MedInfo 2007 Workshop:
MedSemWeb 2007

What Semantics Do We Need for A Semantic Web for Medicine?

How much formality do we need ?

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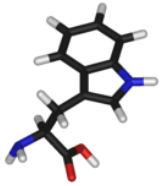
Example

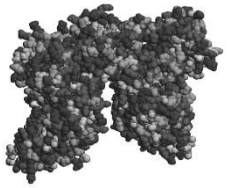
- Using Semantic Web standards (OWL-DL)
- Using Biomedical Ontology standards (OBO)
- Terminological Inference

Classes

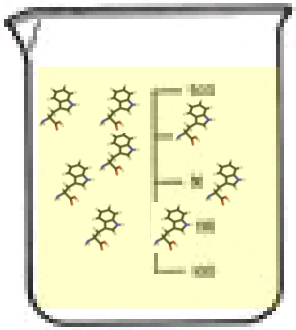
- Amino Acid



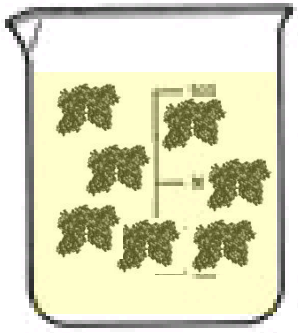
- Protein



- Aminoaciduria



- Proteinuria



Relations (OBO RO)

- *hasPart* / *partOf*

  (parthood in a broad sense):

  relates continuants
- *hasLocation* / *locationOf*

  relates continuants or occurrents with continuants
- transitive, reflexive, antisymmetric

Description Logic $\mathcal{EL}^+$

- Subsumption $\sqsubseteq$
- Equivalence $\equiv$
- Existential quantification $\exists$
- Conjunction $\sqcap$
- transitive roles

Axioms

$$Protein \sqsubseteq \exists hasPart.AminoAcid$$

$$Aminoaciduria \equiv Disorder \sqcap \exists hasLocation.(Body \sqcap \exists hasPart.(PortionOfUrine \sqcap \exists hasPart.AminoAcid))$$

$$Proteinuria \equiv Disorder \sqcap \exists hasLocation.(Body \sqcap \exists hasPart.(PortionOfUrine \sqcap \exists hasPart.Protein))$$

Inference

Proteinuria ⊑ *Aminoaciduria*

(since Proteins have Amino Acids as parts, and partOf is transitive)

- Is this error due to formal underspecification ?
- Is *hasPart* not always transitive?

Formal correctness
but ontological sloppyness

AminoAcid: hidden ambiguity:

- AminoAcidSingleMolecule
- AminoAcidResidue
- AminoAcidSingleMoleculeCollection
  - AminoAcidSingleMoleculeCollectionLowConc
  - AminoAcidSingleMoleculeCollectionHighConc

Corrected Axioms

$$Aminoaciduria \equiv Disorder \sqcap \exists hasLocation.(Body \sqcap \exists hasPart.(PortionOfUrine \sqcap \exists hasPart.AminoAcidSingleMoleculeCollectionHighConc))$$

$$Proteinuria \equiv Disorder \sqcap \exists hasLocation.(Body \sqcap \exists hasPart.(PortionOfUrine \sqcap \exists hasPart.ProteinMoleculeCollectionHighConc))$$

Two sides of the same coin

Formal Correctness

assures consistency

Ontological Correctness

assures adequacy

Conclusion

- Even little formality must be rooted in a correct ontological foundation to prevent unintended models with inadequate inferences
- If we do not know **exactly** what we are formalizing we cannot rely on machine reasoning. In this case we should give preference to informal, thesaurus-like knowledge representations