

# **DATS Specification v2.1**

# NIH BD2K DataMed model annotated with schema.org

#### **Status:**

Version 2.1

## Scope of the Document:

Section 1 of this document introduces the **DatA Tag Suite** (**DATS**) model, its JSON-schemas-based serialization, a mapping between DATS and <u>schema.org</u> and a <u>schema.org</u>-annotated <u>JSON-LD</u> context and a description of the next steps; section 2 provides an overview of the methods leading to the DATS development, including the use cases and the existing schemas and material reviewed.

#### **Abstract:**

The DATS model has been designed to support the intended capability of the NIH BD2K Data Discovery Index prototype, outlined in the bioCADDIE White Paper and named DataMed. The DATS model and its serialization describe the metadata and the structure for datasets needed to populate the DataMed prototype, along the line of the Journal Article Tag Suit (JATS) used by PubMed for literature. The DATS has been designed to have core and extended elements, to progressively accommodate more specialized data types, as needed.

#### **Associated Material:**

Appendix I, II and III (related to the current DATS model v2), Appendix IV (schemas mapping, part of the development process), along with the schema.org-annotated JSON serialization and examples (of core and extended DATS) are available from the <a href="bioCADDIE">bioCADDIE</a> Github repository. Previous versions, presentations and notes on this work can also be found at the <a href="bioCADDIE">bioCADDIE</a> Metadata Working Group (WG) 3 webpage. The schemas and models in Appendix III are also described in the <a href="BioSharing Collection for bioCADDIE">BioSharing Collection for bioCADDIE</a>.

#### Intended Audience:

This document is aimed at: (i) the <u>DataMed development team</u> that will implement and test the model, (ii) prospective data sources that wish to be indexed in the DataMed prototype, and (iii) developers of data harvesting and other metadata tools and catalogs.

#### **Authors and Contact:**

The work has been designed and carried out - as an *open activity* with *community input* - by core bioCADDIE project members (Susanna-Assunta Sansone, Alejandra Gonzalez-Beltran, Philippe Rocca-Serra, with George Alter, Mary Vardigan, Jeffrey Grethe, Hua Xu and other members of the DataMed development team), with contributions from the members of the <u>bioCADDIE Metadata WG 3</u> and <u>WG7</u>. Comments to the DATS can also be added to this Google document, to the <u>Github issue tracker</u> or sent to Susanna-Assunta Sansone (WG3 chair) via biocaddie[at]ucsd.edu (subject line: DATS Spec V2.1).



### **Table of Content**

- 1. The DATS Model
  - 1.1. Brief History Leading to this Version
  - 1.2. General Design
  - 1.3. Core and Extended Elements
  - 1.4. Towards a Schema.org Annotated JSON-LD Serialization
  - 1.5. Detailed Model Description
    - 1.5.1. Cardinality and Requirement Level
    - 1.5.2. Mapping DATS Back to Existing Schemas/Models
- 2. Standard Operating Procedure
  - 2.1. Combined Approaches
    - 2.1.1. Data Discovery Initiatives and Metadata Initiatives
    - 2.1.2. Metamodels
  - 2.2. Top-down Use Cases
    - 2.2.1. Extracting Competency Questions
    - 2.2.2. Identify Entities Attributes and Values
  - 2.3. Bottom-up Mapping
    - 2.3.1. Input Material
      - 2.3.1.1. Generic Metadata Schemas and Models
      - 2.3.1.2. Life Science Metadata Schemas
    - 2.3.2. BioSharing Collection of Schemas/Models



# 1. The DATS Model

This section and related Appendixes describe the DATS model, providing a summary of the previous versions, an overview of the key metadata elements and their relations, and a detailed description of each entity. The DATS model follows the <u>FAIR community best practices</u> for data management, and the recently released <u>W3C Candidate Recommendations on Data on the Web Best Practices</u>.

A full description of the DATS core and extended elements is in a separate <u>"Appendix I - NIH BD2K BioCADDIE DataMed DATS model v2.1 file"</u> available as a Google spreadsheet, and also linked available from the <u>bioCADDIE Github repository</u>. The methods used for its development, including the use cases and the existing schemas and material reviewed, are described in section 2.

### 1.1. Brief History Leading to this Version

In August 2015, the specification v1.0 (DOI:10.5281/zenodo.28019) was released, with the model available as machine readable JSON schemata and several examples. The initial model was tested by the DataMed development team with a variety of data sources. Following the evaluation phase and review by the WG3 members and the larger community, several changes and additions were made leading to the release of the specification v1.1 (DOI:10.5281/zenodo.53078) in March 2016. In June 2016, the specification v2.0 (DOI:10.5281/zenodo.54010) was released; this version represented a further evolution of the model, including the accessibility metadata elements produced by bioCADDIE WG7, a set of JSON-schemas and JSON examples, a schema.org-annotated context file (based on the DATS to schema.org mapping).

The current v2.1 includes: (i) edits based on feedback from the community, including those that attended the <u>DataMed repository workshop</u> in June 2016, and the DataMed development team; and (ii) a schema.org-annotated <u>JSON-LD</u> serialization. The latter activity, set to identifying additional elements to extend the schema.org model, is also carried out under the <u>bioschemas.org</u> umbrella.

## 1.2. General Design

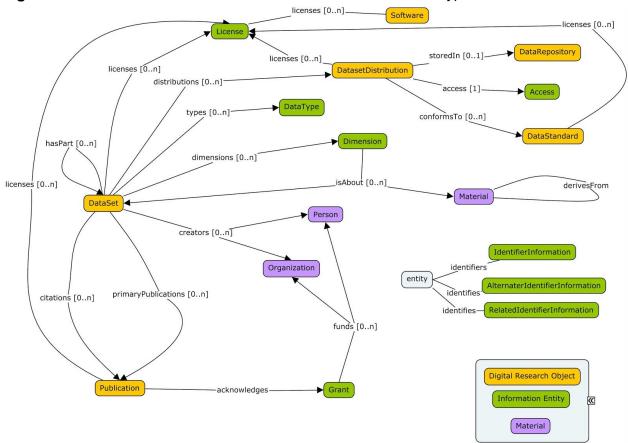
The DATS model is designed around the *Dataset*, an element that intends to cater for any unit of information stored by repositories. *Dataset* covers both (i) experimental datasets, which do not change after deposition to the repository, and (ii) datasets in reference knowledge bases, describing dynamic concepts, such as "genes", whose definition morphs over time. The *Dataset* element is also linked to other digital research objects part of the <u>NIH Commons</u>, such as *Software* and *Data Standard*, which are the focus on other discovery indexes and therefore are not described in detail in this model. The model may appear quite detailed in places as consequence of (i) the combined approaches used to identify the required metadata elements (see section 2), and (ii) the attempt to aim for the maximum coverage of use cases with minimal number of metadata elements. Nevertheless, it is anticipated that not all use cases can be fulfilled, and that it is difficult to foreseen all type of data sources the DataMed prototype should retrieve information from.



#### 1.3. Core and Extended Elements

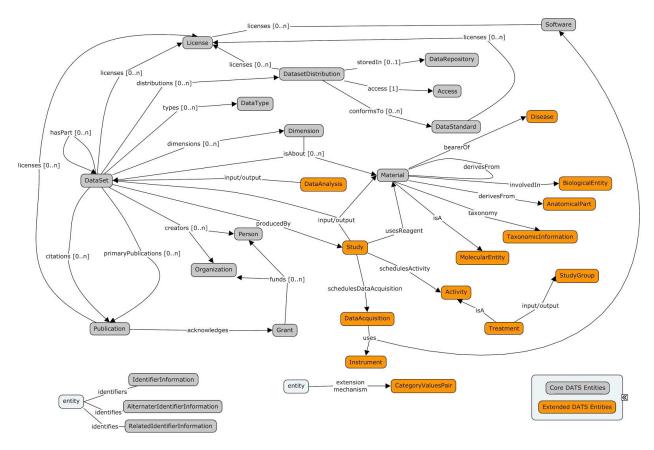
The DATS model has core and extended elements, to progressively accommodate domain-specific metadata for more specialized data types, as needed. Like the JATS, the core DATS elements are generic and applicable to any type of datasets. The extended DATS includes an initial set of elements, some of which are specific for life, environmental and biomedical science domains and can be further extended as needed. Note that the core should by no mean seen as the mandatory set; further information on the requirement level is provided in section 1.5. An overview of the core and extended elements, their types and relations are shown in Figure 1 and 2, respectively.

Figure 1. A schematic overview of the DATS core elements, their types and relations:



**Figure 2.** A schematic overview of the **DATS core and extended elements**, their types and relations:





### 1.4. Schema.org Annotated JSON-LD Serialization

The core and the extended DATS entities are available as machine readable JSON schemata. which are annotated with schema.org elements and made available in the bioCADDIE Github repository, which also includes JSON examples of existing datasets. The use of schema.org annotation provides structured markup. Its use and extension to the life and biomedical areas, is an ongoing discussion under the bioschemas.org umbrella and NIH Commons Working Groups, which the bioCADDIE team is part of. Preliminary discussions indicate that by implementing a schema.org-annotated DATS model, the DataMed prototype will benefit from an increased visibility (by search engines and tools), increased accessibility (via common query interfaces), and possibly, an improve in search ranking. Data repositories indexed by DataMed will also get the benefit of being more visible to search engines through DataMed. The initial "Appendix II -NIH BD2K BioCADDIE DataMed DATS v2.1 mapped to schema.org file" types and properties is available as a Google spreadsheet; this may be subject to change as current schema.org elements and those of health and life science extension evolve, also because the current life science extension is biased towards clinical studies. Gap in coverage have been identified during the DATS annotation process, and are highlighted in the Appendix II file; notifications of these gaps have been submitted to the schema.org github tracker.

## 1.5. Detailed Model Description

The file describes the metadata elements, grouped by types, and the Google <u>JSON</u> <u>style</u> <u>guide</u> has been used to name relevant elements. The descriptors for each metadata element (Entity),



include: Property (describing the Entity), Definition (of each Entity and Property), Value(s) (allowed for each Property).

In Appendix I, the DATS elements are also associated to relevant use cases (detailed in section 2.2) and/or to the existing schema(s)/model(s) used in the development process (detailed in section 2.3), to justify their relevance and provenance.

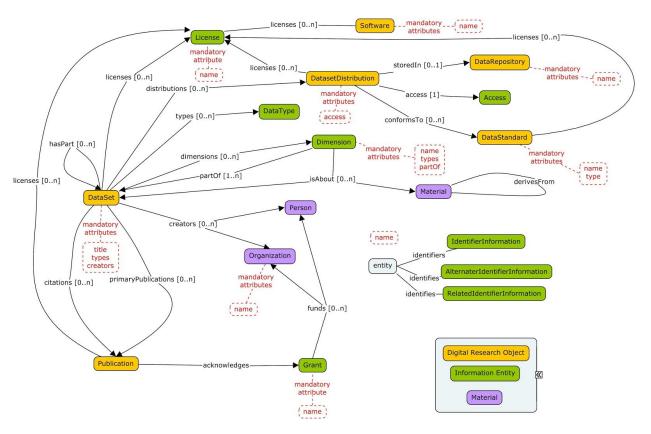
#### 1.5.1. Cardinality and Requirement Level

In both core and extended DATS, Entity are **not** mandatory but applicable only if relevant to the dataset to be described; when an Entity is used, also only few of its Properties are defined as mandatory. Figure 3 provides a view of the core entities highlighting the few properties defined as mandatory.

Cardinality restrictions indicate the number of valid occurrences for an attribute; the initial set of metadata elements are ranked and provisionally associated with a requirement level. The keywords "MUST", "MUST NOT", "REQUIRED", "SHALL", "SHALL NOT", "SHOULD", "SHOULD NOT", "RECOMMENDED", "MAY", and "OPTIONAL" in this document are to be interpreted as described in <a href="RFC 2119">RFC 2119</a> [RFC2119]. The requirement level for a field that is dependent on another one appears in between parenthesis, e.g. 'identifierScheme' MUST be present if 'identifier' is available, and we indicate this with (MUST). While there is some overlap in these specifications (e.g. if the cardinality of an attribute is 1, the requirement level is necessarily MUST), the requirement level adds information about the relative importance of including or not the non-compulsory attributes (either because they are recommended or they are truly optional). Cardinality restrictions will be used for data modelling purposes.

Figure 3. A schematic overview of the DATS core entities and theirs few properties with requirement level "MUST":





#### 1.5.2. Mapping DATS Back to Existing Schemas/Models

To facilitate the use of the DATS model by prospective data sources that wish to be indexed in the DataMed prototype, the DATS elements have been mapped to a number of existing (i) generic and widely used schemas, such as schema.org and DataCite, and (ii) domain specific repositories schemas. This mapping is in a separate "Appendix III - NIH BD2K BioCADDIE DataMed DATS v2.1 mapped to other models file" available as a Google spreadsheet and also linked availbale from the bioCADDIE Github repository. The work is also being carried out by the DataMed development team to inform the implementation of data harvesting converters, pulling data from those repositories.

# 2. Standard Operating Procedure

This section outlines the methods and the process used to identify an initial set of metadata elements, leading to the DATS specification.

# 2.1. Combined Approaches

A variety of data discovery initiatives exists or are being developed; although they have different scope, use cases and approaches, the analysis of their metadata schemas has been a valuable guidance. Several metamodels for representing metadata also exist and have been reviewed. In addition to these, the results of the following approaches has been compared and combined to identify the initial set of metadata elements:

• an analysis of the use cases (top-down approach; section 2.2); and



• a mapping of existing metadata schemas (bottom-up approach; section 2.3 and Appendix III).

### 2.1.1. Data Discovery Initiatives and Metadata Initiatives

This is a **non-comprehensive** list of the data discovery and integrative initiatives analysed, which might have more specific aims and different use cases than the intended capability of the DataMed prototype.

- UK <u>JISC</u> <u>Research</u> <u>Data</u> <u>Registry</u> <u>and</u> <u>Discovery</u> <u>Service</u>: relies on Registry Interchange Format Collections and Services (<u>RIF-CS</u>); related documentations: <u>Github</u> <u>repository</u>, <u>WP3:</u> <u>Metadata</u> <u>Development</u> <u>and</u> <u>Standardisation</u>, <u>Report:</u> <u>metadata</u> <u>mapping</u> <u>schemes</u> <u>/ recommendations</u> (<u>version</u> 9, 2014-05-09).
- 2. <u>Datacite Metadata Search</u> to search datasets registered with Datacite.
- 3. European <u>EUDAT B2FIND</u>: relies in <u>CKAN (CKAN Dataset Model)</u> and harvest data using the Open Archives Initiative Protocol for Metadata Harvesting (<u>OAI-PMH</u>). Other references: <u>documentation</u> and <u>mapping files</u>.
- 4. Research Data Alliance (RDA) Research Data Switchboard relies on OAI-PMH protocol (Github repository).
- 5. National Data Service.
- 6. National Institute of Health's Neuroscience Blueprint funded <u>Neuroscience Information</u> Framework (NIF).
- 7. The National Institute of Diabetes and Digestive and Kidney Diseases' NIDDK Information Network (dkNET).
- 8. <u>Data Documentation Initiative</u> Draft Specification of <u>DDI-RDF Discovery Vocabulary</u> (Disco) for the discovery of microdata sets and related metadata using RDF technologies in the Web of Linked Data (that relies on the <u>Data Cube</u>, <u>DCAT</u> and <u>XKOS</u>).
- Global Alliance for Genomic Health: specifically Data Working Group, Genotype-2-phenotype task team, NIH Office of Director funded Monarch Initiative these are all efforts to develop standardized schemas for genotype-phenotype data integration and data sharing.
- 10. <u>eTRIKS</u> <u>standards</u> <u>starter</u> <u>pack</u>, under a pre-competitive private, public Innovative Medicine Initiative aiming to bridge clinical/CDISC, <u>ISA</u> and other community-based standards.
- 11. RDA Working Group on Data Description Registry Interoperability.
- 12. EBI RDF Platform.
- 13. Content Standard for Digital Geospatial Metadata Part 1: Biological Data Profile, 1999.
- 14. Open PHACTS Discovery Platform integrating pharmacological data resources according to Dataset descriptions for the Open Pharmacological Space, based on W3C VoID for describing Linked Datasets, HCLS Dataset descriptions by the W3C Semantic Web in Health Care and Life Sciences Interest Group.
- 15. <u>Just Enough Results Model (JERM)</u> from the <u>SEEK for Science</u> project.
- 16. Metadata for data citation by Force11 Working group: <u>Achieving human and machine accessibility of cited data in scholarly publications</u>. PeerJ Computer Science, 2015.



- 17. <u>Experimental Metadata Model</u> preliminary work to model metadata about the experiments that produce datasets; collaboration between Elsevier and Oregon Health and Science University.
- 18. WHO Dataset from International Clinical Trials Registry Program.
- 19. <u>VIVO-ISF</u> linking people to scholarly products; it is being aligned and integrated with <u>SCIENCV NIH</u> biosketch system.
- 20. <u>ISO/IEC</u> <u>JTC1</u> <u>SC32</u> <u>WG2</u>: Working Group that develops international standards for metadata and related technologies.
- 21. CERIF and EuroCRIS models.
- 22. Provenance, Annotation and Versioning (PAV) ontology.
- 23. <u>Neuroimaging Data Model</u> (NIDM) a standard that models stages of neuroimaging research including experiment, workflow, and results in RDF using the W3C <u>PROV</u> and <u>HCLS Dataset Descriptors</u> recommendations.

#### 2.1.2. Metamodels

This is a non-comprehensive list of the metamodels analysed, which might have more specific scopes and different use cases than the intended capability of the DataMed prototype.

- 1. ISO/IEC 11179: Metadata Registries Part 3: Registry metamodel and basic attributes.
- 2. <u>ANSI X3.285</u>: <u>Metamodel for the Management of Shareable Data</u>: conceptual model for the specification of a data registry
- 3. DataFairport Profiles.
- 4. Research Object Ontology (based on OAI-ORE for aggregation, W3C Web Annotation Data Model for annotations and W3C PROV for provenance).
- 5. <u>Minimum Information Model ontology</u> a metamodel for describing minimum information model.

## 2.2. Top-down Use Cases

Use cases have been guiding elements throughout the process, in order to define the appropriate boundaries and level of granularity: which queries will be answered in full by the DataMed prototype, which only partially, and which are out of scope.

### 2.2.1. Extracting Competency Questions

The use cases have been: (i) collected at the <u>bioCADDIE Use Cases Workshop</u>, (ii) extracted from the <u>bioCADDIE White Paper</u>, (iii) submitted by the community, and (iv) provided by the NIH to the <u>bioCADDIE Executive Committee</u>. This section describes the methods used to analyse the use cases and derive information on the type of metadata elements needed to support them. From the use cases, a set 'competency questions' have been derived; these are defined as the questions which we want the NIH BD2K DataMed prototype to be able to provide support for.

Subsequently the questions have been abstracted, key concepts highlighted and color-coded and binned in entities, attributes and values categories, to be easily matched with the result of the 'bottom-up approach'. The questions below are grouped according to their source, using an internal code for tracking propose only.



Competency question
Search for <b>disease</b> x <b>data</b> of all <b>types</b> across <b>all databases</b> (Note: these first three use cases are linked; also there is a <b>Common Data Element</b> for the <b>disease</b> x [HD])
Search for <b>data type</b> x related to <b>disease</b> x and <b>disease</b> y to compare <b>behavioral studies</b> (HD and ADHD)
Search for <b>data</b> on <b>diseases</b> c, d, e, and f that mention <b>disease</b> x or the <b>disease</b> x gene
Search for <b>organism x</b> in <b>biological process</b> y (apoptosis) at <b>scale</b> z with an estimate of the <b>reliability of the annotations</b>
Search for new drug x to predict and track biological process y (cardiotoxicity)
Search for <b>data type</b> x ('omics correlates) of biological process for <b>drugs</b> related to <b>drug</b> x
Search for <b>data types</b> a, b, and c (EHR data, self-report, sensor) to determine <u>natural history</u> of patients given <u>drugs similar to drug</u> x
Track responses to treatment to ensure detection of biological process x
Find patient data "like these" with similar treatments, responses to treatment, genetics
Search for <b>studies</b> a-z with <b>patient data</b> with <b>biological process</b> x (e.g, obesity as measured by BMI) and <b>interventions</b> a-z. Then filter on <b>demographic</b> characteristics.
Search for <b>patient data with identifiers</b> linked to <b>data type</b> x (genome) and <b>type</b> z (fMRI) to find variants causal for <b>disease</b> x (autism)
Search for patient data with permission a, size b, demographic characteristic c, biosamples available, and data type d (e.g., imaging) available
Find Publications a-z related to dataset x
Search for studies a-z that tested drug x with agent y and agent role z
Search for data on adverse outcome x (obesity as measured by BMI) and disease y (e.g., diabetes) using standard z with license a and quality indicator b and provenance c
Search for data that was subsetted based on vaccination history
Search for <b>data</b> by <b>NIH</b> researchers with > 100 <b>publications</b> on <b>disease</b> x that were <b>peer reviewed</b>
Search for <b>data</b> that were <b>curated</b> according to <b>standard</b> x by <b>researcher</b> y or project z
Search for data that can be redistributed for free under license x
Search for substance x in groundwater to correlate with outcomes in patients with disease z family <u>history</u>



BGUC5-10	Search for patients with phenotype x and disorder y (e.g., > 4 drinks a day)
BGUC5-11	Search for <b>patients</b> with exposure to substance x correlated with biological process (mutation) in genes a-z
PB1	Search for data type x ( gene expression ) analysis on mouse red blood cells and narrow search results by access statistics
PB2	After search determine which data in result set are most relevant

SPUC1	Search for <b>birth cohort</b> x ( <b>adolescents</b> ) with combination of <b>imaging data types</b> a-z to identify <b>phenotypes</b> a-z predictive of <b>disorders</b> x and y ( <b>alcohol and drug</b> use)
SPUC2	Search for data type x (imaging data), across the lifespan, with deep phenotyping and data type y (GWS data)
SPUC3 PRE3	Search for birth cohort data that are harmonized on variable x (educational attainment) to understand <u>historical</u> impact on biological process y (adult mortality)
SPUC4	Query broader and updated phenotypic categories for generalized enrichment analysis on data type ('omics)
SPUC5	Create virtual networking environment, linking data types x and y and literature to understand biological process (molecular biology of carcinogenic pathway), which is accessible to medical professionals and patients.
SPUC6	Search for constraints of genotypes a-z and phenotypes a-z
SPUC7-1	Search for <b>EHR data</b> to monitor <b>side effects</b> of <b>drug</b> x with <b>condition/context</b> y, <b>data quality</b> z, prevalence of <b>medication</b> use, etc.
SPUC7-2	Link EHR data with knowledge bases a-z (e.g., SemMedDB, DrugBank, etc.)
SPUC7-3	Search for <b>clinical patient data</b> over the course of <b>disease</b> x to study <b>disease</b> progression, treatment change and discontinuation, outcomes, <b>condition</b> (hospital setting)
SPUC8	Search for <b>longitudinal survey data</b> on <b>disorder</b> x (e.g., tobacco use) with <b>data type</b> y (biomarkers)
SPUC9	Search for <b>patterns</b> indicative of <b>drug response</b> in the <b>genome</b> and <b>transcriptome</b> with documented <b>experimental conditions</b>
SPUC10	Search for <b>patients</b> with <b>disorder</b> x (e.g., <b>autism</b> ) and with specific <b>data type</b> (genomic, microbiome and sensor data) profiles; export to <b>big data compute platform</b> .
SPUC11	Search for <b>code snippets</b> in <b>statistical software package</b> x to <b>extract</b> or <b>combine</b> specific <b>variables</b>
SPUC12	Limit searches to <b>datasets</b> with different <b>access requirements</b> (e.g., IRB, DUA, public)
SPUC13	Search for candidate <b>genes</b> a-z associated with biological process x (aging) and validate them



SPUC14	Search for drug-drug interactions through automated extraction of structured metadata in an RDF nanopublication and cite associated paper x
SPUC15	Search for patient data from multiple clinical trials (in academia and industry, with unique IDs for each clinical trial and datasets within them) to combine them
SPUC16	Search for datasets a-z relevant to causal analysis in domains a-z for use with causal discovery algorithms
SPUC17	Search for <u>life histories</u> with <b>data type</b> x (clinical) on outcomes of biological process x (pregnancy) in women with <b>disorder</b> x (Factor 11 deficiency)
SPUC18	Search for <b>pathway</b> x that regulates at least two of the <b>genes</b> in response to <b>cell stress</b> x (e.g., UPR)
SPUC19	Search for clinical trials data with policies x and y (to study transparency)

WPUC1	Search for <b>patients</b> with <b>disease</b> x (Alzheimers) that have <b>data types</b> x, y, and z <b>available</b> (e.g., RNA-seq, behavioral, <b>imaging</b> )
WPUC2	Search for <b>data types</b> x and y related to the same biological process z
WPUC3	Search for <b>data types</b> x (genome data) with <b>biological process</b> (mutations) y and z in <b>species/organism</b> a for <b>phenotype</b> b
WPUC4	Search for <b>data elements</b> and <b>instruments</b> that measure <b>biological process</b> x (stress); use facets to find different types of <b>stressors</b>
WPUC5-p7	Search for dataset x referenced in paper y and determine if dataset x is the <u>latest</u> version
WPUC6-p7	What genes are differentially expressed in the ureteric bud vs. the mesonephric duct? (can be derived from a computation will such services be connected?)
WPUC7-p7	Search for datasets published as a result of grant x (how many?)
WPUC8-p7	Search for <b>datasets</b> produced from <b>funder</b> x (NIH) (how many?)
WPUC9-p7	Search for number of times <b>gene expression</b> x (GSE3114) has been analyzed; is it <b>available</b> in <b>format</b> y?
WPUC10-p7	Which datasets funded by funder x generated the most publications?

UC2	Search for data from author x, from database y, linked to publication z
UC15	Search MIAME standard compliant data, from database x
UC1	Search for data type $x$ (gene expression ) in human cell line $x$ , funded by funder $x$

## 2.2.2. Identify Entities Attributes and Values

The concepts highlighted in the use cases above have been binned in entities, attributes and values categories.



```
material entity
Organization/
        NIH[BGUC5-6,WPUC8-p7]
Biomaterial/
        human cell line [UC1]
organism x [BGUC2,WPUC3]
        mouse [PB1]
        human/Homo sapiens [UC1]
population/cohort [SPUC1,SPUC3]
        family [BGUC5-9]
BGUC5 [BGUC5-1]
groundwater [BGUC5-9]
red blood cells [PB1]
ureteric bud [WPUC6-p7]
mesonephric duct [WPUC6-p7]
Molecular entity/
        gene [BGUC1-3,BGU5-11,SPUC13,SPUC18,WPUC6-p7]
        protein {placeholder}
        nucleic acid{placeholder}
        metabolite {placeholder}
        chemical entity
                 drug/medication [BGUC3-1,BGUC3-2,BGUC3-3,BGUC5-3,SPUC1,SPUC7-1,SPUC14]
         instrument [WPU0
Process
Planned Process
        peer-review [BGUC5-6]
        curation [BGUC5-7]
        publishing [WPUC7-p7]
        distributing [BGUC5-8]
        imaging [SPUC1,SPUC2,WPUC1]
        referencing/citing [WPUC5-p7, SPUC14]
Study
longitudinal survey [SPUC8]
clinical trials [SPUC15,SPUC19]
intervention/experimental condition/stressor/treatment[BGUC3-4,BGUC3-5,WPUC4, SPUC9,SPUC7-1(*)]
        vaccination [BGUC5-5]
analysis/data transformation
        generalized enrichment analysis [SPUC4]
        causal analysis [SPUC16]
        harmonization [SPUC3]
        differential analysis [WPUC6-p7]
        correlation analysis [BGUC5-9,BGUC5-11]
Unplanned Process
Adverse event / Side effect [SPUC7-1]
Property
role/[BGUC5-3]
        researcher [BGUC5-7]
        author [UC2]
        funder [WPUC8-p7,WPUC10-p7, UC1]
```



```
medical professionals[SPUC5]
            [WPUC1,SPUC15,SPUC10,SPUC7-3,SPUC5,
patient
                                                               BGUC4,
                                                                             BGUC5-9,BGUC5-10,BGUC5-11,
BGUC3-3,BGUC3-5,BGUC5,BGUC5-1]
developmental stage
adolescent [SPCU1]
adult [SPUC3]
Phenotype/ [BGUC5-10,WPUC3, SPUC6,SPUC1]
        demographic characteristic [BGUC4,BGUC5-1]
        phenotypic categories [SPUC4]
Disease/ [BGUC1-1,BGUC1-2,BGUC1-3,BGUC5, BGUC5-4,BGUC5-6,BGUC-5-9,SPUC7-3,WPUC1]
        disorder [SPUC1,SPUC-8,SPUC10,SPUC17, BGUC5-10]
        obesity [BGUC5-4,BGUC4]
        autism [BGUC5]
mortality [SPUC3]
availability [BGUC5-1, SPUC5]
quality [SPUC7-1, BGUC5-4]
reliability [BGUC2]
relevance [PB2]
similarity [BGUC3-2,BGUC3-3,BGUC3-5]
compliance [UC15]
provenance [BGUC5-4]
prevalence [SPUC7-1]
Information content entity
Bioinformatic Resource
        knowledge base [SPUC7-2]
statistical software package [SPUC11]
big data compute platform [SPUC10]
pathway [SPUC5,SPUC18]?
identifier [BGUC5, SPUC14]
Publication [UC2-9]
        annotation [BGUC2]
        literature [UC27]
        paper/publication [BGUC5-2,BGUC5-6,SPUC14, UC2,WPUC10-p7]
Specification/Collection of Rules
format [WPUC9-p7]
standard [U15,BGUC5-4,BGUC5-7]
license [BGUC5-4,BGUC5-8]
policy [SPUC19]
permission [BGUC5-1]
version [WPUC5-p7]
Data/Measurement
        gene expression data [uc1]
        imaging data [SPUC1,SPUC2]
deep phenotyping and GWS data [SPUC2]
birth cohort data [SPUC3/PRE3]
genome data [BGUC5,WPUC3]
fMRI data [BGUC5]
omics data [uc26]
eHR data [uc28]
variable [SPUC3,SPUC11]
        educational attainment [SPUC3]
scale [BGUC2]
size [BGUC5-1]
Temporal interval
History [BGUC5-9, BGUC5-5, BGUC3-3, SPUC3, SPUC17, WPUC5-p7]
lifespan [SPUC2]
```



#### 2.3. Bottom-up Mapping

The generic metadata schemas and some life science-specific one that have been mapped to identify common metadata elements are in a separate <u>"Appendix IV - NIH BD2K bioCADDIE WG3 Metadata Mapping File v1.1"</u>.

#### 2.3.1. Input Material

Generic metadata schemas and some life science-specific ones have been mapped to identify common metadata elements. When available, formal representations such XML schema document (XSD) and semantic model (RDF/OWL representations) have been used as input material in the mapping process. The mapping in Appendix III covers the schemas listed below, encompassing both generics and science-specific schemas.

#### 2.3.1.1. Generic Metadata Schemas and Models

- 1. Datacite Metadata Schema
- Schema.org; <u>Dataset class</u>, a collaborative, community activity with a mission to create, maintain, and promote schemas for structured data on the Internet; used by search engines such as Google, Bing and Yahoo.
  - a. The Dataset class in Schema.org, used in this WG mapping, is based upon the W3C Data Catalog Vocabulary (DCAT) and it benefits from collaboration around the DCAT, ADMS and VoID vocabularies; details and mappings.
- 3. Dataset Descriptions: W3C HCLS Community Profile. 2015
  - a. Dataset descriptors identification file
- 4. NLM preliminary work on metadata core set
- 5. Registry Interchange Format Collections and Services (RIF-CS), used in the JISC Research Data Registry and Discovery Service
  - a. Documentation
  - b. Implementation of a profile of ISO 2146.
- 6. Project Open Data Metadata Schema v1.1

#### 2.3.1.2. Life Science Metadata Schemas

- 1. NCBI BioProject / NCBI BioSample
- 2. EMBL-EBI Pride.xsd
- 3. EMBL-EBI/NCBI Short Read Archive xsd
- 4. Nature's *Scientific Data* <u>ISA</u> <u>specification</u> and <u>ISA</u> <u>file</u> (<u>study metadata</u>) <u>as ingested in</u> the article XML
- 5. EMBL-EBI MetaboLights ISA configuration
- 6. NCBI Gene Expression Omnibus MiniML.xsd
- 7. CDISC BRIDG Model 3
- 8. CDISC SDM.xsd, which imports CDISC ODM.xsd
- 9. GA4GH metadata model



In addition, existing mapping and comparisons has also been reviewed and considered:

- 1. An initial comparison by bioCADDIE Development Team's members.
- 2. <u>linkedISA experimental metadata mapped to OBO Foundry OBI and the Semantic science Integrated Ontology (SIO)</u>

### 2.3.2. BioSharing Collection of Schemas/Models

The metadata schemas and models used in the mapping have been described in the <u>BioSharing Collection for bioCADDIE</u>, which will be enriched progressively; the information includes:

- creators and maintainers;
- documentation, including URL where this is located;

#### and when available

- version;
- source of metadata elements (e.g. XSD), including the URL where the model or schema has been sourced.