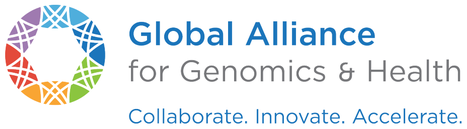
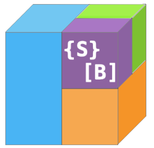


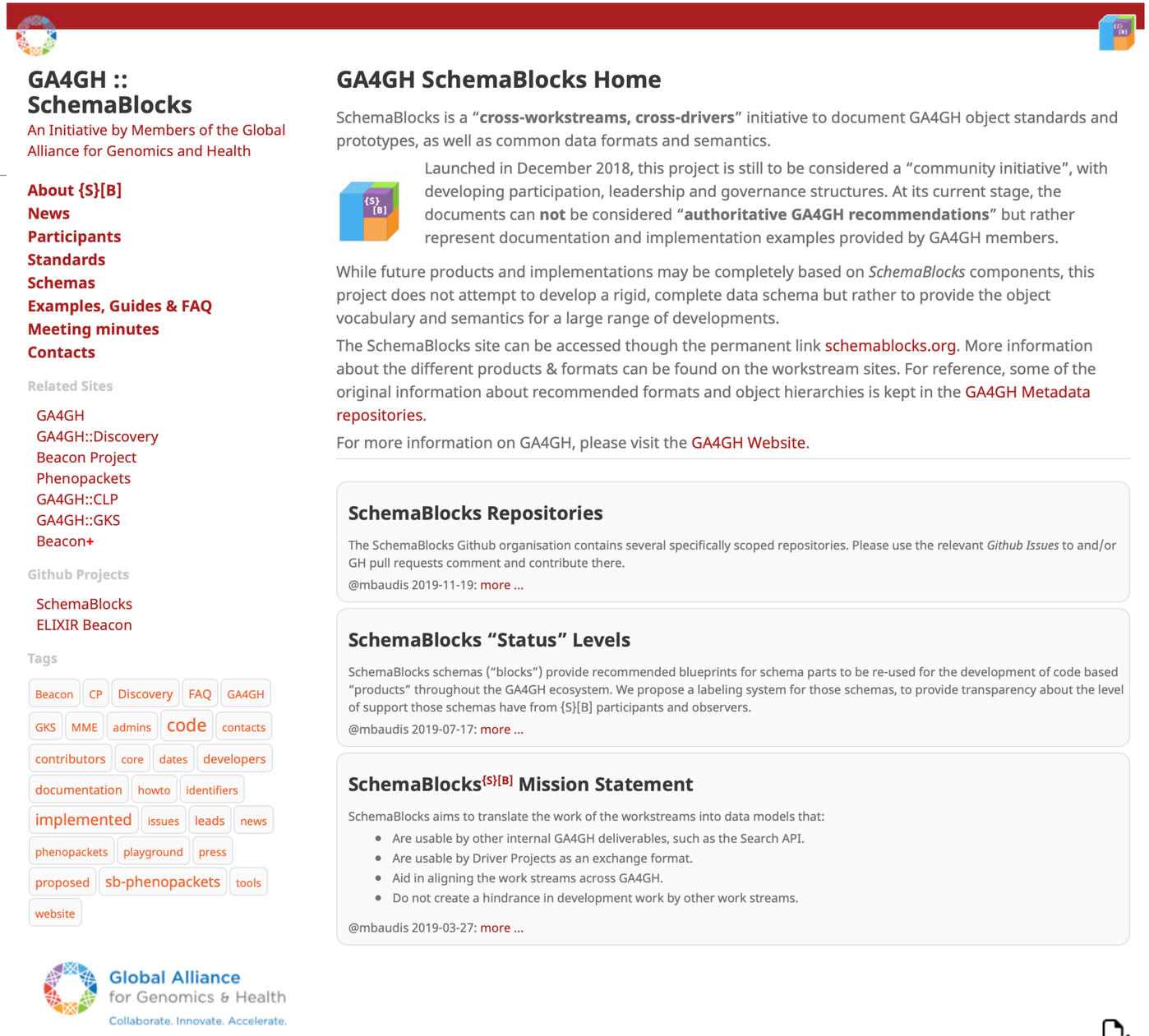

# SchemaBlocks {S}[B] | GA4GH Connect | Michael Baudis | 2021-03-04

## General (Re-)Introduction


# 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 admins code contacts contributors core dates developers documentation howto identifiers implemented issues leads news phenopackets playground press proposed sb-phenopackets tools website

Global Alliance for Genomics & Health
Collaborate. Innovate. Accelerate.

### 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 ...

# SchemaBlocks - A GA4GH Community Initiative

## 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 "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 thiose 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] As a Component Recycler

## "Donor" Schema

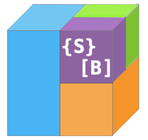
```
Root:
{
  Components: {
    this: {...},
    that: {
      $ref: ./Other
    }
  }
}

Other:
{
  Components: {
    well: {...},
    done: {...}
  }
}
```

## {S}[B] Representation

```
Other:
{
  Meta: {
    donorSchema: "DonorProject",
    donorLink: "donorschema.org",
    status: "core"
  },
  Components: {
    well: {...},
    done: {...}
  }
}
```

## "Acceptor" Schema

```
Root:
{
  Components: {
    my: {...},
    theirs: {
      $ref: "schemablocks.org/other.json"
    },
    more: "..."
  }
}
```



# 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/
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;
...
```

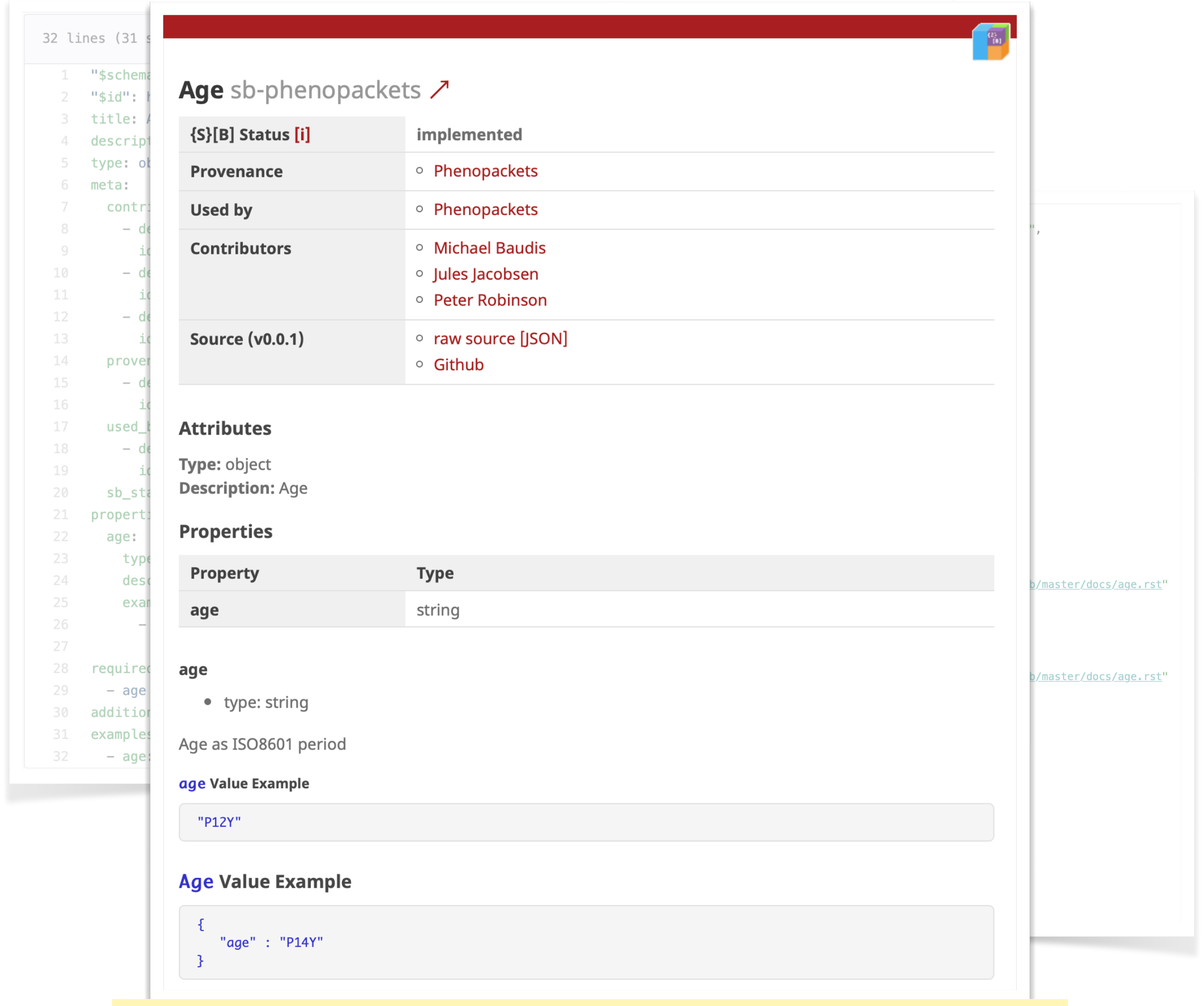
32 lines (31 sloc) | 872 Bytes — Raw | Blame | History

```yaml
"$schema": http://json-schema.org/draft-07/schema#
"$id": https://schemablocks.org/schemas/sb-phenopackets/Age/v0.0.1
title: Age
description: Age
type: object
meta:
  contributors:
    - description: "Michael Baudis"
      id: "orcid:0000-0002-9903-4248"
    - description: "Jules Jacobsen"
      id: "orcid:0000-0002-3265-15918"
    - description: "Peter Robinson"
      id: "orcid:0000-0002-0736-91998"
  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:
  age:
    type: string
    description: Age as ISO8601 period
    examples:
      - 'P12Y'

required:
  - age
additionalProperties: false
examples:
  - 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
    // 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;
...
```




- 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

# 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;
...
```

32 lines (31 sloc) | 872 Bytes | Raw | Blame | History

```
"$schema": http://json-schema.org/draft-07/sc
"$id": https://schemablocks.org/schemas/sb-ph
title: Age
description: Age
type: object
meta:
  contributors:
    - description: "Michael Baudis"
      id: "orcid:0000-0002-9903-4248"
    - description: "Jules Jacobsen"
      id: "orcid:0000-0002-3265-15918"
    - description: "Peter Robinson"
      id: "orcid:0000-0002-0736-91998"
  provenance:
    - description: Phenopackets
      id: 'https://github.com/phenopackets/ph
  used_by:
    - description: Phenopackets
      id: 'https://github.com/phenopackets/ph
  sb_status: implemented
properties:
  age:
    type: string
    description: Age as ISO8601 period
    examples:
      - 'P12Y'

required:
  - age
additionalProperties: false
examples:
  - age: 'P14Y'
```


```
{
  "$id": "https://schemablocks.org/schemas/sb-phenopackets/Age/v0.0.1",
  "$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"
      },
      {
        "description": "Peter Robinson",
        "id": "orcid:0000-0002-0736-91998"
      }
    ],
    "provenance": [
      {
        "description": "Phenopackets",
        "id": "https://github.com/phenopackets/phenopacket-schema/blob/master/docs/age.rst"
      }
    ],
    "sb_status": "implemented",
    "used_by": [
      {
        "description": "Phenopackets",
        "id": "https://github.com/phenopackets/phenopacket-schema/blob/master/docs/age.rst"
      }
    ]
  },
  "properties": {
    "age": {
      "description": "Age as ISO8601 period",
      "examples": [
        "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

# {S}[B] JSON Schemas

## (versioned & current)

- schema definition documents (YAML) are rendered into JSON Schema .json documents
- the latest version is kept under "current", with parallel versioned & archived copy & original YAML, JSON in source repository

*https://schemablocks.org/schemas/ga4gh/current/DataUseLimitation.json*

*https://schemablocks.org/schemas/ga4gh/v0.0.1/DataUseLimitation.json*

*https://github.com/ga4gh-schemablocks/sb-duo/blob/master/generated/json/current/DataUseLimitation.json*


```
{
  "$id": "https://schemablocks.org/schemas/ga4gh/DataUseLimitation/v0.0.1",
  "$schema": "http://json-schema.org/draft-07/schema#",
  "additionalProperties": "",
  "description": "The (GA4GH) Data Use Ontology (DUO) includes terms describing data use
conditions,\nparticularly for research data in the health/clinical/biomedical domain.\n",
  "examples": [
    {
      "modifier": {
        "ontology": {
          "id": "MONDO:0004992",
          "label": "cancer"
        }
      },
      "term": {
        "id": "DUO:0000007",
        "label": "disease specific research"
      }
    },
    {
      "modifier": {
        "text": "UK"
      },
      "term": {
        "id": "DUO:0000022",
        "label": "geographical restriction"
      }
    }
  ],
  "meta": {
    "contributors": [
      {
        "description": "Melanie Courtot",
        "id": "orcid:0000-0002-9551-6370"
      },
      {
        "description": "Michael Baudis",
        "id": "orcid:0000-0002-9903-4248"
      },
      {
        "description": "DUO group"
      },
      {
        "description": "Isuru Liyanage",
        "id": "orcid:0000-0002-4839-5158"
      }
    ],
    "provenance": [
      {
        "description": "DUO",
        "id": "https://github.com/EBISPOT/DUO"
      }
    ],
    "sb_status": "implemented",
    "used_by": [
      {
        "description": "DUO",
        "url": "https://github.com/EBISPOT/DUO"
      }
    ]
  },
  "properties": {
    "description": {
      "description": "free text description mainly to encapsulate those conditions that wouldn't
be described by DUO terms or modifiers\n",
      "type": "string"
    },
    "modifier": {
      "$ref": "./DataUseModifier",
      "description": "modifier/restriction applicable for the DUO"
    },
    "term": {
      "$ref": "https://schemablocks.org/schemas/sb-phenopackets/OntologyClass/v1.0.0",
      "description": "DUO - ontology term subclass of either DUO_0000001 or DUO_0000017"
    }
  },
  "required": [
    "term"
  ],
  "title": "DataUseLimitation",
  "type": "object"
}
```


## DataUseLimitation ga4gh ↗

| | |
|---|---|
| **{S}[B] Status [i]** | **implemented** |
| **Provenance** | ○ DUO |
| **Used by** | ○ DUO |
| **Contributors** | ○ Melanie Courtot<br>○ DUO group<br>○ Isuru Liyanage |
| **Source (v0.0.1)** | ○ raw source [JSON]<br>○ Github |

### Attributes

**Type:** object
**Description:** The (GA4GH) Data Use Ontology (DUO) includes terms describing data use conditions, particularly for research data in the health/clinical/biomedical domain.

### Properties

| Property | Type |
|---|---|
| **description** | string |
| **modifier** | ./DataUseModifier |
| **term** | https://schemablocks.org/schemas/sb-phenopackets/OntologyClass/v1.0.0 |

**description**

- type: string

free text description mainly to encapsulate those conditions that wouldn't be described by DUO terms or modifiers

**modifier**

- type: ./DataUseModifier

modifier/restriction applicable for the DUO

**term**

- type: https://schemablocks.org/schemas/sb-phenopackets/OntologyClass/v1.0.0

DUO - ontology term subclass of either DUO_0000001 or DUO_0000017

### `DataUseLimitation` Value Examples


```
{
   "modifier" : {
      "ontology" : {
         "id" : "MONDO:0004992",
         "label" : "cancer"
      }
   },
   "term" : {
      "id" : "DUO:0000007",
      "label" : "disease specific research"
   }
}
```

```
{
   "modifier" : {
      "text" : "UK"
   },
   "term" : {
      "id" : "DUO:0000022",
      "label" : "geographical restriction"
   }
}
```

## BeaconAlleleRequest beacon ↗

| {S}[B] Status [i] | implemented |
|---|---|
| Provenance | ○ Beacon API |
| Used by | ○ Beacon<br>○ Progenetix database schema (Beacon+ backend) |
| Contributors | ○ Marc Fiume<br>○ Michael Baudis<br>○ Sabela de la Torre Pernas<br>○ Jordi Rambla<br>○ Beacon developers... |
| Source (v1.1.0) | ○ raw source [JSON]<br>○ 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/Chror [HTML] |
| referenceBases | string |
| referenceName | https://schemablocks.org/schemas/beacon/v1.1.0/Chror [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 | ○ vr-spec |
| Used by | ○ vr-spec |
| Contributors | ○ Reece Hart<br>○ Michael Baudis |
| Source (v1.0) | ○ raw source [JSON]<br>○ Github |

### Attributes

**Type:** string
**Pattern:** ^\w[^:]+:.+$
**Description:** A string that refers to an object uniquely. T sender.

VR does not impose any contraints on strings used as ids data, the VR Specification RECOMMENDS that implement

String CURIEs are represented as `prefix:reference` (W `namespace:accession` or `namespace:local id` colloqu

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 wh VR uses CURIEs primarily as a naming mechanism.

Implementations MAY provide CURIE resolution mechan

Using internal ids in public messages is strongly discoura

### `Curie` Value Examples

```
"ga4gh:GA.01234abcde"
```

```
"DUO:0000004"
```

```
"orcid:0000-0003-3463-0775"
```

```
"PMID:15254584"
```

## Biosample sb-phenopackets ↗

| {S}[B] Status [i] | implemented |
|---|---|
| Provenance | ○ Phenopackets |
| Used by | ○ Phenopackets |
| Contributors | ○ GA4GH Data Working Group<br>○ Jules Jacobsen<br>○ Peter Robinson<br>○ Michael Baudis<br>○ Melanie Courtot<br>○ Isuru Liyanage |
| Source (v1.0.0) | ○ raw source [JSON]<br>○ Github |

### Attributes

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

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

Several instances (e.g. technical replicates) or types of experiments (e.g. genomic array as well 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] [HTMI |
| histologicalDiagnosis | https://schemablocks.org/schemas/sb-phenopackets/v1.0.0/OntologyClass.json [SRC] [HTMI |
| 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 | ○ GA4GH DRS (`develop` branch) |
| Used by | ○ GA4GH DRS<br>○ GA4GH TRS |
| Contributors | ○ Susheel Varma |
| Source (v0.0.1) | ○ raw source [JSON]<br>○ 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"
```

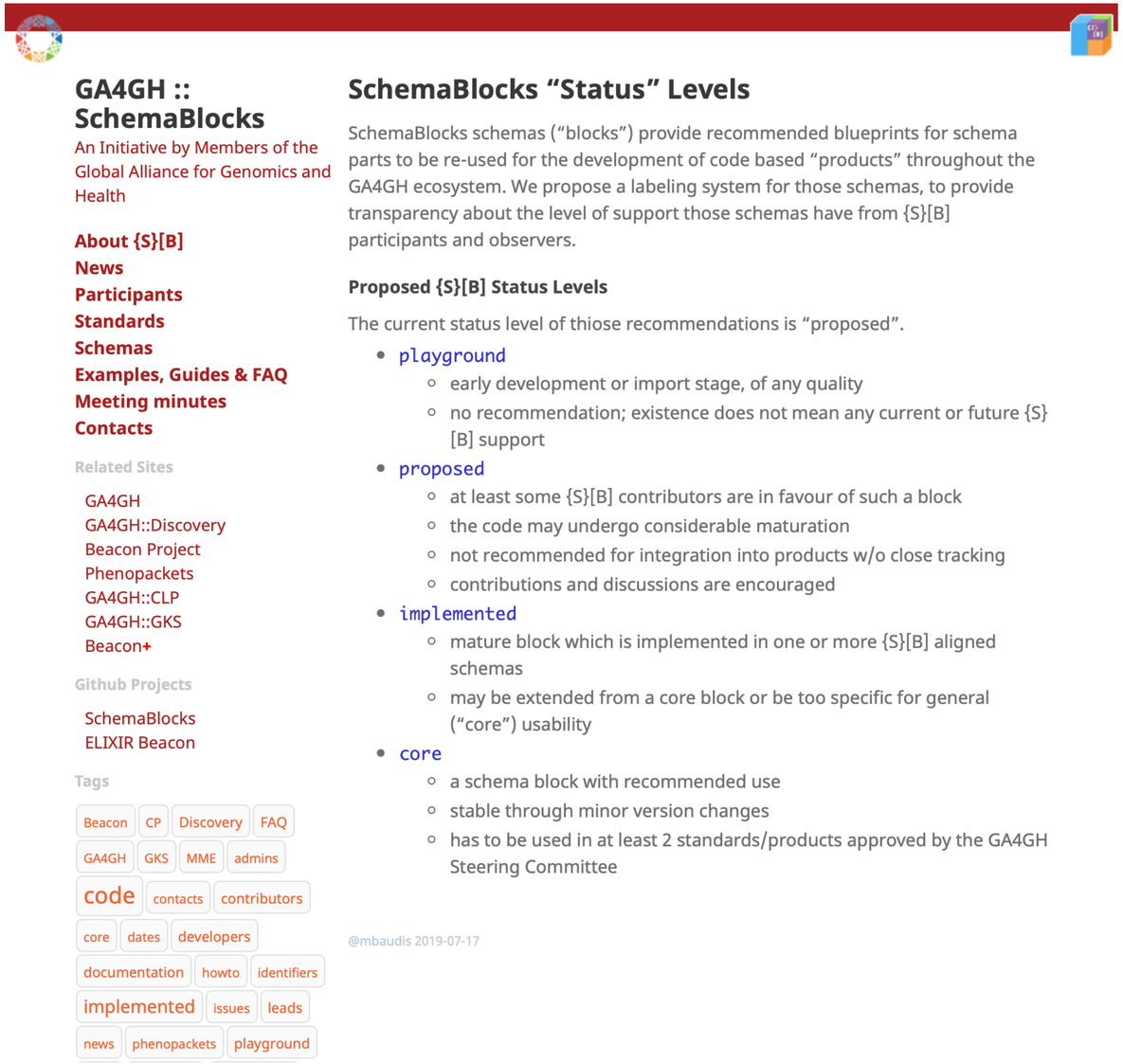
# {S}[B] SchemaBlocks ***JSON Schema*** document format

- {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 t0 the website documents


```yaml
"$schema": http://json-schema.org/draft-07/schema#
"$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'
```

# {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 t0 the website documents



**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 admins code contacts contributors core dates developers documentation howto identifiers implemented issues leads news phenopackets playground

## 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 thiose 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

@mbaudis 2019-07-17

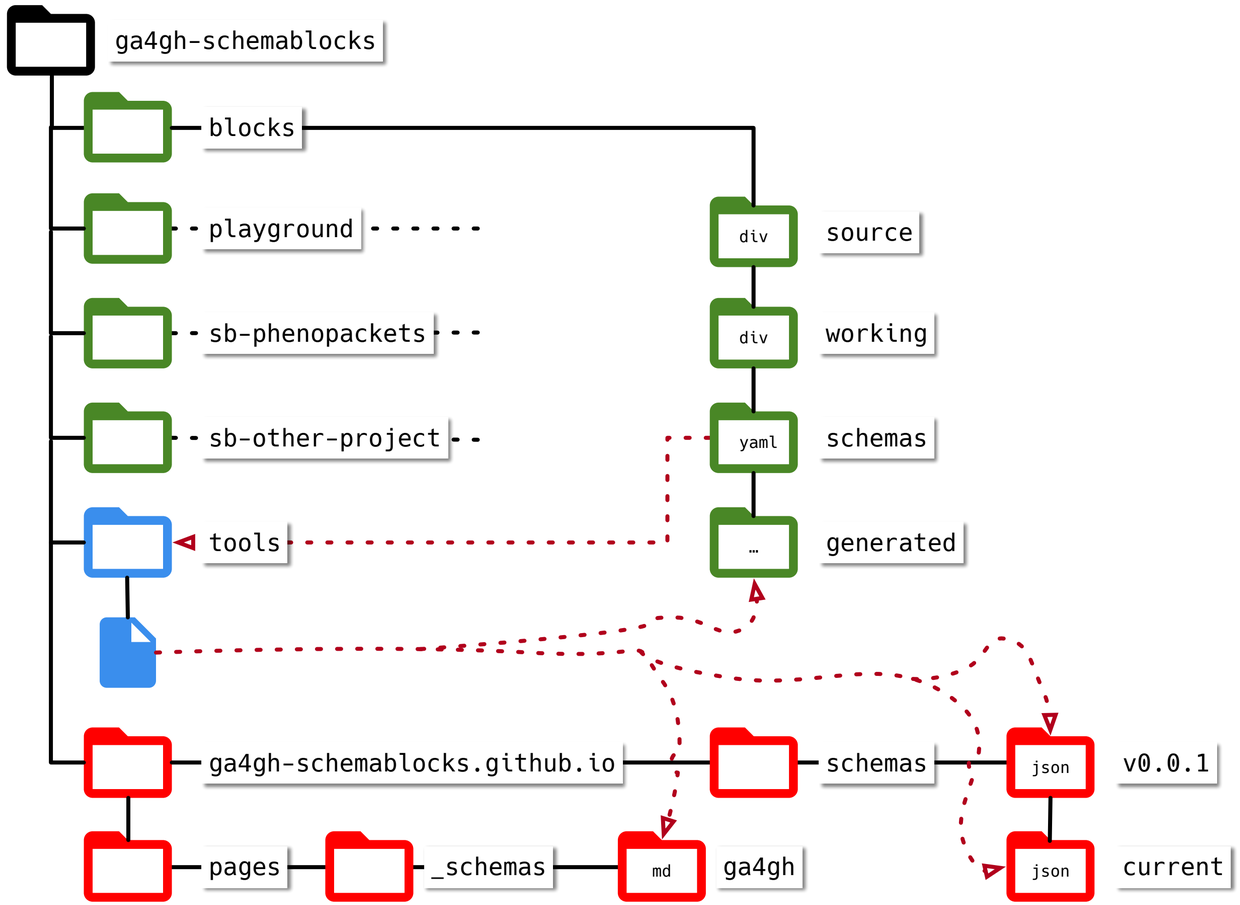
# {S}[B] SchemaBlocks Github Repository Structure

## YAML > Perl > JSON & MD > Jekyll > HTML

blocks repositories

conversion/validation tools

website repository
(Markdown w/ YAML for Github Pages)



# 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?

**Status 2020 ...**

*Technical Alignment Sub Committee

# SchemaBlocks {S}[B] in 2021

## Establishing as key component of the GA4GH standards ecosystem

- 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
  - ➡ demonstration of practical usability of referenced {S}[B] schemas in "acceptor" schemas
- establishing stable repository structures, canoncal endpoint URLs & formats
- participation of more (GA4GH) groups, to expose their standards & products
- GA4GH prodedures - blessed schema status labels?
- **support structure** - maintenance, workflows, stewardship, versioning / updating ...
- tooling / libraries

Global Alliance
for Genomics & Health
Collaborate. Innovate. Accelerate.

# SchemaBlocks {S}[B] | GA4GH Connect | Michael Baudis | 2021-03-04

## Tooling

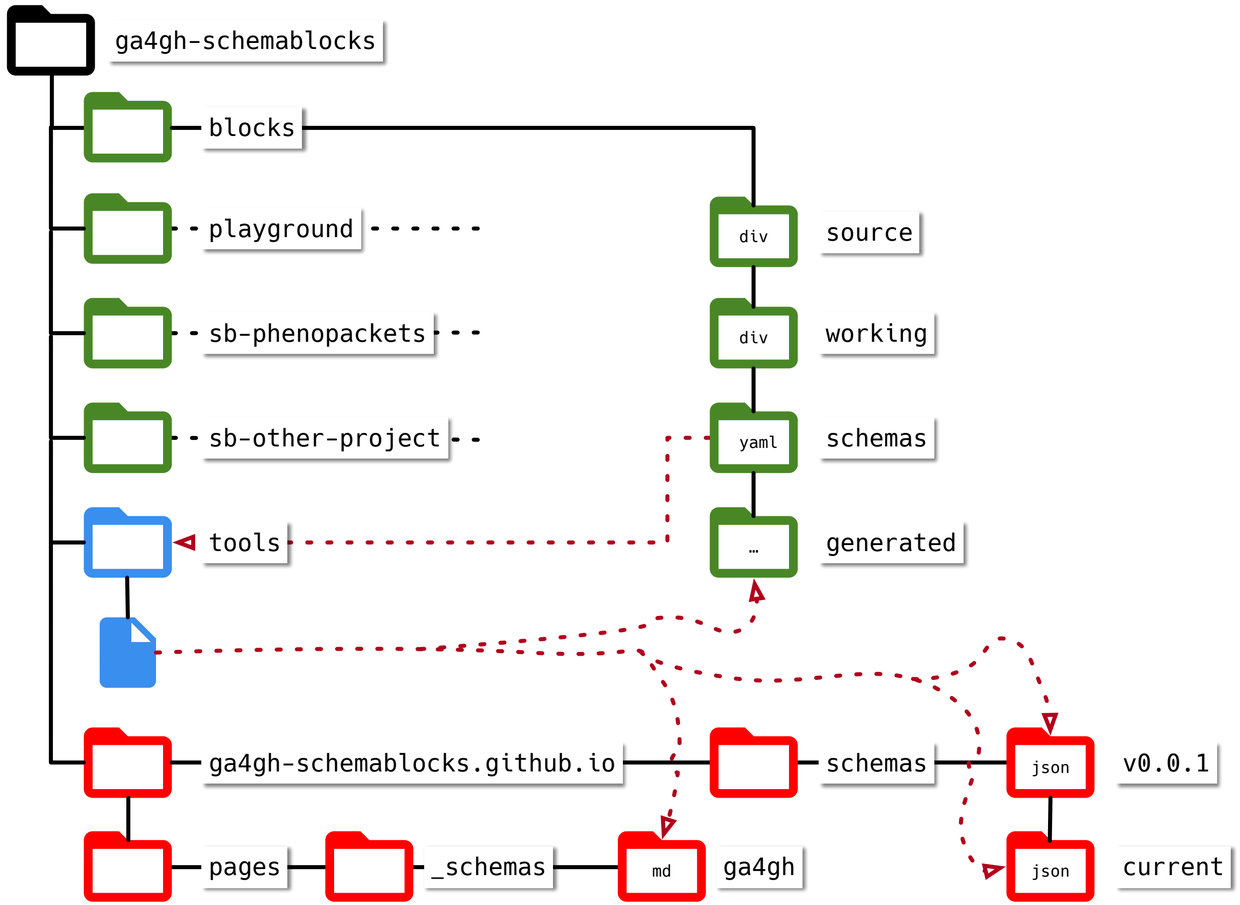
# {S}[B] SchemaBlocks Github Repository Structure

## YAML > Perl > JSON & MD > Jekyll > HTML

blocks repositories

conversion/validation tools

website repository
(Markdown w/ YAML for Github Pages)



# {S}[B] Tooling
## sbSchemaParser

- single tool which provides schema deparsing and output page generation across a *local mirror* of schema repositories and {S}[B] website source directories => "Q & D"
- configuration file for selection & labeling

➡ Works for "all in one" flexible prototyping but should be replaced by modular & safer tools

### tools

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

#### sbSchemaParser

The `sbSchemaParser.pl` script is used to process schema files written in *JSON Schema* (YAML version) into human-readable documentation (e.g. Markdown files for Jekyll based HTML generation) and JSON data files from the embedded examples.

Directives for source and target directories can be modified in the `config.yaml` file in the script's directory. The general repository structures for the repositories which are being parsed by *sbSchemaParser* is shown below.

**{S}[B] Repositories**

{S}[B] code repositories adhere a consistent structure & naming:

```
sb-code           # each of the code repositories
  |
  |- source       # original code
  |- working      # for editing, temporary...
  |- schemas      # JSON Schema files as YAML; read to produce the output files
  |- generated    # contains files generated from main schema YAML files
     |- json     # .json version of the schema
     |- examples # .json example data, from inline examples
     |- doc      # .md documentation, from inline documentation
```

Here

- The `source` and `working` directories are optional.
- The `json`, `examples` and `doc` directories are populated by the *sbSchemaParser*

**Website Files**

The *sbSchemaParser* also generates copies of the `myschema.json` files into the canonical website directory, and a GH-pages version of the Markdown documentation file into the `pages` tree processed by the GH-pages "Jekyll" processing engine. The .md file contains a `permalink` directive in its YAML header, which will lead to GH-pages placing the HTML page at "https://schemablocks.org/schemas/ga4gh/myschema.html".

```
ga4gh-schemablocks.github.io
  |
  |- schemas
  |        |- ga4gh # json version of the schema, generated from YAML
  |                 # => https://schemablocks.org/schemas/ga4gh/myschema.json
  |- pages
        |- _schemas
                |- ga4gh  # the Jekyll Markdown files for the website
```

Global Alliance
for Genomics & Health
Collaborate. Innovate. Accelerate.

{S}
[B]

Melanie Courtot
Michael Baudis
Isuru Liyanage
Melissa Konopko
Jonathan Fuerth
Andy Yates
Ben Hutton
Bo Gao

... and many, many others
in meetings & discussions

elixir

SPHN

Global Alliance
for Genomics & Health
Collaborate. Innovate. Accelerate.

**https://schemablocks.org**

**https://github.com/ga4gh-schemablocks/**