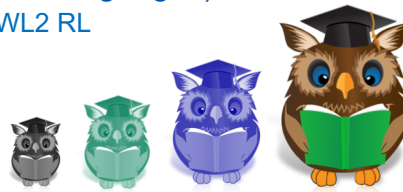


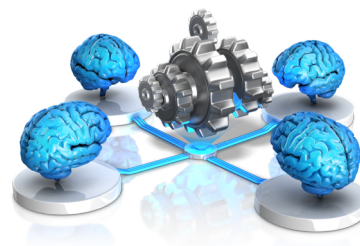
## Description Logic




- Description logics are formal languages designed for knowledge representation and reasoning
- Each DL describes a language
  - Each language differs in expressivity and reasoning complexity
- Expressivity of a DL language is defined by allowing or disallowing different constructs in their language
  - e.g. conjunction, disjunction, negation, quantifiers
- OWL2 is the language used in SNOMED CT
- Different OWL2 profiles (or sublanguages) exist
  - E.g. OWL2 EL, OWL2 QL, OWL2 RL



## Changes to Support Advanced DL Features



## Three Key Goals




- Enable quality improvements
  - More complete definitions can be specified
- Enable improvements to analytics capabilities
  - More complete definitions enable query results to be more complete and more precise
- Increase productivity in concept authoring
  - Support for more complete definitions reduces the need for arbitrary authoring rules



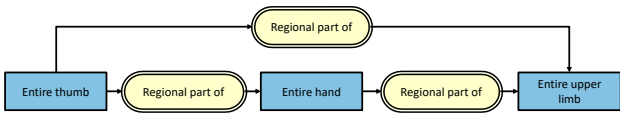


## Examples of Required DL Enhancements

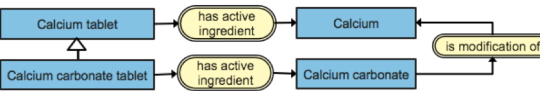


1. Property characteristics
 

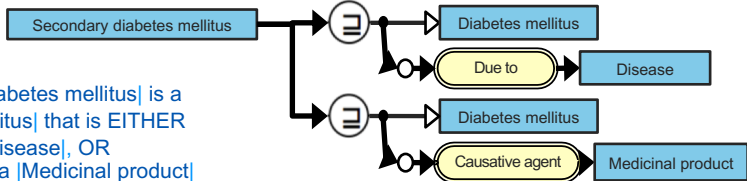
e.g. Declare [regional part of] as transitive


1. Property chains
 


e.g. Declare [active ingredient] chains with [is modification of]


1. General concept inclusions and additional axioms
 

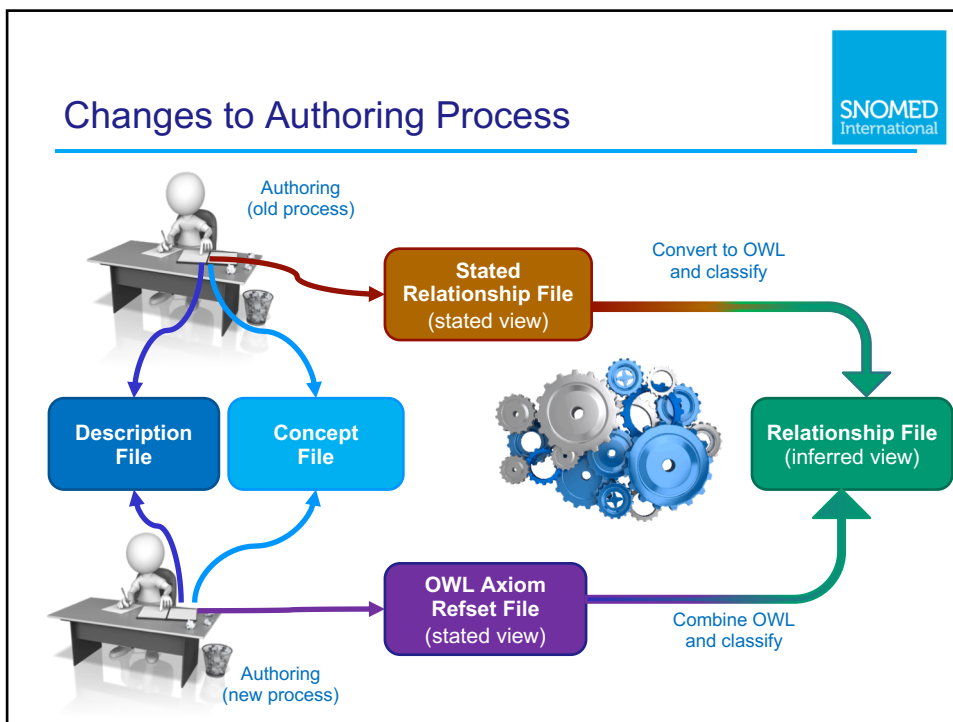

*Example*  
 [Secondary diabetes mellitus] is a [Diabetes mellitus] that is EITHER  
 • due to a [Disease], OR  
 • caused by a [Medicinal product]




## Limitations of the Relationship File Format



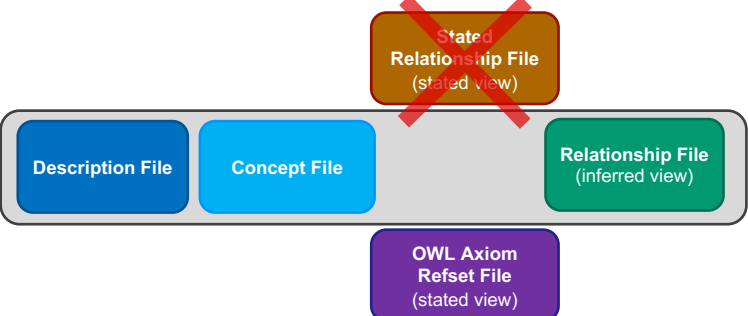
- Cannot fully represent the more expressive semantics of advanced DL profiles
  - For example
    - General concept inclusions (GCIs)
    - Property transitivity
    - Property chains
    - Nested definitions
    - Concrete domains
- Additional RF2 reference sets have been designed to enable SNOMED CT to use this greater expressivity




## No Changes to Other Release Files




- Concept, description and relationship files
  - Have exactly the same structures as before
- Relationship file
  - Still represents inferred relationships in the same way
  - However those inferred relationships are now generated from the stated view represented in the new OWL reference sets



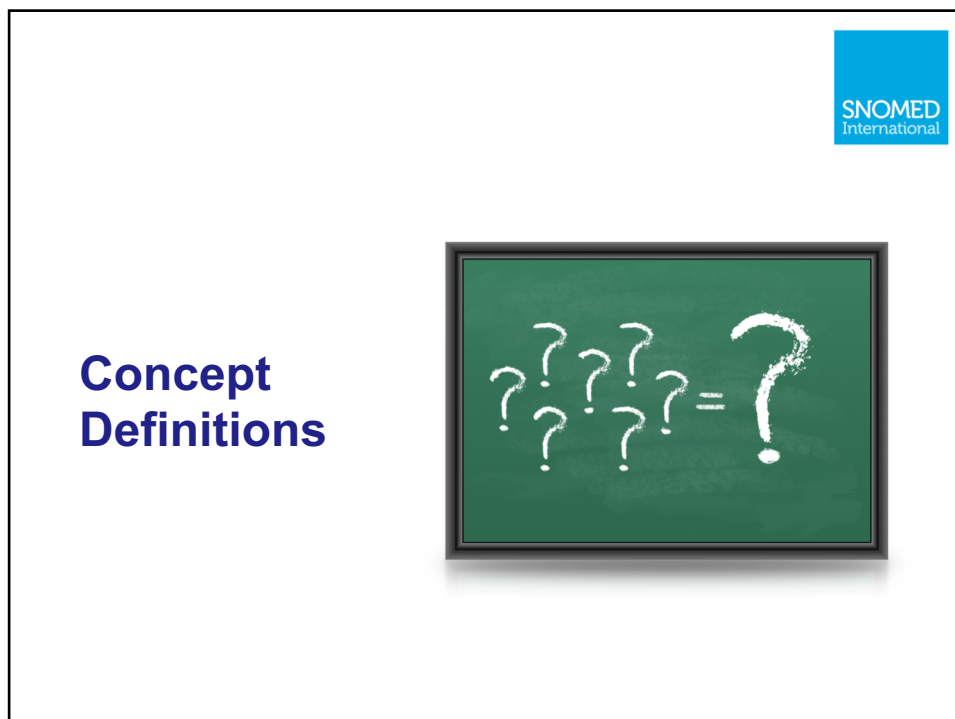
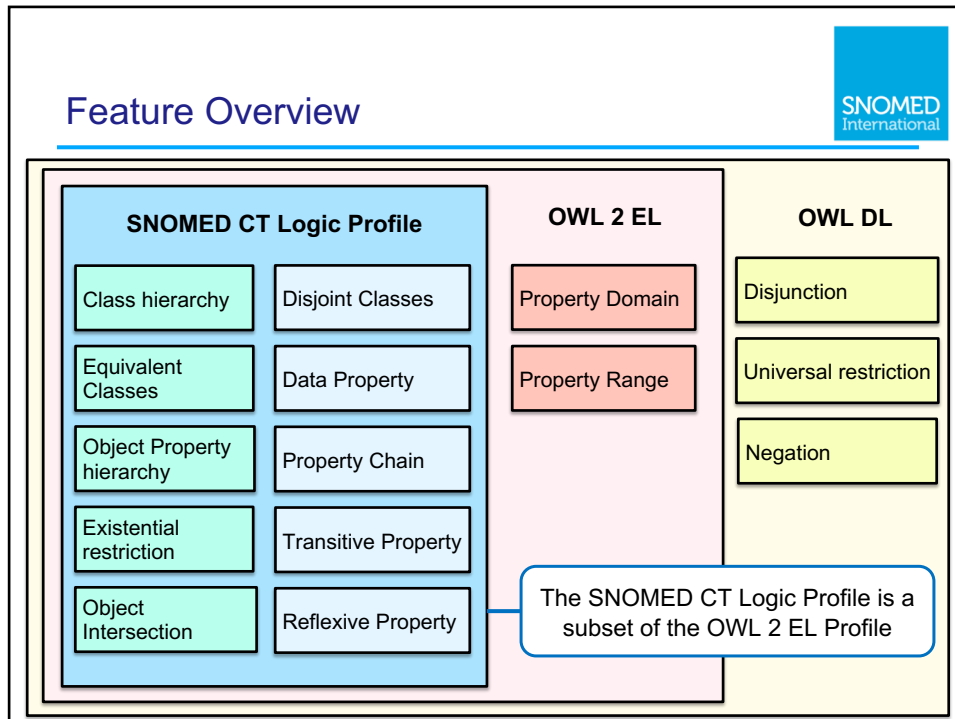
The diagram illustrates the changes to release files. A box at the top contains 'Description File', 'Concept File', and 'Relationship File (inferred view)'. Above this box is a brown box labeled 'Stated Relationship File (stated view)' which is crossed out with a large red 'X'. Below the main box is a purple box labeled 'OWL Axiom Refset File (stated view)'.




## SNOMED CT's DL Profile



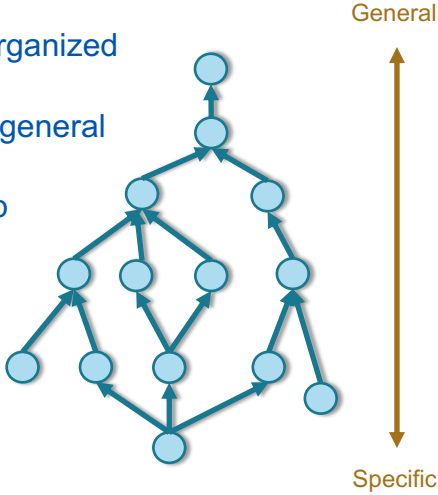
A central graphic consisting of ten colorful lightbulbs arranged in a circle. The colors include blue, green, yellow, orange, red, purple, and cyan.




## Class Hierarchy




- SNOMED CT concepts and expressions correspond to OWL classes
- SNOMED CT concepts are organized in a class hierarchy
- A supertype concept is more general than its subtypes
- The hierarchy can be as deep as needed



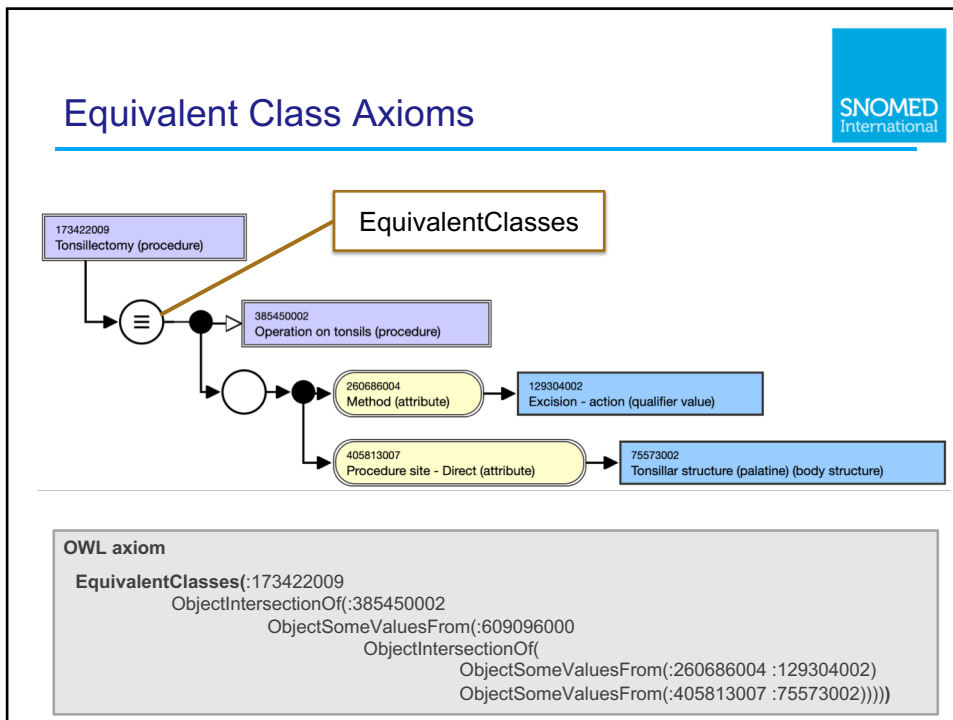
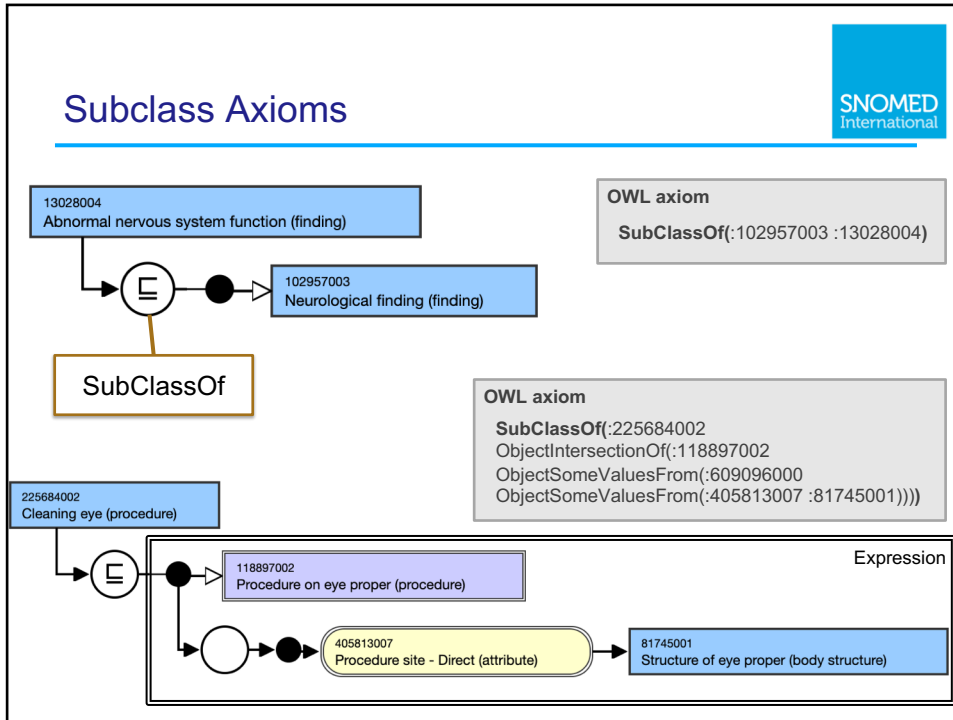
## SubClassOf and EquivalentClasses Axioms




- Concepts in SNOMED CT are defined using OWL axioms
  - SubClassOf axiom
    - When the defining properties do not represent the full semantics of the concept being defined
  - EquivalentClasses axiom
    - When the defining properties represent the full semantics of the concept being defined



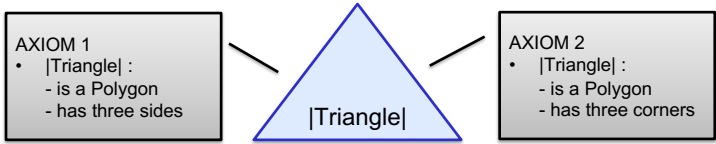




### Multiple Axioms

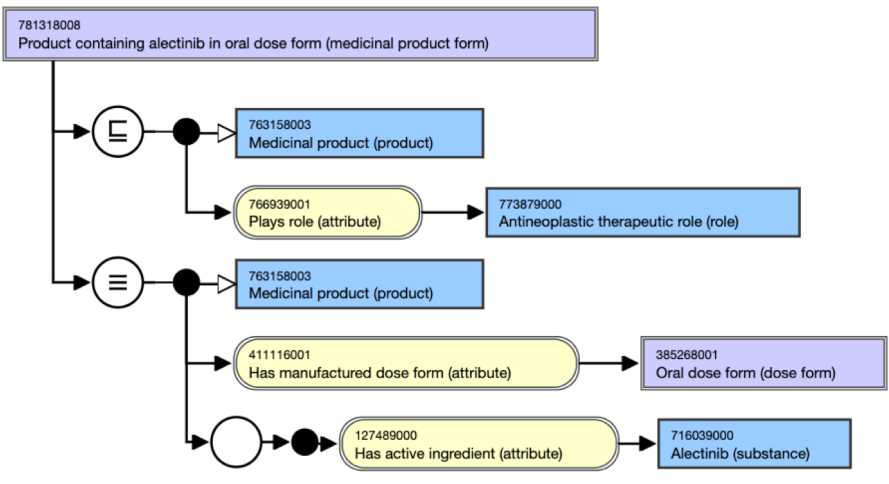




- Additional axioms are used when there is more than one definition of a concept



- Each axiom represents a definition which is always and necessarily true


### Multiple Axioms - Example






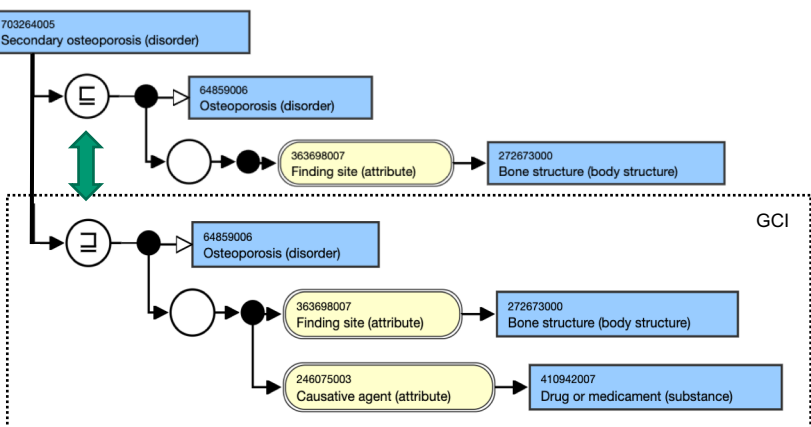
## General Concept Inclusion (GCI)

- Represents sufficient, but not necessary conditions
- Uses SubClassOf axiom from subaxiom to the concept (i.e. the reverse direction to a necessary definition)
- Supports the inference of subtypes for primitive concepts





## GCI – Example



**OWL axiom (GCI)**

```

SubClassOf(ObjectIntersectionOf(:64859006 ObjectSomeValuesFrom(:609096000
ObjectIntersectionOf(ObjectSomeValuesFrom(:246075003 :410942007)
ObjectSomeValuesFrom(:363698007 :272673000)))) :703264005
    
```

## GCI – Example

**Secondary osteoporosis (disorder)** ☆

SCTID: 703264005

703264005 | Secondary osteoporosis (disorder) |

*en* Pathologic osteoporosis

*en* Osteoporosis that results from medical conditions or treatments that interfere with the attainment of peak bone mass, contributing to the structural deterioration of bone tissue.

*en* Secondary osteoporosis (disorder)

*en* Secondary osteoporosis

In the **stated view** of the concept you can see multiple GCIs for the concept

**Axiom**

Finding site → Bone structure

**GCI**

After → Surgical procedure

Finding site → Bone structure

**GCI**

Finding site → Bone structure

Causative agent → Physical force

**GCI**

Finding site → Bone structure

Causative agent → Drug or medicament

**GCI**

Finding site → Bone structure

Due to → Disease

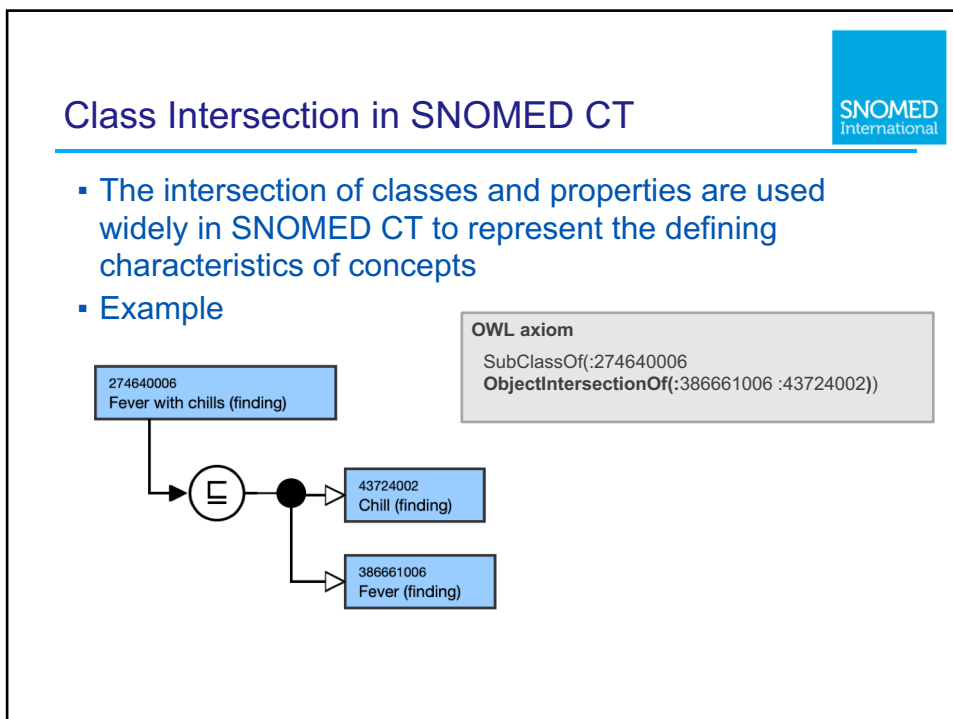
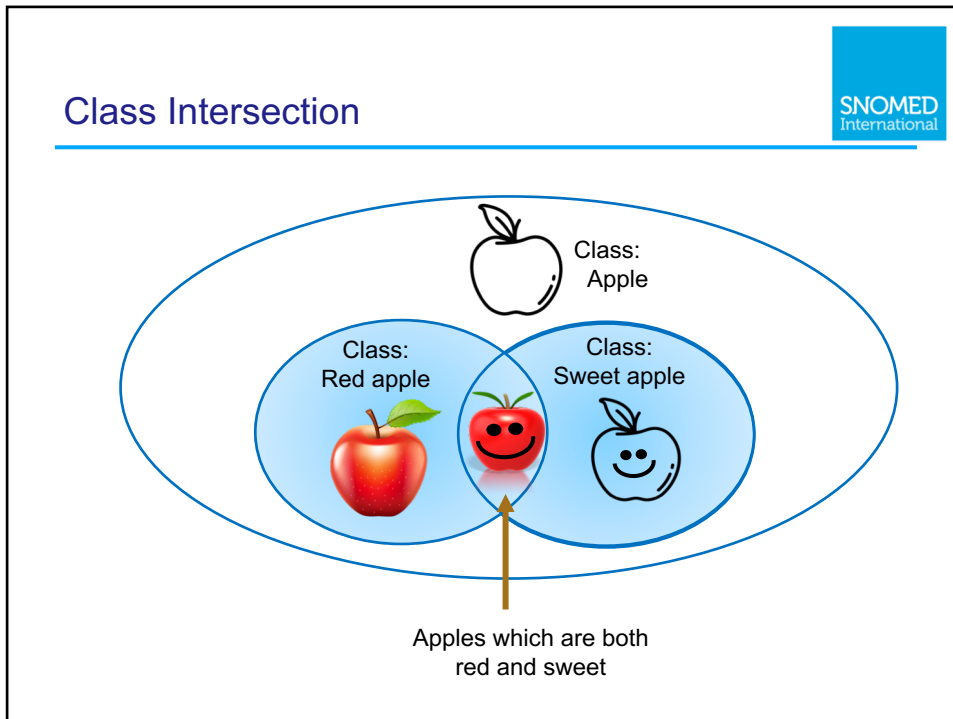
**GCI**

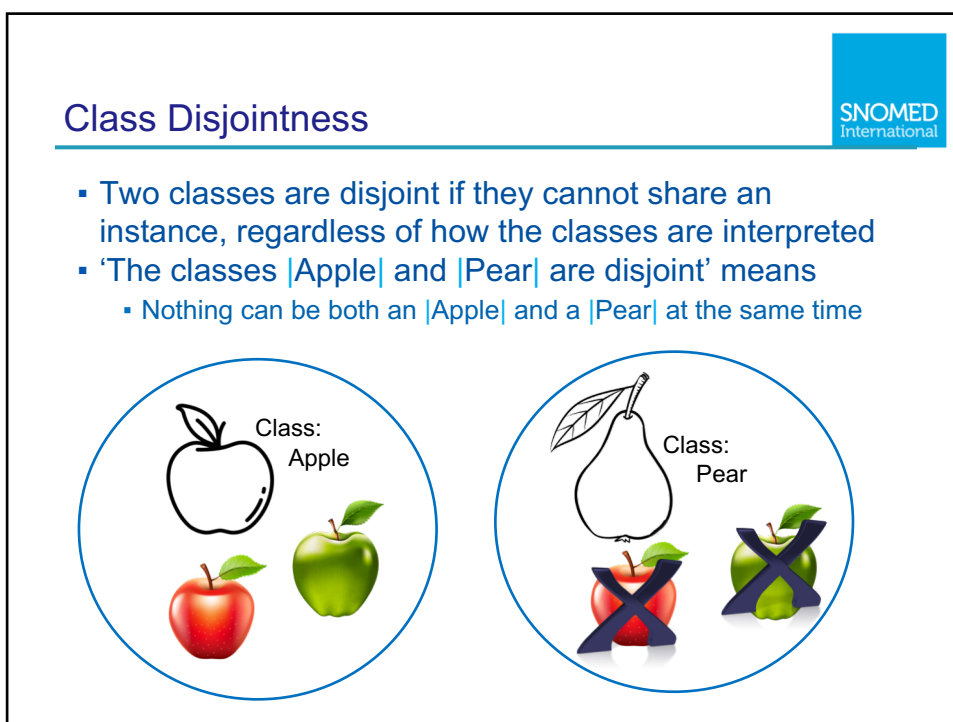
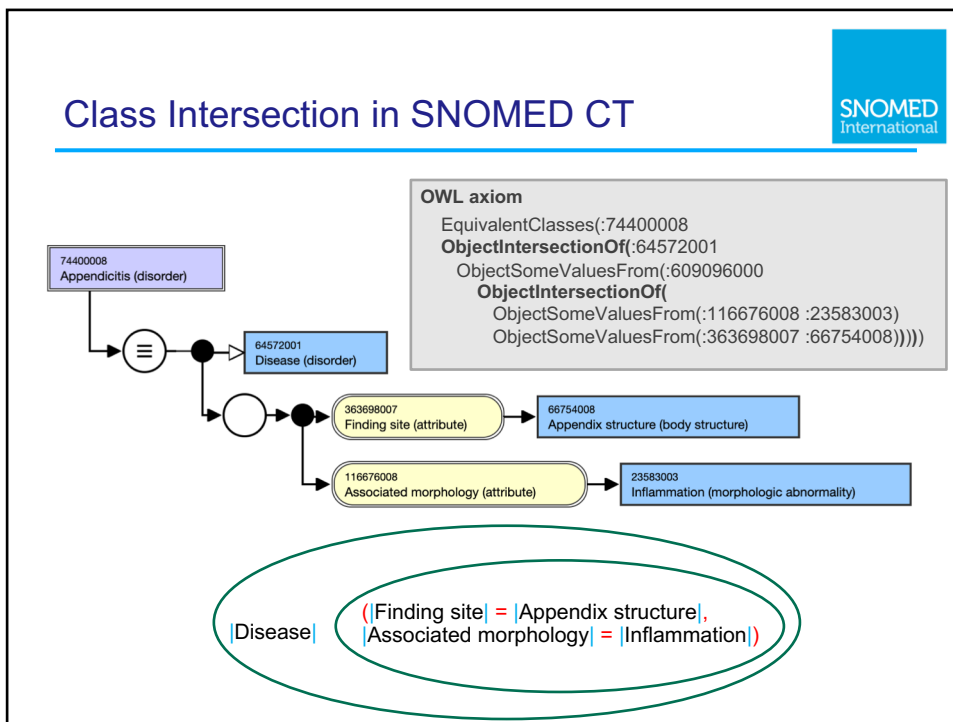
After → Disease

Finding site → Bone structure

## Properties and Restrictions

Properties and Restrictions

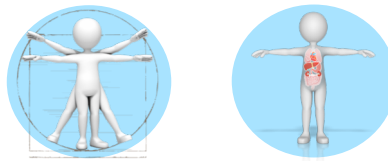




## Class Disjointness in SNOMED CT



- The majority of SNOMED CT concepts have the potential to overlap
- Some concepts are disjoint, i.e. they share no subtypes
- Important for quality assurance, e.g.
  - |Immaterial anatomical entity (body structure)| and |Material anatomical entity (body structure)| are disjoint



- Top-level hierarchies should be disjoint (although not explicitly declared)
  - Exception: |Physical object| and |Pharmaceutical / biologic product|

## Properties

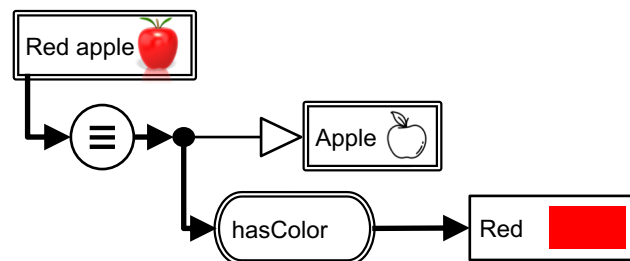


- Represent the characteristics of a class/concept
- In DL - attributes are represented as properties
- In SNOMED CT - attributes are represented as concepts
  - < 410662002 |Concept model attribute (attribute)|
  - Examples
    - 408729009 |Finding context (attribute)|
    - 704327008 |Direct site (attribute) |
    - 127489000 |Has active ingredient (attribute)|
    - 260686004 |Method (attribute)|
- The SNOMED CT concept model restricts the properties which can be used for particular types of concepts

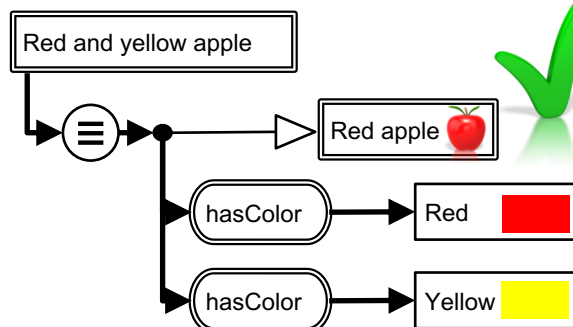
## Existential Restrictions



- A description logic axiom can state that
  - **Some** instances of a property have the specified value
- If a concept is a subtype of |Red apple|
  - It necessarily has at least one |hasColor| property with a value which is either |red|, or a subtype of |red|




## Existential Restrictions

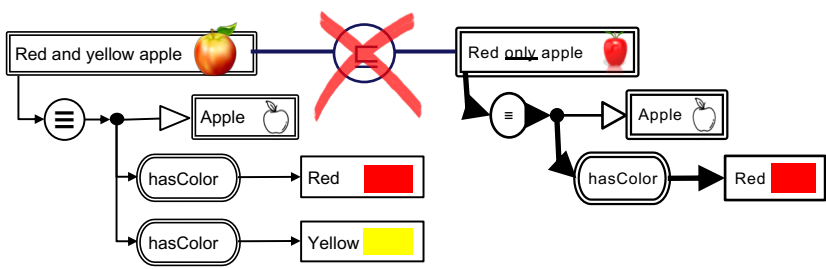





## Universal Restriction

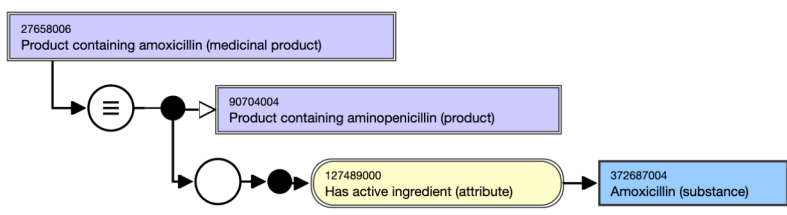


- A Description Logic axiom can state that
  - **All** instances of a property have the specified value
- If a concept is a subtype of **|Red only apple|**
  - If universal restriction is used, then the **only** value allowed for the **|hasColor|** property is **|red|** (or a subtype of **|red|**)
  - Universal restriction by itself does not require a value
    - i.e. There may be no **|hasColor|** property at all



## Existential Restrictions in SNOMED CT

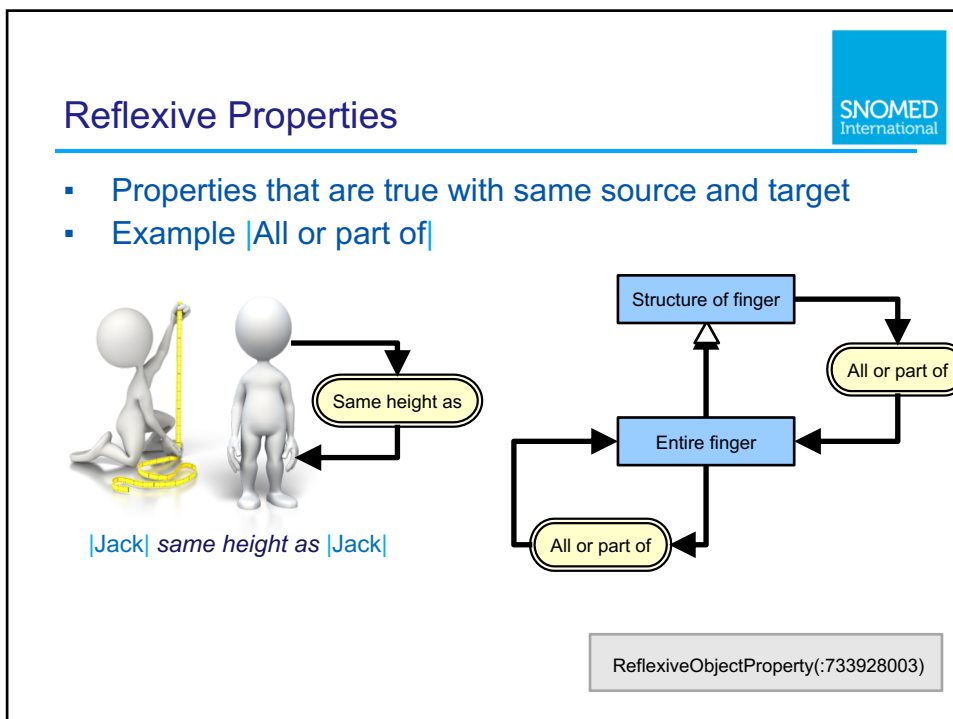
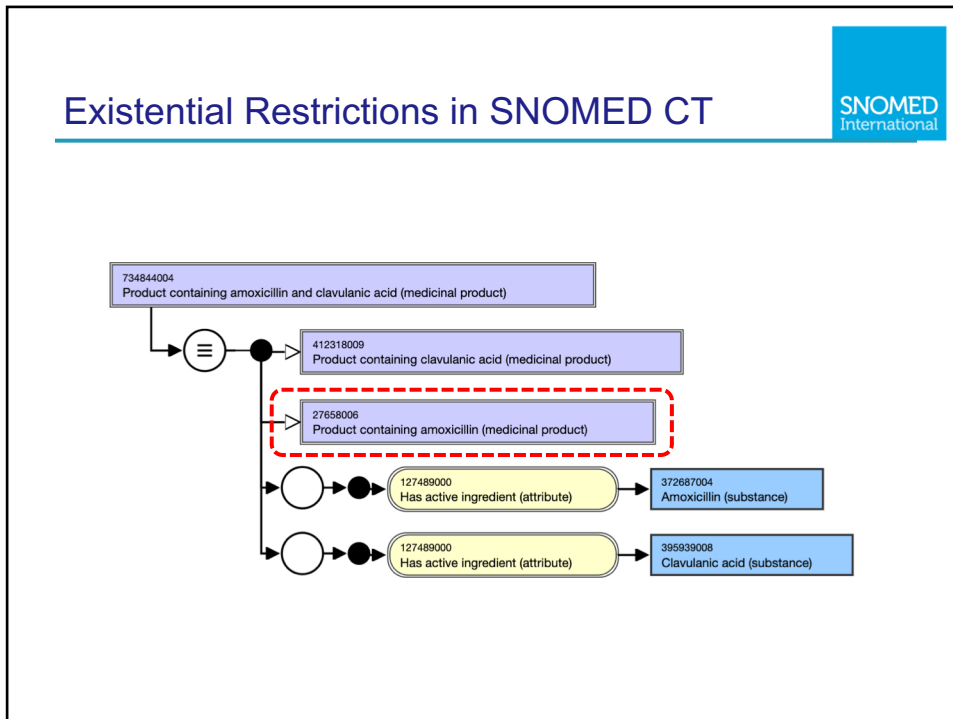




**OWL axiom**

```

EquivalentClasses(:27658006
  ObjectIntersectionOf(:763158003
    ObjectSomeValuesFrom(:609096000
      ObjectSomeValuesFrom(:127489000 :372687004))))
            
```




## SNOMED International

### Transitivity

- Some properties are transitive
  - E.g. |After|, |Regional part of|
- Enables the creation of alternative hierarchies
- For example, if
  - |Y| occurs |after| |X|, and
  - |Z| occurs |after| |Y|
  - ... this implies that
  - |Z| occurs |after| |X|

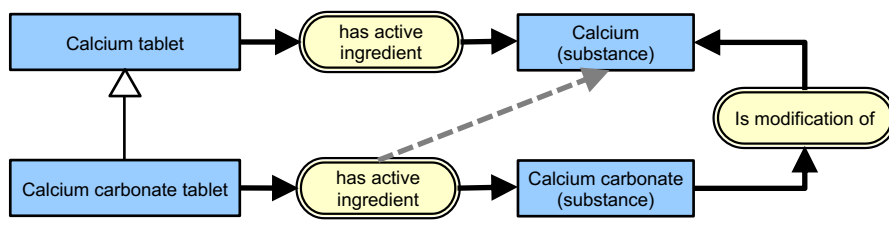
TransitiveObjectProperty(:733930001)




## SNOMED International

### Property Chain



- Similar to transitivity but involves more than one property
- Provides additional reasoning power between relationships for concept modelling
- Example
  - |Has active ingredient| chains with |is modification of|



SubObjectPropertyOf( ObjectPropertyChain(:127489000 :738774007) :127489000 )




## OWL Refsets



## Purpose


- Supports distribution and accurate representation of
  - Stated concept definitions
  - Information about SNOMED CT's OWL ontologies
- Enables the use of DL reasoners
- Supports the validation of OWL expressions
- Enables the creation of OWL ontologies for SNOMED CT




## Syntax for OWL Reference Sets

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- OWL functional-style syntax is recommended syntax
  - E.g. `SubClassOf(:40484003 :138875005)`
- Any compatible syntax may be used to render generated OWL ontology
- Syntax specification: <https://www.w3.org/TR/owl-syntax>






## |OWL expression type reference set|

---



Identification, versioning and modularization information	<b>id</b>	<b>UUID</b>
	<b>effectiveTime</b>	<b>Time</b>
	<b>active</b>	<b>Boolean</b>
	<b>moduleId</b>	<b>SCTID</b>
An identifier of the reference set ( <code>&lt;  OWL expression type reference set </code> )	<b>refsetId</b>	<b>SCTID</b>
The concept to which the OWL expression applies	<b>referencedComponentId</b>	<b>SCTID</b>
The text of the OWL expression to attach to the referenced component	<b>owlExpression</b>	<b>String</b>




## Types

---

- ▲ ● Reference set (foundation metadata concept)
  - ▼ ● OWL expression type reference set (foundation metadata concept)
    - ● OWL axiom reference set (foundation metadata concept)
    - ● OWL ontology reference set (foundation metadata concept)

- |OWL axiom reference set|
  - OWL expressions represent axioms that form the parts of the definition of the concept identified by the referencedComponentId


- |OWL ontology reference set|
  - OWL expressions represent essential information about an ontology



## |OWL ontology reference set|


---

- Represents essential information about the ontology
  - E.g. namespaces, ontology URI, ontology version URI, import statement
- Prefixes declared in the |OWL ontology reference set| minimize the size and improve readability of the |OWL axiom reference set|
  - Prefix ::= <http://snomed.info/id/>




Replaced by

- Following are valid references to the concept 64572001 |Disease|
  - http://snomed.info/id/64572001
  - :64572001



### Example

moduleId	refsetId	Referenced ComponentId	owlExpression
9000000000000012004	762103008	734147008	Ontology(< <a href="http://snomed.info/sct/9000000000000012004">http://snomed.info/sct/9000000000000012004</a> >)
9000000000000012004	762103008	734146004	Prefix(=< <a href="http://snomed.info/id/">http://snomed.info/id/</a> >)
9000000000000012004	762103008	734146004	Prefix(owl:=< <a href="http://www.w3.org/2002/07/owl/#">http://www.w3.org/2002/07/owl/#</a> >)
9000000000000012004	762103008	734146004	Prefix(xml:=< <a href="http://www.w3.org/XML/1998/namespace">http://www.w3.org/XML/1998/namespace</a> >)
9000000000000012004	762103008	734146004	Prefix(xsd:=< <a href="http://www.w3.org/2001/XMLSchema#">http://www.w3.org/2001/XMLSchema#</a> >)




### Example


moduleId	refsetId	Referenced ComponentId	owlExpression
SNOMED CT model component module	OWL ontology reference set	OWL ontology header	Ontology(< <a href="http://snomed.info/sct/9000000000000012004">http://snomed.info/sct/9000000000000012004</a> >)
SNOMED CT model component module	OWL ontology reference set	OWL ontology namespace	Prefix(=< <a href="http://snomed.info/id/">http://snomed.info/id/</a> >)
SNOMED CT model component module	OWL ontology reference set	OWL ontology namespace	Prefix(owl:=< <a href="http://www.w3.org/2002/07/owl/#">http://www.w3.org/2002/07/owl/#</a> >)
SNOMED CT model component module	OWL ontology reference set	OWL ontology namespace	Prefix(xml:=< <a href="http://www.w3.org/XML/1998/namespace">http://www.w3.org/XML/1998/namespace</a> >)
SNOMED CT model component module	OWL ontology reference set	OWL ontology namespace	Prefix(xsd:=< <a href="http://www.w3.org/2001/XMLSchema#">http://www.w3.org/2001/XMLSchema#</a> >)

## |OWL axiom reference set|

---


- 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





## Example

---



moduleId	refsetId	Referenced ComponentId	owlExpression
900000000000207008  SNOMED CT core module	733073007  OWL axiom reference set	404684003  Clinical finding (finding)	SubClassOf( :404684003 :138875005)
900000000000207008  SNOMED CT core module	733073007  OWL axiom reference set	90708001  Kidney disease (disorder)	EquivalentClasses(:90708001 ObjectIntersectionOf(:64572001 ObjectSomeValuesFrom(:609096 000 ObjectSomeValuesFrom( :363698007 :64033007))))
90000000000012004  SNOMED CT model component module	733073007  OWL axiom reference set	733930001  Regional part of (attribute)	TransitiveObjectProperty( :733930001)



### SNOMED International

## SubClassOf Example

moduleId	refsetId	referenced ComponentId	owlExpression
900000000000207008 SNOMED CT core module	733073007 OWL axiom reference set	404684003 Clinical finding (finding)	SubClassOf( :404684003 :138875005)

### SNOMED International

## EquivalentClass Example

moduleId	refsetId	referenced ComponentId	owlExpression
900000000000207008 SNOMED CT core module	733073007 OWL axiom reference set	90708001 Kidney disease (disorder)	EquivalentClasses(:90708001 ObjectIntersectionOf(:64572001 ObjectSomeValuesFrom(:609096000 ObjectSomeValuesFrom(:363698007 :64033007))))

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

### SNOMED International

## EquivalentClass Example

**Syntax: EquivalentClasses (A B)**

The diagram illustrates the equivalent class relationship. On the left, a box contains '90708001 Kidney disease (disorder)'. An arrow points from this box to a central circle containing a horizontal line (the equivalent class symbol). From this central circle, an arrow points to a box containing '64572001 Disease (disorder)'. Another arrow points from the central circle to a larger box containing a sequence of elements: a circle, an arrow, a box with '363698007 Finding site (attribute)', an arrow, and a box with '64033007 Kidney structure (body structure)'.

```


EquivalentClasses(
  90708001 |Kidney disease (disorder)|
  ObjectIntersectionOf(
    :64572001 |Disease (disorder)|
    ObjectSomeValuesFrom(
      :609096000 |Role group (attribute)|
      ObjectSomeValuesFrom(
        :363698007 |Finding site (attribute)|
        ObjectSomeValuesFrom(
          :64033007 |Kidney structure (body structure)|
        )))
  )
  
```

### SNOMED International

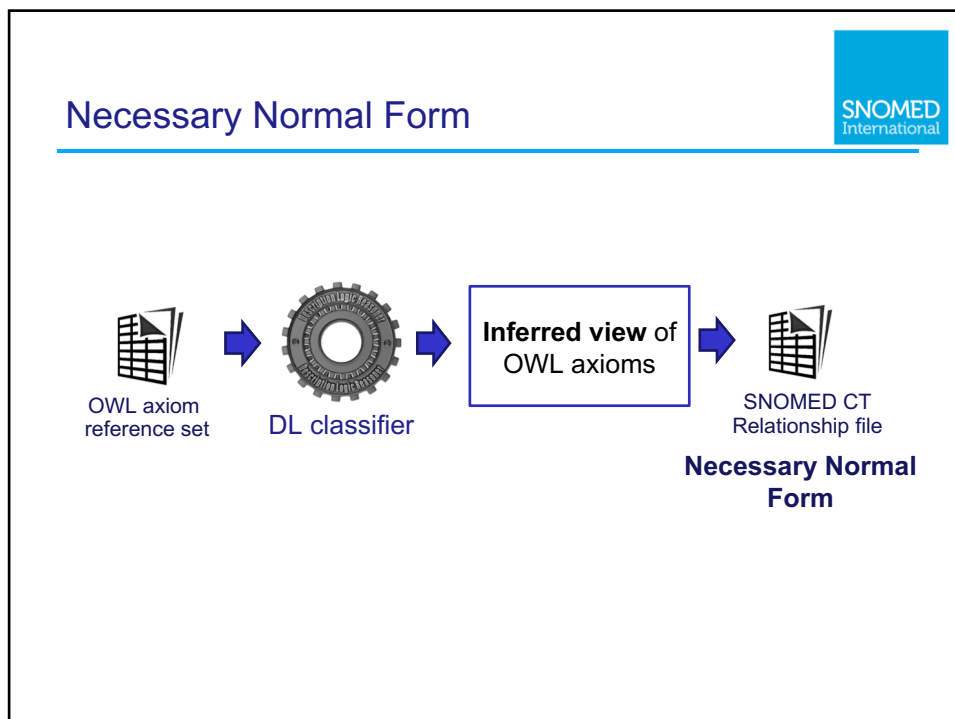

## TransitiveObjectProperty Example

The diagram shows three 3D models representing body parts. From left to right: a finger joint region, a hand joint region, and a full joint region. Dashed arrows labeled 'Regional part of' connect the finger joint to the hand joint, the hand joint to the full joint, and the finger joint directly to the full joint, demonstrating the transitive nature of the property.


moduleId	refsetId	Referenced ComponentId	owlExpression
900000000000012004  SNOMED CT model component module	733073007  OWL axiom reference set	733930001  Regional part of	TransitiveObjectProperty(           :733930001  Regional part of )



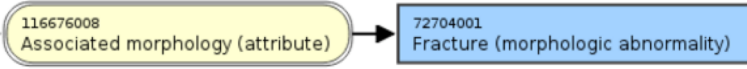
## Understanding the Relationship File



## Necessary Condition




- A characteristic that is *always and necessarily true*
- For example
  - Necessary condition for |Fracture of bone (disorder)|

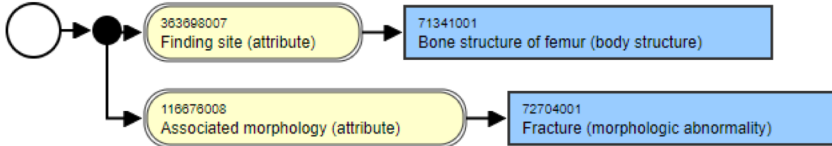


```
graph LR; A(116676008 Associated morphology (attribute)) --> B[72704001 Fracture (morphologic abnormality)]
```

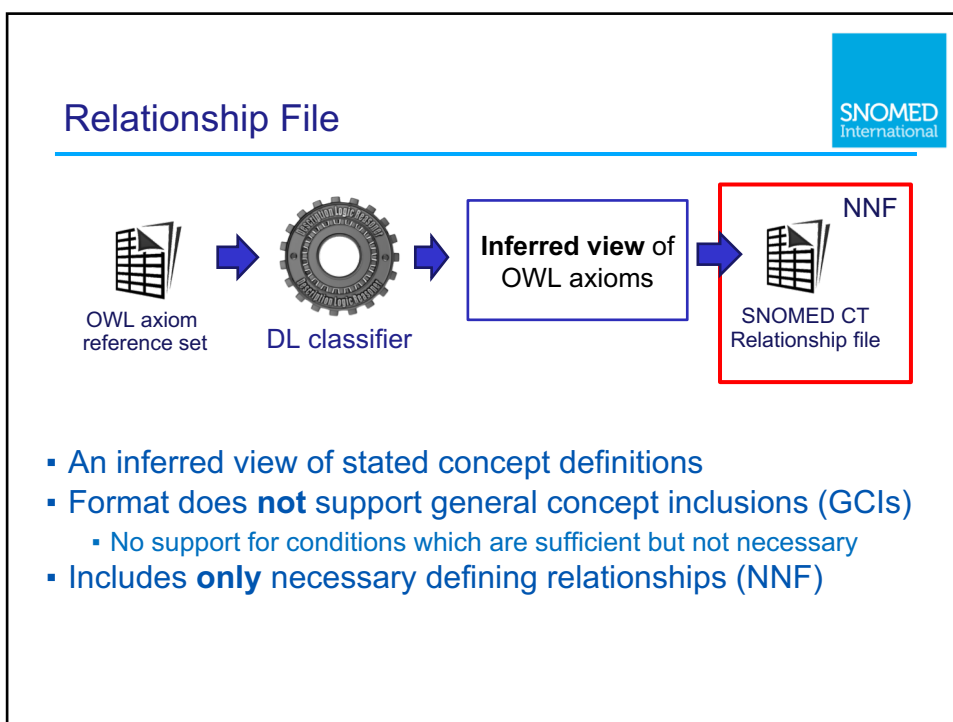
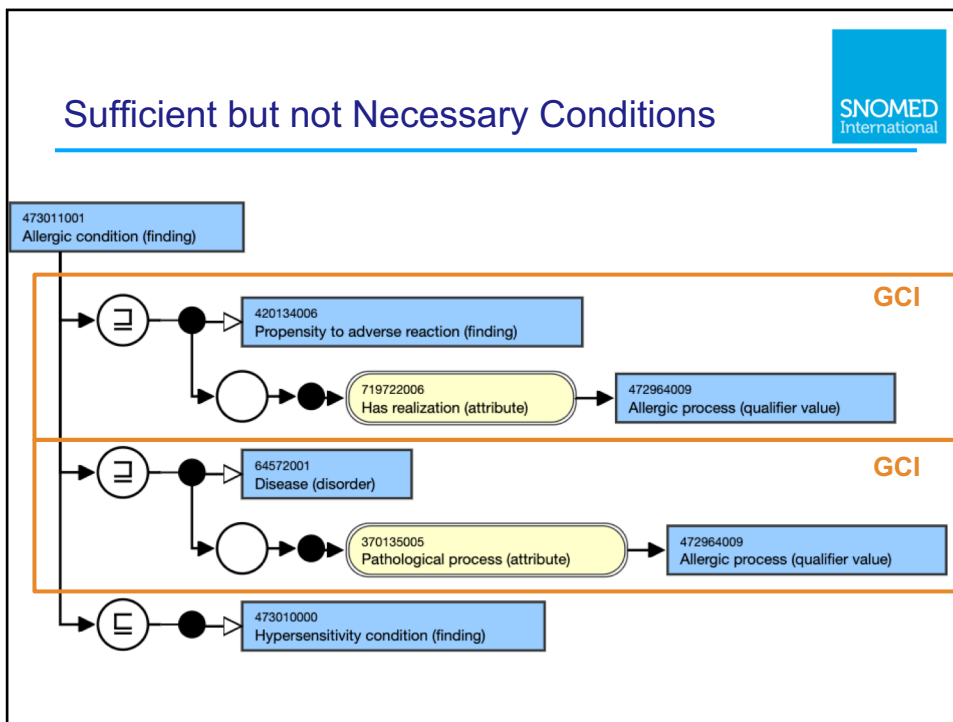
## Sufficient Conditions

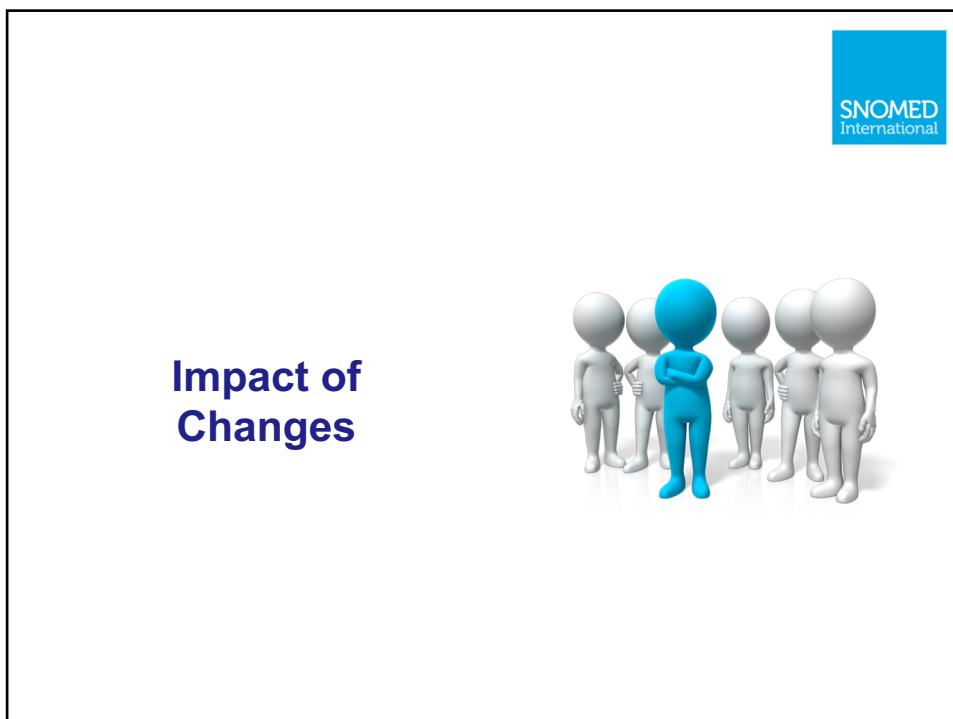
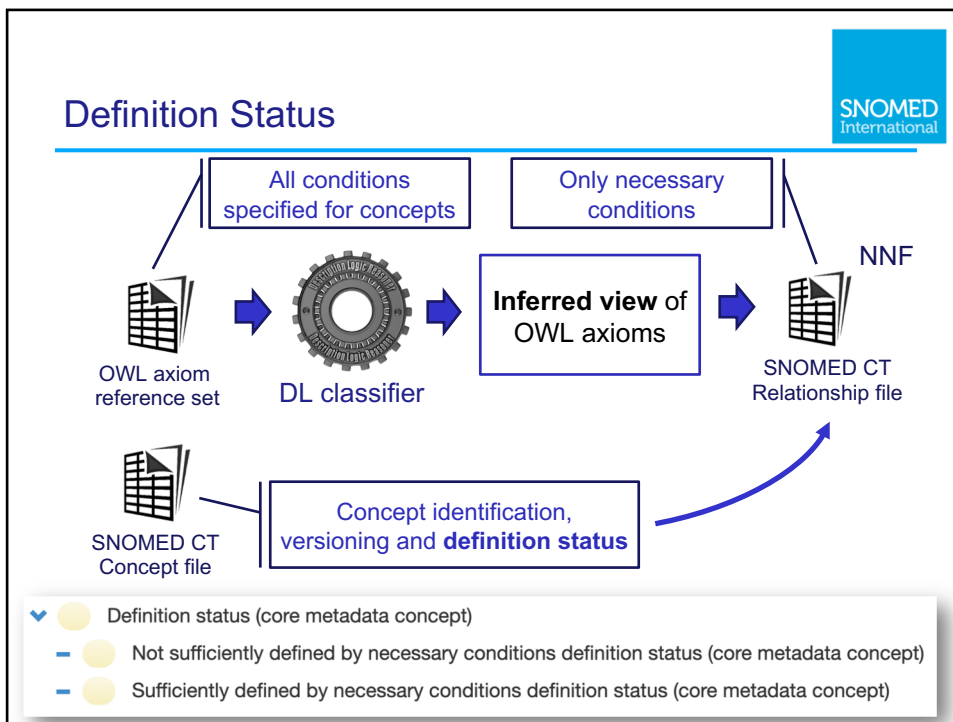


- A set of conditions that are adequate to infer the full meaning of the concept
- For example
  - Sufficient conditions for the concept |Fracture of femur|



```
graph LR; Start(( )) --> Node(( )); Node --> A(363898007 Finding site (attribute)); Node --> B(116676008 Associated morphology (attribute)); A --> C[71341001 Bone structure of femur (body structure)]; B --> D[72704001 Fracture (morphologic abnormality)]
```





## Applications That Use Inferred Relationships



- No impact for most applications
  - Precoordinated content is preclassified before distribution
  - Inferred relationships in relationship file will continue to be available for navigation, display and query
  - Applications will benefit from a more accurate inferred subtype hierarchy in the relationship file
- Exception
  - Testing subsumption of postcoordinated expressions will not be possible using the relationship file
    - Advanced OWL semantics not captured in Relationship file
    - Inferred relationships are accurate but no longer complete
  - A DL classifier must be used to test expression subsumption

## Applications That Use Stated Relationships



- Stated relationship file is no longer available
  - Functions that use the stated relationships will need to be updated to use the OWL axiom refset
  - Using a DL classifier will enable the OWL axiom refset to replace and enhance functions that use stated relationships
- Benefits of migrating to the OWL axiom refset include
  - More accurate classification based on enhanced definitions using OWL Functional Syntax
  - Use of the SNOMED OWL toolkit to combine OWL axioms into a file ready for DL classification

## Extensions



- Extensions with concepts or relationships
  - Stated relationships in extension must be converted to axioms
  - New concepts must be defined by adding to OWL axiom refset
  - Updates to concept definitions require changes to OWL axioms
  - Classification of extension must use OWL axiom refset as input
- Extensions without concepts or relationships
  - Adding descriptions – no impact
  - Adding refset members – no impact
  - However ...
    - All extensions contain at least one module concept
    - Extensions with new refsets need new refset concepts
  - Therefore ...
    - Consult guidance on representing relationships as axioms

## Summary of Impact



- Who will need to change
  - Applications currently using stated relationships
  - Applications testing subsumption of postcoordinated expressions
  - Extensions with concepts or relationships
- Who will **not** be impacted by these changes
  - All other systems using only
    - Concepts
    - Relationships (inferred)
    - Descriptions
    - Reference sets
- Who will benefit from changes
  - ALL users of SNOMED CT

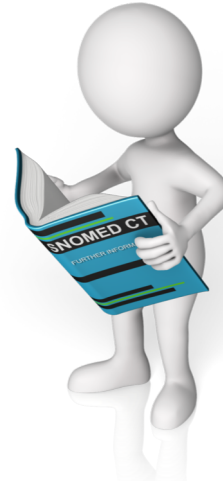




## Further Information

SNOMED  
International

- SNOMED CT OWL guide
  - <http://snomed.org/owl>
- SNOMED CT logic profile specification
  - <http://snomed.org/lps>
- OWL 2 web ontology language primer
  - <https://www.w3.org/TR/owl2-primer/>
- OWL 2 structural specification and functional-style syntax
  - <https://www.w3.org/TR/owl2-syntax/>
- OWL 2 web ontology language profiles
  - <https://www.w3.org/TR/owl2-profiles/>



## Questions and Discussion

SNOMED  
International



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