Reuse and Enrichment for Building an Ontology for Obsessive-Compulsive Disorder

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Abstract

Building ontologies for mental diseases and disorders facilitates effective communication and knowledge sharing between healthcare providers, researchers, and patients. General medical and specialized ontologies, such as the Mental Disease Ontology, are large repositories of concepts that require much effort to create and maintain. This paper proposes ontology reuse and automatic enrichment as means for designing and building an Obsessive-Compulsive Disorder (OCD) ontology. The methods are demonstrated by designing and building an ontology for the OCD. Ontology reuse is proposed through ontology alignment design patterns to allow for full, partial or nominal reuse. Enrichment is proposed through deep learning with a language representation model pre-trained on large-scale corpora of clinical notes and discharge summaries, as well as a text corpus from an OCD discussion forum. An ontology design pattern is proposed to encode the discovered related terms and their degree of similarity to the ontological concepts. The proposed approach allows for the seamless extension of the ontology by linking to other ontological resources or other learned vocabularies in the future. The OCD ontology is available online on Bioportal.

Keywords
Ontology, OCD, Mental health, Conceptual modeling, Ontology enrichment, Ontology reuse,

1. Introduction

Obsessive-Compulsive Disorder (OCD) is a frequently debilitating and often severe mental health disorder that affects approximately 2% of the population[1]. The Royal Collage of Psychiatrists (RCPSYCH[2]) report that approximately 1 in every 50 people suffer from OCD at some point in their lives, amounting to about 1 million people in the UK, affecting men and women equally. It is also noted [1] that people could spend a long time struggling with the disorder, often hiding their symptoms, before they get appropriate help, possibly attributed to the shame or stigma associated with having disturbing thoughts (e.g. ego-dystonic sexual or violent) and compulsive behaviours . Coding information in electronic health records (EHR) using standard medical terminologies, such as the Systematized Nomenclature of Medicine Clinical Terms (SNOMED CT) [2] can facilitate the efficient recording and integration of patient notes, ultimately leading

[1]https://bestpractice.bmj.com/info/

[2]https://www.rcpsych.ac.uk/mental-health/problems-disorders/obsessive-compulsive-disorder

to more effective healthcare management [3]. However, existing clinical terminologies, such as SNOMED CT, and ontologies, such as the Mental Disease Ontology (MDO)[3] are limited with respect to the following dimensions.

1. **Semantic Richness:** Existing ontologies are evolving. No specific ontology or (sub-ontology) exist that delineates OCD, its types and diagnosing symptoms; enough for example to distinguish it from other related disorders, such as *illness anxiety disorder* or *hoarding disorder*[4]. The creation and evaluation of such resources is a costly exercise that requires both domain and technology experts.

2. **Semantic Heterogeneity:** The meaning and provenance of the terms or concepts used in these resources are not usually included or described in sufficient detail to explain the basis of its association with a particular classification hierarchy. For example, in Disease ontology (DOID), OCD is described as a type of *Anxiety Disorder*, whereas this classification has been updated in the DSM-5 revision in 2013, where it is now classified as a type of *Obsessive-Compulsive and Related Disorders (OCRDs)*. Also, different classification hierarchies for the same concept is used in different ontologies. For example, *Obsession* in the MDO is a type of *Pathological Mental Process*, whereas it is a type of *Behavioral Symptom* in the Medical Subject Headings ontology (MeSH)[5], and a type of *Content of Thought* in SNOMED-CT. Understanding and establishing the similarity of concepts across ontologies is a well-known research challenge.

3. **Structural Richness:** Most clinical terminological resources and ontologies are described primarily with subsumption (IS_A) relationships and presented as large class hierarchies of concepts. The uncontrolled use of IS_A relation to signify different types of relations (such as PART_OF, IS_INSTANCE_OF, IS_ASSOCIATED_With, etc.) has been noted, e.g. in SNOMED CT [4], leading to structural overload and possible incorrect subsumption relationships. Also, some modelling constructs such as the use of multiple inheritance, where a concept can have multiple parent types, can lead to complexity in reasoning with the information.

In this paper, we propose the development of an OCD ontology to address some of the issues noted above. The methodology for development follows established proposals in the literature. In particular, structural richness is addressed by making use of rich ontological modelling in the logical definition of concepts, whilst semantic heterogeneity is addressed by the reuse of existing resources directly in the ontology as well as creating explicit reference to related concepts in other resources. Semantic richness is addressed by complementing the definition of concepts in the ontology by the automatic discovery of related concepts from relevant resources using deep learning. Ontology design patterns are proposed to encapsulate the links to other ontologies and the discovered related concepts. The resulting ontology consists of 97 classes (expanded to 1047 classes from other ontologies), 17 object properties (relationships) and 5 data properties. Bio_ClinicalBERT[6] and a text corpus from an OCD discussion forum are used in the ontology enrichment task. This work contributes to the efforts of building biomedical

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[3]http://purl.obolibrary.org/obo/MFOMD.owl
[4]https://www.rcpsych.ac.uk/mental-health/problems-disorders/obsessive-compulsive-disorder
[5]https://www.nlm.nih.gov/mesh/meshhome.html
[6]https://huggingface.co/emilyalsentzer/Bio_ClinicalBERT

ontologies, by providing modelling solutions that allow for the integration, reuse and enrichment of the ontological resources. The developed OCD ontology is available on Bioportal[7]. The remainder of the paper is organized as follows. Some related works on biomedical ontology development, reuse and enrichment are reviewed in section 2. Section 3 describes the proposed OCD ontology and outlines details of its development process. Ontology design patterns for reuse and enrichment are presented in section 4 , followed by conclusions in section 6.

2. Related Work on Ontology Reuse and Enrichment

A brief overview is given here of efforts in the field of biomedical ontologies reuse and enrichment.

An overview of the list of prominent biomedical ontologies is given in [5]. El-Sappagh et al [6] reviews the limitation and complexity of large clinical terminology resources such as SNOMED-CT and proposes its transformation to an ontology by aligning and reusing concepts from the Ontology of General Medical Science (OGMS). Top-level concepts are first manually mapped to OGMS, and then used to compose more refined (pre-coordinated and post-coordinated) concepts. More recently, the integration of ontological resources has become the focus of attention. For example,the Mondo Disease ontology aims to harmonize disease definitions across the world [8] by integrating the classifications and relationships of commonly used disease ontologies into a single semantically coherent resource. It employs a Bayesian approach to ontology structure inference, combining deductive and probabilistic inference, and aims to provide equivalence mappings with precise annotations. Another example of creating a large ontology, demonstrating the complexity and scale of the required effort, is demonstrated in the development of CIDO: the community-based coronavirus infectious disease ontology [7]. The methodology for development of the ontology is based on following the OBO Foundry ontology development principles, and utilizing the eXtensible Ontology Development(XOD) strategy, which prescribes: ontology term reuse, semantic alignment, use of ontology design patterns for new term generation, and community effort. A largely manual effort is ongoing into the development and visual analysis and evaluation of the resulting ontology. The process of encoding logical definitions manually when developing ontologies is a challenging task [8]. Ontology Design Patterns (ODPs) are defined as reusable modeling solutions to frequently occurring ontology design problems and are proposed as a useful tool to address this challenge. The complexity of the ontologies and the need for checking their consistency was investigated in [9]. Using the Foundational Model of Anatomy ontology, they analyzed the musculoskeletal content and show the inconsistencies in the use of relations, lack of definitions of relations, and incomplete definition of the hierarchy. They suggested the definition and use of ODPs to address these issues. Recent approaches are proposing the integration of ontologies by transforming them into a unified knowledge graph, that can be homogeneously queried with SPARQL endpoints [10]. Representing the ontologies as an RDF resource including all its entailment (all consequences of its logical definition) can help in the process of checking the similarity and consistency of the integrated resources.

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[7]https://bioportal.bioontology.org/ontologies/OCD
[8]https://mondo.monarchinitiative.org

Ontology enrichment is a term that has been used in the literature from two points of view: a) enriching the ontology itself; that is extending the ontology by supplementing its existing structure with related terms and metadata, and b) enrichment by ontology; where the ontology is used as a source for discovering related concepts in a particular domain for a particular purpose. A common example of the latter task is the Gene Ontology (GO) term enrichment; a technique for interpreting sets of genes by making use of the GO system of classification, in which genes are assigned to a set of predefined bins depending on their functional characteristics [11].

In this paper, we are concerned with the first view point; enriching the ontology itself. Few research works have addressed this problem by utilizing existing resources, or other generic resources. For example, [12] used UMLS as the source of discovering synonyms for concepts in the ontology. In [13], deep learning with a large corpus of PubMed review articles and veterinary clinical notes was used to discover related terms to some pre-defined terms related to medical conditions. They then use the results to populate the UMLS Semantic Types and Groups ontology[9], whilst relying on specialized ontologies to represent the relatedness (e.g. lexical and provenance) relationships and properties. Utilization of standardised methods of linking ontologies to lexicographic resources[10] is an important aspect of this work. The research area of using ontologies and machine learning is still novel. An overview of how semantic similarity measures and ontology embeddings may be incorporated with ML methods is given in [14]. Further work needs to be done on exploiting the ontology structure, possibly by ontology embedding, in the task of ontology enrichment.

Several methodologies have been proposed to guide the process of developing ontologies. The NeOn [15] methodology places emphasis on collaboration and distributed development. It encourages the modularization of ontology building process, where domain experts contribute to different modules, while ensuring the overall consistency of the ontology. Systematic Approach for Building Ontologies (SABiO) [16] is a related approach that suggests a more guided workflow for the ontology development process, where the design of reference domain and operational ontologies is suggested and the reuse of foundational ontologies and pattern-oriented reuse are encouraged. The Open Biological and Biomedical Ontologies (OBO) Foundry proposed a set of principles and guidelines [17] for the development of ontologies to promote interoperability and standardization in the life sciences domains. OBO ontologies need to adhere to common design patterns and share a foundational set of relations, thereby fostering seamless integration and facilitating collaboration across diverse biomedical domains. The the eXtensible ontology development (XOD) methodology [18] is designed to be flexible and adaptable, allowing ontology developers to extend and customize it according to the unique characteristics of the target domain. By embracing a modular approach, XOD promotes the reuse of existing ontological components, which minimizes duplication efforts and ensures consistency across the ontology. In this paper, we build upon SABiO and XOD to demonstrate the reuse and enrichment of the OCD ontology. We incorporate elements from foundational ontologies, domain-specific ontologies, and external sources to enhance the OCD ontology, thus showcasing the potential of leveraging existing resources to enrich and expand knowledge representation effectively.

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[9]https://lhncbc.nlm.nih.gov/ii/tools/MetaMap/documentation/SemanticTypesAndGroups.html
[10]https://www.w3.org/2019/09/lexicog/

3. The OCD Ontology Development Process

An overview of the ontology development stages is shown in Figure 1. In this paper, we present an overview of ontology building process. The ontology evaluation tasks include, a) evaluating the logical consistency of the ontology; checked using the DL reasoners in Protégé , b) evaluating the capability of the ontology to answer a set of competency questions defined in the knowledge acquisition phase; checked by formulating the questions using the SPARQL query language, and c) expert evaluation to judge the quality and coverage of the ontology. Detailed account of these tasks are left to future publications.
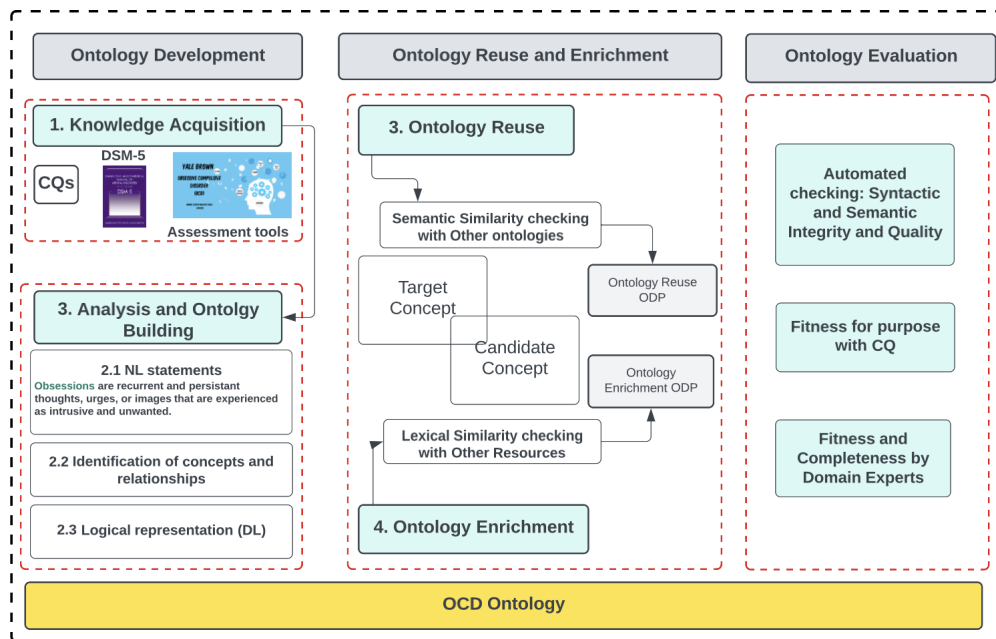

Figure 1: Ontology Development and Evaluation

Knowledge Acquisition: Two primary resources were used to acquire basic knowledge about the nature of the disorder and its diagnosis. These are the Diagnostic and Statistical Manual of Mental Disorder version 5 (DSM-5)[19] and OCD assessments tools, namely, the Yale-Brown Obsessive Compulsive Scale (Y-BOCS)[11] and the Obsessive-Compulsive Inventory (OCI) [12]. Statements describing OCD and its related concepts were manually extracted; 28 statements were extracted. Examples include, "OCD is characterized by obsession, compulsion, or both" and "the definition of obsession as an intrusive thought, image, or urge"; (both examples are from DSM-5). Y-BOCS, and other diagnostic tools, were particularly useful for identifying types of OCD and providing specific values of defined concepts. Eight types of *Obsession* and six types of *Compulsion* were identified. To further refine the definition of concepts that are not described in the primary resources, further relevant resources were employed. For example, the

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[11]https://pcl.psychiatry.uw.edu/wp-content/uploads/2021/12/YBOCS.pdf
[12]https://www.div12.org/wp-content/uploads/2015/07/OCI.pdf

cognitive theory of OCD [20] emphasise that an intrusive thought transforms into an obsession when an additional meaning is attributed to it. A total of 35 statements were extracted (a full list can be found in GitHub repository "OCD-ontology". In this phase, we also compiled a set of competency questions (n=23) from diagnostic and other relevant resources to support the development and evaluation processes. Examples of the competency questions include: What patterns of activities do a person with aggressive thought has? and What is the type of obsession where avoidance behavior is a frequent occurrence?.

The Analysis Phase: Statements in the previous phase were used to identify relevant concepts and relationships. Statements for defining specific concepts were grouped and used to formulate a natural language definition that describe three questions: "what is the concept defined as?", "what are its types?", "what are its symptoms?". During this process, necessary and sufficient criteria for defining the concepts were identified. These refer to the properties (attributes and relationships) that must be present for an instance to be considered a member of the defined class. A natural language statement defining these properties is then formulated to allow for its transformation into logical statements in Description Logic (DL) and further definition in the ontology. An example of this process for the definition of the concept of *Obsession* is shown in Table 1. This approach allowed for a clear definition of the required concepts and for avoiding ambiguity and inconsistency in the representation by utilising the ontology reasoning tools over the defined ontology model. The analysis phase resulted in the definition of a set of 97 concepts and 22 types of relationships. This includes 17 object properties and 5 data properties.

Table 1
Example of the definition of the concept of *Obsession* in the analysis phase.

Statements relevant the concept of Obsession (1) Obsessions are recurrent and persistent thoughts, urges, or images that are experienced as intrusive and unwanted (DSM-5). (2) Individual with obsession attempts to ignore or suppress such thoughts, urges, or images, or to neutralize them with some other thoughts or actions (i.e., by performing a compulsion) (DSM-5) (3) Individual with OCD may experience over-importance of thoughts [20]. (4) There are 8 types of obsession (Y-BOCS).
Natural Language Definition In OCD, an Obsession can be any of: Intrusive Thought, Intrusive Image or Intrusive Impulse or Urge, that causes distress due to the added importance that the individual places on them. In OCD, Obsession is often accompanied by some Compulsions.
Description Logic Expression   ((Obsession ≡ Intrusive thought ⊔ Intrusive image ⊔ Intrusive urge)   ⊓ (∃ hasAssociatedAppraisal.ThoughtAppraisal))
Related Identified Concepts *thought*, *intrusive thought*, *persistent thought*, *mental image*, *urge* and *thought appraisal.*

4. Ontology Design Patterns for Reuse and Enrichment

Reusing Existing Ontologies Related ontologies were identified by searching the NCBO BioPortal [13]; a comprehensive repository of biomedical ontologies. For every concept defined

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[13]https://bioportal.bioontology.org/

in the analysis phase, corresponding concepts were identified in the existing ontologies. The following heuristics were employed in the decision to reuse concepts.

1. The external concept is considered to be fully equivalent to the required OCD concept, if there is a complete overlap between the logical definitions of the two. In this case, the concept is imported directly to the ontology. When a concept is imported, all its related concepts, including its inheritance tree hierarchy, are also imported. For example, the *Activity* and *Symptom* classes are root classes in the Activity of Daily Living (ADL) ontology [21] and Symptom Ontology (SYMP) [14], respectively. Importing both classes in the OCD ontologies implies the use of their complete ontologies as well.

2. The external concept is considered to be partially equivalent to the required OCD concept, if its logical definition can be considered part of the definition of the required concept. For example, "OCD" is defined in the DOID ontology as a subclass of "Anxiety Disorder". No further definition is given in the DOID ontology. This definition is partially sufficient for our ontology and we need to further refine it. Hence, instead of importing the class and redefining it, we align our definition with the external ontology using the OWL:equivalentClass; an example is, ocd:OCD ≡ DOID:OCD (where "ocd" and "DOID" are prefixes for the OCD and the Disease Ontology, respectively). This ontology alignment design pattern allows flexibility of ontology specification, whilst also reusing existing resources. There are 13 OCD concepts aligned as equivalent to external concepts. Figure 2 illustrates the refinement of the definition of the class "Obsession" in the OCD ontology. The definition of the "Obsession" in MDO is defined as: MDO:MFOMD_0000109 ⊑ MDO:Pathological mental process; which is defined as (⊑ OGMS:Pathological bodily process ⊓ (⊑ ∃ manifestationOf.Mental disorder)) . This definition is reused in our ontology as follows: ocd:Obsession ≡ MDO:MFOMD_0000109 (class obsession from MDO). The refinement of ocd:Obsession is presented as follows: ocd:Obsession ≡ Intrusive thoughts ⊔ Intrusive image ⊔ Intrusive urge) ⊓ (∃ hasAssociatedAppraisal.ThoughtAppraisal); ocd:Obsession has 8 sub-classes.

3. The external concept is considered to be nominally similar to the required OCD concept, if there is some overlap between the logical definitions. In this case, we are unable to reuse the external class, but we maintain a link to it using the *Reuse Ontology Design Pattern*, as shown in figure 3.

The set of ontologies that were reused are as follow: Mental Disease Ontology (MDO)[15], Mental Functioning Ontology (MFO)[16], ADL, SYMP, Gene Ontology [17], SNOMED-CT, Experimental Factor Ontol- ogy (EFO), Gender, Sex,and Sexual Orientation(GSSO) ontology, Emotion ontology [18], and the Basic Formal Ontology (BFO)[19].

In the OCD ontology, classes that were not present in existing ontologies were created and mapped to classes in the Basic Formal Ontology (BFO) using the OWL:subClassOf relationship. The mapping process took into account the characteristics of each class in the BFO and

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[14]https://obofoundry.org/ontology/symp.html
[15]http://purl.obolibrary.org/obo/MFOMD.owl
[16]http://purl.obolibrary.org/obo/MF.owl
[17]https://bioportal.bioontology.org/ontologies/GO
[18]http://purl.obolibrary.org/obo/MFOEM.owl
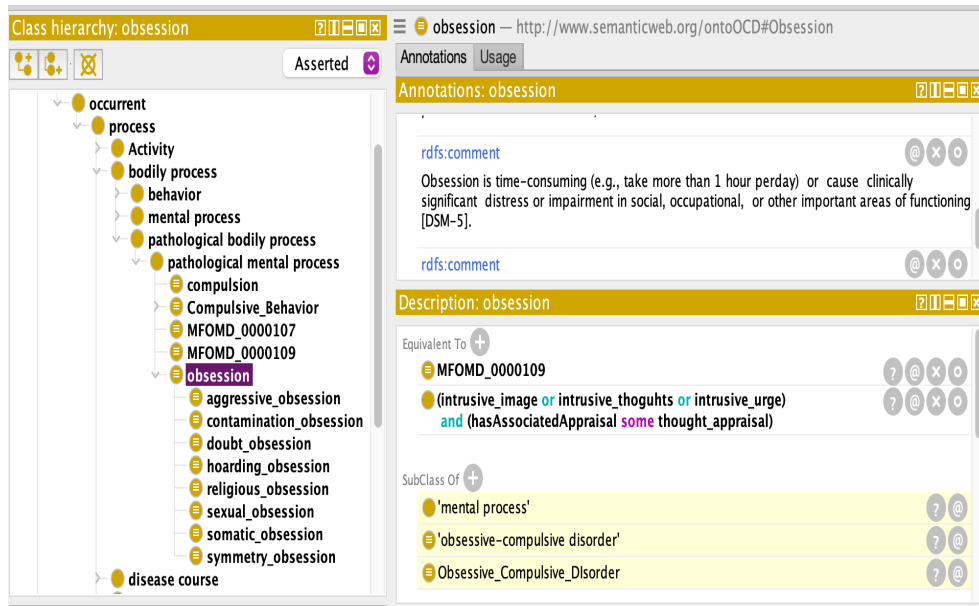[19]http://purl.obolibrary.org/obo/bfo.owl

Figure 2: The representation of the class "Obsession" defined in protégé .
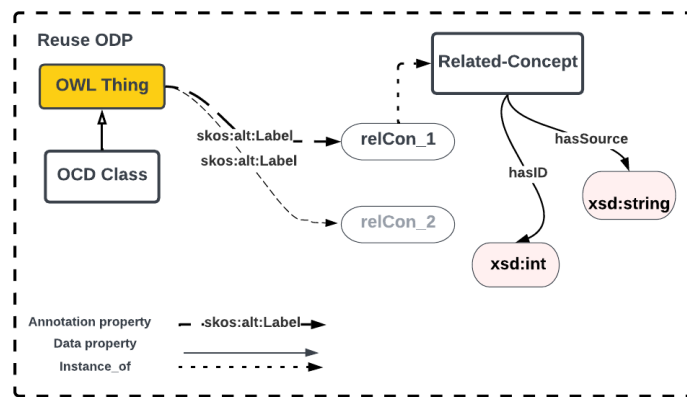

Figure 3: The Reuse Ontology Design Pattern.

determined the most suitable parent class for the OCD classes. A recent study by Emeruem et al. [22] proposed an automatic tool for mapping classes to the BFO. The aforementioned tools were used to guide the mapping of three classes in the OCD ontology: ocd:Severity Level, ocd:Assessment Criteria and ocd:Functional Impairment. Figure 4 illustrates the mapping of class "Functional Impairment" as ⊑ BFO:Quality.

Ontology Enrichment with Deep Learning

The objective here is to demonstrate how to automate the process of extracting related terms using deep learning from a given corpus for integration into the ontology. We first explore the

Figure 4: The representation of class "ocd:Functional impairment" as ⊑ BFO:Quality based on the "Questions History".

efficacy of the BioClinical_BERT langugage model in identifying terms that are semantically similar to target terms in the ontology. BioClinical_BERT leverages contextual embeddings to capture the nuanced meaning of biomedical and clinical terminology. It is trained on a large corpus (2 million) of clinical notes. We then employ a word2vec model trained on an OCD-specific corpus to assess term similarity (cosine similarity). The word2vec model learns the distributional representation of words based on their co-occurrence patterns within the OCD corpus. *Candidate term extraction using BioClinical_BERT*: Target terms are mapped to their corresponding token IDs in the model and their hidden states are retrieved, representing the contextualized embeddings of the tokens. Similarity scores are then computed between the target term's contextualized embedding and the embeddings of other terms in the model. The top (n) terms with the highest similarity scores are then selected. A sample of candidates terms and similarity scores to target terms are listed in table 2. Notably, it was observed that certain target terms such as "compulsion", "intrusive" and "impairment" were not represented in the BioClinical_BERT model. *Candidate terms form the Word2vec model:* Here we utilized the word2vec model with the Continuous Bag of Words (CBOW) architecture to obtain a list of relevant terms for the same target terms in the ontology. Data for this study was collected from an OCD forum[20]. Python Selenium library was employed to gather a substantial dataset comprising 54,410 posts from the forum spanning the period from October 2000 to December 2021 . The experimental set-up; hyperparameter configuration, was as follows. The window size determines the range of neighbouring context words considered for each target word, and was set to 5. This means that words within a distance of 5 from the target word were taken into account. Additionally, a minimum count of 3 was defined, ensuring that words appearing at least three times in the dataset were included during training. Cosine similarity was then

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[20]Online platform dedicated to OCD-related discussions and information exchange https://www.mentalhealthforum.net/forum/forums/obsessive-compulsive-disorder-ocd-forum.46/

computed between the target and candidate terms and the top(n) terms were selected. A sample of results is shown in table 3.

Table 2
Top similar terms to target terms from Bio_Clinnical BERT model

Target term	BioClinical_BERT (Top 6 terms)
obsession	('obsessed', 0.651), ('fascination', 0.636), ('urges', 0.588),('irrational', 0.573,), ('insistence', 0.570) ('preoccupied', 0.569)
urge	('urging', 0.573), ('urgency', 0.535), ('encourages',0.519), ('invite', 0.496), ('desire', 0.49), ('obsession', 0.489)

Table 3
Top similar terms to target terms from Word2Vec model.

Target term	Word2Vec
obsession	('theme', 0.984), ('behaviour', 0.983), ('compulsive', 0.983), ('fear', 0.979), ('trigger', 0.974)('habit', 0.968)
compulsion	('rumination', 0.987), ('pattern', 0.986), ('behaviour', 0.984), ('ritual', 0.979), ('activity', 0.974), ('action', 0.974).
urge	('reflex', 0.988), ('mindfully', 0.984), ('opposite', 0.979), ('reaction', 0.976), ('deliberately', 0.976), ('act', 0.975)
intrusive	('harm', 0.9782), ('triggered', 0.967), ('unwanted', 0.965), ('invasive', 0.965), ('horrific', 0.965), ('disturbing', 0.962)
impairment	('indicative', 0.962), ('distress', 0.954), ('jealousy', 0.944), ('inexplicable', 0.940), ('deliberate', 0.938), ('negativity', 0.917)

An Enrichment ODP is proposed here, as in figure 5, to record the results. As shown in the figure, classes in the ontology can be associated with many alternative labels, whose properties, including, similarity score, method and date are also recorded. The pattern is a simple generic and flexible approach to associating multiple terms to the ontology. A more sophisticated approach to lexical representation of associated terms can also be envisaged, e.g. by employing the OntoLex ontology. This is the subject of ongoing work.

5. Conclusion

The process of defining an ontology is costly in terms of time and effort. Devising means of automating the process to complement the traditional approach will be of benefit to all stakeholders. This paper presents an approach for building an ontology for a specific mental disorder. The aim is to demonstrate how the traditional ontology building process can be complemented with a process of reusing existing ontology resources and enrichment with rich textual resources. The paper considers the building of an ontology for a specific mental disorder, as an example, but the approach proposed is generalisable to other use cases. A uniform approach is presented to enriching the ontology with ontological concepts and lexical terms using ontology design patterns. The degree of similarity of concepts in the ontologies guide the modelling process of the related concepts. Machine learning is used to discover similar terms to concepts
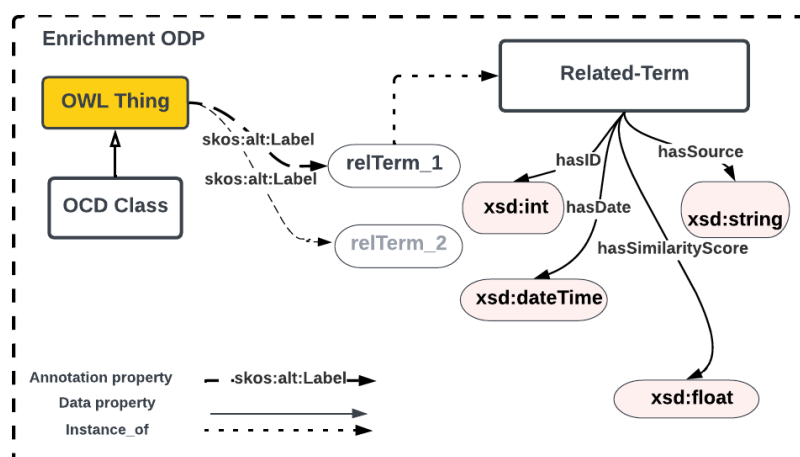
Figure 5: The Enrichment Ontology Design Pattern.

in the ontology using language models trained on large text corpus. The degree of similarity with the lexical terms are explicitly encoded. Results from the application of methods on two different corpora is presented. The paper outlines the approach and sets the way for further work on several fronts; refactoring the patterns to allow for richer modelling of lexical similarity, further refinement of the logical definition of the ontology based on expert evaluation. The detailed processes of building the ontology and its evaluation are the subject of future publications.

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