

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.

A project of	 <p>Schweizerische Akademie der Medizinischen Wissenschaften Académie Suisse des Sciences Médicales Accademia Svizzera delle Scienze Mediche Swiss Academy of Medical Sciences</p>	 <p>Swiss Institute of Bioinformatics</p>	<p>SIB Swiss Institute of Bioinformatics PHI Personalized Health Informatics Group www.sphn.ch dcc@sib.swiss</p>
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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 or concept compositions or inherited	General concept name	General description	Contextualized concept name	Contextualized description	Type	Standard	Value set or subset	Meaning binding	Cardinality for composition
concept	Isolate	a specific individual microbe and its clone separated on a single occasion from a sample taken from a host or culture system	Isolate	a specific individual microbe and its clone separated on a single occasion from a sample taken from a host or culture system	Sample			SNOMED:119303007 [Microbial isolate specimen (specimen)]	
inherited	identifier	unique identifier identifying the concept	identifier	unique identifier identifying the isolate	string				0:n
inherited	material type code	coded information specifying the material type of the concept	material 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	body site	anatomical site or structure associated to the concept	body site	anatomical site or structure associated to the concept	Body Site				0:1
inherited	collection datetime	datetime of collection of the concept	collection datetime	datetime of collection of the isolate	temporal				1:1
inherited	fixation type	fixation or stabilization type	fixation type	fixation or stabilization type	qualitative				0:1

inherited	primary container	type of primary container of the concept	primary container	type of primary container in which the isolate is collected	qualitative				0:1
composedOf	organism	organism associated to the concept	organism	organism that was isolated	Organism				0:1

General concept name	Cardinality for concept to Administrative Case	Cardinality for concept to Data Provider	Cardinality for concept to Subject Pseudo Identifier	Cardinality for concept to Source System
Isolate	0:1	1:1	1:1	1:1

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**