Anatomical Ontologies

How of them many do we need?

Olivier Bodenreider
National Institutes of Health, Bethesda, Maryland, USA
Anatomical ontologies
Coordinated evolution of ontologies

<table>
<thead>
<tr>
<th>RELATION TO TIME</th>
<th>CONTINUANT</th>
<th>OCCURRENT</th>
</tr>
</thead>
<tbody>
<tr>
<td>GRANULARITY</td>
<td>INDEPENDENT</td>
<td>DEPENDENT</td>
</tr>
<tr>
<td>ORGAN AND ORGANISM</td>
<td>Organism (NCBI Taxonomy)</td>
<td>Anatomical Entity (FMA, CARO)</td>
</tr>
<tr>
<td>CELL AND CELLULAR COMPONENT</td>
<td>Cell (CL)</td>
<td>Cellular Component (FMA, GO)</td>
</tr>
<tr>
<td>MOLECULE</td>
<td>Molecule (ChEBI, SO, RnaO, PrO)</td>
<td>Molecular Function (GO)</td>
</tr>
</tbody>
</table>

Open Biomedical Ontologies (OBO) Foundry (ca. 2004)
(Gene Ontology in yellow)
Gene Ontology

- Cellular component hierarchy
- Cellular and subcellular level
- Supports the annotation of gene products in model organisms
- ~4200 classes
- Developed by the GO Consortium for over 20 years
- Public funding from NIH

http://amigo.geneontology.org/
SNOMED CT

- Largest clinical terminology in the world
- Developed by a consortium of over 40 countries
- Used for clinical documentation and analytics
- ~39,000 concepts
- Somewhat similar to FMA*
Uberon – Cross-species ontology

- 15,000 classes
- Species-neutral presentation
- Links to species-centric anatomical ontologies
- Supports integration of model organism and human data

http://uberon.github.io/
Others

• General
  • Medical Subject Headings (MeSH) – “A” tree
  • NCI Thesaurus
  • GALEN
  • [...]

• Specific species
  • Adult Mouse Anatomy (MGI)
  • Zebrafish Anatomy ontology
  • [...]

Anatomy [A]
  Body Regions [A01]
  Musculoskeletal System [A02]
  Digestive System [A03]
  Respiratory System [A04]
  Urogenital System [A05]
  Endocrine System [A06]
  Cardiovascular System [A07]
  Nervous System [A08]
  Sense Organs [A09]
  Tissues [A10]
  Cells [A11]
  Fluids and Secretions [A12]
  Animal Structures [A13]
  Stomatognathic System [A14]
  Hemic and Immune Systems [A15]
  Embryonic Structures [A16]
  Integumentary System [A17]
  Plant Structures [A18]
  Fungal Structures [A19]
  Bacterial Structures [A20]
  Viral Structures [A21]
How many do we need?
Selection criteria for anatomical ontologies

- Human vs. other organisms
- Research vs. clinical
- Gross vs. cellular/subcellular

And...
- Who maintains it?
- Regular updates?
- Intellectual property restrictions?
- Cross-references to other ontologies?

Define use cases first!
If you use more than one

• Terminology integration
  • Unified Medical Language System (UMLS)
    • Integrates FMA, MeSH anatomy, SNOMED CT anatomy, GO Cellular Location
  • BioPortal

• Lexical similarity vs. semantics
  • Prostate in human and mouse: same or different?
  • Different organs with similar functions across species (Uberon)
If you develop a new one

- Ontology development is hard
  - Reuse existing ontologies whenever possible
  - Add cross-references to facilitate integration
  - Partner with ontologists

What is the difference between an ontology and a car?