

# Scoping Review of Ontologies in the Behavioral Sciences

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## Abstract

**Background:** Ontologies have been widely studied in terms of how they are defined, created, evaluated and applied. Less is known about ontologies specific to the behavioral sciences.

**Objectives:** The aim of this scoping review is to illustrate the patterns, trends and volume of literature on ontologies in the behavioral sciences.

**Eligibility criteria:** Published reviews and evaluations, including book chapters in the behavioral sciences. Foreign language papers and grey literature were excluded, as were papers published before 2010.

**Sources of evidence:** Databases searched were: Ovid Medline, Embase, PsycINFO, Web of Science, and Epistemonikos. All searches were conducted on July 6<sup>th</sup>, 2021. Additional searches were conducted in PsycINFO and Web of Science to identify book chapters

**Charting methods:** The charting of documents was conducted independently by a single reviewer using two pre-defined tables to chart characteristics of the publications and ontologies identified in the included studies

**Results:** From 6,257 screened records, 231 full text studies were examined. The final number of included studies was 50, and the number of identified ontologies was 49.

**Conclusions:** This scoping review shows that published reviews and evaluations in the behavioral sciences are relatively few. The studies identified do, however, provide information on the ways in which ontologies have been studied, namely how they have been developed, evaluated and applied. The included studies illustrate the areas of behavioral science which have ontologies, and what their primary characteristics are.

## Introduction

### Rationale

In philosophy, ontology is the study of being or existence. In the world of information and data science, ontologies have been defined by many scholars<sup>1,2</sup> but they are essentially the structured representation of a body of knowledge. They are part of a family of classification systems which includes indexes, thesauri, vocabularies, concept maps and knowledge bases. While some systems provide definitions and a hierarchical structure of terms, ontologies also show the relationships between concepts thus providing a more nuanced picture and additional information to facilitate the understanding of complex bodies of knowledge.<sup>3</sup> In the field of life science and biomedicine, ontologies are used to advance the study of particular fields from the very broad (human disease), to the very specific (interventions for specific conditions). The field of ontology literature is a very large one, covering all aspects from definitions, descriptions of ontologies in specific areas or study, the methods, use of software, programs, and techniques involved in ontology creation and evaluation, and the many specific applications. Biomedicine in particular has been well studied<sup>4</sup>, and ontologies are now making major contributions to research, and in clinical settings. This scoping review aims to extract published reviews and evaluations pertaining to the behavioral sciences.

### Objectives

The aim of this scoping review is to illustrate the patterns, trends and volume of literature on ontologies

in the behavioral sciences. The means of obtaining this information was through the identification of published evaluations, published reviews, and other relevant published papers. This review is part of a wider endeavor being undertaken by an ad hoc Committee of the National Academies of Sciences, Engineering, and Medicine who are drawing upon the broader literature to inform the development of an ontology for the behavioral sciences. The more specific questions this review seeks to answer are: 1) By what methods have ontologies been studied? How might the strengths and limitations of the existing body of work be characterized? 2) What are well supported findings from this work, especially candidate best practices? 3) is it possible to identify categories/types of behavioral ontologies? 4) Are there identifiable patterns in which behavioral fields do/do not have/use ontologies?

### **Methods**

This scoping review follows the reporting guidelines outlined in the PRISMA Extension for Scoping Reviews (PRISMA-ScR) checklist.<sup>5</sup>

#### **Eligibility Criteria**

Articles eligible for inclusion in this review were published reviews and evaluations, including book chapters. Foreign language papers and grey literature were excluded, as were papers published before 2010. Only articles pertaining to the behavioral sciences were sought. Records were screened in two stages: At the title/abstract stage, records were included that were: 1) Relevant to topic of this review and 2) In English. Criteria for including papers at the full text stage were 2) Paper was a published review or evaluation; 3) The review or evaluation was on single or multiple ontologies; 4) The ontology is in the field of behavioral science. Articles were excluded if 1) They were not published; 2) They did not review the characteristics of a specific ontology, or ontologies in general, OR if they did not contain information pertaining to the evaluation of ontologies, and 3) If the paper was from a discipline deemed not relating to the behavioral sciences.

#### **Information Sources**

The following electronic databases were searched for potentially relevant articles: Ovid Medline, Embase, PsycINFO, Web of Science, and Epistemonikos. All searches were conducted on July 6<sup>th</sup>, 2021. Additional searches were conducted in PsycINFO and Web of Science to identify book chapters. Further articles were sought using PubMed Similar Articles. Records from preliminary searches run for this review were also scanned for relevance.

#### **Search**

A combination of subject headings and free text terms were used to represent the concept of ontologies; terms were applied to limit records to published review articles and evaluations. Due to the broad scope of the field of behavioral science no terms were used to narrow the search further, however, searches were limited to English language and from 2010 to August 2021. Copies of all the search strategies with details of database platforms and dates can be found in Appendix 1.

#### **Selection of sources of evidence**

All records were screened in EndNote and assessed for relevance based on the title and abstract. Potentially relevant articles were reviewed in full-text based on the specific eligibility criteria.

#### **Data Charting Process**

All charting of documents was conducted independently by a single reviewer using two pre-defined tables to chart characteristics of the publications and ontologies identified in the included studies.

## Data Items

The elements sought to inform this review were divided into the characteristics of the ontologies identified: 1) Name; 2) category/domain; 3) Number of classes or terms, and the characteristics of evaluations: 1) method of evaluation; 2) strengths and weaknesses, and 3) recommendations or best practices resulting from evaluation.

## Synthesis of results

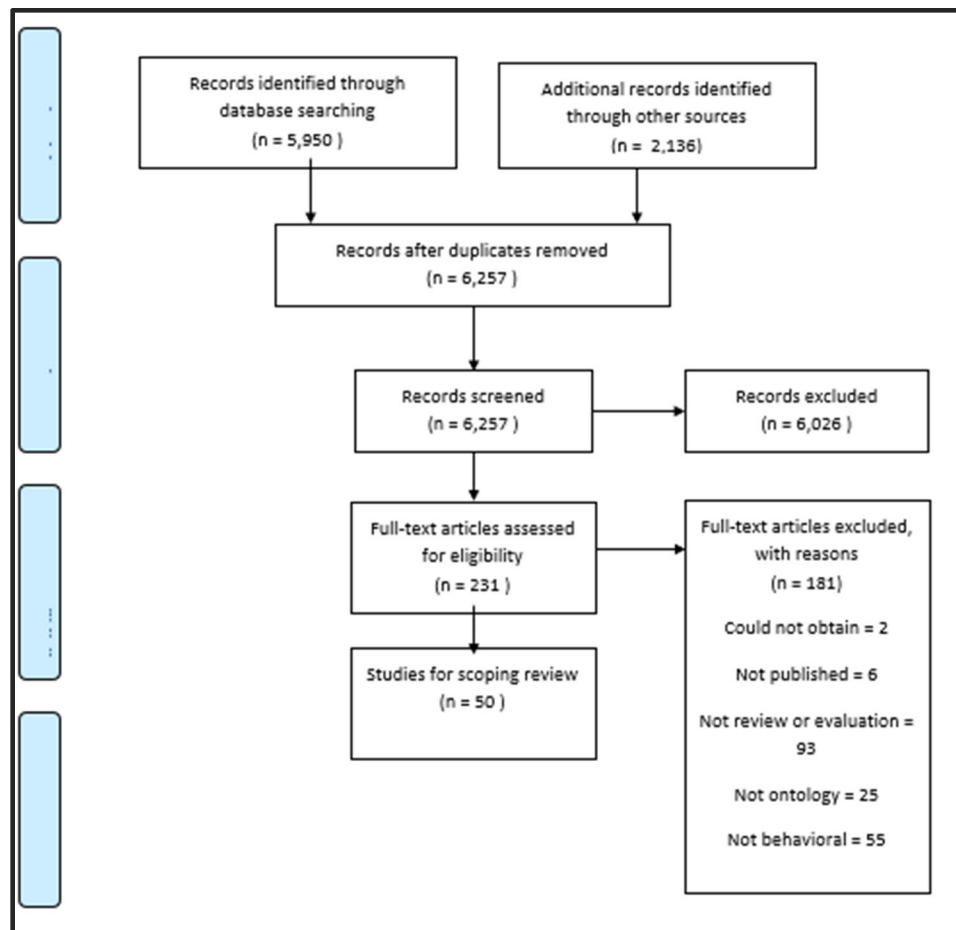
The results are described narratively, summarizing what has been found in terms of 1) the number and characteristics of ontologies found through the reviews and evaluations, 2) the methods used to evaluate ontologies, and 3) what best practices and recommendations for evaluation have been identified.

## Results

### Selection of Sources of Evidence

The number of studies screened in order to obtain the studies summarized in this review is shown in the PRISMA Flow Diagram in Figure 1. The total number of records screened at the title and abstract stage was 6,257, from which 6,026 were excluded and 231 full text studies were examined. The final number of included studies was 50.

**Figure 1:** PRISMA flow chart for the identification of studies



## Characteristics of Sources of Evidence

The number of ontologies retrieved through the studies reviewed was 49 broken down into the following categories: behavioral (n= 16), phenotypes (n=6), disease and mental health conditions (n= 13), genetics (n=4), neuroscience (n= 10). The number of classes in each ontology ranged from 167,138 (National Cancer Institute Thesaurus) to 41 (EmotionsOnto). Table 1 contains a list of all of the ontologies identified through reviewing full text articles. It contains the name and acronym of the ontology, the number of classes where available, and the study from which the ontology was identified (not necessarily the original paper since this scoping review primarily drew upon secondary sources). Figure 2 shows the number of ontologies by category and Table 2 contains the characteristics of the included studies. A brief summary of the results is given here, and then summarized in more detail under the headings of: 1) Reviews 2) Development and Authorship 3) Methods of Evaluation and Assessment 4) Tools and Software 5) Applications of Ontologies 6) Recommendations for Best Practice. Figures 4 and 5 are wordclouds generated from terms in the behavioral ontologies table and from words relating to the evaluation of ontologies.

### Brief summary of included studies

- \* Studies that reviewed ontologies in general were: Aminth 2018,<sup>6</sup> Blanch 2017,<sup>7</sup> Hastings 2012,<sup>8</sup> Konopka 2015,<sup>4</sup> Larsen 2017,<sup>9</sup> Lokker 2015,<sup>10</sup> Norris 2019,<sup>11</sup> Poldrack 2016,<sup>12</sup> Reed 2015,<sup>13</sup> Stancin 2020,<sup>14</sup> and Zhu 2015.<sup>14</sup>
- \* Studies that examined or included methods of evaluation were: Dissanayake 2020,<sup>15</sup> Duque Ramos 2014,<sup>16</sup> Franco 2020,<sup>17</sup> Hastings 2014,<sup>18</sup> Jung 2016,<sup>19</sup> Kronk 2020,<sup>20</sup> Malhotra 2014, Norris 2019,<sup>11</sup> and Quinn 2018.<sup>21</sup>
- \* Studies that described individual ontologies were: Brenas 2019,<sup>22</sup> Eisenberg 2018,<sup>23</sup> Gkoutos 2012,<sup>24</sup> Hastings 2014,<sup>18</sup> Hicks 2016,<sup>25</sup> Jensen 2013,<sup>26</sup> Jung 2016,<sup>19</sup> Kohler 2012,<sup>27</sup> Kronk 2020,<sup>20</sup> Malhotra 2014,<sup>28</sup> Quinn 2018,<sup>21</sup> Win 2019,<sup>29</sup> Woznowski 2018,<sup>30</sup> and Zaveri 2010.<sup>31</sup>
- \* Studies that described ontology tools were: Aminth 2019,<sup>32</sup> Cui 2015,<sup>33</sup> Duque Ramos 2016,<sup>34</sup> Eivazzadeh 2016,<sup>35</sup> Gkoutos 2015,<sup>36</sup> Kohler 2011,<sup>37</sup> Toralf 2011,<sup>38</sup> and van Damme 2018.<sup>39</sup>
- \* Studies that described the development of ontologies were: Brenas 2019,<sup>22</sup> Ceusters 2017,<sup>40</sup> Eisenberg 2018,<sup>23</sup> Gkoutas 2012,<sup>24</sup> Hicks 2016,<sup>25</sup> Jensen 2013,<sup>26</sup> Kohler 2012,<sup>27</sup> Kola 2010,<sup>41</sup> Norris 2021,<sup>42</sup> Rousseau 2018,<sup>43</sup> Vigo 2014,<sup>44</sup> and Win 2019.<sup>29</sup>
- \* The metrics used to describe ontologies was outlined by Yao 2011,<sup>45</sup> and Vigo 2014<sup>44</sup> 2014 described the problems in ontologies and recommendations for correction.

**Table 1: Table of Ontologies Identified from Reviewed Studies**

Ontology	Number of Classes/Terms	Domains	Source
<b>BEHAVIORAL</b>			
Standard Animal Behavior Ontology (SABO)	NA	Animal behavior	Gkoutos 2015
Neuro Behavior Ontology (NBO)	1,036	Behavioral processes and phenotypes	Norris 2019
Health Behaviour Change Ontology (HBCO)	92	Behavior change and automated dialogue systems	Norris 2019
Behaviour Change Techniques (BCT)	110	Behavior Change	Blanch 2017
Persuasion Support Systems (PSS) for Health Behavior Change	NA	Behavior Change	Win 2019
Ontology of Behavior Change Counseling Concepts	NA	Behavior change counseling	Bickmore 2011
Ontology of Self-Regulation	NA	Self-Regulation	Eisenberg 2018
Cognitive Atlas (COGAT)	3,639	Cognitive neuroscience and mental processes	Norris 2019
Cognitive Paradigm Ontology (COGPO)	400	Cognitive and behavioral experiments	Norris 2019
EmotionsOnto	41	Emotions	Norris 2019
Emotion Ontology (MFOEM)	902	Emotions	Norris 2019
Exposure Ontology (ExO)	148	Exposure science, genomics and toxicology	Norris 2019
Lifestyle Ontology	NA	Lifestyle concepts	Benmimoune 2015
OntoPsychia	1,450	Social and environmental determinants for psychiatry	Blanch 2017
Semantic Mining of Activity, Social and Health Data (SMASH)	87	Health-care data and sustained weight loss	Norris 2019
Mental Functioning Ontology (MF)	692	Mental functioning and mental processes	Norris 2019
<b>PHENOTYPES</b>			
Autism Spectrum Disorder Phenotype (ASDPTO)	284	Autism Spectrum Disorder Phenotype	Aminth 2018
Human Phenotype Ontology	13,000	Phenotypes	Gkoutos 2015
Mammalian Phenotype Ontology (MPO)	1,528	Phenotypes	Kohler 2012
Phenotype and Exposures (PhenX)	533	Phenotypes	Blanch 2017

Measurement Method Ontology	701	Methods used to make qualitative and quantitative clinical and phenotype measurement	Yu 2016
Phenotype and Trait Ontology (PATO)	5,607	Biodiversity and Ecology, Plant Phenotypes and Traits	Kohler 2012
<b>DISEASE AND MENTAL HEALTH CONDITIONS</b>			
Disease Ontology	NA	Disease	Gkoutos 2015
Human Disease Ontology (DOID)	12,498	Disease	Norris 2019
Symptom Ontology (SYMP)	942	Symptom and Disease	Norris 2019
Alzheimer's Disease Ontology (ADO)	1565	Alzheimer Disease	Aminth 2018
Bilingual Ontology of Alzheimers Disease and Related Diseases (ONTOAD)	5,899	Alzheimer Disease	Aminth 2018
National Cancer Institute Thesaurus (NCIT)	167,138	Cancer	Blaum 2013
AGCT Master Ontology	NA	Cancer research and management	Brochhausen 2011
Adolescents' Depression Ontology (AdDO)	419	Depression	Jung 2016
Epidemiology Ontology (EPO)	191	Epidemiology	Norris 2019
Epilepsy and Seizure Ontology	NA	Epilepsy and seizure	Yu 2016
Mental Disease Ontology (MFOMD)	1,127	Mental Disease	Norris 2019
Haghighi-Koeda Mood Disorder Ontology	NA	Mood disorder	Yu 2016
Neurological Disease Ontology (ND)	700	Neurological Disease and phenotypes	Jensen 2013
<b>GENETICS</b>			
Gene Ontology (GO)	43850	Genetics	Blaum 2013
Micro Array Gene Expression Data (MGED) Ontology	NA	Microarray data and experiments.	Wu 2014
Ontology for Genetic Susceptibility (OGSF)	127	Genomic and Proteomic, Health	Aminth 2018
Pharmacogenetics Relationships Ontology (PHARE)	229	Pharmacogenetics	Aminth 2018
<b>NEUROSCIENCE</b>			
Biomedical Informatics Research Network Project Lexicon (BIRNlex)	3,580	Neurons and neuronal systems	Hastings 2012
Chemical Entities of Biological Interest (ChEBI)	165,081	Neurotransmitters	Hastings 2012
Consortium for Neuropsychiatric Phenomics (CNP)	NA	Neuropsychiatric Disorders	Blanch 2017

OntoNeuroLOG	1016	Neuroimaging	Blanch 2017
Neural Electromagnetic Ontology (NEMO)	1,851	Biological Process	Blanch 2017
Neuroinformatics Network (NIF)	NA	Neuroinformatics	Gkoutos 2015
Neuroimaging Data Model (NIDM)	161	Neuroimaging	Blanch 2017
Neuropsychological Testing Ontology	NA	Neuropsychological Testing	Gkoutos 2015
NeuroLex	NA	Neurons and neuronal systems	Hastings 2012
Neuroscience Information Framework Ontology (NIF)	1,24337	Neuroscience	Blanch 2017

NA = number not available

**Table 2: Characteristics of Included Studies**

Study	Ontology	Type of Study
Aminth 2018	X	Review
Aminth 2019	X	Tool
Blanch 2017	Multiple	Review
Brenas 2019	Adverse Childhood Experiences Ontology	Development
Brodaric 2010	X	Overview of applications
Carpendale 2014	X	Application
Ceusters 2015	X	Development
Cui 2015	X	Tool
Dissanayake 2020	X	Evaluation (Systematic Review)
Duque Ramos 2014	X	Evaluation
Duque Ramos 2016	X	Tool
Eisenberg 2018	Ontology of Self-Regulation	Development
Eivazzadeh 2016	X	Tool
Franco 2020	X	Evaluation of metrics
Gkoutos 2012	Neurobehavior Ontology	Development
Gkoutos 2015	X	Tools (Commentary)
Hastings 2012	X	Review
Hastings 2014	Emotion Ontology	Evaluation
Hicks 2016	Ontology of Medically Related Social Entities (OMRSE)	Development
Jensen 2013	Neurological Development Ontology	Development
Jung 2016	Adolescents' Depression Ontology	Evaluation
Kohler 2011	X	Tool
Kohler 2012	Human Phenotype Ontology	Development
Kola 2010	X	Development
Konopka 2015	X	Review
Kronk 2020	Gender, Sex, And Sexual Orientation Ontology (GSSO)	Evaluation
Larsen 2017	X	Review
Larsen 2018	Multiple	Application
Lokker 2015	X	Scoping Review
Malhotra 2014	Alzheimer's Disease Ontology	Description and evaluation



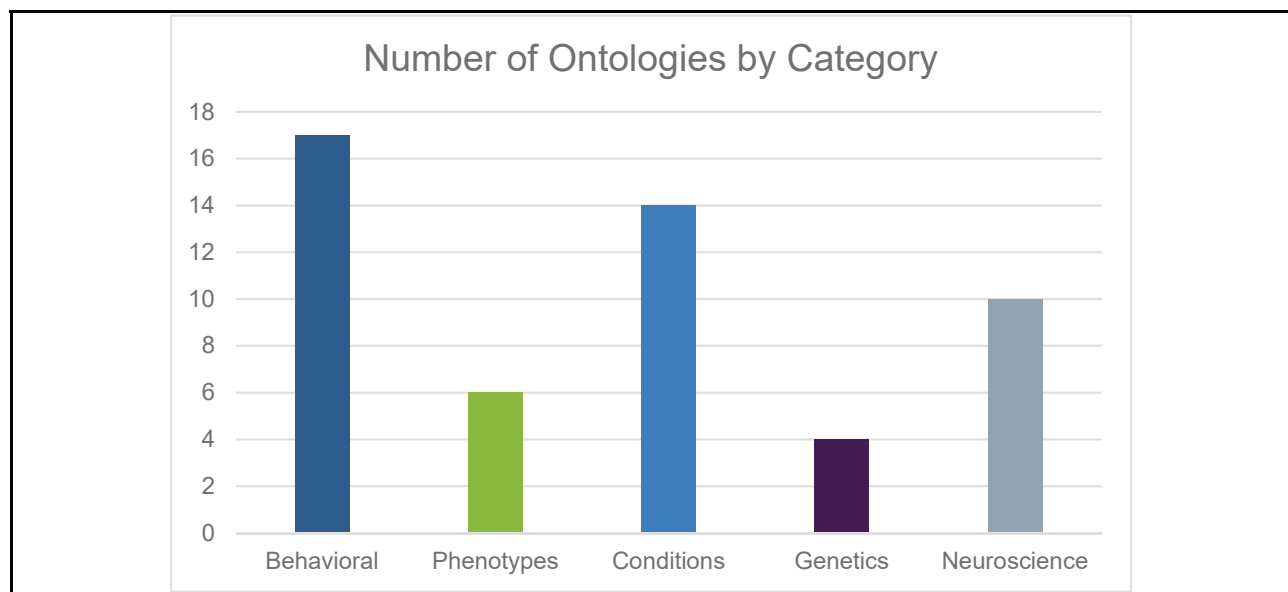
Masuya	X	Application
Michie 2017	X	Application
Mustafa 2019	X	Application
Neumuth 2010	X	Application
Norris 2019	Multiple	Scoping Review
Norris 2021	X	Development
Poldrack 2016	Multiple	Review
Prior 2020	X	Application
Quinn 2018	Personalized education for patients with diabetes or obesity	Evaluation
Reed 2015	Multiple	Development
Rousseau 2018	X	Development
Stancin 2020	Multiple	Review
Toralf 2011	X	Tool
Van Damme 2018	X	Quality assessment
Vigo 2014	X	Development
Win 2019	Persuasion Support Systems (PSS) for health behavior change	Proposal
Woznowski 2018	Ontology for Activities of Daily Living	Development
Yao 2011	X	Metrics
Zaveri 2010	Trial Bank ontology	Application

## Results of individual sources of evidence

### Reviews

There were several reviews pertaining to the field of behavioral science. **Blanch 2017**<sup>7</sup> conducted a literature search of ontologies related to human behavior. They give the results of the 17 included ontologies in table with attributes and narrative description of each, although there are no comments on evaluation. **Larsen 2017**<sup>9</sup> lays the groundwork for the **2019 Norris**<sup>11</sup> scoping review by providing an overview of ontologies and the current efforts in creation of ontologies related to behavior change interventions. The subsequent scoping review includes 15 ontologies relating to human behavior change in their scoping review. In addition to details about the properties of each ontology, the methods by which they were evaluated are also presented. The **Hastings 2012**<sup>8</sup> book chapter looks at ontologies in for the analysis of human behavior. The vocabularies and terminologies relating to human functioning are described (SNOMED CT, ICD and ICF, DSM-IV) followed by two ontologies in this field (Mental Functioning Ontology, the Mental Disease Ontology). The chapter concludes with the applications of ontology to clinical data management and translational research. **Poldrack 2016**<sup>12</sup> outlines the challenges in the field of cognitive neuroscience to delineate brain-mapping cognitions. The role of informatics in overcoming these obstacles, including the use of ontologies as a solution. At a broader level, **Lokker 2015**<sup>10</sup> provides a scoping review of intervention classification schemes and taxonomies in healthcare (including behavior science) which can be used in knowledge translation and the integration of research evidence. The review resulted in identifying 51 classification schemes including 23 taxonomies, 15 frameworks, 8 intervention lists, 3 models and 2 other formats. While ontologies were not explicitly discussed, this review provides valuable background information and references. Both **Konopka 2015**<sup>4</sup> and **Stancin 2020**<sup>14</sup> give good overviews of ontologies in the fields of biomedicine and education respectively, including criteria for development and applications. Lastly, **Zhu 2015** gives a bibliometric analysis of global ontology research progress. It outlines the trends in output, geographical placement of authors, and categories of frequently used keywords.

**Figure 2: Number of Ontologies by Category**



## Development and Authorship

There are 2 methods of developing ontologies, either reusing existing ontologies or classification systems, or by building using an original ontological model. Several studies describe the development of single ontologies: **Brenas 2019**,<sup>22</sup> the Adverse Childhood Experiences Ontology); **Gkoutos 2012**,<sup>24</sup> the Neurobehavior Ontology; **Hicks 2016**<sup>25</sup> (Ontology of Medically Related Social Entities (OMRSE); **Jensen 2013**,<sup>26</sup> the Neurological Development Ontology, **Kohler 2012**, the Human Phenotype Ontology, and **Woznowski 2018**<sup>30</sup> the Ontology for Activities of Daily Living. **Eisenberg 2018**<sup>23</sup> describes the research program to inform an ontology of self-regulation, and **Kola 2010**<sup>41</sup> discusses the need for an ontology in psychosis and proposes methodology for building one. **Win 2019**<sup>29</sup> outlines a proposal of an ontology of Persuasion Support Systems for health behavior change, and describes the literature used to develop the ontology. **Larsen 2017**<sup>9</sup> provides the background for work to build ontologies in the field of behavior change interventions, and gives an overview of why they are needed, how they are created and their potential in advancing the field of behavioral medicine. Three ontologies are described by **Larsen 2018**<sup>46</sup> (The Mental Functioning Ontology, The Emotion Ontology, and The Mental Disease Ontology. The article further discusses how all three ontologies might be used to build a broad framework which will capture the complex interrelations in the area of psychiatric disorders. **Ceusters 2017**<sup>40</sup> reports a wider initiative which aims to classify the field of oro-facial pain (including psychosocial aspects) and specifies design recommendations for a new system to bring the field up to date. **Norris's 2021**<sup>42</sup> review makes a case for including expert stakeholders in the development of ontologies. Methods of how this might be achieved are outlined and examples are given from the social and behavioral sciences. **Reed 2015**<sup>13</sup> proposes a framework for building cognition ontologies using three tools (WordNet, FrameNet and Suggested Upper Merged Ontology (SUMO)). The article also uses psychoses and emotion as examples to illustrate the objectives of science ontologies.

While not specifically in the behavioral sciences field, several studies provide valuable information for ontology development and authorship. At a very high level **Rousseau 2018**<sup>43</sup> describes a concept map which will become an Ontology of Systemology and has some principles about ontology development. Finally, **Vigo 2014**<sup>44</sup> identifies problems reported by the authors of ontologies and the strategies they use to address them. The findings are “mapped to a set of key design recommendations”, thus providing a tool to support authors and to improve the ontology development process.

## Methods of Evaluation and Assessment

There were 6 evaluations on single ontologies, namely, the Ontology of Self-Regulation (**Eisenberg 2018**),<sup>23</sup> Adolescents' Depression Ontology (**Jung 2016**),<sup>19</sup> Emotion Ontology (**Hastings 2014**),<sup>18</sup> Gender, Sex, And Sexual Orientation Ontology (**Kronk 2020**),<sup>20</sup> Alzheimer's Disease Ontology (**Malhotra 2014**),<sup>28</sup> and Personalized Education for Patients with Diabetes or Obesity Ontology (**Quinn 2015**).<sup>21</sup>

Although this review has identified several behavioral ontologies which have been evaluated, and whose methods are outlined below, much of the established criteria for evaluation is reported in the wider literature and covers multiple disciplines. It was therefore felt to be important to cover some of these principles here. The evaluation of ontologies ensures that they are fit for the purpose they were designed for, and to determine whether or not they meet quality standards. The literature describes the methods used, including tools in the form of models, methodology or software, and the metrics used in such evaluations.

Both the **Dissanayake 2020**<sup>15</sup> systematic review and the **Aminth 2018**<sup>6</sup> review provide valuable introductions and review of ontology evaluation, as well as quality assurance methods. They cite some established authors and works in this area. Aminth summarizes their work and outlines the different criteria by which ontologies may be assessed.<sup>6</sup> A number of other papers covering aspects of evaluation, standards and benchmarking but were not in the behavioral field were retrieved; while they

are not summarized here, they are referenced in the list of excluded studies.

Two papers that provide metrics for ontology evaluation are **Yao 2011**<sup>45</sup> who introduces metrics that describe the extent to which an ontology represents its knowledge domain. **Franco 2020**<sup>17</sup> is a report of an evaluation of a “set of reproducible and objective ontology structural metrics”. The study used 197 ontologies to look at the correlations between metrics, and the stability and structure of metric clusters, using 19 metrics which each measured a facet of ontology.

### **Behavioral Evaluations.**

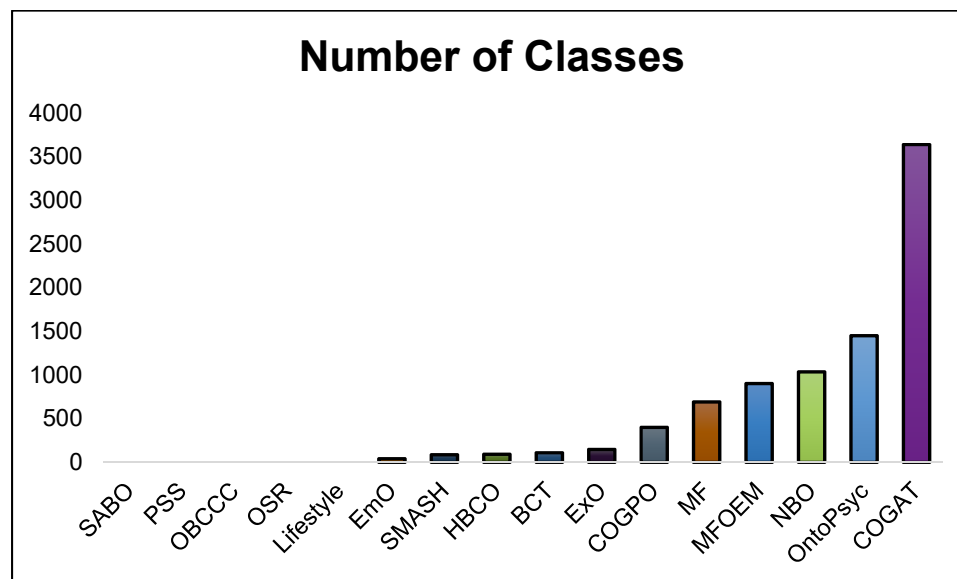
The **Norris 2019**<sup>11</sup> scoping review assessed the quality of included behavioral ontologies under the following categories:

- Ontology methods
  - Uses existing taxonomies
  - Uses existing terminologies
  - Uses existing ontologies
  - User feedback
  - Data driven
  - Unique URIs (Uniform Resource Identifiers)
  - Clear definitions
- Quality Assessment
  - Clear structure
  - Logically consistent
  - Evaluated
  - Maintained

In addition to the above items, criteria reported for the evaluation of single ontologies not included in the above list were found in several studies. **Eisenberg 2018**<sup>23</sup> used domain coverage and task orientation in testing the Ontology of Self-Regulation. **Kronk’s 2020**<sup>20</sup> evaluation of the Gender, Sex, And Sexual Orientation Ontology (GSSO) assessed computational efficiency, mappings to existing terminologies and used query-based and randomly selected term tagging. **Jung 2016**<sup>19</sup> evaluated the Adolescents’ Depression Ontology by looking at the frequency of ontology concepts in social data, and the scope using competency questions. **Malhotra 2014**<sup>28</sup> latter divided the evaluation of the Alzheimer's Disease Ontology into three area: structural (topological and logical properties), functional (“how widely and precisely ontological concepts represent the semantic space for the indicated knowledge domain”) and expert evaluation. The use of human experts to evaluate ontologies appears in many papers and appears to be a widely used despite the subjective nature of this method. **Quinn 2018**<sup>21</sup> describes the development and evaluation of an ontology for personalized education for patients with diabetes or obesity. This paper gives a good summary of evaluation methods, and then describes the 2 methods used to evaluate their ontology: domain expert evaluation, and application-based evaluation using a test patient profile.

**Hastings 2014**<sup>18</sup> reports on an experiment used to evaluation the Emotion Ontology. Figure 3 shows the range in the number of classes in the behavioral ontologies. (Note: where there is no established acronym for an ontology, one has been created for the purposes of this chart.)

**Figure 3: Behavioral Ontologies by Number of Classes**



## Tools and Software

Only one paper focusing on the behavioral sciences presented tools to aid in the design and use of ontologies, namely the **Gkoutos 2015**<sup>36</sup> commentary. As with other aspects of ontology development, evaluation, and quality assurance there are undoubtedly many studies of models, methods, tools and programs in the wider literature. Several examples identified during the course of this review are outlined here.

**Duque-Ramos 2016**<sup>34</sup> describes OQuaRE, an ontology evaluation framework based on quantitative metrics. The methodology presented by **Eivazzadeh 2016**<sup>35</sup> is UVON (Unified eValuation using Ontology; UVON) which was developed for authors to build ontologies; it enables them to “organize, unify, and aggregate the quality attributes of several health information systems into a tree-style ontology structure”. **Aminth 2019**<sup>32</sup> introduces OntoKeeper, a tool for automating quality scoring for ontology developers. This paper tests OntoKeeper and assesses it in terms of useability. **Cui 2015**<sup>33</sup> describes COHeRE (Cross-Ontology Hierarchical Relation Examination) a quality assessment tool for ontology engineers working across multiple ontologies to detect inconsistent relationships in hierarchical relations. **Kohler 2011**<sup>27</sup> illustrates how the Java software tool (GULO) can be used to improve ontology structures by detecting errors and disagreements. In a similar vein, **Van Damme 2018's**<sup>39</sup> work aims to contribute to the quality assurance of biomedical ontologies and terminologies by proposing an approach which automatically analyzes lexical content. On a broad level, GOMMA described by **Toralf 2011**<sup>38</sup> is an infrastructure used to manage and analyze life science ontologies and their expansion.

## Applications of Ontologies

In addition to the role of ontologies in representing the concepts and relationships that make up an area of knowledge, ontologies can then be used in a lot of different practical applications. The applications found from the studies in this review are wide ranging from research to clinical applications to education. **Hastings 2012**<sup>8</sup> gives an overview of applications to clinical data management and

translational research, and while **Blanch 2017**<sup>7</sup> does not discuss specific applications ontologies are described as “key instruments to stimulating a collaborative effort among researchers from a multidisciplinary approach”. The **Masuya 2012**<sup>47</sup> review paper provides an historical background of data integration in biomedical science and the role of ontologies in the field of experimental animal science. The studies in which research applications are described. **Michie 2017**<sup>48</sup> and **Larsen 2017**<sup>9</sup> both highlight the support of efficient knowledge accumulation, the organization and structuring of evidence, enhanced evidence synthesis, and the automation of meta-analysis. **Larsen 2018**<sup>46</sup> gives examples of the use of several applied ontologies, namely the Gene Ontology’s use in research such as analyzing raw data and integrating findings across domains and subject areas. They also highlight the benefits of using a multi-ontology framework for psychiatric disorders which draws upon a wealth of data to refine diagnostic categories, interrogate clinical information systems, and translate research results across a range of disciplines. **Lokker 2015**<sup>10</sup> gives examples of the use of classification schemes in compiling lists of behavior change techniques, implementation strategies, and interventions to be included in a Cochrane Collaboration systematic review.

The **Konopka 2015**<sup>4</sup> review gives examples clinical applications in 4 areas: annotation (e.g. automated annotation of radiology images), terminology mapping (e.g. mapping terminology to phenotypic clinical data to advance knowledge of genetic diseases), the use of natural language processing to code text from clinical documents, and query enhancement i.e. the use of search terms to recognize context and thus provide synonyms and additional terms to enhance the query. **Poldrack 2016**<sup>12</sup> reviews the role of ontologies in helping to “clarify, refine, and test theories of brain and cognitive functions”, while **Neumuth 2010**<sup>49</sup> outlines the advantages bestowed by the method of ontology-supported recording for complex behaviors in the modelling of surgical processes. **Zaveri 2010**<sup>31</sup> describes the use of the Trial Bank ontology to code RCTs in neurosurgery.

In the area of learning and education, **Stancin 2020**<sup>14</sup> reviews the use of ontologies to develop educational software to enrich the description of e-learning content, to personalize and recommend additional content, to design curricula, and to assess the learning process. **Brodaric 2010**<sup>50</sup> summarizes uses of ontologies in online science activities and **Mustafa 2019**,<sup>51</sup> the use of ontologies to generate content for preschool cognitive skills learning. **Prior 2020**<sup>52</sup> proposes the use ontologies for setting age-appropriate passwords. **Carpendale 2014**<sup>53</sup> examines the impact of ontology use in biological data visualization.

Although **Zhu 2015**<sup>54</sup> does not give specific examples the reporting of author keywords used in the bibliometric analysis gives some insight into how ontologies are being used. For example, bioinformatics, information extraction, machine learning, metadata, description logics, and knowledge management.

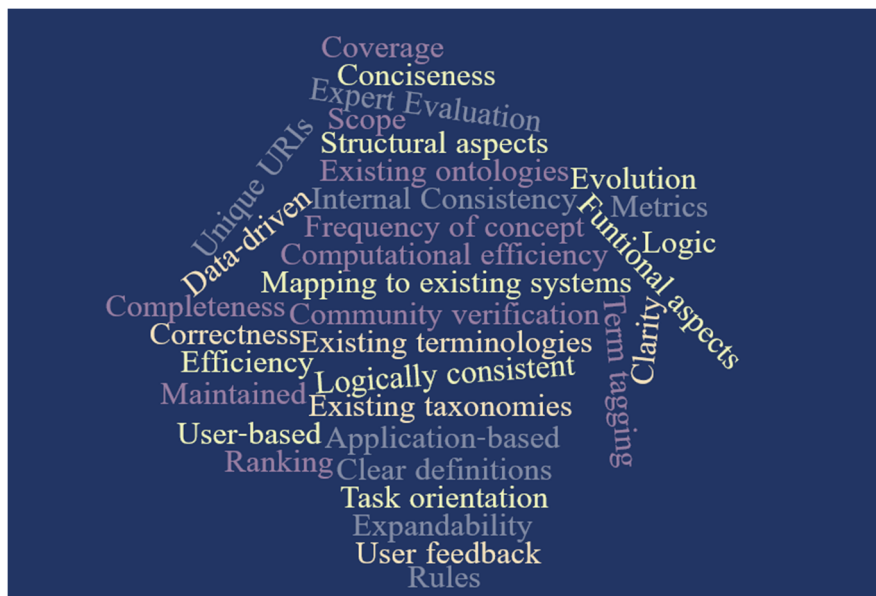
## Recommendations for Best Practice

Several papers included sections with explicit recommendations. **Ceusters 2015**<sup>40</sup> makes recommendations for developing ontologies, and **Duque Ramos 2014**<sup>16</sup> evaluates the Good Ontology Design Guideline, although they don’t make specific recommendations of their own. **Norris 2019**<sup>11</sup> identifies some best practices for ontology development from their review of methods and evaluation of the studies included in their scoping review. In a follow up paper, **Norris 2021**<sup>42</sup> advocates for the engagement of domain experts in ontology development. Finally, **Vigo 2014**<sup>44</sup> give insights into design and give recommendations. While there was not an abundance of best practice recommendations, most of the studies provided lessons learned of some description which can be used to inform the overall picture of ontology design and evaluation.

Figure 4: Wordcloud generated from behavioral ontologies identified.



Figure 5: Wordcloud generated from words relating to the evaluation of ontologies



## **Conclusions**

### **Summary of Evidence**

There were 50 studies included in this scoping review, and 49 ontologies identified as belonging to the behavioral sciences. The large number of records retrieved through the searching is evidence of the extent of literature on ontology, including ontology as a philosophical concept. It also shows the many different disciplines which have studied and applied ontologies, giving a wealth of studies on all aspects of ontologies from overviews and definitions to very specific methods of development and evaluation. While this scoping review has revealed a relatively small number of published reviews and evaluations specific to the behavioral sciences, it is clear that the broader biomedical literature is much larger and contains a wealth of information for developers. It is evident that the top level and large biomedical ontologies are often used to test out methods of development, evaluation, updating and expansion. It is also clear that there are a large number of ontologies in existence from the very large and very broad, to the very small and specific, depending upon the objectives and intended use. The number of behavioral ontologies included here could be added to depending on how behavioral is defined, and repositories of ontologies would likely identify more. It should also be noted that many of the large biomedical ontologies will invariably include concepts on animal and human behavior, but these were considered outside of the scope of this review. Evaluations were shown to have a variety of methods and tools involving software programs, modeling, human expert opinion and knowledge, guidelines and benchmarks. However, despite several systematic and scoping reviews being found, there was no study which conducted a meta-analysis of evaluation methods. The broad array of applications for which ontologies are being used was also revealed. Not only do they contribute to the understanding of a body of knowledge, they enable researchers to work across disciplines and standardize definitions and relationships of concepts. On a practical level they are being used in research in fields such as information science, computer science, medical education, healthcare research, and in clinical settings. Broad principles can be taken from the overall body of work to guide development as well as the ongoing evolution and improvement of ontologies.

### **Strengths and Limitations**

The main strength of this scoping review is the large number of references screened to obtain the studies which were included. The results were from 2 sources, a very broad search of literature relating to ontologies, and a second, comprehensive but structured and targeted search designed to hone in on published reviews and evaluations. The combined set enabled the identification of studies to fit the eligibility criteria, but also a view of the literature as a whole, from which general principles and methods can be drawn given the relative paucity of specific information to the behavioral sciences. The information has been organized narratively and visually to enable readers navigate the information, see overall themes, and access the main points from each of the sections relating to the objectives of the review.

The first limitation is that although several databases were searched, and additional searches conducted, the search terms used may have resulted in potentially relevant studies being missed. Time constraints did not allow for the potentially very large set of results to be screened had a more comprehensive strategy been used. Similarly, although some additional searching was conducted around the most relevant studies found, the planned extensive follow up of references was not done, and thus studies may have missed that were in the scope of this review.



Secondly the review was conducted by one person, and decisions made about what should be included in terms of behavioral science, and which studies should be included or excluded may have been different with a second reviewer. Thirdly, no formal appraisal of studies was conducted and while the essential elements of the included studies have been summarized, they have not been examined in depth.

## References

1. Gruber TR. Toward principles for the design of ontologies used for knowledge sharing? *International Journal of Human-Computer Studies*. 1995;43(5):907-928.
2. Kless D, Milton S, Kazmierczak E, Lindenthal J. Thesaurus and ontology structure: Formal and pragmatic differences and similarities. *Journal of the Association for Information Science and Technology*. 2015;66(7):1348-1366.
3. Hoehndorf R, Schofield PN, Gkoutos GV. The role of ontologies in biological and biomedical research: a functional perspective. *Briefings in bioinformatics*. 2015;16(6):1069-1080.
4. Konopka BM. Biomedical ontologies - A review. *Biocybernetics and Biomedical Engineering*. 2015;35(2):75-86.
5. Tricco AC, Lillie E, Zarin W, et al. PRISMA Extension for Scoping Reviews (PRISMA-ScR): Checklist and Explanation. *Annals of Internal Medicine*. 2018;169(7):467-473.
6. Amith M, He Z, Bian J, Lossio-Ventura JA, Tao C. Assessing the practice of biomedical ontology evaluation: Gaps and opportunities. *Journal of Biomedical Informatics*. 2018;80:1-13.
7. Blanch A, García R, Planes J, et al. Ontologies about human behavior: A review of knowledge modeling systems. *European Psychologist*. 2017;22(3):180-197.
8. Hastings J, Schulz S. Ontologies for Human Behavior Analysis and Their Application to Clinical Data. In: Chesler EJ, Haendel MA, eds. *Bioinformatics of Behavior: Part 1*. Vol 103.2012:89-107.
9. Larsen KR, Michie S, Hekler EB, et al. Behavior change interventions: the potential of ontologies for advancing science and practice. *Journal of Behavioral Medicine*. 2017;40(1):6-22.
10. Lokker C, McKibbin KA, Colquhoun H, Hempel S. A scoping review of classification schemes of interventions to promote and integrate evidence into practice in healthcare. *Implementation Science*. 2015;10:27.
11. Norris E, Finnerty AN, Hastings J, Stokes G, Michie S. A scoping review of ontologies related to human behaviour change. *Nature Human Behaviour*. 2019;3(2):164-172.
12. Poldrack RA, Yarkoni T. From Brain Maps to Cognitive Ontologies: Informatics and the Search for Mental Structure. *Annual Review of Psychology*. 2016;67:587-612.
13. Reed SK, Pease A. A framework for constructing cognition ontologies using WordNet, FrameNet, and SUMO. *Cognitive Systems Research*. 2015;33:122-144.
14. Stancin K, Posic P, Jaksic D. Ontologies in education – state of the art. *Education and Information Technologies*. 2020;25(6):5301-5320.
15. Dissanayake PI, Colicchio TK, Cimino JJ. Using clinical reasoning ontologies to make smarter clinical decision support systems: a systematic review and data synthesis. *Journal of the American Medical Informatics Association*. 2020;27(1):159-174.
16. Duque-Ramos A, Boeker M, Jansen L, Schulz S, Iniesta M, Fernandez-Breis JT. Evaluating the Good Ontology Design Guideline (GoodOD) with the ontology quality requirements and evaluation method and metrics (OQuARE). *PLoS ONE [Electronic Resource]*. 2014;9(8):e104463.

17. Franco M, Vivo JM, Quesada-Martinez M, Duque-Ramos A, Fernandez-Breis JT. Evaluation of ontology structural metrics based on public repository data. *Briefings in Bioinformatics*. 2020;21(2):473-485.
18. Hastings J, Brass A, Caine C, Jay C, Stevens R. Evaluating the Emotion Ontology through use in the self-reporting of emotional responses at an academic conference. *Journal of Biomedical Semantics*. 2014;5:38.
19. Jung H, Park HA, Song TM. Development and Evaluation of an Adolescents' Depression Ontology for Analyzing Social Data. *Studies in Health Technology and Informatics*. 2016;225:442-446.
20. Kronk CA, Dexheimer JW. Development of the Gender, Sex, and Sexual Orientation ontology: Evaluation and workflow. *Journal of the American Medical Informatics Association*. 2020;27(7):1110-1115.
21. Quinn S, Bond R, Nugent C. A two-staged approach to developing and evaluating an ontology for delivering personalized education to diabetic patients. *Informatics for Health & Social Care*. 2018;43(3):264-279.
22. Brenas JH, Shin EK, Shaban-Nejad A. Adverse Childhood Experiences Ontology for Mental Health Surveillance, Research, and Evaluation: Advanced Knowledge Representation and Semantic Web Techniques. *JMIR Mental Health*. 2019;6(5).
23. Eisenberg IW, Bissett PG, Canning JR, et al. Applying novel technologies and methods to inform the ontology of self-regulation. *Behaviour Research and Therapy*. 2018;101:46-57.
24. Gkoutos GV, Schofield PN, Hoehndorf R. The Neurobehavior Ontology: An Ontology for Annotation and Integration of Behavior and Behavioral Phenotypes. In: Chesler EJ, Haendel MA, eds. *Bioinformatics of Behavior: Part 1*. Vol 103.2012:69-87.
25. Hicks A, Hanna J, Welch D, Brochhausen M, Hogan WR. The ontology of medically related social entities: recent developments. *Journal of Biomedical Semantics*. 2016;7:47.
26. Jensen M, Cox AP, Chaudhry N, et al. The neurological disease ontology. *Journal of Biomedical Semantics*. 2013;4(1):42.
27. Kohler S, Doelken SC, Rath A, Ayme S, Robinson PN. Ontological phenotype standards for neurogenetics. *Human Mutation*. 2012;33(9):1333-1339.
28. Malhotra A, Younesi E, Gundel M, Muller B, Heneka MT, Hofmann-Apitius M. ADO: a disease ontology representing the domain knowledge specific to Alzheimer's disease. *Alzheimer's & dementia : the journal of the Alzheimer's Association*. 2014;10(2):238-246.
29. Win KT, Ramaprasad A, Syn T. Ontological Review of Persuasion Support Systems (PSS) for Health Behavior Change through Physical Activity. *Journal of Medical Systems*. 2019;43(3):49.
30. Woznowski PR, Tonkin EL, Flach PA. Activities of Daily Living Ontology for Ubiquitous Systems: Development and Evaluation. *Sensors*. 2018;18(7):20.
31. Zaveri A, Cofiel L, Shah J, et al. Achieving high research reporting quality through the use of computational ontologies. *Neuroinformatics*. 2010;8(4):261-271.
32. Amith M, Manion F, Liang C, et al. Architecture and usability of OntoKeeper, an ontology evaluation tool. *BMC Medical Informatics and Decision Making*. 2019;19(Suppl 4):152.
33. Cui L. COHeRE: Cross-Ontology Hierarchical Relation Examination for Ontology Quality Assurance. *AMIA Annual Symposium Proceedings AMIA Symposium*. 2015;2015:456-465.
34. Duque-Ramos A, Quesada-Martinez M, Iñiesta-Moreno M, Fernandez-Breis JT, Stevens R. Supporting the analysis of ontology evolution processes through the combination of static and dynamic scaling functions in OQuaRE. *Journal of Biomedical Semantics*. 2016;7(1):63.
35. Eivazzadeh S, Anderberg P, Larsson TC, Fricker SA, Berglund J. Evaluating Health Information Systems Using Ontologies. *JMIR Medical Informatics*. 2016;4(2):e20.
36. Gkoutos GV, Hoehndorf R, Tsaprouni L, Schofield PN. Best behaviour? Ontologies and the formal description of animal behaviour. *Mammalian Genome*. 2015;26(9-10):540-547.

37. Kohler S, Bauer S, Mungall CJ, et al. Improving ontologies by automatic reasoning and evaluation of logical definitions. *BMC Bioinformatics*. 2011;12:418.
38. Toralf K, Gross A, Hartung M, Rahm E. GOMMA: a component-based infrastructure for managing and analyzing life science ontologies and their evolution. *Journal of Biomedical Semantics*. 2011;2:1-24.
39. van Damme P, Cornet R, Fernandez-Breis JT, Quesada-Martinez M. From lexical regularities to axiomatic patterns for the quality assurance of biomedical terminologies and ontologies. *Journal of Biomedical Informatics*. 2018;84:59-74.
40. Ceusters W, Michelotti A, Raphael KG, Durham J, Ohrbach R. Perspectives on next steps in classification of oro-facial pain - part 1: role of ontology. *Journal of Oral Rehabilitation*. 2015;42(12):926-941.
41. Kola J, Harris J, Lawrie S, Rector A, Goble C, Martone M. Towards an ontology for psychosis. *Cognitive Systems Research*. 2010;11(1):42-52.
42. Norris E, Hastings J, Marques MM, Mutlu ANF, Zink S, Michie S. Why and how to engage expert stakeholders in ontology development: insights from social and behavioural sciences. *Journal of Biomedical Semantics*. 2021;12(1).
43. Rousseau D, Billingham J, Calvo-Amodio J. Systemic Semantics: A Systems Approach to Building Ontologies and Concept Maps. *Systems*. 2018;6(3).
44. Vigo M, Bail S, Jay C, Stevens R. Overcoming the pitfalls of ontology authoring: Strategies and implications for tool design. *International Journal of Human-Computer Studies*. 2014;72(12):835-845.
45. Yao L, Divoli A, Mayzus I, Evans JA, Rzhetsky A. Benchmarking ontologies: Bigger or better? *PLoS Computational Biology*. 2011;7(1):e1001055.
46. Larsen RR, Hastings J. From affective science to psychiatric disorder: Ontology as a semantic bridge. *Frontiers in Psychiatry*. 2018;9:13.
47. Masuya H. Roles and applications of biomedical ontologies in experimental animal science. *Experimental Animals*. 2012;61(4):365-373.
48. Michie S, Johnston M. Optimising the value of the evidence generated in implementation science: the use of ontologies to address the challenges. *Implementation Science*. 2017;12.
49. Neumuth T, Kaschek B, Neumuth D, et al. An observation support system with an adaptive ontology-driven user interface for the modeling of complex behaviors during surgical interventions. *Behavior Research Methods*. 2010;42(4):1049-1058.
50. Brodaric B, Gahegan M. Ontology use for semantic e-Science. *Semantic Web*. 2010;1(1-2):149-153.
51. Mustafa G, Abbas MA, Hafeez Y, Khan S, Hwang G-J. Effectiveness of ontology-based learning content generation for preschool cognitive skills learning. *Interactive Learning Environments*. 2018.
52. Prior S, Renaud K. Age-appropriate password “best practice” ontologies for early educators and parents. *International Journal of Child-Computer Interaction*. 2020;23-24:12.
53. Carpendale S, Chen M, Evanko D, et al. Ontologies in biological data visualization. *IEEE computer graphics and applications*. 2014;34(2):8-15.
54. Zhu Q, Kong X, Song H, Li J, He Z. Global ontology research progress: a bibliometric analysis. *Aslib Journal of Information Management*. 2015;67(1):27-54.

## List of Excluded Studies

1. Ali S, Shah K. POEM: Practical ontology engineering model for semantic web ontologies. *Cogent Engineering*. 2016;3(1).
2. Alloni A, Quaglini S, Panzarasa S, Sinforiani E, Bernini S. Evaluation of an ontology-based system for computerized cognitive rehabilitation. *International Journal of Medical Informatics*. 2018;115:64-72.
3. Alobaidi M, Malik KM, Hussain M. Automated ontology generation framework powered by linked biomedical ontologies for disease-drug domain. *Computer Methods and Programs in Biomedicine*. 2018;165:117-128.
4. Amith M, Tao C. Modulated evaluation metrics for drug-based ontologies. *Journal of biomedical semantics*. 2017;8(1):17.
5. Andarmoyo S, Syandri G, Nguyen PT, Lydia EL, Shankar K. Need of ontology based systems in healthcare system. *Opcion*. 2019;35(Special Issue 20):1214-1228.
6. Andrade AQ, Blonde W, Hastings J, Schulz S. Process attributes in bio-ontologies. *BMC Bioinformatics*. 2012;13:217.
7. Andronache AS, Simoncello A, Della Mea V, Daffara C, Francescutti C. Semantic Aspects of the International Classification of Functioning, Disability and Health Towards Sharing Knowledge and Unifying Information. *American Journal of Physical Medicine and Rehabilitation*. 2012;91(2):S124-S128.
8. Arbabi A, Adams DR, Fidler S, Brudno M. Identifying Clinical Terms in Medical Text Using Ontology-Guided Machine Learning. *JMIR Medical Informatics*. 2019;7(2):e12596.
9. Arguello-Casteleiro M, Stevens R, Demetriou G, et al. Exploring semantic deep learning for building reliable and reusable one health knowledge from PubMed systematic reviews and veterinary clinical notes. *Journal of Biomedical Semantics*. 2019;10(Supplement 1):22.
10. Baars EW, Hamre HJ. Whole Medical Systems versus the System of Conventional Biomedicine: A Critical, Narrative Review of Similarities, Differences, and Factors That Promote the Integration Process. *Evidence - Based Complementary and Alternative Medicine*. 2017;2017.
11. Bada M, Baumgartner WA, Hunter LE, Vasilevsky N, Haendel M. Gold-standard ontology-based anatomical annotation in the CRAFT Corpus. *Database*. 2017;2017.
12. Badre D. Defining an ontology of cognitive control requires attention to component interactions. *Topics in Cognitive Science*. 2011;3(2):217-221.
13. Bandrowski A, Brinkman R, Brochhausen M, et al. The Ontology for Biomedical Investigations. *PLoS One*. 2016;11(4).
14. Baronchelli A, Ferrer-i-Cancho R, Pastor-Satorras R, Chater N, Christiansen MH. Networks in cognitive science. *Trends in Cognitive Sciences*. 2013;17(7):348-360.
15. Bastian FB, Chibucos MC, Gaudet P, et al. The Confidence Information Ontology: a step towards a standard for asserting confidence in annotations. *Database*. 2015;2015.
16. Batet M, Sanchez D, Valls A. An ontology-based measure to compute semantic similarity in biomedicine. *Journal of Biomedical Informatics*. 2011;44(1):118-125.
17. Benmimoune L, Hajjam A, Ghodous P, Andres E, Talha S, Hajjam M. Ontology-based Medical Decision Support System to Enhance Chronic Patients' Lifestyle within E-care Telemonitoring Platform. *Studies in Health Technology and Informatics*. 2015;213:279-282.
18. Berardini TZ, Li D, Muller R, et al. Assessment of community-submitted ontology annotations from a novel database-journal partnership. *Database*. 2012;2012(0).
19. Berges I, Bermudez J, Illarramendi A. SSDOnt: An Ontology for Representing Single-Subject Design Studies. *Methods of Information in Medicine*. 2018;57(1):55-61.

20. Bickmore TW, Schulman D, Sidner CL. A reusable framework for health counseling dialogue systems based on a behavioral medicine ontology. *Journal of Biomedical Informatics*. 2011;44(2):183-197.
21. Birnbaum S. Moving Beyond the Behavior-Change Framework for Smoking Cessation: Lessons for a Critical Ontology From the Case of Inpatient Psychiatric Units. *Journal of the American Psychiatric Nurses Association*. 2019;25(4):289-297.
22. Blaum WE, Jarczweski A, Balzer F, Stotzner P, Ahlers O. Towards Web 3.0: taxonomies and ontologies for medical education -- a systematic review. *GMS Zeitschrift Fur Medizinische Ausbildung*. 2013;30(1):Doc13.
23. Boeker M, Schober D, Seddig-Raufie D, et al. Effects of Guideline-Based Training on the Quality of Formal Ontologies: A Randomized Controlled Trial. *PLoS ONE*. 2013;8(5):e61425.
24. Boeker M, Tudose I, Hastings J, Schober D, Schulz S. Unintended consequences of existential quantifications in biomedical ontologies. *BMC Bioinformatics*. 2011;12:456.
25. Borlowsky TB, Lele O, Payne PRO. Research-IQ: development and evaluation of an ontology-anchored integrative query tool. *Journal of Biomedical Informatics*. 2011;44 Suppl 1:S56-S62.
26. Bouarab-Dahmani F, Comparot C, Si-Mohammed M, Charrel P-J. Ontology based teaching domain knowledge management for E-learning by doing systems. *Electronic Journal of Knowledge Management*. 2015;13(2):155-170.
27. Boyer P, Barrett HC. Intuitive ontologies and domain specificity. In: Buss DM, ed. *The handbook of evolutionary psychology: Foundations., Vol. 1, 2nd ed*. Hoboken, NJ: John Wiley & Sons, Inc.; 2016:161-179.
28. Bright TJ, Yoko Furuya E, Kuperman GJ, Cimino JJ, Bakken S. Development and evaluation of an ontology for guiding appropriate antibiotic prescribing. *Journal of Biomedical Informatics*. 2012;45(1):120-128.
29. Brochhausen M, Bona J, Blobel B. The Role of Axiomatically-Rich Ontologies in Transforming Medical Data to Knowledge. *Studies in Health Technology and Informatics*. 2018;249:38-49.
30. Brochhausen M, Spear AD, Cocos C, et al. The ACGT Master Ontology and its applications--towards an ontology-driven cancer research and management system. *Journal of Biomedical Informatics*. 2011;44(1):8-25.
31. Brown W, III. *Ontology-based semantic harmonization of HIV-associated common data elements for integration of diverse HIV research datasets*, ProQuest Information & Learning; 2017.
32. Burns GA, Turner JA. Modeling functional Magnetic Resonance Imaging (fMRI) experimental variables in the Ontology of Experimental Variables and Values (OoEVV). *Neuroimage*. 2013;82:662-670.
33. Calvo H, Figueroa-Nazuno J, Mandujano Á. Natural ontologies with elastic matching for elicited knowledge comparison. *Journal of Intelligent and Fuzzy Systems*. 2020;39(2):2291-2303.
34. Cardoso SD, Pruski C, Da Silveira M. Supporting biomedical ontology evolution by identifying outdated concepts and the required type of change. *Journal of Biomedical Informatics*. 2018;87:1-11.
35. Ceusters W, Jensen M, Diehl AD. Ontological Realism for the Research Domain Criteria for Mental Disorders. *Studies in Health Technology and Informatics*. 2017;235:431-435.
36. Ceusters W, Smith B. A unified framework for biomedical terminologies and ontologies. *Studies in Health Technology and Informatics*. 2010;160(PART 1):1050-1054.
37. Chalk SJ. SciData: a data model and ontology for semantic representation of scientific data. *Journal of Cheminformatics*. 2016;8(1):1-24.
38. Chen HW, Du J, Song H-Y, Liu X, Jiang G, Tao C. Representation of Time-Relevant Common Data Elements in the Cancer Data Standards Repository: Statistical Evaluation of an Ontological Approach. *JMIR Medical Informatics*. 2018;6(1):e7.

39. Chen J, Scholz U, Zhou R, Lange M. LAILAPS-QSM: A RESTful API and JAVA library for semantic query suggestions. *PLoS Computational Biology*. 2018;14(3):e1006058.
40. Chen Y, Zhang Z, Huang J, Xie Y. Toward a scientific ontology based concept of function. *Artificial Intelligence for Engineering Design, Analysis and Manufacturing: AIEDAM*. 2013;27(3):241-248.
41. Chua WWK, Kim J-J. BOAT: automatic alignment of biomedical ontologies using term informativeness and candidate selection. *Journal of Biomedical Informatics*. 2012;45(2):337-349.
42. Cubric M, Tosic M. Design and evaluation of an ontology-based tool for generating multiple-choice questions. *Interactive Technology and Smart Education*. 2020;17(2):109-131.
43. D'Mello S, Franklin S. A cognitive model's view of animal cognition. *Current Zoology*. 2011;57(4):499-513.
44. Dahdul W, Manda P, Cui H, et al. Annotation of phenotypes using ontologies: a gold standard for the training and evaluation of natural language processing systems. *Database (Oxford)*. 2018;01:01.
45. del Mar Sánchez-Vera M, Fernández-Breis JT, Castellanos-Nieves D, Frutos-Morales F, Prendes-Espinosa MP. Semantic Web technologies for generating feedback in online assessment environments. *Knowledge-Based Systems*. 2012;33:152-165.
46. Dos Reis JC, Dinh D, Pruski C, Da Silveira M, Reynaud-Delaitre C. The influence of similarity between concepts in evolving biomedical ontologies for mapping adaptation. *Studies in Health Technology and Informatics*. 2014;205:1003-1007.
47. Dragisic Z, Ivanova V, Li H, Lambrix P. Experiences from the anatomy track in the ontology alignment evaluation initiative. *Journal of Biomedical Semantics*. 2017;8(1):56.
48. Duclos C, Soualmia LF, Krivine S, Jamet A, Lillo-Louet A. Modeling, building and evaluating an ontology for the automatic characterization of adverse drug effects during pharmacovigilance. *Studies in Health Technology and Informatics*. 2010;160(Pt 2):1005-1009.
49. Eilbeck KL, Lipstein J, McGarvey S, Staes CJ. Evaluation of need for ontologies to manage domain content for the Reportable Conditions Knowledge Management System. *AMIA Annual Symposium Proceedings*. 2014;2014:496-505.
50. El Kadiri S, Kiritsis D. Ontologies in the context of product lifecycle management: State of the art literature review. *International Journal of Production Research*. 2015;53(18):5657-5668.
51. Elhanan G, Ochs C, Mejino JLV, Jr., Liu H, Mungall CJ, Perl Y. From SNOMED CT to Uberon: Transferability of evaluation methodology between similarly structured ontologies. *Artificial Intelligence in Medicine*. 2017;79:9-14.
52. Elkin PL, Beuscart-Zephir MC, Pelayo S, Patel V, Nohr C. The usability-error ontology. *Studies in Health Technology and Informatics*. 2013;194:91-96.
53. Engeman J, Mabee PM, Dahdul WM, et al. Evolutionary characters, phenotypes and ontologies: Curating data from the systematic biology literature. *PLoS ONE*. 2010;5(5):e10708.
54. Faria D, Pesquita C, Mott I, Couto FM, Martins C, Cruz IF. Tackling the challenges of matching biomedical ontologies. *Journal of Biomedical Semantics*. 2018;9(1):4.
55. Faria D, Pesquita C, Santos E, Couto FM, Cruz IF. Automatic background knowledge selection for matching biomedical ontologies. *PLoS ONE*. 2014;9(11):e111226.
56. Freitas F, Santana da Silva F, Jansen L, Schulz S. Ontological interpretation of biomedical database content. *Journal of Biomedical Semantics*. 2017;8(1):24.
57. Funk C, Baumgartner Jr W, Garcia B, et al. Large-scale biomedical concept recognition: an evaluation of current automatic annotators and their parameters. *BMC Bioinformatics*. 2014;15:59.
58. Gainotti G, Spinelli P, Scaricamazza E, Marra C. The evaluation of sources of knowledge underlying different conceptual categories. *Frontiers in Human Neuroscience*. 2013(FEB).
59. Gan M, Dou X, Jiang R. From ontology to semantic similarity: Calculation of ontology-based

- semantic similarity. *The Scientific World Journal*. 2013;2013:793091.
60. Gao W. *An approach to formalizing ontology driven semantic integration: Concepts, dimensions and framework*, ProQuest Information & Learning; 2013.
  61. García del Valle EP, Lagunes García G, Prieto Santamaría L, Zanin M, Menasalvas Ruiz E, Rodríguez-González A. Disease networks and their contribution to disease understanding: A review of their evolution, techniques and data sources. *Journal of Biomedical Informatics*. 2019;94.
  62. Garla VN, Brandt C. Semantic similarity in the biomedical domain: an evaluation across knowledge sources. *BMC Bioinformatics*. 2012;13:261.
  63. George G, Lal AM. Review of ontology-based recommender systems in e-learning. *Computers & Education*. 2019;142.
  64. Grando A, Schwab R. Building and evaluating an ontology-based tool for reasoning about consent permission. *AMIA Annual Symposium Proceedings*. 2013;2013:514-523.
  65. Guardia GDA, Vencio RZN, de Farias CRG. A UML profile for the OBO relation ontology. *BMC Genomics*. 2012;13 Suppl 5:S3.
  66. Gulzar Z, Anny Leema A. Ontology based classification of computer science domain to support personalization. *Journal of Advanced Research in Dynamical and Control Systems*. 2017;9(Special Issue 6):493-511.
  67. Gundel M, Younesi E, Malhotra A, et al. HuPSON: the human physiology simulation ontology. *Journal of Biomedical Semantics*. 2013;4(1):35.
  68. Guo-Qiang Z, Yan H, Licong C. Can SNOMED CT Changes Be Used as a Surrogate Standard for Evaluating the Performance of Its Auditing Methods? *AMIA Annual Symposium proceedings AMIA Symposium*. 2017;2017:1903-1912.
  69. Gutierrez F. Semantic Technologies and Bio-Ontologies. In: Huang J, Borchert GM, Dou D, et al., eds. *Bioinformatics in Microrna Research*. Vol 1617. 2017:83-91.
  70. Hackett PMW. Facet theory and the mapping sentence as hermeneutically consistent structured meta-ontology and structured meta-mereology. *Frontiers in Psychology*. 2016;7:6.
  71. Harrow I, Jimenez-Ruiz E, Splendiani A, et al. Matching disease and phenotype ontologies in the ontology alignment evaluation initiative. *Journal of Biomedical Semantics*. 2017;8(1):55.
  72. Hastings J, Jeliaskova N, Owen G, et al. eNanoMapper: harnessing ontologies to enable data integration for nanomaterial risk assessment. *Journal of Biomedical Semantics*. 2015;6:10.
  73. Hoehndorf R, Dumontier M, Gennari JH, et al. Integrating systems biology models and biomedical ontologies. *BMC Systems Biology*. 2011;5:124.
  74. Hoehndorf R, Dumontier M, Gkoutos GV. Evaluation of research in biomedical ontologies. *Briefings in Bioinformatics*. 2013;14(6):696-712.
  75. Hoehndorf R, Haendel M, Stevens R, Rebholz-Schuhmann D. Thematic series on biomedical ontologies in JBMS: challenges and new directions. *Journal of Biomedical Semantics*. 2014;5:15.
  76. Hoehndorf R, Schofield PN, Gkoutos GV. The role of ontologies in biological and biomedical research: a functional perspective. *Briefings in Bioinformatics*. 2015;16(6):1069-1080.
  77. Hofer P, Neururer S, Haufler H, Insam T, Zeilner A, Gobel G. Semi-automated evaluation of biomedical ontologies for the biobanking domain based on competency questions. *Studies in Health Technology and Informatics*. 2015;212:65-72.
  78. Hulsege B, Smits MA, te Pas MFW, Woelders H. Contributions to an animal trait ontology. *Journal of Animal Science*. 2012;90(6):2061-2066.
  79. Ivanova T. E-Learning resource reuse, based on bilingual ontology annotation and ontology mapping. *International Journal of Advanced Computer Research*. 2019;9(45):351-364.
  80. Jackson RC, Overton JA, Balhoff JP, Douglass E, Harris NL, Mungall CJ. ROBOT: A Tool for Automating Ontology Workflows. *BMC Bioinformatics*. 2019;20(1):407.

81. Jaziri W, Chaabane S, Sassi N. Geont: Geo-ontologies integration tool. *International Journal of Metadata, Semantics and Ontologies*. 2017;12(2-3):155-166.
82. Kamdar MR, Tudorache T, Musen MA. A Systematic Analysis of Term Reuse and Term Overlap across Biomedical Ontologies. *Semantic Web*. 2017;8(6):853-871.
83. Katayama T, Wilkinson MD, Aoki-Kinoshita KF, et al. BioHackathon series in 2011 and 2012: penetration of ontology and linked data in life science domains. *Journal of Biomedical Semantics*. 2014;5:1-13.
84. Keen E. Grounding the Process of Ontology Concept Formulation in a Multi-perspective Social Context: Developing Ontology to Support Enhanced Collaboration in Community Tourism Event Management. *International Journal of Technology, Knowledge and Society*. 2012;8(3):55-68.
85. Kennedy MT, Fiss PC. An ontological turn in categories research: From standards of legitimacy to evidence of actuality. *Journal of Management Studies*. 2013;50(6):1138-1154.
86. Khan SA, Qadir MA, Abbas MA, Afzal MT. OWL2 benchmarking for the evaluation of knowledge based systems. *PLoS ONE [Electronic Resource]*. 2017;12(6):e0179578.
87. Kiourtis A, Mavrogiorgou A, Kyriazis D. A Semantic Similarity Evaluation for Healthcare Ontologies Matching to HL7 FHIR Resources. *Studies in Health Technology and Informatics*. 2020;270:13-17.
88. Ko YM, Song MS, Lee SJ. Construction of the structural definition-based terminology ontology system and semantic search evaluation. *Library Hi Tech*. 2016;34(4):705-732.
89. Kocbek S, Kim J-D. Exploring biomedical ontology mappings with graph theory methods. *PeerJournal*. 2017;5:e2990.
90. Kohler S, Carmody L, Vasilevsky N, et al. Expansion of the Human Phenotype Ontology (HPO) knowledge base and resources. *Nucleic Acids Research*. 2019;47(D1):D1018-D1027.
91. Lamprecht D, Strohmaier M, Helic D, et al. Using ontologies to model human navigation behavior in information networks: A study based on Wikipedia. *Semantic Web*. 2015;6(4):403-422.
92. Laurini R. Geographic Ontologies, Gazetteers and Multilingualism. *Future Internet*. 2015;7(1):1-23.
93. Le Franc Y, Davison AP, Gleeson P, et al. Computational Neuroscience Ontology: A new tool to provide semantic meaning to your models. *BMC Neuroscience*. 2012;13(SUPPL. 1).
94. Legaz-Garcia MdC, Martinez-Costa C, Minarro-Gimenez JA, Fernandez-Breis JT, Schulz S, Menarguez-Tortosa M. Ontology patterns-based transformation of clinical information. *Studies in Health Technology and Informatics*. 2014;205:1018-1022.
95. Lenartowicz A, Kalar DJ, Congdon E, Poldrack RA. Towards an ontology of cognitive control. *Topics in Cognitive Science*. 2010;2(4):678-692.
96. Liu H, Xu F. Semantic Data Mining, An Ontology-Based Approach. *Terminology*. 2018;24(2):289-294.
97. Liu K, Mitchell KJ, Chapman WW, et al. Formative evaluation of ontology learning methods for entity discovery by using existing ontologies as reference standards. *Methods of Information in Medicine*. 2013;52(4):308-316.
98. Livet P, Müller JP, Phan D, Sanders L. Ontology, a mediator for agent-based modeling in social science. *JASSS*. 2010;13(1).
99. Livingston KM, Bada M, Baumgartner WA, Hunter LE. KaBOB: ontology-based semantic integration of biomedical databases. *BMC Bioinformatics*. 2015;16:126.
100. Liyanage H, Liaw ST, Kuziemy C, et al. The Evidence-base for Using Ontologies and Semantic Integration Methodologies to Support Integrated Chronic Disease Management in Primary and Ambulatory Care: Realist Review. Contribution of the IMIA Primary Health Care Informatics WG. *Yearbook of Medical Informatics*. 2013;8:147-154.



101. Lomas T, Hefferon K, Ivtzan I. The LIFE Model: A Meta-Theoretical Conceptual Map for Applied Positive Psychology. *Journal of Happiness Studies*. 2015;16(5):1347-1364.
102. Lopez-Garcia P, Lependu P, Musen M, Illarramendi A. Cross-domain targeted ontology subsets for annotation: the case of SNOMED CORE and RxNorm. *Journal of Biomedical Informatics*. 2014;47:105-111.
103. Lozano-Rubi R, Munoz Carrero A, Serrano Balazote P, Pastor X. OntoCR: A CEN/ISO-13606 clinical repository based on ontologies. *Journal of Biomedical Informatics*. 2016;60:224-233.
104. Lundh L-G. Psychological science within a three-dimensional ontology. *Integrative Psychological & Behavioral Science*. 2018;52(1):52-66.
105. Maarouf H, Taboada M, Rodriguez H, Arias M, Sesar A, Sobrido MJ. An ontology-aware integration of clinical models, terminologies and guidelines: an exploratory study of the Scale for the Assessment and Rating of Ataxia (SARA). *BMC Medical Informatics and Decision Making*. 2017;17(1):159.
106. Mahdavi A, Taheri M, Schuss M, Tahmasebi F, Glawischnig S. Structured building data management: Ontologies, queries, and platforms. In: Wagner A, O'Brien W, Dong B, eds. *Exploring occupant behavior in buildings: Methods and challenges*. Cham: Springer International Publishing; 2018:261-286.
107. Malone J, Brown A, Lister AL, et al. The Software Ontology (SWO): a resource for reproducibility in biomedical data analysis, curation and digital preservation. *Journal of Biomedical Semantics*. 2014;5:1-13.
108. Marques MM, Carey RN, Norris E, et al. Delivering Behaviour Change Interventions: Development of a Mode of Delivery Ontology [version 1; peer review: awaiting peer review]. *Wellcome Open Research*. 2020.
109. Martinez-Costa C, Schulz S. Ontology content patterns as bridge for the semantic representation of clinical information. *Applied Clinical Informatics*. 2014;5(3):660-669.
110. Martinez-Romero M, O'Connor MJ, Egyedi AL, et al. Using association rule mining and ontologies to generate metadata recommendations from multiple biomedical databases. *Database*. 2019;2019.
111. Martinez-Romero M, Vazquez-Naya JM, Pereira J, Pazos A. BiOSS: A system for biomedical ontology selection. *Computer Methods and Programs in Biomedicine*. 2014;114(1):125-140.
112. Mate S, Kopcke F, Toddenroth D, et al. Ontology-based data integration between clinical and research systems. *PloS One*. 2015;10(1):e0116656.
113. Megan Kong Y, Xiang Q, Qian Y, Dahlke C, Scheuermann RH, Karp D. Toward an ontology-based framework for clinical research databases. *Journal of Biomedical Informatics*. 2011;44(1):48-58.
114. Mi H, Thomas PD. Ontologies and standards in bioscience research: for machine or for human. *Frontiers in Physiology*. 2011;2:5.
115. Michie S, West R, Finnerty AN, et al. Representation of behaviour change interventions and their evaluation: Development of the Upper Level of the Behaviour Change Intervention Ontology [version 1; peer review: awaiting peer review]. *Wellcome Open Research*. 2020.
116. Mohamad UH, Ahmad MN, Benferdia Y, Shapi'i A, Bajuri MY. An Overview of Ontologies in Virtual Reality-Based Training for Healthcare Domain. *Frontiers in Medicine*. 2021;8.
117. Morán-Reyes AA. Contribution to the Ontological Status of Information: Development of the Structural-Attributive Approach. *Library Trends*. 2015;63(3):574-590.
118. Mortensen JM, Horridge M, Musen MA, Noy NF. Applications of ontology design patterns in biomedical ontologies. *AMIA Annual Symposium proceedings / AMIA Symposium*

- AMIA Symposium*. 2012;2012:643-652.
119. Munteanu CR, Dorado J, Matei-Ilfoveanu I, Nita SA. Regulatory affairs issues and legal ontologies in drug development. *Frontiers in Bioscience (Elite edition)*. 2013;5:446-460.
  120. Ning W, Yu M, Kong D. Evaluating semantic similarity between Chinese biomedical terms through multiple ontologies with score normalization: An initial study. *Journal of Biomedical Informatics*. 2016;64:273-287.
  121. Ninomiya K, Takatsuki T, Kushida T, Yamamoto Y, Ogishima S. Choosing preferable labels for the Japanese translation of the Human Phenotype Ontology. *Genomics & informatics*. 2020;18(2):e23.
  122. Norris E, Marques MM, Finnerty AN, et al. Development of an Intervention Setting Ontology for behaviour change: Specifying where interventions take place [version 1; peer review: awaiting peer review]. *Wellcome Open Research*. 2020.
  123. Notaro M, Valentini G, Schubach M, Robinson PN. Prediction of Human Phenotype Ontology terms by means of hierarchical ensemble methods. *BMC Bioinformatics*. 2017;18(1):449.
  124. Nowroozi M, Mirzabeigi M, Sotudeh H. The comparison of thesaurus and ontology. *Library Hi Tech*. 2018;36(4):665-684.
  125. Noy N, Tudorache T, Nyulas C, Musen M. The ontology life cycle: Integrated tools for editing, publishing, peer review, and evolution of ontologies. *AMIA Annual Symposium Proceedings*. 2010;2010:552-556.
  126. Oliveira D, Pesquita C. Improving the interoperability of biomedical ontologies with compound alignments. *Journal of Biomedical Semantics*. 2018;9(1).
  127. Ongenaes F, Myny D, Dhaene T, et al. An ontology-based nurse call management system (oNCS) with probabilistic priority assessment. *BMC Health Services Research*. 2011;11:26.
  128. Osofisan A. KNOWREM: Formal definitions and ontological framework for knowledge reconciliation in economic intelligence. In: Usoro A, Majewski G, Ifinedo P, Arikpo II, eds. *Leveraging developing economies with the use of information technology: Trends and tools*. Hershey, PA: Information Science Reference/IGI Global; 2012:29-38.
  129. Ozgur A, Hur J, He Y. The Interaction Network Ontology-supported modeling and mining of complex interactions represented with multiple keywords in biomedical literature. *BioData Mining*. 2016;9(1):41.
  130. Papadouris N, Constantinou CP. Integrating the epistemic and ontological aspects of content knowledge in science teaching and learning. *International Journal of Science Education*. 2017;39(6):663-682.
  131. Perez W, Tello A, Saquicela V, Vidal M-E, La Cruz A. An automatic method for the enrichment of DICOM metadata using biomedical ontologies. *Conference proceedings : Annual International Conference of the IEEE Engineering in Medicine and Biology Society IEEE Engineering in Medicine and Biology Society Annual Conference*. 2015;2015:2551-2554.
  132. Peroni S, Shotton D, Vitali F. Tools for the automatic generation of ontology documentation: A task-based evaluation. In: Khosrow-Pour M, ed. *Computational linguistics: Concepts, methodologies, tools, and applications, Vols. I - III*. Hershey, PA: Information Science Reference/IGI Global; 2014:839-865.
  133. Pertsas V, Constantopoulos P. Scholarly Ontology: modelling scholarly practices. *International Journal on Digital Libraries*. 2017;18(3):173-190.
  134. Phelps C, Heidl R, Wadhwa A. Knowledge, networks, and knowledge networks: A review and research agenda. *Journal of Management*. 2012;38(4):1115-1166.
  135. Popejoy LL, Khalilia MA, Popescu M, et al. Quantifying care coordination using natural language processing and domain-specific ontology. *Journal of the American Medical Informatics*

- Association : JAMIA. 2015;22(e1):e93-e103.
136. Prodanov D. Data Ontology and an Information System Realization for Web-Based Management of Image Measurements. *Frontiers in Neuroinformatics*. 2011.
  137. Qasim I, Alam M, Khan S, et al. A comprehensive review of type-2 fuzzy ontology. *Artificial Intelligence Review*. 2020;53(2):1187-1206.
  138. Qiu J, Wen L. Constructing an information science resource ontology based on the Chinese Social Science Citation Index. *Aslib Journal of Information Management*. 2014;66(2):202-218.
  139. Quesada-Martinez M, Fernandez-Breis JT, Stevens R, Mikroyannidi E. Prioritising lexical patterns to increase axiomatisation in biomedical ontologies. The role of localisation and modularity. *Methods of information in medicine*. 2015;54(1):56-64.
  140. Quinn S, Bond R, Nugent C. Ontological modelling and rule-based reasoning for the provision of personalized patient education. *Expert Systems: International Journal of Knowledge Engineering and Neural Networks*. 2017;34(2):1-11.
  141. Quinn TP, Lee SC, Venkatesh S, Nguyen T. Improving the classification of neuropsychiatric conditions using gene ontology terms as features. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*. 2019;180(7):508-518.
  142. Ragab MAF, Arisha A. Knowledge management and measurement: A critical review. *Journal of Knowledge Management*. 2013;17(6):873-901.
  143. Rani M, Srivastava KV, Vyas OP. An ontological learning management system. *Computer Applications in Engineering Education*. 2016;24(5):706-722.
  144. Riley WT. Behavioral and Social Sciences at the National Institutes of Health: Methods, Measures, and Data Infrastructures as a Scientific Priority. *Health Psychology*. 2017;36(1):5-7.
  145. Ruiz-Martínez JM, Valencia-García R, Martínez-Béjar R, Hoffmann A. BioOntoVerb: A top level ontology based framework to populate biomedical ontologies from texts. *Knowledge-Based Systems*. 2012;36:68-80.
  146. Rzhetsky A, Evans JA. War of Ontology Worlds: Mathematics, Computer Code, or Esperanto? *Plos Computational Biology*. 2011;7(9).
  147. Sahoo SS, Ramesh P, Welter E, et al. Insight: An ontology-based integrated database and analysis platform for epilepsy self-management research. *International Journal of Medical Informatics*. 2016;94:21-30.
  148. Sahoo SS, Zhang G-Q, Lhatoo SD. Epilepsy informatics and an ontology-driven infrastructure for large database research and patient care in epilepsy. *Epilepsia*. 2013;54(8):1335-1341.
  149. Sanchez D, Batet M. Semantic similarity estimation in the biomedical domain: An ontology-based information-theoretic perspective. *Journal of Biomedical Informatics*. 2011;44(5):749-759.
  150. Sanchez D, Sole-Ribalta A, Batet M, Serratosa F. Enabling semantic similarity estimation across multiple ontologies: an evaluation in the biomedical domain. *Journal of Biomedical Informatics*. 2012;45(1):141-155.
  151. Santana F, Schober D, Medeiros Z, Freitas F, Schulz S. Ontology patterns for tabular representations of biomedical knowledge on neglected tropical diseases. *Bioinformatics (Oxford, England)*. 2011;27(13).
  152. Schulz S, Schober D, Daniel C, Jaulent M-C. Bridging the semantics gap between terminologies, ontologies, and information models. *Studies in Health Technology and Informatics*. 2010;160(Pt 2):1000-1004.
  153. Shen F, Lee Y. Knowledge discovery from biomedical ontologies in cross domains. *PLoS ONE*. 2016;11(8):e0160005.

154. Shen Y, Chen D, Lei K, Tang B, Yang M. EAPB: entropy-aware path-based metric for ontology quality. *Journal of Biomedical Semantics*. 2018;9(1):20.
155. Soldatova LN, Collier N, Oellrich A, et al. Special issue on bio-ontologies and phenotypes. *Journal of Biomedical Semantics*. 2015;6:40.
156. Stoutenburg SK, Kalita J, Ewing K, Hines LM. Scaling alignment of large ontologies. *International Journal of Bioinformatics Research and Applications*. 2010;6(4):384-401.
157. Sweeney TE, Mortensen JM, Januszyk M, et al. Using the wisdom of the crowds to find critical errors in biomedical ontologies: A study of SNOMED CT. *Journal of the American Medical Informatics Association*. 2015;22(3):640-648.
158. Sy MF, Ranwez S, Montmain J, Regnault A, Crampes M, Ranwez V. User centered and ontology based information retrieval system for life sciences. *BMC Bioinformatics*. 2012;13 Suppl 1.
159. Tahar K, Xu J, Herre H. Expert2OWL: A Methodology for Pattern-Based Ontology Development. *Studies in Health Technology and Informatics*. 2017;243:165-169.
160. Tang Z, Xue X, Hang Z. Interactive biomedical ontology matching. *PLoS ONE*. 2019;14(4):e0215147.
161. Tao C, Pathak J, Solbrig HR, Wei W-Q, Chute CG. Terminology representation guidelines for biomedical ontologies in the semantic web notations. *Journal of Biomedical Informatics*. 2013;46(1):128-138.
162. Tsuji S, Fukuda A, Yagahara A, et al. An ontology design for visualizing implicit knowledge of the radiotherapy risk. *Studies in Health Technology and Informatics*. 2013;192:994.
163. Uribe GA, Lopez DM, Blobel B. Towards automated biomedical ontology harmonization. *Studies in Health Technology and Informatics*. 2014;200:62-68.
164. Vanharanta H, Einolander J, Kantola J, Markopoulos E, Sivula A. Phronetic leadership style evaluation with a fuzzy logic application. *Theoretical Issues in Ergonomics Science*. 2020.
165. Viola M, Zanin E. The standard ontological framework of cognitive neuroscience: Some lessons from Broca's area. *Philosophical Psychology*. 2017;30(7):945-969.
166. Vishnyakova D, Pasche E, Lovis C, Ruch P. Utilization of ontology look-up services in information retrieval for biomedical literature. *Studies in Health Technology and Informatics*. 2013;186:155-159.
167. Walls RL, Deck J, Guralnick R, et al. Semantics in support of biodiversity knowledge discovery: An introduction to the biological collections ontology and related ontologies. *PLoS ONE*. 2014;9(3).
168. Wang KC. Standard Lexicons, Coding Systems and Ontologies for Interoperability and Semantic Computation in Imaging. *Journal of Digital Imaging*. 2018;31(3):353-360.
169. Wimmer H, Chen L, Narock T. Ontologies and the Semantic Web for Digital Investigation Tool Selection. *The Journal of Digital Forensics, Security and Law : JDFSL*. 2018;13(3):20-45.
170. Wimmer H, Yoon V, Rada R. Integrating knowledge sources: An ontological approach. *International Journal of Knowledge Management*. 2013;9(1):60-75.
171. Wright AJ, Norris E, Finnerty AN, et al. Ontologies relevant to behaviour change interventions: a method for their development [version 2; peer review: 1 not approved]. *Wellcome Open Research*. 2020.
172. Wu H, Yamaguchi A. Semantic Web technologies for the big data in life sciences. *Bioscience Trends*. 2014;8(4):192-201.
173. Wynden R, Weiner MG, Sim I, et al. Ontology mapping and data discovery for the translational investigator. *Summit on translational bioinformatics*. 2010;2010:66-70.
174. Yu AC, Cimino JJ. A comparison of two methods for retrieving ICD-9-CM data: The effect

- of using an ontology-based method for handling terminology changes. *Journal of Biomedical Informatics*. 2011;44(2):289-298.
175. Yu C, Shen B. XML, Ontologies, and Their Clinical Applications. In: Shen B, Tang H, Jiang X, eds. *Translational Biomedical Informatics: A Precision Medicine Perspective*. Vol 939. 2016:259-287.
  176. Yu F, Qiu J, Lou W. Library resources semantization based on resource ontology. *The Electronic Library*. 2014;32(3):341-362.
  177. Yu T, Chen H, Mi J, Gu P, Wu T, Pan JZ. DartWiki: A semantic wiki for ontology-based knowledge integration in the biomedical domain. *Current Bioinformatics*. 2012;7(3):278-288.
  178. Zarri GP. RDF and OWL for knowledge management. In: Khosrow-Pour M, ed. *Organizational learning and knowledge: Concepts, methodologies, tools and applications, Vols. 1-4*. Hershey, PA: Business Science Reference/IGI Global; 2012:1096-1114.
  179. Zaveri A, Shah J, Pradhan S, et al. Center of excellence in research reporting in neurosurgery--diagnostic ontology. *PLoS ONE [Electronic Resource]*. 2012;7(5):e36759.
  180. Zhang S, Zhao M, Li W, Chen G. Matching biomedical ontologies based on formal concept analysis. *Journal of Biomedical Semantics*. 2018;9(1):11.
  181. Zhang Y-F, Tian Y, Zhou T-S, Araki K, Li J-S. Integrating HL7 RIM and ontology for unified knowledge and data representation in clinical decision support systems. *Computer Methods and Programs in Biomedicine*. 2016;123:94-108.

## Appendix 1: Search Strategies

All searches were run on August 6, 2021.

### Ovid Medline

MEDLINE (Ovid) And Ovid MEDLINE(R) In-Process & Other Non-Indexed Citations  
1946 to July Week 5 2021, 1946 to August 5, 2021

- 1 ontolog\$.ti.
- 2 (informatic\$ adj2 (infrastructure\$ or architecture\$)).tw.
- 3 ((semantic or terminolog\$) adj2 (relationship\$ or mining or network\$ or model\$)).tw.
- 4 taxonom\$.tw.
- 5 (domain adj2 (content or coverage)).tw.
- 6 (knowledge adj2 (organi?ation or organi?e\$ or represent\$)).tw.
- 7 relational database\$.tw.
- 8 or/1-7
- 9 meta-analysis.pt.
- 10 meta-analysis.sh.
- 11 (meta-analys\$ or meta analys\$ or metaanalys\$).tw,sh.
- 12 systematic review.pt.
- 13 (review or overview or evaluat\$ or critique\$).ti
- 14 or/9-13
- 15 8 and 14
- 16 limit 15 to (english language and yr="2010 -Current")

### Embase (Embase.com)

1980 to August 6, 2021

- #16. #15 AND (2011:py OR 2012:py OR 2013:py OR 2014:py OR 2015:py OR 2016:py OR 2017:py OR 2018:py OR 2019:py OR 2020:py OR 2021:py)
- #15. (#9 OR #13) AND ([article]/lim OR [article in press]/lim OR [review]/lim) AND [english]/lim
- #14. #9 OR #13
- #13. #8 AND #12
- #12. #10 OR #11
- #11. review:ti OR overview:ti OR evaluat\*:ti OR critiqu\*:ti
- #10. 'meta analys\*':ab,ti OR 'meta analysis':ab,ti OR 'meta-analyses':ab,ti OR 'meta analytic':ab,ti OR metaanalys\*:ab,ti
- #9. (#1 OR #2 OR #3 OR #4 OR #5 OR #6 OR #7) AND ([systematic review]/lim OR [meta analysis]/lim)
- #8. #1 OR #2 OR #3 OR #4 OR #5 OR #6 OR #7
- #7. 'relational database':ab,ti OR 'relational databases':ab,ti
- #6. (knowledge NEAR/2 (organi?ation OR organi?e\* OR represent\*)):ab,ti
- #5. (domain NEAR/2 (content OR coverage)):ab,ti
- #4. taxonom\*:ab,ti
- #3. ((semantic OR terminolog\*) NEAR/2 (relationship\* OR mining OR network\* OR model\*)):ab,ti
- #2. (informatic\* NEAR/2 (infrastructure\* OR architecture\*)):ab,ti
- #1. ontolog\*:ti

### PsycINFO (EBSCOhost)

1597 to August 2021

S1 TI ontolog\*  
 S2 TI ( (informatic\* N2 (infrastructure\* OR architecture\*)) ) OR AB ( (informatic\* N2 (infrastructure\* OR architecture\*)) )  
 S3 TI ( ((semantic OR terminolog\*) N2 (relationship\* OR mining OR network\* OR model\*)) ) OR AB ( ((semantic OR terminolog\*) N2 (relationship\* OR mining OR network\* OR model\*)) )  
 S4 TI taxonom\* OR AB taxonom\*  
 S5 TI ( (domain N2 (content OR coverage)) ) OR AB ( (domain N2 (content OR coverage)) )  
 S6 TI ( (knowledge N2 (organi?ation OR organi?e\* OR represent\*)) ) OR AB ( (knowledge N2 (organi?ation OR organi?e\* OR represent\*)) )  
 S7 TI ( "relational database" OR "relational databases" ) OR AB ( "relational database" OR "relational databases" )  
 S8 S1 OR S2 OR S3 OR S4 OR S5 OR S6 OR S7

### **Epistemonikos**

Ontolog\* in Title or Abstract limited to last 10 years and Systematic Review