

Realist representation of the medical practice: an ontological and epistemological analysis

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***Abstract.** Realist ontologies organize knowledge by strict adherence to philosophical principles, ensuring robustness and coherence. According to those principles, only entities empirically verifiable can be represented. Our study aimed to analyze medical records to evaluate which kinds of entities should be represented for physicians. We classified the entities and found several entities that cannot be represented in realist ontologies. After due analysis, results suggest that a categorization that distinguishes reality from medical knowledge about reality and observations under both of them are useful to describe entities present in medical records.*

1. Introduction

Information structuring in electronic health records (EHR) is essential for the development of health applications, due to its ability to exchange information between different applications and healthcare professionals. Structured records are amenable to use in several situations, such as: a) scientific discoveries; b) use of recorded data by other professionals; c) healthcare facility management and quality control; d) prevention of epidemics and health policy development.

System interoperability (the capacity of communication between systems without human intervention) requires shared semantics of terms used in both systems. Recently, the use of ontologies for semantic representation is being studied in several domains, like the biomedicine [Rubin et al. 2008]. Particularly, the development and wide adoption of the realist stance for ontology creation allows for an explicit, stable and language independent vocabulary definition, which promotes communication without ambiguities [Smith and Ceusters 2010].

Even though such methodology aims to describe scientific knowledge, such as gene and protein biological functions [Hill et al. 2008], it actually limits the representation of natural language terms that have no direct referent in the world. This “non-ontological” terminology is important to clinical records [Stenzhorn et al. 2008]. For instance, a clinician may use the term “hepatitis” to refer to a real hepatitis, but can also refer to a clinical suspicion (that the patient may have the disease), or to a preventive action (like vaccine prescription). In this paper we generically refer to those terms as epistemological [Bodenreider et al. 2004].

Besides, Schulz and colleagues [2009] argue that the attempt to code probabilistic and default knowledge using ontologies is likely to create incorrect models. In fact, the realist approach seems incapable to represent statements that are not universally true, such as “suspected fever”, “past history of fever” and “no fever”.

This research aims to evaluate real medical records, to analyze which entities are amenable to representation by the so-called realist ontologies, as defined by [Smith 2006]. The description of medical record entities by ontological and epistemological principles, part of an ongoing research project, is being used to create a set of procedures that will guide the analysis and create a generic framework that will improve understanding of medical systems specificities.

This paper is structured as follows. In section 2 we describe the advantages and limitations of realist ontologies for medical knowledge representation. In section 3, we present a critical evaluation regarding the relationship of formal ontologies and clinical reasoning. In section 4 we present the methodology used, aimed at identifying information contained in real medical records. In section 5 we present results, in section 6 we discuss the results and in section 7 we present our final remarks.

2. Ontologies

Ontologies are being used in large scale in varied domains like architecture, geography, [Bittner 2010], medicine and biology [Bittner and Donnelly 2007], whether as support for legacy classification systems, or as way of adequately representing a domain. In the following sections we describe applications in biomedical ontologies (section 2.1), as well foundational principles of realist ontologies, widely used in biomedicine (section 2.2).

2.1. Biomedical ontologies

Ontologies have been successfully used in the biology and medical domain around the world. Several initiatives were gathered in the *Open Biomedical Ontologies Foundry* (OBO), a repository of accessible, interoperable ontologies, described in uniform syntax and unequivocal identification [Smith et al. 2007]. Considering the OBO group, some ontologies are worth mentioning, due to innovation and intense use in scientific research. Among them, the *Gene Ontology*, an ontology that describes basic characteristics of genes; the *Foundational Model of Anatomy*¹, which describes the prototypical human anatomy; the *Cell-Type Ontology*, which describes cell types from some living; the *Protein Ontology*², which describes the relationship between proteins and classes that represent protein evolution; and the *Chemical Entities of Biological Interest*³.

Besides these big foundational ontologies, several others are still under evaluation and available at OBO, such as the *Disease Ontology*⁴ [Cowell and Smith 2010], the *Ontology for Biological Investigations* [Brinkman et al. 2010], the *Ontology for General Medical Science*⁵ [Scheuermann et al. 2009].

¹ <http://fma.biostr.washington.edu/>

² <http://pir.georgetown.edu/pro/>

³ <http://www.ebi.ac.uk/chebi>

⁴ http://do-wiki.nubic.northwestern.edu/index.php/Main_Page

⁵ <http://code.google.com/p/ogms/>

2.2. Realistic ontologies

The term “realism” in Philosophy is widely used and controversial [Miller 2010]. We have to emphasize that realism, while philosophical discipline, can disclose different flavours. Indeed, there are issues under unending debate among people which declare themselves as being realists. Defining universals, a main tenet of realism, is an example of issue on which there is no agreement [MacLeod 2005]. In this paper, we take the “ontological realism” as a methodology for ontology development – said “realist ontologies” – based on principles of the philosophical realism. It is a methodology widely used in biomedicine [Baker et al. 1999][Grenon et al. 2004] grounded at the following generic tenets: [Munn and Smith 2008]: i) there is a real world; ii) the reality in which we live in is part of this world; iii) we are capable of knowing the world and reality, even if just in an approximate way.

One of the assumptions of the ontological realism is the theory of universals, which states that in reality there are particular and universal entities. Particulars are entities described by the observation of the real world, e.g. a clinic or a laboratory. Universals represent that which is common to every correspondent particular - e.g. the characteristic of having a head that is common to every human being – which is invariant in reality [Smith 2004][Smith 2006]. Since ontological realism is based on reality and proposes that the best way to describe it is through science, universals are those entities chosen to be used in the formulation of scientific theories.

According to the ontological realism, the unrestricted creation of classes to represent every possible entity leads to inconsistencies. Classes are human creations – e.g. every human being that is a man and likes swimming – and may be interpreted in different ways [Munn and Smith 2008]. To avoid that, the realist methodology restricts the possible classes to those defined by the scientific community. However, the precise distinction between universals and classes is not always trivial. While universals are grouped by what they are, classes are grouped by how they are [Smith e Ceusters, 2010].

The realist methodology uses an upper-level ontology to organize universals with a top-down approach. Examples of upper-level ontologies are the BFO [Grenon et al. 2004], DOLCE, the SUMO, among others. In the BFO, adopted in the ontological realism stance, we can find structuring divisions made by generic universals called continuants and occurrents. This division is based on the notion of SNAP and SPAN [Grenon et al. 2004]. SPAN entities, called occurrent or perdurants, are universals that possess a determined beginning and end, and encompass process (e.g. “the life of an organism”) and spatiotemporal regions (e.g. “the eighties”). SNAP entities, also called continuants or endurants, are universals for particular that maintain their identities through time (e.g. a “human being”). Continuants may be dependent (e.g. “the color of an object”), independent (e.g. “a table”) or spatial regions (e.g. a “point”). To explain the different treatments for high-level entities in other ontologies abovementioned is beyond the goals of the present paper.

The use of the same upper-level ontology as starting point to create domain ontologies increases the chance that its universals are compatible and, therefore, the chance that they are amenable to integration.

3. The limitations of realist ontologies for representing medical practice

The extension of realist biomedical ontologies to the medical practice is an alternative for medical information organization. However, considering institutions of most countries, medical documentation is usually made of barely structured documents, sometimes even handwritten, containing heterogeneous information. Even so, the medical record is an essential work tool for the clinician. The record is used for medico-legal reasons, as a tool to support care plan creation and as a support to find information required for clinical decision-making.

The realist ontology approach has been the target of many criticisms, which usually argue against the proposal of universals as a sine qua non condition to the creation of good ontologies [Merrill 2010a][Merrill 2010b] [Rector 2010] [Cimino 1998][Cimino 2006] [Dumontier and Hoehndorf 2010]. Such approaches for biomedical ontologies emphasizes the importance of language, communication and medical reasoning and puts under suspicion the obligation in considering [Merrill 2010a]. In many cases, such approaches have been labeled as “epistemological”.

Conceptual approaches, a variant of idealism, are closer to medical everyday language, since they use terms not referenced in reality which are commonly present in new and yet not fully comprehended clinical situations. In the medical practice, diagnoses are usually presumptive and based on incomplete data, making it difficult to identify a particular and the corresponding universal. In fact, statements in such context are constantly revised and do represent truths, but the physicians grounded opinion.

Realist-oriented researchers argue that the creation of ontologies around concepts is based on language and, therefore, is subject to ambiguities and differences of understanding and interpretation by different individuals [Smith 2006]. These researchers consider that ontologies are artifacts made for use by computers and that any natural language-derived ambiguity harms interoperability efforts. This is particularly important in natural sciences representation such as biology in which, despite the enormous volume of data, there is consistency in observation by different institutions. Also in medicine, anatomical and physiological statements are consensual when attributed to universals.

This is made clear by comparing the statements “AIDS is spreading quickly through Asia” and “AIDS is caused by the HIV”. The term AIDS in the former is a class, while it correspond to a universal in the latter. Classes are arbitrary sets and can result in representation that cannot be understood and interpreted. By restricting the ontological commitment to reality as described by science, the ontological realism promotes consensus.

Another relevant aspect to be considered is the distinction between ontology and epistemology. Epistemology is the study of how cognoscent beings come to the truth about some event in reality. The difference between the terms can be shown by evaluating how entities are defined in ontology and in epistemology. Ontology is about an object, process, event, whole, part, determination, dependence, composition, etc. Epistemological statements are about the way we know things and is about belief, truth, probability, confirmation, knowledge and its variations [Poli 2010]. While ontology is a theory of things, epistemology is a theory of knowledge.

The interdependence between the existence of an entity and the knowing about it frequently blurs the distinction between ontology and epistemology. Bodenreider and colleagues classify epistemological terms usually identified in biomedical terminologies in four categories [Bodenreider et al. 2004]:

- Terms containing classification criteria: terms that do not represent universals, but that intend to convey information. For example, the distinction between “febrile seizure” and “afebrile seizure” is not a distinction between characteristics of the seizure itself, but conveys information about probable cause and prognosis.
- Terms reflecting detectability, modality, uncertainty, and vagueness: since complete understanding of a clinical situation is very difficult, physician usually express this incomplete knowledge of the patient condition by modal and approximate statements. E.g. “possible cancer”, “probable cancer”, “unspecified chest pain”.
- Terms created in order to obtain a complete partition of the domain: contain terms that intend to encompass entities not described by other classes. E.g. “Other” and “Pneumonia not otherwise specified”.
- Issues related to normality and to fiat boundaries: terms that intend to convey instructions about how the information should be interpreted, not about the entity itself. E.g. “normal height”, “enlarged liver”. It is important to point out that part of the medical knowledge is based on historical events which had an almost arbitrary definition of normality [Vickers et al. 2008].

The fact that clinical observations are necessary is not opposed to the realist methodology: information about opinions are fundamentally different from information about objects [Munn and Smith 2008] and both have a place in an descriptive ontology⁶. However, the medical practice requires the recording of information of both natures, named here ontological and epistemological, including impressions, plans, suggestions, etc. We intend to pursue this issue while searching for a complementary approach that helps in understanding the medical reality.

4. Methodology

This ongoing research objective is to evaluate the representation of health information in real medical records, through the use of realist ontologies. We intend to determine its limitations and propose new ways of representing non-ontological information. For example, administrative data, which at first had no counterpart in realist reference ontologies, has to be represented through the creation of other ontologies for dealing with such entities, like the Information Artifact Ontology [IAO 2011]. The methodology is composed by the following steps:

1. Record creation based on real clinical cases: The analysis must consider the way health professionals record medical events. We studied two complete records, created by two Internal Medicine specialists, based on common presentations of real patients. No identification data was recorded, such as name, age,

⁶ “concerns the collection of such prima facie information on types of items either in some specific domain of analysis or in general” [Poli 2010, pg.2]

geographical location, health facility, dates and identification and contact numbers, according to recommendations by [Meystre et al. 2010].

2. Transcription of records for information identification: In order to identify information unities, a domain expert transcribed the records in sentential fragments. The domain expert was asked to identify the reason for recording those entities and the information that is being conveyed by the representation. The transcription used the principles of logic and controlled languages described [Fuchs et al. 2005][Fuchs et al. 1999] , which allowed clear identification of entities recorded in natural language, outside the particular context in which the event took place [Vickers et al. 2008]. Since the objective of this paper is to analyze the content of the text, syntactical and markup aspects pertinent to automatic processing are omitted. We hereafter call those information unities as entities, despite their physical existence.
3. Analysis and classification of the record’s information items, according to ontological realism guidelines: The information entities were analyzed according to the tenets of the ontological realism [Grenon et al. 2004] , to verify if they were suitable to ontological representation. This analysis was guided by pre-established criteria aimed to classifying the entity in some upper BFO class. Some examples can be found in table 1. Each entity was tested against the set criteria, respecting the BFO class hierarchy. E.g. the first test separates entities in continuants and occurrent; after this distinction, specific criteria are used for each class. The entities that don’t belong to any BFO class are analyzed according to realist principles and their use in everyday medical practice. We selected some cases for further discussion, presented in section 6.

Table 1. Distinction between continuants (EMT) and occurrents (ECT)

Distinction (I)	Entities that maintain their identity through time (EMT)	Entities that change through time (ECT)
Characteristics	a) The entity exists completely in any given period of time in which it is present b) The entity has no temporal parts.	a) The entity unfolds through a period of time.

5. Results

The records analyzed represent outpatient visits. The first one describes the consultation of a patient with an unexplained chest pain and the second a post-discharge consultation due to dyspnea. The records make use of routine record organization, such as “Complaint”, “History”, “Physical examination”, etc. In table 2 we present a small extract of one of the documents. Partial results can be seen below in Table 3:

Table 2. Extract of an outpatient record of a fictitious patient

QP: Chest pain and abdominal pain. HMA: Six months ago, the patient felt severe precordial pain in addition to nausea and dyspnea. She attempted medical care in the Hospital X, where received isordil + AAS 300mg. Enzymes: CKT 262 CKMB 30. She was not aware of previous pathologies. It was prescribed: Captopril, HCTZ e AAS. Last month, the patient felt severe pain again and sought for medical care in a different place. Then, it

was prescribed: Losartan, AAS, Sinvastatina e Nebilet.
 She sought for medical care in other occasions because of the precordial pain. In addition to the medicine mentioned, she uses Metoprolol - 100 mg 12/12 h.
 She reports diffuse and intermittent abdominal pain, which becomes worse in case of stress. It is not related with bowel movement alterations. She also reports rare burning epigastric pain that improves with water drinking.

Table 3. Example of mapped and non-mapped entities to realist ontologies

1- Aspects that represent entities IN REALITY (some examples)	
Continuant	Occurrent
-Chest pain -Abdominal pain, Precordial pain, Epigastric pain -Nausea, Dyspnea -Enzyme -Captopril, Losartan	-Were prescribed -Makes use -Bowel movements -Moment of first occurrence of pain (six months) -Moment of re-incidence of pain (one month ago)
2- Aspects that represent useful constructs for medical practice NOT empirically verifiable	
- Severe (precordial) heavy pressure (pain)	- Diffuse and intermittent (abdominal pain) - Rare burning (epigastric pain)
3- Aspects that represent observations ABOUT reality (not reality itself)	
-CKT 262 -CKMB 30 -Left ventricle ejection fraction: 68%	
4- Aspects that represent observations ABOUT the physician understanding of the clinical situation (not about reality)	
-Previous consultations and prescriptions -Not related to bowel movement alterations	-Previous diseases - (Diffuse and intermittent abdominal pain) that worsens with stress - (Rare burning epigastric pain) that improves with water drinking

6. Discussion

The medical record is a complex document used for several purposes in healthcare processes. According to the Brazilian Medical Council, it is “a single document made of a set of recorded information, signs and images, created after (events) about the patient health and care provided, of a legal, private and scientific character, that allows communication between the multi-professional team and continuity of care provided to the individual” [Conselho Federal de Medicina 2002, art 1º]. To live up to those expectations, the professional uses the flexibility of natural language expressions to represent the clinical situation, his clinical reasoning process and the relevant context of the health event.

In our research, we drew terms from records trying to fit them to constraints imposed by realist ontologies. Then, we created two main sets: in the first one, we included the entities that could be represented in realist ontologies; the second one gathers entities that can not be represented in realist ontologies as we defined them in the context of this paper (vide section 1) Terms that can be used in realist ontologies are presented as the first group of table 3. Arguably, realism has been shown capable of representing diseases, disorders [Scheuermann et al. 2009] and symptoms [Smith et al. 2009], as evidenced by the *Ontology for General Medical Science* (OGMS). The existence of diseases – defined as a “disposition (i) to undergo pathological processes that (ii) exists in an organism because of one or more disorders in that organism” [Scheuermann et al.

2009, pg.3] is well known by medical science, and its representation is robust and homogenous. Likewise, symptoms can be seen as body characteristics that a patient experiences. In this case, we represent the body alteration considering its scientific description. On the other hand, the diagnosis itself is not a patient attribute, but rather “a conclusion of an interpretive process that has as input a clinical picture of a given patient and as output an assertion (diagnostic statement) to the effect that the patient has a disease of such and such a type.” [Scheuermann et al. 2009, pg.5])

We observed that the realist methodology is incapable of defining symptoms qualities. In order to evaluate a patient, each symptom must be described according to its seven characteristics [Bickley and Szilagyí 2009]: Location; Quality; Quantity or severity; Timing; Setting in which it occurs; Remitting or exacerbating factors; Associated manifestations. These characteristics can be classified in three groups, according to their relation with realist ontologies.

Formal ontologies are capable of precise representation of symptom location and temporality, through the description of body structures – organs and systems – or spatiotemporal regions. This first group can be described by upper level classes of the BFO, as continuants – independent continuants and spatiotemporal regions – and occurrents – the temporal region occupied by the symptom and, eventually, the symptom itself. For example, “chest pain” and nausea”. As stated in the Methodology section, this analysis was based on the BFO, but different upper ontologies may suggest different approaches. This is markedly true in the case of qualities [Masolo and Borgo 2005], defined by the BFO as “a specifically dependent continuant that is exhibited if it inheres in an entity or entities at all (a categorical property)” [Basic Formal Ontology].

The second group of table 3, containing the characteristics of quality and quantity/severity, describes attributes of the symptoms, its temporal evolution, qualities, dispositions, functions and roles. The qualities refer to symptom types as described by scientific knowledge of common clinical presentation of diseases. There are regional and national variations of such typology, but classical symptom description is fairly constant. For instance, the term “crushing pain” is commonly interpreted as a cardiac originated pain. The quality “crushing” of the precordial pain has no direct and unequivocal relation with the subjacent disorder, but the history of pain is similar in patients with the same kind of disorder. In this case, we argue that the term is not a realist universal, but can be described by concepts in a coherent fashion. The same criteria applies to severity (“Severe pain”), which shows the same linguistic ambiguity (how much is severe?). These terms must be described by non-ontological artifacts to avoid reasoning and classification errors, since the distinction between types of pain cannot be empirically verifiable – e.g. distinguishing a crushing from a heavy pressure pain. Another example would be “diffuse abdominal pain”, which should not be treated as a single ontological entity.

In the fourth group of table 3, we find the aspects referring to the situation in which the symptom was experienced by the patient, according to the medical record description. The setting of occurrence describes the state of affairs at the moment when the symptom was perceived, what the patient was doing, climate and environment conditions, events that preceded the symptom, etc. Remitting or exacerbating factors describe entities that, according to the patient’s or physician’s interpretation, changed the natural course of the symptom. This interpretation may be motivated by previous knowledge (e.g. causes of

chest pain may be distinguished by their relation with physical exercise), temporal coincidence or unjustified beliefs. Finally, associated manifestations may be represented by any other symptom, or the absence of symptoms, as long as they aid medical reasoning and the definition of a diagnosis. The representation of this group through realist ontologies is mostly ambiguous. In the cited examples, the occurrent “drinking water” and the “epigastric pain” intensity decrease are temporally related, but the causality cannot be empirically determined. Rather, they reflect the understanding of the situation, so that there is a belief that both entities are causally related.

Besides symptoms, several other entities were found, such as medications, laboratory test results, physical examination findings, among others. Entities like life signs measurements and lab test results do not directly refer to patient qualities, but to observations about those qualities. For example, the CKMB (creatinine phosphokinase MB) refers to the enzyme blood concentration at the exact moment of blood sample collection. It is, therefore, empirically verifiable. However, the value of the measurement is arbitrarily determined (in this case, unity per liter) and does not refer to the existence of the enzyme in the real world. Besides, this information is not analyzed using logic operations, but used in a sequence of pre-established thinking rules, according to clinical training. In this clinical case, the value 30 U/L is just above the normal value (26 U/L) and, therefore, leads the physician to question the hypotheses of myocardial infarction, suggested by the initial presentation of chest pain. The presence of continua in the real world requires fiat delimitations, which are justified by pragmatic reasons. [Schulz and Johansson 2007]. We argue that this information should be distinguished from direct referents, since it refers to a representation of an observation about reality. Moreover, it will be interpreted according to reasoning structure, not according to the structure of reality itself.

Several solutions to this problem can be found. Despite conceptualism shortcomings, the restrict use of concepts to represent epistemological information expands the scope of those representations. To improve the meaning of concept and avoid misuse of the term, we will use the definition put forward by [Klein and Smith 2010, pg.722]: “concept should be used exclusively to refer (1) to the meaning of a corresponding general term, this meaning being (2) unique and (3) agreed upon by responsible persons in the given disciplinary field.”

The use of concepts to represent clinical information, though subject to inconsistencies, is closer to language, since it represents term meaning and does not denote an entity (universal or particular). The definition of each term can be made through formal languages or natural language description, depending on the heterogeneity of interpretations given to a term: e.g. concepts such as “up” and “down” are intuitive and interpreted in a constant way, while the term “AIDS” requires precise explanation. The relation between terms should be done through semantic relations “broader_than” and “narrower_than”, considering the term meaning. Additionally, we can consider the relation “related_to”, as proposed in the W3C *Simple Knowledge Organization System* (SKOS) standard.

While the proposed typology encompasses real and epistemological entities, it still needs improvement. The description of knowledge, thought as an attribute of the cognoscent being, does not describe a real entity, but says something about it. For instance, it is false to represent a “canceled surgery” as a “surgery”, for it never

happened. A solution is to represent the “canceled surgery” as an information artifact, a plan that is about the “surgery”. In this case, the surgery will never come to be, but the plan existed through a defined and verified temporal region [Schulz et al. 2010]. These and other hypothetical entities, like instructions (“in case of recurrence, do X”) and goals (“the patient should try to lose at least 2 kg with this diet within 2 months”) can be placed in more than one category – it may be seen as a real information entity ABOUT another not yet instantiated real entity, or as a mental model simulation on the part of the physician, stating an algorithmic behavior in the form IF X THEN Y. These cases make evident that further effort in refining the model must be done.

7. Final remarks

The proposed categorization suggest that understanding reality representation in four levels – reality itself, the perception of reality by the being, and the recording of reality [Smith et al. 2006] – makes clear the connection between representation methodologies. This connection will be important for proper computerizing of medical records.

During this research project, we intend to expand this categorization in a framework connecting ontological and non-ontological entities that promotes representation of entities required in medical practice without compromising interoperability and automatic inferences. We intend to explore information models such as the HL7 v3 and the OpenEHR, since they offer a great opportunity to understand the relation between concepts and ontological entities.

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