

SNOMED CT OWL Axioms

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SNOMED
International





Learning Objectives

1. Understand how and why SNOMED CT in OWL is provided using a RefSet
2. Gain insight into generation of OWL-files from the SNOMED CT OWL RefSet
3. Gain insight into role and use of the OWL-file in semantic web applications
4. Understand the value of using OWL for Members and Affiliates



OWL - Web Ontology Language

- A Semantic Web language to represent rich and complex knowledge (things, groups of things, and relations between things).
- A computational logic-based language
 - OWL ontologies provide classes, properties, individuals and data values and are stored as Semantic Web documents
- One of the distinguishing features of OWL is that it can be used to express extremely complicated and subtle ideas about your data.
- Primary uses
 - Fast and flexible data modeling
 - Efficient automated reasoning



Before July 2019 - OWL conversion

OWL-version of SNOMED CT can be generated from RF2 tables

- Concept
- (Stated) Relationship
- Description

Two available transformations

- Spackman OWL script `tls2_StatedRelationshipsToOwlKRSS_INT.pl`
- SNOMED OWL Toolkit <https://github.com/IHTSDO/snomed-owl-toolkit>



Before July 2019 - Drawbacks

- Limited expressiveness in RF2 tables
- Implicit knowledge included in OWL-transformation scripts
 - Role chains, e.g.,
 - 127489000 |Has active ingredient (attribute)| o 738774007 |Is modification of (attribute)| \sqsubseteq 127489000 |Has active ingredient (attribute)|
 - Transitivity of relationships, e.g.,
 - 733928003 |All or part of (attribute)|
 - 738774007 |Is modification of (attribute)|
 - Reflexivity, e.g.,
 - 733928003 |All or part of (attribute)|
 - 738774007 |Is modification of (attribute)|



From July 2019 - Possibilities

In theory:

Full OWL expressiveness

In practice:

- Multiple axioms
- Role chains; transitivity; reflexivity
- Generalized Concept Inclusions (GCI's)
- Use of concrete domains (mainly: numbers)



Multiple axioms

Parents

- ● Medicinal product (product)
- ● Medicinal product (product)

☰ Product containing drospirenone and ethinylestradiol (medicinal product) ★ ↗

SCTID: 416617005

416617005 | Product containing drospirenone and ethinylestradiol (medicinal product) |

Axiom

Plays role → Contraceptive therapeutic role

Axiom

Has active ingredient → Drospirenone

Has active ingredient → Ethinylestradiol



Axiom 1

sct2_sRefset_OWLExpressionFull_INT_20190731.txt

id	95587240-b1c1-4767-a8c5-84ad8cbb8253
effectiveTime	20190731
active	1
moduleId	SNOMED CT core module
refsetId	OWL axiom reference set
referencedComponentId	416617005 Product containing drospirenone and ethinylestradiol (medicinal product)
owlExpression	SubClassOf(:416617005 ObjectIntersectionOf(:763158003 ObjectSomeValuesFrom(:766939001 :773895000)))

Plays role (attribute)

Contraceptive therapeutic role (role)

Medicinal product (product)



Axiom 2

sct2_sRefset_OWLExpressionFull_INT_20190731.txt

id	9f84a281-0e62-43f8-8055-66f7599f7894
effectiveTime	20190731
active	1
moduleId	SNOMED CT core module
refsetId	OWL axiom reference set
referencedComponentId	416617005 Product containing drospirenone and ethinylestradiol (medicinal product)
owlExpression	EquivalentClasses(:416617005 ObjectIntersectionOf(:763158003 ObjectSomeValuesFrom(:609096000 ObjectSomeValuesFrom(:127489000 :126097006)) ObjectSomeValuesFrom(:609096000 ObjectSomeValuesFrom(:127489000 :410919000))))



Role chains, transitivity, reflexivity

- Role chains, e.g.,
 - 127489000 | Has active ingredient (attribute) | o 738774007 | Is modification of (attribute) | \sqsubseteq 127489000 | Has active ingredient (attribute) |
→ `SubObjectPropertyOf(ObjectPropertyChain(:127489000 :738774007) :127489000)`
- Transitivity
 - `TransitiveObjectProperty(:738774007)`
- Reflexivity
 - `ReflexiveObjectProperty(:738774007)`



Generalized Concept Inclusions (GCI's)

- 'Propensity to adverse reaction (finding)' AND
('Role group (attribute)' some (
 'Has realization (attribute)' some
 'Hypersensitivity process (qualifier value)'
)
) **SubClassOf**
 'Hypersensitivity condition (finding)'



OWL RefSet - benefits

- Versioning of OWL axioms
 - Challenging in OWL-only
- Maintaining RF2 infrastructure



How to get “pure” OWL-file

- SNOMED OWL Toolkit <https://github.com/IHTSDO/snomed-owl-toolkit>
- Do it yourself
 - From a snapshot, grab all **active** OWL axioms
 - Get all the Prefix statements
 - Get the Ontology declaration
 - Get all other axioms
 - Create AnnotationAssertions from active Descriptions
 - Polish



“Pure” OWL

```
Prefix(owl:=<http://www.w3.org/2002/07/owl#>)  
Prefix(xml:=<http://www.w3.org/XML/1998/namespace>)  
Prefix(rdf:=<http://www.w3.org/1999/02/22-rdf-syntax-ns#>)  
Prefix(rdfs:=<http://www.w3.org/2000/01/rdf-schema#>)  
Prefix(xsd:=<http://www.w3.org/2001/XMLSchema#>)  
Prefix(:=<http://snomed.info/id/>)  
Ontology(<http://snomed.info/sct/900000000000207008>  
...  
SubClassOf(:59770006 :106170009)  
AnnotationAssertion(rdfs:label :59770006 "Dyslexia (finding)"@en)  
...  
)
```



What's the use

Class hierarchy: Family Strigidae (organism)

Family Steatornithidae (organism) Family Strigidae (organism) Subfamily Asioninae (organism) Genus Asio (organism) Asio flammeus (organism) Asio otus (organism) Asio otus wilsonianus (organism) Genus Nesasio (organism) Genus Pseudoscops (organism) Pseudoscops clamator (organism) Subfamily Striginae (organism) Genus Bubo (organism) Genus Ciccaba (organism) Genus Gymnoglaux (organism) Genus Jubula (organism) Genus Ketupa (organism) Genus Lophotrix (organism) Genus Megascops (organism) Megascops asio (organism) Megascops kennicottii (organism) Genus Mimizuku (organism) Genus Otus (organism) Otus flammeolus (organism) Otus scops (organism) Genus Ptilopsis (organism) Genus Pulsatrix (organism) Pulsatrix perspicillata (organism) Genus Pyrroglaux (organism) Genus Scotopelia (organism) Scotopelia peli (organism) Genus Strix (organism) Strix aluco (organism) Strix nebulosa (organism) Strix occidentalis (organism) Strix varia (organism) Subfamily Surininae (organism) Genus Aegolius (organism) Aegolius acadicus (organism) Genus Athene (organism) Athene cunicularia (organism) Athene noctua (organism) Genus Glaucidium (organism) Glaucidium gnoma (organism) Glaucidium gnoma californicum (organism) Genus Heteroglaux (organism) Genus Micrathene (organism) Micrathene whitneyi (organism) Genus Ninox (organism)

Family Strigidae (organism) — http://snomed.info/id/107109004

Class Annotations Class Usage

Annotations: Family Strigidae (organism)

Annotations +

rdfs:label [type: xsd:string]

Family Strigidae (organism)

Description: Family Strigidae (organism)

Equivalent To +

SubClass Of +

- Order Strigiformes (organism)

General class axioms +

SubClass Of (Anonymous Ancestor)

Instances +

Target for Key +

Disjoint With +

Disjoint Union Of +



Protege

Description: Product containing drospirenone and ethinylestradiol (medicinal product)

Equivalent To 




- **'Medicinal product (product)'**
and ('Role group (attribute)' some ('Has active ingredient (attribute)' some 'Ethinylestradiol (substance)'))
and ('Role group (attribute)' some ('Has active ingredient (attribute)' some 'Drospirenone (substance)'))

SubClass Of 

- **'Medicinal product (product)'**
and ('Plays role (attribute)' some 'Contraceptive therapeutic role (role)')



Viewing, editing, reasoning!













 Transplanted organ failure (disorder)  

SCTID: 269295009

269295009 | Transplanted organ failure (disorder) |




en Transplanted organ failure
en Transplanted organ failure (disorder)

Children (6)

-   Bone marrow transplant failure (disorder)
-   Cardiac transplant failure (disorder)
-   Failed renal transplant (disorder)
-   Liver transplant failure (disorder)
-   Transplant failure of cornea of left eye (disorder)
-   Transplant failure of cornea of right eye (disorder)



Viewing, editing, reasoning!

 Transplanted organ failure (disorder)  













SCTID: 269295009

269295009 | Transplanted organ failure (disorder) |

en Transplanted organ failure
en Transplanted organ failure (disorder)

 Transplant failure of cornea

Children (6)

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-   Cardiac transplant failure (disorder)
-   Failed renal transplant (disorder)
-   Liver transplant failure (disorder)
-   Transplant failure of cornea of left eye (disorder)
-   Transplant failure of cornea of right eye (disorder)



Viewing, editing, reasoning!

● Transplanted organ failure (disorder) ☆

SCTID: 269295009

269295009 | Transplanted organ failure (disorder) |

en Transplanted organ failure

en Transplanted organ failure (disorder)

Children (6)

- ☰ Bone marrow transplant failure (disorder)
- ☰ Cardiac transplant failure (disorder)
- ☰ Failed renal transplant (disorder)
- ☰ Liver transplant failure (disorder)
- ☰ Transplant failure of cornea of left eye (disorder)
- ☰ Transplant failure of cornea of right eye (disorder)

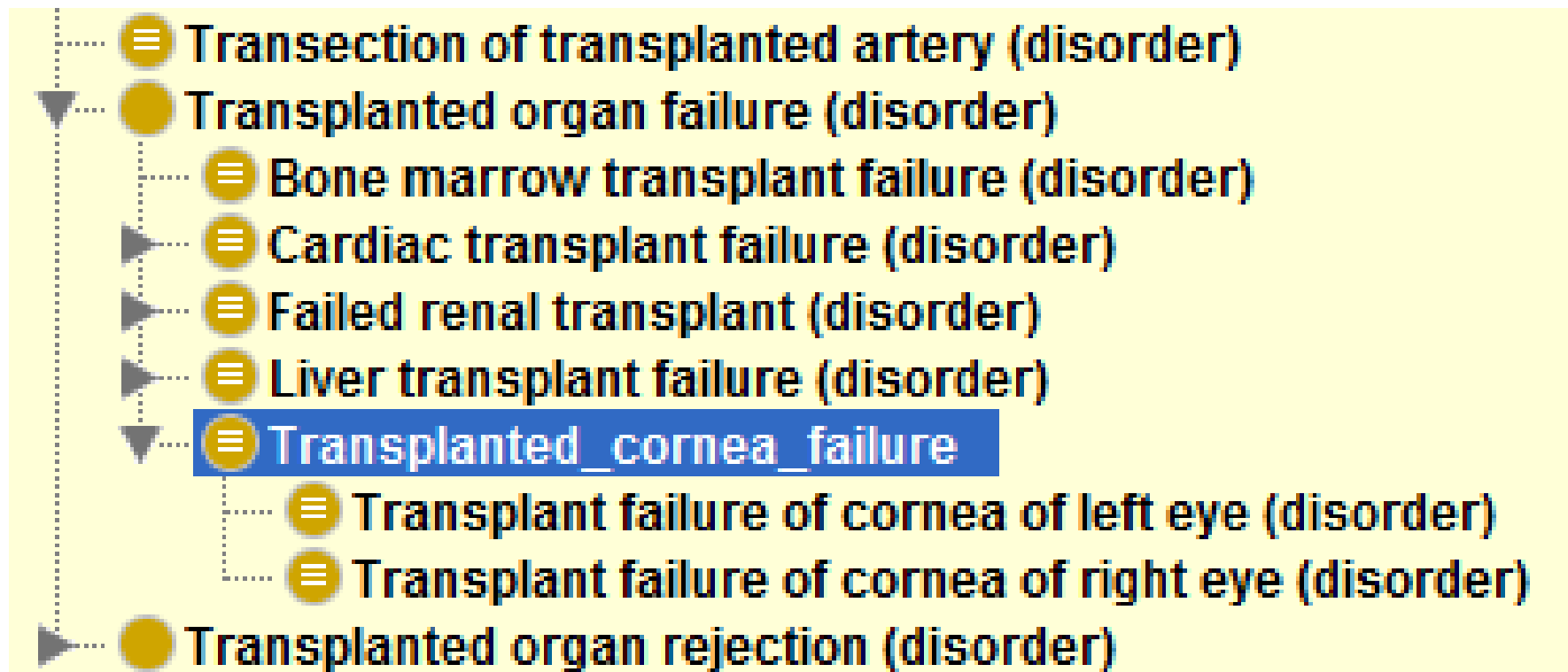
☰ Transplant failure of cornea

OWL:

EquivalentClass('Transplant failure of cornea (disorder)' 'Transplanted organ failure (disorder)' and ('Role group (attribute)' some ('Associated with (attribute)' some 'Corneal transplant (procedure)')) and ('Role group (attribute)' some ('Finding site (attribute)' some 'Structure of transplanted cornea (body structure)'))

NB: ECL =

```
<< 269295009 |Transplanted organ failure (disorder)| :  
    47429007 |Associated with (attribute)| =  
    << 60656008 |Corneal transplant (procedure)| ,  
    363698007 |Finding site (attribute)| =  
    << 10481004 |Structure of transplanted cornea  
(body structure)|
```



Equivalent To 

- 'Transplanted organ failure (disorder)'
 - and ('Role group (attribute)' some ('Associated with (attribute)' some 'Corneal transplant (procedure)'))
 - and ('Role group (attribute)' some ('Finding site (attribute)' some 'Structure of transplanted cornea (body structure)'))



12th International SWAT4HCLS Conference

Semantic Web Applications and Tools for Health Care and Life Sciences



December 9-12, 2019
Edinburgh, Scotland

<http://swat4hcls.org>



Summary

- You don't need to use OWL, but it is the “source of truth” from which RF2 Relationships table is derived
- OWL makes knowledge explicit as axioms, no “hidden semantics”
- OWL in RF2 format enables versioning with well-know principles
- RF2 OWL-refset can be easily transformed into an OWL-file
- OWL-files are widely used in the semantic web community, many tools around: reasoners, editors, etc.
- Opening up possibilities for maintenance and use of SNOMED CT



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- More on Friday November 1st, 13:30-15:00 “[Understanding SNOMED CT's Description Logic Enhancements](#)” by *Linda Bird and Anne Højen (SNOMED International)*



Thank you

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