# BINF 8211/6211
# Design and Implementation of Bioinformatics Databases
# Lecture #18

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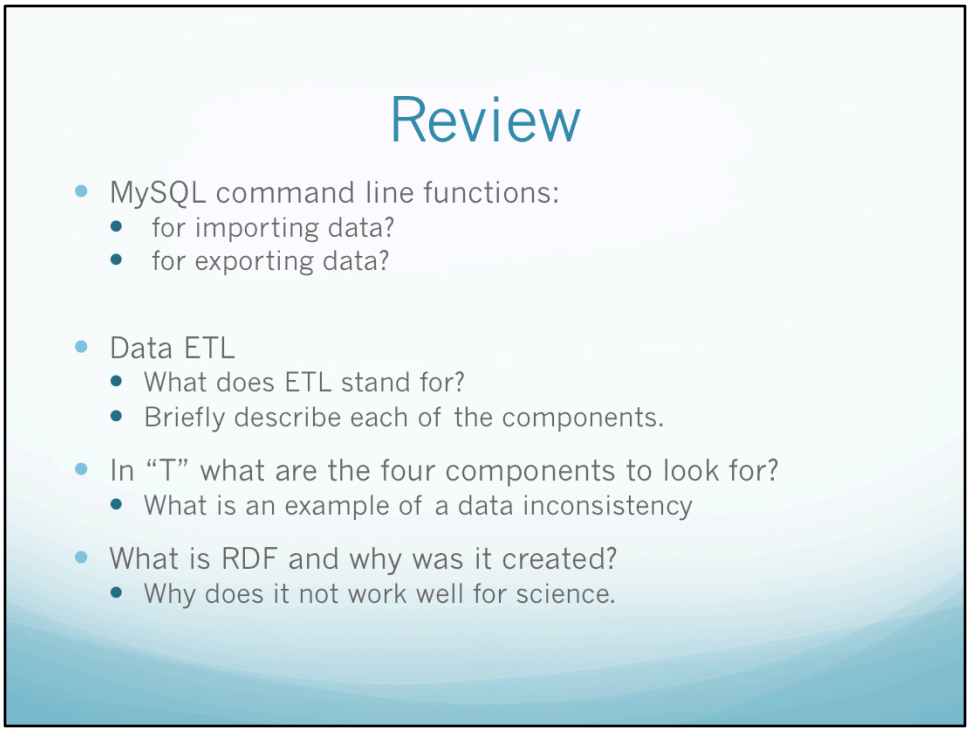
Review Relational Model




1) mysqlimport --columns='head -n 1 $yourfile' --ignore-lines=1 dbname $yourfile
2) SELECT * FROM Cars INTO OUTFILE '/tmp/cars.csv' FIELDS TERMINATED BY ',';
3) Extract, Transform, Load

Acquisition, QC, Import

4) Transform can be thought of as four components: Your QC should look for incomplete data, incorrect data, incomprehensible data and inconsistencies in the data.
5) Data where the units do not align, variation in coded meaning.

Illumina read FASTQ scores are different depending on the software version that was run. Same code can imply a different code.

6) Resource Description Framework was developed by the W3C to have a manner in which to build a Semantic Web.
7) Known issues include these issues include 'too few entailments', 'contradiction classes' and 'size of the universe'

# Lecture Schedule

- MySQL tools
- OWL
- OBO part II

# SQL Topics (MySQL)

- String tools
- Date and Time
- Math functions

# MySQL String tools

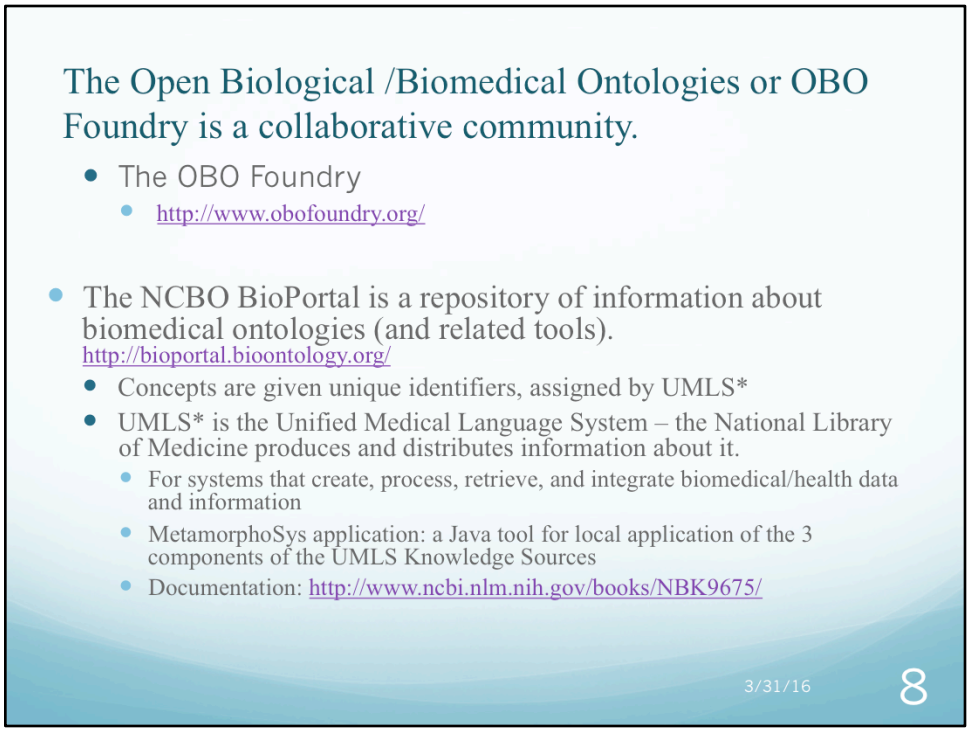
- Compare strings — strcmp(*string1*,*string2*)
- Convert to lower case — lower(*string*)
- Convert to upper case — upper(*string*)
- Left-trim whitespace (similar right) — ltrim(*string*)
- Substring of string — substring(*string*,*index1*,*index2*)
- Encrypt password — password(*string*)
- Encode string — encode(*string*,*key*)
- Decode string — decode(*string*,*key*)

# MySQL (Data Time)

- Get date
  - curdate()
- Get time
  - curtime()
- Extract day name from date string
  - dayname(*string*)
- Extract day number from date string
  - dayofweek(*string*)
- Extract month from date string
  - monthname(*string*)

# MySQL (Math Functions)

- Count rows per group
  COUNT(*column* | *)
- Average value of group
  AVG(*column*)
- Minumum value of group
  MIN(*column*)
- Maximum value of group
  MAX(*column*)
- Sum values in a group
  SUM(*column*)
- Absolute value
  abs(*number*)
- Rounding numbers
  round(*number*)
- Largest integer not greater
  floor(*number*)
- Smallest integer not smaller
  ceiling(*number*)
- Square root
  sqrt(*number*)
- *n*th power pow(*base*,*exponent*)
- random number *n*, $0<n<1$
  rand()
- sin (similar cos, etc.)
  sin(*number*)



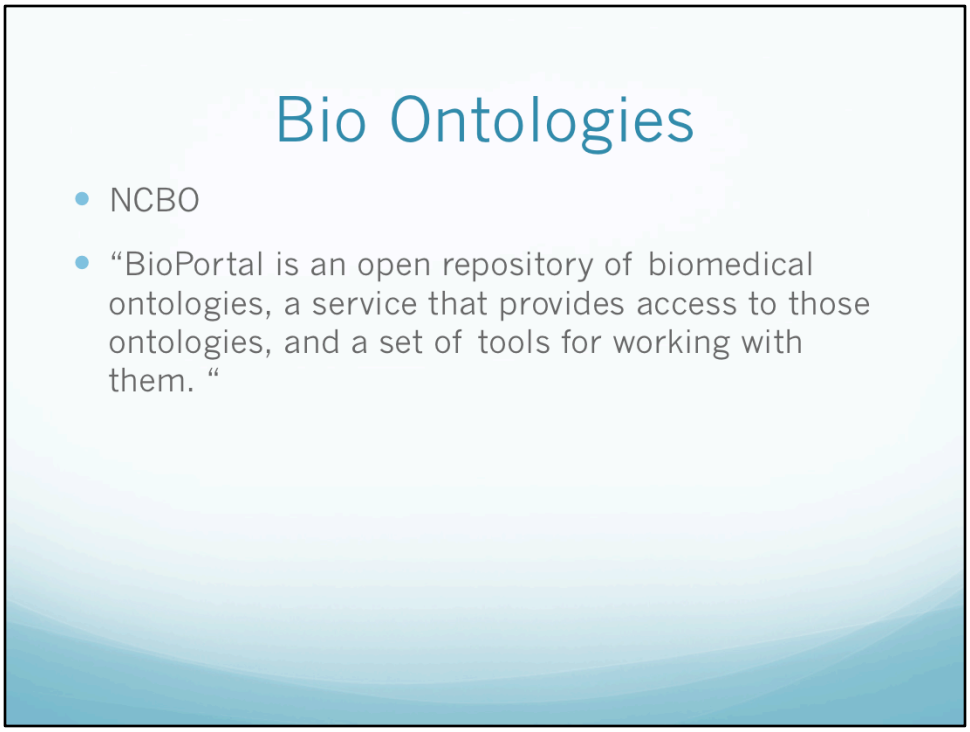

The tools help you build well-formed and correct structures – they help you find mistakes, and also layout existing structures, and compare existing structures.

is a collective of ontology developers that are committed to collaboration and adherence to shared principles. The mission of the OBO Foundry is to develop a family of interoperable ontologies that are both logically well-formed and scientifically accurate. To achieve this, OBO Foundry participants voluntarily adhere to and contribute to the development of an evolving set of principles including open use, collaborative development, non-overlapping and strictly-scoped content, and common syntax and relations, based on ontology models that work well, such as the Gene Ontology (GO).

Some of the NCBO ontologies are private – a subscription is required. It uses a REST API.

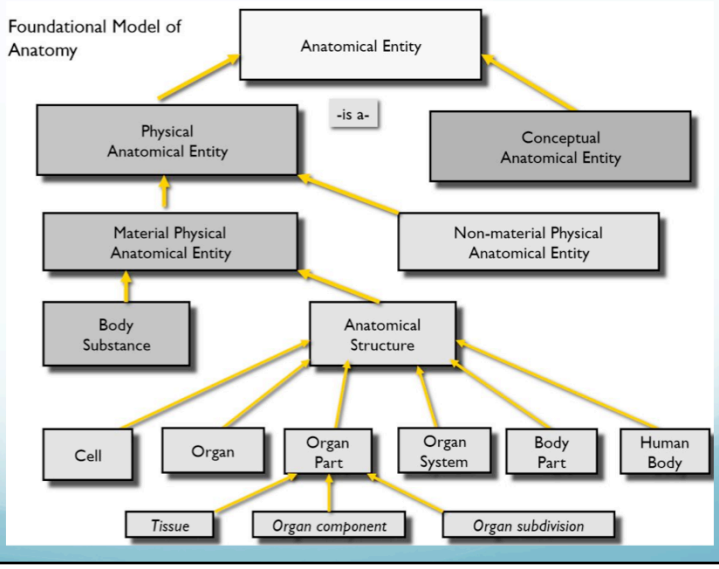
Patient records, scientific literature (like MeSH), guidelines, public health data.

Associated resources include the Semantic Network, the SPECIALIST lexicon, and Lexical Tools.
There is a Meta thesaurus that must be licensed since it includes proprietary content.




National Center for Bio-Ontologies
Largely Medically related…

# Bio-Ontology Example, the Foundational Model of Anatomy

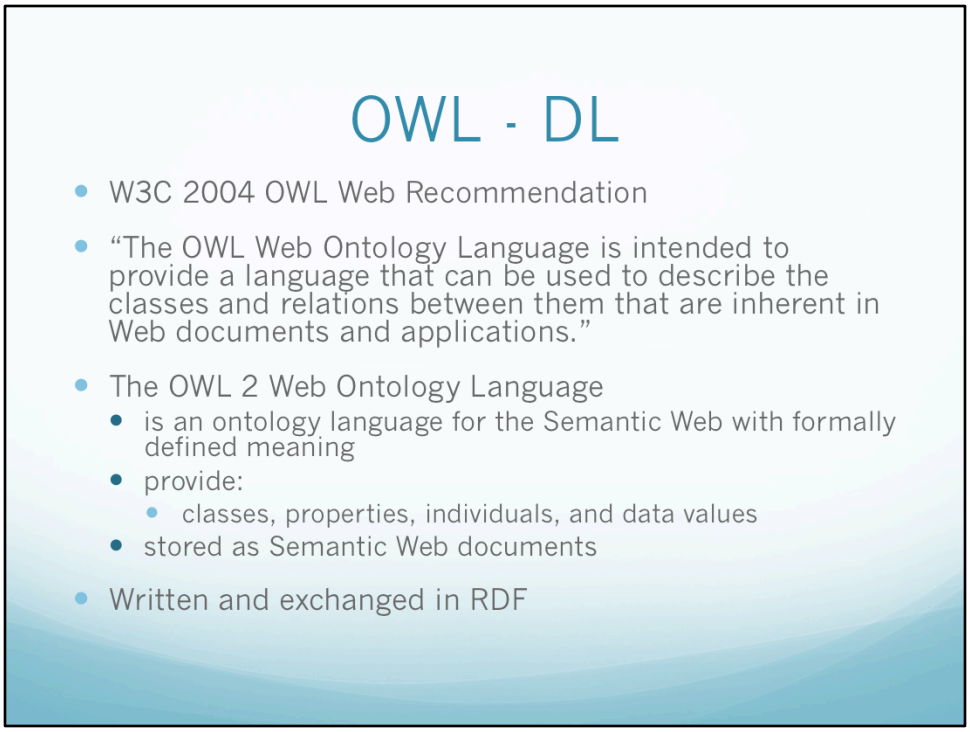


## Bio-Ontology Research Activities

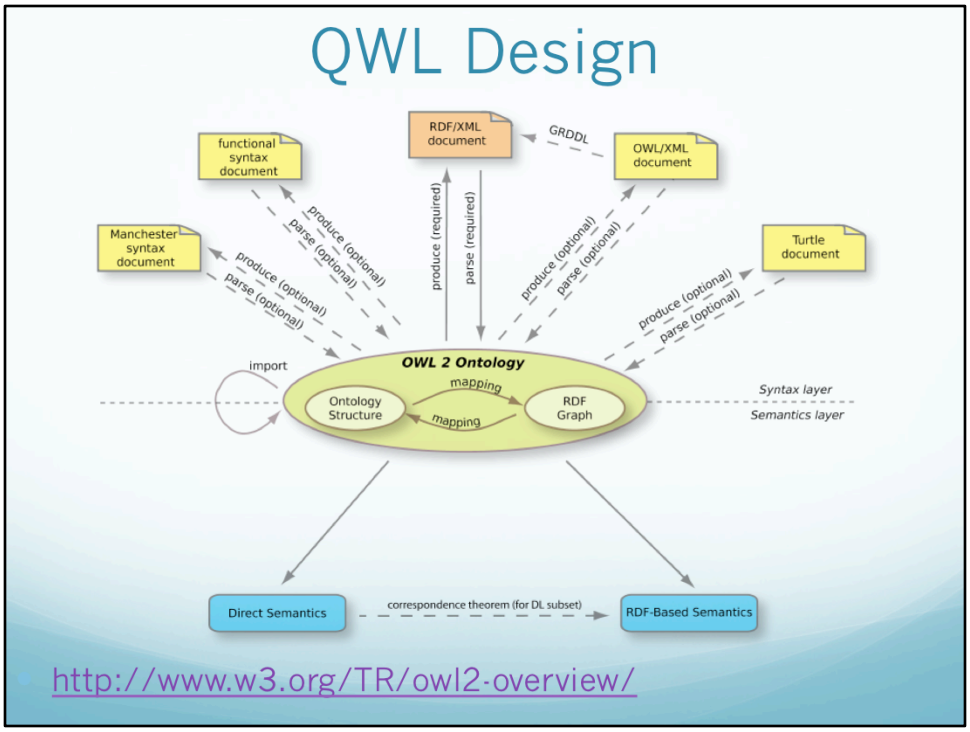
- Develop the controlled vocabularies
  - Terms
  - Rules
- Schema Definition
  - Conceptual model
  - Implementation-specific schemas
- Domain maps are created
- Database query mechanisms are developed
- Resolution rules are defined
- More applications are developed to extend the utility and computational access of the resource


"Ontology is the philosophical study of the nature of being, becoming, existence, or reality, as well as the basic categories of being and their relations. Traditionally listed as a part of the major branch of philosophy known as metaphysics, ontology often deals with questions concerning what entities exist or may be said to exist, and how such entities may be grouped, related within a hierarchy, and subdivided according to similarities and differences. Although ontology as a philosophical enterprise is highly theoretical, it also has practical application in information science and technology, such as ontology engineering." ~Wikipedia

Onto ~ "Being, That which is"



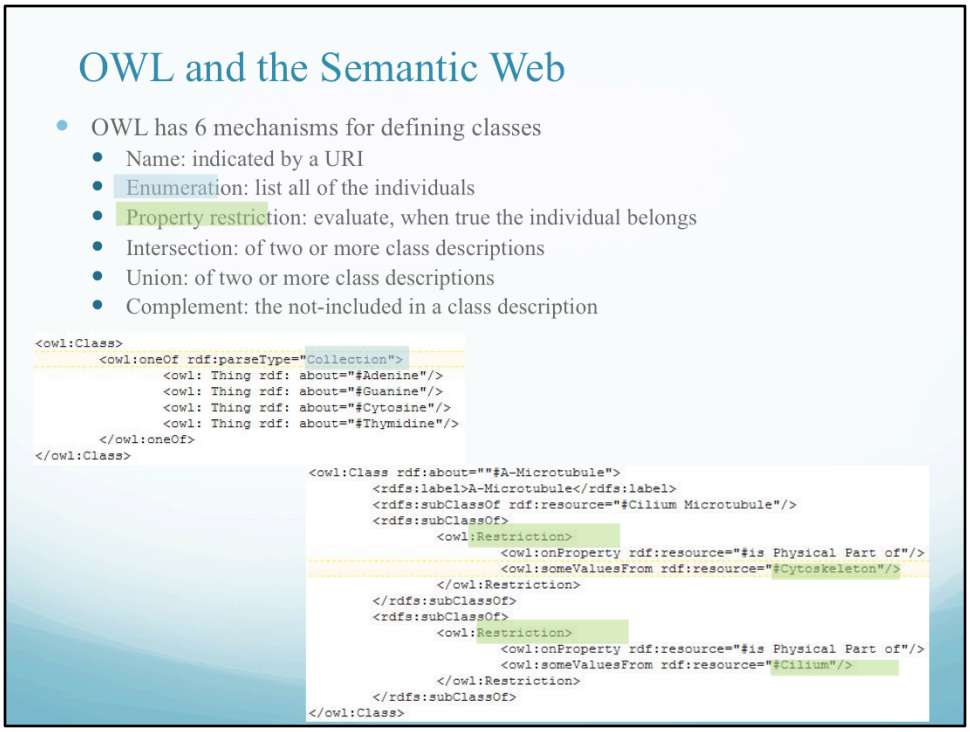

"The World Wide Web as it is currently constituted resembles a poorly mapped geography. Our insight into the documents and capabilities available are based on keyword searches, abetted by clever use of document connectivity and usage patterns. The sheer mass of this data is unmanageable without powerful tool support. In order to map this terrain more precisely, computational agents require machine-readable descriptions of the content and capabilities of Web accessible resources. These descriptions must be in addition to the human-readable versions of that information."




"The OWL 2 Structural Specification document defines the abstract structure of OWL 2 ontologies, but it does not define their meaning. The Direct Semantics [*OWL 2 Direct Semantics*] and the RDF-Based Semantics [*OWL 2 RDF-Based Semantics*] provide two alternative ways of assigning meaning to OWL 2 ontologies, with a correspondence theorem providing a link between the two. These two semantics are used by reasoners and other tools, e.g., to answer class consistency, subsumption and instance retrieval queries."


The class that contains this rdfs:subClassOf element is a subclass of an anonymous class of the form 'all instances for which all values of property P are from class V'. This does not mean that an instance of the class must have the property P only that if it has P then it must have a value from V.

In the second example we define a class by stating what it is **not** – using the Cell Type Ontology CL, and we restrict the definition of cultured cells that do not express the CD8 receptor on the surface using the keyword owl:complementOf.

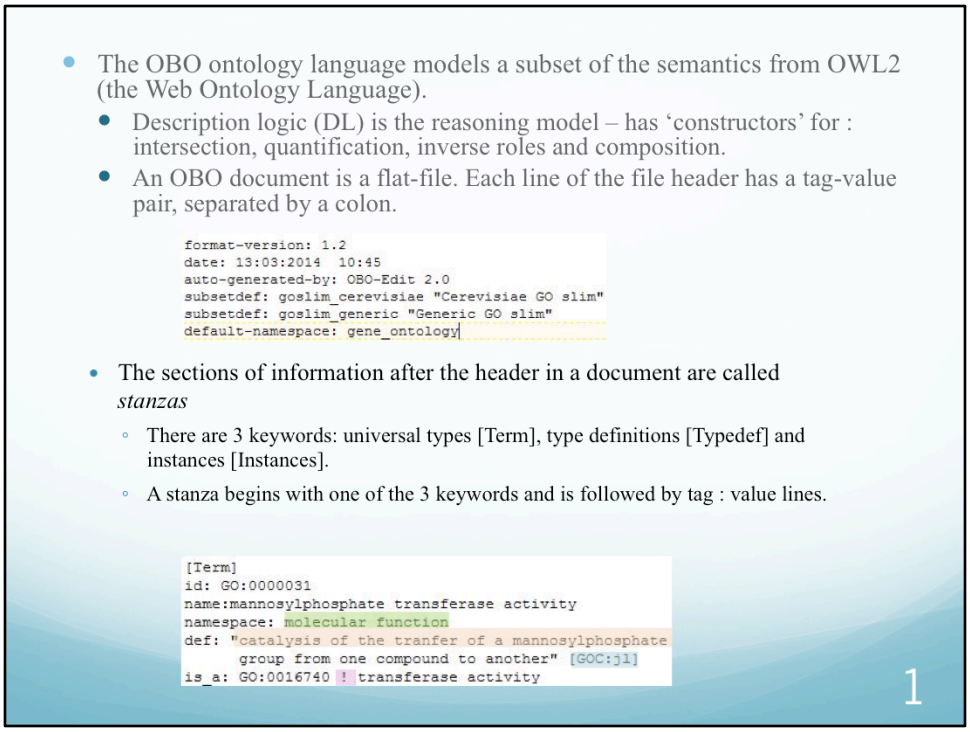



The Web Ontology Language (OWL) was developed as a very generic language to enable interoperability of anything that can appear on the web. This required availability, structure rules, definition rules and consistency in applying the rules.
OWL uses the Resource Description Framework (RDF) data model and language.

The logical model lets us reason about and extend existing relationships.
Definitions can be constructed using a keyword intersection_of also.

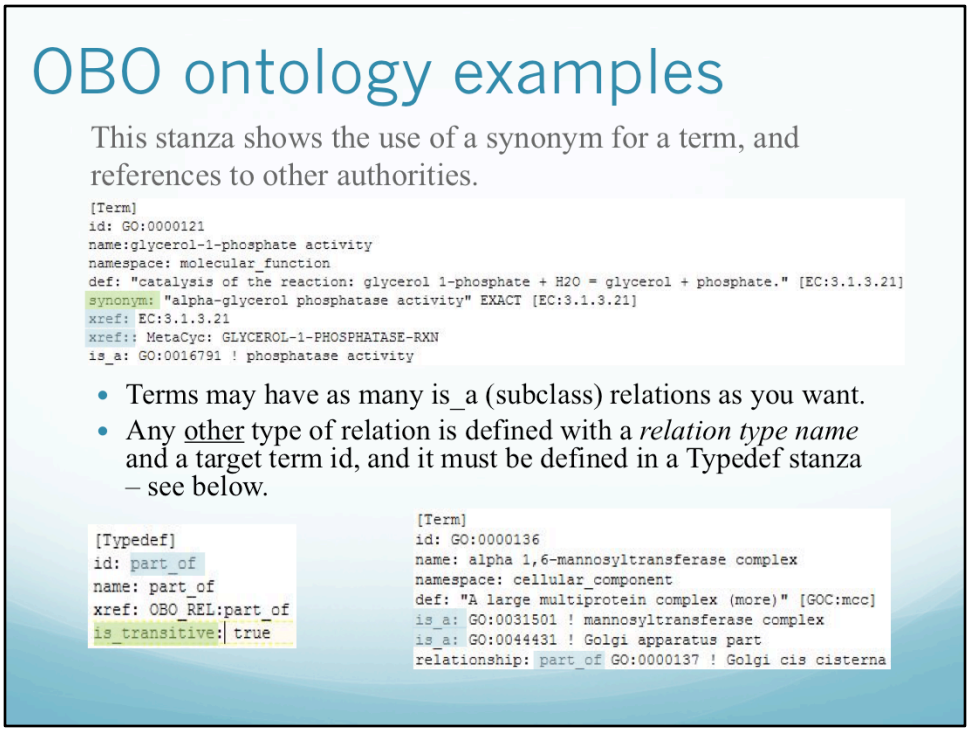
Classes are defined by placing restrictions on the things that belong to the class. Property restrictions are often used (second example).

Here an A-Microtubule is a complete cylindrical microtubule that is part of a microtubule doublet in a cilia.
A-Microtubule is a subclass of Cilium Microtubule
The two subClass of restrictions define that is must be a part of the Cytoskeleton and that is must be a physical part of the Cilium.

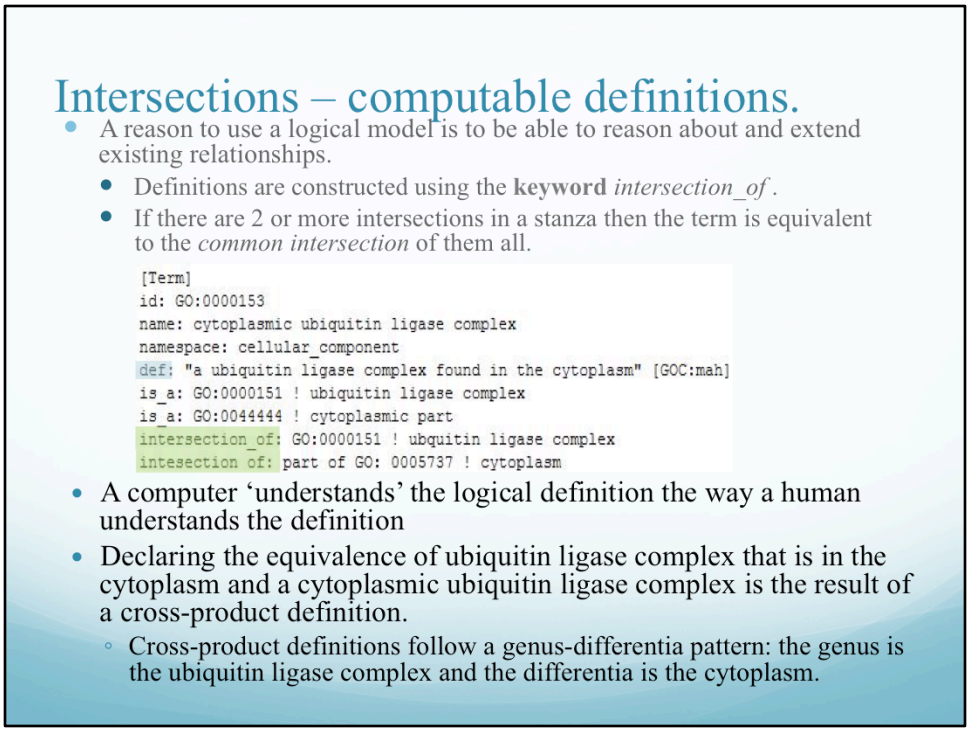



Format-version is a tag (or an element) and it's value is after the colon (here it is '1.2')
Auto-generated-by tells you what editor was used to create the file. This particular tool is widely used by the GO consortium, here we are looking at a GoSlim document describing GO terms for yeast.
Subsetdef tag tells you which dictionary (the cv terms) that will be used in the following information.

You can add empty lines for better human readability – parsers ignore them.
However a NL or CR is interpreted as a new line, so you would not include that in real text.
The id is a unique identifier *within the dictionary* being used.
Each term has a name.
Within the default namespace you can have a sub-category.
In the Gene Ontology there are 3 related categories, Molecular Function, Biological Process and Cellular Component - the namespace in this part of the hierarchy tells which of these you are in.
The definition defines the function and the authority – here the authority is GOC, standing for the Gene Ontology Curator responsible for the definition – this gives provenance.
The type definition belongs to s controlled vocabulary within the ontology, here we have is_a: and a value, and a comment. Note that comments are noted with a ! (see




So for the is_a relationship type you do not have to declare the type (it is the default) but for any others you must declare it - here the part_of relationship type is cross-referenced to the OBO Relationship namespace, and a further characteristic is stated, which can be used for reasoning on new elements (that it is transitive, that is, if a is part of b and b is part of c then a is part of c is true).




By computable we mean the logic is something that can be described to a computer = in this case a comparison is carried out that finds what is common between two sets, one a molecular process and the other a cellular component. The set is the collection of related items that your statements define.

Genus+ Differentia are the two parts of a definition. A genus is a family, which is an existing definition and a differentia is the portion of the new definition that is not provided the original definition. You are creating further subgroups, usually through a distinguishing characteristic and according to the accepted definitions of types.

Composing these two parts follows rules. Some of these are quite general – don't create a circular definition, use a property that is an essential attribute, find a property that classifies a group of things and not a single individual, and declare a perperty in terms that are unambiguous. Ideally the property should be declared in a positive sense rather than in the does –not-have (negative) sense.


This began as GO-Edit, then DAG-Edit, then finally OBO-Edit.
It can output a number of file formats, including its own OBO format, and the GO format, and the OWL format.
A term is called a Class in this system – the entities and processes that you want to define and related.
Relationship types are the allowed relationships (whose logic can be completely and accurately defined) such as is_a and part_of types – the ways that child classes descend from parent classes.
Relationships are the specific relationships that are assigned to a given parent and child term.
The OBO format was developed to track the large amount of meta-data that biological scientists had to manage, as well as the provenance that is essential to the scientific process. Some of the semantics are quite complicated compared to the Semantic Web as well.

# Cardinality restrictions

- Cardinality can be constrained to an exact number, a minimum number and a maximum number using owl:cardinality, owl:minCardinality and owl:maxCardinality.

```
<owl:Class rdf:ID="Wine">
<rdfs:subClassOf rdf:resource="&food;PotableLiquid"/>
rdfs:subClassOf>
        <owl:Restriction>
                <owl:onProperty rdf:resource=#hasMaker"/>
                <owl:cardinality>1</owl:cardinality>
        </owl:Restriction>
</rdfs:subClassOf>
<rdfs:subClassOf>
        <owl:Restriction>
                <owl:onProperty rdf:resource=#hasMaker"/>
                <owl:allValuesFrom rdf:resource="#Winery"/>
        </owl:Restriction>
</rdfs:subClassOf>
<rdfs:subClassOf>
        <owl:restriction>
                <owl:onProperty rdf:resource="#madeFromGrape"
                <owl:minCardinality>1</owl:minCardinality>
        </owl:Restriction>
</rdfs:subClassOf>
</owl:Class>
```

```
<owl:Class rdf:ID="WhiteBurgundy">
        <owl:intersectionOf rdf:parseType="Collection">
                <owl:Class rdf:about="#Burgundy"/>
                <owl:Class rdf:about="WhiteWine"/>
        </owl:intersectionOf>
</owl:Class>

<owl:Class rdf:about="#WhiteBurgundy">
        <rdfs:subClassOf>
                <owl:Restriction>
                        <owl:onProperty rdf:resource="#madeFromGrape"/>
                        <owl:hasValue rdf:resource="#ChardonnayGrape"/>
                </owl:Restriction>
        </rdfs:SubClassOf>
        <rdfs:subClassOf>
                <owl:Restriction>
                        <owl:onProperty rdf:resource="#madeFrom Grape"/>
                        <owl:maxCardinality>1</owl:maxCardinality>
                </owl:Restriction>
        </rdfs:subCLassOf>
</owl:Class>
```

This is a class definition for a wine varietal
It is an intersection of classes White Wine (a subclass of wine) and Burgundy (a region and a style of wine that is made from exactly one grape variety, Chardonnay)
Because we made a constraint on madeFromGrape to have a minimum cardinality of 1 inherited from the class Wine, and a maximum cardinality of 1 for the class White Burgundy, this type of wine can be made from exactly 1 varietal of grape.

# OWL Property Restrictions and functions

- In addition to IntersectionOf, OWL has a UnionOf operator that lets you *combine* 2 collections
- Property characteristics are defined so that logical rules can be evaluated
  - Owl:Transitive Property (Mitochondria in Cytoplasm and Cytoplasm in Cell so Mitochondrion in Cell)
  - owl:SymmetricProperty (hasSameColorAs) expresses equivalence
  - owl:inverseOf lets you express if an edge has two arrows (has_part and part_of)
  - Owl:disjointWith (for classes). Use differentFrom (for two individuals), distinctMembers (for two sets of individuals)
- You can define a function with owl:functionalProperty as the example shows.

```
<owl:ObjectProperty rdf:ID="encodes Amino Acid">
        <rdf:type rdf:resource="&owl;FunctionalProperty"/>
        <rdfs:domain rdf:resource="#Codon"/>
        <rdfs:range rdf:resource="#Amino Acid"/>
</owl:ObjectProperty>
```

The function declares that an amino acid is always coded for by a specific codon. The inverse is not true since the code is redundant.

## Editing OWL with Protégé

- Protégé is an editor application used to build ontologies that has a graphical user interface and can display ontologies in a number of ways. It is free and open source, and frequently updated.
  - http://protege.stanford.edu
- For a tutorial on getting started, see the University of Manchester Web site
  - http://owl.cs.manchester.ac.uk
- Most OWL ontologies are developed with Protégé.

## Summary

- The OBO ontologies have been designed to support science, especially the biomedical sciences – the emphasis has been on use of the ontology.
  - Most are taxonomies, classifications of a domain of biology using is_a and part_of compositional relations for the most part.
  - OBO has been extending its syntax to include structures like the intersections.
  - OBO is a significant part of the OW-DL (data definition language)
- OWL ontologies have been developed by computer scientists who have focused on the frameworks, algorithms and software
  - The RDF model and RDF Schema language are the basis of the OWL framework.
  - There is ongoing work to be able to readily interconvert between OWL and OBO.