

Data Submission - Perspectives and solutions from different Gen3 systems

Gen3 Community Forum
10 July 2024

The Agenda

- Introduction
- Data Submission into the Aotearoa Genomic Data Repository - Nathalie Giraudon, Claire Rye, New Zealand eScience Infrastructure (NeSI)
- COVID-19 Data: 7 Million Records in PRC - Plamen Martinov, Open Commons Consortium
- MIDRC: Ingesting Medical Images from Multiple Contributors - Chris Meyer, Center for Translational Data Science, University of Chicago
- g3t: Gen3 Tracker – User Driven Submissions - Jordan Lee and Liam Beckman, Oregon Health and Science University
- Data Modelling in Gen3 - Joshua Harris, Australian BioCommons

Data submission into the Aotearoa Genomic Data Repository

Nathalie Giraudon, Claire Rye
New Zealand eScience Infrastructure (NeSI)

Data submission into a Gen3

A Case study of the Aotearoa Genomic Data Repository



**Nathalie Giraudon, Claire Rye
NeSI / University of Auckland**

10/11 July 2024

Agenda

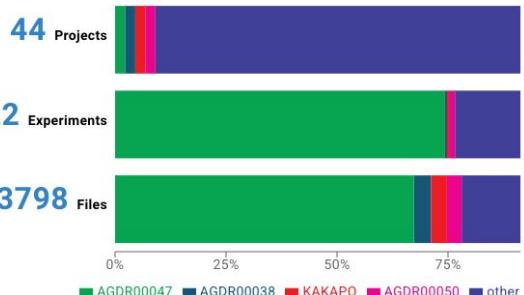
- What is AGDR (Aotearoa Genomic Data Repository)?
- Process of data submission
 - metadata spreadsheet
 - metadata validator
 - Reasoning
 - Principles
 - Ingestion
- Demonstration
- Issues
- Next steps



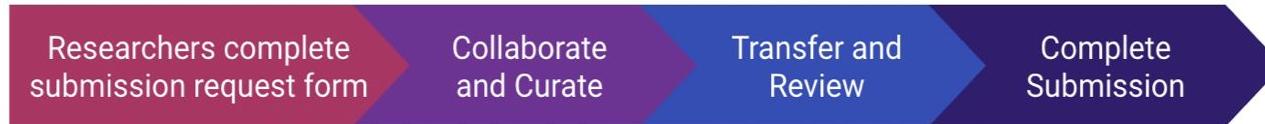
kākāpō chick photo by Dianne Mason, 2009 CC2.0

Aotearoa Genomic Data Repository

The Aotearoa Genomic Data Repository provides secure within-nation storage, management and sharing of non-human genomic data generated from biological and environmental samples originating in Aotearoa New Zealand. This resource has been developed to follow the principles of Māori Data Sovereignty, and to enable kaitiakitanga (guardianship), so that iwi, hapū and whānau (tribes, kinship groups and families) can effectively exercise their responsibilities as guardians over biological entities that are taonga (precious or treasured). While the repository is designed to facilitate the sharing of data – making it findable by researchers and interoperable with data held in other genomic



Data Submission



- Suitability assessment
- Metadata template spreadsheet
- Metadata standard, community built, MIxS compliant
- Upload data
- Metadata reviewed
- Project viewable in AGDR
- DOI provided for use in publications



Spreadsheet template

AGDR Metadata Template - 2024_03_25

File Edit View Insert Format Data Tools Extensions Help

A1:B1 Project Information

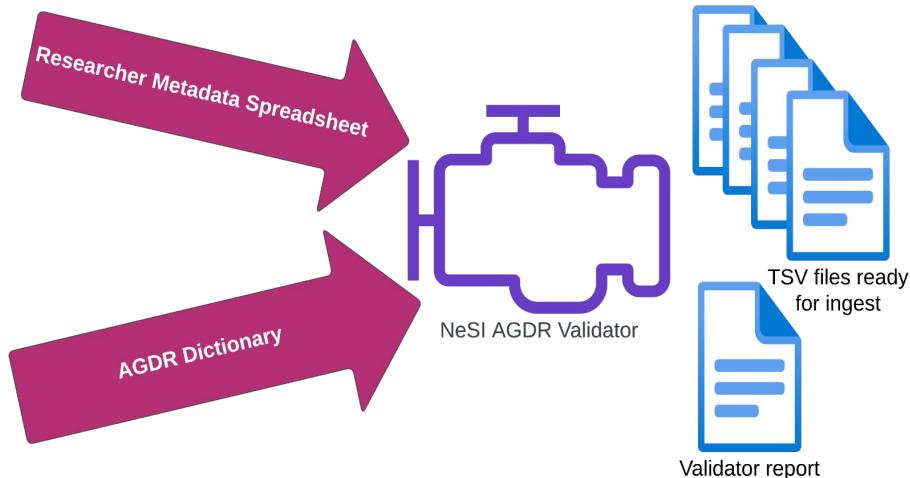
A	B	C	D	E	F
1	Project Information				
2	The research project you have been working on. If you have already submitted a Google Forms, you can skip to the next tab, but please read the instructions below first.				
3					
4	Field	name	date_collected	details	investigator_affiliation
5	Required field?	Required	Required		Required
6	Description	Name of the project	The date or date range in which the project data was collected.	More detailed description of the project.	The investigator's affiliation with respect to a research institution.
7	Example input		e.g. 1997-2000	A couple of paragraphs describing the project.	e.g. School of Biomedical Sciences, University of Otago
8	Your input				e.g. Prof Peter Dearde
9					
10	Instructions and tips				
11					
12	Please fill out this form to submit your data/metadata into Aotearoa Genomic Data Repository.				
13	This is a template, so please make a copy of this spreadsheet before submitting your input. To make a copy, press 'File' on the menu and 'Make a copy'.				
14	Once you have made a copy of this document, please fill in all the fields as much as you can under 'Your input'. Please note that there are multiple tabs which you can access via the buttons at the bottom.				
15	You can press Alt+Enter for multiline answers if needed.				
16	Once you have completed filling in the details, please remember to share your copied spreadsheet with us with claire.rye@nesi.org.nz, jun.huh@nesi.org.nz, and eirian.perkins@nesi.org.nz; and NeSI staff will help enter these data into the system.				
17	Please fill in all the 'Required' field and as many optional fields as possible. The required fields are highlighted in green.				
18					
19	Please feel free to contact us at gasupport@nesi.org.nz for any help.				
20					
21					

Metadata Validation

Reasons:

- Simplification (no need to know the dictionary, submitter_id meaning not obvious...)
- Data files are not loaded with the Gen3 client tool
- Consistency checks for the values
- Early checks before ingest

Principles:



Demonstration of the validator/ingest

- Small dataset with spreadsheet and dictionary errors
- Small dataset with no validator errors
 - Then ingest of the tsv files on our test system
- Large dataset and ingest

```
VALIDATOR VERSION: 1.2.Nat_try.2024_03_25

        Parsing AGDR spreadsheet |████| 3 in 0.6s (5.35/s)
        Loading data dictionary |██████████| 54 in 0.0s (2617.64/s)
        Building metadata graph |██████████| 100% in 1.9s (0.53/s)

PERFORMING VALIDATION...
    FILE: AGDR00057_Validation_Report_2024-07-01.txt
        Validating schema |██████████| 11/11 [100%] in 5.6s (1.86/s)
...VALIDATION COMPLETE

GENERATING TSV FILES...
    DIRECTORY: AGDR00057_TSV_Output_2024-07-01
        Writing metadata to TSVs |██████████| 2794/2794 [100%] in 0.1s (14671.91/s)
```

- Ingest of project via UI
- Problem with large datasets error
- Special characters support - issues (Excel?)
- Robustness of the validator
 - true/TRUE/'true -> can only ingest boolean in lower case...

```
- AGDR00057_bool_Validation_Report_2024-06-26.txt
1 organism [AGDR00057_BOOL_ORGANISM_17]
2   sex
3     Expecting value to be in ['male', 'female', 'unknown'], but received [UnknownN] instead
4 organism [AGDR00057_BOOL_ORGANISM_139]
5   sex
6     Expecting value to be in ['male', 'female', 'unknown'], but received [UnknownN] instead
7 organism [AGDR00057_BOOL_ORGANISM_326]
8   sex
9     Expecting value to be in ['male', 'female', 'unknown'], but received [UnknownN] instead
10 READ_GROUP
11   ERROR: no REQUIRED link found connecting parent [aliquot] link to child [read_group:EC_1_RG_R1]
12   ERROR: no REQUIRED link found connecting parent [aliquot] link to child [read_group:EC_2_RG_R1]
13   ERROR: no REQUIRED link found connecting parent [aliquot] link to child [read_group:EC_3_RG_R1]
14   ERROR: no REQUIRED link found connecting parent [aliquot] link to child [read_group:EC_4_RG_R1]
15   ERROR: no REQUIRED link found connecting parent [aliquot] link to child [read_group:EC_5_RG_R1]
16   ERROR: no REQUIRED link found connecting parent [aliquot] link to child [read_group:EC_6_RG_R1]
17   ERROR: no REQUIRED link found connecting parent [aliquot] link to child [read_group:WC_1_RG_R1]
18   ERROR: no REQUIRED link found connecting parent [aliquot] link to child [read_group:WC_2_RG_R1]
19   ERROR: no REQUIRED link found connecting parent [aliquot] link to child [read_group:WC_3_RG_R1]
20   ERROR: no REQUIRED link found connecting parent [aliquot] link to child [read_group:WC_5_RG_R1]
21   ERROR: no REQUIRED link found connecting parent [aliquot] link to child [read_group:WC_6_RG_R1]
22   ERROR: no REQUIRED link found connecting parent [aliquot] link to child [read_group:EC1_1_RG_R1]
23 read_group [EC1_1_RG_R1]
24   library_selection
25     Expecting value to be in ['Affinity Enrichment', 'Hybrid Selection', 'Other', 'PCR', 'Poly-T Enrichment',
26   ERROR: no REQUIRED link found connecting parent [aliquot] link to child [read_group:EC1_10_RG_R1]
```

Next steps

- Update of Elastic Search/Etlmapping versions
- Last validator improvements before the first release
 - Support of multi links
- Validator release and training with the researchers
- Automation of the ingest via API

≡ AGDR00056_2_Validation_Report_2024-06-21.txt

1	NO ERRORS DETECTED
2	



**genomics
aotearoa**



**MINISTRY OF BUSINESS,
INNOVATION & EMPLOYMENT**
HĪKINA WHAKATUTUKI



NeSI
New Zealand eScience
Infrastructure

Thank you!

- GA Team:
 - Bioinformatics project: Libby Liggins (Massey), Rudi Brauning (AgResearch), Mik Black (Otago), Tracey Godfrey (Otago), Tanis Goodwin (Otago)
- NeSI team (Auckland):
 - Jun Huh, Eirian Perkins, Claire Rye, Nathalie Giraudon, Rui Chen (Carvin)
- Former team members:
 - Miles Benton (ESR/Oxford Nanopore), Ben Te Aika (Otago), Ben Curran (Auckland), Brian Flaherty, Thomas Berger, Kenny Zhao (NeSI).

COVID-19 Data: Case Study of submitting 7 Million Subjects Health Data in PRC

Plamen Martinov
Mikisha Patel
Urvi Sheth
Open Commons Consortium



Plamen Martinov,
Chief Technology and Information Security
Officer

A Collaborative Data Ecosystem to Improve Outcomes for COVID-19

- a. COVID19 shut the world down
- b. OCC expertise in Data Commons and Data Meshes took charge
- c. Working shoulder to shoulder with the University of Chicago, Center for Transportation Data Science we created, first of it's kind Chicagoland region COVID-19 Data Commons
- d. Using the expertise from all teams we forged agreements (aka Common Legal Agreements) with regional health care organizations to bring valuable data for ongoing research
- e. Using the FAIR model as a framework we created a secure space using Gen3 Data Commons for organizing and sharing data related to COVID-19

Background

Progress to date:

- a. We started and defined a data dictionary that help answers questions related to the Case Fatality Ratio eventually making this data public
- b. We integrated a system developed by SIU that tracked mobility of COVID-19 cases
- c. We are now working on Long COVID-19 analysis systemantics through a devolved governance structure
- d. Now we have 7 million subject level records from members of the group to continue the ongoing research

Mikisha Patel,
Technical Project Manager

Data Submission Process

How OCC works with different organizations & Universities to collect data

- Complete Contracts & Legal Documentation
- IRB Approval
- Establishing a clear Data Dictionary
- Onboarding member to PRC
- Upload/submit data to PRC Data Commons

Complete Contracts & Legal Documentation



Establish contracts and legal documentation with member organization and universities for the data submission

<https://pandemicresponsecommons.org/governance/legal-agreements/>

Currently we have 4 member organization who submits the data quarterly:

- Rush
- Northshore
- UIC
- UChicago

IRB Approval

Each organization and university must obtain IRB approval to upload the data fields

Align to the Data Dictionary



- Coordinate with the different organizations and universities to align on a data dictionary to be collected
- Chicagoland COVID-19 Commons dictionary has 41 nodes and 1245 properties
- The data Dictionary can be viewed here:
<https://chicagoland.pandemicresponsecommons.org/DD>

Member Onboarding to PRC

- Providing access to PRC Data Commons
- OCC will create a project to host the data, and provide access to the members accordingly

Urvi Sheth,
Sr. Software Engineer

Upload/Submit Data to PRC Data Commons



- Secure Data Submission or Retrieval
 - Ensuring secure methods for data submission or retrieval.
- Data Pre-processing and Validation
 - Cleaning and validating data to meet data commons standards.
- Data Modeling
 - Creating structured models based on data dictionary.
- Stakeholder Training (optional)
 - Offering training on data upload.
- Data Upload
 - Uploading data to designated nodes within the project.
- Data Utilization and Visualization
 - Enabling data access for analysis and various visualizations in Gen3 Commons.

MIDRC Data Ingestion

Wrangling Imaging Data from Multiple Sources

Chris Meyer, PhD

Center for Translational Data Science

The University of Chicago

Gen3 Community Forum

10 July 2024

MIDRC Initiative Overview



MIDRC was launched in 2020 and aims to accelerate medical machine learning innovation by providing a high-quality, curated data resource, which includes medical imaging studies and associated clinical data.

MIDRC is funded by NIBIB, is hosted at UChicago, and is co-led by ACR, RSNA, and AAPM.



<https://midrc.org>

Most data to date are COVID-19-related clinical and imaging studies, but MIDRC is expanding to other diseases, like cancer and long COVID.

The MIDRC Data Commons

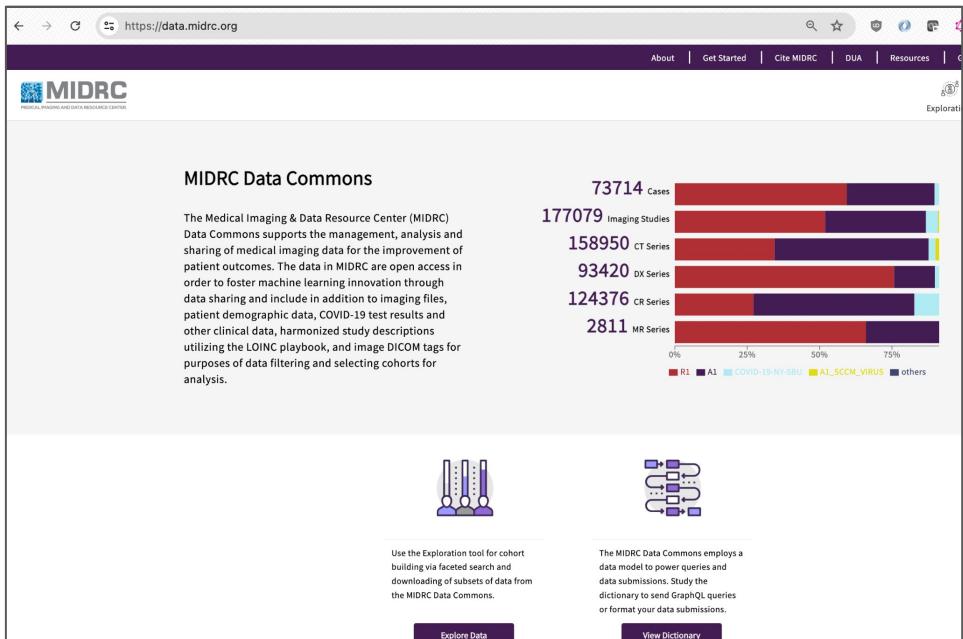


MIDRC operates four Gen3 environments:

- Production
 - **data.midrc.org** (Open)
 - **validate.midrc.org** (Sequestered)
- Staging
 - **staging.midrc.org** (Open Staging)
 - **validatestaging.midrc.org**
(Sequestered Staging)

The **open data are for training** AI algorithms
and the **sequestered data are for testing**
against a demographically balanced subset.

Data are ingested in staging environments then
copied to production after QC.



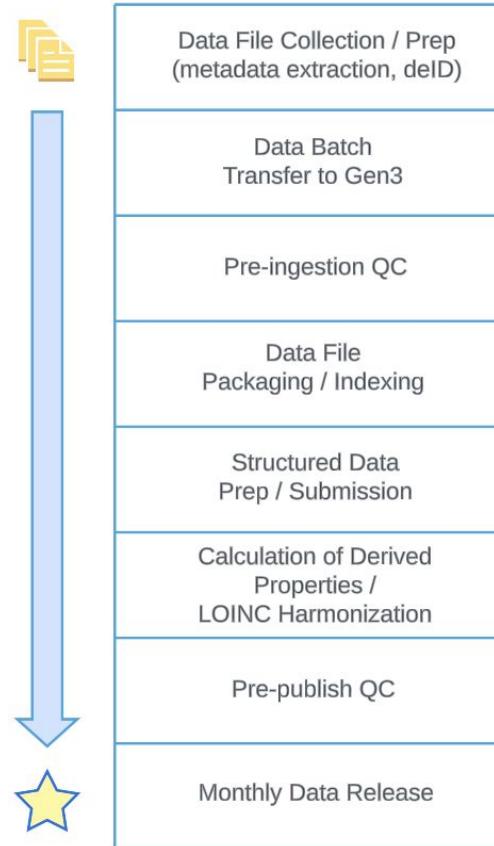
<https://data.midrc.org>

MIDRC Data Ingestion Overview

MIDRC publishes new batches of data on a **monthly release cadence** (data, services, and config changes).

The Gen3 team QC's data on initial receipt and before it's published (copied from staging environment to production).

SOP documents exist for all of these processes, and where we can, **processes are scripted**, e.g., in Python or Jupyter Notebooks (Data QC, preparation/submission, and release).



Collection of Clinical and Imaging Data

Data Continuously Flows into **2 Primary Data**

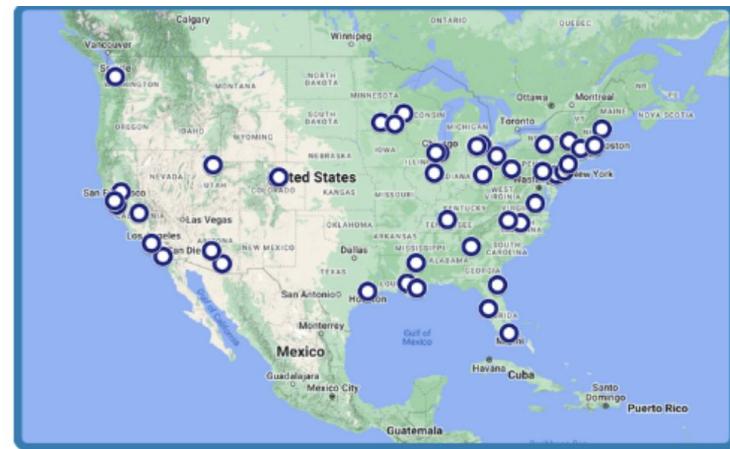
Intake Portals from Contributing Medical Sites:

- American College of Radiology (ACR) **COVID Imaging Research Registry (CIRR)**
- Radiological Society of North America (RSNA) **International COVID-19 Open Radiology Database (RICORD)**



The RSNA and ACR teams:

- Collect clinical and imaging data from medical centers
- De-Identify structured EHR data and Images
- Provide Gen3 team access to batches of data for ingestion into the MIDRC data commons.



<https://www.midrc.org/donate>

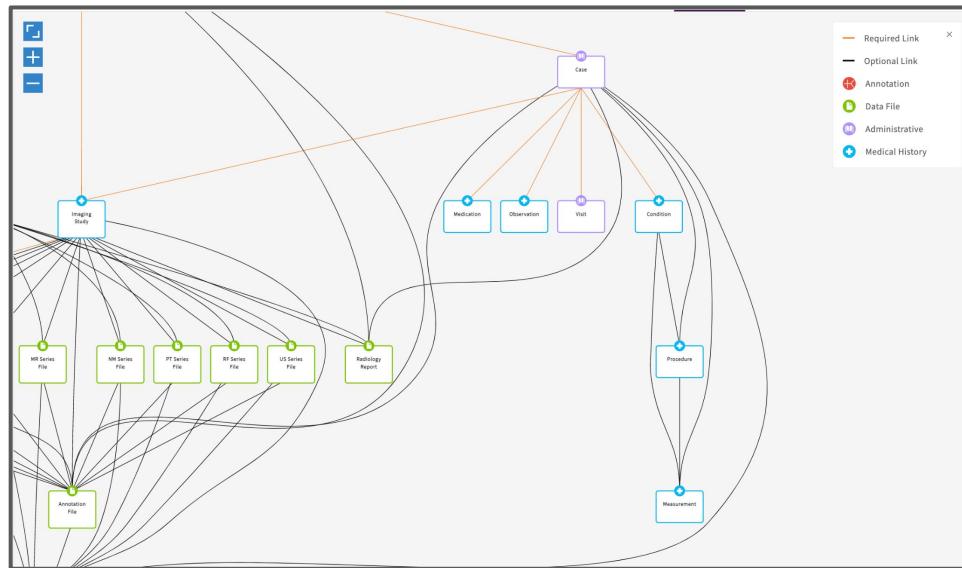
Data Modeling and Harmonization

MIDRC has a subcommittee that collaboratively develops the Gen3 graph data model: **Data Standards and Information Technology (DSIT)**.

ACR and RSNA are members and work closely with the Gen3 team to **implement a data model that best supports queries for cohort building** using patient EHR and image DICOM metadata.

ACR and RSNA extract the DICOM metadata from batches of images and organize it into Gen3 submission TSVs that conform to the data model.

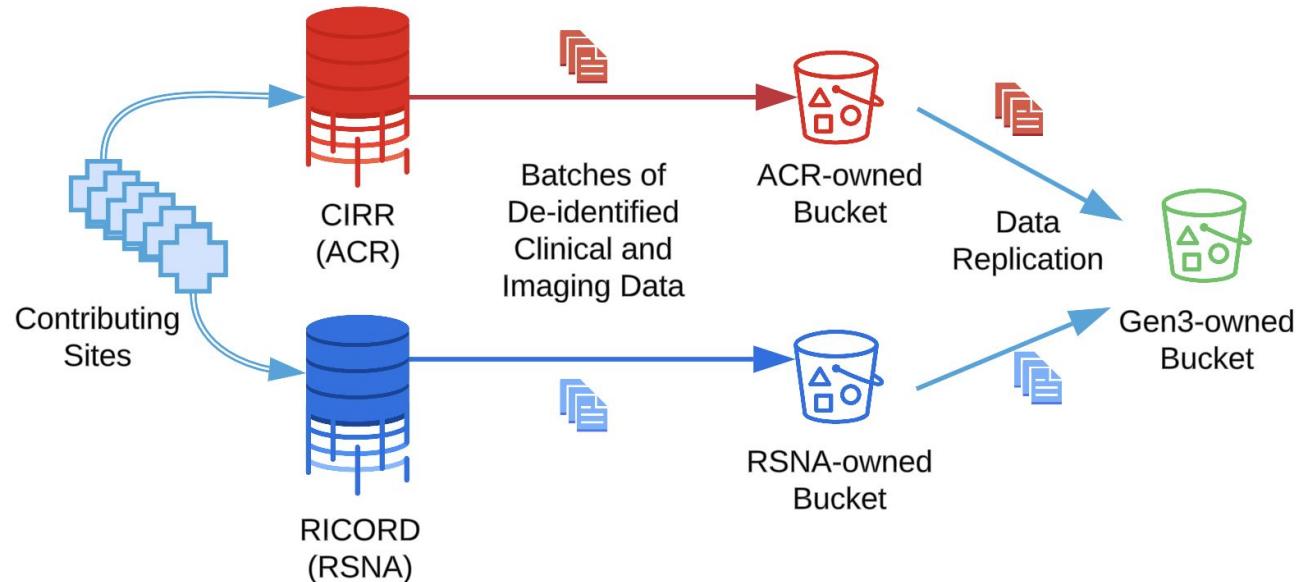
Associated clinical data is similarly extracted from EHR platforms and organized into clinical TSVs.
<https://www.midrc.org/subcommittees>



Data Dictionary Viewer:
<https://data.midrc.org/dd>

Dictionary in GitHub:
https://github.com/uc-cdis/midrc_dictionary

Data Transfer to Gen3



- RSNA and ACR periodically make batches of de-identified data available to the Gen3 Team.
- A batch consists of: structured data/submission TSVs, image files, and an image manifest.
- The Gen3 team copies each batch in order to ingest it into the MIDRC data commons.

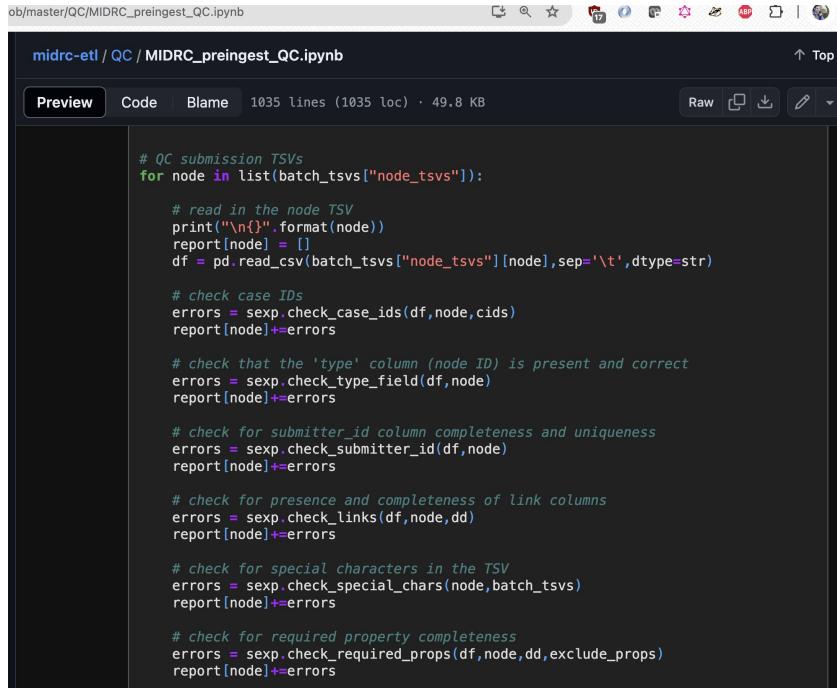
Pre-ingestion QC

Before data are ingested, Gen3 runs a “Pre-ingestion QC Checklist” to ensure completeness and proper formatting.

Checks are implemented in a Jupyter notebook to:

- Confirm reported numbers of patients, imaging studies, and files (in the MIDRC External Gen3 Data Release Tracker) match data received.
- Check that all required data fields are present and complete; report on completeness of optional data fields.
- Check submission TSV formatting.

If the data batch fails any checks, Gen3 notifies the data contributor and requests the batch be corrected.



The screenshot shows a Jupyter notebook interface with the following details:

- Title:** midrc-etl / QC / MIDRC_preingest_QC.ipynb
- Preview tab:** Active
- Code tab:** Available
- Blame tab:** Available
- Statistics:** 1035 lines (1035 loc) · 49.8 KB
- Toolbar:** Includes icons for file operations, search, and help.
- Code Content (Preview):**

```
# QC submission TSVs
for node in list(batch_tsvs["node_tsvs"]):
    # read in the node TSV
    print("\n{}\n".format(node))
    report[node] = []
    df = pd.read_csv(batch_tsvs["node_tsvs"][node], sep="\t", dtype=str)

    # check case IDs
    errors = sexp.check_case_ids(df, node, cids)
    report[node]+=errors

    # check that the 'type' column (node ID) is present and correct
    errors = sexp.check_type_field(df, node)
    report[node]+=errors

    # check for submitter_id column completeness and uniqueness
    errors = sexp.check_submitter_id(df, node)
    report[node]+=errors

    # check for presence and completeness of link columns
    errors = sexp.check_links(df, node, dd)
    report[node]+=errors

    # check for special characters in the TSV
    errors = sexp.check_special_chars(node, batch_tsvs)
    report[node]+=errors

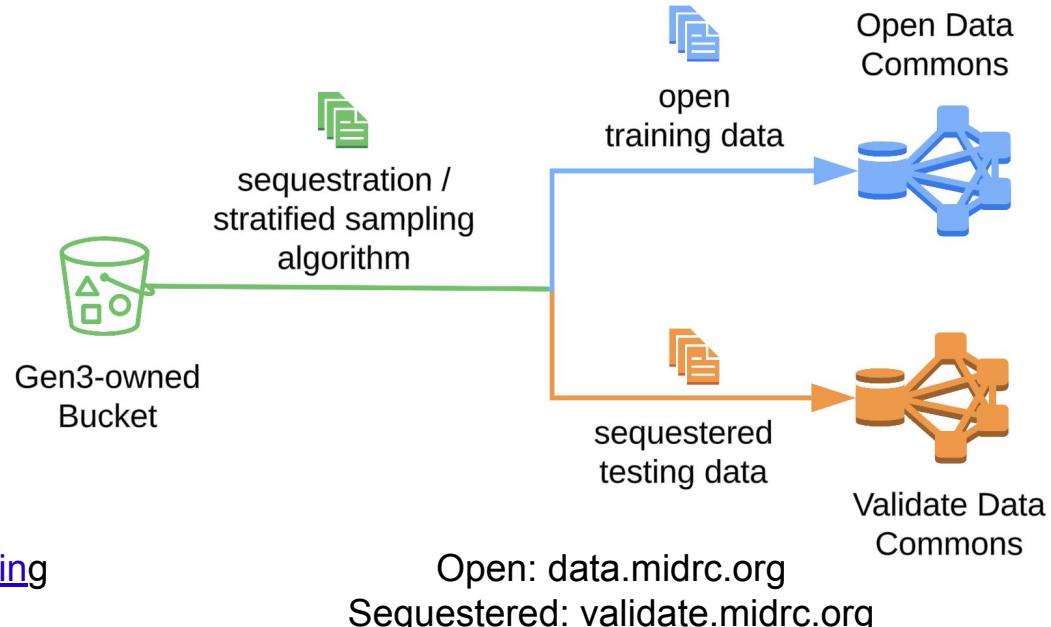
    # check for required property completeness
    errors = sexp.check_required_props(df, node, dd, exclude_props)
    report[node]+=errors
```

https://github.com/uc-cdis/midrc-etl/blob/master/QC/MIDRC_preingest_QC.ipynb

Patient Sequestration

Before ingestion, new patients are split between the **Open (80%)** and **Sequestered (20%)** commons by performing a **stratified sampling algorithm** that attempts to create patient cohorts that are balanced with respect to:

- Patient
 - Age
 - Ethnicity
 - Race
 - Sex
 - Care Site ID
 - COVID-19 status
- Imaging Study
 - Modality
 - Description
 - Body Part Examined



https://github.com/MIDRC/Stratified_Sampling
<https://doi.org/10.1111/1.JMI.10.6.064501>

Open: data.midrc.org
Sequestered: validate.midrc.org

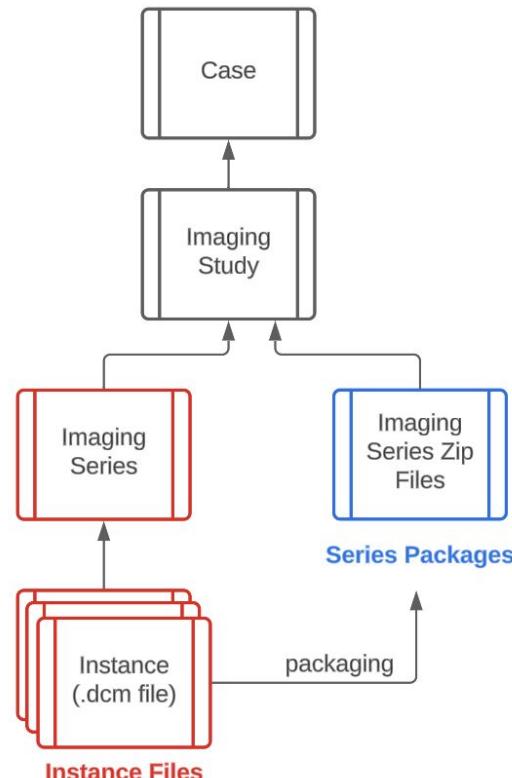
Image File Packaging and Indexing

MIDRC data is in DICOM format, the base-level of which is the image “instance”, which is a .DCM file.

For an x-ray, instances are single images, but for volumetric imaging (MR, CT, etc.) image instances are “slices” in a 3-dimensional image stack comprised of hundreds of instances.

In order to accelerate searches and download speeds for volumetric imaging modalities, **image instances are packaged into series-level zip files** and the zip files are indexed in indexd.

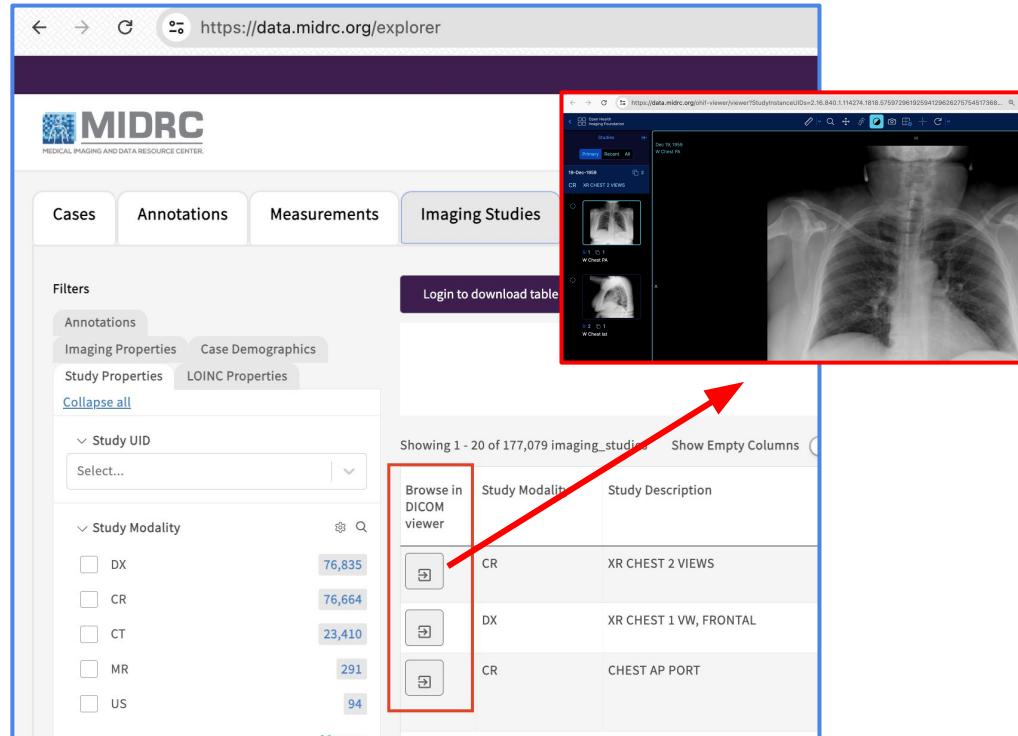
<https://github.com/uc-cdis/midrc-etl/tree/master/packaging>



Upload Instances to DICOM Viewer Server

Imaging studies in the MIDRC data explorer feature a button that links to a page where imaging series can be viewed in the OHIF DICOM Viewer.

In order for this to work, the image instance files are copied to an Orthanc Server, which organizes the instances (slices) into series and studies for viewing.



https://data.midrc.org/ohif-viewer/viewer?StudyInstanceUIDs=<imaging_study.submitter_id>

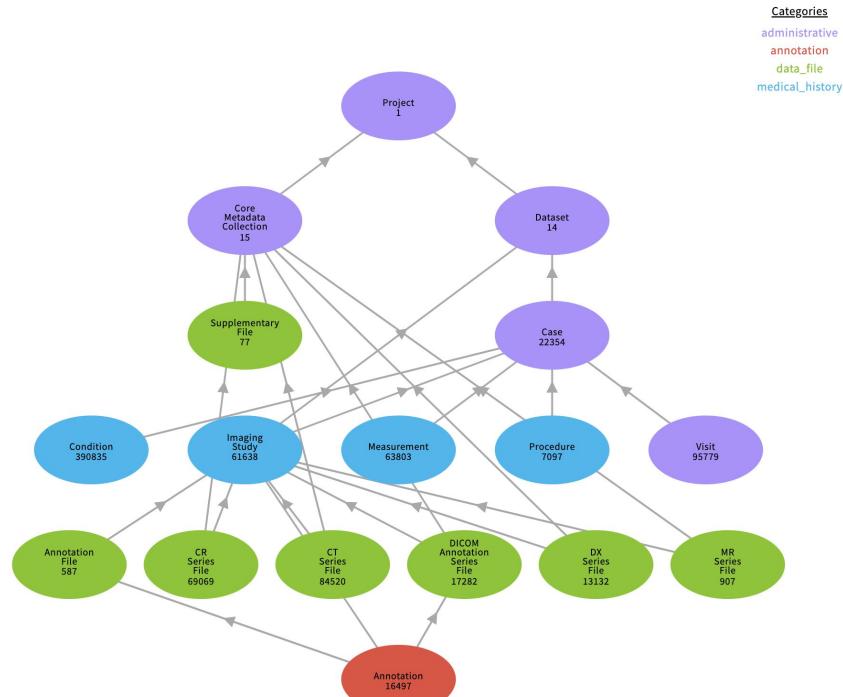
Structured Data Submission

Once the image files are packaged and indexed, image package GUIDs are joined to the imaging series TSVs and the structured data TSVs are submitted to the graph via sheepdog using the Gen3SDK “Submission” class function:

`Gen3Submission.submit_file()`.

- Retries API requests on service failures.
- Returns error messages for troubleshooting
- Returns lists of records by success / failure for faster and simpler retries / resubmissions.

<https://github.com/uc-cdis/gen3sdk-python/blob/dbf607b4e91263ea435be27fefedd42fb83daa42/gen3/submission.py#L509>



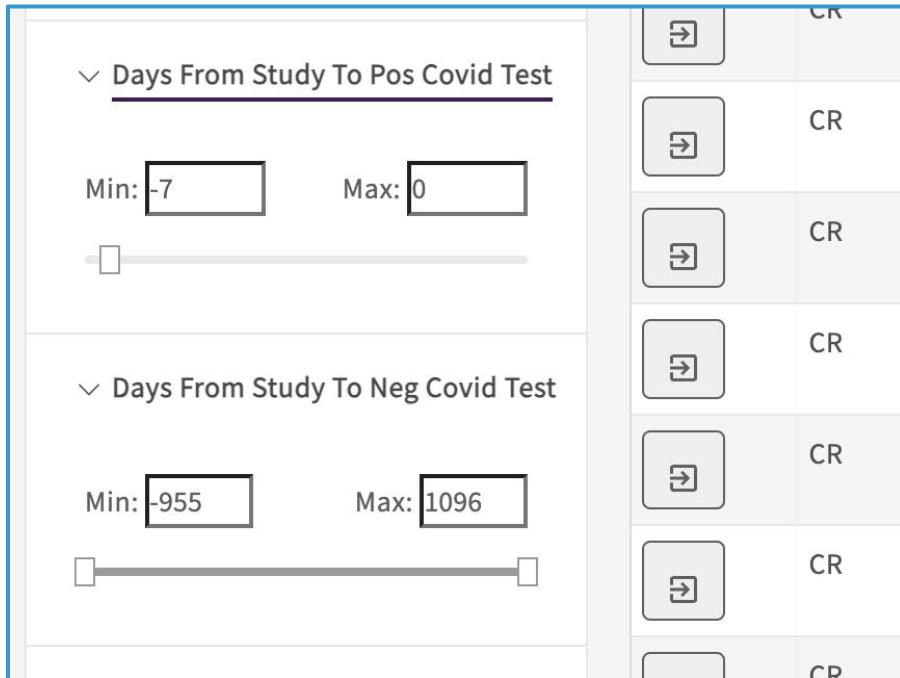
<https://data.midrc.org/Open-A1>

Calculation of Derived Properties

Certain properties in the MIDRC data model are derived from the raw data and these are calculated and submitted for all relevant records in Staging prior to release via Jupyter Notebook.

Two examples are the number of days between each imaging study and a positive or negative COVID test:

- `days_from_study_to_neg_covid_test`
- `days_from_study_to_pos_covid_test`



https://github.com/uc-cdis/midrc-etl/blob/master/temporal/calculate_days_from_study_to_covid_test.ipynb
<https://data.midrc.orc/explorer>

LOINC Mapping / Harmonization

The Data Quality and Harmonization Subcommittee (**DQH**) has used the LOINC Standard to harmonize over 1,700 disparate imaging study descriptions to only 75 LOINC codes, which encompass the following:

- Study Description
- Modality
- Contrast Indicator
- Body Part Examined

Prior to release of new data to production environments, we perform LOINC mapping and sheepdog update via a Jupyter Notebook.

The screenshot shows the MIDRC Explorer interface at the URL <https://data.midrc.org/explorer>. The top navigation bar includes links for About, Get Started, and Cite MIDRC. On the right, there are buttons for Exploration and a Help icon. Below the navigation is a header with tabs for Cases, Annotations, Measurements, Imaging Studies, and Data Files. The Imaging Studies tab is selected, indicated by a blue background. A large number '177,079' is displayed next to the tab. To the left of the main content area, there is a sidebar with filters for Case Demographics, Annotations, Imaging Properties, Study Properties, and LOINC Properties. The LOINC Properties filter is highlighted with a red box. The main content area displays a table of imaging studies. The columns are labeled 'Browse in DICOM viewer', 'Study Modality', 'Study Description', and 'Body Part Examined'. The table lists several entries, such as 'CR' under Study Modality, 'XR CHEST 2 VIEWS' under Study Description, and 'CHEST' under Body Part Examined. The table also includes a 'Download Table' button and a 'Download File Manifest for Imaging Studies' link. At the bottom of the table, there is a note 'Showing 1 - 20 of 177,079 imaging_studies' and a 'Show Empty Columns' toggle switch.

https://github.com/MIDRC/midrc_dicom_harmonization

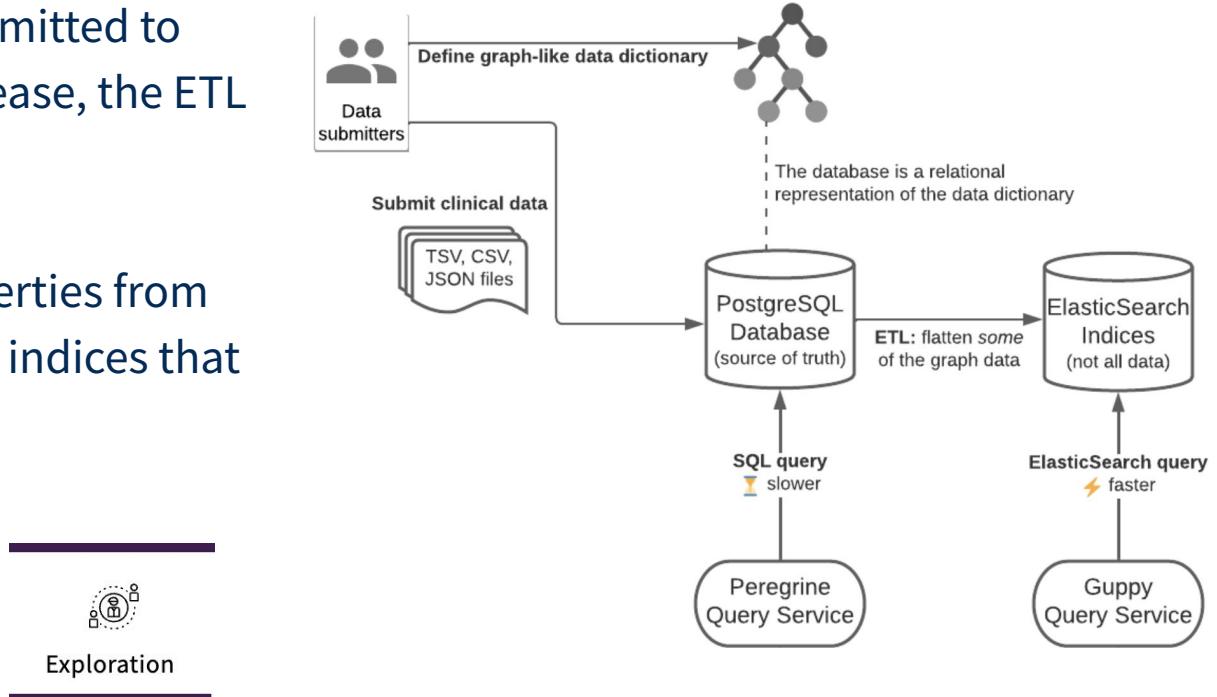
<https://loinc.org/kb/users-guide/loinc-rsna-radiology-playbook-user-guide/>

Running the ETL

Once all data have been submitted to the graph prior to a data release, the ETL process is performed.

The ETL flattens select properties from the graph into ElasticSearch indices that can be queried by guppy.

Guppy indices / queries power the data explorer GUI.



Pre-release QC

Prior to releasing new data from Staging to Production,
Gen3 performs a Pre-release QC Checklist.

- Calculate derived properties / LOINC mapping.
- Confirm ETL has been run.
- Confirm counts of files and metadata entities submitted match expectations.
- Confirm UI components / file downloads working.
- Confirm data dictionary versions are up-to-date and match between staging environments.
- Confirm software versions are up-to-date and match between staging environments.
- New tutorial Jupyter notebooks are added to resource browser.

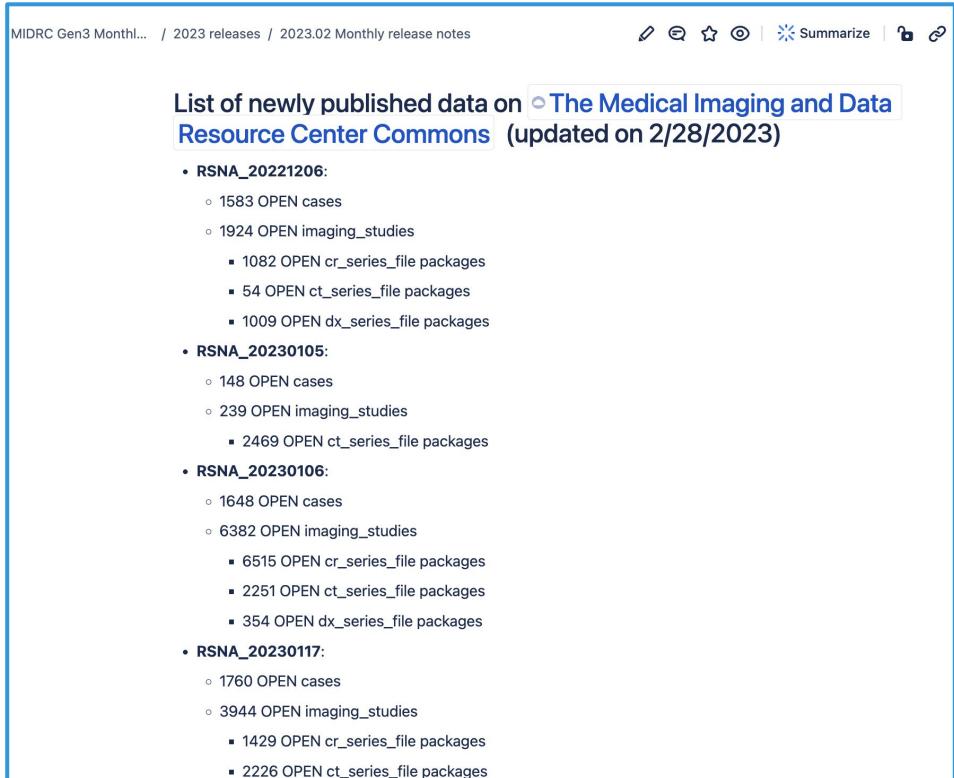
	A	B	C	D
1	Batches	ACR_20230830	RSNA_20230825	
2	All submission TSVs in Box ingested?	Yes	Yes	
3	Instances in manifest	Yes	Yes	
4	Instances removed (deleted, held back)	0	0	
5	Instances packaged	Yes	Yes	
6	Packages published to staging (indexed)	Yes	Yes	
7	Packages published to validate (indexed)	Yes	Yes	
8	Cases - TSV	4818	1558	
9	Imaging Studies - TSV	4818	2872	
10	CR Series Files - TSV	3196	1375	
11	CT Series Files - TSV	4540	0	
12	DX Series Files - TSV	2129	3596	
13	MR Series Files - TSV	0	0	
14	NM Series Files - TSV	0	0	
15	RF Series Files - TSV	0	0	
16	Condition - TSV	0	0	
17	Measurement - TSV	0	3697	
18	Procedure - TSV	0	0	
19	Visit - TSV	0	0	
20	<i>Cases Removed</i>	0	0	

https://github.com/uc-cdis/midrc-etl/blob/master/QC/MIDRC_QC_prerelease_workflow.ipynb

Data Release / Publication Process

At the end of every month, Gen3 performs a “release”:

- Staging indexd, MDS, and sheepdog **databases are copied** from staging to production.
- Relevant **data-portal config is copied to prod** (gitops.json, manifest.json and ETL mapping).
- Finally, **ETL is run in production** to update guppy indices.



MIDRC Gen3 Monthl... / 2023 releases / 2023.02 Monthly release notes

List of newly published data on [The Medical Imaging and Data Resource Center Commons](#) (updated on 2/28/2023)

- **RSNA_20221206:**
 - 1583 OPEN cases
 - 1924 OPEN imaging_studies
 - 1082 OPEN cr_series_file packages
 - 54 OPEN ct_series_file packages
 - 1009 OPEN dx_series_file packages
- **RSNA_20230105:**
 - 148 OPEN cases
 - 239 OPEN imaging_studies
 - 2469 OPEN ct_series_file packages
- **RSNA_20230106:**
 - 1648 OPEN cases
 - 6382 OPEN imaging_studies
 - 6515 OPEN cr_series_file packages
 - 2251 OPEN ct_series_file packages
 - 354 OPEN dx_series_file packages
- **RSNA_20230117:**
 - 1760 OPEN cases
 - 3944 OPEN imaging_studies
 - 1429 OPEN cr_series_file packages
 - 2226 OPEN ct_series_file packages

Example release notes from a MIDRC monthly release.

Thank You!

- Gen3 / Center for Translational Data Science
 - Robert Grossman (co-PI)
 - PMs
 - Ao Liu
 - Lynette Lilly
 - Karen Hyatt
 - Devin Grant-Keane
 - User Services Team
 - Johnbright Anyaibe
 - Eric Giger
 - Dan Biber
 - Tara Lichtenberg
 - Technical Leads
 - Pauline Ribeyre
 - Sai Shanmukha Narumanchi
 - Andrew Prokhorenkov
 - Thanh Nguyen
- MIDRC Central Admin / UChicago
 - Maryellen Giger (co-PI)
 - Katie Pizer (Lead Admin)
 - Erin Mueller (Lead Admin)
 - Nick Gruszauskas (HIRO)
- RSNA
 - Curtis Langlotz (co-PI)
 - Adam Flanders (co-PI)
 - Chris Carr (Data Lead)
- ACR
 - Charles Apgar (co-PI)
 - Michael Tilkin (co-PI)
 - Tao Wang (Data Lead)
 - Brian Bialecki (Data Lead)
- AAPM
 - Maryellen Giger (co-PI)
 - Paul Kinahan (co-PI)
- **And many many more!**

<https://www.midrc.org/midrc-team>

g3t: Gen3 Tracker – User Driven Submissions

Jordan Lee and Liam Beckman

Development By: Brian Walsh, Matthew Peterkort, Nasim Sanati, and Quinn Wai Wong

Ellrott Lab, Oregon Health and Science University

"What is the biggest open challenge in biology?"

- Getting people to share data.
- Structuring, organizing, and annotating data with metadata so it's useful.
- Building higher-level abstractions so people can efficiently work with big data.

[Vince Buffalo](#)



Findable Accessible Interoperable Reusable



- **Findable (F)**

- **Metadata:** Ensure data is accompanied by rich metadata for easy discovery.
- **Unique Identifier:** Assign a unique and persistent identifier to the dataset.
- **Searchable:** Enhance findability through search engines and repositories.

- **Accessible (A)**

- **Open Access:** Make data openly accessible to a wide range of users.
- **Permissions:** Clearly define access rights and provide necessary permissions.
- **Formats:** Ensure data is available in multiple formats for different user needs.

- **Interoperable (I)**

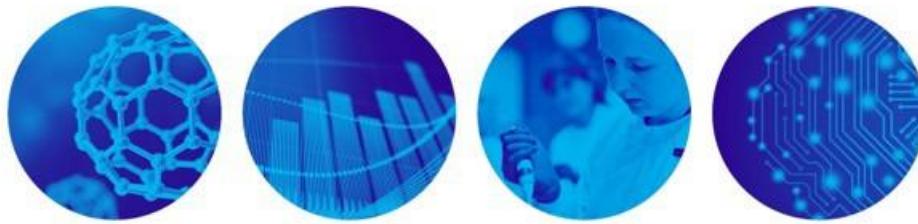
- **Standards:** Use common data standards and formats to facilitate interoperability.
- **Linkage:** Enable linkage with other datasets to derive additional insights.
- **APIs:** Provide Application Programming Interfaces (APIs) for seamless integration.

- **Reusable (R)**

- **Documentation:** Provide comprehensive documentation for easy understanding.
- **Licenses:** Clearly specify the terms of use and licensing agreements.
- **Citations:** Encourage and facilitate proper citation for data reuse.

ACED

INTERNATIONAL
ALLIANCE FOR
CANCER EARLY
DETECTION

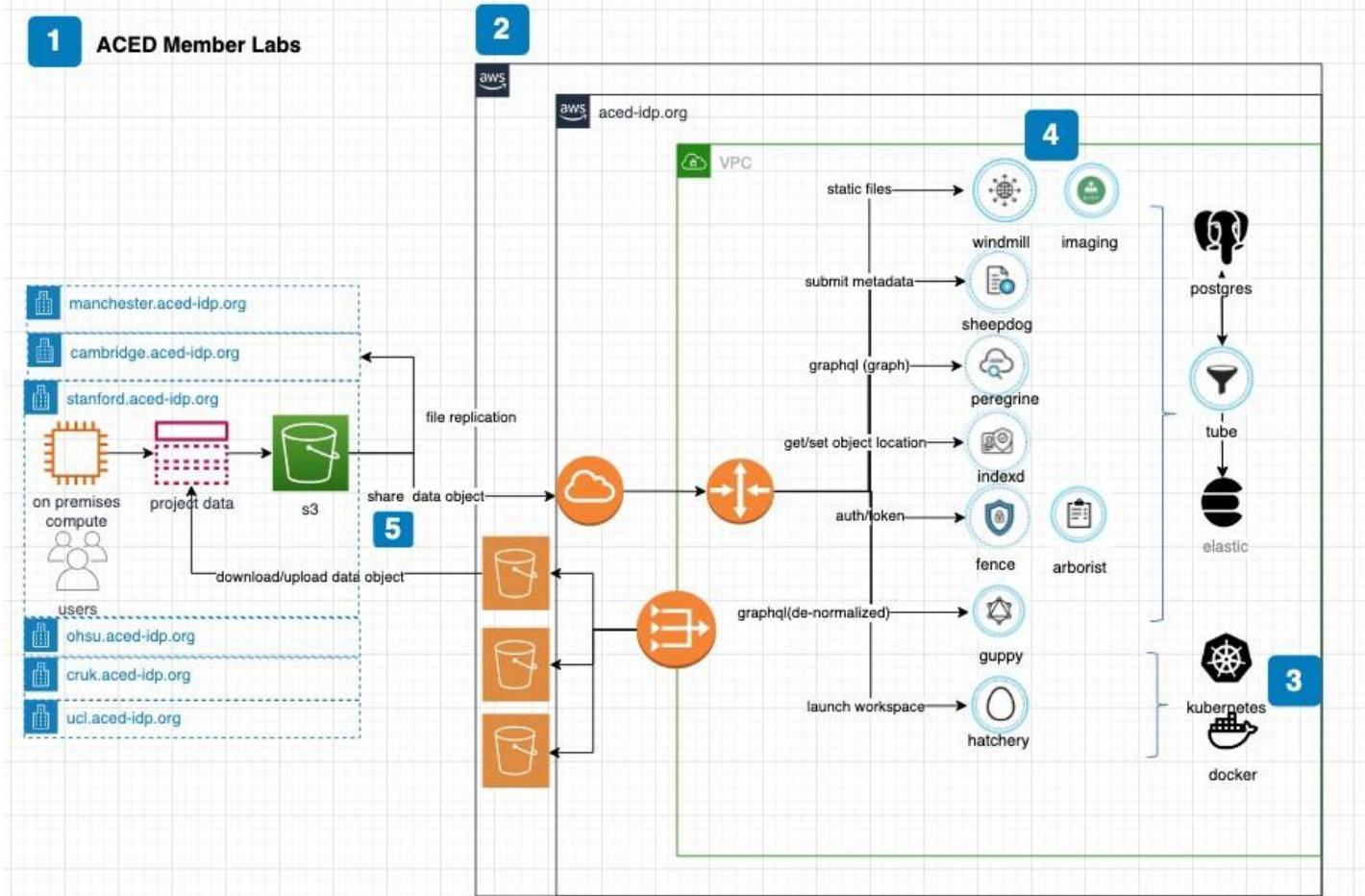


We are uniting world leading researchers to tackle the biggest challenges in early detection, an important area of unmet clinical need. Scientists in the Alliance are working together at the forefront of technological innovation to translate research into realistic ways to **improve cancer diagnosis**, which can be **implemented into health systems** and meaningfully benefit people with cancer.

cancerresearchuk.org

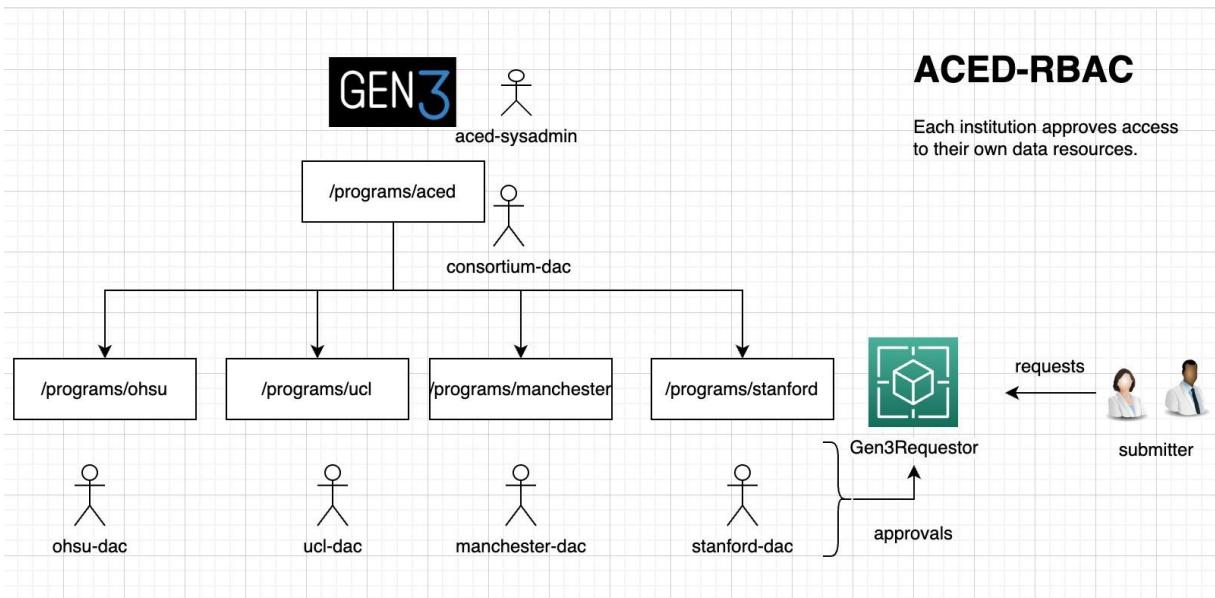
ACED IDP

High Level Architecture



Data Stewardship

As an aced data steward, in order to understand my role in creating projects and granting access, I need way to understand and implement my role and responsibilities.



Accessible
 Open Access, Permissions : A distributed team controls **data stewardship** to grant and revoke access over their institution's data.

[aced-rbac](#)

Data Stewardship: Project Creation

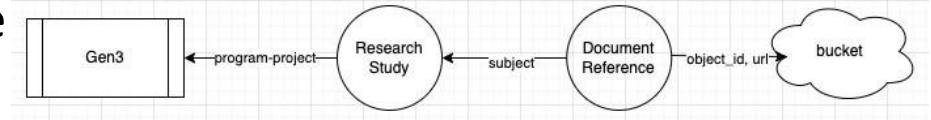
```
# as a data submitter
g3t init {program}-{project}

# as a data steward
g3t collaborator approve --request_id {request ID}

# as a system administrator
g3t projects create
```

Minimal Viable Study

As a data submitter, in order to share data, I want to upload a set of files



```
# repeat for each file
g3t add PATH [--size,--<hash>,--mime]

# create metadata
g3t meta init

# add to repository
g3t commit -m "My study's files"
g3t push

# view upload status (pending, complete)
g3t status
```

Minimal Viable Study

```
$ ls -1 META/
```

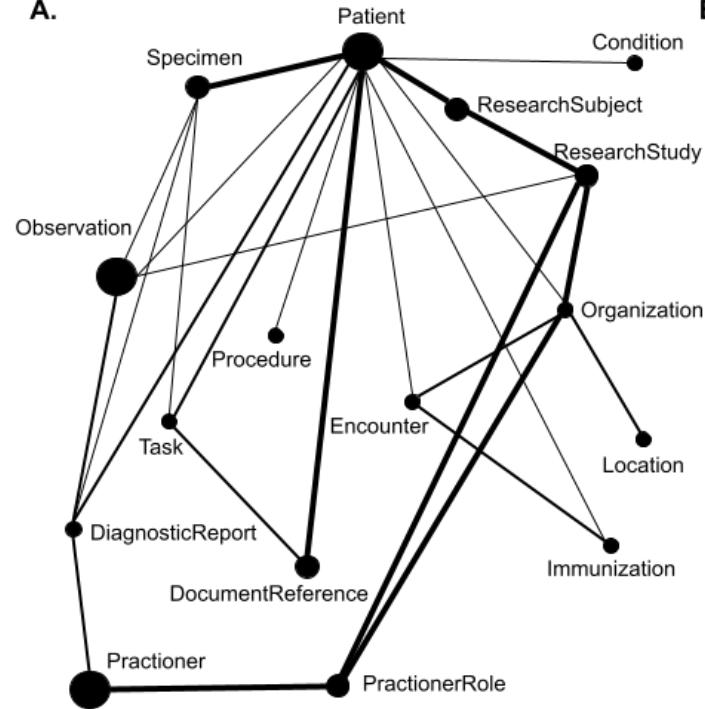
```
DocumentReference.ndjson  
ResearchStudy.ndjson
```

The screenshot shows the ACED (International Alliance for Cancer Early Detection) web application. At the top right, there are links for "About ACED", "beckman@ohsu.edu", and "Logout". Below the header, there are two buttons: "Exploration" and "Profile". The main area displays a search interface with three tabs: "Patient" (selected), "File", and "Project Id". The "Project Id" tab shows a search result for "aced-example" with a count of 1. The "File" tab shows a search result for "no data" with a count of 1. The "Patient" tab is currently inactive. To the right, a "Filter by Value" section is used to refine the search results. The filters applied are "PROJECT ID: ACED-EXAMPLE" and "TITLE: EXAMPLE.TXT". A "Download Manifest" button is available. The results table lists one item: "example.txt" with "file:///data/example.txt" as the source URL, a size of 14 bytes, and a creation date of 2024-07-09T18:05:27.63395. There are also "Show Charts" and "1 Files" buttons. At the bottom, there are pagination controls for "Rows per Page" (set to 10), and navigation icons for "1" and "2".

Title	Source Url	Size	Creation
example.txt	file:///data/example.txt	14	2024-07-09T18:05:27.63395

Data Model

A.



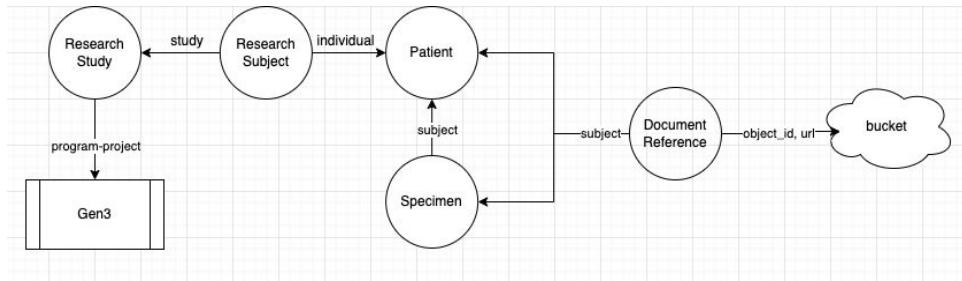
B.

	AnVIL	Cohesive DataSet	Genomics Reporting	Kids First	NCPI	Synthea	dbGap
Condition		705				4690	
DiagnosticReport			1			14127	
DocumentReference	9609	119		38394	1	8161	
Encounter		10603				8161	
Immunization						1738	
Observation	1	47869	10	664	1	51545	46
Organization	4				1	1	
Patient	3202	45	1	1765	2	122	83
Practitioner	1			1	1	1	
PractitionerRole	1				1	1	
ResearchStudy	1	1			1	1	
ResearchSubject	3202	45		1765	1		83
Specimen	3202	23	1	2281	1		
Task	3202	23				1	

Aggregated demonstration datasets. A) A Graph representation of the unified schema, with vertex sizes representing the relative number of data sets that contain that vertex type and the thickness of the edges representing the number of datasets that implemented that relationship. B) A table of the total vertex type counts across the reported datasets.

Study with Tagged Patients [specimens, etc]

As a data submitter, in order to share data, I want to upload a set of files, each tagged with any of patient, specimen, task, etc.



```
# for each file: ...
g3t add PATH --patient my-patient-identifier [--specimen, --size, --hash <hash>]

# create metadata
g3t utilities meta create
# optional: edit generated metadata

# add to repository
g3t commit -m "my study's files, subjects and/or specimens"
g3t push
```

Study with Tagged Patients [specimens, etc]

```
$ ls -1 META/
```

```
DocumentReference.ndjson  
Patient.ndjson  
ResearchStudy.ndjson  
ResearchSubject.ndjson
```

The screenshots illustrate the ACED web application's interface for managing patient and study data. The top screenshot shows a general search and filter interface, while the bottom screenshot provides a detailed view of a specific project's data.

Top Screenshot (Search Interface):

- Project Id:** Filtered by "aced-example".
- Gleason Grade:** Filtered by "no data".
- Identifier:** Filtered by "150".
- Procedure Identifier:** Filtered by "1".
- Specimen Identifier:** Filtered by "1".
- Gleason Grade:** Filtered by "17.918".

Bottom Screenshot (Project Detail View):

- Project ID:** Filtered by "aced-example".
- Type:** Filtered by "no data".
- Category:** Filtered by "1".

File Details:

Title	Source Url	Size	Creation
sample.txt	file:///data/sample.txt	25	2024-07-09T18:14:35.715636



Deep Dive

```
{  
  "resourceType": "Patient",  
  "id": "f027d9b9-da61-5f48-9378-f4dc0e6b85e6",  
  "identifier": [  
    {  
      "use": "official",  
      "system": "https://aced-idp.org/test-one_patient",  
      "value": "P1"  
    }  
  ]  
}
```



- **Findable (F)**
- **Metadata:** The aced-idp system encourages and facilitates the creation of metadata over a wide variety of [use cases](#)
- **Unique Identifier:** The system requires and maintains a submitter driven [identifier](#) and well as location independent, [idempotent ids](#) for all metadata resources. File objects are also registered as DRS (GAGH Data Repository Service) [uris](#)
- **Searchable:** All of the above keys are searchable via the portal or API. The system defaults [CodeableConcept](#) attributes to submitter provided values and encourages additional tagging with standard ontology terms

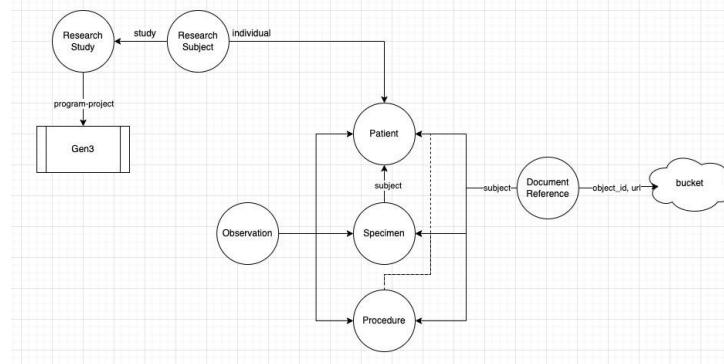
Study with Rich Set of Measurements

Transform submission CSVs to FHIR

A	B	C	D	E	F	G	H	I	J	K	L	M
csv_column_name	csv_description	csv_type	csf	fhir_resource	coding_system	coding_code	coding_display	c	observatio	uom_system	uom_code	uom_unit
id	Patient ID	string		Patient, Specimen, Condition								
align	Aligned lesion	string		Binary Observation					Condition			
ageDiagM	Age at Diagnosis in Months	integer		Condition.age								
ageDiagY	Age at Diagnosis in Years	integer		Observation	https://loinc.org/	63932-8	Age at diagnosis		Condition	http://unitsofmeasure.org	mo	month
ppsa	Presenting PSA at diagnosis	float		Observation	http://snomed.info/sct/	63476009	Prostate specific antigen measurer	Procedure		http://unitsofmeasure.org	/a	/ year
BxPreDiag	Biopsy before diagnosis	integer		Observation					Procedure	http://unitsofmeasure.org	ng/mL	nanograms per milliliter (ng/mL)
psaBx	PSA at Biopsy A or B	float		Observation	http://snomed.info/sct/	63476009	Prostate specific antigen measurer	Procedure	http://unitsofmeasure.org	ng/mL	nanograms per milliliter (ng/mL)	
months.diag	Months that elapsed since prostate cancer diagnosis	integer		Observation					Procedure	http://unitsofmeasure.org	mo	month
gleason	Gleason grade	string		Observation	http://snomed.info/sct/	372278000	Gleason score	Procedure		http://unitsofmeasure.org		
mcc1	Maximum Cancer Core Length in mm	integer		Observation	http://snomed.info/sct/	399598003	Length of core in specimen obtaine	Procedure		http://unitsofmeasure.org	millimeter	mm
uc1	UCL Definition	string		Observation					Procedure			
pvrl	Prostate volume on MRI	float		Observation	https://loinc.org/	15325-4	Prostate specific Ag/Prostate volum	Procedure		http://unitsofmeasure.org	mL	milliliter
side	Sampled area side (Left or Right)	string		Observation					Procedure			
zone	Sampled area zone (Peripheral, Transition, Both)	string		Observation					Procedure			
loc	Sampled area location (Posterior, Anterior or combinations)	string		Observation					Procedure			
level	Sampled area level (Base, Mid-gland, Apex or combinations)	string		Observation					Procedure			
likert	Likert score of sampled MRI area	integer	1-5	Observation	http://snomed.info/sct/	273575009	likert scale (assessment scale)	Procedure				
pirads	PI-RADS v2 score of sampled MRI area	integer	1-5	Observation	http://dicom.nema.org/reso	130564	PI-RADS v2.0	Procedure				
precise	PRECISE score of sampled MRI area (only for timepoint B)	integer	1-5	Observation					Procedure			
adcMean	Mean apparent diffusion coefficient of sampled MRI area	float		Observation	http://snomed.info/sct/	46638006	Diffusion	Procedure		http://unitsofmeasure.org	m2/s	square meters per second
adcn	Mean apparent diffusion coefficient of sampled MRI area (norm	float		Observation					Procedure	http://unitsofmeasure.org	m2/s	square meters per second
adcu	Mean apparent diffusion coefficient of sampled MRI area (norm	float		Observation					Procedure	http://unitsofmeasure.org	m2/s	square meters per second
focality	Lesion focality	string	Binary	Observation					Procedure			
best	MR1 sequence on which lesion is best seen	string		Observation	http://snomed.info/sct/	396199003	Tumour focality	Procedure				
bestVol	Volume of lesion on best sequence (ml)	float		Observation					Procedure	http://unitsofmeasure.org	mL	milliliter
t2Vol	Lesion volume on T2 (ml)	float		Observation					Procedure	http://unitsofmeasure.org	mL	milliliter
Epi_Count	Total number of epithelial cells within all tissue areas on H&E	integer		Observation	http://snomed.info/sct/	593942000	Epithelial cell count	Procedure				
Stroma_Count	Total number of stromal cells within all tissue areas on H&E	integer		Observation	http://snomed.info/sct/	74765001	Lymphocyte	Procedure		http://unitsofmeasure.org	mL	milliliter
Lymphocyte_Count	Total number of lymphocytes within all tissue areas on H&E	integer		Observation	http://snomed.info/sct/	271036002	Lymphocyte percent differential cou	Procedure		http://unitsofmeasure.org		
Lymphocyte_Percent	% of lymphocytes within all tissue areas on H&E	float		Observation					Procedure			
Irani_Gscore	Irani score (number of lymphocytes in largest inflammatory clust	integer		Observation					Procedure			
Tissue_Area	Tissue area (square mm)	float		Observation					Procedure	http://unitsofmeasure.org	mm2	square millimeter
Epithelial_Area	Epithelial area (square mm)	float		Observation					Procedure	http://unitsofmeasure.org	mm2	square millimeter
Stromal_Area	Stromal area (square mm)	float		Observation					Procedure	http://unitsofmeasure.org	mm2	square millimeter
Inflammatory_Area	Inflammation area (square mm)	float		Observation					Procedure	http://unitsofmeasure.org	mm2	square millimeter
Epithelial_Area_Perce	% epithelial area (epithelial area fraction)	float		Observation					Procedure			
Stromal_Area_Percent	% stromal area (stromal area fraction)	float		Observation					Procedure			
Inflammatory_Area_Pe	% inflammation area (inflammation area fraction)	float		Observation					Procedure			
Epithelial_Stromal_Rat	Epithelial area/Stromal area (square mm)	float		Observation					Procedure	http://unitsofmeasure.org	mm2	square millimeter
Lumen_Area	Total area detected as lumen within all tissue areas (square mm	float		Observation					Procedure	http://unitsofmeasure.org	mm2	square millimeter
Lumen_Density	Lumen area/tissue area	float		Observation					Procedure	http://unitsofmeasure.org	mm2	square millimeter

Study with Rich Set of Measurements

As a data submitter, in order to share data, I want to upload a set of files accompanied with a rich set of observations



```
# for each file: ...
g3t add PATH --patient my-patient-identifier [--specimen --size, --hash <hash>]

# create metadata using a transformer
G3T_PLUGIN=my_project.transformer g3t_etl transform

#add to repository
g3t commit -m "my study's files, subjects and/or specimens"
g3t push
```

Study with Rich Set of Measurements

```
$ ls -l META/
```

```
Condition.ndjson  
Observation.ndjson  
Patient.ndjson  
Procedure.ndjson  
ResearchStudy.ndjson  
ResearchSubject.ndjson
```

The screenshot shows the ACED web application interface. At the top, there's a navigation bar with links for 'About ACED', 'beckman@ohsu.edu', and 'Logout'. On the right side of the header are 'Exploration' and 'Profile' buttons. The main content area has a blue header bar with 'Filter by Value' and 'Clear All' buttons. Below this is a search bar containing 'PROJECT ID' and 'DHSU-TCGA_LUAD'. A large table below the search bar displays patient data with columns for Identifier, Procedure Identifier, Specimen Identifier, Gleason Grade, and Maximum Cancer Core Length in mm. The Gleason Grade column shows values like '17,918 Patients'. The Maximum Cancer Core Length in mm column shows values like '13'. The table lists several patient entries, such as TCGA-55-8301, TCGA-49-4506, etc., with their corresponding specimen identifiers and lengths.



Next Steps

Potential Improvements to data upload and **g3t** on our Roadmap (suggestions welcome!):

- Add ability to upload multiple files in parallel (either as an entire directory or other specified set)
- User Friendliness
 - Expand documentation and overall UX based on data analysts experiences
 - Add lessons/tutorials for easier and more gradual adoption
- Learning and Sharing with the Gen3 Community
 - Alternative ways to manage Gen3 data, different Use Cases
- Continue integration with gen3-client + Frontend Framework

Development + Contributions

g3t itself is hosted on a [public repo](#) (with a [Contributor guide](#)) — Issues + PR's welcome!

The screenshot shows a GitHub repository page for 'gen3_util'. The top navigation bar includes 'Edit Pins', 'Unwatch', 'Fork', and 'Starred' buttons. Below the header, there are dropdowns for 'development' branches (15 available), '0 Tags', and a search bar with 'Go to file' and 'Add file' buttons. A green 'Code' button is also present. On the right, there's an 'About' section with a gear icon. The main content area displays a list of recent commits:

Commit	Message	Date
matthewpeterkort Merge pull request #77 from ACED-IDP/feature/git	adds quickstart	2 months ago
matthewpeterkort Merge pull request #78 from ACED-IDP/teslajoy-simplifies	fastq mime	2 weeks ago
.flake8	Initial checkin	last year
.gitignore	chore/cleanup (#36)	7 months ago
.pre-commit-config.yaml	Adds meta, project and files cp (upload)	last year

The 'About' section on the right contains the following information:

- Collection of command line tools to interact with a Gen3 instance
- Readme
- MIT license
- Activity
- Custom properties
- 2 stars
- 4 watching
- 1 fork
- Report repository

ANY
QUESTIONS?



schema management

```
# This limits the top level objects the system will
render dependency_order:
# gen3 scaffolding required objects
- _definitions.yaml
- _terms.yaml
- Program
- Project
# FHIR objects
- Organization
- Practitioner
- PractitionerRole
- ResearchStudy
- Patient
- ResearchSubject
- Substance
- Specimen
- Observation
- DiagnosticReport
- Condition
- Medication
- MedicationAdministration
- Procedure
- DocumentReference
- Task
- ImagingStudy
- FamilyMemberHistory
- BodyStructure
```

The [iceberg schema tools](#) project enables the developer to manage schema “scope” and link to research entities.

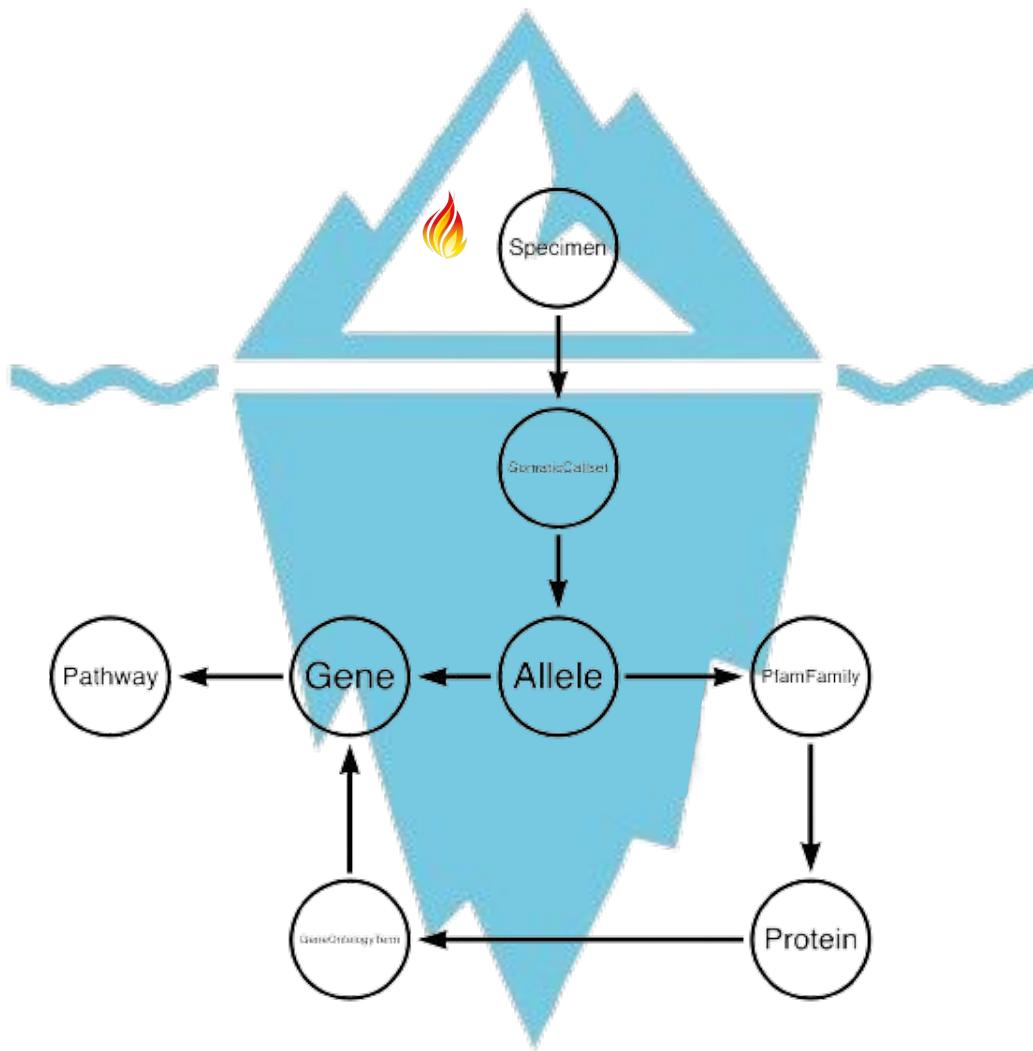
```
{  
  "resourceType": "Observation",  
  "id": "b5820487-f77e-54b2-ae7b-2d3ea6c0d891",  
  "identifier": [  
    {  
      "use": "official",  
      "system": "https://aced-idp.org/test-stavriniides",  
      "value": "123-123/0_A/609-adcMean"  
    }  
  ],  
  . . .  
  "code": {  
    "coding": [  
      {  
        "system": "https://aced-idp.org/test-demo",  
        "code": "adcMean",  
        "display": "Mean apparent diffusion coefficient of sampled MRI area"  
      },  
      {  
        "system": "http://snomed.info/sct",  
        "code": "46638006",  
        "display": "Diffusion"  
      }  
    ],  
    "text": "Mean apparent diffusion coefficient of sampled MRI area"  
  },  
  "subject": {  
    "reference": "Patient/8a92f890-6544-5c88-a27e-78e181c8dca8"  
  },  
  "focus": [  
    {  
      "reference": "Procedure/b8431407-8b39-58ff-96a4-c6981219c7c6"  
    }  
  ],  
  "valueQuantity": {  
    "value": 652.4,  
    "unit": "square meters per second",  
    "system": "http://unitsofmeasure.org",  
    "code": "m2/s"  
  }  
}
```



• Findable (F)

- **Metadata:** The aced-idp system encourages and facilitates the creation of metadata over a wide variety of [use cases](#)
- **Unique Identifier:** The system requires and maintains a submitter driven [identifier](#) and well as location independent, [idempotent ids](#) for all metadata resources. File objects are also registered as DRS (GA4GH Data Repository Service) [uris](#)
- **Searchable:** All of the above keys are searchable via the portal or API. The system defaults [CodeableConcept](#) attributes to submitter provided values and encourages additional tagging with standard ontology terms

schema scope



Example: Installing g3t

g3t releases are hosted on [PyPi](#) and can be installed with your Python package manager of choice!

```
# (Optional) Set up virtual environment
python3 -m venv venv && source venv/bin/activate

# Install latest version
pip install gen3-tracker==0.0.4rc40

g3t --version
g3t, version 0.0.4rc40

export G3T_PROFILE=aced
g3t ping
msg: 'Configuration OK: Connected using profile:production'
endpoint: https://aced-idp.org
username: user@ohsu.edu
```

Example: Uploading Files

Adapted from the [ACED Quickstart Guide](#)

```
# Initialize a new project
g3t init aced-example

# Add files
g3t add folder/file.tsv
g3t add folder/file2.tsv

# Create metadata
g3t utilities meta create

# Commit files
g3t commit -m "Adding files"

# Push to the Gen3 System
g3t push
```

Example: Downloading Files

Adapted from the [ACED Quickstart Guide](#)

[gen3-client](#) is used to download files from our Gen3 system:

```
# Single file download via GUID
gen3-client download-single --profile=aced --guid=f623df8f-5dad-5bce-a8ca-a7b69b7805a5

# Multiple file download via file manifest
gen3-client download-multiple --profile=aced --manifest=file-manifest.json
```

Example: Utilities

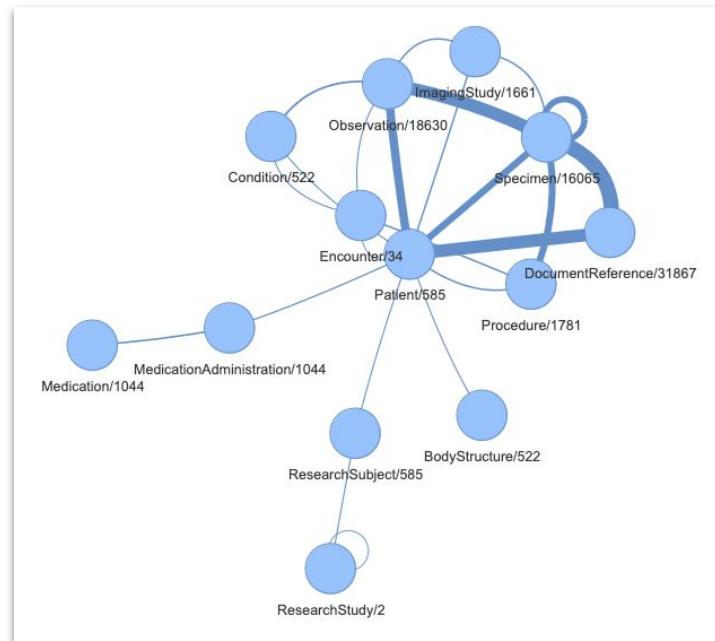
Adapted from the [ACED Quickstart Guide](#)

g3t includes commands to clone projects, manage access, and view + validate metadata:

```
# Clone an existing project
g3t clone ohsu-TCGA_LUAD

# View metadata as a graph
g3t meta graph

# Validate metadata
g3t meta validate
{'summary': {'DocumentReference': 31867,
'Procedure': 1781, 'Specimen': 16065, 'Medication':
1044, 'Observation': 18630, ... 'Patient': 585}}
```



Data Modelling in Gen3

Joshua Harris, PhD - Research Data Manager
Australian BioCommons

Acknowledgement of Country

I would like to show my respect and Acknowledge the Traditional Custodians of the Land, of Elders past and present, on which this meeting takes place.



Australian
BioCommons

Mission

To sustain **strategic leadership** in bioinformatics and bioscience **data infrastructure** nationally, support life science research with advanced **digital infrastructure**, provide sophisticated **analysis services**, ensure enduring access to essential digital tools, and offer comprehensive bioinformatics **training and support**.

Australian Cardiovascular disease Data Commons

Australian Cardiovascular disease Data Commons

This data sharing platform supports the management, analysis and sharing of Australian Coronary Artery Disease (CAD) cohorts as part of the Australian Cardiovascular Alliance (ACvA) Precision Medicine flagship.

26080 Subjects

80 Samples

80 Files

0% 25% 50% 75% 100%

■ AusDiab ■ FIELD ■ BIOHEART-CT ■ simulated

View Studies

Use the Study Explorer to view summary information about the information collected across the ACDC cohorts and apply for access.

Explore Data

The Data Explorer allows you to explore and filter data by the harmonised variables. Detailed information is only available after gaining access to a particular study.

Understand Variables

The platform has a harmonised data dictionary that describes the captured variables across all studies. Please study the dictionary before you start browsing.

Explore studies

Explore data

Explore variables

Table 1. Cohorts with available data and profiling.

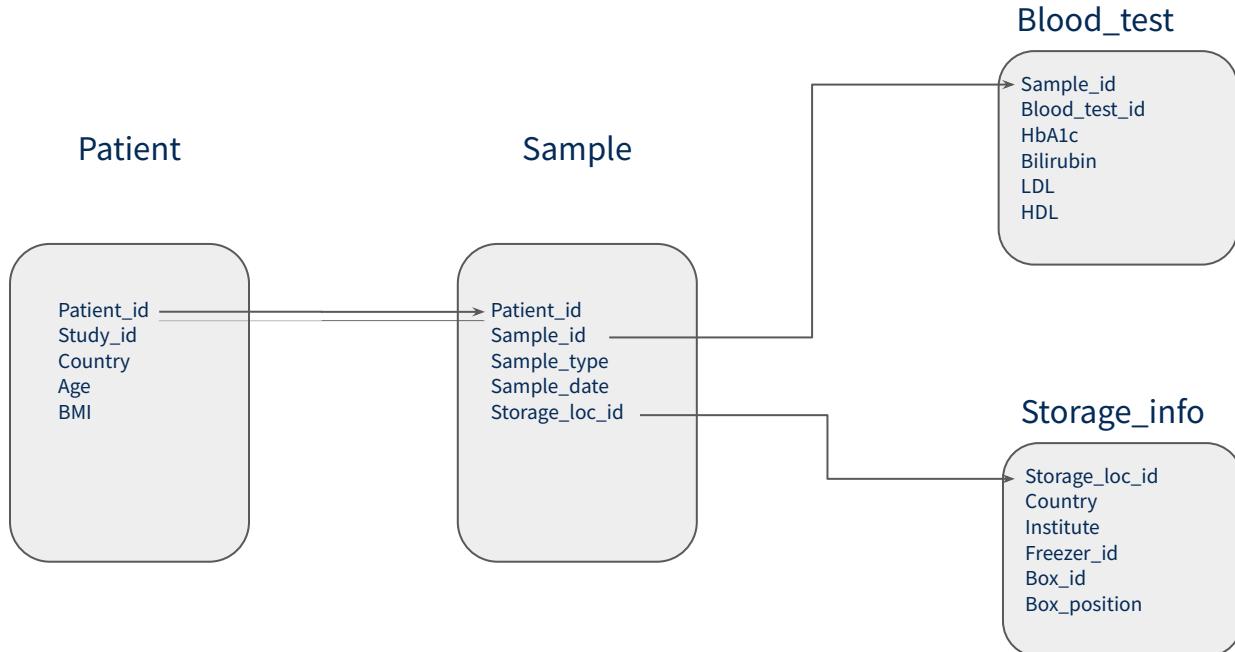
Study	Outcomes (follow-up)	Total numbers ²	Available data ¹		
			Genomic	Lipidomic	other biomarkers
AusDiab	CVD (>15 yr)	11000	0	10000	Yes
FIELD	CVD (>10 yr)	10000	5000	5000	Yes
BIOHEART-CT	CTCA/CVD (<3 yr)	5000	2000	2000	proteomic, metabolomic
Busselton	CVD (>20 yr)	4492	4492	4492	WGS on 1,000
ASPREE	CVD (~5 yr)	14000	14000	4000	WGS on 2,000, Yes
LIPID	CVD (>20 yr)	10000	0	6000	Yes
45 and UP	CVD	267000	5000		WGS on 2,000
BIOHEART-MI	CVD (<3 yr)	2000	2000	2000	proteomic, metabolomic
MCCS	CVD (>20 yr)	41513	12105	3000	
Baker Biobank	CVD (>15 yr)	6000	6000	0	
Caught-CAD	CTCA/CVD (<3 yr)	1000	1000	1000	
EDCAD-PMS	CTCA/CVD (<3 yr)	1000	1000	1000	
PREDICT	CVD	2500	0	0	
CDAH	CVD (>20 yr)	4947	0	0	Yes, metabolomics, imaging
ADVANCE	CVD (<5 yr)	11140	0	3779	
PROPHECY (Indigenous)	CVD (<3 yr)	1386	1386	0	proteomic, metabolomic, epigenetic
BIRCH (Indigenous)	CVD (<3 yr)	490	0	466	
DaVinci	CVD (<3 yr)	600	600		
Total		394068	54583	44737	

¹ Available data (including ongoing profiling activities to be completed by December 2022)

² Represents total numbers of participants for which some (but not all) data is available.

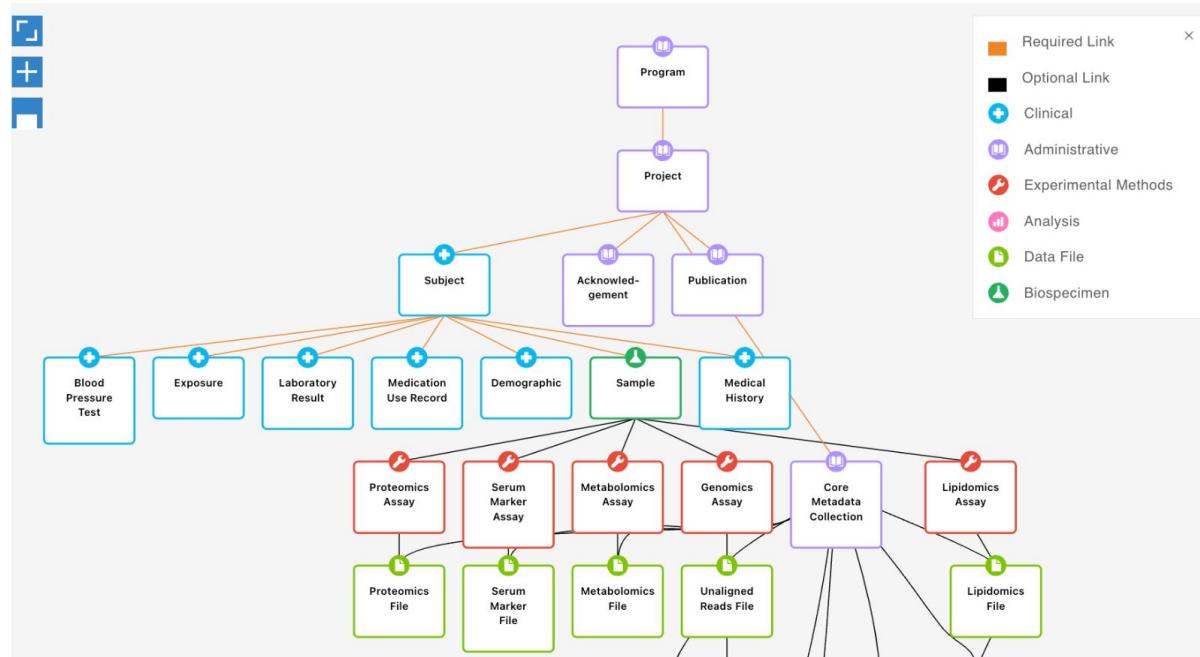
Gen3 data modelling background

Conceptual Entity Relationship Graph

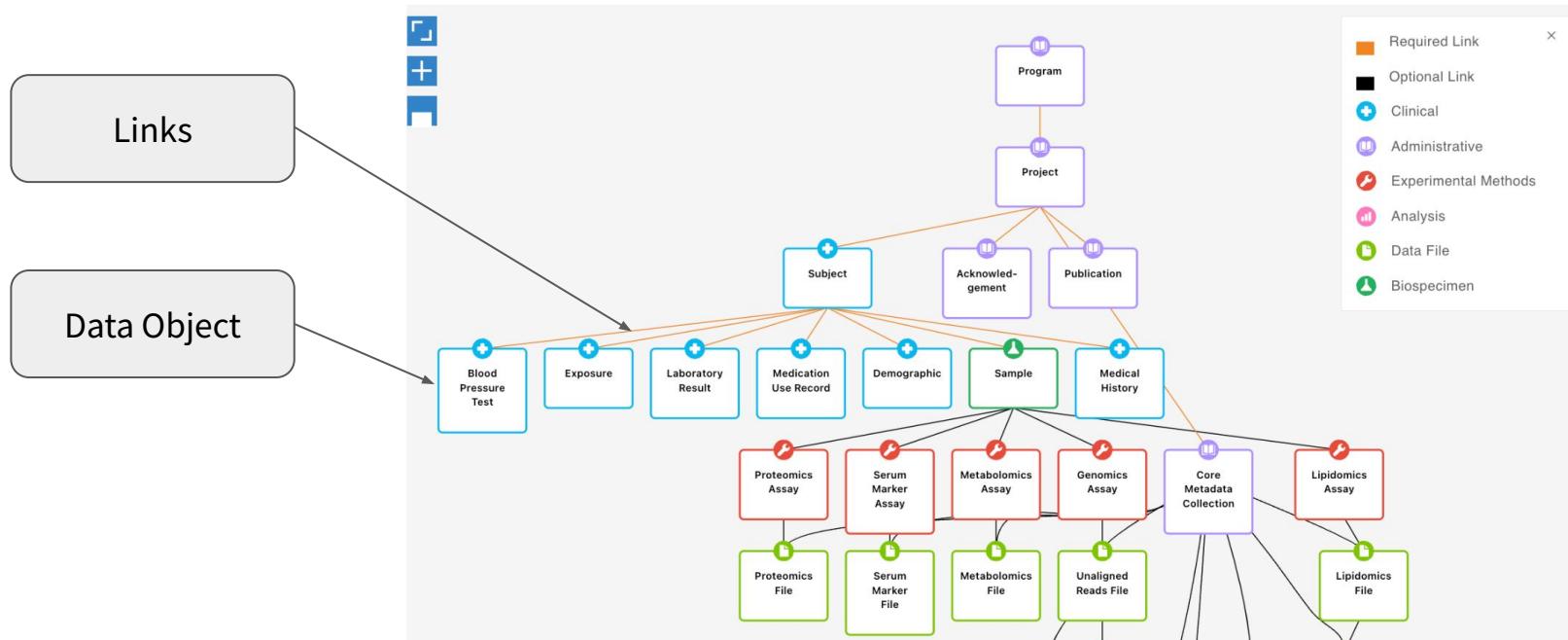


Gen3 data modelling background

Gen3's Graph View Provides a conceptual overview of a data model



Gen3 data modelling background



Gen3 data modelling background

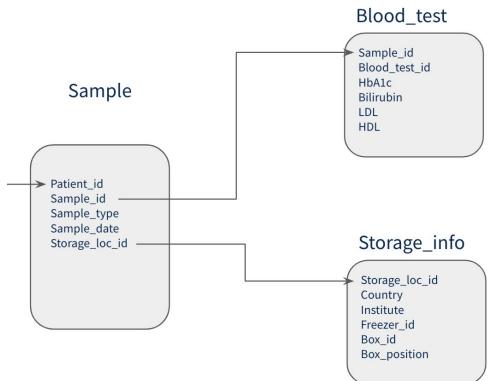
The screenshot shows the Gen3 Data Model Structure interface. On the left, three entities are highlighted with red boxes and arrows pointing to specific parts of the interface:

- Data type**: Points to the "Data Model Structure" section, specifically the "unaligned_reads_file" entry.
- Enums**: Points to the "Close properties" button and the "Download templates" button.
- Property**: Points to the "genomics_assay" property in the "data_file" schema table.

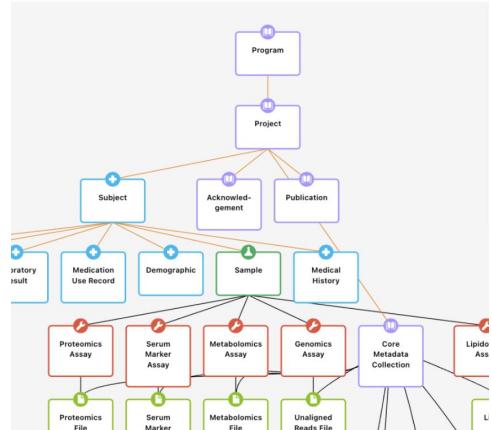
The central part of the interface displays the "data_file" schema for the "Unaligned Reads File". The schema includes the following properties:

Property	Type	Required	Description	Term
submitter_id	• string	★ Required	A project-specific identifier for a node. This property is the calling card/nickname/alias for a unit of submission. It can be used in place of the UUID for identifying or recalling a node.	
type	• string	★ Required	No Description	
baseline_timepoint	• boolean	★ Required	Does the data reflect a baseline measurement?	
data_category	• analysis • sequencing • reads • single nucleotide variation • transcriptome profiling • supplemental	★ Required	Broad categorization of the contents of the data file.	
data_format	• fastq	★ Required	Format of the data files.	
data_type	• unaligned reads • aligned reads • variants annotation • clinical supplement	★ Required	Specific content type of the data file.	
genomics_assay	• array • object	No	No Description	
core_metadata_collections	• array • object	No	No Description	
run_id	• string	No	Sequencing run ID associated with file	
alternate_timepoint	• string	No	If the data is not a baseline measurement, the timepoint name is defined here.	
file_name	• string	No	The name (or part of a name) of a file (of any type).	
file_size	• integer	No	The size of the data file (object) in bytes.	

Gen3 data modelling background

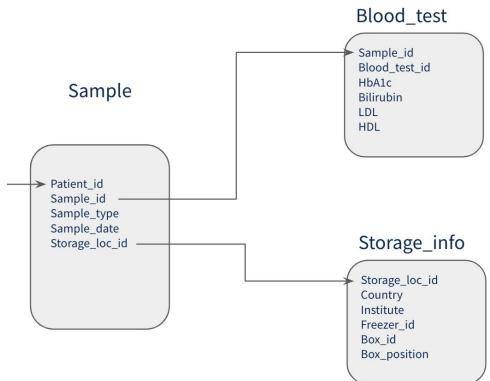


```
json
{
  "$schema": "http://json-schema.org/draft-07/schema#",
  "title": "User Profile",
  "type": "object",
  "properties": {
    "id": {
      "type": "integer"
    },
    "name": {
      "type": "string"
    },
    "email": {
      "type": "string",
      "format": "email"
    },
    "age": {
      "type": "integer",
      "minimum": 0
    }
  },
  "required": ["id", "name", "email"]
}
```

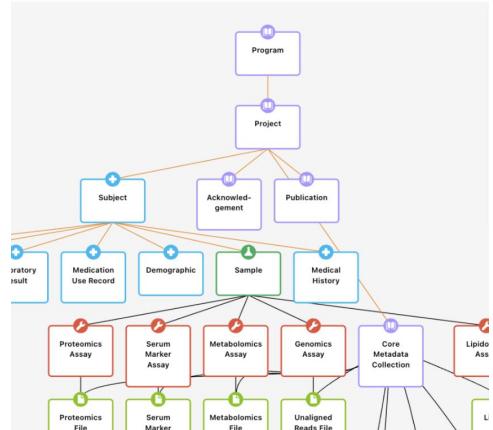


Gen3 data modelling background

???



```
json
{
  "$schema": "http://json-schema.org/draft-07/schema#",
  "title": "User Profile",
  "type": "object",
  "properties": {
    "id": {
      "type": "integer"
    },
    "name": {
      "type": "string"
    },
    "email": {
      "type": "string",
      "format": "email"
    },
    "age": {
      "type": "integer",
      "minimum": 0
    }
  },
  "required": ["id", "name", "email"]
}
```



Thoughts and Experiences with data modelling in Gen3



Thoughts and Experiences with data modelling in Gen3



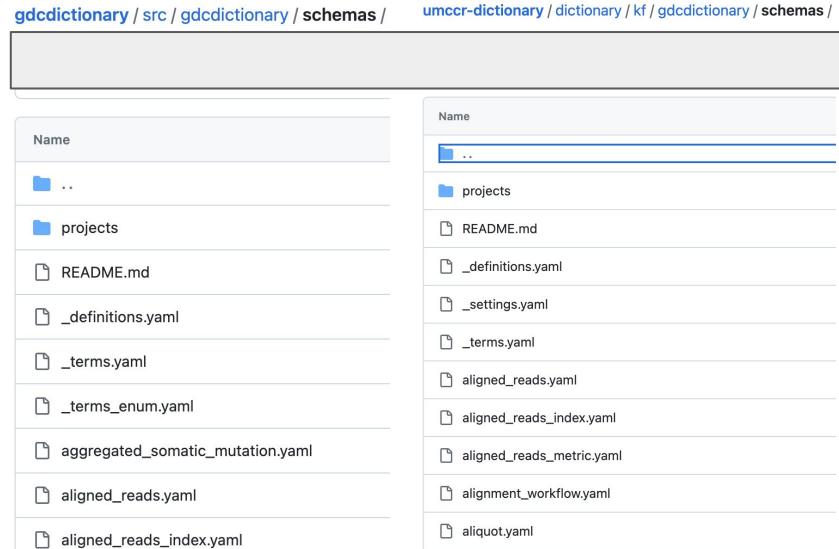
From a former bioinformatician and beginner data modeller.....

Re-using Data Objects from Other Gen3 Dictionaries

Thoughts and Experiences with data modelling in Gen3

Re-using Data Objects from Other Gen3 Dictionaries

- Advantages:
 - Potential for efficiency and consistency
 - Interoperability
 - Many schemas already available in repositories such as
- Challenges:
 - In some cases, this approach leads to dependency and reference complications in the schema



Utilising Common Data Models (CDMs)

Utilising Common Data Models (CDMs)

- Advantages:

- Adopting ontologies can help other users familiar with that ontology identify groups of data
- Can promote interoperability with other health data systems

- Limitations

- Requires a high level of expertise and domain knowledge
- Utilising a CDM in Gen3 requires conversion tools, e.g. `pfb_fhir` to jsonschema



Building Custom Data Models

Building Custom Data Models

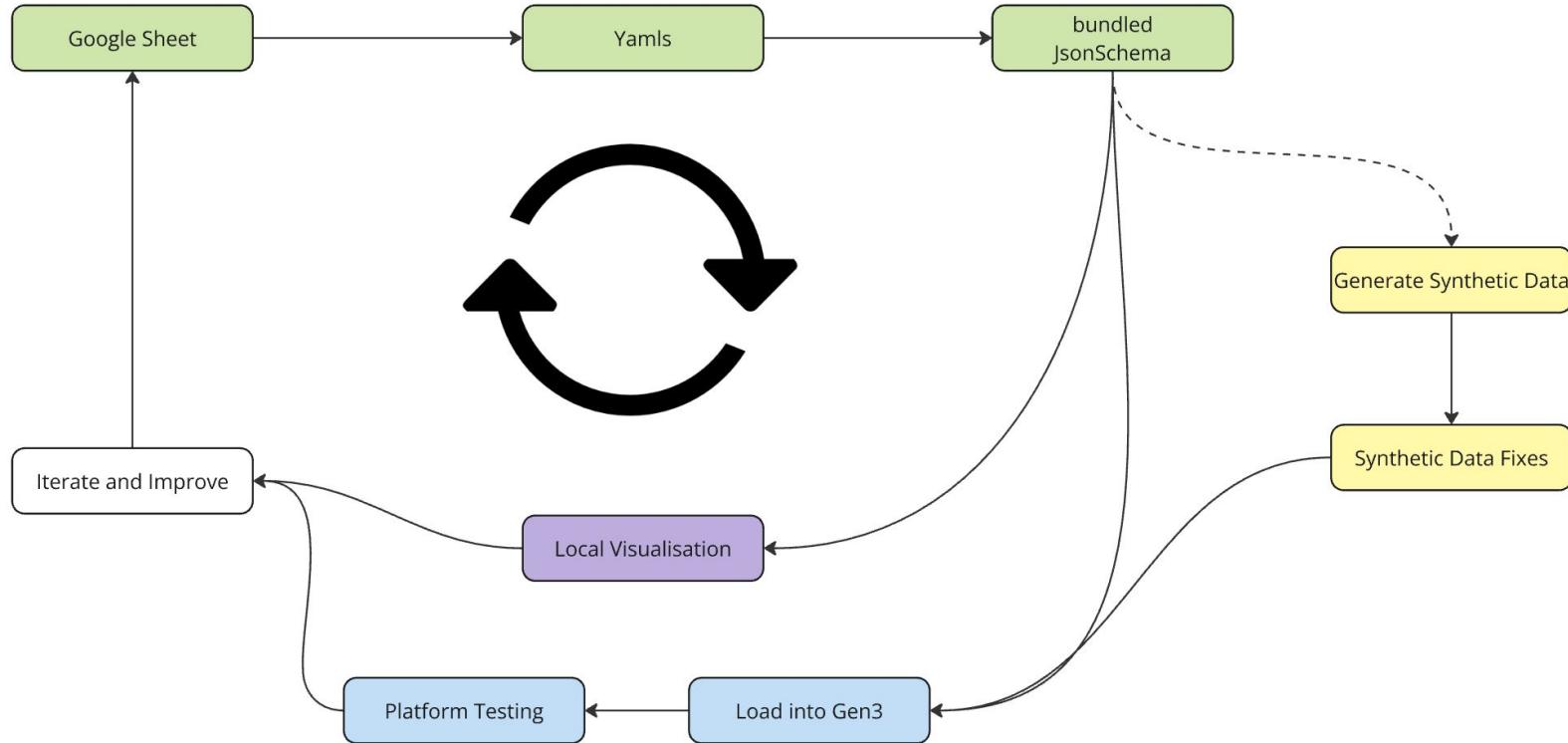
- Efficiency:
 - Building custom data models has provided the fastest and most efficient way of data modelling for our purposes so far
- Pipeline Development:
 - We have set up a data model development pipeline that allows for flexible and frequent updates and testing of our data model prior to acquiring real data

Need for Entry-Level Data Modelling Tools

Need for Entry-Level Data Modelling Tools

- Target Audience:
 - Teams of medical researchers or bioinformaticians without extensive experience in data modelling principles and techniques will struggle to adopt gen3
- Challenges with Current Tools:
 - Data modelling with raw JSON schema can be overwhelming for new users
 - We have devised a simplified approach to lower the barrier to entry

Overview of our data modelling process



Australian Biocommons - Gen3schemadev - Git repo



A screenshot of a GitHub repository page for "gen3schemadev".

The repository details:

- Owner: AustralianBioCommons
- Name: gen3schemadev
- Status: Public
- Last commit by: JoshuaHarris391
- Last commit message: added function to generate guid manifest file for recently submitted data
- Last commit time: 19 hours ago
- Commits: 187
- Branches: 4
- Tags: 0

The main repository page shows a list of recent commits:

File / Commit Message	Date
configs add mapping.json	2 years ago
docs added documentation to data_submitter.py script	3 weeks ago
file_type_templates first working version of synthetic data file generator	2 weeks ago
gen3schemadev added function to generate guid manifest file for recently ...	19 hours ago
jupyter added disclaimer about running the notebook standalone	3 months ago
schema added more stuff to ipynb	3 months ago
scripts minor script edits	2 years ago
users/_pycache_ moved usersync to own branch	5 months ago
.gitignore added .DS_Store to gitignore	3 months ago
LICENSE Create LICENSE	last year
Readme.md added documentation to data_submitter.py script	3 weeks ago

Contributors (3):

- mshadbolt Marion
- uvwxyz Uwe Winter
- JoshuaHarris391 Joshua Harris

Repository statistics:

- Code: 187 commits
- Issues: 0
- Pull requests: 0
- Actions: 0
- Projects: 0
- Wiki: 0
- Security: 0
- Insights: 0

Repository details sidebar:

- About: Gen3 Schema Development tools
- gen3
- Readme
- Apache-2.0 license
- Activity
- Custom properties
- 1 star
- 2 watching
- 0 forks
- Report repository

Releases:

- No releases published
- Create a new release

Packages:

- ...

Entry Level Data Modelling in Google Sheets

Data Modelling in google sheets utilises 4 main sheets:

object_definitions ▾

2 link_definitions ▾

1 property_definitions ▾

enum_definitions ▾

Entry Level Data Modelling in Google Sheets

Creating Object Nodes

D31 | fx

	A	B	C	D	E
1	ID	TITLE	CATEGORY	DESCRIPTION	DEFINITION_REFS
2	project	Project	administrative	▼ The study the data is coming from	
3	publication	Publication	administrative	▼ Publication for a project	
4	acknowledgement	Acknowledgement	administrative	▼ Acknowledgement of an individual or group involved in a pro	
5	sample	Sample	biospecimen	▼ Biospecimen information that links subjects to samples inclu	
6	subject	Subject	clinical	▼ An individual participant in the study with baseline measurer	
7	lab_result	Laboratory Result	clinical	▼ Measurements obtained from blood or other laboratory tests	
8	demographic	Demographic	clinical	▼ Data for the characterization of the patient by means of segm	
9	medical_history	Medical History	clinical	▼ Medical history of the participant	
10	exposure	Exposure	clinical	▼ Clinically relevant patient information relating to environment	
11	medication	Medication Use Record	clinical	▼ Records about historical or current medication use.	
12	blood_pressure_test	Blood Pressure Test	clinical	▼ Blood pressure reading (insert method here).	
13	aligned_reads_file	Aligned Reads File	data_file	▼ Data file containing aligned reads from a sequencing experi [data_file_properties]	
14	aligned_reads_index_file	Aligned Reads Index File	data_file	▼ Data file containing an index for a set of aligned reads [data_file_properties]	
15	unaligned_reads_file	Unaligned Reads File	data_file	▼ Data file containing raw reads from a sequencing experim [data_file_properties]	
16	genomics_assay	Genomics Assay	experimental_methods	▼ Details about the methods used to produce genomic output	
17	lipidomics_file	Lipidomics File	data_file	▼ Data file containing lipidomics data [data_file_properties]	

Entry Level Data Modelling in Google Sheets



Defining Links between Objects

Entry Level Data Modelling in Google Sheets

Properties

H24	A	B	C	D	E	F	G	H	I
1	VARIABLE_NAME	OBJECT	REQUIRED	TYPE	DESCRIPTION	PREFERRED	FORMAT	PATTERN	TERM_REF
2	contact_type	acknowledgement	TRUE	enum_role	The type of contact or role in the project, e.g. Principal				
3	orcid	acknowledgement	FALSE	string	The ORCID number for the acknowledgee			$^{\wedge}[0\{4\}-\text{d}\{4\}-\text{d}\{4\}-(\text{d}\{3\}\text{X})\text{d}\{4\}\$}$	
4	acknowledgee	acknowledgement	TRUE	string	Name of the individual or group to be acknowledged.				
5	data_type	aligned_reads_file	TRUE	enum_data_type	x				data_type
6	data_format	aligned_reads_file	TRUE	enum_align_data_form	Format of the data files.				data_format
7	data_category	aligned_reads_file	TRUE	enum_seq_data_cat	Broad categorization of the contents of the data file.				data_category
8	run_id	aligned_reads_file	FALSE	string	Sequencing run ID associated with file				
9	reference_genome_build	aligned_reads_file	FALSE	enum_ref_genome	Reference genome used e.g. GRCh37.			$^{\wedge}\text{GRCh}[0-9]\{2\}\$$	
10	consent_codes	aligned_reads_file	FALSE	array	Data Use Restrictions that are used to indicate permit				
11	baseline_timepoint	aligned_reads_file	TRUE	boolean	Based on the Data Use Ontology : see http://www				
12	alternate_timepoint	aligned_reads_file	FALSE	string	Does the data reflect a baseline measurement?				
13	data_type	aligned_reads_index	TRUE	enum_data_type	If the data is not a baseline measurement, the timepoi				data_type
14	data_format	aligned_reads_index	TRUE	enum_index_data_form	Specific content type of the data file.				
15	data_category	aligned_reads_index	TRUE	enum_seq_data_cat	Format of the data files.				data_category
16	baseline_timepoint	aligned_reads_index	TRUE	boolean	Broad categorization of the contents of the data file.				
17	alternate_timepoint	aligned_reads_index	FALSE	string	Does the data reflect a baseline measurement?				
18	workflow_type	alignment_workflow	TRUE	enum_align_work	If the data is not a baseline measurement, the timepoi				
19	workflow_end_datetime	alignment_workflow	FALSE	string	Type of read aligner used				
20	workflow_link	alignment_workflow	FALSE	string	A combination of date and time of day in the form [-]C	date-time		$^{\wedge}\text{d}\{4\}-\text{d}\{2\}-\text{d}\{2\}\text{T}\text{d}\{2\}:\text{d}\{2\}:\text{d}\{2\}(?:\.\text{d}+)?(?:\text{Z} [+-]\text{d}\{2\}:\text{d}\{2\}:\text{d}\{2\})$	
21	workflow_start_datetime	alignment_workflow	FALSE	string	Link to Github hash for the CWL workflow used.				
22	workflow_version	alignment_workflow	FALSE	string	A combination of date and time of day in the form [-]C	date-time		$^{\wedge}\text{d}\{4\}-\text{d}\{2\}-\text{d}\{2\}\text{T}\text{d}\{2\}:\text{d}\{2\}:\text{d}\{2\}(?:\.\text{d}+)?(?:\text{Z} [+-]\text{d}\{2\}:\text{d}\{2\}:\text{d}\{2\})$	
					Version of the workflow used				

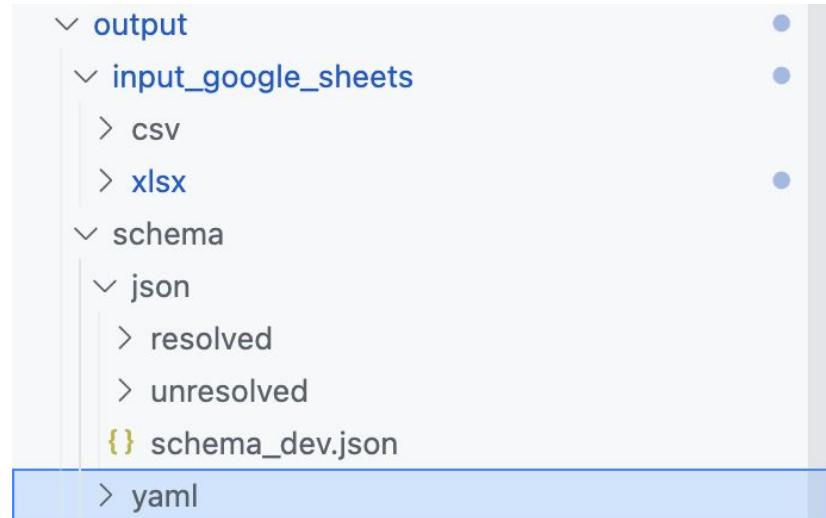
Entry Level Data Modelling in Google Sheets

Enums

A1	type_name	enum	enum_definition	source	term_id	version
1	type_name	enum				
2	enum_activity	sufficient	sufficient	Active Australia		
3	enum_activity	insufficient	insufficient	Active Australia		
4	enum_activity	sedentary	sedentary	Active Australia		
5	enum_activity	not collected				
6	enum_activity	not stated or inadequately described				
7	enum_collection	baseline				
8	enum_collection	first follow-up				
9	enum_collection	second follow-up				
10	enum_seq_data_cat	analysis				
11	enum_seq_data_cat	sequencing reads				
12	enum_seq_data_cat	single nucleotide variation				
13	enum_seq_data_cat	transcriptome profiling				
14	enum_seq_data_cat	supplemental				

Entry Level Data Modelling in Google Sheets

- **sheet2yaml-CLI.py** Reads google sheets and converts to yaml
- **gen3schemadev** library has functions to also download the current state of the google sheet for your records and reproducibility
- **umCCR-g3po** for compiling yaml to jsonschema
-



Validation and Visualisation of Data Model

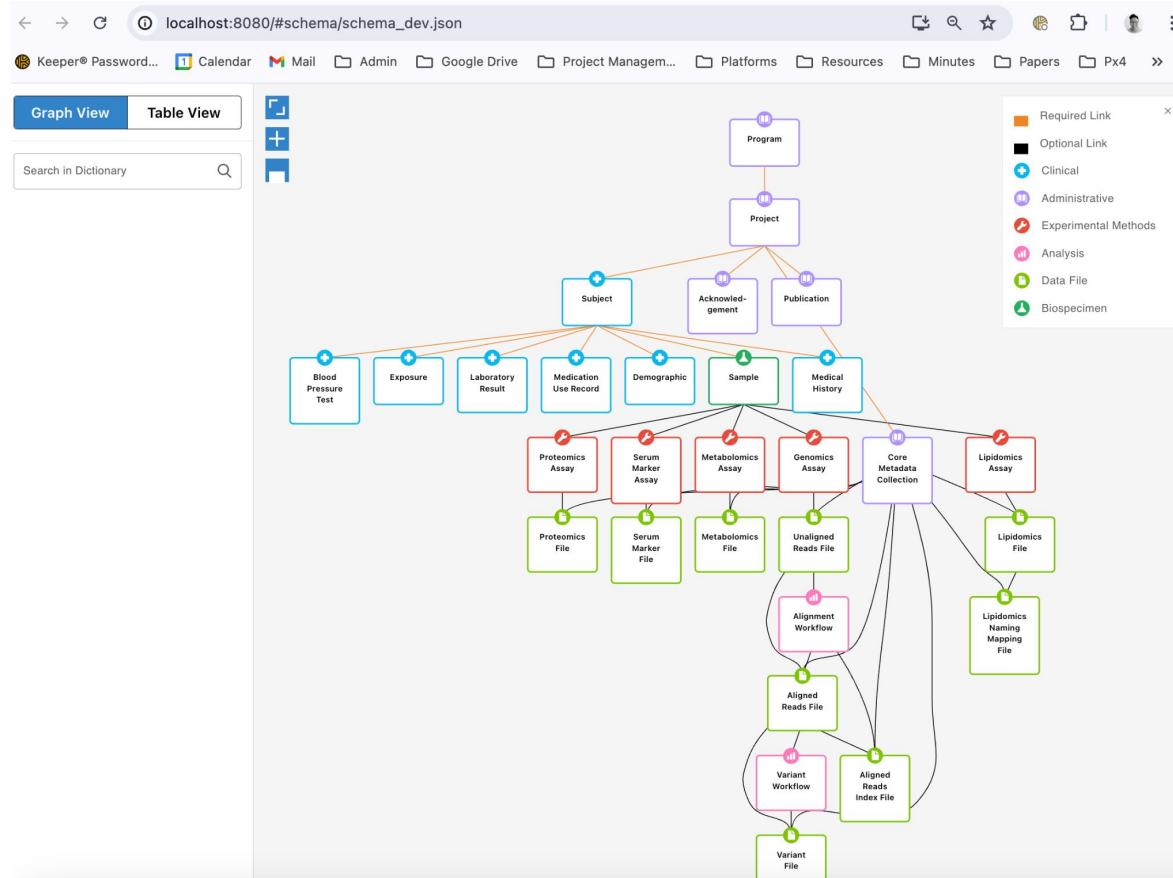
- Uc-cdis: data-simulator

```
1 # Running Validation
2 !cd umCCR-dictionary && make validate program=schema_dev
[2] ✓ 1.8s
...
Using .env-sample
Validating Data Dictionary: schema_dev
[2024-07-10 02:34:42,654] [data-simulator] [ INFO] Data simulator initialization...
[2024-07-10 02:34:42,656] [data-simulator] [ INFO] Loading dictionary from url http://ddvis/schema/schema\_dev.json
[2024-07-10 02:34:42,738] [data-simulator] [ INFO] Initializing graph...
[2024-07-10 02:34:42,738] [data-simulator] [ INFO] Validating...
[2024-07-10 02:34:42,740] [data-simulator] [ INFO] Done!
```

▷ ▾

```
1 # Visualising data dictionary
2 !open http://localhost:8080/#schema/schema\_dev.json
[1] ✓ 0.5s
```

Validation and Visualisation of Data Model



Validation and Visualisation of Data Model

localhost:8080/#schema/schema_dev.json

Graph View Table View

Search in Dictionary

Data Model Structure

- program
- project
- 4 nodes with 6 links
- unaligned_reads_file

[Close properties](#) [Download templates](#)

data_file Data file containing raw reads from a sequencing experiment.

Unaligned Reads File

Property	Type	Required	Description	Term
submitter_id	• string	★ Required	A project-specific identifier for a node. This property is the calling card/nickname/alias for a unit of submission. It can be used in place of the UUID for identifying or recalling a node.	
type	• string	★ Required	No Description	
baseline_timepoint	• boolean	★ Required	Does the data reflect a baseline measurement?	
data_category	• analysis • sequencing reads • single nucleotide variation • transcriptome profiling • supplemental	★ Required	Broad categorization of the contents of the data file.	
data_format	• fastq	★ Required	Format of the data files.	
data_type	• unaligned reads • aligned reads • variants annotation • clinical supplement	★ Required	Specific content type of the data file.	
genomics_assay	• array • object	No	No Description	
core_metadata_collections	• array • object	No	No Description	
run_id	• string	No	Sequencing run ID associated with file	
alternate_timepoint	• string	No	If the data is not a baseline measurement, the timepoint name is defined here.	
file_name	• string	No	The name (or part of a name) of a file (of any type).	
file_size	• integer	No	The size of the data file (object) in bytes.	

Synthetic Data Creation



uc-cdis: data-simulator

```
1 # Generating synthetic metadata using umccr-dictionary
2 !cd umccr-dictionary && make simulate program=schema_dev project=AusDiab max_samples=110
3 !cd umccr-dictionary && make simulate program=schema_dev project=BioHEART-CT max_samples=50
4 !cd umccr-dictionary && make simulate program=schema_dev project=FIELD max_samples=100
```

7]

```
[2024-07-09 08:02:42,505] [data-simulator][ INFO] Data simulator initialization...
[2024-07-09 08:02:42,506] [data-simulator][ INFO] Loading dictionary from url http://ddvis/schema/schema\_dev.json
[2024-07-09 08:02:42,556] [data-simulator][ INFO] Initializing graph...
[2024-07-09 08:02:42,557] [data-simulator][ INFO] Generating data...
[2024-07-09 08:02:42,559] [data-simulator simulate][ INFO] Simulating data for node project
[2024-07-09 08:02:42,707] [data-simulator simulate][ INFO] Simulating data for node subject
[2024-07-09 08:02:42,745] [data-simulator simulate][ INFO] Simulating data for node demographic
[2024-07-09 08:02:42,805] [data-simulator simulate][ INFO] Simulating data for node sample
[2024-07-09 08:02:42,880] [data-simulator simulate][ INFO] Simulating data for node serum_marker_assay
[2024-07-09 08:02:42.922] [data-simulator simulate][ INFO] Simulating data for node aenomics assay
```

Synthetic Data Creation

Then **gen3schemadev** - plausible_data_gen.py

object	property	data_type	schema_type	mean	sd	median	first_quart	third_quart	proportion	range_start	range_end	source	enum
blood_pressure_test	bp_systolic	mean	number	129.4	18.7							Barr et al. 2007	
blood_pressure_test	bp_diastolic	mean	number	70.2	11.8							Barr et al. 2007	
demographic	year_birth	range	number							1955	1984		
demographic	month_birth	range	number							1	12		
demographic	baseline_age	mean	integer	51.4	14.2							Barr et al. 2007	
demographic	bmi_baseline	mean	number	27	5							Barr et al. 2007	
demographic	height_baseline	mean	number	1.7	0.25								
demographic	weight_baseline	mean	number	80	5								
exposure	cigarettes_per_day	mean	integer	10.7	6							ABS	
lab_result	total_cholesterol	mean	number	5.66	1.07							Barr et al. 2007	
lab_result	hdl	mean	number	1.42	0.38							Barr et al. 2007	
lab_result	ldl	mean	number	3.984	1.06							calculated from TC, HDL & trigs	
lab_result	triglycerides	median	number				1.28	0.89	1.9			Barr et al. 2007	
lab_result	glucose_fasting	mean	number	5.5	1							Dunstan et al. 2010	
lab_result	hba1c_ngsp	mean	number	5.5	0.1							AHS 2013	
lab_result	hba1c_ifcc	mean	number	36.62	1.09							Conversion NGSP-->IFCC = (10.9:	
lab_result	creatinine_serum_enzymatic	mean	number	93.71	19.05							Odden et al. 2009	
lab_result	creatinine_urinary	mean	number	12	6.3							Cocker et al. 2011	
lab_result	age_at_collection	mean	integer	51.4	14.2							Barr et al. 2007	
lab_result	egfr_baseline	mean	number	85.5	0.1							AHS 2013	
medical_history	hypertension	proportion	string						0.325			Barr et al. 2007	enum_yes_no
medical_history	incident_diabetes	proportion	string						0.032			Dunstan et al. 2011	enum_yes_no
medication	lipid_lowering_medication	proportion	string						0.086			Barr et al. 2007	enum_yes_no
medication	antihypertensive_meds	proportion	string									AusDiab	enum_yes_no
medication	diabetes_therapy	proportion	string									AusDiab	enum_anti_diabet

Data Model and Synthetic Data Version Management



A screenshot of a GitHub repository page for "AustralianBioCommons / ACDCSchemaDev".

The repository is a private template.

Key statistics shown:

- Code: 4
- Issues: 0
- Pull requests: 0
- Actions: 0
- Projects: 0
- Security: 0
- Insights: 0
- Settings: 0

Branches: main (selected), 3 Branches, 5 Tags

Commits:

Author	Message	Time
gen3schemadev @ a6d09cb	Generated linked synthetic data files, updated DD to only ...	2 weeks ago
output	node links to core metadata are now last in order, change...	last week
screenshots	added screenshot of dd	last month
umccr-dictionary @ 550396f	added git ignore to umccr	3 months ago
.DS_Store	not final, but all synth metadata has passed validation	3 weeks ago
.gitignore	fixed entity relationships and regen synth data	last week
.gitmodules	node links to core metadata are now last in order, change...	last week
LICENSE	Update LICENSE	3 months ago
README.md	updated readme	3 weeks ago
generate_synthetic_data.ipynb	node links to core metadata are now last in order, change...	last week
schema_dev_framework.ipynb	node links to core metadata are now last in order, change...	last week
submit_data.ipynb	node links to core metadata are now last in order, change...	last week
synth_data_transformations.ipynb	node links to core metadata are now last in order, change...	last week

Watch: 4 | Fork: 0 | Starred: 1 | Use this template

About

No description, website, or topics provided.

Readme | Apache-2.0 license | Activity | Custom properties

1 star | 4 watching | 0 forks

Releases 5

v0.1.3 (Latest) 2 weeks ago

+ 4 releases

Packages

No packages published [Publish your first package](#)

Languages

Data Model and Synthetic Data Version Management



We manage our schema version and matching synthetic data batches with git releases

3 weeks ago
JoshuaHarris391
v0.1.1
797ce31
Compare ▾

v0.1.1

Full Changelog: [v0.1.0...v0.1.1](#)

Summary:

- Fixed data dictionary by adding back compulsory gen3 properties (data_type, data_format, data_category)
- Fixed ISO8601 regex format pattern in workflow nodes
- This release now has a batch of synthetic metadata that passed validation (using my gen3 metadata validator)
- No dummy files generated yet, will still need to write the scripts to better generate them.

Assets 2

- Source code (zip) 3 weeks ago
- Source code (tar.gz) 3 weeks ago



Jun 4
JoshuaHarris391
v0.1.0
94649f0
Compare ▾

v0.1.0

Release v0.1.0: UAT Data Dictionary

Hi Team,

We have released version 0.1.0 of our data dictionary. This data dictionary version will be loaded onto the UAT test system and facilitate the generation, transformation, and loading of synthetic data onto the UAT ACDC platform.

Reason for Release

Benefits and Limitations of our method



- Advantages:
 - Easy to use
 - Low barrier of entry
 - Good for prototyping
 - Can help you create the bulk of your data model before working explicitly with jsonschema
 - google sheets used to compile the json schema is saved for reproducibility
- Limitations
 - Not reverse compatible (json schema -> google sheet)
 - Can only incorporate CDM elements or other gen3 data objects after compilation to jsonschema

Future Work

- Potentially package this workflow and tools into an open source project
- Reverse engineer jsonschema back to google sheet
- Finalise tools for gen3 data model node/object ingestion

Acknowledgements

Funders:

- BPA - Bioplatforms Australia



- MRFF - Medical Research Future Fund - Australia



Acknowledgements

- **Speakers**
 - Nathalie Giraudon, New Zealand eScience Infrastructure (NeSI)
 - Plamen Martinov, Open Commons Consortium
 - Chris Meyer, Center for Translational Data Science, University of Chicago
 - Liam Beckman, Oregon Health and Science University
 - Joshua Harris, Australian BioCommons
- **Gen3 Forum Steering Committee**
 - Robert Grossman - Center for Translational Data Science, University of Chicago
 - Steven Manos - Australian BioCommons
 - Claire Rye - New Zealand eScience Infrastructure
 - Plamen Martinov - Open Commons Consortium
 - Michael Fitzsimons - Center for Translational Data Science, University of Chicago