The Philosophy of Medicine
Die Philosophie der Medizin

31 July & 1 August 2009 Internationales Begegnungszentrum

Bergstraße 7a, 18057 Rostock

What do SNOMED CT Concepts Represent?

Stefan Schulz

University Medical Center, Freiburg, Germany

Why ontology matters for medicine

Natural and Political

OBSERVATIONS

Mentioned in a following INDEX

and made upon the

Bills of Mortality.

RY

Capt. 70HN GRAUNT,

Fellow of the Royal Society.

With reference to the Government, Religion, Trade, Growth, Air, Diseases, and the several Changes of the said CITY.

— Non, me ut miretur Turba, laboro, Contentus paucis Lectoribus. —-

The Third EDITION, much Enlarged.

Printed by John Martyn, and James Allestry,
Printers to the Royal Society, and are to be sold at the
sign of the Bell in St. Pauls Church-yard.
MDC LX V

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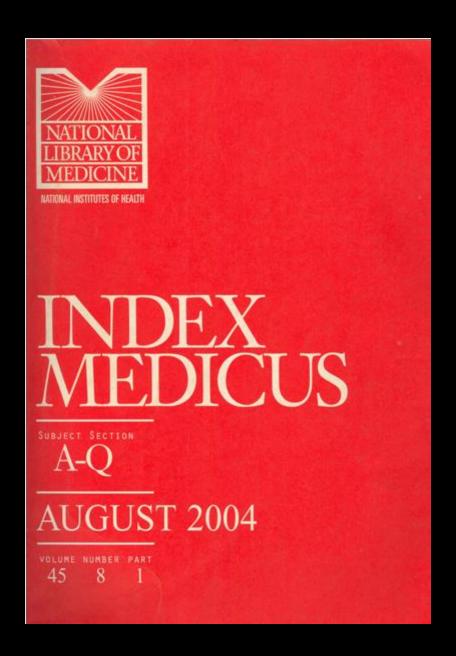
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International Classification of Diseases



Why ontologies matter for medicine

- Create taxonomies of natural kinds
 - classify instances in the world
 - basic for health statistics
 - exemplified in disease & procedure classification systems



Medical Subject Headings



National Library of Medicine - Medical Subject Headings

2006 MeSH

MeSH Descriptor Data

Return to Entry Page

MeSH Heading	Staphylococcus aureus
Tree Number	B03.510.400.790.750.100
Annotation	infection = STAPHYLOCOCCAL INFECTIONS & do not bother to coord with S. aureus unless particularly discussed (index IM); DF: STAPH AUREUS
Scope Note	Potentially pathogenic bacteria found in nasal membranes, skin, hair follicles, and perineum of warm-blooded animals. They may cause a wide range of infections and intoxications.
Allowable Qualifiers	CH CL CY DE EN GD GE IM IP ME PH PY RE UL VI
Entry Version	STAPH AUREUS
Previous Indexing	Staphylococcus (1966-1974)
Online Note	use STAPHYLOCOCCUS AUREUS to search MICROCOCCUS PYOGENES 1975-91; use STAPHYLOCOCCUS 1966-74
History Note	76; was MICROCOCCUS PYOGENES see under STAPHYLOCOCCUS 1963-75; MICROCOCCUS PYOGENES was see STAPHYLOCOCCUS AUREUS 1976-91
Unique ID	D013211

MeSH Tree Structures

Bacteria [B03]

Gram-Positive Bacteria [B03.510]

Gram-Positive Cocci [B03.510.400]

Staphylococcaceae [B03.510.400.790]

Staphylococcus [B03.510.400.790.750]

Staphylococcus aureus [B03.510.400.790.750.100]

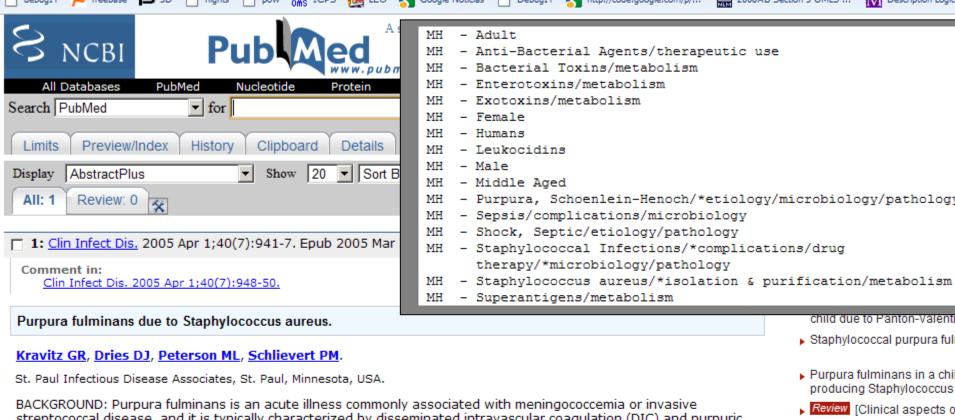
Staphylococcus epidermidis [B03.510.400.790.750.343]

Staphylococcus haemolyticus [B03.510.400.790.750.400]

Staphylococcus hominis [B03.510.400.790.750.425]

Return to Entry Page

Link to NLM Cataloging Classification



streptococcal disease, and it is typically characterized by disseminated intravascular coagulation (DIC) and purpuric skin lesions. In this article, we report the first 5 cases (to our knowledge) of purpura fulminans directly associated with Staphylococcus aureus strains that produce high levels of the superantigens toxic shock syndrome toxin-1 (TSST-1), staphylococcal enterotoxin serotype B (SEB), or staphylococcal enterotoxin serotype C (SEC). METHODS: Cases were identified in the Minneapolis-St. Paul, Minnesota, metropolitan area during 2000-2004. S. aureus infection was diagnosed on the basis of culture results, and susceptibility to methicillin was determined. The ability of the isolated organisms to produce TSST-1, SEB, SEC, and Panton-Valentine leukocidin (PVL) was determined. TSST-1, SEB, and SEC levels were also quantified after in vitro growth of the organisms. RESULTS: In 3 of the 5 cases, the infecting S. aureus strain was isolated from the blood cultures. In 2 of the 5 cases, the infecting S. aureus strain was isolated only from the respiratory tract, indicating that purpura fulminans and toxic shock syndrome resulted from exotoxin and/or other host factors, rather than septicemia. One of these latter 2 patients also had necrotizing pneumonia, and the isolated S. aureus was a methicillin-resistant strain that produced both SEC and PVL. Only 2 of the 5 patients survived, and 1 of the survivors received activated protein C. CONCLUSIONS: Staphylococcal purpura fulminans may be a newly emerging illness associated with superantigen production. Medical practitioners should be aware of this illness.

PMID: 15824983 [PubMed - indexed for MEDLINE]

- Review Menstrual toxic she
- case report and review.

Cited by 9 PubMed Central a

- Review Pathogenesis of m
- Superantigen profile of Sta steroid-resistant atopic der
- Alpha and beta chains of h aureus exotoxins.

Recent Activity

















































































Why ontologies matter for medicine

- Create taxonomies of natural kinds
 - classify instances in the world
 - basic for health statistics
 - exemplified in disease & procedure classification systems

- Creating common vocabularies / terminologies
 - normalization of word meanings
 - annotation of research data
 - facilitate document and fact retrieval

Why ontologies matter for medicine

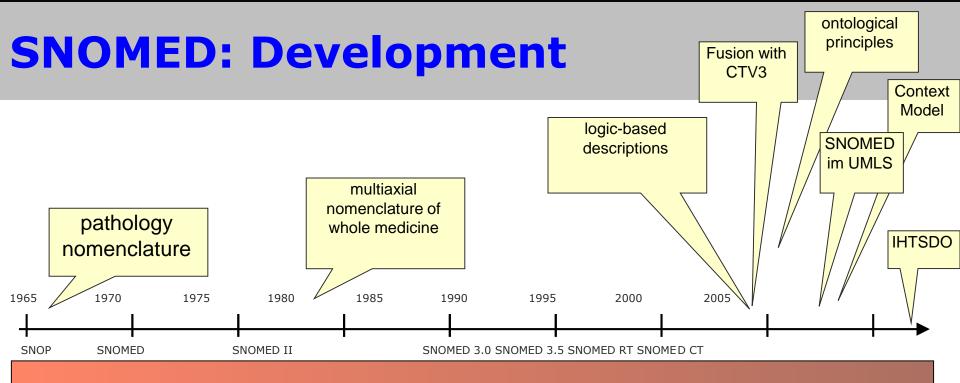
- Create taxonomies of natural kinds
 - classify instances in the world
 - basic for health statistics
 - exemplified in disease & procedure classification systems

- Creating common terminologies
 - normalization of word meanings
 - annotation of research data
 - facilitate document and fact retrieval

linguistics

set theory extensionality real things

intensionality concepts



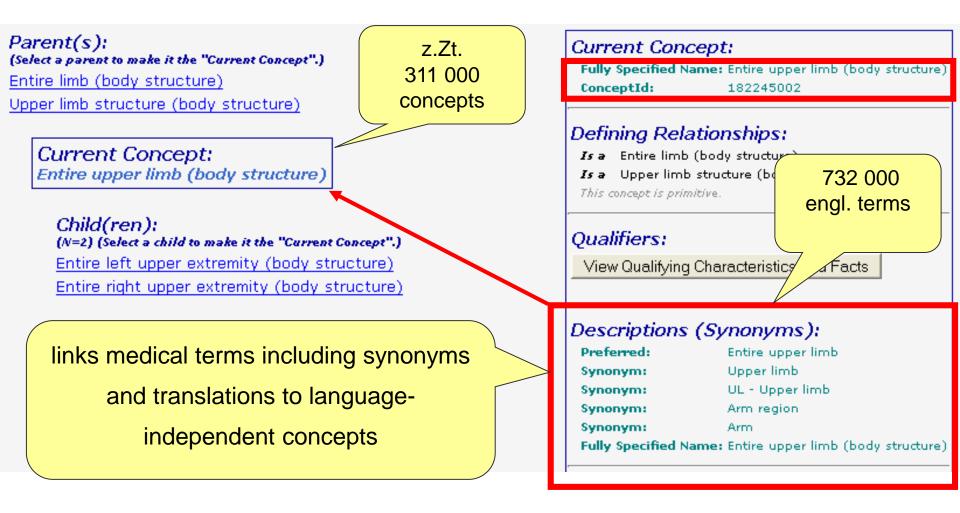




SNOMED CT

- "Standardized Nomenclature of Medicine Clinical Terms"
- Comprehensive clinical terminology
 (> 300,000 representational units)
- Devised to represent the meaning of clinical terms for whole range of health and clinical care
- Increasingly guided by ontological design principles
- Using a formal language: (Basic) Description Logics EL:
 - equivalence (≡) , subsumption (⊑)
 - existential role restriction (∃), conjunction (□)

SNOMED CT as a controlled vocabulary



SNOMED CT as a formal system

Parent(s): (Select a parent to make it the "Current Concept".) Disorder of appendix (disorder)

Inflammation of large intestine (disorder)

Current Concept: Appendicitis (disorder)

Child(ren):

(N=14) (Select a child to make it the "Current Concept".)

There are 5 Retired Children. Show Retired Children

Acute appendicitis (disorder)

Amebic appendicitis (disorder)

Appendicitis of a pelvic appendix (disorder)

Atypical appendicitis (disorder)

Catarrhal appendicitis (disorder)

Chronic appendicitis (disorder)

Complicated appendicitis (disorder)

Focal appendicitis (disorder)

hierarchies: strict

specialization (is-a)

urrent Concept:

Fully Specified Name: Appendicitis (disorder)

ConceptId: 74400008

efining Relationships:

Is a Disorder of appendix (disorder)

Is a Inflammation of large intestine (disorder)

Group 1

Associated morphology (attribute)
Finding site (attribute)

Inflammation (morphologic abnormality)
Appendix structure (body structure)

This concept is fully defined.

Qualifiers:

View Qualifying Characteristics and Facts

Descriptions (Synonyms):

Preferred: Appendicitis

Fully Specified Name: Appendicitis (disorder)
Synonym: Appendicitis, NOS

SNOMED CT as a formal system

Parent(s): (Select a parent to make it the "Current Concept".) Disorder of appendix (disorder) Inflammation of large intestine (disorder) Current Concept: Appendicitis (disorder)

Child(ren):
(N=14) (Select a child to make it the "Current Concept".)
There are 5 Retired Children. Show Retired Children

Acute appendicitis (disorder)
Amebic appendicitis (disorder)

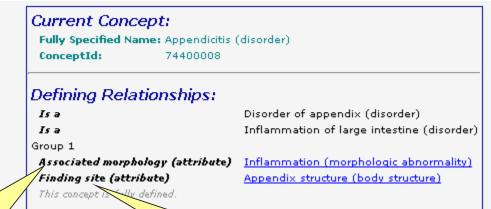
Appendicitis of a pelvic appendix (disorder

Atvnical annendicitis (disorder)

restrictions based on simple description logics:

C1 - Rel - C2 interpreted as:

 $\forall x: instanceOf(x, C1) \Rightarrow \exists y: instanceOf(C2) \land Rel(x,y)$



Qualifiers:

View Qualifying Chy

Descriptions (.
Preferred:
Fully Specified Nam
Synonym:

Relations (Attributes): z.B.
Associated morphology
Finding site
(50 relation types)

SNOMED CT als formales System

Current Concept:

Fully Specified Name: Entire upper limb (body structure)
ConceptId: 182245002

Defining Relationships:

Is a Entire limb (body structure)

Is a Upper limb structure (body structure)

This concept is primitive.

defined vs. primitive concepts

Current Concept:

Fully Specified Name: Appendicitis (disorder)

ConceptId: 74400008

Defining Relationships:

Is a Disorder of appendix (disorder)

Is a Inflammation of large intestine (disorder)

Group 1

Associated morphology (attribute)
Finding site (attribute)

Inflammation (morphologic abnormality)
Appendix structure (body structure)

This concept is fully defined.

Oualifiers:

View Qualifying Characteristics and Facts

Descriptions (Synonyms):

Preferred: Appendicitis

Deficit of previous non-formal SNOMED versions

D5-46210

Acute appendicitis, NOS

D5-46100

Appendicitis, NOS

G-A231

Acute

M-41000

Acute inflammation, NOS

G-C006

In

T-59200

Appendix, NOS

- Unterschiedliche
 Beschreibungen desselben
 Sachverhalts sind nicht
 aufeinander abbildbar
- Aneinanderreihung von Konzepten und Relationen nicht eindeutig interpretierbar

G-A231 <u>Acute</u>

M-40000 Inflammation

G-C006 <u>In</u>

Introduction

T-59200 Appendix, NOS

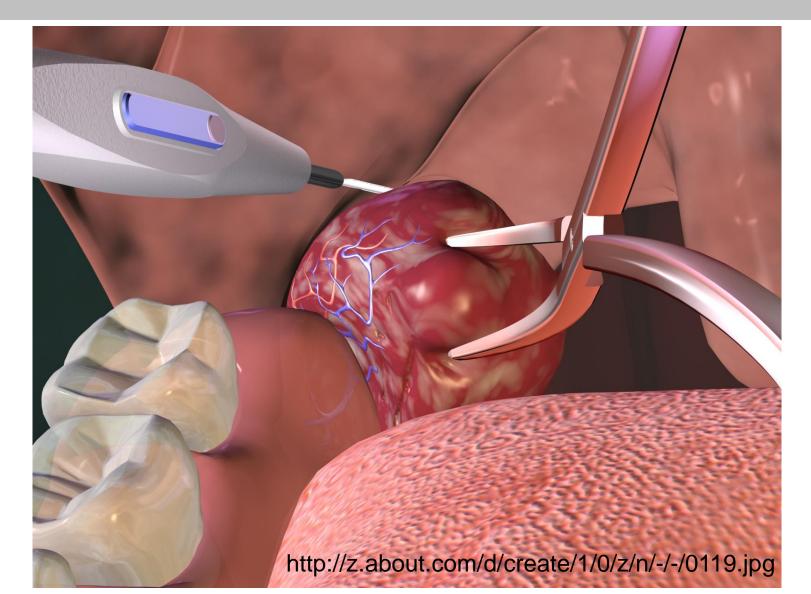
SNOMED INTERNATIONAL

Ontological commitment

- "Agreement about the ontological nature of the entities being referred to by the representational units in an ontology" (modified definition following Gruber 93)
- Formal ontologies: subsumption and equivalence statements are either true or false
- Problem: change of truth-value of axioms and sentences according to resulting competing interpretations

Conclusions Introduction Examples Discussion

Tonsillectomy



1. Tonsillectomy planned ≡

```
∃ rg.( ∃ associatedProcedure.Tonsillectomy ⊓
```

```
∃ procedureContext.Planned ⊓
```

- ∃ subjectRelationshipContext.SubjectOfRecord ⊓
- ∃ temporalContext.CurrentOrSpecifiedTime)

1. Tonsillectomy planned ≡

```
∃ rg.( ∃ associatedProcedure.Tonsillectomy ⊓
```

```
∃ procedureContext.Planned ⊓
```

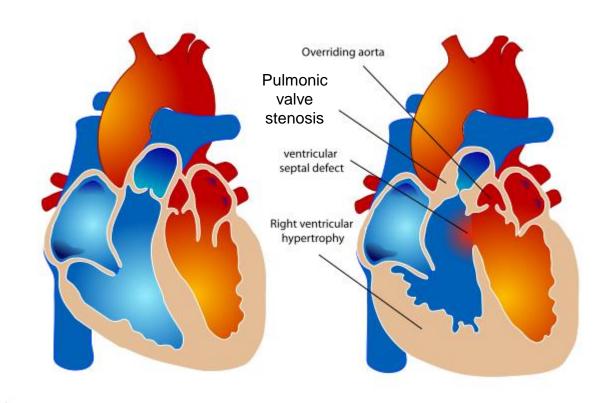
- ∃ subjectRelationshipContext.SubjectOfRecord ⊓
- ∃ temporalContext.CurrentOrSpecifiedTime)

1. Tonsillectomy planned ≡

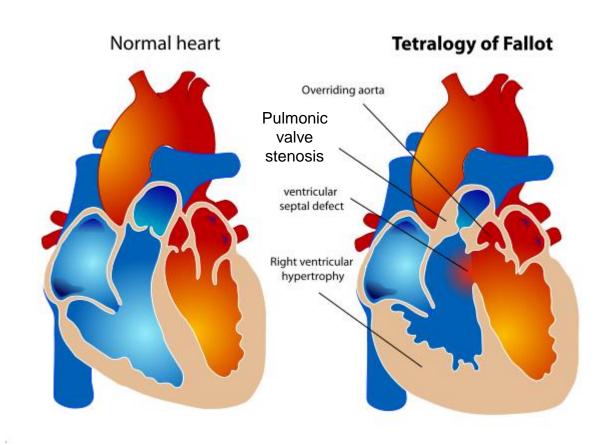
```
∃ rg.(∃ associatedProcedure.Tonsillectomy ☐
∃ procedureContext.Planned ☐
∃ subjectRelationshipContext.SubjectOfRecord ☐
∃ temporalContext.CurrentOrSpecifiedTime)
```

2. Denied tonsillectomy ≡

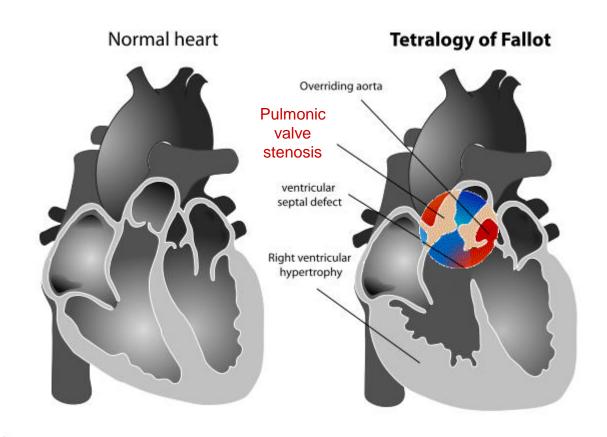
Tonsillectomy ⊓ ∃ Priority. Denied



Tetralogy of Fallot



Tetralogy of Fallot



SNOMED CT Examples

1. Tonsillectomy planned ≡

```
∃ rg.(∃ associatedProcedure.Tonsillectomy ⊓
∃ procedureContext.Planned ⊓
∃ subjectRelationshipContext. SubjectOfRecord ⊓
∃ temporalContext.CurrentOrSpecifiedTime)
```

2. Denied tonsillectomy ≡

3. Tetralogy of Fallot ≡

"every instance of "Tonsillectomy planned" implies some tonsillectomy"

- 1. Tonsillectomy planned =
 - ☐ rg.(☐ associatedProcedure.Tonsillectomy □
 - ∃ procedureContext.Planned ⊓
 - ∃ subjectRelationshipContext. SubjectOfRecord ⊓
 - ∃ temporalContext.CurrentOrSpeci

"every denied tonsillectomy is a tonsillectomy"

2. Denied tonsillectomy(≡)

Tonsillectomy ⊓ ∃ Priority.Denied

3. Tetralogy of Fallot (≡)

"every Fallot is also a Pulmonic Valve Stenosis"

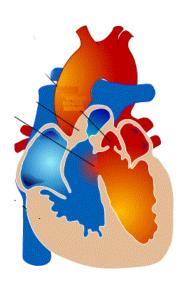
PulmonicValveStenosis π VentricularSeptaiDefect π

OverridingAorta

RightVentricular hypertrophy

Problems



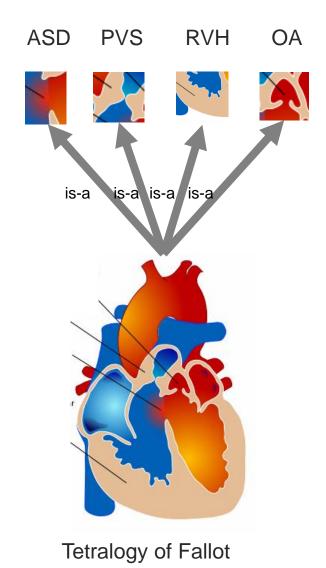


- The negation of a process is a specialization of this process
- A plan is defined such as its realization is implied
- A (definitional) proper part of a compound entity is its taxonomic parent

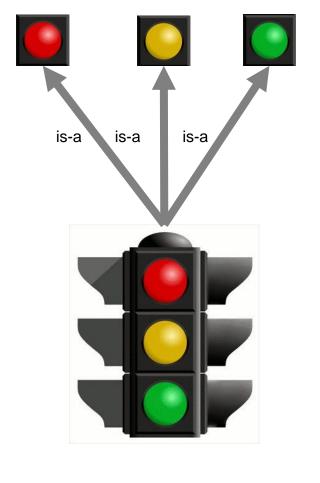
Proper parts of taxonomic parents?

Example from Harold Solbrig

Conclusions



Red Light Yellow Light Green Light



Traffic Light

Relevance

- The three examples are not accidental errors they represent systematic architectural patterns of SNOMED CT
 - for 50,000 procedure concepts, "denied" subconcepts can be created
 - hundreds of concepts have properties like "planned",
 "suspected" or "known absent" in their definition
 - 77,000 "procedure" or "finding" concepts have their constituent parts as parent concepts (side effect of role group constructor)
- Hypothesis: they represent different and competing ontological commitments strongly influenced by the practice of clinical coding and documentation

Alternative interpretations?

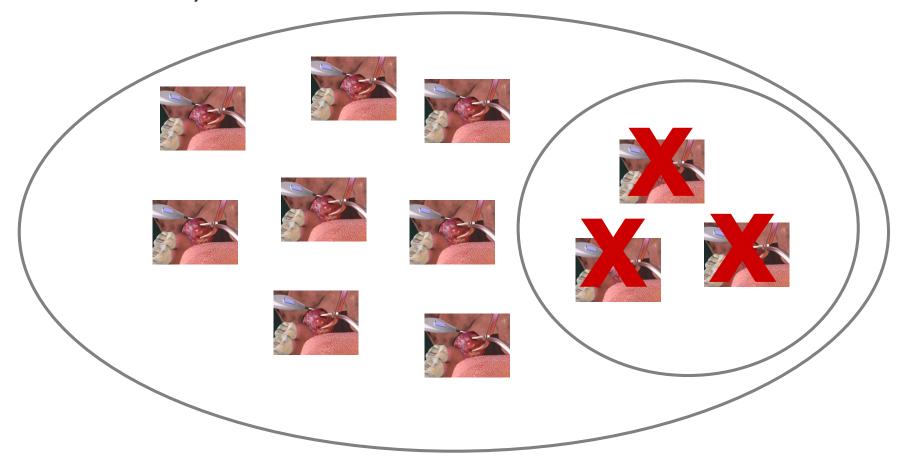
Information Artifact

T	≯H ∜			OPERATING	G RO	OM PL	ANNE	R		Aitiid
	Sched		Pat.Rm	Procedure & Remarks	Surgeon	Attending	Anesthetist	An.Type	Nurse	mer Staff
4		#388827		Bil. Tonsillectomy	AB	ОВ	AR	Int	CM	
4		#445321		Adenoidectomy	AB	OB	AR	Int	CN	
4	9:00	#200334	1023	Dil. Tonsillectomy	OD	AD	AR	Int	CN	suspended
4	9:45	#889881	1001	Mastoidectomy	AB	ОВ	AR	Int	CN	
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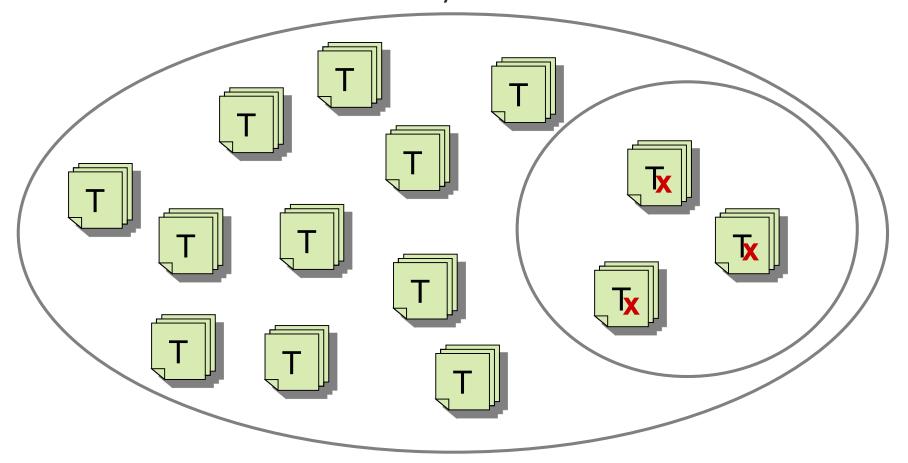
SNOMED CT concepts are instantiated by representational artifacts as contained in an electronic patient record

- A documentation artifact of a certain kind is created for each patient scheduled for an operation
- The class of these information artifacts includes subclasses of information artifacts that include values such as "planned", "executed", "denied" etc.
- An expression such as ∃ associatedProcedure.Tonsillectomy can be seen representing a plan (but ∃ is false anyway)
- → ∃ Priority. Denied refines the class of information artifacts but not the class of tonsillectomies

Extension of "Tonsillectomy" includes extension of "Denied Tonsillectomy": **FALSE**



Extension of "Record of Tonsillectomy" includes extension of "Record of Denied Tonsillectomy": **TRUE**



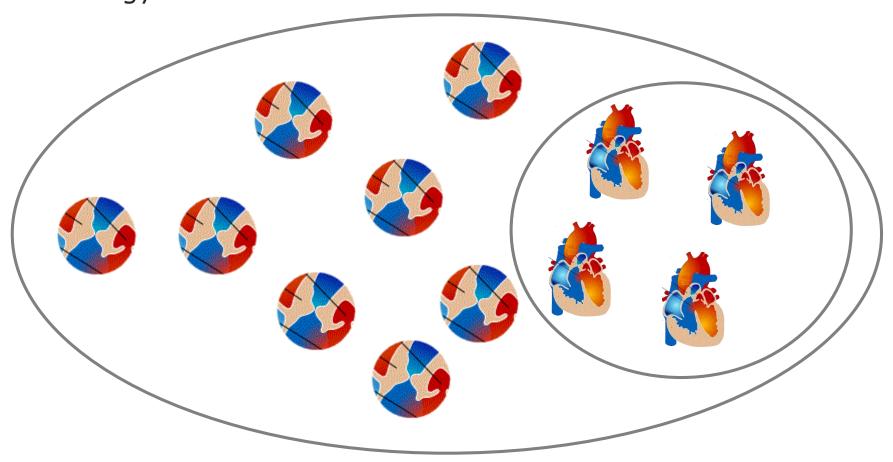
SNOMED CT concepts are instantiated by patients or clinical situations.

- Pulmonic Valve Stenosis stands for "Patient with a pulmonic valve stenosis"
- Tetralogy of Fallot stands for "Fallot Patient"
- All Fallot patients are also patients with pulmonic valve stenosis because every instance of Tetralogy of Fallot has one instance of pulmonic valve stenosis as part

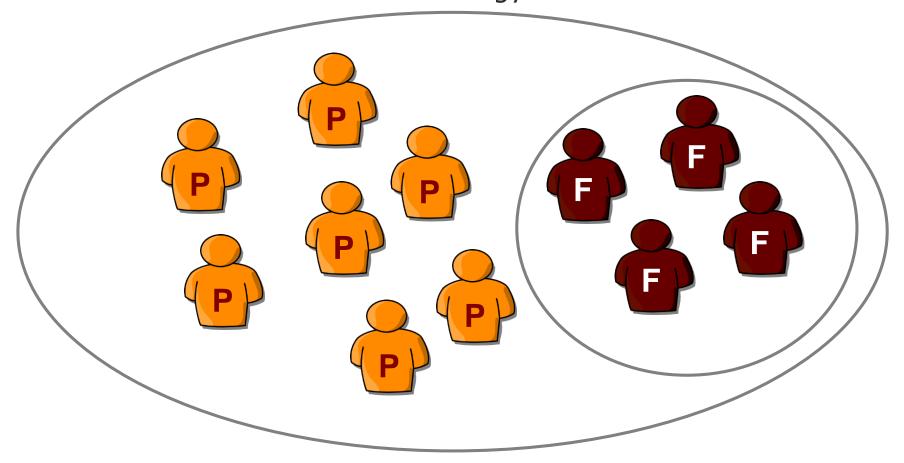
Consequence:

 Finding and procedure concepts extend to classes of patients but not to classes of findings or procedures

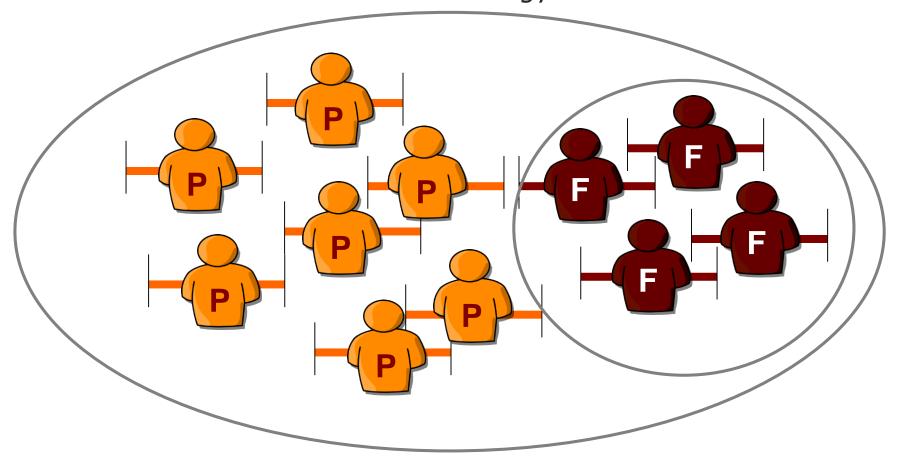
Extension of "Pulmonic Valve Stenosis" includes extension of "Tetralogy of Fallot": **FALSE**



Extension of "Patient with Pulmonic Valve Stenosis" includes extension of "Patient with Tetralogy of Fallot": TRUE



Extension of "Situation with Pulmonic Valve Stenosis" includes extension of "Situation with Tetralogy of Fallot": TRUE



Conclusions

- SNOMED CT's ontological commitment is heterogeneous
- SNOMED CT's alternative interpretations are implicit, thus leaving burden of interpretation to the user.
- The alternative interpretations reflect clinicians' reasoning patterns
- SNOMED mixes elements of an ontology with elements of information models (information artifacts)
- Use of SNOMED CT as an ontology depends on agreement about its ontological commitment