Spatial Representation and Reasoning in Biomedical Ontologies: How a formal spatial theory can help increase clarity and reasoning capabilities

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Outline

- Examples of containment assertions in the FMA and GALEN
- Different types of individual-level containment relations
- Different types of relations among classes
- How formal theories can be used to improve biomedical ontologies

I. Examples of containment assertions in the FMA and GALEN

Anatomical Classes

The FMA and GALEN are comprised of assertions about relations between anatomical classes, not assertions about relations between anatomical individuals.

Example:

Heart contained_in *Middle Mediastinal Space*

NOT:

Tom's heart contained_in Tom's middle mediastinal space

Note: Tom's heart is an instance of the class *Heart*.

Examples of the FMA's contained_in Assertions

Right Ovary contained_in *Abdominopelvic Cavity*

Urinary Bladder contained_in *Cavity of Female Pelvis*
Urinary Bladder contained_in *Cavity of Male Pelvis*
Blood contained_in *Cavity of Cardiac Chamber*

Urinary Bladder contained_in *Pelvic Cavity*
Uterus contained_in *Cavity of Female Pelvis*
Prostate contained_in *Cavity of Male Pelvis*
Heart contained_in *Middle Mediastinal Space*
Blood contained_in *Lumen of Cardiovascular System*

Examples of GALEN's isContainedIn Assertions

Ovarian Artery isContainedIn *Pelvic Cavity*
Uterus isContainedIn *Pelvic Cavity*

Uterus isContainedIn *Female Pelvic Cavity*
Mediastinum isContainedIn *Thoracic Space*
Larynx isContainedIn *Neck*

Pleural Cavity isContainedIn *Pleural Membrane*
Tooth isContainedIn *Tooth Socket*

Two ways of distinguishing the use of containment relations in these assertions

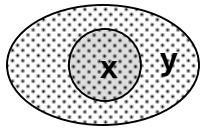
- Differences in the way in the relations between anatomical classes (*Heart*, *Middle Mediastinal Space*, etc) depend on relations between their individual instances.
- Differences in the underlying spatial configurations holding between the appropriate pairs of individuals.

Differences in the Dependence of Class-Level Relations on Individual-Level Relations

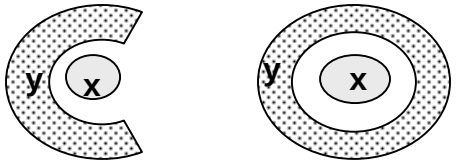
- *Heart* contained_in *Middle Mediastinal Space*
  All hearts are contained in a middle mediastinal space AND every middle mediastinal spaces contains a heart.
- *Right Ovary* contained_in *Abdominopelvic Cavity*
  All right ovaries are contained in an abdominiopelvic cavity, but not all abdominopelvic cavities contain ovaries.
- *Urinary Bladder* contained_in *Cavity of Male Pelvis*
  Not all urinary bladders are contained in the cavity of a male pelvis, but every cavity of a male pelvis contains a urinary bladder.

Differences in the underlying spatial configurations

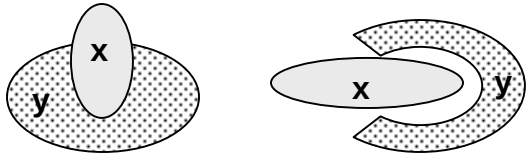
- *Uterus* isContainedIn *Pelvic Cavity* (region containment)



- *Pleural Cavity* isContainedIn *Pleural Membrane* (surrounds)



- *Tooth* isContainedIn *Tooth Socket* (partial-containment)



II. Different types of individual-level containment relations

Formal Tools

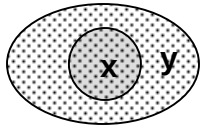
Four different types of containment relations among individuals are distinguished in a first-order theory which includes:

- A *parthood* relation (P) where Pxy means x is part of y (e.g. my heart is part of my body).
- A *region* function (r) which maps each individual to the spatial region it exactly occupies (e.g. r(my heart) is the heart-shaped region exactly occupied by my heart).
- A *convex hull* function (ch) which maps each individual to the smallest convex region which extends over it (e.g. the convex hull of my pleural membrane extends over both the membrane and the space inside it; the convex hull of the wall of my stomach extends over both the wall of my stomach and my stomach cavity).

Region Containment

x is *region-contained* in y if x's region is part of y's region:

$CNT\text{-}IN_r xy =: Pr(x)r(y)$.



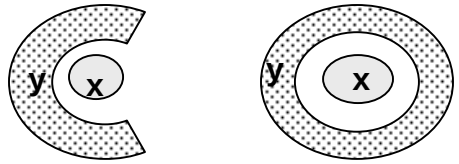
Examples: my uterus is region-contained in my pelvic cavity, my heart is region-contained in my middle mediastinal space, my larynx is region-contained in my neck.

NOTE: parthood entails region containment but not vice versa.

Surround Containment

x is *surrounded* by y if x's region is part of y's convex hull and x's region does not overlap y's region:

CNT-IN$_s$xy =: Pr(x)ch(y) & ~Or(x)r(y)

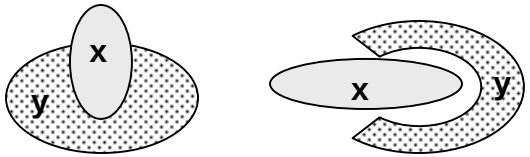


Examples: My pleural space is surrounded by my pleural membrane and the cavity of my stomach is surrounded by the wall of my stomach.

Partial Containment

x is *partially contained* in y if x's region overlaps y's convex hull:

P-CNT-INxy =: Or(x)ch(y).



Examples: My tooth is partially contained in its socket; my esophagus is partially contained in my thoracic cavity.

Basic Logical Properties of these Containment Relations

	reflexive	irreflexive	symmetric	asymmetric	transitive
Region Containmt	yes	no	no	no	yes
Surround Containmt	no	yes	no	no	no
Partial Containmt	yes	no	no	no	no

Interaction with the parthood relation

- Parthood entails both region containment and partial containment.

- Parthood is *incompatible* with surround containment. Example: if x is surrounded by the wall of my stomach (in the sense of CNT-IN_s), then x cannot be part of the wall of my stomach.

Compositional Reasoning using Parthood and Containment Relations

	y is a proper part of z
CNT-IN$_r$xy	CNT-IN$_r$xz
CNT-IN$_s$xy	
P-CNT-INxy	P-CNT-INxz

	x is a proper part of y
CNT-IN$_r$yz	CNT-IN$_r$xz
CNT-IN$_s$yz	CNT-IN$_s$xz
P-CNT-INyz	

Examples: Given that my heart is r-contained in my middle mediastinal space and my left atrium is a proper part of my heart, it follows that my left atrium is also r-contained in my middle mediastinal space.

But given that my tooth is partially contained in its socket and my tooth's crown is part of my tooth, it does NOT follow that the crown is partially contained in the tooth socket.

III. Different types of relations among classes

Three types of spatial relations among classes

Let *R* be any individual-level spatial relation.

- R^1(A, B) =: $\forall$x (Inst(x, A) $\rightarrow$ $\exists$y(Inst(y, B) & *R*xy))

(every A is stands in relation *R* to some B)

- R^2(A, B) =: $\forall$y (Inst(y, B) $\rightarrow$ $\exists$x(Inst(x, A) & *R*xy))

(for each B there is some A that stands in relation *R* to it)

- R^{12}(A, B) =: R^1(A, B) & R^2(A, B)

(every A stands in relation *R* to some B and for each B there is some A that stands in relation *R* to it)

Examples Using Region Containment

- CNT-IN$_r^1$ (A, B) =: Every A is region-contained in some B.

Example: Every uterus is region-contained in some pelvic cavity.
CNT-IN$_r^1$ (*Uterus*, *Pelvic Cavity*)

- CNT-IN$_r^2$ (A, B): Every B region-contains some A.

Example: Every male pelvic cavity region-contains some urinary bladder.
CNT-IN$_r^2$ (*Urinary Bladder*, *Male Pelvic Cavity*)

- CNT-IN$_r^{12}$ (A, B) =: Every A is region-contained in some B and every B region-contains some A.

Example: Every brain is region-contained in some cranial cavity and every cranial cavity region-contains a brain.CNT-IN$_r^{12}$ (*Brain*, *Cranial Cavity*)

Properties of relations among individuals vs. properties of relations among classes

Among Individuals	**Among Classes**		
R is...	R^1 must also be...?	R^2 must also be...?	R^{12} must also be...?
Reflexive	Yes	Yes	Yes
Irreflexive	No	No	No
Symmetric	No	No	Yes
Asymmetric	No	No	No
Antisymmetric	No	No	No
Transitive	Yes	Yes	Yes

Interaction between class-level spatial relations and the Is_a class subsumption relation

	Is_a(C, A)	Is_a(A, C)	Is_a(C, B)	Is_a(B, C)
R^1(A, B)	R^1(C, B)			R^1(A, C)
R^2(A, B)		R^2(C, B)	R^2(A, C)	
R^{12}(A, B)	R^1(C, B)	R^2(C, B)	R^2(A, C)	R^1(A, C)

Examples:

1.every lung is region-contained in some thoracic cavity

2. Is_a(*Right Lung*, *Lung*),

imply:

3. every right lung is region-contained in some thoracic cavity.

Examples

1. every ovarian artery is region contained in some pelvic cavity
2. Is_a(*Male Pelvic Cavity*, *Pelvic Cavity*)

do NOT imply:

3. every ovarian artery is region contained in some male pelvic cavity.

IV. How formal theories can be used to improve biomedical ontologies

Clarifying the intended meanings of the assertions in biomedical ontologies

- It is obviously important for the intended interpretation of relations used in biomedical ontologies to be as clear as possible for both users and curators.
- Clarity is especially important for projects that involve information from multiple sources (e.g. multiple biomedical ontologies)– different sources may have different intended meanings for the same spatial terms.

The FMA's contained_in vs. GALEN's isContainedIn

The FMA's contained_in:

- the underlying individual-level relation is a restricted version of the region containment relation which holds only between material contents (e.g. my heart) and immaterial containers (e.g. my middle mediastinal space);
- the class-level contained_in is used in some contexts as an R^1 relation:

Right Ovary contained_in *Abdominopelvic Cavity*

in other contexts as an R^2 relation:

Urinary Bladder contained_in *Cavity of Male Pelvis*

and in other contexts as an R^{12} relation:

Heart contained_in *Middle Mediastinal Space*

The FMA's contained_in vs. GALEN's isContainedIn

GALEN's isContainedIn:

- the underlying individual-level relation is in some contexts a version of the region containment relation:

Uterus isContainedIn *Pelvic Cavity*

Larynx isContainedIn *Neck*

in other cases a version of the surrounds relation:

Pleural Cavity isContainedIn *Pleural Membrane*

and in others a version of the partial containment relation:

Tooth isContainedIn *Tooth Socket*

- the class-level isContainedIn is generally used as an R^1 or R^{12} relation.

Detecting Invalid Automated Reasoning in Biomedical Ontologies

- The precise logical properties of the FMA's contained_in and GALEN's isContainedIn can be determined only when these relations are satisfactorily treated in a formal theory. But we can already see that the following are problematic:
- GALEN implements transitivity reasoning on isContainedIn. If this relation is supposed to include *partial containment*, then it should not be transitive.
- GALEN includes the assertion:

Male Pelvic Cavity Contains *Ovarian Artery*

which seems to be inferred from

Pelvic Cavity Contains *Ovarian Artery*

Male Pelvic Cavity Is_a *Pelvic Cavity*.

This is an example of invalid reasoning over class-level spatial relations and the Is_a relation stemming from a confusion between R^1 and R^2 relations.

Extending Automated Reasoning in Biomedical Ontologies

- Once the logical properties of the class-level spatial relations are clearly worked out, more extensive automated reasoning could be added to the ontologies.
- For example, the FMA includes the assertions:

Right Ovary contained_in *Abdominopelvic Cavity*

Left Ovary contained_in *Abdominopelvic Cavity*

but no containment information for *Ovary*. This could be generated automatically from the assertions above with appropriate reasoning schemas.

Conclusions

- Spatial relation terms do not yet have clear formal semantics in existing biomedical ontologies.
- Possibilities for expanding the inference capabilities of biomedical ontologies are limited, in part because relations with different logical properties are not explicitly distinguished.
- Containment relations are relatively simple. Much more work will be required to clear up more complex spatial relations used in biomedical ontologies (e.g. adjacency, continuity, etc).