Towards an Ontology of Schizophrenia

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***Abstract*—The paper presents an Ontology of Schizophrenia (OS) that is designed to provide a formal representation of schizophrenia-related entities crucial to the treatment and study of schizophrenia. OS is developed in accordance with the OBO (Open Biomedical Ontology) Foundry principles and constructed in compliance with Basic Formal Ontology (BFO) as its upper-level ontology. It uses mid-level and domain ontologies built in conformity with BFO such as the Ontology for General Medical Science (OGMS) and the Mental Functioning Ontology (MFO). OS is developed using Protégé 4.3 and is implemented in OWL2. Having imported BFO2.0 OWL and the development version of OGMS, OS adds approximately 30 schizophrenia-related terms and attempts to provide both formal and textual definitions for each. The terms in OS are in addition annotated with ontology metadata such as labels, textual definitions, definition sources, term editors and editor notes.**

Keywords—OBO Foundry; Schizophrenia; Ontology

I. INTRODUCTION

In biomedical ontology research, the ontological analysis of disease has been much discussed, since disease is of paramount importance for biomedicine. However, little research has been done on the ontology of mental disease. Consequently, data and information about mental disease have not yet been exploited using ontology-based approaches.

Among mental diseases, schizophrenia has been a focus of attention because of its unique features. A rigorous representation of the entities relevant to the study of schizophrenia would provide a new avenue for its prevention and cure [9]. However, the definitions or general descriptions of schizophrenia vary greatly from one domain expert to another [1, 7, 12]. For instance: "Schizophrenia is a chronic and severe disorder that affects how a person thinks, feels, and acts." (National Institute of Mental Health); "Schizophrenia is a mental disorder that makes it hard to tell the difference between what is real and not real." (Medical Encyclopedia); and "Schizophrenia is a serious brain illness." (National Library of Science). An attempt to create an ontology of schizophrenia is therefore a challenging but potentially valuable task.

Furthermore, efforts to deal with schizophrenia in existing ontologies are not fully adequate. For instance, schizophrenia is textually defined in the Experimental Factor Ontology (EFO) [5] as follows: "A major psychotic disorder characterized by abnormalities in the perception or expression of reality." We assume that the phrases 'abnormalities in the perception or expression of reality' is an attempt to refer to common clinical signs and symptoms of schizophrenia such as delusions and hallucinations. But in the absence of definitions of terms such as these, EFO's characterization of schizophrenia is uninformative. Additionally, the EFO class 'schizophrenia' falls under the class 'mental or behavioural disorder', which in turn falls under the class 'brain disease'. But EFO does not specify what it is that makes a brain disease can qualify as a mental or behavioral disorder.

II. PURPOSE AND METHOD

The Ontology of Schizophrenia (OS) aims to provide a formal representation of schizophrenia-related entities that are crucial to the treatment and study of schizophrenia. OS is developed in accordance with the OBO (Open Biomedical Ontology) Foundry principles and constructed in compliance with Basic Formal Ontology (BFO) as upper-level ontology and with the mid-level and domain ontologies that are built in conformance with BFO such as the Ontology for General Medical Science (OGMS) and Mental Functioning Ontology (MFO). OS is developed according to an initial proposal in [4] to ontologize mental disease on the basis of BFO, OGMS, and MFO.

As the embodiment of the realist methodology, BFO has been widely used in various domains, specifically in biomedical fields [2]. OGMS is intended to be used primarily in the development of clinical application ontologies, and focuses on the main types of entities involved in a clinical encounter such as 'disease', 'disorder', and 'disease course' [6]. MFO aims to represent all the aspects of mental functioning, including mental processes such as cognition and perception and qualities such as intelligence [8].

OS is developed using Protégé 4.3 and is implemented in OWL2. Having imported BFO2.0 OWL and the development version of OGMS, OS adds approximately 30 schizophrenia-related terms such as 'schizophrenia', 'pathological schizophrenia process', 'schizophrenia disorder' and 'schizophrenia course' (Figure 1).

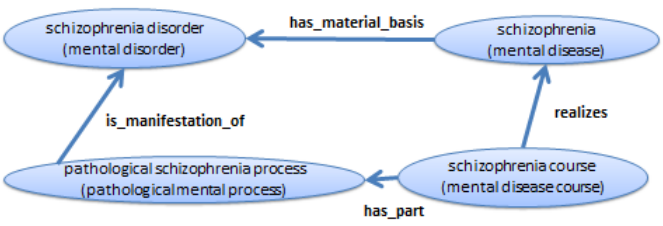

Fig. 1. Core OS terms and relations between them

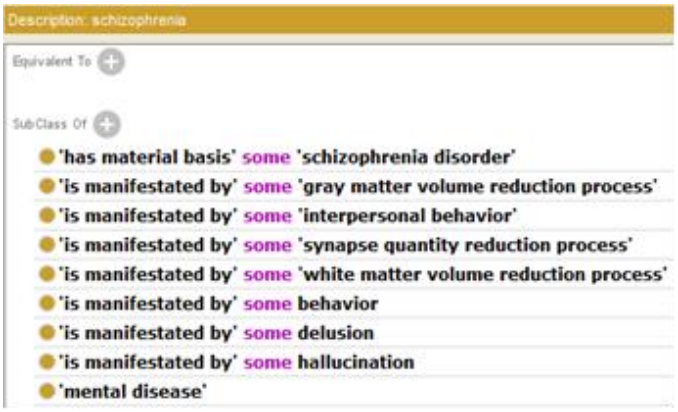

Fig. 2. Screenshot from Protégé showing the formal definition for the OS class 'schizophrenia'

OS attempts to provide both formal and textual definitions for each of these terms. Currently, some OS terms cannot be defined by providing a statement of necessary and sufficient conditions. For instance, given OS's framework, the class 'schizophrenia' cannot currently be defined in this way due to a lack of scientific consensus. The class is however specified via a formal definition through a set of subclass axioms (Figure 2). The classes 'gray matter volume reduction process', 'white matter volume reduction process', and 'synapse quantity reduction process' are created to represent excessive loss of gray and white matter and reduced numbers of synaptic structures on neurons, which have been repeatedly observed traits with schizophrenia [12, 13].

The classes 'delusion' and 'hallucination' fall into the class 'non-referring cognitive representation'. OS's way of handling cognitive representation and non-referentiality draws on recent discussions on the realist approach to aboutness [14].

OS terms are in addition annotated with ontology metadata such as labels, textual definitions, definition sources, term editors and editor notes. For instance, the class 'hallucination' is textually defined as follows: "A non-referring cognitive representation that is experienced without an external stimulus as if there were something perceived."

The current version of OS focuses exclusively on schizophrenia in part because many related kinds of mental disease – for instance schizoaffective disorders – cannot be specified in the ontology until schizophrenia itself has received an adequate treatment. However, many of the classes discussed so far are applicable to schizoaffective disorders.

III. CONCLUSION

A qualitative assessment of work on OS thus far might read as follows: OS unifies different existing definitions of schizophrenia into a set of manifestations of schizophrenia. In addition, the ontological characterization of schizophrenia in OS is more coherently aligned with BFO than is the case with other BFO-based ontologies, such as EFO. As an extension of BFO and OGMS, OS also fits well with the Neurological Disease Ontology (ND) [11]. More generally, OS has made a positive contribution to biomedical ontologies in the sense of being one of the few ontologies of mental disease.

Further development of OS includes: continued enlargement, especially adding terms relating to the treatment of schizophrenia; the specification of OS's relation with ND; and a quantitative assessment such as to what extent schizophrenia-related data can be mapped onto OS. With further bottom-up approaches, OS is highly expected to assist medical practitioners and researchers. For instance, OS has the potential to promote the treatment of schizophrenia patients by enhancing the International Psychopharmacology Algorithm Project (IPAP) Schizophrenia Algorithm [10], given that the BFO framework is able to capture, in a rigorous and computationally tractable way, all the types of entities represented in the IPAP Schizophrenia Algorithm [3].

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