

The Center for Expanded Data Annotation and Retrieval: *Better Metadata Through Domain-Specific V*

Mark A. Musen, M.D., Ph.D.
Stanford University
musen@Stanford.EDU



Open
data
is about
MORE
THAN
DISCLOSURE
it must be
“Fair”

- Findable
- Accessible
- Interoperable
- Reusable

Open
data
is about
MORE
THAN
DISCLOSURE
it must be
Fair

- Findable
- Accessible
- Interoperable
- Reusable

Problem #1: Describe what exists in the world completely and consistently using ontologies



Gene Ontology

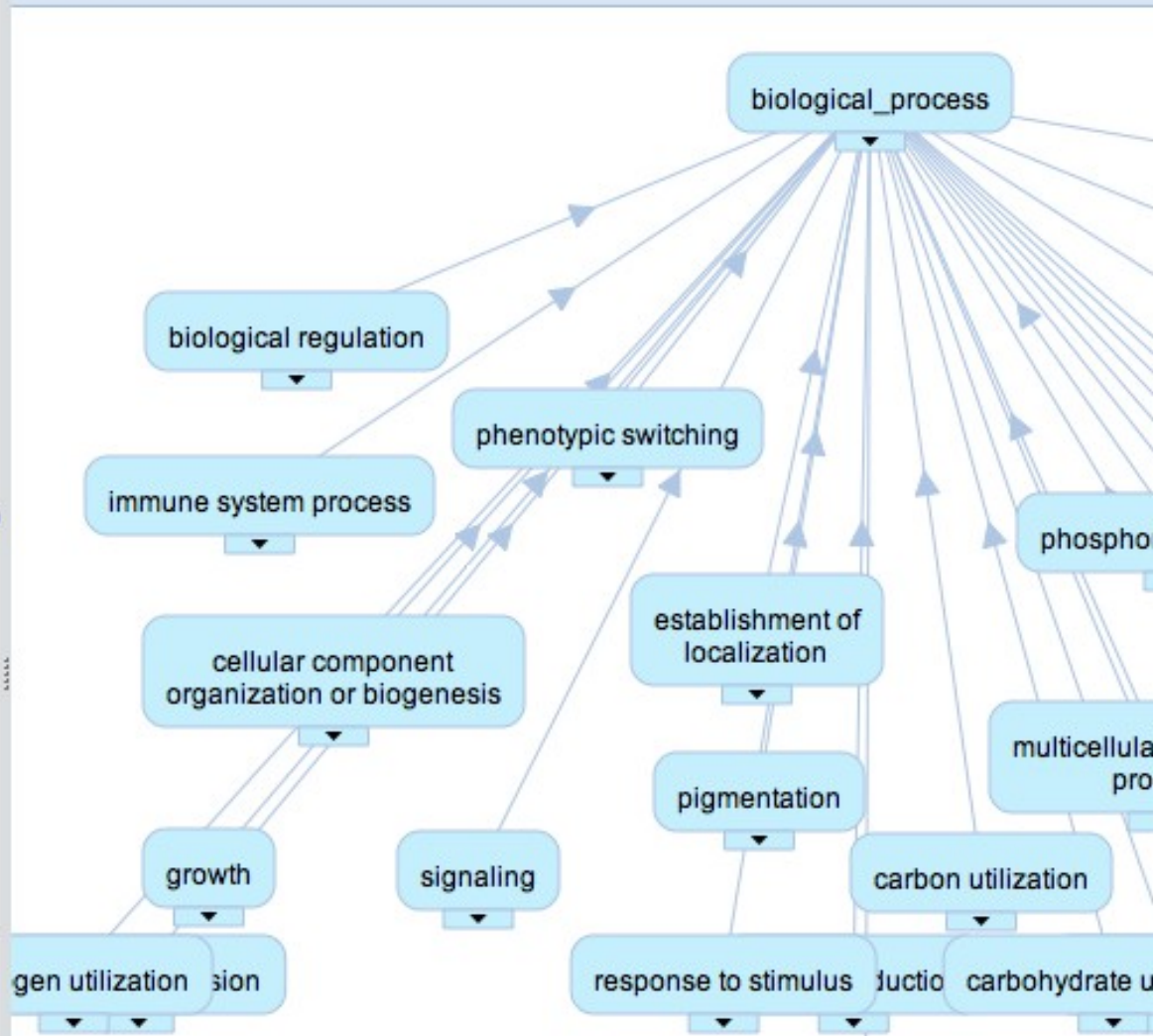
Terms ▾

Jump To:

Details Visualization Notes (0) Term Mappings (31) Term Resources

path to root ▾

- biological_process
- cellular_component
- molecular_function
 - antioxidant activity
 - binding
 - catalytic activity
 - channel regulator activity
 - chemoattractant activity
 - chemorepellent activity
 - electron carrier activity
 - enzyme regulator activity
 - metallochaperone activity
 - molecular transducer activity
 - morphogen activity
 - nucleic acid binding transcription factor activity
 - nutrient reservoir activity
 - protein binding transcription factor activity
 - protein tag
 - receptor activity
 - receptor regulator activity
 - structural molecule activity
 - translation regulator activity
 - transporter activity





SNOMED Clinical Terms

Terms ▾

Jump To:

Details

Visualization

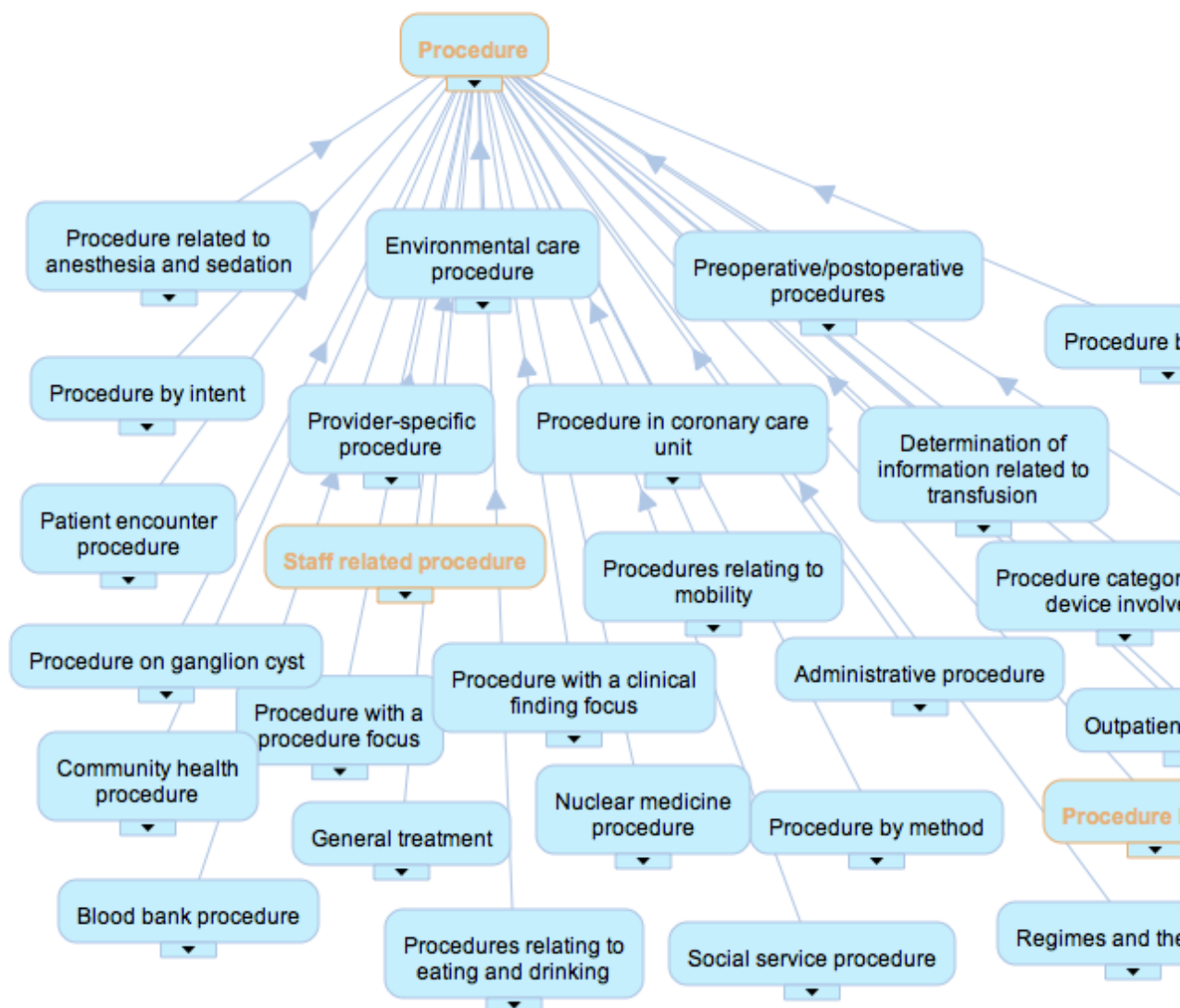
Notes (0)

Term Mappings (31)

Term Resources

path to root ▾

- ⊕ Body structure
- ⊕ Clinical finding
- ⊕ Environment or geographical location
- ⊕ Event
- ⊕ Linkage concept
- ⊕ Observable entity
- ⊕ Organism
- ⊕ Pharmaceutical / biologic product
- ⊕ Physical force
- ⊕ Physical object
- ⊖ **Procedure**
 - ⊕ Administrative procedure
 - ⊕ Blood bank procedure
 - ⊕ Community health procedure
 - ⊕ Determination of information related to transfusion
 - ⊕ Environmental care procedure
 - ⋮ General treatment
 - ⊕ Laboratory procedure
 - ⊕ Nuclear medicine procedure
 - ⊕ Obstetric procedure
 - ⋮ Outpatient procedure
 - ⊕ Patient encounter procedure
 - ⊕ Preoperative/postoperative procedures
 - ⊕ Procedure by intent
 - ⊕ Procedure by method
 - ⊕ Procedure by priority
 - ⊕ Procedure by site
 - ⊕ Procedure categorized by device involved
 - ⋮ Procedure in coronary care unit
 - ⊕ Procedure on ganglion cyst
 - ⊕ Procedure related to anesthesia and sedation
 - ⊕ Procedure related to breastfeeding
 - ⊕ Procedure with a clinical finding focus



Welcome to BioPortal, the world's most comprehensive repository of biomedical ontologies

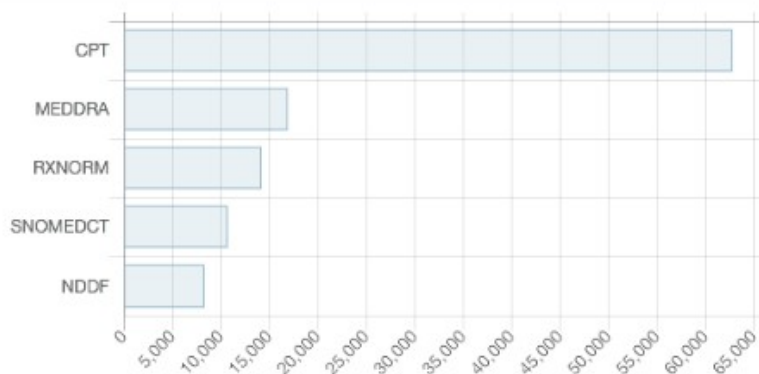
Search for a class


[Advanced Search](#)

Find an ontology


[Browse Ontologies](#)

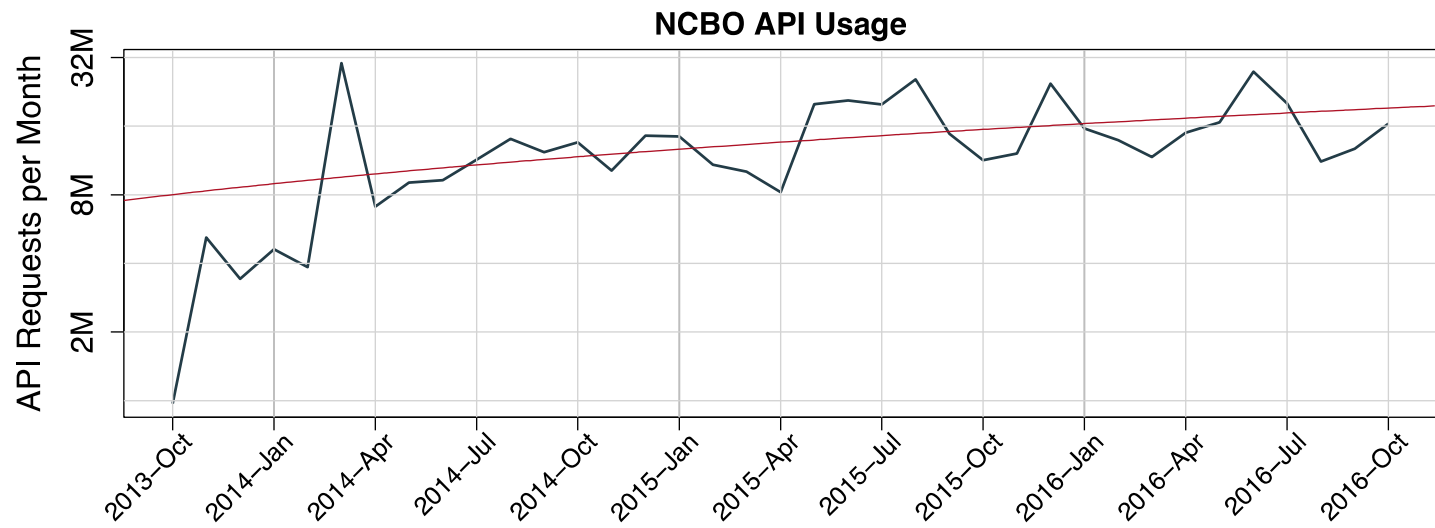
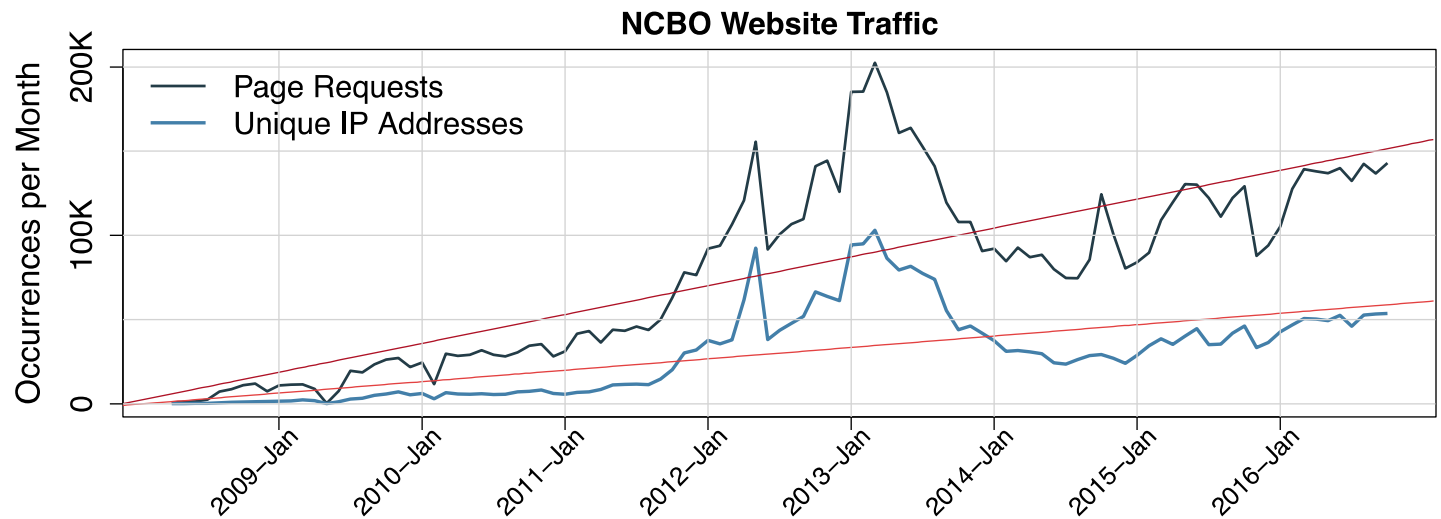
Ontology Visits (January 2017)


[More](#)

BioPortal Statistics

Ontologies	537
Classes	7,725,748
Resources Indexed	48
Indexed Records	39,537,360
Direct Annotations	95,468,433,792
Direct Plus Expanded Annotations	144,789,582,932

<http://bioportal.bioontology.org>



Who is using NCBO technology?



The collage includes logos from the following institutions and organizations:

- openMDR
- Labortree
- OpenClinica
- OASIS
- Technische Universität Braunschweig
- SilicoTryp
- ababase
- Integrated Rice Science Database
- SEEK
- Gene Wiki
- BRISQ
- University of Stavanger
- OntoFactory
- eurerca
- University of Leicester
- EMBL-EBI
- European Bioinformatics Institute
- isacreator
- GWAS CENTRAL
- Synapse
- ODISSea
- i2b2
- WIKI HOM Home
- Galaxy Tool Shed
- iPlant Collaborative
- MASSACHUSETTS GENERAL HOSPITAL
- BIRN
- BioScholar
- MGI
- CTSA
- Clinical & Translational Science Awards
- Translating Discoveries to Medical Practice
- CBiB
- BIO SITEMAPS EDITOR
- BUCK INSTITUTE
- FOR RESEARCH ON AGING
- BioLit
- RIKEN
- ontogator
- ele MAP
- caNanoLab
- Leibniz Institute of Plant Biochemistry
- IO
- MIND INFORMATICS
- MAYO CLINIC
- S3DB
- Kunnskapsegget
- sswap.info
- eagle-i
- SE-mantic
- M-medical
- I-mage
- A-notation
- RGXPress
- ontocat
- UTHealth
- The University of Texas
- RSNA Informatics
- RadLex
- RED FLY
- The DXBrain Project
- nbc
- Nutritional Phenotype Database
- Imperial College London
- Microsoft Research
- BioDAG Builder
- Ontology Add-in for Word
- gminer
- NMC Data Support Platform
- cancergrid
- biosharing
- Toxicogenomics CENTRE
- STANFORD ENGINEERING
- NuGO
- Nutrigenomics Organisation
- SIDR
- UCSF
- MEDICO RadSpeech
- RightField
- W3C
- CardioVascular Research Grid
- University of Bergen
- SCDE
- STEM CELL DISCOVERY ENGINE
- MG-RAST
- metagenomics analysis server

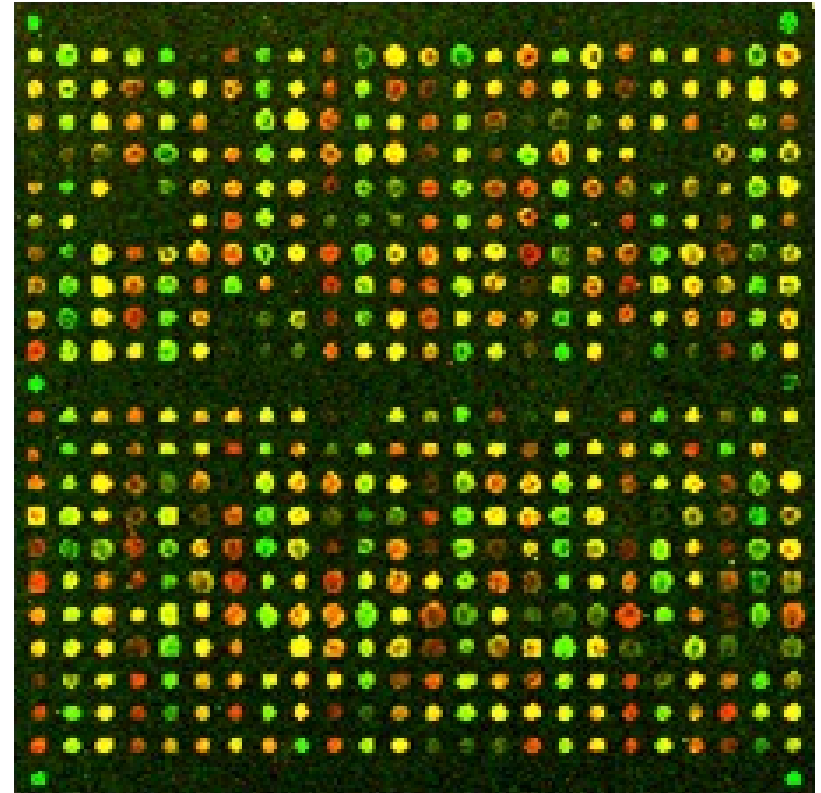
Open
data
is about
MORE
THAN
DISCLOSURE
it must be
Fair

- Findable
- Accessible
- Interoperable
- Reusable

Problem #2: Describe properties of experiments completely and consistently

The microarray community took the lead in standardizing metadata

- What was the substrate of the experiment?
- What array platform was used?
- What were the experimental conditions?



- DNA
Microarra

Minimum Information About a Microarray Experiment - MIAME

MIAME describes the **Minimum Information About a Microarray Experiment** that is needed to enable the interpretation of the results of the experiment unambiguously and potentially to reproduce the experiment. [[Brazma et al., Nature Genetics](#)]

The six most critical elements contributing towards MIAME are:

1. The raw data for each hybridisation (e.g., CEL or GPR files)
2. The final processed (normalised) data for the set of hybridisations in the experiment (study) (e.g., the gene expression data matrix used to draw the conclusions from the study)
3. The essential sample annotation including experimental factors and their values (e.g., compound and dose in a dose response experiment)
4. The experimental design including sample data relationships (e.g., which raw data file relates to which sample, which hybridisations are technical, which are biological replicates)
5. Sufficient annotation of the array (e.g., gene identifiers, genomic coordinates, probe oligonucleotide sequences or reference commercial array catalog number)
6. The essential laboratory and data processing protocols (e.g., what normalisation method has been used to obtain the final processed data)

For more details, see [MIAME 2.0](#).

[article](#)[discussion](#)[view source](#)[history](#)

MIBBI portal

- [Registration form](#) for the MIBBI Portal (please return to [chrisftaylor\[@\]gmail.com](mailto:chrisftaylor[@]gmail.com))
- [Summary spreadsheet](#) of all registered projects
- [XML document](#) containing all registered projects (from [this schema](#), same information as the Excel spreadsheet)

Bioscience projects registered with MIBBI

CIMR	Core Information for M etabolomics R eporting
GIATE	Guidelines for Information A bout T herapy E xperiments
MIABE	Minimal Information A bout a B ioactive E ntity
MIABIE	Minimum Information A bout a B iofilm E xperiment
MIACA	Minimal Information A bout a C ellular A ssay
MIAME	Minimum Information A bout a M icroarray E xperiment
MIAPA	Minimum Information A bout a P hylogenetic A nalysis
MIAPAR	Minimum Information A bout a P rotein A ffinity R eagent
MIAPE	Minimum Information A bout a P roteomics E xperiment
MIAPepAE	Minimum Information A bout a P eptid e A rray E xperiment
MIARE	Minimum Information A bout a R NAi E xperiment
MIASE	Minimum Information A bout a S imulation E xperiment
MIASPPE	Minimum Information A bout S ample P reparation for a P hosphoproteomics E xperiment
MIATA	Minimum Information A bout T Cell A ssays
MICEE	Minimum Information about a C ardiac E lectrophysiology E xperiment

Open
data
is about
MORE
THAN
DISCLOSURE
it must be
Fair

- Findable
- Accessible
- Interoperable
- Reusable

Problem #3: Make it palatable to
describe experiments
completely and consistently

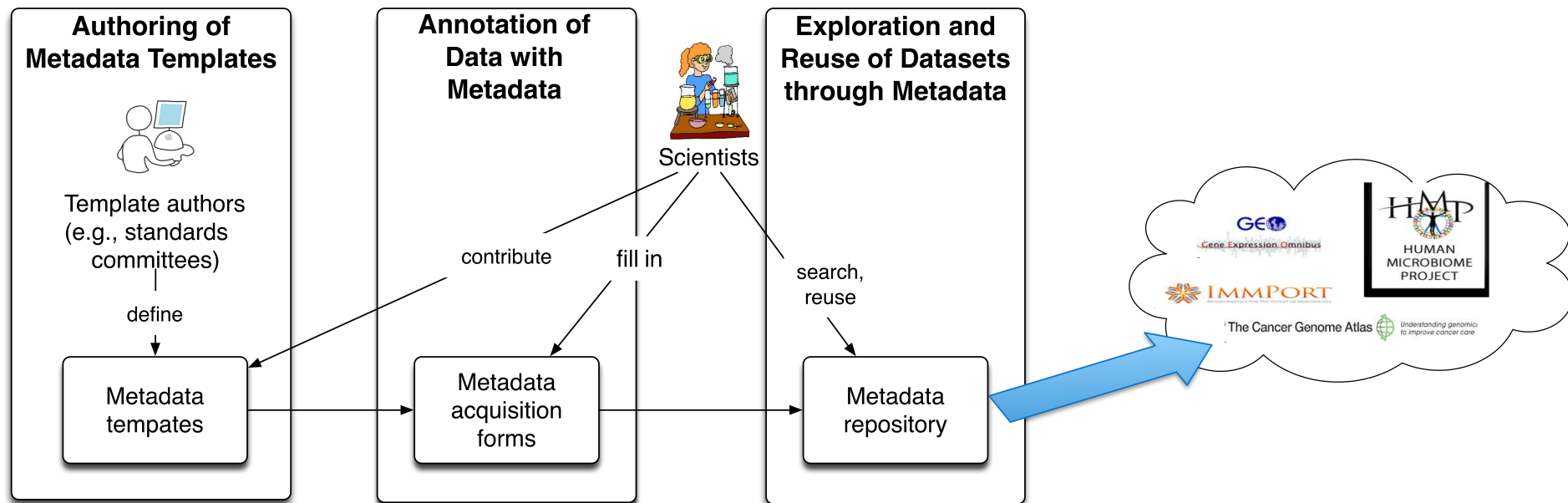
<http://metadatacenter.org>



CEDAR

CENTER FOR EXPANDED DATA ANNOTATION
AND RETRIEVAL

The CEDAR Approach to Metadata











FILTER

RESET

TYPE











	Title	Created	Modified
	BIBFRAME	11/5/16 1:37 PM	11/5/16 1:37 PM
	CDEs	8/21/16 10:22 AM	8/21/16 10:22 AM
	Sample templates	11/4/16 8:19 PM	11/4/16 8:19 PM
	BioSample Human	11/17/16 3:56 PM	11/17/16 3:56 PM
	ImmPort Study	5/2/16 12:38 PM	5/2/16 12:38 PM
	Investigation	5/7/16 2:03 PM	5/7/16 2:03 PM
	LINCS Antibody	10/12/16 3:42 PM	10/12/16 3:42 PM
	LINCS Cell Line	10/12/16 3:57 PM	10/12/16 3:57 PM

FILTER

RESET

TYPE



	Title	Created	Modified
	BIBFRAME	11/5/16 1:37 PM	11/5/16 1:37 PM
	CDEs	8/21/16 10:22 AM	8/21/16 10:22 AM
	Sample templates	11/4/16 8:19 PM	11/4/16 8:19 PM
	BioSample Human	11/17/16 3:56 PM	11/17/16 3:56 PM
	ImmPort Study	5/2/16 12:38 PM	5/2/16 12:38 PM
	Investigation	5/7/16 2:03 PM	5/7/16 2:03 PM
	LINCS Antibody	10/12/16 3:42 PM	10/12/16 3:42 PM
	LINCS Cell Line	10/12/16 3:57 PM	10/12/16 3:57 PM

Populate

Edit

Info

Share...

Move to...

Rename...

Make a copy

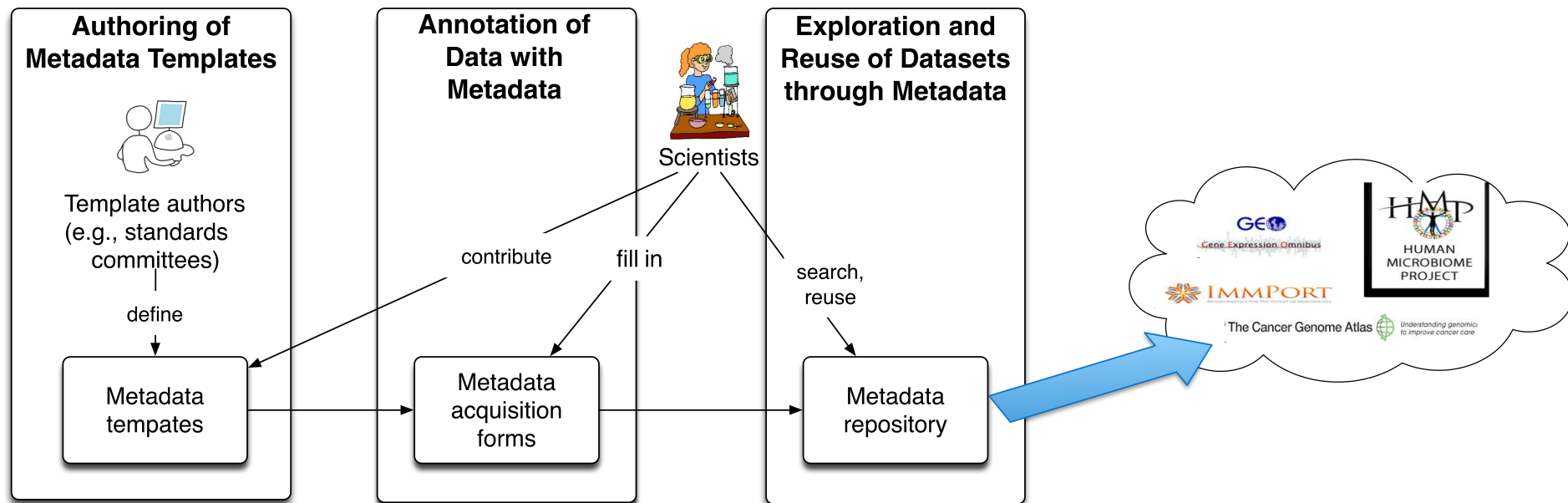
Delete






BioSample Human



Sample Name	056
Organism	Homo sapiens
Tissue	skin of body
Sex	male
Isolate	N/A
Age	74 years
Biomaterial Provider	Life Technologies
Optional Attribute (1)	
Name	disease
Value	dermatitis
Optional Attribute (2)	
Name	description
Value	Cell line was cultured until the 5th passage
Optional Attribute (3)	
Name	treatment
Value	350mg brodalumab


The CEDAR Approach to Metadata







 **a** TEXT * 



 **a** TEXT * 



 **a** TEXT * 

 FIELD TYPE
  **VALUES**
MULTIPLE
 REQUIRED
 SUGGESTIONS

Name	Type	Source	Identifier	No. Values
<div>SEARCH</div>				

a





☒







CLEAR

CANCEL

SAVE TEMPLATE

Find terms in BioPortal or [Create New Terms](#) to constrain the values of the 'Tissue' field

[Start Over](#)

Search in BioPortal

Tissue



500 results for the query 'Tissue'. Click on a term below to select it

TERM	DEFINITION	TYPE	SOURCE	ID
tissue	Multicellular anatomical structure that consists of many cells of one or a few types, arranged in an extracellular...	Class	UBERON	UBERON_0000479
tissue	-	Class	MA	MA_0003002
Tissue	-	Class	NIFSTD	birnlex_19
tissue	Anatomical structure, that consists of similar cells and intercellular matrix, aggregated according to genetically...	Class	TAO	CARO_0000043

Ontology: UBERON

- ⊖ Multicellular Organism
 - ⊖ Tissue
 - Mole
 - Roof Plate Of Metencephalon
 - ⊕ Macula
 - Intervillous Pockets
 - Purkinje Cell Layer Corpuscles
 - Mossy Fiber
 - Pars Basilaris
 - Dermis Of Feather Follicle
 - Upper Oral Valve
 - ⊕ Anlage
 - Anterior Lateral Plate Mesoderm
 - Molecular Layer Valvula Cerebri

TERM DETAILS		ONTOLOGY DETAILS
Name	tissue	
Id	http://purl.obolibrary.org/obo/UBERON_0000479	
Definition	Multicellular anatomical structure that consists of many cells of one or a few types, arranged in an extracellular matrix such that their long-range organisation is at least partly a repetition of their short-range organisation.	

TERM

BRANCH

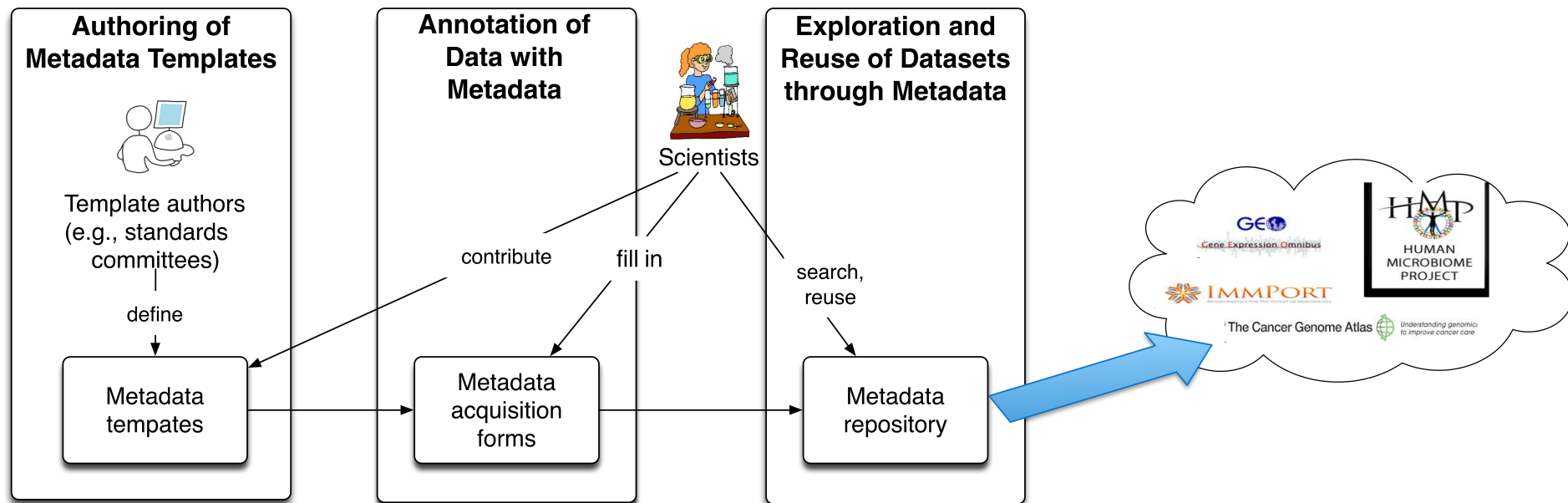
ONTOLOGY

Term Id	http://purl.obolibrary.org/obo/UBERON_0000479
Term Name	tissue

Click to add all the descendants of the selected term

ADD

The CEDAR Approach to Metadata



BioSample Human

- Sample Name
- Organism
- Tissue
- Sex
- Isolate
- Age
- Biomaterial Provider
- Optional Attribute
 - Name
 - Value

CANCEL

VALIDATE


SAVE


BioSample Human


- Sample Name
- Organism
- **Tissue**
- Sex
- Isolate
- Age
- Biomaterial Provider
- Optional Attribute
 - Name
 - Value


056


Homo sapiens





 blood (UBERON) (50%)


 liver (UBERON) (9%)

 bone marrow (UBERON) (6%)

 breast (UBERON) (6%)

 lymph node (UBERON) (6%)

 lung (UBERON) (5%)

 colon (UBERON) (5%)



BioSample Human

Sample Name	056
Organism	Homo sapiens
Tissue	brain
Sex	male
Isolate	N/A
Age	74 years
Biomaterial Provider	Life Technologies
Optional Attribute	



Name
Value

disease



- Parkinson's disease (DOID) (39%)
- central nervous system lymphoma (DOID) (27%)
- autistic disorder (DOID) (22%)
- melanoma (DOID) (5%)
- Edwards syndrome (DOID) (2%)
- schizophrenia (DOID) (1%)

BioSample Human

Sample Name	056
Organism	Homo sapiens
Tissue	lung
Sex	male
Isolate	N/A
Age	74 years
Biomaterial Provider	Life Technologies
Optional Attribute	

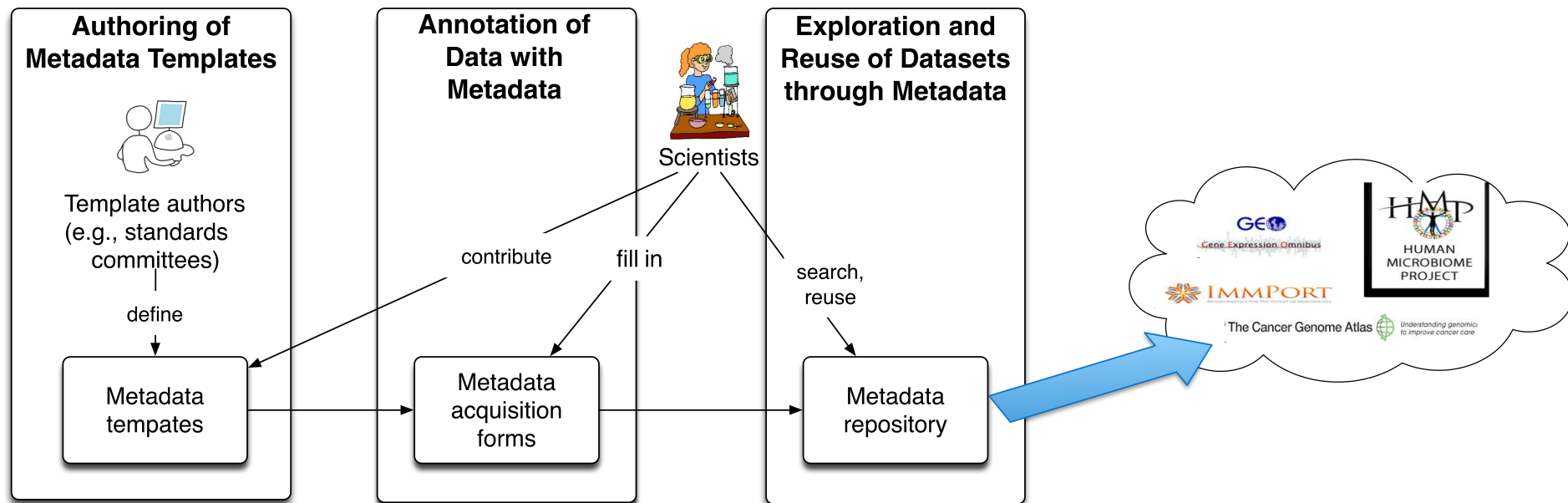


Name	disease
Value	



- 🔍 lung cancer (DOID) (61%)
- 🔍 chronic obstructive pulmonary disease (DOID) (13%)
- 🔍 lung squamous cell carcinoma (DOID) (5%)
- 🔍 idiopathic pulmonary fibrosis (DOID) (4%)
- 🔍 lung adenocarcinoma (DOID) (4%)
- 🔍 adenocarcinoma (DOID) (3%)
- 🔍 carcinoma (DOID) (2%)

The CEDAR Approach to Metadata



Authoring metadata using domain-specific ontologies and templates ...

- Aids dataset
 - Discovery
 - Exploration
 - Integration
 - Secondary use
- Aids communication of scientific results
- Aids knowledge management within research organizations
- Is making data more FAIR



CEDAR

CENTER FOR EXPANDED DATA ANNOTATION
AND RETRIEVAL