

May 9, 2019
Indiana University
Bloomington, Indiana



HuBMAP
Human BioMolecular Atlas Program

COMMON COORDINATE
FRAMEWORK WORKSHOP
CCFWS-01

Anatomical Ontologies

How of them many do we need?

Olivier Bodenreider

National Institutes of Health, Bethesda, Maryland, USA



U.S. National Library of Medicine



Anatomical ontologies



Coordinated evolution of ontologies

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)	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)	

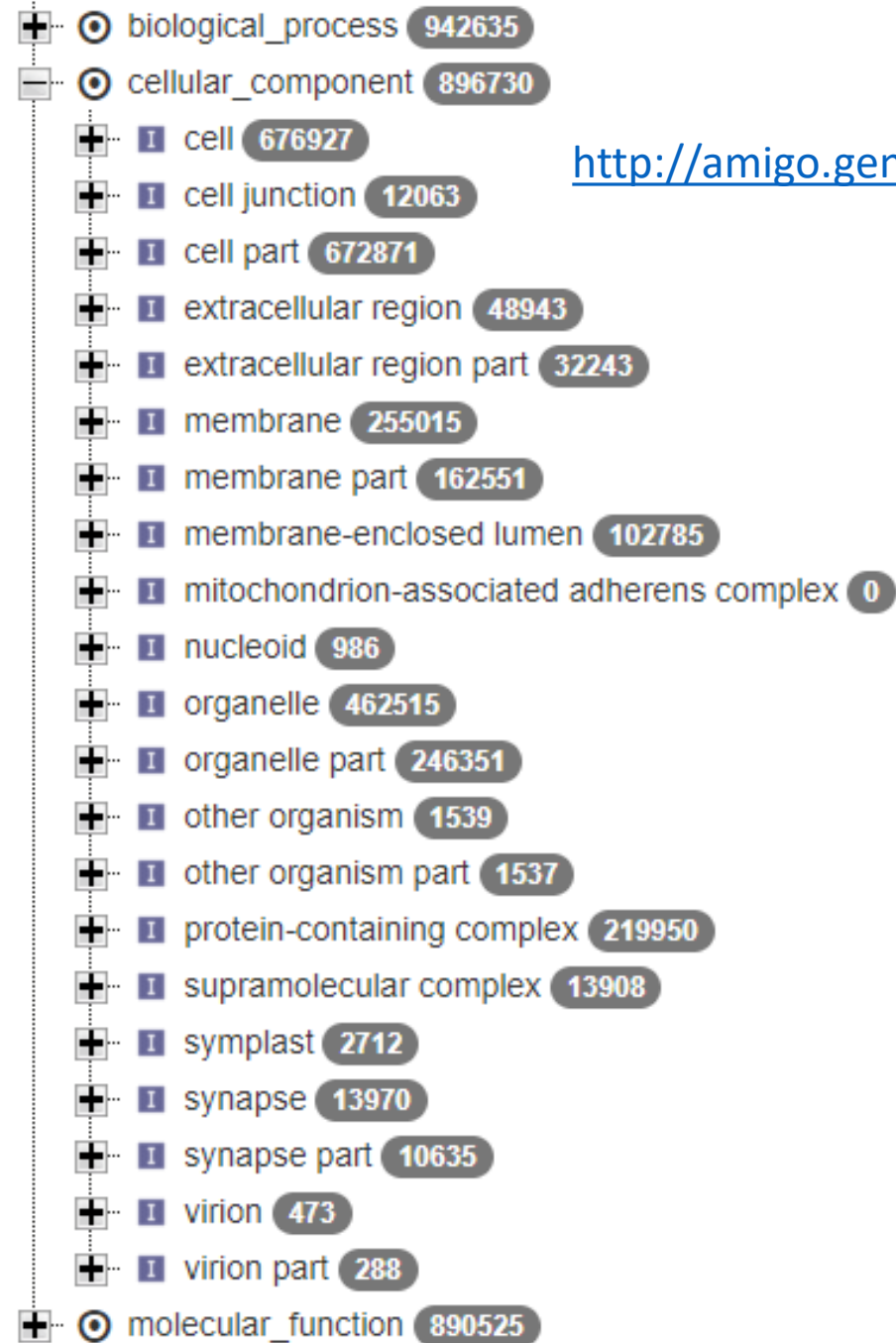
Open Biomedical Ontologies (OBO) Foundry (ca. 2004)
(Gene Ontology in yellow)



Borrowed from Barry Smith

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

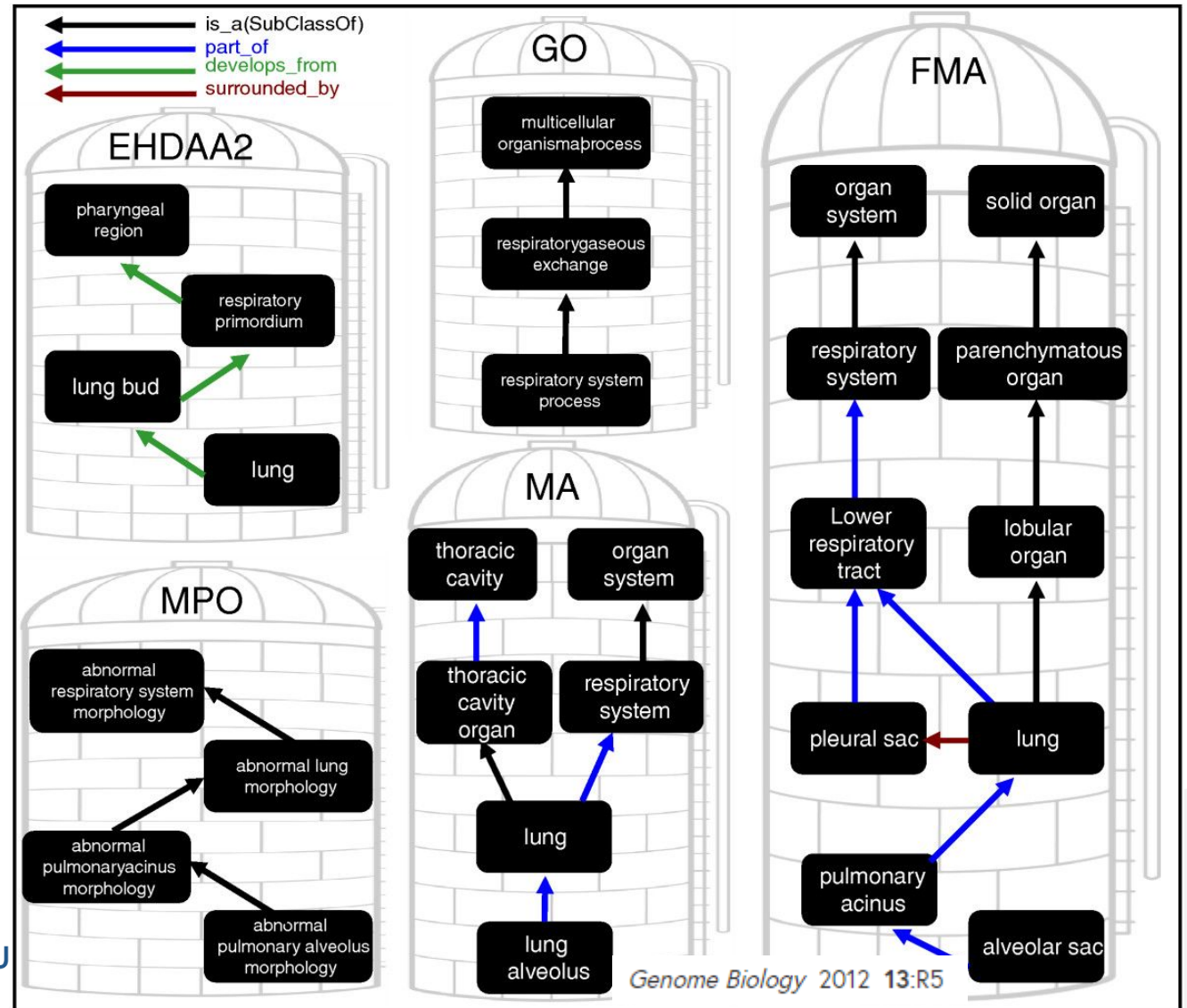
- Largest clinical terminology in the world
- Developed by a consortium of over 40 countries
- Used for clinical documentation and analytics
- ~39,000 concepts
- Somewhat similar to FMA*

The screenshot displays the SNOMED CT browser interface. At the top, a box labeled "Parents" contains a single concept: "Anatomical or acquired body structure (body structure)". Below this, a blue box highlights the selected concept: "Anatomical structure (body structure)" with a star icon and a download icon. It includes the SCTID: 91723000 and a list of languages: "91723000 | Anatomical structure (body structure) | en Anatomical structure en Anatomical structure (body structure)". To the right of this box is a light blue box labeled "No attributes". Below the highlighted box is a list of "Children (17)" concepts, each preceded by a right-pointing arrow and a yellow circle. The children listed are: Anatomical junction (body structure), Anatomical space structure (body structure), Body organ structure (body structure), Body region structure (body structure), Body system structure (body structure), Body tissue structure (body structure), Body wall structure (body structure), Cell structure (cell structure), Developmental body structure (body structure), Entire physical anatomical entity (body structure), Gland structure (body structure), and Intercellular anatomical structure (body structure). A vertical scrollbar is visible on the right side of the children list. At the bottom right, a URL is provided: <https://browser.ihtsdotools.org/>

Uberon – Cross-species ontology

<http://uberon.github.io/>

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



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?



olivier.bodenreider@nih.gov



U.S. National Library of Medicine