Ontology-based Convergence of Medical Terminologies: SNOMED CT and ICD-11

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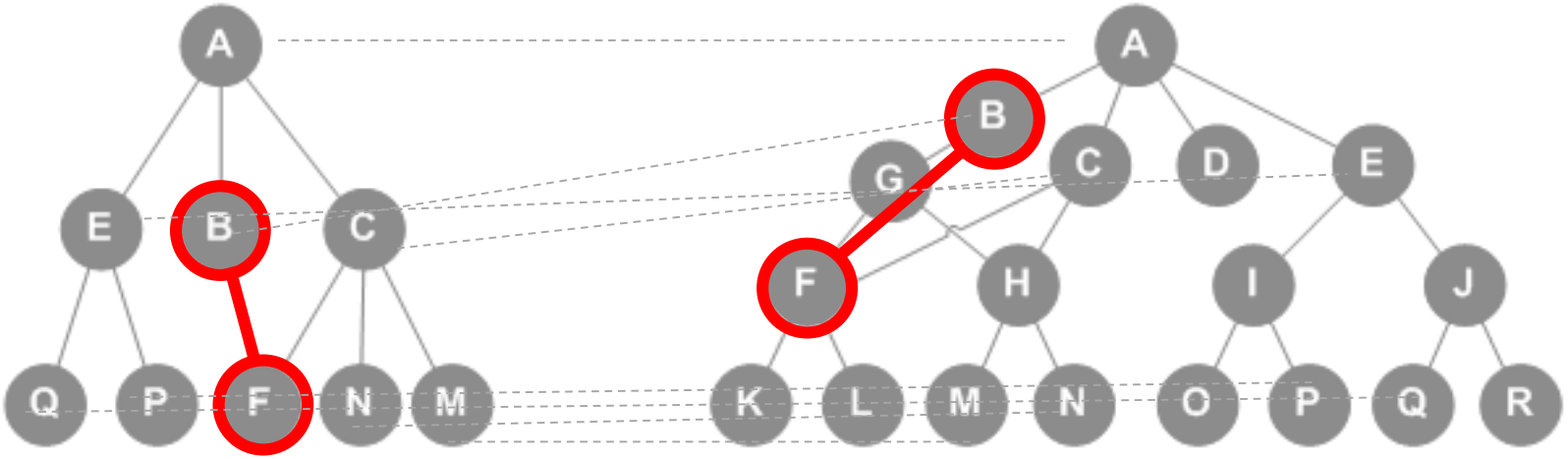
WHO – IHTSDO Joint Advisory Group (JAG) for the harmonisation between ICD-10, ICD-11 and SNOMED CT

ICD-11 – SNOMED CT Harmonization

- Background:
  - ICD: disease classification maintained by WHO (World Health Organization) ICD-11 revision process ongoing
  - SNOMED CT: ontology-based clinical terminology maintained by IHTSDO (International Health Terminology Standards Development Organisation)
  - Size: SNOMED CT >> ICD,
  - Coverage: ICD only diseases, SNOMED CT: all EHR content
- Institutional agreement between WHO and IHTSDO:
  - Goal: common ontological basis for both the ICD-11 foundation component (FC) and SNOMED CT
  - Practical implications:
    - Each class in the ICD-11 foundation component will correspond to exactly one class in SNOMED CT.
    - The transitive closure of taxonomic (subclass) relations in ICD-11-FC is included in the transitive closure of these relations in SNOMED CT.

ICD - SNOMED CT Mapping principle



ICD 11 Foundation Component (multihierarchical)

SNOMED CT

- Taxonomies are main construction principle for both terminologies
- Edges correspond to subclass links. Each ICD class corresponds to exactly one SNOMED class (same letter).
- Subclass links contained in ICD but not SNOMED must be obtained by transitive closure.

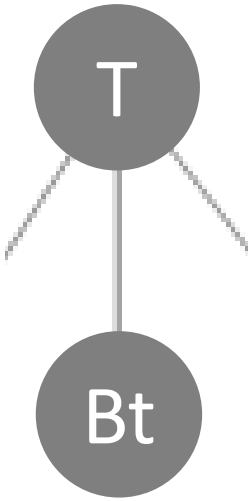
Two Principles of ontology-based mapping of SNOMED CT and ICD-11

1. The semantics of the subclass relation is shared

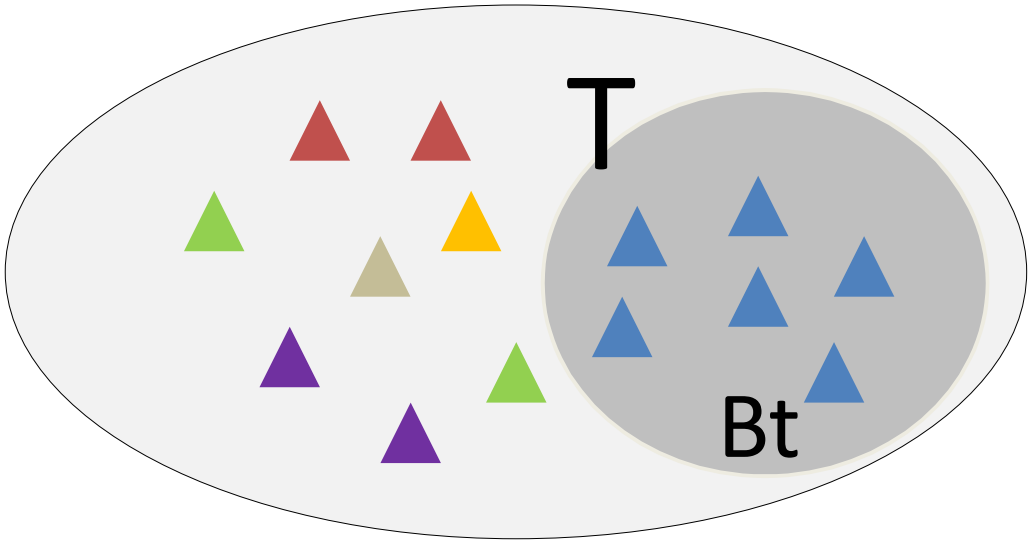
2. Classes to be aligned denote the same types of entities

Meaning of subClassOf

$$subClassOf\ (X, Y) =_{def} \forall i: i \in X \rightarrow i \in Y$$



Graph



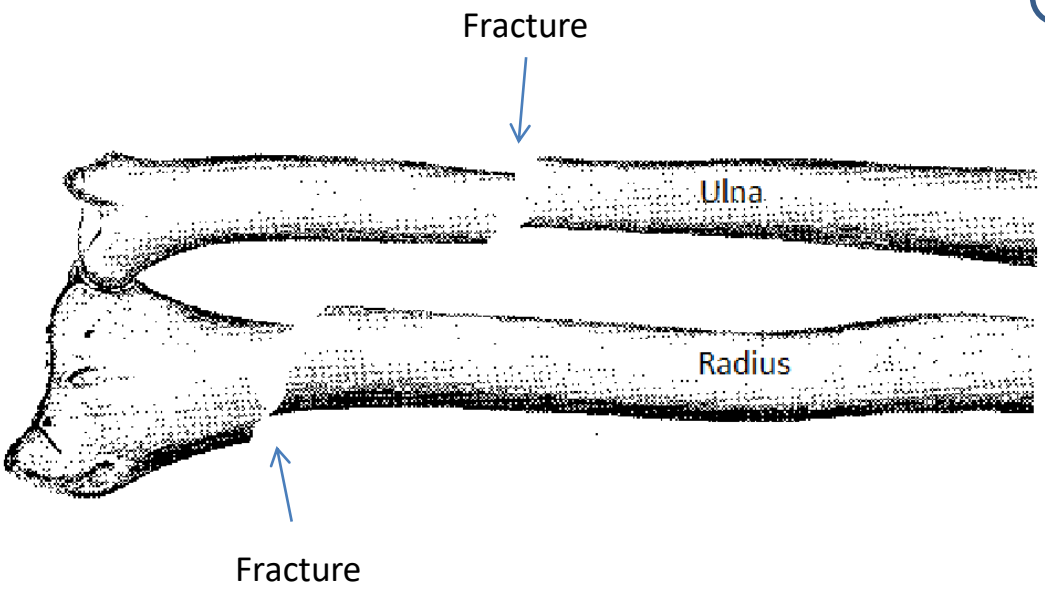
Corresponding Venn diagram

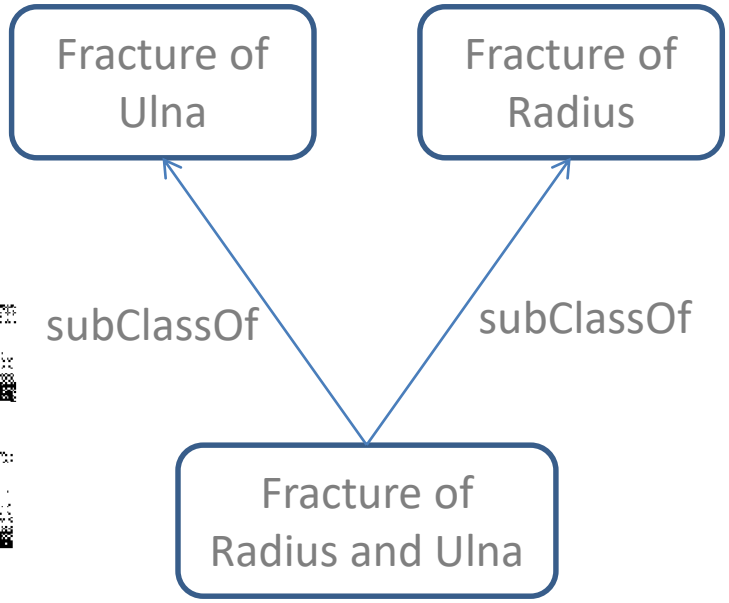
Two Principles of ontology-based alignment of SNOMED CT and ICD-11

1. The semantics of the subclass relation is shared √

2. Classes to be aligned denote the same types of entities ?

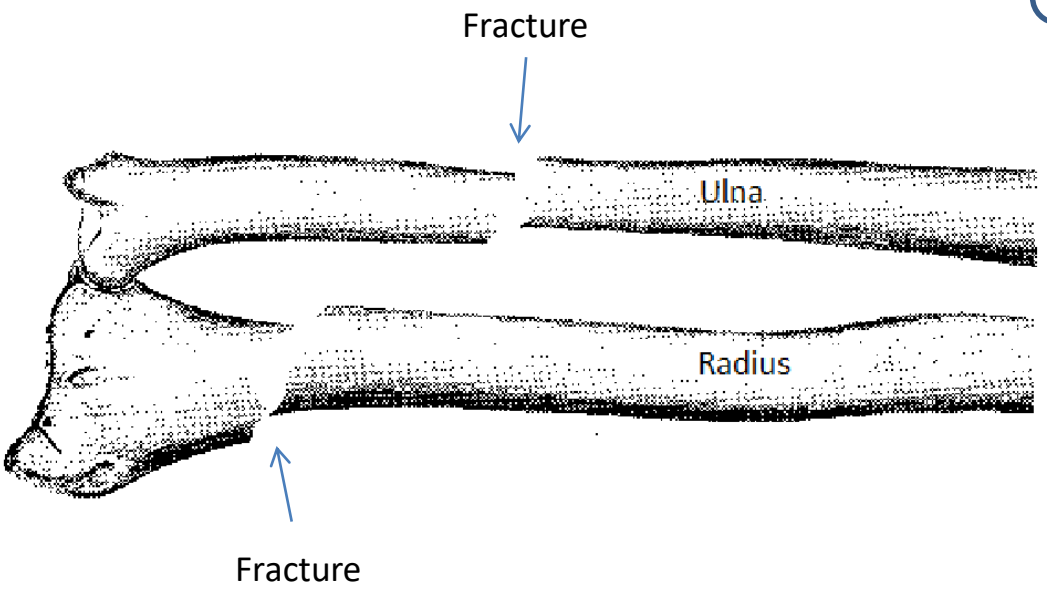
Is this correct?

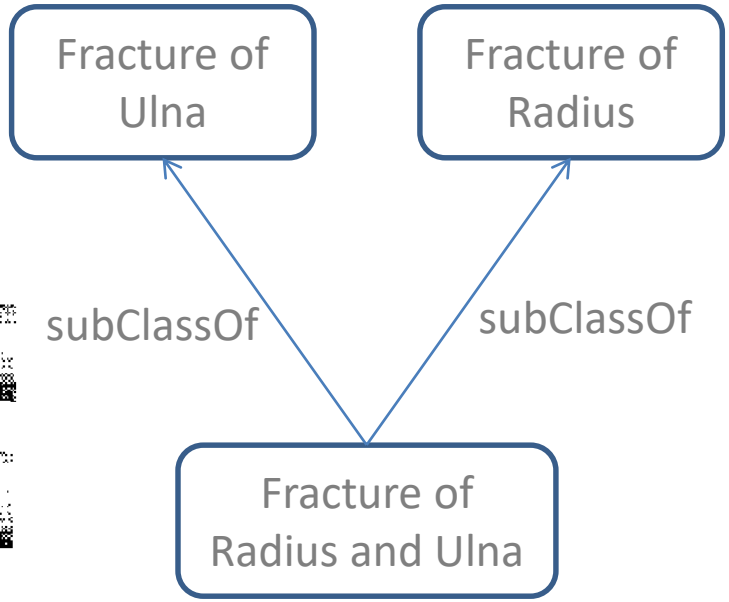




SNOMED CT Example

Is this correct?



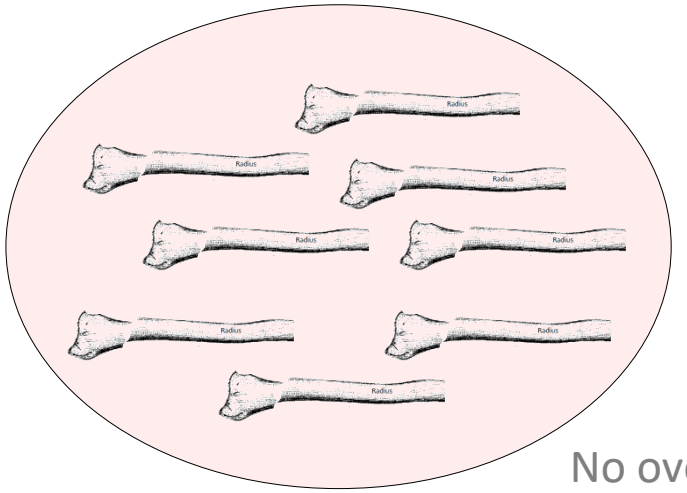


SNOMED CT Example

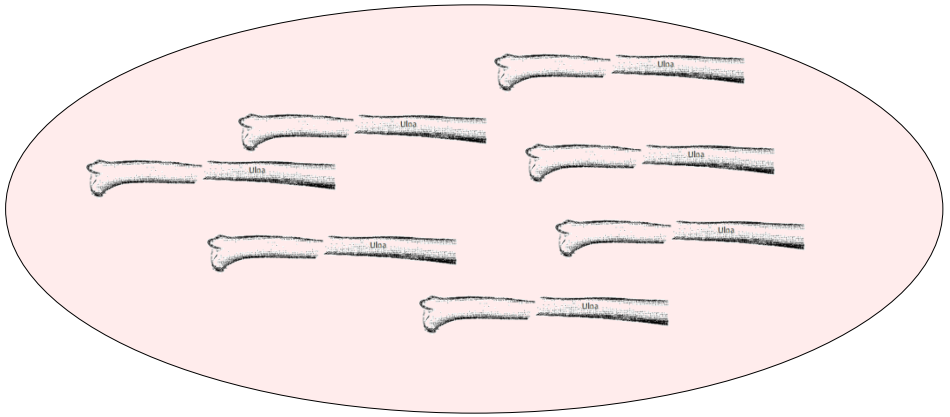
- **No**, if "clinical condition": the combined fracture is composed by the two single fractures, not a subtype
- **Yes**, if "clinical situation":
  "situation with X" or "patient having X"

Clinical condition view

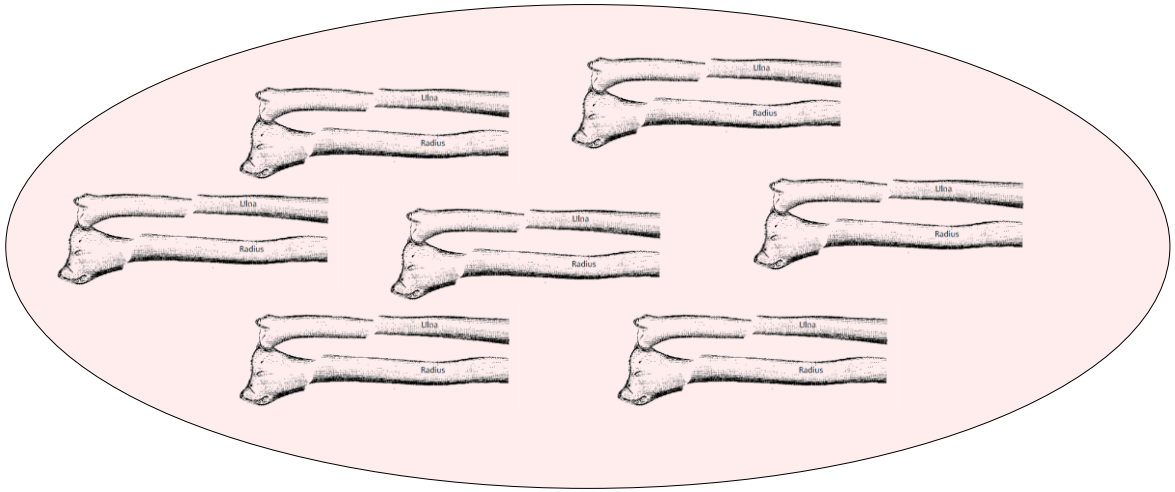
Fractured radius



Fractured ulna

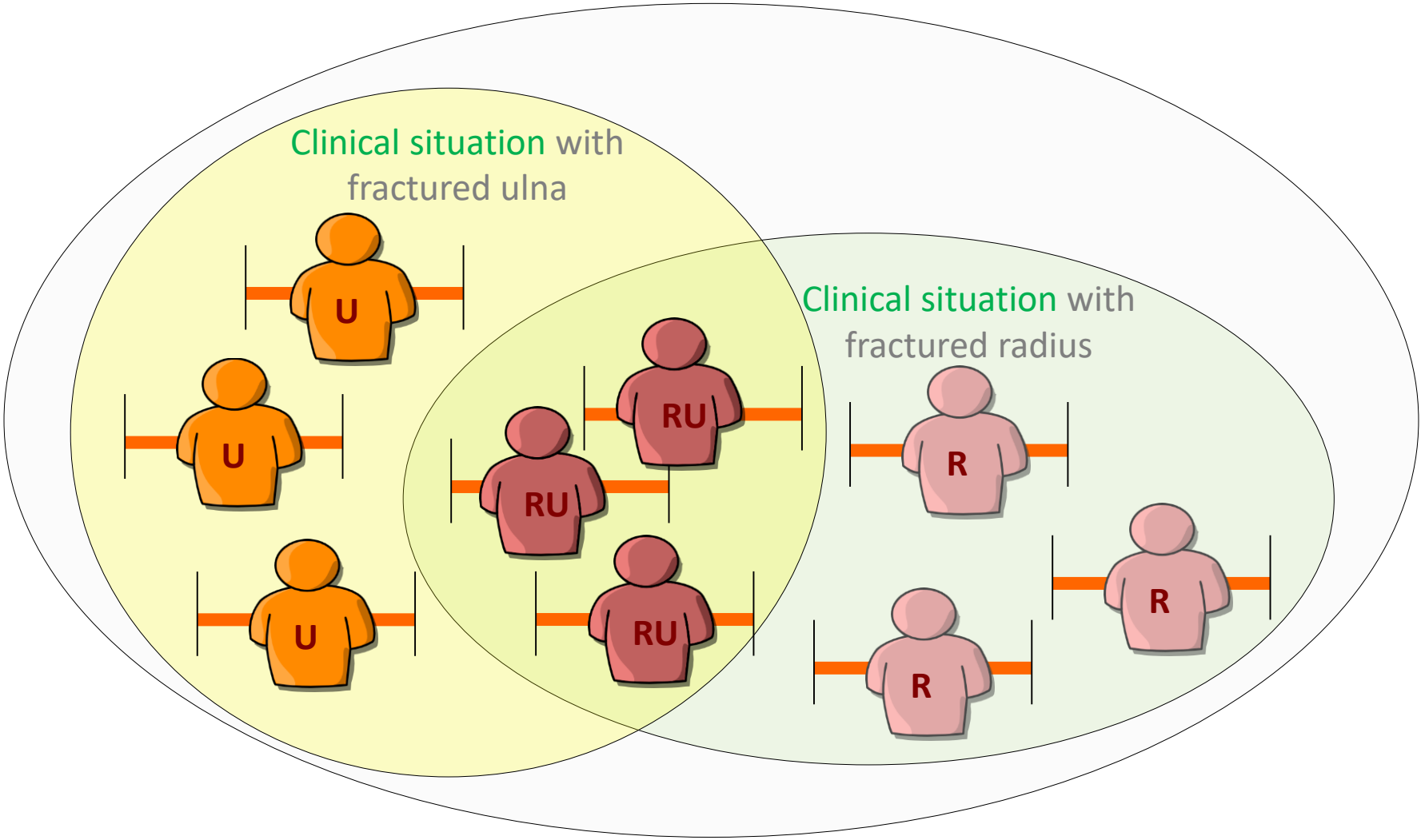


No overlap / inclusion of classes



Fractured radius + ulna

Clinical situation view



Current axiomatization in SNOMED CT

'*Fracture of radius* (*disorder*)' equivalentTo

'*Fracture of forearm* (*disorder*)' and '*Injury of radius* (*disorder*)' and

Group some ('**Associated morphology**' some '*Fracture* (*morphologic abnormality*') and

'**Finding site**' some '*Bone structure of radius* (*body structure*)')

'*Fracture of ulna* (*disorder*)' equivalentTo

'*Fracture of forearm* (*disorder*)' and '*Injury of ulna* (*disorder*)' and

Group some ('**Associated morphology**' some '*Fracture* (*morphologic abnormality*') and

'**Finding site**' some '*Bone structure of ulna* (*body structure*)')

'*Fracture of radius AND ulna* (*disorder*)' equivalentTo

'*Fracture of radius* (*disorder*)' and '*Fracture of ulna* (*disorder*)' and

Group some ('**Associated morphology**' some '*Fracture* (*morphologic abnormality*') and

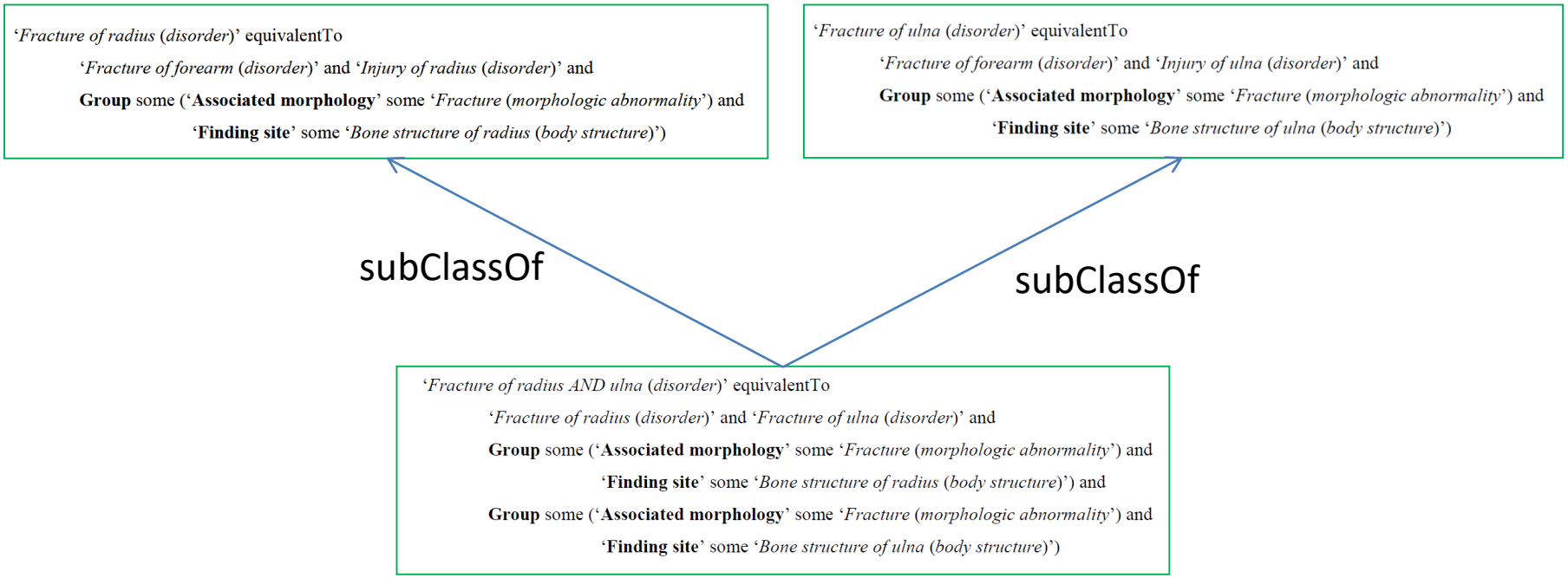
'**Finding site**' some '*Bone structure of radius* (*body structure*)') and

Group some ('**Associated morphology**' some '*Fracture* (*morphologic abnormality*') and

'**Finding site**' some '*Bone structure of ulna* (*body structure*)')

Current axiomatization in SNOMED CT

Inferred taxonomic links



Facts / Hypotheses

- "Problematic" subclass links between SNOMED CT classes result from formal (description logics) definitions
- It can be shown:

A_{cond} subClassOf B_{cond} entails:

A_{sit} subClassOf B_{sit}

A_{cond} subClassOf **hasPart** B_{cond} entails:

A_{sit} subClassOf B_{sit}

→ More subClassOf relations between situation classes

Review of 400 sample SNOMED CT disorder concepts

- Four experts:

  K. Spackman, A. Rector, J.-M. Rodrigues, S. Schulz
- Assessment of a sample of 400 SNOMED disorder concepts
  - Fully specified names
  - Formal definitions
  - Parent classes
  - Child classes
- Evidence for "clinical situation" reading

Schulz S, Rector A, Rodrigues JM, Spackman K. Competing Interpretations of Disorder Codes in SNOMED CT and ICD. Submitted to AMIA 2012

Results

- ~ 11% of disorder evidence that they represent situations and not conditions (such as *Fracture of radius and ulna*)
- For the rest, both interpretations are possible
- Agreement difficult – fuzzy boundary between what should be interpreted as a clinical condition and what as a clinical situation .

Possible actions

1. Redesigning the SNOMED CT disorder hierarchy to exclude interpretation a clinical situations: huge effort, difficult boundary decisions
2. Leaving disorder classes uncommitted: should support condition interpretation: many existing subclass relations wrong
3. Considering all SNOMED CT disorder codes as denoting clinical situations:
   - more robust
   - consistent with current state of the disorder hierarchy
   - agreement with ICD view on the meaning of the code
   - compatible with clinical use cases

Foundations of ontology-based alignment of SNOMED CT and ICD-11

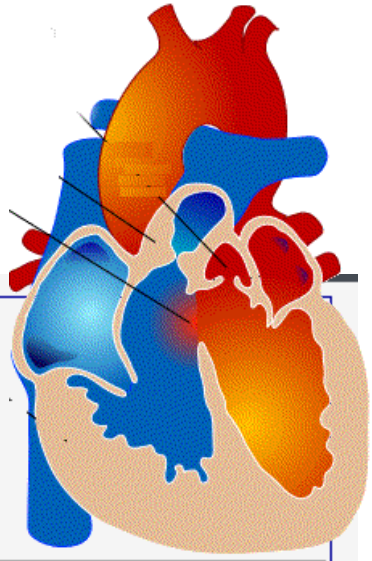
1. The semantics of the subclass relation is shared √

$$subClassOf\ (X, Y) =_{def} \forall i: i \in X \rightarrow i \in Y$$

2. Classes to be aligned denote the same types of entities √

SNOMED CT disorder codes and ICD-11 classes denote:
Clinical Situations

Example 2



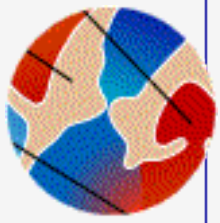
Current Concept:

Fully Specified Name: Tetralogy of Fallot (disorder)

ConceptId: 86299006

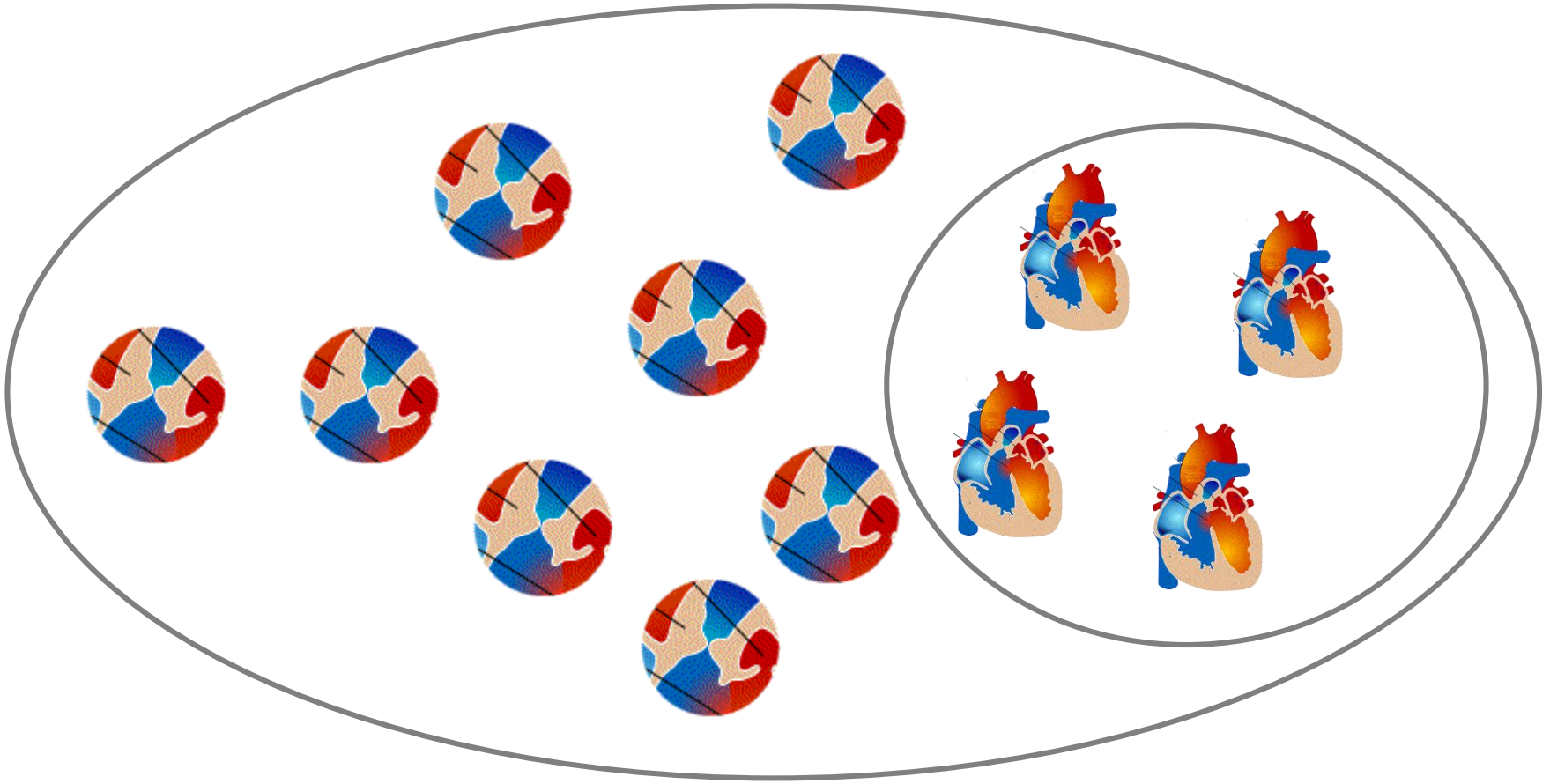
Defining Relationships:

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)



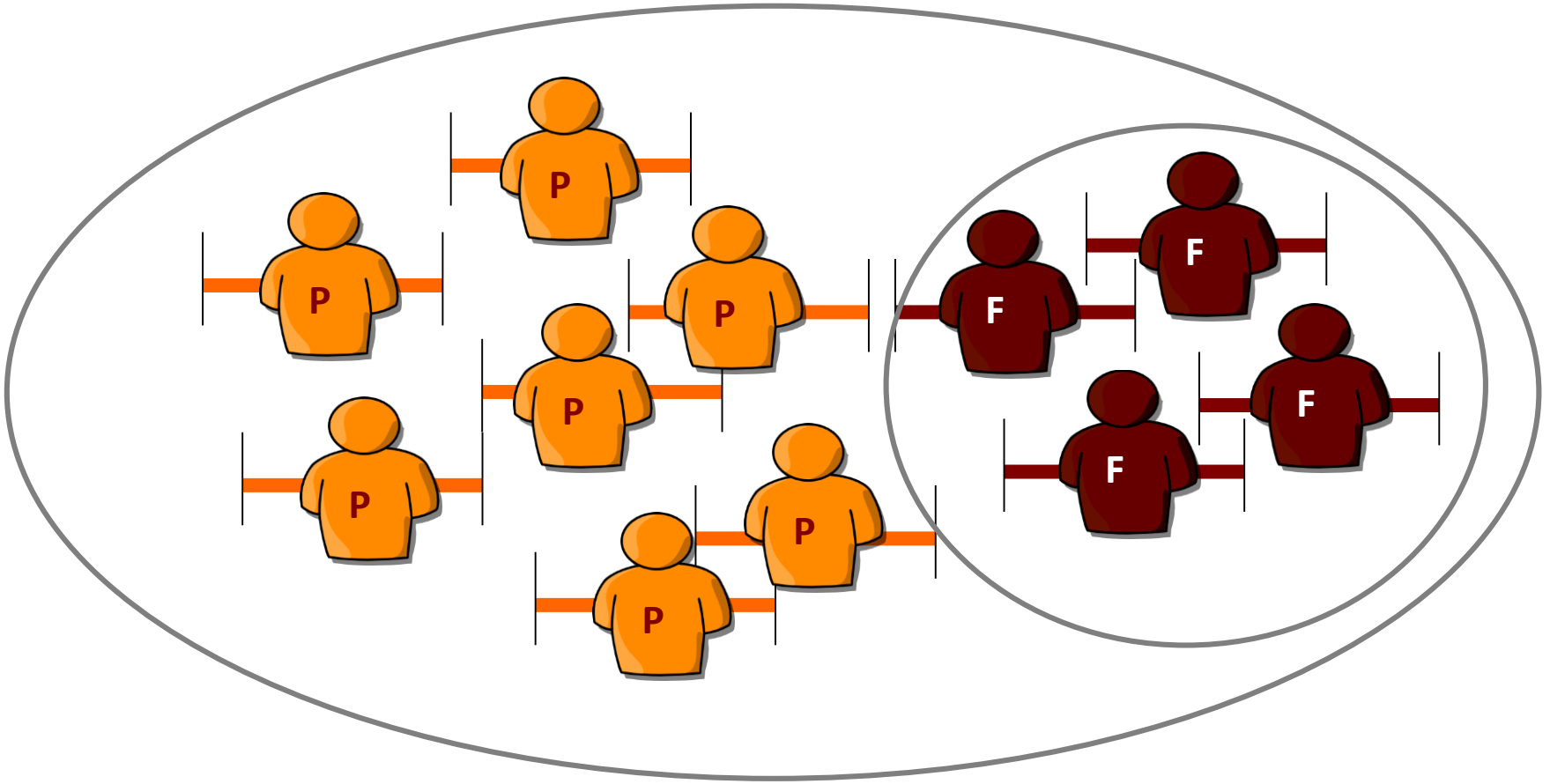
Example 2

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



Example 2

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



Two diverging interpretations of disorder terms in SNOMED CT and ICD:

- They denote patient-borne **Conditions** such as body processes, states, dispositions, or (patho-) anatomical structures, which are reportable in the context of medical records
- They denote Clinical **Situations**, which are defined as phases of a patient's life, during which he/she is bearer of (some combination of) pathological conditions.

Situations, conditions and role groups

'*Fracture of radius AND ulna* (*disorder*)' equivalentTo

'*Fracture of radius* (*disorder*)' and '*Fracture of ulna* (*disorder*)' and

Group some ('**Associated morphology**' some '*Fracture* (*morphologic abnormality*') and

'**Finding site**' some '*Bone structure of radius* (*body structure*)') and

Group some ('**Associated morphology**' some '*Fracture* (*morphologic abnormality*') and

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'*Fracture of forearm* (*disorder*)' and '*Injury of ulna* (*disorder*)' and

Group some ('**Associated morphology**' some '*Fracture* (*morphologic abnormality*') and

'**Finding site**' some '*Bone structure of ulna* (*body structure*)')

Facts / Hypotheses

- Most SNOMED CT disorder concepts contain role groups
- The role group link can be interpreted as a relation that links a situation with a condition
- It can be shown:
  - ‘A_cond subClass of B_cond’ entails: ‘A_sit subClass of B_sit’
  - ‘A_cond subClass of hasPart B_cond’ entails: ‘A_sit subClass of B_sit’

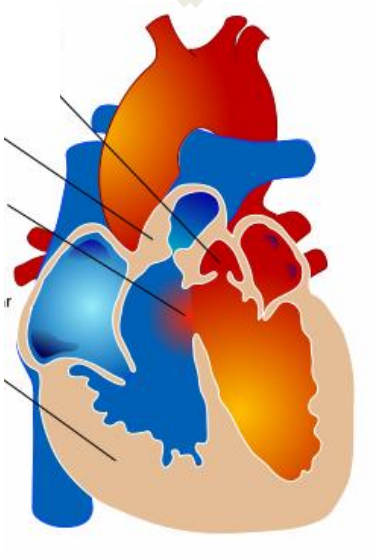
Schulz S, Rector A , Rodrigues JM ,Chute C , Üstün B , Spackman K . ONTOLOGY-BASED CONVERGENCE OF MEDICAL TERMINOLOGIES: SNOMED CT AND ICD-11. In: Schreier G, Hayn D, Hörbst A, Ammenwerth E, editors. Proceedings of the eHealth2012. 2012 Mai 10-11; Vienna, Austria. OCG; 2012.

Proper parts or taxonomic parents ?

Example from Harold Solbrig

VSD PVS RVH OA

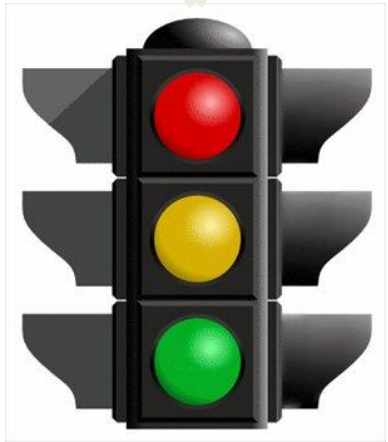
is-a is-a is-a is-a



Tetralogy of Fallot

Red Light Yellow Light Green Light

is-a is-a is-a

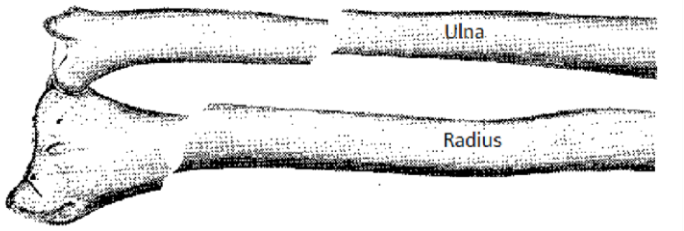


Traffic Light

Proper parts or taxonomic parents ?

Example from Harold Solbrig

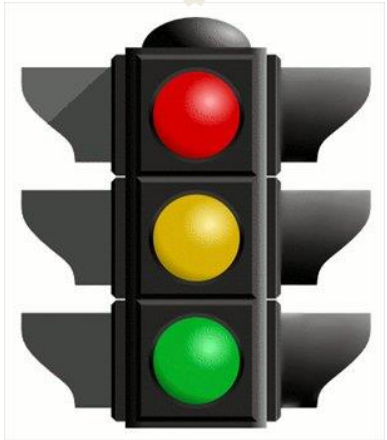
subClassOf



Combined fracture

Red Light Yellow Light Green Light

subClassOf



Traffic Light