New concept proposal

Reference Sequence, Reference Value

Author	**Vasundra Touré**	**Date last updated**	**07.11.2023**
Project	General importance	**Contact person**	DCC
Dataset release	2024.1	**Consulted expert**	-

1 Rationale

Semantically, a reference can have different meanings and can be used in different contexts (bibliographic reference, reference range, reference value, an acknowledgement, reference genome, etc.).
In 2023.2 SPHN Dataset, the concepts related to references were insufficiently delineated.

In the 2024.1 release of the SPHN Dataset, we attempt to fix the meaning and detangle the representations of the different kinds of references that are needed.

Change Request:

- The 2023.2 meaning of Reference concept actually refers to a reference sequence used for comparison to the studied sequence and therefore the concept is renamed in 2024.1 to “Reference Sequence”.

New concept:

- Reference Value is added as a new concept for enabling the representation of numerical references that have only one threshold or data point used for comparison and interpretation of measurements, test results, etc. This differs with the Reference Range in the way that the reference range defines a lower and upper boundary for doing the comparison.

2 Comparison to other standards/data models

3 Concept information

3.1 Reference Value (new 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 composed Of
concept	Reference Value	numeric data point serving as reference for comparison and interpretation	Reference Value	numeric data point serving as reference for comparison and interpretation					
composedOf	quantity	an amount or a number of the concept	value	the actual threshold of the reference value	Quantity				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
Reference Value				

3.2 Reference Sequence (change request)

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	Reference Sequence	reference construct used for comparison purposes	Reference Sequence	reference construct used for comparison purposes				GENO:0000017 |reference sequence|	
composedOf	code	coded information specifying the concept	reference sequence identifier	identifier of a reference sequence deposited in a repository	Code	NCBI GenBank or other			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
Reference Sequence	-	-	-	-

4 Impact on the SPHN Dataset

The meaning of Reference is changed to Reference Sequence.

5 Discussion

The SPHN Dataset 2024.1 is now composed of three different concepts for representing reference elements:

- a Reference Range for intervals used for interpreting of results
- a Reference Value for single value thresholds used for comparison and interpretation of results
- a Reference Sequence for omics-related reference constructs used for comparison purposes.

The Reference Range and Reference Value are used in the 2024.1 updated Lab Result concept to inform about numerical reference information that applies to Lab Tests and their specific results. They are also used in the Reference Interpretation to infer some findings about a Result.

6 Example

Example of a reference value for a CRP test

Reference Value
    quantity:
        value: 1
        unit: mg/dL