Development of an Ontology for Supporting Diagnosis in Psychiatry

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Abstract. Psychiatry is the medical specialty responsible for the diagnosis and treatment of mental disorders, in an attempt to secure mental health in the human beings. However, the human brain is a very complex organ, which makes the diagnosis of this kind of illnesses a hard task. Considering this fact, an ontology was conceived with the task of inferring one or more psychiatric disorders depending on the symptoms presented by a patient.

1 Introduction

The human being possesses a very developed brain when compared to others species. Due to its extreme complexity, this organ can be striken by a large number or mental disorders like mood, personality and eating disorders. Psychiatry is the medical specialty that focuses on the diagnostic and treatment of this kind of problems. Unlike other physical disorders, mental problems can be hard do diagnose, considering some aspects like the absence of specific and easily detectable symptoms as well as the subjectivity of the physician responsible for the diagnosis[2].

While the various technical supporting solutions are promising, they proved to be insufficient due to several factors such as the difficulty in automating the decision-making processes or the identification of the various factors that lead to the onset of psychiatric disorders.

In an attempt to mitigate such situations, we argue that ontologies can have an important role in supporting the diagnosis of this type of problems, particularly when using automated reasoners to, given a set of symptoms for a particular patient, obtain a set of possible disorders affecting that individual. Furthermore, it is our conviction that the ontologies will facilitate the description of the diagnosis rational. These hypotheses arise from the claim that ontologies are the best answer for intelligent systems operating close to the human conceptual level[11].

Considering these facts, the goal of this project consists in creating an ontology-based system capable of supporting the psychiatrists' work by facilitating the diagnosis of mental disorders, specifically disorders of bipolar and depressive

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aetiology. In particular this paper presents the team's efforts to develop a rich-expressive ontology that will be the base for such system either for capturing knowledge expertise, to perform reasoning over the patient's symptoms and for explaining/describing the diagnosis. The authors decided in a first phase to adopt well-established documents on the domain instead of capturing the knowledge from experts (i.e. clinical). Two standard documents describe the psychiatric problems and diagnosis: ICD-10 (International Classification of Diseases, 10th revision) – from Mental Health Organization – and DSM-IV (Diagnostic and Statistical Manual of Mental Disorders, 4th Edition) – from American Psychiatric Association[3,2]. In this work the DSM-IV was adopted as our core source of conceptual knowledge and diagnostic rules. In fact, contrarily to ICD-10, DSM-IV possesses a group of rules and text-based decision trees that describe the diagnosis process, that we believe is expressible in *OWL DL* ontologies.

This document is structured as follows: initially, we will be presenting some information on the state of the art, going out as well as a brief explanation of the rules used by the DSM-IV in the diagnosis of Mental Disorders. The next step consists of the presentation of the ontology development process adopted as well as the writing of the results obtained in this study.

2 State of the Art

Through time, various authors have developed *AI* tools-based systems capable of helping health care professionals in the diagnosis of one or more pathologies in the various medical specialties.

In 2009 Nunes, Pinheiro and Pequeno[10] used DSM-IV as a reference to create a study about some of the *AI* techniques used to support diagnosis of Obsessive-Compulsive Disorder. In particular, these authors focused their efforts on developing a support system for multi-criteria decision (called *MACBETTH* method),which allows the attribution of a grade to an alternative based on pair comparisons; the program is therefore responsible for analysing both the cardinality and semantics and suggesting a way of solving the problem in cases of inconsistence. This is achieved by suggesting a range of grades and intervals susceptible to changes without making the problem inconsistent. In addition to the *MACBETH* method, Nunes and colleagues were based on the *SINTA* expert system[14], which consists of a visual tool for the development of expert systems based on the input rules of the domain. Although these authors use several *AI* tools in order to achieve their goal[10], they also consider that it is insufficient when seeking for fully automated decision-making processes based on multi-criteria analysis.

Later, in 2008, Kola and peers[11] have proposed the development of an ontology-based system capable of representing and storing information regarding to the various psychotic disorders. However, the existence of different data models regarding neuro-scientific research and clinical practice hinders their interoperability, representing an obstacle when considering the closeness between both fields. The main goal of this ontology is to serve as a connection bond in

the domain of psychosis although without going into great details in terms of "dimensionality" and "disorder definition". This ontology should be viewed as a logical approach of some psychiatric concepts due to the lack of consensus inside Psychiatry itself.

That same year, and assuming that most of the information related to mental health is computerized, Hadzic, Chen and Dillon[5] proposed the development of a *Mental Health Ontology (MHO)* capable of modelling concepts related to mental health and relationships that could be used to create a semantic structure appropriate to the storage and retrieval of information; this ontology consists of three sub ontologies, each one representing one of three categories: types of disorders, factors and treatments:

1. By collecting information from both ICD-10 and DSM-IV, Hadzic and peers were able to identify thirteen types of Mental Disorders as well as their subclasses and represent them in the sub ontology related to "Types of Disorders"
2. The second sub ontology is responsible for capturing and representing knowledge related to the factors affecting an individual's Mental Health; the authors considered the existence of five types of factors: Genetic, Physical, Environmental, Personal and Microorganic
3. Finally, the third and last sub ontology of *MHO* is related to Treatments like, for example, psychotherapy, pharmacotherapy, group and familiar therapy as well as electroconvulsive therapy

Later, Lin and colleagues[12] proposed an approach which consists of a distribution network throughout the body whose function is to provide vital energy to all parts of the body, strongly linked to physiological functions and the maintenance of health in humans. This solution interacts with human users via a decision tree algorithm in order to obtain the meridians necessary for diagnosis as well as design of a domain ontology (responsible for storing static knowledge on the meridian system) and a task ontology (used to describe the rules of psychiatric diagnosis based on the indicated meridians, as well as for inferring upon those rules – dynamic knowledge). The goal of this work is to infer the task-ontology rules based on the decision tree, regarding case history data from patients and the training data used. Despite resembling with this work, our approach is based on a reference expert-oriented documents such that the rules are much more complicated than the rules inferred in [12] (mostly based on scalar values).

3 Classification of Mood Disorders According to DSM-IV

In DSM-IV, the section related to Mood Disorders includes a number of diseases that are divided into three distinct parts: Mood Episodes, Mood Disorders and Specifiers relating to disorders of this aetiology that describe either the most recent episode or the course of recurrent episodes.

DMS-IV's Mood Episodes are sets of symptoms for a particular patient and they cannot be considered actual disorders. However, it can be stated that a given patient having all symptoms and characteristics of that Mood Episode can be diagnosed as having that Mood Episode. The DSM-IV defines four types of Mood Episodes: Major Depressive, Manic, Hypomanic and Mixed Episodes[2]. In turn, a Mood Disorder can be seen as a set of Mood Episodes (and other isolated symptoms) while specifiers characterize a particular Mood episode/disorder, such as the type or severity.

Considering the Dysthymic disorder (defined and characterized in DSM-IV), it can be assumed that it is a type of Depressive Disorder where the patient presents with depressed mood for at least two years. Additionally, Dysthymic disorder features by a minimum of two symptoms presented in the following listing:

- Increased or decreased appetite
- Insomnia or hypersomnia
- Fatigue or low energy
- Low self-esteem
- Difficulty concentrating and decision making
- Feelings of hopelessness

Concomitantly, the patient must have the following symptoms:

- Lack of any Major Depressive Episodes in the last two years and of any other Mood Episode throughout the course of the disorder
- Present severe difficulties or impairment in work, social and other daily-life activities
- Lack of association of symptoms with substance abuse, Cyclothymic Disorder or Chronic Psychotic Disorder
- Symptoms are not caused by a Chronic Major Depressive Disorder or a Major Depressive Disorder in partial remission[2]

The definition of the Dysthymic disorder is dependent on the definition of other disorders and vice-versa, eventually, which causes considerable definition and reasoning difficulties.

As mentioned earlier, the aim of the project is to create an ontology for supporting the task of diagnosing mental disorders afflicting the patient, taking into account the information entered by the user (like the physician, the patient or other) on the symptoms (and their characteristics such as the duration, stage, nature, etc.). It is then intended that the episodes and disorders captured in the ontology/KB give rise to the more specific diagnosis.

4 Ontology Development

In Computer Science, ontologies are engineering artefacts capable of formally modelling a system's structure, i.e., the most relevant entities and relations

considered useful for the modelling and axioms that supply interpretation restrictions and reasoning capabilities to reasoning engines. This section describes the team's effort developing the ontology following the incremental and iterative process *KMP – Knowledge Meta Process*[1], which consists of five basic steps, which will be described in the following subsections as adopted by the team for this ontology.

4.1 System's Economic and Technical Feasibility Study

In this project, the feasibility study consisted in a critical analysis of the rules used by the DSM-IV to make the diagnosis of mood episodes and disorders. Although the analysis of these rules have served as a warning to the impossibility of achieving the initial goals of the project, it played an important role in the developed work since it allowed the identification of several primitive classes which make up the ontology; these classes have served as the basis for the organization of knowledge captured during the reading of the document.

4.2 Kick-off

During the Kick-off phase the team identified and developed the primitive classes and the basic hierarchy of the ontology. Since its main purpose is to diagnose a certain disorder that affects a certain patient (considering the information entered by the user about both the symptoms and their characteristics presented by this same patient), one of the main characteristics of this ontology is the reclassification of mood episodes and disorders considering the input data.

In order to represent knowledge related about each of the Bipolar and Depressive disorders represented in DSM-IV, the ontology conceived in this project followed a basic hierarchy of classes that will be explained below:

- Patient - Represents an individual patient in consultation
- Symptom - Represents all aspects of the symptoms presented by the patient, including the duration of symptoms, the four types of mood episodes identified by the DSM-IV[2] and its aetiology
- Affective Disorder - Identifies and characterizes the various conditions considered by the DSM - IV
- Cause -Identifies the causes of the symptoms, like other medical condition or health problem or the abuse of substances
- Consequence - Identifies the consequences of symptoms, which could consist of social distress or impairment, hospitalization or when symptoms are notorious to others
- Feature - Consist of the specifiers defined by DSM-IV[2]
- Stage - identifies the stage of the disorder (e.g. Chronic or In Remission)
- Severity - Mild, Moderate or Severe (according to DSM-IV[2])

In addition to the above classes, eight properties have been create in order to design rules for defining some of these classes (especially classes related to disorders and episodes). Figure 1 tries to explain the reader a better understanding of these rules.

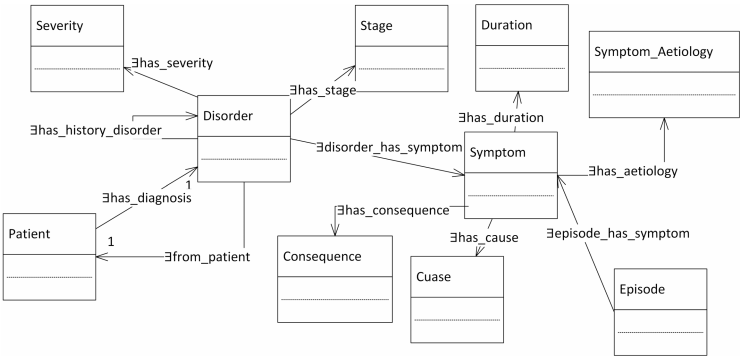

Fig. 1. Ontology primitive classes and their relationships

Unlike what usually happens in the Kick-off phase of the *KMP*, the *ORSD (Ontology Requirements Specification Document)* was not formally created as the goals of the ontology had been previously defined i.e., the capture of decision trees presented in the ontology for future inference of diagnostics.

4.3 Refinement

During the refinement of the ontology we chose to use a top-down approach in the creation of subclasses of primitive classes. Such approach allowed starting from generalized concepts and refine them in order to obtain a class hierarchy increasingly closer to the real needs of the ontology. Figure 2 tries to explain how this refinement was made.

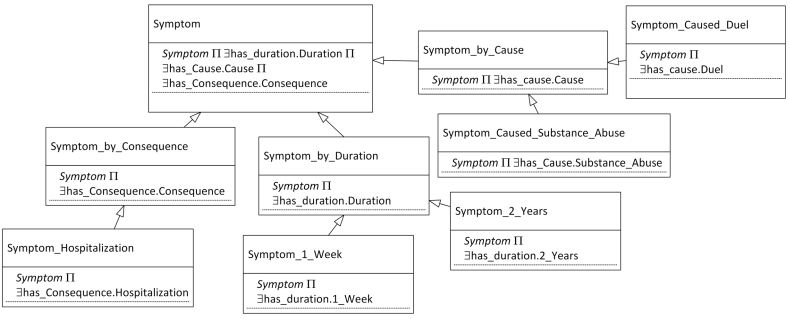

Fig. 2. Example of refinement

Although the vast majority of diagnostic rules explained in DSM-IV were captured and formalized in *OWL DL* considered in early iterations, during the

evaluation process the team noticed that some of them were not representable in *OWL*, leading to the adoption of *SWRL*[13]. One of the rules is the following:

Rule 1: Distúrbio(?d), Paciente(?p), de paciente(?d, ?p), paciente tem sintoma(?p, ?s) $\rightarrow$ *distúrbio tem sintoma(?d, ?s)*

This can be read as follows: given a specific disorder d and a patient p, if it is stated that the patient p has symptom s, then it is inferred that the disorder d will also have the symptom s.

5 Evaluation

According to the *KMP* the ontology evaluation is done at three levels: syntactic (related to the linguistic conformity of the ontology properties), semantic (related to the consistency of the ontology, i.e., how it captures the rules represented in DSM-IV's decision trees) and of technological properties (e.g. interoperability or scalability).

We assumed that the testing and debugging of ontology designed in this project fall under the evaluation phase of the *KMP*. In order to test and debug the ontology, several individuals were created in a bottom-up approach. This approach allows the testing of the rules previously defined, allowing early detection of both syntactic and semantic inconsistencies in the ontology. The evaluation was repeated for all the created individuals in order to test all the classes contained in the ontology, which allowed the development of a both semantic and syntactically valid ontology according to the DSM–IV.

6 Conclusions and Future Work

The work described in this paper seeks, in some way to help health professionals in their arduous task of making accurate diagnoses in clinical Psychiatry since it can be useful in suggesting the diagnosis to be made to a particular patient. However, the diagnosis rules adopted in DSM-IV are relatively complex and difficult to formalize in *OWL DL* and *SWRL*. Currently the team doubts about the causes since we are aware of the difficulties in understanding/interpreting certain text-driven ambiguities of the rules, which would have been prevented by a domain-expert participation in the ontology development process.

The evaluation phase allowed concluding that the expressivity capabilities of *OWL* and *SWRL* associated with standard reasoners are sufficient for formalizing and reasoning upon one patient's data. Yet, early efforts for validation and verification of the ontology by domain experts (i.e. psychiatrist) were not successful. This was especially due to the development environment adopted, which inhibits a non-technical stakeholder to full-understand the semantically rich ontology, preventing a full-commitment with the verification and validation of the ontology.

Despite the near future team's efforts will be focused in the domain-expert verification and validation of the ontology, the efforts will not address the development of a domain-expert-oriented user-interface, but the validation and

verification with minimal expert intervention. For that, real patient's data will be adopted in the evaluation phase so that the reasoner's inferences (i.e. the diagnosis) can be compared with the psychiatrist' diagnosis. This will allow the revision and refinement of the DSM-IV-based ontology's rules based on the psychiatrist's rules, thus focusing in the core of the system.

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