Beacon v.2 default schemas v0.2

Dataset

datasetId CATEGORICAL VALUE Dataset reference ID

datasetSource CATEGORICAL VALUE Reference to dataset source, e.g. “DECIPHER”, “DisGenNET”

datasetType CATEGORICAL VALUE Type of dataset: “case-level” or “variant-level” (aggregated)

Variant 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 CATEGORICAL VALUE (ONTOLOGY TERM) Type of variant. Value from Sequence Ontology, e.g. “SNV” (SO:0001483), “structural variant” (SO:0001537)

Variant 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) categorical value(s) (ontology term) Classification(s) of the variant 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** categorical value (ontology term) 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) categorical value(s) (ontology term) 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 variant" (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*"

Subject

subjectId alphanumeric value Subject reference ID (external accession or internal ID)

datasetId alphanumeric value Reference to dataset ID

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

sex categorical value (ontology term) Sex of subject. Value from NCIT General Qualifier ontology (NCIT:C27993): "UNKNOWN" (not assessed or not available) (NCIT:C17998), "FEMALE" (NCIT:C46113), "MALE" (NCIT:C46112) or "OTHER SEX" (NCIT:C45908)

ethnicity categorical value (ontology term) Ethnic background of subject. Value from NCIT Race ontology (NCIT:C17049). e.g "Latin American" (NCIT:C126531)

geographicOrigin categorical value (ontology term) Subject'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 subject, defined by phenotype, date, type or age of onset and level/ severity. (List of:)

> **phenotypeId** categorical value (ontology term) 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** categorical value (ontology term) Onset type. Value from HPO Onset ontology (HP:0003674), e.g. "congenital onset" (HP:0003577), "adult onset" (HP:0003581)
>
> **ageOfOnset** Subject's age at onset/observation of phenotype
>
> > **age** alphanumeric value (ISO8601 duration format) Age, e.g. "P32Y6M"
> >
> > **ageGroup** categorical value (ontology term) Age group. Value from NCIT Age Group ontology, e.g. "NCIT:C27954" (Adolescent)
>
> **level/severity** categorical value (ontology term) 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 subject, defined by disease ID, date, type or age of onset, stage, level/severity and the presence of family history. (List of:)

diseaseId categorical value (disease code /ontology term) 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 categorical value (ontology term) Onset type. Value from HPO Onset ontology (HP:0003674), e.g. "congenital onset" (HP:0003577), "adult onset" (HP:0003581)

ageOfOnset Subject's age at onset/ diagnosis of disease

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

ageGroup categorical value (ontology term) Age group. Value from NCIT Age Group ontology, e.g. "NCIT:C27954" (Adolescent)

stage categorical value (ontology term) 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 categorical value (ontology term) 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

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

pedigreeId alphanumeric value Reference to pedigree ID (external accession)

pedigreeDisease categorical value (disease code /ontology term) 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 categorical value (ontology term) Pedigree role, defined as relationship 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 subject in disease of pedigree: "affected" or "unaffected"

numSubjects numeric value Number of subjects in pedigree

Biosample

biosampleId alphanumeric value Biosample reference ID (external accession)

subjectId Reference to subject ID

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

biosampleStatus categorical value (ontology term) 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) Subject's age at the time of biosample collection, e.g. "P72Y6M19D"

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

sampleOriginType CATEGORICAL VALUE (ONTOLOGY TERM) 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 CATEGORICAL VALUE (ONTOLOGY TERM) 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 Investigations (OBI) or Cell Line Ontology (CLO), e.g. "cerebellar vermis" (UBERON:0004720), "HEK-293T cell" (BTO:0002181), "nasopharyngeal swab specimen" (OBI:0002606), "cerebrospinal fluid specimen" (OBI:0002502)

obtentionProcedure CATEGORICAL VALUE (ONTOLOGY TERM) 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 CATEGORICAL VALUE (ONTOLOGY TERM). 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 Neoplasm" (NCIT:C84509)

tumorGrade CATEGORICAL VALUE (ONTOLOGY ID) Tumor grading. Value from Tumor Grading Characteristic ontology (Mondo Disease Ontology MONDO:0024488), e.g. "Grade 3" (MONDO:0024493)

Run

runId ALPHANUMERIC VALUE Run reference ID (external accession or internal ID)

biosampleId ALPHANUMERIC VALUE Reference to sample 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. "RANDOM", "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"

Analysis

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"

Variant in Sample

variantId ALPHANUMERIC VALUE Reference to variant ID

analysisId Reference to analysis ID

biosampleId Reference to biosample ID

variantFrequency NUMERIC VALUE Variant/allele frequency in biosample

zigosity CATEGORICAL VALUE (ONTOLOGY TERM) Zigosity in which variant is present in the sample. Value from the Zigosity Ontology (GENO:0000133), e.g. "heterozygous" (GENO:0000135)

alleleOrigin CATEGORICAL VALUE (ONTOLOGY TERM) 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** CATEGORICAL VALUE (ONTOLOGY TERM) Descriptor of phenotype found associated to variant in the present study. Value from Human Phenotype Ontology (HPO)
>
> **phenotypeEffect** CATEGORICAL VALUE (ONTOLOGY TERM) 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** CATEGORICAL VALUE (ONTOLOGY TERM) 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** CATEGORICAL VALUE (DISEASE CODE/ ONTOLOGY TERM) 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** CATEGORICAL VALUE (ONTOLOGY TERM) Clinical effect classification. Value from Sequence types and features ontology (SO) variant phenotype (SO:0001769), e.g. "disease causing variant" (SO:0001772)
>
> **evidenceType** CATEGORICAL VALUE (ONTOLOGY TERM) Type of evidence supporting variant-disease association from the Evidence & Conclusion Ontology (ECO), e.g. "experimental evidence"

Variant 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** CATEGORICAL VALUE (ONTOLOGY TERM) Descriptor of phenotype associated. Value from Human Phenotype Ontology (HPO)
>
> **phenotypeEffect** CATEGORICAL VALUE (ONTOLOGY TERM) Phenotypic effect classification. Value from Sequence types and features ontology (SO) variant phenotype (SO:0001769), e.g. "benign variant" (SO:0001770)
>
> **alleleOrigin** (LIST OF) CATEGORICAL VALUE(S) (ONTOLOGY TERM) Annotation(s) on allele origins in which the variant has been found in association to phenotype. 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:17024373"

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

diseaseId categorical value (disease code/ ontology term) 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" (ICD10CM:E73)

clinicalEffect categorical value (ontology term) Clinical effect classification. Value from Sequence types and features ontology (SO) variant phenotype (SO:0001769), e.g. "disease associated variant" (SO:0001771)

alleleOrigin (List of) categorical value(s) (ontology term) 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"

Interactor

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

relationType categorical value (ontology term) Type of relation with Subject. Value from Infectious disease Ontology(IDO), e.g. "host" (IDO:0000531), "commensal" (IDO:0000525), "infectious agent" (IDO:0000596)

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