INTERNATIONAL HEALTH TERMINOLOGY
STANDARDS DEVELOPMENT ORGANISATION

WHO – IHTSDO: SNOMED CT – ICD-11 coordination: Conditions vs. Situations

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Principles of ICD-SNOMED mapping within WHO IHTSDO JAG

- Goal: common ontological basis for both the (polyhierarchical) ICD-11 foundation component and SNOMED CT
- Each class (categories) in the ICD-11 foundation component will correspond to exactly one class in SNOMED CT. Exceptions: navigational classes, should be clearly kept distinct from ontological classes.
- The equivalence in meaning between these class pairs will be assured by common text definitions.
- The transitive closure of taxonomic (subclassOf) relations in ICD-11-FC is included in the transitive closure of subClassOf relations in SNOMED CT.

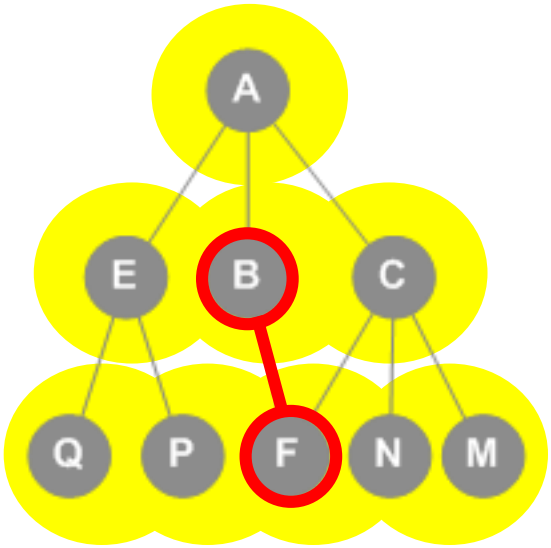
Summary Alan Rector's paper (I) + discussion

- **ICD Foundational Layer**: contains much more knowledge than just ontology
  - **Ontological component**:
    - definitional / universal: "All x has some y"
    - What is shared with SNOMED CT is just the ontological "spine", mainly the is-a hierarchies + axioms
    - text definitions
  - **Content model component**:
    - Non-ontological knowledge, e.g. Leukemia can be treated by Metotrexate, or
    - Those non-ontological pieces of knowledge are in the "content model": probabilistic knowledge, default knowledge,
    - Refer to SNOMED CT concepts as value sets, but not in terms of DL logic
    - navigational classes
  - **Metadata component**
  - **Linguistic component**: Labeling information (linguistic), including multilingual issues

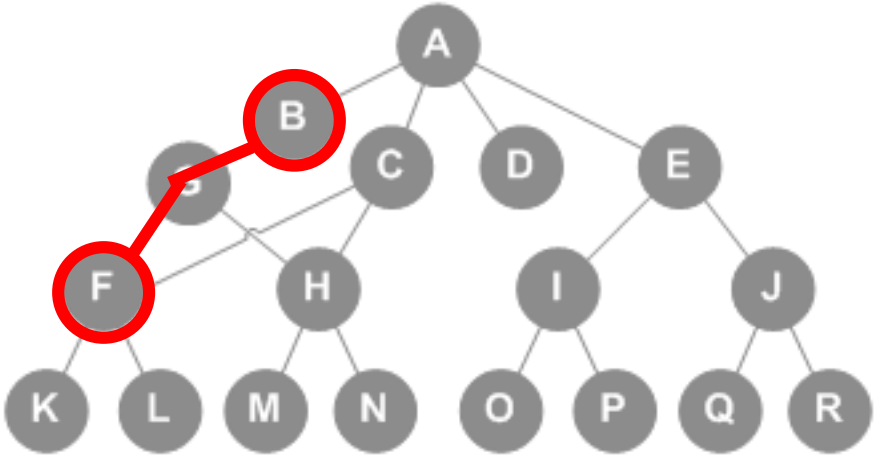
Summary Alan Rector's paper (II)

- URIs: joint / separate
- Distribution form
- "Projection" (more an SW engineering issue)
  - Static version: no need to replicate
  - Dynamic version: replicate

Mapping principle



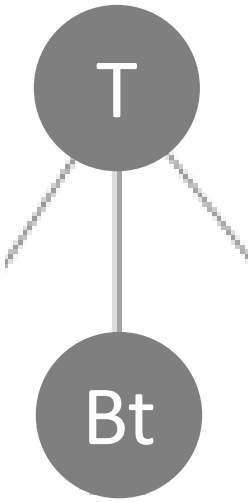
ICD 11 FC



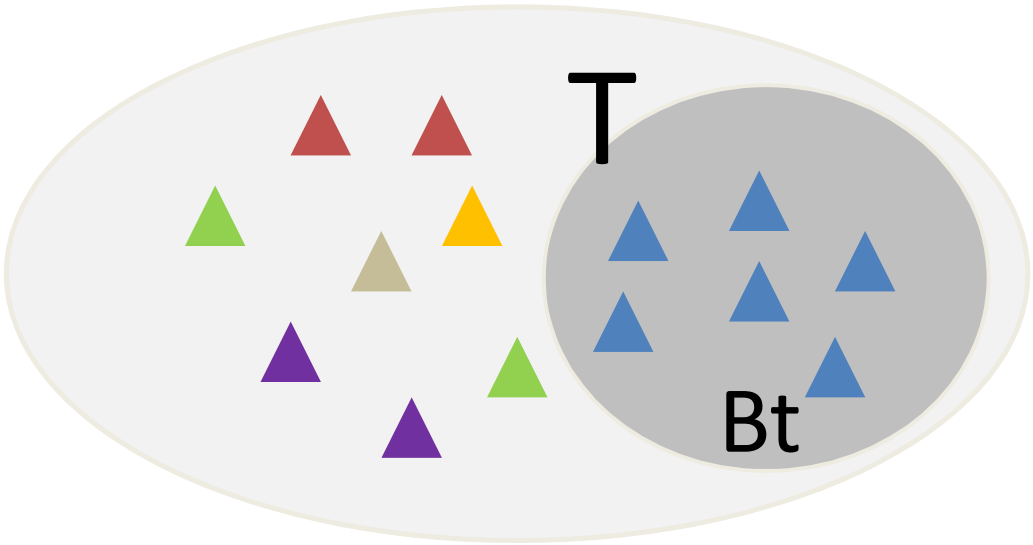
SNOMED CT

Edges correspond to subClassOf links. Each ICD class corresponds to exactly one SNOMED class (same letter).
SubClassOf - links contained in ICD but not SNOMED can be obtained by transitive closure.

Meaning of subClassOf (is-a)



Graph

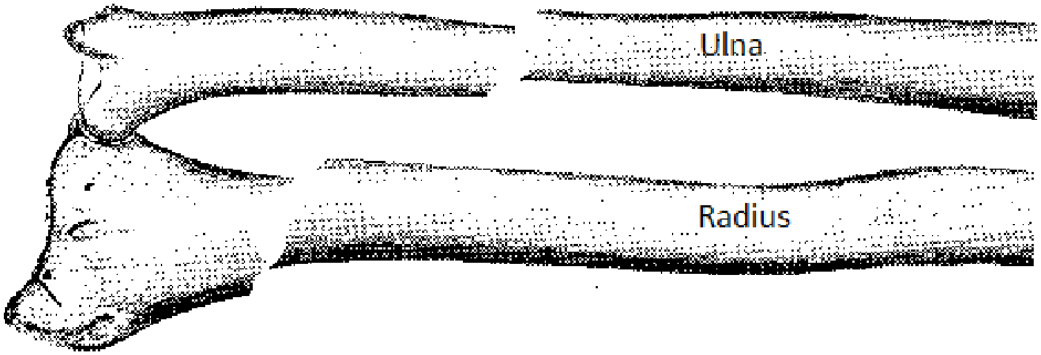


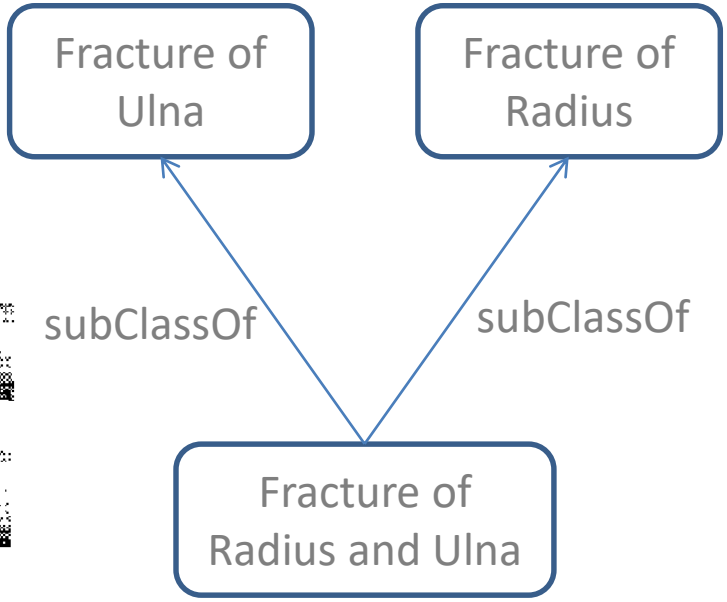
Corresponding Venn diagram

Alignment of SNOMED CT and ICD11 requires that in both systems

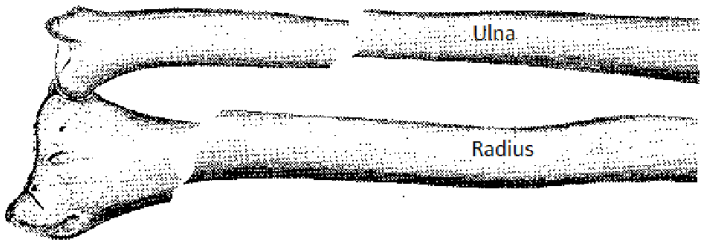
1. The semantics of the subclass relation is shared
2. Classes to be aligned denote the same entities

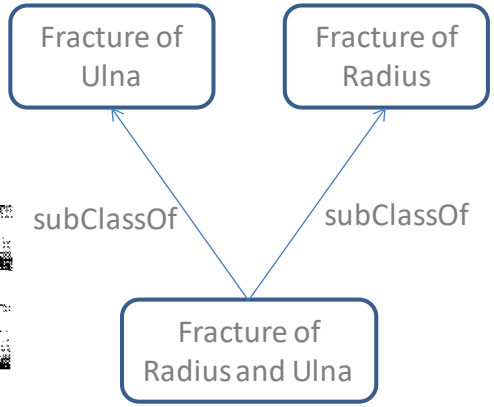
Example 1


Ulna
Radius


Fracture of Ulna
Fracture of Radius
subClassOf
subClassOf
Fracture of Radius and Ulna

Is this True?

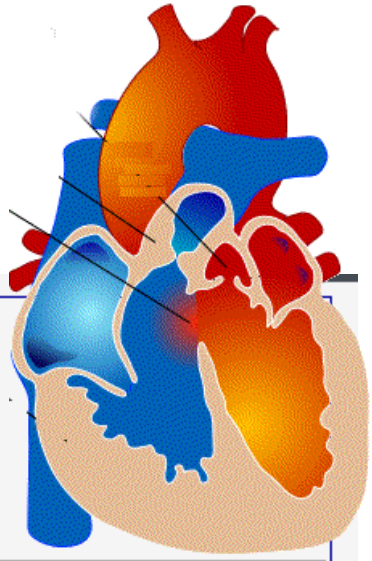




- FALSE, if X means “pathological entity”
- TRUE, if X means “situation with X” or “patient having X” (“additivity”)

Schulz S, Spackman K, James A, Cocos C, Boeker M. **Scalable representations of diseases in biomedical ontologies.** J Biomed Semantics. 2011 May 17;2 Suppl 2:S6.

Example 2



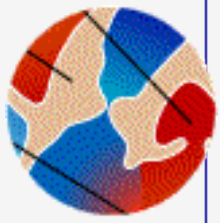
Current Concept:

Fully Specified Name: Tetralogy of Fallot (disorder)

ConceptId: 86299006

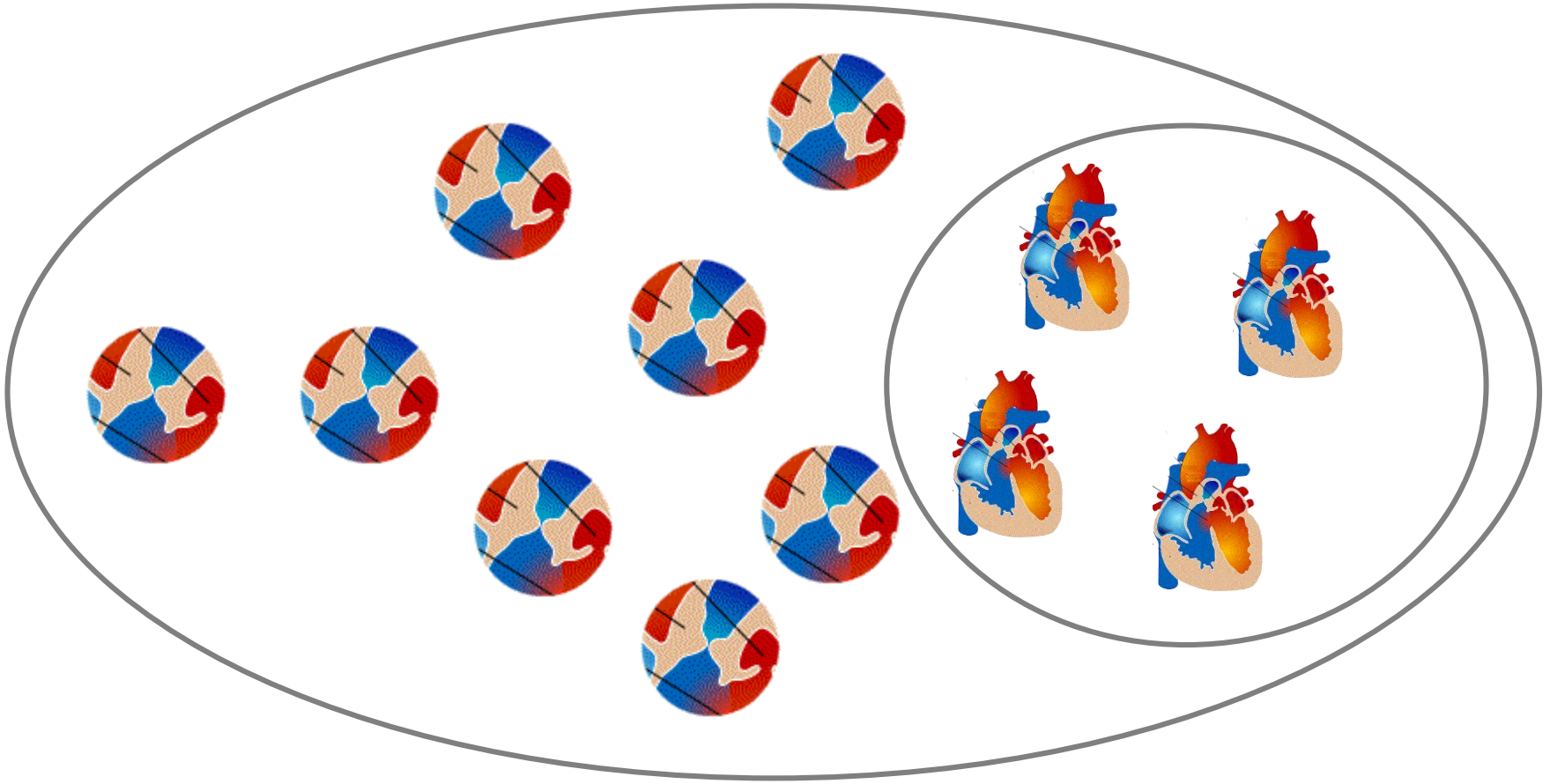
Defining Relationships:

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)



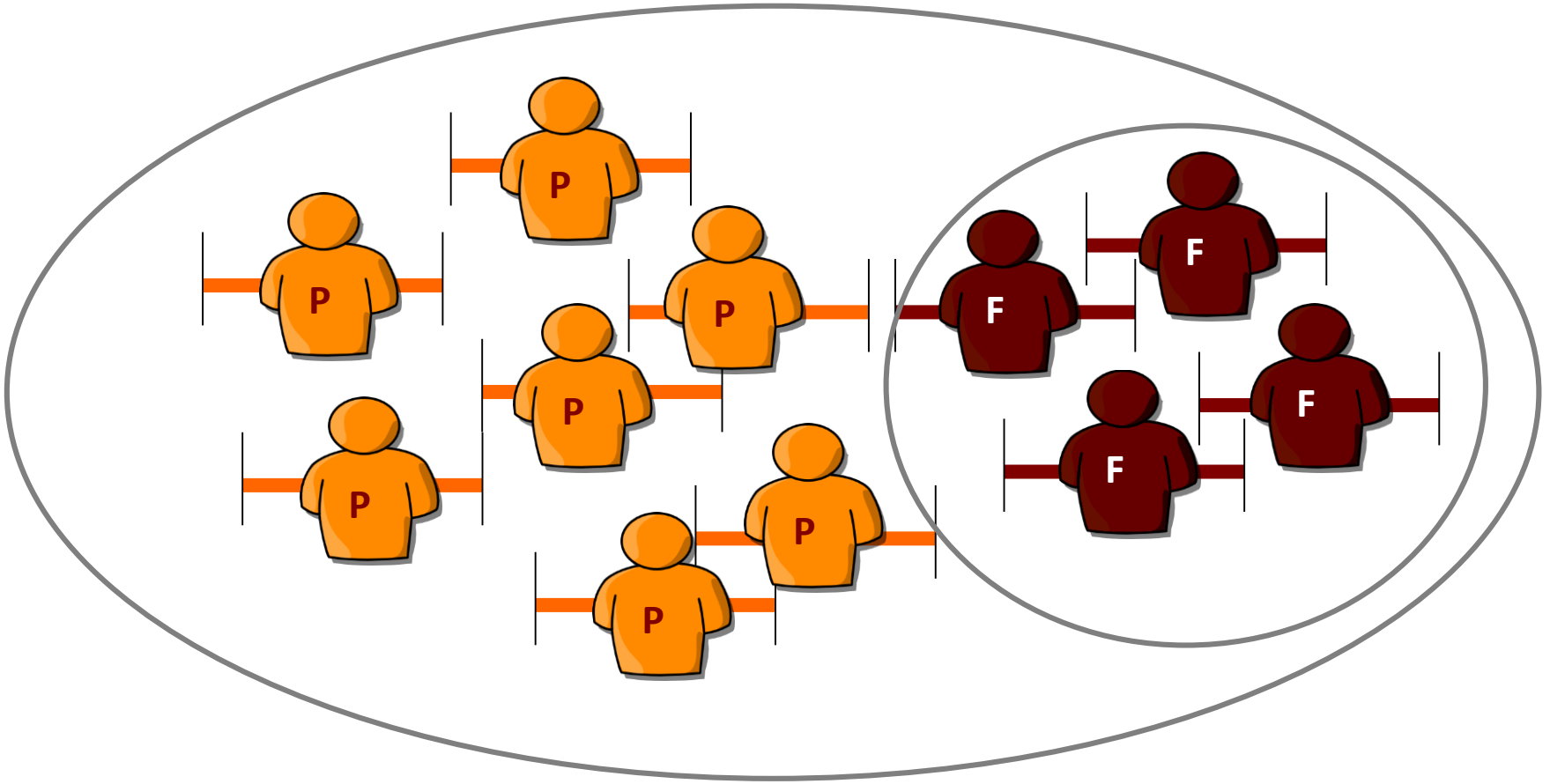
Example 2

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**

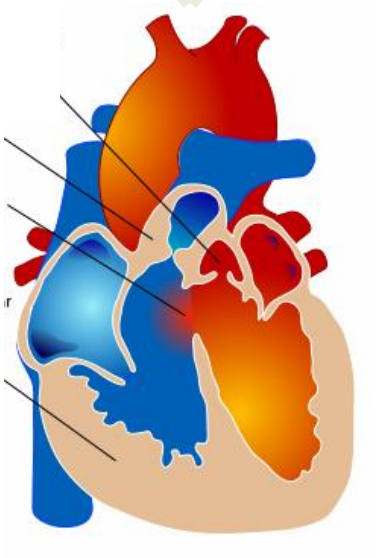


Proper parts or taxonomic parents ?

Example from Harold Solbrig

VSD PVS RVH OA

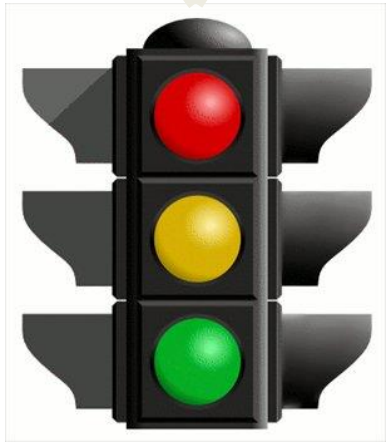
is-a is-a is-a is-a



Tetralogy of Fallot

Red Light Yellow Light Green Light

is-a is-a is-a



Traffic Light

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

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

'*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*)')

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.

Review of 400 sample disorder concepts

- 4 experts: Kent Spackman, Alan Rector, Jean-Marie Rodrigues, Stefan Schulz
- Assessment of
  - FSN
  - Formal definitions
  - Children

  Of a sample of disorder concepts

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

Table 1. Stratification of sample by distance from the root of the SNOMED CT hierarchy

Distance from root	Number of concepts	Proportion	Number in Sample	Stratified distance	Proportion in Stratum	Concepts in Stratum
0	1	0.0%	0	1	14,2%	56
1	78	0.1%	0	1		
2	2127	3.3%	13	1		
3	7090	10.8%	43	1		
4	15657	23.9%	96	2	23.9%	96
5	17602	26.9%	108	3	26.9%	108
6	13457	20.6%	82	4	20.6%	82
7	6392	9.8%	39	5	14,4%	58
8	2319	3.5%	14	5		
9	577	0.9%	4	5		
10	92	0.1%	1	5		
11	4	0.01%	0	5		

Table 2. Rating results

	C1: positive additivity of parent concepts	C2: Child concepts with additivity (count)	C2':At least one child concept evident for Situation	C3: Evidence of Situation by fully specified name (ratio of positive ratings)	At least one positive rating at the concept level (C1 or C3), ratio of pos. ratings	At least one positive rating at concept + Child level (C1, C2',C3), ratio of pos. ratings
Sample size	400	559	400	400	400	400
Number in sample	43	223	72	64	88	143
Ratio	10.8%	39.9%	18.0%	16.0%	22.0%	35.8%
Cohen's Kappa for binary ratings	0.32	-	0.18	0.61	0.56	0.26
Normalized to all disorder concepts (total = 65396)	7,030	26,088	11,771	10,463	14,387	23,379
With 95% confidence interval	[± 1,962]	[± 2,001]	[± 2,425]	[± 2,341]	[± 2,648]	[± 3,061]

Table 3. Influence of role groups

Number of role groups (inherited plus asserted)	Number of concepts in the sample	Positive ratings at concept level (C1 or C3)	Percentage of positive ratings
0	19	2	10.5%
1	194	39	20.1%
2	124	29	23.3%
3	52	15	28.8%
4 and more	11	3	27.3%

Table 4. Influence of depth (stratified, see Table 1)

Hierarchical level	Number of concepts in the sample	Positive ratings at concept level (C1 or C3)	Percentage of positive ratings
1	56	12	21.4%
2	96	21	21.8%
3	108	24	22.2%
4	82	19	23.1%
5	58	12	20.7%

Table 5. Influence of topology

Topology	Number of concepts in the sample	Positive ratings at concept level (C1 or C3)	Percentage of positive ratings
Terminal concepts (without children)	269	69	25.6%
Non-terminal concepts (with children)	131	19	14.5%

Results

- ~ 11% of disorder concepts represent situations rather than conditions
- For the rest, both interpretations are possible
- Agreement difficult – fuzzy boundary between what should be interpreted as a condition and what as a situation

Conclusions

- Redesigning the disorder hierarchy to exclude situation interpretation: huge effort, difficult decisions
- Leaving disorder code uncommitted: many existing subclass relations wrong
- Considering all disorder codes as denoting situation: consistent with current state of the disorder hierarchy, only rationale for concepts with single role groups
- If explicit reference to conditions: post-coordination