Design of an Ontology on Cerebral Aneurysms: Representing the Conceptual Space of the @neurIST Project

Susanne Hanser[a], Martin Boeker[a], Kai Kumpf[b], Stefan Schulz[a]

[a] *Department of Medical Informatics, University Hospital, Freiburg University, Germany*
[b] *Fraunhofer Institute for Algorithms and Scientific Computing SCAI, FhG St. Augustin, Germany*

Abstract

Ontologies are designed to represent knowledge, and to support the semantic interoperability among heterogeneous, distributed sources (e.g. databases or web sites) by providing machine-readable descriptions of entity types (concepts) in a domain. We describe our approach to a domain ontology which is currently under development within the scope of the European project @neurIST, intended to serve as a module in a complex architecture aiming at the provision of a better understanding and management of intracranial aneurysms.

Keywords

Medical Informatics Applications; Ontology design; Intracranial aneurysm; Terminology

Introduction

The aim of the European project @neurIST [1] is the provision of an integrated information structure related to intracranial aneurysms and subarachnoid hemorrhage. The expected benefit for clinicians and scientists includes the support of diagnosis and treatment planning, particularly with regard to aneurysm rupture risk, and an easier access to knowledge in the field, provided by a set of elaborate software tools and platforms. In this paper, we describe our approach to the design of an ontology binding together the different levels of description of the disease intracranial aneurysm (e.g. from a clinicians or a geneticist's view) and different sources of information (e.g. literature, clinical databases, image databases).

Materials and Methods

Following the current state of the art of Semantic Web technology we use the ontology editor Protegé and OWL DL as a description language. The logical consistency of the ontology during the editing process is ensured by the RacerPro OWL reasoner . In the first place, we intend to reuse existing terminology sources (as the UMLS) and widely acknowledged ontologies like the Foundational Model of Anatomy.

Acquisition of types and relations

The first step to the identification of relevant entity types ("concepts") consists in collecting terms, terms describing disease and patient as well as terms related to technical devices and data structures used in the process of description, therapy and prognosis. Sources of relevant entity types in the domain were 1. clinical databases and information models (CRIM), 2. literature (high frequency domain specific terms extracted from literature), 3. domain experts, 4. the UMLS metathesaurus, and 5. open source databases (e.g. in the molecular biology field)

Ontology Design

Top level: Particular

We chose the *Descriptive Ontology forLinguistic and Cognitive Engineering (DOLCE*) [2] as the basic ontological framework of our ontology and included its DOLCE-Lite-Plus version via the OWL import mechanism. The root of DOLCE's top level ontology is named "particular". This reflects a consensus among top-level ontologies: The ontology is a system of semantic types which define classes of particulars (individuals or instances), and not of concepts in the sense of entities of mind. According to DOLCE, "particulars" are further subdivided in terms of:

- Endurant (including object- and substance-like entities, e.g. "Internal Carotid Artery", "Blood")
- Perdurant (event- and state-like entities, e.g. "Angioplasty", "Hypertensive Disease")
- Quality (individual attributes, e.g. "Blood Pressure", "Sex") and
- Abstract (mainly conceptual "regions" for structuring attributes – "values", e.g. "Male" as a possible value for the quality "Sex")

Results

Coverage

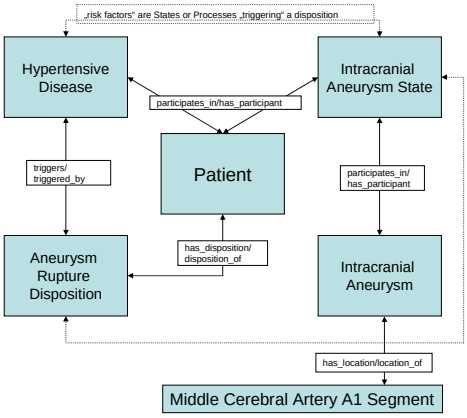
The actual representation of the domain covered by the @neurIST project includes over 2000 entity types, which

mostly belong to the realm of clinical medicine. They are classified throughout as perdurants, endurants, qualities and value ranges. Logical descriptions link nodes from different branches of the taxonomy, for example: The relation "participates_in/ has_participant" links e.g. patient ("human"), pathologic anatomical entity, and disease.

Model

Fig.1 shows a model of an exemplary clinical situation: A "patient", for example, participates in a disease ("Intracranial aneurysm state" which is the pathologic state of bearing an aneurysm), an "intracranial aneurysm (which is an anatomical abnormality) participates, too. The patient suffers from hypertensive disease; a fact which according to epidemiological knowledge leads to an increased probability for aneurysm rupture and subarachnoid hemorrhage.

Fig. 1: Model of a possible clinical situation in the project



Usability

A synonym database linking to the ontology classes provides a variety of names and acronyms used by partners in their own research areas. - We introduced a second hierarchy for the representation of entity types in certain application-related contexts of human knowledge (Particular in Context), including the possibility to extract ontology subtrees according to user's needs, for example for text mining purposes. - Finally, a web browser is under development which will provide easy access to the ontology content.

Discussion

Are top level categories necessary? In contrast to 'lightweight' ontologies, which focus on a minimal terminological structure (often just a taxonomy) fitting the needs of a specific community, the main purpose of foundational ontologies like DOLCE is to *negotiate* meaning, either for enabling effective cooperation among multiple artificial agents, or for *establishing consensus* in a mixed society where artificial agents cooperate with human beings. It is our experience that the use of the upper level categories facilitated a classification of the @neurIST entity types in the difficult field of clinical terminology with its often vaguely defined vocabulary. Our choice to use DOLCE depended on the estimation that an ontology with a "cognitive bias" was more appropriate for the representation of a conceptual space covering several scientific domains with their different views on the disease, than the more realistic, fundamental approach of the BFO (Basic Formal Ontology). While DOLCE aims at capturing the ontological categories underlying natural language and human commonsense, BFO claims that "each ontology (*SNAP* and *SPAN*) represents some partition of reality" [3]. In addition, DOLCE has been applied in the restructuring of the WordNet Lexicon with good results. [4]

Conclusion

In our opinion, a classification of the entity types from the @neurIST CRIM (describing mainly the clinical aspects of intracranial aneurysms) according to the Descriptive Ontology for Linguistic and Cognitive Engineering results in a sound classification of the domain, while a the separate presentation of human knowledge meets user's needs of a classification and concept representation in certain contexts.

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Address for correspondence

Susanne Hanser, Department of Medical Informatics, University Hospital, Stefan-Meier-Str. 26, D-79104 Freiburg, Germany, e-mail: hanser@mi.ukl.uni-freiburg.de