

# BIOMEDIN210, Homework #2

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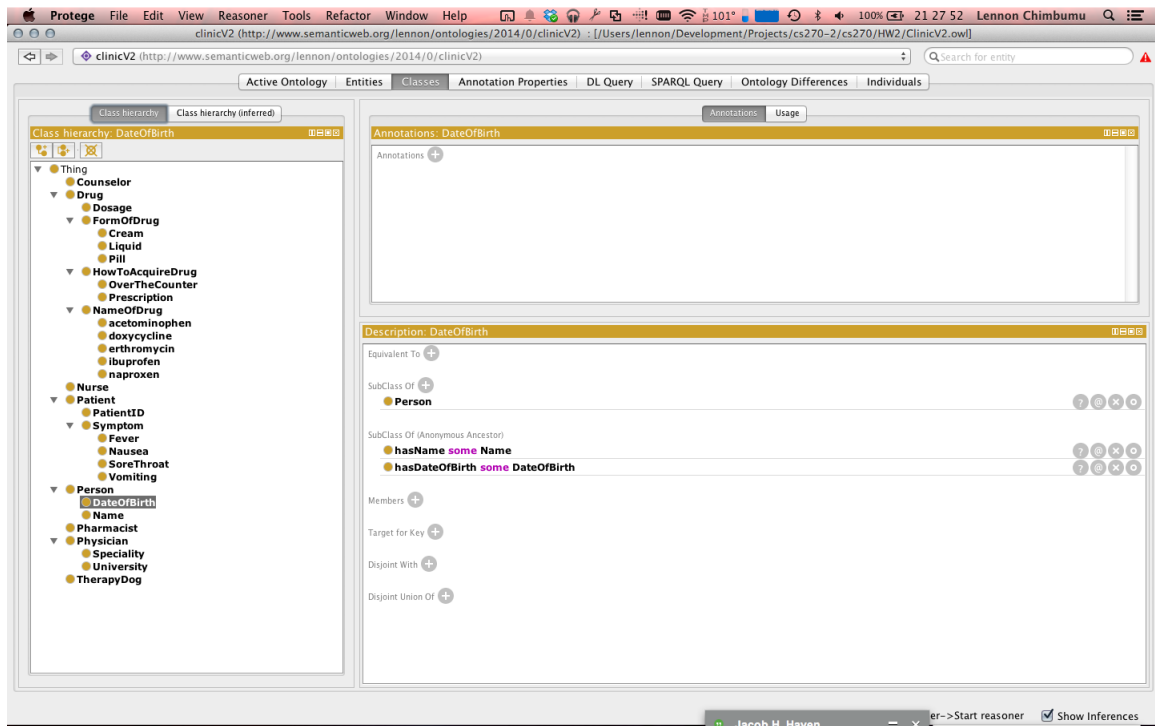
January 31, 2014

## Problem 1 (Considerations in modeling)

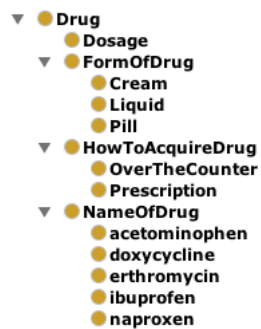
1. The closed-world assumption is that anything that is not known to be true must be considered false.
2. The open-world assumption is that failure to derive a conclusion does not mean the opposite is true. It is the assumption that the truth-value of a statement is actually independent of whether or not it is known by any single observer or agent to be true.
3. The unique-name assumption is the assumption that different names refer to different entities.
4. In Frames, if two objects have different names, they are assumed to be different. So the unique name assumption applies to Frames.
5. The open-world assumption applies to OWL.
6. These assumptions affect the choice of ontological language we might choose in modeling a certain domain.
7. After discussion, we believe that both open and closed-world assumptions have their place. Take the simple example: if I do not have information about whether person X is a spy, it is not safe to assume that they are a spy (open), or that they are not a spy (closed). For the NSA, it is safer for them to assume that the world is “negative,” as in, assume “everyone” is a spy and only trust those who have been specifically white-listed. Thus, it makes more sense for the NSA to use an open-world assumption, especially if they are interested in modeling who has contact with whom and who knows what information. It is “safer” for the NSA to assume that if no information is available, that there is contact between parties and that information is being transferred and thus to be conservative in their actions, which falls under an open-world assumption which states that under circumstances when a conclusion cannot be derived, we cannot assume that the opposite is true.

## Problem 2 (Modeling with Protege)

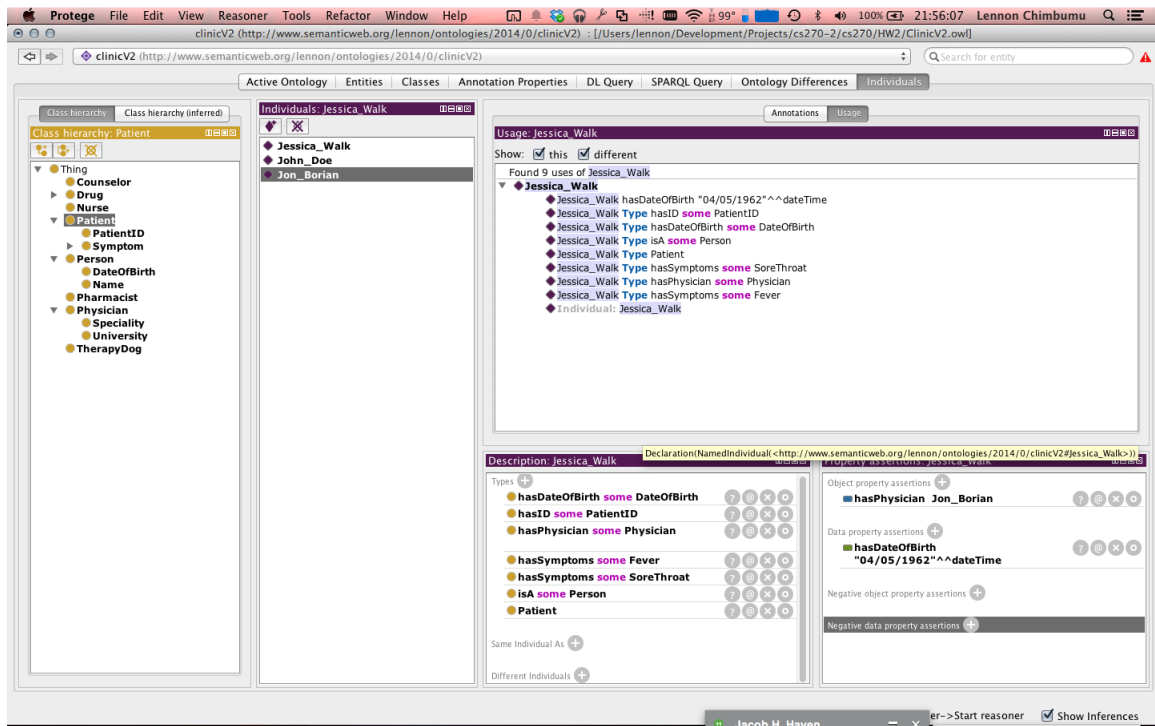
1. The class structure for the data provided looks like:



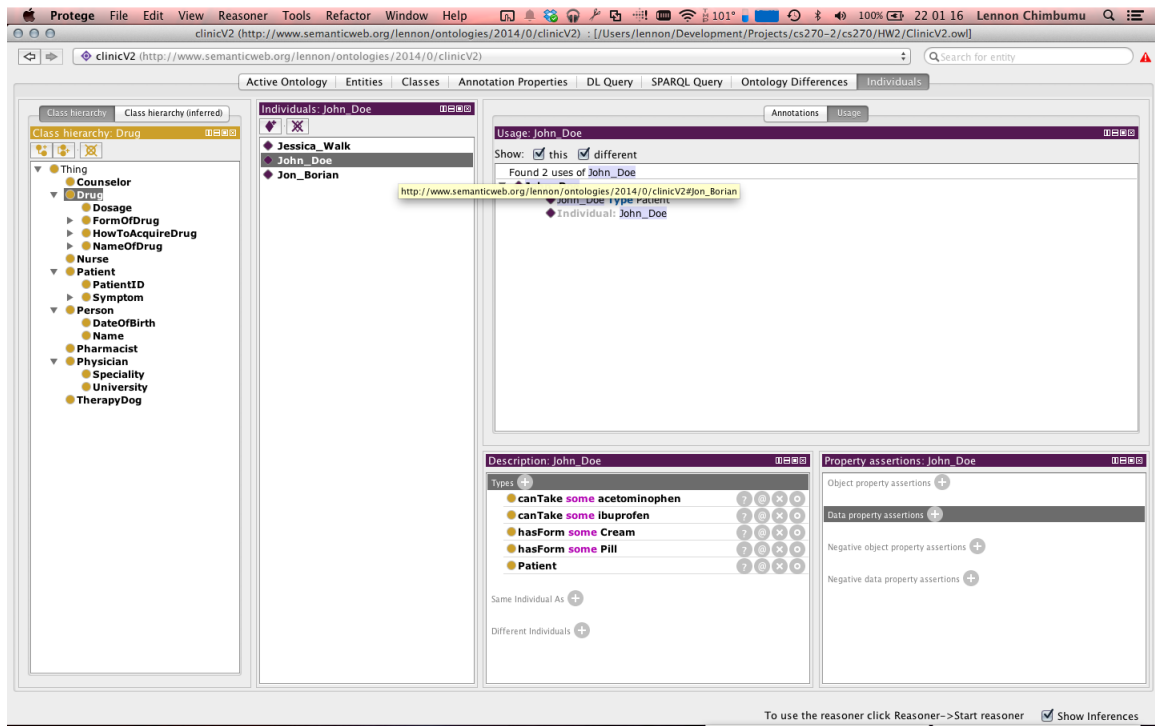
The **Drug** class looks like:



2. The Hierarchy classifies sickle cell anemia as an **autosomal recessive disease** which is a subclass of **autosomal genetic disease** which is a subclass of **monogenetic disease** which is a subclass of **genetic disease** which is a subclass of **disease**. This hierarchy may be useful for determining the best treatment for a disease. In the case of sickle cell anemia, we may want to use methods in gene therapy to try and treat it.
3. Jessica Walk's record looks like:



John Doe's record looks like:



### Problem 3

1. A class is unsatisfiable if there is a contradiction in the ontology that implies that the class cannot have any instances. In OWL, such a class would entail to **owl:nothing**.
2. An ontology is inconsistent if it is impossible to interpret the axioms in the ontology such that there is at least one class which has an instance.
3. An ontology is incoherent if it contains at least one unsatisfiable class.
4. The OWL2 Profiles are: OWL 2 EL, OWL 2 QL and OWL 2 RL. **OWL 2 EL** was designed for biomedical ontologies because it can handle very large ontologies and has very fast reasoning capabilities, both of which are essential when dealing with biomedical ontologies.
5. An entailed axiom directly follows from an ontology because it has been explicitly asserted in that ontology. Inferred axioms arise from the relationships among the classes. These two are the same because OWL automatically maintains the relationships between classes as you enter them.
6. i. The **satisfiable** classes are: A, B, C and D. The only **unsatisfiable** class is E

ii.	A SubClassOf D	entailed
	A SubClassOf B	entailed
	A SubClassOf B or D	entailed
	E SubClassOf E	entailed
	E SubClassOf B and not B	not entailed

#### Problem 4 (The Semantic Web)

```

1. PREFIX actor: <http://data.linkedmdb.org/resource/actor/>
PREFIX movie: <http://data.linkedmdb.org/resource/movie/>
PREFIX country: <http://data.linkedmdb.org/resource/country/>
PREFIX capital: <http://data.linkedmdb.org/resource/movie/country_capital/>
SELECT distinct ?all_capitals
FROM <http://xmlns.com/foaf/0.1/> #not used
{
  SERVICE <http://data.linkedmdb.org/sparql>
  {
    #replace Patrick Stewart with actor name
    ?all_actors movie:actor_name "Patrick_Stewart".
    ?all_movies movie:actor ?all_actors.
    ?all_movies movie:country ?all_countries.
    ?all_countries movie:country_capital ?all_capitals.
  }
}

```

- Assume that all actors have starred in movies and all movies have actors.
- Assume that a movie is shot in a particular country.
- Assume that all countries have capitals.

#### Problem 5 (Ontology Applications)

	Term	Domain
	aerobic respiration	Biological Process
	Epithelial Cell Differentiation	Biological Process
1.	Enzyme Binding	Molecular Function
	Synovial Bursa	Not Classified
	Mad-Cow Disease	Not Classified
	Endoplasmic Reticulum	Cellular Component

Synovial bursa and mad-cow disease are not classified in GO because they are not Gene Ontology terms. A GO term by definition is an initiative to unify the representation of a gene and gene product attribute across all species. Neither Synovial Bursa nor Mad-Cow fit this specification.

- The 5 resulting products of fruit morphogenesis are in the screenshot below:

AmiGO: Term Association

amigo.geneontology.org/cgi-bin/amigo/term-assoc.cgi?term=GO:0048530&session\_id=8418amigo1391057621

Download all association information in: [gene association format](#) [RDF/XML](#)

**Filter associations displayed**

Filter by Gene Product: Gene Product Type: ☐ All ☐ complex ☐ gene ☐ gene product Data source: ☐ All ☐ ASAP ☐ AspGD ☐ CGD Species: ☐ All ☐ Arabidopsis thaliana ☐ Aspergillus fumig... ☐ Aspergillus fumig... Filter by Association: Evidence Code: ☐ All ☐ IBA ☐ IKR ☐ IRD View associations: ☒ All ☐ Direct associations [Set filters](#) [Remove all filters](#)

**fruit morphogenesis ; GO:0048530** [\[show def\]](#) [\[view in tree\]](#)

Symbol, full name	Information	Qualifier	Evidence	Reference	Assigned by
<input type="checkbox"/> ABO1 ABA-OVERLY SENSITIVE 1	20 associations <a href="#">[BLAST]</a> protein from <i>Arabidopsis thaliana</i>		IMP	PMID:20836892	TAIR
<input type="checkbox"/> AN AT1G01510	23 associations <a href="#">[BLAST]</a> protein from <i>Arabidopsis thaliana</i>		IMP	PMID:19843316	UniProtKB (via TAIR)
<input type="checkbox"/> fas fasciated	1 association <a href="#">[BLAST]</a> gene from <i>Solanum lycopersicum</i>		IEP	PMID:18469814	SGN
<input type="checkbox"/> GOA AT1G31140	8 associations protein from <i>Arabidopsis thaliana</i>		IMP	PMID:20598091	TAIR
<input type="checkbox"/> RHA1 AT5G45710	17 associations <a href="#">[BLAST]</a> protein from <i>Arabidopsis thaliana</i>		IMP	PMID:18381353	TAIR

☐ [Select all](#) [Clear all](#) Perform an action with this page's selected gene products... [Go](#)

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AmiGO version: 1.8  
Try AmiGO Labs

GO database release 2014-01-25  
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And the terms associated with ABO1 are in the screenshots below

AmiGO: ABO1 Association

amigo.geneontology.org/cgi-bin/amigo/gp-assoc.cgi?gp=TAIRlocus:2173199&session\_id=8418amigo1391057621

Accession, Term	Ontology	Qualifier	Evidence	Reference	Assigned by
<input type="checkbox"/> GO:0080178 : 5-carbamoylmethyluridine metabolic process	2 gene products <a href="#">view in tree</a> biological process		IMP	PMID:20836892	TAIR
<input type="checkbox"/> GO:0008283 : cell proliferation	11716 gene products <a href="#">view in tree</a> biological process		IMP	PMID:15894610	TAIR
<input type="checkbox"/> GO:0071215 : cellular response to abscisic acid stimulus	281 gene products <a href="#">view in tree</a> biological process		IMP	PMID:16943431	UniProtKB (via TAIR)
<input type="checkbox"/> GO:0009560 : embryo sac egg cell differentiation	139 gene products <a href="#">view in tree</a> biological process		RCA	PMID:22589469	TAIR
<input type="checkbox"/> GO:0048530 : fruit morphogenesis	5 gene products <a href="#">view in tree</a> biological process		IMP	PMID:20836892	TAIR
<input type="checkbox"/> GO:0009965 : leaf morphogenesis	219 gene products <a href="#">view in tree</a> biological process		IMP	PMID:20836892	TAIR
<input type="checkbox"/> GO:0006312 : mitotic recombination	469 gene products <a href="#">view in tree</a> biological process		RCA	PMID:22589469	TAIR
<input type="checkbox"/> GO:0031538 : negative regulation of anthocyanin metabolic process	3 gene products <a href="#">view in tree</a> biological process		IMP	PMID:19500300	UniProtKB (via TAIR)
<input type="checkbox"/> GO:0035265 : organ growth	812 gene products <a href="#">view in tree</a> biological process		IMP	PMID:15894610	TAIR
<input type="checkbox"/> GO:0008284 : positive regulation of cell proliferation	5001 gene products <a href="#">view in tree</a> biological process		IMP	PMID:19500300	UniProtKB (via TAIR)
<input type="checkbox"/> GO:0009787 : regulation of abscisic acid-activated signaling pathway	62 gene products <a href="#">view in tree</a> biological process		IMP	PMID:16943431	UniProtKB (via TAIR)
<input type="checkbox"/> GO:0010928 : regulation of auxin mediated signaling pathway	44 gene products <a href="#">view in tree</a> biological process		IMP	PMID:20080602	UniProtKB (via TAIR)
<input type="checkbox"/> GO:0032784 : regulation of DNA-templated transcription, elongation	642 gene products <a href="#">view in tree</a> biological process		ISS With	TAIR:Communication:501714663	TIGR (via TAIR)

GO Term	Gene Products	Biological Process	Molecular Function	PMID	Database
GO:0008284 : <a href="#">positive regulation of cell proliferation</a>	<a href="#">5001 gene products</a> <a href="#">view in tree</a>	<a href="#">biological process</a>	<a href="#">IMP</a>	<a href="#">PMID:19500300</a>	UniProtKB (via TAIR)
GO:0009787 : <a href="#">regulation of abscisic acid-activated signaling pathway</a>	<a href="#">62 gene products</a> <a href="#">view in tree</a>	<a href="#">biological process</a>	<a href="#">IMP</a>	<a href="#">PMID:16943431</a>	UniProtKB (via TAIR)
GO:0010928 : <a href="#">regulation of auxin mediated signaling pathway</a>	<a href="#">44 gene products</a> <a href="#">view in tree</a>	<a href="#">biological process</a>	<a href="#">IMP</a>	<a href="#">PMID:20080602</a>	UniProtKB (via TAIR)
GO:0032784 : <a href="#">regulation of DNA-templated transcription, elongation</a>	<a href="#">642 gene products</a> <a href="#">view in tree</a>	<a href="#">biological process</a>	<a href="#">ISS With Pfam:PF04762</a>	<a href="#">TAIR:Communication:501714663</a>	TIGR (via TAIR)
GO:2000024 : <a href="#">regulation of leaf development</a>	<a href="#">20 gene products</a> <a href="#">view in tree</a>	<a href="#">biological process</a>	<a href="#">IMP</a>	<a href="#">PMID:19500300</a>	UniProtKB (via TAIR)
GO:0009737 : <a href="#">response to abscisic acid</a>	<a href="#">670 gene products</a> <a href="#">view in tree</a>	<a href="#">biological process</a>	<a href="#">IMP</a>	<a href="#">PMID:19500300</a>	UniProtKB (via TAIR)
GO:0006979 : <a href="#">response to oxidative stress</a>	<a href="#">5105 gene products</a> <a href="#">view in tree</a>	<a href="#">biological process</a>	<a href="#">IMP</a>	<a href="#">PMID:19500300</a>	UniProtKB (via TAIR)
GO:0006400 : <a href="#">tRNA modification</a>	<a href="#">1739 gene products</a> <a href="#">view in tree</a>	<a href="#">biological process</a>	<a href="#">IMP</a>	<a href="#">PMID:20836892</a>	TAIR
GO:0005829 : <a href="#">cytosol</a>	<a href="#">30016 gene products</a> <a href="#">view in tree</a>	<a href="#">cellular component</a>	<a href="#">IDA</a>	<a href="#">PMID:21166475</a>	TAIR
GO:0033588 : <a href="#">Elongator holoenzyme complex</a>	<a href="#">119 gene products</a> <a href="#">view in tree</a>	<a href="#">cellular component</a>	<a href="#">IDA</a>	<a href="#">PMID:20080602</a>	UniProtKB (via TAIR)
GO:0005634 : <a href="#">nucleus</a>	<a href="#">83664 gene products</a> <a href="#">view in tree</a>	<a href="#">cellular component</a>	<a href="#">ISM</a>	<a href="#">TAIR:AnalysisReference:501750651</a>	TAIR

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3. The Classification of interleukin-1 is in the diagram below:

- [GO:0005488 binding \[244065 gene products\]](#)
- [GO:0003674 molecular function \[577197 gene products\]](#)
- [GO:0005515 protein binding \[68359 gene products\]](#)
- [GO:0030545 receptor regulator activity \[257 gene products\]](#)
- [GO:0005102 receptor binding \[11404 gene products\]](#)
- [GO:0030547 receptor inhibitor activity \[73 gene products\]](#)
- [GO:0005126 cytokine receptor binding \[1781 gene products\]](#)
- [GO:0048019 receptor antagonist activity \[53 gene products\]](#)
- [GO:0005152 interleukin-1 receptor antagonist activity \[15 gene products\]](#)
    - [GO:0045352 interleukin-1 Type I receptor antagonist activity \[5 gene products\]](#)
    - [GO:0045353 interleukin-1 Type II receptor antagonist activity \[5 gene products\]](#)

Blocks the binding of interleukin-1 to the interleukin-1 receptor complex.