NIH BD2K bioCADDIE Data Discovery Index WG3 Metadata Specification v1

Status:

Version 1 - 12th August 2015

Working Group Goals:

The NIH BD2K bioCADDIE Metadata Working Group (WG) 3 is tasked to define a set of metadata specifications that support intended capability of the NIH BD2K Data Discovery Index prototype - as outlined in the bioCADDIE White Paper. This is a joint activity between bioCADDIE and CEDAR, another BD2K centre focused on metadata. This joint Metadata WG3 operates in two phases to produce a 'core metadata' (phase 1), designed to be future-proofed for progressive extensions to accommodate 'domain-specific metadata' for more specialized data types (phase 2), as needed.

Scope of the Document:

This document describes the process, the material reviewed (section 1 and Appendix I) and the use cases (section 2) used to identify an initial set of metadata elements and create a JSON schemata (section 3 and Appendix II); this work will be iteratively reviewed, following a test phase.

Associated Material:

This document, its appendixes and the associated code are available from the <u>bioCADDIE</u> <u>Github repository</u> and the <u>bioCADDIE</u> <u>Metadata WG3</u> webpage; the latter also serves the original working files, the presentations and notes from the WG3 activities. The schemas and models in Appendix I are also listed in the <u>BioSharing Collection for bioCADDIE</u>.

Intended Audience:

This document is primarily aimed at the <u>bioCADDIE</u> <u>Core</u> <u>Development</u> <u>Team</u> that will implement and test this model; however this is also informative for other parties such as prospective data sources for the Data Discovery Index and developers of data harvesting and other tools.

Contact:

Questions and comments to Susanna-Assunta Sansone (WG3 chair) and WG3 members - adding them directly to the <u>live Google document</u> or sending them to <u>biocaddie[at]ucsd.edu</u>.

Table of Content:

1. Standard Operating Procedure

- 1.1. Combined Approaches
 - 1.1.1. Data Discovery Initiatives and Metadata Initiatives
 - 1.1.2. Metamodels
- 1.2. Top-down Use Cases
- 1.3. Bottom-up Mapping
 - 1.3.1. Input Material
 - 1.3.1.1. Generic Metadata Schemas and Models
 - 1.3.1.2. Life Science Metadata Schemas
 - 1.3.2. BioSharing Collection of Schemas and Models
- 1.4. Phase 1 Outputs, Evaluation and Next Steps

2. Use Cases and Derived Metadata

- 2.1. Methodology
 - 2.1.1. Competency Questions
 - 2.1.2. Entities Attributes and Values

3. Initial Set of Metadata Elements

- 3.1. Overview and General Considerations
- 3.2. JSON Schemas
- 3.3. Detailed Description
 - 3.3.1. Cardinality and Requirement Level
 - 3.3.2. Links to Use Cases and Schemas/Models
- 4. Appendix I Metadata Mapping File
- 5. Appendix II Metadata Elements File

1. Standard Operating Procedure

Authors: Alejandra Gonzalez-Beltran¹, Philippe Rocca-Serra¹, Susanna-Assunta Sansone¹ and WG3 members.

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

1.1. 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 (section 1.1.1). Several metamodels for representing metadata also exist and have been reviewed (section 1.1.2). 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 1.2); and
- a mapping of existing metadata schemas (bottom-up approach; section 1.3 and Appendix I).

1.1.1. Data Discovery Initiatives and Metadata Initiatives

This is 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 NIH BD2K Data Discovery Index prototype.

- UK <u>JISC Research Data Registry and Discovery Service</u>: relies on Registry Interchange Format Collections and Services (<u>RIF-CS</u>); related documentations: <u>Github repository</u>, <u>WP3: Metadata Development and Standardisation</u>, <u>Report: metadata mapping schemes</u> / recommendations (version 9, 2014-05-09).
- 2. Datacite Metadata Search 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> <u>Framework (NIF).</u>
- 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>).

3

¹ University of Oxford, Oxford e-Research Centre, UK; bioCADDIE and CEDAR partner.

- 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. <u>Open PHACTS</u> Discovery Platform integrating pharmacological data resources according to <u>Dataset descriptions for the Open Pharmacological Space</u>, based on W3C <u>VolD</u> for describing Linked Datasets, <u>HCLS Dataset descriptions</u> by the W3C Semantic Web in Health Care and Life Sciences Interest Group.
- 15. Just Enough Results Model (JERM) from the SEEK for Science 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.

1.1.2. 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 NIH BD2K Data Discovery Index prototype.

- 1. <u>ISO/IEC 11179: Metadata Registries</u> 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.

1.2. 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 NIH BD2K Data Discovery Index prototype, which only partially, and which are out of scope. From a selection of competency questions derived from different sources key metadata elements have been highlighted and color-coded to be easily matched with the metadata resulting from the bottom-up mapping approach, described below. This top-down approach is detailed in section 2.

1.3. Bottom-up Mapping

1.3.1. Input Material

Generic metadata schemas and some life science-specific one 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 is available as Appendix I - NIH BD2K bioCADDIE WG3 Metadata Mapping v1 (section 4). The mapping for this phase 1 covers the schemas listed below; additional life science-specific schemas will be considered in phase 2, when domain-specific metadata for more specialized data types are tackled.

1.3.1.1. 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 <u>ISO 2146</u>.

1.3.1.2. 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 <u>ISA configuration</u>

- 6. NCBI Gene Expression Omnibus MiniML.xsd
- 7. CDISC BRIDG Model 3
- 8. <u>CDISC SDM.xsd</u>, which imports CDISC <u>ODM.xsd</u>
- 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</u> mapped to OBO Foundry OBI and the <u>Semantic science Integrated Ontology (SIO)</u>

1.3.2. BioSharing Collection of Schemas and 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.

1.4. Phase 1 Outputs, Evaluation and Next Steps

The approach described in the sections above has delivered an initial set of metadata elements, which have also been ranked and provisionally associated with a requirement level, as per the RFC 2119; the metadata model is available as machine readable JSON schemata. These outputs are described in section 3.

To evaluate this initial set of metadata elements, the work will continue as part of the bioCADDIE Core Development Team: the metadata elements will formally be represented - adopting the FAIR principles - and tested with several data sources. This initial set of metadata elements will be iteratively reviewed, as needed, and domain specific extensions identified as part of the phase 2 activities of the WG3.

2. Use Cases and Derived Metadata

Authors: Philippe Rocca-Serra¹, Mary Vardigan² and WG3 members.

As outlined in section 1, the analysis of the use cases is referred to as top-down approach that combined to the bottom-up approach (section 1.3 and Appendix I) - has been used to identify the initiate set of metadata elements. The use cases have been: (i) collected at the bioCADDIE Use Cases Workshop, (ii) extracted from the bioCADDIE White Paper, (iii) submitted by the community, and (iv) provided by the NIH to the bioCADDIE Executive Committee. This section

6

² ICPSR, University of Michigan, USA; bioCADDIE partner.

describes the methods used to analyse the use cases and derive information on the type of metadata elements needed to support them.

2.1. Methodology

From the use cases, a set 'competency questions' have been derived; these are defined as the questions which we want the NIH BD2K Data Discovery Index 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'.

2.1.1. Competency Questions

The questions below are grouped according to their source, using an internal code for tracking propose only.

propose only.	
Internal bioCADDIE code	Competency question
BGUC1-1	Search for disease x data of all types across all databases (Note: these first three use cases are linked; also there is a Common Data Element for the disease x [HD])
BGUC1-2	Search for data type x related to disease x and disease y to compare behavioral studies (HD and ADHD)
BGUC1-3	Search for data on diseases c, d, e, and f that mention disease x or the disease x gene
BGUC2	Search for organism x in biological process y (apoptosis) at scale z with an estimate of the reliability of the annotations
BGUC3-1	Search for new drug x to predict and track biological process y (cardiotoxicity)
BGUC3-2	Search for data type x ('omics correlates) of biological process for drugs related to drug x
BGUC3-3	Search for data types 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
BGUC3-4	Track responses to treatment to ensure detection of biological process x
BGUC3-5	Find patient data "like these" with similar treatments, responses to treatment, genetics
BGUC4	Search for studies a-z with patient data with biological process x (e.g, obesity as measured by BMI) and interventions a-z. Then filter on demographic characteristics.
BGUC5	Search for patient data with identifiers linked to data type x (genome) and type z (fMRI) to find variants causal for disease x (autism)
BGUC5-1	Search for patient data with permission a, size b, demographic characteristic c, biosamples available, and data type d (e.g., imaging) available
BGUC5-2	Find Publications a-z related to dataset x
BGUC5-3	Search for studies a-z that tested drug x with agent y and agent role z

BGUC5-4	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
BGUC5-5	Search for data that was subsetted based on vaccination <u>history</u>
BGUC5-6	Search for data by NIH researchers with > 100 publications on disease x that were peer reviewed
BGUC5-7	Search for data that were curated according to standard x by researcher y or project z
BGUC5-8	Search for data that can be redistributed for free under license x
BGUC5-9	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 patients 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 birth cohort x (adolescents) with combination of imaging data types a-z to identify phenotypes a-z predictive of disorders x and y (alcohol and drug use)
SPUC2	Search for data type x (imaging data), <u>across</u> <u>the lifespan</u> , 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 EHR data to monitor side effects of drug x with condition/context y, data quality z, prevalence of medication use , etc.
SPUC7-2	Link EHR data with knowledge bases a-z (e.g., SemMedDB, DrugBank, etc.)
SPUC7-3	Search for clinical patient data over the course of disease x to study disease progression, treatment change and discontinuation, outcomes, condition (hospital setting)
SPUC8	Search for longitudinal survey data on disorder x (e.g., tobacco use) with data type y (biomarkers)

SPUC9	Search for patterns indicative of drug response in the genome and transcriptome with documented experimental conditions
SPUC10	Search for patients with disorder x (e.g., autism) and with specific data type (genomic, microbiome and sensor data) profiles; export to big data compute platform .
SPUC11	Search for code snippets in statistical software package x to extract or combine specific variables
SPUC12	Limit searches to datasets with different access requirements (e.g., IRB, DUA, public)
SPUC13	Search for candidate genes 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 data type x (clinical) on outcomes of biological process x (pregnancy) in women with disorder x (Factor 11 deficiency)
SPUC18	Search for pathway x that regulates at least two of the genes in response to cell stress x (e.g., UPR)
SPUC19	Search for clinical trials data with policies x and y (to study transparency)

WPUC1	Search for patients with disease x (Alzheimers) that have data types x, y, and z available (e.g., RNA-seq, behavioral, imaging)
WPUC2	Search for data types x and y related to the same biological process z
WPUC3	Search for data types x (genome data) with biological process (mutations) y and z in species/organism a for phenotype b
WPUC4	Search for data elements and instruments that measure biological process x (stress); use facets to find different types of stressors
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 datasets produced from funder x (NIH) (how many?)
WPUC9-p7	Search for number of times gene expression x (GSE3114) has been analyzed; is it available in format 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				

2.1.2. 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 [WPUC4]
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
Iongitudinal 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]
patient
            [WPUC1,SPUC15,SPUC10,SPUC7-3,SPUC5,
                                                                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,SPUC17,WPUC5-p7]
lifespan [SPUC2]

3. Initial Set of Metadata Elements

Authors: Alejandra Gonzalez-Beltran¹, Philippe Rocca-Serra¹, Susanna-Assunta Sansone¹ and WG3 members.

This section describes the initial set of metadata elements derived from the bottom-up approach - as they were present in more than one of the schemas analysed (section 1.3 and Appendix I) - and the use cases-driven top-down approach (section 2). An high level overview of the key metadata elements and and their relations is provided (section 3.1), along with a detailed description of each entity (section 3.2 and Appendix II).

3.1. Overview and General Considerations

A schematic overview of some of the metadata elements, their types and relations is shown in the Figure 1. All elements, along with their definition, relations and further details are described in section 3.3. This overview also illustrates that the model is designed around the *Dataset* object. This entity is also linked to other digital research objects part of the NIH Commons, such as *Software* and *Data Standard*, which are the focus on other discovery indexes and therefore 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, 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 competency questions can be answered in full and that these may not be representative of all kind of data sources the NIH BD2K Data Discovery Index prototype should retrieve information from. This initial model will be implemented by the bioCADDIE Core Development Team and tested with a variety of sources. Subsequently, it will be iteratively reviewed (modified, simplified and/or enriched) as needed, as part of the phase 2 activities of the WG3.

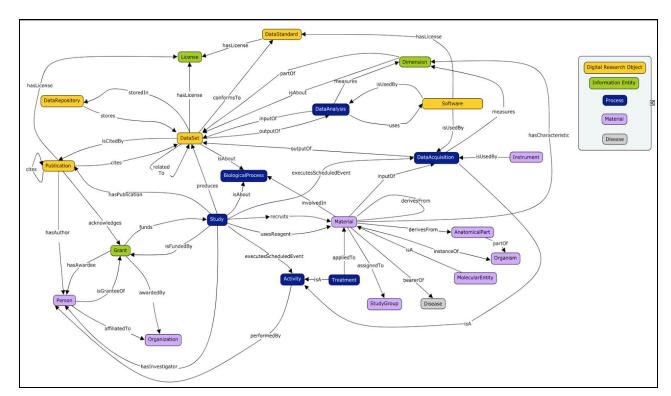


Figure 1. A schematic overview of some of the metadata elements, their types and relations.

3.2. JSON Schemas

The metadata elements are also available as machine readable JSON schemata from bioCADDIE Github repository, along with example JSON files.

3.3. Detailed Description

A full description of the metadata elements, grouped by types and color coded (to match Figure 1) in provided in Table 1, which is also available as a separate: Appendix II - NIH BD2K BioCADDIE WG3 Metadata Elements File v1 (section 5); the <u>Google JSON</u> style guide 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); others are detailed below.

3.3.1. Cardinality and Requirement Level

Cardinality restrictions indicate the number of valid occurrences for an attribute; the RFC 2119
requirement levels indicates if a particular entity MUST/MUST NOT/SHOULD/SHOULD
NOT/MAY be present, i.e. if it is compulsory, recommended, optional. 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. The requirement levels will be iteratively reviewed and used to evaluate if a data/database source

'complies' to the NIH BD2K Data Discovery Index prototype's metadata, and therefore if it has the potential to fulfil the relevant competency questions.

3.3.2. Links to Use Cases and Schemas/Models

The metadata elements are associated to relevant use cases and competency questions (section 2.1.1). In Appendix II, for those entities and/or properties were no specific competency questions are indicated, links to the relevant schema(s)/model(s) are also provided, to justify their relevance and provenance.

Table 1. A full description of the metadata elements grouped by types and color coded to match Figure 1.

ENTITY	PROPERTY	DEFINITION	TYPE	CARDINALITY	REQUIREM ENT LEVEL	COMPETENCY QUESTION
Dataset		A set of dimensions about an entity being observed. A collection of data, published or curated by a single agent, and available for access or download in one or more formats (from DCAT: http://www.w3.org/TR/vocabdcat/#Class:_Dataset) A body of structured information describing some topic(s) of interest (from: http://schema.org/Dataset)				BGUC5-2;BGUC5-4; BGUC5-5;UC2;UC15; WPUC5-p7;WPUC7-p 7;WPUC8-p7;WPUC1 0-p7
	identifier	a code uniquely identifying the dataset	IRI	0n	SHOULD	BGUC5
	identifierSche me	if an identifier is supplied, the identifier scheme represent information about how identifiers are formed, maintained and minted	string	1, if identifier is available	(MUST)	
	title	one sentence, usually short description of an entity, here the dataset	string	1	MUST	
	description	a textual narrative, comprised of one or more statements describing the dataset	string	01	SHOULD	_
	dataType	a term, ideally from a controlled terminology, identifying the nature of the data, placing it in a typology	string or IRI	1n	MUST	BGUC1-1;BGUC1-2; BGUC3-2;BGUC3-3; BGUC5;BGUC5-1;W PUC1;WPUC2;WPUC 3;WPUC9-p7;UC1
	conformsTo	a property used to indicate if the dataset meets the requirements and constraints	DataStandard	0n	MAY	BGUC5-7;WPUC9-p7

DataStandard		A format, reporting				BGUC5-7;UC15;WPU
	relatedDataset	a property to indicate if the dataset relies on other	Dataset	0n	MAY	
	size	a property used to specify the size of the dataset	number	01	MAY	BGUC5-1
	isAboutBiolog ical Process	a property used to specify the biological processes relevant to the study, ideally from a controlled vocabulary/ontology	Biological Process	0n	SHOULD	
	version	an optional property used to specify a release point for the dataset when applicable	string	01	SHOULD	WPUC5-p7
	dateType	a property qualifier to the 'data' attribute. The type of date, used to specify the process which is being timestamped by the date attribute value, ideally comes from a controlled terminology.	string or IRI	1, if date is available	(MUST)	
	date	a property used to specify any date relevant to the dataset	date	1n	MUST	
	creator	a property to list the person(s) or organization(s) which contributed to the creation of the dataset.	Person or Organization	1n	MUST	UC2
	producedBy	a property used to associate a study process which generated a given dataset, if any	Study	01	SHOULD	
	downloadURL	a property used to specify a URL where the dataset can be obtained	URL	0n	SHOULD	
	license	a property used to specify relevant terms of usage and license	License	0n	SHOULD	BGUC5-1;BGUC5-4; BGUC5-8
	isCitedBy	a property used to specify links to relevant publication(s)	Publication	0n	MAY	BGUC5-2
	hasPartDimen sion	a property used to identify, specify and list the different dimensions (granular components) making up a dataset.	Dimension	On	MAY	BGUC2;BGUC5-4
	storedin	a property used to specify the data repository(ies) hosting the dataset	DataRepositor y	0n	MAY	BGUC1-1;UC2
		defined by specific community data standard				

	identifier identifierSche me	used to indicate whether the dataset conforms to a particular community norm or specification. a code uniquely identifying an entity locally to a system or globally if an identifier is supplied, the identifier scheme represent information about how identifiers are formed,	IRI string	0n 1, if identifier is available	SHOULD (MUST)	
		maintained and minted			NUICE	
	name .	the name of the standard	string	1	MUST	
	homepage	URL of the homepage of the standard	URI	1	MUST	
	dataStandardT ype	a descriptor (ideally from a controlled vocabulary) providing information about the nature of the information resource	string or IRI	1	MUST	WPUC9-p7
	license	a property used to specify to the terms of use of the standard	License	0n	SHOULD	BGUC5-4
	version	an optional property used to specify a release point for the repository when applicable	string	01	SHOULD	
DataRepository		A repository or catalog of dataset				BGUC1-1;UC2;UC15
	identifier	a code uniquely identifying the data repository	IRI	0n	SHOULD	
	identifierSche me	if an identifier is supplied, the identifier scheme represent information about how identifiers are formed, maintained and minted	string	1, if identifier is available	(MUST)	
	name	the name of the data repository	string	1	MUST	BGUC1-1;UC2
	homepage	URL of the homepage of the data repository	URI	1	MUST	
	dataRepositor yDataType	a descriptor (ideally from a controlled vocabulary) providing information about the nature of the datasets in the repository	string or IRI	0n	1n	SPUC1;SPUC7-2
	license	a property used to specify to the terms of use of the standard	License	0n	SHOULD	BGUC5-4

	version	an optional property used to specify a release point for the repository when applicable	string	01	SHOULD	
	creator	a property used to specify the person(s) or organization(s) responsible for the repository	Person or Organization	1n	MUST	
Software		A digital entity containing sets of instructions and operation, which allows computation and operation of and by computer				SPUC11;SPUC10
	identifier	a code uniquely identifying an entity locally to a system or globally	IRI	0n	SHOULD	
	identifierSche me	if an identifier is supplied, the identifier scheme represent information about how identifiers are formed, maintained and minted	string	1	(MUST)	
	name	a property to specify the name of the software program	string	1	MUST	
	license	a property used to specify to the terms of use of the software	License	0n	SHOULD	
	version	an optional property used to specify a release point for the software when applicable	string	01	SHOULD	
Publication		A digital document made available by a publisher.				BGUC5-2;WPUC5-p7 ;WPUC10-p7;UC2
	identifier	a code uniquely identifying an entity locally to a system or globally.	IRI	0n	SHOULD	BGUC5-6;BGUC5-2;S PUC14;SPUC5;UC2
	identifierSche me	if an identifier is supplied, the identifier scheme represent information about how identifiers are formed, maintained and minted	string	1, if identifier is available	(MUST)	
	publicationTy pe	type of publication type, delegated to an external vocabulary/resource	string or IRI	01	SHOULD	
	title	a property to specify the title of the publication	string	1	MUST	
	date	a property to specify the date of the publication	date-time	1	MUST	
	author		Person or Organization	1n	MUST	BGUC5-6

		a property to specify the				
		person(s) responsible for the publication				
	acknowledges	a property to specify the grant(s) which funded and supported the work reported by the publication	Grant	0n	SHOULD	
	cites	a property to specify a reference to a dataset or publication	Dataset or Publication	0n	MAY	BGUC5-2
	license	a property used to specify to the terms of use of the publication	License	0n	SHOULD	
Grant		An allocated sum of funds given by a government or other organization for a particular purpose				BGUC5-6
	identifier	a code uniquely identifying an entity locally to a system or globally.	IRI	0n	SHOULD	
	identifierSche me	if an identifier is supplied, the identifier scheme represent information about how identifiers are formed, maintained and minted	string	1, if identifier is available	(MUST)	
	name	a property to specify the name of the grant and its funding program	string	1	MUST	
	funds	a property to specify the study (or dataset) supported by the grant	Study (or Dataset)	0n	SHOULD	
	funder	a property to specify the organization(s) which has awarded the funds supporting the project	(Person or Organization) and role funder	1n	MUST	BGUC5-6;WPUC7-p7 ;WPUC8-p7;WPUC10 -p7;UC1
	awardee	a property to specify the person(s) or organization(s) which received the funds supporting the project	Person or Organization	0n	SHOULD	
License		A legal document giving official permission to do something with a Resource. It is assumed that an external vocabulary will describe with sufficient granularity the permission for redistribution, modification, derivation,				BGUC5-4,BGUC5-8

		reuse, etc. and conditions for citation/acknowledgment.				
	identifier	a code uniquely identifying an entity locally to a system or globally.	IRI	0n	SHOULD	
	identifierSche me	if an identifier is supplied, the identifier scheme represent information about how identifiers are formed, maintained and minted	string	1, if identifier is available	(MUST)	
	name	a property to specify the name of the license	string	1	MUST	
	version	an optional property used to specify the version of the license	string	01	SHOULD	
	creator	a property to specify the person(s) or organization(s) responsible for writing the license	Person or Organization	0n	SHOULD	
Dimension		A feature of an entity, i.e. an individual measurable property (both quantitative or qualitative) of the entity being observed				BGUC2;BGUC4;BGU C5-1;BGUC5-4;PB1
	identifier	a code uniquely identifying the dimension	IRI	01	SHOULD	
	identifierSche me	if an identifier is supplied, the identifier scheme represent information about how identifiers are formed, maintained and minted	string	1, if identifier is available	(MUST)	
	name	the name of the dimension measured or observed during the data acquisition process (e.g. signal intensity, standard deviation), ideally from a controlled terminology	string or IRI	1	MUST	BGUC5-10,WPUC3, SPUC6,SPUC1
	dimensionTyp e	a term, ideally from a controlled terminology, identifying the nature of the dimension, placing it in a typology	string or IRI	1n	MUST	
	values	a property used to specify the actual collections of values collected for that dimension	set	0n	SHOULD	BGUC2
	unit	an optional property used to specify a reference measurement unit associated with scalar dimensions. Ideally,	string or IRI	01	MAY	

		unit should be coming from a reference controlled terminology.				
	isAbout	an optional property used to specify what the dimension is about, it could be about material (the heights of the patients) or about a dataset (the standard deviation or the set of outliers or a quality indicator of a dataset)	Dataset or Material	0n	MAY	BGUC5-4;WPUC9-p7 ;PB1
	partOf	a property used to specify the dataset(s) this dimension belongs to	Dataset	1n	MUST	
Activity	Superclass for Study, DataAnalysis, DataAcquisitio n	A type of process scheduled in a study				BGUC5-5;BGUC5-7; BGUC5-6
	identifier	a code uniquely identifying an entity locally to a system or globally	IRI	0n	SHOULD	
	identifierSche me	if an identifier is supplied, the identifier scheme represent information about how identifiers are formed, maintained and minted	string	1, if identifier is available	(MUST)	
	title	a property to specify the title/name of the process	string	1	MUST	
	startDate	a timestamp to record the starting point of the process	date	01	SHOULD	
	endDate	a timestamp to record the ending point of the process	date	01	SHOULD	
	duration	a property to specify the duration of the activity	string	01	MAY	
	location	a property to specify where the process is performed	string or IRI	01	SHOULD	
	performedBy	a property to specify the person(s) or organisation(s) responsible for executing the process	Person or Organization	0n	SHOULD	BGUC5-7
Study	(subclassOf Activity)	Process to acquire data on a sample and attempt to draw conclusions about the population the sample has been selected from, executing a plan and design				BGUC4;BGUC5-3

identifier	a code uniquely identifying an entity locally to a system or globally.	IRI	0n	SHOULD	SPUC8; BGUC5-3; BGUC4;SPUC15;SPU C19
identifierSche me	if an identifier is supplied, the identifier scheme represent information about how identifiers are formed, maintained and minted	string	1, if identifier is available	(MUST)	BGUC1-2
title	one sentence, usually short description of an entity, here the study	string	1	MUST	
schedulesActi vity	a property used to specify the activities scheduled by the study	Activity	0n	SHOULD	
schedulesDat aAcquisition	a property used to specific the kind of techniques and response variables used during a study for acquiring signal	DataAcquisitio n	1n	MUST	
studyType	a property used to specify the type of study, e.g. intervention or observation or meta-analysis	string or IRI	1	MUST	BGUC1-2;SPUC19
hasStudyGrou p	a property used to list the different study groups associated with a study	StudyGroup	0n	MAY	
recruits	a property used to specify which materials are the subjects and participants of the study	Material	0n	MAY	BGUC4
usesReagent	a property used to specify which materials are used as reagents (but not subjects) of the study	Material	0n	MAY	
isAboutBiolog ical Process	a property used to specify the biological processes relevant to the study (ideally from a controlled vocabulary/ontology)	Biological Process	0n	SHOULD	BGUC2;BGUC4
startDate	a timestamp to record the starting point of the process	date	01	SHOULD	
endDate	a timestamp to record the ending point of the process	date	01	SHOULD	
duration	a property to specify the duration of the activity	string	01	MAY	
location	a property to specify where the process is performed.	string or IRI	01	SHOULD	
performedBy	a property to specify the person(s) or organisation(s) responsible for executing the process	Person or Organization	0n	SHOULD	

	keywords	a property used to provide a list of terms providing insights into the main topic and feature of the study	string or IRI	0n	MAY	
	resultsin	a property used to specify the collection of data during assays over the course of a study	Dataset	1n	MUST	
Treatment	(subclassOf Activity)	Process, part of a study, consisting in exposing participants to the study to different conditions or group those participants into different categories based on specific criteria and compare their outcomes				BGUC3-4;BGUC3-5; BGUC4;BGUC5-3;BG UC5-11;SPUC8;SPU C9;BGUC5-10
	identifier	a code uniquely identifying an entity locally to a system or globally	IRI	0n	SHOULD	
	identifierSche me	if an identifier is supplied, the identifier scheme represent information about how identifiers are formed, maintained and minted	string	1, if identifier is available	(MUST)	
	agent	a property used to specify the nature of the perturbation or intervention used in the study	MolecularEntit y;Material;Acti vity;IRI;string	01	SHOULD	BGUC3-1;BGUC3-3; BGUC5-3
	intensity	a property used to specify how acute the perturbation is	string if qualitative, double if quantitative	01	SHOULD	
	title	one sentence, usually short description of the study	string	1	MUST	
	startDate	a timestamp to record the starting point of the process	date	01	SHOULD	
	endDate	a timestamp to record the ending point of the process	date	01	SHOULD	
	duration	a property to specify the duration of the activity	string	01	MAY	
	location	a property to specify where the process is performed	string or IRI	01	SHOULD	
	performedBy	a property to specify the person(s) or organisation(s) responsible for executing the process	Person or Organization	0n	SHOULD	
	concomitance	a property used to specify if more that one perturbations are applied at the same time to the same subject	boolean	01	MAY	

	order	a property used to specify the rank in which perturbations are being applied to study subjects	integer	01	MAY	
DataAcquisition	(subclassOf Activity)	Process of generating data through measurement made with specific techniques				
	identifier	a code uniquely identifying an entity locally to a system or globally	IRI	0n	SHOULD	
	identifierSche me	if an identifier is supplied, the identifier scheme represent information about how identifiers are formed, maintained and minted	string	1, if identifier is available	(MUST)	
	title	a property to specify the name of the technology used to acquire signal	string	1	MUST	
	uses	a property to specify the instrument(s) used to acquire the signal	Instrument or Software	0n	MAY	
	startDate	a timestamp to record the starting point of the process	date	01	SHOULD	
	endDate	a timestamp to record the ending point of the process	date	01	SHOULD	
	duration	a property to specify the duration of the activity	string	01	MAY	
	location	a property to specify where the process is performed	string or IRI	01	SHOULD	
	performedBy	a property to specify the person(s) or organisation(s) responsible for executing the process	Person or Organisation	0n	SHOULD	
	measures	a property to specify the dimension(s) being acquired as signal	Dimension	1n	MUST	BGUC2
Data American	(autala Of					ODUCA ODUCAC VITU
DataAnalysis	(subclassOf Activity)	Process of transforming data and producing data				SPUC4,SPUC16,WPU C6-p7,BGUC5-9,BGU C5-11;WPUC9-p7
	identifier	a code uniquely identifying an entity locally to a system or globally	IRI	0n	SHOULD	
	identifierSche me	if an identifier is supplied, the identifier scheme represent information about how identifiers are formed, maintained and minted	string	1, if identifier is available	(MUST)	

	1					
	title	a property to specify the name of the analysis being performed	string	1	MUST	
	startDate	a timestamp to record the starting point of the process	date	01	SHOULD	
	endDate	a timestamp to record the ending point of the process	date	01	SHOULD	
	duration	a property to specify the duration of the activity	string	01	MAY	
	location	a property to specify where the process is performed	string or IRI	01	SHOULD	
	uses	a property to specify any software package that may have been using in the data analysis process	Software	0n	MAY	
	input	a property to specify the dataset used as input	Dataset	1n	MUST	
	output	a property to specify the dataset resulting from applying the technology	Dataset	1n	MUST	
Biological Process		A biological process is a recognized series of events or molecular functions (from: http://geneontology.org/page/biological-process-ontology-guidelines)				BGUC2;BGUC3-1;BG UC3-2;BGUC3-4;BG UC5-11;SPUC13;WP UC2;WPUC3;WPUC4
	identifier	a code uniquely identifying an entity locally to a system or globally	IRI	0n	SHOULD	
	identifierSche me	if an identifier is supplied, the identifier scheme represent information about how identifiers are formed, maintained and minted	string	1, if identifier is available	(MUST)	
	name	a property to specify the name of the biological process	string	1	MUST	BGUC2
Material		A physical entity, part of collection or used in a study				BGUC3-3;BGUC3-5; BGUC5;BGUC5-1;BG UC5-9;BGUC5-11;PB 1;SPUC13;WPUC6-p 7
	identifier	a code uniquely identifying an entity locally to a system or globally	IRI	0n	SHOULD	
	identifierSche me	if an identifier is supplied, the identifier scheme represent	string	1, if identifier is available	(MUST)	

		information about how				
		identifiers are formed, maintained and minted				
	name	a property to specify the name of the material	string	1	MUST	
	derivesFrom	a property to indicate that this material originated from another material	Material	0n	MAY	
	anatomicalPar t	a property to indicate that this material originated from an anatomical part (ideally from a controlled vocabulary/ontology)	AnatomicalPar t	0n	MAY	
	bearerOfDisea se	a property to specify the pathology affecting the material used in the study or refered to in the dataset (ideally from a controlled vocabulary/ontology)	Disease	0n	MAY	BGUC1-1;BGUC1-2; BGUC1-3;BGUC5,BG UC5-4,BGUC5-6,BGU C5-8,BGUC-5-9,SPU C7-3,WPUC1
	instanceOfOrg anism	a property to specify the organism this is an instance of - when the whole organism is considered and participates in the study (ideally from a controlled vocabulary/ontology)	Organism	0n	MAY	BGUC2
	derivesFrom Organism	a property to specify the organism this material derives from - for instance when samples are taken from an organism (ideally from a controlled vocabulary/ontology)	Organism	0n	MAY	BGUC2
	involvedInBiol ogicalProcess	a property to specify that the material is involved in a particular biological process (ideally from a controlled vocabulary/ontology)	Biological Process	0n	MAY	BGUC2;BGUC3-1;BG UC3-2;BGUC4;SPUC 18
i	inputOf	a property to specify the pathology affected the material used in the study or refered to in the dataset	DataAcquisitio n	0n	MAY	
	hasCharacteri stic	a property to specify the characteristic information denoting the material	Dimension	0n	MAY	BGUC2
	role	a property to specify the role played by a material in a study	string or IRI	0n	SHOULD	
StudyGroup		A collection of entities known as study subjects based on a set of specified criteria and rules; synonyms: population, cohort				

	identifier	a code uniquely identifying an entity locally to a system or globally	IRI	0n	SHOULD	
	identifierSche me	if an identifier is supplied, the identifier scheme represent information about how identifiers are formed, maintained and minted	string	1, if identifier is available	(MUST)	
	name	a property to specify the name of the study group	string	1	MUST	
	size	a property to specify the number of members in the group	integer	01	MAY	
	member	a property to list references entities of type material make up a group	Material	1n	MUST	
	selectionCriter ia	a property to list the attributes of material and the values those should meet in order to be part of the group	string or IRI	0n	SHOULD	BGUC5-10;BGUC5-9; BGUC3-5;BGUC5-11; SPUC1;SPUC3;PRE3
MolecularEntity	(subclass of Material)	A physical entity of molecular scales such as proteins, nucleic acids, chemical materials. They can be abiotic, biological or synthetic origin				BGUC1-3;SPUC18;W PUC6-p7
	identifier	a code uniquely identifying an entity locally to a system or globally.	IRI	0n	SHOULD	
	identifierSche me	if an identifier is supplied, the identifier scheme represent information about how identifiers are formed, maintained and minted	string	1	(MUST)	
	name	a property to specify the name of the molecular entity	string	1	MUST	BGUC1-3;BGUC3-1; BGUC3-2;BGUC3-3; BGUC5-3;BGUC5-11
	structure	a property to specify the primary sequence of the molecular entity	string	0n	MAY	
	role	a property to specify the role played by a molecular entity in a study	string or IRI	0n	SHOULD	
Person		A human being				UC2
	identifier	a code uniquely identifying an entity locally to a system or globally	IRI	0n	SHOULD	

	identifierSche me	If an identifier is supplied, the identifier scheme represent information about how identifiers are formed, maintained and minted	string	1, if identifier is available	(MUST)	
	firstName	a string denoting the given name of a person	string	1	MUST	
	lastName	a string denoting the family name of a person	string	1	MUST	
	email	a string denoting an SMTP electronic mail address, contains @ character	email	0n	SHOULD	
	affiliation	a string or crossreference denoting a legal entity or physical entity corresponding to a business or administration	Organization	0n	SHOULD	
	role	roles assumed by a person, defined in an external resource	string or IRI	0n	SHOULD	(has_role author) BGUC5-6, UC2
Organization		Legal or physical entity corresponding to a business or administration				
	identifier	a code uniquely identifying an entity locally to a system or globally	IRI	0n	SHOULD	
	identifierSche me	If an identifier is supplied, the identifier scheme represent information about how identifiers are formed, maintained and minted	string	1, if identifier is available	(MUST)	
	name	a property to specify the name of the organization	string	1	MUST	
	abbreviation	a property to specify the shortname, abbreviation associated to the organization	string	01	MAY	
	postalAddress	a property to specify the postal, street address associated to an organization	string	01	MAY	
		A property that identifies the role of the organization (ideally from a controlled				
	role	vocabulary/ontology)	string or IRI	0n	SHOULD	UC1; SPUC5
In the second		A selection of the sele				
Instrument		A physical entity produced by humans to perform specific tasks or functions				
	identifier	A code uniquely identifying an entity locally to a system or globally	IRI	0n	SHOULD	

	identifierSche me	if an identifier is supplied, the identifier scheme represent information about how identifiers are formed, maintained and minted	string	1, if identifier is available	(MUST)	
	name	a property to specify the name of the instrument	string	1	MUST	WPUC4;SPUC10
	instrumentTyp e	a property to specify the type of the instrument/technology	string or IRI	01	SHOULD	WPUC4;SPUC10
	isUsedBy	a property to specify the activity that makes use of this instrument	DataAcquisitio n	0n	MAY	
	manufacturer	a property to specify the organisation which produced the instrument	Person or Organization	0n	MAY	
Organism		A living entity				BGUC2
	identifier	a code uniquely identifying a taxonomic information entity locally to a system or globally	IRI	0n	SHOULD	
	identifierSche me	if an identifier is supplied, the identifier scheme represent information about how identifiers are formed, maintained and minted	string	1, if identifier is available	(MUST)	
	name	a property to specify the scientific name of an organism	string	1	MUST	
AnatomicalPart		A structure that is part of a multicellular organism				
	identifier	a code uniquely identifying an anatomical entity locally to a system or globally	IRI	0n	SHOULD	
	identifierSche me	if an identifier is supplied, the identifier scheme represent information about how identifiers are formed, maintained and minted	string	1, if identifier is available	(MUST)	
	name	a property to specify the name of the anatomical entity or anatomical part	string	1	MUST	
Disease		A disorder of structure or function in a human, animal, or plant, that produces specific symptoms or that affects a specific location.				BGUC1-1,BGUC1-2,B GUC1-3,BGUC5,BGU C5-4,BGUC5-6,BGUC 5-8,BGUC-5-9,SPUC7 -3,WPUC1

identifier	a code uniquely identifying an entity locally to a system or globally.	IRI	0n	SHOULD	
identifierSche me	if an identifier is supplied, the identifier scheme represent information about how identifiers are formed, maintained and minted	string	1, if identifier is available	(MUST)	
name	a property to specify the name of the disease	string	1	MUST	

4. Appendix I

Authors: Alejandra Gonzalez-Beltran¹, Philippe Rocca-Serra¹ and WG3 members.

NIH BD2K bioCADDIE WG3 Metadata Mapping File v1, also downloadable from the bioCADDIE Github repository. The file describes the generic metadata schemas and some life science-specific one that have been mapped to identify common metadata elements.

5. Appendix II

Authors: Alejandra Gonzalez-Beltran¹, Philippe Rocca-Serra¹, Susanna-Assunta Sansone¹ and WG3 members.

NIH BD2K bioCADDIE WG3 Metadata Elements File v1, also downloadable from the bioCADDIE Github repository. The file describes the metadata elements, grouped by types, providing details on their: definition, attributes, requirements, cardinality and requirement level.