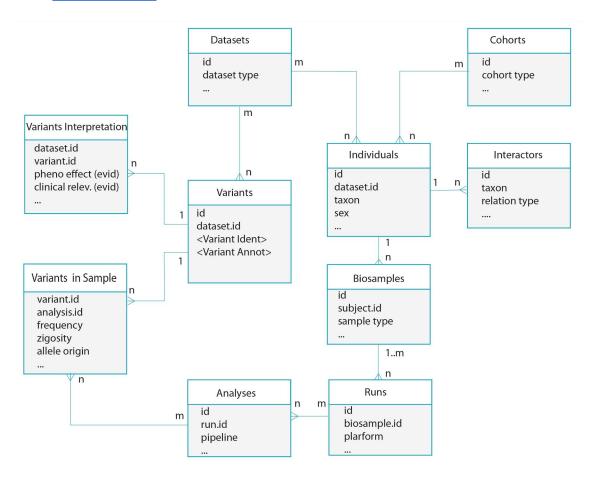
# Beacon v.2 default schemas draft

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For comments> link to this document



-Check the issue with Dataset object vs Beacon original one

# **OntologyTerm**

This object describes how to format fields which contain ontology terms. It is not a querable entity.

id Ontology term ID using CURIE syntax (SO:0001483).

**label** This is the "preferred label" for this ontology term (SNV).

### **Datasets**

datasetId alphanumeric value Dataset reference ID

datasetSource categorical value Reference to dataset source, e.g. "DECIPHER", "DisGenNET"

### Variants Identification

variantId alphanumeric value Reference to variant ID

assemblyId categorical value Genomic assembly accession and version as RefSqeq assembly accession (e.g. "GCG 000001405.39") or a versioned assembly name or synonym such as UCSC Genome Browser assembly (e.g. "hg38") or Genome Reference consortium Human (e.g. "GRCh38.p13") names

refseqId categorical value Reference sequence Refseq ID and version for genomic reference sequence in which variant coordinates are given, e.g. "NC 000009" for human chromosome 9. Alternatively, names, synonymous or aliases e.g. "Chr9" when **assemblyId** is given. For organisms with a single reference sequence covering the genome, the versioned Refseq ID can be given here as an alternative to the assembly ID and version in **assemblyId**, e.g. "NC 045512.2" for SARS-CoV2 full-length genome reference sequence.

**start** *numeric value* Start position of variant

end numeric value End position of variant

**ref** alphanumeric value Reference sequence in start-end coordinates

alt alphanumeric value Alternative sequence in start-end coordinates

variantType OntologyTerm object Type of variant. Value from Sequence Ontology, e.g. "SNV" (SO:0001483), "structural variant" (SO:0001537)

### **Variants Annotation**

variantId alphanumeric value Reference to Variant ID

variantAlternativeIds (List of) alphanumeric value(s) Cross-referencing ID(s) (CURIE(s)) for the variant in the original databases or variant-level (aggregated) databases for previously described variants (e.g. clinVarId, ClinGen, COSMIC), e.g. "VCV000055583.1", "CA003602"

genomicHGVSId alphanumeric value HGVSId descriptor at genomic level (recommended, referred to genome assembly defined in Variant Identification), e.g. "NC 000017.10:g.41199678C>A"

transcriptHGVSIds (List of) alphanumeric value(s) HGVSId descriptor at transcript level: "NC 000023.10(NM 004006.2):c.357+1G

proteinHGVSIds (List of) alphanumeric value(s) HGVSId descriptor(s) at protein level (for protein-altering variants), e.g. "NP 009225.1:p.Glu1817Ter" or "LRG 199p1:p.Val25Gly" (preferred)

**genomicRegions** (List of) OntologyTerm object(s) Classification(s) of the vari-

ant according to the genomic region affected (all that apply, relative to each feature affected). Value from Ensembl

Glossary (ENSGLOSS) Variant consequence ontology (ENSGLOSSARY:00000134, e.g. "3UTR" (ENSGLOSSARY:0000159), "coding sequence variant" (ENSGLOSSARY:0000159), "upstream gene variant" (ENSGLOSSARY:0000164), "intergenic variant" (ENSGLOSSARY:0000174), "intron variant" (ENSGLOSSARY:0000161), "non-coding transcript variant" (ENSGLOSSARY:0000163)

**genomicFeatures** Genomic feature(s) affected by the variant. (List of:)

class OntologyTerm object(s) Class of feature affected by the variant. Value from ENSGLOSS Biotype (ENSGLOSSARY:0000025) ontology, e.g. "protein coding gene", "non-coding RNA", "long non-coding RNA'

**featureId** (alphanumeric value) ID /accession/name of feature affected by the variant, matching **class**, e.g. "TP53", "GeneID:43740578"

molecularEffects (List of) OntologyTerm object(s) Predicted effect at nucleotide level for protein affecting variants. Value from Sequence ontology (SO) coding sequence variant (SO:0001580) ontology, e.g. "synonymous variant" (SO:0001819), "nonsynonymous vari- ant" (SO:0001992) (and classifications therein, such as "stop gained" (SO:0001587), "missense variant" (SO:0001583), "inframe indel" (SO:0001820))

**aminoacidChanges** (List of) categorical value(s) Change at aminoacid level for protein affecting missense variants e.g. "V304\*"

### **Individuals**

**individualId** alphanumeric value Individual reference ID (external accession or internal ID)

datasetId alphanumeric value Reference to dataset ID

taxonId alphanumeric value Taxon ID of Individual, e.g. "9606"

**sex** OntologyTerm object Sex of Individual. Value from NCIT General Qualifier ontol- ogy (NCIT:C27993): "UNKNOWN" (not assessed or not available) (NCIT:C17998), "FEMALE" (NCIT:C46113), "MALE" (NCIT:C46112) or "OTHER SEX" (NCIT:C45908)

**ethnicity** OntologyTerm object Ethnic background of Individual. Value from NCIT Race ontology (NCIT:C17049). e.g. "Latin American" (NCIT:C126531)

**geographicOrigin** OntologyTerm object Individual's country or region of origin (birthplace or residence place regardless of ethnic origin). Value from GAZ Geographic Location ontology (GAZ:00000448), e.g. "United States of America" (GAZ:00002459)

**phenotypicFeatures** Phenotypic feature(s) observed in the Individual, defined by phenotype, date, type or age of onset and level/ severity. (List of:)

**phenotypeId** OntologyTerm object Phenotypic feature observed. Value from Human Phenotype Ontology (HPO)

**dateOfOnset** alphanumeric value (ISO8601 date format) Date of onset/observation of phenotype, e.g. "2010-07-10"

**onsetType** OntologyTerm object Onset type. Value from HPO Onset ontology (HP:0003674), e.g. "congenital onset" (HP:0003577), "adult onset" (HP:0003581)

**ageAtOnset** Individual's age at onset/observation of phenotype

age alphanumeric value (ISO8601 duration format) Age, e.g. "P32Y6M"

**ageGroup** OntologyTerm object Age group. Value from NCIT Age Group ontology, e.g. "NCIT:C27954" (Adolescent)

**level/severity** OntologyTerm object Level/severity when and as applicable to phenotype observed. Value from Human Phenotype Ontology (HPO) Severity ontology (HP:0012824), e.g. "severe" (HP:0012828)

**diseases** Disease(s) been diagnosed to the Individual, defined by disease ID, date, type or age of onset, stage, level/severity and the presence of family history. (List of:)

**diseaseId** OntologyTerm object Disease ID. Value from ICD10 disease codes or ontology terms from disease ontologies such as HPO, OMIM, Orphanet, MONDO, e.g. "lactose intolerance" (HP:0004789)

**dateOfOnset** alphanumeric value (ISO8601 date format) Date of onset/diagnosis of disease, e.g. "1987-10-28"

**onsetType** OntologyTerm object Onset type. Value from HPO Onset ontology (HP:0003674), e.g. "congenital onset" (HP:0003577), "adult onset" (HP:0003581)

ageOfOnset Individual's age at onset/diagnosis of disease

**age** alphanumeric value (ISO8601 duration format) Age, e.g. "P6M"

**ageGroup** OntologyTerm object Age group. Value from NCIT Age Group ontology, e.g. "NCIT:C27954" (Adolescent)

**stage** OntologyTerm object Stage of disease. Value from Ontology for General Medical Science or Disease Stage Qualifier ontology (NCIT:C28108), e.g. " acute onset" (OGMS:0000119)

**level/severity** OntologyTerm object Level/severity when and as applicable to disease course. Value from Human Phenotype Ontology (HPO) Severity ontology (HP:0012824), e.g. "mild" (HP:0012825)

**familyHistory** boolean indicating determined or self-reported presence of family history of the disease

**treatments** Treatment(s) or Medication(s) been prescribed/administered to Individual, defined by treat ment ID, date and age of onset, dose, schedule and duration. (List of:)

**treatmentId** OntologyTerm object Treatment ID. Value from NCIT Drug, Food, Chemical or Biomedical Material.

**treatmentRoute** OntologyTerm object Route of treatment. Value from NCIT Route of Administration ontology.

**TreatmentStartDate** alphanumeric value (ISO8601 duration format) Date of the beginning of treatment

**ageAt**Treatment Individual's age at the beginning of treatment

age alphanumeric value (ISO8601 duration format) Age

**ageGroup** OntologyTerm object Age group. Value from NCIT Age Group ontology, e.g. "NCIT:C27954" (Adolescent)

treatmentDose Treatment dose

doseValue numeric value Quantity value of Dose

doseUnits OntologyTerm object Treatment dose units. Value from Units of measurement Ontology (UO)

**schedule** OntologyTerm object Treatment schedule. Value from NCIT Schedule Frequency ontology, e.g "weekly" (NCIT:C67069)

duration alphanumeric value (ISO8601 duration format) Treatment duration

**interventions** Intervention(s) been practiced on Individual, defined by intervention ID), date and age of onset, dose, schedule and duration. (List of:)

**interventionId** OntologyTerm object Intervention ID. Value from Medical Action Ontology or code from OPCS Classifications of Interventions and Procedures.

**modifier** Modifier of the intervention. Value from TBD.

**interventionDate** alphanumeric value (ISO8601 date format) Date of intervention

**ageAtIntervention** Individual's age at the date of intervention, as age or age range

**age** alphanumeric value (ISO8601 duration format) Age

**ageGroup** OntologyTerm object Age group value, from NCIT Age Group ontology, e.g. "NCIT:C27954" (Adolescent)

**measures** Measures(s) been taken from Individual, defined by measure ID, date and/or age at measure ment, unit and value. (List of:)

measureId OntologyTerm object Measure ID. Value from Clinical Measurement ontology (CMO)

#### **measureQuantity**

measureValue numeric value Quantity value of measurement

measureUnit OntologyTerm object Measure units

**modifier** Modifier of the measurement. Value from TBD.

measureDate alphanumeric value (ISO8601 date format) Date of measurement

**ageAtMeasure** Individual's age at the date of measurement, as age or age range

age alphanumeric value (ISO8601 duration format) Age

**ageGroup** OntologyTerm object Age group value, from NCIT Age Group ontology, e.g. "NCIT:C27954" (Adolescent)

**exposures** Exposures(s) occurred to or practiced (lifestyle, behavioural exposures) by Individual, defined by exposure ID, date and age of onset, dose, schedule and duration. (List of:)

**exposureId** OntologyTerm object Intervention ID. Value from Environment Exposure Ontology.

**exposureDose** Exposure dose

exposureValue numeric value Quantity value of exposure

exposureUnit OntologyTerm object Exposure units

**modifier** Modifier of the exposure. Value from TBD. e.g frequency

**exposureDate** alphanumeric value (ISO8601 duration format) Date of exposure

**ageAtExposure** Individual's age at the date of exposure, as age or age range

age alphanumeric value (ISO8601 duration format) Age

**ageGroup** OntologyTerm object Age group value, from NCIT Age Group ontology, e.g. "NCIT:C27954" (Adolescent)

duration alphanumeric value (ISO8601 duration format) Exposure duration

**pedigrees** Pedigree(s) to which the Individual belongs. (List of:)

pedigreeId alphanumeric value Reference to pedigree ID (external accession)

**pedigreeDisease** categorical value (disease code/ontology term object) Disease ID. Value from ICD10 disease codes or ontology terms from disease ontologies such as HPO, OMIM, Orphanet, MONDO, e.g. "lactose intolerance" (HP:0004789)

**pedigreeRole** OntologyTerm object Pedigree role, defined as relation- ship to proband. Value from Family member ontology (NCIT:C41256), e.g. "proband" (NCIT:C64435), "identical twin" (NCIT:C73429), "mother" (NCIT:C25189)

**affectedStatus** categorical value Affected status of Individual in disease of pedigree: "af- fected" or "unaffected"

**numSubjects** numeric value Number of subjects in pedigree

## **Biosamples**

**biosampleId** alphanumeric value Biosample reference ID (external accession)

individual Id Reference to Individual ID

**description** free text Any relevant info about the biosample that does not fit in any field in the schema

biosampleStatus OntologyTerm object Classification of biosample based on their

role in study. Value from Experimental Factor Ontology (EFO) Material Sample ontology (OBI:0000747) Classification of the sample in "abnormal sample" (EFO:0009655) or "reference sample" (EFO:0009654)

**collectionDate** alphanumeric value (ISO8601 date format) Date of biosample collection, e.g. "2019-12"

**subjectAgeAtCollection** alphanumeric value (ISO8601 duration format) Individual's age at the time of biosample collection, e.g. "P72Y6M19D"

**sampleOriginDescriptors** Descriptor(s) of origin of biosample. (List of:)

**sampleOriginType** OntologyTerm object Category of sample origin. Value from Ontology for Biomedical Investigations (OBI) material entity (BFO:0000040) ontology, e.g. "specimen from organism" (OBI:0001479), "xenograft" (OBI:0100058), "cell culture" (OBI:0001876), "cell specimen" (OBI:0001468), "environmental swab specimen" (OBI:0002613)

**sampleOriginDetail** OntologyTerm object Specific instance of sample origin matching the category set in **sampleOriginType**. Value from Uber-anatomy ontology (UBERON) or BRENDA tissue / enzyme source (BTO), Ontology for Biomedical Investi- gations (OBI) or Cell Line Ontology (CLO), e.g. "cerebellar vermis" (UBERON:0004720), "HEK-293T cell" (BTO:0002181), "nasopharyngeal swab specimen" (OBI:0002606), "cere- brospinal fluid specimen" (OBI:0002502)

**obtentionProcedure** OntologyTerm object Ontology ID from NCI Thesaurus (NCIT) Intervention or Procedure (NCIT:C25218) ontology. e.g. "biopsy" (NCIT:C15189)

cancerFeatures Values specifying cancer-specific features, including progression and tumor grade

**tumorProgression** OntologyTerm object Descriptor of tumor progression. Value from Neoplasm by Special Category ontology (NCIT:C7062). Tumor progression category indicating primary, metastatic or recurrent progression e.g. "Primary Malignant Neo- plasm" (NCIT:C84509)

**tumorGrade** OntologyTerm object Tumor grading. Value from Tumor Grading Characteristic ontology (Mondo Disease Ontology MONDO:0024488), e.g. "Grade 3" (MONDO:0024493)

#### Runs

runId alphanumeric value Run reference ID (external accession or internal ID)

biosampleId alphanumeric value Reference to Biosample ID

**runDate** alphanumeric value (ISO8601 date format) Date at which run was performed, e.g. "2019-12-28"

librarySource categorical value Sequencing library source, e.g. "Metagenomic", "Viral RNA"

libraryStrategy categorical value Sequencing library strategy, e.g. "WGS"

**librarySelection** categorical value Selection method for sequencing library preparation, e.g. "RAN- DOM", "RT-PCR"

libraryLayout categorical value Sequencing library layout, e.g. "PAIRED", "SINGLE"

platform categorical value Sequencing technology, e.g. "Illumina", "Oxford Nanopore Technologies"

**platformModel** categorical value Sequencing platform model, e.g. "Illumina MiSeq", "GridION"

## **Analyses**

analysisId alphanumeric value Analysis reference ID (external accession or internal ID)

runId alphanumeric value Reference to run ID

analysisDate alphanumeric value (ISO8601 date format) Date at which analysis was performed, e.g. "2020-01-10"

**pipelineName** categorical value Analysis pipeline and version, e.g., "Isaac Whole Genome Sequencing; version 3.0.0.0"

**pipelineRef** Link to Analysis pipeline resource

**aligner** categorical value Mapping/Alignment software e.g. "bwa-0.7.8"

variantCaller categorical value Variant calling software/pipeline, e.g. "GATK4.0"

# **Variants in Sample**

variantId alphanumeric value Reference to variant ID

analysis Id Reference to analysis ID

**biosampleId** Reference to biosample ID

variantFrequency numeric value Variant/allele frequency in biosample

**zigosity** OntologyTerm object Zigosity in which variant is present in the sample.

Value from the Zigosity Ontology (GENO:0000133), e.g. "heterozygous" (GENO:0000135)

**alleleOrigin** OntologyTerm object Allele origin of variant in sample. Value from the Variant Origin (SO:0001762), e.g. "somatic variant", "germline variant", "de novo variant".

**phenotypicEffects** Observed effect(s) of variant on phenotype. (List of:)

**phenotypeId** OntologyTerm object Descriptor of phenotype found associated to variant in the present study. Value from Human Phenotype Ontology (HPO)

**phenotypeEffect** OntologyTerm object Phenotypic effect classification determined in the present study. Value from Sequence types and features ontology (SO) variant phenotype (SO:0001769), e.g. "quantitative variant" (SO:0001774)

**evidenceType** OntologyTerm object Type of evidence supporting variant-phenotype association from the Evidence & Conclusion Ontology (ECO), e.g. "experimental evidence"

**clinicalRelevances** Observed effect of variant on disease. (List of:)

**diseaseId** OntologyTerm object Descriptor of disease associated. Value from ICD10 disease codes or ontology terms from disease ontologies such as HPO, OMIM, Orphanet, MONDO, e.g. "lactose intolerance" (HP:0004789)

**clinicalEffect** OntologyTerm object Clinical effect classification. Value from Sequence types and features ontology (SO) variant phenotype (SO:0001769), e.g. "disease causing variant" (SO:0001772)

**evidenceType** OntologyTerm object Type of evidence supporting variant- disease association from the Evidence & Conclusion Ontology (ECO), e.g. "experimental evidence"

## **Variants Interpretation**

variantId alphanumeric value Reference to Variant ID

datasetId alphanumeric value Reference to dataset ID source of variant interpretation

**phenotypicEffects** Annotated effect(s) of variant on any phenotypic feature other than a disease. (List of:)

**phenotypeId** OntologyTerm object Descriptor of phenotype associated. Value from Human Phenotype Ontology (HPO)

**phenotypeEffect** OntologyTerm object Phenotypic effect classification. Value from Sequence types and features ontology (SO) variant phenotype (SO:0001769), e.g. "benign variant" (SO:0001770)

**alleleOrigin** (List of) OntologyTerm object(s) Annotation(s) on allele origins in which the variant has been found in association to phenotype. Value(s) from Se-quence types and features ontology (SO) variant origin (SO:0001762) ontology, e.g. "somatic variant" (SO:0001777), "germline variant" (SO:0001778), "de novo variant" (SO:0001781)

**references** (List of) publication identifier(s) Reference(s) to publication(s) describing association, as PMID(s), e.g. "PMID:17024373"

**clinicalRelevances** Annotated effect(s) of variant on disease. (List of:)

**diseaseId** OntologyTerm object Disease. Value from ICD10 disease codes or ontology terms from disease ontologies such as HPO, OMIM, Orphanet, MONDO. e.g. "lactose intolerance" (ICD10CM:E73)

**clinicalEffect** OntologyTerm object Clinical effect classification. Value from Sequence types and features ontology (SO) variant phenotype (SO:0001769), e.g. "dis- ease associated variant" (SO:0001771)

**alleleOrigin** (List of) OntologyTerm object(s) Annotation(s) on allele origins in which the variant has been in association to condition. Value(s) from Sequence types and features ontology (SO) variant origin (SO:0001762) ontology, e.g. "somatic variant" (SO:0001777), "germline variant" (SO:0001778), "de novo variant" (SO:0001781)

**references** (List of) publication identifier(s) Reference(s) to publication(s) describing association, as PMID(s), e.g. "PMID:19793655"

#### **Interactors**

This is an organism/agent whose phenotypic data is collected in association with the Individual, but which is not the sequenced/genotyped Individual itself. This block accounts for the 'extended phenotype' of variants in organisms/agents other than the one harboring them.

**interactorId** alphanumeric value Interactor organism/ agent reference ID (external accession or internal ID)

**relationType** OntologyTerm object Type of relation with Individual. Value from Infectious disease Ontology(IDO), e.g. "host" (IDO:0000531), "commensal" (IDO:0000525), "in-fectious agent" (IDO:0000596)

[...] All the rest of objects from Individual

## Cohorts (<u>link</u>)

**cohortType** categorical value Cohort type by its definition. One of: study-defined, beacon-defined or user-defined. If a cohort is declared study-defined or beacon-defined, criteria are to be entered in **cohortInclusionCriteria**; if a cohort is declared 'user-defined' **cohortInclusionCriteria** will be automatically populated from the parameters used to perform the query.

**cohortId** alphanumeric value Cohort identifier. For 'study-defined' or 'beacon-defined' cohorts this field is set by the implementer. For 'user-defined' this unique identifier could be generated upon the query that defined the cohort, but could be later edited by the user

**cohortName** alphanumeric value Cohort name. For 'user-defined' this field could be generated upon the query, e.g. a value that is a concatenation or some representation of the user query

**cohortInclusionCriteria** Inclusion criteria. For 'beacon-defined cohorts, cohorts matching the whole dataset will not apply criteria for if cohorts are subsets of Datasets the criteria used to defined them can be added in these fields). (List of:)

cohortLocations (list of) OntologyTerm object Geographic location(s) in cohort inclusion criteria

cohortGenders (list of) OntologyTerm object Gender(s) in cohort inclusion criteria

cohortEthnicities (list of) OntologyTerm object Ethnicity(ies) in cohort criteria

cohortDiseases (list of) OntologyTerm object Disease(s) in cohort inclusion criteria

cohortPhenotypicFeatures (list of) OntologyTerm object Phenotypic feature(s) in in cohort inclusion criteria

cohortAgeRange categorical value Individual age range in cohort inclusion criteria

start alphanumeric value (ISO8601 duration format) Min age accepted in cohort criteria

end alphanumeric value (ISO8601 duration format) Max age accepted in cohort criteria

cohortExclusionCriteria Exclusion criteria. Same structure as cohortInclusionCriteria

cohortLicense TBD

cohortRight TBD

cohortContact TBD

**cohortSize** integer Count of unique Individuals in cohort (individuals meeting criteria for 'user-defined' cohorts). If not previously known, it could be calculated by counting the individuals in the cohort.

**cohortDataTypes** Data types available from the Cohort. (List of:)

**dataType** OntologyTerm object Data types included in cohort (GECKO, e.g. Genomic data (GECKO:0000032), Clinical Measurements (CMO:0000000), Lifestyle (GECKO:0000067), etc. **num** paired to id field, number of individuals with this data type in the cohort

**collectionEvents** Collection events/ data points (List of:)

#### collectionEvent

**eventNum** integer Cardinality of the collection event/data point in a series

**eventDate** ISO8601 date and time format date of collection event/data point.

**eventTimeline** Aggregated information of dates of visit/diagnostic/inclusion in study obtained from individual level info in database. Will coincide with collection event date for multi-time

**start** alphanumeric value (ISO8601 date format) Earliest date of event

end alphanumeric value (ISO8601 date format) Latest date of event

**eventSize** integer Count of individuals in cohort at data point (for 'user-defined' cohorts, this is individuals meeting criteria) obtained from individual level info in database.

eventCases integer count of cases

**eventControls** integer Count of controls

eventLocations Aggregated information of geographic location obtained from individual level info in

database.

availability boolean availability

availabilityCount integer count of individuals with data available

distribution list of: [unique values in field, count]

eventGenders (Aggregated information of gender(s) obtained from individual level info in database.

availability boolean data availability

availabilityCount integer count of individuals with data available

**distribution** (List of) [unique values in field, count]

**eventEthnicities** Aggregated information of ethnicity obtained from individual level info in database.

availability boolean data availability

availabilityCount integer count of individuals with data available

**distribution** (List of) [unique values in field, count]

eventAgeRange Individual age range, obtained from individual level info in database

**start:** alphanumeric value (ISO8601 duration format) Min age in collection event **end** alphanumeric value (ISO8601 duration format) Max age in collection event

**eventDiseases** Aggregated information of disease/condition(s) obtained from individual level info in database.

availability boolean data availability

availabilityCount integer count of individuals with data available

**distribution** (List of) [unique values in field, count]

**eventPhenotypes** (GECKO disease or disorder: MONDO:0000001) Aggregated information of phenotype(s) obtained from individual level info in database.

availability boolean data availability

availabilityCount integer count of individuals with data available

distribution (List of) [unique values in field, count]

**eventDataTypes** (list of) Aggregated data type information available for each cohort data type as declared in cohort data types, and obtained from individual level info

availability boolean data availability

availabilityCount integer count of individuals with data available

distribution (List of) [unique values in field, count]