

Naming pattern for components of clinical syndromes (X with Y) - Feedback on SEAG 2040_08_19

<https://confluence.ihtsdotools.org/display/editorialag/2024-09-23+SNOMED+Editorial+Advisory+Group+Conference+Call>

SEAG ISSUE: SNOMED currently has extensive editorial guidance on terming patterns for disease combinations that involve the distinction of causal and temporal relationships between conditions. One scenario that has not been adequately addressed is the representation of variably present clinical manifestations of a syndrome. Syndromes often have multiple associated clinical manifestations that are variably present during the disease process or progression. It is important to be able to specifically call out these conditions as being extant at the time of recording, i.e. they are not definitional for the syndrome in general, but are definitional at the time of clinical presentation. An example is Bechet's disease, for which the pathognomic clinical presentation is oral ulcers, but other inflammatory conditions can occur with the disease (e.g. skin rashes, uveitis, arthritis, etc.).

Current editorial guidance (Disorder Combination Modeling) suggests the use of "X with Y" as the FSN terming pattern. Prior discussions have suggested that the use of terms such as "X with Y" and "X in Y" are too vague to represent the association of the condition with the syndrome. The use of "X due to Y" has also been considered inappropriate as the condition is not caused by the syndrome but is a variably present component of the syndrome.

We are seeking advice on a proper terming pattern for this scenario. Some suggested patterns include:

- "X as component of Y"
- "X as manifestation of Y"
- "Y-related X"

SEAG Discussion:

The current editorial guidance is already complicated. Better to clean up the existing combination disorders. [Monique van Berkum](#) has presented examples. It may be better to reallocate these to components of an information model as opposed to try and represent them within the concept model.

Need more clarity on the use of Concurrent with. Would like to see some terms as examples where the current modeling is not sufficient. The desire is to create a relationship between a disorder and its clinical manifestation. e.g. Lyme uveitis

Consensus is that the current guidance is too complex, and the benefits are outweighed by the inconsistency in its application. E.g. 1269223003 |Paraneoplastic uveitis (disorder)|

There is a challenge in creating a terminology that is ontologically precise, yet clinically viable.

Example concepts that would be affected by any terming changes are listed below:

- 230150008 |Meningitis in Lyme disease (disorder)|
- 34253008 |Myopathy in Addison's disease (disorder)|
- 53509000 |Myopathy in Cushing's disease (disorder)|
- 197143009 |Megacolon in Chagas' disease (disorder)|
- 230601003 |Neuropathy in thromboangiitis obliterans (disorder)|
- 142001000119106 |Depressed mood in Alzheimer's disease (disorder)|
- 198231000 |Ulceration of vulva in Behcet's disease (disorder)|
- 192795000 |Cerebral degeneration in Hunter's disease (disorder)|
- 192791009 |Cerebral degeneration in Gaucher's disease (disorder)|
- 230280008 |Progressive aphasia in Alzheimer's disease (disorder)|
- 193177003 |Polyneuropathy in collagen vascular disease (disorder)|

MvB Comments:

SNOMED already has challenges making distinctions between sequelae, complications, associated with, due to, etc. Attempts to “sufficiently define” these concepts have been difficult. In my opinion, we should not add more variations of associations between concepts, or at least we should accept that we cannot sufficiently define content of this nature and may not be able to name it consistently.

Examples of some current SNOMED Definitions:

(https://confluence.ihtsdotools.org/display/DOCEG/Complication+and+Sequela+Modeling_

Sequelae - *A sequela is a disorder that is a consequence, but not an unexpected outcome, that follows after another disorder, procedure, or event. These conditions are often described with the words following, after, post, sequela(e), or late effects.*

Complication - *The ubiquitous use of complication is not defined in relation to criteria; a complication is really just a disorder due to another disorder or procedure.*

From the SNOMED definitions above, it is hard to tell whether diabetic retinopathy is a sequela, a complication, or both, or whether sequela are also complications. Some sequelae in SNOMED become complications others do not. I would advise against adding to the confusion with new distinctions like “as a component of” or “as a manifestation of”.

With respect to the example of Uveitis as a manifestation of Behcet’s, uveitis is one of a set of possible criteria (a set of signs, symptoms, and tests) used to diagnose a person with Behcet’s. Per https://eyewiki.org/Behcet_Disease#:~:text=The%20criteria%20specifies%20a%20requirement,vasculitis%2C%20or%20cells%20in%20vitreous:

The International Study Group (ISG) published the preferred criteria for Behcet’s Disease in 1990 ^[23], which today still are the most widely used and accepted criteria among experts in Behcet’s disease. The criteria specifies a requirement for the presence of recurrent oral aphthae (at least three times in one year) plus two of the following in the absence of other systemic diseases:

- *Recurrent genital ulcers*
- *Ocular lesions, which include anterior or posterior uveitis, retinal vasculitis, or cells in vitreous*
- *Cutaneous lesions, which include erythema nodosum, pseudo-vasculitis, papulopustular lesions, or acneiform lesions consistent with Behcet’s*
- *Positive pathergy test*

IMO, it is very hard to distinguish between the following:

- Uveitis as a manifestation of Behcet’s
- Uveitis as a component of Behcet’s
- Behcet’s-related uveitis
- Uveitis due to Behcet’s
- Uveitis co-occurrent and due to Behcet’s
- Uveitis associated with Behcet’s
- Uveitis as a complication of Behcet’s (NOTE: Unlike Diabetes, which can be diagnosed via lab tests, Behcet’s disease has **no** pathognomonic laboratory tests for its diagnosis. Therefore, a diagnosis is made based on clinical findings. However, if there was a diagnostic lab test for Behcet’s, would uveitis then be merely a complication of Behcet’s in the same way that retinopathy can be a complication of Diabetes?)

As we consider FSN improvements for existing content, I am not sure that we can focus only on FSNs like “X in Y”. However, I would not add more of the “X in Y” variant. We already have more variants than we can clearly understand or define.

EXAMPLES:

Can we clearly articulate and create concept definitions to represent the difference between the following naming variations (which are similar to patterns seen in SNOMED)?

- a. 230150008 |Meningitis **in** Lyme disease (disorder)|
- b. Meningitis AND Lyme disease
- c. Meningitis due to Lyme disease
- d. Meningitis caused by Borrelia
- e. Borrelia infection of the meninges

230150008 |Meningitis in Lyme disease (disorder)| has a parent of 1269516003 |Lyme neuroborreliosis (disorder)|. It’s hard to determine what the difference is between 1269516003 |Lyme neuroborreliosis (disorder)| and its parent 406563003 |Borrelia infection of central nervous system (disorder)|.

Below is a comparison of the inferred definitions of 1269516003 |Lyme neuroborreliosis (disorder)| and its parent 406563003 |Borrelia infection of central nervous system (disorder)|. The only thing that is preventing these 2 concepts from being detected as equivalent, appears to be the the stated primitive parent of 23502006 |Lyme disease (disorder)| on 1269516003 |Lyme neuroborreliosis (disorder)|. This is a good example of two FSNS that likely mean the same thing and are both sufficiently defined but not in exactly the same way.

In general, I would avoid adding new variants for concepts that link “X” and “Y” especially when the relationship between them is difficult to define. Many concept definitions that attempt to represent this complexity and sufficiently define content are not interpretable. For example, the concept hidden by the yellow box below is sufficiently defined but I cannot guess the FSN based on the concept definition.

Summary | Details | Diagram | Expression | Refsets | Members | History | References | Stated | Inferred

Parents

- Language disorder associated with thought disorder (disorder)
- Sequelae of injuries of head (disorder)
- Traumatic injury (disorder)

After → Major head injury

Associated with → Disturbance in thinking

Occurrence → Period of life beginning after birth and ending before death

Interprets → Ability to perform functions related to communication

Has interpretation → Abnormal

Due to → Traumatic event

I think it is unlikely that anyone would come up with 229675006 [Post-traumatic mutism (disorder)] as the FSN for the above Sufficiently Defined concept. It has parents that include patterns like “sequelae of X”, “X associated with Y”, and attributes that include “After”, “Associated with”, and “Due to”.

[Post-traumatic mutism (disorder)] has a synonym of “Mutism after head trauma”. It is hard to know which of the following choices this concept was intended to mean:

- Mutism due to head trauma
- Mutism associated with head trauma
- Mutism after head trauma

Summary | Details | Diagram | Expression | Refsets | Members | History | References | Stated | Inferred

Parents

- Language disorder associated with thought disorder (disorder)
- Sequelae of injuries of head (disorder)
- Traumatic injury (disorder)

Post-traumatic mutism (disorder) ☆

SCTID: 229675006

229675006 | Post-traumatic mutism (disorder) |

en Post-traumatic mutism (disorder)

en Post-traumatic mutism

en Mutism after head trauma

After → Major head injury

Associated with → Disturbance in thinking

Occurrence → Period of life beginning after birth and ending before death

Interprets → Ability to perform functions related to communication

Has interpretation → Abnormal

Due to → Traumatic event