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Ontologies for terminological purposes: the EndoTerm project

Abstract

In today’s digital society, characterized by the Semantic Web and by Linked Data, ontologies, in the sense of Knowledge Engineering, have paved the way for new perspectives for Terminology, namely in what concerns the operationalization of terminological products. The collaborative work involving Terminology and ontologies has led to the emergence of new theoretical perspectives, one of which being Ontoterminology. This approach aims to reconcile Terminology’s linguistic and conceptual dimensions whilst maintaining their fundamental differences and, in addition, enables the construction of a computer-readable representation of a given conceptualization. Bearing this in mind, this paper presents the EndoTerm project, a multilingual resource within the medical domain – with <Endometriosis> as the core concept – that comprises both verbal and non-verbal representations and that can be computationally represented and manipulated. The presentation of micro-concept systems based on these verbal and non-verbal representations will support a reflection upon the role of the latter in terminology work.

1 Introduction

Today’s digital society has paved the way for new perspectives and opportunities for Terminology. In a context characterized by the Semantic Web¹ and by Linked Data², the need for the operationalization of terminologies, i.e. a computational representation of their concept system, has become increasingly important. In this respect, ontologies, in the sense of Knowledge Engineering (KE) – “a formal, explicit specification of a shared conceptualization”³ –, constitute, according to Roche (2015: 129), “one of the most promising paths towards operationalizing terminologies”. Granting a key status to ontology in terminology work implies, nevertheless, rethinking Terminology’s theoretical and methodological principles and acknowledging the existence of a double dimension – linguistic and conceptual – that may enhance Terminology’s role as a scientific discipline in its own right.

In the recent years, this joint work involving Terminology and ontology has led to the development of numerous resources in various areas of knowledge, one of them being Medicine. The current challenges concerning the way medical information and knowledge are produced, used, stored and shared require efficient and reliable

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¹ Berners-Lee et al. (2001); Shadbolt et al. (2006).
² Berners-Lee (2006); Bizer et al. (2009)
³ Even though Gruber’s definition – “explicit specification of a conceptualization” (1993: 199) – prevails in the literature as the most widely quoted, for the purpose of this paper, ontology in the sense of KE will be regarded bearing in mind Studer et al.’s proposal (1998) quoted above, as it introduces three critical features: the fact that this specification should be explicit, i.e. the type of concepts used and the constraints on their use are explicitly defined, and formal, i.e. machine-readable; and that the conceptualization should be shared, i.e. an ontology should capture knowledge that is consensual among a given community. These authors have merged Gruber’s definition and the one put forward by Borst (1997) – “a formal specification of a shared conceptualization”. For further information, see Guarino et al. (2009)
solutions, in a society that demands immediate and multi-platform access to all digital content.

eHealth, defined by the World Health Organization (WHO) as “the cost-effective and secure use of information and communications technologies in support of health and health-related fields, including health-care services, health surveillance, health literature, and health education, knowledge and research”, has been considered a top priority by national and international institutions worldwide, with several action plans and programs focusing on expert collaboration, patient empowerment and interoperability.

In order to be achieved, these and other goals may greatly benefit from the input provided by an approach combining the operationalization potential of ontologies with Terminology’s vital contribution to specialized knowledge as regards its representation, organization and dissemination.

In short, this paper aims to reflect on the role of ontologies in supporting the creation of concept systems for terminological purposes, particularly in the subject field of Medicine. Within Medicine, special attention will be given to Obstetrics and Gynecology, namely to the concept of Endometriosis, a chronic, inflammatory disease of gynecological nature that is yet relatively unknown, even among the expert community.

This paper will be structured as follows: section 2 will focus on the theoretical background, specifically in what concerns Terminology’s double dimension perspective and the notion of Ontotermology. Section 3 will be dedicated to the role of ontologies and/or terminological systems in the biomedical domain. Section 4 will provide a brief overview of the EndoTerm project, presenting a case study around the concept of Laparoscopic single-site surgery, a type of surgery currently being used within the context of endometriosis. Based on verbal and non-verbal representation, as well as on the input of subject field experts, a set of conceptual maps will be put forward. The final section will consist of some concluding remarks.

2 Terminology and ontology

2.1 Terminology’s double dimension

This approach, which encompasses both a linguistic and conceptual dimension that are interrelated, has been more recently described by Roche (2012, 2015), Costa (2013) and Santos & Costa (2015). According to Roche (2015: 136), Terminology is “both a science of objects and a science of terms”. For Costa (2013), it is precisely this double dimension, and the study of the relationship between one and the other that makes Terminology assume its role as an autonomous scientific subject.

This double dimension approach implies, therefore, that both the experts’ conceptualization of a given subject field and the discourses produced by them must be taken into account. The cornerstone of this approach lies in the complementarity of these two fundamentally different dimensions. Understanding the relationship between the two dimensions is crucial in terminology work, as it will contribute to define a methodology that will not compromise the main goal of a terminological project as it is understood in this paper, which is to represent, organize and share the knowledge from a domain, based on the way it is conceptualized by a community of experts.

Consequently, it is believed that experts are indispensable to terminology work, working collaboratively with the terminologist in the different steps of the project, in order to identify the key concepts of the subject field, as well as the way they relate to each other and how they are represented (cf. Costa et al., 2012).

Nonetheless, and bearing in mind what was described in the introductory section, it is of paramount importance that the terminological products may, at some point, be operationalized, i.e. have a computational representation, and thus a more effective impact on the everyday life of the different target groups within the various subject fields.

The rising interest in the aforementioned conceptual and linguistic dimension, as well as in the subsequent synergies involving Terminology
and ontologies has led to the emergence of new theoretical perspectives\(^7\), one of which being Ontotermontology.

2.2 Ontotermontology: a new approach to Terminology?

Proposed by Roche \textit{et al.} (2009), Ontotermontology aims to reconcile Terminology’s linguistic and conceptual dimensions while maintaining their fundamental differences. Defined as a “terminology whose conceptual system is a formal ontology” (Roche \textit{et al.}, 2009: 325), this approach considers the conceptualization of a given subject field as the starting point of any terminological project, thus corroborating ISO 704’s view that “producing a terminology requires an understanding of the conceptualization that underpins human knowledge in a subject area” (2009: 3).

As mentioned in 2.1, the expert plays an essential role throughout the process. However, Roche (2007) believes there may be risks inherent to the extraction of ontologies directly from texts, since very often, and due to inconsistencies, ellipses, metaphors and other phenomena, the lexical networks extracted from texts may not match the conceptual systems created with the help of the experts – hence, the discourse about knowledge should not be confused with knowledge itself: “Saying is not Modelling” (2007).

This is not to say, though, that natural language should be excluded from terminology work. In fact, “to conceptualize one must verbalize” (Roche, 2015: 149). Resorting to specialized texts is indeed relevant\(^8\), although it must be taken into account that texts do not contain concepts \textit{per se}, but the linguistic usages of the terms that designate them. All in all, specialized texts constitute an invaluable resource to the terminologist, especially in their first contact with a given domain, and the experts can – and should – play a critical role in advising the terminologist as to the texts that are deemed representative and/or mandatory in a given area.

Access to both linguistic and extra-linguistic knowledge is essential to any terminological project, provided the text selection is supported by rigorous criteria and the methodology/ies used are consistent with the type of resource being created, its purpose(s), target group(s) and respective needs\(^9\).

Instead of making them incompatible, the Ontotermontology approach aims to integrate the linguistic and the conceptual dimensions whilst preserving their core identities. This is visible in Roche’s (2012) extension of the classical semiotic triangle by Ogden and Richards (1923), called the “double semiotic triangle” (Figure 1).

![Double semiotic triangle](Figure 1: Double semiotic triangle (Roche, 2012))

In this diagram, it becomes clear that even though both dimensions are present in ontotermological projects, they rely on two distinct semiotic systems and should therefore not be confused. By separating signified (meaning) and signifier (term) – related to Linguistics and natural language – from the concept and its name (identifier) – part of a formal system, Ontotermontology acknowledges a distinction between the definition of the term, written in natural language, and the definition of the concept, written in a formal language\(^10\).

This distinction can be particularly important in subject fields where concepts can be both represented and defined in a non-verbal way\(^11\). Med-

\(^7\)“Termontography” has been developed by the CVC in Brussels within the scope of the FF Poirot European Project and seeks to integrate ontologies in terminology work by combining Ontology Engineering, Terminography and Corpus Linguistics (Kerremans \textit{et al.} (2004); Kerremans & Temmerman (2004); Temmerman & Kerremans (2003)). Despite the fact they do not share the same goals and are based on a different theoretical and methodological framework, comparing these approaches is not the purpose of this paper.

\(^8\)And, in some cases, even indispensable, especially in the legal field, where texts are the pillar of expert knowledge and communication (see Costa \textit{et al.} 2011, 2013).

\(^9\)Santos & Costa (2015) advocate a mixed methodology in terminological work (onomasiological and semasiological), although they argue that the order “is not arbitrary” (p. 176). For knowledge representation purposes, a concept-based approach may constitute a more adequate starting point.

\(^10\)The formal language supporting concept definitions should allow these to be objective (not depending on an individual interpretation), consistent and constructive (allowing the conceptualization to be computationally manipulated) (Roche, 2015).

\(^11\)A more thorough analysis on the role of the non-verbal in terminology and knowledge representation may be found, for instance, in Galinski & Picht (1997); Picht (1999, 2011);
icine is one of such domains: Figure 2 depicts the female reproductive system of a woman suffering from endometriosis, and it includes the extent and location of the disease in terms of lesions and adhesions.

Figure 2: Stage-IV endometriosis (Classification of Endometriosis by the American Society for Reproductive Medicine, 1997)

Far from seeing a mere illustration, a subject field expert would immediately recognize a case of Stage-IV (severe) endometriosis. Rather than being regarded as signs from a Saussurean perspective, the terms that can be identified here (“peritoneum”, “culdesac”, “deep endo”, “complete obliteration”, “dense adhesions”, etc.) should be perceived as signs in the sense of William of Ockham, for whom a sign is “tout ce qui, étant appréhendé, fait connaître quelque chose d’autre” (cf. 1988: 7).12

The potential of the ontoterminological approach, supported by the acknowledgement of Terminology’s double dimension, provides an opportunity to make a contribution to the subject field of Medicine, in particular to <Endometriosis>, allowing the creation of EndoTerm, a multilingual resource that comprises both verbal and non-verbal representation and that can be computationally represented and manipulated.13

### 3 Terminological resources in Medicine

As mentioned above, Medicine is currently undergoing significant changes in what concerns the production, use, storage and dissemination of medical information and, subsequently, medical knowledge. Nowadays, it is somewhat difficult to conceive – at least in some parts of the world – the practice of medicine without computerized medical records, prescriptions, examinations or even procedures, especially with the advent of robotic surgery.

Due to the increasing needs and challenges that have characterized this area over the last few decades, a new discipline has emerged, in the confluence of Information Science, Computer Science and Healthcare: Health Informatics has been defined as “the interdisciplinary study of the design, development, adoption and application of IT-based innovations in healthcare services delivery, management and planning.”

In order to facilitate the computer-based processing and exchange of medical or clinical information among all the stakeholders, that information is represented and organized via a number of terminological products, often grouped under the notion of “terminological system”, with several typologies having been proposed throughout the years (see Table 1).

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Table 1: Typologies of terminological systems.

Used by the ISO/TC215 “Health Informatics”, this umbrella term is characterized as a “set of designations within the domain of health care with, when appropriate, any associated rules, relationships and definitions” (ISO 1828: 2012). Albeit relevant, this definition does not fit the purposes of this paper and the project it aims to present, as it does not address the conceptual di-

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12 Madsen (forthcoming); Roche (forthcoming); Prieto-Velasco (forthcoming).

13 A first glimpse of which can be see in Section 4.
mension of terminological resources and, hence, their ongoing evolution from “simple code-name-hierarchy arrangements, into rich, knowledge-based ontologies of medical concepts”, as noted by Cimino (2001)\(^{15}\).

Concept orientation has been presented in the literature as one of the key principles underlying the creation of today’s (bio)medical terminological resources (see, for example, Chute et al. (1996); Coiera (2003); Duclos et al. (2014); etc.), and was, in fact, one of the twelve requirements, also known as desiderata, that Cimino (1998) believed should support all terminological systems within the medical context in the 21st century\(^{16}\).

In recent years, many (bio)medical terminological resources have been designed or redesigned, in order to incorporate ontology-based elements, such as formal concept definitions, which, in turn, will enable both the operationalization and the aspired interoperability in this field. Yet each resource serves a specific purpose, which, in turn, determines their epistemological principles, core structure, the organization of the various concepts, as well as the language(s) of expression.

One of the initial stages of the EndoTerm project included extensive research of a set of representative (bio)medical resources (e.g. International Classification of Diseases (ICD), Medical Subject Headings (MeSH), Human Disease Ontology (DOID), Unified Medical Language System (UMLS)), to be used as a starting point in the creation of a thorough concept map of the domain in question. One of the following subsections will contain an example of one of these resources and its respective results concerning <Endometriosis>, namely the Systematized Nomenclature of Medicine – Clinical Terms (SNOMED-CT). Firstly, however, it is important to contextualize the concept of departure within our research project.

3.1 Endometriosis: facts and figures

Endometriosis is defined as “the presence of endometrial-like tissue outside the uterus, which induces a chronic, inflammatory reaction” (Kennedy et al., 2005). The exact prevalence of the disease is unknown, but it is believed to affect an estimated 176 million women of reproductive age worldwide (Adamson et al., 2010). While its etiology is uncertain, it is likely to be multifactorial, including genetic, immunological, endocrinological and environmental influences.

Women with endometriosis typically have a range of pain-related symptoms, such as dysmenorrhea, dyspareunia, dyschezia, dysuria, non-cyclical pelvic pain, as well as chronic fatigue (Dunselman et al., 2014). A recent study conducted in 10 countries throughout the world has reported an overall diagnostic delay of 6.7 years (Nnooham et al., 2011). Moreover, the World Endometriosis Research Foundation (WERF) EndoCost study (Simons et al. (2012) has shown that the costs arising from women with endometriosis treated in referral centers are substantial (an average annual total cost per woman of €9579), an economic burden that is at least comparable to the costs of other chronic diseases, such as diabetes, Crohn’s disease, or rheumatoid arthritis.

Surgical procedures play a key role in the diagnosis and treatment of the disease and are often depicted in the form of videos, which is why they were chosen as the focus of the case study to be presented in Section 4\(^{17}\).

3.2 Endometriosis in SNOMED-CT

The Systematized Nomenclature of Medicine – Clinical Terms (SNOMED-CT) is a comprehensive, multilingual healthcare terminology, resulting from the merge of the Systematized Nomenclature of Pathology (SNOP), published by the College of American Pathologists, and the Clinical Terms Version 3 (former Read Codes), designed by the UK’s National Health Service\(^{18}\). When implemented in an application, and due to the Description Logic foundation of this tool, SNOMED-CT enables the representation of clin-

\(^{15}\) It should be mentioned, though, that the boundaries among these different types of resources have become more and more blurred, in such a way that the term “ontology” is often being used indistinctly to refer to all of them. Grabar et al. (2012: 376-377) list several examples from the (bio)medical domain that illustrate “the lack of precise distinction among semantic resources in the literature”.

\(^{16}\) Check Cimino (1998, 2006) for further information on the Desiderata.

\(^{17}\) Laparoscopy plus histology of resected endometriosis is, in fact, considered the “gold standard” in the diagnosis of this condition (Dunselman et al., 2014).

\(^{18}\) It is currently owned and distributed by the International Health Terminology Standards Development Organisation (IHTSDO).
ical content in electronic health formats (e.g. EHR) in a consistent, reliable and computer-readable way.\textsuperscript{19}

The building blocks of this resource are the: i) \textbf{concepts}, representing clinical meanings and organized into hierarchies, ranging from general to specific (with 19 top-level concepts); ii) \textbf{descriptions}, which link appropriate human-readable terms to concepts; and iii) \textbf{relationships}, connecting concepts to other related concepts.\textsuperscript{20} Each one of these three components has their own unique numeric identifier. Figure 3 illustrates the results obtained for \textit{<Endometriosis>} in SNOMED-CT.

In the blue box on the top left corner, it can be seen that \textit{<Endometriosis (disorder)>} is the concept name, and it coincides with the so-called Fully Specified Name (FSN), whereas “Endometriosis (clinical)” is the preferred synonym and “Endometriosis” the acceptable synonym. The Parents and Children elements refer to the “supertypes” and the “subtypes” of the concept in question, linked via |Is a| relationships.

\textsuperscript{19}For more information, see: http://www.ihtsdo.org/snomed-ct; https://elearning.ihtsdotools.org

\textsuperscript{20} The relationships in SNOMED-CT express defining characteristics of a concept and they can be divided into: a) subtype hierarchy relationships (Is a); or b) attribute relationships, which have a particular value provided by another concept, i.e. procedure concepts are linked, for instance, to certain sites.

It should be noted that the subtype concepts are mainly related to the different organs or body parts where the disease can be located (e.g. bladder, intestine, etc.). There are further subdivisions in some of the Children that have not been included due to space constraints.

The final diagram represents the two types of concept relationships associated to \textit{<Endometriosis (disorder)>}, distinguished by colours and types of arrows. The purple concept is an upper-level SNOMED-CT concept, linked to the initial concept by a |Is a| relationship. The yellow bubbles display an attribute relationship [has Associated morphology] and [has definitional manifestation] between the initial concept and \textit{<Endometriosis (morphological abnormality)>} and \textit{<Pain (finding)>}, respectively.

4 Terminology and Knowledge Organization

4.1 The EndoTerm Project

As previously mentioned, the EndoTerm project aims at the creation of a multilingual\textsuperscript{21} terminological resource based around the concept of \textit{<Endometriosis>}. This will be destined to future experts and to experts of other, related domains, mainly for training purposes. One of the objectives is to integrate the resource in an e-learning platform.

Since there are very few specialized texts about this disease in European Portuguese (EP), as most experts publish in English, it is believed a contribution could also be made to enrich the domain terminology in EP and, simultaneously, to improve linguistic quality criteria, which, in

\textsuperscript{21} In English, European Portuguese and French. German might be included at a later stage of the project.
the future, might be applied to other projects involving information retrieval.

Although the inclusion of both verbal and non-verbal elements had already been foreseen in the project, due to the importance of the latter in this particular subject field, the group of experts that have been collaborating in this endeavour suggested the analysis of a type of resource that is becoming more and more important within the medical community: the video article.22

By combining verbal (narration from the expert(s), slides with text, etc.) and non-verbal elements (2D or 3D images, animations, surgery footage), video articles constitute a noteworthy resource to take into account in the light of Terminology’s double dimension. As a new type of scholarly communication that seems to be here to stay, its inclusion in a specialized corpus in a medical terminology project may become inevitable, which will, in turn, pose interesting theoretical and methodological challenges.

4.2 <LESS surgery>: the case study

The case study presented in this paper is based on a video article entitled “Single port laparoscopy”23, which portrays a gynecological procedure – in this case, a hysterectomy, commonly seen as a last resort in cases of severe endometriosis – using a relatively recent type of surgery called single port laparoscopy.

The further study of the concept <Single port laparoscopy> pointed towards a lack of terminological consensus among the expert community. In fact, more than 20 acronyms used to designate this concept have been identified in the literature.24

In order to solve this problem, a multidisciplinary medical consortium25 gathered in 2008 and decided that the term “laparoendoscopic single-site surgery” (also known as LESS surgery) most accurately depicted the surgical procedure in question.

Based on information provided by textual sources, some of which cited below, by the aforementioned video article and others on the same topic, as well as by the feedback from two senior expert gynecologists who are also surgeons, a concept modeling proposal based on <LESS surgery> was created using a software environment for concept system building called OTe (Ontotermology engine) Soft, supported by ontotermological principles (see Section 2.2.).

Designed by the Condillac research team26, this tool has a clear concept orientation, even though the user can also incorporate terms and, thus, the linguistic dimension. OTe Soft is structured around concepts, perceived as knowledge of a plurality of things that “help organize reality by grouping similar objects through what they have in common (Roche, 2015) (e.g. <Laparoscope>). One or more terms may be assigned to each concept, in various languages: i) natural (e.g. “laparoscope” (EN); “laparoscope” (FR); etc.; or ii) formal (e.g. programming language). In addition, a concept may be qualified by attributes, which have a given value, and be assigned one or more instances, also called “things”, i.e. representations of elements in reality (Check Figure 6).

Concepts are linked to each other via concept relations: subsumption (is a) (generic) and composition (part_of) (partitive) are presented by default. However, the tool allows the user to create new concept relations, as long as the logical principles are maintained (e.g. two concepts cannot be linked by the instance of relation). These relations are represented by different colours, in order to facilitate the graph’s visual readability. The final “product” is called model, or semantic network, which can be exported in various formats (json, RDFS or OWL).

The following figures (4, 5 and 6) present examples of micro-concept maps built around the concept of <LESS surgery>: due to possible visual constraints, only partial views are shown here. The first micro-map (Figure 4) aims to po-

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22 For a more detailed description of this new type of resource, see Carvalho et al. (forthcoming).
23 Available at: http://www.fertstert.org/article/S0015-0282%2812%2900387-1/fulltext
25 Called the Laparoendoscopic Single-Site Surgery Consortium for Assessment and Research (LESSCAR), that published a consensus statement with the main conclusions of that meeting (Gill et al., 2010).
26 www.condillac.org
27 One of the challenges of creating a concept-modeling proposal lies, in fact, in defining other types of concept relations that do not fall under the generic or partitive categories. The ISO standards (1087-1:2000 and 704: 2009) lack diversity and systematization, by classifying all the remaining relations as “non-hierarchical” (cf. Nuopponen 2011, 2014).
sition <LESS surgery> within the broader concept of <Surgery>.

Figure 4: The concept of <LESS surgery>

There is a first subdivision presenting <Open surgery> and <Minimally invasive surgery> as subordinate concepts of <Surgery>. In the latter subtype, the subsequent hierarchy-based modeling was constructed through specific differentiation, bearing in mind the Aristotelian definition of genus + differentia: i) with/without external incision; ii) with one incision/with more than one incision. Besides other advantages, such as the operationalization potential mentioned before, this concept modeling strategy constitutes a valuable starting point for the terminologist in the construction of natural language definitions.

On the upper left side, the linguistic dimension is also visible, and it includes the terms associated to the <LESS surgery> concept. In this case, it was decided to list some of the synonyms of the concept identified in the literature: Single-Incision Laparoscopic Surgery – SILS; Single-Site Laparoscopy (SSL); Single-Port Access (SPA); Single-Port Laparoscopy (SPL). Although the image does not show that, the user has the possibility of navigating through the concept network via concepts, terms, or relations. The three images in Figure 4 were added afterwards, as the current version of the OTe Soft tool does not yet allow the user to upload external resources (e.g. images, videos, diagrams, etc.)

Figure 5 explores the types of umbilical incisions that may occur in a LESS surgery, being that the single incision in the umbilicus (navel) is regarded by the expert community as the essential characteristic of the concept, i.e. the characteristic which makes the concept what it is and constitutes its essence (cf. ISO 1087-1: 2000). In this figure, the metaphoric use attributed to the <Omega incision> should also be emphasized.

Figure 5: Types of <Umbilical incision>

Figure 6 contains a more detailed insight on the types of laparoscopes that exist, one of which - <Flexible video laparoscope> - is currently being used to perform LESS surgeries. In this case, the EndoEYE is presented as an instance of this concept.

Figure 6: Types of <Laparoscope>

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28 These, along with other Aristotelian categories, are explored in Porphyry’s Isagoge (2003).

29 This also applies to the images in the remaining figures.
There is also a basic distinction within the expert community between flexible and rigid laparoscopes, as depicted by the map.

5 Concluding remarks

As a scientific discipline in the confluence of several others (e.g. logic, information science, cognitive sciences, linguistics, etc.), Terminology brings an unquestionable added value not only to the study of specialized language in various subject fields but, ultimately, also to the study of how knowledge is represented, organized and shared among a community of practice.

Nowadays, though, that contribution can only be further enhanced if the results of terminological work can be operationalized, i.e. represented in a computational format. Ontologies, in the sense of KE, represent a promising pathway that, however, must be based on collaborative work and on solid theoretical and methodological approaches. By acknowledging Terminology’s linguistic and conceptual dimension and by applying that principle to the creation of tools, multidisciplinary teams integrating both “linguist-terminologists” and “engineer-terminologists” will be able to respond more effectively to the growing needs of expert communities – and, increasingly, of society as a whole.

Medicine is one of the fields where changes are more constant and substantial, and where terminological resources can play an even more vital role. Due to today’s technological progress, it is likely that a sort of “multimedia knowledge base” may become a more and more common instrument in patient and expert education, which is why it is believed that the study and inclusion of non-verbal elements in these resources would represent an important qualitative leap in the joint research involving Terminology and Knowledge Engineering.

References


