Representation of Part-Whole Relationships in SNOMED CT

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Outline

- Part/Whole Relationship Effect on Classification of Disease in SNOMED CT
- Review Approaches for Representing this Pattern in OWL-DL for SNOMED CT
- Present Five Representational Approaches Based on Translations on Current Approach
- Conclusions / Future Work

Introduction

- Representing and reasoning on relationships such subclass-of (for generalization) and part-of (for aggregation) is crucial for medical information systems.
- Is *part-of* transitive?
  - Usage must be consistent
- Generally transitive for anatomy
  - The femur is part of the leg, leg is part of the body therefore femur is part of the body
- **Part-Whole Specialization**
  - Inheritance of roles along a part-whole taxonomies
  - Fracture of the femur is a fracture of the leg
  - A disease of the heart valve is a disease of the heart
- Representation and reasoning must handle where this sort of inference does not always hold:
  - Transplantation of the aorta ***is not*** a transplantation of the heart
  - Amputation of the finger ***is not*** an amputation of the hand

Primarily 3 Methods to Represent This Pattern

1. **Propagation Across Transitive Properties**
   - In earlier languages achieved by right identities (Sterns et al., 2001) and refined by (Rogers and Rector, 2000).
   - In this case Heart disease is defined simply as ‘Disorder that has locus some Heart’.
     (x has_locus y y part_of z x has_locus z)
   - Originally thought to be intractable, but since shown to be not (Horrocks and Sattler, 2004) and within EL++.
2. **Explicit Definition of Disease as Disjunctions**
   - Heart disease is defined explicitly as ‘Disease that has locus some Heart OR some part of Heart’.
   - Explicitly represents Method 3.
   - Within ALC, but outside EL++.
3. **Use of Structure-Entity-Part (SEP) Triples**
   (Hahn, Schulz, and Romacker, 1999).
   - For SEP, part-whole reasoning is implicitly done via subsumption reasoning.
   - Within ALC, does not require transitive properties, disjunctions or property paths.

SNOMED CT History

- SNOMED CT originally developed using a variant of #1.
- SNOMED CT converted to # 3 (SEP triples).
- Recently re-examined in the light of experience.
- #2 experimental, one format being considered a variant of #1.
- Here explore variants on the three methods in the light of modern description logics
  - Focus on formal, rather than cognitive aspects.

SEP Triples

- 'S' stands for a Structure Class
  - *Heart*$_S$: A part of the heart or a whole heart
- 2 Asserted Subclasses: 'E' and 'P' classes for Entire and Part Class
  - *Heart*$_E$: A heart
  - *Heart*$_P$: A part of a heart

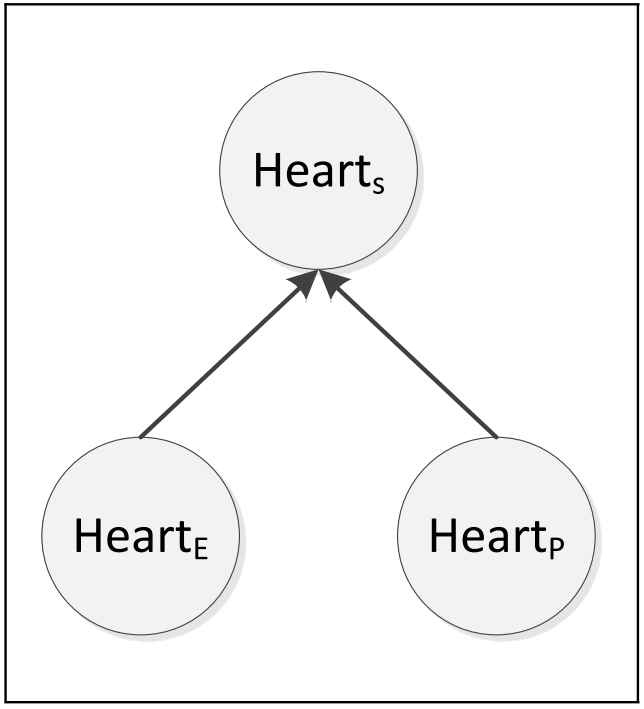

Figure: SEP Triple for Heart

SEP Triples

- SEP triples given for all anatomical entities
- e.g., Myocardium (*Myocardium*$_S$, *Myocardium*$_E$, *Myocardium*$_P$)
- Myocardium$_S$ is an asserted subclass of Heart$_P$

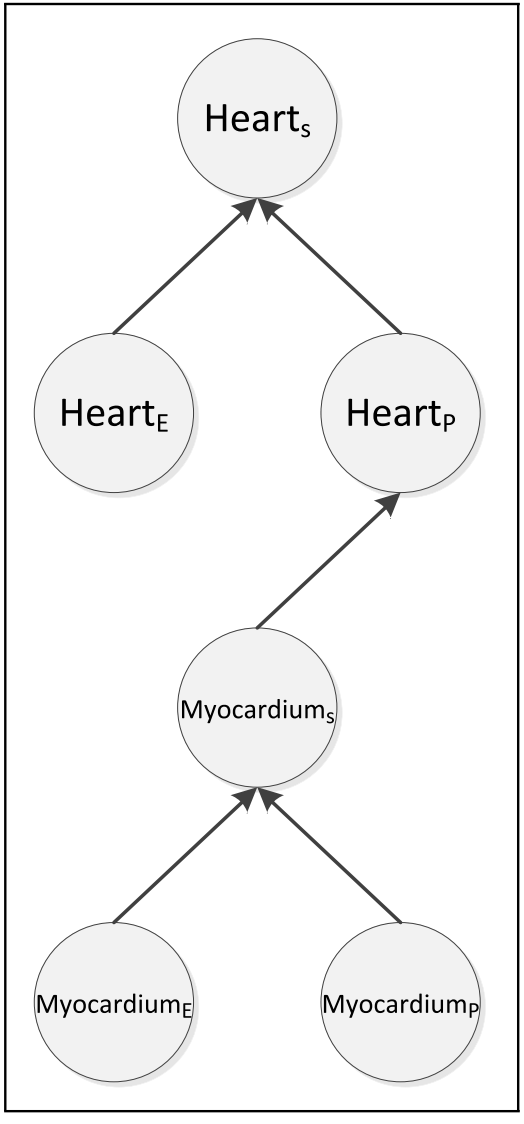

Figure: Additional SEP triples

SEP Triples

- Also, Heart_S is a subclass of Body_P
- $Myocardium_E \sqsubseteq Myocardium_S \sqsubseteq$

  $Heart_P \sqsubseteq Heart_S \ldots \sqsubseteq Body_P \sqsubseteq Body_S$
- $Heart_E \sqsubseteq Heart_S \ldots \sqsubseteq Body_P \sqsubseteq Body_S$
- Therefore:
  - A part of a myocardium or a whole myocardium is a part of some heart, a part of a heart or a whole heart is a part of some body.
  - A part of a myocardium or a whole myocardium is a part of some body.

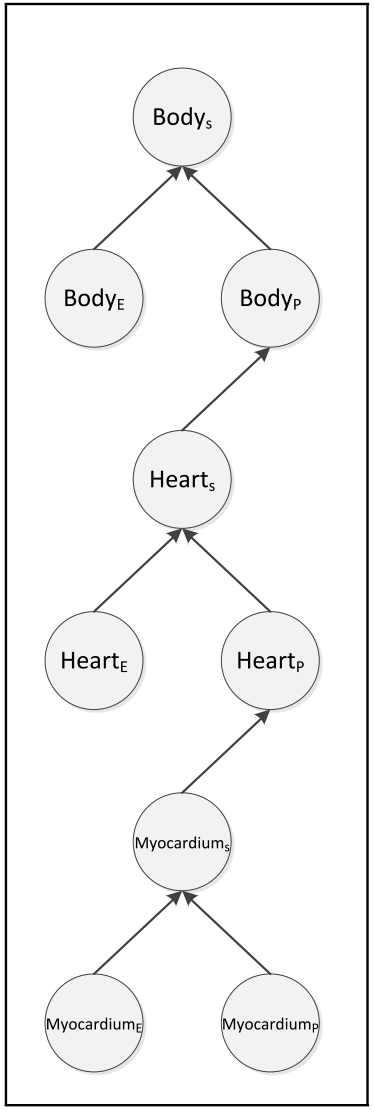

Figure: Additional SEP triples

SEP Triples

- $Myocarditis \equiv$
  $Inflammation \sqcap$
  $\exists has\_locus.Myocardium_S$
- $Carditis \equiv$
  $Inflammation \sqcap \exists has\_locus.Heart_S$
- $\models Myocardium_S \sqsubseteq Heart_S$ (subsumption)
- $\models Myocarditis \sqsubseteq$
  $Inflammation \sqcap \exists has\_locus.Heart_S$
- $\models Myocarditis \sqsubseteq Carditis$

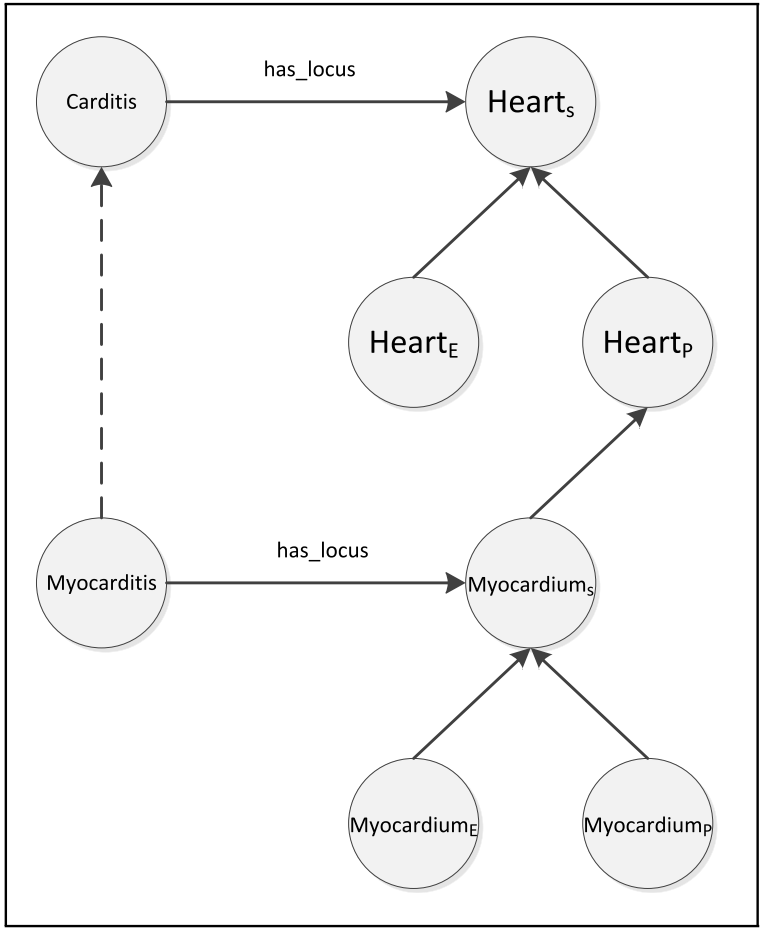

Figure: Entailment Given Part-Whole Relationship

SEP Triples

- $Pancarditis \equiv$
  $Inflammation \sqcap \exists has\_locus.Heart_E$
- $Myocarditis \equiv$
  $Inflammation \sqcap \exists has\_locus.Myocarditis_S$
- $\nvDash Myocardium_S \sqsubseteq Heart_E$
- $\nvDash Myocarditis \sqsubseteq Pancarditis$

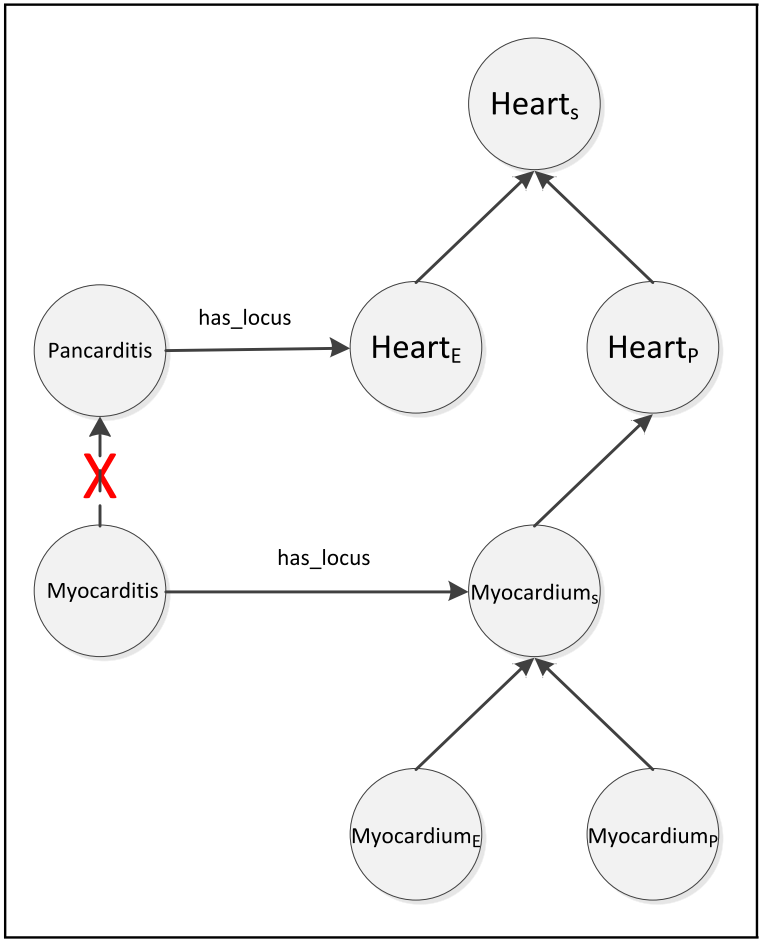

Figure: Entailment Given Part-Whole Relationship

Approaches

1. (A) Original SEP Triples Approach (just given)
2. (A1) SEP fully defined using *part_of*
3. (A2) A1 with removal of SEP triple classes
4. (A3) Variation on A2: still uses disjunctions, introduces *proper_part_of*
5. (A4) Variation on A3: eliminates disjunctions, still uses *proper_part_of*, introduces property path
6. (A5) Variation on A4: still uses *proper_part_of*, introduces *has_locus_entire*, introduces a different property path

Conventions

- SNOMED's set of classes C to be partitioned into:

  $C_n \cup C_S \cup C_E \cup C_P$
- $C_S \cup C_E \cup C_P$ are specific to (human) anatomy.
- X_S for class names in C_S, X_E for class names in C_E, and X_P for class names in C_P.
- We assume that in any occurrence of X_S, X_E, or X_P in an axiom, 'X' refers to the same term, e.g., *Heart*.

Adaptation of the SEP triples, Approach **(A1)**

- $SNOMED \cup$
  $\{X_S \equiv X_E \sqcup \exists part\_of.X_E \mid X_S \in C_S, X_E \in C_E\} \cup$
  $\{X_P \equiv \exists part\_of.X_E \mid X_P \in C_P\}$
- *part_of* is transitive and reflexive
- Adapted Triples for Myocardium and Heart:
  1. $Myocardium_S \equiv Myocardium_E \sqcup \exists part\_of.Myocardium_E$
  2. $Myocardium_P \equiv \exists part\_of.Myocardium_E$
  3. $Myocardium_S \sqsubseteq Heart_P$ (Connecting triples axiom)
  4. $Heart_S \equiv Heart_E \sqcup \exists part\_of.Heart_E$
  5. $Heart_P \equiv \exists part\_of.Heart_E$
- Yields:
  - $\models Myocardium_E \sqcup \exists part\_of.Myocardium_E \sqsubseteq \exists part\_of.Heart_E$ (1, 3, 5)
  - $\models Myocarditis \sqsubseteq Inflammation \sqcap \exists has\_locus.Heart_S$ (3, 5, 4)
  - $\models Myocarditis \sqsubseteq Carditis$ (by def of *Carditis*)

Alternative Approach **(A2)**

- Based on A1 and takes away SEP triple classes:
  1. Remove all axioms of the form $X_E \sqsubseteq X_S$ and $X_P \sqsubseteq X_S$.
  2. Replace all connecting axioms of the form $X_S \sqsubseteq Y_P$ (where X and Y are different) with $X \sqsubseteq \exists part\_of.Y$.
  3. Replace every occurrence of X_S of a class name in C_S with $X \sqcup \exists part\_of.X$ and every occurrence of X_E of a class name in C_E with X.
- **Applying 2**: $Myocardium \sqsubseteq \exists part\_of.Heart$ (Conn. Axiom)
- **Applying 3:**
  - $Myocarditis \equiv Inflammation \sqcap \exists has\_locus.(Myocardium \sqcup \exists part\_of.Myocardium)$
  - $Carditis \equiv Inflammation \sqcap \exists has\_locus.(Heart \sqcup \exists part\_of.Heart)$
  - $Pancarditis \equiv Inflammation \sqcap \exists has\_locus.Heart$
- $\models \exists part\_of.Myocardium \sqsubseteq \exists part\_of.Heart$ (trans.)
- $\models Myocarditis \sqsubseteq Inflammation \sqcap \exists has\_locus.(Heart \sqcup \exists part\_of.Heart)$ (previous, OR Intr)
- $\models Myocarditis \sqsubseteq Carditis$ (by def of *Carditis*)

Alternative Approach **(A3)**

1. Remove all axioms of the form $X_E \sqsubseteq X_S$ and $X_P \sqsubseteq X_S$.
2. Replace all connecting axioms of the form $X_S \sqsubseteq Y_P$ (where X and Y are different) with $X \sqsubseteq \exists proper\_part\_of.Y$.
3. Replace every occurrence of X_S of a class name in C_S with $X \sqcup \exists part\_of.X$ and every occurrence of X_E in a class name in C_E with X.
4. Add $proper\_part\_of \sqsubseteq part\_of$.

A3 differs from A2 in that in (2) *proper_part_of* replaces *part_of* (for the connecting axiom), and also (4) is an additional step in A3. These are required because parthood between anatomical entities is defined by the *proper_part_of* property.

Alternative Approach **(A4)**

Alternative Approach 4 (A4) applies the *proper_part_of* property, repeats Step (1) and (2) from A3, and includes the following steps:

3. Replace every occurrence of X_S of a class name in C_S with $\exists part\_of.X$ and every occurrence of X_E of a class name in C_E with X.
4. Add $proper\_part\_of \sqsubseteq part\_of$.
5. Add $part\_of \circ proper\_part\_of \sqsubseteq proper\_part\_of$. (left identity)

- A4 differs from A3 because for (3) $part\_of.X$ replaces $X \sqcup part\_of.X$, and step (5) introduces a left identity axiom, allowing us to infer:
- $\models \exists part\_of.Myocardium \sqsubseteq \exists proper\_part\_of.Heart$
- $\models Myocarditis \sqsubseteq \exists has\_locus.\exists proper\_part\_of.Heart$
- **# 2**: $Myocardium \sqsubseteq \exists proper\_part\_of.Heart$ (Conn. Axiom)
- **# 3**: $Carditis \equiv Inflammation \sqcap \exists has\_locus.\exists part\_of.Heart$

  $Myocarditis \equiv Inflammation \sqcap \exists has\_locus.\exists part\_of.Myocardium$

Alternative Approach **(A5)**

Alternative Approach 5 (A5) introduces the *has_locus_entire* property, a subproperty of *has_locus*, which expresses when a finding is located in some X_E class. A5 repeats Step (1) and (2) from A3 also, and includes the following steps:

1. Replace every occurrence of X_S of a class name in C_S with $\exists part\_of.X$ and every occurrence of $\exists has\_locus.X_E$ with $\exists has\_locus\_entire.X$.
2. Add $proper\_part\_of \sqsubseteq part\_of$.
3. Add $has\_locus \circ part\_of \sqsubseteq has\_locus$.

- **# 1**: $Myocarditis \equiv Inflammation \sqcap \exists has\_locus.Myocardium$
  $Carditis \equiv Inflammation \sqcap \exists has\_locus.Heart$
  $Pancarditis \equiv Inflammation \sqcap \exists has\_locus\_entire.Heart$

Conclusions

- We introduced 3 major methods for representing part-whole relationships, by applying: (1) transitive properties (2) disjunctions and (3) SEP triples.
- We introduced the logic underlying the current approach in SNOMED CT, and the logic underlying five alternative approaches.
- The approach used in SNOMED CT currently, A, applies (3), which is within ALC expressivity. A1 applies both (2) and (3), and A2 and A3 corresponds to just (2), because SEP triple classes are removed for both.
- Due to disjunctions the first three alternative approaches are beyond EL++ expressivity.
  A4 and A5 apply (1) only, therefore fall within EL++.
- A / A1-A5 Difference Propositional vs. Relational Representation of Parthood.
- User Navigation Issues of a Rendering Issue
- Prelim Performance Testing / Future Work

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