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on Biomedical Ontology**

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SNOMED CT's Ontological Commitment

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Basic questions for ontology developers

- What are the instances ?
- What are the entities they are dependent on (without what they can't exist) ?

Alan Ruttenberg

(Tutorial on the Information Artifact Ontology)

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
- Case study: SNOMED CT

Introduction

Examples

Discussion

Conclusions

SNOMED CT

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 \mathcal{EL} :
 - equivalence (\equiv), subsumption (\sqsubseteq)
 - existential role restriction (\exists), conjunction (\sqcap)

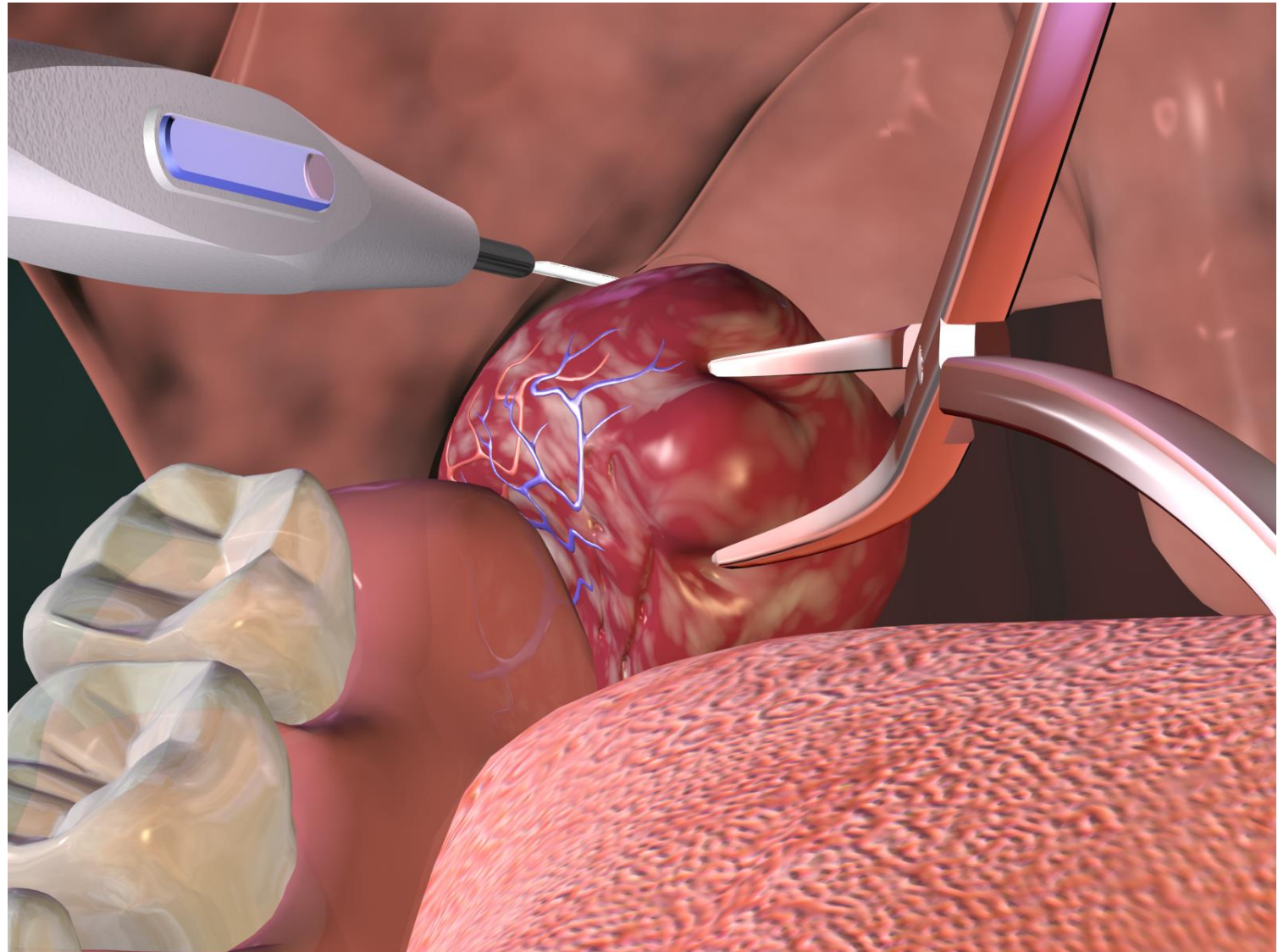
Introduction

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Tonsillectomy



SNOMED CT Examples

1. Tonsillectomy planned \equiv

$\exists rg. (\exists associatedProcedure. Tonsillectomy \sqcap$
 $\exists procedureContext. Planned \sqcap$
 $\exists subjectRelationshipContext. SubjectOfRecord \sqcap$
 $\exists temporalContext. CurrentOrSpecifiedTime)$

SNOMED CT Examples

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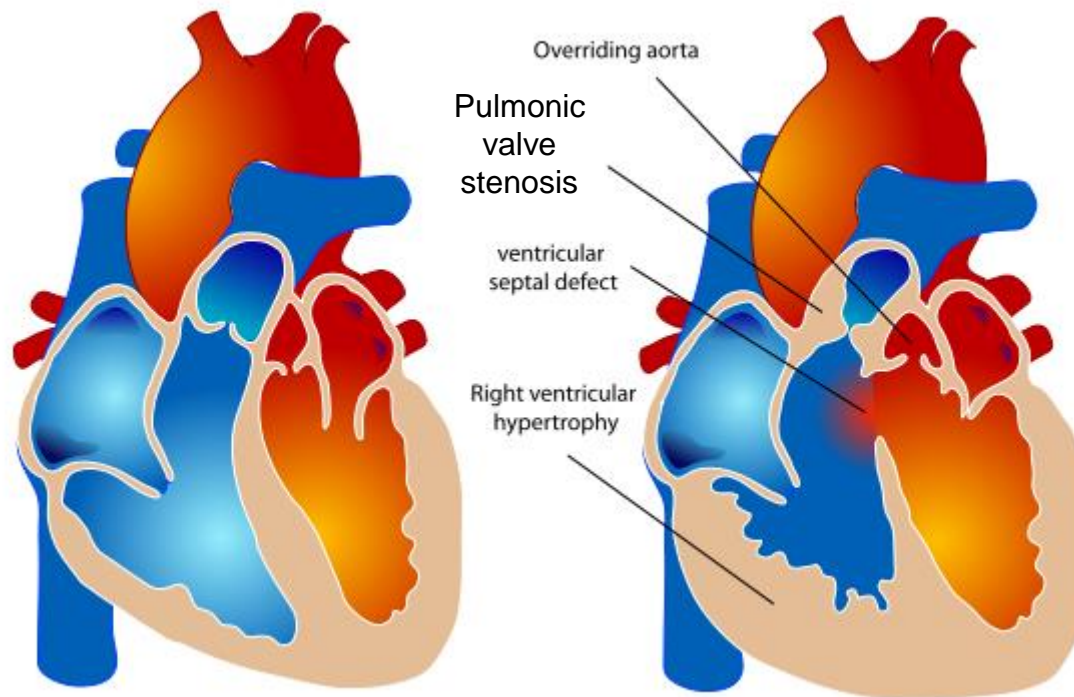
SNOMED CT Examples

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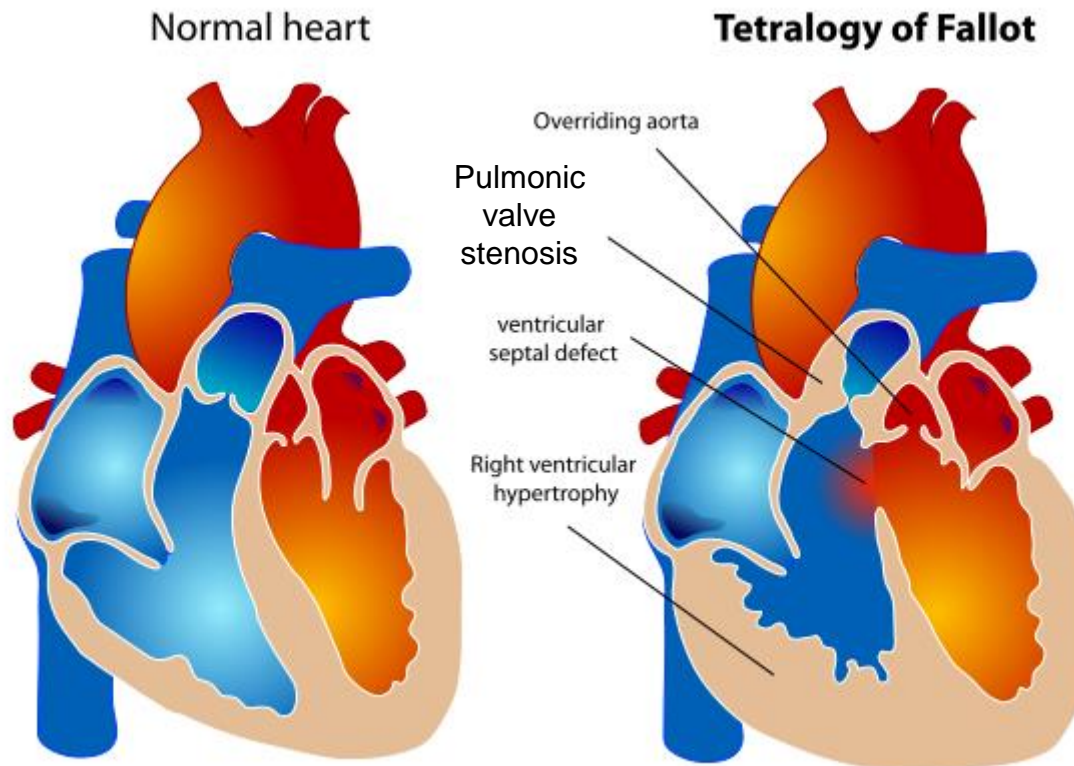
$\exists rg. (\exists associatedProcedure. Tonsillectomy \sqcap$
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2. Denied tonsillectomy \equiv

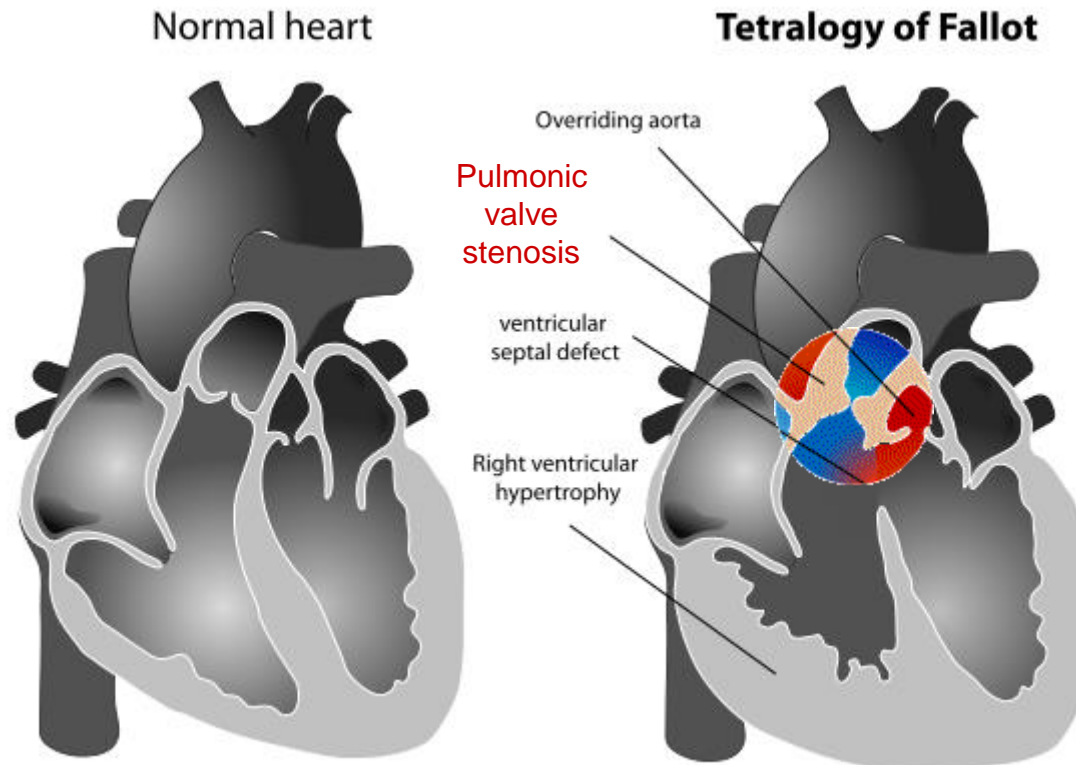
$Tonsillectomy \sqcap \exists Priority. Denied$



Tetralogy of Fallot



Tetralogy of Fallot



obstruction to flow from the right ventricle (RV) to the pulmonary artery ..

SNOMED CT Examples

1. Tonsillectomy planned \equiv

$\exists rg.(\exists associatedProcedure.Tonsillectomy \sqcap$
 $\exists procedureContext.Planned \sqcap$
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2. Denied tonsillectomy \equiv

$Tonsillectomy \sqcap \exists Priority.Denied$

3. Tetralogy of Fallot \equiv

$PulmonicValveStenosis \sqcap VentricularSeptalDefect \sqcap$
 $OverridingAorta \sqcap RightVentricular hypertrophy$

SNOMED CT Examples

1. Tonsillectomy planned \equiv

\exists arg. (\exists associatedProcedure.Tonsillectomy \sqcap
 \exists procedureContext.Planned \sqcap
 \exists subjectRelationshipContext. SubjectOfRecord \sqcap
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“every instance of
“Tonsillectomy
planned” implies some
tonsillectomy”

2. Denied tonsillectomy \equiv

Tonsillectomy \sqcap \exists Priority.Denied

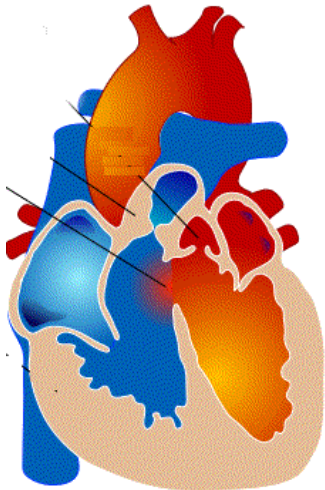
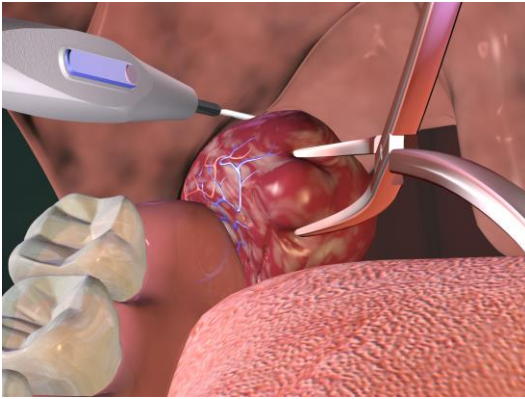
“every denied
tonsillectomy is a
tonsillectomy”

3. Tetralogy of Fallot \equiv

PulmonicValveStenosis \sqcap VentricularSeptalDefect \sqcap
 OverridingAorta \sqcap RightVentricular hypertrophy

“every Fallot is also
a Pulmonic Valve
Stenosis”

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

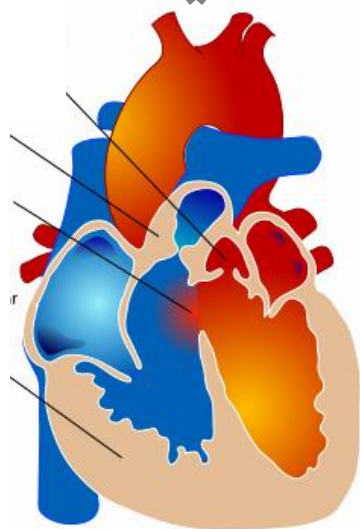
Proper parts of taxonomic parents ?

Example from Harold Solbrig

ASD PVS RVH OA



is-a is-a is-a is-a

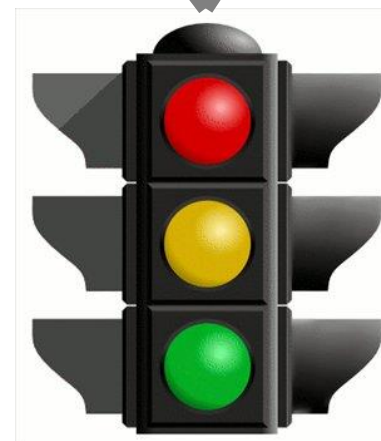


Tetralogy of Fallot

Red Light Yellow Light Green Light



is-a is-a is-a



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 ?

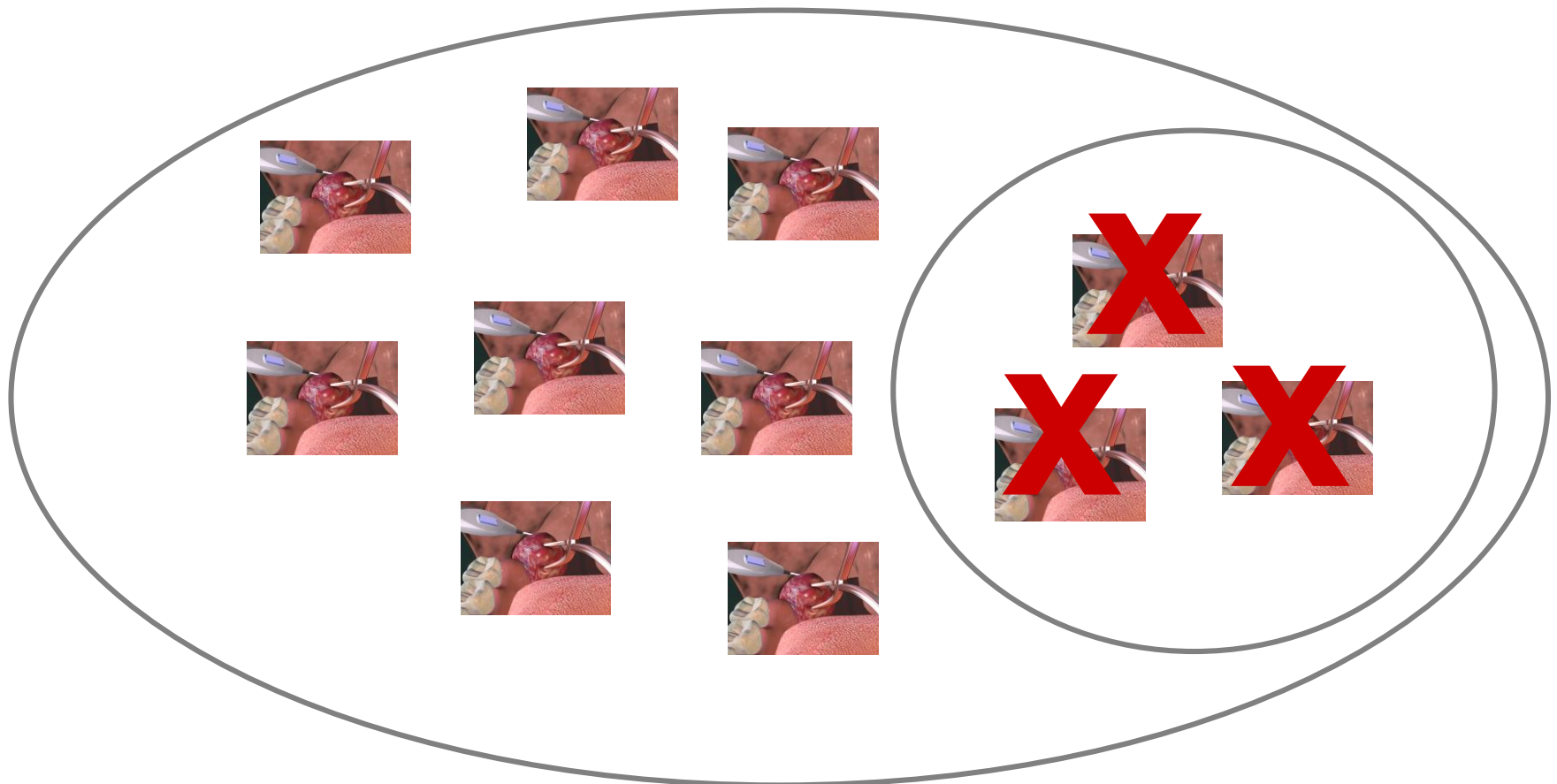
Alternative interpretation (I)

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 \exists *associatedProcedure.Tonsillectomy* can be seen representing a plan (but \exists is false anyway)
- \exists *Priority.Denied* refines the class of information artifacts but not the class of tonsillectomies

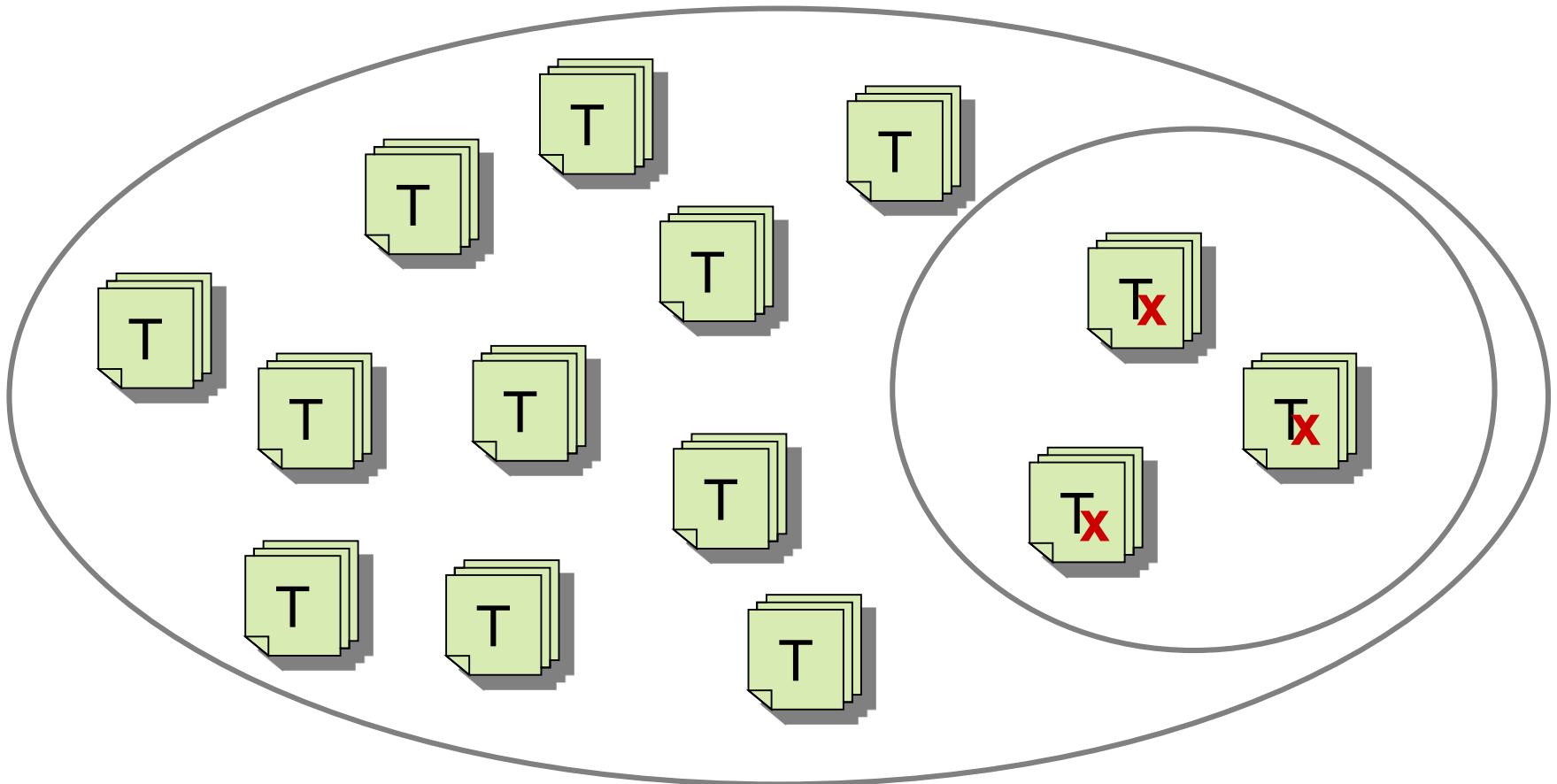
Alternative interpretation (I)

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



Alternative interpretation (I)

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



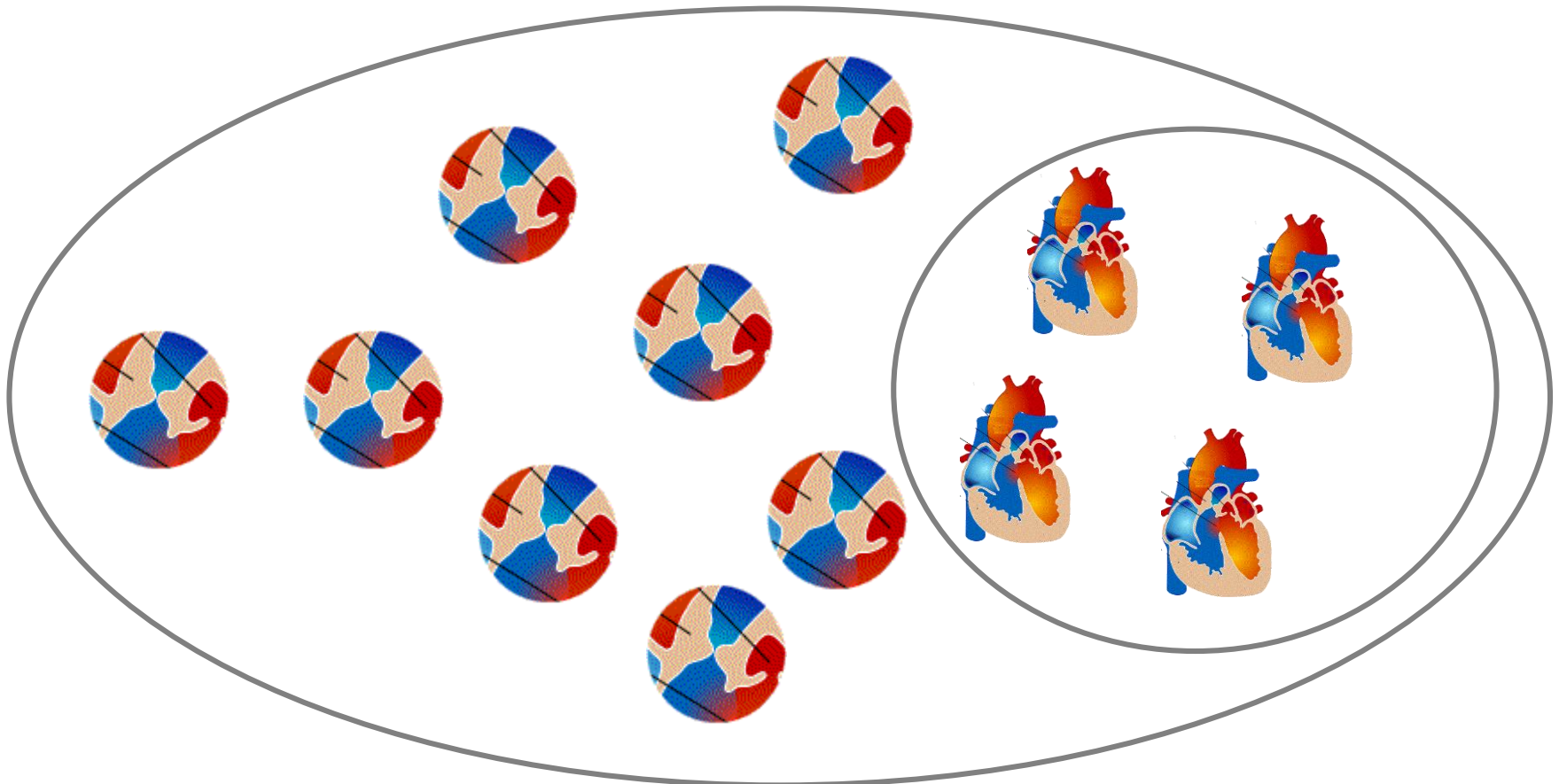
Alternative interpretation (II)

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 (pathologic structure) 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

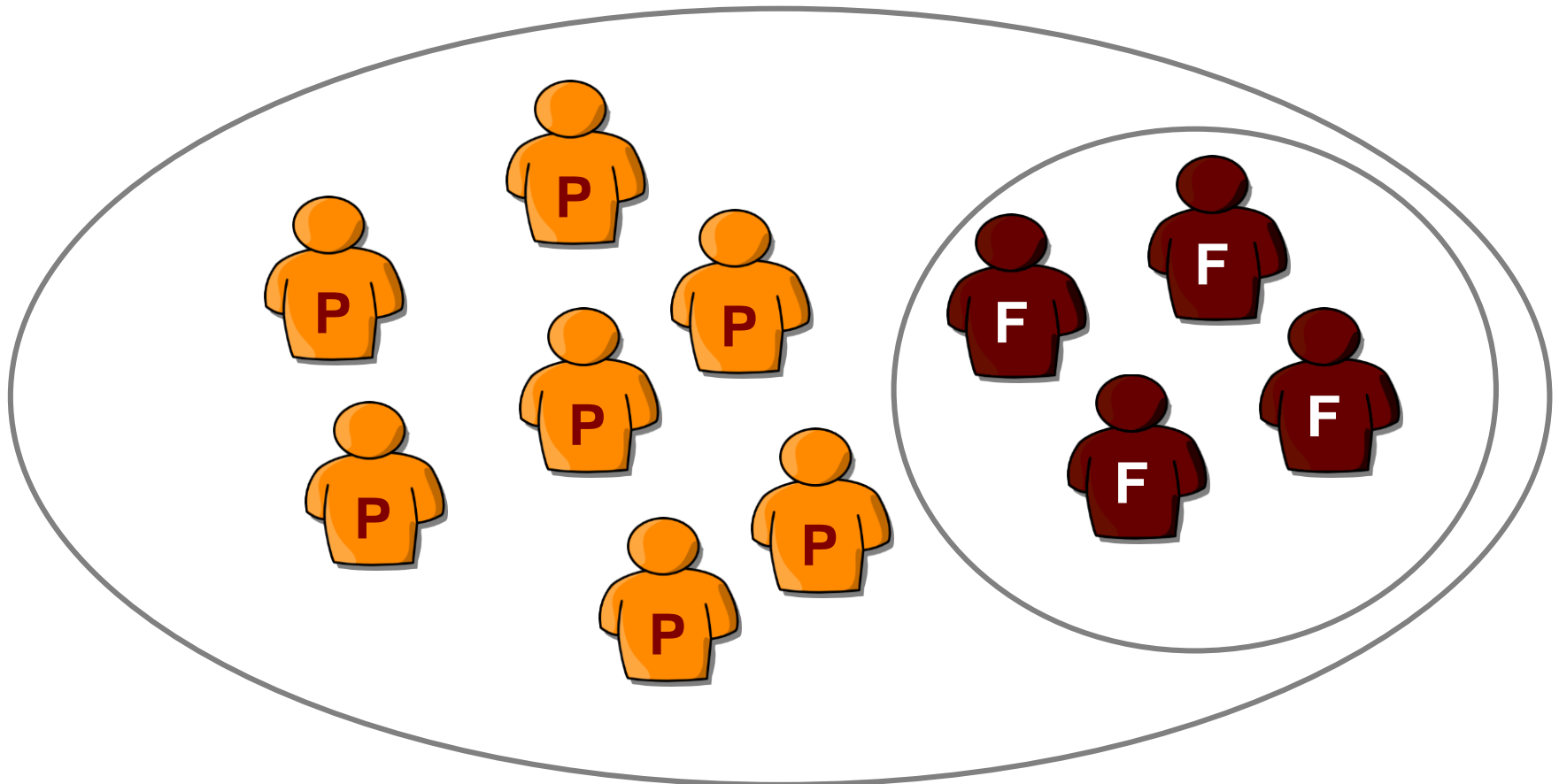
Alternative interpretation (II)

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



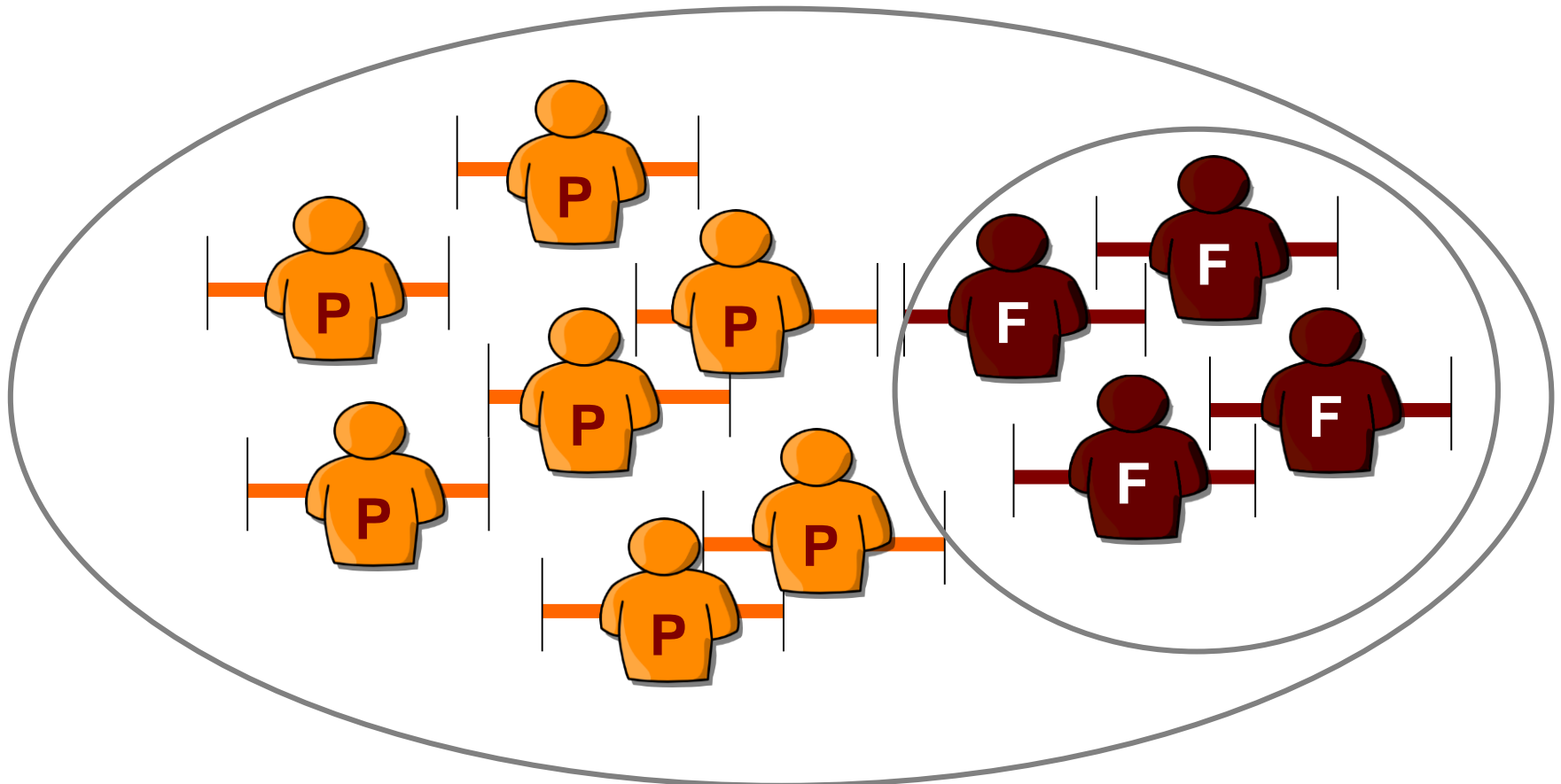
Alternative interpretation (II)

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



Alternative interpretation (II)

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



Conclusions

- Many hierarchies and definitions SNOMED CT suggest that SNOMED CT's ontological commitment is heterogeneous
- SNOMED CT's alternative commitments are completely implicit, thus leaving burden of interpretation to the user.
- But the alternative interpretations shed light on clinicians' reasoning
- 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

Conclusions

- What are the instances of SNOMED CT concepts?
- What are the entities they are dependent on (without what they can't exist) ?