Anna Maria Masci Background

Immunology wet lab experience for more than 15 years

 Working in Biomedical ontology since 2008
 Working with Gene Ontology, Cell Ontology, Protein Ontology, Infectious Disease Ontology, Ontology for Biomedical Investigators Since 2008







Ontology as a branch of philosophy is the science of what is, of the kinds and structures of the objects, properties and relations in every area of reality.

From onto-logos, the science of being.



Confusion with the word ontology

Controlled Vocabulary:

Is a restricted list of words used for labeling, indexing or categorizing

Taxonomy:

Is a classification system. The taxonomy in a controlled vocabulary indicates a hierarchical structure based on an IsA relationship

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Ontology:

Is a controlled vocabulary with a taxonor complex than complex than just IsA relati/





ONTOLOGY

List of terms in a specific domain that have different type of relationships in addition to IsA relation and have textual and logical definitions.



What is an ontology (in Computer Science)

- Provides a schema for organizing data
- Data annotation (e.g. NLP) and reusability
- Underlying concept for semantic web (World Wide Web Consortium Web 3C)
- Allows semantic reasoning by computer systems (Computable)
- Results in improved web searchers (e.g. Google, Microsoft, Facebook)

In biomedical research used for: classification of data, representation of standards, enhanced searches



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Ontology Key Points

- Ontology provides unique identifiers for classes and relations that represent a phenomena within a specific domain.
- Ontology provides labels for classes and relations.
- Ontology provides metadata associated with classes and relations that allow human users to understand their meaning and contribute to consistent use in annotation and other applications.
- Ontology provides axioms and formal definitions that enable computational access to some aspects of the meaning of the classes and relations.
- Combining the four main features of ontology facilitates semantic integration of heterogeneous multimodal data within and across domains and enable novel data mining methods that span traditional boundaries between domains and data types.



Biomedical Ontology

Scientific ontologies have special features Every term must be such that the developers of the ontology believe it to refer to some entity on the basis of the best current scientific evidence (Important role of instances that we can observe in the laboratory)



A methodology for building and testing ontologies applied thus far in the biomedical domain on:

- FMA
- FuGO
- GO + other OBO Ontologies
- HL7-RIM
- ICF (International Classification of Functioning, Disability and Health)
- ISO Terminology Standards
- NCI Thesaurus
- SNOMED
- UMLS Semantic Network



Upper Level ontologies are ontologies that consist of very general terms common across all domains. Application ontologies are ontologies that consist of very specific term in a particular domain

Formal ontologies

<u>Upper ontology</u> <u>BFO – Basic Formal Ontology</u> <u>BORO – Business Objects Reference Ontology</u> <u>CIDOC Conceptual Reference Model</u> <u>Cyc (Cyc is not just an upper ontology, it also contains</u> <u>many mid-level and specialized ontologies as well)</u> <u>UMBEL – Upper Mapping and Binding Exchange Layer, a</u> <u>subset of OpenCyc</u> <u>DOLCE – Descriptive Ontology for Linguistic and Cognitive</u> <u>Engineering</u> <u>GFO – General Formal Ontology</u> <u>SUMO – Suggested Upper Merged Ontology</u>

Upper Level ontologies are ontologies that consist of very general terms common across all domains. Application ontologies are ontologies that consist of very specific term in a particular domain



BFO BASIC FORMAL ONTOLOGY

The Basic Formal Ontology is a small, upper level ontology that is designed for use in supporting information retrieval, analysis and integration in scientific and other domains. BFO is a genuine upper ontology. Thus it does not contain physical, chemical, biological or other terms which would properly fall within the coverage domains of the special sciences. BFO is used by more than 250 ontology-driven endeavors throughout the world. <u>http://ifomis.uni-saarland.de/bfo/</u>



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BFO STRUCTURE





National Cancer Institute Thesaurus

Summary Classes Properties Notes Mappings Widgets

Ju	imp ⁻	Го:	Details Visu	ualization	Notes (0) Class Mappings (27)					
	(Dbjective Dbserver	Preferred Name	e ł	Histology					
	ė. (Decupation or Discipline	Synonyms	1	Microanatomy					
	¢.	Benavioral Sciences Psychology Biological Sciences Anatomical Sciences Biochemistry	Definitions	t i	The study of the structure of the cells and their arrangement to constitute tissues and, finally, the association among these to form organs. In pathology, the microscopic process of identifying normal and abnormal morphologic characteristics in tissues, by employing various cytochemical and immunocytochemical stains.					
		Biology	ID	ł	http://ncicb.nci.nih.gov/xml/owl/EVS/Thesaurus.owl#C16681					
		Histology	ALT_DEFINITIO	N T	The study of tissues and cells under a microscope.					
		Meurosciences	code	(C16681					
		Physiology Physiology	Contributing_Source		FDA					
	¢	Psychobiology Complementary and Alternative Medicine Alternative Medical System Chinese Meridian Theory Lifestyle and Disease Provention			The study of the structure of the cells and their arrangement to constitute tissues and, finally, the association among these to form organs. In pathology, the microscopic process of identifying normal and abnormal morphologic characteristics in tissues, by employing various cytochemical and immunocytochemical stains.					
		Mind-Body System	FULL_SYN	I	Microanatomy					
	Þ	Health Sciences	label	ŀ	Histology					
		Clinical Sciences	Legacy_Concep	ot_Name H	Histology					
		Dentistry Environmental Health Sciences	Preferred_Nam	e I	Histology					
		Nursing	prefixIRI	(C16681					
	Þ	Medical Science	Semantic_Type		Biomedical Occupation or Discipline					
		Cancer Science Immunooncology	UMLS_CUI	(C0019638					
	ė.	Medicine Occupation	subClassOf	E	Biological Sciences					
		Allied Health Profession								

Medical Occupation



Ontology for Biomedical Investigations

Summary Classes Properties Notes Mappings Widgets

ump 1	To:		Details Visualization	Notes (0) Class Mappings (33)
		animal feeding assay	Preferred Name	histology
		3D structure determination assay activated partial thromboplastin time (aPTT) assay	Synonyms	histopathology
		applification of intermethylated sites (AIMS) assay	Definitions	the visual examination of cells or tissue (or images of them) with an assessment regarding a quality of the cells or tissue. Parts are: staining, imaging, judgement
		array based nucleic acid structure mapping assay	ID	http://purl.obolibrary.org/obo/OBI_0600020
		 assay detecting IFN-gamma production assay for transposase-accessible chromatin using se bacterial and burded 	achieves_planned_objective	assay objective
		Bernoulli trial	alternative term	histopathology
		 binding assay comet assay conv number variation profiling 	curator note	need to incorporate partsn\nThis is a very vague term, it should be in the same place as transcriptomics, proteomics metaboloimcs. It is the 'study' of tissues, not the process of studying tissues\n
		- cytochalasin-induced inhibition of actin polymerizatic		PRS:20090901: addition of alternative term = histopathology
		er cytometry assay er detection of molecular label	definition source	OBI branch derived
		DNA replication timing by array assay DNA sequence feature detection	editor preferred label	histology
		DNA sequence variation detection efficacy of epitope intervention experiment	example of usage	the counting of the number of cells with fluorescent label at their surface to determine the percentage of the population which was activated
		electrophoretic mobility shift assay	has curation status	http://purl.obolibrary.org/obo/IAO_0000125
		epigenetic modification assay		staining
		far-Western blot fluorescence detection assay	1	imaging assay
		gene dosage assay	has part	collecting specimen from organism
		gene knock-down assay glucose tolerance test		histological sample preparation
		handedness assay	label	histology
		histology	prefixIRI	OBI:0600020
		imaging assay	prefLabel	histology
		immune epitope assay immune response assay	term editor	PERSON:Compiled by Helen Parkinson for Transcriptomics thanks to Adam Witney
		- in container assay		the visual examination of cells or tissue (or images of them) with an assessment regarding a quality of the cells or tissue. Parts are: staining,
		in live cell assay in live organism assay	textual definition	imaging, judgement
		in situ hybridization in vivo intervention experiment	subClassOf	assay
		infectious agent detection assay		
		intra cellular electrophysiology reco		

Superclasses & Asserted Axioms

- <u>assay</u>
- has part some imaging assay
- has part some histological sample preparation
- has part some staining
- · has part some collecting specimen from organism
- <u>has specified_output</u> some (information content entity and (is about some (cell or organ section)))
- <u>achieves_planned_objective</u> some assay objective

Ontologies that use the Class



I erm Browse	r	EVS Enterprise Vocab	ulary Ser
Terminologies Value (Sets Mappings		
	1111	specimen	Search
NOUI			<u>_</u>
NCIT	esaurus	Back to search results	Advanced Se
Version: 17.06d (Release da	ate: 2017-06-26)	Hierarchy I Value Sats I Visited Concents	Auvanceu Se
	The second second		
		QUICK LINKS	
Specimen (Cod	0 010157)	View in Hierarchy View History View Graph Add to Cart Si	Suggest Char
Specimen (Cod	le C19157)		
Terms & Properties	Synonym Details Relationships Ma	appings View All	
Terms & Properti	ies		
Preferred Name: Specime	en		
Definition: A part of a thin	ng, or of several things, taken to demonstrate	or to determine the character of the whole, e.g. a substance, or portion of material ob	btained for u
in tooling, oxanination, or	orady, particularly, a proparation of toodo of	boary had taken for examination of diagnosic.	
Label: Specimon			
Label: Specimen			
Label: Specimen NCI Thesaurus Code: C1	19157 (Search for linked caDSR metadata) (search value sets)	
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Ontology for Biobanking									
Keywords:	Search terms								
Class: specimen	Class: specimen								
Term IRI: http://purl.obolibrary.org/obo/	OBI_0100051								
Definition: A material entity that has the specim	ien role.								
Annotations									
 editor note: Note: definition is in specimen creatinput during an investigation. definition editor: Philippe Rocca-Serra, James definition source: GROUP: OBI Biomaterial Br example of usage: Biobanking of blood taken a bas curation status: ready for release 	tion objective which is defined as an objective to obtain and store a material entity for potential use as an Malone anch and stored in a freezer for potential future investigations stores specimen.								

Equivalents

• material entity and (has role some specimen role)

Class Hierarchy

Thing + entity

+ continuant

- + independent continuant
 - + material entity
 - fiat object
 - + object aggregate
 - + object
 - + molecular entity
 - + <u>cell</u>
 - material information bearer
 - + collection packet
 - medication material
 - + processed material
 - + population

 - + organization
 - + material sample
 - + manufacturer - molecular-labeled material
 - + collection of specimens
 - target of material addition
 - disorder
 - post-office box

 - street
 - aggregate of organizations
 - more...
 - specimen
 - + material sample
 - + processed specimen
 - + specimen with known storage state
 - + specimen from organism
 - microdissected specimen
 - specimen meeting specification
 - normal specimen

Superclasses & Asserted Axioms

material entity



Examples of BFO-based ontology frameworks in other areas

UNEP Ontology Framework	United Nations Environment Programme
CIA Ontology Framework	Central Intelligence Agency
USGS National Map	United States Geological Survey
Joint Doctrine Ontologies	US Air Force Research Labs
Common Core Ontologies (CCO)	US Army / I2WD and ARL, IARPA, JIDO, ONR, AFRL
TRIP Ontologies	Federal Highway Administration (FHWA) Transportation Research Informatics Platform (TRIP)

Overview of Siri Technology





Ontology Development Process





Ontology Repositories for Life Sciences

Ontobee
 <u>http://www.ontobee.org/</u>



 Bioportal project <u>http://bioportal.bioontology.org/</u>



 Ontology Look up service <u>http://www.ebi.ac.uk/ols/index</u>



Best practices for developing ontologies @ OBO foundry: http://www.obofoundry.org/



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The OBO Foundry : a step-bystep, principles-based approach

- Developers commit in advance to collaborating with developers of ontologies in adjacent domains and
- To working to ensure that, for each domain, there is community convergence on a single ontology



OBO FOUNDRY PRINCIPLES

- COMMON GOVERNANCE
- COMMON TRAINING
- ROBUST VERSIONING
- COMMON ARCHITECTURE



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top level							
mid-level	Information Artifact Ontology (IAO)			ntology for Bior Investigation (OBI)	nedical ns	Onto M	ology of General edical Science (OGMS)
	Anato (FM	my Ontology A*, CARO) Cellular		Environment	Infectious Disease Ontology (IDO*) Phenotypic Quality		
domain level	Ontology (CL)	Component Ontology (FMA*, GO*	t ')	(EnvO)			Biological Process
	Subcellu	lar Anatomy (Onto	ology (SAO)	Ontolo (PaTC	ogy D)	Ontology (GO*)
		Sequence Ont (SO*)	tolo	gy	Molecu	ılar	
		Protein Onto (PRO*)	olog	y.	(GO	*)	

OBO Foundry Modular Organization



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Ontologies adopting the BFO structure

Domain

Ontology for Adverse Events (OAE)

Ontology for Biobanking (OBIB)

biomedical investigations

Ontology for Autism Spectrum Disorder

Ontology for Dengue Fever (IDODEN)

Ontology for Energy Investigations (OEI)

Ontology for Drug Discovery Investigations (DDI)

Ontology for Functionally Graded Materials (OFGM)

· Ontology for Genes and Genomes - Mouse (OGG-MM)

Ontology for Genetic Disease Investigations (OGDI)

Ontology for Genetic Susceptibility Factor (OGSF)

· Ontology for Guiding Appropriate Antibiotic Prescribing

Ontology for MIcroRNA Target Prediction (OMIT) (here)

Ontology for Newborn Screening and Translational Research (ONSTR)

Ontology for Next Generation Sequencing Experiments (NGS Ontology)

Ontology for Pain and Related Disability, Mental Health and Quality of Life (OPMQoL)

Ontology for Parasite LifeCycle (OPL): Reference ontology for parasite life cycle stages

Ontology-Driven Information System (ODIS)/Ontology-Driven Scenario Generator (ODSG)

· Proteomics data and process provenance ontology (ProPreO): bioinformatics for glycan expression, integrated

Protein Ontology (PRO): protein types and modifications classified on the basis of evolutionary relationships

Semantic Electronic Health Record (SEHR) Model from Linked2Safety (see also here and here)

Ontology for Laparoscopic Surgeries (LapOntoSPM)

Ontology for Microbial Phenotypes (OMP)

Ontology for Periodontitis (PERIO)

Ontology of Data Mining (OntoDM)

Ontology of Social Participation (OPS)

Ontology-Based Data Access (OBDA)

Ontology of Vaccine Adverse Events (OVAE)

Ontology-Based eXtensible Data Model (OBX)

Patient Safetry Categorial Structure (PS-CAST)

Plant Experimental Assay Ontology (PEAO)

Population and Community Ontology (PCO)

Protein-Ligand Interaction Ontology (PLIO)

technology resource for biomedical glycomics

Schistosomiasis Process Ontology (IDOSCHISTO)

Semanticscience Integrated Ontology (SIO)

Sleep Domain Ontology (SDO) (see also here)

Subcellular Anatomy Ontology (SAO) of NCMIR

Special Nuclear Materials Detection Ontology (SNM-DO)

Suggested Ontology for Pharmacogenomics (SO-Pharm)

Taxonomy for rehabilitation of knee conditions (TRAK)

Situation Awareness Ontology (SAO)

Skin Physiology Ontology (SPO)

Software Ontology (SWO)

Statistics Ontology (STATO)

Surface Water Ontology (SWO)

RNA Ontology (RnaO): RNA features, interactions and motifs

Scientific Evidence and Provenance Information Ontology (SEPIO)

· Senselab Ontology with applications to NeuronDB and BrainPharm

Sequence Ontology (SO): features and properties of nucleic sequences

SMART Protocols: SeMAntic RepresenTation for Experimental Protocols

Spatiotemporal Ontology for the Administrative Units of Switzerland (SONADUS)

Population Health Record (PopHR)

· Phenotypic Quality Ontology (PaTO): qualities of biomedical entities

Oral Health and Disease Ontology (OHD)

Parasite Experiment Ontology (PEO)

Petrochemical Ontology

Plant Ontology (PO)

Porifera Ontology (PORO)

Pre-Eclampsia Ontology (PEO)

Proper Name Ontology (PNO)

Quality of Service (QoS) Ontology

Role Ontology (ROLEO)

Saliva Ontology (SALO)

Ontology of Clinical Research (OCRe)

Ontology of Biobanking Administration (OMIABIS)

· Ontology for Rehabilitation (Traumatic Brain Injury)

Ontology of Datatypes (OntoDT) (see also here)

Ontology of Biological and Clinical Statistics (OBCS)

· Ontology of Experimental Variables and Values (OOEVV)

Ontology of Medically Related Social Entities (OMRSE)

Ontology for General Medical Science (OGMS)

· Ontology for Genes and Genomes (OGG)

Ontology for Genetic Interval (OGI)

OncoCL-KB: a Knowledgebase for Integration of Clinical and Molecular Cancer Data

· OntoForinfoScience: a Detailed Methodology for Construction of Ontologies and its Application in the Blood

Ontology for Biomedical Investigations (OBI): design, protocol, instrumentation, and analysis applied in

OntoAlign++: a Combined Strategy for Improving Ontologies Alignment

Ontologized Minimum Information About Blobank data Sharing (OMIABIS)

Time Event Ontology (TEO)

Translational Medicine Ontology (TMO)

(Microbial) Typing Ontology (TypOn)

Universal Core Semantic Laver

Vaccine Ontology (VO)

• Vital Sign Ontology (VSO)

Xenopus Anatomy Ontology (XAO)

Institutions, Groups and Projects

Brain Operation Database (BODB)

L3 Data Tactics Corporation (see also)

DOQS: Data-Oriented Quality Solutions

Darwin Core (DwC) (see also)

Elsevier Smart Content Strategy

Good Ontology Design (GoodOD)

Influenza Research Database (IRD)

INRIA Lorraine Research Unit

Language and Computing

Bought by Nuance

OntoCAT

OntoCOG

OntoMaton

openEHR

REMINE

Securboration

SMART Protocol:

Skeletome

also here)

disciplines

(see also here)

Open PHACTS

Extensible Neuroimaging Archive Toolkit (XNAT)

National Center for Multi-Source Information Fusion

Qualitative Spatiotemporal Reasoning Unit

Open Access Journals Integrated service System Project (GoOA)

· Praxis: EMR research on interoperability and query of clinical records

· University of Arkansas, Biomedical Informatics, Medical Center, Little Rock

University of Texas Southwestern Medical Center (see also here)

University of Washington - Structural Informatics Group (SIG)

Saitama University - Faculty of Liberal Arts (see also here)

Ontology, Logic and Technology Unit

· Referent Tracking Unit

Science Commons - Neurocommons

University Hospital Erlangen - Radiology

Virus Pathogen Resource (VIPR)

CTSAconnect

DSpace at NTNUA

Dumontier Lab

eNanoMapper

EuPathDB

HIGHFLEET

Tumour-Node-Metastasis Ontology (TNM-O)

Vaccination Informed Consent Ontology (VICO)

YAMATO (Yet Another More Advanced Top-Level Ontology)

· yOWL: ontology-driven knowledge base for yeast biologists

Applied Physics Laboratory (APL), Johns Hopkins University

AstraZeneca - Clinical Information Science (see also here)

Berkeley Bioinformatics Open-Source Projects (BBOP)

Air Force Research Laboratory, Rome, New York

Zebrafish Anatomical Ontology (ZAO): anatomical structures in D. rerio

Biomedical Knowledge Engineering Lab at Seoul National University (SNU BIKE)

· eagle-i Consortium: a biomedical research resource discovery network

· Kobe University, Graduate School of Medicine, Department of Sociomedical Informatics

· Medical University Graz - Institute of Medical Informatics, Statistics and Documentation

. Ontohow Research Group at the New York State Center of Excellence in Bioinformatics & Life Sciences

U.S. Geological Survey (USGS) - Center of Excellence for Geospatial Information Science (CEGIS)

University of Augsburg - Computer Science - Software Methodologies for Distributed Systems

. University of South Alabama - School of Computing - South Biomedical Informatics (SBI) Research Group (see

· VIVO: An interdisciplinary national network enabling collaboration and discovery between scientists across all

United Nations SDG (Sustainable Development Goals) Ontology Framework

BFO Basic Formal Ontology

Home GitHub Guidebook Publications FOL Users

Users

Below you find an alphabetic list of ontologies and institutions/groups using BFO. If you want to be added, just contact us

Ontologies

- · ACGT Master Ontology (ACGT MO): represent the domain of cancer research and management in a computationally tractable manner
- Algorithm-Implementation-Execution Ontology Design Pattern
- Animals in Context Ontology (ACO)
- Autism-DSM-ADI-R Ontology (ADAR) Alzheimer Disease Ontology (ADO)
- Adverse Event Reporting Ontology (AERO)
- AFO Foundational Ontology
- Actionable Intelligence Retrieval System (AIRS)
- Apollo Structured Vocabulary (Apollo-SV)
- Bacterial Clinical Infectious Diseases Ontology (BCIDO)
- Behavior Perspective Model (BPM)
- Bank Ontology
- Battle Management Ontology (BMO)
- Beta Cell Genomics Application Ontology (BCGO)
- BioAssay Ontology
- Bioinformatics Web Service Ontology (OBIWS) (see also here)
- Biological Collections Ontology (BCO)
- Biomedical Ethics Ontology
- · Biomedical Grid Terminology (BiomedGT, retired): open, collaboratively developed terminology for translational
- research Biomimetic Ontology
- BioTop: A Biomedical Top-Domain Ontology
- BIRNLex: controlled terminology for annotation of BIRN data sources
- Blood Ontology (BLO)
- Body Fluids Ontology (BFLO)
- Bone Dysplasia Ontology (BDO) (see also here)
- Cancer Cell Ontology (OncoCL)
- Cancer Chemoprevention Ontology (CanCo)
- Cardiovascular Disease Ontology (CVDO) (Winner of FOIS 2014 Ontology Competition)
- Cell Behavior Ontology (CBO)
- Cell Culture Ontology (CCONT)
- Cell Cycle Ontology: application ontology for the representation and integrated analysis of the cell cycle process · Cell Expression, Localization, Development and Anatomy Ontology (CELDA)
- Cell Line Ontology (CLO)
- · Cell Ontology: designed as a structured controlled vocabulary for cell types Cellular Microscopy Phenotype Ontology (CMPO)
- Chemical Analysis Ontology (CAO)
- . Chemical Entities of Biological Interest (ChEBI): freely available dictionary of molecular entities focused on .small. chemical compounds
- Chemical Information Ontology (CHEMINF)
- Chemical Methods Ontology (CHMO)
- CHRONIOUS Ontology Suite
- Cigarette Smoke Exposure Ontology (CSEO)
- Clusters of Orthologous Groups (COG) Analysis Ontology (CAO)
- Cognition-Guided Surgery (CGS) Ontology
- Cognitive Paradigm Ontology (COGPO)
- · Common Anatomy Reference Ontology (CARO): anatomical structures in all organisms Common Core Ontologies (CCO)
- Communication Standards Ontology (CSO) Comparative Data Analysis Ontology (CDAO)
- Computational Neuroscience Ontology (CNO)
- Computer Aided Engineering Modeling Language Ontology (CAEMLOnto)
- Computer-Based Patient Record Ontology (CPRO)
- Conceptual Model Ontology (CMO)
- Core Data Integration Model (CDIM)
- Coriell Cell Line Ontology
- CPR Ontology for the Institute of Medicine's (IOM) Computer-Based Patient Record Ontology
- Diabetes mellitus Diagnosis Ontology (DDO)
- Document Act Ontology (D-Acts)
- Drug Interaction Ontology (DIO): ontology-driven inferences of possible drug-drug Interactions
- Drug Ontology (DrOn)
- Drug-drug Interaction Evidence Ontology (DIDEO)
- Drug-drug Interaction Ontology (DINTO)
- Dynamic Earth Sciences Ontologies: Process and Event Ontologies
- Eagle-I Research Resource Ontology (ERO)
- Economics Ontology (EcO)

- Electrocardiography Ontology (ECG Email ontology
- · Emotion Ontology (EMO): an ontology to describe affective phenomena
- ENanoMapper (ENM)
- . Environment Ontology: an ontology that supports the annotation of the environment of any organism or biological sample
- Epidemiology Ontology (EPO)
- Epilepsy and Seizure Ontology (EPSO)
- Event-Based Functional Behavior Ontology (EFBO)
- Evolution Ontology (EO)
- Experimental Factor Ontology (EFO)
- (EXperimental ACTions) Biomedical Protocol Ontology (EXACT2)
- Exposé: An Ontology for Data Mining Experiments (see also here)
- Flybase Drosophila Anatomy Ontology (DAO)
- Fission Yeast Phenotype Ontology (FYPO)
- · Flower-Visiting Domain Ontology (FV), Known Flower-Visiting Group Domain Ontology (KFG), Flower-Visiting Behavior Application Ontology (FVB) and Observation-Date Application Ontology (OBD)
- Food Ontology (FoodOn)
- · Foundational Model of Anatomy (FMA): structure of the mammalian and in particular the human body (Further reading)
- Gastrointestinal Endoscopy Ontology (GIEO)
- · Gene Regulation Ontology (GRO)
- General Information Model (GIM)
- Genomic Epidemiology Ontology (GenEpiO)
- Genomic Feature and Variation Ontology (GFVO)
- · Gestalt: Federated Access to Cyber Observables for Detection of Targeted Attack

International Center for Food Ontology, Operability, Data and Semantics (IC-FOODS)

uc Milk: An ontology for the characterization of mammalian milks

Major histocompatibility complex (MHC) Restriction Ontology (MRO)

Minimum Information Model for Patient Safety (MIMPS)

Middle Laver Ontology for Clinical Care (MLOCC)

miRNAO: An Ontology Unfolding the Domain of microRNAs

Nanoparticle Ontology (NPO): Ontology for Cancer Nanotechnology Research

Neuroscience Information Framework Cell Ontology (NIFCELL)

Neuroscience Information Framework Dysfunction Ontology (NIFDYS)

Neuroscience Information Framework (NIF) Subcellular Ontology (NIFSUBCELL)

NMR-Instrument Component of Metabolomics Investigations Ontology

o uc_Eating: An ontology for the characterization of eating, drinking and otherwise consuming foods

· uc Sense: An ontology for the characterization of sensory perception, qualities, and attributes

· MIRO and IRbase: IT Tools for the Epidemiological Monitoring of Insecticide Resistance in Mosquito Disease

Neuroscience Information Framework Standard Ontology (NIFSTD): a collection of OWL modules covering

· Neural Electromagnetic Ontologies (NEMO): Ontology-based Tools for Representation and Integration of Event-

· uc FoodDrugInteractionOntology: An ontology for the characterization of interactions between foods and

o uc Processing: An ontology for the characterization of processing methods and the products they

- Health Data Ontology Trunk (HDOT)
- Hemocomponents and Hemoderivatives Ontology (HEMONTO)
- Host Pathogen Interactions Ontology (HPO)
- Human Experience Realist Ontology (HERO)
- Human Interaction Network Ontology (HINO)
- . Human Physiology Simulation Ontology (HuPSON) (see also here)
- Infectious Disease Ontology

Interdisciplinary Prostate Ontology Project (IPOP)

- Information Artifact Ontology (IAO)
- Informed Consent Ontology (ICO)

Intracranial aneurysm (ICA) Ontology

Malaria Ontology (IDOMAL)

Materials Ontology (MatOnto)

Mental Functioning Ontology

Microbiology Ontology (MicrO)

Military Ontology (see publication)

Military Scenario Ontology (MSO)

Model for Clinical Information (MCI)

Mouse Pathology Ontology (MPATH)

Neomark Oral Cancer Ontology (NEOMARK4)

Neuroscience Information Framework (see also here)

distinct domains of biomedical reality

· Neuroscience Information Framework (NIF) Cell Ontology

Next Generation Sequencing Ontology (NGSONTO)

NeuroPsychological Testing Ontology (NPT)

Name Reaction Ontology (RXNO)

related Brain Potentials

New Upper Level Ontology

Non-Coding RNA Ontology (NCRO)

Mental Disease Ontology

Knowledge Base Of Biomedicine (KaBOB)

Interaction Network Ontology (INO)

drugs

Lipid Ontology

OWL: W3C Web Ontology Language

Key features of OWL is automatic reasoning (aka classification) with a reasoner using Description Logic (DL) which can perform several tasks including:

Subsumption, automatically organizes classes in an ontology into a hierarchy of sub/super-classes

Consistency, highlight inconsistent statements e.g. a given compound can't be organic and inorganic at the same time because these are disjoint

Querying, answering questions about an ontology (e.g. query languages like SPARQL and SPARQ-DL)



<!-- http://purl.obolibrary.org/obo/RO_0002201 -->

<owl:ObjectProperty rdf:about="http://purl.obolibrary.org/obo/RO_0002201">
 <rdfs:subPropertyOf rdf:resource="http://www.w3.org/2002/07/owl#topObjectProperty"/>
 <rdf:type rdf:resource="http://www.w3.org/2002/07/owl#FunctionalProperty"/>
 <obo:IAO_0000114 rdf:resource="http://purl.obolibrary.org/obo/IAO_0000125"/>
 <obo:IAO_0000115>inverse of has phenotype</obo:IAO_0000115>
 <obo:IAO_0000117 xml:lang="en">Chris Mungall</obo:IAO_0000117>
 <obo:IAO_0000117 xml:lang="en">Chris Mungall</obo:IAO_0000117>
 <oboInOwl:inSubset rdf:resource="http://purl.obolibrary.org/obo/RO_0002259"/>
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</owl:ObjectProperty>

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 <code:inverseOf rdf:resource="http://purl.obolibrary.org/obo/RO_0002203"/>
 <rdfs:domain rdf:resource="http://purl.obolibrary.org/obo/BFO_0000004"/>
 </rdfs:domain rdf:resource="http://purl.obolibrary.org/obo/BFO_000004"/>
 </p

evelops from y</obo:IAO_0000115>

<obo:IAO_0000117 xml:lang="en">Chris Mungall</obo:IAO_0000117> <obo:IAO_0000117 xml:lang="en">David Osumi-Sutherland</obo:IAO_0000117> <obo:IAO_0000117>Melissa Haendel</obo:IAO_0000117> <obo:IAO_0000117 xml:lang="en">Terry Meehan</obo:IAO_0000117> <rdfs:comment>This is the transitive form of the develops from relation</rdfs:comment> <rdfs:label xml:lang="en">develops from </rdfs:label>

</owl:ObjectProperty>

<!-- http://purl.obolibrary.org/obo/RO_0002203 -->

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 <rdfs:subPropertyOf rdf:resource="http://purl.obolibrary.org/obo/RO_0002388"/>
 </rdf:type rdf:resource="http://purl.obolibrary.org/obo/RO_0002388"/>
 <obo:IAO_0000114 rdf:resource="http://purl.obolibrary.org/obo/IAO_0000125"/>
 <obo:IAO_0000115>inverse of develops from</obo:IAO_0000115>
 <obo:IAO_0000117 xml:lang="en">Chris Mungall</obo:IAO_0000117>
 <obo:IAO_0000117 xml:lang="en">David Osumi-Sutherland</obo:IAO_0000117>
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 <obo:IAO_0000117</obo/IAO_0000259"/>
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 </obo/IAO_000259"/>



Same word can have different meanings

Same meaning can be express using different words



What is Blood?

Tissue

Fluid



What is wrong here?



Login Tools 🗹 Support 🕞

Ontology of Clinical Research

Summary Classes Properties Notes Mappings Widgets

Jump To:	Details Visual	ization N	otes (0)	Class Mappings (146)							
e Entity e Event	Preferred Name	Blood									
GeneriallyDependentContinuant Role Statistical concept	Definitions	A tissue wi plasma. Blo	A tissue with red blood cells, white blood cells, platelets, and other substances suspended in f plasma. Blood takes oxygen and nutrients to the tissues, and carries away wastes.								
Value set	ID	http://purl	.org/net/O	CRe/OCRe.owl#OCRE400034							
Biospecimen availability Biospecimen preservation method	definition	A tissue with red blood cells, white blood cells, platelets, and other substances suspended in fluid called plasma. Blood takes oxygen and nutrients to the tissues, and carries away wastes.									
- Blood	definition editor	Simona									
- Bodily fluid - Cells	definition source	NCI Glossary									
Tissue Boolean operator	label	Blood									
Comparison operator	prefixIRI	OCRE4000	34								
Domain term	prefLabel	Blood									
Pronty Recruitment status	subClassOf http://purl.org/net/OCRe/OCRe.owl#OCRE400080										
Semantic relation Study characteristic											
 Study status Telecommunication scheme 											
Time interval comparison operator											



https://bioportal hioontology org/sea

=√&query=blood&button=



Controlled vocabularies Particular concept is assign to a single term



What does it mean that Ontology has relationships?



Hierarchy







Example Lymph node Part_of conventional dendritic cell Part_of TLR4 Part_of TIR domain



ID	OBO_REL:part_of
Name	part_of
Relation properties	[transitive] [reflexive] [anti-symmetric]
inverse_of	OBO_REL:has_part (Instance level)
Definition	For continuants: C part_of C' if and only if: given any c that instantiates C at a time t, there is some c' such that c' instantiates C' at time t, and c *part_of* c' at t. For processes: P part_of P' if and only if: given any p that instantiates P at a time t, there is some p' such that p' instantiates P' at time t, and p *part_of* p' at t. (Here *part_of* is the instance-level part-relation.)
Comments	Parthood as a relation between instances: The primitive instance-level relation p part_of p1 is illustrated in assertions such as: this instance of rhodopsin mediated phototransduction part_of this instance of visual perception. This relation satisfies at least the following standard axioms of mereology: reflexivity (for all p, p part_of p); anti-symmetry (for all p, p1, if p part_of p1 and p1 part_of p then p and p1 are identical); and transitivity (for all p, p1, p2, if p part_of p1 and p1 part_of p2, then p part_of p2). Analogous axioms hold also for parthood as a relation between spatial regions. For parthood as a relation between continuants, these axioms need to be modified to take account of the incorporation of a temporal argument. Thus for example the axiom of transitivity for continuants will assert that if c part_of c1 at t and c1 part_of c2 at t, then also c part_of c2 at t. Parthood as a relation between classes: To define part_of as a relation between classes we again need to distinguish the two cases of continuants and processes, even though the explicit reference to instants of time now falls away. For continuants, we have C part_of C1 if and only if any instance of C at any time is an instance-level part of some instance of C1 at that time, as for example in: cell nucleus part_ of cell.

http://bioportal.bioontology.org/ontologies/OBOREL?p=properties



Which is the problem we are going to solve by using ontology



How to find your data? How to reason with data when you find it? How to understand the significance of the data you collected 3 years earlier? How to integrate with other people's data? Part of the solution must involve consensus based, standardized terminologies and coding schemes



http://ontology.buffalo.edu/08/TrainingCourse

How Ontology can help researcher







Ontology annotation



How to create annotation

Manual curation

Computer inference



SNCBI Resources 🖸	How To 🖂		
Publiced.gov US National Library of Medicine National Institutes of Health	PubMed 🛟	Advanced	earch

Abstract -

Send to: -

BMC Genomics. 2008 Feb 27;9:102. doi: 10.1186/1471-2164-9-102.

Protein abundance profiling of the Escherichia coli cytosol.

Ishihama Y¹, Schmidt T, Rappsilber J, Mann M, Hartl FU, Kerner MJ, Frishman D.

Author information

Abstract

BACKGROUND: Knowledge about the abundance of molecular components is an important prerequisite for building quantitative predictive models of cellular behavior. Proteins are central components of these models, since they carry out most of the fundamental processes in the cell. Thus far, protein concentrations have been difficult to measure on a large scale, but proteomic technologies have now advanced to a stage where this information becomes readily accessible.

RESULTS: Here, we describe an experimental scheme to maximize the coverage of proteins identified by mass spectrometry of a complex biological sample. Using a combination of LC-MS/MS approaches with protein and peptide fractionation steps we identified 1103 proteins from the cytosolic fraction of the Escherichia coli strain MC4100. A measure of abundance is presented for each of the identified proteins, based on the recently developed emPAI approach which takes into account the number of sequenced peptides per protein. The values of abundance are within a broad range and accurately reflect independently measured copy numbers per cell. As expected, the most abundant proteins were those involved in protein synthesis, most notably ribosomal proteins. Proteins involved in energy metabolism as well as those with binding function were also found in high copy number while proteins annotated with the terms metabolism, transcription, transport, and cellular organization were rare. The barrel-sandwich fold was found to be the structural fold with the highest abundance. Highly abundant proteins are predicted to be less prone to aggregation based on their length, pl values, and occurrence patterns of hydrophobic stretches. We also find that abundant proteins tend to be predominantly essential. Additionally we observe a significant correlation between protein and mRNA abundance in E. coli cells.

CONCLUSION: Abundance measurements for more than 1000 E. coli proteins presented in this work represent the most complete study of protein abundance in a bacterial cell so far. We show significant associations between the abundance of a protein and its properties and functions in the cell. In this way, we provide both data and novel insights into the role of protein concentration in this model organism.

PMID: 18304323 [PubMed - indexed for MEDLINE] PMCID: PMC2292177 Free PMC Article

Gene Product Associations

Free text filtering	Found entitie	es												
	Total: 24; showing 1-10 Results count 10 +													
4	14 14													
Your search is pinned to these filters														
 document_category: annotation 	Gene/produ	ct Gene/product Qualifie	r Direct annotation Annotation	Assigned by	Taxon	Evidence	Evidence with	PANTHER	Isoform	Reference	Date			
 bioentity: UniProtKB:P30850 	- donorprodu	name	extension	Assigned by	Tuxon	Endonioo		family	10010111	The for the for	Pato			
No current user filters.	🔲 rnb	ribonuclease	rRNA processing	GO_CENTRAL	Escherichia	IBA	PANTHER:PTN000599170	ribonuclease	PR:000023787	GO_REF:0000033	20150212			
Source	📄 mb	ribonuclease	exosome (RNase	GO_CENTRAL	Escherichia	IBA	PANTHER:PTN000599170	ribonuclease	PR:000023787	GO_REF:0000033	20150212			
Assigned by	🔲 rab	ll	complex)	GO CENTRAL	coli K-12 Escherichia	IBA		pthr23355 ribonuclease	PB-000023787	GO_REE-0000033	20150212			
Ontology (aspect)	II e		exoribonuclease activity		coli K-12		F		111.000020707	00_1121.0000000	23100272			
Evidence type	🔲 rnb	ribonuclease	RNA	GO_CENTRAL	Escherichia	IBA	PANTHER:PTN000599170	ribonuclease	PR:000023787	GO_REF:0000033	20150212			
PANTHER family		II	phosphodiester bond hydrolysis,		coli K-12			pthr23355						
Qualifier	🔲 rnb	ribonuclease	cytosol	EcoCyc	Escherichia	IDA		ribonuclease	PR:000023787	PMID:18304323	20140602			
Taxon	C. anh	II.	anatala biadhaa	1-14-1	coli K-12	101		pthr23355	DD-00000707	DUID-04504554	00110510			
Direct annotation	un u	ll	protein binding	IntAct	coli K-12	IPI	UNIPROTEB: PUA/W1	pthr23355	PR:000023787	PMID:24001004	20140512			
Inferred annotation	🔲 mb	ribonuclease II	protein binding	IntAct	Escherichia coli K-12	IPI	UniProtKB:P0A7W1	ribonuclease pthr23355	PR:000023787	PMID:15690043	20140512			
Annotation extension	🔲 rnb	ribonuclease II	hydrolase activity	UniProtKB	Escherichia coli K-12	IEA	SP_KW:KW-0378	ribonuclease pthr23355	PR:000023787	GO_REF:0000037	20150523			
	🔲 mb	ribonuclease II	RNA catabolic process	InterPro	Escherichia coli K-12	IEA	InterPro:IPR011804	ribonuclease pthr23355	PR:000023787	GO_REF:0000002	20150523			
	🔲 rnb	ribonuclease II	exonuclease activity	UniProtKB	Escherichia coli K-12	IEA	SP_KW:KW-0269	ribonuclease pthr23355	PR:000023787	GO_REF:0000037	20150523			



Evidence code

A simple controlled vocabulary used to record evidence that supports the association between the gene and the GO term

- Inferred from Experiment (EXP)
- Inferred from Direct Assay (IDA)
- Inferred from Physical Interaction (IPI)
- Inferred from Mutant Phenotype (IMP)
- Inferred from Genetic Interaction (IGI)
- Inferred from Expression Pattern (IEP)
- Inferred from Sequence or structural Similarity (ISS)
- Inferred from Sequence Orthology (ISO)
- Inferred from Sequence Alignment (ISA)
- Inferred from Sequence Model (ISM)
- Inferred from Genomic Context (IGC)
- Inferred from Biological aspect of Ancestor (IBA)
- Inferred from Biological aspect of Descendant (IBD)
- Inferred from Key Residues (IKR)
- Inferred from Rapid Divergence(IRD)
- Inferred from Reviewed Computational Analysis (RCA)
- Traceable Author Statement (TAS)
- Non-traceable Author Statement (NAS)
- Inferred by Curator (IC)
- No biological Data available (ND) evidence code
 Automatically-Assigned evidence code is:
 - Inferred from Electronic Annotation (IEA)



Why develop an Ontology?

- •To share common understanding of the entities in a specific domain
- •Among people
- •Among software
- •Among people and software



Legend for maps





Ontologies are legends for data

mbol	Ensembl ID									10.00				 					
002511.1	ENSG00000185897							10 10 00											
YAP1R1	ENSG0000078549								12.1										
DIPOR1	ENSG00000159346				d (1														
DIPOR2	ENSG0000008831								T										
ADORA1	ENSG00000163485				11 81	1.10	1 1 1	10 1	0.000				41	1000		10 10		1 11 1	
DORA2A	ENSG00000128271																1		
ORA2B	ENSG00000170425								100			1000				- 10			11
DORAS	ENSG00000121933																		
ADRA1A	ENSG00000120907																		
ADRA18	ENSG00000170214				1 11		1111												
DRA1D	ENSG00000171873			1 T			1.1		1 111										
DRA2A	ENSG00000150594														i II	110	1	1111	11
DRA2C	ENSG00000184160								60 M H	1 1					1111	111		110	Ш.,
ADRB1	ENSG0000043591		<u> - 1 - 1 - 1 - 1 - 1 - 1 - 1 - 1 - 1 - </u>				_			1 I I				 - 111		- 111		111	11
ADRB2	ENSG00000169252													 					
ADRB3	ENSG00000188778																		
AGTR1	ENSG00000144891																		
AGTR2	ENSG00000180772	A DESCRIPTION OF THE OWNER OF THE																	
ASL	ENSG00000128522																		
WPR1A	ENSG00000166148				1 1							10.07							
WPR1B	ENSG00000198049			and the second s															
AVPR2	ENSG00000126895	and the second of																	
BAI1	ENSG00000181790																		
BAI2	ENSG00000121753		A CONTRACTOR			110			1 81		21			10					
BAI3	ENSG00000135298																		111
DKRB1	ENSG00000100739											7 7 7							
DKRB2	ENSG00000168398																		
BRS3	ENSG00000102239	THE REPORT			1 1														
14orf100	ENSG0000050130																	6.4.4	



NCBO

• Dissemination and Ontology Best Practices of the National Center for Biomedical Ontology

• <u>http://bioontology.org</u>



Ontologies are modular Example: The Protein Ontology



Toll-Like Receptor Signaling in Vertebrates: Testing the Integration of Protein, Complex, and Pathway Data in the Protein Ontology Framework. Masci et al.2015



PRO



http://research.bioinformatics.udel.edu/pro/entry/PR%3A000001155/

http://pingeorgetown.edu/cgi-bin/pro/browser_pro?ids=PR:000001155

BIOSTATISTICS C BIOINFORMATICS <u>.ttp://pir.georgetown.edu/cgi-bin/ipcSFrid=PIRSF800008</u>

Sequence ontology









A strength of OBI is modeling the processes that connect biological source material to the data generated about it



Measurement of Glucose concentration in blood





Ontologies are Modular Example: The Cell Ontology



An improved ontological representation of dendritic cellsas a paradigm for all cell type. Masci et al. 2009



Ontologies are modular Example: The Liver Immunology Ontology



Bridging Multiple Ontologies: Representation of the Liver Immune Response. Masci et al. 2011



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NGS ontology Summary Classes Properties Notes Mappings Widgets

Jump To:	Details Visualization No	utes (0) Class Mappings (22) σ^{p}
Agent device	Preferred Name	DNA sequencer
entity 	Definitions	A DNA sequencer is an instrument that determines the order of deoxynucleotides in deoxyribonucleic acid sequences.nntnnhttp://purl.obolibrary.org/obo/OBL_0400103
enerically dependent continuant	ID	http://purl.obolibrary.org/obo/OBI_0400103
e data item	comment	A DNA sequencer is an instrument that determines the order of deoxynucleotides in deoxyribonucleic acid
		sequences.nntnnhttp://purl.obolibrary.org/obo/OBI_0400103
	Belong To	Agent
i∎ software	definition source	МО
specifically_dependent_continuant	editor preferred label	DNA sequencer
a quality	example of usage	ABI 377 DNA Sequencer, ABI 310 DNA Sequencer
Execution step	has curation status	http://purl.obolibrary.org/obo/IAO_0000122
NGS Workflow step	imported from	http://purl.obolibrary.org/obo/obi.owl
realizable_entity	label	DNA sequencer
function	prefixIRI	OBI:0400103
	prefLabel	DNA sequencer
material_entity	term editor	Trish Whetzel
DNA sequencer	textual definition	A DNA sequencer is an instrument that determines the order of deavunucleatides in deavuribanucleic acid
G Message	textual definition	sequences.
- contig	subClassOf	material_entity
DNA extract		
Library		
ial material sample ial read		
- object		
object_aggregate		
bject_boundary site		
spatial_region		
three_dimensional_region		
two_dimensional_region		
ian processual entity		
fiat_process_part		
□ process		
 Next Generation Sequencing Process Data processing 		
Filtering		
Sequence cutting		
DNA extraction		
DNA sequencing		
Sequence assembly		
Sequence assembly (de-novo assembly)		
Sequence assembly (mapping assembly)		

Example ontologies Basic Formal Ontology (BFO) http://ifomis.uni-saarland.de/bfo/ Common Anatomy Reference Ontology (CARO) http://www.bioontology.org/wiki/index.php/CARO:Main_Page Environment Ontology (EnvO) http://environmentontology.org/ Foundational Model of Anatomy (FMA) http://sig.biostr.washington.edu/projects/fm/ Gene Ontology (GO) http://geneontology.org/ Protein Ontology (PRO) http://pir.georgetown.edu/pro/ Cell Ontology (CL) http://cellontology.org/ Infectious Disease Ontology (IDO) http://infectiousdiseaseontology.org/page/Main_Page Ontology for Biomedical Investigations (OBI) http://obi-ontology.org/page/Main_Page Phenotypic Quality Ontology (PATO)



Relation Ontology (RO)

Ontologies and terminologies

SNOMED

http://www.ihtsdo.org/snomed-ct

Unified Medical Language System

http://www.nlm.nih.gov/research/umls/

National Cancer Institute Thesaurus

https://ncit.nci.nih.gov/ncitbrowser/

HL7 Reference Information Model

http://www.hl7.org/implement/standards/product_brief.cfm?product_id=78

International Classification of Diseaseas ICD http://www.who.int/classifications/icd/en/



Can we reasoning on this?



glioma ecosystem



GUTMANN DH., NEURO ONCOL 2015

• To enable reuse of data and information

• Avoid re-invention of the wheel

Introduce standards to allow interoperability and automatic reasoning



Helping comunication between researchers





annamaria.masci@duke.edu

