Specific Disorder Modeling

Ischemic disorder

Ischemic disorders are defined by a morphology of ischemic structural change. This need not be permanent, but it is assumed that all ischemia results in some structural alterations at the molecular level.

Ischemic heart disease

Ischemic heart disease includes myocardial infarction, myocardial ischemia (without infarction), angina, and other disorders of the heart that have ischemic structural change (reversible or non-reversible) as a defining characteristic.

Coronary arteriosclerosis can, of course, be present without causing ischemia, so coronary arteriosclerosis is not a subtype of ischemic heart disease.

Likewise, there are causes of myocardial ischemia and infarction other than coronary arteriosclerosis, so ischemic heart disease is not a subtype of coronary arteriosclerosis.

Arrhythmia

Cardiologists noted confusion in the placement of Conduction disorder of the heart (disorder) as a broad grouping that subsumed arrhythmias and heart blocks. In common usage arrhythmia refers to a broad set of conditions that include conduction disorders, under which are heart blocks. The concept Cardiac arrhythmia (disorder) is a parent of Conduction disorder of the heart (disorder), and the active referent of the inactive concepts named dysrhythmia or arrhythmia.

For example,

- Arrhythmias, like 72654001 | Supraventricular arrhythmia (disorder), are under 698247007 | Cardiac arrhythmia (disorder).

Conduction disorders include heart block, AV block, bundle branch block, conduction delay, and conduction defect, like 418341009 | Atrioventricular conduction disorder (disorder). Other arrhythmias were moved from under 44808001 | Conduction disorder of the heart (disorder) and placed under 698247007 | Cardiac arrhythmia (disorder).

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 116676008 | Associated morphology (attribute) = 49755003 | Morphologically abnormal structure (morphologic abnormality). For procedures, use 405816004 | Procedure morphology (attribute).

Functional lesions should not be modeled using values from the Morphologically abnormal structure hierarchy.
Trauma, injury

The word *trauma* has multiple meanings. The first is physical damage to the body (versus emotional trauma). We assume *trauma* means physical damage unless accompanied by words that make clear it is emotional.

Injury is not synonymous with trauma since injuries, caused by stroke, drowning, or toxins, for example, are non-traumatic. Thus the concept, 417746004 |Traumatic AND/OR non-traumatic injury (disorder)|.

**Traumatic injury (disorder)**

417746004 |Traumatic injury (disorder)| is defined as any disorder with a morphology of 19130008 |Traumatic abnormality (morphologic abnormality)|.

Friction injury, abrasion

An injury due to friction can be represented using 400152004 |Friction injury (morphologic abnormality)|, in which case it will not classify as a kind of wound.

For example,

- 47222000 |Friction injury of tooth (disorder)|
- 400068007 |Mechanical irritation (morphologic abnormality)|

However, most disorders that are named abrasion imply that skin or other tissue has been abraded (scraped or worn away). Thus, they are also considered wounds and will correctly classify as wounds after assigning the correct morphology, 400061001 |Abrasion (morphologic abnormality)|.

For example,

- 211039006 |Abrasion, chest wall (disorder)|

Rupture

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. 239731008 |Rupture of lateral collateral ligament of knee (disorder)|). *Rupture*, when applied to an internal organ, may be either traumatic or spontaneous (e.g. 4240001 |Rupture of aorta (disorder)|, 88294009 |Rupture of ovary (disorder)|, etc).

*Rupture* has the subtype morphologies, 415747007 |Traumatic rupture (morphologic abnormality)| and 125672000 |Nontraumatic rupture (morphologic abnormality)|. It is important to make this distinction, at a minimum, in order to support queries related to the effects of trauma. Modelers choose:

415747007 |Traumatic rupture (morphologic abnormality)| as the value of associated morphology for concepts involving anatomical sites, such as muscles and tendons, where trauma is involved (in the absence of a specific lesion).

For example,

- 209765005 |Rupture of tendon of thumb (disorder)|

125671007 |Rupture (morphologic abnormality)| as the value of associated morphology for concepts involving anatomical sites, such as internal organs, where rupture may be traumatic or spontaneous.

For example,

- 46126003 |Rupture of artery (disorder)|

125672000 |Nontraumatic rupture (morphologic abnormality)|, when it is stated as such.

For example,

- 268002004 |Non-traumatic tendon rupture (disorder)|

Traumatic disorders

If an *injury* or *traumatic disorder* does not have a morphology which is a sub-class of 19130008 |Traumatic abnormality (morphologic abnormality)|, then an additional relationship group is added to express this relationship. The relationship is only required for traumatic injury concepts.

These disorders often have an FSN prefixed by *injury* or explicitly prefixed by *traumatic*.

Examples of FSNs,
• Injury of brachial plexus trunk (disorder)

Most traumatic concepts have the relationship groups, Finding site and Associated morphology.

For example, 721347007 Fracture of third cervical vertebra (disorder) has

• Finding site (attribute) = Bone structure of C3 (body structure)
• Associated morphology (attribute) = Fracture (morphologic abnormality)

Usually, as in the above example, the morphology is a sub-class of 119130008 | Traumatic abnormality (morphologic abnormality) | and thus auto-classifies appropriately. However, some morphologies are not currently sub-classes of Traumatic abnormality, and a traumatic variant does not exist. These morphologies will not auto-classify as an injury.

For example, 722628000 Traumatic hemorrhage of subdural space of infratentorial region (disorder)

• Associated morphology (attribute) = Hemorrhage (morphologic abnormality) and
• Finding site (attribute) = Structure of subdural space of infratentorial region (body structure)

In these cases an additional relationship group is added.

For example,

• Finding site (attribute) = Structure of subdural space of infratentorial region (body structure) and
• Associated morphology (attribute) = Traumatic abnormality (morphologic abnormality)

Tumor vs. neoplasm

The word tumor has two primary meanings, a mass, regardless of whether it is neoplastic or not, or a neoplastic mass. Neoplasm is preferred since it is less ambiguous than tumor.

For example,

• 92385005  Benign neoplasm of small intestine (disorder)

Primary vs secondary neoplastic disorders

SNOMED CT follows ICD-O and ICD-10, where secondary malignant neoplasm of (site X) is uniformly interpreted to mean that metastasis has occurred to site X.

For example,

• 94521000  Secondary malignant neoplasm of rib (disorder)

For concepts that describe metastasis from a malignant neoplasm, SNOMED CT explicitly uses the word from.

For example,

• 315006004  Metastasis from malignant tumor of lung (disorder)

In SNOMED CT, metastases are modeled with two relationship groups, each with an appropriate morphology and site.

For example,

• 712849003  Primary malignant neoplasm of prostate metastatic to bone (disorder):
  • IS A (attribute): Disease (disorder)
  • Finding site (attribute): Bone structure (body structure) and Associated morphology (attribute): Neoplasm, metastatic (morphologic abnormality)
  • Associated morphology (attribute): Malignant neoplasm, primary (morphologic abnormality) and Finding site (attribute): Prostatic structure (body structure)

Neoplasia

When modeling neoplasia, distinguish structure from process. Do not use neoplasia in the FSN to identify the structure (even though it implies it). Use 126537000  Neoplasm of bone (disorder), not neoplasia of bone.

Neoplastic disease refers to the process of neoplasia, leading to the formation of a neoplasm.

Where the definition is primary, the associated morphology: 86049000  Malignant neoplasm, primary (morphologic abnormality) is used.

Where the definition is primary or secondary, the morphology: 367651003  Malignant neoplasm of primary, secondary, or uncertain origin (morphologic abnormality) is used.
Where the concept expresses a specific morphology the FSN always contains the word *primary*.

For example,

9541000119105 Primary adenocarcinoma of gallbladder (disorder)
- Finding site (attribute): Gallbladder structure (body structure)
- Associated morphology (attribute): Adenocarcinoma (morphologic abnormality)

**Neoplasm versus hamartoma**

A neoplasm is defined as a growth of tissue no longer under normal control. A hamartoma is defined as a benign, self-limiting growth of disorganized mature cells normally found in the region, representing faulty development. SNOMED CT has disorder (and morphologic abnormality) concepts and subtypes representing neoplasia, hamartomas, and tumors.

The SNOMED CT concept 399981008 Neoplasm and/or hamartoma (disorder) has six subtypes:
- angiomatosis
- hamartoma
- hemangioma
- lymphangioma
- melanocytic nevus
- neoplastic disease

The SNOMED CT concept 400177003 Neoplasm and/or hamartoma (morphologic abnormality) also has six subtypes:
- angiomatosis
- blood vessel tumor
- hamartoma
- lymphatic vessel tumor
- melanocytic nevus
- neoplasm

**Nevus**

The word *nevus* has many different meanings. The differences are generally based on answers to the following questions:

- Is it necessarily on the skin? Or can it be located in mucosal sites or other sites?
- Is it necessarily visible? Or can it be in internal locations such as gastric mucosa, etc?
- Is it necessarily present at birth? Or can it occur later in life?
- Is it necessarily dark and made of melanocytes? Or can it be non-pigmented, or made of other types of cells?
- Is it necessarily made of tissue that is normally present at the site? Or can it be ectopic?
- Does it exclude benign neoplasms?

Some common meanings of nevus based on some combinations of answers to the questions are as follows:

- A birthmark, that is, any visible spot on the skin or oral mucosa present since birth, regardless of tissue of origin, excluding benign neoplasms.
- Any benign cluster of melanocytes, regardless of location, and regardless of pigmentation, whether present since birth or appearing later.
- 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 CT FSN containing the word *nevus* may be ambiguous. For example, the term *vascular nevus* may mean:

- Congenital blood vessel tumor in the skin
- Congenital blood vessel hamartoma or neoplasm that is visible somewhere (not only the skin, but also the mucosa, whether visible externally or not)
- Congenital blood or lymphatic vessel tumor in the skin
- Congenital blood or lymphatic vessel hamartoma or neoplasm that is visible somewhere
- 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 need to inactivate the description or the concept.

**Infectious disease and inflammatory disorder**

The concepts 40733004 Infectious disease (disorder) and 1281390000 Inflammatory disorder (disorder) are siblings; not all infectious diseases are inflammatory.
Infectious disease has a Causative agent relationship to the appropriate organism.

For example,

- The causative agent for 4834000 |Typhoid fever (disorder)| is Salmonella Typhi (organism)

If inflammation (or a subtype) is always present for an infectious disease, then the associated morphology attribute, with the appropriate inflammatory morphologic abnormality or one of its subtypes, is also included.

128139000 |Inflammatory disorder (disorder)| has an 116676008 |Associated morphology (attribute)| of 409774005 |Inflammatory morphology (morphologic abnormality)| or one of its subtypes.

For example,

- The associated morphology for 81077008 |Acute rheumatic arthritis (disorder)| is Acute inflammation (morphologic abnormality)

Infectious/inflammatory disorders may or may not have the pathological process attribute.

For example, 37298004 |Acute suppurative arthritis caused by bacteria (disorder)| does have a pathological process attribute, while 236624004 |Acute cystitis - culture-negative (disorder)| does not.
Pneumonia vs. Pneumonitis

The terms _pneumonia_ and _pneumonitis_ are often used interchangeably. In SNOMED CT, pneumonia should be used for infectious causes, and pneumonitis should be used for noninfectious causes.

Pneumonia is a type of pneumonitis, as inflammation is present in both. The distinguishing feature between the two disorders is the presence of infection in pneumonia. Pneumonia should have a pathological process of infectious process, pneumonitis should not.

Consolidation is a feature of most forms of pneumonia. It may not be a feature of some atypical pneumonias, e.g. mycoplasma pneumonia.

Except as noted above, the morphologic abnormality for 233604007 |Pneumonia (disorder)| is 707496003 |Inflammation and consolidation (morphologic abnormality)|.

The morphologic abnormality for 205237003 |Pneumonitis (disorder)| is 409774005 |Inflammatory morphology (morphologic abnormality)|.

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.

Bacterial disorders with organism and/or toxin

In modeling some bacterial disorders, there will be situations where either the organism or the toxin (substance), or both values, are required for the causative agent attribute. The decision is often determined by whether or not the bacteria are considered endotoxins or exotoxins. The most common exotoxins are:

- Botulinum Toxin
- Enterotoxin
- Cholera Toxin
- Diphtheria Toxin
- Tetanospasmin

Exotoxins are more lethal in comparison to endotoxins, but there are vaccines against many exotoxins whereas there are no vaccines against endotoxins. There can be instances where an infection is present but the disease-causing toxins are not; in this case, model the concept only with the organism and not the toxin substance.

Example,

276202003 |Infection caused by Clostridium tetani (disorder)| is modeled with a causative agent of 30917009 |Clostridium tetani (organism)| only.

In the situation where a disease is caused by both the infection and the associated toxin, model with both the causative agent and the toxin substance.

Example,
Hypersensitivity disorders

473010000 Hypersensitivity condition (disorder) is a primitive concept. It subsumes 473011001 Allergic condition (finding) and 609405001 Pseudoallergic condition (disorder).

473010000 Hypersensitivity condition (disorder) is a direct descendant of 404684003 Clinical finding (finding).

473011001 Allergic condition (finding) and 609405001 Pseudoallergic condition (disorder) are both primitive concepts. Each has three descendants representing:

- Diseases/disorders: abnormal structures
- Processes: allergic and nonallergic hypersensitivity (pseudoallergic) reactions
- Dispositions: propensities to develop allergic and nonallergic hypersensitivity (pseudoallergic) reactions; they do not have pathophysiologic manifestations prior to allergic and nonallergic hypersensitivity (pseudoallergic) processes, i.e. reactions

Diseases/disorders and reactions, but not dispositions, are defined by underlying pathological processes.

Pathological process (qualifier value) hierarchy

In order to fully describe the full range of hypersensitivity responses, there are qualifier values in the Pathological process (qualifier value) hierarchy. (See also Qualifier Value page).

Disorders of immune function

Allergic reaction

419076005 Allergic reaction (disorder) has a causative agent (attribute) of 105590001 Substance (substance).

Figure 1: Allergic reaction (disorder) modeling

Modeling 414029004 Disorder of immune function (disorder) with 769247005 Abnormal immune process (qualifier value) allows allergic and autoimmune disorders to correctly classify as disorders of immune function.

Allergic and nonallergic hypersensitivity (pseudoallergic) disease

Allergic and nonallergic hypersensitivity (pseudoallergic) diseases represent manifestations of pathologic processes that result in abnormal structures. Modeling an allergic and nonallergic hypersensitivity (pseudoallergic) disease includes the following relationship groups:

116676008 Associated morphology (attribute) and 363698007 Finding site (attribute) representing the abnormal structure

Pathological process: 472963003 Hypersensitivity process (qualifier value), or one of its descendants
Figure 2: Allergic and nonallergic hypersensitivity (pseudoallergic) disease model

For example,
Allergic and nonallergic hypersensitivity (pseudoallergic) disposition

Allergic and nonallergic hypersensitivity (pseudoallergic) dispositions are propensities to develop allergic and nonallergic hypersensitivity (pseudoallergic) reactions; they do not have pathophysiologic manifestations prior to reactions. They are considered clinical findings, not disorders. This further distinguishes them from allergic and nonallergic hypersensitivity (pseudoallergic) reactions.

Figure 3: Allergic and nonallergic hypersensitivity (pseudoallergic) disposition model
For example,

**Figure 4: Allergic disposition (finding) modeling**

**Allergic and nonallergic hypersensitivity (pseudoallergic) reaction**

Allergic and nonallergic hypersensitivity (pseudoallergic) reactions are adverse reactions and allergic conditions. Like diseases/disorders, they are defined by underlying pathological processes.

**Figure 5: Allergic and nonallergic hypersensitivity (pseudoallergic) reaction model**

For example,
Contact hypersensitivity

Contact hypersensitivity occurs when the skin, or a mucous membrane, comes in contact with a substance. The resulting response may be immune mediated (allergic) or nonimmune (irritant). Allergic responses may be immunoglobulin E mediated, as in 402304007 Allergic contact urticaria (disorder), or nonimmunoglobulin E mediated, as in 238575004 Allergic contact dermatitis (disorder). Contact hypersensitivity concepts are modeled as 473010000 Hypersensitivity condition (disorder), using the pathological processes illustrated below:

For example,
Intolerance to substance

An intolerance is the propensity to develop an adverse reaction to a substance. The adverse reaction may be associated with various pathological processes, but specifically excludes hypersensitivity reactions.

It may be difficult to define the pathological process and to associate the substance with the propensity to develop a reaction. Consequently, 47429007 Associated with (attribute) is used to model intolerance to substances.

Figure 6: Stated view of |Intolerance to substance (finding)| model

For example, Iatrogenic
Adding to the iatrogenic disorder hierarchy is discouraged. An iatrogenic disorder should remain as a primitive concept. It should be a child of 116223007 Complication (disorder). iatrogenic is not available as a value of 263547006 Pathogenesis (attribute).

Subtypes of 12456005 iatrogenic disorder (disorder) that have FSNs without the word iatrogenic, should be remodeled by inactivating the IS_A relationship to 12456005 iatrogenic disorder (disorder).

Concepts with iatrogenic in the FSN should be modeled with an IS_A relationship to 12456005 iatrogenic disorder (disorder).

For example,

![Figure 7: Inferred view of 232081005 |iatrogenic glaucoma (disorder)| using IS_A 12456005 |iatrogenic disorder (disorder)|](image)

**Congenital**

The concept 66091009 Congenital disease (disorder), means present at birth. Though the word congenital may be applied to genetic disorders, the term genetic is preferred for those disorders.

Model congenital disorders with the following relationship group:

Associated morphology = X (morphologic abnormality)
Occurrence = congenital (qualify value)
Finding site = X (body structure)
Pathological process = pathological development process (qualifier value)

- The relationship, pathological process = pathological development process (qualifier value), is required when there is a morphologic abnormality.

The following guidelines apply:

A disorder with the word congenital in the FSN should classify under 66091009 Congenital disease (disorder). Do not make a stated assertion that the parent is 66091009 Congenital disease (disorder); instead, allow the classifier to infer this relationship.

Occurrence should be in the same relationship group as Associated morphology and Finding site because the morphology is located at the site and the occurrence applies to the combined morphology-site pair.
For general congenital anomaly disorder grouper concepts, such as 9904008 [Congenital anomaly of cardiovascular system (disorder)], the preferred value for Associated morphology (attribute) is 49755003 [Morphologically abnormal structure (morphologic abnormality)] with the additional Pathological process relationship.

Congenital X (morphologic abnormality) terms should not be used if there is a non-congenital supertype of that morphologic abnormality.

- For example, 102283003 [Congenital misalignment (morphologic abnormality)] should not be used as a value because there is a non-congenital supertype concept of 399898009 [Misalignment (morphologic abnormality)].

Previously, when modeling congenital disorders, the Associated morphology = congenital X (morphologic abnormality) was used. Congenital X (morphologic abnormality) concepts are being inactivated. When revising concepts that contain a [Congenital X (morphologic abnormality)], the congenital morphology is replaced with the general morphology. For example, 385297003 [Congenital deformity (morphologic abnormality)] should be replaced with 6081001 [Deformity (morphologic abnormality)]. The relationship group should also include Occurrence and Pathological process attributes as per guidance.

### Congenital versus acquired

Generally, disorders may be either congenital or acquired. The acquired form should only be used when needed to differentiate from the congenital form, but not when a concept refers to a finding that is only acquired.

**Congenital disorders** are modeled using 246454002 [Occurrence (attribute)] of 255399007 [Congenital (qualifier value)]. If the FSN does not include congenital, it should not be modeled as congenital. The precise meaning of the FSN should be followed (e.g. many hereditary disorders have congenital appearances).

For example, 35387008 [Congenital aphakia (disorder)] is modeled with 246454002 [Occurrence (attribute)] of 255399007 [Congenital (qualifier value)]

Acquired disorders are those which originate and manifest after birth. The disorders are associated with a period of life, as opposed to a specific process or structure. All diseases (disorders) that occur after birth are considered acquired.

There is a Period of life concept, 767023003 [Period of life beginning after birth and ending before death (qualifier value)].

All concepts that explicitly state acquired in the FSN or in a synonym should be modeled with Occurrence = 767023003 | Period of life beginning after birth and ending before death (qualifier value) | that is if an Occurrence relationship does not already exist, i.e. there is not already another period of life that is a value for Occurrence, which would infer the parent. This allows many primitive concepts to be sufficiently defined.

For example, 240253004 [Acquired abduction deformity of foot (disorder)]
Hereditary

It may be a challenge to classify a condition as a Hereditary disease (disorder). Hereditary requires case-by-case definition; it cannot be applied to broad categories. Nevertheless, the names by which many diseases are known include the term, and it is permitted, as long as it does not introduce ambiguity.

Familial

The term familial may also be ambiguous when used for broad categories. It may mean that the disorder is found in higher proportions in the immediate or extended family compared to other groups. Or, it may mean there is a possibility of a disease being inherited. It may be used; however, it may require clarification of meaning from the requestor. It should not be used as a synonym for genetic.

Developmental

Developmental is a useful label for disorders that affect developing structures or functions that may occur during pre- or postnatally. They may be present at birth or develop later.

Genetic, developmental, congenital, and physical

The following figure shows the structure of genetic, developmental, and congenital categories, along with non-genetic, non-developmental, and postnatal categories. A dimension, called extrinsic physical force, is included to distinguish deformations from malformations. The sections of the diagram represent categories formed from the combination of the dimensions, each of which represents the answer to one of the following questions:

- Is it genetic or not?
- Is it developmental or not?
- Is it present at birth or not?
- Is it due to an extrinsic physical force or not?

According to the American Medical Association, the periods of life in the postnatal period include all periods after birth including the neonatal or immediate postpartum period. It may be challenging to differentiate a congenital disorder from a neonatal disorder. A condition may be present at birth, i.e. congenital; however, clinical manifestations may take longer to appear, i.e. during the neonatal period (e.g. 14332004 Alloimmune neonatal neutropenia (disorder)).
Figure 8: The relationships of genetic, congenital, developmental, and acquired disorders

Explanation of Figure

The sections with 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 sections with blue crossing lines represent congenital malformations; they may be either genetic or non-genetic. For example, congenital infectious malformations.

The red circle represents congenital genetic malformations.

The blue sections represent acquired, i.e. disorders that are non-genetic and not present at birth. For example, Vitamin D deficiency (rickets) in children is a non-genetic, non-congenital, developmental malformation.

The white sections represent genetic congenital or genetic postnatal disorders. For example, Huntington's disease is a genetic disease that is neither congenital nor developmental. The gene defect is present at birth, but the disease does not manifest until adulthood.

Arrows leading from the sections point to examples of disorders for the category.

Malformation, deformation, anomaly

A deformation is a structural abnormality that is due to an extrinsic physical force. Newly created concepts representing a deformity should be considered disorders.
Malformations are structural abnormalities that result from intrinsically disordered development. The word *anomaly* is, by itself, ambiguous. It may mean: any abnormality including non-structural ones; malformation; both malformation and deformation. Concepts with the word *anomaly* must be evaluated for ambiguity.

For example,

- Congenital anomaly of <x structure> is definitely structural, but is not the same as congenital malformation (structural abnormality due to intrinsically disordered development present at birth). Therefore, it can be regarded as having the more general meaning of structural abnormality present at birth.

### Hematologic, lymphatic

There is more than one meaning of *hematologic*. A definition based on hematological system *structure* includes 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 *clinical usage by hematologists* is broader. Disorders of hemostasis and thrombosis are often 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 is used to define a *reference set* that includes disorders of blood and blood forming organs, as well as disorders of hemostasis and thrombosis, depending on what is intended.

### Hematologic disorders, lymphoid and myeloid neoplasms

*Hematologic disorders* may refer to disorders of: hematopoietic cell origin; blood forming organs (bone marrow, lymph nodes, spleen, thymus, and other lymph tissues); cellular components of blood; or function of hemostatic and thrombotic systems.

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

The morphology (neoplastic diseases, at a minimum, include those morphologies covered by neoplasms in the International Classification of Diseases for Oncology, ICD-O).

For example,

- 118599009 | Hodgkin's disease (disorder) | has 128930002 | Hodgkin lymphoma - category (morphologic abnormality). The body site involved (especially specific lymph node groups or skin sites).

For example,

- 400122007 | Primary cutaneous T-cell lymphoma (disorder) | has Finding site, skin structure (body structure)

For some disorders, like T-cell lymphomas, and plasma cell and immunosecretory disorders, it is important to distinguish those defined by morphology, site, or manifestation.

T-cell lymphomas can be subcategorized according to the primary site, a lymph node, the skin, or other extranodal site. This means that a *site* of lymphoid structure cannot be the defining characteristic of the parent concept *T-cell lymphoma*. Its defining attribute should be morphology alone. Plasma cell and immunosecretory disorders (e.g. monoclonal gamopathy, heavy chain disease, Waldenstrom’s macroglobulinemia) are defined by their manifestations, i.e. the type of monoclonal protein they secrete. Others (e.g. myeloma, plasmacytoma) are defined by their morphology, regardless of whether or not they are secretory.

Immunosecretory disorders may have a morphology of *plasma cell neoplasm*, even though no mass has 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 morphologies, but without a site. Leukemias and myelodysplastic syndromes are modeled with Finding Site, bone marrow structure (body structure).

### Coagulation, hemostasis, thrombosis

There is more than one meaning of *coagulation*. A broad meaning, to stop bleeding, is better described as *hemostasis*. A more narrow definition, limited to the formation of the fibrin clot, might exclude certain components of hemostasis (e.g. 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 their bodies are able to stop bleeding. Therefore, *coagulation disorders* are kinds of *hemostatic disorders*.

### Hernias

Hernias involve two body structures, one is the hernial opening and the other is the herniated structure. When modeling hernias, use two role groups to represent the body structures and the associated morphology for each site. If the herniated structure is not explicit, use the supertype concept for the finding site.

For example, the concept 50063009 | Femoral hernia (disorder) | is modeled with finding site = 19203006 | Structure of abdominopelvic viscus (body structure) to represent the herniated structure.
Osteoarthritis

396275006 [Osteoarthritis (disorder)] is regarded as a degenerative disease, despite the -itis in its name. Because of this, 396275006 [Osteoarthritis (disorder)] is not a subtype of arthritis in the disorder hierarchy but instead, the more general, 399269003 [Arthropathy (disorder)]. Arthritis is inflammatory by definition, but osteoarthritis has a subclass in the medical literature called non-inflammatory osteoarthritis. In fact, according to many authoritative sources, osteoarthritis is usually regarded as a non-inflammatory disease, and therefore it is not strictly a subtype of arthritis.

Structuring the hierarchy this way does not imply that there are no cases of osteoarthritis with inflammation, nor does it rule out inflammation as an etiologic or contributory factor. It is well established that inflammation often occurs in osteoarthritis, and treatment with anti-inflammatory agents has been more effective than pure analgesics in many cases. Despite growing evidence of the role of inflammatory cytokines in osteoarthritis, it is not always necessarily an inflammatory disorder of the joint.

Multisystem disorders

Multisystem disorders are often rare conditions. There may be limited information about such disorders, so they should be carefully modeled.

When determining parent concepts:

A multisystem parent concept should be included.

Genetic or inherited disorders should be modeled in the same way as other genetic and inherited disorders.

The manifestations of the disorder must always necessarily be true before assigning the relevant parents.

Attributes must also always necessarily be true.

For example,

- In 702410002 Iris coloboma with ptosis, hypertelorism, and mental retardation (disorder), the coloboma of the iris is not always present. This would not be explicitly modeled in the relationships.

Some multisystem disorders can be named by their manifestations. The FSN should be descriptive rather than just a list of names.

For example,

- 717909004 Bilateral microtia with deafness and cleft palate syndrome (disorder)

A multisystem disorder with an eponymous syndrome name should be included as a synonym only.

Mental health disorders
Dependence-related concepts which express the current existence of abuse are acceptable.

For example,

- 191816009 Drug dependence (disorder)

Dependence-related concepts which express the pattern as either continuous or episodic are not acceptable.

Unacceptable patterns,

- X with single episode
- X with multiple episodes
- Current episode of X
- First episode of X
- X with continuous pattern

Unacceptable legacy concepts,

- Drug abuse, continuous (disorder)
- Episodic drug abuse (disorder)

Concepts describing full or partial remission are acceptable but not the phase of the remission. The patterns are:

- X in full remission
- X in partial remission

For example,

- 46244001 Recurrent major depression in full remission (disorder)
- 5703000 Bipolar disorder in partial remission (disorder)

Unacceptable examples,

- X in early full remission
- X in sustained full remission
- X in sustained partial remission

Conditions with associated symptoms should be expressed and modeled like combined disorders. Co-occurrent and Due to, and Due to situations are acceptable, but not simple Co-occurrent.

For example,

- 724665004 Perceptual disturbances co-occurrent and due to sedative withdrawal (disorder)

Concepts containing X without Y are considered on a case-by-case basis.

For example,

- 724735003 Oppositional defiant disorder without chronic irritability-anger (disorder)

Unacceptable example,

- Bipolar type II disorder with current episode moderately depressive without psychotic symptoms

Death

Death is an event, not a disorder.

Sudden cardiac death

Sudden cardiac death is a term used in clinical practice. It refers to an arrhythmia that results in sudden loss of cardiac function which, if not quickly reversed, will lead to actual death. The FSN Sudden cardiac death (disorder) is modeled as a subtype of 127337006 [Acute heart disease (disorder)]. It should not be classified as death. Individuals with sudden cardiac death have not necessarily been declared dead and are frequently revived. It is regarded as a subtype of cardiac dysrhythmia.

Poisoning

When modeling poisoning disorders, ensure that the disorder being described is caused by the substance or active ingredient in the product selected as the causative agent (attribute) value. Do not add poisoning disorders if the causative agent is a product constituent (e.g. adjuvant, carrier, preservative, flavoring, stabilizer, or other inactive ingredient) that cannot be identified as the causative agent.

Vaccine-related overdose
For the January 2020 Release, vaccine-related overdose concepts in the Clinical Finding/Disorder hierarchy were inactivated. They were replaced with concepts in the Event hierarchy, see 788094008 |Excessive dose of vaccine administered (event)| and subtypes.

When authoring, determine whether the concept describes an overdose, which is a disorder, or the administration or ingestion of an excessive dose, which is an event.

Vaccine-related poisoning
Vaccine-related poisoning concepts have been inactivated.

Obstruction
Since an obstruction describes blockage inside the space of a tubular structure, the Finding site of obstruction concepts should be a value from the 113342003 |Structure of lumen of body system (body structure)| subhierarchy. For example, when modeling gastrointestinal tract obstruction concepts, the Finding site value should be a value from the 432899004 |Structure of lumen of gastrointestinal tract (body structure)| hierarchy as the site obstructed is the lumen of the tract. At present, some but not all anatomy content exists to support this model for tracts, ducts and blood vessels beyond the gastrointestinal tract but is expected in the future.

Combining Morphologic Abnormalities to Prevent Multiple Groups
When modeling a concept requiring two role groups with the same body structure but two different morphologies (because a combined morphology does not exist), then those morphologic abnormalities can be combined to create a single |(morphologic abnormality)| concept. Keep the newly-created morphologic abnormality concept primitive as all morphologic abnormality concepts are primitive.

<table>
<thead>
<tr>
<th>Example: Modeling</th>
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<tbody>
<tr>
<td>Concept</td>
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<td>400067002</td>
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Another example is 1076491000119102 |Nontraumatic complete rupture of muscle or tendon structure of rotator cuff of left shoulder (disorder)|. If this disorder had the same finding site of |Structure of rotator cuff of left shoulder (body structure)| with two different morphologic abnormalities of |Nontraumatic rupture| and |Complete rupture|, then those two morphologic abnormality concepts can be combined to create a single, primitive, morphologic abnormality concept of |Nontraumatic complete rupture (morphologic abnormality)|. This will prevent modeling with two relationship groups.

Instead of this: