



University of
Zurich^{UZH}

arrayMap



EMBL-EBI



Global Alliance
for Genomics & Health



GA4GH Metadata & Beyond

*Implementing the GA4GH schema specifications
on top of the arrayMap cancer genome resource*

Michael Baudis | ELIXIR All Hands 2017 | 21-23 March | Rome | Italy



GA4GH API promotes sharing

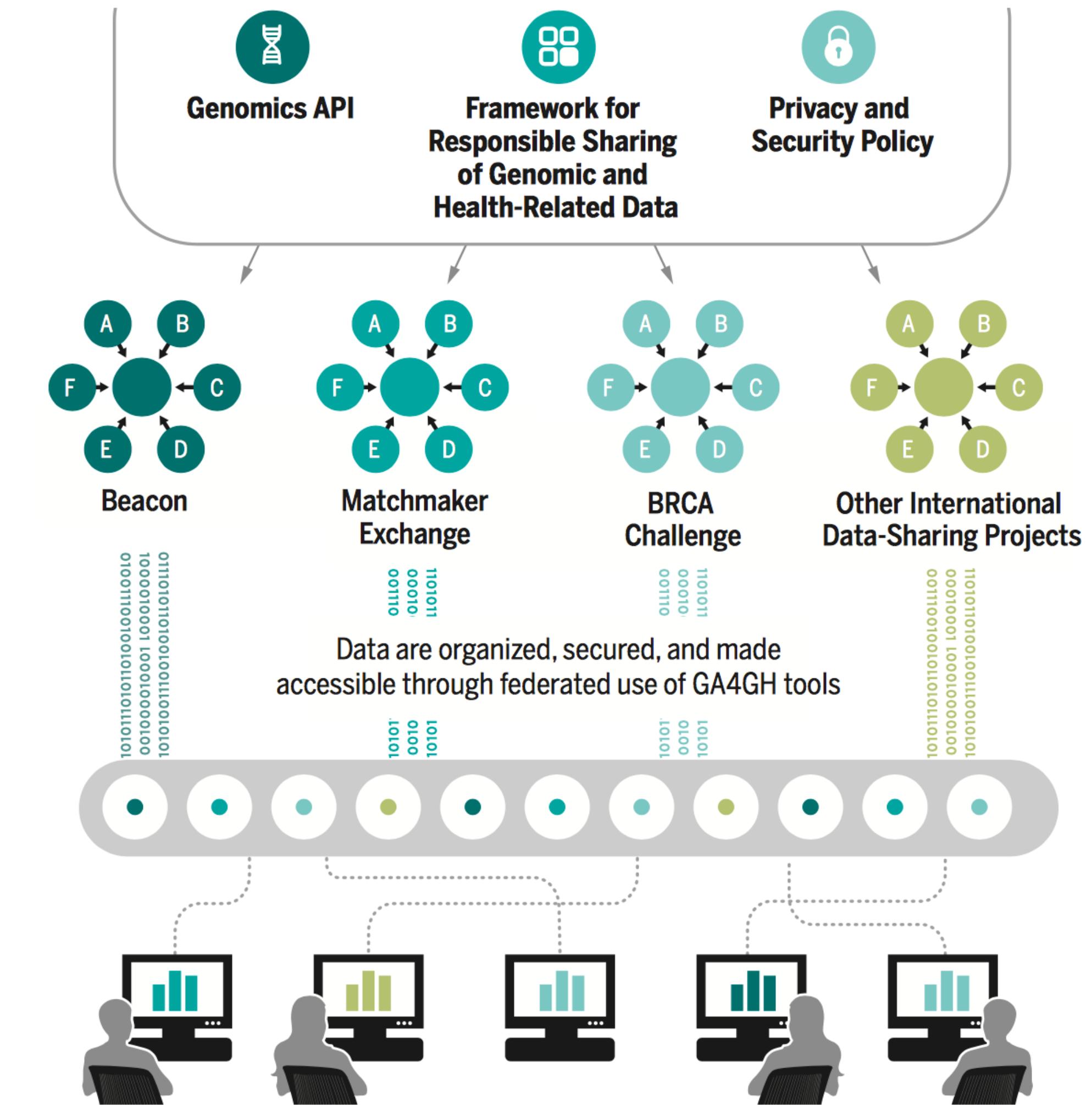
A federated data ecosystem. To share genomic data globally, this approach furthers medical research without requiring compatible data sets or compromising patient identity.



GENOMICS

A federated ecosystem for sharing genomic, clinical data

Silos of genome data collection are being transformed into seamlessly connected, independent systems





This repository

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ga4gh / schemas

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Work on data models and APIs for Genomic data. <http://ga4gh.org/#/api>

1,102 commits

17 branches

16 releases

46 contributors

Apache-2.0

Branch: [metadata-integ...](#) ▾[New pull request](#)[Create new file](#)[Upload files](#)[Find file](#)[Clone or download](#) ▾

This branch is 15 commits ahead, 3 commits behind master.

[Pull request](#) [Compare](#)**mbaudis** Merge branch 'master' into metadata-integration

Latest commit 077c2c7 2 days ago



Merge branch 'master' into metadata-integration

2 days ago



Add constraints file

2 days ago



Utilize new common methods in schemas

2 days ago



Merge branch 'master' into metadata-integration

13 days ago



Utilize new common methods in schemas

2 days ago



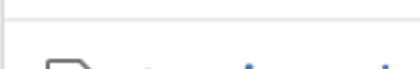
Merge branch 'master' into metadata-integration

13 days ago



Remove protoc call from install path (#781)

7 days ago



Add constraints file

2 days ago



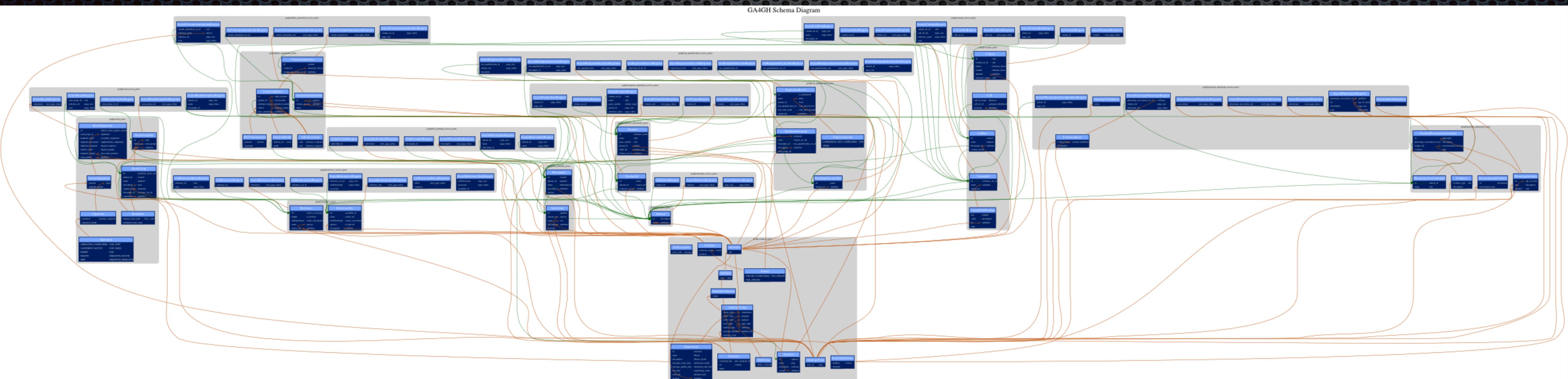
Convert Avro -> proto3.

10 months ago

The State of the Schema, Feb 2017



Global Alliance for Genomics & Health





Reference Resources for Cancer Genome Profiling

- curated reference resources for cancer genome profiling data and related information
- basis for own research activities, collaborative projects and external use
- structured information serves for implementing GA4GH concepts

	progenet	arrayMap
techniques	cCGH, aCGH, WES, WGS	aCGH (+?)
scope	sample (e.g. combination of several experiments)	experiment
content	>31000 samples	>60000 arrays
raw data presentation	no (link to sources if available)	yes (raw, log2, segmentation if available)
per sample re-analysis	no; supervised result (mostly as provided through publication)	yes (re-segmentation, thresholding, size filters ...)
final data	annotated/interpreted CN status for GP and cytogenetic regions	unsupervised CN status for GP and cytogenetic regions
main purposes	<ul style="list-style-type: none">Distribution of CNA target regions in most tumor types (>350 ICD-O)Cancer classification	<ul style="list-style-type: none">Gene specific hitsGenome feature correlation (fragile sites ...)

arrayMap

Resource for copy number variation data in cancer

arrayMap 

visualizing cancer genome array data @ arraymap.org

arrayMap is a curated reference database and bioinformatics resource targeting copy number profiling data in human cancer. The arrayMap database provides an entry point for meta-analysis and systems level data integration of high-resolution oncogenomic CNA data.

The current data reflects:

- 63060 genomic copy number arrays
- 763 experimental series
- 145 array platforms
- ICD-O** 141 ICD-O cancer entities
- 554 publications (Pubmed entries)

 University of Zurich UZH

Citation User Guide Registration & Licensing People External Links ↗ FOLLOW US ON [twitter](#)

 130.60.23.21

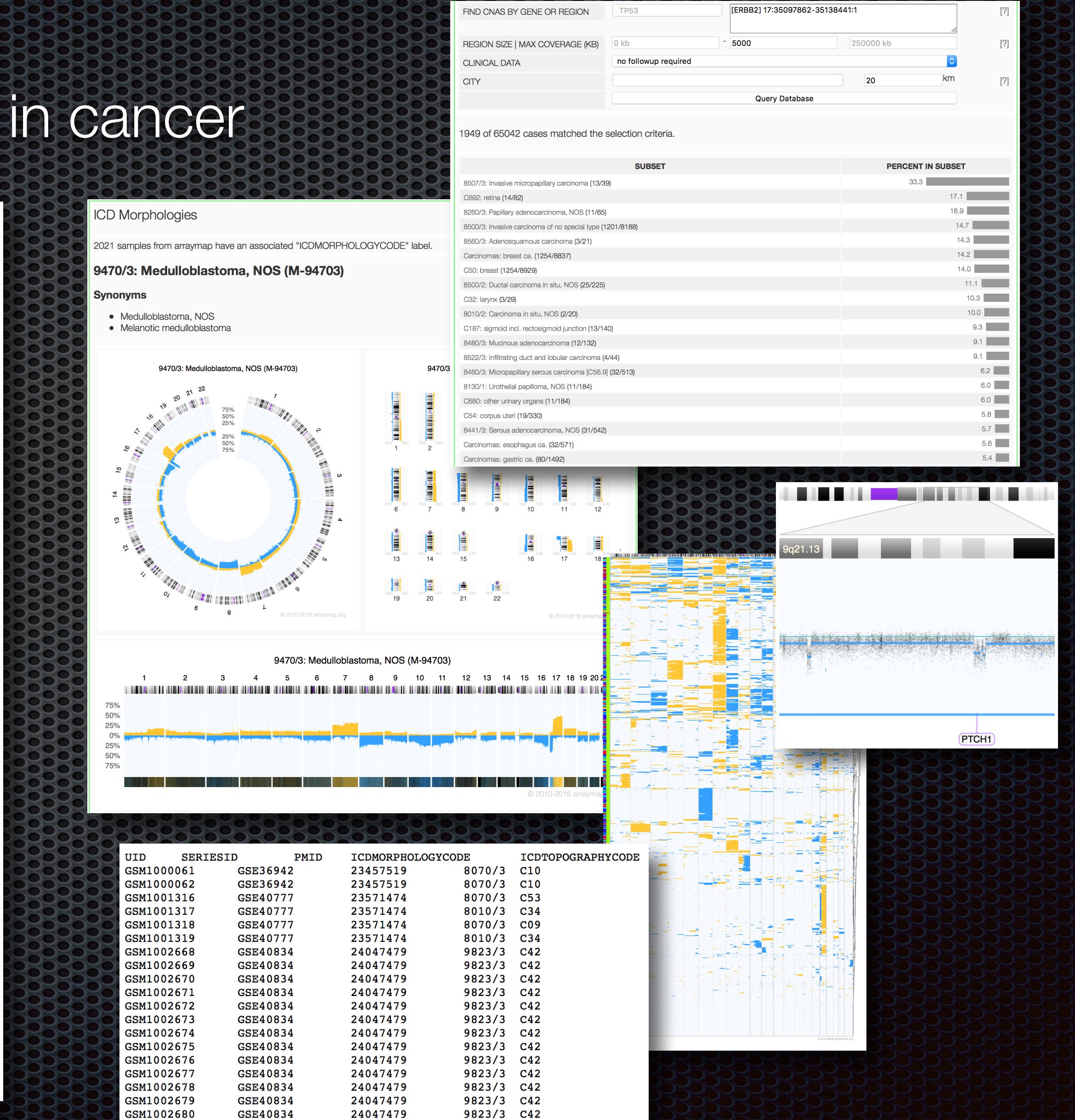
For the majority of the samples, probe level visualization as well as customized data representation facilitate gene level and genome wide data review. Results from multi-case selections can be connected to downstream data analysis and visualization tools, as we provide through our Progenetix project.

arrayMap is developed by the group "Theoretical Cytogenetics and Oncogenomics" at the Institute of Molecular Life Sciences of the University of Zurich.

BRAIN TUMOURS	5653 samples ↗	[?]
BREAST CANCER	8329 samples ↗	[?]
COLORECTAL CANCER	3238 samples ↗	[?]
PROSTATE CANCER	991 samples ↗	[?]
STOMACH CANCER	1062 samples ↗	[?]
ARRAYMAP NEWS	2016-08-03: SVG graphics 2016-05-17: Transitioning to Europe PMC More news ...	

Feel free to use the data and tools for academic research projects and other applications. If more support and/or custom analysis is needed, please contact Michael Baudis regarding a collaborative project or a special license.

© 2000 - 2016 Michael Baudis, refreshed Mon, 19 Sep 2016 10:20:09 GMT in 6.87s on server 130.60.240.68. No responsibility is taken for the correctness of the data presented nor the results achieved with the Progenetix tools.



Developing the GA4GH Metadata Schema

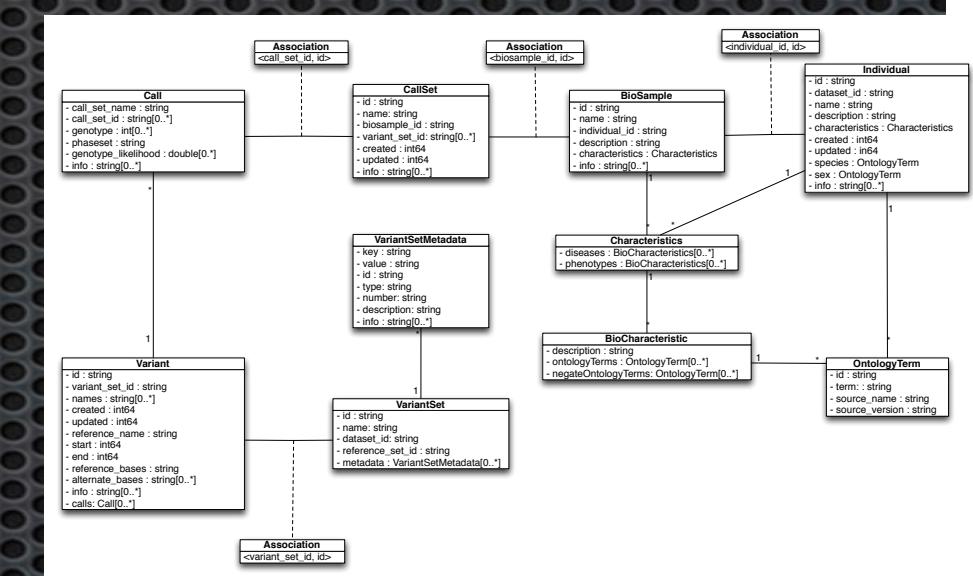
▶ arrayMap for GA4GH

- metadata schema development through implementation of arrayMap resource data
 - OntologyTerm objects for biodata
 - implementation w/ ontology services

Driving Beacon Development

▶ Beacon+

- CNV/CNA as first type of structural variants
 - disease specific queries
 - quantitative reporting



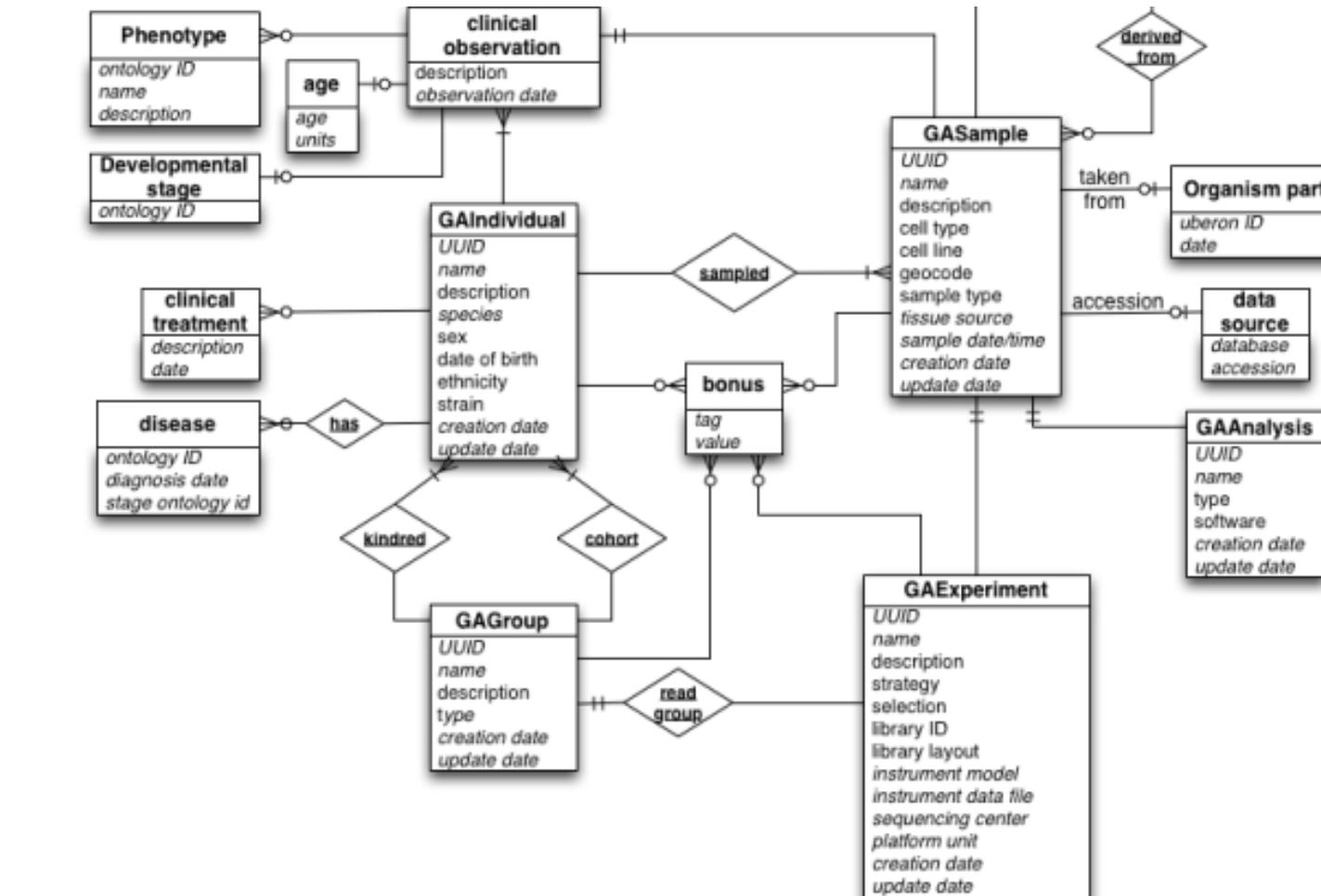
```
{  
    "_id" : ObjectId("58297ca32ca4591e5a0df054"),  
    "id" : "AM_V_1778741",  
    "variant_set_id" : "AM_VS_HG18",  
    "reference_name" : "10"  
    "start" : 579049,  
    "end" : 17236099,  
    "alternate_bases" : "DUP",  
    "reference_bases" : ".",  
    "info" : {  
        "svlen":16657050,  
        "cipos": [  
            -1000,  
            1000  
        ],  
        "ciend": [  
            -1000,  
            1000  
        ]  
    },  
    "calls" : [  
        {  
            "genotype" : [  
                ".",  
                ".  
            ],  
            "call_set_id" : "AM_CS_TCGA-61-1917-01A-01D-0648-01",  
            "info" : {  
                "segvalue" : 0.5491  
            }  
        }  
    ],  
    "created" : ISODate("2016-11-14T08:33:58.202Z"),  
    "updated" : ISODate("2016-11-14T08:33:58.202Z"),  
}
```

Meta Data: Everything but the sequence

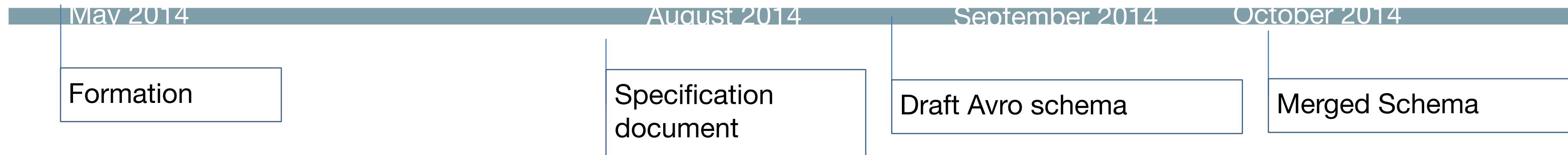


Frameworks for consistent description of annotated data domains:

- *GAExperiment* Technical data - bridge to Reads Task Team
- *GAIIndividual* Clinical data - bridge to Clinical and Regulatory & Ethics
- *GAIIndividualGroup* Static or dynamically generated semantic collections
- *GAAnalysis* Interpretation & methodology of one or more
- *GASample* Biological information using common ontologies
 - Species neutral but focus on human use cases
 - Standardised ontologies for feature annotations
 - Next steps:
 - ontology recommendations (with CWG)
 - model refinement through implementation
 - validation and data transformation tools

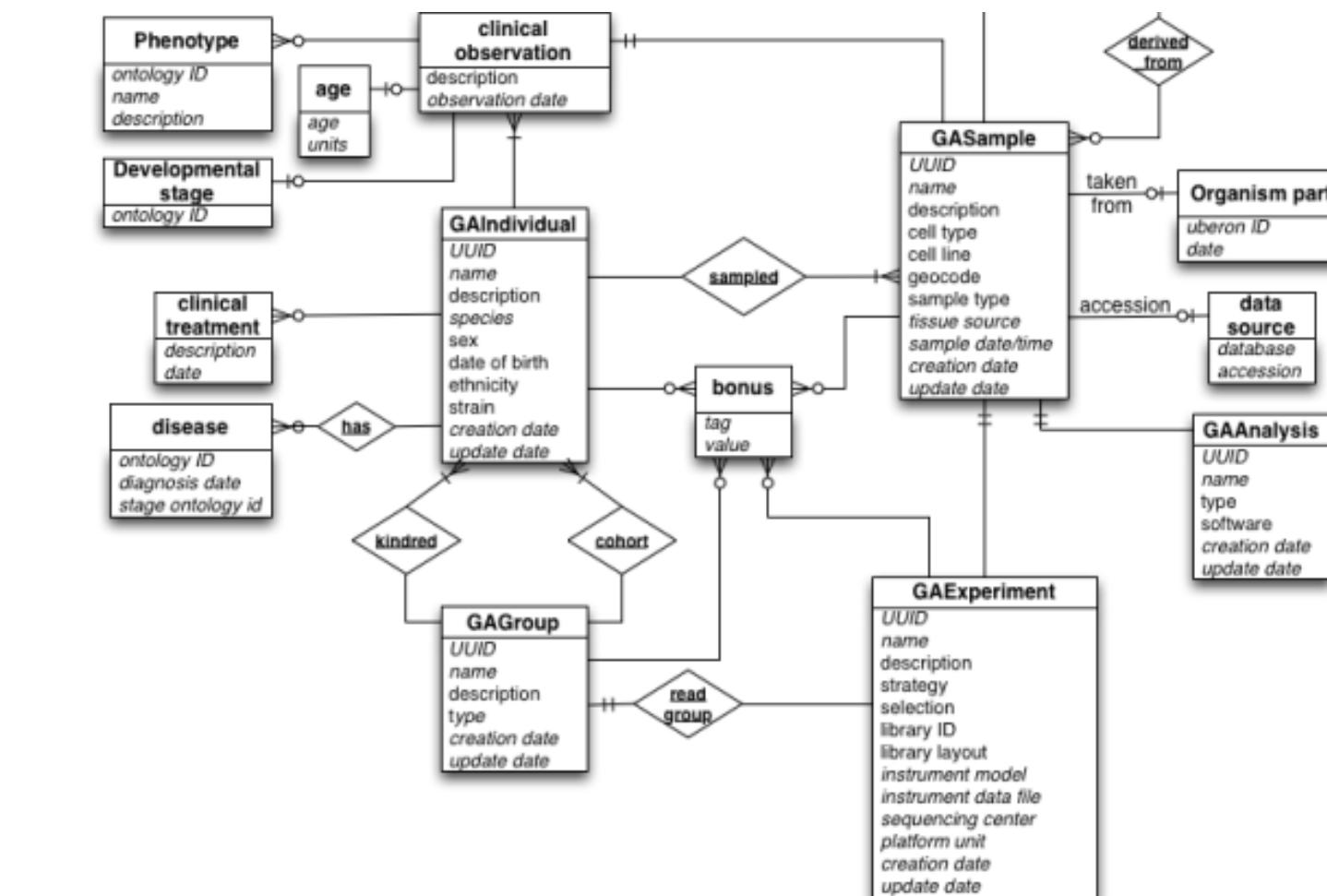


Meta Data: Everything but the sequence



Frameworks for consistent description of annotated data domains:

- *GAExperiment* Technical data - bridge to Reads Task Team
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 - Next steps:
 - ontology recommendations (with CWG)
 - model refinement through implementation
 - validation and data transformation tools



“Metadata” Objects

metadata branch, December 2015

Individual
accessions
dateOfBirth
description
developmentalStage
diseases
ethnicity
geographicLocation
guid
id
info
interventions
name
observations
phenotypes
recordCreateTime
recordUpdateTime
sex
species
strain

BioSample
accessions
ageAtSampling
cellLine
cellType
description
geographicLocation
guid
id
individualIds
info
interventions
name
observations
organismPart
preservationMethod
recordCreateTime
recordUpdateTime
samplingDate
sex
species
specimenType

Experiment
description
id
info
instrumentDataFile
instrumentModel
molecule
name
platformId
platformName
preparationId
processingFacility
recordCreateTime
recordUpdateTime
runTime
selection
strategy

```
record OntologyTerm {  
    union { null, string } ontologySourceID = null;  
    union { null, string } ontologySourceName = null;  
    union { null, string } ontologySourceVersion = null;  
}  
  
record GeographicLocation {  
    union { null, string } description = null;  
    union { null, float } elevation = null;  
    union { null, float } latitude = null;  
    union { null, float } longitude = null;  
}  
  
record Observation {  
    union { null, string } ageAtObservation = null;  
    union { null, string } dateTimeObserved = null;  
    union { null, string } id = null;  
    OntologyTerm observation;  
    union { null, string } unit = null;  
    OntologyTerm value;  
}  
record Evidence {  
    union { null, string } description = null;  
    OntologyTerm evidenceType;  
}  
  
record Dataset {  
    array<string> accessions;  
    union { null, string } description = null;  
    union { null, string } guid = null;  
    string id;  
    array<string> memberIds;  
    union { null, string } name = null;  
}  
  
record Disease {  
    }  
    union { null, string } ageOfOnset = null;  
    union { null, string } dateTimeDiagnosis = null;  
    OntologyTerm disease;  
    union { null, OntologyTerm } stageAtDiagnosis = null;  
}
```

State of the schema

Biosample from arrayMap

2017-02-17

```
1  {
2      "_id" : ObjectId("589dfa5109d374e4f3655aee"),
3      "name" : "AM_BS_GSM322223",
4      "individual_id" : "PGIND_GSM322223",
5      "id" : "AM_BS_GSM322223",
6      "characteristics" : {
7          "diseases" : [
8              {
9                  "ontologyTerms" : [
10                     {
11                         "termLabel" : "Chronic Lymphocytic Leukemia",
12                         "termId" : "NCIT:C3163"
13                     },
14                     {
15                         "termLabel" : "B-cell chronic lymphocytic leukemia/small lymphocytic lymphoma",
16                         "termId" : "SNMI:M-98233"
17                     },
18                     {
19                         "termLabel" : "B-cell chronic lymphocytic leukemia/small lymphocytic lymphoma",
20                         "termId" : "ICDOM:9823_3"
21                     },
22                     {
23                         "termLabel" : "hematopoietic and reticuloendothelial systems",
24                         "termId" : "ICDOT:C42"
25                     }
26                 ],
27                 "negatedOntologyTerms" : [ ],
28                 "description" : "Chronic Lymphocytic Leukemia"
29             }
30         ],
31         "phenotypes" : [ ]
32     },
33     "description" : "Chronic Lymphocytic Leukemia",
34     "info" : {
35         "tnm" : "T1",
36         "death" : "0",
37         "country" : "Sweden",
38         "geo_long" : 17.64,
39         "redirected_to" : "null",
40         "followup_months" : 68,
41         "geo_lat" : 59.86,
42         "pubmed_id" : "18484635",
43         "sex" : "female",
44         "age" : 59,
45         "city" : "uppsala"
46     },
47     "updated" : ISODate("2017-02-10T17:15:02.380Z"),
48     "created" : ISODate("2017-02-10T17:15:02.380Z")
49 }
```

- object model instead of named attributes
- referencing of ontologies instead of text descriptors

- need for **ontologies** & mappings
- **these** are no “real” open ontologies

- curating phenotypic data into ontologies
- fallback to key:value map for unassigned data; this should disappear over time

```
"_id" : ObjectId("589dfa5109d374e4f3655aee"),
"name" : "AM_BS_GSM322223",
"individual_id" : "PGIND_GSM322223",
"id" : "AM_BS_GSM322223",
"characteristics" : {
  "diseases" : [
    {
      "ontologyTerms" : [
        {
          "termLabel" : "B-cell chronic lymphocytic leukemia/small lymphocytic lymphoma",
          "termId" : "SNMI:M-98233"
        },
        {
          "termLabel" : "B-cell chronic lymphocytic leukemia/small lymphocytic lymphoma",
          "termId" : "ICDOM:9823_3"
        }
      ],
      "negatedOntologyTerms" : [ ],
      "description" : "Chronic Lymphocytic Leukemia"
    }
  ],
  "phenotypes" : [ ],
  "description" : "Chronic Lymphocytic Leukemia",
  "info" : {
    "tnm" : "T1",
    "death" : "0",
    "country" : "Sweden",
    "geo_long" : 17.64,
    "redirected_to" : "null",
    "followup_months" : 68,
    "geo_lat" : 59.86,
    "pubmed_id" : "18484635",
    "sex" : "female",
    "age" : 59,
    "city" : "uppsala"
  },
  "updated" : ISODate("2017-02-10T17:15:02.380Z"),
  "created" : ISODate("2017-02-10T17:15:02.380Z")
```

Working towards ontologies: Mapping >55'000 samples from ICD-O to NCIt (neoplasm core)

example_dx	ICDMORPHOLOGY	ICDOM	ICDTOPOGRAPHY	ICDOT	NCIT:CODE
malignant melanoma [metastatic cell line MaMel19]	Malignant melanoma NOS	8720/3	skin	C44	C3224
malignant melanoma [vagina]	Malignant melanoma NOS	8720/3	vagina and labia	C510	C3224
malignant melanoma [uvea metastasized]	Malignant melanoma NOS	8720/3	retina	C692	C3224
meningioma	Meningioma NOS	9530/0	meninges cerebral spinal	C700	C3230
mesothelioma	Mesothelioma NOS	9050/3	lung and bronchus	C34	C3234
pleural mesothelioma	Mesothelioma NOS	9050/3	pleura	C384	C3234
mesothelioma	Mesothelioma NOS	9050/3	connective and soft tissue NOS	C499	C3234
multiple myeloma	Plasma cell myeloma	9732/3	hematopoietic and reticuloendothelial system	C42	C3242
Mycosis fungoides	Mycosis fungoides	9700/3	skin	C44	C3246
Myelodysplastic syndrome	Myelodysplastic syndrome NOS	9989/3	hematopoietic and reticuloendothelial system	C42	C3247
Acute myeloblastic leukemia with maturation [FAB M2]	Acute myeloblastic leukemia with maturation [FAB M2]	9874/3	hematopoietic and reticuloendothelial system	C42	C3250
neuroblastoma	Neuroblastoma NOS	9500/3	peripheral nerves incl. autonomous	C47	C3270
Cerebral neuroblastoma [cerebral region midline frontal lobe]	Neuroblastoma NOS	9500/3	cerebrum	C710	C3270
neuroblastoma [adrenal gland cell line]	Neuroblastoma NOS	9500/3	adrenal gland	C76	C3270
Cutaneous neurofibroma	Neurofibroma NOS	9540/0	skin	C44	C3272
Plexiform neurofibroma	Neurofibroma NOS	9540/0	Nervous system NOS	C729	C3272
Oligodendrogioma [Supratentorial Frontal Lobe]	Oligodendrogioma NOS	9450/3	cerebrum	C710	C3288
oligodendrogioma	Oligodendrogioma NOS	9450/3	Brain NOS	C719	C3288
oligodendrogioma	Oligodendrogioma NOS	9450/3	brain nos	c719	C3288
Paraganglioma	Paraganglioma NOS	8680/1	Nervous system NOS	C729	C3308
paraganglioma	paraganglioma NOS	8680/1	adrenal cortex	C740	C3308
Pheochromocytoma	Pheochromocytoma NOS	8700/0	adrenal cortex	C740	C3326
polycythemia vera	Polycythemia vera	9950/3	hematopoietic and reticuloendothelial system	C42	C3336
pediatric rhabdomyosarcoma	Rhabdomyosarcoma NOS	8900/3	connective and soft tissue NOS	C499	C3359
Sezary syndrome	Sezary syndrome	9701/3	hematopoietic and reticuloendothelial system	C42	C3366
sezary syndrome	sezary syndrome	9701/3	skin	C44	C3366
Synovial sarcoma	Synovial sarcoma NOS	9040/3	connective and soft tissue NOS	C499	C3400
essential thrombocythemia	Essential thrombocythemia	9962/3	hematopoietic and reticuloendothelial system	C42	C3407
carcinosarcoma	Carcinosarcoma NOS	8980/3	connective and soft tissue NOS	C499	C34448
Carcinosarcoma [breast cell line HS578T]	Carcinosarcoma NOS	8980/3	breast	C50	C34448
acute monocytic leukemia	Monocytic leukemia NOS	9860/3	hematopoietic and reticuloendothelial system	C42	C3171
leiomyoblastoma	Epithelioid leiomyoma	8891/0	kidney	C649	C3157
colon mucosa [low grade dysplastic tumor; myhmut]	atypical adenoma	8140/1	large intestine excl. rectum and rectosigmoid	C189	C7559

State of the schema

Biosample from arrayMap

2017-03-20

```
1 {
2     "id" : "PGX_AM_BS_GSM510730",
3     "individual_id" : "PGX_IND_GSM510730",
4     "name" : "PGX_AM_BS_GSM510730",
5     "description" : "breast carcinoma",
6     "bio_characteristics" : [
7         {
8             "description" : "breast carcinoma",
9             "ontology_terms" : [
10                 {
11                     "term_id" : "NCIT:C4017",
12                     "term_label" : "Ductal Breast Carcinoma"
13                 },
14                 {
15                     "term_id" : "SNMI:M-85003",
16                     "term_label" : "invasive carcinoma of no special type"
17                 },
18                 {
19                     "term_id" : "PGX:ICDOM:8500_3",
20                     "term_label" : "invasive carcinoma of no special type"
21                 },
22                 {
23                     "term_id" : "PGX:ICDOT:C50",
24                     "term_label" : "breast"
25                 },
26                 {
27                     "term_id" : "PGX:SEER:26000",
28                     "term_label" : "Breast"
29                 }
30             ],
31             "negated_ontology_terms" : [ ],
32         }
33     ],
34     "individual_age_at_collection" : "P47Y",
35     "attributes" : {
36         "tnm" : {
37             "values" : [
38                 {
39                     "string_value" : "T1N0M0"
40                 }
41             ]
42         },
43         "death" : {
44             "values" : [
45                 {
46                     "string_value" : "0"
47                 }
48             ]
49         },
50         "country" : {
51             "values" : [
52                 {
53                     "string_value" : "Norway"
54                 }
55             ]
56         },
57         "city" : {
58             "values" : [
59                 {
60                     "string_value" : "Oslo"
61                 }
62             ]
63         },
64         "geo_lat" : {
65             "values" : [
66                 {
67                     "double_value" : 59.91
68                 }
69             ]
70         },
71         "geo_long" : {
72             "values" : [
73                 {
74                     "double_value" : 10.75
75                 }
76             ]
77         },
78     },
79     "external_identifiers" : [
80         {
81             "database" : "Pubmed",
82             "identifier" : "20592421"
83         },
84         {
85             "database" : "GEO",
86             "identifier" : "GSM510730"
87         },
88         {
89             "database" : "GEO",
90             "identifier" : "GSE20394"
91         }
92     ],
93     "created" : ISODate("2017-03-20T08:37:07.771Z"),
94     "updated" : ISODate("2017-03-20T08:37:07.771Z")
95 }
```

- drop of diseases | phenotypes wrapper used to type characteristics (discussion with code integration team at UCSC)
- **TODO**: introduce alternative characteristic_type label?
- introduction of “PGX” prefix for “ontologized” local versions of ICD-O 3 etc.

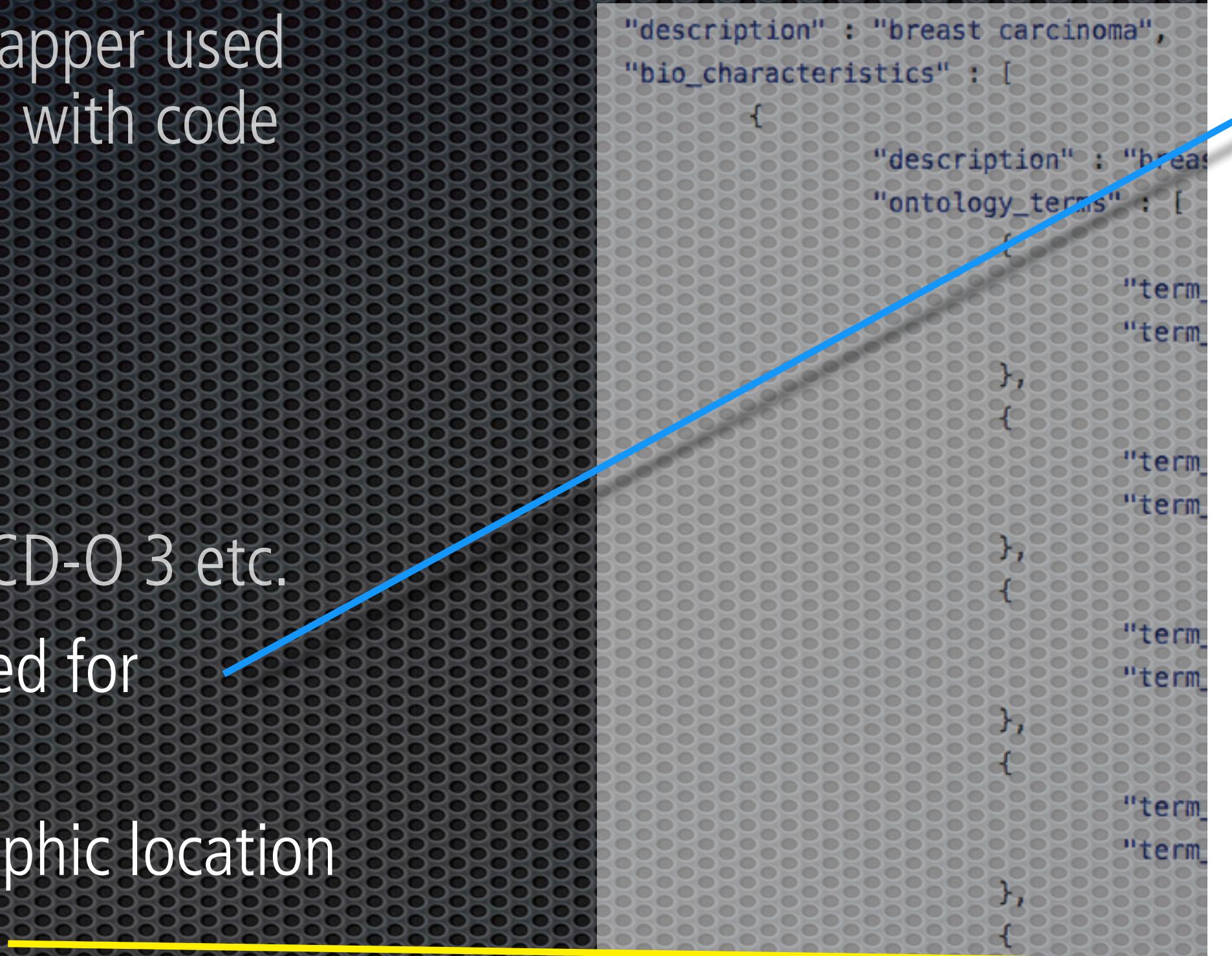
```

1 {
2   "id" : "PGX_AM_BS_GSM510730",
3   "individual_id" : "PGX_IND_GSM510730",
4   "name" : "PGX_AM_BS_GSM510730",
5   "description" : "breast carcinoma",
6   "bio_characteristics" : [
7     {
8       "description" : "breast carcinoma",
9       "ontology_terms" : [
10         {
11           "term_id" : "NCIT:C4017",
12           "term_label" : "Ductal Breast Carcinoma"
13         }
14       ]
15     }
16   ],
17   "negated_ontology_terms" : [ ],
18 },
19 ],
20 ]
21
22
23
24
25
26
27
28
29
30
31
32
33
34
35
36
37
38
39
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59
60
61
62
63
64
65
66
67
68
69
70
71
72
73
74
75
76
77
78
79   "external_identifiers" : [
80     {
81       "database" : "Pubmed",
82       "identifier" : "20592421"
83     },
84     {
85       "database" : "GEO",
86       "identifier" : "GSM510730"
87     },
88     {
89       "database" : "GEO",
90       "identifier" : "GSE20394"
91     }
92   ],
93   "created" : ISODate("2017-03-20T08:37:07.771Z"),
94   "updated" : ISODate("2017-03-20T08:37:07.771Z")
95 }

```

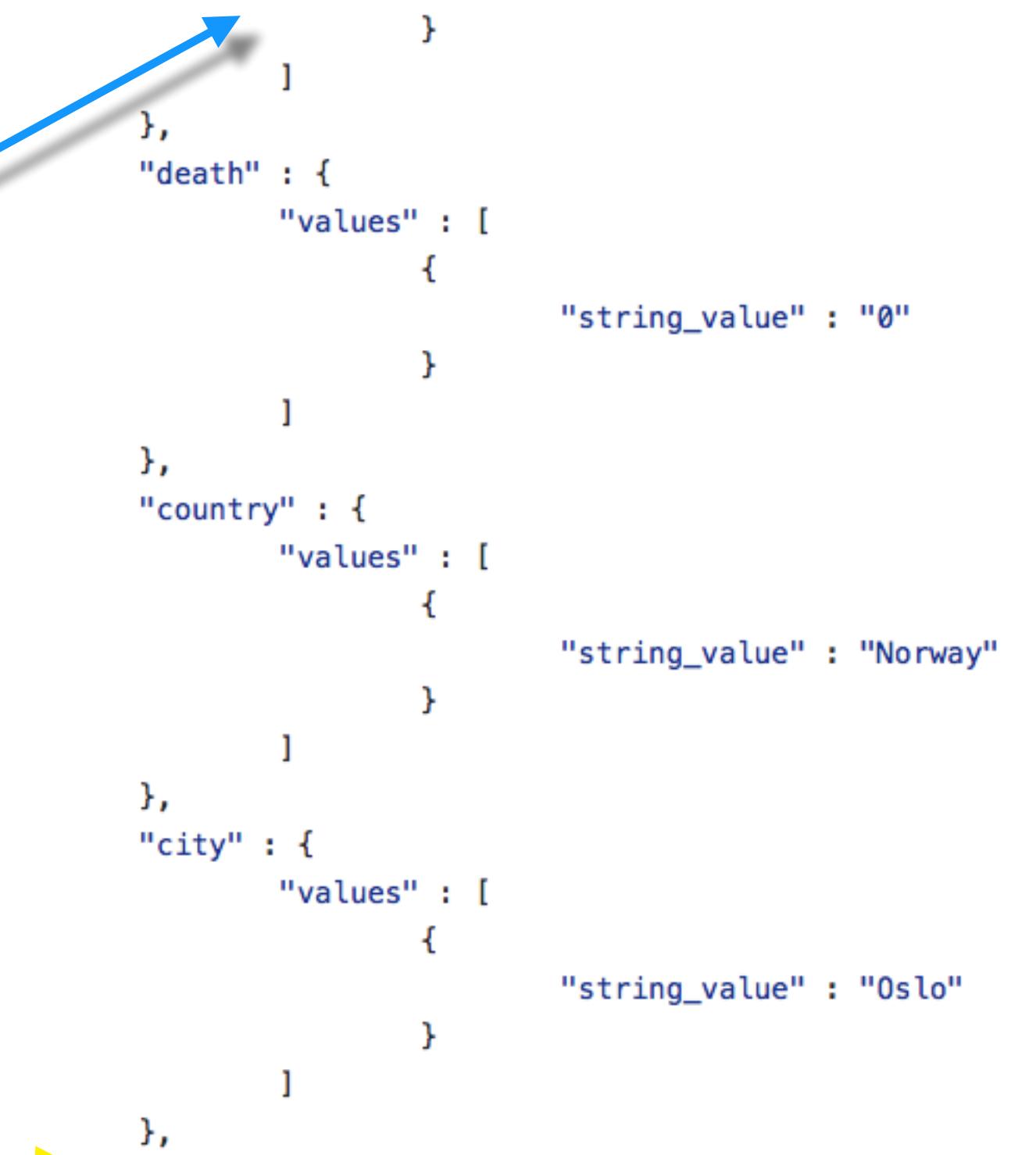
The diagram shows a flow from a list of tasks on the left to a JSON schema on the right. A blue arrow points from the first task (drop of diseases) to the 'bio_characteristics' section of the schema. A yellow arrow points from the third task ('ontologized' local versions of ICD-O 3 etc.) to the 'ontology_terms' section of the schema.

- drop of diseases | phenotypes wrapper used to type characteristics (discussion with code integration team at UCSC)
- **TODO:** introduce alternative characteristic_type label?
- introduction of “PGX” prefix for “ontologized” local versions of ICD-O 3 etc.
- typed constructors now being used for “arbitrary” attributes
- **TODO:** Finish design of a geographic location object



```

{
  "description" : "breast carcinoma",
  "bio_characteristics" : [
    {
      "description" : "breast carcinoma"
    }
  ],
  "ontology_terms" : [
    {
      "term" : "T1N0M0"
    },
    {
      "term" : "0"
    }
  ],
  "country" : {
    "values" : [
      {
        "string_value" : "Norway"
      }
    ]
  },
  "city" : {
    "values" : [
      {
        "string_value" : "Oslo"
      }
    ]
  },
  "geo_lat" : {
    "values" : [
      {
        "double_value" : 59.91
      }
    ]
  },
  "geo_long" : {
    "values" : [
      {
        "double_value" : 10.75
      }
    ]
  }
}
  
```



```

{
  "id" : "PGX_AM_BS_GSM510730",
  "individual_id" : "PGX IND GSM510730",
  "attributes" : {
    "tnm" : {
      "values" : [
        {
          "string_value" : "T1N0M0"
        }
      ]
    },
    "death" : {
      "values" : [
        {
          "string_value" : "0"
        }
      ]
    }
  },
  "country" : {
    "values" : [
      {
        "string_value" : "Norway"
      }
    ]
  },
  "city" : {
    "values" : [
      {
        "string_value" : "Oslo"
      }
    ]
  },
  "geo_lat" : {
    "values" : [
      {
        "double_value" : 59.91
      }
    ]
  },
  "geo_long" : {
    "values" : [
      {
        "double_value" : 10.75
      }
    ]
  }
}
  
```

94 "updated" : ISODate("2017-03-20T08:37:07.771Z"),
95 }

GeoLocation object proposal

- general consensus about some sort of **geographic attribution**, for a yet to be determined subset of GA4GH records/objects
- GeoJSON itself is not a good option for storing data, but rather for indicating objects on a maplocal **obfuscation** approaches will be needed for privacy protection
- destructive obfuscation can recode addresses to (random) points in higher level administrative boundaries
- the combination of *lat*, *long* with a location name seems a good compromise, with a “**precision level**” providing additional features (e.g. possibility to randomize point locations in a given boundary)

```
message GeoLocation {  
    // a text representation, preferably using standard geographic identification  
    // elements, of the corresponding latitude,longitude(),altitude()  
    // This representation serves the purposes to  
    // - capture standard data entry parameters  
    // - provide a sanity check for latitude,longitude values  
    // Example:  
    // - 34 Washington Blvd, Venice Beach, Los Angeles, CA, United States  
    // - Str Marasesti 5, 300077 Timisoara, Romania  
    // - Heidelberg, Deutschland  
    string geo_label = 1;  
  
    // an optional indication of the maximum precision to be derived from the  
    // latitude,longitude values  
    // Example:  
    // Given a street address "Winterthurerstrasse 190, 8057 Zürich, Switzerland",  
    // a privacy driven (destructive) obfuscation approach could recode this  
    // to  
    // "latitude": 47.37, "longitude": 8.54  
    // while providing  
    // "geo_precision":"city", "geo_label": "Zürich, Switzerland"  
    // ... indicating that the original location could correspond to any  
    // latitude,longitude point value inside the administrative boundaries of  
    // the city of Zürich, Switzerland  
    string geo_precision = 2;  
  
    // signed decimal degrees (North, relative to Equator)  
    double latitude = 3;  
  
    // signed decimal degrees (East, relative to IERS Reference Meridian)  
    double longitude = 4;  
  
    // optional, e.g. for environmental samples  
    double altitude = 5;  
}
```


- sex described here, not at Biosample level
- referencing of ontologies instead of text descriptors

- diseases & phenotypes which are associated with the individual are recorded here
- diseases can also include those for which biosample records exist (independent of the specific annotation there)

```
{  
    "id" : "PGX_IND_GSM847445",  
    "name" : "PGX_IND_GSM847445_edited",  
    "species" : {  
        "term_id" : "NCBITaxon:9606",  
        "term_label" : "Homo sapiens"  
    },  
    "sex" : {  
        "term_id" : "PATO:0020002",  
        "term_label" : "female genotypic sex"  
    },  
    "description" : "individual with Li-Fraumeni syndrome",  
    "bio_characteristics" : [  
        {  
            "description" : "Li-Fraumeni syndrome carrier",  
            "ontology_terms" : [  
                {  
                    "term_id" : "DOID:3012",  
                    "term_label" : "Li-Fraumeni syndrome",  
                },  
                {  
                    "term_id" : "NCIT:C9325",  
                    "term_label" : "Adrenal Cortex Carcinoma",  
                }  
            ]  
        },  
        {  
            "description" : "adrenocortical carcinoma",  
            "ontology_terms" : [  
                {  
                    "term_label" : "Adrenal Cortex Carcinoma",  
                    "term_id" : "NCIT:C9325",  
                }  
            ]  
        }  
    ],  
    "attributes" : null,  
    "created" : ISODate("2017-03-20T08:37:07.771Z"),  
    "updated" : ISODate("2017-03-20T08:37:07.771Z"),  
}  
}
```

Structural Variants from arrayMap

- name here could be e.g. an rsid
- calls are embedded in the variant set
- alternative option?
- “info” provides an intensity measurement
- could e.g. provide QC, copy number count ...
- Housekeeping needed?

```
{  
  "id" : "AM_V_3110636",  
  "name" : null,  
  "reference_name" : "11",  
  "reference_bases" : ".",  
  "start" : 75085926,  
  "alternate_bases" : ".",  
  "end" : 75744338,  
  "variant_type" : "DEL",  
  "svlen" : 658412,  
  "info" : {},  
  "calls" : [  
    {  
      "call_set_id" : "AM_CS_GSM902433",  
      "info" : {  
        "segvalue" : -0.3881  
      },  
      "genotype" : [ ".", "." ]  
    },  
    {  
      "call_set_id" : "AM_CS_GSM902435",  
      "info" : {  
        "segvalue" : -0.4112  
      },  
      "genotype" : [ ".", "." ]  
    }  
  "updated" : ISODate("2017-02-10T17:15:02.380Z"),  
  "created" : ISODate("2017-02-10T17:15:02.380Z"),  
}
```

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Schema examples

21 commits

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 mbaudis	edited individual entry	...	Latest commit 34234a7 6 hours ago
 data	changing the project structure		2 months ago
 examples	edited individual entry		6 hours ago
 README.md	links		21 days ago

 README.md

Implementation of the GA4GH schema based on genome profiles and metadata from arrayMap

This repository will contain data and information regarding the arrayMap based implementation of a GA4GH schema structure. While it is not expected that GA4GH compliant resources mirror the schema in their internal structure, this

Beacon+ Concept

- Implementation of cancer beacon prototype, backed by arrayMap data
- structural variations
- quantitative queries
- metadata
- current version uses GA4GH schema compatible, non-SQL database backend (MongoDB)

Heinz Stockinger, Séverine Duvaud & SIB Technology Group

Beacon arrayMap

Beacon v0.4 implementation for arrayMap.



Reference name	<input type="text" value="9"/>
Start	<input type="text" value="42049214"/>
Length	<input type="text" value="1000"/>
Assembly ID	<input type="text" value="GRCh36"/>
Dataset Ids	<input type="text" value="(9440/3) 9440/3: Glioblastoma, NOS (2047)"/>
Alternate bases	<input type="text" value="DEL (Deletion)"/>
Confidence Interval (Start position)	<input type="text" value="500"/>
Confidence Interval (End position)	<input type="text" value="500"/>
Match type	<input type="text" value="Complete"/>

SIB

Beacon Query **Beacon Info**

arrayMap

GA4GH Metadata

“Everything but the sequence”

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