Analysis of propagation along transitive roles: Formalisation of the GALEN experience with Medical Ontologies

Alan Rector
Department of Computer Science, University of Manchester
Email: rector@cs.man.ac.uk

1 Introduction

Anatomical, functional and causal relations are fundamental to knowledge representation in bioscience and medicine. Their represenation requires dealing with the interaction of partitive and other transitive roles with each other and with other non-transitive roles. Padgham and Labrix [5] identify a number of schemas of which the two most commonly occurring in biomedical applications are illustrated by the examples below.

Schema 1 A "disease located in a part" is a kind of "disease located in the whole", *e.g.* a "Fracture located in the shaft of the femur" is a kind of "Fracture located in the Femur", and conversely a "Femur with a fracture located in its shaft" is a kind of "Femur which is the location of a fracture".

Schema 2 A "layer of a division" is a part of the corresponding "layer of the whole", *e.g.* "the skin of the hand" is a part of "the skin of the upper extremity"

Both schemas are essential to the representation of anatomy and its relation to disease, causation, and function. Good engineering solutions suitable for large (>100,000 concept) ontologies are needed urgently if description logics, and formalisms based on them such as DAML+OIL/OWL[1]. are to play a significant role in Bio- and Medical Informatics.

Schema 1, which is the subject of this paper, is particularly important. We shall label it the "*propagates_via* " schema — *e.g. has_location propagates_via is_part_of*. It is often described as "inheritance across transitive roles" — *e.g.* "location is inherited across partonomy".

[1] www.daml.org/2001/03/daml+oil-index

One solution is to allow axioms with the description logic itself of the form:

$$has\_location \circ is\_part\_of \rightarrow has\_location \tag{1}$$

which is an example a general schema and its equivalent converse of the form:

$$\mathsf{R} \circ \mathsf{S} \rightarrow \mathsf{R} \qquad \mathsf{S}^{-} \circ \mathsf{R}^{-} \rightarrow \mathsf{R}^{-} \tag{2}$$

A complete and decidable algorithm for the general schema is one of the unsolved problems in description logics [1](Horrocks, Personal communications). A recent report suggests that a more general problem is undecidable [10].

Nonetheless this is the approach taken by the two largest efforts at clinical ontologies, OpenGALEN [8][6] and SNOMED-CT/RT[9], albeit using incomplete classifiers, and has resulted in large ontologies in use for significant applications. *Open*Galen allows knowledge engineers to to use multiple instances of the axiom schema. When multiple axioms effect a single role, the effect is that a graph or path of roles implies that role. We shall shall here call such a graph a "*role-path*". Note that the definition of the *role-path* is restricted to implication rather than equivalence. We do not want to collapse the path into the single role, but only to prove subsumption by it. For example, we need to say that "Murmur caused by flow restriction caused by encrustation located on cusp which is part of the valve which is part of the ventricle which is part of the heart" is a kind of "Disorder located in the heart"; therefore the "*role-path*" should include

$$caused\_by^{*} \circ has\_location \circ is\_part\_of^{*} \rightarrow has\_location \tag{3}$$

Producing efficient representations which have the effect of such "*role-paths*" is a major barrier to converting the existing large ontologies in GALEN and SNOMED-CT/RT, and applications using them, to modern complete description logics. It is also the major barrier to being able to advocate expressing the largest authoritative model of anatomy — the Digital Anatomist Foundational Model of Anatomy [4]— formally in a description logic.

One approach to understanding the nature of the problem is to analyse the potential "*role-paths*" which could be generated by combinations of such axioms. A second is to analyse the "*role-paths*" which have, empirically, been generated by knowledge engineers' in practical development to address otherwise difficult problems[7]. Since the knowledge engineers tend to think of the axioms individualy, analysing the global consequences of the combination of axioms used in the form of "*role-paths*" has also proved a valuable aid in rationalising the ontological schemas.

In this paper we:

- Outline the derivation of a general form for the "*role-path*" of a role in a knowledge base containing multiple *propagates_via* axioms. The result is a restricted regular expression of roles; further restrictions which might lead to tractable algorithms are also suggested.

- Display example *role-paths* that have resulted from practical use in *Open*Galen of axioms following the *propagates_via* schema and show how this analysis has helped to rationalise the use of such axioms in *Open*Galen.
- Discuss the relation of this approach to Shulz and Hahn's recently suggested alternative which involves rewriting the knowledge base in terms of triples — the whole, the part, and the whole or the parts — which they term "SEP triples"[2].

It is hoped that this work may motivate others to further efforts to provide better practical engineering solutions to this urgent problem.

2 Outline of derivation of the expression for the *role-path* for R: the function $rp(\mathsf{R})$

2.1 Preliminary results

An important feature of the *propagates_via* schema is that in the context of a R *propagates_via* S, S always behaves as if it were transitive since

$$\text{If } \mathsf{R} \circ \mathsf{S} \rightarrow \mathsf{R} \text{ then } \mathsf{R} \circ \mathsf{S} \circ \mathsf{S} \rightarrow \mathsf{R} \circ \mathsf{S} \rightarrow \mathsf{R} \tag{4}$$

Generalising by induction, it follows that

$$\text{If } \mathsf{R} \circ \mathsf{S} \rightarrow \mathsf{R} \text{ then } \mathsf{R} \circ \mathsf{S}^* \rightarrow \mathsf{R} \tag{5}$$

Hence it is preferable, from the point of view of understandability and software engineering, but not logically necessary, to restrict the *propagates_via* axioms to the case were S is transitive.

It follows immediately from the above that *propagates_via* self is equivalent to transitivity.

$$\mathsf{R} \circ \mathsf{R} \rightarrow \mathsf{R} \text{ if and only if } \mathsf{R} \text{ is transitive} \tag{6}$$

Hence any instance of the schema of the form R *propagates_via* R is redundant in a description logic which supports transitive roles, and it is bad practice, in the light of what follows, to include axioms of the form R*propagates_via* S where $\mathsf{R} \rightarrow \mathsf{S}$.

By contrast, whether or not R is transitive maters.

$$\text{If } \mathsf{R} \text{ is transitive and } \mathsf{R} \circ \mathsf{S} \rightarrow \mathsf{R} \text{ then } \mathsf{R} \circ (\mathsf{R} \vee \mathsf{S})^* \rightarrow \mathsf{R} \tag{7}$$

Proof is by induction based on the observation that if R is transitive, then any chain of Rs before an S can be collapsed to a single R which can be combined with the S to produce yet another R, etc.

For this reason it is useful to define the function $\tau(\mathsf{R},\mathsf{S})$ where R is a role and S can be either a role or a *role-path*:

$$\text{if } \mathsf{R} \text{ is transitive then } \tau(\mathsf{R},\mathsf{S}) = (\mathsf{R} \vee \mathsf{S}) \text{ otherwise } \tau(\mathsf{R},\mathsf{S}) = \mathsf{S} \tag{8}$$

With respect to the role hierarchy, if R *propagates_via* S, then it also propagates by any role subsumed by S.

$$\text{If } \mathsf{S}_1 \rightarrow \mathsf{S} \textit{ and } \mathsf{R} \circ \mathsf{S} \rightarrow \mathsf{R} \text{ then } \mathsf{R} \circ \mathsf{S}_1 \rightarrow \mathsf{R} \circ \mathsf{S} \rightarrow \mathsf{R} \tag{9}$$

Therefore, any axioms of the form R *propagates_via* S_1, where $\mathsf{S}_1 \rightarrow \mathsf{S}$ are redundant and irrelevant in constructing the *role-path* for R.

Unfortunately, the same is not true where R *subsumes* R_1, *i.e.* If $\mathsf{R}_1 \rightarrow \mathsf{R}$ and $\mathsf{R}_1 \circ \mathsf{S} \rightarrow \mathsf{R}_1$ Then, even if it is already known that $\mathsf{R} \circ \mathsf{S} \rightarrow \mathsf{R}$, the new axiom cannot be derived from the old. However, such axioms do affect the *role-path* of R because it follows by simple substitution that:

$$\text{If } \mathsf{R}_1 \rightarrow \mathsf{R} \text{ and } \mathsf{R}_1 \circ \mathsf{S} \rightarrow \mathsf{R}_1 \text{ then } \mathsf{R}_1 \circ \mathsf{S} \rightarrow \mathsf{R} \tag{10}$$

2.2 Expression for the *role-path* of R: the function $rp(\mathsf{R})$

The basic structure is most easily seen in the simplified case where there is at most one *propagates_via* axiom for each role and at most one for its converse.

If $\mathsf{R} \circ \mathsf{S}_{\mathsf{d}_1} \rightarrow \mathsf{R}$, $\mathsf{S}_{\mathsf{d}_1} \circ \mathsf{S}_{\mathsf{d}_2} \rightarrow \mathsf{S}_{\mathsf{d}_1}, \ldots, \mathsf{S}_{\mathsf{d}_{n-1}} \circ \mathsf{S}_{\mathsf{d}_n} \rightarrow \mathsf{S}_{\mathsf{d}_{n-1}}$ and none of R or the $\mathsf{S}_{\mathsf{d}_i}$ is transitive, then

$$\mathsf{R} \circ (\mathsf{S}_{\mathsf{d}_1} \circ (\mathsf{S}_{\mathsf{d}_2} \circ \ldots \circ \mathsf{S}^*_{\mathsf{d}_n})^*)^* \rightarrow \mathsf{R} \tag{11}$$

Or in other words, a *role-path* for R consists of an R followed by zero or more $\mathsf{S}_{\mathsf{d}_1}s$, which if present can be followed by zero or more $\mathsf{S}_{\mathsf{d}_2}s$, etc. Proof is by induction on n using the definition in (2). If any roles may be transitive, then the function τ defined in (8) above must be used:

$$\mathsf{R} \circ (\tau(\mathsf{R},\mathsf{S}_{\mathsf{d}_1}) \circ (\tau(\mathsf{S}_{\mathsf{d}_1},\mathsf{S}_{\mathsf{d}_2}) \circ \ldots \circ \tau(\mathsf{S}_{\mathsf{d}_{n-1}},\mathsf{S}_{\mathsf{d}_n})^*)^*)^* \rightarrow \mathsf{R} \tag{12}$$

Analogously, adapting the results above to cover coverses using (2),

if $\mathsf{R}^- \circ \mathsf{S}_{\mathsf{c}_1} \rightarrow \mathsf{R}^-$, $\mathsf{S}^-_{\mathsf{c}_1} \circ \mathsf{S}_{\mathsf{c}_2} \rightarrow \mathsf{S}^-_{\mathsf{c}_1}, \ldots, \mathsf{S}^-_{\mathsf{c}_{n-1}} \circ \mathsf{S}_{\mathsf{c}_n} \rightarrow \mathsf{S}^-_{\mathsf{c}_{n-1}}$ then

$$((\tau(\mathsf{S}^-_{\mathsf{c}_{n-1}},\mathsf{S}^-_{\mathsf{c}_n})^* \circ \ldots \circ \tau(\mathsf{S}^-_{\mathsf{c}_1},\mathsf{S}^-_{\mathsf{c}_2}))^* \circ \tau(\mathsf{R}^-,\mathsf{S}^-_{\mathsf{c}_1}))^* \circ \mathsf{R}^- \rightarrow \mathsf{R}^- \tag{13}$$

These results may be combined in a single recursive formula using "I" for the identity role.

$$\begin{aligned} rpSingle(\mathsf{I}) &= \mathsf{I} \\ rpSingle(\mathsf{R}) &= \tau(\mathsf{R}, rpSingle(Sc^-))^* \circ \mathsf{R} \circ \tau(\mathsf{R}, rpSingle(Sd))^* \end{aligned} \tag{14}$$

To procede to the general case requires only noting that both multiple *propagates_via* axioms and *propagates_via* axioms regarding subsumed roles add additional disjuncts.

If $\mathsf{R} \circ \mathsf{S}_{\mathsf{d}_1} \to \mathsf{R},\ \mathsf{R} \circ \mathsf{S}_{\mathsf{d}_2} \to \mathsf{R},\ \ldots\ ,\ \mathsf{R} \circ \mathsf{S}_{\mathsf{d}_n} \to \mathsf{R}$ then

$$\mathsf{R} \circ (\mathsf{S}_{\mathsf{d}_1} \vee \mathsf{S}_{\mathsf{d}_2} \vee \ldots \vee \mathsf{S}_{\mathsf{d}_n}) \to \mathsf{R} \tag{15}$$

If for each role $\mathsf{R}_{\mathsf{s}_i}$ and $\mathsf{S}_{\mathsf{ds}_i}$, $\mathsf{R}_{\mathsf{s}_i} \to \mathsf{R}$, and $\mathsf{R}_{\mathsf{s}_i} \circ \mathsf{S}_{\mathsf{ds}_i} \to \mathsf{R}_{\mathsf{s}_i}$ then

$$(\mathsf{R}_{\mathsf{s}_1} \circ \mathsf{S}_{\mathsf{ds}_1}) \vee (\mathsf{R}_{\mathsf{s}_2} \circ \mathsf{S}_{\mathsf{ds}_2}) \vee \ldots \vee (\mathsf{R}_{\mathsf{s}_n} \circ \mathsf{S}_{\mathsf{ds}_n}) \to \mathsf{R} \tag{16}$$

These results can be combined with (14) and generalised by induction to give the definition for the function for the *role-path* of R, $rp(\mathsf{R})$, in terms of three mutually recursive functions.

Given a role R and roles $\mathsf{S}_{\mathsf{d}_i}$, $\mathsf{S}_{\mathsf{c}_j}$, $\mathsf{R}_{\mathsf{s}_k}$ none equal to the identity role, I, and covering all cases satisfying
$\mathsf{R} \circ \mathsf{S}_{\mathsf{d}_i} \to \mathsf{R}$, $\mathsf{R}^- \circ \mathsf{S}_{\mathsf{c}_j} \to \mathsf{R}^-$,
$\mathsf{R}_{\mathsf{s}_k} \to \mathsf{R} \wedge \exists \mathsf{S}((\mathsf{R}_{\mathsf{s}_k} \circ \mathsf{S} \to \mathsf{R}_{\mathsf{s}_k}) \vee (\mathsf{R}^-_{\mathsf{s}_k} \circ \mathsf{S} \to \mathsf{R}^-_{\mathsf{s}_k}))$
then

$$\begin{aligned} rpself(\mathsf{R}) &= \bigvee_{all\ i,\ all\ j} (\ \tau(\mathsf{R}, rp(\mathsf{S}^-_{\mathsf{c}_j}))^* \circ \mathsf{R} \circ \tau(\mathsf{R}, rp(\mathsf{S}_{\mathsf{d}_i}))^*\ ) && (17)\\ rpSubs(\mathsf{R}) &= \bigvee_{all\ k} rpSelf(\mathsf{R}_{\mathsf{s}_k}) && (18)\\ rp(\mathsf{R}) &= rpSelf(\mathsf{R}) \vee rpSubs(\mathsf{R}) && (19) \end{aligned}$$

Note that $rpSubs(\mathsf{R})$ need recur only on *rpSelf* rather than *rp* because any roles subsumed by any $\mathsf{R}_{\mathsf{s}_k}$ are also subsumed by R. Note that these functions can give rise to correct but redundant disjuncts which are straightforward to simplify.

3 Experience in GALEN

Using this analysis, the function $rp(\mathsf{R})$ has been implemented to display role paths derived from multiple *propagates_via* axioms in a knowledge base. It has been used to identify errors, cycles and overcomplicated *role-paths* in the GALEN ontology. For example, the *role-path* for the key *hasLocation* role which links pathology to anatomy includees two roles *isPhysicalPartOf* which is self explanatory, and *isTransitiveMultipleOf* which is the relation of a 'rash' to the 'spots' which constitute it, etc:

$$\begin{aligned} &rp(hasLocation) = && (19)\\ &\qquad hasLocation \circ\ ((hasLocation \vee isTransitiveMultipleOf)^* \vee \\ &\qquad\qquad (hasLocation \vee isPhysicalPartOf)^* \end{aligned}$$

This analysis suggests that *isPhysicalPartOf* and *isTransitiveMultipleOf* both be made children of a single parent even though they seem intuitively different.

Other roles have paths which we have not been able to simplify. For example "Collagen *is a constituent of* interstitial tissue which *makes up in part* the dermis which *is a layer of* the skin".

$$\begin{aligned} &rp(isConstituentOf) = \\ &\quad isConstituentOf \circ \\ &\quad\; (makesUpInPart \circ (makesUpInPart \vee isLayerOrDivisionOf)^*)^* \end{aligned} \tag{20}$$

4 Comparison with SEP Triples

Schulz and Hahn [2] have recently proposed the mechanism of SEP triples to deal with the same engineering problem but using a slightly different semantics from *propagates_via* axioms. In this approach, rather than embedding the reasoning within the logic itself, the burden is placed on the transformation between the users' notions and the description logic representation. In the description logic, three separate concepts are created for each for each user notion: a) "the thing as a whole", b) "the parts of the thing", and c) "the thing or its parts". If using $\mathcal{ALC}$, as do Schulz and Hahn, each of the three concepts must be asserted explicitly as a primitive. Using a description logic with transitive roles such as SHIQ [3], the SEP triples can be rewritten as:

Thing
(some part_of Thing)
(or Thing (some part_of Thing))

These are precisely the constructs required for the most natural approximate translation of the *role-paths* described here into SHIQ, *e.g.* to approximate the effect of $\mathsf{R} \circ \mathsf{S} \rightarrow \mathsf{R}$, where R corresponds to *hasLocation* and S corresponds to *isPartOf* rewrite as:

$$\begin{aligned} (\textit{some}\ \mathsf{R}\ Thing) &\Rightarrow (\textit{some}\ \mathsf{R}\ (\textit{or}\ Thing\ (\textit{some}\ \mathsf{S}\ Thing))) \\ (\textit{all}\ \mathsf{R}\ Thing) &\Rightarrow (\textit{all}\ \mathsf{R}\ (\textit{or}\ Thing\ (\textit{some}\ \mathsf{S}\ Thing))) \\ (\textit{some}\ \mathsf{R}^-\ Subj) &\Rightarrow (\textit{or}\ (\textit{some}\ \mathsf{R}^-\ Subj)(\textit{some}\ \mathsf{S}^-\ (\textit{some}\ \mathsf{R}^-\ Subj))) \\ (\textit{all}\ \mathsf{R}^-\ Subj) &\Rightarrow (\textit{all}\ \mathsf{R}^-\ Subj) \\ &\ (\textit{implies}\ (\textit{some}\ \mathsf{S}^-\ \mathsf{Top})\ (\textit{all}\ \mathsf{R}^-\ Subj)) \end{aligned}$$

However, more complicated chains or forms involving disjunctions arising from transitive roles as in (7) cannot be expressed without role disjunction, although they can be approximated by consructing "pseudo disjuncts" or "disjunctive orbits" — *i.e.* asserted common parents in the role hiearchy.

The semantics of SEP triples has the further advantage that they can be used selectively. OpenGALEN requires separate roles for "function of the whole" from "function of the whole or its parts", for example, to represent the difference between " 'Pumping' *is a function of* 'the Heart as a whole' " and " 'Conduction' *is a function of* 'The conduction system of the heart or any of its parts' " SEP triples transfer this distinction from the roles to the concepts, which seems more natural. On the other hand, if used strictly within $\mathcal{ALC}$ as do Schulz and Hahn, there are no inverse roles, and inverse roles are essential for many applications — *e.g.* to be able to represent that the "hands have subdivisions fingers" as well as that "fingers are subdivisions of hands". If rewritten to SHIQ, the presence of inverses can give rise to scaling problems under circumstances which are not yet fully explored.

5 Discussion

Our goal is a sound tractable engineering solution for part-whole and related reasoning in biomedicine. With respect to the *propagates_via* alternative, the key question is whether efficient classifier algorithms supporting the restricted regular expression form the *role-path* of R given in (19) are feasible, possibly with further restrictions. The two most obvious further restrictions are a) that no *role-path* contain cycles and b) that no *role-path* contain both a role and its inverse. Analysis of GALEN's experience suggests that neither restriction would unduly limit expressiveness and that both could be easily enforced. Expressions containing cycles are probably undecidable, and even simple expressions of the form $\mathsf{R} \circ \mathsf{R}^{-}$ are often ambiguous, *e.g.* is *"fracture located in a bone which is the location of trauma"* tautologous (because *trauma* subsumes *fracture*) or should it be interpreted as meaning *"other trauma"*, including possibly another fracture — a notion not expressible in SHIQ.

In general, the post fix expressions in (17) due to direct *propagates_via* axioms form a tree fanning out from the role R with a modest branching factor at each step. By contrast the prefix expressions resulting from converse *propagates_via* axioms form a tree fanning in often giving a large initial branching factor which is potentially expensive computationally. This is confirmed for GRAIL's network based classifier but the effect for tableaux based classifiers is unknown.

Schulz and Hahn's SEP triples appear to provide an alternative. Using SEP triples requires significant rewriting of the original knowledge base and has subtly different, at times advantageous, semantics. It is a reasonable hypothesis that any expression required in a 'sensible' ontology can be achieved by such rewriting combined with judicious use of the role hierarchy. Initial attempts to reformulate the GALEN ontology in this way are promising formally but performance problems remain. Whether new algorithms can improve the performance of either or both approaches — or whether yet further alternatives can be suggested to achieve a similar engineering result — is an urgent question anyone wanting to use description logics for practical applications in medicine and bioscience.

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