The Role of Ontologies for an Effective and Unambiguous Dissemination of Clinical Guidelines.

Domenico M. Pisanelli, Aldo Gangemi, Geri Steve

CNR, Istituto di Tecnologie Biomediche
Viale Marx 15, 00137 Roma, Italy

{pisanelli, gangemi, steve} @itbm.rm.cnr.it
http://saussure.irmkant.rm.cnr.it

Abstract. Guidelines for clinical practice are being introduced in an extensive way in more and more different fields of medicine They have the potential to improve the quality and cost-efficiency of care in a complex health care delivery environment. Computerization may increase the effectiveness of both the information retrieval of guidelines and the management of guideline-based care. The scenario is evolving from stand-alone workstations to telematics applications that enable guidelines development and dissemination. However, such a knowledge sharing requires the definition of formal models for guidelines representation. The models should have a clear semantics in order to avoid ambiguities. The role of ontologies is that of making explicit the conceptualizations behind a model. In this paper we present our library of generic and domain ontologies and point out its role for integrating existing guideline models and defining standard representations. In particular, we stress the distinction –often collapsed within existing guideline models– between the conceptualization of actual procedures, the conceptualization of planning, and the conceptualization behind the diagrammatic representation of plans.

1. Introduction

Guidelines for clinical practice are being introduced in an extensive way in more and more different fields of medicine [1][2]. They have the goal of indicating the most appropriate decisional and procedural behavior optimizing health outcomes, costs and clinical decisions.

Guidelines can be expressed in a textual way as recommendations or in a more formal and rigid way as protocols or flow diagrams. In different contexts they can be either a loose indication for a preferred set of choices or they can be considered a normative set of rules.

Clinical practice guidelines are seen as a tool for improving the quality and cost-efficiency of care in an increasingly complex health care delivery environment. It has been proved that adherence to plans may reduce cost of care up to 25% [3].

However the overwhelming number of guidelines available makes it difficult to select the right one. Just to give an idea of the figures, it is reported that there are 855 different guidelines for British GPs ranging from a single page to small booklets of more than 15 pages [4].

Computerization may increase the effectiveness of both the information retrieval of guidelines and the delivery of guideline-based care. In an optimal scenario they are integrated with the information systems operational at the point of care. The full potentialities of computerized systems can be exploited in such an environment where different processes are executed in parallel on several patients. In this context such systems must be able to retrieve the updated situation of every patient, as well as to give an overall report on the ward, freeing the physicians to concentrate more on clinical decisions. Keeping track of the parallel activities performed, they should avoid unnecessary duplication of tasks and prevent possible omissions.

Several research projects deal with the computer representation and implementation of guidelines. In the next paragraph we review some of the most relevant ones. The scenario is evolving from stand-alone workstations to telematics applications that - utilizing e.g. the Internet - not only support the use of guidelines, but also enable their development and dissemination.

Such a knowledge sharing requires the definition of formal models for guidelines representation. The models should have a clear semantics in order to avoid ambiguities. The role of ontologies is that of making explicit the conceptualizations behind a model. In particular an ontology contains the formal description of the entities to which a model makes a commitment and of the relations holding among the entities.

In this paper we present our library of ontologies and point out its role for integrating existing guideline models and defining standard representations.

2. Applications of Clinical Guidelines

Many efforts have been devoted in the last few years in realizing computerized tools for guidelines management (see for example: Vissers and co-workers [5] and Ertle and co-workers [6]).

The European PRESTIGE project (Guidelines for healthcare: faster implementation of health care standards) is mainly dedicated to supplying information and modelling technology to guidelines in the context of the IV Framework Programme (1994-1999) [7]. PRESTIGE aims to produce telematic applications for a faster implementation of new standards of quality in clinical practice in Europe. It is therefore directed more towards the clinical part, neglecting the organisational and administrative components.

The PRO*forma* knowledge representation language and associated software tools are designed to support the dissemination of medical knowledge by means of electronic publishing [8]. It is also a method for specifying clinical guidelines and protocols in a form which can be executed by a computer in order to support the management of medical procedures and clinical decision making.

Web based tools for supporting clinical care are becoming increasingly popular (see for example [9-11]). The system SMART allows users to access its database - storing information on patients following a guideline-based care - by means of the Internet [12].

COLLATE is a WWW-enabled workgroup environment designed to support dispersed collaborative groups throughout the complete guideline development cycle [13]. COLLATE supports document management, collaborative authoring and editing over the Internet, and provides methods for accessing and browsing multimedia databases of systematic literature reviews and guideline documents, and can provide links to external applications such as on-line bibliographic database packages.

Recently, increasing attention has been paid to formal model of guidelines and protocols. For instance, EON is a computational model of treatment protocols [14]. The EON framework consists of three different components: the domain knowledge base of medical concepts that will specify the application and temporal-abstraction knowledge necessary for reasoning, one or more applications that use problem-solving methods formulated as collections of CORBA objects and the Tzolkin subsystem that performs temporal abstraction and temporal pattern matching.

This model was the basis for implementing the protocol-based decision-support module for the already existing T-HELPER system [15]. This system was originally aimed at supporting AIDS care, but, because its architecture was domain independent, it was possible to substitute the AIDS knowledge base with another one regarding breast cancer. The Protégé-II tool was employed to create an ontology of concepts related to the management of breast cancer [16]. Such a reuse of knowledge allowed the Stanford researchers to implement their prototype system in less than one week.

The successful experience of the EON model demonstrates that knowledge-based systems cannot be constructed in isolation from development in the softwareengineering community. Their approach, oriented to modularity and to the definition of ontologies, facilitated knowledge re-use.

3. An Excerpt of our Ontology Library

Ontologies not only make knowledge re-use easier, they are also the foundation of standardization efforts since they make explicit the conceptualizations behind a terminology or a model. The actual demand is not for a unique conceptualization, but for an unambiguous communication of complex and detailed concepts (possibly expressed in different languages), leaving each user free to make explicit his/her conceptualization.

We developed ONIONS, a methodology for integrating domain terminologies by exploiting a library of generic theories [17]. By means of this methodology we realized the library of ontologies ON9.2 (available in Ontolingua at: http://saussure.irmkant.rm.cnr.it), including both general and domain specific ontologies [18]. The ontologies related to guidelines, which we will sketch out in the next paragraph, are part of this library.

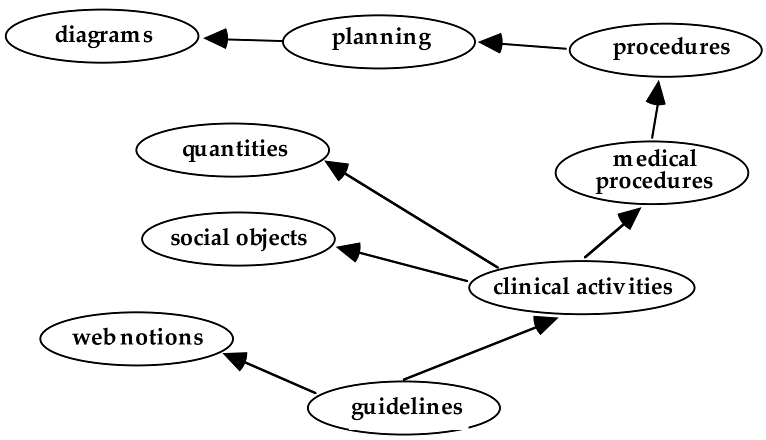

Fig. 1. An excerpt from the ontology library ON 9.2, showing the included theories nearest to the guidelines theory.

Figure 1 reports a fragment of the library architecture. Each oval represents an ontology, i.e. a module which embodies the formal definition of related concepts and relationships.

Arrows denote inclusion between ontologies: the specific one includes the generic one (e.g. "medical procedures" includes "procedures").

"Clinical activities" is one ontology included by "guidelines", meaning that its concepts and relationships are used by the former one. It consists of the main types of entities involved in clinical activities, including: "patient", "patient group", "health care operator", "medical device", "health care structure", "medical sign" and "health condition", and some actor-like relations that divide into: 1) treatment relations, i.e. "treats" (between a "health care operator" and a "health condition"), and other composed relations (i.e. chaining "treats" with other relations) that account for different senses of "treats", e.g. "treatment-device" (between a "medical device" and a "health condition"); 2) diagnosis relations, i.e. "diagnoses" (between a "health care operator" and a "health condition"), and some composed relations; 3) care relations, i.e. "cares for" (holding between a "health care operator" and a "patient" or "patient group"), and some composed relations.

"Medical procedures" contains the definition of the main types of medical procedures, deriving from the integration of the UMLS Semantic Network [19] and the types defined by the Institute of Medicine. A medical procedure is a procedure having health-care activities as temporal parts. Five main kinds of medical procedures are defined: 1) screening and prevention procedures; 2) surgical procedures; 3) care of clinical conditions procedures; 4) diagnostic procedures; 5) laboratory procedures.

Procedures 1) to 3) share the need for some "bearer" patient or patient group, some pharmacologic resource, and some medical device; 4) are characterized by a patient or patient-group "target" and by their "diagnostic action" carried out on clinical conditions; 5) are characterized by the fact that they "analyze" some chemical or body substance, possibly assessing their effect. Relations such as: "bearer", "target", "analyzes", and "diagnostic action" are defined according to the generic ontology "actors", which describes the relations holding between the entities participating in processes and situations with a particular role (e.g. "performer", "embodier", "instrument", "goal", etc.).

To implement our ontology we adopted both Ontolingua [20], which is a very expressive language, and the Loom knowledge representation system [21] which supports automatic classification and semantic consistency check. Both languages allow HTML translation and browsing facilities. In particular the Ontosaurus [22], an interface to Loom through the CL-HTTP server, is appropriate for allowing collaborative development of ontologies [23].

What is peculiar to our library is its integration of medical domain and generic (domain-independent) ontologies. Examples of generic ontologies include: "mereology" or theory of parts, "topology" or theory of wholes and connexity, "morphology", or theory of form and congruence, "localization", "time", "actors", "planning", etc.

The relevance of generic theories to the development of ontologies is not always recognized. There are a number of significant experiences showing that an ontological analysis can profit from theories which are philosophically and linguistically grounded [24]. Our position is that generic theories are essential to the development of ontologies and to a rigorous conceptual integration of heterogeneous models [25].

For example, available formal models of guidelines make commitments to various entities and relations associated with guideline specification: plans, diagrammatic charting, information requests, actions, decisions, situations, tasks, etc.

These formal models do not sort out the entities by means of their ontological nature, but only on the basis of system design and efficiency issues.

For example, the Asbru model defines some guideline properties, but it splits them in two sets: one (plan status) includes "rejected", "ready", etc., another (plan state) includes "aborted", "completed", etc. [26] .

Understanding such properties within an ontological framework requires at least understanding the difference between a plan and a procedure: a plan is an abstract entity (a "script") which acts as the method of a procedure, which is a process actually occurring in the real world.

Such distinction can be drawn with the use of some generic theories that define a "plan" as a special kind of "abstract entity", a "procedure" as a special kind of "process", a "method" relation as a special kind of "actor" relation, and so on.

Once plan and procedure are kept disjoint in our ontology, one can easily infer why the two sets of Asbru properties actually draw that distinction: only a plan can be "rejected", and only a procedure can be "aborted".

If one disagrees with the plan/procedure distinction only has to dismiss the commitment to the generic theories mentioned, possibly providing other theories that support a different conceptualization.

4. The Ontology of Guidelines

In this paragraph we present the main features of the "guidelines" ontology and related concepts.

Guidelines are distinguished in "paper guidelines" and "web guidelines". Some common concepts - like "author" - pertain to both of them, whereas "URL" and "last-checked" are peculiar of the web guidelines.

They are also categorized in five different kinds, as defined in the Guideline Interchange Format standard (GLIF) [27]: "guideline for care of clinical condition", "screening and prevention", "diagnosis and prediagnosis management of patients", "indications for use of surgical procedures", "appropriate use of specific technologies and tests".

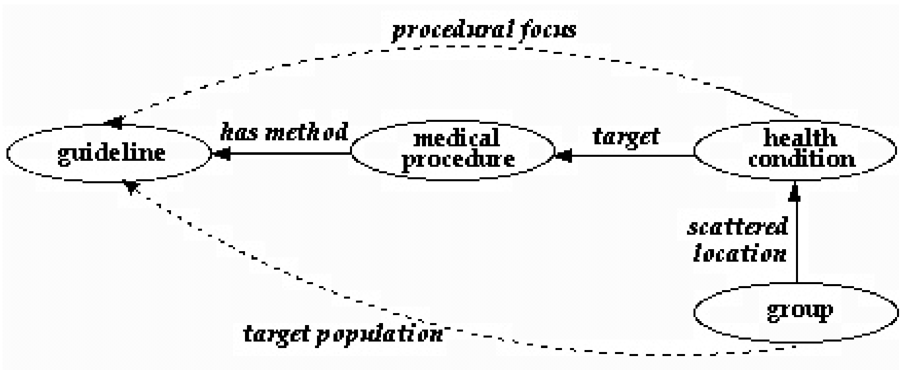

Fig. 2. The relations among basic concepts concerning guidelines (grey arrows stand for composite relations); concepts belong to different ontologies in the ontology library.

Such classification is furtherly specialized by us: for example a "guideline for care of clinical condition" may be a "therapy assessment", a "pharmacologic therapy" or a "disease management".

Figure 2 shows the relations among the basic concepts concerning guidelines. More specific concepts inherit these relations holding at general level. As an example, let us consider "diabetic patients" (concept more specific than "group", defined as any collection of individuals that carry a diabetic health condition). Such patients are the "scattered location" (this is a special location relation accounting for naïve localization holding between distributed object, defined in the generic ontology "localization") of "diabetes" (a "health condition"), which is the "target" of "diabetes therapy" (a "medical procedure") which "has method" a given guideline.

As far as the formal representation of guidelines is concerned, our ontology integrates some of the most relevant modeling efforts so far produced: notably PROforma [8], EON [14], Asbru [26] and GLIF [27]. It is also an evolution of a model previously defined in the context of the SMART system [12].

A "guideline" is a kind of "plan" which is a method of a "procedure", and it is represented by a "flowchart" (fig. 3).

The concept of "flowchart" pertains to the "diagrams" ontology. It is defined as a set of nodes and edges like an ordinary graph with some restrictions. Every flowchart has a first and a last node, only four kinds of nodes are allowed: single nodes, branching nodes, synch nodes and cycle nodes. Moreover the flowchart ontology allows for recursion, i.e. a node may be expanded into a flowchart.

This ontology accounts for the structural part of a guideline, but no semantics for actions is attached to it. The semantics for the actions involved pertains to the *planning* ontology, where simple nodes represent elementary actions and branching nodes enquiries and decisions. The recursion allowed in the flowchart domain, where a node of a flowchart may be expanded into a flowchart, is isomorph to the planning ontology, where an elementary action may be refined into a plan.

We believe that in our model it is appropriate to capture the distinction between the structural part of a guideline, represented by the flowchart, and its action semantics, represented by the plan. A third level is that of the procedure, i.e. what is actually performed.

Figure 4 reports an excerpt from the *planning* ontology (the representation language is Loom). Such an approach can put in evidence - at formal level - the equivalences among the various modeling approaches. We defined the *GL-mapping* ontology in order to account for such equivalences (figure 5).

Therefore this ontology integrates some of the most relevant work in the guideline modeling field. It is GLIF-compliant, i.e. each concept defined in GLIF is represented in it (e.g. the "synch" node after parallelization of activities). It takes into account the ProForma task ontology which categorizes tasks into: actions, enquiries and decisions and allows recursive definition of them (a plan is made of tasks, a task may be a plan).

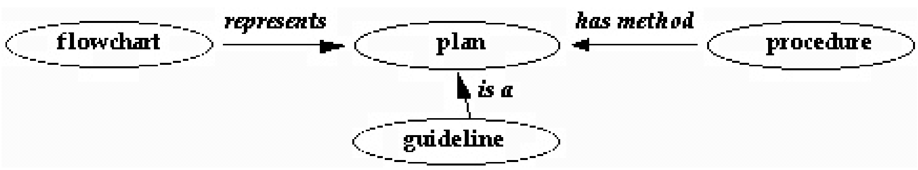

Fig. 3. The main concepts concerning a guideline. A guideline is a plan, and planning should be kept distinct from both diagrammatic representation and reference to actual procedures.

5. Conclusions

It has been proven that the introduction of guidelines can significantly decrease the costs of care and this make them a "hot topic" in the agenda of health care professionals.

Guidelines are mushrooming and computers can help in retrieving them and can give assistance during their execution. However such a widespread diffusion poses new problems, not only in terms of credibility and acceptability, but also concerning non-ambiguity in knowledge dissemination. Formal models with a clear semantics should be defined in order to represent guidelines and facilitate their diffusion.

The definition of ontologies - i.e. the formal description of the entities to which a model makes a commitment and of the relations holding among the entities - is the groundwork for making a standard model acceptable and sharable. An ontology library is not normative, but allows an inter-subjective, explicit and formal agreement on the semantics of the primitives of a model, by referring to more generic primitives (generic theories).

In this paper we presented our work in terms of the definition of an ontology of guidelines which is part of a larger ontology library containing both domain and generic theories. We believe that such an approach can facilitate the standardization process by allowing an explicit mapping in a formal ontology of the concepts represented in the heterogeneous models proposed so far.

References

1. Woolf SH. Practice Guidelines, a New Reality in Medicine: Impact on Patient Care. *Arch Intern Med,* 1993; 153: 2646-2655.
2. Grimshaw JM, Russell IT. Effect of Clinical Guidelines on Medical Practice: a Systematic Review of Rigorous Evaluations. *Lancet,* 1993; 342: 1317-1322.
3. Clayton PD, Hripcsak G. Decision support in healthcare. *Int. J. of Bio-Medical Computing,* 1995; 39: 59-66.
4. Hibble A., Kanka D., Penheon D., Pooles F: Guidelines in general practice: The new Tower of Babel? *British Medical Journal,* 1998; 317: 862-3.
5. Vissers MC, Hasman A, van der Linden CJ. Protocol Processing System (ProtoVIEW) to Support Residents at the Emergency Ward. *Proc. MIE 94,* 1994: 138-143.
6. Ertle AR, Campbell EM, Hersh WR. Automated Application of Clinical Practice Guidelines for Asthma Management. *Proc. AMIA 96,* 1996: 552-556.
7. Gordon CJ et al. Telematics for Clinical Guidelines: A Conceptual Modeling Approach. *Proc MIE 97,* 1997.

8. Fox J, Johns N, Rahmanzadeh A. Disseminating Medical Knowledge: The PROforma Approach *Artificial Intelligence in Medicine,* 1998; 14.
9. Barnes M, Barnett GO. An Architecture for a Distributed Guideline Server.*Proc. 19th SCAMC,* 1995: 233-237.
10. Cimino JJ, Socratous SA. Automated Guidelines Implemented via the World Wide Web.*Proc. 19th SCAMC,* 1995: 941.
11. Liem EB, Obeid JS, Shareck EP, Sato L, Greenes RA. Representation of Clinical Practice Guidelines through an Interactive World-Wide-Web Interface.*Proc. 19th SCAMC,* 1995.
12. Pisanelli DM, Consorti F, Merialdo P. SMART: A System Supporting Medical Activities in Real Time, *Proc. MIE 97,* 1997. .
13. http://antipodes.cec.fr/collate/co-home.nsf
14. Musen MA, Tu SW, Das AK, Shahar Y. EON: A component-based approach to automation of protocol-directed therapy. *JAMIA,* 1996; 3.
15. Musen MA, Carlson CW, Fagan LM et al. T-HELPER: Automated support for community-based clinical research. *Proc. 16th SCAMC,* 1992.
16. Gennari JH, Altman RB, Musen MA. Reuse with Protégé-II. Proceedings of the Symposium on Software Reuse, Seattle, 1995.
17. Steve G, Gangemi A, Pisanelli DM, "Integrating Medical Terminologies with ONIONS Methodology", in: *Info.Modelling and Knowledge Bases VIII,* Amsterdam, IOS Press 1998.
18. Pisanelli DM, Gangemi A, Steve G, "A Medical Ontology Library that integrates the UMLS Metathesaurus", *Proc. AI in Medicine,* 1999.
19. National Library of Medicine, *UMLS Knowledge Sources,* 1998 edition, available from the NLM, Bethesda, Maryland.
20. Gruber TR, "A translation approach to portable ontology specification", *Knowledge Acquisition,* 5 (2), 1993: 199-220.
21. MacGregor RM, "A Description Classifier for the Predicate Calculus" *Proceedings of AAAI 94, Conference,* 1994.
22. Swartout B, Patil R, Knight K, Russ T, "Toward Distributed Use of Large-Scale Ontologies", *Proceedings of Knowledge Acquisition Workshop,* Banff, participants edition, 1996.
23. Pisanelli DM, Gangemi A, Steve G, "WWW-available Conceptual Integration of Medical Terminologies", *Proc. AMIA 97,* 1997.
24. Guarino N, Carrara M, Giaretta P, An Ontology of Meta-Level Categories. *Proc. of KR94*. San Mateo, CA, Morgan Kaufmannn, 1994.
25. Pisanelli DM, Gangemi A, Steve G, "An Ontological Analysis of the UMLS Metathesaurus", *Proc. AMIA 98,* 1998.
26. Shahar Y, Miksch S, Johnson P. The Asgaard project, *Artificial Intelligence in Medicine,* 1998; 14.
27. Ohno-Machado L et al. The Guideline Interchange Format, *JAMIA,* 1998; 6.

```
(defcontext planning
  :theory (unrestricted-time diagrams meronymy))

(defconcept generic-plan
  :annotations ((DOCUMENTATION "A generic plan is a script that
contains the method for executing or performing a procedure or
a stage of a procedure."))
    :is-primitive (:and script
                   (:all method
                    (:or procedure procedures^stage)))
    :implies (:and (:some represented-by document)
               (:some has-component generic-plan)
               (:all component generic-plan)
               (:all serial-value serial-value-filler)
               (:all has-title symbolic-string)
               (:all has-precondition situation)
               (:all authored-by plan-author)
               (:all duration-value number)
               (:all has-description symbolic-string)
               (:all has-postcondition situation))
    :partitions $generic-plan$)

  (defconcept elementary-plan
  :annotations ((DOCUMENTATION "A plan that does not contain
compacted plans (ie, subplans represented by a simple node that
can be expanded into a flow-chart)."))
    :is-primitive
     (:and generic-plan
      (:all has-component Incoherent generic-plan)))

  (defconcept complex-plan
    :is (:and generic-plan
              (:at-least 2 has-component generic-plan)))

  (defconcept branching-plan
    :annotations ((DOCUMENTATION "A plan that subdivides in a
set of plans."))
    :is-primitive
     (:and elementary-plan
      (:at-least 2 plan-direct-predecessor))
    :implies (:and (:some represented-by fork-node)
                   (:some method planning-transition))
    :in-partition $generic-plan$)
```

```
(defconcept case-plan
   :annotations ((DOCUMENTATION "A plan branched to a set of
plans not executable in parallel."))
   :is-primitive
    (:and branching-plan
     (:at-least 2 plan-direct-predecessor
      (:and generic-plan
       (:at-least 1 has-precondition)
       (:all co-exist Incoherent generic-plan))))
   :implies (:some represented-by fork-node))
```

Fig. 4. An excerpt from the *planning* ontology.

```
(defcontext  GL-Mapping  :theory (guidelines))

(defrelation  maps
  :is-primitive  extrinsic-structuring-relation)

(defrelation  mapped-by
  :is (:inverse  maps))

(defrelation  equimaps
  :is-primitive maps)

(defrelation  equimapped-by
  :is (:inverse  equimaps))

(defrelation  partly-maps
  :is-primitive  maps)

(defrelation  partly-mapped-by
  :is (:inverse  partly-maps))

;;;ProForma
(defconcept  Proforma-entity
  :is-primitive  (:and  document
                        (:some mapped-by  document)))

(defconcept  Proforma-task
  :is-primitive  (:and  Proforma-entity
                  (:the  equimapped-by  generic-plan)))
```

```
(defconcept  Proforma-plan
  :is (:and  Proforma-task
             (:the equimapped-by  complex-plan)))

(defconcept  Proforma-action-task
  :is-primitive
   (:and Proforma-task
    (:the subsumapped-by  elementary-plan)
    (:all has-description  symbolic-string)))

;;;GLIF
(defconcept  Glif-step
 :is-primitive  (:and  Glif-entity
                 (:the equimapped-by  generic-plan)))

(defconcept  Glif-guideline-step
  :is (:and Glif-entity
            (:the equimapped-by  complex-plan)
            (:all has-description  symbolic-string)))

(defconcept  Glif-action-step
  :is-primitive  (:and  Glif-step
                  (:the subsumapped-by  elementary-plan))
  :implies
   (:some has-description  Glif-action-specification))
```

Fig. 5. An excerpt from the *GL-mapping* ontology.