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An Ontological Analysis of Reference in Health Record Statements

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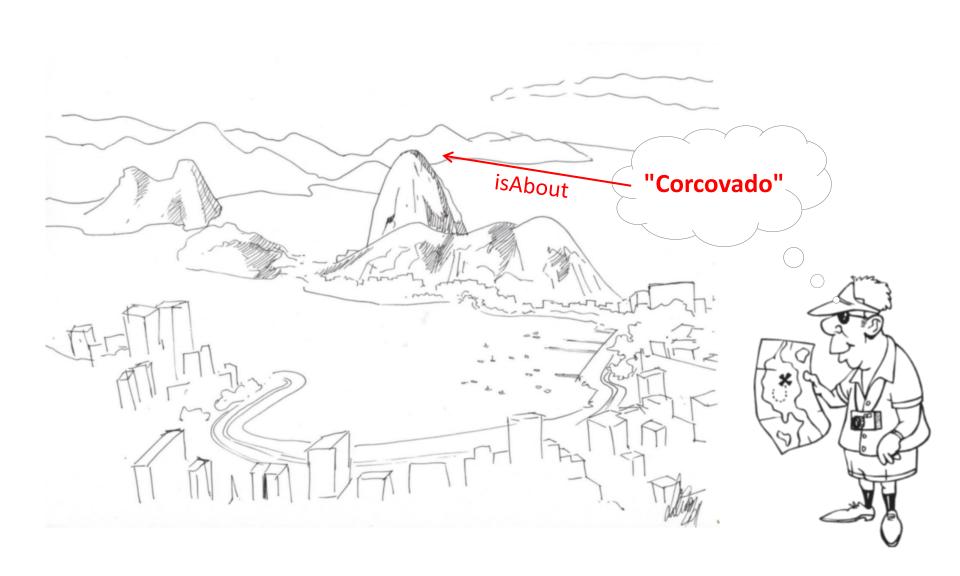
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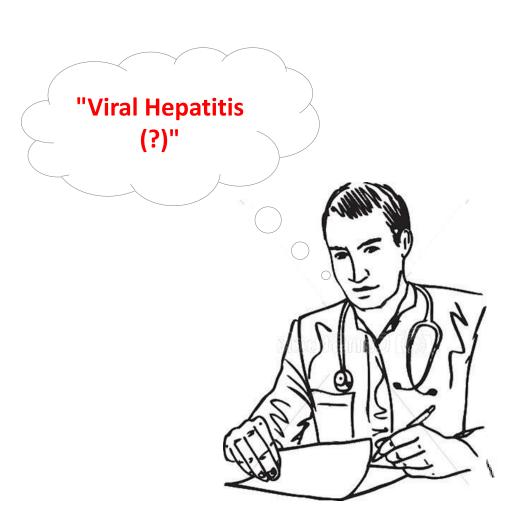


Correct reference?



Correct reference?





- "patient with **possible viral hepatitis**" (has alcoholic hepatitis)
- "patient scheduled for heart transplant" (dies before operation)
- "planned pregnancy"

 (unfortunately never gets pregnant)
- "omeprazol given to prevent **gastric ulcer**" (works, therefore patient won't get an ulcer)
- "patient drinks socially" (in fact a heavy drinker)
- "patient reports **severe back pain**" (patient simulates)
- "patient denied **hemodialysis**" (survives without hemodialysis)
- "father died from myocardial infarction"

 (died from ruptured aneurysm, son did not remember)

Nonreferring
expressions
in health
records

Adjectival modifiers in diagnostic statements

- "It is unlikely that the patient has hepatitis B"
- "It is confirmed that the patient has hepatitis B"
- "It is excluded that the patient has hepatitis B"

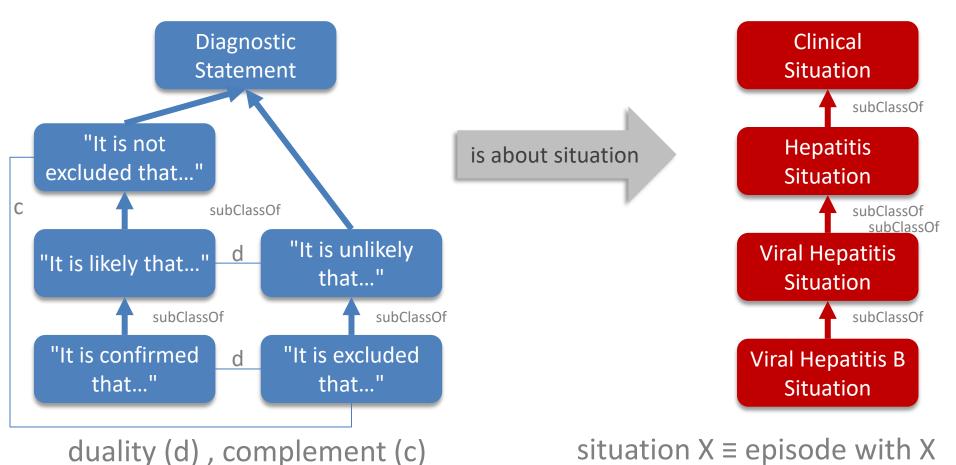


Goal

- Develop appropriate OWL-DL patterns that allow for expressing reference with different (qualitative) gradings of certainty
- Create a gold standard of examples of commonly agreed plausible inferences
- Validate the ontology patterns by comparing machine inferences to gold standard using DL reasoner (HermiT)

Example

Diagnostic statement: "The diagnosis of the condition X is confirmed / likely / not excluded / unlikely / excluded."



Plausible inferences

| | | Bei | ng sai | d to ha | ve h | epatit | is (H) | vira | al hep | atitis (v | vH) / | ' viral | hepati | tis B | (vHB | 3) is |
|---------------|-------------|-----------|--------|---------|--------|--------|--------|--------------|--------|-----------|----------|---------|--------|----------|------|-------|
| Precondition: | | confirmed | | | likely | | | not excluded | | | unlikely | | | excluded | | |
| Entailment: | Entailment: | | vH | vHB | Н | vH | vHB | Н | vH | vHB | Н | vH | vHB | Н | vH | vHB |
| | Н | X | X | X | | | | | | | | | | | | |
| confirmed | vH | | X | X | | | | | | | | | | | | |
| | vHB | | | X | | | | | | | | | | | | |
| likely | Н | X | X | X | X | X | X | | | | | | | | | |
| | vH | | X | X | | X | X | | | | | | | | | |
| | vHB | | | X | | | X | | | | | | | | | |
| | Н | X | X | X | X | X | X | X | X | X | | | | | | |
| not excluded | vH | | X | X | | X | X | | X | X | | | | | | |
| | vHB | | | X | | | X | | | X | | | | | | |
| | Н | | | | | | | | | | X | | | X | | |
| unlikely | vH | | | | | | | | | | X | X | | X | X | |
| | vHB | | | | | | | | | | X | X | X | X | X | X |
| | Н | | | | | | | | | | | | | X | | |
| excluded | vH | | | | | | | | | | | | | X | X | |
| | vHB | | | | | | | | | | | | | X | X | X |

Five OWL patterns

- "Existential" (using OWL existential restrictions)
- "Universal" (using OWL universal restrictions)
- "Punning" (using the same OWL entities as classes and individuals)
- "Two-Level" (introducing universals as A-Box inhabitants)
- "Query" (expressing reference as SPARQL queries on OWL models)

Five OWL patterns

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OWL pattern "Existential"

```
BeingSaidToHaveXisConfirmed equivalentTo
        DiagnosticStatement and (hasCertainty only isConfirmed)
                             and (isAboutSituation some Xsituation)
BeingSaidToHaveXisLikely equivalentTo
        DiagnosticStatement and (hasCertainty only isLikely)
                             and (isAboutSituation some Xsituation)
BeingSaidToHaveXisNotExcluded equivalentTo
        DiagnosticStatement and (hasCertainty only IsNotExcluded)
                             and (isAboutSituation some Xsituation)
BeingSaidToHaveXisUnlikely equivalentTo
        DiagnosticStatement and
                 (hasCertainty only isLikely) and
                 (isAboutSituation some (ClinicalSituation and not Xsituation))
BeingSaidToHaveXisExcluded equivalentTo
        DiagnosticStatement and
                 (hasCertainty only isConfirmed) and
                 (isAboutSituation some (ClinicalSituation and not Xsituation))
```

Entailments of "Existential"

| - | | Bei | ng sai | d to ha | ve h | epatit | tis (H) | / vira | al hep | atitis (| vH) / | viral | hepati | tis B | (vHB | 3) is |
|---------------|-----|-----------|--------|---------|--------|--------|---------|--------------|--------|----------|----------|-------|--------|----------|------|-------|
| Precondition: | | confirmed | | | likely | | | not excluded | | | unlikely | | | excluded | | |
| Entailment: | | Н | vH | vHB | Н | vH | vHB | Н | vH | vHB | Н | vH | vHB | Н | vH | vHB |
| | Н | X | X | X | | | | | | | | | | | | |
| confirmed | vH | | X | X | | | | | | | | | | | | |
| | vHB | | | X | | | | | | | | | | | | |
| likely | Н | X | X | X | X | X | X | | | | | • | | • | | |
| | vH | | X | X | | X | X | | | | | | | | | |
| | vHB | | | X | | | X | | | | | | | | | |
| | Н | X | X | X | X | X | X | X | X | X | | | | | | |
| not excluded | vH | | X | X | | X | X | | X | X | | | | | | |
| | vHB | | | X | | | X | | | X | | | | | | |
| | Н | | | | | | | | | | X | | | X | | |
| unlikely | vH | | | | | | | | | | X | X | | X | X | |
| | vHB | | | | | | | | | | X | X | X | X | X | X |
| | Н | | | | | | | | | | | | | X | | |
| excluded | vH | | | | | | | | | | | | | X | X | |
| | vHB | | | | | | | | | | | | | X | X | X |

Problem with "Existential" pattern

- Existential import: for each statement about X there is
 - some instance of XSituation , or
 - some instance of 'Clinical Situation and not XSituation'
- Conflicting statements would produce logical contradictions
- Solution: universal quantifier ("only") instead of existential quantifier ("some")
- "isAboutSituation only SituationX" → in case there a reference exists, then it is of the type SituationX

OWL pattern "Universal"

```
BeingSaidToHaveXisConfirmed equivalentTo
        DiagnosticStatement and (hasCertainty only isConfirmed)
                             and (isAboutSituation only Xsituation)
BeingSaidToHaveXisLikely equivalentTo
        DiagnosticStatement and (hasCertainty only isLikely)
                             and (isAboutSituation only Xsituation)
BeingSaidToHaveXisNotExcluded equivalentTo
        DiagnosticStatement and (hasCertainty only IsNotExcluded)
                             and (isAboutSituation only Xsituation)
BeingSaidToHaveXisUnlikely equivalentTo
        DiagnosticStatement and
                  (hasCertainty only isLikely) and
                  (isAboutSituation only (ClinicalSituation and not Xsituation))
BeingSaidToHaveXisExcluded equivalentTo
        DiagnosticStatement and
                  (hasCertainty only isConfirmed) and
                  (isAboutSituation only (ClinicalSituation and not Xsituation))
```

Entailments of "Universal"

| | | Bei | ng sai | d to ha | ve h | epatit | tis (H) | vira | al hep | atitis (| vH) / | viral | hepati | tis B | (vHB | 3) is |
|---------------|-------------|-----------|--------|---------|--------|--------|---------|--------------|--------|----------|----------|-------|--------|----------|------|-------|
| Precondition: | | confirmed | | | likely | | | not excluded | | | unlikely | | | excluded | | |
| Entailment: | Entailment: | | vH | vHB | Н | vH | vHB | Н | vH | vHB | Н | vH | vHB | Н | vH | vHB |
| | Н | X | X | X | | | | | | | | | | | | |
| confirmed | vH | | X | X | | | | | | | | | | | | |
| | vHB | | | X | | | | | | | | | | | | |
| likely | Н | X | X | X | X | X | X | | · | | | | | | | |
| | vH | | X | X | | X | X | | | | | | | | | |
| | vHB | | | X | | | X | | | | | | | | | |
| | Н | X | X | X | X | X | X | X | X | X | | | | | | |
| not excluded | vH | | X | X | | X | X | | X | X | | | | | | |
| | vHB | | | X | | | X | | | X | | | | | | |
| | Н | | | | | • | | | | | X | | | X | | |
| unlikely | vH | | | | | | | | | | X | X | | X | X | |
| • | vHB | | | | | | | | | | X | X | X | X | X | X |
| | Н | | | | | | | | | | | | | X | | |
| excluded | vH | | | | | | | | | | | | | X | X | |
| | vHB | | | | | | | | | | | | | X | X | X |

Problem with "Universal" pattern

"isAboutSituation only SituationX" ≡ not (isAboutSituation some (not SituationX)

- contradicts ground axiom of IAO
- Known issues with this kind of statements in DL

 unexpected entailments (probably not relevant here
 (due to strict range restriction of the isAboutSituation
 relation), but doubts persist
- Expression is still "about" something, viz. the type SituationXType

OWL pattern "Two level"

Type subClassOf owl:Thing

Particular subClassOf owl:Thing

Type subClassOf hasInstance some Particular

X EquivalentTo isInstanceOf value x_Type

x_Type type Type and **hasInstance** only X

X subclassOf isInstanceOf value x_Type

every member of the class X is an instance of the type \mathbf{x} _Type. The type \mathbf{x} _Type has only instances that are members of the class X

isAboutSituation o isSubtypeOf subPropertyOf isAboutSituation

OWL pattern "Two Level"

```
BeingSaidToHaveXisConfirmed equivalentTo
        DiagnosticStatement and (hasCertainty only isConfirmed)
                             and (isAboutSituation value XsituationType)
BeingSaidToHaveXisLikely equivalentTo
        DiagnosticStatement and (hasCertainty only isLikely)
                             and (isAboutSituation value XsituationType)
BeingSaidToHaveXisNotExcluded equivalentTo
        DiagnosticStatement and (hasCertainty only IsNotExcluded)
                             and (isAboutSituation value XsituationType)
BeingSaidToHaveXisUnlikely equivalentTo
        DiagnosticStatement and
                 (hasCertainty only isUnlikely) and
                 (isAboutSituation value XsituationType)
BeingSaidToHaveXisExcluded equivalentTo
        DiagnosticStatement and
```

(hasCertainty only isExcluded) and

(isAboutSituation value XsituationType)

Entailments of "Two Level"

| 1:4: | | | u to 110 | 110 11 | сраш | is (H) | VIII | и пер | auus (| VII <i>) 1</i> | vii ai | перац | 113 D | (VIID | <i>y</i> 18 | |
|---------------|---|--|---|--------|---------------|--------|------|---|---|----------------|--|-------|-------|----------|-------------|--|
| Precondition: | | confirmed | | | likely | | | not excluded | | | unlikely | | | excluded | | |
| Entailment: | | vH | vHB | Н | vH | vHB | Н | vH | vHB | Н | vH | vHB | Н | vH | vHB | |
| Н | X | X | X | | | | | | | | | | | | | |
| vH | | X | X | | | | | | | | | | | | | |
| vHB | | | X | | | | | | | | | | | | | |
| Н | X | X | X | X | X | X | | | | | | | | | | |
| vH | | X | X | | X | X | | | | | | | | | | |
| vHB | | | X | | | X | | | | | | | | | | |
| Н | X | X | X | X | X | X | X | X | X | | _ | | | | | |
| vH | | X | X | | X | X | | X | X | | | | | | | |
| vHB | | | X | | | X | | | X | | | | | | | |
| Н | | | | | | | | | | X | X | X | X | X | X | |
| vH | | | | | | | | | | | X | X | | X | X | |
| vHB | | | | | | | | | | | | X | | | X | |
| Н | | | | | | | | | | | | - | X | X | X | |
| vH | | | | | | | | | \ | | | | | X | X | |
| vHB | | | | | | | | | | | | | | | X | |
| | | . — | | | · | · —— | | · | | | · | . —— | | · —— | | |
| | | | | | | | | | | | | | | | | |
| | vH vHB H vH vHB H vHB H vH vHB H vHB | vH vHB H x vH vHB H x vH vHB H vH vHB H vH vHB H vH vHB H vH vHB | H X X X VHB X YHB YHB YHB YHB | H | H | H | H | H x x x x x x x x x x x x x x x x x x x | H x x x x x x x x x x x x x x x x x x x | H | H x x x x x vH vHB x x x x x x x x x x x x x x x x x x x | H | H | H | H | |

Problem with "Two Level" pattern

- No inversions with negative statements
- The sentence "Hepatitis B excluded" is still a statement about the type Hepatitis, whereas it does not claim the existence of an instance of the type hepatitis
- Possible solution: combine "Universal" with "Two Level", but removing the axiom

isAboutSituation o isSubtypeOf subPropertyOf isAboutSituation

• Is there any practical usefulness of maintaining parallel, isomorphic hierarchies of OWL classes in the T-box and OWL types in the A-box?

Open issues / Outlook

- Related work from philosophy, e.g. dummy entities like "subfactuals" (Meinong)
- Relax assumptions of ontological realism
- Relation to alternative approaches of representing the content of health records, e.g. Referent tracking (Ceusters)
- Relation to other logics (higher-order, modal logics)
- Relation to models of probability
- Empirical assessment of computational behaviour (theoretically, OWL DL is NExpTime-complete)

Thank you



Download sample ontologies from https://biotop.googlecode.com/svn/trunk/apps/FOIS2014/