Biological Data Resources

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4/1/2015 CBPSS Panel

http://cherylab.stanford.edu

- The original model organism database for the first eukaryotic genome (*Saccharomyces cerevisiae)* to be sequenced
- Stores genes and biological annotations extracted from scientific literature *via* manual curation

http://www.yeastgenome.org/ … since 1993

SGD Annotations

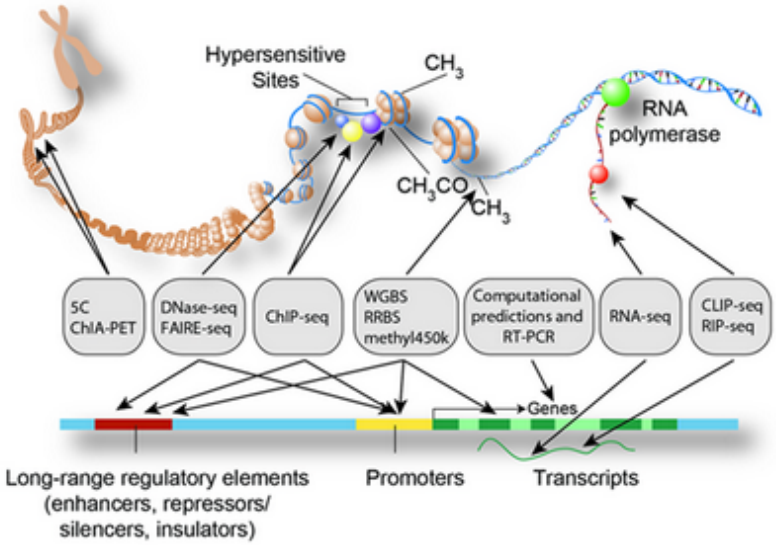
GO: FUS3 encodes a MAP protein kinase as shown by direct enzyme assay in Bao et al. (2004).

Phenotype: A null mutant of VAC14 has abnormal vacuolar morphology in S288C strain background as shown in Alghamdi et al. (2013).

Bioentity (what is it)	**Bioconcept** (what it does)	**Reference** (who said it)	**Experiment** (how is it known)	**Strain** (details)
FUS3	GO: MAP kinase activity	PMID:15620357	Direct enzyme assay	
VAC14	Phenotype: Vacuolar morphology	PMID: 23389034	Classical genetics	S288C

Additional properties (e.g., allele, conditions, qualifier) can be attached to any annotation.

ENCODE: Encyclopedia of DNA Elements



The ENCODE (Encyclopedia of DNA Elements) Consortium is an international collaboration of research groups funded by the National Human Genome Research Institute (NHGRI). The goal of ENCODE is to build a comprehensive parts list of functional elements in the human genome, including elements that act at the protein and RNA levels, and regulatory elements that control cells and circumstances in which a gene is active.

Image credits: Darryl Leja (NHGRI), Ian Dunham (EBI), Michael Pazin (NHGRI)

Data

To find and download ENCODE Consortium data:

- Click the Data toolbar above and browse data

News

Sept 12, 2014: Data release: 23 human and 5 mouse datasets. [read more]

August 28, 2014: modENCODE and ENCODE comparison papers

What is the ENCODE Consortium?

Production Groups

- **A** Broad Institute
- **B** Cold Spring Harbor; Centre for Genomic Regulation (CRG);
- **C** University of Connecticut Health Center; UCSD
- **D** HudsonAlpha; Pennsylvania State; UC Irvine; Duke; Caltech
- **E** UCSD; Salk Institute ; Joint Genome Institute; Lawrence Berkeley National Laboratory; UCSD
- **F** Stanford; University of Chicago; Yale
- **G** University of Washington; Fred Hutchinson Cancer Research Center; University of Massachusetts Medical School

Data Coordination Center

- **H** Stanford; UCSC

Data Analysis Center

- **I** University of Massachusetts Medical School; Yale; MIT; Stanford; Harvard; University of Washington

Technology Development Groups

- **J** MIT
- **K** Washington University, St. Louis
- **L** USC; Ohio State University; UC, Davis
- **M** University of Washington
- **N** Sloan-Kettering; Weill Cornell Medical College
- **O** Princeton; Weizmann
- **P** University of Michigan
- **Q** Broad Institute
- **R** University of Washington; UCSF
- **S** Advanced RNA Technologies, LLC
- **T** Harvard

Computational Analysis Groups

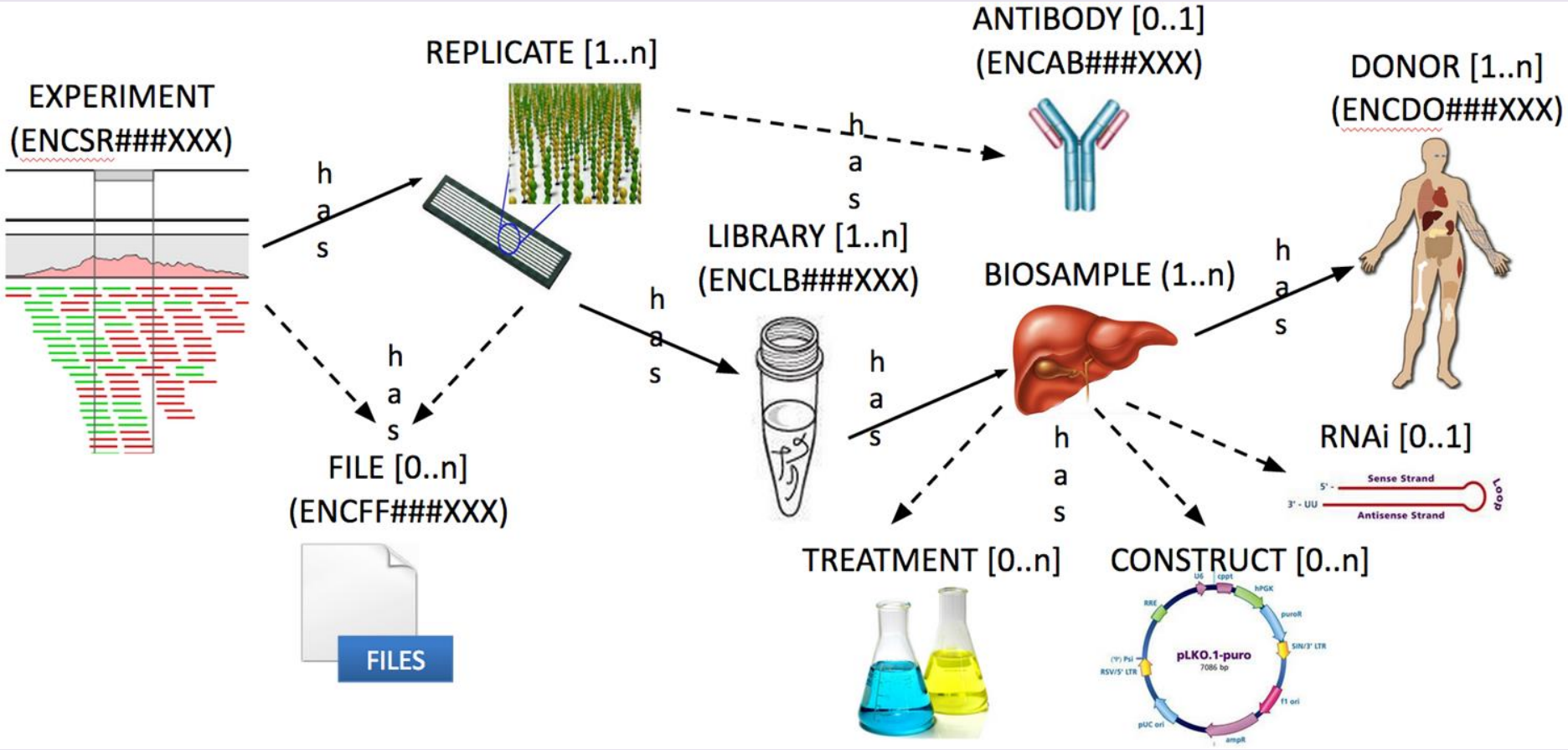
- **U** Berkeley; Wayne State University
- **V** MIT
- **W** University of Wisconsin
- **X** Sloan-Kettering; Broad Institute
- **Y** Stanford
- **Z** UCLA

Affiliated Groups

- **1** Wellcome Trust Sanger Institute
- **2** Florida State University

Image credit: NHGRI

Metadata model



Metadata submission

Metadata db

REST API

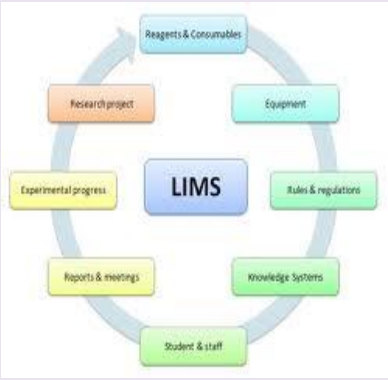
JSON Objects (GET, PATCH, POST)

Scripts

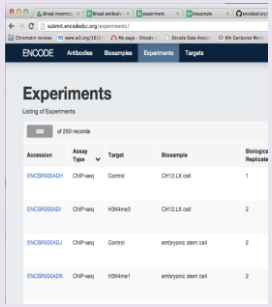
Wrangler Spreadsheets

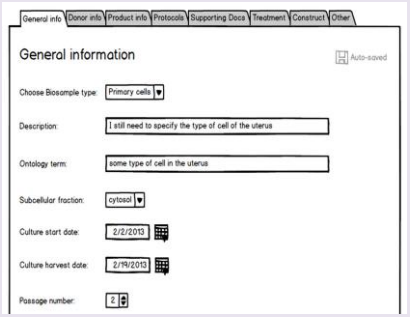
```
SERVER = 'http://184.169.162.173'
AUTHID = 'RD2YMWZD'
AUTHPW = '76bttrhni2lspbz5'

def get_json(url):

    headers = {'content-type': 'applicatio
    r = requests.get(url, auth=(AUTHID, AU
    if not r.status_code == requests.codes
        print r.text
        #r.raise_for_status()
    return r.json()

def post_json(url, payload):

    headers = {'content-type': 'applicatio
    r = requests.post(url, auth=(AUTHID, A
    if not r.status_code == requests.codes
        print r.text
        #r.raise_for_status()
        return False
    else:
        return True
```

Code



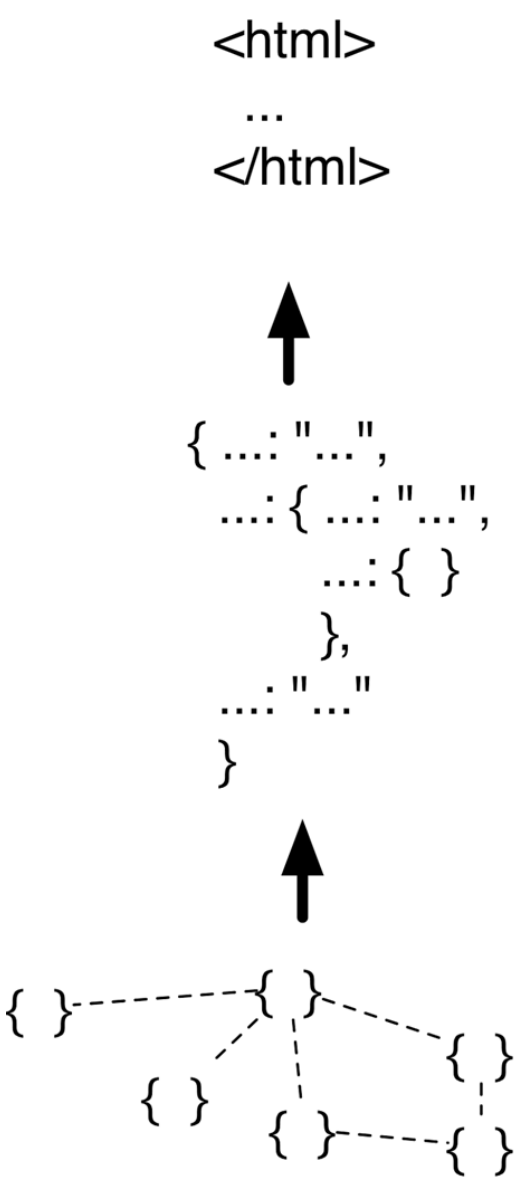
Web App

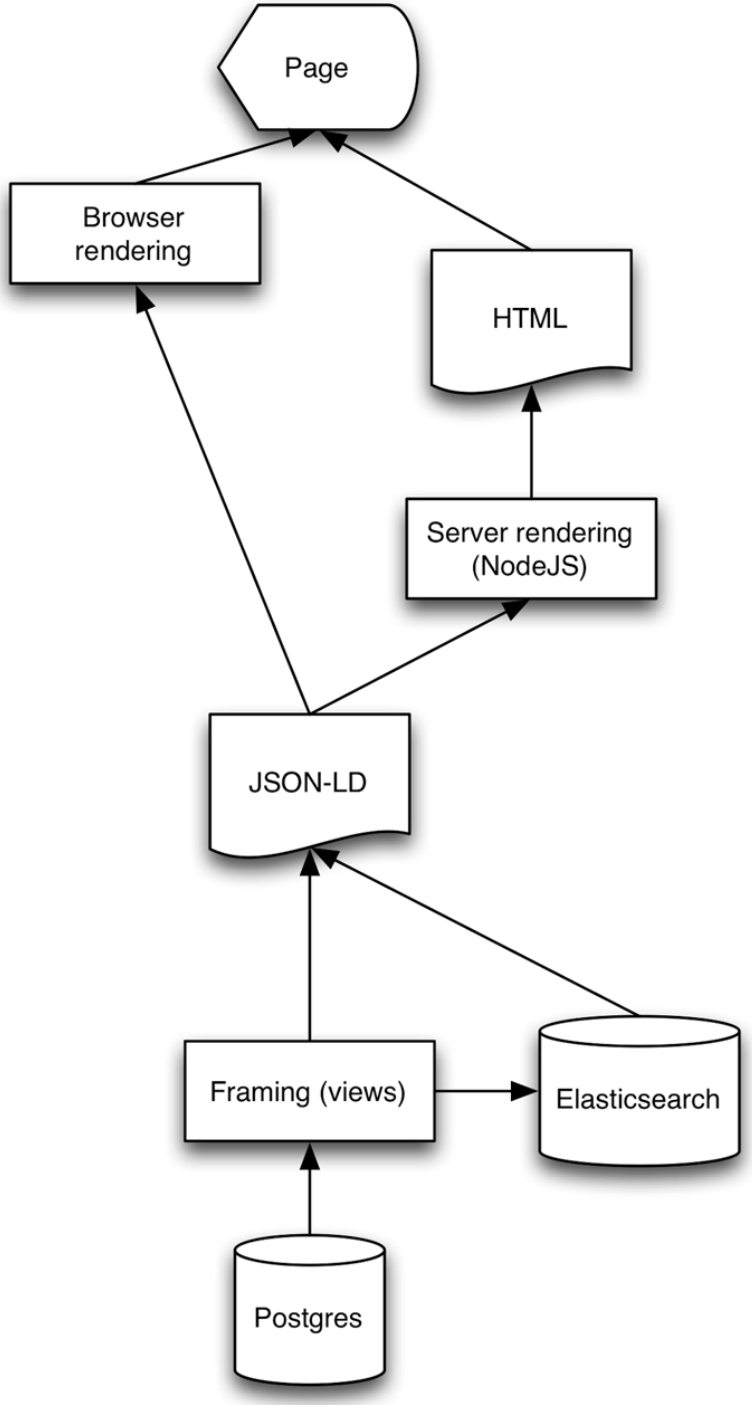




Web Forms

Software Stack

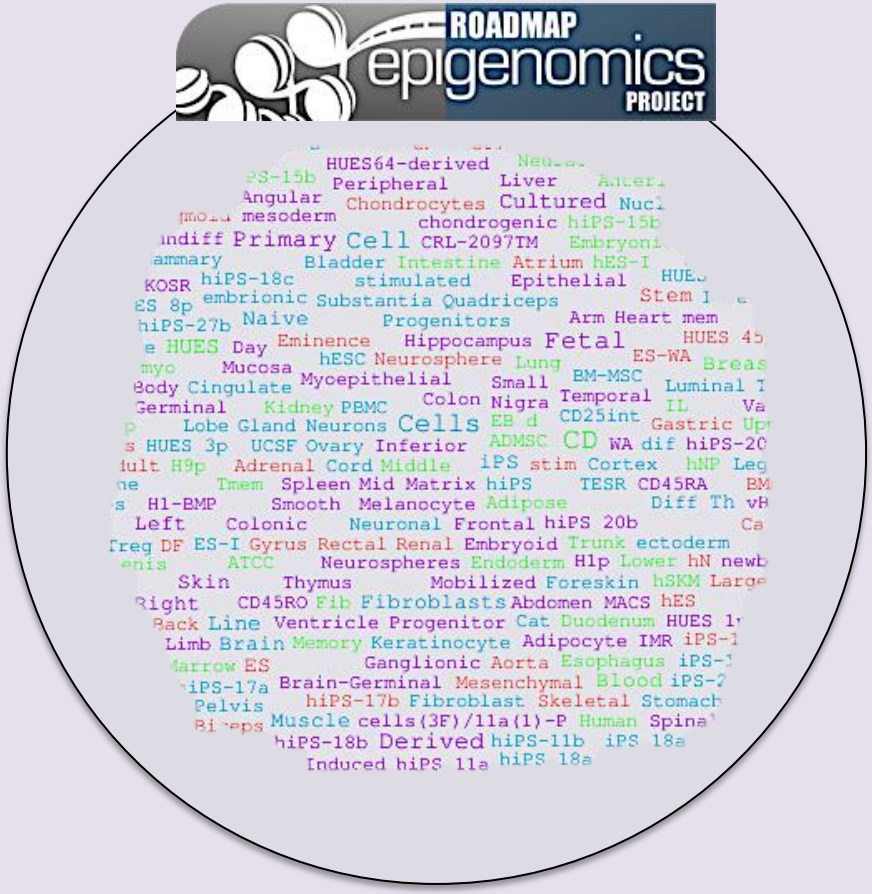




Find Common Biosamples Between Two Consortia

356 terms

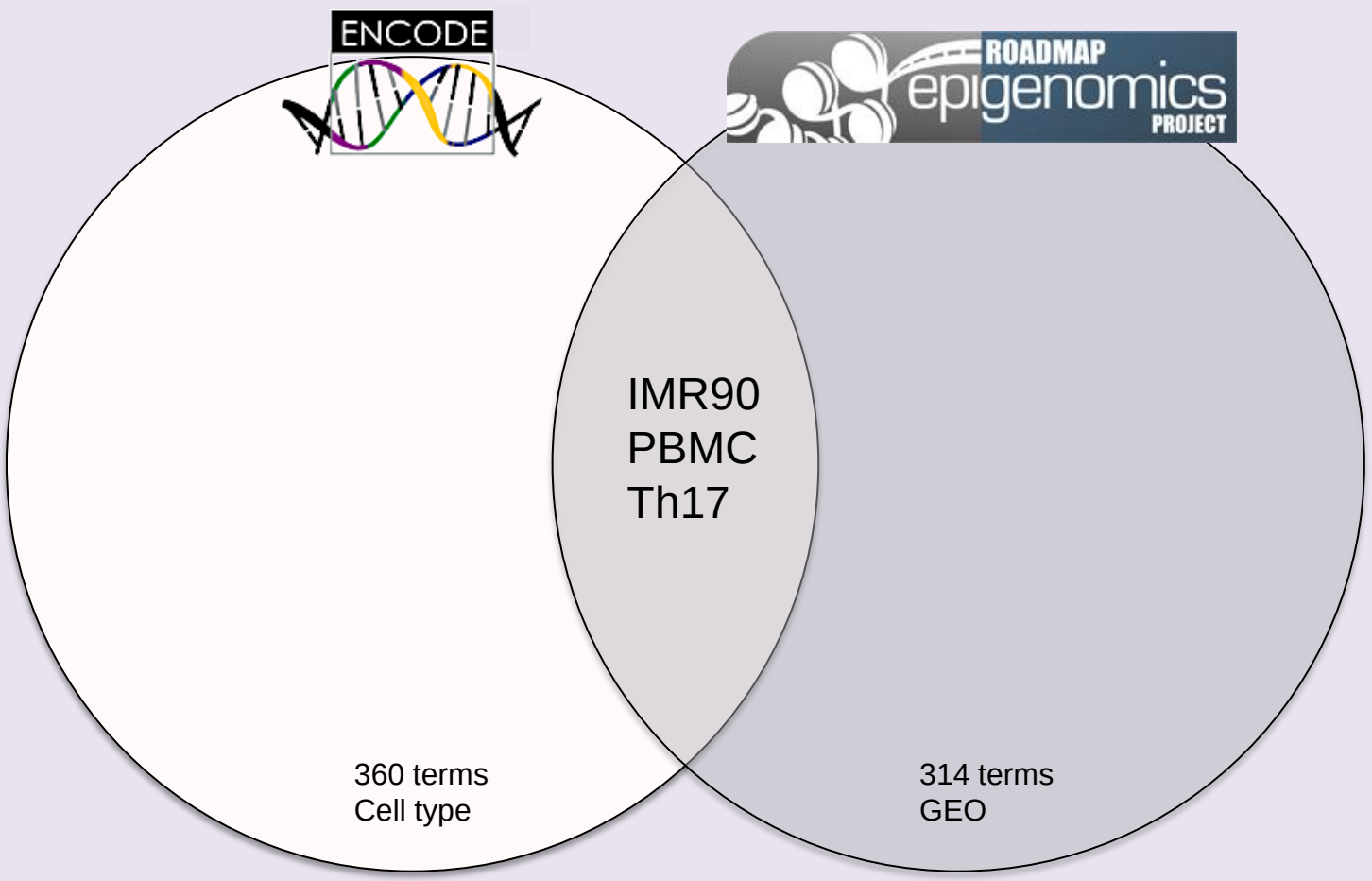
http://genome.ucsc.edu/ENCODE/cellTypes.html



314 terms

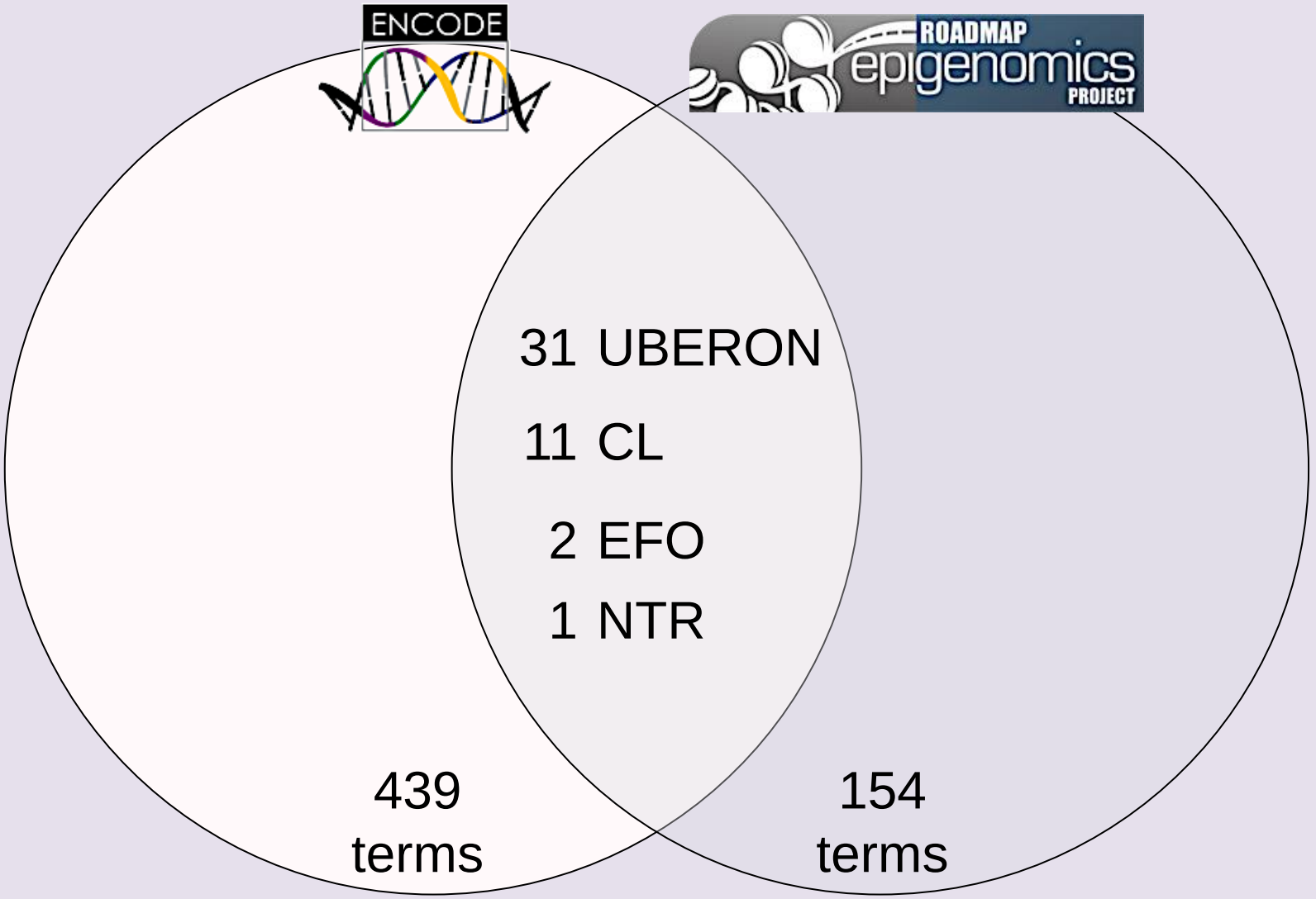
GEO characteristics: common_name, tissue_type, cell_type, lines

Labs were internally consistent



… but only 3 biosample names match exactly between projects

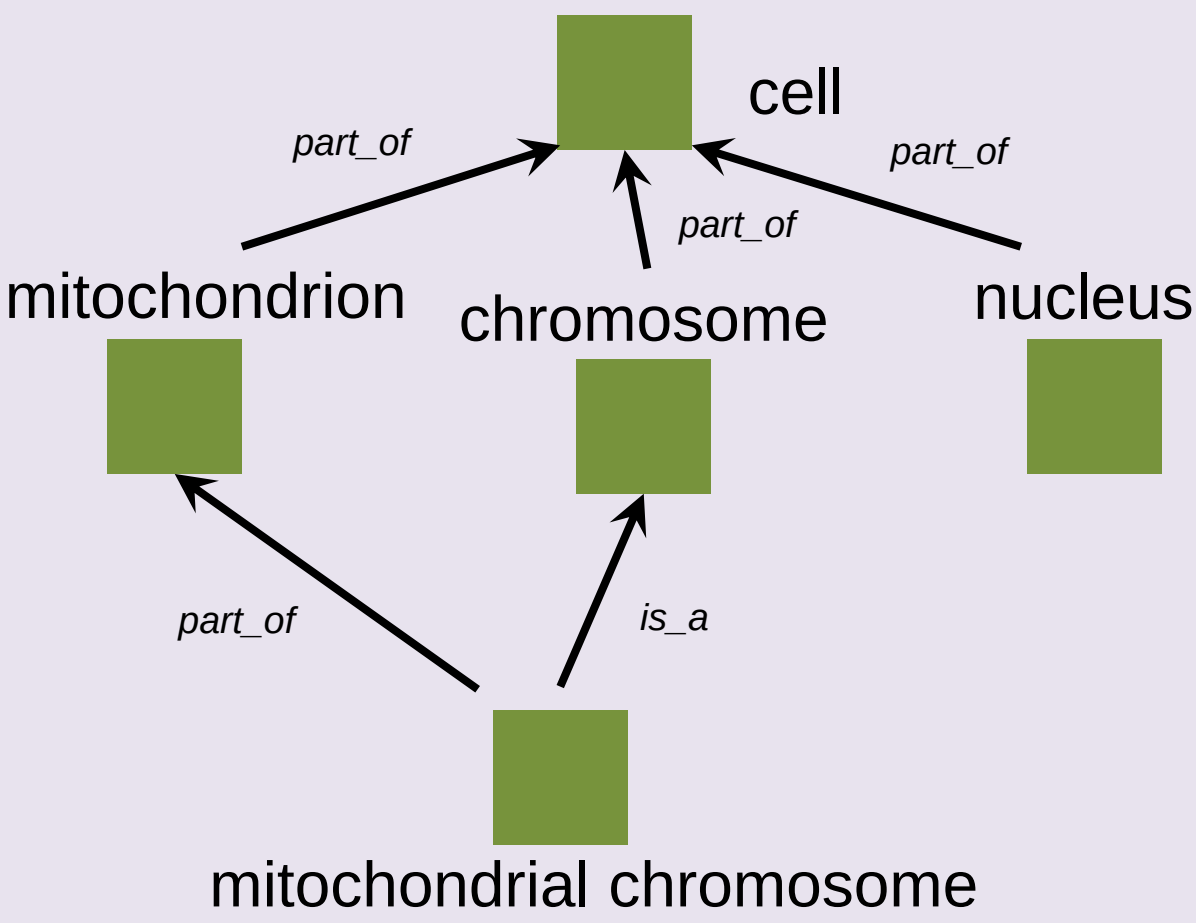
45 Biosamples in Common Between Current ENCODE & REMC



An ontology is a set of words...

An ontology is a set of words...

.. with different types of relationships to each other.



Why use ontologies?

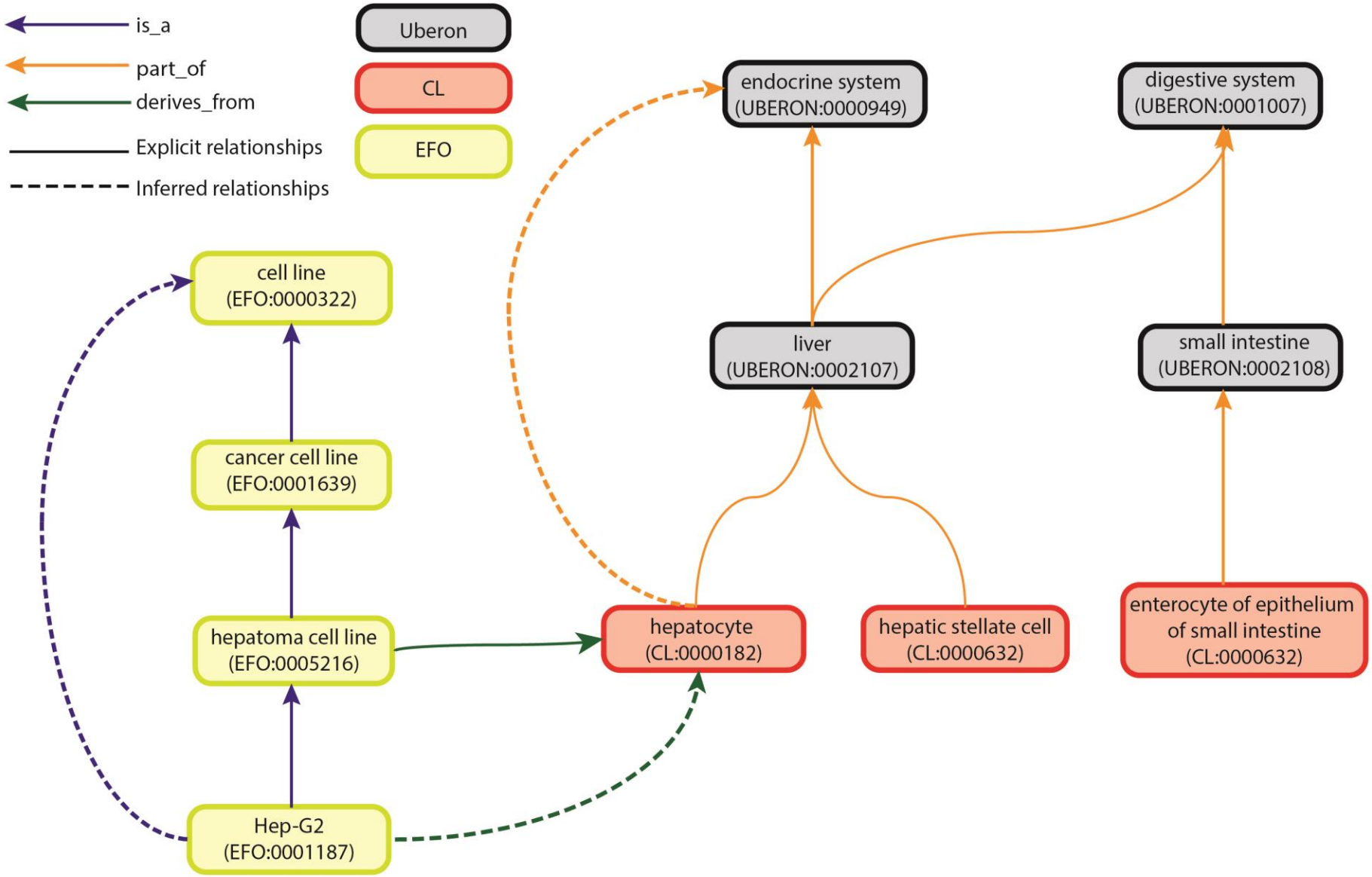
- Consistency of language and identifiers facilitates identification of data programmatically. Alternative spellings & phrases are synonyms. Independent of a particular data model.

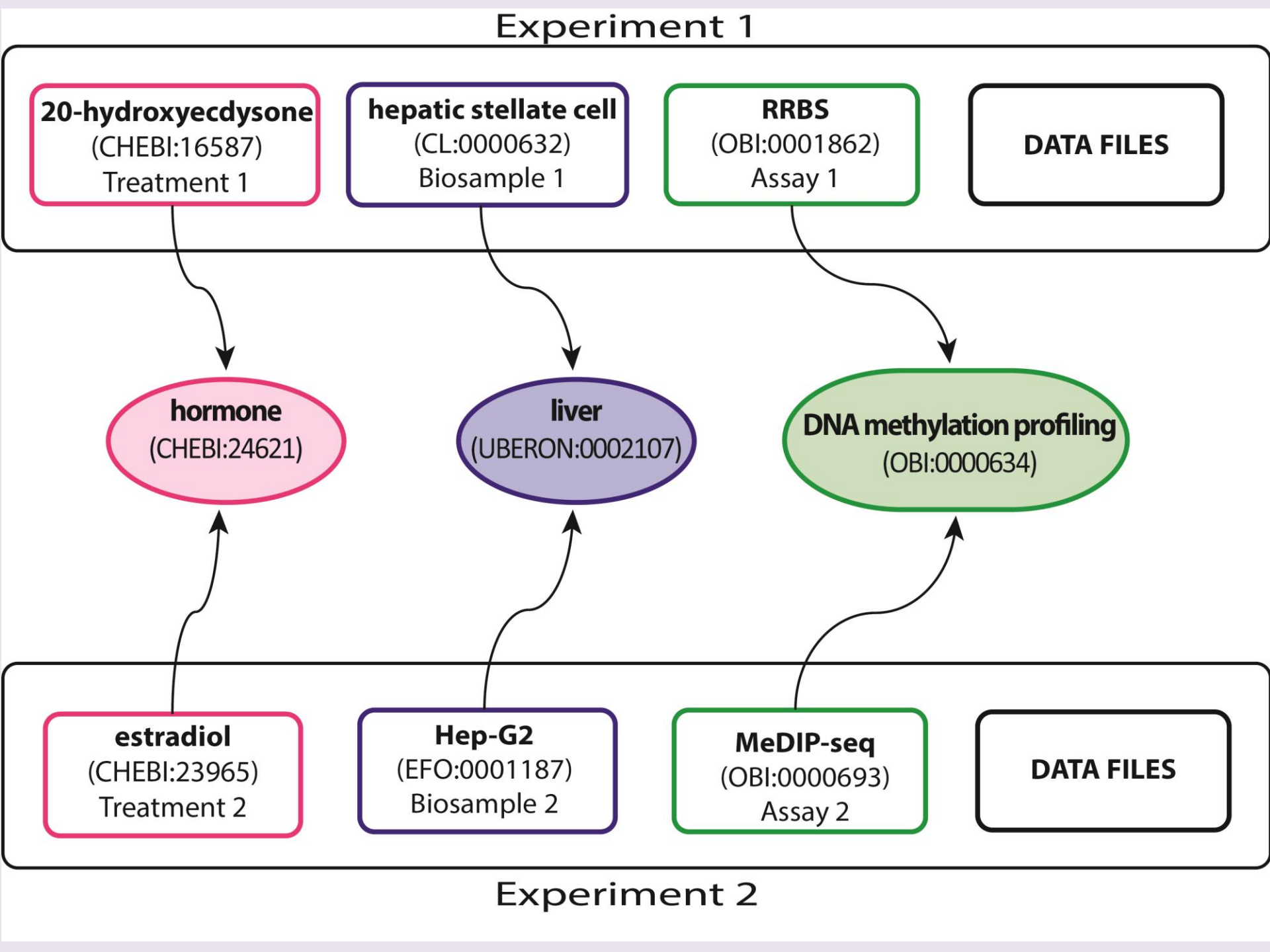
F ≠ f ≠ Female ≠ female

- Biological concepts are defined to provide scope

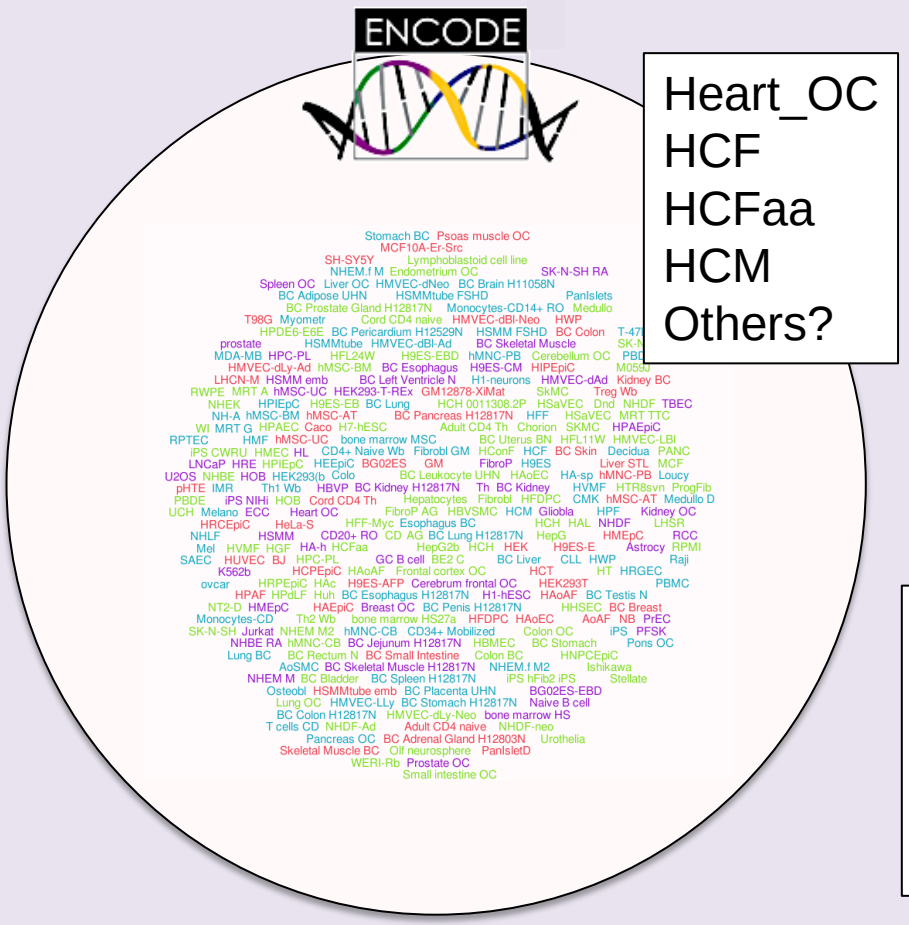
> *Mitochondria: A semiautonomous, self replicating organelle that occurs in varying numbers, shapes, and sizes in the cytoplasm of virtually all eukaryotic cells. It is notably the site of tissue respiration.*

- Relationships between terms can be computed to provide additional annotation details for grouping, searching, or analysis

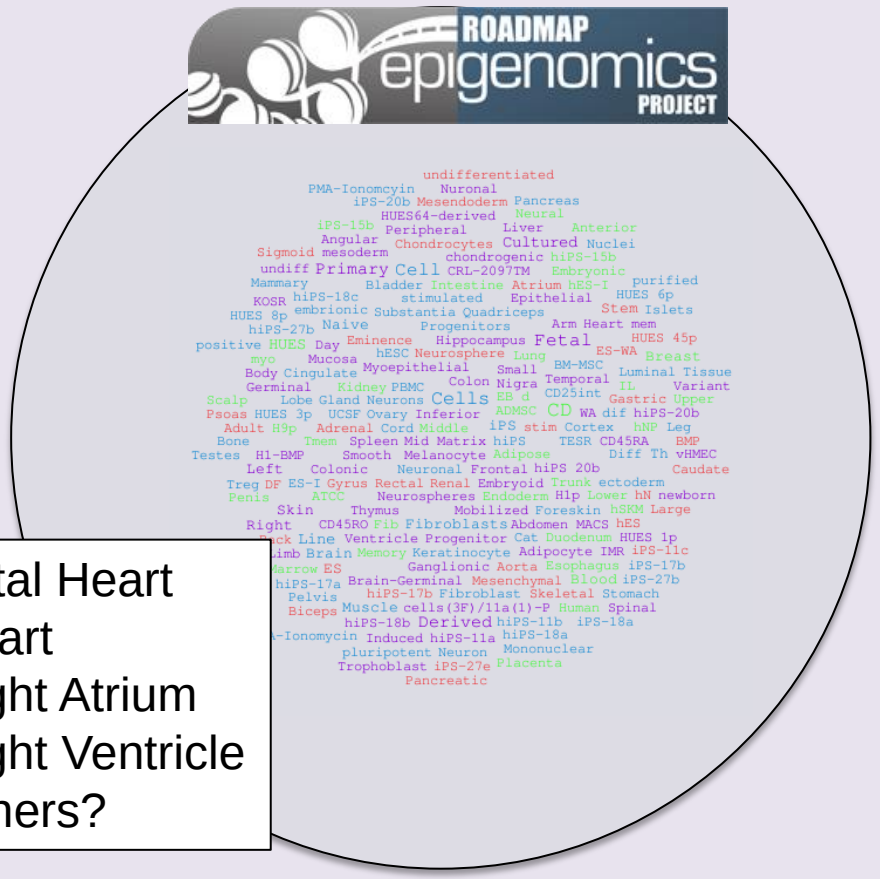

is_a
part_of
derives_from
Explicit relationships
Inferred relationships
Uberon
CL
EFO
endocrine system
(UBERON:0000949)
digestive system
(UBERON:0001007)
cell line
(EFO:0000322)
liver
(UBERON:0002107)
small intestine
(UBERON:0002108)
cancer cell line
(EFO:0001639)
hepatoma cell line
(EFO:0005216)
hepatocyte
(CL:0000182)
hepatic stellate cell
(CL:0000632)
enterocyte of epithelium
of small intestine
(CL:0000632)
Hep-G2
(EFO:0001187)


Experiment 1
20-hydroxyecdysone
(CHEBI:16587)
Treatment 1
hepatic stellate cell
(CL:0000632)
Biosample 1
RRBS
(OBI:0001862)
Assay 1
DATA FILES
hormone
(CHEBI:24621)
liver
(UBERON:0002107)
DNA methylation profiling
(OBI:0000634)
estradiol
(CHEBI:23965)
Treatment 2
Hep-G2
(EFO:0001187)
Biosample 2
MeDIP-seq
(OBI:0000693)
Assay 2
DATA FILES
Experiment 2

Challenge: Find all heart-related tissues?



Heart_OC
HCF
HCFaa
HCM
Others?



Fetal Heart
Heart
Right Atrium
Right Ventricle
Others?

Organism
- *Homo sapiens* 10
- *Mus musculus* 6

Biosample status
- in progress 7
- released 7
- deleted 2

Biosample type
- tissue 8
- primary cell 7
- in vitro differentiated cells 1

Organ
- heart 14

Sex
- male 8
- unknown 6
- female 2

Life stage
- fetal 7
- adult 6
- unknown 3

Source
- John Stamatoyannopoulos 7
- BDRL 6

Showing 16 of 16

heart (*Mus musculus*, adult 8 week) Biosample
Type: tissue
Source: John Stamatoyannopoulos
ENCBS536YRO **deleted**

heart (*Homo sapiens*, fetal 80 day) Biosample
Type: primary cell
Source: BDRL
ENCBS913ULP **in progress**

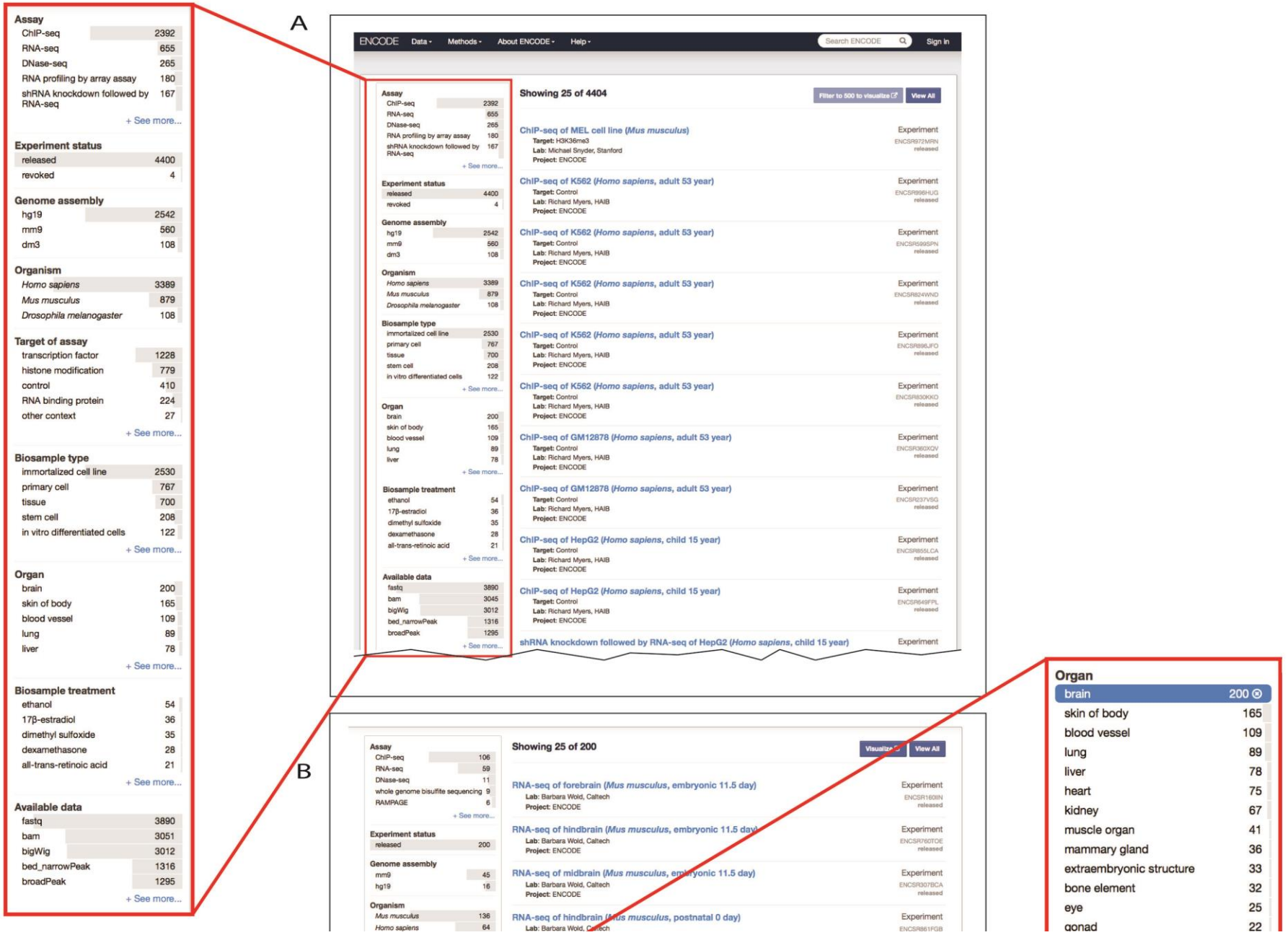
heart (*Homo sapiens*, fetal 76 day) Biosample
Type: primary cell
Source: BDRL
ENCBS953MIB **in progress**

heart (*Mus musculus*, adult 8 week) Biosample
Type: tissue
Source: John Stamatoyannopoulos
ENCBS331ENC **released**

cardiac fibroblast (*Homo sapiens*) Biosample
Type: primary cell
Source: ScienCell
ENCBS307AAA **released**

heart (*Mus musculus*, adult 8 week) Biosample
Type: tissue
Source: John Stamatoyannopoulos
ENCBS846GWQ **released**

heart (*Mus musculus*, adult 8 week) Biosample


A
B
ENCODE
Data
Methods
About ENCODE
Help
Search ENCODE
Sign In
Showing 25 of 4404
Filter to 500 to visualize
View All
Assay
ChIP-seq 2392
RNA-seq 655
DNase-seq 265
RNA profiling by array assay 180
shRNA knockdown followed by RNA-seq 167
+ See more...
Experiment status
released 4400
revoked 4
Genome assembly
hg19 2542
mm9 560
dm3 108
Organism
Homo sapiens 3389
Mus musculus 879
Drosophila melanogaster 108
Biosample type
immortalized cell line 2530
primary cell 767
tissue 700
stem cell 208
in vitro differentiated cells 122
Organ
brain 200
skin of body 165
blood vessel 109
lung 89
liver 78
Biosample treatment
ethanol 54
17β-estradiol 36
dimethyl sulfoxide 35
dexamethasone 28
all-trans-retinoic acid 21
Available data
fastq 3890
bam 3045
bigWig 3012
bed_narrowPeak 1316
broadPeak 1295
ChIP-seq of MEL cell line (Mus musculus)
Target: H3K36me3
Lab: Michael Snyder, Stanford
Project: ENCODE
Experiment
ENCSR972MRN
released
ChIP-seq of K562 (Homo sapiens, adult 53 year)
Target: Control
Lab: Richard Myers, HAIB
Project: ENCODE
Experiment
ENCSR996HUG
released
ChIP-seq of K562 (Homo sapiens, adult 53 year)
Target: Control
Lab: Richard Myers, HAIB
Project: ENCODE
Experiment
ENCSR599SPN
released
ChIP-seq of K562 (Homo sapiens, adult 53 year)
Target: Control
Lab: Richard Myers, HAIB
Project: ENCODE
Experiment
ENCSR824WND
released
ChIP-seq of K562 (Homo sapiens, adult 53 year)
Target: Control
Lab: Richard Myers, HAIB
Project: ENCODE
Experiment
ENCSR896JFO
released
ChIP-seq of K562 (Homo sapiens, adult 53 year)
Target: Control
Lab: Richard Myers, HAIB
Project: ENCODE
Experiment
ENCSR830KKD
released
ChIP-seq of GM12878 (Homo sapiens, adult 53 year)
Target: Control
Lab: Richard Myers, HAIB
Project: ENCODE
Experiment
ENCSR360XQV
released
ChIP-seq of GM12878 (Homo sapiens, adult 53 year)
Target: Control
Lab: Richard Myers, HAIB
Project: ENCODE
Experiment
ENCSR237VSG
released
ChIP-seq of HepG2 (Homo sapiens, child 15 year)
Target: Control
Lab: Richard Myers, HAIB
Project: ENCODE
Experiment
ENCSR855LCA
released
ChIP-seq of HepG2 (Homo sapiens, child 15 year)
Target: Control
Lab: Richard Myers, HAIB
Project: ENCODE
Experiment
ENCSR649FPL
released
shRNA knockdown followed by RNA-seq of HepG2 (Homo sapiens, child 15 year)
Experiment
Showing 25 of 200
Visualize
View All
ChIP-seq 106
RNA-seq 59
DNase-seq 11
whole genome bisulfite sequencing 9
RAMPAGE 6
Experiment status
released 200
Genome assembly
mm9 45
hg19 16
Organism
Mus musculus 136
Homo sapiens 64
RNA-seq of forebrain (Mus musculus, embryonic 11.5 day)
Lab: Barbara Wold, Caltech
Project: ENCODE
Experiment
ENCSR160IIN
released
RNA-seq of hindbrain (Mus musculus, embryonic 11.5 day)
Lab: Barbara Wold, Caltech
Project: ENCODE
Experiment
ENCSR760TOE
released
RNA-seq of midbrain (Mus musculus, embryonic 11.5 day)
Lab: Barbara Wold, Caltech
Project: ENCODE
Experiment
ENCSR307BCA
released
RNA-seq of hindbrain (Mus musculus, postnatal 0 day)
Lab: Barbara Wold, Caltech
Experiment
ENCSR861FGB
Organ
brain 200
skin of body 165
blood vessel 109
lung 89
liver 78
heart 75
kidney 67
muscle organ 41
mammary gland 36
extraembryonic structure 33
bone element 32
eye 25
gonad 22


+ See more...
Biosample treatment
ethanol 54
17β-estradiol 36
dimethyl sulfoxide 35
dexamethasone 28
all-trans-retinoic acid 21
+ See more...
Available data
fastq 3890
bam 3051
bigWig 3012
bed_narrowPeak 1316
broadPeak 1295
+ See more...
B
Assay
ChIP-seq 106
RNA-seq 59
DNase-seq 11
whole genome bisulfite sequencing 9
RAMPAGE 6
+ See more...
Experiment status
released 200
Genome assembly
mm9 45
hg19 16
Organism
Mus musculus 136
Homo sapiens 64
Biosample type
tissue 160
primary cell 40
Organ
brain 200
skin of body 165
blood vessel 109
lung 89
liver 78
heart 75
kidney 67
muscle organ 41
mammary gland 36
extraembryonic structure 33
bone element 32
eye 25
gonad 22
stomach 22
placenta 19
bronchus 18
small intestine 18
mouth 16
spleen 14
thymus 13
large intestine 11
prostate gland 11
esophagus 10
pancreas 8
spinal cord 8
urinary bladder 6
adrenal gland 5
thyroid gland 3
tongue 3
trachea 3
lymphatic vessel 1
- See fewer
Showing 25 of 200
Visualize
View All
RNA-seq of forebrain (Mus musculus, embryonic 11.5 day)
Lab: Barbara Wold, Caltech
Project: ENCODE
Experiment
ENCSR160IIN
released
RNA-seq of hindbrain (Mus musculus, embryonic 11.5 day)
Lab: Barbara Wold, Caltech
Project: ENCODE
Experiment
ENCSR760TOE
released
RNA-seq of midbrain (Mus musculus, embryonic 11.5 day)
Lab: Barbara Wold, Caltech
Project: ENCODE
Experiment
ENCSR307BCA
released
RNA-seq of hindbrain (Mus musculus, postnatal 0 day)
Lab: Barbara Wold, Caltech
Project: ENCODE
Experiment
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released
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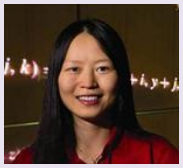
Lab: Barbara Wold, Caltech
Project: ENCODE
Experiment
ENCSR527RFK
released
RNA-seq of midbrain (Mus musculus, postnatal 0 day)
Lab: Barbara Wold, Caltech
Project: ENCODE
Experiment
ENCSR026ZRP
released
RNA-seq of hindbrain (Mus musculus, postnatal 0 day)
Lab: Thomas Gingeras, CSHL
Project: ENCODE
Experiment
ENCSR749BAG
released
RNA-seq of midbrain (Mus musculus, postnatal 0 day)
Lab: Thomas Gingeras, CSHL
Project: ENCODE
Experiment
ENCSR255SDF
released
RNA-seq of forebrain (Mus musculus, postnatal 0 day)
Lab: Thomas Gingeras, CSHL
Project: ENCODE
Experiment
ENCSR723SZV
released
RNA-seq of forebrain (Mus musculus, embryonic 11.5 day)
Lab: Barbara Wold, Caltech
Project: ENCODE
Experiment
ENCSR000OXO
released
RNA-seq of Purkinje cell (Homo sapiens, adult 20 year)
Lab: Barbara Wold, Caltech
Project: ENCODE
Experiment
ENCSR417EDR
released
RNA-seq of Purkinje cell (Homo sapiens, adult 20 year)
Lab: Barbara Wold, Caltech
Project: ENCODE
Experiment
ENCSR672QFA
released
RNA-seq of Purkinje cell (Homo sapiens, adult 20 year)
Lab: Barbara Wold, Caltech
Project: ENCODE
Experiment
released
Organ
brain 200
skin of body 165
blood vessel 109
lung 89
liver 78
heart 75
kidney 67
muscle organ 41
mammary gland 36
extraembryonic structure 33
bone element 32
eye 25
gonad 22
stomach 22
placenta 19
bronchus 18
small intestine 18
mouth 16
spleen 14
thymus 13
large intestine 11
prostate gland 11
esophagus 10
pancreas 8
spinal cord 8
urinary bladder 6
adrenal gland 5
thyroid gland 3
tongue 3
trachea 3
lymphatic vessel 1
- See fewer

ENCODE DCC

https://www.encodeproject.org/

Eurie Hong, Mike Cherry (PI), Jim Kent (co-PI), Ben Hitz

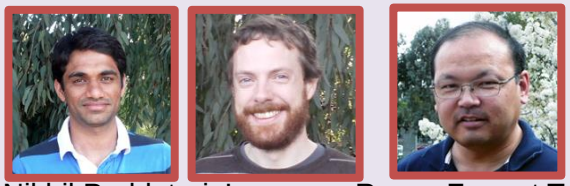


Zhiping Weng, ENCODE DAC

Data Wranglers

Esther Chan, Jean Davidson, Venkat Malladi, Cricket Sloan, J. Seth Strattan

Software Engineers



Nikhil Podduturi, Laurence Rowe, Forrest Tanaka

QA, administration, biocuration

Brian Lee, Stuart Miyasato, Matt Simison, Zhenhua Wang, Marcus Ho

Funding Source: NHGRI

Malladi et al. ; Database, 2015, 1–11 doi: 10.1093/database/bav010

@encodedcc encode-help@lists.stanford.edu https://github.com/ENCODE-DCC/

Using ontologies for metadata annotation

1. **Uber Anatomy ontology** (UBERON; http://uberon.org/)
   - tissues: heart, blood, brain

2. **Cell Ontology** (CL; http://cellontology.org/)
   - primary cell types: hepatocyte, cardiomyocyte

3. **Experimental Factor Ontology** (EFO; http://www.ebi.ac.uk/efo/)
   - immortalized cell lines: K562, HepG2, MCF-7

4. **Ontology for Biomedical Investigations** (OBI; http://obi-ontology.org/page/Main_Page)
   - experimental assays: RNA-seq, CLIP-seq, ChIP-seq, etc

5. **Chemical Entities of Biological Interest** (ChEBI; http://www.ebi.ac.uk/chebi/)
   - chemical treatments: estradiol, ethanol, etc

6. **Sequence Ontology** (SO; http://www.sequenceontology.org/)
   - nucleic acid being sequenced: microRNA, poly-A+ mRNA, etc

7. **Gene Ontology** (GO; http://www.geneontology.org/)
   - group gene products that are targets of ChIP-seq or RNAi experiments