

# Agenda · Day 2

- An ontological introduction to biomedicine: Defining organism, function and disease
- The Gene Ontology (GO), the Foundational Model of Anatomy (FMA) and the Infectious Disease Ontology (IDO)
- **The OBO Foundry: A suite of biomedical ontologies to support reasoning and data integration**
- Applications of ontology outside biomedicine

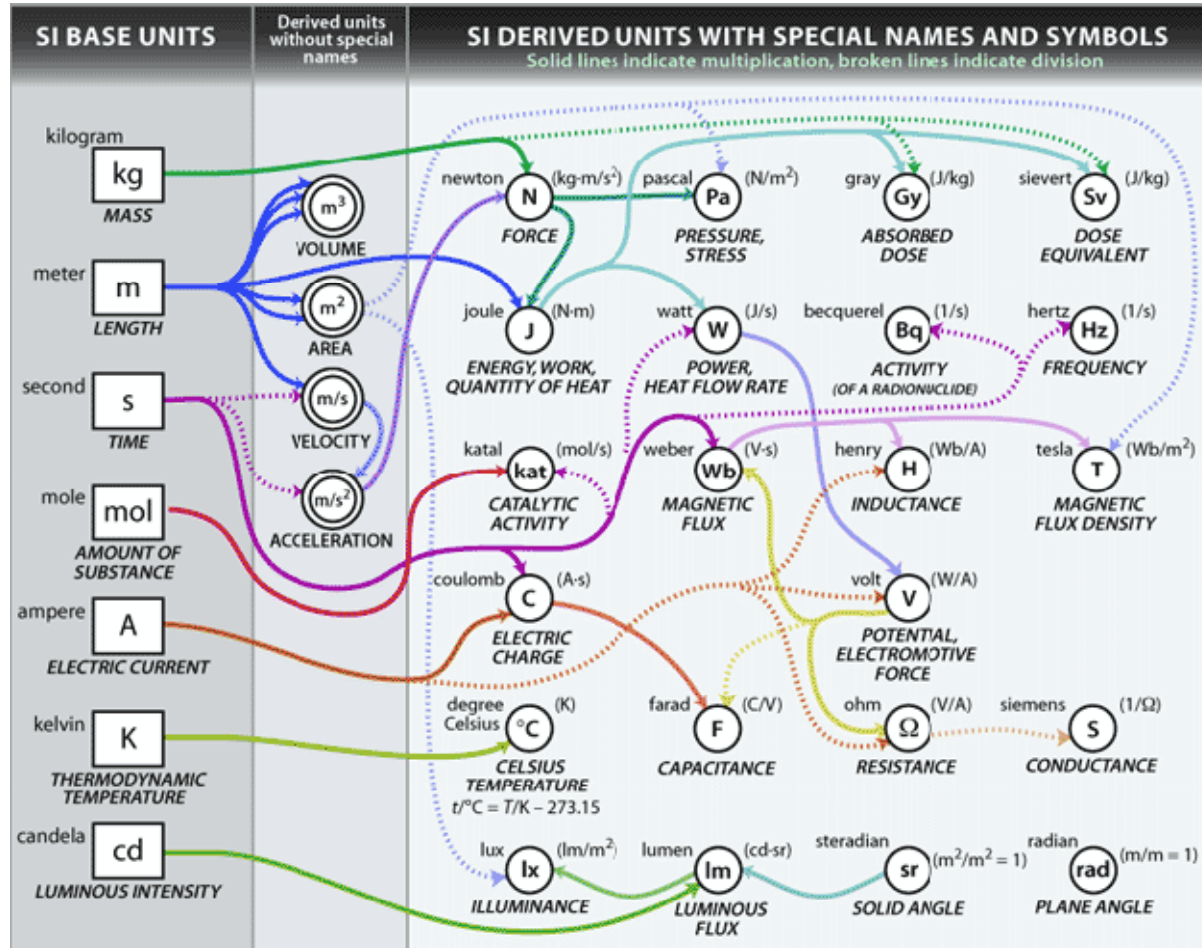
# In the olden days

people measured lengths using inches, ulnas, perches, king's feet, Swiss feet, leagues of Paris, etc., etc.

on June 22 1799  
everything changed



# we now have the International System of Units



# The SI is a Controlled Vocabulary

Each SI unit is represented by a symbol, not an abbreviation. The use of unit symbols is regulated by precise rules.

The symbols are designed to be the same in every language.

Use of the SI system makes scientific results comparable

# The SI is an Ontology

Quantities are universals

one each for each 'quantitative' dimension  
of reality

(= dimension which can be apportioned  
into homogeneous units, and thus  
associated with quantitative measures)

# Goal of OBO Foundry

to provide a suite of controlled structured vocabularies for the calibrated annotation of data to support integration and algorithmic reasoning across the entire domain of biomedicine

current list of Foundry ontologies:

<http://obofoundry.org>

see also Coordinated Evolution of Ontologies to Support Biomedical Data Integration, Nature Biotechnology 25 (2007)

RELATION TO TIME GRANULARITY	CONTINUANT			OCCURRENT	
	INDEPENDENT		DEPENDENT		
ORGAN AND ORGANISM	Organism (NCBI Taxonomy)	Anatomical Entity (FMA, CARO)	Organ Function (FMP, CPRO)	Phenotypic Quality (PaTO)	<b>Biological Process (GO)</b>
CELL AND CELLULAR COMPONENT	Cell (CL)	<b>Cellular Component (FMA, GO)</b>	Cellular Function (GO)		
MOLECULE	Molecule (ChEBI, SO, RNAO, PRO)		<b>Molecular Function (GO)</b>		Molecular Process (GO)



RELATION TO TIME	CONTINUANT		OCCURRENT		
	INDEPENDENT	DEPENDENT			
ORGAN AND ORGANISM	Organism (NCBI Taxonomy)	Anatomical Entity (FMA, CARO)	Organ Function (FMP, CPRO)	Phenotypic Quality (PaTO)	Organism-Level Process (GO)
CELL AND CELLULAR COMPONENT	Cell (CL)	Cellular Component (FMA, GO)	Cellular Function (GO)		Cellular Process (GO)
MOLECULE	Molecule (ChEBI, SO, RNAO, PRO)		Molecular Function (GO)		Molecular Process (GO)

rationale of OBO Foundry coverage  
(homesteading principle)

# OBO FOUNDRY CRITERIA

The ontology is **open** and available to be used by all.

The ontology is in a **common formal language**.

The developers of the ontology agree in advance to **collaborate** with developers of other OBO Foundry ontology where domains overlap.

# OBO FOUNDRY CRITERIA

UPDATE: The developers of each ontology commit to its **maintenance in light of scientific advance**, and to soliciting community feedback for its improvement.

# OBO FOUNDRY CRITERIA

IDENTIFIERS: The ontology possesses a **unique identifier** space within OBO.

VERSIONING: The ontology provider has procedures for identifying distinct successive **versions**.

The ontology includes **textual definitions** for all terms.

# OBO FOUNDRY CRITERIA

CLEARLY BOUNDED: The ontology has a clearly specified and **clearly delineated content**.

DOCUMENTATION: The ontology is **well-documented**.

USERS: The ontology has a plurality of **independent users**.

# OBO FOUNDRY CRITERIA

ORTHOGONALITY: They commit to working with other Foundry members to ensure that, for any particular domain, there is community **convergence on a single controlled vocabulary.**

# OBO FOUNDRY CRITERIA

**COMMON ARCHITECTURE:** The ontology uses relations which are unambiguously defined following the pattern of definitions laid down in the **OBO Relation Ontology**

# How to submit **ontologies** to the Foundry

First step is to join one or more mailing lists  
(<http://obofoundry.org>)

1.to become familiar with the Foundry's  
collaborative methodology

2.to identify members with overlapping  
expertise

3.submit new ontology resources for informal  
consideration by existing members



# How to submit *single terms* to Foundry ontologies

Submit to ontology trackers/editor(s)

Orthogonality brings division of labor; so almost all development decisions can be made by the authors of single ontologies.

In cases of overlap, editors of involved ontologies will negotiate

In cases where these negotiations bring no satisfactory outcomes, OBO Foundry editors adjudicate

All decisions are revisable

# PROPOSED NEW CRITERIA

- OBO Foundry Ontologies should be organized in such a way as to reflect the top-level categories of dependent and independent / continuant and occurrent
- INSTANTIABILITY: Every term in an ontology should correspond to instances in reality
- Use singular nouns

# PROPOSED NEW CRITERIA

- Use terms which form part of ordinary (including technical) English; do not use phrases like EV-EXP-IGI
- Use Aristotelian definitions (An A =def. a B which Cs)
- Employ cross-products and compositionality in building terms and definitions

# THESE CRITERIA

provide **guidelines (traffic laws)** to new groups of ontology developers in ways which can ensure coordination of effort and provide for cumulation of benefits of lessons learned

The OBO Foundry map provides a navigational guide for those who need to find ontology resources

RELATION TO TIME	CONTINUANT				OCCURRENT
	INDEPENDENT		DEPENDENT		
GRANULARITY					
ORGAN AND ORGANISM	Organism (NCBI Taxonomy / placeholder)	Anatomical Entity (FMA, CARO)	Organ Function (placeholder)	Phenotypic Quality (PATO)	Biological Process (GO)
CELL AND CELLULAR COMPONENT	Cell (CL)	Cellular Component (FMA, GO)	Cellular Function (GO)		
MOLECULE	Molecule (ChEBI, SO, RNAO, PRO)		Molecular Function (GO)		Molecular Process (GO)

building out from this original map

ORGAN AND ORGANISM	Organism (NCBI Taxonomy / placeholder)	Anatomical Entity (FMA, CARO)	Organ Function (placeholder)			
CELL AND CELLULAR COMPONENT	Cell (CL)	Cellular Component (FMA, GO)	Cellular Function (GO)	Phenotypic Quality (PATO)	Disease (DO)	Biological Process (GO)
MOLECULE	(ChEBI, SO, RNAO, PRO)		Molecular Function (GO)			Molecular Process (GO)

ORGAN AND ORGANISM	Organism (NCBI Taxonomy / placeholder)	Anatomical Entity (FMA, CARO)	Organ Function (placeholder)	Phenotypic Quality (PATO)	Disease (DO)	Biological Process (GO)
CELL AND CELLULAR COMPONENT	Cell (CL)	Cellular Component (FMA, GO)	Cellular Function (GO)		Cellular Pathology ????	
MOLECULE	(ChEBI, SO, RNAO, PRO)		Molecular Function (GO)		Molecular Process (GO)	

ORGAN AND ORGANISM	Organism (NCBI Taxonomy / placeholder)	Anatomical Entity (FMA, CARO)	Organ Function (placeholder)	Phenotypic Quality (PATO)	Disease (DO)	Biological Process (GO)
CELL AND CELLULAR COMPONENT	Cell (CL)	Cellular Component (FMA, GO)	Cellular Function ??? (GO???)		Cellular Pathology ???	
MOLECULE	(ChEBI, SO, RNAO, PRO)		Molecular Function (GO)		Molecular Process (GO)	



ORGAN AND ORGANISM	Organism (NCBI Taxonomy / placeholder)	Anatomical Entity (FMA, CARO)	Organ Function (placeholder)			
CELL AND CELLULAR COMPONENT	Cell (CL)	Cellular Component (FMA, GO)	Cellular Function (GO)	Phenotypic Quality (PATO)	Disease (DO)	Biological Process (GO)
MOLECULE	(ChEBI, SO, RNAO, PRO)		Molecular Function (GO)			Molecular Process (GO)

ORGAN AND ORGANISM	Organism (NCBI Taxonomy / placeholder)	Anatomical Entity (FMA, CARO)	Organ Function (placeholder)			
CELL AND CELLULAR COMPONENT	Cell (CL)	Cellular Component (FMA, GO)	Cellular Function (GO)	Phenotypic Quality (PATO)	Disease (DO)	Biological Process (GO)
MOLECULE	2- and 3-D Structure (RNAO)	(PRO)	Molecular Function (GO)		Molecular Process (GO)	
	Small Molecule (ChEBI)	1-D Sequence (SO)				

ORGAN AND ORGANISM	Organism (NCBI Taxonomy / placeholder)	Anatomical Entity (FMA, CARO)	Organ Function (placeholder)			
CELL AND CELLULAR COMPONENT	Cell (CL)	Cellular Component (FMA, GO)	Cellular Function (GO)	Phenotypic Quality (PATO)	Disease (DO)	Biological Process (GO)
MOLECULE	2- and 3-D Structure (RNAO)	(PRO)	Molecular Function (GO)			Molecular Process (GO) ?????
	Small Molecule (ChEBI)	1-D Sequence (SO)				Reactome

ORGAN AND ORGANISM	Organism (NCBI Taxonomy / placeholder)	Anatomical Entity (FMA, CARO)	Organ Function (placeholder)			
CELL AND CELLULAR COMPONENT	Cell (CL)	Cellular Component (FMA, GO)	Cellular Function (GO)	Phenotypic Quality (PATO)	Disease (DO)	Biological Process (GO)
MOLECULE	2- and 3-D Structure (RNAO)	(PRO)	Molecular Function (GO)	Phenotypic Quality of Molecule ????		Molecular Process (GO) ?????
	Small Molecule (ChEBI)	1-D Sequence (SO)				Reactome

RELATION TO TIME	CONTINUANT			OCCURRENT	
	INDEPENDENT		DEPENDENT		
GRANULARITY					
ORGAN AND ORGANISM	Organism (NCBI Taxonomy)	Anatomical Entity (FMA, CARO)	Organ Function (FMP, CPRO)	Phenotypic Quality (PaTO)	<b>Biological Process (GO)</b>
CELL AND CELLULAR COMPONENT	Cell (CL)	<b>Cellular Component (FMA, GO)</b>	Cellular Function (GO)		
MOLECULE	Molecule (ChEBI, SO, RNAO, PRO)		<b>Molecular Function (GO)</b>		Molecular Process (GO)

Building out from the original GO

# clinical data includes

clinical records

clinical trial data

demographic data

National Hospital Discharge Survey

National Ambulatory Medical Care Surveys

MEDPAR

Medicare's national claims data base

# Community / Population Ontology

- family, clan
- ethnicity
- religion
- diet
- social networking
- education (literacy ...)
- healthcare (economics ...)
- household forms
- demography
- public health
- ...

RELATION TO TIME	CONTINUANT				OCCURRENT
	INDEPENDENT		DEPENDENT		
GRANULARITY					
<b>ORGAN AND ORGANISM</b>	Organism (NCBI Taxonomy)	Anatomical Entity (FMA, CARO)	Organ Function (FMP, CPRO)	Phenotypic Quality (PaTO)	<b>Biological Process (GO)</b>
CELL AND CELLULAR COMPONENT	Cell (CL)	<b>Cellular Component</b> (FMA, GO)	Cellular Function (GO)		
MOLECULE	Molecule (ChEBI, SO, RnaO, PrO)		<b>Molecular Function</b> (GO)		Molecular Process (GO)



RELATION TO TIME GRANULARITY	CONTINUANT			OCCURRENT	
	INDEPENDENT		DEPENDENT		
ORGAN AND ORGANISM	<b>Family, Community, Deme, Population</b>		Organ Function (FMP, CPRO)	Phenotypic Quality (PaTO)	<b>Biological Process (GO)</b>
	Organism (NCBI Taxonomy)	Anatomical Entity (FMA, CARO)			
CELL AND CELLULAR COMPONENT	Cell (CL)	Cellular Component (FMA, GO)	Cellular Function (GO)		
MOLECULE	Molecule (ChEBI, SO, RnaO, PrO)		<b>Molecular Function (GO)</b>		Molecular Process (GO)

RELATION TO TIME	CONTINUANT			OCCURRENT
	INDEPENDENT		DEPENDENT	
GRANULARITY				
COMPLEX OF ORGANISMS	<b>Family, Community, Deme, Population</b>		<b>Population Phenotype</b>	<b>Population Process</b>
ORGAN AND ORGANISM	Organism (NCBI Taxonomy)	Anatomical Entity (FMA, CARO)	Organ Function (FMP, CPRO)	Phenotypic Quality (PaTO)
CELL AND CELLULAR COMPONENT	Cell (CL)	Cellular Component (FMA, GO)	Cellular Function (GO)	
MOLECULE	Molecule (ChEBI, SO, RnaO, PrO)		<b>Molecular Function (GO)</b>	Molecular Process (GO)

# The Environment Ontology



OBO Foundry

Genomic Standards Consortium

National Environment Research Council (UK)

USDA, Gramene, J. Craig Venter Institute, ...

<http://environmentontology.org/>

RELATION TO TIME  GRANULARITY	CONTINUANT			OCCURRENT		
	INDEPENDENT		DEPENDENT			
<b>COMPLEX OF ORGANISMS</b>	<b>Family, Community, Deme, Population</b>		<b>E N V I R O N M E N T</b>	<b>Population Phenotype</b>	<b>Population Process</b>	
ORGAN AND ORGANISM	Organism (NCBI Taxonomy)	(FMA, CARO)		Organ Function (FMP, CPRO)	Phenotypic Quality (PaTO)	Biological Process (GO)
CELL AND CELLULAR COMPONENT	Cell (CL)	Cell Component (FMA, GO)		Cellular Function (GO)		
MOLECULE	Molecule (ChEBI, SO, RnaO, PrO)			<b>Molecular Function (GO)</b>		

RELATION TO TIME	CONTINUANT	
	INDEPENDENT	
GRANULARITY		
COMPLEX OF ORGANISMS	<b>Family, Community, Deme, Population</b>	
ORGAN AND ORGANISM	Organism (NCBI Taxonomy)	(FMA, CARO)
CELL AND CELLULAR COMPONENT	Cell (CL)	Cell Component (FMA, GO)
MOLECULE	Molecule (ChEBI, SO, RnaO, PrO)	

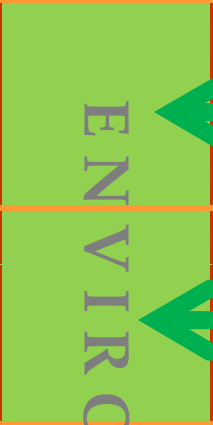
ENVIRONMENT

**Environment of population**

**Environment of single organism**

**Environment of cell**

**Molecular environment**

RELATION TO TIME	CONTINUANT	
	INDEPENDENT	
GRANULARITY		
COMPLEX OF ORGANISMS	Family, Community, Deme, Population	 <b>Environment of population</b>
ORGAN AND ORGANISM	Organism (NCBI Taxonomy)	(FMA, CARO) <b>Environment of single organism*</b>

\* The sum total of the conditions and elements that make up the surroundings and influence the development and actions of an individual.

RELATION TO TIME	CONTINUANT	
	INDEPENDENT	
GRANULARITY		
COMPLEX OF ORGANISMS	ENVIRONMENT	biome / biotope, territory, habitat, neighborhood, ...
ORGAN AND ORGANISM		work environment, home environment; host/symbiont environment; ...
CELL AND CELLULAR COMPONENT		extracellular matrix; chemokine gradient; ...
MOLECULE		hydrophobic surface; virus localized to cellular substructure; active site on protein; pharmacophore ...

# Applications of EnvO in biology

- Support the annotation of meta-data related to:
  - Data about biological samples produced from various technologies
    - Metagenomics, Metabolomics, Proteomics, Transcriptomics, Genomics...
  - Data produced from remote sensing equipment
  - Images
    - Web 2.0, tagging
  - Physical holdings
    - Museum artifacts, (preserved) biological samples / organisms
  - ...anything that has an ***environment***





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Search on : *Str containB141* [NEW SEARCH](#)

Species name	Staphylococcus aureus Rosenbach 1884 AL
Strain number	B141
Other collections number	<a href="#">See other collection numbers</a>
Original substract	Swimming pool water
Country of origin	Morocco
Collector name	Benabdellah
Isolator name	ONEP
Determinator name	ONEP
Redeterminator	LMG
Name as redetermined	Staphylococcus aureus subsp.aureus
Depositor name	ONEP
History	<-2000, S. Benabdellah ONEP Rabat (Staphylococcus aureus). (19... pool water
Media and temperature	1 <a href="#">Details</a> 37°C

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Species name	Staphylococcus aureus Rosenbach 1884 AL
Strain number	B13
Other collections number	<a href="#">See other collection numbers</a>
Original substract	Sputum
Country of origin	Morocco
Collector name	A. Alaoui
Isolator name	FM Rabat
Determinator name	FM Rabat
Redeterminator	LMG
Name as redetermined	Staphylococcus aureus subsp.aureus
Depositor name	FM Rabat
History	<-2000, A. Alaoui FM Rabat (Staphylococcus aureus). (1999), Hôpital human, sputum
Media and temperature	<a href="#">1Details</a> 37°C



# to enhance coordination of research

Obesity, Physical Activity & Built Space in New York City
Environmental and Family Influences on Adolescent Overweight
Walking on Campus: Correlates & Web Tools
Measuring Physical Activity Affordances in Preschool Outdoor Environments
Environmental Predictors of Elderly Obesity
Neighborhood Food Environment, Diet & Health: Quasi-Experimental Study

# How EnvO currently works for information retrieval

**Retrieve all experiments on organisms obtained from:**

- deep-sea thermal vents
- arctic ice cores
- rainforest canopy
- alpine melt zone

**Retrieve all data on organisms sampled from:**

- hot and dry environments
- cold and wet environments
- a height above 5,000 meters

**Retrieve all the omic data from soil organisms subject to:**

- moderate heavy metal contamination

Environment = totality of circumstances external to a living organism or group of organisms

- pH
- evapotranspiration
- turbidity
- available light
- predominant vegetation
- predatory pressure
- nutrient limitation ...

# extending EnvO to the clinical domain

- dietary patterns (Food Ontology: FAO, USDA) ... allergies
- neighborhood patterns
  - built environment, living conditions
  - climate
  - social networking
  - crime, transport
  - education, religion, work
  - health, hygiene
- disease patterns
  - bio-environment (bacteriological, ...)
  - patterns of disease transmission (links to IDO)

# a new type of patient data

a patient's *environmental history*

use EnvO and the community ontology to  
mine relations between disease  
phenotypes and environmental patterns  
and patterns of community behavior

e.g. for cows



# Another way the OBO Foundry is being used

The Senselab/NeuronDB\* comprehends three types of neuronal properties:

- voltage gated conductances

- neurotransmitter receptors

- neurotransmitter substances

Many questions immediately arise: what *are* receptors? Proteins? Protein complexes? The Foundry framework provides an opportunity to evaluate such choices.

\* <http://senselab.med.yale.edu/>

# Senselab/NeuronDB

The GO Molecular Function (MF) ontology already has classes such as *receptor activity* (GO\_0004872) plus subclasses describing receptor activities already referred to in NeuronDB.

This provides a roadmap for further development. Review the 130 receptor classes to see if they exist in MF, where not, create subclasses and submit to GO for future inclusion. We can then e.g. take advantage of GO Annotations to find the proteins that correspond to these receptor classes in different species.

