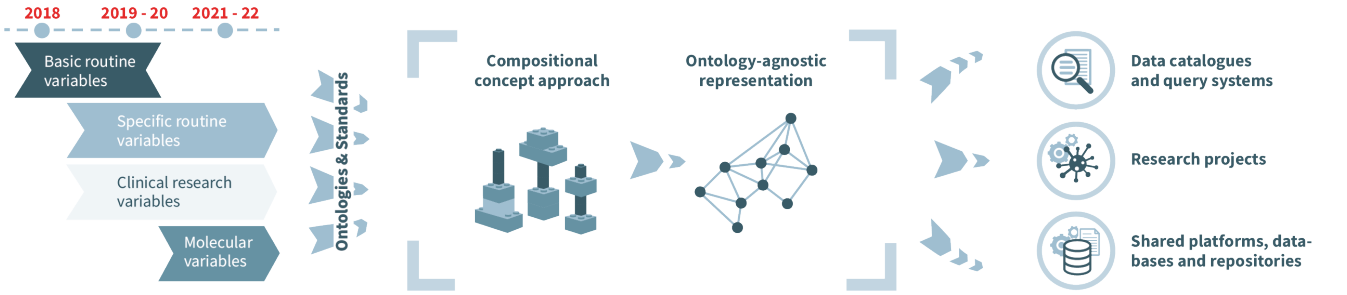
2022 Fact sheet
The SPHN Semantic Interoperability Framework

1. Summary

Health-related data, ranging from hospital routine data to biomedical research data, are quite diverse, stored in different databases and formats, and often coded in local standards. This fragmentation and diversity make it very time-consuming to combine data from different sources for doing research on a particular topic. It is generally difficult to understand data and the intended meaning due to the lack of common standards, metadata, or a common data dictionary. Following the FAIR (findable, accessible, interoperable and reusable) principles, SPHN builds an infrastructure to overcome these hurdles and enables collaborative research by making the meaning of health-related data understandable to humans and machines. This allows for an easy combination of the data from different sources, thus simplifying the use and exploration of data across Switzerland. Experience shows that the diverse needs and requirements of the different use cases make it impossible to agree on one single data model. Not only the required data elements differ between projects, but also the level of information granularity required for a single datapoint can vary significantly. Therefore, SPHN developed a framework based on a strong semantic layer of information (pillar 1), and graph technologies for the exchange layer, which can be extended by the individual projects to fit their purposes (pillar 2). Thus, a universal exchange language for healthcare is established, using the "words" from various international standard vocabularies (such as SNOMED CT or LOINC), a simple "grammar" (subject-predicate-object; expressed in RDF), and additional SPHN guidelines and rules to establish good practices for FAIR data.



Project needs

Variables and associated information needs are collected from the SPHN Projects. In a close dialogue with all stakeholders a global understanding for a data-driven representation is developed. Individual research needs are aligned across projects and translated into a common semantic representation.

01

Semantic representation

Strong semantic definitions are conceptualized in building blocks, so-called "concepts". Concept definitions take into account existing medical ontologies and standards. In a modular way, concepts can be reused and combined into more comprehensive concepts.

02

Data transport and storage

A flexible data exchange format, able to carry the meaning of data, is recommended for data transport and storage.

This format is capable to support various data-driven research scenarios in personalized health.

03

Use cases

Diverse use cases and applications in healthcare, biomedical research and data science can be served with a flexible, multi-dimensional semantic data architecture, which makes it easy to transform data into desired, use case specific data models.

A project of SAMW ASSM
Schweizerische Akademie der Medizinischen Wissenschaften
Académie Suisse des Sciences Médicales
Accademia Svizzera delle Scienze Mediche
Swiss Academy of Medical Sciences

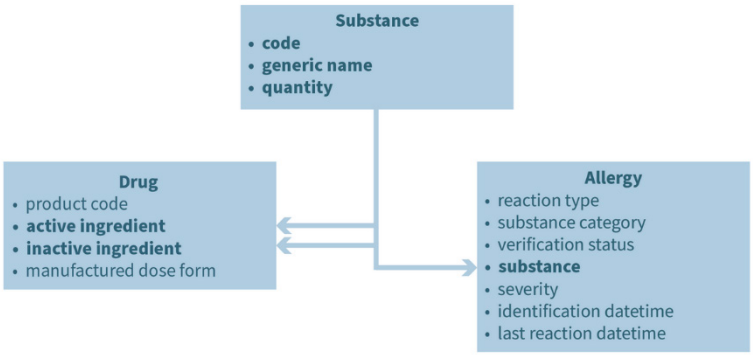
SIB Swiss Institute of Bioinformatics

SIB | Swiss Institute of Bioinformatics
PHI | Personalized Health Informatics Group
www.sphn.ch | dcc@sib.swiss

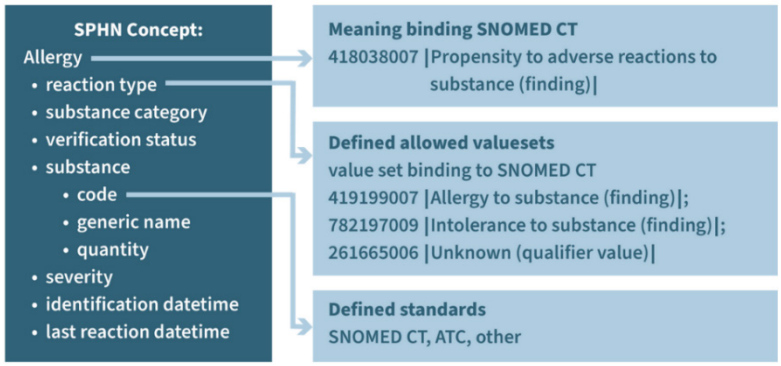
2. How it is implemented

Semantic representation (Pillar 1)

SPHN concepts are generalizable building blocks, which can be used in different contexts. Each concept contains all information necessary to understand it, and concepts can be combined to composed concepts, which again can be combined to more complex compositions. It is important to find the right level between abstraction and granularity to optimize the power of expression. The approach can be illustrated with the example of "substance". A substance can be an active or inactive ingredient of a drug or it can be the substance someone is allergic to. Therefore, we can abstract "substance" as a concept on its own. The concept of substance is composed of three concepts: "code", "generic name" and "quantity". These concepts describe a substance no matter if it is the active ingredient of a drug (that is defined as the type "substance") or the substance to which someone is allergic to.



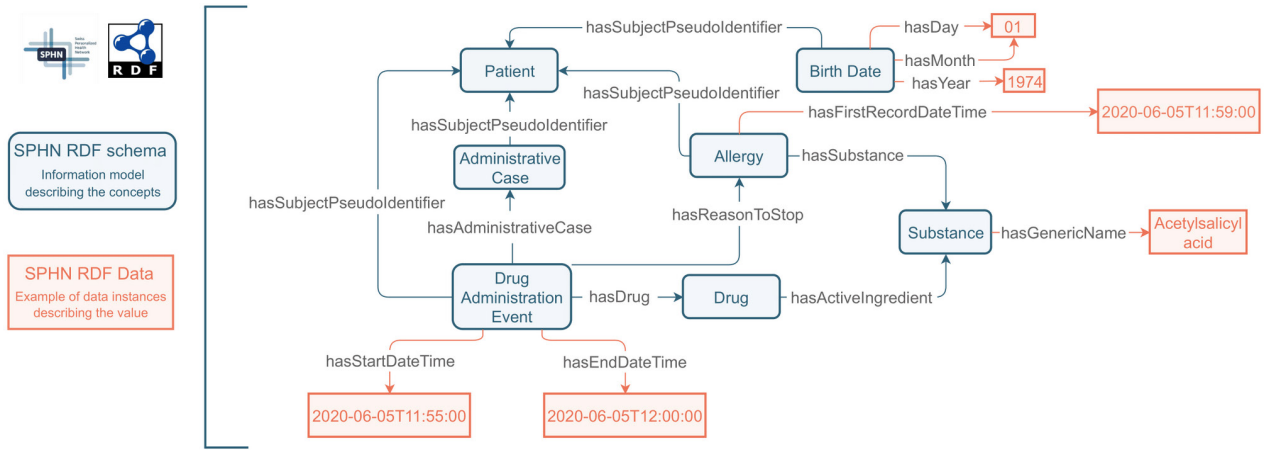
To make the SPHN concepts comparable nationally and internationally, we express their meaning using existing semantic standards (controlled vocabularies), by creating a meaning binding wherever possible to SNOMED CT and/or LOINC. The data element of a concept can be expressed using one or several recommended standards (e.g. LOINC, SNOMED CT, ICD-10, ICD-O-3, CHOP, ATC). For example, the instance of substance code under the concept "allergy" can be an ATC, a SNOMED CT, or a code form another semantic standard. If needed value sets are defined and if possible, a value set binding to SNOMED CT is added. Descriptions for concepts and value sets as well as standards are, whenever possible, aligned with national and international sources.



Data transport and storage (Pillar 2)

SPHN concepts (blue) and their instances (orange) can easily be mapped from/to other data representations or merged with other RDF data sets without losing their semantics. The relations between different concepts and the data are expressed in the form of triples composed of a "subject", a "predicate", and an "object". The example shows a patient born on the 1st of January 1974 which has an Administrative Case registered with a Drug Administration Event. During this administration, a Drug was given to the patient on the 05.06.2020 at 11:55.

The Substance administered was “Acetylsalicyl acid”. The Drug Administration Event has stopped five minutes later, at 12:00 for the reason that an Allergy was observed at 11:59 to that Substance. RDF is quite flexible and enable us to make use of different existing semantic standards, and value sets as defined in the SPHN Dataset.



In RDF, subjects, predicates and (sometimes) objects have a Unique Resource Identifier (URI) that enables the unique identification of these elements. In our example, the concept Allergy has the URI https://bio-medit.ch/rdf/sphn-ontology/sphn/Allergy, which uniquely and unambiguously identifies it in the context of SPHN. The SNOMED CT meaning binding to the equivalent meaning of ‘Allergy’ is introduced by linking the URI of the SPHN Allergy class to the corresponding URI in SNOMED CT (http://snomed.info/id/418038007) in the SPHN RDF schema.

Use cases (Pillar 3)

Based on these needs, researchers can:

1. use the RDF files directly as input into their analysis software, e.g. RDFLib in Python,
2. extract data into a flat file, e.g. Excel or CSV,
3. load the data into other existing data models with adequate converters, e.g. i2b2, OMOP or a data management software.

3. Selected SPHN semantic tool services

– **The SPHN Schema Visualization Tool** generates a human-readable HTML document describing the project's RDF schema.
– **DCC Terminology Service** provides SPHN compatible, machine-readable versions of national (CHOP or ICD-10 GM) and international (SNOMED CT, LOINC, ATC, UCUM) terminologies and classifications in RDF
– **The SHACLer** is a Python tool that extracts SHACL rules from an SPHN-compliant input ontology for facilitating data validation.

– **SPHN Quality Check Tool** facilitates the validation process of SPHN RDF data at the data provider level. Based on the SHACL file generated by the SHACLer and statistical queries in SPARQL, it generates a human-friendly report with information about data conformance to the schema and some basic statistics.

4. How does this strategy help to address the following FAIR criteria:

Findable

F1. (Meta)data are assigned a globally unique and persistent identifier
→ SPHN classes and properties are defined with URIs in the following namespace https://biomedit.ch/rdf/sphn-ontology/sphn#
→ data use the following namespace https://biomedit.ch/rdf/sphn-resource/ and must be identified with a unique identifier for distinct data points

F2. Data are described with rich metadata
→ each SPHN concept contains a set of information to describe the data element
→ administrative metadata (e.g., date of extraction, data provider) is included directly in the RDF data files

F3. Metadata clearly and explicitly includes the identifier of the data they describe
→ data and metadata are linked via RDF properties

F4. (Meta)data are registered or indexed in a searchable resource
→ the SPHN RDF schema is openly available and searchable on the web (biomedit.ch)

Accessible

A1. (Meta)data are retrievable by their identifier using a standardized communications protocol
→ the SPHN RDF schema URIs are resolvable on the web (accessible via https://)
→ SPARQL is the official RDF Query Language which is a standard protocol of the World Wide Web Consortium (W3C). It facilitates the exploration of data in RDF

Interoperable

I1. (Meta)data use a formal, accessible, shared, and broadly applicable language for knowledge representation
→ RDF is the standard language used for knowledge representation

I2. (Meta)data use vocabularies that follow FAIR principles
→ SNOMED CT, LOINC and other terminologies are used as controlled vocabulary in the RDF schema and the data

I3. (Meta)data include qualified references to other (meta)data
→ when possible, the SPHN URIs of concepts and instances are linked to the URIs of the external resources like SNOMED CT, LOINC, ATC, ICD-10

Reusable

R1. (Meta)data are richly described with a plurality of accurate and relevant attributes
→ the SPHN RDF schema is published under the CC BY-NC-SA 4.0 license

5. Further information

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