

# **Ontological Analysis, Ontological Commitment, and Epistemic Contexts**

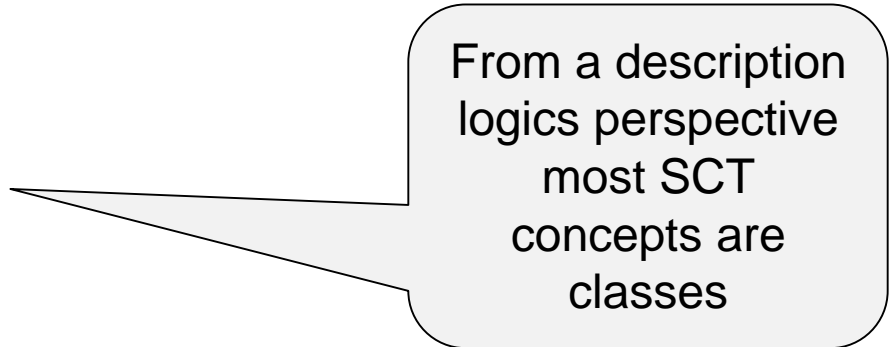
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WHO – IHTSDO Joint Advisory Group  
First Face-to-Face Meeting  
Heathrow, London  
15-16 December 2010

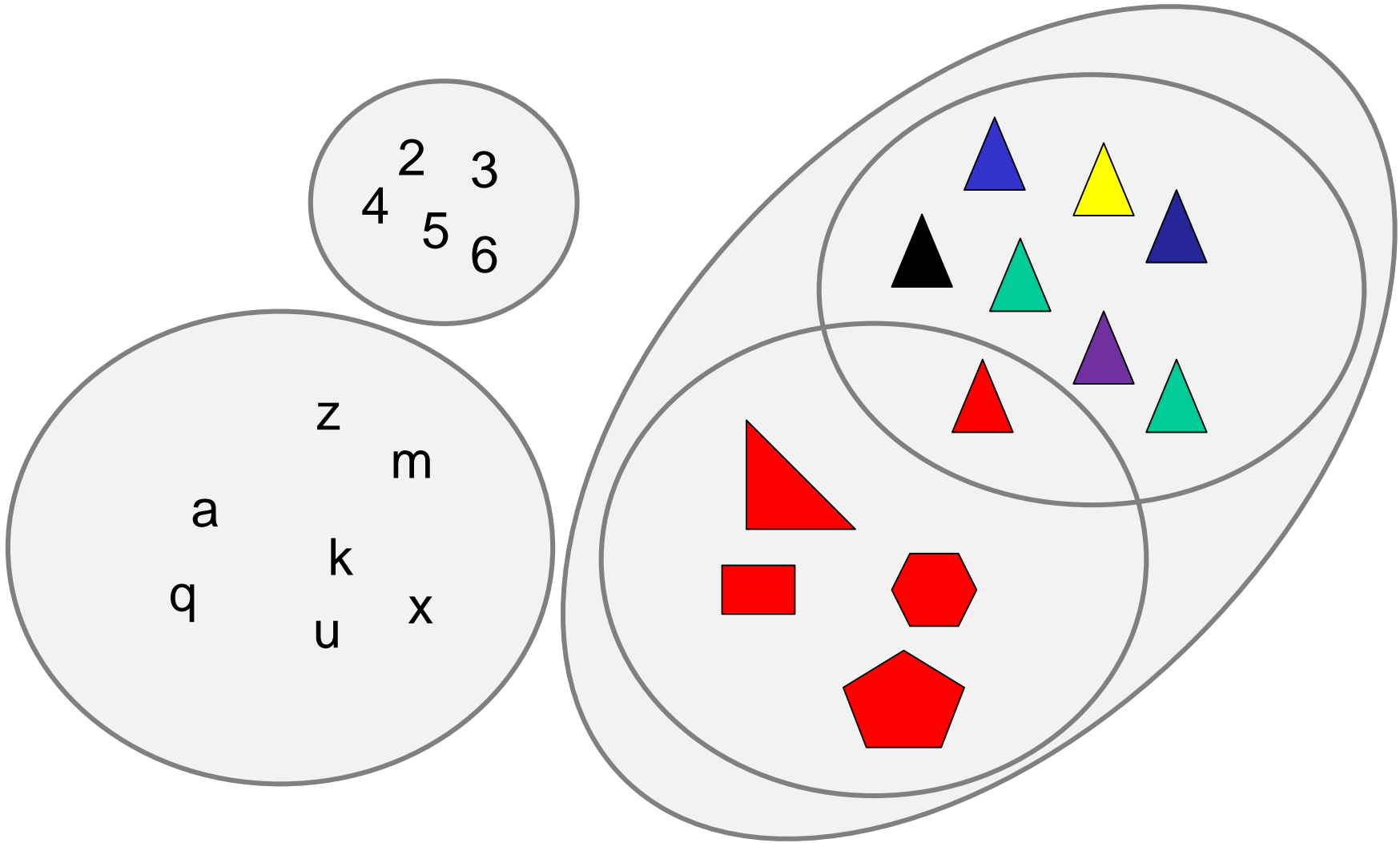
# Ontological analysis

- What do the representational units in a representational artifact represent?
  - members of classes
  - instances of concepts
  - denotation of terms
- What are the entities they are dependent on (without what they can't exist) ?
- In which upper level categories do they belong ?



From a description logics perspective most SCT concepts are classes

# Classes and their extensions



# Example

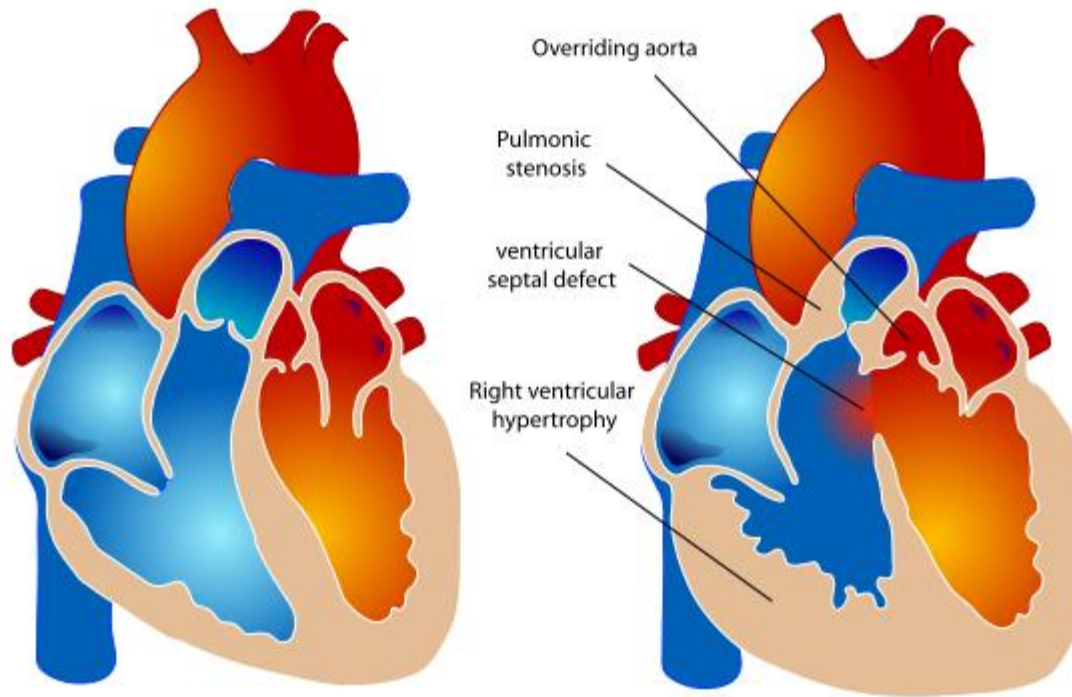
- Representational unit (class, concept, term): Melanoma
- Member, instance: e.g. basal cell carcinoma at left cheek of patient #12334
- Dependencies: every basal cell carcinoma is located in some skin
- Upper level Categories:
  - Material entity?
  - Process?
  - both?
  - what is the ontological commitment of "basal cell carcinoma"?



# 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
- Example: Tetralogy of Fallot in SNOMED CT and ICD10

# Tetralogy of Fallot



<b>Q21</b>	<b>Congenital malformations of cardiac septa</b> <i>Excludes:</i> acquired cardiac septal defect ( <a href="#">I51.0</a> )
<b>Q21.0</b>	<b>Ventricular septal defect</b>
<b>Q21.1</b>	<b>Atrial septal defect</b> Coronary sinus defect Patent or persistent: · foramen ovale · ostium secundum defect (type II) Sinus venosus defect
<b>Q21.2</b>	<b>Atrioventricular septal defect</b> Common atrioventricular canal Endocardial cushion defect Ostium primum atrial septal defect (type I)
<b>Q21.3</b>	<b>Tetralogy of Fallot</b> Ventricular septal defect with pulmonary stenosis or atresia, dextroposition of aorta and hypertrophy of right ventricle.
<b>Q21.4</b>	<b>Aortopulmonary septal defect</b> Aortic septal defect Aortopulmonary window
<b>Q21.8</b>	<b>Other congenital malformations of cardiac septa</b> Eisenmenger's defect Pentalogy of Fallot <i>Excludes:</i> Eisenmenger's · complex ( <a href="#">I27.8</a> ) · syndrome ( <a href="#">I27.8</a> )
<b>Q21.9</b>	<b>Congenital malformation of cardiac septum, unspecified</b> Septal (heart) defect NOS

### Current Concept:

**Fully Specified Name:** [Tetralogy of Fallot \(disorder\)](#)

**ConceptId:** 86299006

### Defining Relationships:

**Is a** Congenital abnormality of ventricles and ventricular septum (disorder)

**Is a** Overriding aorta (disorder)

**Is a** Pulmonic valve stenosis (disorder)

**Is a** Right ventricular hypertrophy (disorder)

**Is a** [Ventricular septal defect \(disorder\)](#)

**Occurrence** [Congenital \(qualifier value\)](#)

Group 1

**Associated morphology** [Congenital anomaly \(morphologic abnormality\)](#)

**Finding site** [Cardiac ventricular structure \(body structure\)](#)

Group 2

**Associated morphology** [Defect \(morphologic abnormality\)](#)

**Finding site** [Interventricular septum structure \(body structure\)](#)

Group 3

**Associated morphology** [Stenosis \(morphologic abnormality\)](#)

**Finding site** [Pulmonary valve structure \(body structure\)](#)

Group 4

**Associated morphology** [Overriding structures \(morphologic abnormality\)](#)

**Finding site** [Thoracic aorta structure \(body structure\)](#)

Group 5

**Associated morphology** [Hypertrophy \(morphologic abnormality\)](#)

**Finding site** [Right ventricular structure \(body structure\)](#)

*This concept is primitive.*

### Current Concept:

**Fully Specified Name:** Ventricular septal defect (disorder)

**ConceptId:** 30288003

### Defining Relationships:

**Is a** Disorder of cardiac ventricle (disorder)

**Is a** Structural disorder of heart (disorder)

Group 1

**Associated morphology** [Defect \(morphologic abnormality\)](#)

**Finding site** [Interventricular septum structure \(body structure\)](#)

*This concept is fully defined.*

### Qualifiers:

[View Qualifying Characteristics and Facts](#)

### Descriptions (Synonyms):

**Fully Specified Name:** Ventricular septal defect (disorder)

**Preferred:** Ventricular septal defect

**Synonym:** Interventricular septal defect

**Synonym:** VSD - Ventricular septal defect

**Synonym:** Ventricular septal abnormality

**Synonym:** Roger's disease

**Synonym:** Absence of interventricular septum

### Related Concepts:

- [All "Is a" antecedents](#) -

- [All descendents and related subtypes](#) -

Every heart disorder that includes a defect of an interventricular septum structure is a ventricular septum defect.

Therefore tetralogy of Fallot is a kind of ventricular septum defect



# Tetralogy of Fallot definition

## **SNOMED CT:**

*TetralogyOfFallot* equivalentClass

*PulmonicValveStenosis* and *VentricularSeptalDefect* and  
*OverridingAorta* and *RightVentricularHypertrophy*

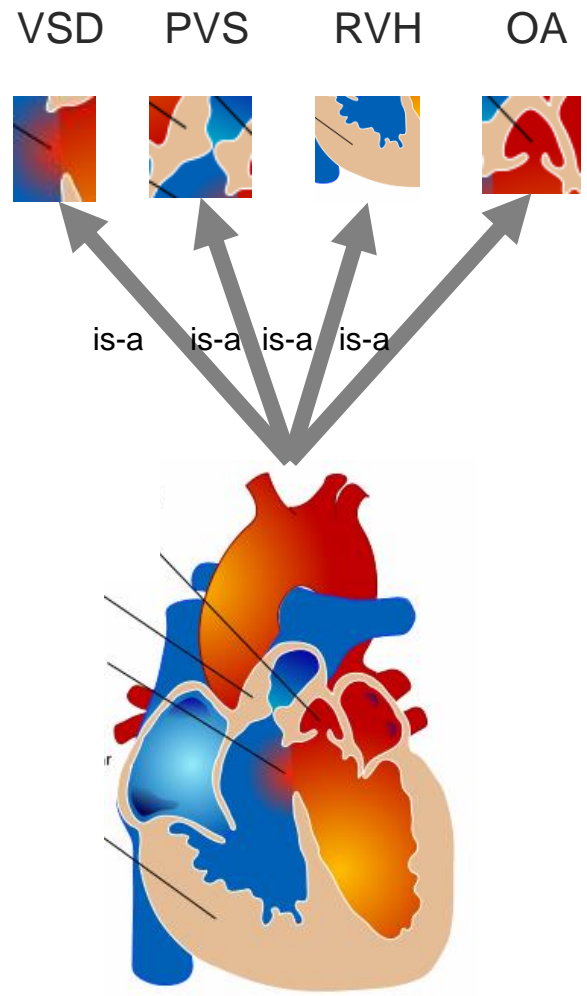
*TetralogyOfFallot* is a child concept of *VentricularSeptalDefect*

## **ICD10:**

Tetralogy of Fallot is a child of “congenital malformations of cardiac septa” and a sibling of “ventricular septal defect”

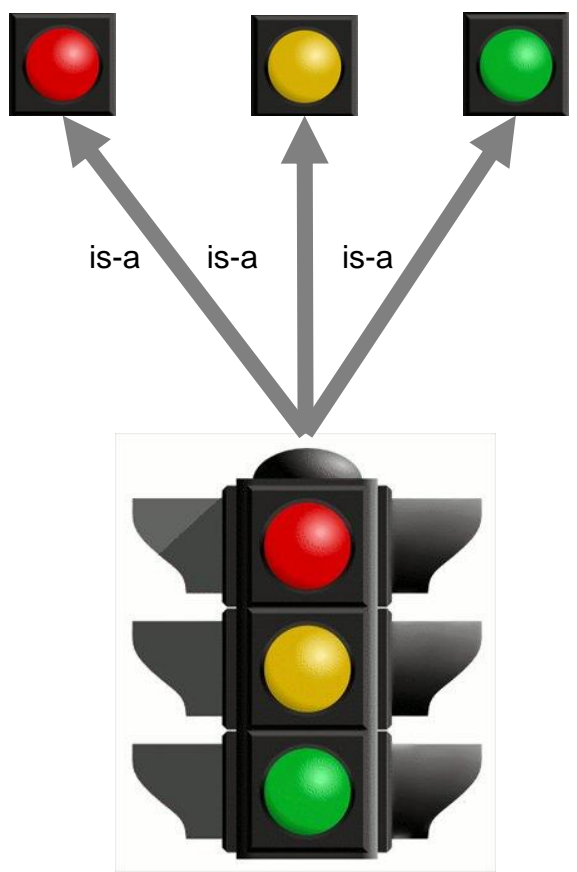
# Proper parts or taxonomic parents ?

Example from Harold Solbrig



Tetralogy of Fallot

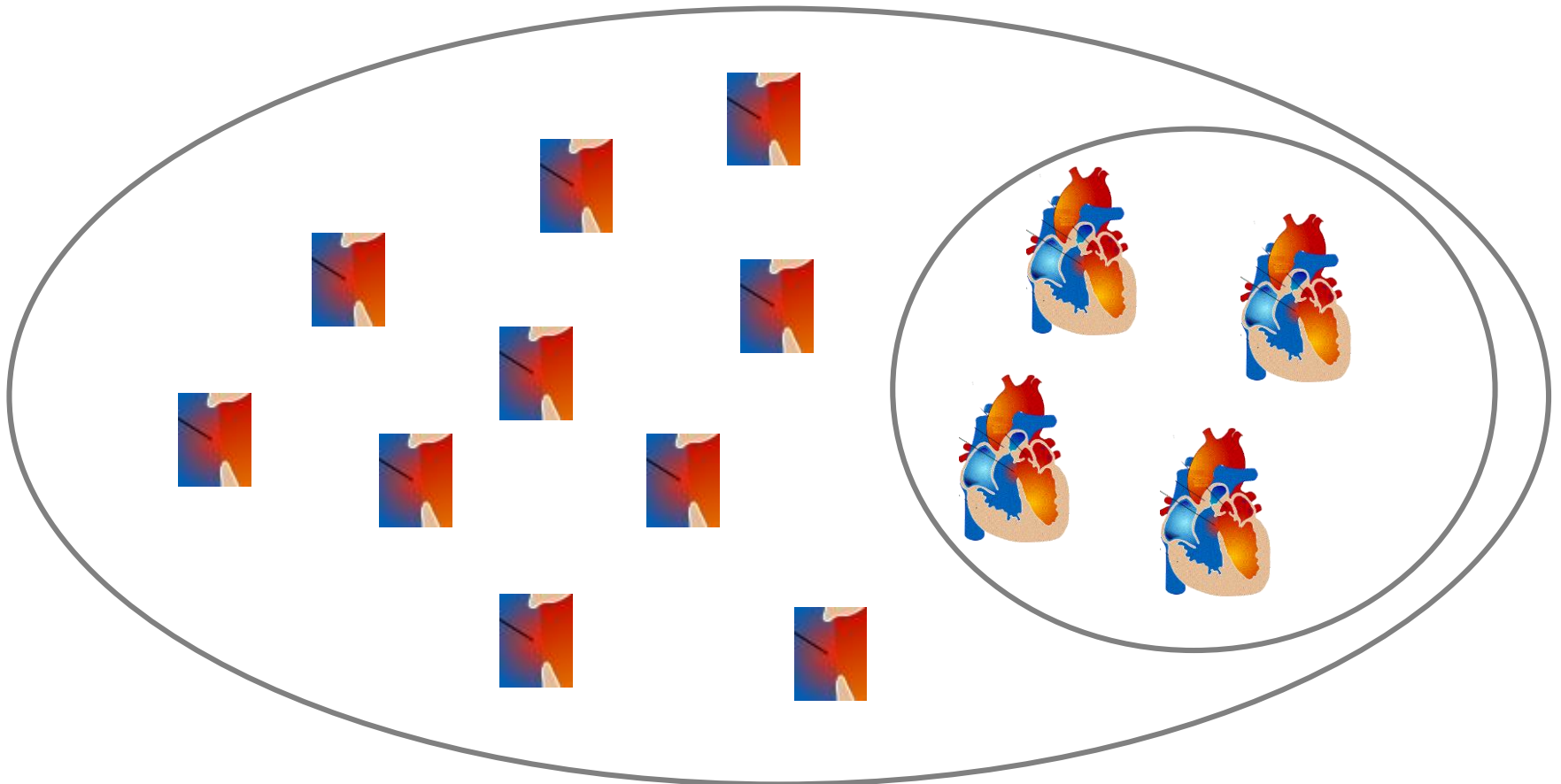
Red Light Yellow Light Green Light



Traffic Light

# Ontological Commitment 1

Extension of "*Ventricular Septal Defect*" includes extension of "*Tetralogy of Fallot*": **FALSE**



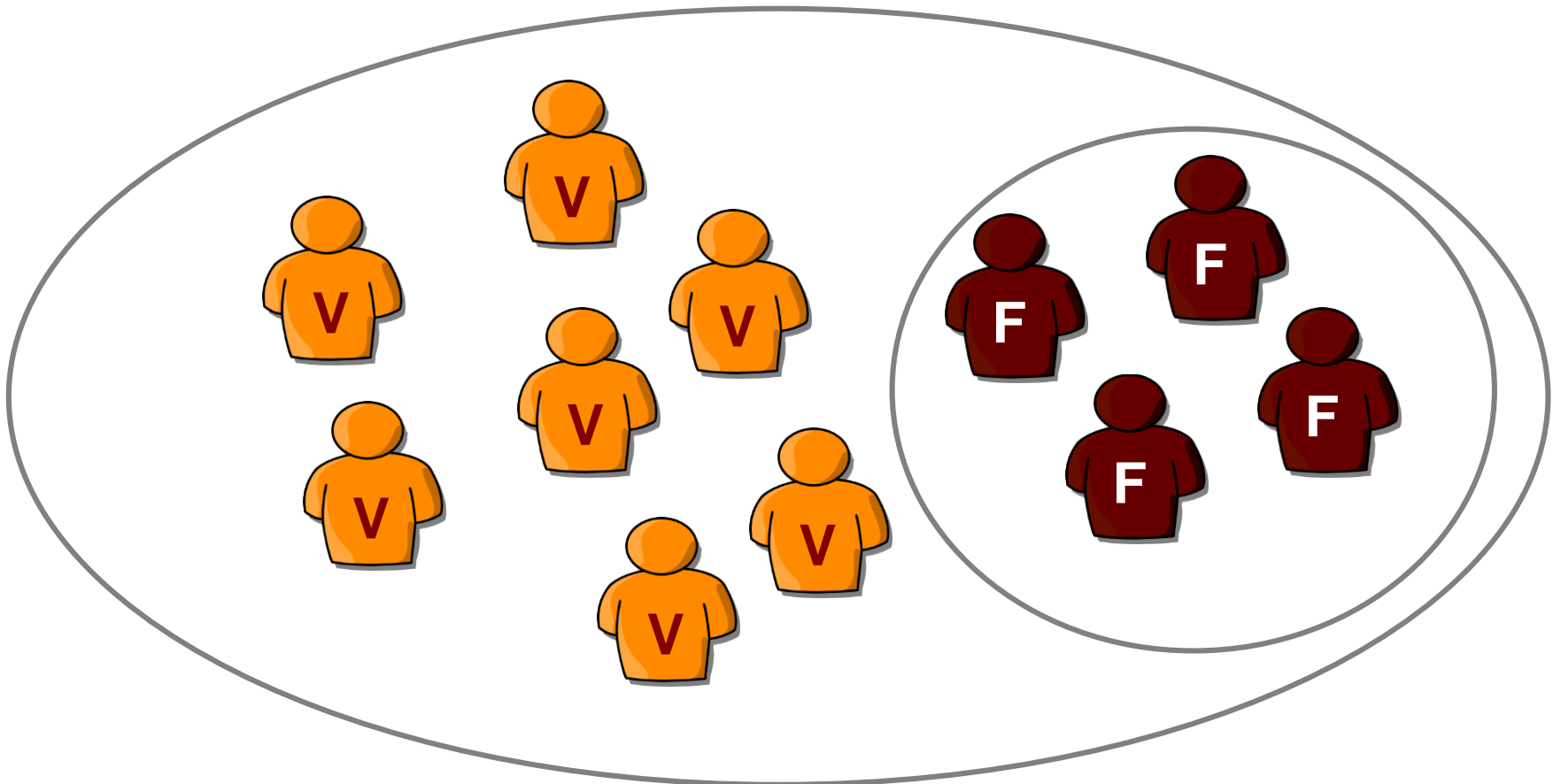
# Ontological Commitment 2

## **SNOMED CT concepts are instantiated by patients or clinical situations.**

- *VentricularSeptalDefect* stands for “Patient with a ventricular septum defect”
- *Tetralogy of Fallot* stands for “Fallot Patient”
- All Fallot patients are also patients with ventricular septum defect because every instance of Tetralogy of Fallot (pathologic structure) has one instance of ventricular septum defect as part
- Consequence:
  - Finding and procedure concepts extend to classes of patients but not to classes of findings or procedures

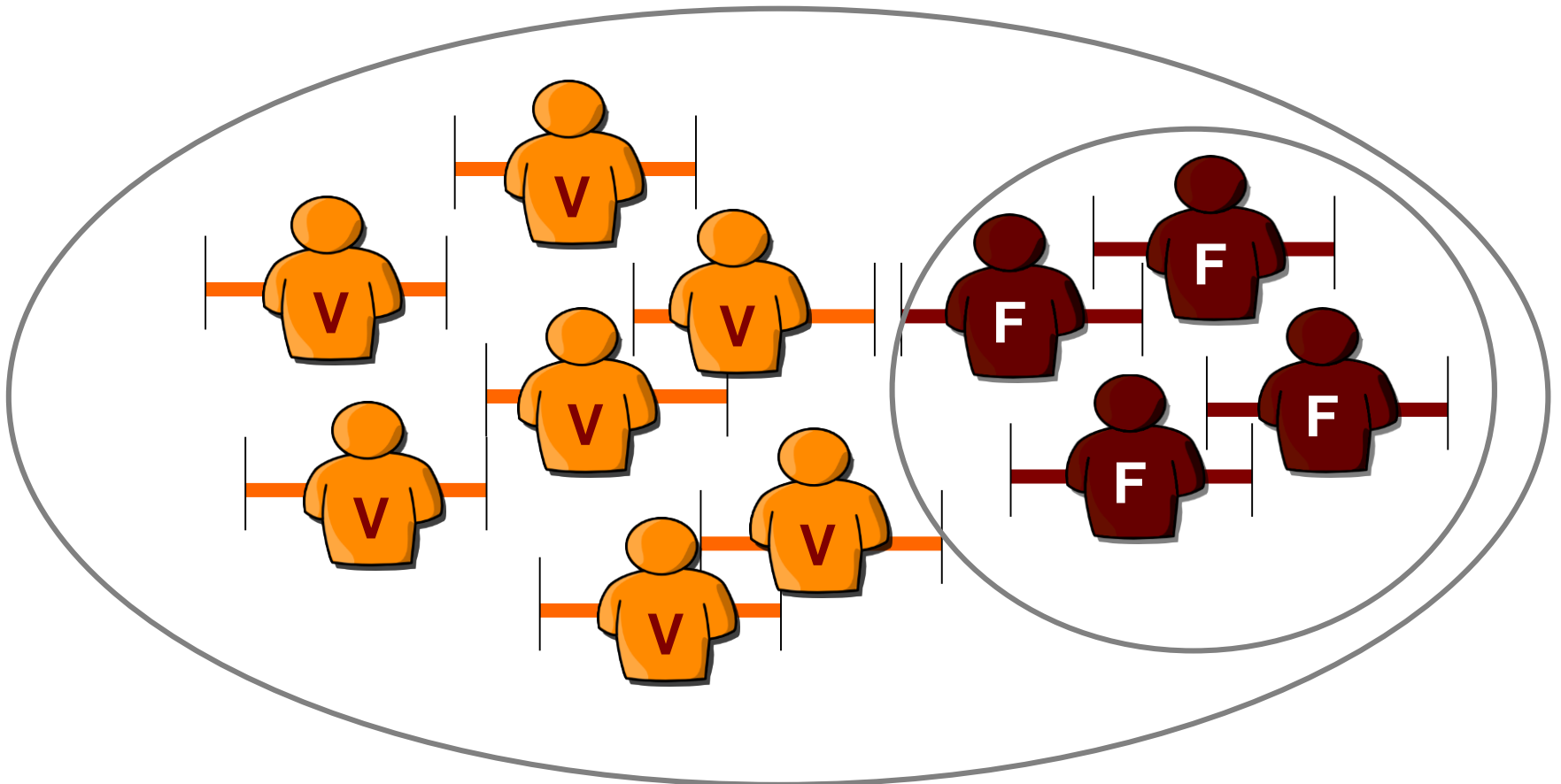
# Ontological Commitment 1

Extension of "**Patient with** *Ventricular Septal Defect*" includes extension of "**Patient with** Tetralogy of Fallot": **TRUE**



# Ontological Commitment 3

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



# Problem

- The same term can be used to denote pathological structures, patients, or situations
- Difficulties with classes that have compositional objects as members

To what extends "A with B":

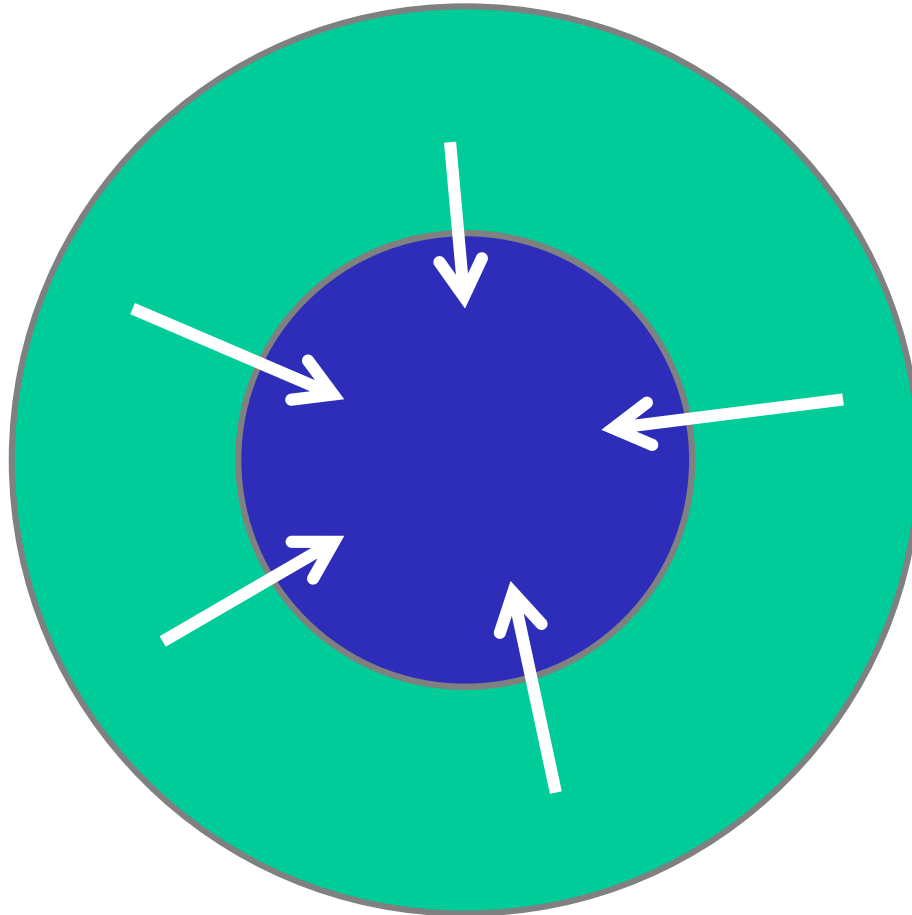
- a mereological sum  $A + B$ ?
- A kind of A which is located in an organism which is also the location of some B ?
- A kind of B which is located in an organism which is also the location of some A ?
- the organism?
- the situation?

# Epistemic contexts

- Pregnancy, not (yet) confirmed
- Diarrhoea of presumed infectious origin
- Atypical squamous cells of uncertain significance, probably benign
- Natural death with probable cause suspected
- Family history of dementia
- Absent foot

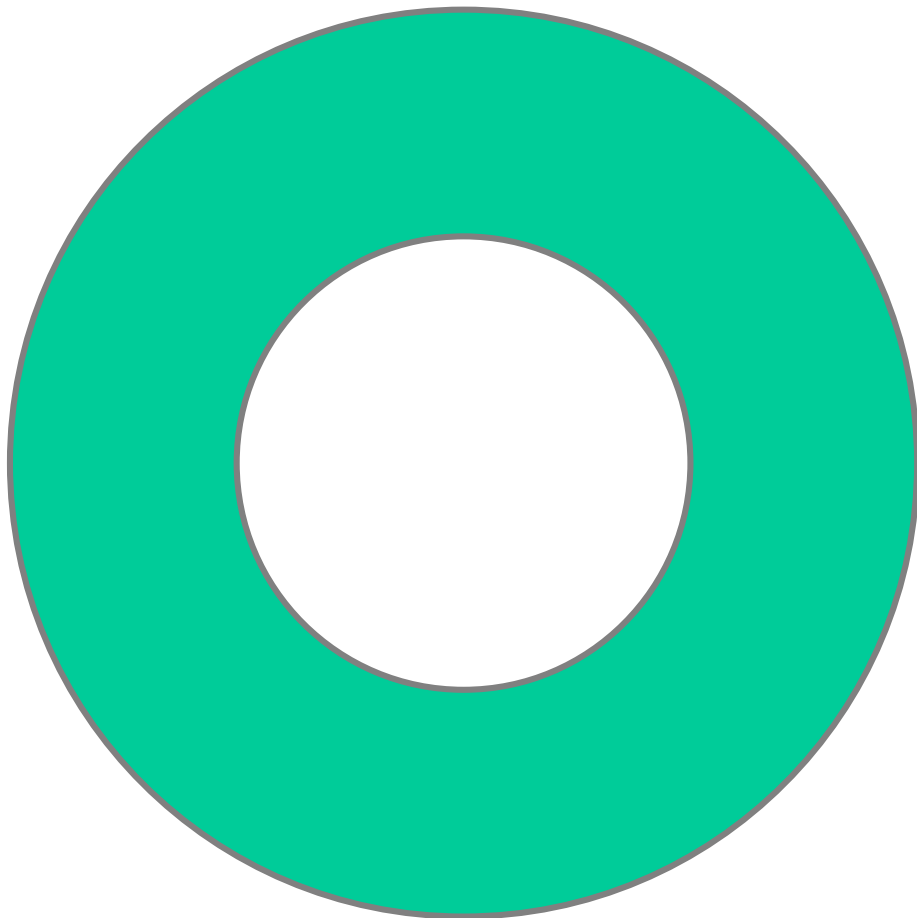


**both in SNOMED CT and ICD**

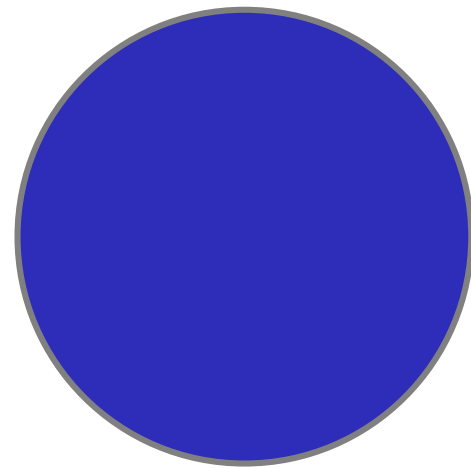


# both in SNOMED CT and ICD

Information entities,  
Diagnostic statements



Context-free representation of  
diseases, disorders



Ontological core

# Other example of conflicting meanings

**Parent(s):**  
 (Select a parent to make it the "Current Concept".)  
[Absence of hand \(finding\)](#)  
[Injury of hand \(disorder\)](#)  
[Traumatic amputation of arm \(disorder\)](#)

**Current Concept:**  
[Traumatic amputation of hand \(disorder\)](#)

← Child

**Child(ren):**  
 (N=3) (Select a child to make it the "Current Concept".)  
[Traumatic amputation of both hands \(disorder\)](#)  
[Traumatic amputation of digit of hand \(disorder\)](#)  
[Traumatic amputation, hand, through metacarpals \(disorder\)](#)

**Current Concept:**  
 Fully Specified Name: Traumatic amputation of hand (disorder)  
 ConceptId: 95856002

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**Defining Relationships:**

**Is a** Absence of hand (finding)  
**Is a** Injury of hand (disorder)  
**Is a** Traumatic amputation of arm (disorder)

Group 1  
**Associated morphology** [Traumatic amputation \(morphologic abnormality\)](#)  
**Finding site** [Hand structure \(body structure\)](#)  
*This concept is fully defined.*

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**Qualifiers:**

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**Descriptions (Synonyms):**  
 Fully Specified Name: Traumatic amputation of hand (disorder)  
**Preferred:** Traumatic amputation of hand  
**Synonym:** Traumatic amputation of hand, NOS

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**Related Concepts:**  
 - All "Is a" antecedents -  
 - All descendants and related subtypes -

↙ Sibling

**S68**

- S68.0** Traumatic amputation of wrist and hand
- S68.1** Traumatic amputation of thumb (complete)(partial)
- S68.2** Traumatic amputation of other single finger (complete)(partial)
- S68.3** Traumatic amputation of two or more fingers alone (complete)(partial)
- S68.4** Combined traumatic amputation of (part of) finger(s) with other parts of wrist and hand
- S68.8** Traumatic amputation of hand at wrist level
- S68.9** Traumatic amputation of other parts of wrist and hand
- S68.9** Traumatic amputation of wrist and hand, level unspecified

**S58**

- S58.0** Traumatic amputation of forearm
- S58.1** **Excludes:** traumatic amputation of wrist and hand ( [S68.-](#) )
- S58.1** Traumatic amputation at elbow level
- S58.9** Traumatic amputation at level between elbow and wrist
- S58.9** Traumatic amputation of forearm, level unspecified

# 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
- Both SNOMED CT and ICD10 mix elements of an ontology with elements of information models (information artifacts)

# The Ontology-Epistemology Divide: A Case Study in Medical Terminology

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**Abstract.** Medical terminology collects and organizes the many different kinds of terms employed in the biomedical domain both by practitioners and also in the course of biomedical research. In addition to serving as labels for biomedical classes, these names reflect the organizational principles of biomedical vocabularies and ontologies. Some names represent invariant features (classes, universals) of biomedical reality (i.e., they are a matter for ontology). Other names, however, convey also how this reality is perceived, measured, and understood by health professionals (i.e., they belong to the domain of epistemology). We analyze terms from several biomedical vocabularies in order to throw light on the interactions between ontological and epistemological components of these terminologies. We identify four cases: 1) terms containing classification criteria, 2) terms reflecting detectability, modality, uncertainty, and vagueness, 3) terms created in order to obtain a complete partition of a given domain, and 4) terms reflecting mere fiat boundaries. We show that epistemology-loaded terms are pervasive in biomedical vocabularies, that the "classes" they name often do not comply with sound classification principles, and that they are therefore likely to cause problems in the evolution and alignment of terminologies and associated ontologies.

## Consolidating SNOMED CT's Ontological Commitment

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### Abstract

*SNOMED CT is a clinical terminology that provides terms with meaning by logical axioms. This enforces precise agreements about the ontological nature of the entities referred to, commonly described as ontological commitment. We demonstrate that SNOMED CT implicitly supports at least three different kinds of commitments, viz. the reference to (i) independently existing entities, (ii) to representational artifacts, and (iii) to clinical situations. Our analysis shows how the truth-value of a sentence changes according to one of these perspectives. We argue that a clear understanding of to what kind of entities SNOMED CT concepts extend is crucial for the proper use and maintenance of SNOMED CT. We argue that the three kinds of commitment can co-exist but need to be clearly distinguished.*