New concept proposal

Sequencing Instrument

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

The instrument that was used to conduct a sequencing assay is essential information to understand and evaluate the experimental context and data generation process. Sequencing data may be generated using a range of instruments. Different instruments may vary in sensitivity, accuracy, or precision, and recording the instrument used allows researchers to assess data quality and identify potential sources of variability. Knowing the instrument that was used to produce a dataset enables researchers to assess compatibility and potential biases when performing cross-platform comparisons.

2 Comparison to other standards/data models

2.1 FAIR Genomes

FAIR Genomes stores information about an instrument that was used for sequencing in the 'SequencingInstrumentModel' and 'SequencingMethod' elements of the Sequencing module. It also defines lookup values for these properties. We propose to compose instrument properties into their own concept, thereby avoiding duplication and allowing for instrument identity that can be referenced, reused, and provided with additional information.

2.2 OBI and EFO

OBI defines a 'Device' class (OBI:0000968), which has 'instrument' as synonym, that is defined as "a material entity that is designed to perform a function in a scientific investigation, but is not a reagent. A 'measurement device' (OBI:0000832) is "a device in which a measure function inheres". DNA sequencers are a type of 'measurement device'. EFO uses 'instrument' to indicate "devices which provide a mechanical or electronic function" (EFO:0000548). The range of subclasses is quite broad, from sequencers to braces. For a few subclasses, details about instruments are defined in the name of the class (e.g. Illumina Genome Analyzer).

2.3 NCIT

NCIT has defined ‘Instrumentation’ as “any object, or item of electrical or electronic equipment, which is designed to carry out a specific function or set of functions” (NCIT:C16742). Mass spectrometers are also a type of ‘Instrumentation’, but sequencers are not defined.

2.4 ENA

ENA has two Experiment properties for instrument information: ‘Sequencing Platform’ and ‘Sequencing Instrument’. ‘Sequencing Platform’ holds only the name of the platform, e.g. ‘Illumina’, or ‘Oxford Nanopore’ while ‘Sequencing Instrument’ also holds the model of the instrument that was used, e.g. ‘Illumina MiniSeq’, or ‘Illumina NovaSeq 6000’.

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 composedOf
concept	Sequencing Instrument	A sequencing instrument that is used in a sequencing assay	Sequencing Instrument	A sequencing instrument that is used in a sequencing assay				EFO:0000548 |instrument|	
composedOf	code	coded information specifying the concept	instrument type	code specifying the type of sequencing instrument	Code	OBI, EFO or other	for OBI: descendant of: OBI:0400103 |DNA sequencer|; for EFO: descendant of: EFO:0003739 |sequencer|		1:1

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
Sequencing Instrument	-	1:1	-	-

4 Impact on the SPHN Dataset

Currently, the SPHN model includes a *Medical Device* concept, with *Lab Analyzer* as a child concept. However, *Medical Device* is not always applicable in the omics use cases. The existing SPHN concept for *Medical Device* could fall under the same parent as *Sequencing Instrument* by introducing an *Instrument* concept. Note that a sequencing instrument, like other instruments, could be medical devices or not, depending on the context.

5 Discussion

Providing the instrument type will be required for the *Sequencing Instrument* concept.

6 Example

Example of Illumina NovaSeq 6000 sequencer.

code: OBI:0002630 |Illumina NovaSeq 6000|