New concept proposal

Organism

Author	**Jan Armida**	**Date last updated**	**10/05/2022**
Project	Genomic Concepts	**Contact person**	Jan Armida
Status	Accepted	**Consulted expert**	-

1 Rationale

The concept of *Organism* serves as general reference for the specification of the organism of origin of a given concept element. When combined with other concepts such as *Gene* or *Protein,* it allows retrieval of the information about the species for which nucleic acid or protein sequence have been determined via its taxon identifier.

2 Comparison to other standards/data models

2.1 SNOMED CT

SNOMED CT provides a specific code for “Organism”, 410607006 | Organism (organism) |. The descendants of Organism encompass all domains including: archaea, bacteria, eukarya, prions and viruses.

2.2 GA4GH VRS

In GA4GH VRS, it is possible to specify the organism associated with a certain chromosome location within the element *ChromosomeLocation.* The attribute species_id makes use of the NCBI species taxonomy identifiers.

Field	Type	Limits	Description
_id	CURIE	0..1	Location Id. MUST be unique within document.
type	string	1..1	MUST be "ChromosomeLocation"
species_id	CURIE	1..1	CURIE representing a species from the NCBI species taxonomy. Default: "taxonomy:9606" (human)
chr	string	1..1	The symbolic chromosome name. For humans, For humans, chromosome names MUST be one of 1..22, X, Y (case-sensitive)
interval	CytobandInterval	1..1	The chromosome region defined by a CytobandInterval

3 Concept information

Contextualized concept name	Contextualized description	Type	Standard	Value set	Meaning binding
Organism	living system capable of replicating or reproducing, growth and maintenance. An organism may be unicellular or multicellular				SNOMED CT: 410607006 | Organism (organism) |
organism identifier	unique organism id representing a species, e.g. taxon ID or SNOMED CT code for organism	Code	SNOMED CT, NCBI Taxon	for SNOMED CT: descendant of : 410607006 | Organism (organism) |	

4 Impact on the SPHN Dataset

The addition of *Organism* does not require any further change in the current SPHN Dataset release.

5 Discussion

The possibility to refer to an organism as a separate instantiation and not only when associated with another concept increases the flexibility and reusability of the concept in different contexts.