

New concept proposal

Isolate

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|-----------------|---------------------------------------|-------------------|------------|
| Project | General interest | Contact | DCC |
| Dataset release | 2024.1 | Consulted expert | - |

1 Rationale

Pathogens can be isolated from samples resulting in a so-called 'isolate'. Therefore, they can be considered a subclass of *Sample*. To capture information about specific isolates and their characterization, we propose this *Isolate* concept.

2 Comparison to other standards/data models

2.1 SNOMED CT

SNOMED CT defines a 'Microbial isolate specimen (specimen)' (SNOMED:119303007) which is equivalent to the proposed *Isolate* concept. It also defines a 'Microbial isolate (substance)' (SNOMED:386126001), however, the first SNOMED CT concept is more in line with the SPHN design, since it has a notion of specimen taking (similar to the use of *Sample*), instead of being the substance itself.

2.2 FAIR Genomes

FAIR Genomes defines an 'Isolate' (fg:Material_Biospecimen_type_Isolate) which is aligned with/imported from NCIT 'Isolate' (NCIT:C53471). NCIT defines the term as "A specific individual microbe and its genetically identical progeny separated on a single occasion from a sample taken from a host or culture". It is a type of 'Biospecimen', which is itself a type of 'Specimen'. CDISC is the contributing source of this NCIT term.

2.3 OBI

OBI includes the 'selectively maintained organism' class (OBI:0001185), with synonym 'strain', that is defined as "An organism that is bred to have some uniform behavioural, morphological, physiological, or genetic characteristics with similarly bred organisms", which is not compatible with our intended use case since it implies that the isolation is done not just to examine but to retrieve an organism to be used for its characteristics.









2.4 EFO

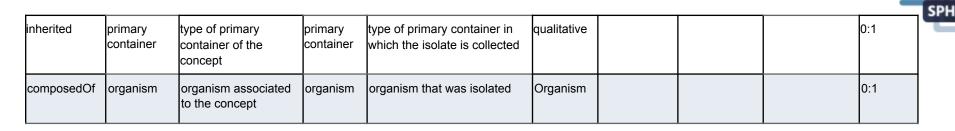
EFO has the 'genetically isolated population' with the synonym 'population isolate'. However, this class is defined as "a population with increased genetic homogeneity and reduced genetic variation due to cultural or geographic isolation", which doesn't fit our case.



3 Isolate concept

| | concept | | Contextua lized concept name | Contextualized description | Туре | Standard | Value set or subset | Meaning binding | Cardinalit y for compose dOf |
|-----------|---------|--|---------------------------------------|--|-------------|-----------|--|---|---------------------------------------|
| concept | | a specific individual microbe and its clone separated on a single occasion from a sample taken from a host or culture system | | a specific individual microbe and its clone separated on a single occasion from a sample taken from a host or culture system | Sample | | | SNOMED:11930 3007 Microbial isolate specimen (specimen) | |
| inherited | | unique identifier identifying the concept | | unique identifier identifying the isolate | string | | | | 0:n |
| | code | coded information specifying the material type of the concept | type code | material (structure, substance, device) removed from a source (patient, donor, physical location, product) | Code | SNOMED CT | descendant of: 123038009 Specimen (specimen) | | 0:n |
| inherited | , | anatomical site or structure associated to the concept | | anatomical site or structure associated to the concept | Body Site | | | | 0:1 |
| inherited | | datetime of collection of the concept | collection datetime | datetime of collection of the isolate | temporal | | | | 1:1 |
| inherited | 1 ,. | fixation or stabilization type | fixation type | fixation or stabilization type | qualitative | | | | 0:1 |

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Swiss Personalized Health Network

| - | - | | | Cardinality for concept to Source System |
|---------|-----|-----|-----|--|
| Isolate | 0:1 | 1:1 | 1:1 | 1:1 |

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4 Impact on the SPHN Dataset

One or more pathogens can be isolated from a single sample.

5 Discussion

In contrast to Sample, an Isolate is always defined by a species and strain of the pathogen/microbe that is isolated and not from the host. Body site, however, is irrelevant for isolates. As the cardinality for body site of the Sample concept is 0:1, it was discussed and decided to accept this inconsistency and still make Isolate a subtype of Sample. Although the phenotypic characterization in bacteriology is usually done at the moment the isolate is collected, this might not always be the case in all circumstances and domains, for instance in virology.

6 Example

Example of an isolate with organism (species, strain), material type code, and collection datetime mentioned

identifier: ExampleIsolate01

organism: code:

name: Oscillibacter ruminantium GH1

coding system and version: NCBI Taxonomy - 2023-07-28

identifier: 1007096

material type code: 429951000124103 |Bacterial isolate specimen|

collection datetime: 2023-07-26



