# Ontologies, Disorders and Prototypes

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**Abstract**. As it emerged from philosophical analyses and cognitive research, most concepts exhibit typicality effects, and resist to the efforts of defining them in terms of necessary and sufficient conditions. This holds also in the case of many medical concepts. This is a problem for the design of computer science ontologies, since knowledge representation formalisms commonly adopted in this field (such as, in the first place, the Web Ontology Language - OWL) do not allow for the representation of concepts in terms of typical traits. The need of representing concepts in terms of typical traits concerns almost every domain of real world knowledge, including medical domains. In particular, in this article we take into account the domain of mental disorders, starting from the DSM-5 descriptions of some specific disorders. We favour a hybrid approach to concept representation, in which ontology oriented formalisms are combined to a geometric representation of knowledge based on conceptual space. As a preliminary step to apply our proposal to mental disorder concepts, we started to develop an OWL ontology of the schizophrenia spectrum, which is as close as possible to the DSM-5 descriptions.

**Keywords:** Representation of concepts, Formal ontologies, Conceptual spaces, Medical ontologies, Mental Disorders, DSM-5.

## 1. Representing Concepts: Some Problems Raised by Medicine

In philosophy and cognitive sciences, different theories about the nature of concepts have been proposed. According to the traditional view, known as "classical", concepts can be simply defined in terms of sets of necessary and sufficient conditions. This theory was dominant since the times of Aristotle until the mid'70s of the last century, when the philosophical analyses by Ludwig Wittgenstein (Wittgenstein 1953) and the experimental results obtained by Eleonor Rosch (Rosch 1975) showed that, for most of the common-sense concepts, this position does not hold since conceptual structures are mainly characterized by "typical" category membership cues and are organized in human mind in terms of prototypes. Since then, different positions and theories on the nature of concepts have been proposed in order to explain the aspects concerning conceptual "typicality". Usually, they are grouped in three main classes, namely: prototype views, exemplar views and theory-theories (see e.g. Murphy 2002; Machery 2009). All of them are assumed to account for (some aspects of) prototypical effects in conceptualization.

According to the prototype view (introduced by Rosch), knowledge about categories is stored in terms of prototypes, i.e. in terms of some representation of the "best" instances of the category. For example, the concept CAT should coincide with a representation of a prototypical cat. In the simpler versions of this approach, prototypes are represented as (possibly weighted) lists of features.

According to the exemplar view, a given category is mentally represented as a set of specific exemplars explicitly stored within memory: the mental representation of the concept CAT is the set of the representations of (some of) the cats we encountered during our lifetime.

Theory-theory approaches adopt some form of holistic point of view about concepts. According to some versions of the theory-theories, concepts are analogous to theoretical terms in a scientific theory. For example, the concept CAT is individuated by the role it plays in our mental theory of zoology. In other version of the approach, concepts themselves are identified with micro-theories of some sort. For example, the concept CAT should be identified with a mentally represented micro-theory about cats.

These approaches turned out to be not mutually exclusive. Rather, they seem to succeed in explaining different classes of cognitive phenomena, and many researchers hold that all of them are needed to explain psychological data (see again Murphy 2002; Machery 2009).

The case of some medical concepts, such as the general concept of DISEASE and the various individual disease concepts (such as PNEUMONIA, BREAST CANCER, SCHIZOPHRENIA, CATATONIA, and so on) show the same "problems" presented by most common-sense concepts, as they can hardly be represented in terms of individually necessary and jointly sufficient conditions. Faced with the issues raised by the many attempts to find a traditional definition for the general concept of DISEASE (Amoretti 2015), some philosophers of medicine have thus proposed to regard the concept of DISEASE and those of individual diseases as non-classical ones. In this vein, on the grounds of the great variability among individual diseases, new theories based on family resemblances, prototypes or exemplars have been proposed (see e.g. Sadegh-Zadeh 2011, 2000, 2008, Lilienfeld and Marino 1995, 1999, Pickering 2013, McNally 2011).

In the first case, there is no common feature that all individual diseases must have, but any two of them should share at least one feature. In the second case, there is a set of properties that represents the best instance of the disease category, that is an ideal and abstract construction of the general concept of disease, the prototype, to which any individual disease must approximate to some degree, sharing with it a goodly number of properties. In the third case, some individual diseases are regarded as particularly relevant, as the exemplars of the disease category, and thus all other diseases must exhibit a goodly number of their specific features.

These views are obviously different: embracing the family resemblances theory implies that there is no specific set of properties, determined by the prototype or the exemplars, that individual diseases must meet to some degree; the prototype is an abstract construction that doesn't need to correspond to any individual disease, while the exemplars are concrete members of the category. Nevertheless they are often conflated or muddled in the relevant literature. For example, McNally (2011, p. 212) refers to Wittgenstein saying that "Examples of most useful concepts bear only a family resemblance to one another. Most have some overlapping attributes without sharing an essence present in every case"; but clearly he has in mind the prototype view, as he continues specifying that "The more attributes a given case has, the better an example it is of the concept". A similar confusion is made by Cooper (2007, p. 41), who mentions family resemblances saying that "While

there need not be any one feature that all family members possess, any two members will be similar in a variety of ways"; however, she unpacks this idea through the exemplar view: "whether a condition counts as a mental disorder depends on its degree of resemblance to prototypical cases, such as schizophrenia and psychotic depression. Conditions that are sufficiently like these central cases get counted as disorders". Again, Sadegh-Zadeh (2008, p. 119) seems to conflates prototypes and exemplars claiming that "A concept determines a category […] by exhibiting the relational structure of the category that is characterized by best examples, called prototypes, such that other category resemble them to different extents".

The above confusions can be partially explained by the fact that all three views offer a plausible way to deal with conceptual "typicality", that is the evidence that some instances of the general category of disease, namely some individual diseases, are regarded as more representative than others. Moreover, all three views agree that there is no set of properties shared by all and only individual diseases: no specific property is individually necessary and no fixed number of them is sufficient to characterize the general concept of DISEASE. On the contrary, overall similarities among different set of properties should encompass the absence of any particular shared property – such as, as it is often claimed, dysfunction (Boorse 1976, Wakefield 1992, 1999). Many scholars adopting one of the above strategies do not attempt to better explicate the similarities relationship among individual diseases (Lilienfeld and Marino 1995, 1999); others think that fuzzy logic is the best, and possibly the only, way do the job (Seising and Tabacchi 2013, Sadegh-Zadeh 2000, 2008, 2011) – but, of course, some important alternatives to represent non-classical concepts have been proposed in the general literature, especially because fuzzy-logic faces some unavoidable difficulties in handling compositionality (on this aspect see Frixione and Lieto 2014).

As sketched above, approaches based on family resemblances, prototypes, and exemplars have been used to characterize the general concept of DISEASE, but they seem particularly suited to handle the general concept of MENTAL DISORDER (Lilienfeld and Marino 1995, 1999) as well as the various concepts of individual mental disorders. This more restricted class of medical concepts will be the focus of our present work.

The DSM (the Diagnostic and Statistic Manual of Mental Disorders), which is published by the American Psychiatric Association and represents a sort of "bible" for psychiatrists and scholars within the field of mental pathology, has in fact a merely descriptive approach: it rarely incorporates theoretical information regarding the causes of individual mental disorders, and classifies them using a list of operational diagnostic criteria. As a consequence, and somehow differently to what usually happens with individual somatic diseases included in ICD (the International Classification of Disease), individual mental disorders are typically identified not by their etiology or underlying pathological cause (a few exceptions being, for example, the different types of neurocognitive disorders), but through their syndromes, that is through a catalogue of their characterizing symptoms and signs, none of which is individually necessary and no fixed number of which is sufficient to determine membership to a certain individual disorder category. Moreover, in most cases these syndromes are not supposed to be reified, as to correspond to some kind of entity or mechanism (such as an underlying dysfunction).

Let's see, for instance, an oversimplified version of the diagnostic criteria for schizophrenia and catatonia given by the DSM-5: "Two (or more) of the following […] At least one of these must be (1), (2), or (3): 1. Delusions; 2. Hallucinations; 3. Disorganized speech […] 4. Grossly disorganized or catatonic behavior; 5. Negative symptoms" (DSM-5, p. 99); "The clinical picture is dominated

by three (or more) of the following symptoms: 1. Stupor […] 2. Catalepsy […] 3. Waxy flexibility […] 4. Mutism […] 5. Negativism […] 6. Posturing […] 7. Mannerism […] 8. Stereotypy […] 9. Agitation […] 10. Grimacing 11. Echolalia […] 12. Echopraxia" (DSM-5, p. 119). Similar operational criteria, introduced in DSM-III (1982), were meant to replace what psychiatrists dub as "prototypes", that is short descriptions of paradigmatic cases that would serve as standards of comparison to evaluate and diagnose any single patient. Here, as an example, the category of schizophrenic reactions according to DSM-I (1952): "It represents a group of psychotic reactions characterized by fundamental disturbances in reality relationships and concept formations, with affective, behavioral, and intellectual disturbances in varying degrees and mixtures. The disorders are marked by strong tendency to retreat from reality, by emotional disharmony, unpredictable disturbances in stream of thought, regressive behavior, and in some, by a tendency to deterioration", (DSM-I, p. 26).

Even if the operational structure of DSM-5 coincides neither with the prototype nor the exemplar views as they are developed by cognitive psychologists, it may still suggest to incorporate some features of these approaches in the representations of the various concepts of individual mental disorders (such as, SCHIZOPHRENIA, CATATONIA, etc.) as well as the general concept of MENTAL DISORDER, as like non-classical concepts they cannot be possibly defined through necessary and sufficient conditions, and clearly exhibit prototypical effects.

In order to address this problem from a computational perspective, we have analyzed the field of logic-oriented knowledge representation systems and, in particular, the class of formalisms known as formal ontologies. We provide below a brief overview of this class of systems by showing that, also in this artificial context, we face the problem of representing typical or "non-classical" information of medical concepts.

## 2. Formal Ontologies and Common-Sense Representations

In the last decades the problem of concept representation received a great deal of attention within the field of artificial intelligence (AI), and in particular in knowledge representation, due to its relevance for semantic technologies and for the development of formal ontologies.

In the AI tradition, an ontology is "an engineering artifact, constituted by a specific vocabulary used to describe a certain reality, plus a set of explicit (axiomatic) assumptions regarding the intended meaning of the vocabulary words" (Guarino 1998). The representation languages adopted for the development of formal ontologies stemmed from the tradition of so-called structured inheritance semantic networks – the first system in this line of research was KL-ONE (Brachman and Schmolze 1985). These formalisms are known today as description logics (DLs), and the main formal ontological languages such as OWL and OWL 2 belong to this class. The main constructs of such languages are concepts (or classes), roles (or properties), and individuals.

DLs are are logical systems (usually, they are subsets of first order predicate calculus). They can perform a series of automatic inferences, such as categorization (the process of attributing a specific individual as a member to a class), classification (the process through which new class-subclass relations are inferred) and consistency checking (the process of testing the logical coherence of a given ontology).

As logical systems, DLs have a model theoretic, Tarskian style semantics associated to them (Horrocks et al. 2003). This fact causes a problem: Tarskian semantics is fully compositional, and typicality effects are hard to accommodate with compositionality. As a consequence, DLs do not allow the representation of concepts in prototypical terms (on this aspects see Frixione and Lieto, 2012). DLs allow the representation of concepts exclusively in terms of sets of necessary and/or sufficient conditions. This is a severe drawback from the standpoint of the representation of many classes of concepts.

In particular, this strong bias towards the representation of concepts in terms of necessary and/or sufficient conditions alone is a problem in the field of biomedical ontologies. Most of them, indeed, (including SNOMED, the largest biomedical ontology currently available: http://www.ihtsdo.org/ snomed-ct) are conditioned by the adoption of formalisms that do not allow to represent concepts in typical terms. This possibility should be of crucial importance for representing both such general concepts as DISEASE or MENTAL DISORDER, and the concepts of individual diseases and mental disorders. Consider for example the concepts of individual mental disorders. In DSM-5 they are characterized in terms of syndromes and operational criteria. However, at the level of specific mental disorders, it is impossible to individuate sets of symptoms and criteria that are individually necessary or jointly sufficient to determine membership.

In this perspective, we propose to integrate typicality effects in computational representations of concepts. More precisely, we focus on prototypical and exemplar based approaches, and propose to combine them in a hybrid model. (For the time being, we do not take into consideration here theory-theory approaches, since them are in some sense more vaguely defined if compared to the other two positions.)

Following the approach proposed in Frixione and Lieto (2013, 2014b), we propose a hybrid architecture (fig. 1) combining a "classical" component (in which concepts are represented, as far as it is possible, in terms of necessary and/or sufficient conditions) with a "typicality-oriented" component, allowing both prototype and exemplar-based representations.
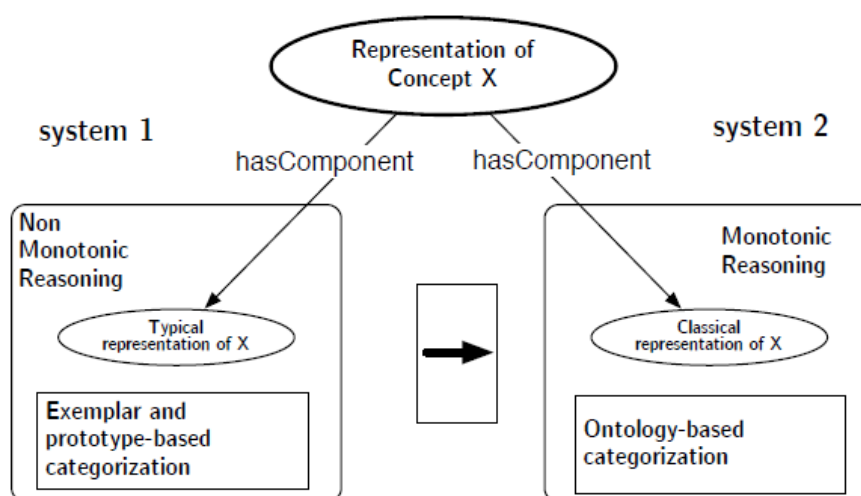


Figure 1. The Conceptual Architecture.

The "classical" component is demanded to some standard ontological formalism; the "typicality-oriented" component to a conceptual space, where conceptual spaces are a geometric framework for knowledge representation proposed by Peter Gärdenfors (2014).

In a conceptual space concepts are described in terms of a number of quality dimensions. In some cases, such dimensions are directly related to perception; examples could be temperature, weight, brightness, pitch. In other cases, dimensions can be more abstract in nature. To each quality dimension is associated a geometrical (topological or metrical) structure. The central idea behind this approach is that the representation of knowledge can take advantage from the geometrical structure of the spaces. Instances (or exemplars) are represented as points in a space, and their degree of similarity can be calculated in a natural way according to some suitable distance measure. Concepts correspond to regions, and regions with different geometrical properties correspond to different kinds of concepts. Prototypes and typicality effects have a natural geometrical interpretation: a prototype corresponds to the geometrical center of the region representing a concept (provided that the concept corresponds to a convex region). Thus, given a concept, a degree of centrality can be associated to each point that falls within the corresponding region. This degree of centrality can be interpreted as a measure of its typicality. Conversely, given a set of $n$ prototypes represented as points in a conceptual space, a tessellation of the space in $n$ convex regions can be determined in the terms of the so-called Voronoi diagrams. An example is shown in Figure 2, where the centre of each region corresponds to the prototype of a given concept, and where different exemplars can be represented as points in a conceptual region. The similarity between exemplars, or between prototypes and exemplars is obtained by calculating the metric distances in the underlying space.
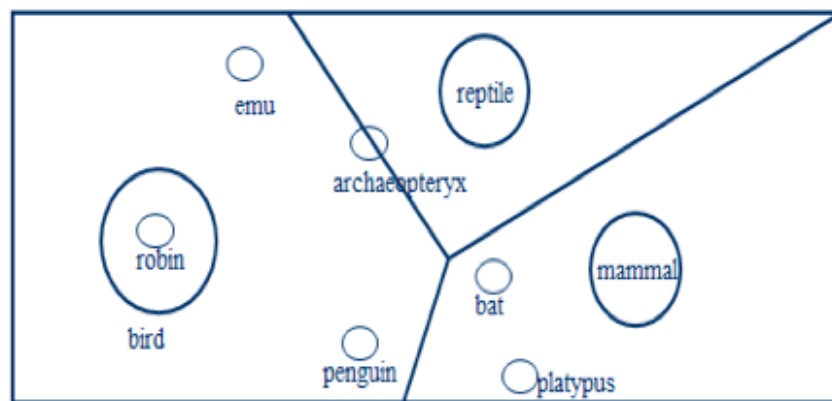


Figure 2. Example of the Voronoi tessellation of a conceptual spaces (from Gärdenfors and Williams, 2001)

In sum, the appeal of conceptual spaces consists in the fact that they provides a natural way of representing typicality effects, and that their geometrical structure provides a natural way of calculating the semantic relations between concepts, prototypes and exemplars in terms of metrical distance.

## 3. Some Preliminaries of a Case Study: The Schizophrenia Spectrum

As a preliminary step to implement a system based on the above described conceptual architecture, we are currently developing a formal ontology that tries to overcome some limitations of the existing ontological representations of individual mental disorders. In particular, with respect to the

existing taxonomies, we are currently building a representation that aims to be closer to the DSM-5 nosology and rationales. Some important remarks must be done here.

Our assumption that the general concept of MENTAL DISORDER and the various concepts of individual mental disorders – as they are currently described and categorized by DSM-5 – should be treated as non-classical ones is preliminary tested by developing an ontology based on the OWL-DL (Ontology Web Language Description Logic) dialect. This first step may seem to contrast with what we claimed in the previous two sections. However, we believe it is a necessary stage in order to verify and evaluate the exact limits of a classical approach to the formal representation of the concepts of individual mental disorders, as we suspect that some problems encountered by the already available formal ontologies might be due to an oversimplification of the structure and rationales of DSM-5 descriptive nosology.

On this respect, we take seriously the DSM-5 definition of the concept of MENTAL DISORDER, according to which a mental disorder is primarily a syndrome, that is a set of symptoms and signs. This means, for example, that the classes of *Mental_Disorder* and *Symptom* must be linked through an appropriate property (it must be remembered that, in OWL terminology, *properties* correspond to roles, or two place relations). Making the relationships between mental disorders and pattern of symptoms explicit might also help to clear out some classification disputes about where to place some controversial mental disorders among DSM-5 chapters.

Moreover, even if the DSM-5 definition of the concept of MENTAL DISORDER requires a dysfunction being in place, there is also the widespread conviction that syndromes should not be reified. The possibility to discover that basic dimensions of functioning, and thus dysfunctioning, cut across traditional syndrome-based diagnostic categories is actually envisaged – as the NIMH Research Domain Criteria (RDoC) project seems to corroborate. This means, for example, that the class of *Mental_Disorder* must be conceived in non realist terms and the concept of MENTAL DISORDER clearly distinguished from the concept of DISEASE.

Broadly speaking, the rationale we have followed to build our DSM-5 compliant ontology can be summarized in 4 steps, as shown in Figure 3:

- Identification of main concepts;

- Formalization of classes and properties;

- Implementation;

- Comparison between symptoms and evaluation (i.e. modelling decision about the taxonomical position and the related axioms that need to be added).
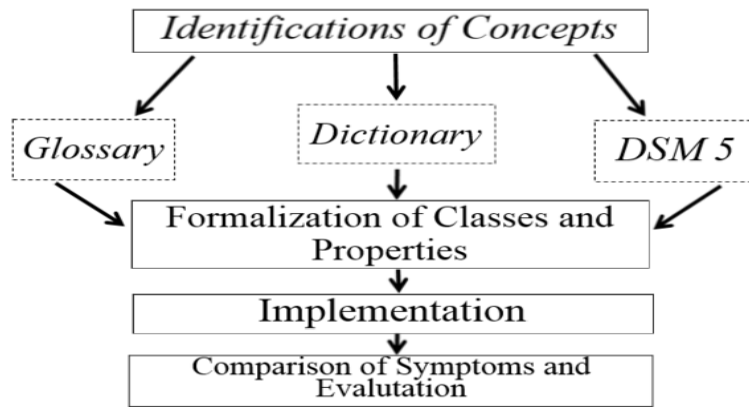
Figure 3. The process of building a DSM-5 compliant ontology

The goal of the first phase was identifying, organizing and structuring all the main concepts of the domain by using an abstract model, e.g. graphs or schemes. Initially, we focused on the chapter of Schizophrenia Spectrum only, and defined relevant concepts and properties through a glossary or dictionary written in natural language. Afterwards, with the second phase we used description logics to formalize all the concepts and properties previously identified and thus to obtain the adequate terminological domain knowledge. The third phase aimed at encoding and implementing a formal ontology using Protégé, a widespread ontology editor. In the fourth and last phase we compared various symptoms among ontologies and different disorders. Moreover, the process of evaluation can be also driven in parallel with the previous three steps.

The Schizophrenia_Spectrum ontology that we have developed is currently composed by 58 classes, 5 properties and 191 axioms. As already mentioned, the ontology has been developed by adopting the OWL-DL (Ontology Web Language-Description Logic) dialect.

The three main classes are *Mental_Disorder*, *Patient* and *Symptom*. The top level of the ontology, which focuses on the various classes of the Schizophrenia Spectrum category and the associated symptoms, is shown in Figure 4 below.
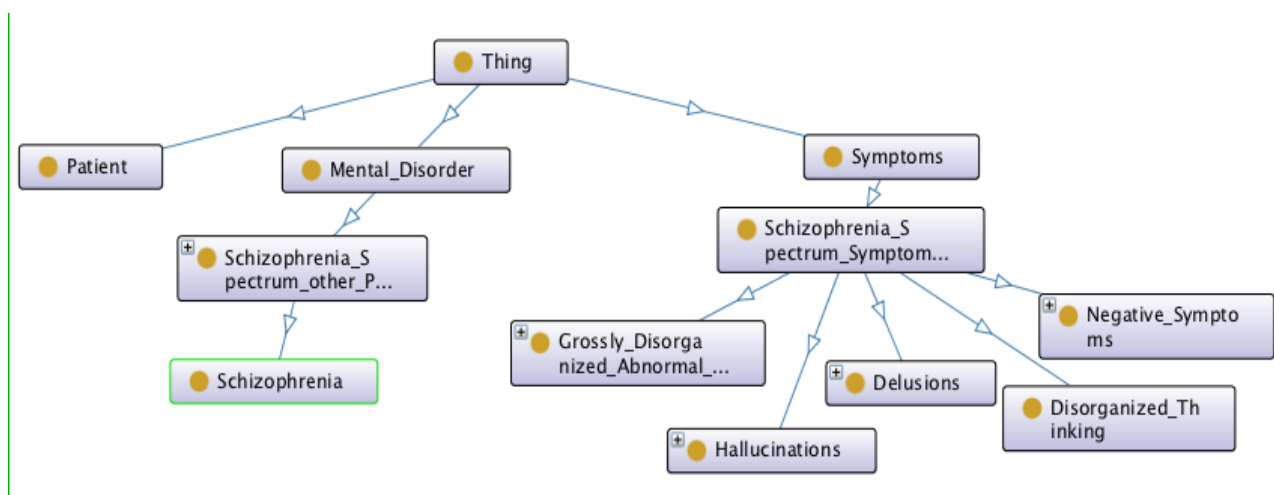


Figure 4. Top Level of the Schizophrenia Spectrum Ontology

The top-level classes chosen by the adopted modelling not only allow that each mental disorder might be identified through quite different set of symptoms (as it is clearly demanded by the DSM-5 diagnostic criteria), but also address comorbidity (a phenomenon which is still common in DSM-5 meaning that each patient showing a certain set of symptoms might be diagnosed with more than one mental disorder).

The class of *Patient* allows to model many different patient instances, which is useful in order to include personal information regarding individuals (such as age, sex, gender, ethnicity, etcetera).

Finally, the class *Symptom* currently contains the following main subclasses, which have been built in accordance with the DSM-5 criteria: *Delusions*, *Disorganized Thinking***,** *Grossly Disorganized Abnormal Motor Behavior***,** *Hallucinations***,** *Negative Symptoms***.**

The class of *Symptom* and its subclasses are disjointed from the *Schizophrenia Spectrum other Psychotic Disorder* and its subclasses as this guarantees the separation from symptoms that involves other mental disease.

The current version of the ontology (which is still subject to revisions and extensions) is available in a navigable format at: http://www.di.unito.it/~lieto/Schizophrenia_Spectrum.html.

Even if we developed a formal ontology which is more DSM-5 compliant than others (for instance, Ceusters and Smith 2010), as we predicted it is still unable to handle the representation and reasoning of common-sense cues. For this reason, and in accordance with our theoretical reflections advanced in the previous two sections, we plan to integrate the current ontology-based symbolic structure with a conceptual space component.

One possible application of this integration would be the realization of an artificial system that, given a set of typical traits characterizing the different symptoms, would be able to provide the correct identification of a given mental disorder.

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