Towards an ontology for Psychological Disorders

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ABSTRACT

Psychological diagnosis is the process by which mental health professionals determine if problems that affect a person meet all specific criteria for a psychological disorder. In the last years decision support systems (DSS) have helped practitioners in the field of psychological clinical diagnosis with notable results. Given that ontologies are created, among other goals, to allow different sorts of formal reasoning, they are seen as valid artifacts to support a new generation of DSS in mental health professionals. Although in recent years some initiatives have emerged in order to build realist ontologies of mental diseases, this field presents a remarkable heterogeneity of data and two different clinical classification systems that is why authors present an ontology with the aim of reusing existing works and serving as the key element of a clinical DSS in the field of mental disorders.

Keywords: Psychological Disorders; Semantics; Ontologies; ICD-10

INTRODUCTION

The diagnosis of psychological disorders implies a set of practices such as interviews or personality tests that tries to gather all information and data from patients with the objective of getting an accurate evaluation and diagnosis. Trained mental-health professionals have to manage all this information and the sheer mass of data using traditional storage and classification methods that prevent new ways of exploiting this huge amount of knowledge with regards to professional's expertise. Nevertheless in the mental-health domain, several efforts have developed classification schemes to compare collected information and data to existing knowledge organization systems such as the Diagnostic and Statistical Manual of Mental Disorders (DSM-IV). The DSM was designed with the broad objective of sharing statistics among a wide range of mental health professionals (clinicians and researchers) in different contexts such as biological, psychodynamic, cognitive, behavioral, interpersonal or family/systems. For instance, the DSM provides a system of three major components (the diagnostic classifications, the diagnostic criteria sets and the descriptive text) which use identifiers coming from the coding system used by all health care professionals in the United States, known as the International Classification of Diseases, Tenth Edition (ICD-10). In the case of diagnostic criteria, DSM aligns symptoms, disorders and conditions to provide a concise description of each disorder easing the diagnosis process and generating more accurate diagnostics. Finally, each disorder is described using a heading text that improves the understanding of the diagnosis. Some authors (e.g. Bradford, 2010) and initiatives like DSM present their flaws, they underline that there are appropriate reasons to professionally follow and reference these initiatives.

The aforementioned situations led us to a realm in which a huge amount of information and data coming from different sources (patients, existing classification schemes, etc.) with multilingual and multicultural features emerges. Existing knowledge organization systems such as thesauri, taxonomies and classifications systems developed by specific communities and institutions tries to code all this information using standards and bridging the gap between data extracted from the patients out and the domain knowledge, but a common sharing data model and format to automatically exploit this information, apart from exchanging statistics, through standard tools and methods is missing, e.g. one of the key points lies in the explanation and description of the diagnosis.

On the other hand, the Semantic Web initiative through a common sharing data model (RDF) and format tries to raise the meaning of information resources by means of coding domain knowledge and data into ontologies using the web ontology language (OWL) and other specific RDF-based vocabularies. More specifically, OWL enables the use of different logic formalisms to automatically process domain knowledge providing added-value reasoning services for classifying individuals, checking consistency of knowledge bases and inferring new types in the taxonomy. For instance, existing classification systems can be expressed as OWL-DL ontologies with the SKOS and SKOS-XL, RDF-based vocabularies designed to represent taxonomies, controlled vocabularies and thesauri including labeling properties. Moreover, in recent years the Linked Data initiative have emerged as a practical implementation of the Semantic Web vision to connect, publish and share pieces of data, information and knowledge using URIs and RDF on the web and it has been successfully applied to diverse domains such as e-Government, e-Health or e-Tourism supporting the implementation of new enhanced services and linking together information and data from different sources.

Taking into account the requirements and needs of psychological disorders diagnosis and the advantages of using certain technologies in broad diagnosis processes, this paper presents the ontology behind PsyDis (Casado-Lumbreras et al., 2012), a system that supports the decision-making process in mental disorder diagnosis based on the use of semantic technologies.

This paper is structured as follows. In “Related Work”, authors review the different approaches for mixing diagnosis processes and semantic web technologies. In the next Section, the complete description of the ontology for psychological disorders is presented. Finally, the paper ends with a discussion of research findings, limitations and concluding remarks.

RELATED WORK

The term "Semantic Web" was coined by Berners-Lee, Hendler and Lassila (2001) in order to describe the evolution from a document-based web towards a new paradigm that includes data and information for computers to manipulate. Semantic technologies, based on ontologies (Fensel, 2002), provide a common framework that enables data integration, sharing and reuse from multiple sources. In other words, ontologies provide information systems with a semantically rich knowledge base for the interpretation of unstructured content (Mikroyannidis & Theodoulidis, 2010). This approach facilitates the integration of data coming from a broader non-relational domain of data, which, additionally, might be distributed and lie outside enterprise boundaries and control (García, 2010). Taking this into account, according to Alani et al. (2008), Semantic applications are beginning to be pragmatic in several industrial settings (e.g. Colomo-Palacios et al., 2010; García-Crespo et al., 2012; González-Carrasco et al., 2012; López-Cuadrado et al., 2012). This movement can be extended to medical domains. In this way, modern formal ontology facilitates the creation of knowledge-based systems for managing medical information (Sicilia et al., 2009). Moreover, ontologies constitute an important enhancement in the field, since they allow a better representation of biomedical data, enabling more effective queries, statistical analysis and semantic web searching (Viti et al., 2011).

On the other hand Decision support systems (DSS) are computer technology solutions that can be used to support complex decision-making and problem-solving (Shim et al., 2002). DSS uses knowledge and theory from diverse areas such as database research, artificial intelligence, decision theory, economics, cognitive science, management science, mathematical modeling, and others (Kou, Shi & Wang, 2011). The fundamental task for modern DSS is to help decision-makers in building up and exploring the implications of their judgments (French, 2000). In DSS scenario, Clinical Decision Support Systems (CDSS) are information systems designed to improve clinical decision-making (Amit et al., 2005). A formal definition of CDSS can be found in the works of Sim et al. (2001) and is as follows ‘‘software that is designed to be a direct aid to clinical decision-making in which the characteristics of an individual patient are matched to a computerized clinical knowledge base, and patient-specific assessments or recommendations are then presented to the clinician and/or the patient for a decision’’. When using CDSS, the role of the clinical expert is fundamental; CDSS provide support to the decision-making process, but do not indicate the decision to be taken (Ocampo et al., 2011). However, and in spite of the tradition and soundness of such systems, only a few evaluations have been conducted and no definitive conclusions have been reached from the CDSS (Suhasini, Palanivel & Ramalingam, 2011).

Mixing the two aforementioned concepts, namely ontologies and DSS, authors can state that the use of semantic technologies in the field of Medical Diagnostic Decision Support Systems (MDSS) has become a valuable aid in improving the accuracy of medical diagnosis (Rodríguez-González et al., 2012a). Examples of the interaction of semantic technologies and MDSS can be found in the recent literature (e.g. García-Crespo et al., 2010; Rodríguez-González et al., 2011b; Splendiani et al., 2011).

On the other hand, the development of biomedical ontologies is already an active field, and one that is relatively mature in terms of use cases, infrastructures and methodologies (Smith & Scheuermann, 2011). Thus, there are several efforts following this path; Basic Formal Ontology (BFO) (Smith, 2004), the Open Biomedical Ontology (OBO) Foundry (Smith et al., 2007) and The Relation Ontology (RO) (Smith et al., 2005).

From a psychological point of view, the increasing trend of psychological morbidity (Wang et al., 2007) certainly adds to the burden of mental health care providers to offer timely and quality services so as to maintain the health of the community (Wang & Cheung, 2011). Given that mental health problems require costly investigation before diagnosis is reached (Salmon, Dowrick, & Ring, 2004), the need of automated support tools to give faster responses along with cost-cutting is more than evident.

Stemming from this need, several recent and relevant works aim to provide CDSS in the fields of psychology and psychiatry (e.g. Baumgartner, Ferrari & Palermo, 2008; Delgado et al., 2005; Razzouk et al., 2006; Suhasini, Palanivel & Ramalingam, 2011; Toro et al., 2012; Trivedi et al., 2004; Wang & Cheung, 2011; Wang et al., 2007). In this scenario, the importance and influence of ontologies in DSS scenario could provide improve psychological diagnosis.

AN ONTOLOGY FOR PSYCHOLOGICAL DISORDERS

Ontologies are the main cornerstone of Semantic Web technologies. The importance of ontologies lies in the fact that their design allows to define in a formal way a concrete domain. The definition of the domain implies the identification of its entities and their relationships and this is an important feature that should be taken into account when researchers are modeling knowledge representations in the medical domain.

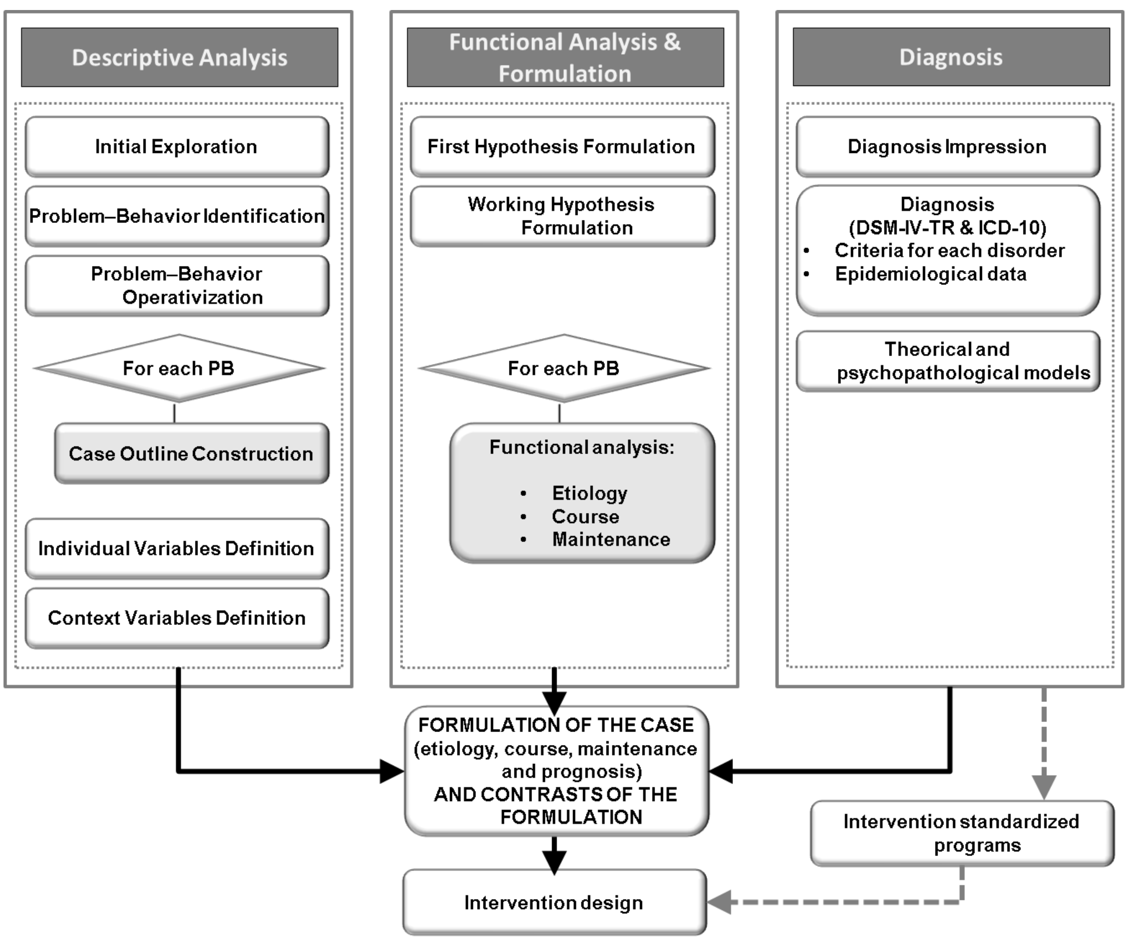
In the case of psychological domain, as have been stated in the related works, there is a lack of standard representation of entities which take part in the psychological disorder diagnosis process and the relation between them. For hence, in this paper, a new model based on ontologies for the representation of these entities is proposed. After studying the related literature in the area, authors can conclude that this lack of representation schemes is a problem in the field of psychology. The identification of main entities and their formal representation can provide several benefits to researchers in the area, having a tool to model and work with the psychological information which takes part in the process of diagnose a psychological disorder.

The work proposed in this paper is the definition of a new medical ontology for the domain of diagnosis in the area of psychology. As was defined in the state of the art, current ontologies for the healthcare domain not always are ready to be reused in other domain and fields due to their complexity or size among other factors. The creation of a light ontology to only represent the medical information which is necessary to perform the process of diagnosis in psychological field is for hence necessary.

As will be shown, the entities which take part in the process of psychological diagnosis are complex and have several relations between them. Furthermore, these entities represent a concept themselves. This means that it is possible to reuse the knowledge of separate entities in future domains or tools. This aspect is quite important because several of the ontologies which have been developed in the last years (not only in medical domain), present this lack: the ontologies are designed in such a way that it is not possible to reuse part of this knowledge. They are designed as a whole. Authors, based on previous works (Neches et al., 1991; Rodríguez-González et al., 2012b) consider that the representations where all the knowledge is presented as an unique entity have a lower reusability than the scheme where the knowledge have been divided in subdomains.

KNOWLEDGE REPRESENTATION

The psychological assessment process proposed by Muñoz (2003), presented in Figure 1, makes use of three different blocks in the process of diagnosing psychological pathologies. These three blocks represents the different type of analysis which can be done in the steps which should be made just before the diagnosis process. These two blocks are important in the decision making process of diagnose a psychological disease. The functional analysis represents an application on the laws of operant conditioning to investigate the relationships between factors and a behavior-problem. This analysis consists in identifying the antecedent and consequent variables, internal and external, which control a problem or disorder, and to establish the relationships between such variables and such disorder. Identifying the problem-behavior´s antecedent and consequences events facilitates the modeling of this phase. On the contrary, descriptive or topographic analysis consists in identifying an individual behavior in the three systems of response that human beings present: behavioral, physiological and cognitive. This identification must include the parameter formulation of such response: frequency, duration and intensity. Therefore, topographic analysis is a complex process that makes modeling task difficult. As a consequence of this, authors have focused on functional analysis, assuming that the problem-behaviors are already defined by means of a full topographical analysis.



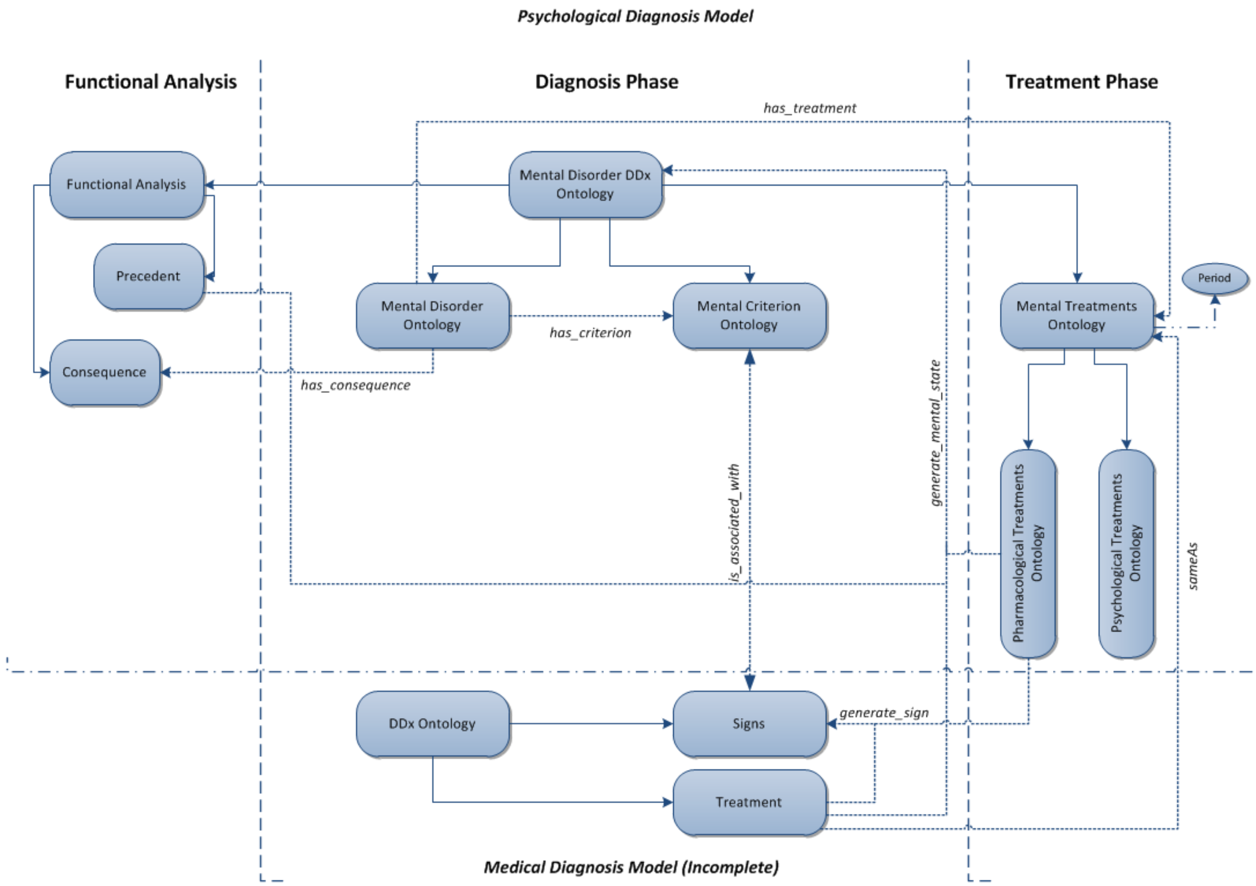
A representation of the whole diagnosis block, with all the analysis that should be made in the diagnosis process of a psychological disease represents a very complex task that involves the use and creation of very specialized concepts and knowledge representation elements such as terminologies, taxonomies or ontologies.

In this paper we have decided to perform a simplification of the model just using, from the psychological domain, the functional analysis and formulation and the diagnosis process. Functional analysis enables us to formulate hypotheses about the etiology, course and maintenance of the behavior-problem. The hypothesis formulation is a consequence of functional analysis in each conventional therapeutic process; but such formulation is not reflected in the model developed in this work. The reason for that is rooted on the difficulty of modeling the formulation process. However, the formulation of hypotheses will used in the diagnostic phase.

An additional phase that should be taken into account given that is quite related with the diagnosis process itself is the treatment phase. This phase establish the adequate pharmacological and psychological treatment which should be followed by a patient. However, the treatment not always implies the end of a psychological disorder, given that some complications can occur. For hence, a psychological treatment can also have relation with a new diagnosis process which is based on the previous diagnosis and the treatment proposed. An important novelty that should be remarked is that the knowledge involved in the proposed ontology also makes use of medical information as will be explained in the next paragraphs.

ONTOLOGY MODEL

The proposed ontology model is depicted in Figure 2. The ontology model has been divided in three main parts which represents some of the blocks of psychological assessment process proposed by Muñoz (2003) (horizontal perspective).



From the vertical perspective we can also see that the proposed model is divided in two models: psychological diagnosis model and medical diagnosis model. In the following paragraphs an explanation about the represented models will be presented, including a definition of the different concepts which appears in the model and their relations.

Models

Psychological diagnosis model: This model represents some of the blocks of the psychological assessment process proposed by Muñoz (2003), concretely, functional analysis and diagnosis phase. This is a high-level semi-complete representation of the main elements which takes part in the diagnosis process, including the treatment phase that as was mentioned before, can take part in the diagnosis process. Some elements such as the sub-relations of the treatment (*has_treatment*) relation such as *has_psychological_treatment* or *has_pharmacological_treatment* have been omitted.

Medical diagnosis model: This is an incomplete model about the medical diagnosis process and the entities which takes part in this process. The medical diagnosis model is based in several works about the development of MDSS (e.g. Shortliffe, 1976; Adlassing et al., 1985; García-Crespo et al., 2010; Rodríguez-González et al., 2011). In this case, the representation is incomplete because only signs concept is represented. The idea behind this representation is that a relation can be established between the consumption of a pharmacological treatment and the appearance of a physic sign. The sign, in this case, is a medical finding which is classified under different schemas than psychological criteria's, and for hence, should be represented using medical models.

Concepts

Mental Disease DDx Ontology: It represents a meta-ontology which imports the remaining ontologies which takes part in the diagnosis process following the work provided by Rodríguez et al. (2012b). Basically, this meta-ontology makes use of the following ontologies:

- **Mental Diseases Ontology**: It defines the psychological disorders.
- **Mental Criteria Ontology**: It defines the psychological criterions used in the diagnosis process.

Functional Analysis: It defines the two main components of the functional analysis in psychological diagnosis process.

- **Antecedent**: It defines the antecedents that can cause a disorder. The antecedents are those events that occur immediately prior to the appearance of behavior-problem, and which are functionally related to it, that is, exerting some influence on it.
- **Consequent**: It defines the consequences that can emerge from the disorder. The consequents are those events that happen immediately after the occurrence of the behavior-problem, and that favors that the problem is not resolved.

For the modeling of the functional analysis, a new meta-ontology with the content of the concepts associated to antecedents and consequences have been designed. The model has been designed using a hierarchy where the antecedents and consequences are mainly categorized represented by its type (intern and extern). Nowadays there is no classification of the antecedents and consequences; for hence, a list of some antecedents and consequences based on their description has been added to the current model. The items of the functional model also contains information such as if the item is cognitive, physiological or motor.

Mental Treatments: It defines the two main types of treatments that are available in the diagnosis process.

- **Pharmacological Treatments Ontology**: It defines the pharmacological treatments.
- **Psychological Treatments Ontology**: It defines the psychological treatments.

DDx Ontology: It defines the meta-ontology of medical diagnosis (see Rodríguez-González et al. (2012b)). The only element of this meta-ontology is the concept Signs, which represents the signs in the medical field.

Relations and DataTypes

In the following lines the different relations and data types of the model will be explained. The relations marked with an asterisk (*) are not represented in the model because a high-level representation has been made; however, in the developed ontology these relations have been created. These relations are sub relations of another relation which is represented in the diagram.

- *has_treatment*: It defines the relation between a disorder and its treatment. It is a non-functional relation because a disorder can have several treatments. It has two sub-relations to define the type of treatment.

- *has_pharmacological_treatment (*)*: It is used to identify those relations between a disorder and a pharmacological treatment.
- *has_psychological_treatment (*):* It is used to identify those relations between a disorder and a phsychological treatment.

- *has_criterion*: It defines the relation between a disorder and the criterions that help to identify the disorder. It is a non-functional relation because a disorder can have several criterions.
- *is_associated_with*: It defines the relation between a psychological criterion and a physical criterion (sign) and vice versa. It is a non-functional relation due to a sign can be associated to several psychological criterions. The idea of this relation is allow identifying a psychological criterion from a physical reaction. For example, a dyspnea process can be related with an anxiety state.
- *generate_sign*: It defines the relation between a treatment (pharmacological[1] or medical) and a physical criterion (sign). The idea of this relation is allow knowing that a pharmacological treatment (from the medical or psychological perspective) can trigger a physical reaction on the patient. This reaction can be confused or related with a psychological mental criterion. With these relations it is possible to identify that, for example, a new criterion on a patient which is under treatment could be caused by a physical reaction to the treatment, and not because it is a new problem. It is a non-functional relation because a treatment can generate several signs.
- *generate_mental_state*: It defines the relation between a treatment (pharmacological or medical) or antecedent and a mental state (psychological criterion or psychological disorder). The idea of this relation is allow knowing that a treatment (pharmacological or medical) can trigger a new psychological problem (from the treatment perspective). From the functional analysis perspective, this relation is used to establish a relation between a precedent and a mental state. With this relation it is possible to identify that, for example, a new criterion on a patient which is under treatment could be caused by a psychological reaction to the treatment, and not because it is a new problem (in fact, it is a new problem, but the origin can be isolated). A good example in this case is the use of Alprazolam as pharmacological treatment for anxiety. An adverse effect of this treatment is the major depressive disorder. It is a non-functional relation because a treatment can generate several mental states.
    - *generate_mental_criterion (*)*: It is used to identify those relations between a treatment or antecedent and a psychological criterion. For example: A treatment that generates anxiety.

[1] When authors make reference to pharmacological treatment we are referring to the use of drugs in the treatment of a psychological disorder. In the case of medical treatment we are making an assumption about the use of drugs for the treatment of a concrete disease.

    - *generate_mental_disorder (*)*: It is used to identify those relations between a treatment or antecedent and a psychological criterion. For example: A treatment that generates depression (see example of Alprazolam).

- *sameAs*: The built-in OWL property *owl:sameAs* links an individual to an individual. Such an *owl:sameAs* statement indicates that two URI references actually refer to the same thing: the individuals have the same "identity" (W3C, 2004). In this case we use *sameAs* to identify pharmacological treatments (drugs) in the psychological domain that are the same than in the medical domain.

- *has_consequence*: Relation to indicate that a mental disorder can have one or several consequences from the functional analysis point of view. It is a non-functional relation because a mental disorder can have several consequences.

With regards to DataType properties, only one property have been defined: *Period*

Period property is used to represent the period where the treatment was prescribed. The intention of this property is allow when a concrete treatment was prescribed in order to know if for example new mental states can be as a consequence of this treatment and for hence avoid new prescriptions of the same treatment.

Table 1 is a summary of the relations, including their domain, range and type (functional or non-functional).

Relation	*Domain*	*Range*
has_treatment	Mental Disorder	Mental Treatment
has_pharmacological_treatment	Mental Disorder	Pharmacological Treatment
has_psychological_treatment	Mental Disorder	Psychological Treatment
has_criterion	Mental Disorder	Mental Criterion
is_associated_with	Mental Criterion Signs	Signs Mental Criterion
generate_sign	Pharmacological Treatment Treatment	Signs
generate_mental_state	Treatment Antecedent Pharmacological Treatment	Mental Disorder DDx
generate_mental_criterion	Treatment Antecedent Pharmacological Treatment	Mental Criterion
generate_mental_disorder	Treatment Antecedent Pharmacological Treatment	Mental Disorder
has_consequence	Mental Disorder	Consequence

CONCLUSIONS AND FUTURE WORK

Heterogeneity of data in the mental health domain is a combination of a plethora of assessment methods and two clinical classification systems with no formal method of interconversion (Kola et al., 2010). Recent and relevant works proposed mental disease ontologies aimed to be consistent with other biomedical ontologies (Ceusters & Smith, 2010). In this work, authors present an ontology for psychological disorders that is aimed to act as the cornerstone of PsyDis (Casado-Lumbreras et al., 2012), a clinical diagnosis DSS. This paper presents the ontology underlying PsyDis illustrating the main features of its design and implementation.

Authors firmly believe that a well-designed ontology can serve as a good starting point in addressing some of the interoperability challenges that many actual systems present as well as a more coordinated diagnosis and treatment among practitioners.

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