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Purpose of this document

This document describes editorial policies regarding the intended meanings of the clinical findings content in SNOMED CT. It is intended to describe the editorial policies and previous decisions about meanings that are reflected in the current logic-based models. To the extent that there are inconsistencies between the stated policy in this document and the implemented logic-based definitions, these inconsistencies should be resolved through a consensus-based process. For short-term decision-making, the policies in this document should be adhered to. However, this is a working document, subject to change and revision. The intention is to support communication among those who are actively creating definitions, as well as those who are advising, consulting or providing feedback in a variety of capacities.

Status

The document is a working draft. Its contents have in part been derived from several historical sources, including the SNOMED RT Users Guide, Clinical Terms Version 3 documentation, minutes of the SNOMED CT Content Working Group, Concept Model Working Group, Kaiser CMT modelers meetings, and SNOMED Editorial Board / SNOMED International Standards Board meetings.



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1 Clinical Findings

Clinical findings may be simply defined as observations, judgments or assessments about patients. The problem with the terms “finding” and “observation” is that they seem to refer to the judgment of the observer rather than to the actual state of the body. “Organism state” has been suggested as a more neutral name, but it would need to be delimited from a “course of disease.” Examples of clinical findings include: difficulty swallowing, nose bleed, diabetes, headache, and so forth. More precise and reproducible definitions of clinical findings, and the precise boundaries between findings and events, between findings and observables, between findings and situations, and the distinction between “finding” and “disorder”, remain ongoing challenges at the margins. The distinction between a disorder and an observation has proven to be difficult to define in a reproducible manner across the tens of thousands of concepts included under clinical findings. Nevertheless, there are several reliable characteristics of each sub-category (disorders and findings):

1.1 Disorders

- 1) Disorders necessarily are abnormal.
- 2) They have temporal persistence, with the (at least theoretical) possibility of their manifestations being treated, in remission, or quiescent even though the disorder itself still present.
- 3) They necessarily have an underlying pathological process.

1.2 Findings

- 1) Findings may be normal (but not necessarily); no disorders may.
- 2) Some findings may exist only at a single point in time (e.g. a serum sodium level); no disorders may.
- 3) Findings cannot be *temporally* separate from the observing of them (you can't observe them and say they are absent, nor can you have the finding present when it is not capable of being observed).
- 4) They cannot be defined in terms of an underlying pathological process that is present even when the observation itself is not present.

Disorders may be present as a propensity for certain abnormal states to occur, even when treatment mitigates or resolves those abnormal states. In some cases the disease process is irrefutable, e.g. meningococcal meningitis. In others an underlying disease process is assumed based on the temporal and causal association of the disorder and its manifestation, e.g. nystagmus disorder is different from the finding/observation of nystagmus, which can be a normal physiological response to rotation of the head. If you spin around and around and then have nystagmus (the finding) you still do not have nystagmus disorder. And someone can have a nystagmus disorder without currently manifesting nystagmus. Similarly, deafness disorder is different from the symptom (observation) of reduced hearing, which can be due to a number of temporary causes such as excessive ear wax.



2 The SNOMED CT Concept Model for Clinical Findings / Disorders

2.1 Attributes used to define Clinical Findings and Disorders

NOTE: Permissible values for these attributes include the concepts listed and their descendants.

Table 4.1: Approved Clinical Finding attributes summary table

Defining Attribute	Permissible Values (Concepts listed and their descendants)	
FINDING SITE	<i>Anatomical structure</i> 91723000	<i>Acquired body structure</i> 280115004
ASSOCIATED MORPHOLOGY	<i>Morphologically abnormal structure</i> 49755003	
PATHOLOGICAL PROCESS	<i>Autoimmune</i> 263680009	
HAS DEFINITIONAL MANIFESTATION	<i>Clinical finding</i> 404684003	
ASSOCIATED WITH	<i>Clinical Finding</i> 404684003	<i>Physical object</i> 260787004
	<i>Procedure</i> 71388002	<i>Physical force</i> 78621006
	<i>Event</i> 272379006	<i>Pharmaceutical/biologic product</i> 373873005
	<i>Organism</i> 410607006	<i>SNOMED CT Concept</i> 138875005*
	<i>Substance</i> 105590001	* For this concept, only the concept and not all of its descendants is allowed as a value.
CAUSATIVE AGENT	<i>Organism</i> 410607006	<i>Physical force</i> 78621006
	<i>Substance</i> 105590001	<i>Pharmaceutical/biologic product</i> 373873005
	<i>Physical object</i> 260787004	* For this concept, only the concept and not all of its descendants is allowed as a value.
	<i>SNOMED CT Concept</i> 138875005*	
DUE TO	<i>Clinical Finding</i> 404684003	<i>Event</i> 272379006
AFTER	<i>Clinical Finding</i> 404684003	<i>Procedure</i> 71388002
SEVERITY	<i>Severities</i> 272141005	
CLINICAL COURSE	<i>Courses</i> 288524001	
EPISODICITY	<i>Episodicities</i> 288526004	
INTERPRETS	<i>Observable entity</i> 363787002	<i>Evaluation procedure</i> 386053000
	<i>Laboratory Procedure</i> 108252007	
HAS INTERPRETATION	<i>Findings values</i> 260245000	<i>Result comments</i> 281296001
OCCURRENCE	<i>Periods of life</i> 282032007	
FINDING METHOD	<i>Procedure</i> 71388002	
FINDING INFORMER	<i>Performer of method (person)</i> 420158005	<i>Subject of record (person)</i> 410604004
	<i>Provider of history other than subject (person)</i> 420058008	<i>Subject of record or other provider of history (person)</i> 419358007



2.2 Finding site

This attribute specifies the body site affected by a condition.

Permissible values include the following concepts and their descendants:

Anatomical structure (body structure) 91723000
Acquired body structure (body structure) 280115004

Examples:

- (1) *Kidney disease (disorder)*
FINDING SITE *Kidney structure (body structure)*
- (2) *Appendicitis (disorder)*
FINDING SITE *Appendix structure (body structure)*

2.3 Associated morphology

This attribute specifies the morphologic changes seen at the tissue or cellular level that are characteristic features of a disease.

Permissible values include the following concept and its descendants:

Morphologically abnormal structure (morphologic abnormality) 49755003

Examples:

- (1) *Bone marrow hyperplasia (disorder)*
ASSOCIATED MORPHOLOGY *Hyperplasia (morphologic abnormality)*
- (2) *Pancreatitis (disorder)*
ASSOCIATED MORPHOLOGY *Inflammation (morphologic abnormality)*

2.4 Pathological process

This attribute provides information about the underlying pathological process for a disorder, but only when the results of that process are not structural and cannot be represented by the ASSOCIATED MORPHOLOGY attribute. Permissible values include the following single concept:

- *Autoimmune (qualifier value)* 263680009

Example:

Autoimmune parathyroiditis (disorder)
PATHOLOGICAL PROCESS *Autoimmune (qualifier value)*

Pathological process must not be used for values that could overlap with ASSOCIATED MORPHOLOGY. Inflammatory processes result in inflammation (by definition), and our preference is to model these disorders using their morphology. At present we have only one pathological process that has no defined morphology: autoimmune. But it seems likely that others may be found to be necessary, such as infectious and metabolic processes.



2.5 Has definitional manifestation

This attribute links disorders to the manifestations (observations) that define them. It can only be applied to disorders.

Permissible values include the following concept and its descendants:

- *Clinical finding (finding)* 404684003

Example:

Seizure disorder (disorder)

HAS DEFINITIONAL MANIFESTATION *Seizure (finding)*

Hypertensive disorder, systemic arterial (disorder)

HAS DEFINITIONAL MANIFESTATION *Finding of increased blood pressure (finding)*

2.6 Associated with

This attribute asserts an interaction between two concepts beyond simple co-occurrence in the patient. ASSOCIATED WITH represents a clinically relevant association between concepts without either asserting or excluding a causal or sequential relationship between the two.

Permissible values include the following concepts and their descendants:

- *Clinical finding (finding)* 404684003
- *Procedure (procedure)* 71388002
- *Pharmaceutical/biologic product (product)* 373873005
- *Substance (substance)* 105590001
- *Organism (organism)* 410607006
- *Physical object (physical object)* 260787004
- *Physical force (physical force)* 78621006
- *Event (event)* 272379006
- *SNOMED CT Concept* 138875005 (For this concept, only the concept and not all of its descendants is allowed as a value)

ASSOCIATED WITH subsumes the following, more specific, attributes in what is called a role hierarchy (explained in the section on Role Hierarchies):

- AFTER
- DUE TO
- CAUSATIVE AGENT

2.6.1 After

This attribute is used to model concepts in which a clinical finding occurs after another clinical finding or procedure. Neither asserting nor excluding a causal relationship, it instead emphasizes a sequence of events.

Permissible values include the following concepts and their descendants:

- *Clinical finding (finding)* 404684003
- *Procedure (procedure)* 71388002



Example:

Post-viral disorder (disorder)
AFTER *Viral disease (disorder)*

This example can be paraphrased as: “every post-viral disorder occurs after some viral disease.”

2.6.2 Due to

This attribute is used to relate a *Clinical finding* directly to its cause. If a clinical finding merely predisposes to or worsens another disorder, rather than causing it directly, then the more general attribute ASSOCIATED WITH is used instead.

Permissible values include the following concepts and their descendants:

- *Clinical finding (finding)* 404684003
- *Event (event)* 272379006

Example:

Acute pancreatitis due to infection (disorder)
IS_A *Acute pancreatitis (disorder)*
DUE TO *Infectious disease (disorder)*

This example can be paraphrased as: “every acute pancreatitis due to infection is due to some infectious disease.” DUE TO is not a sub-attribute of AFTER because DUE TO can take values that are not events; and AFTER implies a clear temporal sequence, whereas causative events may effectively appear at the same time and be temporally co-existent with the thing they cause.

2.6.3 Causative agent

This attribute identifies the direct causative agent of a disease. It does not include vectors, e.g., a mosquito that transmits malaria.

Permissible values include the following concepts and their descendants:

Organism (organism) 410607006
Substance (substance) 105590001
Pharmaceutical/biologic product (product) 373873005
Physical object (physical object) 260787004
Physical force (physical force) 78621006
SNOMED CT Concept 138875005 (For this concept, only the concept and not all of its descendants is allowed as a value)



Examples:

(1) *Bacterial endocarditis (disorder)*

CAUSATIVE AGENT *Bacterium (organism)*

(2) *Fentanyl allergy (disorder)*

CAUSATIVE AGENT *Fentanyl (substance)*

(3) *Electrical burn of skin (disorder)*

CAUSATIVE AGENT *Electricity (physical force)*

The first example can be paraphrased as “every bacterial endocarditis has some bacterium as causative agent.”

2.7 Severity

This attribute is used to subclass a *Clinical finding* concept according to its severity; however, caution is encouraged because this use is said to be *relative*. By relative, it is meant that it is incorrect to assume that the same degree of disease intensity or hazard is implied for all *Clinical findings* to which this attribute is applied. There are three reasons.

First, “severe” could be interpreted differently depending on what other values are available to choose for severity. Thus severity is relative to the other values in the value set. Consider the different meaning of “severity” in each of the following two sets of values:

Mild / Moderate / Severe

Minimal / Mild / Moderate / Severe / Very Severe

Second, the severity is defined relative to the expect degree of intensity or hazard of the *Clinical finding* that is being qualified. A common cold has a baseline intensity or hazard much less than that of a more serious disease like lupus erythematosus or pneumonia; thus a severe cold might be considered less intense or hazardous than a mild pneumonia.

Third, some disorders that are life-threatening do not ordinarily have a severity assigned to them. Cancer, for example, is generally not subclassed according to mild, moderate and severe types, but rather is subclassed according to stage or grade.

For these reasons, the SEVERITY attribute cannot be relied on to retrieve all *Clinical findings* with serious or life-threatening import. Nevertheless, it is still useful for subclassing certain concepts and differentiating between different severities of a single disorder.

Current permissible values of SEVERITY include:

Descendants of *Severities (qualifier value)* 272141005 which include but are not limited to:

Mild (qualifier value)

Moderate (severity modifier) (qualifier value)

Severe (severity modifier) (qualifier value)

2.8 Clinical course

This attribute is used to represent both the course and onset of a disease. Many conditions with an acute (sudden) onset also have an acute (short duration) course. Few diseases with a chronic (long-term) course would need to have their onset sub-divided into rapid or gradual subtypes, and thus there



is no clear need for separating the rapidity of onset from the duration of a disease; based on testing by implementers and modelers, a single attribute with values that combine these meanings has clearly been more reproducible and useful than two attributes that attempt to separate the meanings.

Permissible values include the following concept and its descendants:

- *Courses (qualifier value)* 288524001

Examples:

(1) *Acute amebic dysentery (disorder)*

CLINICAL COURSE *Sudden onset AND/OR short duration (qualifier value)*

(2) *Chronic fibrosing pancreatitis (disorder)*

CLINICAL COURSE *Chronic clinical course (qualifier value)*

The word “acute” has more than one meaning, and the meanings are often overlapping or unclear. Acute may imply rapid onset, short duration, or high severity; in some circumstances it might be used to mean all of these. For morphological terms it may also imply the kind of morphology associated with the speed of onset. *Acute inflammation (morphologic abnormality)* does not necessarily have CLINICAL COURSE *Sudden onset AND/OR short duration*, but rather implies polymorphonuclear infiltration; likewise *Chronic inflammation (morphologic abnormality)* implies lymphocytic infiltration, not necessarily a chronic course, although inflammation with a chronic course is highly correlated with a lymphocytic infiltration.

2.9 Episodicity

EPISODICITY is used to represent episodes of care provided by a physician or other care provider, typically a general practitioner, *not* episodes of disease experienced by the patient (see notes in section 6.2 regarding origin of the attribute). For example, asthma with EPISODICITY = *first episode* represents the first time the patient presents to their GP with asthma. EPISODICITY is not used to model any concepts pre-coordinated in the International Release but it can still be used in post-coordination as a qualifier.

Current permissible values include the following concept and its descendants:

- *Episodicities (qualifier value)* 288526004

2.10 Interprets

This attribute refers to the entity being evaluated or interpreted, when an evaluation, interpretation or “judgment” is intrinsic to the meaning of a concept. This attribute was created specifically to be grouped with the HAS INTERPRETATION attribute in order to form a clinical finding definition.

Permissible values include the following concepts and their descendants:

- *Observable entity (observable entity)* 363787002



- *Laboratory procedure (procedure)* 108252007
- *Evaluation procedure (procedure)* 386053000

Example:

Decreased muscle tone (finding)

INTERPRETS *Muscle tone (observable entity)*

HAS INTERPRETATION *Decreased (qualifier value)*

2.11 Has interpretation

This attribute is designed to be grouped with the attribute INTERPRETS, and designates the judgment aspect being evaluated or interpreted for a concept (e.g., presence, absence, degree, normality, abnormality, etc.), applied to the observable entity referenced by INTERPRETS.

Permissible values include the following concepts and their descendants:

- *Findings values (qualifier value)* 260245000
- *Result comments (qualifier value)* 281296001

Example:

Decreased muscle tone (finding)

INTERPRETS *Muscle tone (observable entity)*

HAS INTERPRETATION *Decreased (qualifier value)*

2.12 Occurrence

This attribute refers to the specific period of life during which a condition first presents. Multiple values of OCCURRENCE for a single concept would be an error. This does not mean the condition cannot persist beyond the period of life in which it first presents.

Permissible values include the following concept and its descendants:

- *Periods of life (qualifier value)* 282032007

Example:

Childhood phobic anxiety disorder (disorder)

OCCURRENCE *Childhood (qualifier value)*

2.13 Finding method

This attribute specifies the means by which a clinical finding was determined. This attribute is frequently used in conjunction with FINDING INFORMER. Findings that specify that they were determined by examination of the patient (e.g., *On examination - ankle clonus (finding)*) should have a value for both FINDING METHOD and FINDING INFORMER.

Permissible values include the following concept and its descendants:

- *Procedure (procedure)* 71388002



Example:

Finding by palpation (finding)

FINDING METHOD *Palpation (procedure)*

2.14 Finding informer

This attribute specifies the person or other entity from which the clinical finding information was obtained. This attribute is frequently used in conjunction with FINDING METHOD.

Permissible values include the following concepts and their descendants:

Subject of record or other provider of history (person) 419358007

Subject of record (person) 410604004

Provider of history other than subject (person) 420058008

Performer of method (person) 420158005

Examples:

(1) *On examination - ankle clonus (finding)*

FINDING INFORMER *Performer of method (person)*

(2) *Complaining of a headache (finding)*

FINDING INFORMER *Subject of record or other provider of history (person)*

It is accepted that an information model should permit identification of a particular individual who provides information; FINDING INFORMER is not about the particular individual. It is about the *category or type* of informer, which is used to differentiate self-reported symptoms from provider-observed signs. Granted, this permits inclusion of “epistemology-loaded terms” (cf. Bodenreider et al., FOIS 2004), but health care is full of such terms, and they are (or at least can be) understandable, reproducible and useful.

3 Specific disorder types

3.1 Congenital, hereditary, familial, developmental, genetic

3.1.1 Congenital:

We apply OCCURRENCE = *congenital* to those disorders that are present at birth. Although the word “congenital” is often applied to genetic disorders, we prefer the term “genetic” for those disorders that arise from abnormalities of the genes.

3.1.2 Hereditary:

It is difficult to cleanly define “hereditary” because it either may or may not include random mutations; the offspring of genetically normal parents may have a genetic disease, but there may be confusion about whether this is to be classified as a hereditary disease. It may be hereditary to the proband’s offspring, but was not inherited from the proband’s parents. Because of this ambiguity, “hereditary” is not a reproducible label for a category. See “known issues” below for current status.



3.1.3 Familial:

The term “familial” is also somewhat ambiguous. It may be interpreted broadly, meaning that the disorder is found in higher proportions in the immediate or extended family than in other groups. It should not be used as a synonym for “genetic”. See “known issues” below for current status.

3.1.4 Developmental:

“Developmental” is a useful label for disorders that occur during development (both before and after birth) and that affect structures or functions that are in the process of developing. Some of these may be present at birth, and others may only manifest themselves post-natally.

The following diagram lays out the general logical structure of genetic, developmental and congenital categories, along with non-genetic, non-developmental and post-natal categories. A final dimension, called “extrinsic physical force”, is necessary to distinguish deformations from malformations. The various boxes in the diagram represent categories formed from the combination of the four dimensions each of which represents the answer to one of the following four questions:

- 1) is it genetic or not?
- 2) is it developmental or not?
- 3) is it present at birth or not?
- 4) is it due to an extrinsic physical force or not?

The diagonal hashed lines represent combination categories that do not occur. For example, there are no genetic disorders that are due to an extrinsic physical force. Likewise, there are no congenital disorders that are considered non-developmental. The blue lines represent congenital malformations; they may be either genetic or non-genetic in origin. The red circle represents those that are genetic in origin. Finally the green area represents the meaning of “acquired”, i.e. any disorder that is non-genetic and not present at birth.

Arrows leading from each of the non-hashed boxes in the central diagram point to examples of disorders that typify that category. For example, Huntington’s disease is the typical example of a genetic disease that is neither congenital nor developmental. Vitamin D deficiency rickets is a typical example of a non-genetic, non-congenital developmental malformation.

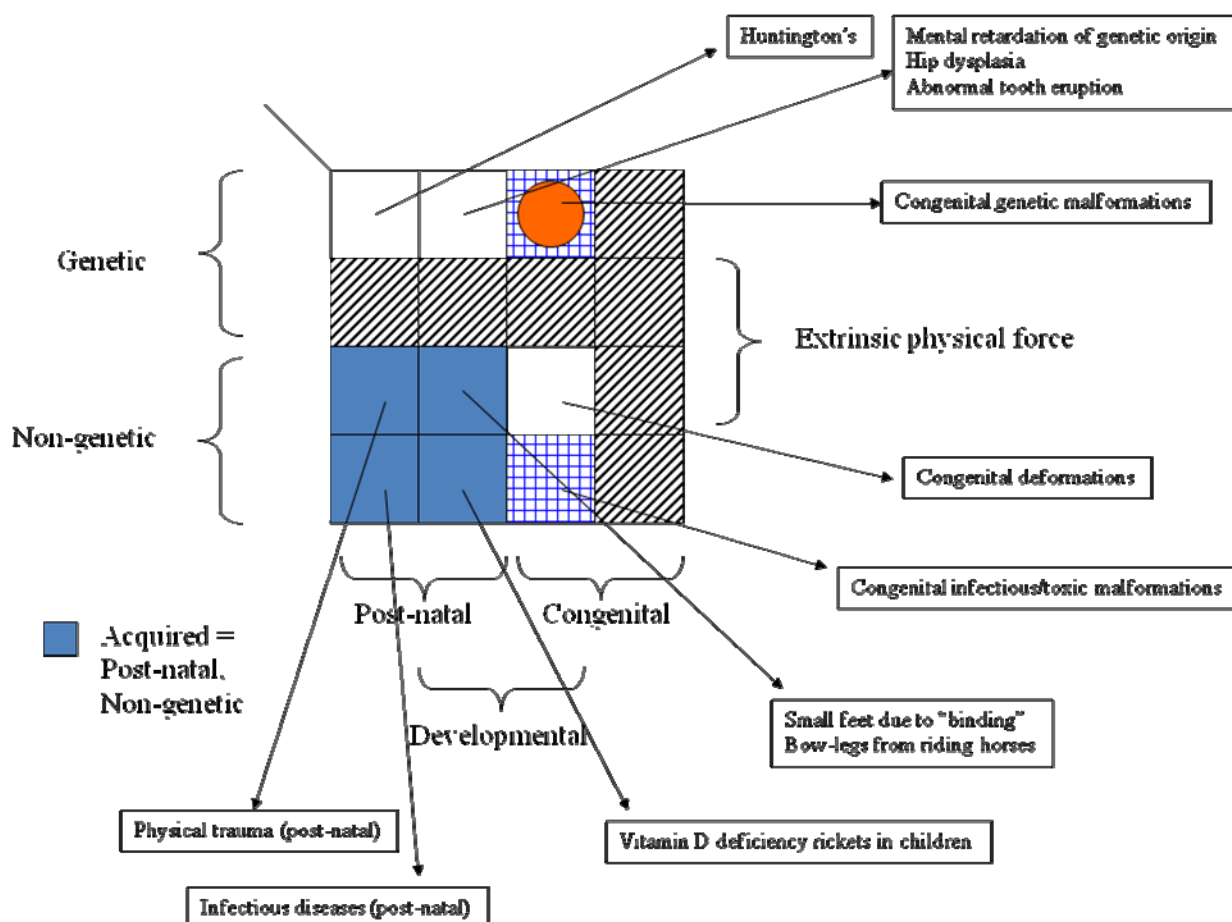


Figure 1: Diagram of the relationships of genetic, congenital, developmental, and acquired

3.2 Congenital vs. acquired

The general rule is that disorders in general may be either congenital or acquired, and congenital disorders are specifically modeled using OCCURRENCE = congenital, but there is no attribute for modeling the fact that a disorder is acquired. If the FSN does not mention either “congenital” or “acquired”, then we do not model the concept as being under “congenital disorder”, and there is nothing in the concept model to specifically indicate that it is necessarily acquired. There are a few concepts that have “acquired” in their FSN, but they remain primitive.

3.2.1 Congenital vs acquired syphilis

As an exception to the general rule that “acquired” is never assumed, syphilis is a disorder in which an FSN that does not mention “congenital” might be assumed to imply acquired. ICD seems to follow such a rule, but there is no rule to that effect in SNOMED. In the absence of such a rule, the precise meaning of the FSN should be followed. If “acquired” is not stated in the FSN, the concept means the general category that subsumes both congenital and acquired forms.



3.3 Malformation, deformation, anomaly

As illustrated in figure 1 and described in section 4.1, a deformation is a structural abnormality that is due to an extrinsic physical force. Malformations are those structural abnormalities that result from intrinsically disordered structural development. The word “anomaly” is, by itself, ambiguous because it may be used to mean any abnormality including non-structural ones, or it may be used to mean malformation, or it may be a general term that includes both malformation and deformation. Terms that contain the word “anomaly” must therefore be examined to see whether the additional words provide sufficient specificity to overcome the inherent ambiguity. “Congenital anomaly of <x structure>” is definitely structural but is not the same as congenital malformation, and therefore it can be regarded as having the more general meaning of “structural abnormality present at birth.”

3.4 Lesion

The word “lesion” can be used to refer to both structural and functional abnormalities. If a disorder or procedure refers to a “lesion” in a way that makes it clear that it is a generic term for a **structural** abnormality, then the correct modeling approach is to use ASSOCIATED-MORPHOLOGY = *morphologically abnormal structure* (for disorders) or PROCEDURE-MORPHOLOGY (for procedures). Functional-only lesions obviously should not be modeled using values from the *morphologically abnormal structure* hierarchy.

3.5 Tumor vs. Neoplasm

The word “tumor” has two main meanings:

- 1) a mass, regardless of whether it is neoplastic or not, or
- 2) a neoplastic mass

“Neoplasm” is preferred since it is less ambiguous than “tumor”.

3.6 Primary vs secondary neoplastic disorders

SNOMED follows the usage in ICD-O, ICD-9 and ICD-10, where “secondary malignant neoplasm of (site x)” is uniformly interpreted to mean that metastasis has occurred *to* site x. The alternative reading (*from* site x) is not what is intended. If you want to code a metastasis from a lung tumor, then SNOMED also has codes that explicitly use the word “from”, such as 315006004 “metastasis from malignant tumor of lung”.

Detailed information about metastases (primary at site x, metastatic to site y) could possibly be recorded using one of two different styles: either a style using *two* expressions – placed in two statements in the clinical record – one statement for the primary and one statement for the secondary; or a style using only *one* expression. The two-expression style would be required to code the case with ICD9 or ICD10 codes, and it would be valid to use this style with SNOMED expressions also. The one-statement style would have to use one SNOMED expression with two role groups – each with a morphology and site that is appropriate to the level of detail required.

An example of the one-statement style: primary malignant neoplasm of breast metastatic to lymph node.

64572001|disease|:



```
{116676008|associated morphology|=86049000|neoplasm, malignant (primary)|  
,363698007|finding site|=76752008|breast structure|},  
{116676008|associated morphology|=14799000|neoplasm, metastatic|  
,363698007|finding site|=59441001|lymph node structure|}
```

The morphology code in the role group differentiates the primary from the secondary site. This style of modeling, using a single statement/expression, does not cleanly permit one to differentiate between *just* a metastasis from the lung, versus *both* a primary lung tumor and a metastasis, at a particular instance/point in time. The recommended solution to this problem is the two-statement style, with a third statement that links them, using the information model to accomplish the linkage, rather than trying to do it all in the terminology. This recommended style permits users to attach time-stamps and other instance identifiers to the primary neoplasm, and separate time-stamps and other instance identifiers to the metastasis. This appears more flexible and semantically robust. On the other hand, if a user just wants a broad category expressing the site of the metastasis and the site of the primary, in the same statement, the one-expression style does allow that, as accommodated by some pre-coordinated codes that are modeled with two role groups as described above.

3.7 Neoplasm vs hamartoma

A neoplasm is defined as an abnormal growth of tissue no longer under normal control. A hamartoma is defined as a benign self-limited growth of disorganized mature cells normally found in the region, representing faulty development. Since the cells in hamartomas are mature cells whose growth is under normal control, a hamartoma is not a neoplasm.

SNOMED attempts to sort out the disorder concepts that get confused in the area of tumors, neoplasms and hamartomas by making a “neoplasm and/or hamartoma” disorder category, with five subtypes:

- 1) hamartoma
- 2) neoplastic disease
- 3) hemangioma
- 4) lymphangioma
- 5) melanocytic nevus

Likewise in the morphologic abnormality hierarchy, we have “neoplasm and/or hamartoma” with five subtypes:

- 1) hamartoma
- 2) neoplasm
- 3) hemangioma - category
- 4) lymphatic vessel tumor
- 5) melanocytic nevus - category

Hemangiomas, lymphangiomas and melanocytic nevi can be either hamartomas (these are usually present at birth) or neoplasms (these usually develop later in life). All of the subtypes of hemangioma, lymphangioma or melanocytic nevus can thus be aggregated under these upper-level generalizations,



immediately under “neoplasm and/or hamartoma”, without necessarily having to first attempt to (incorrectly) categorize them as either neoplasms or hamartomas.

3.8 Nevus

The word “nevus” has many different meanings. The differences generally hinge on answers to the following questions:

- a) is it necessarily on the skin? Or can it be located in mucosal sites or other sites?
- b) is it necessarily visible? Or can it be in internal locations such as gastric mucosa, etc?
- c) is it necessarily present at birth? Or can it make its appearance later in life?
- d) is it necessarily dark and made of melanocytes? Or can it be non-pigmented, or made of other types of cells?
- e) is it necessarily made of tissue that is normally present at the site? Or can it be ectopic?
- f) does it exclude benign neoplasms?

Here are some common meanings of “nevus” based on some combinations of answers to these questions:

- 1) a birthmark, that is, any visible spot on the skin or oral mucosa present since birth, regardless of tissue of origin, excluding benign neoplasms.
- 2) any benign cluster of melanocytes, regardless of location, and regardless of pigmentation, whether present since birth or appearing later.
- 3) any cutaneous hamartoma. This excludes non-cutaneous sites, and excludes neoplasms and ectopic tissue such as choristomas.

As a result of this wide variation in meaning, any SNOMED FSN containing the word “nevus” is prone to being ambiguous. For example, consider “vascular nevus”. This term might mean:

- 1) congenital blood vessel tumors in the skin,
- 2) congenital blood vessel hamartomas or neoplasms that are visible somewhere (not just in the skin, but also including mucosa, whether visible externally or not),
- 3) congenital blood or lymphatic vessel tumors in the skin,
- 4) congenital blood or lymphatic vessel hamartomas or neoplasms that are visible somewhere,
- 5) any of the above but not necessarily congenital

A better FSN for “vascular nevus (morphologic abnormality)” would be “vascular hamartoma (morphologic abnormality)”. Likewise a better FSN for “congenital vascular nevus (disorder)” would be “congenital vascular hamartoma (disorder)”. In those cases where common clinical usage of a term containing “nevus” is unambiguous, there is no call for the term (or concept described using the term) to be retired.

3.9 Hematologic

There is more than one meaning of "hematologic". A structural definition based on "hematological system structure" would include hematopoietic and lymphoid structures (including bone marrow, spleen, thymus, lymph nodes, etc) as well as the cellular components of blood. Hematologic neoplasms clearly fit this definition.



A definition based on what hematologists do is broader. Disorders of hemostasis and thrombosis are managed by hematologists, but these do not have a common structural overlap with the lymphoid and hematopoietic systems (with the exception of platelets and megakaryocytes). For clarity, "hematologic disorder" is a navigational concept that could be used to define a reference set that would include disorders of blood and blood forming organs, as well as disorders of hemostasis and thrombosis, depending on what is intended.

If a patient says they have a "hematologic disorder" or a "blood disorder," the navigational concept could be used to record and capture what they said, but the variability in meaning is too great to assign necessary and sufficient conditions to this phrase.

3.9.1 Hematologic disorders, lymphoid and myeloid neoplasms

When a clinician or patient (or the literature) says "hematologic disorder," they could be referring to disorders with a morphology of hematopoietic cell origin, disorders affecting the blood forming organs (bone marrow, lymph nodes, spleen, thymus, and other lymph tissues), disorders of the cellular components of blood, and/or disorders of the function of hemostatic and thrombotic systems.

Within SNOMED CT, diseases of the cellular components of blood are most readily defined in terms of their definitional manifestations (for example, *anemia (disorder)* HAS DEFINITIONAL MANIFESTATION *erythropenia(finding)*), because there is no clearly defined body site (a cell type is not considered a body site) and there may be no defined morphology.

Diseases of the blood forming organs (bone marrow, lymph nodes, etc.) can be defined in terms of any one or a combination of the following:

- 1) The morphology. For neoplastic diseases this is understood, at a minimum, to include those morphologies covered by the neoplasms listed in ICD-O.

Example:

Hodgkin's disease (disorder) ASSOCIATED MORPHOLOGY *Hodgkin lymphoma (category)* (*morphologic abnormality*)

- 2) The body site involved – especially specific lymph node groups or skin sites.

Example:

primary cutaneous T-cell lymphoma (disorder) FINDING SITE *skin structure*

- 3) The definitional manifestations of the disease - for those diseases without a specific neoplastic morphology and/or without a specific topographical site of involvement other than the blood-forming organs in general.

Example:

toxic neutropenia (disorder) HAS DEFINITIONAL MANIFESTATION *neutropenia (finding)*

Important examples of where it is important to distinguish disorders defined by morphology versus site versus manifestation include the T-cell lymphomas and disorders of plasma cells/immunosecretory disorders. T-cell lymphomas can be subcategorized according to the site of the primary: a lymph node versus the skin or other extranodal site. This means that a site of "lymphoid structure" *cannot* therefore be the defining characteristic of the parent concept "T-cell lymphoma". Its defining attribute should be morphology alone. In the case of plasma cell disorders/immunosecretory disorders, some of them (monoclonal gammopathy, heavy chain disease, Waldenstrom's, etc) are defined in terms of their manifestation, i.e. the type of monoclonal protein they secrete, while others (myeloma,



plasmacytoma) are defined in terms of their morphology, regardless of whether they are secretory or not. It would be incorrect to add "HAS DEFINITIONAL MANIFESTATION = monoclonal paraproteinemia" to myeloma, because not all myelomas are secretory. However, we can safely give immunosecretory disorders a morphology of "plasma cell neoplasm", even though no mass may have been identified and the monoclonal protein may be the only evidence that there is a clonal neoplasm. In general, lymphoid and myeloid neoplasms *can* be modeled with their morphology alone, without a site. Leukemias and myelodysplastic syndromes are, *in addition*, modeled with site of bone marrow structure. Hairy cell leukemia has site bone marrow and site spleen, because both are uniformly involved.

For reference see Harris NL et al, "World Health Organization Classification of Neoplastic Diseases of the Hematopoietic and Lymphoid Tissues: Report of the Clinical Advisory Committee Meeting-Airlie House, Virginia, November 1997" in J. Clinical Oncology, Vol.17, No 12 (December), 1999: pp 3835-3849.

For additional information on the classification of the myeloid neoplasms, see Vardiman JW, Harris NL and Brunning RD. "The World Health Organization (WHO) Classification of the Myeloid Neoplasms", Blood, 1 October 2002, Volume 100, Number 7, pp 2292-2302.

3.9.2 Coagulation, Hemostasis and Thrombosis

There is more than one meaning of "coagulation". A broad sense of "coagulation" as the stopping of bleeding is better described as hemostasis. A more narrow definition limited to the formation of the fibrin clot might exclude certain components of hemostasis, such as the ability to stop hemorrhage through the actions of blood vessels, collagen, endothelial cells, and platelets, in the absence of clotting. Individuals with congenital fibrinogen deficiency cannot form fibrin clots, yet they are able to stop bleeding. Therefore, coagulation disorders are kinds of hemostatic disorders.

3.10 Post-infectious disorders

Post-infectious disorders are not subtypes of infectious disorders. The AFTER attribute is used for linking post-infectious disorders with their associated infections.

3.11 Infectious disease vs. Inflammatory disorder

Infectious disease and inflammatory disorder are siblings. It might seem more intuitive for infectious disease to be a child of inflammatory disorder. However, not all infectious disorders are inflammatory. These concepts will remain siblings. Infectious disease and its subtypes have a CAUSATIVE AGENT relationship to the organism that is infecting. Inflammatory disorder has an ASSOCIATED MORPHOLOGY relationship to *inflammation (morphologic abnormality)* or one of its subtypes.

3.12 Trauma, injury, damage

The word "trauma" has multiple senses. The first distinction is physical damage to the body versus psychic trauma. We assume "trauma" means physical damage unless accompanied by words that make clear it is psychic.



Traumatic injury (disorder) is defined as any disorder with a morphology of "traumatic abnormality". See known issues (below) for a discussion of the known problems with traumatic morphologies.

There is a problem that occurs if we attempt to require "injury" to be synonymous with "trauma" which can be best illustrated by the example of the very common usage of the word "injury" when referring to damage to the brain. An internet search for the phrase "non-traumatic brain injury" will show that this refers to brain damage that is the result of asphyxiation, stroke, drowning, toxic injury, etc., and not due to direct physical impact to the skull (the traumatic brain injuries). We needed a broad category that would allow us to categorize injuries broadly including non-traumatic ones. The concept created for this purpose is *traumatic and/or non-traumatic injury (disorder)*.

3.12.1 Laceration, incised wound, rupture, traumatic rupture, spontaneous rupture:

The word "lacerated" has two meanings, which can be succinctly summarized as "torn" vs "cut". Common clinical usage equates "laceration" with "incised wound". For example, a common emergency room problem is accidental cuts of fingers with kitchen knives. These are routinely called "lacerations". On the other hand, most dictionaries insist that "laceration" implies a wound with ragged edges as a result of tearing. Obstetrical lacerations carry this latter meaning. When structures are torn or ruptured, the edges are usually irregular.

There are two morphologies with a synonym of "laceration": "incised wound", and "traumatic rupture". Modelers must choose which of these two meanings is intended when the word "laceration" or "lacerated" appears in a concept description from the "injury" hierarchy.

More generally, ruptures can occur either as a result of injury or spontaneously. The word "rupture", when applied to muscles and tendons, implies a traumatic injury (e.g. "rupture of collateral ligament of the knee"). But "rupture" when applied to an internal viscus may be either traumatic or spontaneous (e.g. rupture of aorta, rupture of ovary, etc).

"Rupture" has subtype morphologies "traumatic rupture" and "nontraumatic rupture". It is important to make this distinction, at a minimum, in order to support queries related to the effects of trauma. Modelers should choose "traumatic rupture" as the value of Associated-morphology for concepts using the word "rupture" with anatomical sites (such as muscles and tendons) where rupture requires trauma, in the absence of a specific lesion. Modelers should choose "rupture" as the value of Associated-morphology for concepts using the word "rupture" with sites (such as internal organs) where both traumatic and spontaneous rupture are seen. Nontraumatic rupture is usually stated to be so, but may also be inferred if the thing rupturing is a lesion which ordinarily leads to spontaneous rupture in the absence of trauma (e.g. rupture of inflamed appendix).

3.13 Death

"Death" is an event, not a disorder. Concepts like "relatives died," "death of companion" go under "life events - finding" which is under "social and personal history finding."

3.13.1 Sudden Cardiac Death

"Sudden cardiac death" is a term used in clinical practice to refer to an arrhythmia that results in sudden loss of cardiac function which, if not quickly reversed, will lead to *actual* death (as opposed to a high risk of imminent death). This concept needs an FSN that indicates it is not a kind of death, and it should not be classified under "death" because individuals to whom this label is applied have not



necessarily been officially declared dead, and are frequently revived. It is regarded as a subtype of "cardiac dysrhythmia."

3.14 Hernia, herniated structure, and hernial opening

Hernias involve two different structures, the structure herniated and the structure through which the hernia passes. Each of these might need to be described by different morphologies. There are two morphology codes, 414403008: *herniated structure (morphologic abnormality)*, and 414402003: *hernial opening (morphologic abnormality)*. The "herniated structure" morphology should be the value of ASSOCIATED-MORPHOLOGY, and this should be grouped with a FINDING-SITE that has as its value the code for the anatomical structure that herniates.

Example:

Intestinal hernia (disorder)

Group 1 { ASSOCIATED MORPHOLOGY = herniated structure,
FINDING SITE = intestinal structure},

Group 2 { ASSOCIATED MORPHOLOGY = hernial opening,
FINDING SITE = abdominal structure}.

The "hernial opening" morphology should be grouped with a FINDING-SITE attribute, with a value which is the code for the anatomical structure through which the hernia passes. It can be a general concept such as "abdominal structure" if the concept is non-specific about what is being herniated through. One or the other of these role groups should be omitted if the hernia does not necessarily entail a particular herniated structure or a particular hernial opening. For example, "abdominal wall hernia" specifies the hernial opening but not the herniated structure. In this case, the definition should omit the group with a morphology of herniated structure.

4 Known issues and problems

4.1 Attribute overlap or interaction issues

4.1.1 Morphology and occurrence

ASSOCIATED MORPHOLOGY *Congenital morphology* versus OCCURRENCE *congenital*.

It has been proposed to eliminate all "congenital morphology" concepts since congenital is interpreted to mean "at birth". This proposal still requires analysis.

4.1.2 Morphology and pathologic process

An inflammatory process yields an inflammatory morphology. We don't need to define inflammatory disorders with both attributes, therefore we don't use PATHOLOGIC PROCESS for defining inflammatory disorders. However, some users may wish to see a fully populated set of pathological processes, in addition to (and coordinated with) the morphologies.

4.1.3 Morphology and After

ASSOCIATED MORPHOLOGY *traumatic morphology* versus AFTER *injury*



It may be possible to define a general concept inclusion that allows inference that a morphology that occurs after injury is a traumatic morphology, and likewise that a traumatic morphology implies a disorder that occurred after injury.

4.2 Distinguishing continuants, continuant potentialities, and occurrents

Many of the difficulties that were found in trying to make a clean split between findings and disorders probably resulted from asking the wrong question. We clearly understood that epilepsy is different from a seizure, and that diabetes is different from elevated blood sugar, and that hypertensive disorder is different from elevated blood pressure. But we did not have the right definitions and reproducible guidance to enable a thoroughgoing application of the definitions so that each meaning could clearly be placed where it belongs.

It now appears that a better distinction is between continuants and occurrents, and in particular a definition of disease (not disorder) that identifies it as a kind of continuant potentiality. A continuant is something that exists at any single point in time, and continues across time. A potentiality is something that is not necessarily manifest but always has a real potential to be manifest – with a probability above that of random chance. Epilepsy is a continuant potentiality for being manifest by a seizure. Hypertensive disorder is a continuant potentiality for being manifest by high blood pressure. On the other hand, a fractured femur is just a continuant – the fracture is just there, and once the fracture is healed it is not a fracture anymore. Therefore we don't need two codes for fractures – only one. But we do need a code for epilepsy that is different from the code for seizure, and a code for migraine headache disorder (the continuant potentiality to be manifest by a migraine headache) that is different from the code for migraine headache itself.

These definitions and distinctions may need to be thoroughly applied to the clinical findings hierarchy, with elimination of dual codes where there is only one continuant, and creation and linking of dual codes when there is a continuant potentiality separate from its manifestation.

4.3 Event, state, process and function

In SNOMED CT clear distinctive criteria between event, process, state, and function are still missing. This would, however, mean a major effort of alignment with an upper ontology. Unfortunately, current upper ontologies (e.g. BFO, DOLCE) do not coincide in this respect.

4.4 Interprets and has-interpretation

The model of “decreased muscle tone” with INTERPRETS = *muscle tone (observable entity)* and HAS-INTERPRETATION = *decreased* is illustrative because it shows that findings are medical judgments that interpret observable entities (which are qualities of entities in the world). The problem is that decreased muscle tone can be thought of as a kind of muscle tone, and it would appear to be easier to define decreased muscle tone in the following way:

Decreased muscle tone IS-A muscle tone and HAS QUALITY decreased.



4.5 Recurrent

Does “recurrent” mean that the patient currently has the disorder, or does it just imply that the patient has been having multiple episodes but doesn't currently have the disorder? A clear set of definitions will be required.

4.6 Non-morphological morphologies

The traumatic morphologies are subdivided according to extent, number, mechanism, morphology, intention (accidental, intentional, self-inflicted) and timing. Most of these are not really morphological distinctions, and these values do not really belong in morphology. Using morphology alone to define injury is inadequate. Injury needs to be defined according to the cause, not the morphology. However, many morphologies are inherently traumatic (such as laceration), so disorders that are defined in terms of a traumatic morphology should be classifiable with disorders that are defined in terms of a traumatic cause. Sometimes the traumatic morphology implies a particular traumatic cause, and vice versa, and in these cases we need an appropriate mechanism for recognizing them as equivalent. GCIs (general concept inclusions), also known as “necessarily” conditions, may be one way to accomplish this.

4.7 Hereditary disease, familial disease, congenital disease

The disorder hierarchy contains a grouper called “hereditary disease”, with synonyms “genetic disease” and “inherited disease”. Obviously some genetic diseases are not inherited, so this concept (“hereditary disease”) needs to be retired and replaced with a concept called “genetic disease.” Likewise the disorder hierarchy contains a grouper called “familial disease”. The subtypes of this concept need to be critically examined to see whether they are in fact types of genetic disease; if they are, they should be moved to a genetic disease hierarchy, leaving only diseases that are “familial but not genetic (not known to be genetic)” in this category.

It has been suggested that phrases containing “congenital” be replaced by phrases containing “present at birth”. The name is clear but introduces the word “present”, which creates confusion between findings and situations.

It has been pointed out that we already have an attribute called PATHOGENESIS designed to go with a value hierarchy under *pathogeneses (qualifier value)*, including values like hereditary, familial, single gene defect, etc. Perhaps this attribute should be introduced into the approved list.

4.8 Fetal, perinatal, neonatal

It is often difficult to sort out the differences between fetal, perinatal and neonatal disorders. At present there are some disorders that use FINDING SITE *Fetal structure* and others that use OCCURRENCE *fetal period*. For most of these disorders, both are true. The decision to use one, or the other, or both, can be obviated once we adopt a description logic that allows “also necessarily” conditions (so-called General Concept Inclusions or GCIs). Meanwhile the best approach is to model both when both are true.

In addition, we need clear definitions of fetal, perinatal and neonatal to be added to this style guide.



5 Changes and historical notes

5.1 ONSET and COURSE retired

In earlier releases, there were two attributes named ONSET and COURSE. These were retired because they could not be used reproducibly. While ONSET was intended to specify the rapidity of onset or the temporal pattern of presentation for a given condition, it was easily confused with the attribute COURSE used to represent the duration of a condition. There was not consistent agreement between observers making this distinction.

5.2 EPISODICITY no longer modeled in active content

EPISODICITY originated in the National Health Service Clinical Terms Version 3 where it was used not to specify the first episode of a disease for a patient but rather, the first time a patient presented to their general practitioner (GP) for a particular disorder. A first episode of asthma was not intended to represent the first time a patient had asthma, but rather the first time a patient presented to their GP with asthma. EPISODICITY has been removed from existing concepts and is no longer used in pre-coordinated definitions. It can still be used in post-coordination as a qualifier.