New concept proposal
Excluded Disorder

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Dataset release	2024.1	**Consulted expert**	-

1   Rationale

The knowledge of the absence of certain disorders is relevant for diagnosis and prognosis purposes, and the determination of absence of these disorders can be undertaken. Not having any observation about a disorder in the dataset means that it was not observed, but may have still occurred as no action was performed to determine its absence. Excluded Disorder is an observation that clearly establishes the absence of the disorder, at least at the date of this observation.

2   Comparison to other standards/data models

2.1   SNOMED CT

Element 315215002 | Disorder excluded (situation) | represents the known absence of disorder. The list of descendants to specify the excluded disorder is too long to be represented here, but the following list contains 10 children of this concept:

- 473139002 | Acanthosis nigricans absent (situation) |
- 473143003 | Chronic disease absent (situation) |
- 315216001 | Diabetes mellitus excluded (situation) |
- 688232241000119100 | Disease caused by severe acute respiratory syndrome coronavirus 2 absent (situation) |
- 371622005 | Elevated blood pressure reading without diagnosis of hypertension (situation) |
- 394926003 | Heart disease excluded (situation) |
- 704240001 | Hydrops fetalis not present (situation) |
- 733740006 | Ischemic ulcer absent (situation) |
- 473136009 | Lipodystrophy absent (situation) |
- 274621000 | Lymphadenopathy absent (situation) |

3   Concept information

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 of ComposedOf
concept	Excluded Disorder	determination of the absence of a disease	Excluded Disorder	determination of the absence of a disease				SNOMED CT:315215002 |Disorder excluded (situation)|	
composedOf	code	coded information specifying the concept	code	coded information specifying the absent disease	Code	SNOMED CT; ICD-10; ICD-O-3 Topography; ICD-O-3 Morphology; NANDA; ORPHA; or other			1:n
composedOf	record datetime	datetime the concept was recorded	record datetime	datetime the excluded disorder was recorded	temporal				1:1

General concept	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
Excluded Disorder	0:1	1:1	1:1	1:1

4   Impact on the SPHN Dataset

Excluded Disorder should not impact other concepts of the SPHN Dataset.

5   Discussion

5.1 Naming

The first name of this concept was Exclusion of Diagnosis. The diagnosis is defined as the determination, an interpretative judgement, of the presence of a disorder or its distinction from other disorders based on symptoms and possibly several objective measurements. Since we want to define a concept that captures the exclusion of the disorder itself and not the exclusion of the judgement stating the presence of the disorder, Exclusion of Diagnosis is a metonymy and Exclusion Disorder is more suitable.
Besides, this previous naming could lead to another confusion with the concept of diagnosis by exclusion.

5.2 Current structure

The concept "Excluded Disorder" in its current form represents an initial idea to describe data from cancer registries in the context of the SPHN-financed SPO-project. It needs to be extended further in the future depending on the needs of prospective users, for example with attributes who (medical doctor, study nurse, data engineer), when, on what basis (laboratory test, physical examination, …), etc. the respective diagnosis was excluded.

6   Example

Atopy and lupus erythematosus (an immune disease) are absent from a patient history

Excluded Disorder

> code
>> identifier: L20.-
>> name: Atopic dermatitis
>> coding system name and version: ICD-10-GM-2022
> record datetime: 2015-09-18

Excluded Disorder

> code
>> identifier: L90.-
>> name: Lupus erythematosus
>> coding system name and version: ICD-10-GM-2022
> record datetime: 2015-09-18

A project of SAMW ASSM
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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
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Same example, but SNOMED CT is used instead

Excluded Disorder

    code:
        identifier: 428448003
        name: History of atopy (situation)
        coding system name and version: SNOMED-CT-2023-04-30
    record datetime: 2015-09-18

Excluded Disorder

    code:
        identifier: 200936003
        name: Lupus erythematosus (disorder)
        coding system name and version: SNOMED-CT-2023-04-30
    record datetime: 2015-09-18