

PsyDis: towards a diagnosis support system for psychological disorders

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ABSTRACT

Psychological diagnosis is not a simple task. In spite of the amount of information and decision support systems available, mental disorders are still difficult to diagnose due to their intrinsic lack of formal boundaries. This paper presents PsyDis, a tool aimed to support the decision-making process in mental disorder diagnosis. PsyDis combines ontologies and logical inference mechanisms to offer decision support in the field of psychological clinical diagnosis. The system has been evaluated by means of two different studies. Results show notable accuracy of the system in terms of precision, recall and f measures.

Keywords: Decision Support System; Psychological Disorders; Semantics; Ontologies

1. INTRODUCTION

Psychological problems present a huge burden of illness in our community (Hutton & Gunn, 2007). However, most mental disorders lack a single, universally-acknowledged pathogenesis, which in the past led to unreliability among clinicians in diagnosis (Kim & Ahn, 2002). The diagnosis can be defined as the “description of a health problem in terms of known diseases” and the diagnostic process as a “set of actions needed to obtain the diagnosis” (Van Bemmelen & Musen, 1997). Nowadays there are several efforts that, in order to help diagnosis, code diseases and signs, symptoms, abnormal findings, complaints, social circumstances and external causes of injury or diseases in the psychological arena (e.g. World Health Organization, 1992). However, in spite of these efforts, the work of diagnosis is still a complicated task. For instance, according to the American Psychiatric Association (2000) there can be no assumption that each category of mental disorder is a completely discrete entity with absolute boundaries dividing it from other mental disorders or from non mental disorder. Mental disorder diagnosis, according to Widiger and Samuel (2005) presents several dilemmas that decision-support systems are aimed to mitigate. This paper presents PsyDis, a psychological disorder diagnosis support system based on semantic technologies and logic-based inference. PsyDis can be seen as a relevant contribution

since the area of psychology systems has not been widely mentioned in the literature of diagnosis systems, in contrast with other sets of pathologies.

The paper consists of six sections and is structured as follows. Section 2 reviews the relevant literature. Next psychological diagnosis process is depicted. Section 4 discusses the main features of PsyDis, including the architecture and internals. Section 5 describes the evaluation of tool performance including a description of the sample, the method, results and discussion. Finally, the paper ends with a discussion of research findings, limitations and concluding remarks.

2. RELATED WORKS

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).

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. Consequently, all organizations, including medical organizations, need to constantly improve their competitive advantage and respond faster to changing markets by reducing costs, improving quality and increasing productivity (O’Sullivan & Dooley, 2010).

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; Trivedi et al., 2004; Wang & Cheung, 2011; Wang et al., 2007). Therefore, the importance and influence of semantic technologies in DSS scenario could provide wider and more accurate solutions to psychological diagnosis.

The “semantic”, as an IT research field, was born in the early 2000’s (Álvarez Sabucedo et al., 2010) prompted by Tim Berners-Lee (Berners-Lee, Hendler & Lassila, 2001). Semantic technologies, based on ontologies (Fensel, 2002), provide a common framework that enable data

integration, sharing and re-use from multiple sources. The use of semantic support in IT-based solutions allows the introduction of “intelligence” in software based systems, making it possible to introduce computer based reasoning enabling process automatization (Álvarez Sabucedo et al., 2010) and the performance of sophisticated tasks (Durguin & Sherif, 2008).

Several authors have highlighted the importance of semantic technologies in organizations. Ding (2010) stated that the semantic web is fast moving in a multidisciplinary way and Breslin et al. (2010) confirmed that industry is adopting semantic technology applications. Thus, the health domain is permeable to this movement. There are several attempts in the literature to describe the use of semantic technologies in the field (e.g. García-Sánchez et al., 2008; Jalali & Borujerdi, 2011; Patel et al., 2010; Valencia-García et al., 2008). Specifically, according to Fuentes-Lorenzo, Morato & Gómez-Berbís (2009), semantic technologies can be exploited to reveal machine-readable latent relationships within specific diagnostic-related information in the medical field, where the homogeneity of terminology is particularly problematic. In this way, modern formal ontology facilitates the creation of knowledge-based systems such as those required for managing medical information (Sicilia et al., 2009). Thus, the use of semantic technologies in the field of Medical Diagnostic Decision Support Systems (MDSS) has become a valuable aid for improving the accuracy of medical diagnosis (Rodríguez-González et al., 2012b). 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., 2011) to cite some of the most recent and relevant cases. However, to date, no efforts have been devoted to combining the advantages of semantic technologies with the clinical psychology domain.

The aim of this paper is to present PsyDis, a tool that combines Semantic Technologies (and, specifically, ontologies) and inference mechanisms to offer decision support in the field of clinical psychology diagnosis.

3. PSYCHOLOGICAL DIAGNOSIS PROCESS

Psychological Assessment can be defined as the process of evaluation and measurement of psychological factors, biological and social relationships in a person or group of persons with possible psychological disorders. Specifically, diagnosis is the process by which practitioners determine if the problems that affect a person meet all specific criteria for a psychological disorder, which are specified in the diagnostic manuals (DSM-IV-TR and ICD-10). Therefore, diagnosis is understood as an aspect of the overall process of psychological evaluation. In other words, evaluation includes diagnosis as one of its activities.

This differentiation between the overall assessment process and the diagnostic process is relevant to the work presented in this paper. Specifically, PsyDis provides support during the diagnostic activity.

Psychological Assessment is a complex process that has received many influences throughout its history. Currently, two initiatives strongly influence this process. On the one hand the Diagnostic and Statistical Manual of Mental Disorders (DSM) (American Psychiatric Association, 2000), sponsored by the American Psychiatric Association (APA), and on the other hand The International Statistical Classification of Diseases and Related Health Problems which in its last revision is known as ICD-10 (World Health Organization, 1992). These two manuals have decisively changed the evolution of the classification of mental disorders and dramatically influenced the psychological assessment process.

In these manuals, for each disorder there is a group of basic (mandatory) criteria, along with other possible criteria that a given disorder could present. Therefore, we speak of criteria rather than symptoms, and emphasize the proper observance of these criteria. This approach greatly facilitates the conjugation of nomothetic diagnostic categories along with idiographic diagnostic applications.

The literature has reported some criticism of these two initiatives (e.g. Beutler & Malik, 2002), however their advantages outweigh the disadvantages. In fact, the level of agreement reached by professionals on the use of DSM-IV-TR and ICD-10 is higher than that achieved by any previous method. Therefore, the use of any of these classification systems is essential for any global clinical evaluation process. In PsyDis, given that there are documented correspondences between both initiatives, the authors chose ICD-10.

The psychological assessment process proposed in Figure 1 represents an integrative model based on the works of Muñoz (2003). This model presents three main components: Descriptive analysis, Functional analysis & formulation and, finally, Diagnosis. The differentiation between the three proposed components cannot be taken sequentially. In fact, the activities under each of them develop in a dynamic and continuous process.

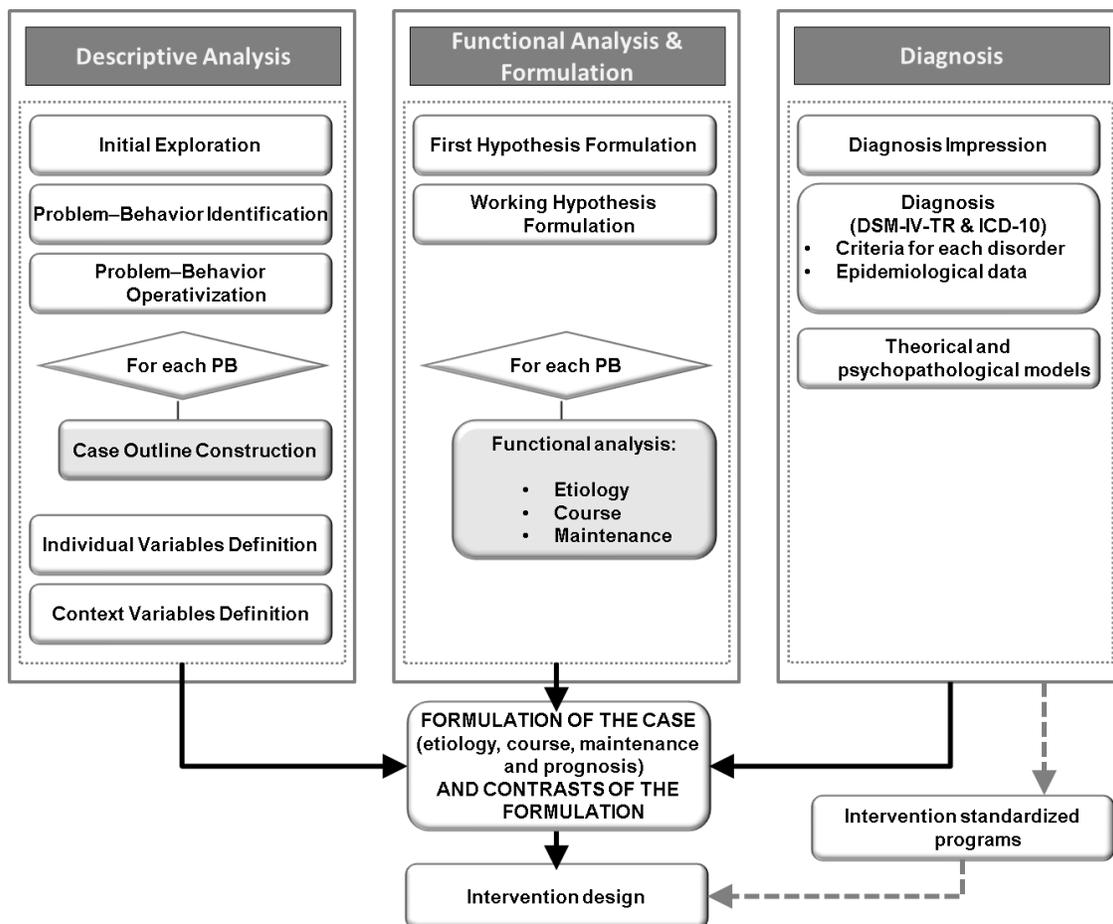


Figure 1. Psychological assessment process

PsyDis focuses upon the Diagnosis component. More precisely on the Diagnosis process itself. This tool, by means of semantic technologies, taking into account criteria described in ICD-10, suggests a disorder or set of disorders that could fit criteria provided.

4. PSYDIS: INTERNALS AND ARCHITECTURE

Ontologies are the main cornerstone of Semantic Web technologies. The importance of ontologies resides in the fact that their design allows formal definition of a concrete domain. The definition of the domain implies the identification of its entities and their relationships. This is an important feature that should be taken into account when researchers are modeling domains.

For this reason, PsyDis is based on the premise that Semantic technologies and ontologies in particular are the perfect scenario for modeling psychological entities and their relations. Moreover, the use of ontologies as knowledge-base allows us to use Semantic reasoners to make inferences and obtain new knowledge in the form of diagnosis procedure.

The main architecture of PsyDis is based in the components depicted in Figure 2.

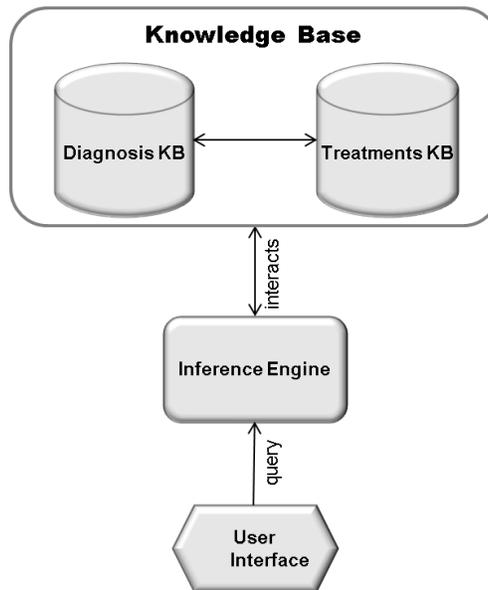


Figure 2. PsyDis main architecture

As can be seen in Figure 2, the architecture of PsyDis is quite simple. Our approach is focused on a more detailed and complex organization of the knowledge base (which will be decomposed in the following sections) than in having a complex main architecture. Figure 3 shows an extended approach of the architecture, including the relations with the treatments which should be consulted using a SPARQL query.

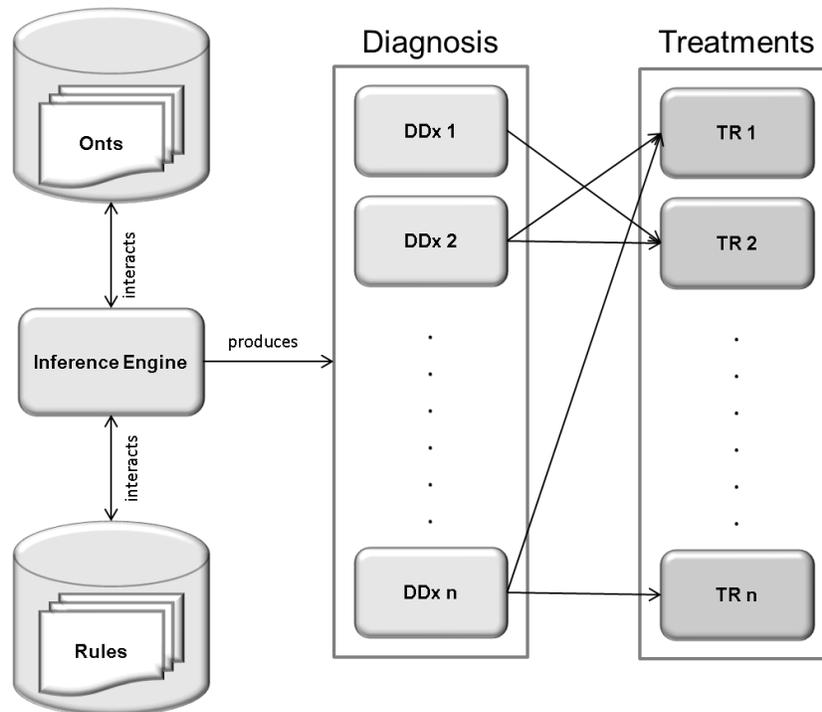


Figure 3. PsyDis extended architecture

User Interface (UI) component represents the component which is shown to the final user. This component allows the introduction of the clinical data which is stored in the knowledge base.

Inference engine is the component which is in charge of reception of the inputs introduced in UI and generates the queries that will be thrown against the Knowledge Base component.

Finally, the Knowledge Base component represents two main Knowledge Bases that should be explained separately. These knowledge bases contain all the clinical information that plays a part in the diagnosis of a psychological pathology and information about the treatments which should be followed depending on the type of pathology diagnosed.

DIAGNOSIS KNOWLEDGE REPRESENTATION

The representation of the knowledge involved in the presented domain is one of the main challenges of this paper. It is important to define the clinical entities which take part in the diagnostic process of a psychological pathology, as well as to identify their associated treatments in order to correctly define the relations which exist between the different entities.

Another important aspect is the definition of the hierarchy which will define the taxonomy of the concepts that have previously been isolated, providing an ontological model of the domain.

The aim of this section of the paper is to show the main decisions made in the modeling of the knowledge involved in the diagnosis process. These decisions include ontology design and diagnosis process design (multiple diagnosis and unique diagnosis).

DOMAIN ONTOLOGY MODELING

There are several approaches which have been followed in the representation of medical information using semantic technologies. The main approaches, which are widely known, are mainly OBO-Foundry (Smith et al., 2007) and Open GALEN (Rector et al., 2003) initiatives. These initiatives, arising from the intention of covering the majority of existing knowledge in the medical domain, have been widely used, given the vast amount of information that they cover. However, it is precisely this vast amount of information that could be considered one of the main failings in these initiatives.

As has been set out in previous studies (Wroe, 2006), there are several sub-domains where only a small fragment of knowledge from bigger knowledge representations (such as OBO-Foundry or Open GALEN) is needed to achieve a better understanding and representation of the domain.

For this reason, following previous studies such as that presented by Rodríguez et al. (2012a), the ontology modeling aspect will be focused upon the division of the psychological diagnosis domain into several sub-domains where each domain will represent the different entities that participate in the diagnosis process.

The main approach found in the representation of psychological disorders from an ontological perspective is the work done by Hadzic et al. (2005) where taxonomy of mental health disorders has been developed.

In the case of the present work, we have developed a new approach to cover more psychological entities than in previous studies. As previously mentioned, there are several entities which have been identified in the diagnosis of mental health disorder processes and, for this reason, a new design of current ontology models is needed.

In the proposed model, we follow the main design principles presented by Rodríguez et al. (2012a), adapting our domain to several sub-domains, linking each one to the entities involved in the diagnosis process. These entities are the following:

- **Disorder:** In classical medical diagnosis the entity disorder is represented by other names such as pathology or disease, among others. The disorder is the final entity which will represent the disease suffered by a patient. In the case of mental health (psychology or psychiatry domain), we represent this entity with the name of disorder.
- **Criteria:** The criteria entity is the representation of a finding which allows the physician to discover the pathology affecting a patient. In classical medical diagnosis this entity is normally represented as “finding” entity. This entity normally covers other more specialized entities such as symptom or sign.
- **Treatment:** The treatment entity represents a psychological or psychiatric treatment. A treatment makes reference to the attempted remedy of a health problem. The treatments are applied after the diagnosis process.

Figure 4 shows the main ontology design hierarchy.

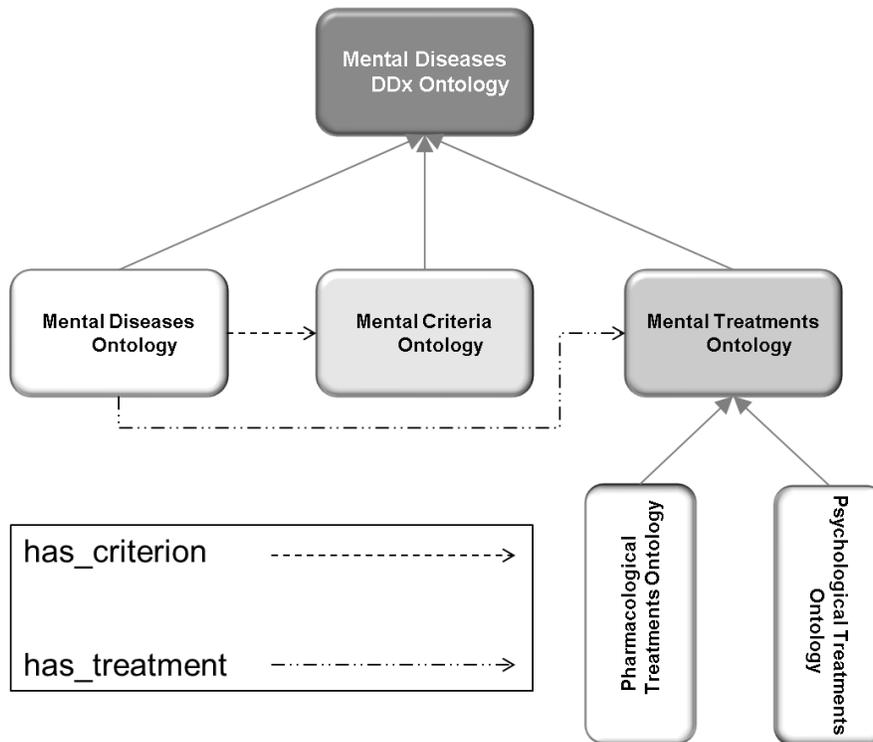


Figure 4. Ontology hierarchy model

DIAGNOSIS MODELING

The modeling of the diagnosis process could be addressed from two main points of view: unique diagnosis and multiple diagnoses.

Each type of modeling could be carried out using Description Logics (Baader et al., 2003), which are part of the main languages usefully defining restrictions and modeling knowledge in Semantic Technologies. However, the main problem of this type of modeling is the difficulty of correctly defining the restrictions associated with the model.

In the following sections the modeling associated with each available viewpoint will be explained.

An important preliminary re this modeling that should be taken into account is that the modeling of the entities which participate in the reasoning process from Description Logics perspective was addressed introducing restrictions on the classes. As was mentioned in the domain ontology modeling section, the ontology has been designed according to two main conditions:

1. The entities which represent a certain mental disorder should be represented by a class, not as an instance.
2. The criteria will be represented as instances in the ontology. Classes will only be used to define the root elements to which the instances (criteria) belong.

These two conditions will be applied in the modeling of multiple and unique diagnosis.

Another important feature that should be taken into account from the OWL modeling perspective is that all the restrictions applied should be carried out using necessary and sufficient conditions.

In OWL modeling we have two choices when we are describing a class (this description will allow the reasoner to compute the new hierarchy of the model or classify instances depending on the restrictions applied). The first one, necessary conditions, says that If an individual is a member of 'NC' (NC being the class we are describing), then they must satisfy the conditions (the descriptions applied). However, if some individual satisfies these necessary conditions, we cannot say that they are a member of 'NC'. In other words, A sufficient condition is one that, if satisfied, assures the statement's truth.

However, in necessary and sufficient conditions, if an individual is a member of 'NC' then they must satisfy the conditions. If some individual satisfies the conditions, then the individual must be a member of 'NC'.

This second definition about necessary and sufficient conditions is the one which is interesting in this case. We want to ensure that if some (random) individual satisfies the conditions defined in the class NC, then, this individual should be a member of 'NC' and then the classifying process will give us the desired results.

MULTIPLE DIAGNOSIS

In the context of modeling a psychological entity, with the relations to the main elements (criteria) that allow diagnosis, we have the concept of multiple diagnoses. This concept could be defined as a modeling paradigm where the knowledge base that contains the relations between elements which intervene in a psychological diagnosis process have been modeled in such a way that the system is capable of diagnosing more than one element. In this definition we should include as a key concept that the appearance of extra criteria does not act as a discriminant to discard diseases.

The concept of a criterion acting as discriminant has been mentioned in previous works where diagnosis entities have been modeled (García-Crespo et al., 2010; Rodríguez-González et al., 2011). The idea of the use of discriminants in this kind of modeling is based on non-monotonic logic. A non-monotonic logic is a formal logic whose consequence relation is not monotonic. Most studied formal logics have a monotonic consequence relation, meaning that adding a formula to a theory never produces a reduction of its set of consequences.

An example of this in the studied domain is shown in figure 5.

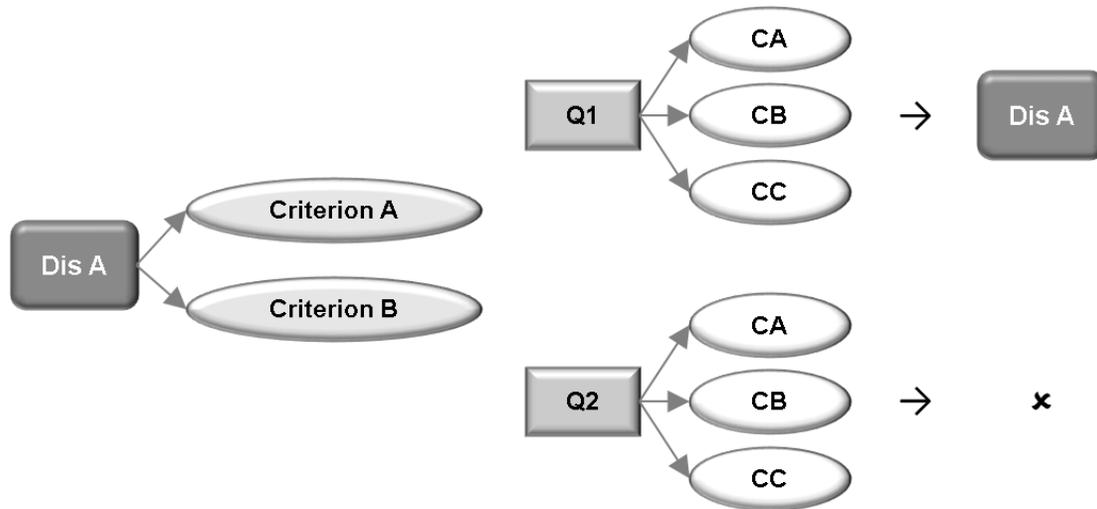


Figure 5. Discriminant example.

In Figure 3 we can see a hypothetical definition of a concrete disease (Dis A) which is based on two criteria (Criterion A (CA) and Criterion B (CB)). If the system was designed to work with discriminants, the result of a hypothetical query which contains as input parameters the criteria A, B and C will return no results. However, if the system was designed not using these kinds of discriminant, it will return Disease A as a result.

The idea behind the use of discriminants is to allow the system to perform multiple diagnoses. When the discriminant concept is not used we are stating that a criterion cannot be used to discard pathologies. This is quite useful if we wish, for example, to design a system which will be capable of diagnosing multiple diseases. Behind the concept of multiple disease diagnosis it is necessary to clarify that this concept refers to the ability of the system to diagnose more than one possible pathology occurring at the same time in a specific patient.

However, it should be noted that the ability of a system to discover multiple pathologies in a patient is a really hard task that should be improved from this schema. The schema provided could be used to establish the bases for developing this kind of system, but cannot be directly used in real environments.

Description Logics modeling

Given the conditions previously described re the ontology modeling performed, we can explain the special features that describe the modeling of multiple diagnosis approach using Description Logics.

In the case of multiple diagnoses, we are using Open World Assumption (OWA), which comes by default with OWL. In formal logic, the open world assumption is the assumption that the truth-value of a statement is independent of whether or not it is known by any single observer or agent to be true. It is the opposite of the closed world assumption, which holds that any statement that is not known to be true is false. The OWA is used in knowledge representation to codify the informal notion that in general no single agent or observer has complete knowledge, and therefore cannot make the closed world assumption.

Given that we are not using discriminants, we are still working with OWA, and this implies that if we don't describe the concrete set of criteria related with a mental disorder (specifying that

only these criteria and any other are part of the mental disorder), the reasoner assumes by default that other criteria could be part of the mental disorder, and therefore allows the inference of more than one disorder even using discriminants.

In the Psychological diagnosis process section, we state that following the information provided by WHO and DSM-IV-TR, a mental disorder can only be diagnosed when it fulfills concrete requirements. Between these requirements a restriction exists that establishes that the patient should present at least five criteria (in the case of DSM-IV-TR) or three criteria (in the case of WHO) from a criterion list related to the mental disorder for diagnosis.

Thus, the main restriction on diagnosis of a mental disorder is that the patient should present at least three criteria which are pre-defined. In fact, the WHO and DSM-IV-TR also establish a fixed list of criteria which are related with any mental disorder, but, for simplification purposes we will not mention them here.

This restriction could be modeled from a Description Logic point of view using qualified cardinality restrictions (QCR) from OWL2.

The modeling consists of creating a class that will embrace the criteria that can determine a concrete disorder. Imagine for example that we have a mental disorder called “MDA” which has as diagnosis criteria the following criteria: CA, CB, CD, ... CF.

The graphical representation of the cardinality restriction (at least 3 elements) to be able to diagnose the disorder MDA is represented in Figure 6.

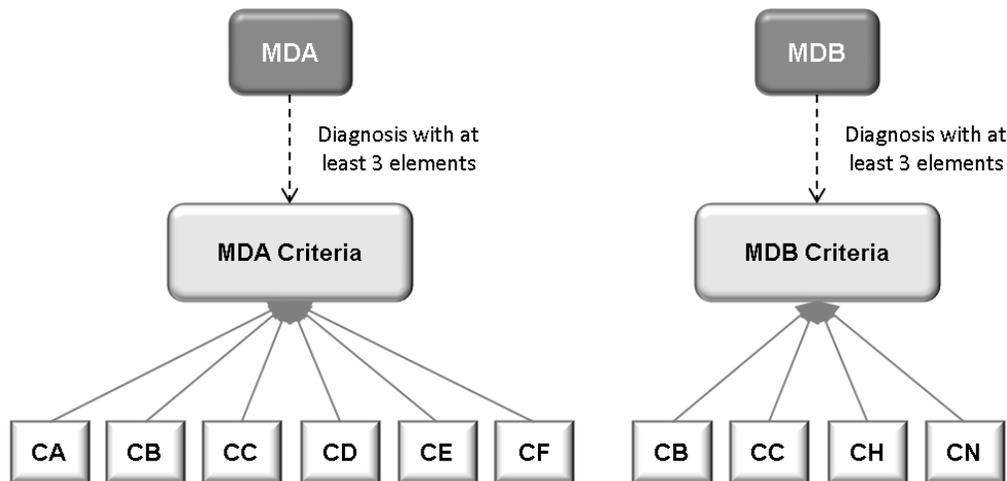


Figure 6. MDA and MDB Diagnosis example

In this case, CA to CF are common criteria which can be applied to any mental disorder. The definition of MDA implies that to diagnose MDA we need to have at least five of the elements contained by “MDA Criteria”. However, this does not imply that the elements CA to CF cannot belong to other criterion, as we saw in the definition of MDB. MDB in this case is defined by some of the elements used by MDA (CB, CC) and some other elements (CH, CN).

This representation allows us to show that the criteria are defined as belonging, at least, to a concrete class (Criteria class, which is defined in Criteria ontology). However, the criteria can also belong to other classes. This is quite important because it is necessary to define a super class which will embrace the specific criteria that will be used to define a certain mental disorder.

Coming back to the representation of this in Description Logics, this should be carried out as is presented in Code Listing 1 and Code Listing 2.

Code Listing 1 shows the formal definition using description logics of the class “MDA” to restrict that an instance will only be classified (inferred, diagnosed) as belonging to MDA only if it fulfills the cardinality restriction.

```
has_criterion min 3 "MDA Criteria"
```

Code Listing 1. MDA Criteria Restriction

```
has_criterion min 3 "MDB Criteria"
```

Code Listing 2. MDB Criteria Restriction

Note: In formal OWL Syntax “MD* Criteria” class should be written as “MD*_Criteria” or similar, with no spaces.

We should apply this schema to all the disorders stored in the knowledge base. It is important to define a “Criteria” class to establish that a concrete criterion (instance) belongs to this class, and then, link this criteria class with the disorder through the cardinality restriction shown in Code Listing 1 and Code Listing 2.

The building blocks of DL knowledge bases are concepts (or classes) representing a set of objects, roles (or properties) to establish relationships between objects and individuals in order to define specific entities. In this modeling approach, concepts such as Criteria, Treatment or Disease are called atomic. The use of concept constructors allows us to model complex concepts that describe the conditions of concept membership. For instance, the concept *∃has-criteria.Histrionic* describes those entities that are related through the *has-criteria* role with an object from the concept Histrionic. DLs provide a rich set of concept constructors such as Boolean connectives, existential and universal quantification and number restrictions (qualified). A DL knowledge base O is typically comprised of a TBox T and ABox A . A TBox contains axioms about the general structure of all allowed worlds similar to a database schema. An ABox contains axioms describing the structure of particular worlds. A DL knowledge base can be given semantics by translating it into first-order logic with equality. Atomic concepts are translated into unary predicates, complex concepts into formulae with one free variable and roles into binary predicates. In the case of multiple diagnoses, the following TBox, see Code Listing 3, composed of a hierarchy to define criteria, personality disorders and their relationships through the role *has-criteria* has been defined:

```
Criteria ⊆ CriteriaT ⊆ ∃ has-criteria.Criteria
```

```
Cognitive ⊆ Criteria
```

```
Cognitive ⊆ ¬ Emotional,
```

```
Cognitive ⊆ ¬ Interpersonal
```

```
Emotional ⊆ Criteria
```

```
Emotional ⊆ ¬Interpersonal
```

```
Emotional ⊆ ¬Cognitive
```

```

Interpersonal  $\sqsubseteq$  Criteria
Interpersonal  $\sqsubseteq \neg$  Emotional
Interpersonal  $\sqsubseteq \neg$  Cognitive

AdultPersonalityDisorder  $\sqsubseteq$  MentalBehaviouralDisorder
SpecificPersonalityDisorder  $\sqsubseteq$  AdultPersonalityDisorder

 $\exists$  has-criteria.T  $\sqsubseteq$  MentalBehaviouralDisorderT  $\sqsubseteq \forall$  has-criteria.Criteria

HistrionicCriteria  $\sqsubseteq$  SpecificPersonalityDisorder Criteria
Histrionic  $\sqsubseteq$  SpecificPersonalityDisorder
Histrionic  $\equiv \geq 3$  has-criteria.HistrionicCriteria  $\sqcap \neg$  ParanoidCriteria  $\sqcap \neg$  ObsessiveCompulsiveCriteria

ObsessiveCompulsiveCriteria  $\sqsubseteq$  SpecificPersonalityDisorder Criteria
ObsessiveCompulsive  $\sqsubseteq$  SpecificPersonalityDisorder
ObsessiveCompulsive  $\equiv \geq 3$  has-criteria.ObsessiveCompulsiveCriteria  $\sqcap \neg$  ParanoidCriteria  $\sqcap \neg$ 
HistrionicCriteria

Paranoid  $\sqsubseteq$  SpecificPersonalityDisorder
ParanoidCriteria  $\sqsubseteq$  SpecificPersonalityDisorder Criteria
Paranoid  $\equiv \geq 3$  has-criteria.ParanoidCriteria  $\sqcap \neg$  ObsessiveCompulsiveCriteria  $\sqcap \neg$  HistrionicCriteria

```

Code Listing 3. TBox complete representation for multiple and unique diagnosis.

On the other hand, the ABox contains instances of criteria depending on type (Cognitive, Emotional or Interpersonal) and these criteria can also be re-used in the different disorders criteria. Thus, they are classified by type and grouped by the kind of disorder enabling the possibility of explaining the reasoning process behind diagnosis.

```

Cognitive(PC00001)
Cognitive(PC00007)
Cognitive(PC00021)
...
Emotional(PC00004)
Emotional(PC00006)
Emotional(PC00008)
...
Interpersonal(PC00002)
Interpersonal(PC00003)
Interpersonal(PC00005)
..

HistrionicCriteria(PC00008)
HistrionicCriteria(PC00009)
HistrionicCriteria(PC00010)
..

ObsessiveCompulsiveCriteria(PC00014)
ObsessiveCompulsiveCriteria(PC00015)
ObsessiveCompulsiveCriteria(PC00016)
...

ParanoidCriteria(PC00001)
ParanoidCriteria(PC00002)
ParanoidCriteria(PC00003)

```

```
PC00001 ≠ PC00002 ≠ PC00003 ≠ PC00004 ≠ PC00005 ≠ PC00006 ≠ PC00007 ≠ PC00008 ≠ PC00009 ≠  
PC00010 ≠ PC00011 ≠ PC00012 ≠ PC00013 ≠ PC00014 ≠ PC00015 ≠ PC00016 ≠ PC00017 ≠ PC00018 ≠  
PC00019 ≠ PC00020 ≠ PC00021
```

```
has-criteria(enf1, PC00002)  
has-criteria(enf1, PC00015)  
has-criteria(enf1, PC00003)  
has-criteria(enf1, PC00001)  
has-criteria(enf1, PC00018)  
has-criteria(enf1, PC00017)
```

Code Listing 4. ABox representation

A DL-reasoner is able to perform a classifying process according to these axioms to decide whether a specific set of criteria (*enf1*) matches the definition of a certain disorder or not. In case of multiple disorder matching, the process will return an inconsistency or an upper-concept classification (e.g. AdultPersonalityDisorder) because it will be unable to decide in which specific disorder the instance should be classified. This situation implies strange behavior due to inconsistencies in DL ontologies can be caused by other kind of axioms. In order to avoid this result, a new “closed” version of the ontology has been designed including axioms to support CWA declaring: 1) all criteria individuals as different 2) axioms to define disorders as equivalent classes with disjoints regarding other diseases in the hierarchy. Thus the inference process can uniquely classify disorder instances. Nevertheless, the use of Description Logics has some drawbacks: 1) the performance of OWL-DL reasoners is “practically good” for the intentional level when the size of a TBox is not likely to scale up too much and 2) for the extensional level they are unable to handle instances (ABoxes) of large size or even medium size for basic services like instance checking. The main reasons lie in the current algorithms (defined for intentional tasks and working in main memory) and the lack of real optimization for ABox services. In most cases these limitations make it impossible to use these tools to perform reasoning processes over real data, more specifically for data integration on the web. These limits may be overcome following two approaches: 1) do not reason over ontologies (we should wait for more efficient algorithms) and 2) limit the expressive power of the Description Logics (using OWL tractable fragments). In the second version of OWL called OWL2 (a W3C recommendation on October 2009) a set of profiles is provided called OWL 2 Profiles: OWL 2 QL based on DL-Lite, OWL 2 EL based on EL and OWL 3 RL based on DLP. These profiles enable the possibility of splitting the knowledge base into ontologies and rules to get more efficient reasoning processes. Now the approach of PsyDis is based on OWL-DL (*ALCQ*), due to the use of QCRs, but the axioms can be translated into a FOL program using TPTP syntax (Infrastructure for Automated Reasoning) that can be executed in any FOL inference engine, improving the performance but keeping the expressiveness. This novel approach is ongoing work (Schneider & Sutcliffe, 2011) and only a part of OWL 2 Full Semantics has been translated into TPTP formulae.

Finally, the number and type of treatments can be directly retrieved via a SPARQL query, see Code Listing 5. The disorder URI should be considered as an input parameter of the query. The result of these queries can be checked in Code Listing 6.

```
SELECT DISTINCT ?treatmentValue ?treatmentType WHERE {  
<http://127.0.0.1/psydiag/mdd.owl#F60.0> rdfs:subClassOf ?treatment.
```

```

?treatment rdf:type ?type.
FILTER (?type = owl:Class || ?type = owl:Restriction).
OPTIONAL {
  ?treatment owl:intersectionOf ?setOfTreatments .
  ?setOfTreatments ?x ?individualTreatment .
  ?individualTreatment rdf:type owl:Restriction.
  ?individualTreatment owl:hasValue ?treatmentValue.
  ?individualTreatment owl:onProperty ?treatmentType.
}
OPTIONAL {
  ?treatment owl:hasValue ?treatmentValue.
  ?treatment owl:onProperty ?treatmentType.
}
FILTER (
  ?treatmentType = md:has_psychological_treatment ||
  ?treatmentType = md:has_pharmacological_treatment)
}

SELECT DISTINCT count(distinct ?treatment) WHERE {
  <http://127.0.0.1/psydiag/mdd.owl#F60.0> rdfs:subClassOf ?treatment.
  ?treatment rdf:type ?type.
  FILTER (?type = owl:Class || ?type = owl:Restriction).
}

```

Code Listing 5. SPARQL Query

```

-----
| treatmentValue | treatmentType      |
=====
| mdpst:RCRT0001 | md:has_psychological_treatment |
| mdpt:10756001  | md:has_pharmacological_treatment |
| mdpst:COG0001  | md:has_psychological_treatment |
| mdpst:RAT0001  | md:has_psychological_treatment |
| mdpt:48546005  | md:has_pharmacological_treatment |
-----
|.1 |
=====
| 5 |
-----

```

Code Listing 6. SPARQL Query Results

UNIQUE DIAGNOSIS

In the case of unique diagnosis, we are dealing with the concept of using discriminants. Most of the systems which have been developed earlier with the aim of providing diagnosis are based on this schema. The main idea behind that is that "the more information we have, the more we can focus on possible results ". The extra information provided could be used to discard pathologies and focus better on the results.

As was previously mentioned, in Figure 3 we can see that a system that makes use of discriminant schema (more inputs, fewer outputs) will be able to discard pathologies when the

items do not match those existing in the knowledge base. This is the schema used to obtain a diagnosis system which provides a unique diagnosis.

The problem of unique diagnosis comes from the Open World Assumption (OWA) mentioned before. Given that in OWA it is not possible to assume that a fact is false unless it has been declared as false, to determine that a disorder should not be diagnosed, we need to establish some restrictions on the elements which take part in the diagnosis process. Previous studies have worked on this as that presented by García-Crespo, A. et al. (2010). However, this work was not focused on the main restriction which exists in psychological diagnosis process: we have a cardinality restriction to be able to provide a diagnosis.

For this reason, in this paper we provide a new solution, based on the work provided by García-Crespo, A. et al. (2010), but adding these cardinality restrictions and adapting the whole knowledge base to be used in a different domain (mental disorder diagnosis).

Imagine in this case that we have defined a new knowledge base with three psychological disorders defined as is shown in Figure 7.

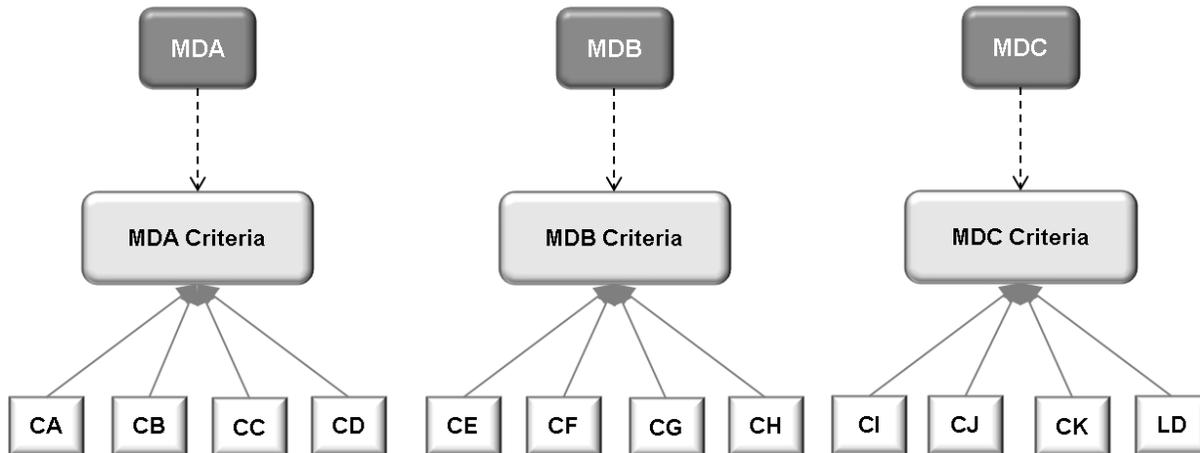


Figure 7. Unique Diagnosis Knowledge Base example

The aim of the description logics which will define each disorder is to “close the world” (make CWA) to ensure that only the disorder could be diagnosed. We also need to define the cardinal restriction to ensure that a disorder will only be diagnosed in the “query” which contains the information (criteria) that the patient suffers when there are enough elements related to the disorder. The Code Listing 5, 6 and 7 shows the restrictions for MDA, MDB and MDC respectively, from an OWL perspective.

```
(not (has_criterion some 'MDB Criteria'))
and (not (has_criterion some 'MDC Criteria'))
and (has_criterion only 'MDA Criteria')

has_criterion min 3 "MDA Criteria"
```

Code Listing 5. MDA Unique Diagnosis Formalization

```
(not (has_criterion some 'MDA Criteria'))
and (not (has_criterion some 'MDC Criteria'))
and (has_criterion only 'MDB Criteria')
```

```
has_criterion min 3 "MDB Criteria"
```

Code Listing 6. MDB Unique Diagnosis Formalization

```
(not (has_criterion some 'MDB Criteria'))  
and (not (has_criterion some 'MDA Criteria'))  
and (has_criterion only 'MDC Criteria')
```

```
has_criterion min 3 "MDC Criteria"
```

Code Listing 7. MDC Unique Diagnosis Formalization

Other restrictions or definitions which should be applied are referred to define disjoint classes. We are working with the unique diagnosis schema. Therefore, we have to define that an instance could only be classified in one of the available disorders. To do this, we need to establish that each class which represents a disorder should be disjoint of the rest.

This new approach could be used for querying the model in order to find the disorder which could affect any one patient.

However, this schema has three main restrictions:

1. It is only possible to query the model using a single individual which will define the current query. If two or more individuals with different queries are defined in the model, we will have inconsistency problems with the ontology. This is a problem that can be safely overcome trying to make single queries to the model.
2. In this model, we have a big limitation re the criteria. In the previous model we saw that a certain criterion (for example, CA) could pertain to more than one disorder. However, in the current model this is not possible. Each criterion will only be related to a concrete disorder. This generates a problem in those disorders which share common criteria because with this model they cannot be modeled.
3. The model has been designed in such a way that if a query does not satisfy any of the conditions/descriptions of the disorders, the ontology becomes inconsistent. Again, this is a similar problem to that commented upon in the first item. It can be overcome, in this case, applying an interface which detects the inconsistency and returns "no results".

EVALUATION

In order to evaluate if PsyDis produces accurate results in real scenarios, an evaluation of the tool has been carried out. This evaluation checked all PsyDis features using real examples of psychological diagnosis. The evaluation of the system consists of the performance of a set of diagnosis based on historical data. The aim is to compare PsyDis results with practitioners' diagnoses by means of sound methods. In order to do so, two different studies were performed. This evaluation consists of two different studies. The first study compares PsyDis with a set of diagnoses made by practitioners in the past (historical data). The second study tests PsyDis diagnosis with practitioners' judgments, but in this case this comparison is made at the same time by both actors and is not based on historical data.

STUDY 1

Research Design

The purpose of this study is to compare a set of diagnoses made in the past by a group of therapists with those made in the present by PsyDis. Thus, the aim is to compare the diagnosis made by three professional therapists on four of their clinical cases with the diagnosis made by the tool for each of six cases from the notes that they documented. Therapists selected four cases each. The therapists had already described, analyzed and diagnosed each of these cases. Therefore, the cases were "closed" (in some cases, the treatment had ended, in others, treatment was in course). Although cases were selected by the participating therapists, researchers asked them to select cases of specific personality disorders such as obsessive-compulsive disorder, histrionic and paranoid disorders (selected to illustrate the present investigation).

Hence, this study compares the diagnosis of a practitioner with PsyDis results for these twelve cases. It should be borne in mind that each of the cases had been diagnosed only once by a single therapist.

Sample

The three participating therapists were carefully selected from a long list of professional clinical activity. Subjects were selected from those who answered positively to a personal invitation sent by the authors. Participants present over 15 years of experience in the clinical area of personality disorders in adults. Two of them hold a Ph.D. in Psychology and are psychological assessment academics. The third participant is specialized in Psychiatry and holds a Ph.D. in Psychology.

Selected cases are all adults, and their evaluation and diagnosis was made in the last three years of clinical activity of the participants. The anonymity of patients and the characteristics of each case are guaranteed by therapists and authors.

With respect to disorders, seven cases of obsessive-compulsive personality disorder were selected, two cases of histrionic personality disorder and three cases of paranoid personality disorder.

Results and Discussion

Results show that Psydis coincides with professionals in ten of the twelve cases. The tool does not produce accurate results in one of the two cases of histrionic personality disorder and in one of the three cases of paranoid personality disorder. In fact, the tool does not perform any diagnosis in both cases, because they do not fit the criteria of any personality disorders described in ICD-10. Thus, in terms of accuracy of the system, the standard precision, recall and F1 are used to measure the performance of PsyDis in this Study 1. The precision calculates the percentage of results that are relevant, while the recall computes the percentage of results that are predicted. In statistics, the F-score (also F1 score or F-measure) is a measure of a test's accuracy. The F1 score can be interpreted as a weighted average of the precision and recall, where an F1 score reaches its best value at 1 and worst at 0, which means the higher the F-score, the more accurate the test (Tsai & Kwee, 2011). These set of measures are used frequently in the literature to assess recommender systems and other knowledge-based tools (e.g. García-Crespo et al., 2011; García-Peñalvo et al., 2011; López-Cuadrado et al., 2012; Tsai & Kwee, 2011). Results of PsyDis are as follows:

$$\text{PRECISION} = 10/10 = 1.000$$

$$\text{RECALL} = 10/12 = 0.833$$

$$\text{F1} = 0.909$$

Although these results show notable performance of the system, there is still room for improvement. Analyzing the roots of the errors, authors believe that it is not easy to asseverate that such differences are errors. From the beginning, we have assumed that the diagnosis of therapists in the first study was correct. However, a diagnosis is performed by a single professional, and therefore does not represent a consensus diagnosis. As a result, authors cannot conclude that PsyDis erred. The tool is faithful to the criteria of ICD-10, and according to its analysis the two cases do not fit the criteria for histrionic and paranoid disorders. However, the tool does match in diagnosing the remaining cases. Perhaps these diagnoses were not entirely appropriate for these two cases? This is a possibility. However, information regarding a case is very broad and complex and, as the professionals say, in many circumstances it is **not** possible to find behavioral disorders that are fully adapted to the criteria established by the manuals. Given these cases, that can be described as "less prototypical" the decision of the professionals is the result of detailed analysis to identify problem behaviors, personal variables, environmental variables, functional analysis, etc. In this sense, the authors might conclude that perhaps these cases do not represent typical cases, and consequently are not suitable for PsyDis. In any case, accuracy results reached by PsyDis are remarkably in line with previous efforts (e.g. García-Crepeo et al., 2010). But given the complexity of the problem the tool is facing, a second study is needed to determine whether in consensus diagnosis scenarios, PsyDis provides accurate results.

STUDY 2

Research Design

In this second study, authors again compare the diagnosis of therapists with the diagnosis made by PsyDis. The difference with Study 1 is that therapists do not select their own cases, but have to evaluate a set of new cases selected by an independent therapist from among his current patients. Researchers proposed three cases differing from those evaluated in the first study. These cases represent three different examples: an obsessive-compulsive disorder (case 1), a histrionic disorder (case 2), and a paranoid disorder (case 3). The substantial difference from the first study is that these cases are diagnosed by another group of therapists, who confirmed the initial diagnosis. Thus, we now have three cases in which diagnosis is agreed by four therapists (the independent therapist "owner" of the cases, and another three professionals who performed the diagnosis, together with PsyDis diagnosis). It is important to note that cases selected were the result of consensus in therapists' diagnosis, discarding those whose diagnostic evaluation was not identical.

The task of the therapist consisted in performing a diagnosis of each of the cases with the help of the ICD-10. This task was performed by these practitioners in isolation. The information provided to therapists consisted of a set of data for a general description of the problem behaviors and the results of tests or assessment instruments used by the independent therapist. Descriptive analysis and functional analysis were not provided.

Thus, therapists must diagnose three cases unknown by them in a "laboratory context". This process was to be completed within a maximum period of 4 hours. The output consisted in the evaluation and diagnosis of cases. They were not asked to perform an intervention design for the cases.

Samples

As already mentioned, the participants in this second study are different from those who took part in Study 1. The study consists of three therapists with more than 15 years of clinical experience in the area of personality disorders. One of them holds a Ph.D. in Psychology and the other two hold a MSc. in Psychology. Participants were not informed that their diagnoses would be compared.

With respect to the two samples of therapists involved in this second study (the one who selects their cases, and those that confirm the diagnosis), they present similar characteristics in terms of training and clinical experience.

Regarding disorders, three cases were selected: obsessive-compulsive personality disorder, histrionic personality disorder and paranoid personality disorder.

Results and discussion

Results show a disagreement among diagnoses. Thus, one of the practitioners diagnoses the case labeled as paranoid disorder as paranoid schizophrenia and another diagnoses the case that the independent therapist diagnosed as histrionic personality disorder as narcissism. Meanwhile, PsyDis matches the diagnosis established in advance by the independent therapist. Table 1 presents results provided by PsyDis and subjects:

Table 1. Study 2 Results.

Case	Independent Therapist	Subject#1	Subject#2	Subject#3	PsyDis
1	Obsessive-compulsive personality disorder				
2	Histrionic personality disorder	Histrionic personality disorder	Histrionic personality disorder	Narcissism	Histrionic personality disorder
3	Paranoid personality disorder	Paranoid schizophrenia	Paranoid personality disorder	Paranoid personality disorder	Paranoid personality disorder

Apparently, therefore, results indicate that Subject#1 and Subject#3 are wrong in some cases while PsyDis is right. Study 2 aimed to provide a test scenario in which diagnoses are agreed. In this particular case this agreement is moderate but, in any case, "typical" cases favor the diagnosis of the tool, which stays true to the criteria of the manuals. As a result, PsyDis would find it more difficult to diagnose more complex or less prototypical cases. However, these findings lead us to believe that, despite the disagreements, the percentage of overlap between judgments is high, indicating that PsyDis may be useful as a support tool in diagnostic decision-making, which is the intended purpose.

CONCLUSIONS AND FUTURE WORK

The main advantage of using Description Logics (a decidable fragment of first-order logic) is its support for formal reasoning (classification, check consistency and type inference). On the other hand, Web Ontology Language (OWL), the standardized language for representing the semantics of information on the Web, enables the development of an extensible framework in which new information, data and facts can be easily added to cover new psychological disorders. Nevertheless, the use of OWL-DL as the underlying representation model is related to the overhead of DL reasoning under changing data, which makes the approach unsuitable for some real-world domains, more specifically when real time capabilities are needed. This situation can be overcome by applying more efficient incremental reasoning, splitting the knowledge base into ontologies and rules, decreasing the expressiveness of the existing model or transforming the DL axioms into more efficient logics in terms of time-processing such as F-Logic.

Future work will be mainly focused on three areas: 1) appliance of DL-extensions such as Pronto for enabling probabilistic reasoning over DL (Lukasiewicz, 2007), allowing establishment of a percentage to the diagnosed diseases; 2) use of other diagnosis criteria such as those provided in DSM-IV-TR as part of the diagnosis process and 3) extension of the diagnosis model to allow a broader diagnosis which includes descriptive analysis, functional analysis and formulation (see figure 1).

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