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SNOMED CT OWL Guide

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This document is a guide to the use of the Ontology Web Language (OWL) in SNOMED CT. It includes detailed information about structure, content and use of OWL reference sets. These OWL reference sets are used to distribute ontology reference information and the axioms that represent the formal logical definitions of SNOMED CT concepts.

Web browsable version: http://snomed.org/owl

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1. Introduction

Background

SNOMED CT is a clinical terminology with a comprehensive coverage of wide clinical specialties and requirements. The development and maintenance of SNOMED CT relies on Description Logics (DL) and its reasoning services. The stated relationship file has been used to represent DL definitions. However, it is unable to fully represent semantics of the DL expressivity due to the limitation of its relational structure.

The new OWL refsets are designed to replace the stated relationship file and it represents the DL definitions for SNOMED CT content by following the international standard of OWL 2 Web Ontology Language.

Purpose

The purpose of this document is to define and describe the OWL Reference Set.

Scope

The document presents the specification of OWL refsets. It also documents the background, design considerations, quality assurance, and distribution format of the OWL refsets.

The implementation of the OWL refsets in the applications, and transformation of it to an OWL ontology, are out of the scope.

Audience

The target audiences of this document include:

- SNOMED CT national release centers;
- SNOMED CT terminology content developers and technical developers;
- SNOMED CT implementers, analysts, and developers of electronic health records and information systems.

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2. Design Considerations for OWL Reference Sets

2.1. Reference Set Type

The |OWL expression type reference set| represents components and their associations to OWL expressions, e.g. definitions specified in OWL expressions for concepts, or information about OWL ontologies of SNOMED CT. This new type of reference set follows the pattern for annotation type reference set, but the content are not mixed with other types of annotations. The new refset supports accurate representation for feeding data into Description Logic reasoners, validation of OWL expressions, and creation of OWL ontologies for SNOMED CT.

2.2. Syntax for the OWL Refsets

The OWL Functional Syntax-Style is the recommended default syntax. The generated OWL ontologies of SNOMED CT may be rendered in any compatible OWL syntax and transformed between syntaxes. The specification of syntax including the BNF grammar published by the W3C can be found at https://www.w3.org/TR/owl-syntax/.

Specializations for SNOMED CT

For the benefits of consistent representation and detection of changes to an expression, the style and rules are recommended though they have no semantic significance. The OWL expressions should be rendered in the form specified by the OWL Functional Syntax-Style but a restricted version may be used for creating and updating the OWL refsets. The following rules should be followed.

Whitespace

OWL Functional Syntax-Style defines that whitespace is a nonempty sequence of space (U+20), horizontal tab (U+9), line feed (U+A), or carriage return (U+D) characters.
The whitespace should only be represented as a single space (U+20) in the OWL refsets.

Comments

OWL Functional Syntax-Style defines that a comment is a sequence of characters that starts with the # (U+23) character and does not contain the line feed (U+A) or carriage return (U+D) characters.
Comments could improve the readability. However, comments should not be included to minimize the size of the OWL refsets and reduce the need to maintain the alignment between an axiom and its comments.

Sorting Order

A standard sort order is very useful to enable fast matching of identical expressions though it has no impact to semantics and is not essential for general purpose use.
Each axiom is a serialization of a tree structure that can be traversed by nodes in the following order:

1. Node that is either a SNOMED CT concept ID or a value of type data property. These nodes are sorted in UTF-8 byte-order (as a sequence of bytes) rather than numbers or strings. The byte-order compares two bytes; one from each sequence, until the values of the bytes are different. Then, the ID or value with the lowest byte value would be sorted as first regardless the length difference of byte arrays;
2. The composite node ObjectSomeValuesFrom(), sorting order by concept id of attribute and then value;
3. The composite node DataHasValue(), sorting order by concept id of attribute and then value.

The Functional-Style Syntax determines the general order of most elements within an OWL axiom. In fact, the order can only be applied to the nodes within EquivalentClasses(), DisjointClasses(), EquivalentDataProperties(), and ObjectIntersectionsOf() for the current SNOMED CT Logic Profile Specification.

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Syntax preference for expression variants

The same semantics can be represented by a single axiom or multiple axioms. When axioms are in the same SNOMED CT module,

- Expression in the OWL refset should be presented as an Equivalent Classes axiom instead of two Subclass axioms where both apply, e.g.

  EquivalentClasses(A C) is recommended. The following two Subclass axioms are not.

  SubClassOf(A C)
  SubClassOf(C A)

- Expression in the OWL refset should be presented as a single Subclass axiom with intersection of Class expressions instead of multiple Subclass axioms where they apply, e.g.

  SubClassOf(A ObjectIntersectionOf(C D)) is recommended. The following two Subclass axioms are not.

  SubClassOf(A C)
  SubClassOf(A D)

2.3. Content for the OWL Ontology Refset

The OWL ontology refset should represent essential information about an ontology, such as the namespaces, ontology URI, ontology version URI, and import statement.
Apart from the standard information for all OWL ontologies, any specific information of an ontology can be included. The OWL ontology refset enables the use of prefixes in the OWL axiom refset.

Namespaces

The namespace declarations cover the standard and SNOMED CT specific prefix names, for all ontologies. The prefix name "sct:" is for SNOMED CT concept identifiers and the namespace URI is http://snomed.info/id/.

The prefix names are associated with SNOMED CT concept 734146004 |OWL ontology namespace (metadata)| as referencedComponentId and examples can be found in table 4-2 OWL Ontology Reference Set example.
The URIs can be represented in full form or using the prefix names. For example, The URI for 64572001 |Disease (disorder)| can be one of the following format.

- http://snomed.info/id/64572001
- :64572001

It is recommended to use the default prefix ":" to minimize the size and improve readability in the OWL refset. The prefix name "sct:" should be declared and used when http://snomed.info/id/ is not the default namespace.

Ontology URI and Version URI

The ontology URI and the version URI together identify a particular version of ontology. According to the convention, an ontology document should be accessible via the ontology URI if it is the current version. Since a release edition contains the current version ontology, the ontology URI alone is sufficient for the OWL ontology refset. The version should be determined from the SNOMED CT release package. The version URI does not need to be included in the OWL ontology refset. For example:

Ontology(<http://snomed.info/sct/900000000000207008>)

SNOMED CT International Edition	http://snomed.info/sct/900000000000207008

However, the version URI should always be included for a standalone ontology file of SNOMED CT to provide accurate version information. The SNOMED CT URI Standard describes how to unambiguously reference a particular version of a SNOMED CT edition. It is also a trivial task to generate the Ontology version URI by the transformation process for a standalone ontology file. For example,

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Ontology(<http://snomed.info/sct/900000000000207008> <http://snomed.info/sct/900000000000207008/version/20180731>)

Edition, Modules and Ontology Import Statement

Each SNOMED CT Edition should be represented as a separate ontology based on the identifier of the most dependent module. The international release is represented by a single ontology and identified by the SNOMED CT core module id as the ontology URI. It includes two modules for terminology content excluding modules for derivatives:

- 900000000000207008 |SNOMED CT core module (core metadata concept)|
- 900000000000012004 |SNOMED CT model component module (core metadata concept)|

The module dependency has specified that 900000000000207008 |SNOMED CT core module (core metadata concept)| depends on 900000000000012004 |SNOMED CT model component module (core metadata concept)|.

The OWL ontology reference set should only have one single active entry of |OWL ontology header| for the ontology declaration. This entry represents the identity of an ontology and the identifier for the ontology URI should be the most dependent module for that particular edition.

A new entry for OWL ontology header should be added with the extension module id and effective time for an extension edition of SNOMED CT. The OWL ontology header for SNOMED CT international release should be inactivated.

The OWL ontology header represents the identity of an ontology. It needs to be updated when the OWL expression refeset represents a different ontology, e.g. ontology of a national extension. Note, the changes to the content, version, and module dependency of an ontology do not require to update the ontology header.

It is possible that modules in SNOMED CT can be directly translated into OWL ontologies, with the module dependency reference set describing how these ontologies are imported. Extensions can import the ontology of SNOMED CT core module since the content in the model component module has already been included. The new OWL ontology header should include the extension module identifier and import statement(s). This approach could avoid accidental changes to the imported ontology. However, the implementation could be more complicated than the representation of an entire edition as a single ontology. Therefore, it is not recommended to use an ontology import statement at the current stage. Instead, each edition will be rendered as one OWL ontology, without any ontology import statement.

2.4. Content for the OWL Axiom Refset

Axioms are statements or propositions which are regarded as being established, accepted, or self-evidently true in the domain. The OWL axioms are in the scope for the OWL axiom refset if they are allowed in the SNOMED CT Logic Profile Specification. Annotation axioms and Class declarations are generally excluded from the OWL axiom refset to avoid duplication to the RF2 files:

- Annotation Axiom - Descriptions can be represented as annotation assertions. They are excluded from the OWL axiom refset to avoid duplication to the RF2 description file.

The transformation from the OWL expression refset to OWL ontology document should process the description file and language refset when descriptions are included. They should be represented by the following annotation properties for a given language refset.

  - rdfs:label - if present, it is RF2 fully specified name
  - skos:prefLabel - if present, it is RF2 preferred term
  - skos:altLabel - if present, it is RF2 acceptable synonym
  - skos:definition - if present, it is RF2 definition

- Declaration
  - Declarations for the built-in entities, e.g. owl:Thing, owl:Nothing, are excluded from the OWL refsets because they are implicitly presented in every OWL 2 ontology.

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- Class and property declarations are excluded from the OWL axiom refset to avoid duplication to the content that is represented by the RF2 concept file. Note, Class and property declarations should be included in cases where an entity type cannot be derived from the existing axiom.

Referenced component for an Axiom

The OWL Axiom Reference Set is designed to cover all logic definitions in SNOMED CT. A concept can be defined by one or more axioms in the same module or different modules. Each axiom is represented by a string in OWL Functional Syntax in the owlExpression field. Each concept is represented by a concept ID in the referencedComponentId in the refset. The following rules should be followed when assigning the referencedComponentId for an axiom:

- If an axiom is in the form of SubClassOf(C D) or EquivalentClasses(C D) where concept C is a precoordinated concept, the concept ID of C should be the referencedComponentId for this axiom.
- If an axiom is in the form of SubPropertyOf(r s) or EquivalentProperties(r s) where attribute r is a precoordinated concept, the concept ID of r should be the referencedComponentId for this axiom.
- If an axiom is in the form of SubClassOf(C D) where concept C is NOT a precoordinated concept and D is a precoordinated concept. This is a General Concept Inclusion (GCI) axiom. Because concept C is a sufficient but not necessary condition for concept D, the concept ID of D should be the referencedComponentId for this axiom.
- If an axiom is in the form of SubObjectPropertyOf(u t) where attribute u is NOT a precoordinated concept and attribute t is a precoordinated concept. The concept ID of t should be the referencedComponentId for this axiom. e.g. SubObjectPropertyOf(ObjectPropertyChain(:r :s) :t)
- If an axiom is in the form of SubClassOf(C D) or EquivalentClasses(C D) where both C and D are NOT precoordinated concepts, this is a GCI axiom and the referencedComponentId should be 733929006 |General concept inclusion axiom (metadata)|.
- If an axiom is in the form of DisjointClasses(C D) where both C and D are precoordinated concepts, the concept ID of C should be the referencedComponentId for this axiom. Then the axiom DisjointClasses(D C) is redundant.
- The disjointness in more than two Classes can be represented as DisjointClasses(C D E …) where all classes are precoordinated concepts. The concept 787776007 |Disjoint classes axiom (OWL metadata concept)| should be the referencedComponentId for this axiom.

The definition status of a concept is |Sufficiently defined by necessary conditions definition status| if the concept has at least one EquivalentClasses axiom, irrespective of whether other SubClassOf or EquivalentClasses axioms exist.

Representation of |is a| Relationships

116680003 |Is a (attribute)| relationships in SNOMED CT are represented by different axioms, SubClassOf, SubObjectProperty, SubDataPropery, or EquivalentClasses in the OWL axiom refset.

- SubClassOf represents the |Is a (attribute)| relationship between concepts in SNOMED CT.
- SubObjectPropertyOf or SubDataPropertyOf represents the |Is a (attribute)| relationship between attributes in SNOMED CT for concept model object attributes and concept model data attributes respectively.
- EquivalentClasses means that two concepts are subclass of each other in description logics , e.g. SubClassOf(C D) and SubClassOf(D C). EquivalentClasses are usually used to represent the equivalence between a precoorrdinated concept(a named class) and an expression such as ObjectIntersectionOf() that has one part of a precoordinated superconcept and the other part is an expression that usually refines the superconcept, e.g. ObjectSomeValuesFrom() or intersection of expressions.

Class and property are all uniquely identified by an IRI in OWL 2 Ontology. It is not allowed to represent |Is a| relationships between SNOMED CT attributes and concepts in the OWL refset.

- 410662002 |Concept model attribute| should be represented as a class in the OWL axiom refset and SNOMED Ontology.

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- 762705008 |Concept model object attribute| and its subconcepts should be represented as an object property in the OWL axiom refset and SNOMED Ontology. The |Is a| relationships among them should be represented as SubObjectProperty();
- 762706009 |Concept model data attribute| and its subconcepts should be represented as a data property in the OWL axiom refset and SNOMED Ontology. The relationship among them should be represented as SubDataProperty();

The metamodeling capacities of OWL 2 allow the punning for Classes and Properties. Both attribute concepts, 762706009 |Concept model data attribute (attribute)| and 762705008 |Concept model object attribute (attribute)|, can be treated as Classes and Properties at the same time to enable the single root node of SNOMED CT. The following two SubClass axioms have been added in addition to SubProperty axioms in the OWL expression refset. These SubClass axioms represent and infer the 116680003 |Is a| relationships to 410662002 |Concept model attribute (attribute)| that is a Class.

- SubClassOf(:762706009 :410662002)
- SubClassOf(:762705008 :410662002)

For inferred relationships in the Necessary Normal Form (NNF), SubClassOf, SubObjectProperty and SubDataProperty in the OWL axiom refset should be represented as |Is a| relationships. The explanation for the Necessary Normal Form and the rules for calculating the NNF can be found in section 2.5. Generating Necessary Normal Form Relationships from the OWL Refsets.

Representation of concrete domains

DataHasValue class expression should be used for a 762706009 |Concept model data attribute (attribute)| with a particular literal value that is specified in the SNOMED OWL Logic profile specification. The DataSomeValuesFrom and DataAllValuesFrom class expressions should not be used. For example, the value of 1142135004 |Has presentation strength numerator value (attribute)| should be represented as:

```
DataHasValue(:1142135004 "50"^^xsd:decimal)
```

Unfortunately, the xsd:boolean data type is out of the scope for the OWL 2 EL profile (https://www.w3.org/TR/owl2-profiles/#Entities). In order to have a consistent representation and clear semantics, the boolean values should be represented by the concept 31874001 |True (qualifier value)| and 64100000 |False (qualifier value)|. The attribute should be a subtype of 762705008 |Concept model object attribute (attribute)| with a constraint in the SNOMED CT Machine Readable Concept Model that only these two values are in the range and the attribute cardinality is 0..1.

Note, trailing zeros are optional for the decimal data type in SNOMED CT. If the fractional part is zero, the period and following zero(es) can be omitted. For example, 2 is equivalent to 2.0. In particular, trailing zeros are prohibited for medicinal products in SNOMED CT because of clinical safety concerns. This 'trailing zero' policy should be applied in the same subject area of SNOMED CT for consistent classifications because the ELK reasoner has an incomplete implementation of concrete domains. However, it is possible that other subject areas, e.g. clinical findings, could allow for trailing zeros if it is desirable.

Attributes

SNOMED CT is based on concepts, with attributes also being represented as concepts. However, attributes need to be represented as properties in the OWL axiom refset. The following rules should be followed.

- The attribute 410662002 |Concept model attribute| should be represented as a Class in the OWL axiom refset.
- The attribute 762705008 |Concept model object attribute (attribute)| and 762706009 |Concept model data attribute (attribute)| should be represented as Classes and Properties as noted above.
- All descendants of 762705008 |Concept model object attribute (attribute)| should be represented as ObjectProperties.
- All descendants of 762706009 |Concept model data attribute (attribute)| should be represented as DataProperties.
- All the rest attributes in SNOMED CT should be represented as Classes.

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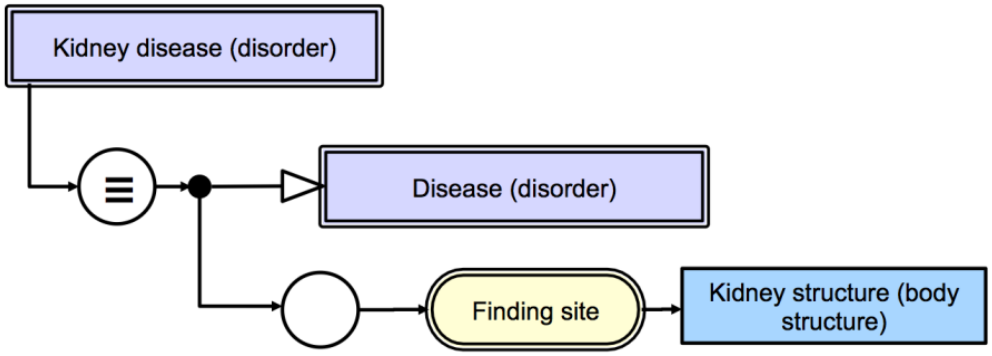
Role Group

Role groups must be explicitly stated and represented by the concept 609096000 |Role group (attribute)| as an object property in the OWL axiom refset. In the diagram of stated relationships in the OWL axiom refset, the attribute 609096000 |Role group (attribute)| should be represented by a circle rather than an object property. The role group should not be omitted for self-group attributes where there is only a single attribute in a role group.

An example for diagram representation of an OWL axiom



Modules and Axioms

The editing of entries in the OWL refsets can only be performed by the owner of that module. The extensions must not modify the OWL refset entries from the dependent modules, such as |International Health Terminology Standards Development Organization maintained module|. No changes are permitted to the content of the International Edition, except for the addition of new versions of this content in a module owned by the extension producer.

- **Axiom addition**

SNOMED CT extensions can add new axioms to the concepts in the international release to support extension content, such as adding an extension concept as an additional parent to a concept in the international release. Each axiom in an extension must have a new UUID, moduleId and effectiveTime from the extension. An international concept may have multiple axioms - one or more axiom from the international edition and zero or more axiom from extension. An axiom addition in an extension can be presented by two alternative forms with the same classification result.

- **Axiom inclusion** - a new axiom **with inclusion** of the axiom from the international release as part of the extension axiom. For example, an extension concept D can be added as a superconcept by adding a new axiom in the extension.
  SubClassOf(A C) is an axiom released in the international release.
  SubClassOf(A ObjectIntersectionOf(C D)) is a new axiom in the extension.

- **Axiom without inclusion** - a new axiom **without inclusion** of the axiom from the international release as part of the extension axiom. For example,
  SubClassOf(A C) is an axiom released in the international release.
  SubClassOf(A D) is a new axiom in the extension.

It is possible that substantive improvements or corrections to the International Edition can be made through axiom overriding in an extension, if they cannot be achieved by the Axiom addition approach.

- **Axiom overriding** - a new axiom has the same UUID from the international release, but a new effectiveTime, module id and OWL expression from the extension.

This approach is different to the Axiom addition. The extension takes over the ownership of an axiom from the International Edition and overrides the axiom.

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Whichever approach is taken by extensions, either Axiom addition or Axiom overriding, it must follow the General Authoring Principles for extensions. Any modifications resulting in changes to the classification of international content must be accompanied by a disclaimer notifying users of the differences between the extension edition and the International Edition. These changes should be forwarded to SNOMED International in a timely fashion to improve the quality of the International Edition for all users. Please note that modifications of this kind pose a risk to the comparability and interoperability of data captured using different SNOMED CT editions.

Versioning for Axioms

The versioning is at the axiom level for a concept in the OWL axiom refset. This means that there is only one effectiveTime for each version of an axiom, which may have multiple relationships. The changes to any relationships in the current editing tool will trigger an "update" of the most recent version of the relevant axiom. The versioning at the axiom level, where each axiom may represent multiple relationships, is different from the versioning of each individual relationship in the relationship file.

Any changes to the NNF should have a corresponding history of changes in the OWL axiom refset. This will ensure that all entries in the NNF can be derived from the OWL refset except for those relationships listed in table 2.4-1. The NNF will still have the computed effectiveTime for each inferred relationship. The version of each inferred relationship can be derived from the OWL refset, but it is not true in reverse.

It is permitted to make modifications to a published axiom without inactivation by the owner of the module, by creating a new version of the axiom with the same UUID and a new effectiveTime. Since any modification to an axiom could potentially alter its meaning, it is not necessary to inactivate an axiom and create a new axiom. It is also not necessary to reinstate an inactivated axiom in the previous release when the same axiom is created as a new expression. This approach can simplify the tooling and authoring process and it is a different approach to the history of changes to the individual relationship.

However, an axiom must be inactivated in the following situations:

1. The concept used as the referenced component is inactivated or changed.
2. Axiom needs to be inactivated without any replacement.
3. Any inactive concept or attribute referenced in the axiom will not be replaced by an active component.

Special notes on axioms during the transitional period from the stated relationship file to the OWL axiom refset

During the transitional period, there should be only one active axiom in the OWL axiom reference set for each concept. This axiom will represent the set of all active attributes (from the existing stated relationship file) for which the given concept is the source concept. This axiom overriding approach should only be used for the transition once. After the transition period, these axioms will be reviewed, and where appropriate will be split into multiple axioms as described in the Axiom addition approach.

If a dependent module (e.g. an extension module) adds defining relationships to a concept, then this will result in a new version of the axiom (which has the new relationship included) being added to the OWL axiom reference set. A new version of the axiom must be created if any of the concept's defining relationships change, regardless of whether or not that change is in the given module. For example (moduleId, sourceId, destinationId, typeId and referencedComponentId are represented by letters for easier readability):

id	effectiveTime	active	moduleId	sourceId	destinationId	typeId
111111	20190731	1	A	X	W	Ra
222222	20190731	1	A	X	Y	Rb

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333333	20191031	1	B	X	Z	Rz

Results in two *versions* of a single OWL reference set entry

UUID	effectiveTime	active	moduleId	referencedComponentId	owlExpression
9c1951e8-bbaa-434b-b9d7-82b460e221de	20190731	1	A	X	EquivalentClasses(:X ObjectIntersectionOf(ObjectSomeValuesFrom(:Ra :W) ObjectSomeValuesFrom(:Rb :Y)))
9c1951e8-bbaa-434b-b9d7-82b460e221de	20191031	1	B	X	EquivalentClasses(:X ObjectIntersectionOf(ObjectSomeValuesFrom(:Ra :W) ObjectSomeValuesFrom(:Rb :Y) ObjectSomeValuesFrom(:Rz :Z)))

2.5. Generating Necessary Normal Form Relationships from the OWL Refsets

The logic definitions are represented by the OWL axiom refset that is a replacement of the RF2 stated relationship file. As a result, the nature of the inferred relationship file in the distribution normal form (DNF) has changed, because the new DL features are not representable in the current relationships file. The inferred relationship file will maintain the same format and structure, but it is no longer equivalent to the stated form (containing all necessary and sufficient conditions). In fact, it is a collection of all the necessary conditions of precoordinated concepts and represents a subset of the full semantics.

Necessary Normal Form

The Necessary Normal Form (NNF) is a replacement for the Distribution Normal Form for inferred relationships. The NNF is a precalculated distribution form for practical purposes, for example, to support the continuity of existing implementations based on relational databases and queries by the expression constraint language.

The NNF consists of the full set of necessary relationships of precoordinated concepts after removal of redundant relationships within a given concept definition. Within the scope of a SNOMED CT terminology, necessary relationships are defined only for precoordinated concepts (aka OWL's named classes). Let C be a precoordinated concept and D be either a precoordinated concept or a complex expression. If an axiom is in the form of SubClassOf(C D) or EquivalentClasses(C D), then all of the derivable and necessary relationships of D are necessary relationships of C.

The NNF does not include class disjointness, transitive properties, reflexive properties and sufficient conditions represented as General Concept Inclusions (GCIs) in the OWL axiom refset.

Inferred relationships for concrete values, e.g. decimal, integer, string, or dateTime, should be included in a separate relationship file.

Rules for Determining Redundant Relationships

Rule 1 - Class and Role inclusions

Given two relationships, A and B, A with r = C and B with s = D, within the same role group, A is redundant if:

- r is the same as or a supertype of s, and

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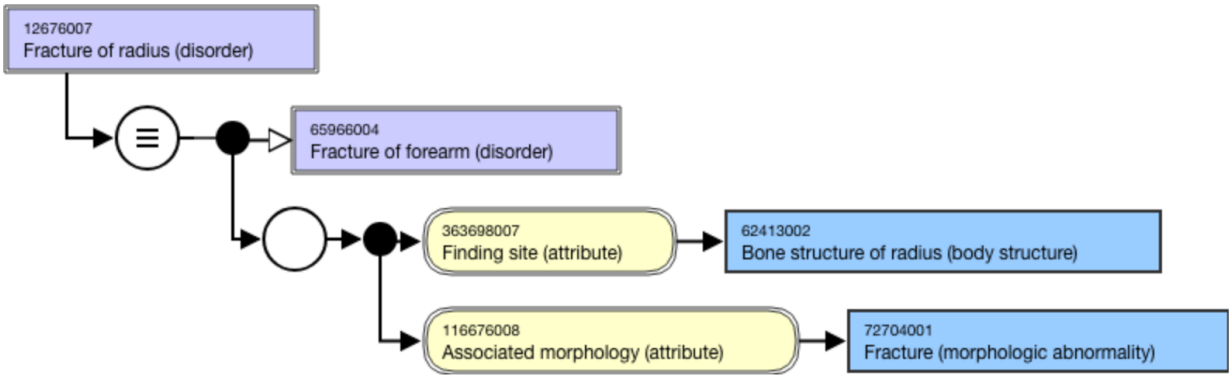
- C is the same as or a supertype of D

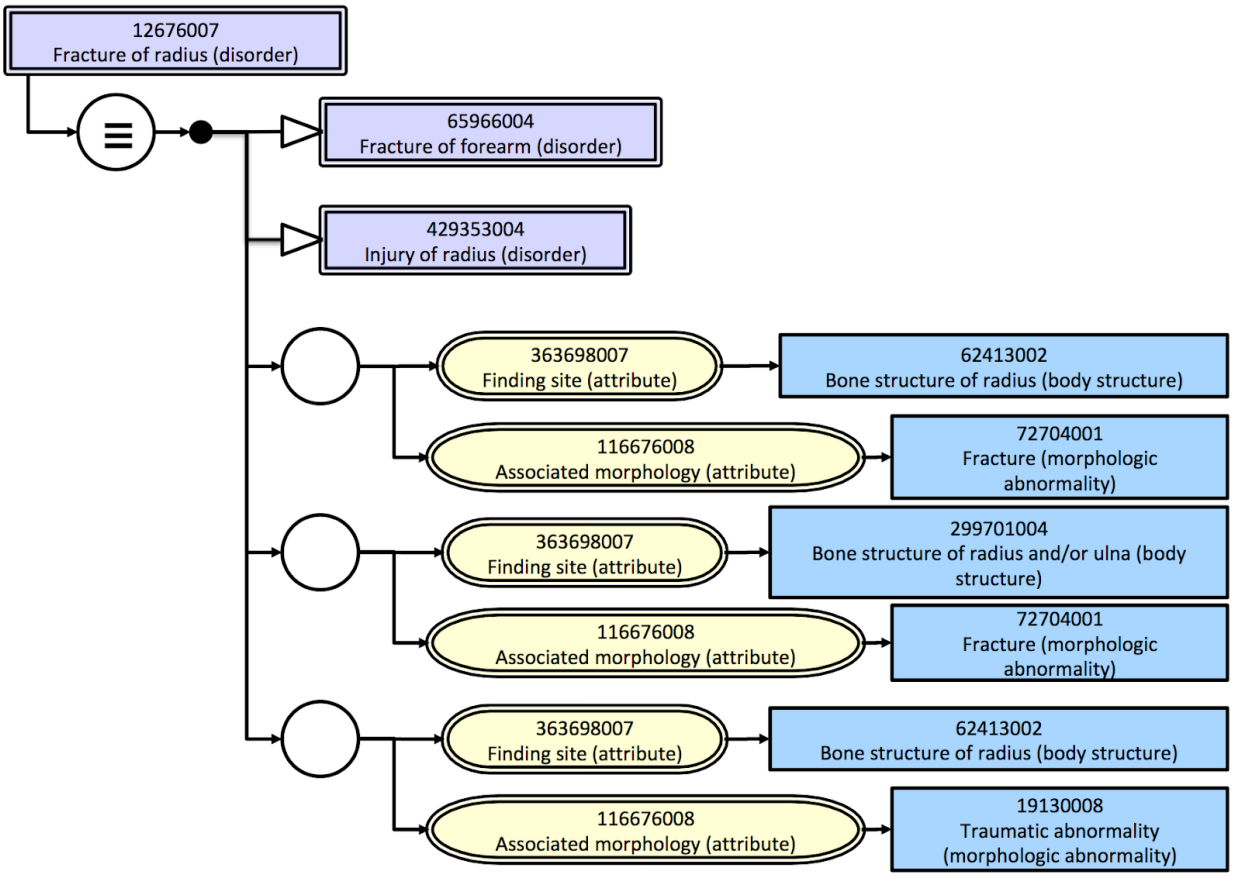
Note, “crossover relationships”, where r is a supertype of s, and C is instead a subtype of D do not result in a redundant relationship.

Example for Class inclusion

Stated relationships



Inferred relationships before the removal of redundant relationships



Inferred relationships after the reduction

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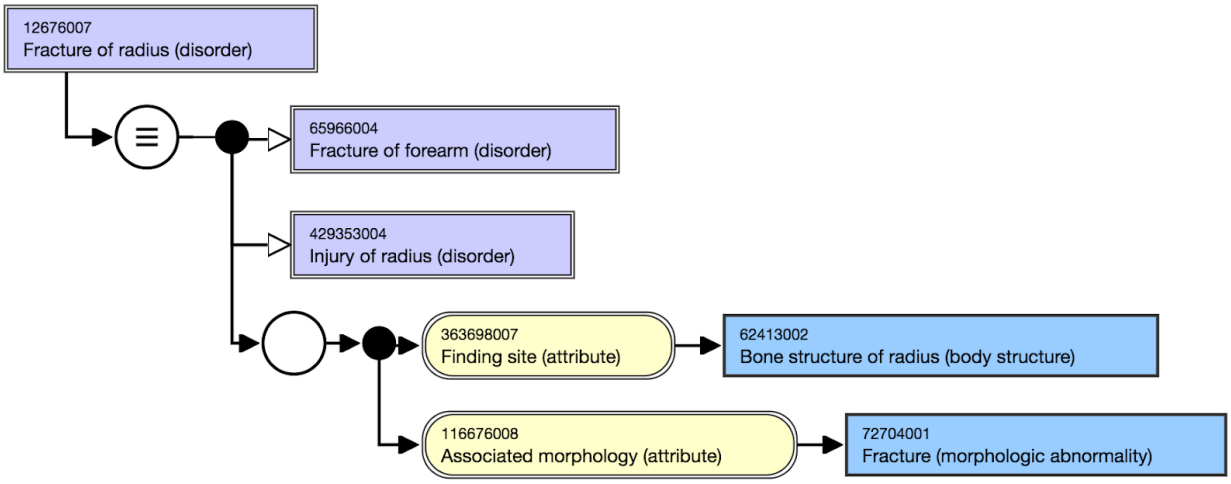

For |Fracture of radius|, the relationship |Finding site| = |Bone structure of radius and/or ulna| is inherited from |Fracture of forearm| , which is a redundant relationship because |Bone structure of radius| is a subtype of |Bone structure of radius and/or ulna|. The relationship |Associated morphology| = |Traumatic abnormality| is inherited from |Injury of radius|, which is a redundant relationship because |Fracture (morphologic abnormality)| is a subtype of |Traumatic abnormality|.

Table 2.5-1: Example in OWL axiom refset and RF2 relationship file (NNF)

referencedComponentId	owlExpression (stated relationships)	Inferred Relationships in Necessary Normal Form			
		sourceId	destinationId	relationshipGroup	typeId
125605004	EquivalentClasses(:125605004 ObjectIntersectionOf(:64572001 ObjectSomeValuesFrom(:609096000 ObjectIntersectionOf(ObjectSomeValuesFrom(:116676008 :72704001) ObjectSomeValuesFrom(:363698007 :272673000)))))	125605004	284003005	0	116680003
		125605004	72704001	1	116676008
		125605004	272673000	1	363698007
12676007	EquivalentClasses(:12676007 ObjectIntersectionOf(:64572001 ObjectSomeValuesFrom(:609096000 ObjectIntersectionOf(ObjectSomeValuesFrom(:116676008 :72704001) ObjectSomeValuesFrom(:363698007 :62413002)))))	12676007	65966004	0	116680003
		12676007	429353004	0	116680003
		12676007	72704001	1	116676008
		12676007	62413002	1	363698007

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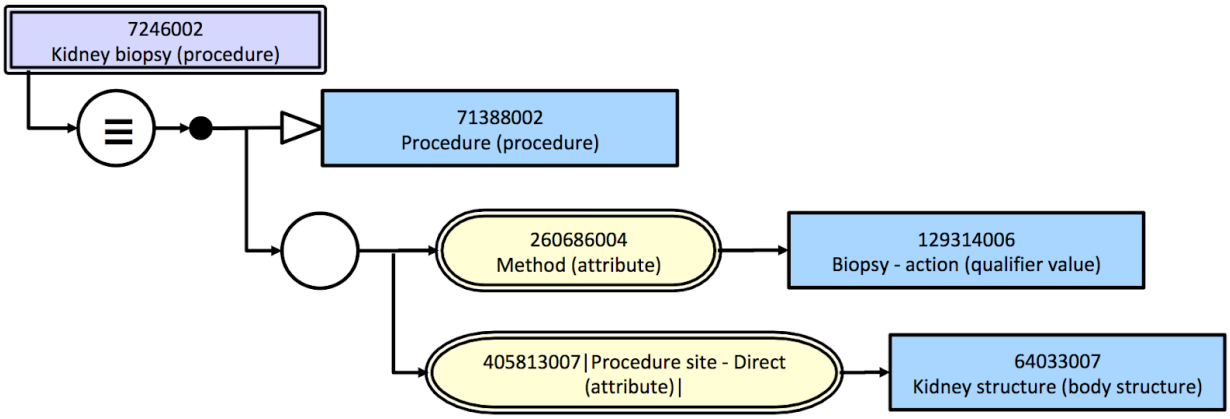
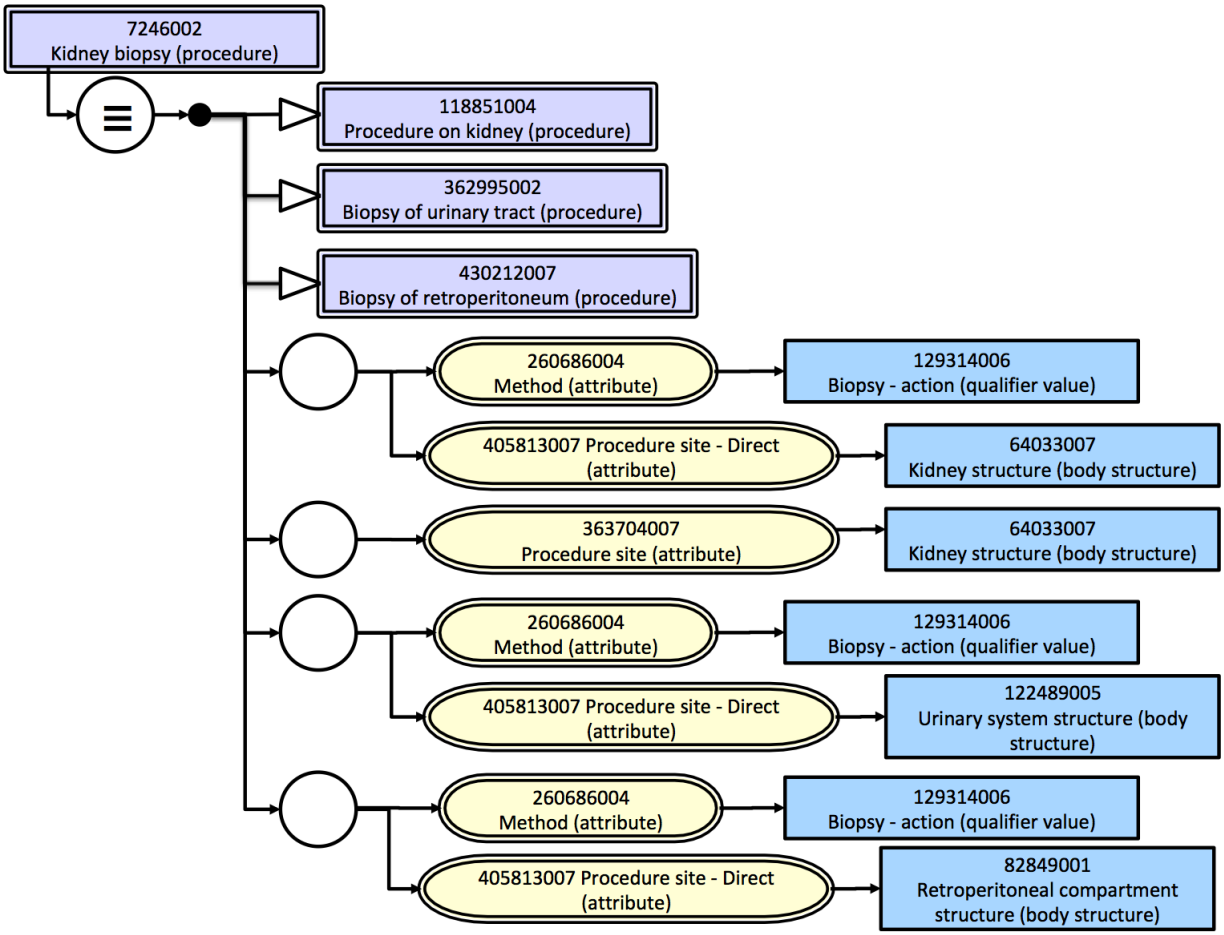
62413002	SubClassOf(:62413002 :299701004)	62413002	299701004	0	116680003

Example for Role inclusion

Stated relationships



Inferred relationships before the removal of redundant relationship



Inferred relationships after reduction

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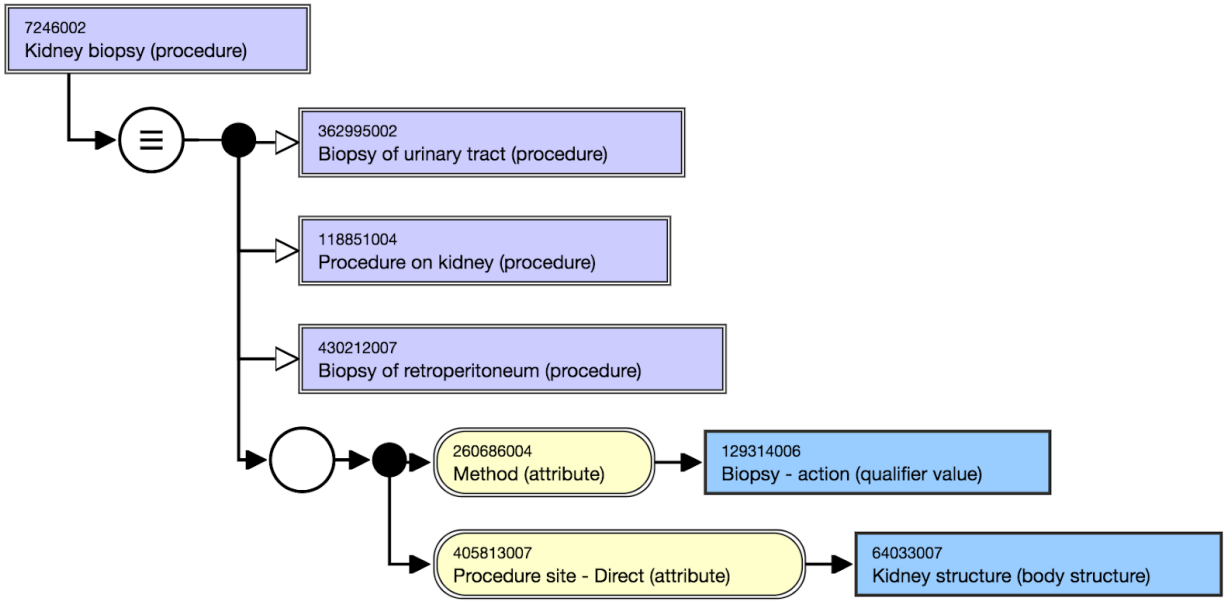

For concept |Kidney biopsy|, the relationship |Procedure site| = |Kidney structure| is inherited from |Procedure on kidney|, which is a redundant relationship to |Procedure site - Direct| = |Kidney structure| because |Procedure site - Direct| is a subtype of |Procedure site|. Because |Kidney structure| is a subtype of |Urinary system structure| and |Retroperitoneal compartment structure|, the inherited relationships for |Procedure site - Direct| are also redundant.

Table 2.5-2: Example in OWL axiom refset and RF2 relationship file (NNF)

referencedComponentId	owlExpression (stated relationships)	Inferred Relationships in Necessary Normal Form			
		sourceId	destinationId	relationshipGroup	typeId
118851004	EquivalentClasses(:118851004 ObjectIntersectionOf(:71388002 ObjectSomeValuesFrom(:609096000 ObjectSomeValuesFrom(:363704007 :64033007))))	118851004	71388002	0	116680003
		118851004	64033007	1	363704007
7246002	EquivalentClasses(:7246002 ObjectIntersectionOf(:71388002 ObjectSomeValuesFrom(:609096000 ObjectIntersectionOf(ObjectSomeValuesFrom(:260686004 :129314006) ObjectSomeValuesFrom(:405813007 :64033007)))))	724602	118851004	0	116680003
		724602	362995002	0	116680003
		724602	430212007	0	116680003

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		72460 02	1293140 06	1	26068 6004
		72460 02	6403300 7	1	40581 3007
405813007	SubObjectPropertyOf(:405813007 :363704007)	40581 3007	3637040 07	0	11668 0003

Rule 2 - Property chains including transitive properties

Given attribute r, s and t with a property chain SubObjectPropertyOf(ObjectPropertyChain(t s) r), and two relationships A and B, A with r = C and B with u = D, within the same role group, A is redundant if:

- Attribute u is the same as or a subtype of t, and
- D has a relationship to C via attribute s

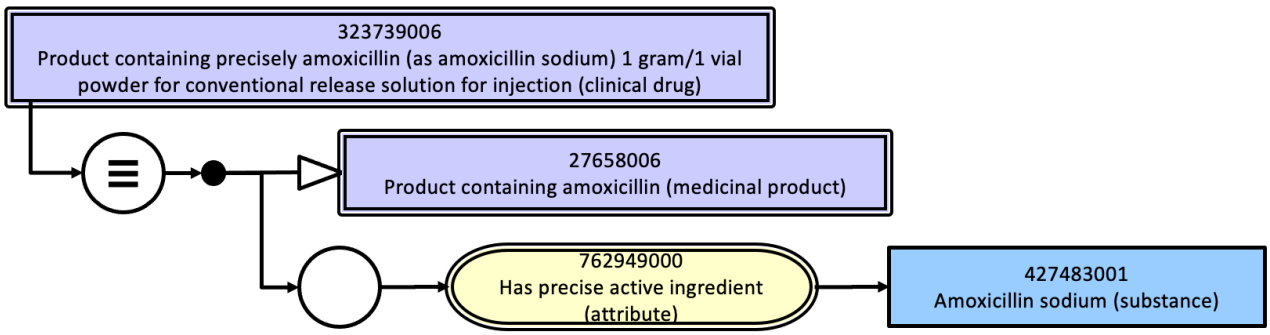
Note the following:

- C does not need to subsume D
- Attribute t does not need to be the same as or a subtype of r
- Transitive properties are defined by a property chain in the form of

SubObjectPropertyOf(ObjectPropertyChain(r r) r) and thus it is a special case of the above.

Example for property chain:

Stated relationships of |Product containing precisely amoxicillin (as amoxicillin sodium) 1 gram/1 vial powder for conventional release solution for injection (clinical drug)| (the other model detail has been omitted):



Stated relationships of |Amoxicillin sodium (substance)|:

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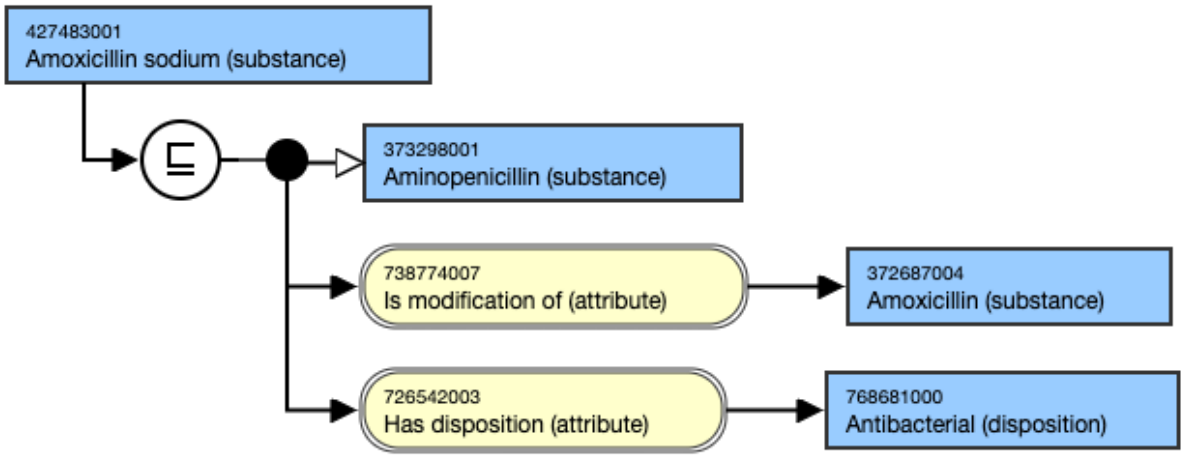
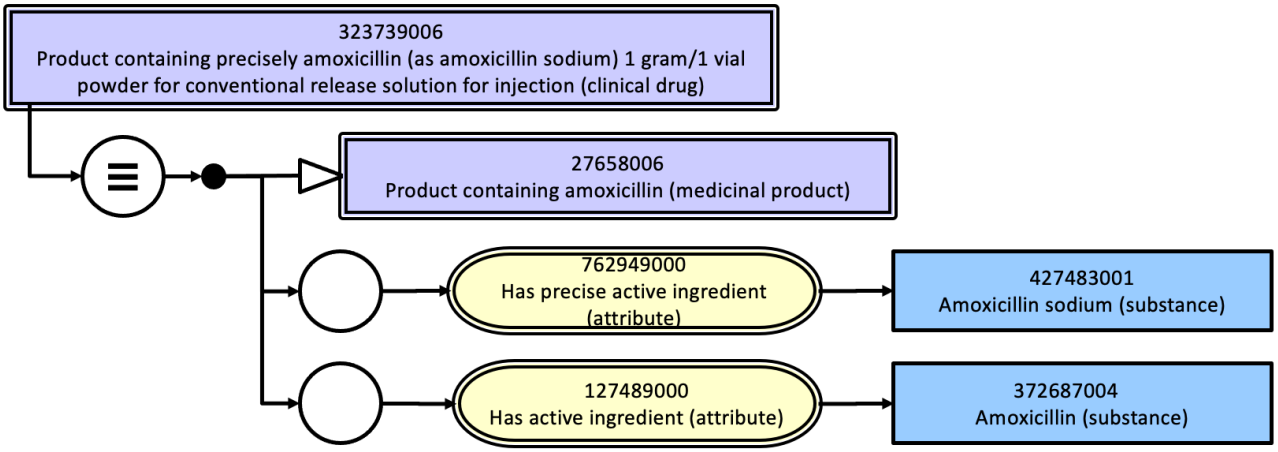
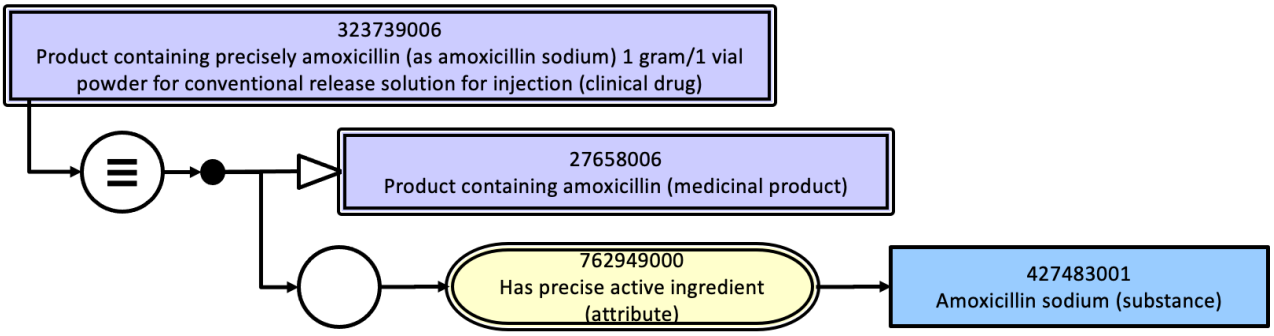


Inferred relationships before the removal of the redundant relationship



Inferred relationships after the reduction



For |Product containing precisely amoxicillin (as amoxicillin sodium) 1 gram/1 vial powder for conventional release solution for injection (clinical drug)|, the relationship |Has active ingredient| = |Amoxicillin| is inherited from |Product containing amoxicillin|. If rule 1 for class inclusion was applied, the relationships would not be considered redundant because |Amoxicillin sodium (substance)| is not a subconcept of |Amoxicillin (substance)|. Since |Amoxicillin sodium| |Is modification of| |Amoxicillin| and property chain of "|Has active ingredient| o |Is modification of|" is a sub-property of |Has active ingredient|, rule 2 actually compares the anonymous concepts for

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subsumption, i.e. |Has active ingredient| = |Amoxicillin| with |Has active ingredient| = |Amoxicillin sodium|. Therefore, the inherited relationship is redundant and can be removed from the NNF. Their relationships and property chain can be demonstrated in the following diagram.

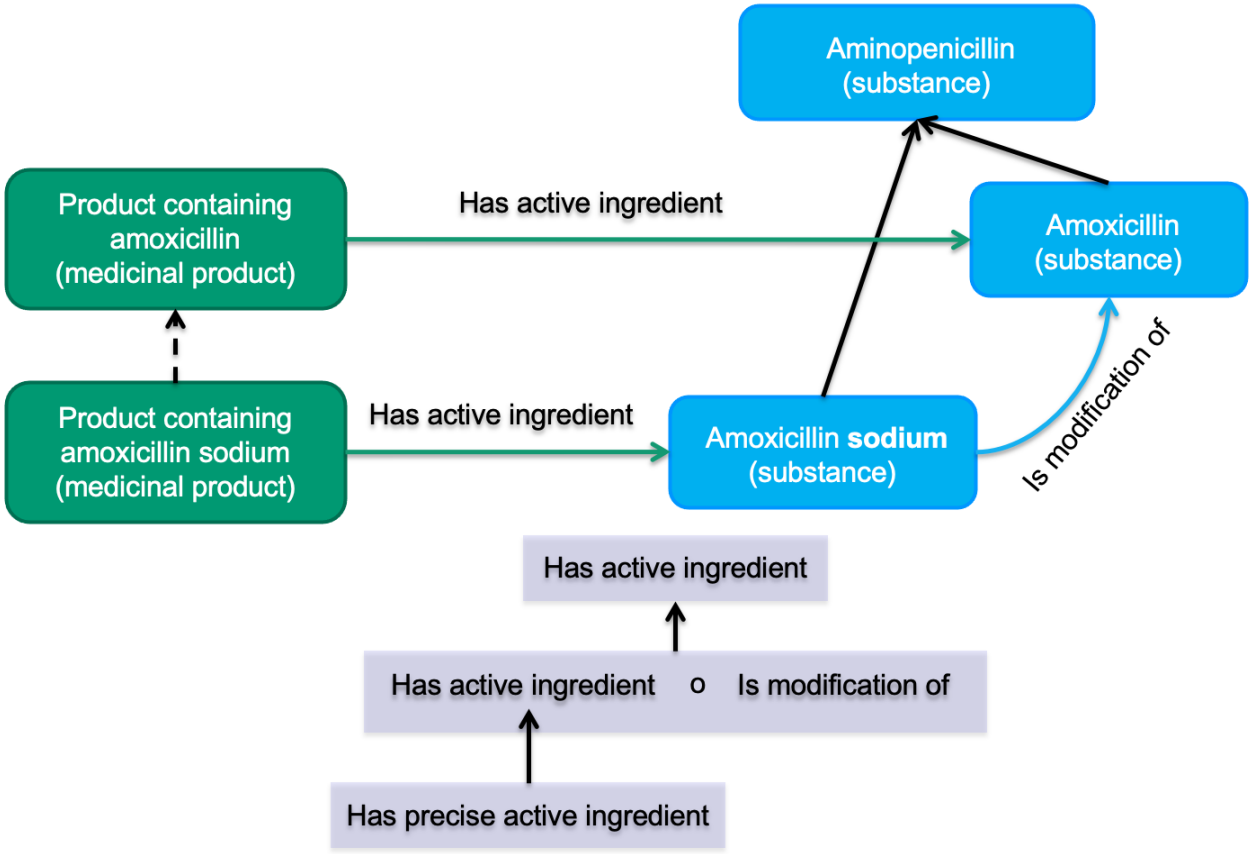

Table 2.5-3: Example in OWL axiom refset and RF2 relationship file (NNF)

referencedComponentId	owlExpression (stated relationships)	Inferred Relationships in Necessary Normal Form			
		sourceId	destinationId	relationshipGroup	typeId
27658006	EquivalentClasses(:27658006 ObjectIntersectionOf(:763158003 ObjectSomeValuesFrom(:609096000 ObjectSomeValuesFrom(:127489000 :372687004))))	27658006	90704004	0	116680003
		27658006	372687004	1	127489000
323739006	EquivalentClasses(:323739006 ObjectIntersectionOf(:763158003 ObjectSomeValuesFrom(:609096000 ObjectSomeValuesFrom(:762949000 :427483001))))	323739006	27658006	0	116680003

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		323739006	427483001	1	762949000
127489000	SubObjectPropertyOf(ObjectPropertyChain(:127489000 :738774007) :127489000))	N/A	N/A		N/A
427483001	SubClassOf(:427483001 ObjectIntersectionOf(:440327007 ObjectSomeValuesFrom(:738774007 :372687004)))	427483001	373298001	0	116680003
		427483001	372687004	0	738774007
		427483001	768681000	0	726542003

Technical Implementation for Calculating the NNF

This fairly complex process uses the stated form and the output of the reasoner to calculate the necessary normal form which is represented in the relationship RF2 file.

The most straightforward way to produce the necessary normal form would be to use the Snomed OWL Toolkit or the Classification Service REST API which is language agnostic.

High Level Process

Classification

1. Read the Stated Form from RF2 files.

a. The following files are required: Concept, OWL Ontology Reference Set, OWL Axiom Reference Set.
b. If the edition has any active stated relationships then the Stated Relationship and MRCM Attribute Domain Reference Set files are also required.

1. Use the OWL API to infer the class hierarchy

a. Build the Ontology object using:

i. Axioms from the OWL Axiom Reference Set, making a note of any Transitive property and Property Chain axioms.
ii. Axioms created by converting Stated Relationships to OWL Axioms using the MRCM Attribute Domain Reference Set for list of attributes which should not be grouped in the given domain.

a. Use a reasoner to pre-compute the class hierarchy.

Necessary Normal Form Calculation

Calculating the necessary normal form happens in two passes of the hierarchy.org.snomed.otf.owltoolkit.normalform

1. Walk the class hierarchy in a top-down, breadth first, order.

a. For each class visited gather the stated attributes of this class and each inferred parent.
b. Compare the attributes and remove those which are found to be redundant because they are less specific in terms of depth in the hierarchy.
c. During this first pass build a hierarchy for property chains and transitive properties.

1. Walk the class hierarchy again in the same order reducing the attributes of each class further.

a. Compare the attributes and remove those which are found to be redundant because they are less specific in terms of depth in one of the alternate hierarchies.

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For fine level detail the best source of information is the Java class org.snomed.otf.owltoolkit.normalform.RelationshipNormalFormGenerator which performs the Necessary Normal Form calculation.

Assignment for Role Group Number

It is important to clearly indicate if an attribute is grouped or not because |Role group (attribute)| has impact to semantics and classification results. |Role group| is represented by an integer in the field of relationshipGroup in the relationship file. In contrast, |Role group| is represented by 609096000 |Role group (attribute)| as an object property in the OWL axiom refset. After the stated relationship file is replaced by the OWL expression refset, role group numbers need to be generated for inferred relationships.

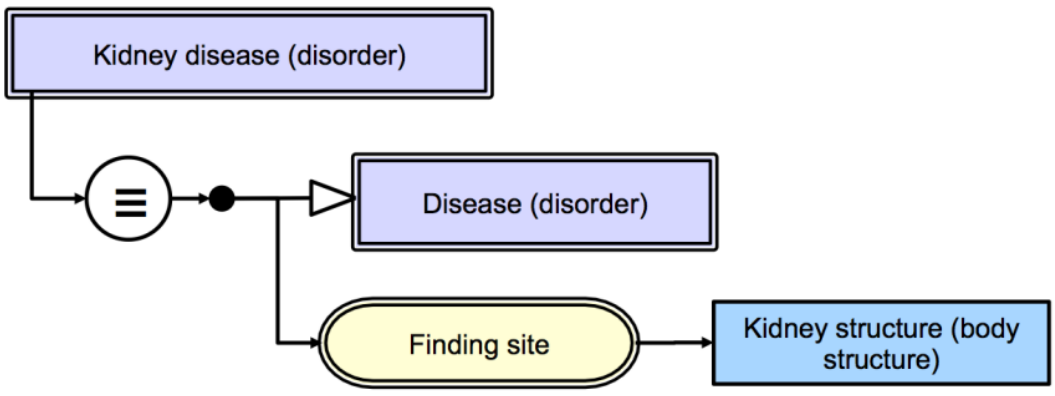
The following rules should be followed in the inferred relationship file to provide consistent representation aligned with the concept model diagram and the OWL axiom refset.

1. All 116680003 |Is a| relationships should be assigned in role group 0;
2. Attribute that is not grouped, not a value of |Role group (attribute)| or grouped=0 in MRCM, should be assigned in role group 0;
3. Attribute that is grouped, value of |Role group (attribute)| or grouped=1 in MRCM, should be assigned a role group number that is not 0. Each |Role group (attribute)| in the OWL axiom should be presented by a unique role group number. Note, role group merging is not covered here.

609096000 |Role group (attribute)| is explicitly represented for self-grouped attributes where there is only a single attribute in a role group in an OWL axiom. However, these self-grouped attributes and values are not explicitly represented in the current relationship files. This representation has caused confusion if an attribute in role group 0 is grouped or not. The following example demonstrates the changes to assignment of role group number after the implementation of the complete OWL axiom refset.

An example for the current diagram representation for attribute in role group 0 in the stated relationship file and concept model diagram

sourceId	destinationId	relationshipGroup	typeId
90708001	64033007	0	363698007



After the complete OWL axiom refset is implemented, |Role group| in the OWL axiom refset and concept model diagram should be represented as following.

referencedComponentId	owlExpression
90708001	EquivalentClasses(:90708001 ObjectIntersectionOf(:64572001 ObjectSomeValuesFrom(:609096000 ObjectSomeValuesFrom(:363698007 :64033007))))

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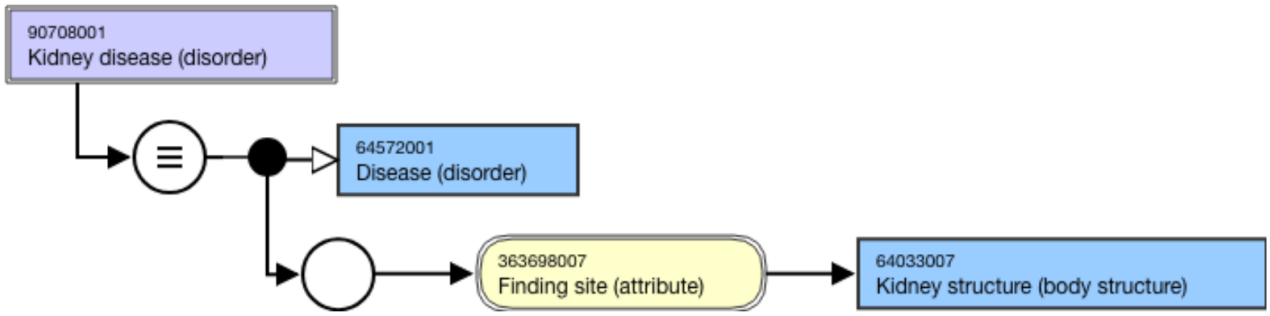
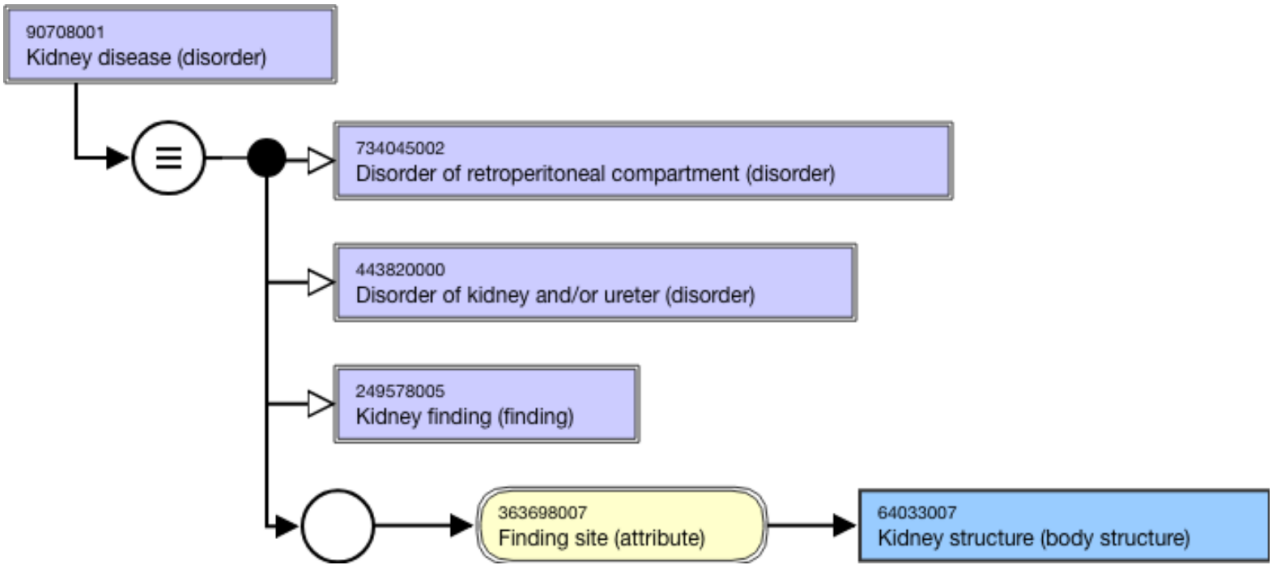


Representation of |Role group| in the NNF relationship file and concept model diagram

sourceId	destinationId	relationshipGroup	typeId
90708001	734045002	0	116680003
90708001	443820000	0	116680003
90708001	249578005	0	116680003
90708001	64033007	1	363698007



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3. Quality Assurance

The following additional quality assurance should be developed for the OWL refsets though this is not a complete list.

1. Validation of expressions following OWL Functional-Style Syntax specification
2. Validation of profile of the OWL ontology generated from the OWL refsets
3. Each active concept should have at least one active axiom
4. Each active concept can only have one declaration of Class, Object property, or Data property
5. Inactive concept must not have active axiom
6. Active axiom must not contain any inactive component
7. Domain and range validation by property types

1. All descendants of |Concept model object attribute| can only have target values from component of concept or expression
2. All descendants of |Concept model data attribute| can only have target values from data types and it must not have values from component of concept[1]

---

[1] Utilise the OWL API 4 or 5 for profile violation check as part of QA
http://owlcs.github.io/owlapi/apidocs_4/org/semanticweb/owlapi/profiles/OWLProfileViolation.html
http://owlcs.github.io/owlapi/apidocs_5/org/semanticweb/owlapi/profiles/OWLProfileViolation.html

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4. OWL Expression Reference Set Specification

Purpose

An 762676003 |OWL expression type reference set| associates description logic statements with SNOMED CT concept in the OWL functional syntax.

The SNOMED CT International Release contains content in two reference sets that follow the 762676003 |OWL expression type reference set| pattern:

- The 733073007 |OWL axiom reference set (foundation metadata concept)|, in which the OWL expressions represent axioms that form the logical definition of the concept identified by the referencedComponentId
- The 762103008 |OWL ontology reference set (foundation metadata concept)|, in which the OWL expressions represent essential information about an ontology. This information includes, namespaces, ontology URI, ontology version URI, and import statements. The 762103008 |OWL ontology reference set (foundation metadata concept)| enables the use of prefixes in the ontology

Data Structure

An 762676003 |OWL expression type reference set| is structured as shown in the following table.

Field	Data type	Purpose	Mutable	Part of Primary Key
id	UUID	A 128 bit unsigned Integer, uniquely identifying this reference set member. Different versions of a *reference set member* share the same id but have different effectiveTime. This allows a *reference set member* to be modified or made inactive (i.e. removed from the active set) at a specified time.	NO	YES (Full / Snapshot)
effectiveTime	Time	The inclusive date or time at which this version of the identified reference set member became the current version. **Note**: In distribution files the effectiveTime should follow the short ISO date format (*YYYYMMDD*) and should not include the hours, minutes, seconds or timezone indicator. The current version of this reference set member at time *T* is the version with the most recent effectiveTime prior to or equal to time *T* .	YES	YES (Full) Optional (Snapshot)
active	Boolean	The state of the identified reference set member as at the specified effectiveTime . If active = 1 (true) the reference set member is part of the current version of the set, if active = 0 (false) the reference set member is not part of the current version of the set.	YES	NO
moduleId	SCTID	Identifies the SNOMED CT module that contains this reference set member as at the specified effectiveTime . The value must be a subtype of 900000000000443000 |Module (core metadata concept)|within the metadata hierarchy.	YES	NO

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refsetId	SCTID	Identifies the reference set to which this reference set member belongs. In this case, a subtype descendant of: 762676003 |OWL expression type reference set (foundation metadata concept)|	NO	NO
referencedComponentId	SCTID	A reference to the SNOMED CT component to be included in the reference set. The concept to which the OWL expression applies. In the case of the 733073007 |OWL axiom reference set (foundation metadata concept)|, the axiom contributes to the definition of the identified concept.	NO	NO
owlExpression	String	The text of OWL expression to attach to the component identified by referencedComponentId.	YES	NO

Metadata

The following metadata supports this reference set:

900000000000454005 |Foundation metadata concept|
  900000000000455006 |Reference set|
    762676003 |OWL expression type reference set|
      762103008 |OWL ontology reference set|
      733073007 |OWL axiom reference set|
  900000000000457003 |Reference set attribute|
    706999006 |Expression|
      762677007 |OWL expression|
    900000000000459000 |Attribute type|
      900000000000465000 |String|
      762678002 |OWL 2 language syntax|

Descriptor Template and Examples

The reference set example tables on this page have been revised as follows to aid clarity and understanding:

- The first four columns which are present in all release files are not shown. The omitted columns (id, effectiveTime, active) are used in the same way in all referenced sets to support identification, versioning and packaging. They do not directly affect the specific features of a particular reference set or reference set type.
- Reference set columns that contain SNOMED CT identifiers are expanded to show details of the concept or description referenced by that identifier. In some cases, the term is shown in the same column using the expression syntax, in other cases an additional column with a name suffix '_term' has been added. In the standard reference set files only the identifier is present in the column and there is no added column for the term. When using reference sets, the term and other details of the component are looked up from the relevant component release files.

Descriptor Template

The table below shows the descriptors that define the structure of the 762676003 |OWL expression type reference set| pattern and examples of the descriptors for specific reference sets that follow this pattern.

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Table 4-1: Descriptor templates for OWL expression reference rets

refsetId	referencedComponentId	attributeDescription	attributeType	attributeOrder
900000000000456007 | Reference set descriptor|	762676003 |OWL expression type reference set|	449608002 |Referenced component|	900000000000461009 |Concept type component|	0
900000000000456007 | Reference set descriptor|	762676003 |OWL expression type reference set|	762677007 |OWL expression|	762678002 |OWL 2 language syntax|	1
900000000000456007 | Reference set descriptor|	762103008 |OWL ontology reference set|	449608002 |Referenced component|	900000000000461009 |Concept type component|	0
900000000000456007 | Reference set descriptor|	762103008 |OWL ontology reference set|	762677007 |OWL expression|	762678002 |OWL 2 language syntax|	1
900000000000456007 | Reference set descriptor|	733073007 |OWL axiom reference set|	449608002 |Referenced component|	900000000000461009 |Concept type component|	0
900000000000456007 | Reference set descriptor|	733073007 |OWL axiom reference set|	762677007 |OWL expression|	762678002 |OWL 2 language syntax|	1

OWL Ontology Reference Set Example

Table 4-2: OWL ontology reference set example

moduleId	refsetId	referencedComponentId	owlExpression
900000000000012004 |SNOMED CT model component module|	762103008 |OWL ontology reference set|	734146004 |OWL ontology namespace|	Prefix(:=<http://snomed.info/id/>)
900000000000012004 |SNOMED CT model component module|	762103008 |OWL ontology reference set|	734146004 |OWL ontology namespace|	Prefix(owl:=<http://www.w3.org/2002/07/owl#>)
900000000000012004 |SNOMED CT model component module|	762103008 |OWL ontology reference set|	734146004 |OWL ontology namespace|	Prefix(rdf:=<http://www.w3.org/1999/02/22-rdf-syntax-ns#>)
900000000000012004 |SNOMED CT model component module|	762103008 |OWL ontology reference set|	734146004 |OWL ontology namespace|	Prefix(xml:=<http://www.w3.org/XML/1998/namespace>)
900000000000012004 |SNOMED CT model component module|	762103008 |OWL ontology reference set|	734146004 |OWL ontology namespace|	Prefix(xsd:=<http://www.w3.org/2001/XMLSchema#>)

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900000000000012004 |SNOMED CT model component module|	762103008 |OWL ontology reference set|	734146004 |OWL ontology namespace|	Prefix(rdfs:=<http://www.w3.org/2000/01/rdf-schema#>)
900000000000012004 |SNOMED CT model component module|	762103008 |OWL ontology reference set|	734147008 |OWL ontology header|	Ontology(<http://snomed.info/sct/900000000000207008>)

OWL Axiom Reference Set Example

Table 4-3: OWL axiom reference set example

moduleId	refsetId	referencedComponentId	owlExpression	Explanatory Notes
900000000000207008 |SNOMED CT core module|	733073007 |OWL axiom reference set|	404684003 | Clinical finding (finding)|	SubClassOf(:404684003 :138875005)	Example of SubClassOf, which is equivalent to an |Is a| relationship between most SNOMED CT concepts. ⚠ A different OWL expression is used to represent |Is a| relationships between attributes. This is shown in the row below. • 404684003 |Clinical finding (finding)| • 138875005 |SNOMED CT Concept (SNOMED RT+CTV3)|
900000000000012004 |SNOMED CT model component module|	733073007 |OWL axiom reference set|	774081006 | Proper part of (attribute)|	SubObjectPropertyOf(:774081006 :733928003)	Example of SubObjectPropertyOf, which is equivalent to an |Is a| relationship between attributes. • 774081006 |Proper part of (attribute)| • 733928003 |All or part of (attribute)|
900000000000207008 |SNOMED CT core module|	733073007 |OWL axiom reference set|	90708001 | Kidney disease (disorder)|	EquivalentClasses(:90708001 ObjectIntersectionOf(:64572001 ObjectSomeValuesFrom(:609096000 ObjectSomeValuesFrom(:363698007 :64033007))))	Example of EquivalentClasses. which is equivalent to stating that a concept is sufficiently defined by a set of necessary relationships. • 90708001 |Kidney disease (disorder)| • 64572001 |Disease| • 609096000 |Role group (attribute)| • 363698007 |Finding site (attribute)| • 64033007 |Kidney structure (body structure)|

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900000000000207008 |SNOMED CT core module|	733073007 |OWL axiom reference set|	126516008 | Neoplasm of skin of upper limb (disorder) |	EquivalentClasses(:126516008 ObjectIntersectionOf(:64572001 ObjectSomeValuesFrom(:609096000 ObjectIntersectionOf(ObjectSomeValuesFrom(:116676008 :108369006) ObjectSomeValuesFrom(:363698007 :371311000)))))	Example of a role group with a conjunction of two relationships as its value. • 126516008 |Neoplasm of skin of upper limb (disorder)| • 64572001 |Disease| • 609096000 |Role group (attribute)| • 116676008 |Associated morphology (attribute)| • 108369006 |Neoplasm (morphologic abnormality)| • 363698007 |Finding site (attribute)| • 371311000 |Skin structure of upper limb (body structure)|
900000000000012004 |SNOMED CT model component module|	733073007 |OWL axiom reference set|	774081006 | Proper part of (attribute)|	TransitiveObjectProperty(:774081006)	Example of a transitive object property. • 774081006 |Proper part of (attribute)|
900000000000012004 |SNOMED CT model component module|	733073007 |OWL axiom reference set|	127489000 | Has active ingredient (attribute)|	SubObjectPropertyOf(ObjectPropertyChain(:127489000 :738774007) :127489000))	Example of a property chain. • 127489000 |Has active ingredient (attribute)| • 738774007 |Is modification of (attribute)|
900000000000207008 |SNOMED CT core module|	733073007 |OWL axiom reference set|	703264005 | Secondary osteoporosis (disorder)|	SubClassOf(ObjectIntersectionOf(:64859006 ObjectSomeValuesFrom(:609096000 ObjectSomeValuesFrom(:255234002 :387713003)) ObjectSomeValuesFrom(:609096000 ObjectSomeValuesFrom(:363698007 :272673000))) :703264005)	Example of a general concept inclusion (GCI). • 703264005 |Secondary osteoporosis (disorder)| • 64859006 |Osteoporosis (disorder)| • 609096000 |Role group (attribute)| • 255234002 |After (attribute)| • 387713003 |Surgical procedure (procedure)| • 363698007 |Finding site (attribute)| • 272673000 |Bone structure (body structure)|

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