

New concept proposal

Assay

Author	Eelke van der Horst, Femke Kopmels	Date last updated	31/10/2023
Project	General interest	Contact	DCC
Dataset release	2024.1	Consulted expert	-

1 Rationale

Assay metadata is essential when sharing (experimental) results: for providing context, ensuring data quality, enabling data integration, and facilitating collaboration and reproducibility in research and clinical settings. An Assay takes a sample and produces data about that sample. For different types of omics research, different types of assays will be relevant, each with their own defining attributes. Therefore, a common Assay concept is proposed, that can be used as-is, or inherited by more specific types of Assay.

2 Comparison to other standards/data models

2.1 OBI / EFO

OBI has the 'assay' class, which is "a planned process that has the objective to produce information about a material entity (the evaluant) by examining it" (OBI:0000070). This class is equal to the proposed Assay concept. EFO imports OBI's 'assay' class and many of its subclasses (though not all), and defines additional assays. Assay types from both terminologies can be used to type the assay concept (using code). EFO 'Assay' cross references NCIT 'Assay' (NCIT:C60819) defined as "A qualitative or quantitative analysis performed to determine the amount of a particular constituent in a sample or the biological or pharmacological properties of a drug".

2.2 EDAM

EDAM includes the procedures used to conduct an experiment as 'Laboratory Techniques' (EDAM: topic_3361). This concept is defined as a 'Topic' rather than an 'Operation', therefore it is considered not relevant for our Assay proposal.

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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3 Concept Information

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 composed Of
concept	Assay	a process with the objective to produce information about a sample by examining it	Assay	a process with the objective to produce information about a sample by examining it				OBI:0000070 [assay]	
composedOf	standard operating procedure	standard operating procedure associated to the concept	standard operating procedure	the standard operating procedure that was followed for this assay	Standard Operating Procedure				0:1
composedOf	predecessor	a preceding process associated to the concept	predecessor	sample processing preceding this assay	Sample Processing			RO:0002087 [immediately preceded by]	0:n
composedOf	code	coded information specifying the concept	code	code specifying the type of assay		EFO, OBI or other	for OBI: descendant of: OBI:0000070 [assay]		1:1
composedOf	identifier	unique identifier identifying the concept	identifier	unique identifier identifying the assay	string				0:1

composedOf	start datetime	datetime at which the concept started	start datetime	datetime at which the assay was first executed	temporal				0:1
composedOf	data file	data file associated to the concept	data file	data file associated to the assay	Data File				0:n
composedOf	sample	any material sample for testing, diagnostic, propagation, treatment or research purposes associated to the concept	sample	material that is being examined by this assay	Sample				0:n

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
Assay	0:n	1:1	0:n	1:n

4 Impact on the SPHN Dataset

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5 Discussion

As a value set we propose “EFO, descendants of OBI:0000070, or other” to facilitate in foreseen assay types. EFO imports many classes from OBI (such as ‘assay’ and subclasses of assay) but also defines their own. Since EFO is an application ontology for EBI data repositories and OpenTargets, there are also terms missing that are present in OBI but have no purpose for these repositories. For instance, ‘Imaging Mass Cytometry’ (ICM) is available in OBI but not in EFO, while EFO holds a concept for “clone by clone sequencing” that is missing in OBI.

It was discussed to call the proposed concept “(Data) generator”. Although assays such as sequencing are data generators, it can also include processes that do not evaluate a sample, such as a machine or data transformation. Additionally, the Assay is more aligned with the existing design of the SPHN model as it provides more specificity with the concept name.

It could be possible that an Assay has more than one Sample as input. This would be common where multiple samples are multiplexed as part of the same sequencing assay. In the case of sequencing assay, the samples can be mapped back via the resulting demultiplexed data file. However, this will not always be the case for all types of assays. Therefore, it was decided to maintain a 0:n cardinality for this more general Assay concept, but subclasses of Assay can be more strict.

When multiple runs are executed for the same Assay, the onset datetime for this concept will be equal to the run datetime of the Run that was first executed.

6 Example

Imaging Mass Cytometry (ICM) assay. The output format is .fcs, which is a binary file format with three main segments. This file format is not a descendant of EDAM:format_1915 and therefore assigned the more general format_2333 code for “binary format”.

```
code: OBI:0003096 |imaging mass cytometry assay|
identifier: example_assay123
start datetime: 2023-06-29
sample:
  identifier: sample_123
  collection datetime: 2023-06-28
  material type code: 127470000 |Tissue specimen from pancreas|
data file:
  format code: EDAM:format_2333 |Binary format|
```