New concept proposal

Chromosome

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Status	Accepted	Consulted expert	SVIP-O, SwissGenVar

1   Rationale

Chromosomes are an organized package of DNA generally found in the nucleus of a cell. The number of chromosomes vary across organisms. In humans, deviations from the normal chromosomal pattern (22 pairs of autosomes, and 1 pair of sex chromosomes) is likely to cause developmental abnormalities and is therefore associated with a number of chromosomal disorders. Furthermore, the ability to refer to a specific chromosomal region is required to correctly annotate causative and non-causative variants. It is therefore important to provide a construct that allowing to refer to a specific chromosome. The *Chromosome* concept is currently targeted towards humans and enables the possibility to refer to a particular chromosome using its name and, in combination with other concepts, to pinpoint specific DNA regions.

2   Comparison to other standards/data models

2.1   SNOMED CT

In SNOMED CT it is possible to refer to a Chromosome directly using the code 91272006 | Chromosome (cell structure). This term has several children that allow to describe single chromosome pairs or groups.

2.2   LOINC

In LOINC the concept of Chromosome is described by the code 48000-4 Chromosome [Identifier] in Blood or Tissue by Molecular genetics method. An answer list is associated to this code and allow do refer to a specific human chromosome using a common Chromosome + number representation:

LL2938-0 List of human chromosomes:

OID   1.3.6.1.4.1.12009.10.1.1880
Name   List of human chromosomes
Description   1-22, X & Y
LOINCs Using This List

A project of

SAMW ASSM
Schweizerische Akademie der Medizinischen Wissenschaften
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Swiss Institute of
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2.3   HUGO Gene Nomenclature Committee

In the HUGO Gene Nomenclature committee (HGNC) the field "Chromosomal location" refers to a specific position within a chromosome using cytogenetic coordinates.

2.4   HPO

In the Human Phenotype Ontology (HPO) there is no code to refer to a precise chromosome.

2.5   GA4GH Phenopackets

In the GA4GH Phenopackets there is no specific building block to represent a Chromosome. Instead, a reference to chromosomal region (cytoband) and chromosome name is used within the *Chromosomal Location* object of VRS.

3   Concept information

Contextualized concept name	Contextualized description	Type	Standard	Value set	Meaning binding
Chromosome	thread-like structure composed of tightly coiled DNA and histones or circular DNA (mitochondrial DNA). These structures act as carriers of genetic information				SNOMED CT: 91272006	Chromosome (cell structure)	; LOINC: 48000-4 Chromosome
chromosome name	symbolic name of the chromosome	Code	SNOMED CT	Descendant of: 91272006	Chromosome (cell structure)		

4   Impact on the SPHN Dataset

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

5 Discussion

The concept of *Chromosome* has been developed as an instrument to easily refer to a specific human chromosome by using a standardized terminology (in this case SNOMED CT). Being the concept very simple it is particularly easy to extend such a concept adding project-specific attributes if needed. This concept can be further expanded to include other organisms.