

{S}[B] SchemaBlocks

GA4GH Standards Documentation and Alignment Initiative



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[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



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for Genomics & Health

[Poking Holes in Genetic Privacy](#)

The New York Times - Gina Kolata
June 16, 2013

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

The Globe and Mail - By André Picard
June 5, 2013

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

GenomeWeb
June 19, 2013

[Our Genes, Their Secrets](#)

The New York Times
June 18, 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

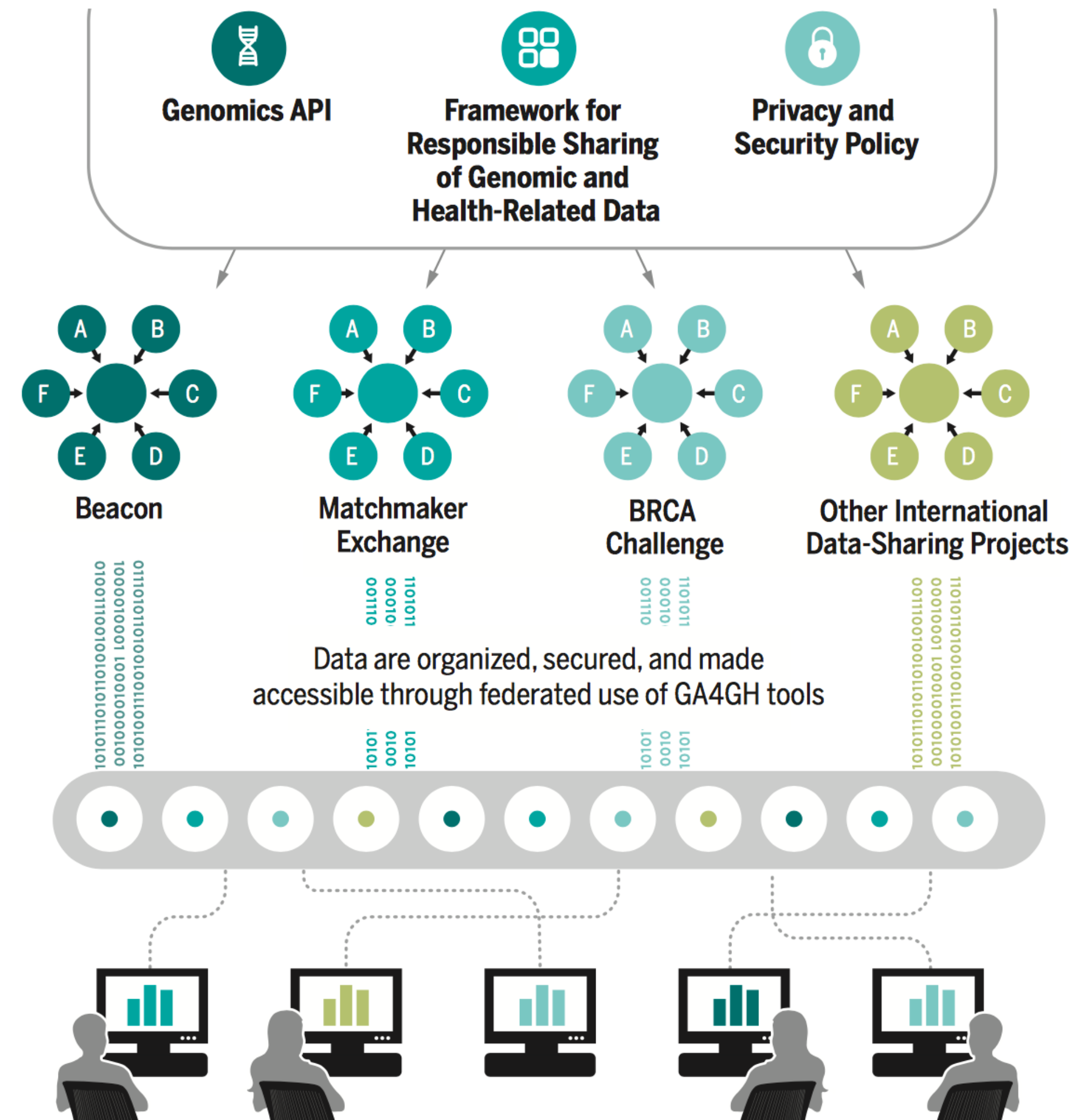


GENOMICS

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.

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

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



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

- [Beacon](#)
- [GA4GH](#)
- [SchemaBlocks](#)
- [admins](#)
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- [contributors](#)
- [developers](#)
- [leads](#)
- [press](#)
- [releases](#)
- [website](#)

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

schemablocks.org



GA4GH :: SchemaBlocks

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

- [About {S}\[B\]](#)
- [News](#)
- [Participants](#)
- [Standards](#)
- [Schemas](#)
- [Examples, Guides & FAQ](#)
- [Meeting minutes](#)
- [Contacts](#)

Related Sites

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

Github Projects

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

Tags

- [Beacon](#)
- [CP](#)
- [Discovery](#)
- [FAQ](#)
- [GA4GH](#)
- [GKS](#)
- [MME](#)
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- [howto](#)
- [identifiers](#)
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- [issues](#)
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- [news](#)
- [phenopackets](#)
- [playground](#)
- [press](#)
- [proposed](#)
- [sb-phenopackets](#)
- [tools](#)
- [website](#)



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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 though 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:

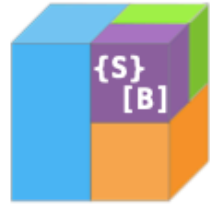
- Are usable by other internal GA4GH deliverables, such as the Search API.
- Are usable by Driver Projects as an exchange format.
- Aid in aligning the work streams across GA4GH.
- Do not create a hindrance in development work by other work streams.

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



GA4GH SchemaBlocks Home

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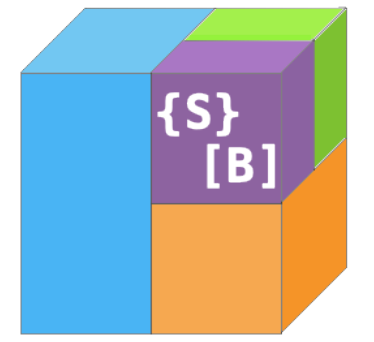


Launched in December 2018, this project is still to be considered a “community initiative”, with developing participation, leadership and governance structures. At its current stage, the documents can **not** be considered “**authoritative GA4GH recommendations**” but rather represent **documentation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

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^{{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.

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

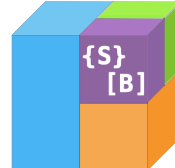
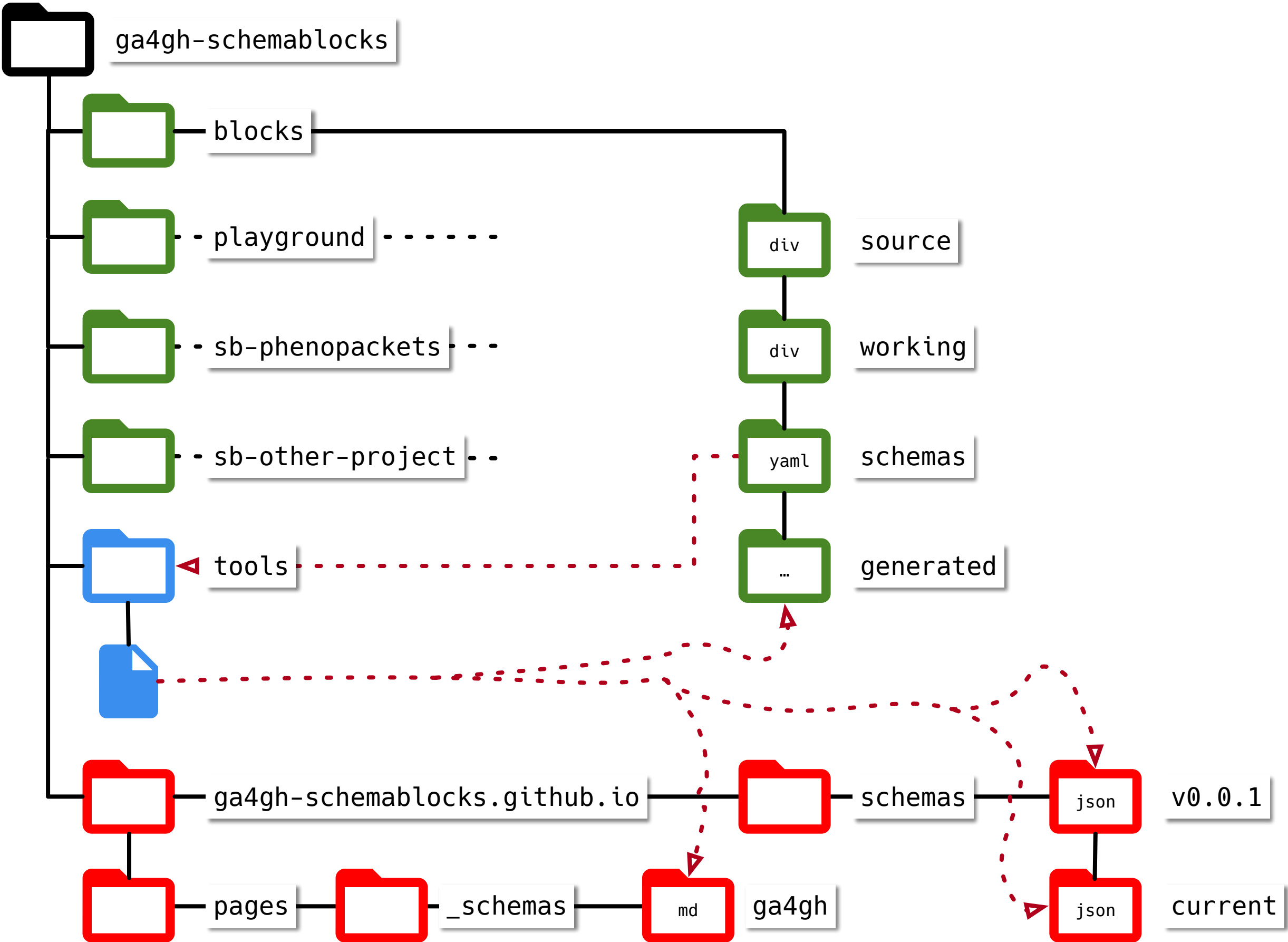
Work streams will continue to create standards proposals and their own coherent project implementations, but will work with the SchemaBlocks group to write the Blocks that will come from their own work and are considered of overarching use. Generally, primary work stream and driver project outputs will 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;

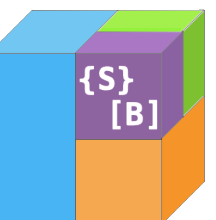
  // The onset of the disease. The values of this will come from the HPO onset hierarchy
  // i.e. subclasses of HP:0003674
  // FHIR mapping: Condition.onset
  oneof onset {
    Age age_of_onset = 2;
    AgeRange age_range_of_onset = 3;
    OntologyClass class_of_onset = 4;
  }

  // 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

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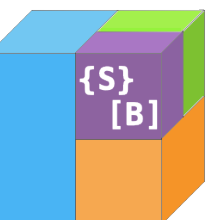
  // The onset of the disease. The values of this will come from the HPO onset hierarchy
  // i.e. subclasses of HP:0003674
  // FHIR mapping: Condition.onset
  oneof onset {
    Age age_of_onset = 2;
    AgeRange age_range_of_onset = 3;
    OntologyClass class_of_onset = 4;
  }

  // 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;
}
```

```
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/
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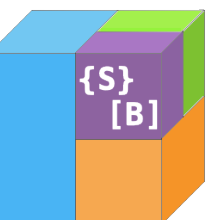
  // Disease staging, the extent to which a disease has developed.
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  // Valid values include child terms of NCIT:C28108 (Disease Stage Qualifier)
  repeated OntologyClass disease_stage = 5;
}
```

32 lines (31 sloc) | 872 Bytes

```
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"
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14  provenance:
15    - description: Phenopackets
16      id: "https://github.com/phenopackets/phenopacket-schema/blob/master/docs/age.rst"
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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  required:
28    - age
29  additionalProperties: false
30  examples:
31    - age: 'P14Y'
32
```

```
{
  "$id": "https://schemablocks.org/schemas/sb-phenopackets/Age/v0.0.1",
  "$schema": "http://json-schema.org/draft-07/schema#",
  "additionalProperties": "",
  "description": "Age",
  "examples": [
    {
      "age": "P14Y"
    }
  ],
  "meta": {
    "contributors": [
      {
        "description": "Michael Baudis",
        "id": "orcid:0000-0002-9903-4248"
      },
      {
        "description": "Jules Jacobsen",
        "id": "orcid:0000-0002-3265-15918"
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      {
        "description": "Peter Robinson",
        "id": "orcid:0000-0002-0736-91998"
      }
    ],
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      {
        "description": "Phenopackets",
        "id": "https://github.com/phenopackets/phenopacket-schema/blob/master/docs/age.rst"
      }
    ],
    "sb_status": "implemented",
    "used_by": [
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        "description": "Phenopackets",
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      }
    ]
  },
  "properties": {
    "age": {
      "description": "Age as ISO8601 period",
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        "P12Y"
      ],
      "type": "string"
    }
  },
  "required": [
    "age"
  ],
  "title": "Age",
  "type": "object"
}
```

- 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/
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  repeated OntologyClass disease_stage = 5;
}
```

The screenshot shows a code editor on the left with 32 lines of schema code. On the right, a rendered website for 'Age sb-phenopackets' is displayed. The website includes a table for provenance and contributors, an 'Attributes' section with type and description, a 'Properties' table with 'age' as a string, and two 'Value Example' sections showing 'P12Y' and a JSON object with 'age' set to 'P14Y'.

{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

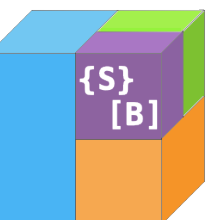
Property	Type
age	string

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



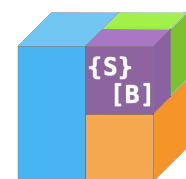
{S}[B] SchemaBlocks **JSON** *Schema* document format

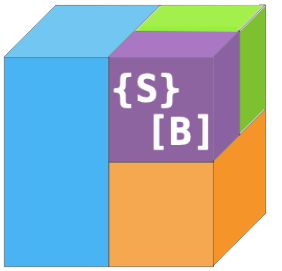
- {S}[B] "blocks" are written in the YAML version of a JSON Schema document format
 - convenience choice - flexibility, readability, tooling ...
 - **not** implying specific semantics beyond some format conventions - extensible for use-case driven requirements
- the **meta** part (itself defined as a 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:  
    allof:  
      "$ref": https://schemablocks.org/schemas/ga4gh/v0.0.1/Age.json  
      description: Age as ISO8601 string or OntologyClass  
      examples:  
        - age: 'P12Y'  
    end:  
    allof:  
      "$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'
```





BeaconAlleleRequest beacon ↗

{S}[B] Status [i]	implemented
Provenance	<ul style="list-style-type: none"> Beacon API
Used by	<ul style="list-style-type: none"> Beacon Progenetix database schema (Beacon+ backend)
Contributors	<ul style="list-style-type: none"> Marc Fiume Michael Baudis Sabela de la Torre Pernas Jordi Rambla Beacon developers...
Source (v1.1.0)	<ul style="list-style-type: none"> raw source [JSON] Github

Attributes

Type: object

Description: Allele request as interpreted by the beacon.

Properties

Property	Type
alternateBases	string
assemblyId	string
datasetIds	array of string
end	integer
endMax	integer
endMin	integer
mateName	https://schemablocks.org/schemas/beacon/v1.1.0/Chrom[HTML]
referenceBases	string
referenceName	https://schemablocks.org/schemas/beacon/v1.1.0/Chrom[HTML]
start	integer (int64)
startMax	integer
startMin	integer
variantType	string

alternateBases

- type: string

The bases that appear instead of the reference bases. Accepted values: [ACGTN]*. N is a wildcard, that denotes the position of any base, and can be used as a standalone base of any type or within a partially known sequence. For example a sequence where the first and last bases are known, but the middle portion can exhibit countless variations of [ACGT], or the bases are unknown: ANNT the Ns can take take any form of [ACGT], which makes both ACCT and ATGT (or any other combination) viable sequences.

Symbolic ALT alleles (DEL, INS, DUP, INV, CNV, DUP:TANDEM, DEL:ME, INS:ME) will be represented in **variantType**.

Optional: either **alternateBases** or **variantType** is required.

alternateBases Value Example

assemblyId

- type: string

Assembly identifier (GRC notation, e.g. **GRCh37**).

assemblyId Value Example

Curie sb-vr-spec ↗

{S}[B] Status [i]	implemented
Provenance	<ul style="list-style-type: none"> vr-spec
Used by	<ul style="list-style-type: none"> vr-spec
Contributors	<ul style="list-style-type: none"> Reece Hart Michael Baudis
Source (v1.0)	<ul style="list-style-type: none"> raw source [JSON] Github

Attributes

Type: string

Pattern: ^\w[^\:]+:.\$

Description: A string that refers to an object uniquely. The sender.

VR does not impose any constraints on strings used as ids data, the VR Specification RECOMMENDS that implement String CURIEs are represented as **prefix:reference** (Where **namespace:accession** or **namespace:local id** colloquially). The VR specification also RECOMMENDS that **prefix** be

The **reference** component is an unconstrained string.

A CURIE is a URI. URIs may *locate* objects (i.e., specify where VR uses CURIEs primarily as a naming mechanism.

Implementations MAY provide CURIE resolution mechanisms. Using internal ids in public messages is strongly discouraged.

Curie Value Examples

"ga4gh:GA_01234abcde"
"DUO:0000004"
"orcid:0000-0003-3463-0775"
"PMID:15254584"

Biosample sb-phenopackets ↗

{S}[B] Status [i]	implemented
Provenance	<ul style="list-style-type: none"> Phenopackets
Used by	<ul style="list-style-type: none"> Phenopackets
Contributors	<ul style="list-style-type: none"> GA4GH Data Working Group Jules Jacobsen Peter Robinson Michael Baudis Melanie Courtot Isuru Liyanage
Source (v1.0.0)	<ul style="list-style-type: none"> raw source [JSON] Github

Attributes

Type: object

Description: A Biosample refers to a unit of biological material from which the substrate molecular genomic DNA, RNA, proteins) for molecular analyses (e.g. sequencing, array hybridisation, mass spectrometry) are extracted.

Examples would be a tissue biopsy, a single cell from a culture for single cell genome sequencing fraction from a gradient centrifugation.

Several instances (e.g. technical replicates) or types of experiments (e.g. genomic array as well as experiments) may refer to the same Biosample.

FHIR mapping: **Specimen**.

Properties

Property	Type
ageOfIndividualAtCollection	https://schemablocks.org/schemas/sb-phenopackets/v1.0.0/Age.json [SRC] [HTML]
ageRangeOfIndividualAtCollection	https://schemablocks.org/schemas/sb-phenopackets/v1.0.0/AgeRange.json [SRC] [HTML]
description	string
diagnosticMarkers	array of https://schemablocks.org/schemas/sb-phenopackets/v1.0.0/OntologyClass.json [SRC] [HTML]
histologicalDiagnosis	https://schemablocks.org/schemas/sb-phenopackets/v1.0.0/OntologyClass.json [SRC] [HTML]
htsFiles	array of https://schemablocks.org/schemas/sb-phenopackets/v1.0.0/HtsFile.json [SRC] [HTML]
id	string
individualId	string
isControlSample	boolean
phenotypicFeature	array of https://schemablocks.org/schemas/sb-phenopackets/v1.0.0/PhenotypicFeature.json [SRC] [HTML]
procedure	https://schemablocks.org/schemas/sb-phenopackets/v1.0.0/Procedure.json [SRC] [HTML]
sampledTissue	https://schemablocks.org/schemas/sb-

Checksum sb-checksum ↗

{S}[B] Status [i]	proposed
Provenance	<ul style="list-style-type: none"> GA4GH DRS (`develop` branch)
Used by	<ul style="list-style-type: none"> GA4GH DRS GA4GH TRS
Contributors	<ul style="list-style-type: none"> Susheel Varma
Source (v0.0.1)	<ul style="list-style-type: none"> raw source [JSON] Github

Attributes

Type: object

Description: Checksum

Properties

Property	Type
checksum	string
type	string

checksum

- type: string

The hexadecimal encoded (**Base16**) checksum for the data

checksum Value Example

"77af4d6b9913e693e8d0b4b294fa62ade6054e6b2f1ffb617ac955dd63fb0182"
--

type

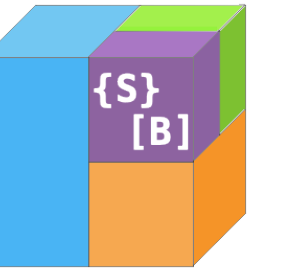
- type: string

The digest method used to create the checksum. The value (e.g. **sha-256**) SHOULD be listed as **Hash Name String** in the **GA4GH Hash Algorithm Registry**. Other values MAY be used, as long as implementors are aware of the issues discussed in **RFC6920**.

GA4GH may provide more explicit guidance for use of non-IANA-registered algorithms in the future.

type Value Example

"sha-256"



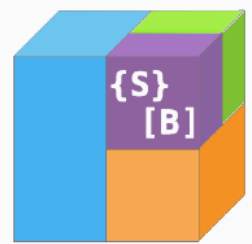
SchemaBlocks {S}[B] - Directions & Contributions

- 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



GA4GH SchemaBlocks {S}[B]

Code and website repositories of the GA4GH SchemaBlocks standards initiative

Earth <http://schemablocks.org>

Repositories 10

Packages

People 12

Teams

Projects

Settings

Find a repository...

Type: All

Language: All

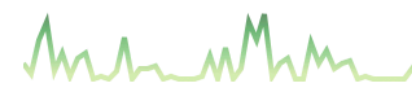
Customize pins

New

ga4gh-schemablocks.github.io

Website of the GA4GH SchemaBlocks Project

HTML 5 0 6 1 Updated 12 days ago



Top languages

Perl Java HTML

sb-checksum

SchemaBlocks Version of GA4GH Checksum Standard

0 0 4 0 Updated on Dec 11, 2019



People

12 >



Invite someone

playground

The playground repository for proposing blocks and docs

0 0 0 0 Updated on Dec 6, 2019



sb-beacon-api

SchemaBlocks version of the GA4GH Beacon API

0 0 0 0 Updated on Dec 6, 2019



sb-phenopackets

THIS IS A DRAFT REPOSITORY to write schemablocks using JSON schema and convert this into markdown.

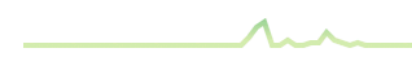
Java 0 0 4 1 Updated on Dec 6, 2019



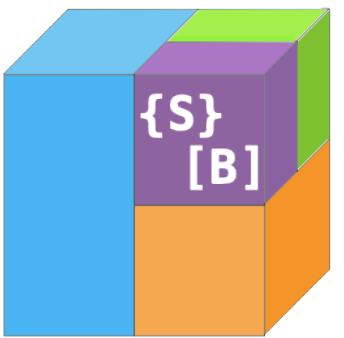
tools

Tools for managing the {S}[B] repositories and website

Perl 0 0 0 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

