 # Proposed beacon v2 schema changes discussed today and maybe a couple other suggestions

Note: whatever is in automatic/black font is kept the same as in current version, things in blue font means additions or changes. Things marked with gray boxes ( ) are fields currently in viral beacon (the mapping of those into the corresponding fields here is in https://github.com/clauw87/virusbeacon/blob/raw_ideas/virus_beacon_schema_v1_to_generic.md)

Variant basic

refAssemblyId
startPos
endPos
ref
alt
variantType Variant classification e.g SNV, indel, CNV, structural variant

Organism

taxonId categorical value (ontology ID). Taxon ID of species from where variants come from, for example, the SARSCoV 2 taxon id and not human's in the case of viral beacons)

Individual

individualId
datasetId
taxon_id categorical value (ontology ID) (reference taxon ID for this individual human, animal or plant)
sex
ethnicity
geographicOrigin
phenotypicFeatures list of
    phenotypeId categorical value (ontology ID) Phenotypic feature observed (not disease)
    level/severity categorical value (ontology ID) Level/severity when and as applicable to phenotype observed e.g "mild", "severe"
diseases list of

diseaseId
dateOfOnset alphanumeric value (ISO8601 duration format) Date of onset/diagnosis of disease
ageOfOnset
age alphanumeric value (ISO8601 duration format)
ageGroup categorical value (ontology ID)
stage categorical value (ontology ID)
outcome categorical value (ontology ID) Outcome of disease e.g fatal or non-fatal
level/severity categorical value (ontology ID) Level/severity when and as applicable to disease observed e.g “mild”, “severe”
familyHistory
treatments list of
treatmentId categorical value (ontology ID) eg. chemotherapy
dateAtOnset alphanumeric value (ISO8601 duration format)
ageOfOnset
age alphanumeric value (ISO8601 duration format)
dose numerical value
units categorical value (ontology ID)
schedule free text for now eg. “/week”
duration alphanumeric value (ISO8601 duration format)
interventions list of
InterventionId categorical value (ontology ID) eg. Vasectomy
date alphanumeric value (ISO8601 duration format)
ageAtIntervention alphanumeric value (ISO8601 duration format)
age alphanumeric value (ISO8601 duration format)

pedigrees list of
pedigreeID
disease disease format
pedigreeRole
numberOfIndividualsTested

info

Biosample

biosampleId
individualId
description
biosampleStatus
collectionDate alphanumeric value (ISO8601 duration format). Date at which sample

is collected.
IndividualAgeAtCollection
sampleOriginType categorical value (ontology ID) Category of sample origin e.g “organism primary tissue”, “organism xenograft”, “organism-derived fluid”, “cell culture”, “environmental sample”
sampleOriginDetail categorical value (ontology ID) Specific instance of sample origin matching the category set in sampleOriginType e.g “HEK293T”, “nasopharyngeal swab”
obtentionProcedure categorical value (ontology ID)
cancerFeatures list of
    tumorProgression
    tumorGrade
info

Variant Annotation

variantId
genomicHGVSId
transcriptHGVSId alphanumeric ID (HGVSId descriptor at transcript level)
proteinHGVSId
genomicRegion list of
    class categorical value(s) (ontology ID) Class of genomic regions altered by the variant eg “protein coding”, “intergenic”, “untranslated region”, “transcript”
    featureID categorical value(s) IDs matching class (of genes, genomic regions, subgenomic regions, transcripts, other RNA species and proteins that are affected by the variant names or genomic region ref seq accessions (NC, NM, YP))
annotationToolVersion alphanumeric value. Tool used for annotation and prediction of variant effects e.g “SnpEffVersion=4.3t (build 2017-11-24 1018)”
molecularEffect categorical value (ontology ID) Predicted effect at nucleotide level eg “STOP_GAINED” as opposed to the description at protein level for protein affecting variants eg. “Nonsense” that goes into molecularConsequence
molecularConsequence
aminoacidChange string. Change at aminoacid level for for protein affecting variants eg. “V304*”
phenotypicEffect categorical value (ontology ID) Annotated effect on any phenotypic feature other than a disease
    phenotypeId Phenotype associated
    phenotypeEffect categorical value (ontology ID). Phenotypic effect classification
    references list of PMIDs
clinicalRelevance list of
    disieaseId
    clinicalEffect previously variantClassification
    references

alleleOrigin list of
info

Run
runId alphanumeric ID (external accession) e.g ”SRR10903401”
librarySource categorical value e.g “Metagenomic”, “Viral RNA”
libraryStrategy categorical value e.g “WGS”
librarySelection categorical value e.g “RANDOM”, “RT-PCR”
libraryLayout categorical value e.g “PAIRED” “SINGLE”
platform categorical value Sequencing platform group e.g “Illumina”, “Nanopore”
platformModel categorical value Sequencing platform model e.g “Illumina MiSeq” , ”GridION"
info (or handover maybe)
    experiment_info
        experimentId alphanumeric ID External experiment accession e.g "SRX7571571"
    study_info
        studyId alphanumeric ID External study reference/accession e.g "SRP242226"
        studyTitle string e.g ”Total RNA sequencing of BALF (human reads removed)”

        studyRef list of PMIDs

Variant in Sample
variantId alphanumeric ID
runId alphanumeric ID
variantCaller categorical value e.g GATK vxx
biosampleId alphanumeric ID
individualId categorical value (ontology ID)
variantFrequency numeric value
zygosity
alleleOrigin
clinicalRelevance list of
    disieaseId categorical value (ontology ID)
    clinicalEffect categorical value (ontology ID)
info

Encounter

encounterID alphanumeric ID

encounterDate alphanumeric value (ISO8601 duration format) Date of encounter/medical visit

ageAtEncounter

- age alphanumeric value (ISO8601 duration format)
  - ageGroup categorical value (ontology ID)

clinicalFindings Non quantifiable or not quantified clinical findings

- finding categorical value (ontology ID) eg: “arrhythmia”
- level/severity categorical value (ontology ID) e.g “mild”

measurements (list of) measurements taken during encounter

- id categorical value (ontology ID)
- value numerical value
- units categorical value (ontology ID)

info