ARTÍCULO ORIGINAL

Terminologies and classification systems in biomedical sciences

Terminologías y sistemas de clasificación en ciencias biomédicas

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ABSTRACT

Standards terminologies emerged as an attempt to reduce the diversity terminology in scientific languages, facilitating good communication, which is the basis of all scientific research. This review explains principles and applications associated with terminologies and classification systems focusing mainly on the field of biomedical sciences. The research was conducted on scientific databases, books and network using the keywords: Terminologies, Classification systems, Medical Informatics, Electronic Health Records systems, Interoperability, Ontologies and Bio-ontologies. This review is intended to explain that terminologies facilitate good communication, reducing terminology diversity and they are not static systems. They can "evolve" to more complex structures like biomedical ontologies, with the aim of being used with multiple purposes beginning with the efficient transfer of information, to the processing of information as a result of biological research for its understanding.

Key words: terminology, classification system, interoperability, ontology.

RESUMEN

Las terminologías surgieron como un intento de reducir la diversidad terminológica en el lenguaje científico, facilitando una buena comunicación, que es la base de toda investigación científica. Esta revisión explica los principios y las aplicaciones asociadas con las terminologías y los sistemas de clasificación, centrándose en el campo de las ciencias biomédicas. La investigación fue realizada en bases de datos científicas, libros e internet, utilizando las palabras clave: Terminología, sistemas de clasificación, interoperabilidad, ontologías y Bio-ontologías. Esta revisión tiene por objeto explicar que las terminologías facilitan una buena comunicación, reduciendo la diversidad terminológica y además explicando que no son sistemas estáticos. Ellas pueden "evolucionar" para formar estructuras complejas como ontologías biomédicas, con el objetivo de ser utilizadas con múltiples propósitos que comienzan con la transferencia eficiente de la información, hasta el procesamiento de información obtenida de la investigación biológica para su comprensión.

Palabras clave: terminología, sistema de clasificación, interoperabilidad, ontología.

INTRODUCTION

One of the most common phenomena about language, including medical and scientific language, talks about the existence of several terms to designate the same concept and the fact that the same term holds several meanings. This word variation has been regarded as an obstacle for scientific communication, which led to the appearance of the modern standard terminology.[1,2] Standard terminologies emerged as an attempt to reduce the terminology diversity in scientific languages achieving an effective communication. The construction of a general nomenclature, as a list of terms and glossaries, with the objective of achieving uniformity of terminology, based on the idea that variation is a disservice to communication, is essential to establishing a unique and acceptable terminology for all professional sectors involved in scientific communication. This terminological uniformity facilitates good communication, which is the basis of all scientific research.[3] To solve this problem, just one term would be prioritized over other equivalents, by choosing a single acceptable term to describe a single concept, or by coding through the classification of terms reducing redundancy. The objective is to minimize the terminological diversity, choosing the more descriptive term with the most simplicity and specificity. Minimizing terminological diversity reduces the time and cost of scientific development and avoids the existence of multiple terminologies that represent the same meaning, which is necessary to ensure proper interoperability. For this reason, some terminologies have been developed in order to study, collect and describe particular terms.[4-6] However, while many terminologies have been developed, no single terminology has been accepted as a universal standard for the representation of scientific concepts. So, the goal of this review is to explain principles and applications associated with terminologies and classification systems focusing mainly on the field of biomedical sciences. Here, we mention that terminologies are not static systems. They can "evolve" to more complex structures like biomedical ontologies, with the aim of being used with multiple purposes beginning the efficient transfer of information, to processing of information as a result of biological research for its understanding.

METHODOLOGY

It is an integrative review of published scientific studies without limit of time because it is intended to cover terminologies from its beginnings and its evolution to classification systems, establishing its main definitions and proposing a classification for them. The searching strategy for the identification and selection of studies was through literature research publications indexed in the following databases: Medical Literature and Retrieval System on Line (MEDLINE) and the Scientific Electronic Library Online (SciELO). Update information was also searched on books and networks, to support the searching. Searching in databases was conducted in March 2014 and the studies that are in more than one database were considered only once. Articles and literature review available in Portuguese, English or Spanish were reviewed, containing in their titles and / or abstracts and / or keywords, the following descriptors: Terminologies, Classification systems, Medical Informatics, Electronic Health Records systems, Interoperability, Ontologies and/or Bio-ontologies. Finally, we proceeded to read each summary of obtained articles, and later full article of those who responded to the proposal in this study, to explain principles and applications associated with terminologies and classification systems on the biomedical sciences, in order to organize and tabulate the data destination.

Definition of terminologies and classification systems

Despite International Organization for Standardization (ISO) defining "terminology" as a set of terms representing the system of concepts of a particular subject field, there is still no consensus or single definition. The problem is that each knowledge area discusses their terms differently. *Terms*, for language is a linguistic sign, to philosophy, it is fundamentally a concept, a representation of reality; and to technical and scientific language, *Terms* are units of expression and communication that transfer a specific thought.[7] However, beyond the differences between disciplines, without being strict we could get a consensus and say that terminologies are words that allow obtaining specialized knowledge. On this basis, many authors agree on defining terminologies as a set of words or phrases, called *terms*, aggregated in a systematic manner to represent the conceptual information that makes up a given knowledge domain.[8-10] But here, it is necessary to clarify differences among concept and term: a *Concept* is defined as a unit of thought constituted through abstraction on the basis of properties common to a set of objects, while a *Term* is a designation of a defined concept in a special language by a linguistic expression.[11] It is important to talk about this difference, because these two words are often used interchangeably, as well as terminology and vocabulary. In order to clarify, terminologies consist of basic and specialized sets of words or terms, which have a specific meaning. It means, each term stands for some defined concept. For example, the concepts ''myocardial infarction'' or ''heart attack'' in a given terminology might represent the meaning ''ischemic injury and necrosis of heart muscle cells resulting from absent or diminished blood flow in a coronary artery''.[8] The fact is that terminologies include knowledge about their own contents (definitions) making them more than simple vocabulary, where you can only find a simple list of terms.[12]

Most languages permit words to have the same or similar meanings to permit some flexibility, allowing the same concept to be named in several different ways, for example in healthcare, the concept "Breathless" is defined by multiple terms such as short of breath, breathless, dyspnoea, etc. However, since several terms may be used for the same concept, it is usual to define a single alphanumeric code for every distinct concept in the language. This gives rise to the process of coding, where a set of words (terms) describing some concept is translated into a code for later analysis.[13]

Thus a terminology should contain a separate coding name for each distinct specialized term, as well as any reasonable synonyms. In the same way, terms and codes considered to be similar are collected together into a single category, but, they can quickly become so large that it is difficult to find individual terms. Consequently, they need to be organized in such a way that the terms can be easily searched through, in a way that permits concept-driven exploration, it means, a terminology needs to be more like a thesaurus than a dictionary, organizing terms into conceptually similar groupings. So, one of the most common ways to assist search is to produce a classification hierarchy.[13] This is how, the classification of terminologies gives rise what is known as classification systems. So, it is important to clarify that a terminology is not exactly a classification system. This is because a classification system brings together similar concepts and groups them into categories. In a terminology there is a separate listing and code for every concept. Considering this, a terminology provides a way to input scientific data into a record to cover a particular subject and include the smallest details, whilst a classification system has the purpose of grouping or categorizing details and it is designed for output.[14] It aggregates the details being designed for reporting. Table 1 shows some differences among terminologies and classification systems.

Table 1. Some differences between terminologies and classification systems[14]

TERMINOLOGY	CLASIFICATION SYSTEM
To facilitate electronic data collection. To send and receive data in an understandable and usable manner. To provide an organized system of data collection and retrieval resulting in the linkage of published research.	To allow collection and reporting of basic scientific data. To provide data that is used in monitoring public health and risks. To make available information that can be used to improve scientific, financial and administrative performances.

So, producing a classification hierarchy is important because it provides a structured grouping of ideas, organized around some set of attributes, serving as a map to help locate unknown terms. In this way, the hierarchy begins to provide some meanings to terms through the way are related to others. Such structure is what we find in taxonomies. Relationships between terms of taxonomies are typically monohierarchical, thus each term has exactly one (or no) parent term (Fig. 1 (a)). It means, each element is assigned exactly one parent term.[15,16] A classic example of a strictly monohierarchical taxonomy is the International Classification of Diseases (ICD).[17]

There are many ways in which terms can relate to one another in a hierarchy, depending upon which attributes of the concept are of interest. Some complex terminologies permit multiple axes of classification within them. Ontologies, for example, may also utilize polyhierarchical structures, so that a single vocabulary term may have multiple parent terms, it means one term in a polyhierarchy may have the ability to hold a relationship with more than one term or terms tree (Fig. 1 (b)).[16] Ontologies are intended to describe some given portions of reality.[18] In building them, we can rely on logically defined formalisms which contain special symbols and constructs making it possible to describe reality without the need to depend on human language descriptions that are often very ambiguous.[18] SNOMED CT is considered an ontology because it includes both a defined vocabulary and defined relationships.[19] Nowadays, ontologies have a wide range of uses in many domains, but mainly in biomedicine.[20-23]

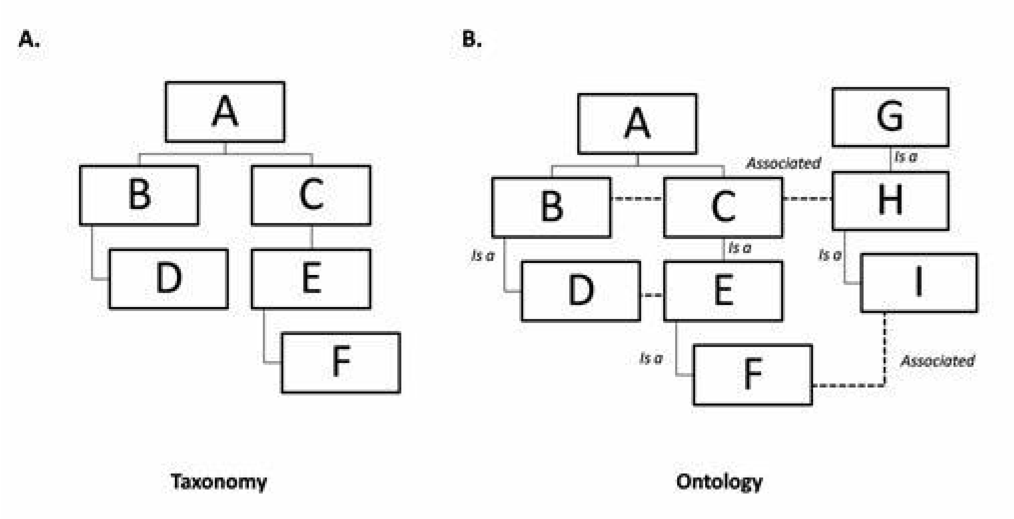

Fig. 1. On taxonomies, each term is assigned exactly one parent term (monohierarchical). In an ontology, one term may be associated with other elements (polyhierarchical, dashed line). A Relationship connect concepts in a hierarchy, while Associated relationships connect concepts in different hierarchies

Then, we are able to say that taxonomies and ontologies provide controlled vocabularies for content description (semantic annotation) of objects through complex data structures that organize conceptual information and establishing relationships among concepts.[23] This relationship between the terms of a vocabulary are stored within taxonomies and ontologies, allowing for an automated search to identify similar content descriptions among a multitude of learning resources, independent of their location. Figure 2 shows briefly an approaching to the classification of terminologies or classification systems mentioned above.

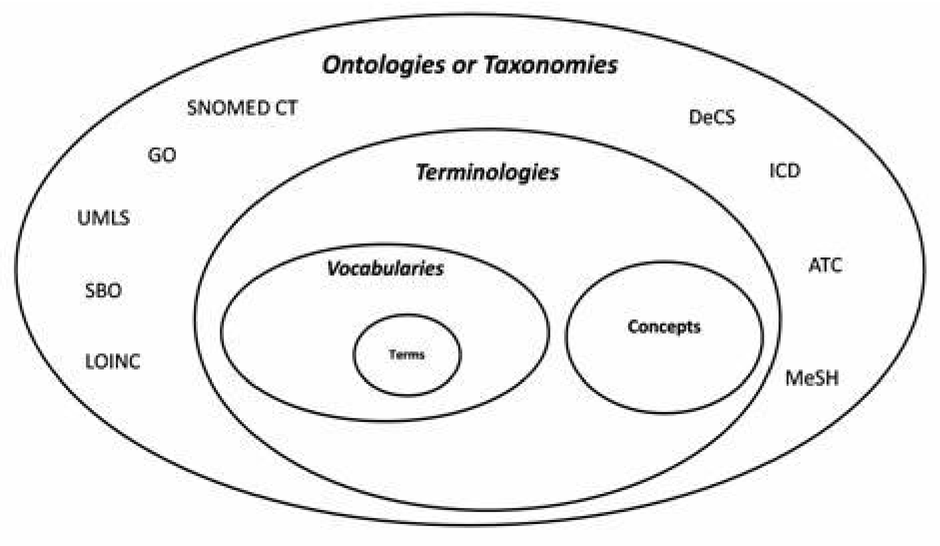

Fig. 2. An approaching to the classification of terminologies or classification systems. Some examples are showed in black color. To the left, there are some examples of ontologies and to the right of taxonomies

In determining the selection or development of a terminology or classification system[14,24,25] it is necessary to take into account that a good terminology or classification system is:

1. Appropriate to its purpose, according to the scientific setting.
2. Implementable to work with existing systems and have appropriate systems for development, maintenance and support.
3. Understandable and acceptable to be used by scientists and other stakeholders.
4. Acceptable to software developers, by supporting key application functions and decision support.
5. Acceptable for statistical and research reporting.
6. Manageable, to improve scientific, financial and administrative performance.
7. Access to complete an accurate scientific data, facilitating electronic data collection.

And with some technical characteristics of:

1. Content, it must include all possible concepts according to the scientific setting.
2. Concept orientation and performance, terms must not be vague, ambiguous or redundant, must be unchangeable.
3. The concept has a unique identifier that carries no other meaning.
4. Formal definitions, there are definitions by association that computers understand.
5. Representation of context, there is formal explicit information about how a concept is used.
6. Changing, it allows for the addition and changing of concepts without breaking rules or making structural changes.

Some clssification systems

While many terminologies and classification systems have been developed, none has been accepted as a universal standard for the representation of biomedical concepts. By contrast, many terminologies and classification systems have been identified by standards organizations as candidates for specific uses. Here, we briefly mention some classification systems, focusing mainly in the field of biomedical sciences and grouping them according to their purpose.

Diagnostic codes

It is healthcare data collected and reported from patients receiving medical care in all settings: inpatient, ambulatory care surgery, observation, outpatient, rehabilitation, skilled nursing, home health, and so on. The clinical health information is converted into coded data that are used for multiple purposes, including planning, making comparisons, determining reimbursement, determining appropriate levels of service, measuring severity of illness. This information allows generating statistics about causes of illness and death. The International Classification of Diseases (ICD) is the world's standard tool to capture mortality and morbidity data. It organizes and codes health information that is used for statistics and epidemiology, health care management, allocation of resources, monitoring and evaluation, research, primary care, prevention and treatment. It helps to provide a picture of the general health situation of countries and populations. The ICD is important because it provides a common language for reporting and monitoring diseases. This allows the world to compare and share data in a consistent and standard way between hospitals, regions and countries and over periods of time. It facilitates the collection and storage of data for analysis and evidence-based decision-making.[17,26]

Procedure codes

It was developed to be used identifying specific health interventions achieved by healthcare professionals. Some examples of procedures codes are the Current Procedural Terminology (CPT) and the **Healthcare Common Procedure Coding System** (HCPCS). The American Medical Association developed CPT as a procedural terminology to describe medical services and procedures performed by physicians and other healthcare providers. CPT is used to report physician services and surgical services. Although it was originally developed as a way to report physician services for reimbursement as accurately as possible, CPT now is used for healthcare trending and planning, benchmarking, and measurement of quality of care as well.[27,28] HCPCS is a collection of codes and descriptors used to represent healthcare procedures, supplies, products and services. HCPCS is divide into three levels, Level I are CPT codes, level II are used for equipment, supplies and services not covered by CPT; level III are local codes that are developed for a geographic region by a Medicare fiscal intermediary when the CPT or national code does not cover a service or supply.[14,29,30]

Pharmaceutical codes

It is used to uniquely identify human drugs, for representing medications, their biological mechanisms of actions, and their physiologic effects. National Drug Codes (NDCs) is a set of medical codes maintained and approved by the Food and Drug Administration (FDA). NDCs are unique number assigned to all drugs and biologics and it is adopted as the standard for reporting drugs and biologics on standard retail pharmacy transactions.[14,31,32]

Another important pharmaceutical code is the Anatomical Therapeutic Chemical (ATC) classification system. The purpose of the ATC system is to serve as a tool for drug utilization research in order to improve quality of drug use. One component of this is the presentation and comparison of drug consumption statistics at international and other levels. In the ATC, the active substances are divided into different groups according to the organ or system on which they act and their therapeutic, pharmacological and chemical properties.[33,34]

Observation codes

The goal is to create different codes for each test, measurement, or observation. It represents a question or assessment which can produce an answer or result. The Logical Observation Identifiers Names and Codes (LOINC) is a common language for clinical and laboratory observations. LOINC covers anything that you can test, measure, or observe about a patient. It has codes for observations like vital signs, hemodynamics, cardiac echo, urologic imaging, gastro endoscopic procedures, pulmonary ventilator management, radiology studies and other clinical observations.[35-37]

Bio-Ontologies

Biology is rapidly changing from a descriptive to a data-driven discipline. As a consequence, ontologies are becoming more and more important for describing the existing biological knowledge, through the large-scale comparison, integration, and sharing of massive data sets.[38]

One of the most well-known ontologies, Gene Ontology (GO), integrates model organism databases to provide descriptions of gene products across organisms using standardized, machine-readable language. The GO project has developed three structured controlled vocabularies (ontologies) that describe gene products in terms of their associated biological processes, cellular components and molecular functions in a species-independent manner. Rather, GO describes how gene products behave in a cellular context.[21,39]

The Systems Biology Ontology (SBO) is a set of controlled and relational vocabularies of terms commonly used in Systems Biology and computational modeling. It provides a set of interrelated concepts that can be used to specify, for instance, the type of component being represented in a model, or the role of those components in systems biology descriptions. It consists of seven orthogonal vocabularies defining: reaction participants roles, quantitative parameters, classification of mathematical expressions describing the system, modelling framework used, the nature of the entity, the type of interaction, as well as a branch to define the different types of metadata that may be present within a model.[40-42]

Other classification systems are not classifiable within the grouping adopted in this paper because they cover a wide range of clinical specialties, include the Systematized Nomenclature of Medicine Clinical Terms (SNOMED CT) and the Unified Medical Language System (UMLS). Here we summarize the purpose of each one.

SNOMED CT is a clinical terminology with global scope covering a wide range of clinical specialties, disciplines and requirements. It is one of the leading biomedical terminologies in use today. This is evidenced, for example, by the fact that it is slated to become an integral component of standardization in health information technology. SNOMED CT provides the core general terminology for the electronic health record (EHR) and contains more than 311,000 active concepts with unique meanings and formal logic-based definitions organized into hierarchies.[43]

The UMLS is a set of files and software that brings together many health and biomedical vocabularies and standards to enable interoperability between computer systems. The UMLS was developed by the National Library of Medicine (NLM) as an effort to overcome two significant barriers to interoperability: the variety of names used to express the same concept and the absence of a standard format for distributing terminologies. The major component of the UMLS is the Meta thesaurus, a repository of inter-related biomedical concepts. The two other knowledge sources in the UMLS are the Semantic Network, providing high-level categories used to categorize every Meta thesaurus concept, and lexical resources including the SPECIALIST lexicon and programs for generating the lexical variants of biomedical terms.[44,45]

The scope of terminologies and classification systems

Terminologies and classification systems are important because they reduce conceptual and terminological ambiguity as they provide a framework for unification. So, they allow exchange of information and facilitating communication between systems with different needs or views according to a particular context. This exchange of data among systems is basically known as interoperability. The National Alliance for Health Information Technology (NAHIT) has defined interoperability as "the ability of different information technology systems, software applications and networks to communicate, to exchange data accurately, effectively and consistently, and to use the information that has been exchange".

Interoperability exists between two applications when one application can accept data from the other and perform a speci?c task in an appropriate and satisfactory manner without an extra operator intervention. To be interoperable, two applications need to agree on: the communication and transport layer, it covers the range from the transport and communication layer protocols; the document layer which involves the format of the exchanged messages and documents, as well as the coding systems used.[46]

There are several real life cases that will benefit from interoperability, such as being able to share Electronic Health Records (EHRs) of patients among different healthcare providers, providing clinical decision support. An EHR is defined as digitally stored healthcare information throughout an individual's lifetime with the purpose of supporting continuity of care, education, and research. The EHRs may include such things as; observations, laboratory tests, medical images, treatments, therapies, drugs administered, patient identifying information, legal permissions, and so on.[47] Whilst the world as a whole is still far from seeing the end to paper records, there has been a very rapid expansion in the last 5-10 years to the point where now in some countries, nearly 90 % of all healthcare records are digital.[48]

Making EHRs interoperable will contribute to more effective and efficient patient care by facilitating the retrieval and processing of clinical information about a patient from different sites and may decrease costs significantly. Transferring patient information automatically between care sites will speed delivery and reduce duplicate testing and prescribing. Moreover, it will reduce errors, improve productivity, and benefit patient care. Furthermore, one of the prominent research directions in the medical field is about using genomics data for improving health knowledge and processes for prevention, diagnosis, treatment of diseases, and personalization of health care.[49]

In recent years, ontologies have become an increasingly important topic in the biomedical field, where they are considered a critical component of multidisciplinary research.[50] Biomedical research is an era of unprecedented large scale data analysis powered by hundreds of public biological databases and hundreds of millions of patient records, so there is a real and urgent need to explore effective methods for biomedical data integration and knowledge management.[51] The necessity of structuring, sharing and reusing the huge volume of data about genes, proteins, diseases, molecular functions, etc, that has been generated during the last years has led to the development of multiple biomedical ontologies.[52] Ontologies are important because they enable true semantic integration across the data sources that they represent, and it is possible to draw wider conclusions from the data and look at the data from several distinct perspectives.[53,54]

This kind of advances in computing and new computational techniques have changed the way researchers approach biology, medicine, and indeed all of science promoting the integration of disparate data sources. Thanks to this, it is now possible to demonstrate computationally correlations among genes, diseases, treatments, and outcomes, to use these correlations to efficiently direct research into potentially fruitful areas, and to translate the insights from this research to the practice of medicine.[55,56]

It is important to highlight that ontologies are usually stored into large-scale repositories available for researchers. One of the best-known biomedical ontology's repositories is NCBO's Bio Portal, which is a Web-based, open repository containing more than 350 biomedical ontologies and terminologies, and this number is continuously growing.[57] The National Center for Biomedical Ontology is another good example. It allows creating and maintaining a repository of biomedical ontologies and terminologies; building tools and web services to enable the use of ontologies and terminologies in clinical and translational research.[58]

Trouble with coding

There are four main obstacles to devising a universal terminological system. The first is the model construction problem, it means that terminologies are simply a way of modeling the world and the world is always richer and more complex than any model human can devise. The second is that terms are subjective; it is the collection of the perceptions, experiences, personal or cultural understandings specific to a person; suggesting that concepts are relative. The third is that terms are context-dependent, so, there is no stable notion of the correct category for objects or events. The best category to describe an object depends on the context within which it is applied. The fourth is that terms evolve over time. Terminologies growth and alteration introduces huge problems of maintenance, and the very real possibility that the system will start to incorporate errors, duplications and contradictions. Introducing changes into a mature terminological system becomes increasingly expensive over time.[13,59]

CONCLUSIONS

It is important for the scientific community to have an uniform and comprehensive approach to the representation of biomedical information, a standard to use in expressing precisely the many details of scientific observation. So, there is a need for a formal language with an associated set of meanings, a syntax that specify how words may be combined, and a semantics that make clear what relations words.

Ontology, considered here as the maximum classification which can reach the terms, can be defined as a formal and explicit specification to represent entities in a particular area and their relationships. The domain ontologies can help eliminate terminological and conceptual confusion generated by specialized languages, but its greatest advantage, unlike terminologies, is the ability to make inferences from explicit knowledge and making possible to draw wider conclusions which can help solve hypothesis or research questions.

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