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Review

Semantic Web and the Future of Health Care Data in Family Practice

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Abstract

New tools in data management have emerged in the last decade, which are going to revolutionize electronic health records and health information systems. Family practitioners are the first, main and continuous contact with the patient. They are the source and endpoint of the circle of information, generated by judicious medical documentation and smart secondary use of medical data. This article is an introduction to health terminologies, ontologies, semantic data, and linked open data, all expressions used by computer scientists, preparing themselves for the next step: semantic web for health care data.

Keywords: Biological Ontologies, Classification, Controlled vocabulary, Family, Physicians, Semantic interoperability

INTRODUCTION

The complex world of patient and family doctors

The acronym GP/FM shows by itself the extent and the complexity of the domain. "General Practice" (GP), used in UK and in continental Europe, refers to what first line doctors are doing in the medical field, while "Family Medicine" (FM), most used in the United States and Canada, is addressing for which people the doctor is working. This apparent opposition between what and for whom, process (practice) and humans (family) reflects the tension between the hard and soft sciences, between biomedical and anthropological views.

GP/FM terminologies address multiple scientific facets of medical knowledge in anthropological/psychological and epidemiological/biological sciences (Van Dormael, 2001). The General Practitioner/Family Physician (GP/FP) becomes an interface between the health system and the patient's world. Being at a crucial network point in the complexity of health care (Plsek and Greenhalgh, 2001), s/he has to use understandable terms (lay terms) to communicate with the patient and his social network (Messai et al., 2008). Links between lay

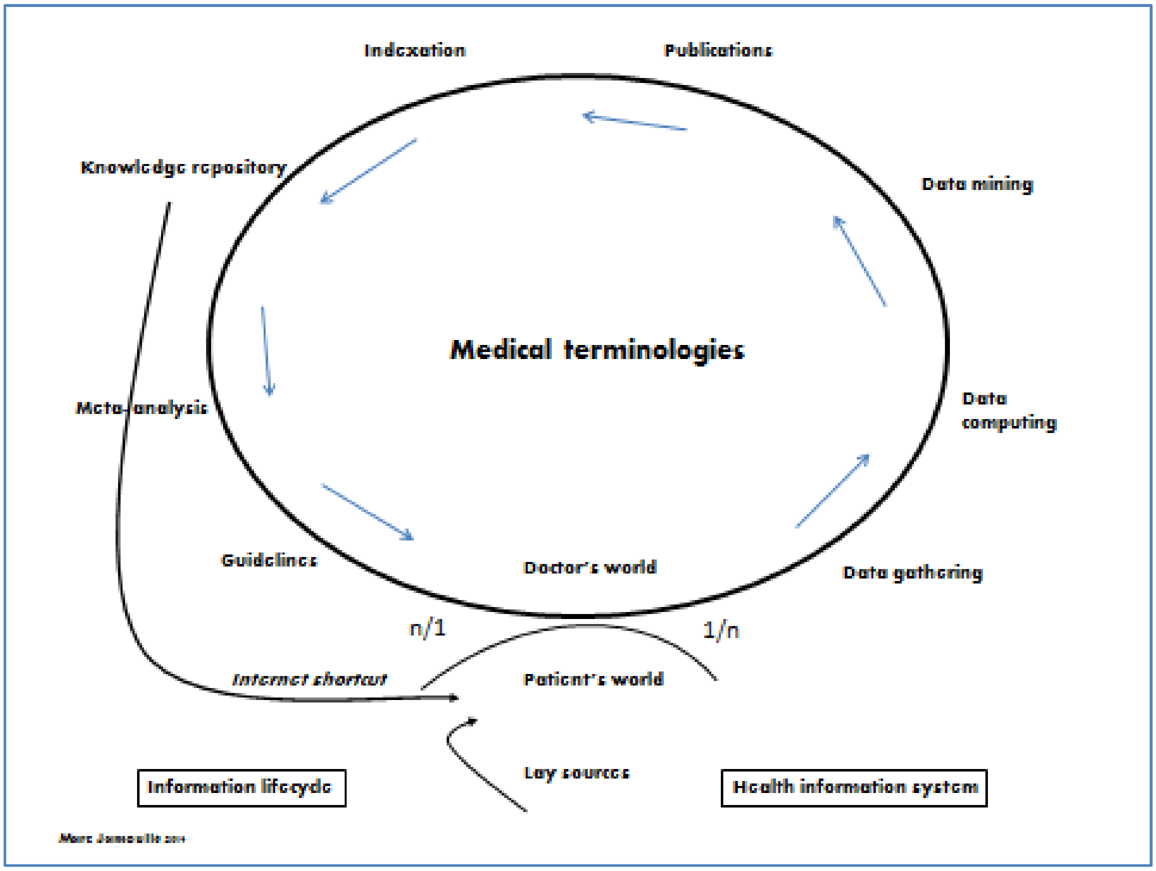

Figure 1. Information lifecycle in medical practice. The consultation is a meeting between patient's and doctor's world of reference

and professional terminologies are becoming a necessity and researchers are working fast in this domain (Cardillo et al., 2014).

Medical data, coming from the health ecosystem, are transformed by research, and finally edited as publications, could create new knowledge and insights (Liyanage et al., 2012). Information used to treat the patient is most often of statistical nature (n/1) while the patient is unique and becomes sources of information (1/n) (Glasziou et al., 1998). In the lifecycle of medical information, guidelines (Woolf et al., 1999) or EBM (Evidence Based Medicine) resources (De Jonghe, 2015) offer a particular way to go back to practice with sets of coherent, comprehensive and consensual recommend-dations which minimize the potential harms to patients (Gray, 2004).

Primary Care terminologies have thus to build bridges between different and distinct worlds: the biomedical world and the patient's world. All this requires a common language or at least common meanings. Hence, there is a crucial role for sophisticated medical terminologies and classifications (Figure 1).

Moreover, patients express their fears and feeling in a particular language, which is sometimes poorly understood by doctors and transformed in preconceived symptoms or disease (Gomes et al., 2015), as shown in Figure 2. This requires a deep professional understanding but also dedicated consumer-oriented terminologies, which bridge the gap between lay language and technical jargon (Cardillo et al., 2010; Marshall, 2000; Smith, 2011).

New tools in information management

Since the seminal proposal of Tim Berners Lee to turn the Internet of documents into an Internet of data (Berners-Lee et al., 2001) (then Internet of information and Internet of knowledge), giant steps have been made by numerous researchers. Gradually the Healthcare Knowledge Management (HKM) field becomes a multidisciplinary area involving family physicians, medical specialists, terminologists, taxonomists, computer scientists, knowledge engineers, and computational linguists, while medical informaticians may often offer the glue between all this heterogeneous competencies.

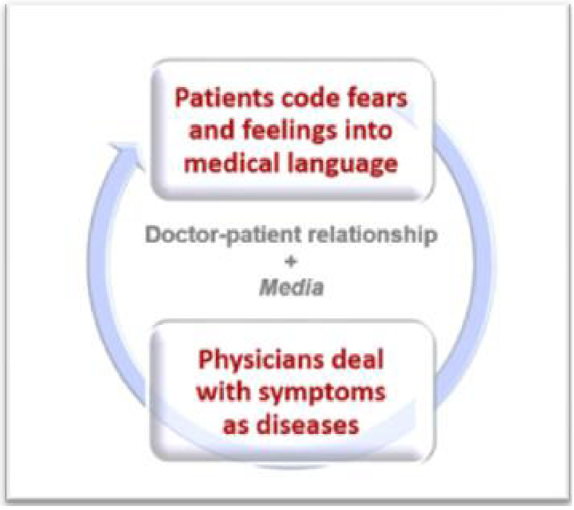

Figure 2. Bias in patient doctor communication

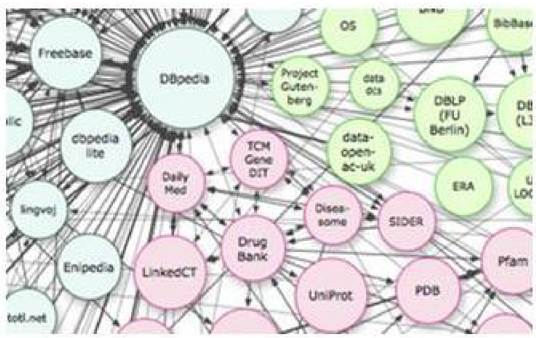

Figure 3. Linked Open Data

The medical semantic web (See Semantic Web Health Care and Life Sciences Interest Group, 2011) and the huge possibility of distributed data have exploded in many different fields: clinical guidelines (Kumar et al., 2004; Camilo et al., 2013; Claudio et al., 2008) consumer information resources (Smith and Fellbaum, 2004; Medline plus http://www.nlm.nih.gov/medlineplus/) mappings between medical classification systems (Cardillo et al., 2008; Ceusters et al., 2005), biomedical data integration (Smith et al., 2007; Ofoghi et al., 2014) semantic interoperability (Qamar, 2008); Linked data (Bizer et al., 2009) e.g. the Bio2RDF framework, to create and provide linked data for the life science (The Bio2RDF frameworkhttp://bio2rdf.org/), medical terminologies and classification systems development or revision (Tudorache et al., 2010; Cramerotti et al., 2015), representation of adverse events (Ceusters et al., 2011), medical education (Blaum et al., 2013), Supervised Machine Learning (SVM) (Pham et al., 2014; Volker et al., 2005-2007), Natural Language Processing (NLP), ontologies (Liu et al., 2011) and many others.

Semantic web and linked data

Semantic web technologies and, above all, Linked data (Figure 3) have emerged as a future solution to exchange scientific data distributed between so many providers as family physicians and colleagues, spread around the globe. The considerable development in the field of medical ontologies shows the vitality of this field of discovery.

Advances in information technology, ontologies, new languages such as the Resource Description Framework (RDF) (Allemang and Hendler, 2008), SKOS (SKOS http://www.w3. org/2004/02/skos/), or SPARQL (Salvadores et al., 2012) for semantic queries, transform the Internet into a huge distributed data base. When associated with Natural Language Processing techniques (Ittoo and Bouma, 2013), what will allow strides in information management in general and family medicine.

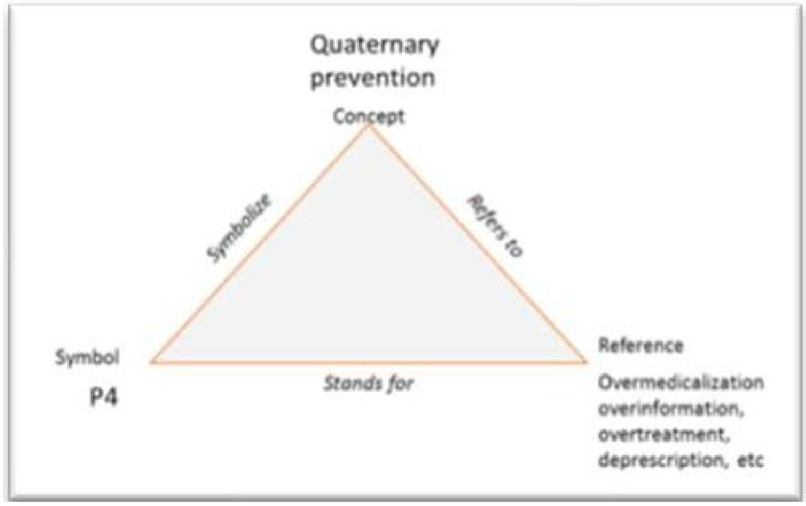

Figure 4. Ogden and Richards triangle (1923) Conceptualization of an item of knowledge.

It brings us closer to fulfill the prediction that "information is to general practice what technology is to specialized medicine" (Van Dormael, 2001).

Ontology, what's in a name?

In its traditional philosophical meaning, Ontology is the study of being or existence (Sadeg-Zadeh, 2015).

In the world of reference of computer scientists, computational linguists, librarians, the word *ontology* has a different meaning. Here it refers to small or huge collections of interlinked concepts, sometimes spread over the Internet, with the aim to describe the knowledge content of a domain in a dedicated machine readable format.

All the fundamental papers in the field are referring to Gruber for a standardized definition: "Ontology is a formal specification of a conceptualization" (Gruber, 1993).

To be informative for general practitioners, this definition needs further explanation of its three terms "formal", "conceptualisation", " and specification".

The first term to understand is the term "formal". Computer scientists are using the term "formal"as a set of symbols understandable by machines. The collections of terms have to be read and understood both by human beings and by machines. Ontologies are not primarily built for human use but for machine use in three types of communications: i) from human to machine, ii) from machine to machine, and finally, iii) from machine to human. So, one can say that ontologies are an arrangement of human knowledge ready to be used by machines.

The second term of Gruber's definition is "conceptualisation". This term has to be understood through the triangle of Ogden and Richards (Figure 4). The concepts, defined as mental representations of reality are symbolized by a machine (and/or human) readable sign, and are finally used to describe specific physical objects or actual practices. All concepts pertaining to a particular domain can be collected and have their relationships defined. Although ontologies often use English labels to describe the concepts to humans, their logical organisation is language-independent, and hence, suitable for multilingual applications. See for further insight the website www.babelnet.org (Navigli and Ponzetto, 2012).

The third and last term of the definition proposed by Gruber is "s*pecification".* It refers to a number of technology tools patiently elaborated by hundreds of computer scientists and computational linguists.

Every single data in a semantic information system receives identification with a Unique Resource Identifier (URI), as proposed by Tim Berners Lee (Berners-Lee, 2002).

Also concepts receive an URI and their origin will be specified as a part of a specific ontological class. For instance, the concept of "feeling tired" will be identified as specific subclass of an OBO Foundry Emotion Ontology (Hastings et al., 2011) on subjective symptoms, perceived by the patient (Figure 5).

Finally, data are no longer recorded in two-dimensional databases of records and fields but are stored in triples. A triple contains a subject, a predicate and an object (Vander Stichele and Dipak, 2015) (Figure 6).

Data structured in triples can be linked by programs. Resource Definition Framework (RDF) is the name of one

```
<!-- http://purl.obolibrary.org/obo/MFOEM_000080->
<owl:Class rdf:about="&obo;MFOEM_000080">
<rdfs:label>feeling tired</rdfs:label>
<rdfs:subClassOf rdf:resource="&obo;MFOEM_000006"/>
<obo:MFOEM_000165>tired</obo:MFOEM_000165>
<obo:IAO_0000115>The subjective emotional feeling
of tiredness, needing sleep.</obo:IAO_0000115>
</owl:Class>
```

Figure 5. The concept « feeling tired » in the Ontology of emotion in RDF (Obo Foundry)

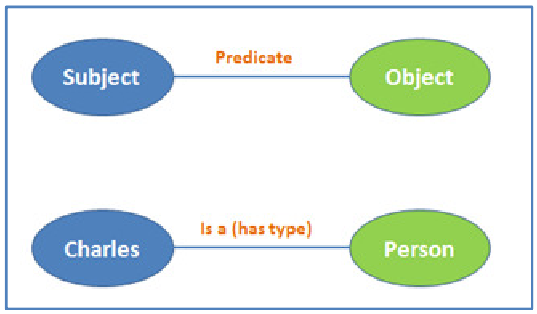

Figure 6. The triplet

of the languages which allow the link of data elements with other data elements. In addition, a whole stack of tools has been build which allow to query, organise and reformat information stored in disparate places on the internet.

Linked data and data differentiation

The linked data cloud on http://linkeddata.org/ shows billions of interlinked data elements published in RDF and submitted to queries by dedicated tools. Whatever is the activity (e.g. business, cars selling, books selling, civil affairs management or health care) there are a lot of sites already managing linked-data.

The difference with the Internet "of documents" is striking. If one queries the BBC linked data web site asking information about an artist, he/she will not receive a link to an already prepared page, but rather it will be created instantly based on semantic data (including semantic relations). This is achieved by retrieving data from the relevant semantic repositories that can be updated independently of the BBC.

The semantic web scientists do not use the concept "retrieved" for this, they use the strange term "dereferencing data" instead. It means that the knowledge needed on a subject is collected based on the URI that identifies it. As URIs is the addressing system of the internet, internet is now the database, or better the "knowledge base"

Practical application in health care terminology: the HeTOP portal

Hetop.eu interactive web site offers cross lingual multi-terminological mappings on a semantic basis (Grosjean et al., 2012). Developed initially on MeSH and French mapping of MeSH for French speaking users, it has evolved towards a dynamic semantic interface supporting a two-dimensional navigation across terminologies/ ontologies and languages. It also provides access to PubMed via the InfoRoute infobutton which is a powerful tool to leverage Medline querying based on the semantic mappings (Thirion et al., 2007) (Figure 7)

The future of health care information systems

The next EHR

Now imagine that the data elements on the patients are not sent by e-mail to GPs electronic medical record (EMR) anymore but that, with the due authorizations and encryption, GP is allowed to dereference the needed data in all the hospitals or primary care settings which store information about his/her patient in the correct format. GP will reconstitute in seconds and only for the time needed the current record of his/her patient. Health-Data becomes Linked-Data (Dowling, 2015). As stated by Pierce et al., "*Semantic Web technologies offer the potential to revolutionize management of health care data*

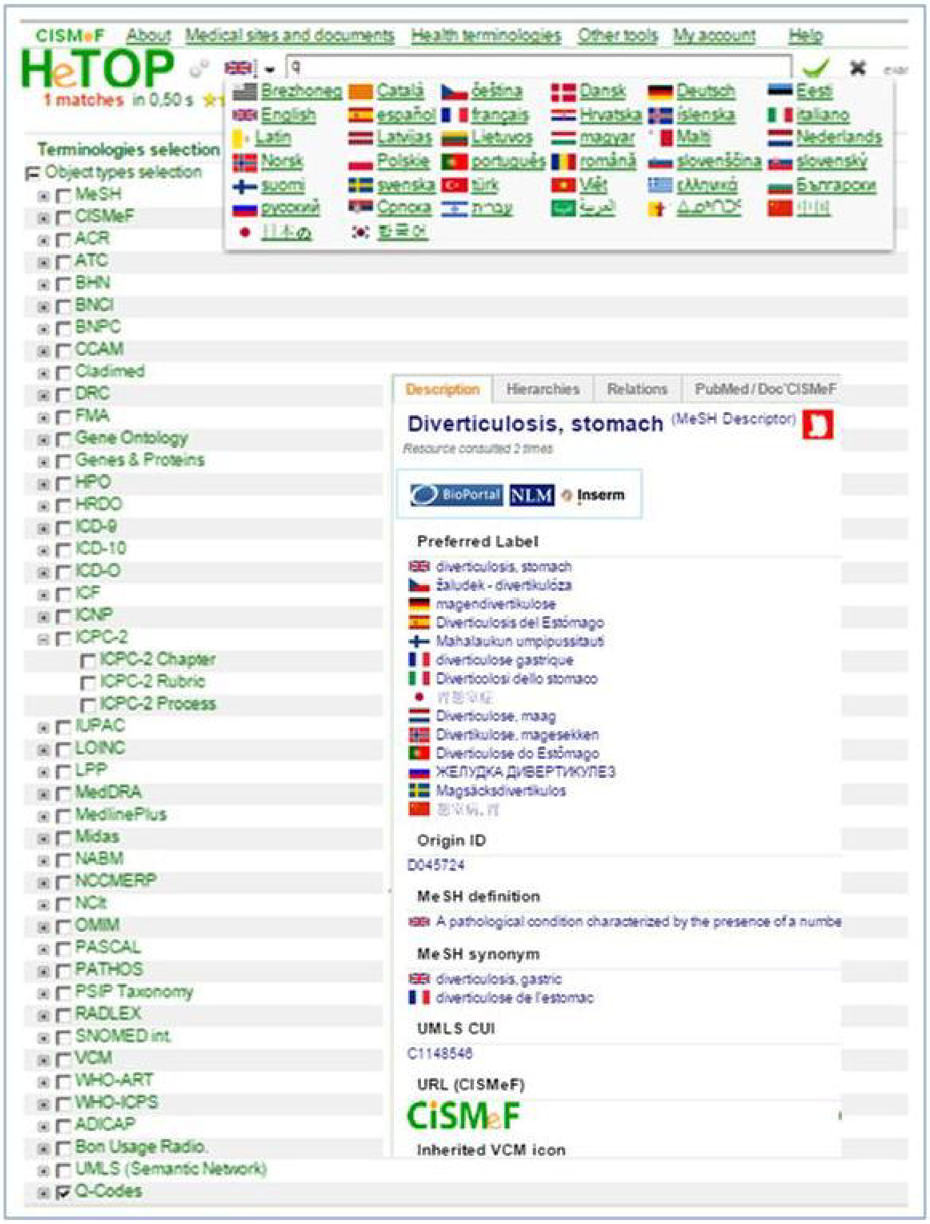

Figure 7. The cross-lingual multi-terminology HeTOP portal and an opening of a MeSH with tabs for access to bibliographic databases

by increasing interoperability and reusability while reducing the need for redundant data collection and storage" (Pierce et al., 2014). This is the future of health care information system and of electronic health records (EHR). This is behind our door (Fernández-Breis et al., 2013). That is why scientists are working so hard to develop medical ontologies like the Open Biomedical Ontologies (OBO) consortium (Smith et al., 2007), National Center for Biomedical Ontology (Musen et al., 2011) or Linking Open Drug Data (LODD) for pharmaceutical research (Samwald et al., 2011).

Natural Language Processing techniques like Information Extraction or Supervised Machine Learning and Data mining can be applied to discover terms (e.g. smoking, cancer) and their semantic relationships (e.g. "causes" as in "smoking causes cancer") from huge text corpora such as patient records. These discovered terms and relationships can be used to populate/augment existing Linked Data ontologies or to create new ones (Fahmi, 2009). EHR warehouses are currently growing all around the world. Multiple research projects are coming with them to ease Information Retrieval and data visualization in EHRs. Some goals are already reached today with complex systems such as I2B2 (Integrating Biology and the Bedside) (Informatics for integrating biology and the bedside. URL: https://www.i2b2.org). These frameworks are not so friendly for physicians because they are dealing with complex models, huge data and heterogeneous information. However, the future is right there with exciting perspectives: intelligent semantic search among EHRs, patient cohorts automatic creation, decision making support, etc (Garde et al., 2007).

Interlinked publications

In the same way, the gray literature and non-published works like more than 50% of abstracts presented by doctors in congresses (Post et al., 2013) could be tagged by semantic specification and stay in the local database of congress organizers or local organizations. Then, a semantic web tool could crawl these websites, dereferencing when needed the asked information through semantically interoperable indexation systems.

One hopes to apply such techniques to an indexing system for communications of family doctors, and link them through the use of dedicated ontologies in a semantic web GP/FM universe.

CONCLUSION

Health knowledge management is a difficult but promising new research domain, in which family physicians should be embedded as the main interface between the healthcare system and the patient, with the Electronic Health Record as the main tool. Family physicians should develop their understanding of and their capacities to be involved in the practical applications of this domain.

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