

{S}[B] SchemaBlocks

GA4GH Standards Documentation and Alignment Initiative



Global Alliance for Genomics & Health

Collaborate. Innovate. Accelerate.

[Scientists Seek Order to Potential Confusion of Gene Data](#)

Bloomberg - Drew Armstrong & Robert Langreth

June 5, 2013

[Q&A: David Altshuler on How to Share Millions of Human Genomes](#)

Science - Jocelyn Kaiser

June 7, 2013

~~[DNA data to be shared worldwide in medical research project](#)~~

The Guardian - Ian Sample

June 5, 2013

[Geneticists push for global data-sharing](#)

Nature - Erika Check Hayden

June 5, 2013

~~[Accord Aims to Create Global Trove of Genetic Data](#)~~

The New York Times - Gina Kolata

June 5, 2013



[New alliance aims to create international system for sharing genomic data](#)

The Globe and Mail - By André Picard

June 5, 2013

[Poking Holes in Genetic Privacy](#)

The New York Times - Gina Kolata

June 16, 2013

[Our Genes, Their Secrets](#)

The New York Times

June 18, 2013

~~[White House Open Science 'Champions' Highlights Genomic Data Pioneers](#)~~

GenomeWeb

June 19, 2013

[Global alliance to create framework for sharing genomic data](#)

The Boston Globe - Carolyn Y. Johnson

June 5, 2013

[Une alliance pour partager les données génomiques et cliniques](#)

Le Monde - Sandrine Cabut

June 14, 2013

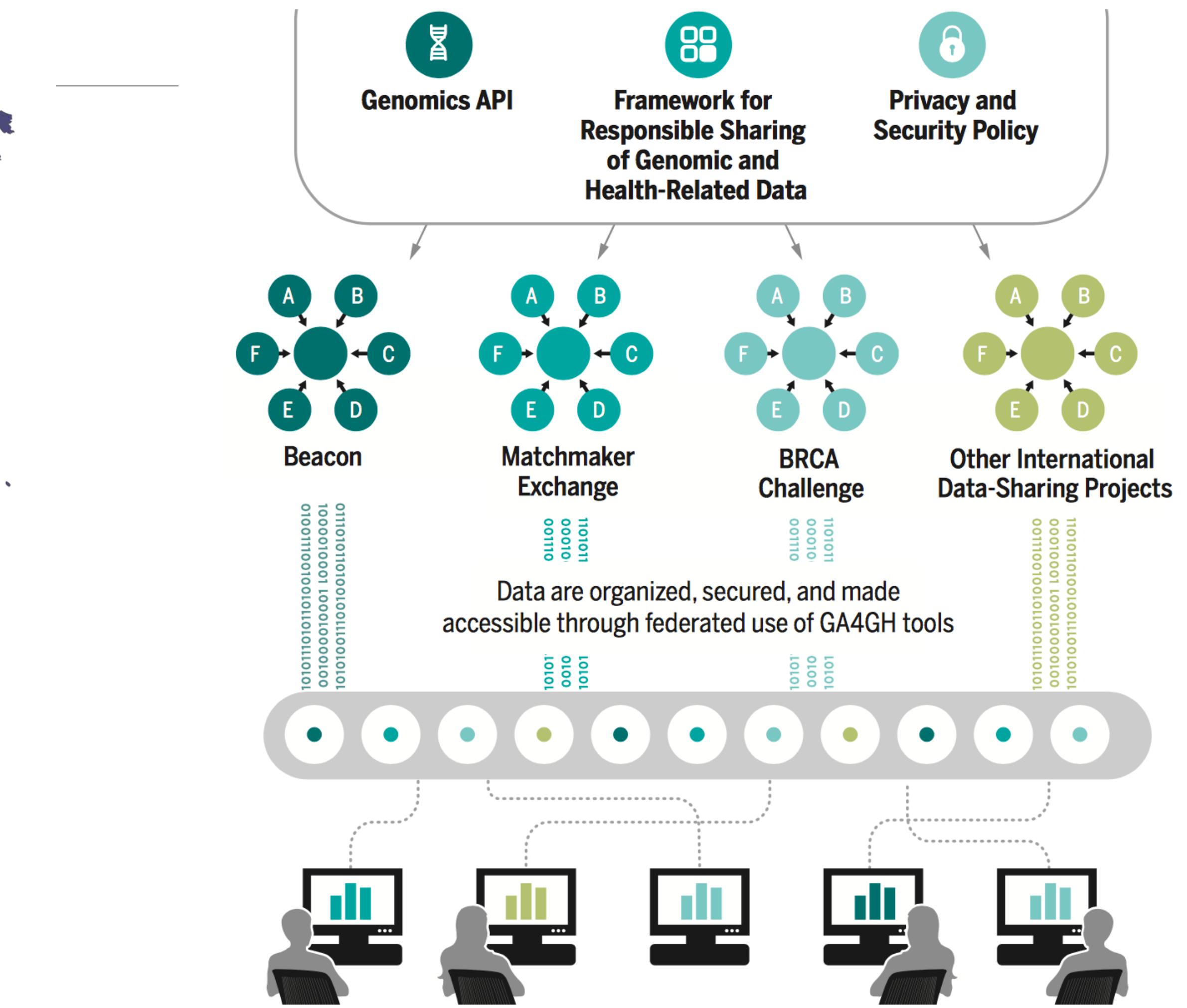


GENOMICS

A federated ecosystem for sharing genomic, clinical data

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

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





Organizational Structure - Work Streams & Driver Projects

GA4GH Driver Projects are **real-world genomic data** initiatives that help guide our development efforts and pilot our tools. Stakeholders around the globe advocate, mandate, implement, and use our **frameworks** and standards in their local contexts.

		Real-World Driver Projects							
		Discovery	Large-Scale Genomics	Data Use & Researcher IDs	Cloud	Genomic Knowledge Standards	Clinical & Phenotypic Data Capture	Regulatory & Ethics	Data Security
Technical Work Streams	Discovery	✓		✓		✓		✓	
	Large-Scale Genomics		✓		✓		✓		✓
	Data Use & Researcher IDs	✓		✓		✓	✓		✓
	Cloud		✓	✓				✓	
	Genomic Knowledge Standards		✓			✓	✓	✓	
	Clinical & Phenotypic Data Capture	✓			✓	✓	✓		✓
	Regulatory & Ethics								
Foundational Work Streams	Data Security								

GA4GH Foundational and Technical Work Streams develop standards and tools that are designed to overcome technical and regulatory hurdles to international genomic data-sharing.

Partner Engagement

The GA4GH Partner Engagement initiative facilitates two-way dialogue with the international community, including national initiatives, major health care centres, and patient advocacy groups.



GA4GH :: Discovery

A Work Stream of The Global Alliance for Genomics and Health

We build standards for federated, secured networks of data and services, forming an “Internet of Genomics”, and asking meaningful questions across it.

- Marc Fiume
 - Discovery Networks
 - Search API / Data Discovery

- Michael Baudis



- Beacon

- SchemaBlocks {S}[B]



GA4GH :: Discovery

News

Participants

Examples, Guides & FAQ

Meeting minutes

Contacts

Workstream Products

Beacon

Discovery Networks

GA4GH SchemaBlocks

Search API

Related Sites

ELIXIR beacon

GA4GH

Beacon⁺

beacon-network.org

GA4GH SchemaBlocks

Github Projects

Discovery

ELIXIR Beacon

SchemaBlocks

Tags



GA4GH Discovery Work Stream

Welcome to the homepage for the GA4GH Discovery Work Stream. We build standards for federated, secured networks of data and services, forming an “Internet of Genomics”, and asking meaningful questions across it.

The Discovery Work Stream is lead by Marc Fiume and Michael Baudis. For details on how this Work Stream operates please read the [Discovery Work Stream Organizational Structure & Vision document](#).

This group meets at a high-level monthly. [Meeting minutes are available to view here](#). In addition, the sub-groups listed below meet on their own schedules.

Participation in these groups require participants to adhere to the [GA4GH Standards for Professional Conduct](#).

For more information on GA4GH, please visit the [GA4GH Website](#).

Products

Product development in GA4GH follows a process outlined in a [GA4GH Product Approval Process Guide, in draft](#). Products developed by the work stream undergo an initial investigation phase, followed by a formal Proposed Product Phase, in which most of the work is done, followed by an formal Approval Phase during which the products gain GA4GH Approval. The formal steps require the approval of the Work Stream leads.

The following products are currently under development for this Work Stream.

Beacon API

A **Beacon** is a federated, web-accessible service that can be queried for information about a specific genomic variant, e.g. a single nucleotide polymorphism (SNP/SNV) or a copy number variation (CNV), and reports about its existence in the queried resources. Future versions of the Beacon protocol will support different usage scenarios and offer the opportunity to link to the matched data using e.g. a [handover](#) scenario.

The Beacon API specification is now coordinated through the [ELIXIR Beacon project](#) and accessible there or directly through its [repository](#).



Discovery Search API

The Discovery Search API aims at developing a component based approach towards the implementation of interfaces for genomic data and related information, for instance for global, federated data sharing through the querying, and subsequent optional processing of the results in a cloud environment. The in-development specification for the *Search API* can be [accessed here](#).

Discovery Networks API



The BeaconNetwork was the first successful implementation of an open, federated API for world-wide querying of genome resources. Current and future developments target especially the integration of user authentication for different access levels, extensions to the query language as provided through the emerging Beacon API and the evaluation of different topologies, especially with respect to security concerns.

GA4GH {S}[B] SchemaBlocks

- “cross-workstreams, cross-drivers” initiative to document GA4GH object standards and prototypes, data formats and semantics
- launched in December 2018
- documentation and implementation examples provided by GA4GH members
- no attempt to develop a rigid, complete data schema
- object vocabulary and semantics for a large range of developments
- currently not “authoritative GA4GH recommendations”
- recognized in GA4GH roadmap as element in “TASC” effort



GA4GH :: SchemaBlocks

An Initiative by Members of the Global Alliance for Genomics and Health

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[Meeting minutes](#)
[Contacts](#)

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[Beacon Project](#)
[Phenopackets](#)
[GA4GH::CLP](#)
[GA4GH::GKS](#)
[Beacon+](#)

[Github Projects](#)

[SchemaBlocks](#)
[ELIXIR Beacon](#)

[Tags](#)

Beacon CP Discovery FAQ GA4GH
GKS MME admins code contacts
contributors core dates developers
documentation howto identifiers
implemented issues leads news
phenopackets playground press
proposed sb-phenopackets tools
website

GA4GH SchemaBlocks Home

SchemaBlocks is a “**cross-workstreams, cross-drivers**” initiative to document GA4GH object standards and prototypes, as well as common data formats and semantics.

Launched in December 2018, this project is still to be considered a “community initiative”, with developing participation, leadership and governance structures. At its current stage, the documents can **not** be considered “**authoritative GA4GH recommendations**” but rather represent documentation and implementation examples provided by GA4GH members.

While future products and implementations may be completely based on *SchemaBlocks* components, this project does not attempt to develop a rigid, complete data schema but rather to provide the object vocabulary and semantics for a large range of developments.

The SchemaBlocks site can be accessed through the permanent link schemablocks.org. More information about the different products & formats can be found on the workstream sites. For reference, some of the original information about recommended formats and object hierarchies is kept in the [GA4GH Metadata repositories](#).

For more information on GA4GH, please visit the [GA4GH Website](#).

SchemaBlocks Repositories

The SchemaBlocks Github organisation contains several specifically scoped repositories. Please use the relevant *Github Issues* to and/or GH pull requests comment and contribute there.

@mbaudis 2019-11-19: [more ...](#)

SchemaBlocks “Status” Levels

SchemaBlocks schemas (“blocks”) provide recommended blueprints for schema parts to be re-used for the development of code based “products” throughout the GA4GH ecosystem. We propose a labeling system for those schemas, to provide transparency about the level of support those schemas have from {S}[B] participants and observers.

@mbaudis 2019-07-17: [more ...](#)

SchemaBlocks {S}[B] Mission Statement

SchemaBlocks aims to translate the work of the workstreams into data models that:

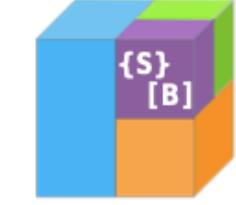
- Are usable by other internal GA4GH deliverables, such as the Search API.
- Are usable by Driver Projects as an exchange format.
- Aid in aligning the work streams across GA4GH.
- Do not create a hindrance in development work by other work streams.

@mbaudis 2019-03-27: [more ...](#)



GA4GH SchemaBlocks Home

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Launched in December 2018, this project is still to be considered a “community initiative”, with developing participation, leadership and governance structures. At its current stage, the documents can **not** be considered “authoritative GA4GH recommendations” but rather represent documentation and implementation examples provided by GA4GH members.

While future products and implementations may be completely based on *SchemaBlocks* components, this project does not attempt to develop a rigid, complete data schema but rather to provide the object vocabulary and semantics for a large range of developments.

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SchemaBlocks “Status” Levels

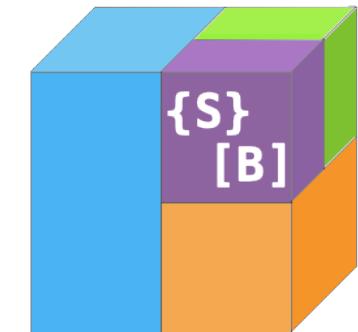
SchemaBlocks schemas (“blocks”) provide recommended blueprints for schema parts to be re-used for the development of code based “products” throughout the GA4GH ecosystem. We propose a labeling system for those schemas, to provide transparency about the level of support those schemas have from {S}[B] participants and observers.

Proposed {S}[B] Status Levels

The current status level of those recommendations is “proposed”.

- [playground](#)
 - early development or import stage, of any quality
 - no recommendation; existence does not mean any current or future {S}[B] support
- [proposed](#)
 - at least some {S}[B] contributors are in favour of such a block
 - the code may undergo considerable maturation
 - not recommended for integration into products w/o close tracking
 - contributions and discussions are encouraged
- [implemented](#)
 - mature block which is implemented in one or more {S}[B] aligned schemas
 - may be extended from a core block or be too specific for general (“core”) usability
- [core](#)
 - a schema block with recommended use
 - stable through minor version changes
 - has to be used in at least 2 standards/products approved by the GA4GH Steering Committee

SchemaBlocks - A GA4GH Community Initiative



SchemaBlocks{S}[B] Mission Statement

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- Are usable by other internal GA4GH deliverables, such as the Search API.
- Are usable by Driver Projects as an exchange format.
- Aid in aligning the work streams across GA4GH.
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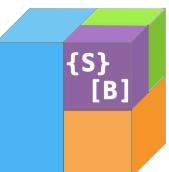
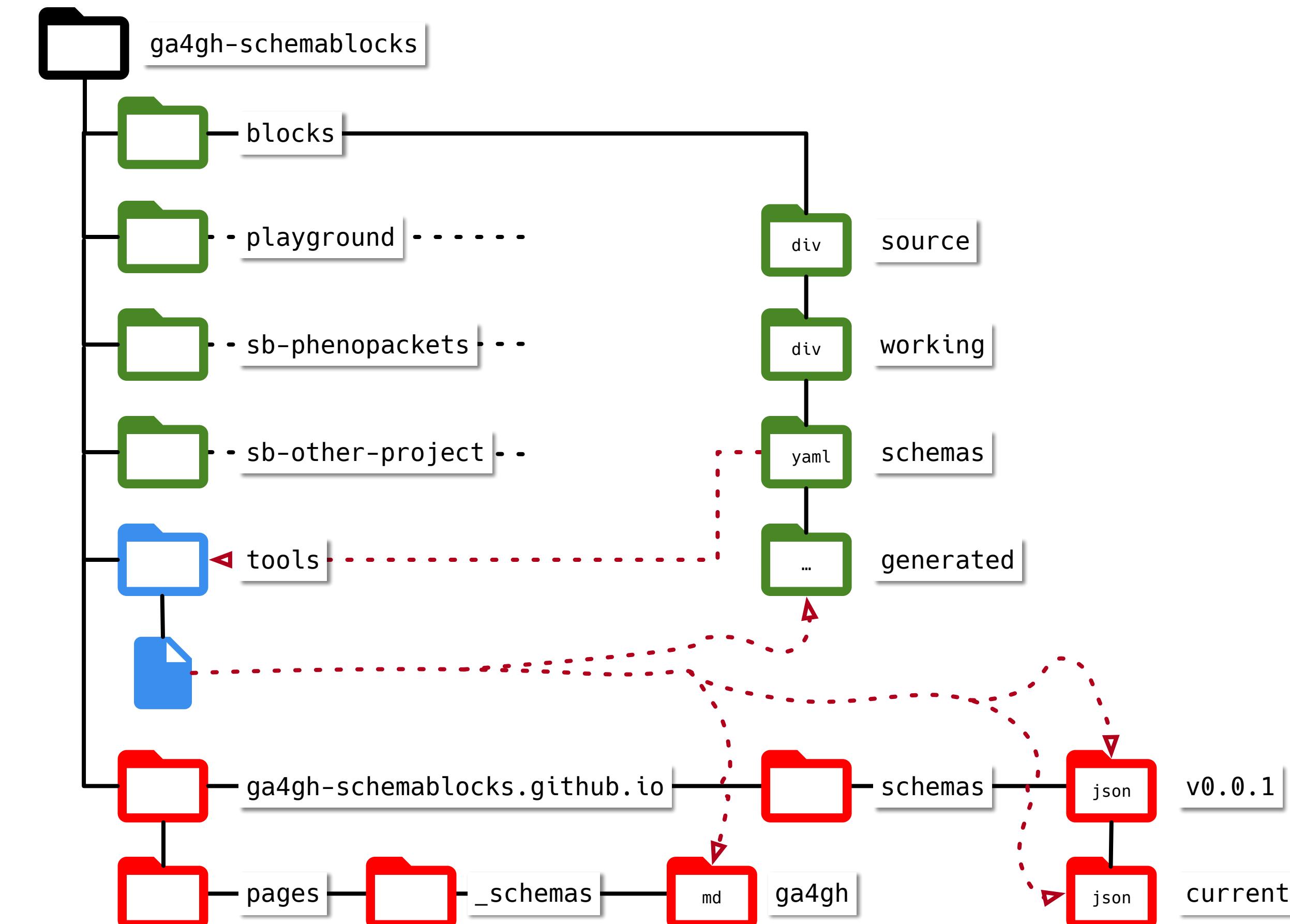
After discussions with stakeholders from GA4GH work streams and driver projects who create data models (such as Phenopackets, Search API) or who would use SchemaBlocks for the development of their APIs and data exchange formats (Beacon, EGA, GeL), the SchemaBlocks team has come up with the following principles for this initiative:

Work Stream Interactions

Work streams will continue to create standards proposals and their own coherent project implementations, but will work with the SchemaBlocks group to write the Blocks that will come from their own work and are considered of overarching use. Generally, primary work stream and driver project outputs will live in their own spaces outside of SchemaBlocks, with shareable, mature elements - code, documentation, implementation snapshots - being represented in {S}[B].

{S}[B] SchemaBlocks Github Repository Structure

blocks repositories
conversion/validation tools
website repository
(Markdown w/ YAML for Github Pages)



Dissection & Transformation

```
// See http://build.fhir.org/datatypes and http://build.fhir.org/condition-definitions.html#Condition.onset_x_
// In FHIR this is represented as a UCUM measurement - http://unitsofmeasure.org/trac/
message Age {

    // The :ref:`ISO 8601<metadata_date_time>` age of this object as ISO8601
    // duration or time intervals. The use of time intervals makes an additional
    // anchor unnecessary (i.e. DOB and age can be represented as start-anchored
    // time interval, e.g. 1967-11-21/P40Y10M05D)
    string age = 1;
}

message AgeRange {
    Age start = 1;
    Age end = 2;
}

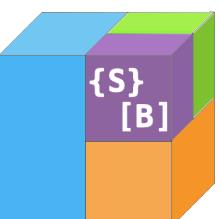
// Message to indicate a disease (diagnosis) and its recorded onset.
message Disease {
    // The identifier of this disease e.g. MONDO:0007043, OMIM:101600, Orphanet:710, DOID:14705 (note these are all equivalent)
    OntologyClass term = 1;

    // The onset of the disease. The values of this will come from the HPO onset hierarchy
    // i.e. subclasses of HP:0003674
    // FHIR mapping: Condition.onset
    oneof onset {
        Age age_of_onset = 2;
        AgeRange age_range_of_onset = 3;
        OntologyClass class_of_onset = 4;
    }

    // Disease staging, the extent to which a disease has developed.
    // For cancers, see https://www.cancer.gov/about-cancer/diagnosis-staging/staging
    // Valid values include child terms of NCIT:C28108 (Disease Stage Qualifier)
    repeated OntologyClass disease_stage = 5;
}
```

- Excerpt from Phenopackets v1.0 Schema
- written in *Protocol Buffers* (Google's data serializing format)
- separate documentation rendered in "ReadTheDocs"

Use Case Transforming Phenopackets objects (here "Age") into JSON Schema documents with (proposed) stable id and address as well as "human readable" documentation & examples.



Dissection & Transformation

```
// See http://build.fhir.org/datatypes and http://build.fhir.org/condition-definitions.html#Condition.onset_x_
// In FHIR this is represented as a UCUM measurement - http://unitsofmeasure.org/trac/
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    oneof onset {
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    }

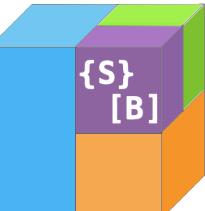
    // Disease staging, the extent to which a disease has developed.
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    repeated OntologyClass disease_stage = 5;
}
```

32 lines (31 sloc) | 872 Bytes

Raw Blame History

```
1  "$schema": "http://json-schema.org/draft-07/schema#"
2  "$id": "https://schemablocks.org/schemas/sb-phenopackets/Age/v0.0.1"
3  title: Age
4  description: Age
5  type: object
6  meta:
7      contributors:
8          - description: "Michael Baudis"
9              id: "orcid:0000-0002-9903-4248"
10         - description: "Jules Jacobsen"
11             id: "orcid:0000-0002-3265-15918"
12         - description: "Peter Robinson"
13             id: "orcid:0000-0002-0736-91998"
14     provenance:
15         - description: Phenopackets
16             id: 'https://github.com/phenopackets/phenopacket-schema/blob/master/docs/age.rst'
17     used_by:
18         - description: Phenopackets
19             id: 'https://github.com/phenopackets/phenopacket-schema/blob/master/docs/age.rst'
20     sb_status: implemented
21 properties:
22     age:
23         type: string
24         description: Age as ISO8601 period
25         examples:
26             - 'P12Y'
27
28 required:
29     - age
30 additionalProperties: false
31 examples:
32     - age: 'P14Y'
```

- Separate {S}[B] repository for parental project
- here "sb-phenopackets"
- individual schema documents for each original object
- (currently) manual re-write into JSON Schema documents (YAML version), including metadata header (id, provenance ...)
- versioned



Dissection & Transformation

```
// See http://build.fhir.org/datatypes and http://build.fhir.org/condition-definitions.html#Condition.onset_x_
// In FHIR this is represented as a UCUM measurement - http://unitsofmeasure.org/trac/
message Age {

    // The :ref:`ISO 8601<metadata_date_time>` age of this object as ISO8601
    // duration or time intervals. The use of time intervals makes an additional
    // anchor unnecessary (i.e. DOB and age can be represented as start-anchored
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    // FHIR mapping: Condition.onset
    oneof onset {
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    // Disease staging, the extent to which a disease has developed.
    // For cancers, see https://www.cancer.gov/about-cancer/diagnosis-staging/staging
    // Valid values include child terms of NCIT:C28108 (Disease Stage Qualifier)
    repeated OntologyClass disease_stage = 5;
}
```

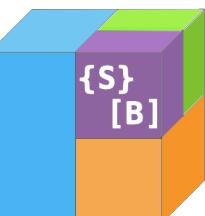
32 lines (31 sloc) | 872 Bytes

Raw Blame History

```
1  "$schema": "http://json-schema.org/draft-07/schema#"
2  "$id": "https://schemablocks.org/schemas/sb-pheno-packets/Age/v0.0.1"
3  title: Age
4  description: Age
5  type: object
6  meta:
7      contributors:
8          - description: "Michael Baudis"
9              id: "orcid:0000-0002-9903-4248"
10         - description: "Jules Jacobsen"
11             id: "orcid:0000-0002-3265-15918"
12             - description: "Peter Robinson"
13                 id: "orcid:0000-0002-0736-91998"
14         provenance:
15             - description: Phenopackets
16                 id: "https://github.com/phenopackets/pheno-packets"
17             used_by:
18                 - description: Phenopackets
19                     id: "https://github.com/phenopackets/pheno-packets"
20         sb_status: implemented
21     properties:
22         age:
23             type: string
24             description: Age as ISO8601 period
25             examples:
26                 - 'P12Y'
27         required:
28             - age
29         additionalProperties: false
30     examples:
31         - age: 'P14Y'
32 }
```

The JSON schema defines the structure of the Age resource. It includes fields for contributors (Michael Baudis, Jules Jacobsen, Peter Robinson), provenance (Phenopackets), and usage (Phenopackets). The 'age' field is a string representing the age as an ISO8601 period (e.g., P12Y). The schema is versioned at v0.0.1 and is available at <https://schemablocks.org/schemas/sb-pheno-packets/Age/v0.0.1>.

- schema documents are programmatically converted into different outputs
- a versioned JSON document serves as canonical reference for integration into other products/schemas



Dissection & Transformation

```
// See http://build.fhir.org/datatypes and http://build.fhir.org/condition-definitions.html#Condition.onset_x_
// In FHIR this is represented as a UCUM measurement - http://unitsofmeasure.org/trac/
message Age {
    // The :ref:`ISO 8601<metadata_date_time>` age of this object as ISO8601
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message AgeRange {
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// Message to indicate a disease (diagnosis) and its recorded onset.
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    // The onset of the disease. The values of this will come from the HPO onset hierarchy
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    // FHIR mapping: Condition.onset
    oneof onset {
        Age age_of_onset = 2;
        AgeRange age_range_of_onset = 3;
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    // Disease staging, the extent to which a disease has developed.
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    repeated OntologyClass disease_stage = 5;
```

32 lines (31 s)

Age sb-phenopackets ↗

{S}[B] Status [I]	implemented
Provenance	◦ Phenopackets
Used by	◦ Phenopackets
Contributors	◦ Michael Baudis ◦ Jules Jacobsen ◦ Peter Robinson
Source (v0.0.1)	◦ raw source [JSON] ◦ Github

Attributes

Type: object
Description: Age

Properties

Property	Type
age	string

age

- type: string

Age as ISO8601 period

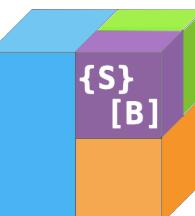
age Value Example

```
"P12Y"
```

Age Value Example

```
{
  "age" : "P14Y"
}
```

- schema documents are programmatically converted into different outputs
- a Markdown document with "Jekyll" header is auto-converted by Github into a complete website document, including inline code examples



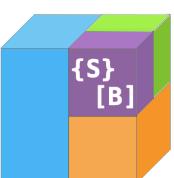
{S}[B] SchemaBlocks **JSON** **Schema** document format

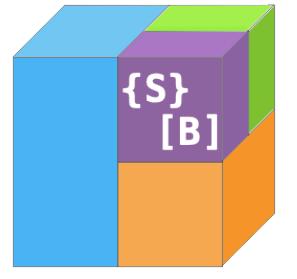
- {S}[B] "blocks" are written in the YAML version of a JSON Schema document format
 - convenience choice - flexibility, readability, tooling ...
 - **not** implying specific semantics beyond some format conventions - extensible for use-case driven requirements
- the **meta** part (itself defined as a schema "block") contains housekeeping information
 - reference address & version
 - provenance & use cases
 - sb_status about "blessing level"
- the properties part defines the attributes including their description and usage examples
 - descriptions & examples provide the core documentation which is deparsed to the website documents

```
 "$schema": http://json-schema.org/draft-07/schema#
"$id": https://schemablocks.org/schemas/ga4gh/AgeRange/v0.0.1
title: AgeRange
description: Age range
type: object

meta:
contributors:
- description: "Jules Jacobsen"
  id: "orcid:0000-0002-3265-15918"
- description: "Peter Robinson"
  id: "orcid:0000-0002-0736-91998"
- description: "Michael Baudis"
  id: "orcid:0000-0002-9903-4248"
- description: "Isuru Liyanage"
  id: "orcid:0000-0002-4839-5158"
provenance:
- description: Phenopackets
  id: 'https://github.com/phenopackets/phenopacket-schema/blob/master/docs/age.rst'
used_by:
- description: Phenopackets
  id: 'https://github.com/phenopackets/phenopacket-schema/blob/master/docs/age.rst'
sb_status: implemented

properties:
start:
alof:
"$ref": https://schemablocks.org/schemas/ga4gh/v0.0.1/Age.json
description: Age as ISO8601 string or OntologyClass
examples:
- age: 'P12Y'
end:
alof:
"$ref": https://schemablocks.org/schemas/ga4gh/v0.0.1/Age.json
description: Age as ISO8601 string or OntologyClass
examples:
- ageClass:
  id: 'HsapDv:0000086'
  label: 'adolescent stage'
- age: 'P16Y6M'
required:
anyof:
- start
- end
examples:
- start:
  age: 'P12Y'
  ageClass:
    id: 'HsapDv:0000086'
    label: 'adolescent stage'
end:
age: 'P18Y'
```





SchemaBlocks {S}[B] - Directions & Contributions

- Recognized need of having a set of recommended standards for integrating into product development
 - no need to work through complex standards/projects like FHIR, Phenopackets ...
 - simplification of development
- SchemaBlocks {S}[B] to assume strategic position in GA4GH *TASC system
 - Inclusion into product approval processes?
 - Management/Support?
- Wish for participation of (GA4GH affiliated) groups & individuals, to **expose** their standards & products
- Most important role is the **community aspect**, the interactive exchange of concepts, ideas, code, knowledge, resources ...
- Technical to-dos:
 - Lifecycle: Versioning and representation of donor schemas?
 - Development of conversion workflows for updated source products?
 - Alternative/conflicting blocks...: Graded recommendations? Name spacing?

Search or jump to... / Pull requests Issues Marketplace Explore

Earth http://schemablocks.org

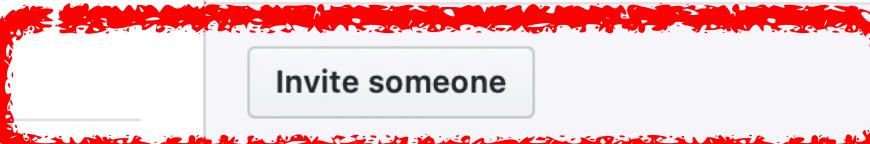
GA4GH SchemaBlocks {S}[B]

Code and website repositories of the GA4GH SchemaBlocks standards initiative

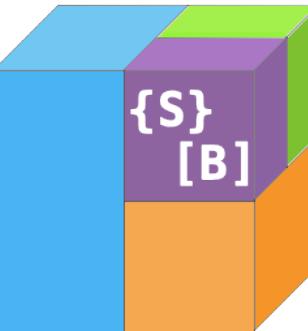
Repositories 10

Find a repository... Type: All Language: All Customize pins New

- ga4gh-schemablocks.github.io**
Website of the GA4GH SchemaBlocks Project
HTML 5 stars 0 issues 6 pull requests 1 updated 12 days ago
- sb-checksum**
SchemaBlocks Version of GA4GH Checksum Standard
0 stars 0 issues 4 pull requests 0 updated on Dec 11, 2019
- playground**
The playground repository for proposing blocks and docs
0 stars 0 issues 0 pull requests 0 updated on Dec 6, 2019
- sb-beacon-api**
SchemaBlocks version of the GA4GH Beacon API
0 stars 0 issues 0 pull requests 0 updated on Dec 6, 2019
- sb-phenopackets**
THIS IS A DRAFT REPOSITORY to write schemablocks using JSON schema and convert this into markdown.
Java 0 stars 0 issues 4 pull requests 1 updated on Dec 6, 2019
- tools**
Tools for managing the {S}[B] repositories and website
Perl 0 stars 0 issues 0 pull requests 0 updated on Dec 2, 2019



{S}[B] Info



Leads

- Melanie Courtot [↗]
- Michael Baudis [↗]

Coordination

- Melissa Konopko

Websites

- schemablocks.org
- github.com/ga4gh-schemablocks/

Meeting minutes

- schemablocks.org/categories/minutes.html

