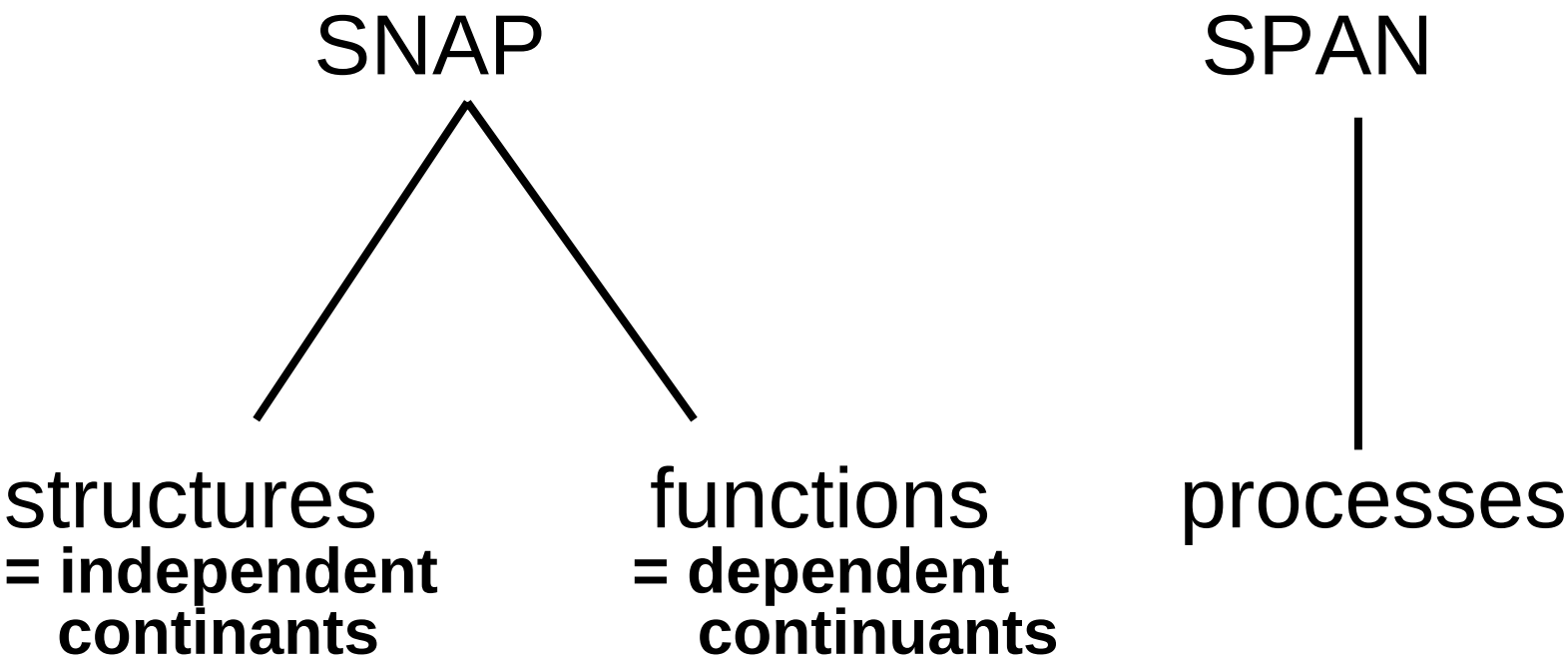
The Epidemic (SNAP)

The *Spread* of an Epidemic (SPAN)

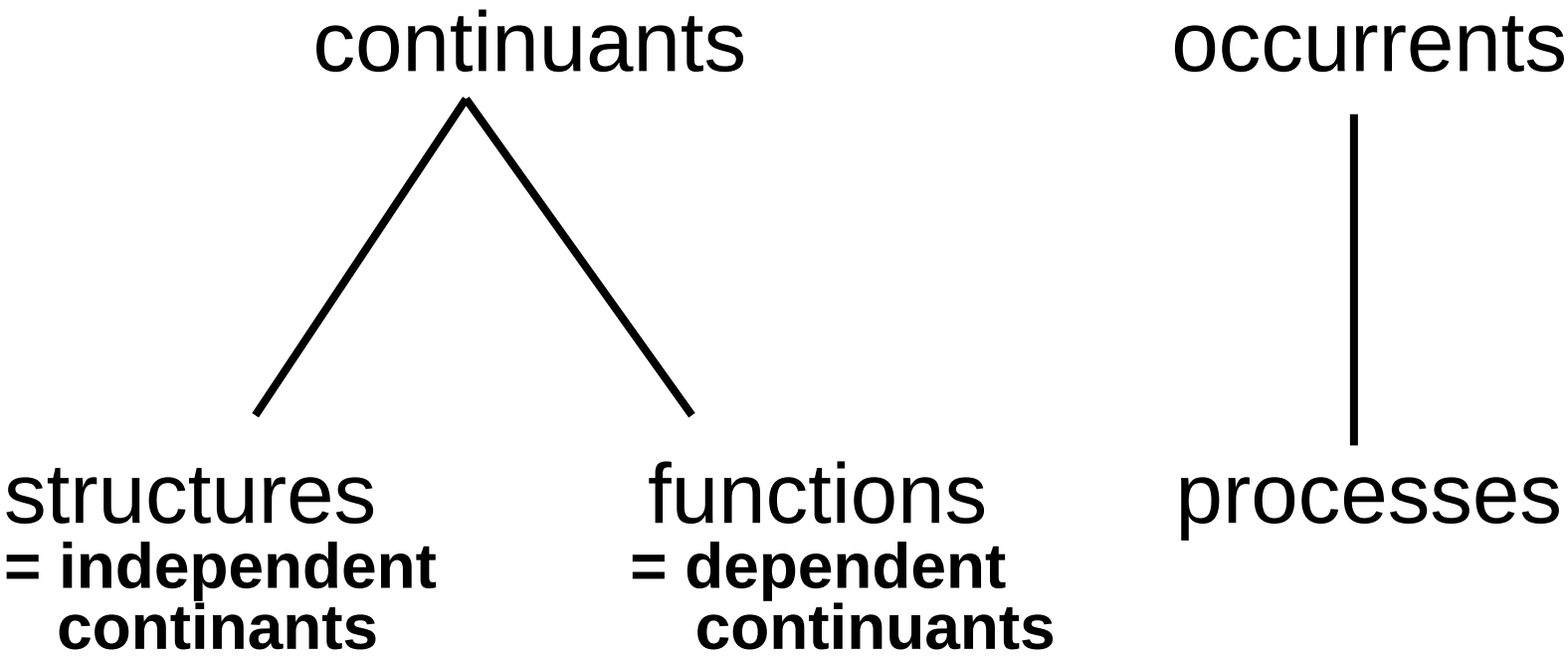
Material examples:

performance of a symphony
projection of a film
expression of an emotion
utterance of a sentence
application of a therapy
increase of temperature

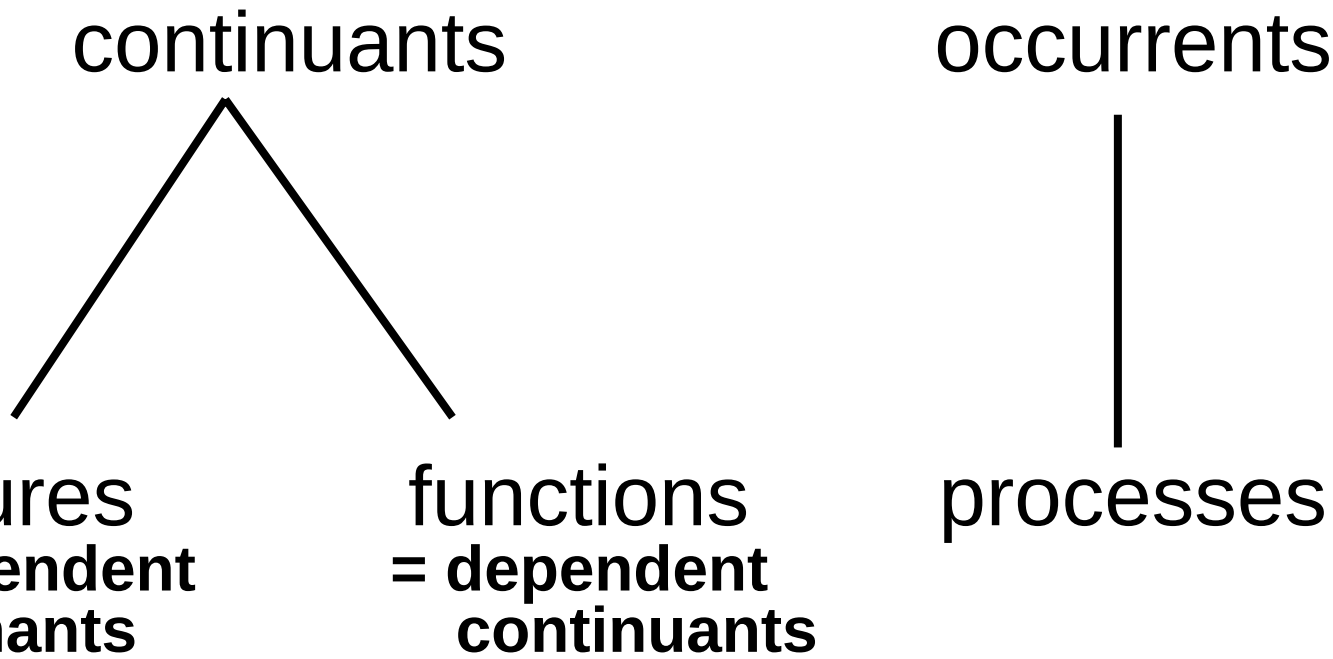
The Tri-Categorial Ontology



The Tri-Categorial Ontology



A Window on Reality



Entities in all three categories exist both as universals and as instances (as tokens and as types)

The function of *your* heart is: to pump blood
The function of *my* heart is: to pump blood

Functions are continuants

The function of your heart begins to exist with the beginning to exist of your heart, and continues to exist, self-identically, until (roughly) your heart ceases to be able to respond if stimulated by your sympathetic and parasympathetic nervous systems

Functions have bearers

The bearer of the function of your heart is: your heart.

Functions are dependent continuants.

The bearers of functions are independent continuants (hearts, screwdrivers …)

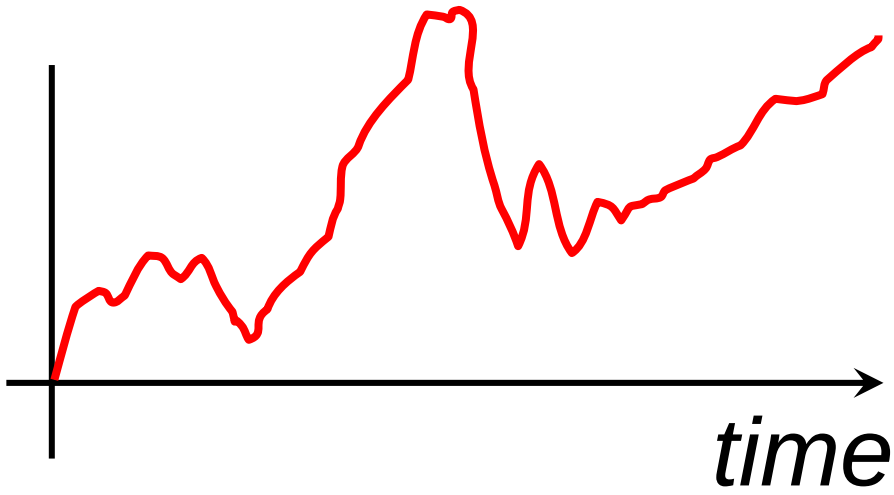
Functions are *realized*

in special sorts of processes called *functionings*

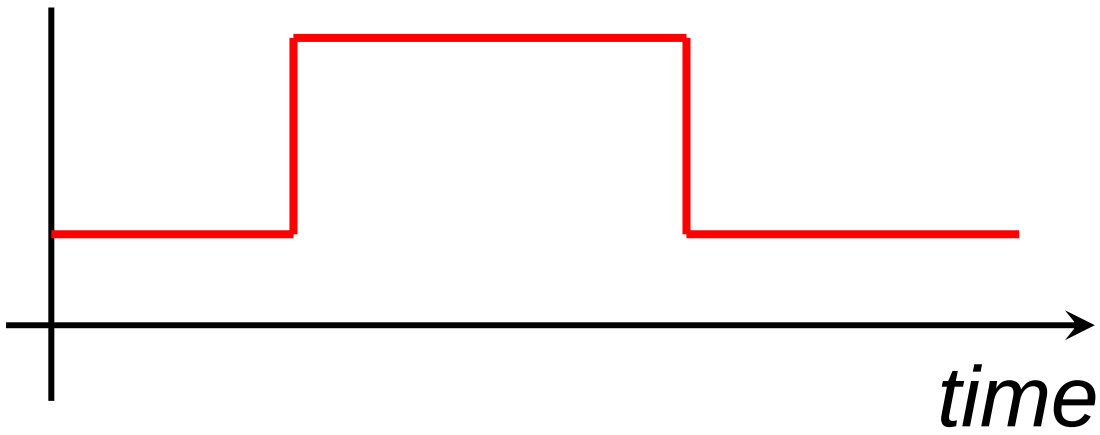
The processes taking place in or involving entities which are bearers of functions can be divided into two types: those which are realizations of their functions (also called *functionings*) and processes of other types (junk processes)

Functions can exist even when they are not being realized

Processes (realizations) are causal-energetic



Functions are historical (they exist in time)
but they are also quasi-Platonic



Compare the relation between temperature,

which is quasi-Platonic

and Brownian motion,

which is causal-energetic

Your temperature at *t* vs. the value of your temperature at *t*

Your temperature is quasi-Platonic

Your temperature as a *determinable* is identical from one moment of your existence to the next

This determinable takes on different *values* at different times

Biological functions are always *constituent functions*

If X has a biological function then there is some Y of which X is a part and X's functioning is in the service of / for the benefit of Y

Functions are beneficial

If an organism has a constituent part X, and if X is the bearer of a function Z, then those processes which are the realizations of the function Z are (in normal circumstances) beneficial to the organism

(≅ such as to sustain the organism in existence)

Functional Genomics

= study of what the genes contribute to the organism in the way of survival
(Bad genes do not *have* functions)

Every oncogene is a proto-oncogene

There is functioning, poor functioning, malfunctioning

There is not having a function at all (and this can be either neutral in the stakes of beneficiality or also positively malignant)

Does this sense of ‘function’ correspond to the way biologists talk?

Clinical vs. biological sense of ‘function’

Biologists sometimes talk about biological structures ‘gaining function’ (= being switched on) even where their functioning is *not* beneficial

Are all functions associated with *mal*functionings?

Health – Disease – Illness

Diseased organ = organ predisposed to malfunction

Its functioning is defective




Part Four: Definitions of Health

World Health Organization:

Health is the state of psychological and physical well-being of humans

Biostatistical Theory

Christopher Boorse

Health is conformity to normal species design (as statistically determined).

Abnormally healthy people are therefore in fact sick (?)

The Vital Goal Theory

Lennart Nordenfelt

Health is the bodily and mental state of a person which is such that he or she has an ability to realize vital goals, given standard or otherwise accepted circumstances.

Disease is a state or process of a person's body or mind that tends to cause ill health in the bearer.

The Ordinary Action Theory

K.W.M. Fulford

Health is being able to do what one ordinarily does in the absence of obstruction or opposition.

Illness is failing to do what one ordinarily does in the absence of obstruction or opposition.

The Abnormality Theory

Lawrie Reznek

Disease is a state of a person which issues in abnormal behavior;
something is an abnormal bodily or mental process if it does standard members of the human species some harm in standard circumstances;
something does a person harm if it makes the person less able to live a good or worthwhile life.

Problems with standard definitions

1. Circularity
2. Make health a social construction
3. Make health a Cambridge property
4. Confuse state and process, disposition and realization, potentiality and actuality
5. Do not apply to organisms other than humans

Circularity

Health is ... well-being

Health is ... being able to live a good or worthwhile life

Disease is a state … that tends to cause ill health in the bearer

Health a social construction

Health is …the ability to realize vital goals, given standard or otherwise accepted circumstances

Illness = what the insurance company will pay to treat

Health a Cambridge Property

Health is conformity to normal species design (as statistically determined).

If everyone in society becomes sicker and you remain the same, then *you* are the person who becomes unhealthy

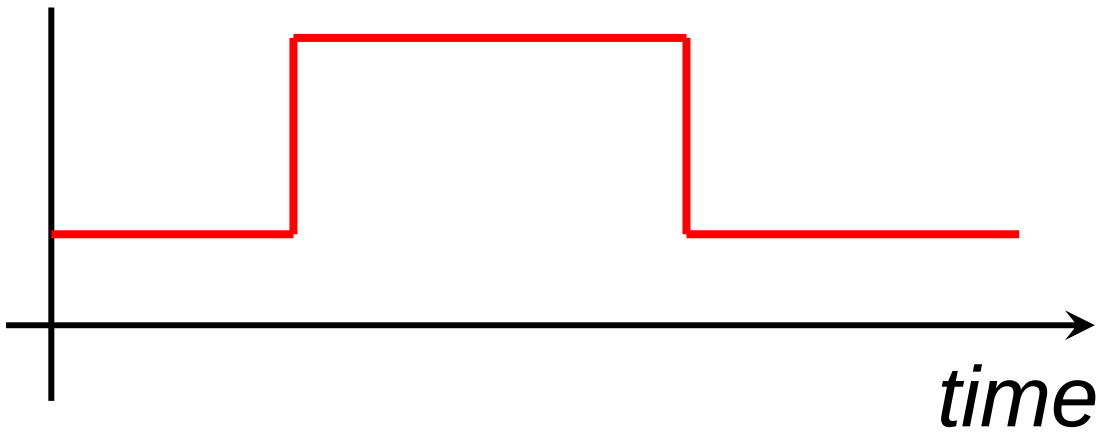
Ontology of Disease

Diseases are, like functions, dependent continuants

They are states or conditions which endure for a certain time and have a **course** or **history,** which is an occurrent

Disease tokens, like roles and functions, do not change through their existence over time

Diseases are both historical and quasi-Platonic



Functions

This is a screwdriver

This is a good screwdriver

This is a broken screwdriver

This is a heart

This is a healthy heart

This is an unhealthy heart

Functions are associated with certain characteristic *process shapes*

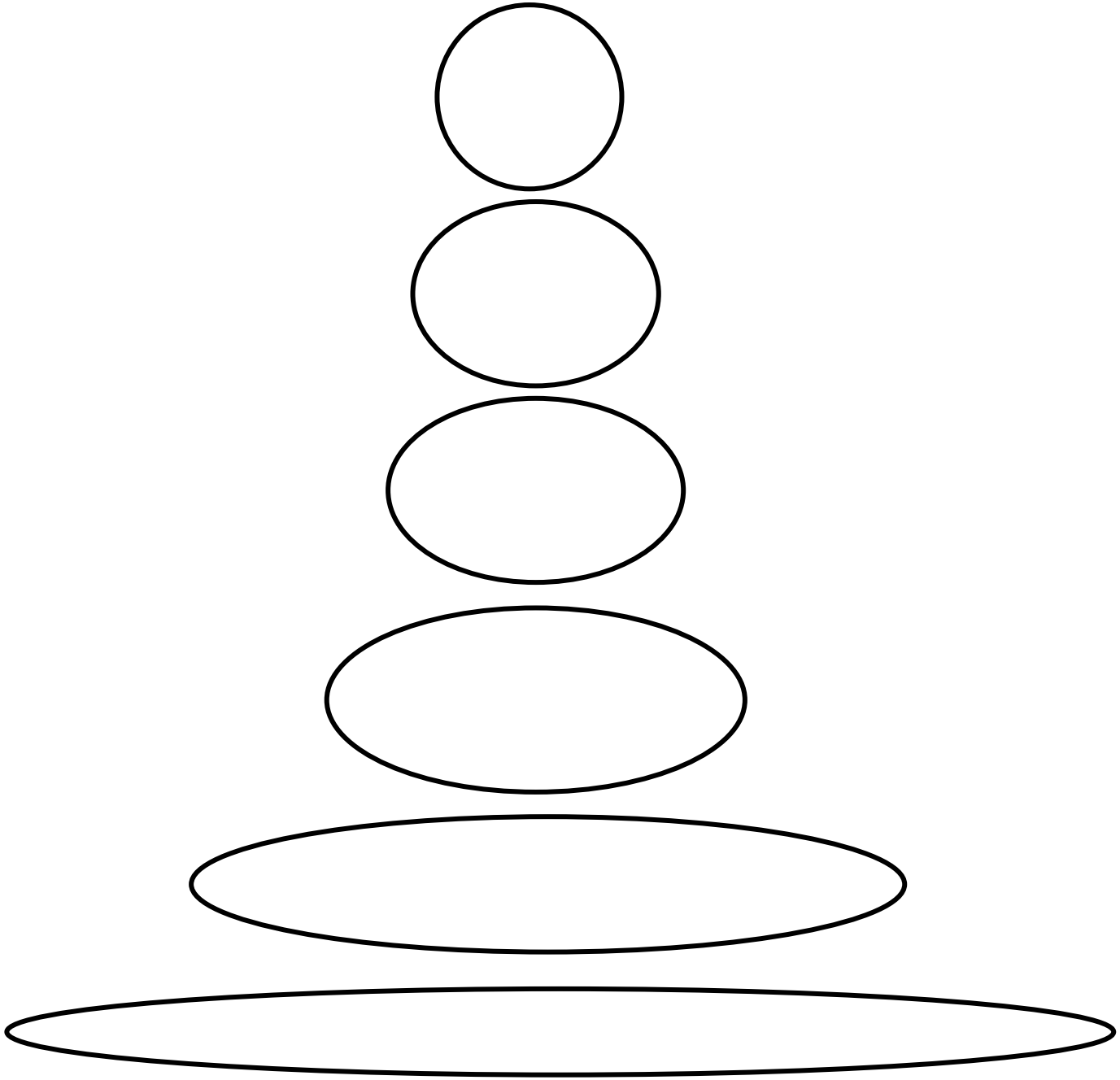
Screwdriver: rotates and simultaneously moves forward simultaneously transferring torque from hand and arm to screw

Heart: performs a contracting movement inwards and an expanding movement outwards simultaneously transferring hydraulic pressure to the blood stored within its chambers

For each function

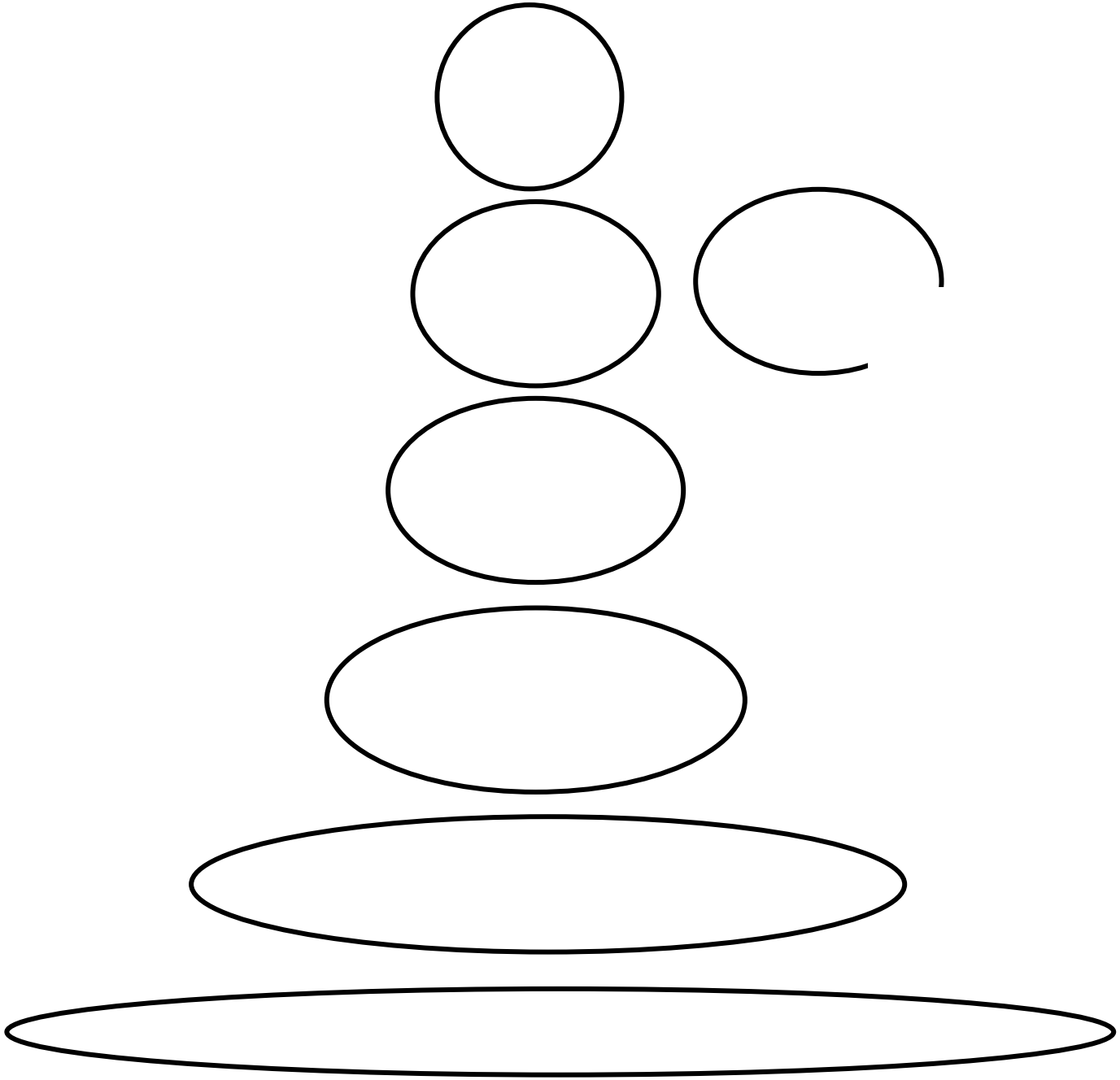
there is an associated *family* of (four-dimensional) process shapes, organized around a core of prototypical process shapes representing good functioning

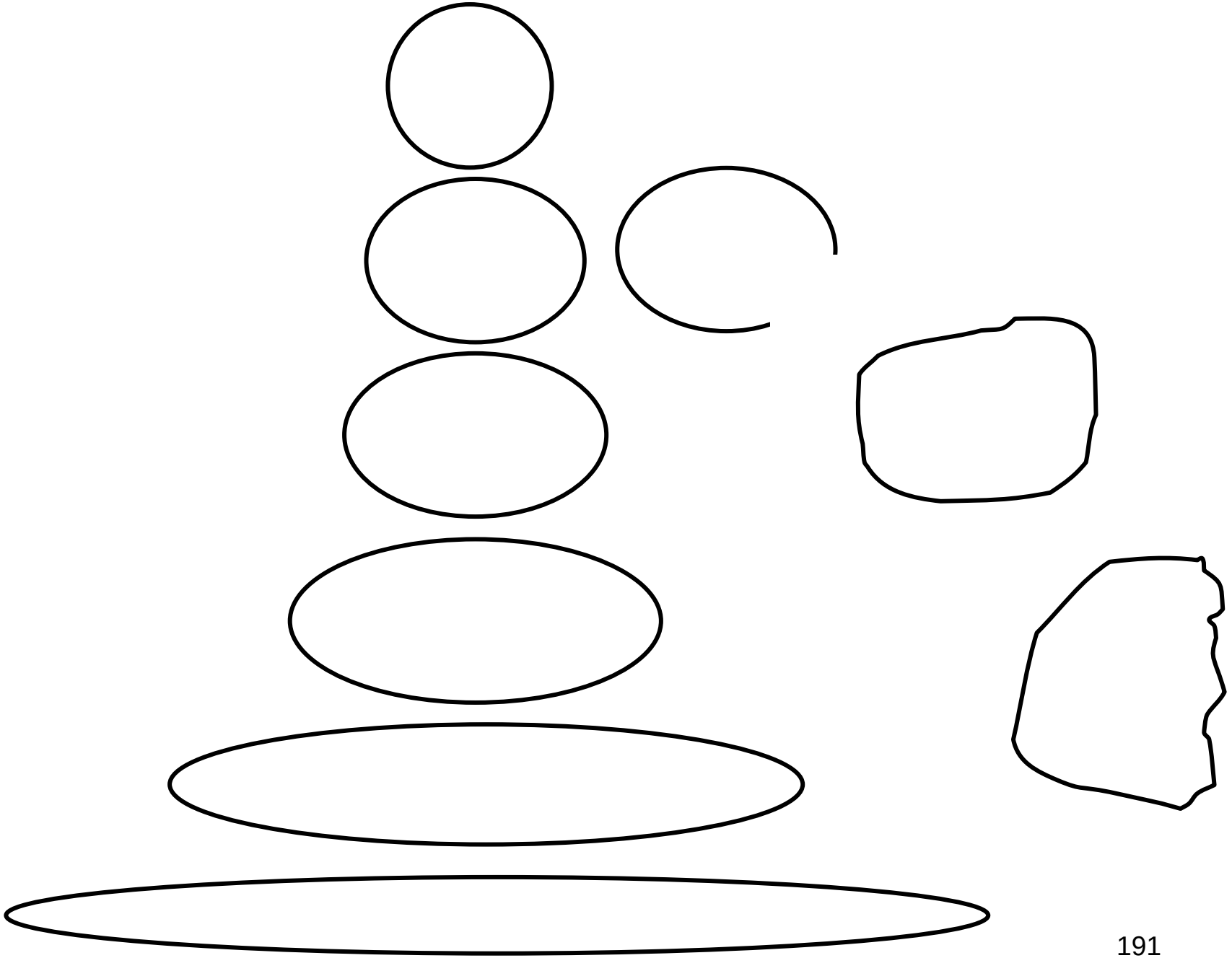
The prototypes play a role analogous to the standard meter rule in the organization of those one-dimensional shapes we call lengths



Outside the core

are process shapes which are not instances of functioning at all

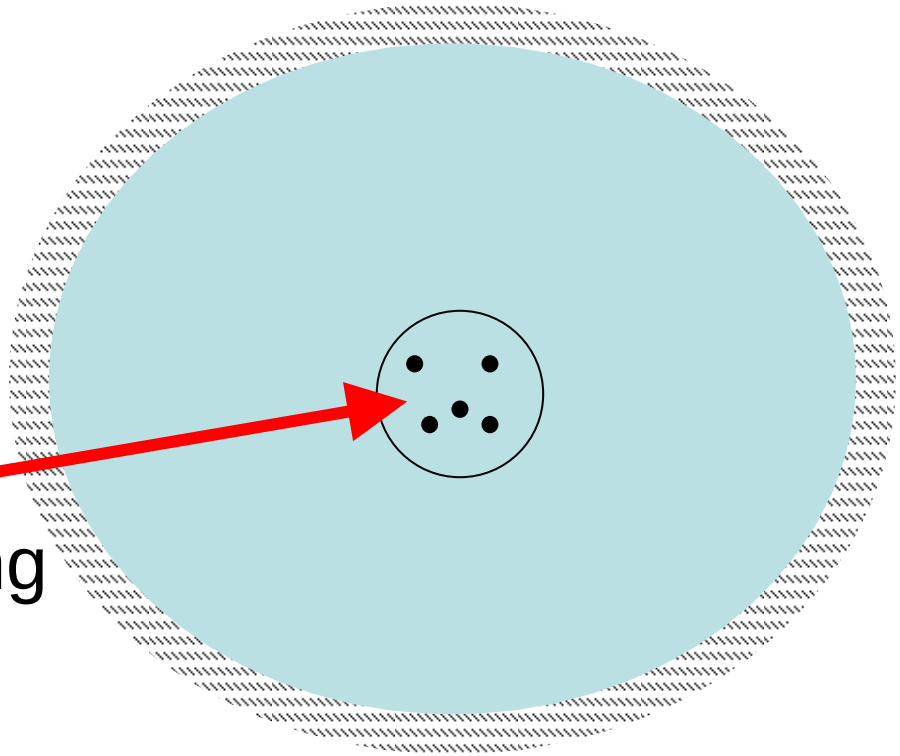




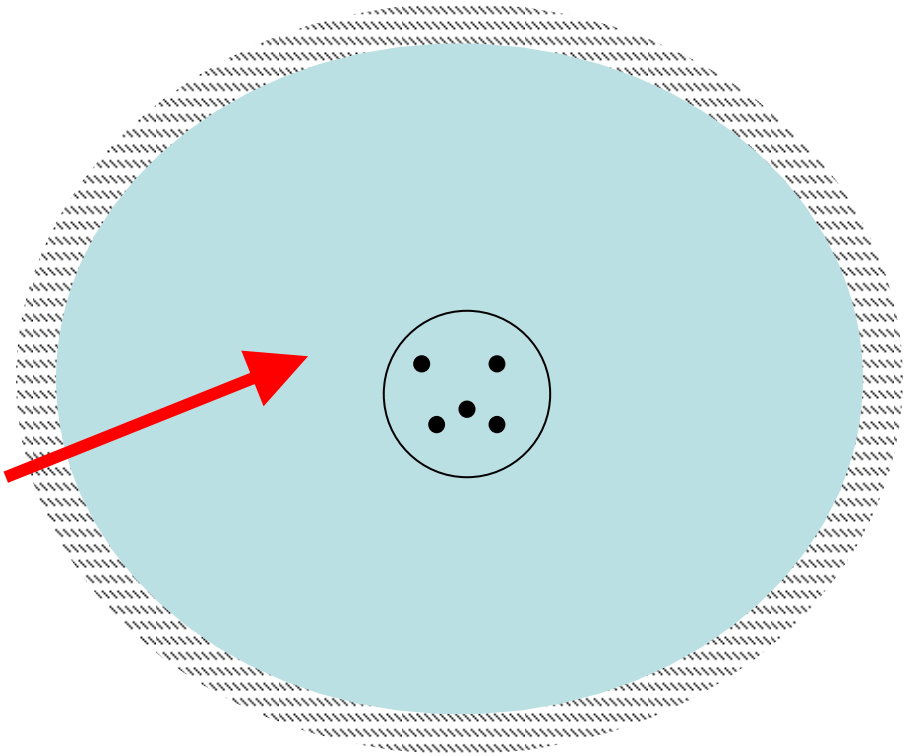
Normal functioning

= functioning (realizing a four-dimensional shape) at or close to the prototype

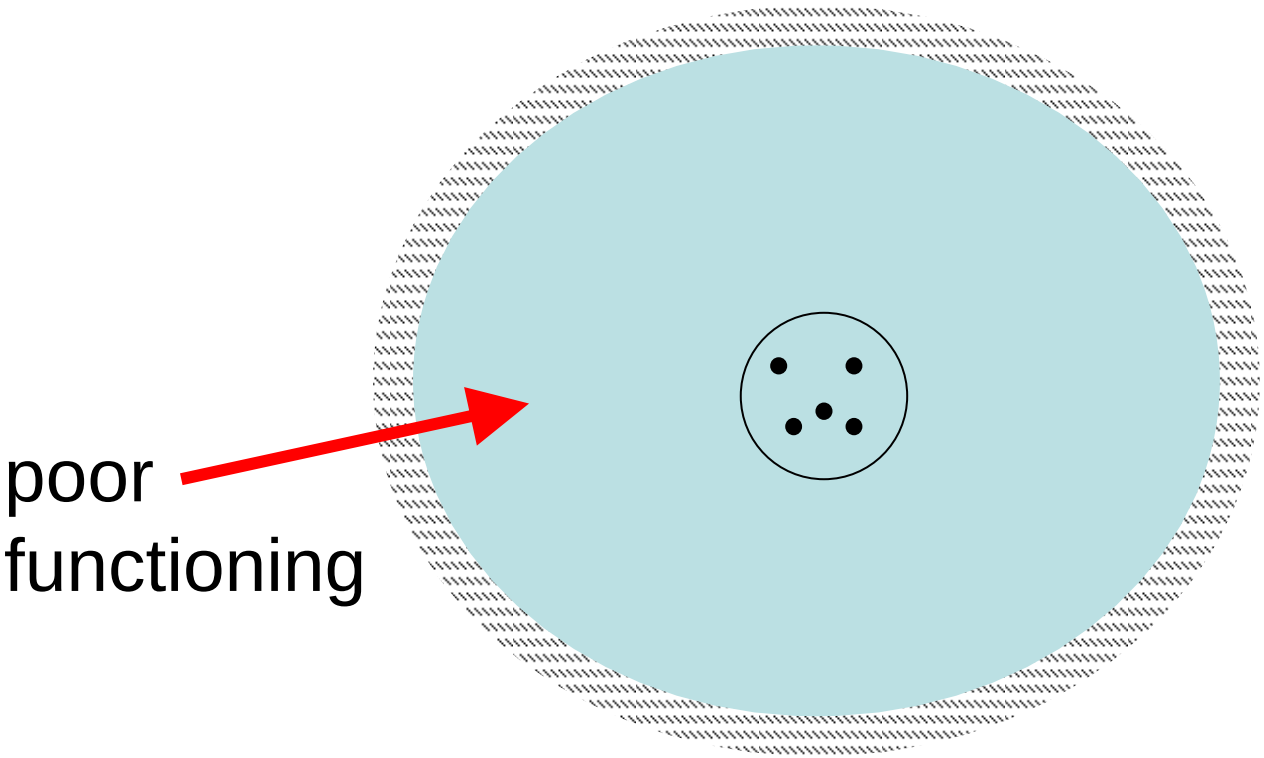
Prototypes



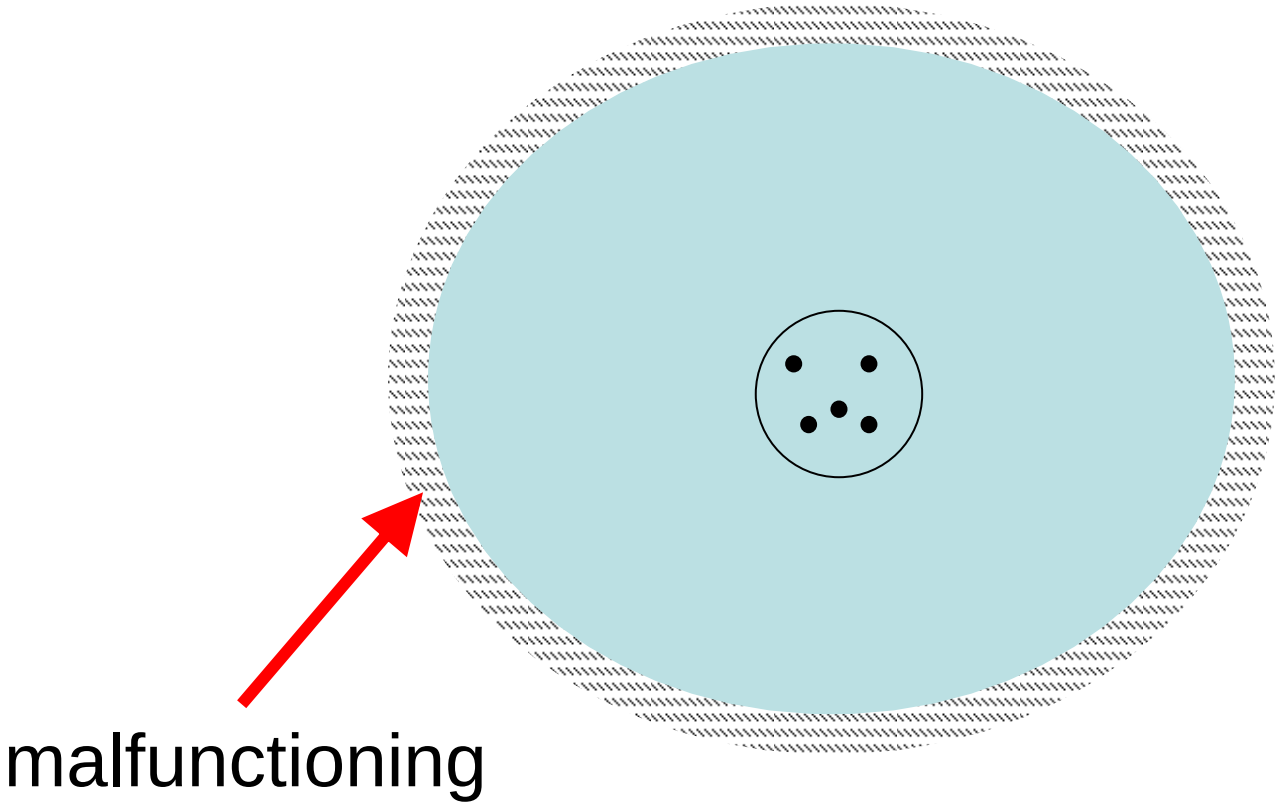
Prototypes



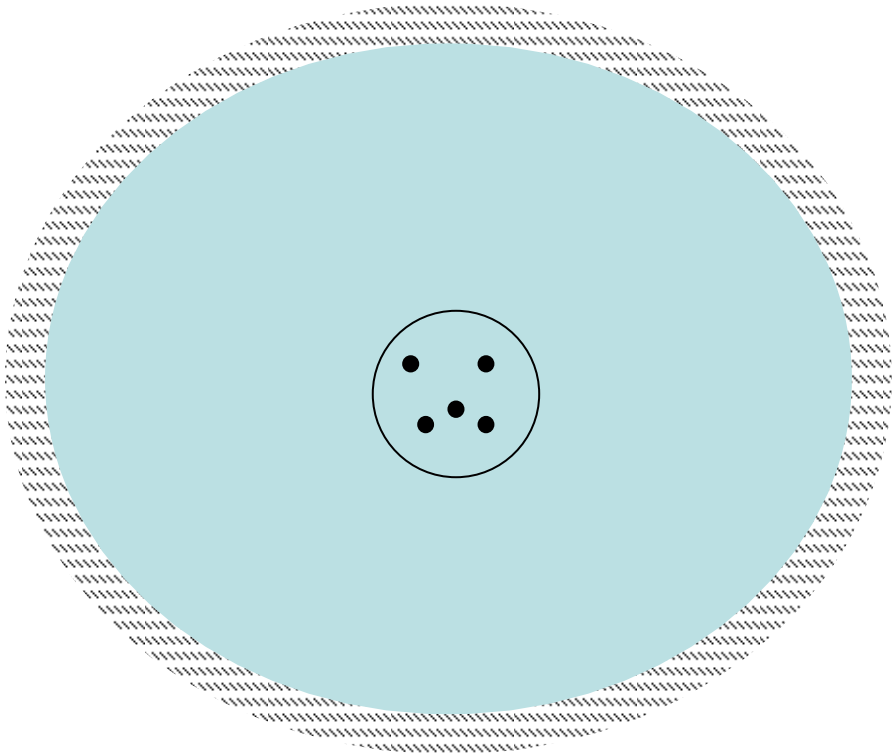
Poor functioning



Malfunctioning



Death?



not functioning at all

Not functioning at all

leads to death *modulo* internal factors:

- plasticity
- redundancy (2 kidneys)
- criticality of the system involved

external factors:

- prosthesis (dialysis machines, oxygen tent)
- special environments
- assistance from other organisms

Relevance of Millikan

Prototypical functioning = exercising what Millikan calls ‘proper function’

(defined historically)

X is the proper function of Y means: 1) Y performs X and 2) Y exists because its predecessors’ performing the function X is responsible for my existing

It is not the function of the nose to hold up spectacles *because this was not selected for*

Millikan = backward looking, focused on whole species

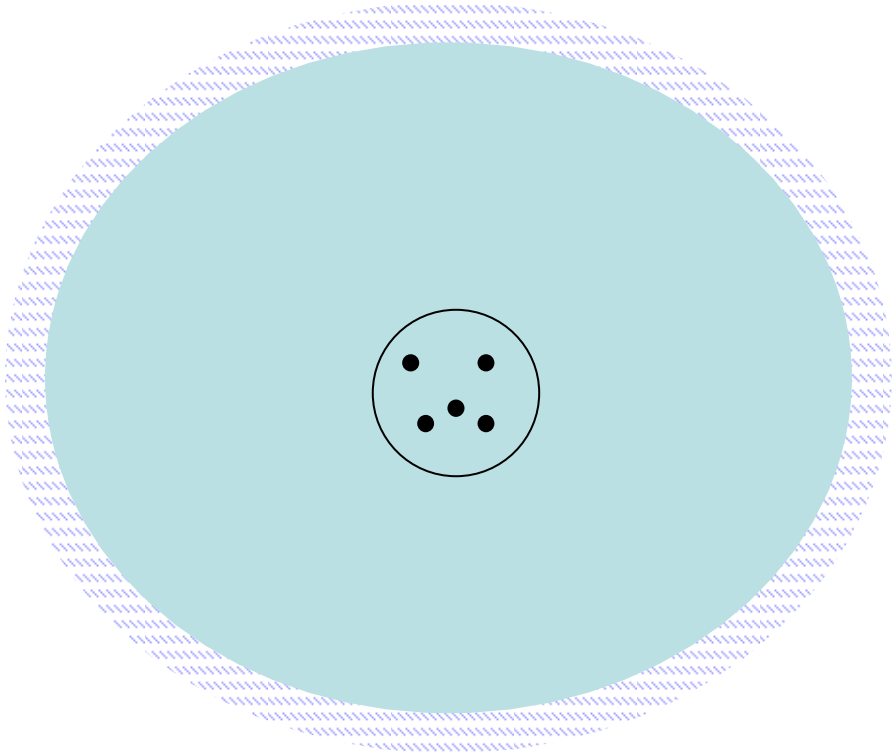
This account = forward looking, focused on single organism

X has a function = (1) X's functioning is beneficial to the organism of which X is a part

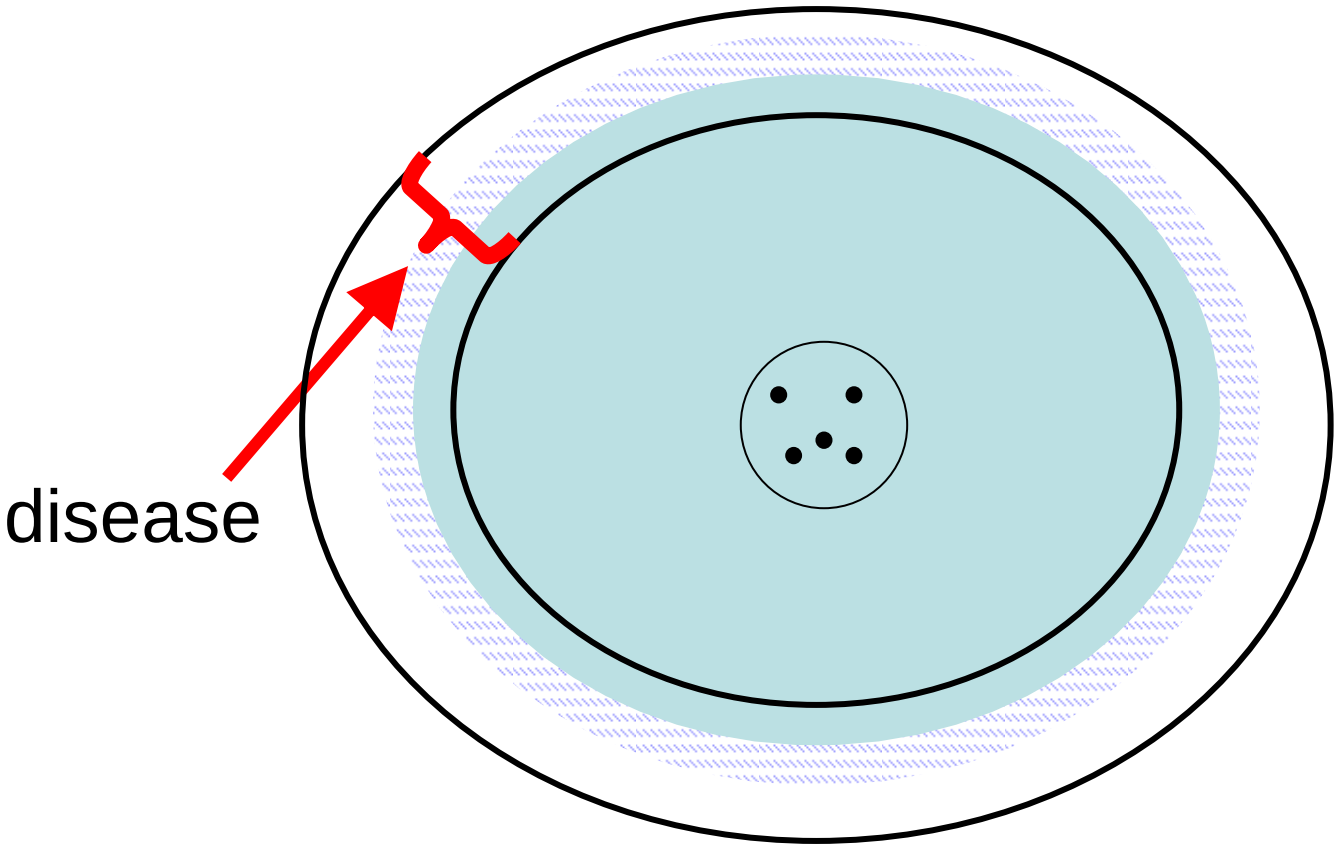
Boorse's Internal Impairment Theory

Disease is an internal state which is an impairment or limitation of normal functional ability.

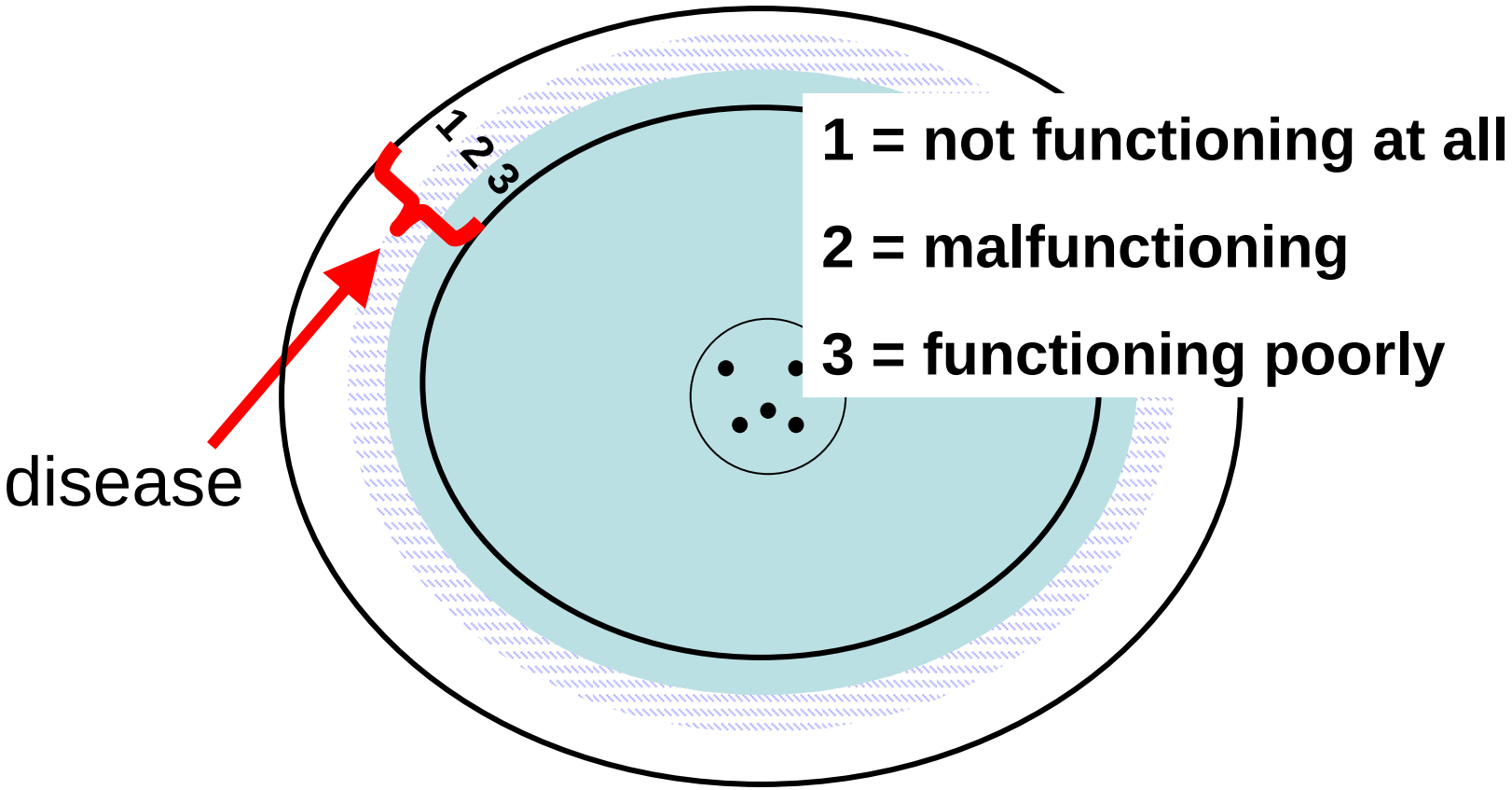
Disease



Disease = remoteness from prototypical functioning



Disease = remoteness from prototypical functioning



Not functioning at all

= death *modulo*:

criticality of the system involved

Biological entities have biological functions only as parts of organisms

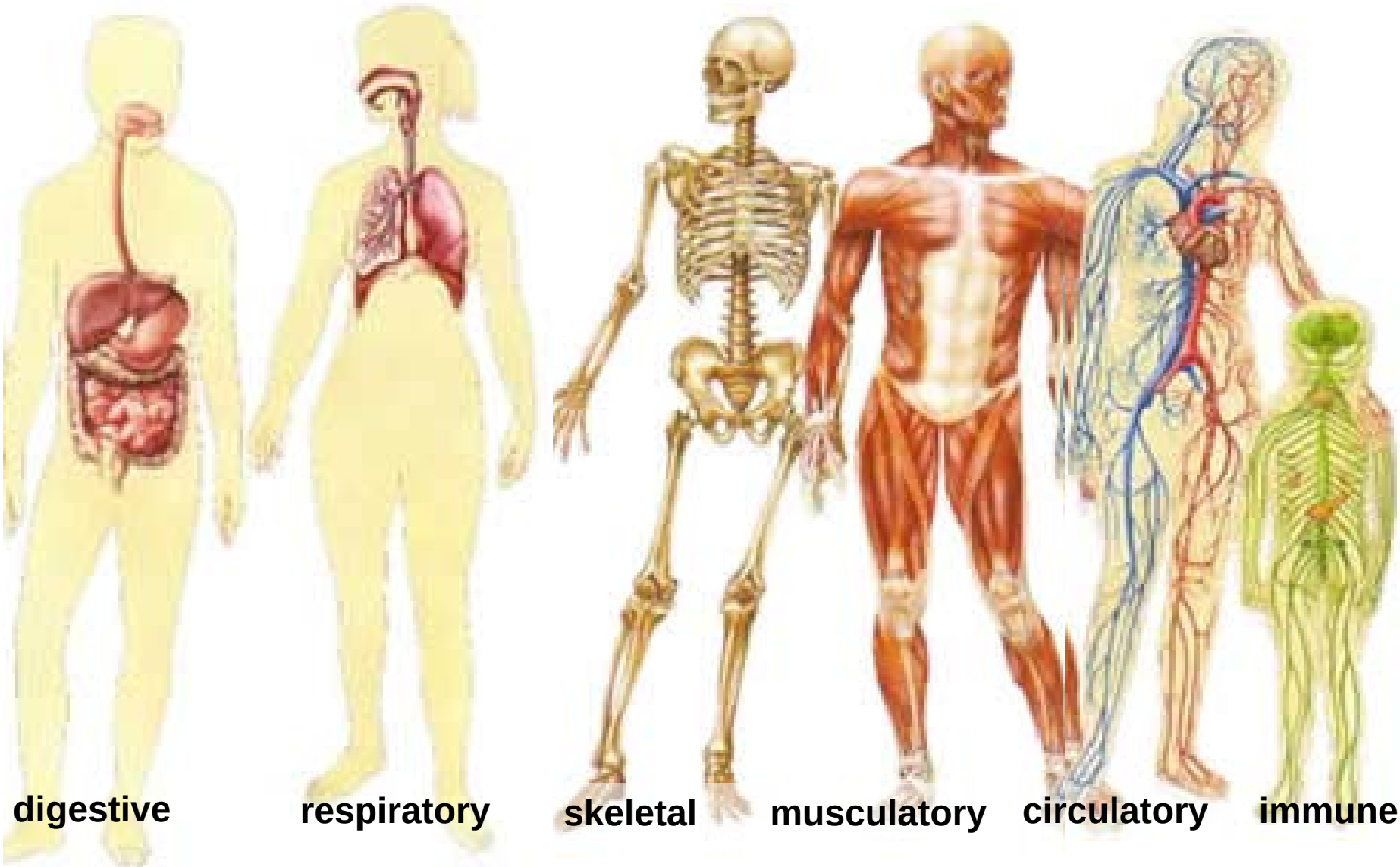
An organic entity functions in the service of the organism of which it is a part

There are immediate parts of the organism – the bodily systems – which function directly in the service of the organism.

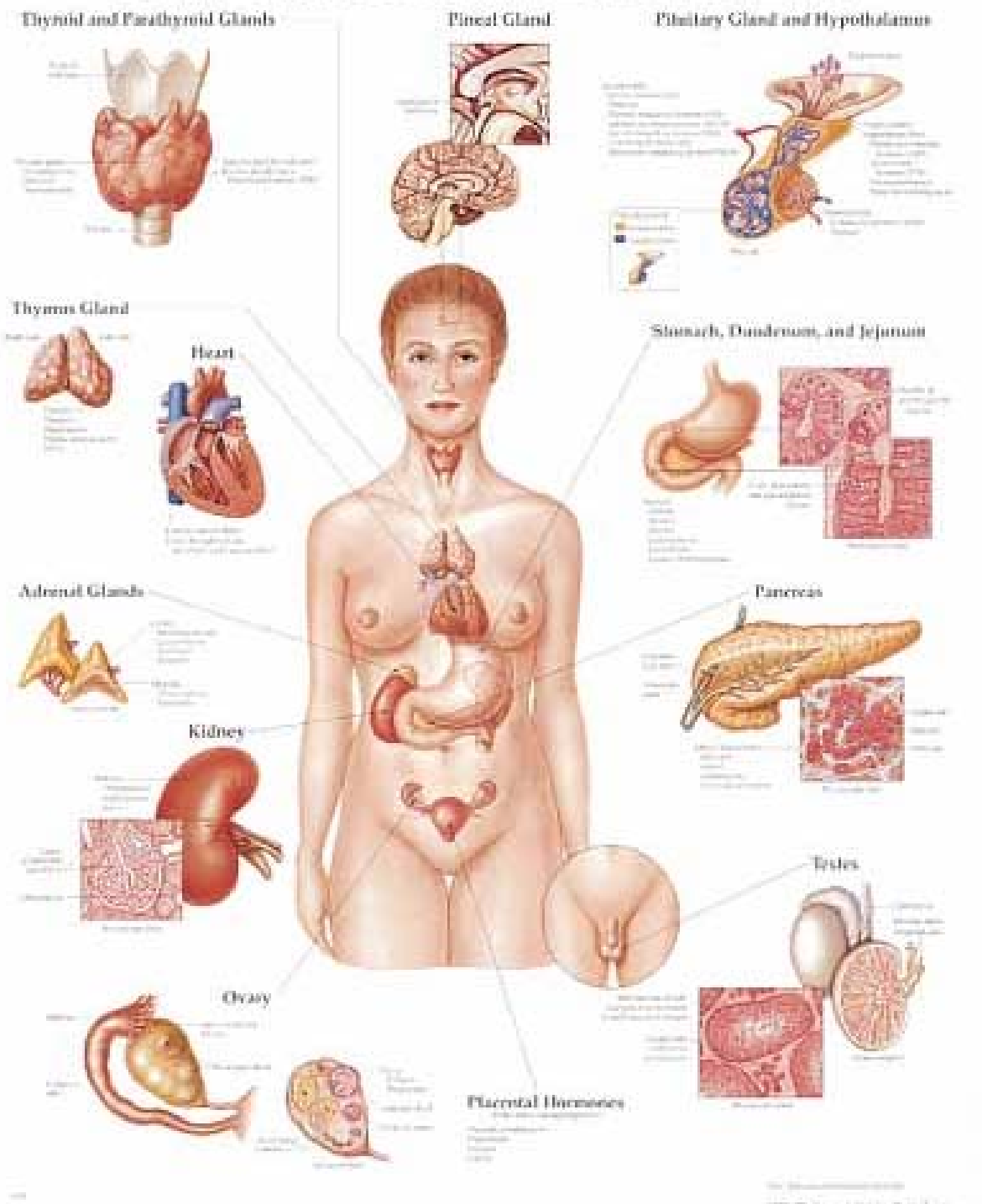
And there are mediate (= smaller) parts of the organism – cells, tissues, organs … -- which function in the service of larger parts

Immediate parts of the organism are more critical

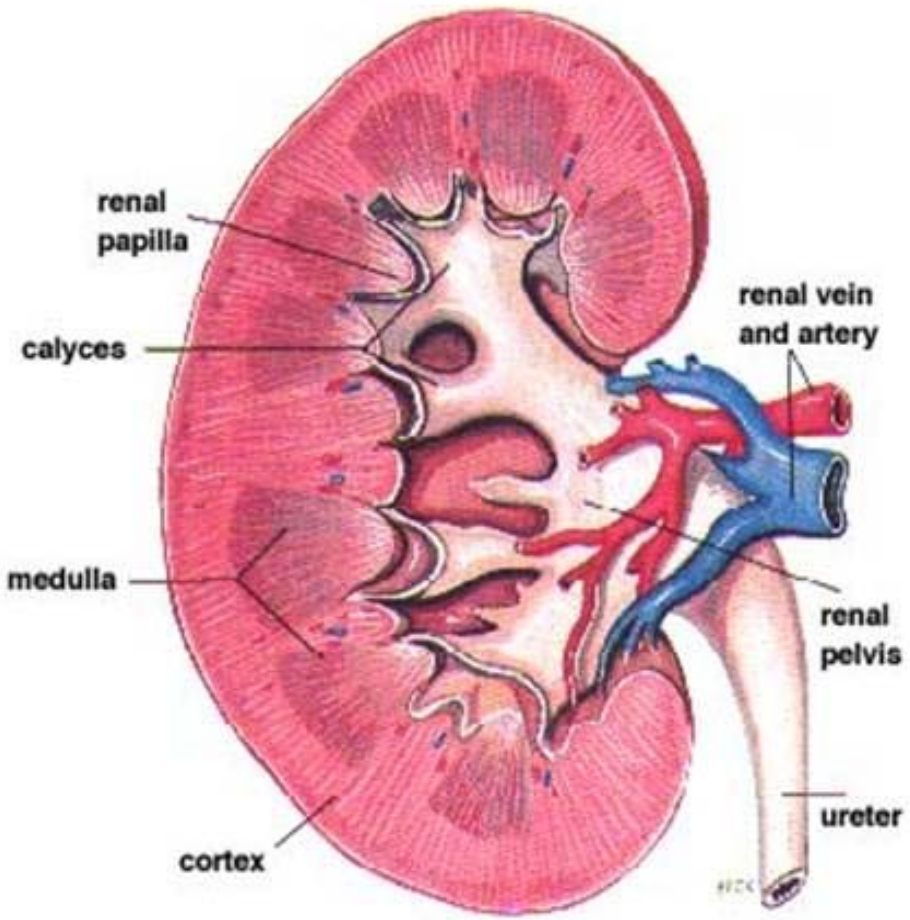
Bodily Systems



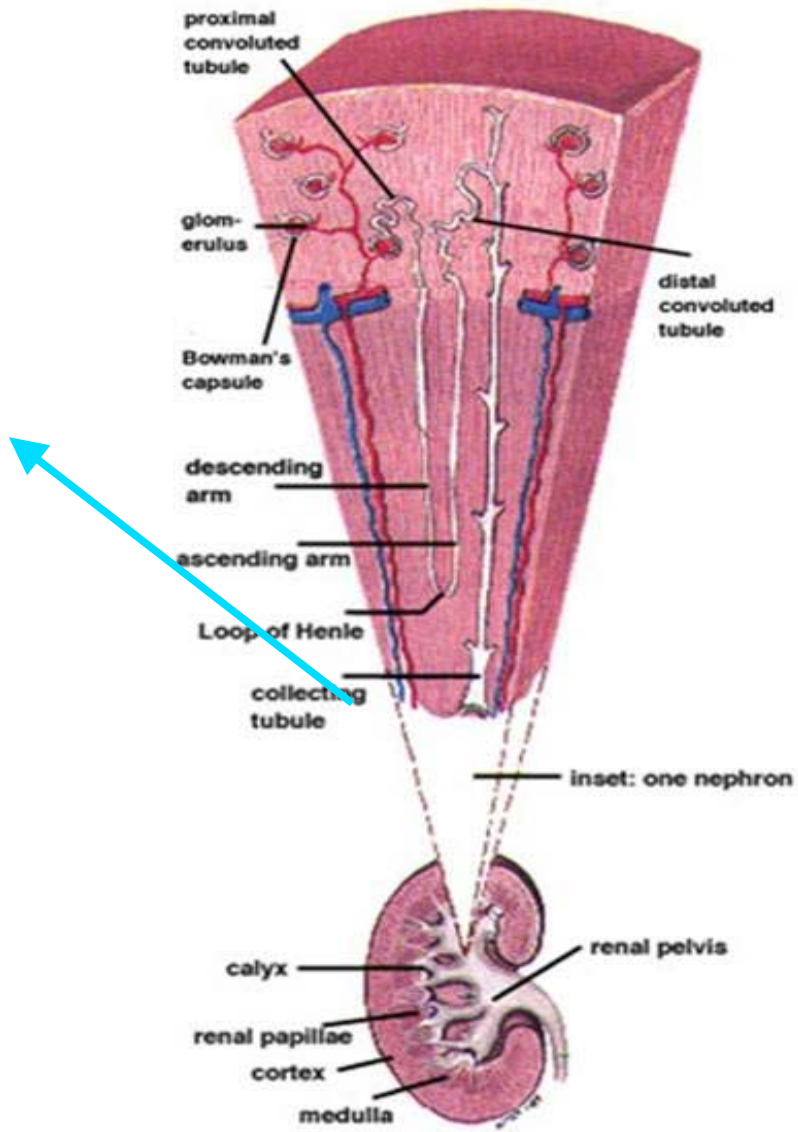
ENDOCRINE SYSTEM



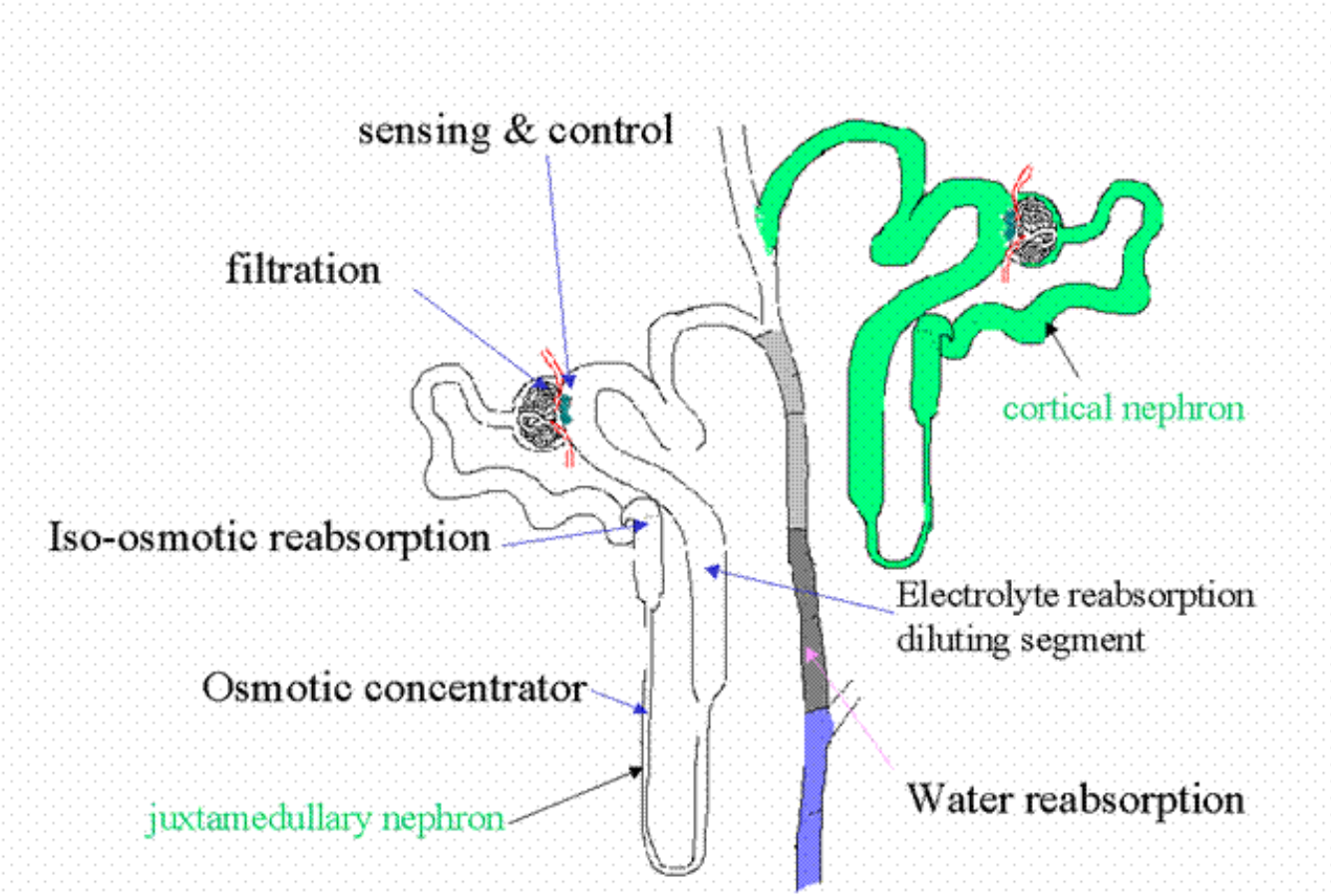
KIDNEY


renal papilla
calyces
medulla
cortex
renal vein and artery
renal pelvis
ureter

NEPHRON

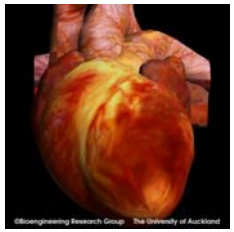

proximal convoluted tubule
glom-erulus
Bowman's capsule
distal convoluted tubule
descending arm
ascending arm
Loop of Henle
collecting tubule
inset: one nephron
calyx
renal pelvis
renal papillae
cortex
medulla

FUNCTIONAL SEGMENTS

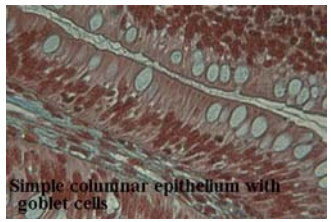


Organism

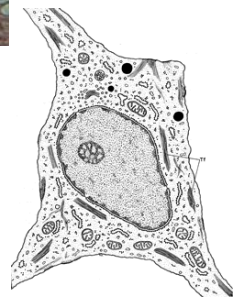
Organ



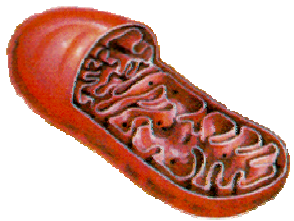
Tissue



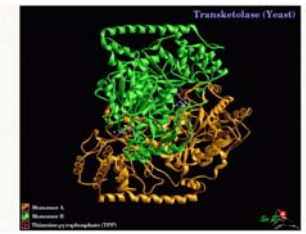
Cell



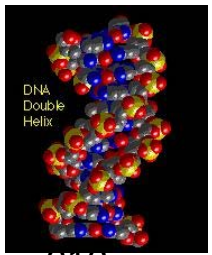
Organelle



Protein



DNA



Coda on ‘Normal’

Normal functioning of the pancreas

Normal functioning of the sexual organs

On the several senses of ‘normal’ in biology

Problem: The Sexual Organs do not have Biological Functions

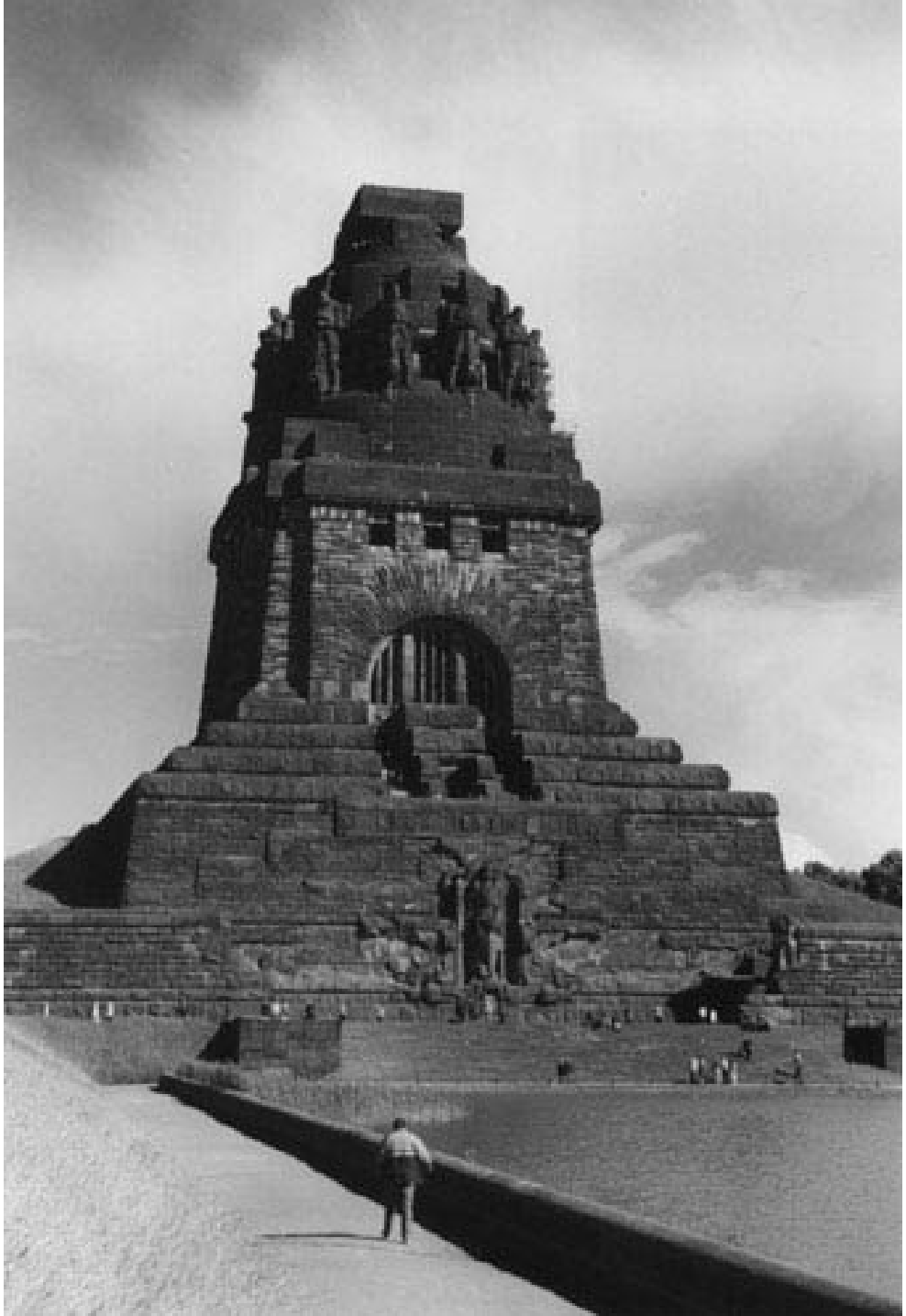
A constituent part of an organism has a function = its functioning is beneficial to the survival of the host organism

– this does not hold for the reproductive system and its parts

Hence the sexual organs do not have functions

Alternatively they have functions in relation to some larger whole (the family, the dynasty …)

Compare the role of worker bees in bee colonies …



The End