New concept proposal

Diagnosis

Author	Kristin Gnodtke	Date last updated	18.05.2022
Project	ALL	Contact person	-
Dataset release	2022.1	Consulted expert	-

1 Rationale

In the SPHN Dataset, there are several concepts related to diagnosis such as *FOPH Diagnosis, ICD-O Diagnosis* and *Nursing Diagnosis*. However, there are even more concepts for types of diagnoses in discussion, e.g. first cancer diagnosis or diagnosis at admission to the Intensive Care Unit (ICU). All these concepts represent a diagnosis and have common features that can be covered on a more general level. A new *Diagnosis* concept would support queries for all types of diagnoses of a patient. The concept *FOPH diagnosis* was included in the SPHN Dataset to represent the "billing" diagnosis, which is encoded in ICD-10 for inpatients in Swiss hospitals. A more general diagnosis concept would represent all kinds of different diagnoses, encoded (also) in other standards such as SNOMED CT or ORPHA. Such semantic standards are in some cases the more appropriate one compared to ICD-10. For example, rare diseases are underrepresented in ICD-10 (only approx. 8% of rare diseases have an ICD-10 code). In addition, the FOPH Diagnosis is only available for inpatients and a broader concept *Diagnosis* would cover diagnoses for all patients – inpatients and outpatients.

2 Comparison to other standards/data models

2.1 HL7 FHIR

In HL7 FHIR a diagnosis is represented in the resource *Condition*. With this resource, general information about the diagnosis can be represented such as onset (can be onset datetime, age), date the diagnosis was first recorded, etc. Further, there is a proposal to capture the "purpose" of the diagnosis with the *DiagnosisRole* (Maturity Level:0 on 09.09.2021).

Code	Display	Definition
AD	Admission diagnosis	
DD	Discharge diagnosis	
CC	Chief complaint	
CM	Comorbidity diagnosis	
pre-op	pre-op diagnosis	
post-op	post-op diagnosis	
billing	Billing	

2.2 UMLS

In UMLS, there are 10 different definitions of diagnosis with the UMLS Concept Unique Identifier(CUI): C0011900 (status of 14.09.2021). The following two definitions are describing the nature of diagnosis in the most comprehensive way.

Definitions (10)

The determination of the nature of a disease or condition, or the distinguishing of one disease or condition from another. Assessment may be made through physical examination, laboratory tests, or the likes. Computerized programs may be used to enhance the decision-making process. **(MSH)**

The investigation, analysis and recognition of the presence and nature of disease, condition, or injury from expressed signs and symptoms; also, the scientific determination of any kind; the concise results of such an investigation. **(NCI)**

3   Concept information

Concept name	Description	Type	Standard	Meaning binding SNOMED CT	Meaning binding LOINC
Diagnosis	determination of the presence of a disease, condition, or injury from expressed signs and symptoms and assessments such as physical examination, laboratory test, or the like			439401001 |Diagnosis (observable entity)|	
code	code, name, coding system and version used to describe the diagnosis	Code	SNOMED CT, ICD, ICD-O-3 Topography, ICD-O-3 Morphology, NANDA, ORPHA or other		
record datetime	datetime the diagnosis was recorded	temporal			
coding datetime	datetime the diagnosis was coded	temporal			
subject age	age of the individual at the time of diagnosis	Age			

4   Impact

4.1   Impact on the SPHN Dataset

For the existing diagnoses concepts *FOPH Diagnosis, ICD-O Diagnosis* and *Nursing Diagnosis,* the type would change to *Diagnosis* (instead of empty), and the properties *code* and *datetime* would need to be adapted, i.e. change to inherited (instead of composedOf).

4.1.1   FOPH Diagnosis

Currently released version of concept *FOPH Diagnosis*

SPHN Dataset version: 2021.1

Unique concept ID: 0000000320

Concept or concept compositions or inherited	Concept name	Description	Type	Standard	Value set	Meaning binding SNOMED CT	Meaning binding LOINC
concept	**FOPH Diagnosis**	discharge diagnosis given respecting the rules of FOPH and used for building the DRGs, e.g. K35 acute appendicitis				89100005 |Final diagnosis (discharge) (contextual qualifier) (qualifier value)|	38999-9 Hospital discharge DRG [Interpretation]
composedOf	code	code, name, coding system and version of the FOPH diagnosis	Code	ICD-10			
composedOf	datetime	datetime at which the diagnosis was given	temporal				
composedOf	diagnosis rank	specifies the level of diagnosis	qualitative		principal; secondary; complementary	106229004 |Qualifier for type of diagnosis (qualifier value)|	

Proposed adaptation of concept *FOPH Diagnosis*

Concept or concept compositions or inherited	Concept name	Description	Type	Standard	Value set	Meaning binding SNOMED CT	Meaning binding LOINC
concept	**FOPH Diagnosis**	discharge diagnosis given respecting the rules of FOPH and used for building the DRGs, e.g. K35 acute appendicitis	Diagnosis			89100005 |Final diagnosis (discharge) (contextual qualifier) (qualifier value)|	38999-9 Hospital discharge DRG [Interpretation]
inherited	code	code, name, coding system and version of the FOPH diagnosis	Code	ICD-10			
inherited	record datetime	datetime the diagnosis was recorded	temporal				
inherited	coding datetime	datetime the code was assigned	temporal				
composedOf	diagnosis rank	specifies the level of diagnosis	qualitative		principal; secondary; complementary	106229004 |Qualifier for type of diagnosis (qualifier value)|	
inherited	subject age	age of the individual at the time of diagnosis	Age				

4.1.2 ICD-O Diagnosis

Currently released version of concept ICD-O Diagnosis

SPHN Dataset version: 2021.1

Unique concept ID: 0000000233

Concept or concept compositions or inherited	Concept name	Description	Type	Standard	Meaning binding SNOMED CT	Meaning binding LOINC
concept	**ICD-O Diagnosis**	ICD-O diagnosis		ICD-O-3		
composedOf	topography code	the topography code that describes the anatomical site of origin of the neoplasm	Code		116677004 |Associated topography (attribute)|	
composedOf	morphology code	the morphology code that describes the characteristics of the tumor itself, including its cell type and biological activity; the code is composed of four digits that indicate the cell type or histology and one digit that indicates the behaviour	Code		116676008 |Associated morphology (attribute)|	
composedOf	datetime	datetime at which the diagnosis was given	temporal			

Proposed adaptation of concept *ICD-O Diagnosis*

Concept or concept compositions or inherited	Concept name	Description	Type	Standard	Meaning binding SNOMED CT	Meaning binding LOINC
concept	**ICD-O Diagnosis**	ICD-O diagnosis	Diagnosis			
composedOf	topography code	the topography code that describes the anatomical site of origin of the neoplasm	Code	ICD-O-3 Topography	116677004 |Associated topography (attribute)|	
inherited	morphology code	the morphology code that describes the characteristics of the tumor itself, including its cell type and biological activity; the code is composed of four digits that indicate the cell type or histology and one digit that indicates the behaviour	Code	ICD-O-3 Morphology	116676008 |Associated morphology (attribute)|	
inherited	record datetime	datetime the diagnosis was recorded	temporal			
inherited	coding datetime	datetime the diagnosis was coded	temporal			
inherited	subject age	age of the individual at the time of diagnosis	Age			

4.1.3 Nursing Diagnosis

Currently released version of concept *Nursing Diagnosis*

SPHN Dataset version: 2021.1

Unique concept ID: 0000000318

Concept or concept compositions or inherited	Concept name	Description	Type	Standard	Meaning binding SNOMED CT	Meaning binding LOINC
concept	**Nursing Diagnosis**	clinical judgment concerning a human response to health conditions/life processes, or a vulnerability for that response, by an individual, family, group or community; a nursing diagnosis provides the basis for selection of nursing interventions to achieve outcomes for which the nurse has accountability			86644006 |Nursing diagnosis (finding)|	
composedOf	code	code, name, coding system and version used to describe the nursing diagnosis	Code	NANDA		
composedOf	datetime	datetime of code assignment	temporal			

Proposed adaptation of concept *Nursing Diagnosis*

Concept or concept compositions or inherited	Concept name	Description	Type	Standard	Meaning binding SNOMED CT	Meaning binding LOINC
concept	**Nursing Diagnosis**	clinical judgment concerning a human response to health conditions/life processes, or a vulnerability for that response, by an individual, family, group or community; a nursing diagnosis provides the basis for selection of nursing interventions to achieve outcomes for which the nurse has accountability	Diagnosis		86644006 |Nursing diagnosis (finding)|	
inherited	code	code, name, coding system and version used to describe the nursing diagnosis	Code	NANDA		
inherited	record datetime	datetime the diagnosis was recorded	temporal			
inherited	coding datetime	datetime the diagnosis was coded	temporal			
inherited	subject age	age of the individual at the time of diagnosis	Age			

5   Discussion

A general concept *Diagnosis* supports queries to retrieve all diagnoses for a patient and allows to see how different types of diagnoses for the same patient evolve during the patient's journey through the healthcare system. The two separate temporal statements (record datetime; coding datetime) allow for correct chronological interpretation of events, e.g., if a medication was prescribed after the doctor diagnosed the patient, but the diagnosis was coded in ICD-10 only after the drug prescription is dated.

6 Example

The following graphic shows the diagnosis of *Myositis ossificans progressiva* coded in different semantic standards.