Personalized Depression Treatment Ontology

Cole Feuer, Gunnar Eastman, Nancy Zhang

Abstract

In this paper, we discuss the Personalized Depression Treatment Ontology, an ontology developed to describe the symptoms, genetic factors, and treatments for Major Depressive Disorder (MDD) and related psychiatric disorders. With ongoing advances in the medical field, new treatments are continually being developed. However, predicting how individual patients will respond to specific treatments remains a challenge due to interactions between different factors such as genetics, environment, and medical history. This ontology seeks to fill that gap by creating a detailed framework that combines medication treatment recommendations with genetic and phenotypic data, both of which are essential for customizing treatment plans. Using this ontology, healthcare professionals will be able to go through a broad range of current medications that will be recommended and make more informed decisions based on both the presenting symptoms and the patient's genetic makeup. Specifically, the system will offer treatment recommendations that take into account genetic markers and alleles associated with better responses to particular medications, allowing for more precise, individualized care. Through the use of this ontology, we hope to contribute to the growing field of personalized medicine, offering a patient-specific, more data-driven approach to the treatment of depression and related mental health conditions. This approach has the potential to significantly improve patient outcomes and reduce trial-and-error prescribing, which often leads to delays in achieving effective treatment.

Introduction

The mental healthcare field has been in rapid development, with a large number of medicinal products being developed, tested, and released to treat similar psychiatric disorders. While this progress offers hope to many, it also challenges clinicians with navigating through the vast array of available treatments to find the most suitable option. Recognizing this complexity, we have developed the Personalized Depression Treatment Ontology (PDTO) which categorizes these treatments and highlights key differences in their effectiveness across various demographic groups. This ontology aims to make it easier to identify treatments that are more likely to work for specific individuals based on their unique characteristics.

The motivation for creating this ontology comes from the realization that depression treatments are not universally effective. Every patient is different, and their responses to medications can vary significantly based on factors such as age, gender, ethnicity, and genetic predispositions. This variation often leads to a frustrating process of trial and error in finding the right treatment. To address this, our ontology integrates data from studies that investigate treatment effectiveness with genetic markers, when available. This ensures that recommendations are evidence-based and tailored to each patient as much as possible, increasing the likelihood of success and reducing unnecessary side effects or delays in finding an effective treatment.

In creating the Personalized Depression Treatment Ontology, our goal is and continues to be to improve the precision of depression treatment and reduce the reliance on trial-and-error approaches. By integrating demographic-specific and genetic data, the ontology provides a more individualized approach to mental healthcare, ultimately helping patients find effective treatments faster and with fewer setbacks. This project reflects the growing need for personalized medicine and offers a pathway toward more effective and patient-centered care in the mental health field.

Related Work

Our work is far from the first to categorize medicinal products, nor is it the first to collect published scientific data. We owe many of the concepts captured by our ontology to prior work in the field.

First, we make use of several terms from the ontology for the Identification of Medicinal Products (IDMP). The central term that appears in both ontologies is Medicinal Product, representing a single drug that the patient would take. The IDMP's representation of a Medicinal Product and its associated concepts go into much more detail than is required for our work. As such, our ontology has reduced the level of detail.

Another ontology that we reference is the Study Cohort Ontology (SCO), which is dedicated to populations for clinical studies. We use this ontology for its representations of clinical trials, which are vital to our ontology as a method by which to provide provenance for the treatments that the system recommends.

Technical Approach

The approach taken for this work involved extensive documentation and iterative maintenance of several key artifacts

throughout the project's lifespan. These included the Use Case Document, the Conceptual Model, and a Terminology List. Each of these artifacts played a vital role in ensuring the ontology's alignment with its intended purpose while allowing for flexibility to adapt to evolving requirements. Over the course of the semester, these documents underwent meticulous refinement to keep pace with the expanding scope and technical details of the project. By maintaining and refining these artifacts, the project established a robust, scalable, and contextually relevant ontology. This technical foundation was essential for addressing the complex and dynamic nature of personalized depression treatment. The resulting ontology is not only practical for clinical use but also extensible to accommodate future advancements in personalized mental healthcare.

Use Case

The Use Case Document served as an important artifact throughout the development process, providing a structured framework to demonstrate the system's intended functionality. It detailed the competency questions, which are high-level queries the system must be able to address, ensuring that our ontology met the needs of end users. Additionally, it outlined the flow of events, mapping how various processes would unfold within the system and ensuring logical consistency. The document also incorporated usage scenarios that depicted real-world applications, enabling us to consider the diverse ways the ontology might be utilized by different stakeholders. Resources, such as datasets and research articles were also recorded in this document, providing a foundation for system design and development. Figure 1 below is an overview diagram from this use case document, showcasing the different ways in which different classes of users might make use of the system.

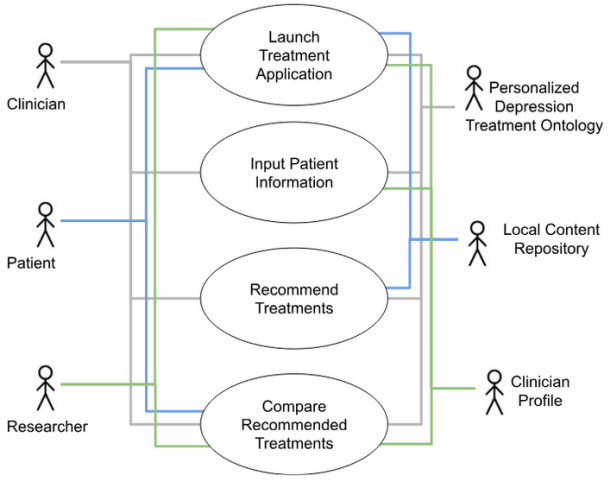

Figure 1: Activity Diagram showcasing use of PDTO

The full use case can be found on our website. [1]

[1]https://personalized-depression-treatment–rpi-ontology-engineering.netlify.app/oe2024/personalized-depression-treatment/usecase

Conceptual Model

The concept model for this project is in the form of a Google Slides presentation, which enabled us to break down the model and keep it readable. The first slide serves as an overview diagram which showcases the ontology's most important concepts: Condition, which was borrowed from the IDMP; Diagnosis; Gene; Patient; Treatment, and Treatment Response. The other 4 slides each go into further detail on a subset of the concepts from the first slide, with the second slide having more detail on the auxiliary concepts and properties of Patient and Condition; the third and fourth elaborating on Treatments and Responses; and the final slide containing more information on the Gene class.

The full conceptual model can be found on our website. [2]

[2]https://personalized-depression-treatment–rpi-ontology-engineering.netlify.app/oe2024/personalized-depression-treatment/ontology

Evaluation

The ontology was evaluated by measuring its ability to answer each of the following competency questions. These four competency questions were converted into SPARQL queries, the results of which were used to judge the success of the project.

1. What are effective treatments for patients with the SLC6A4 long (L) genetic marker?
   Sample Answer:

Treatment	Effectiveness
Citalopram	High
Escitalopram	High
Fluoxetine	High
Fluvoxamine	High
Paroxetine	High
Sertraline	High
Vilazodone	High

Table 1: Effectiveness of Treatments

2. What are effective treatments for a 35 year old patient with depression?
   Sample Answer:

Treatment Name
Citalopram
Escitalopram
Fluoxetine
Paroxetine
Sertraline

Table 2: List of Treatments

3. What alternative treatments are recommended for patients that have experienced poor response to SSRIs?
   Sample Answer:

Treatment	Effectiveness
Bupropion	High

Table 3: Effectiveness of Treatment

4. Which gene alleles impact the effectiveness of SSRIs and what are their impact?
   SPARQL Query and Results:

```
PREFIX pdt: <https://tw.rpi.edu/
    ontology-engineering/oe2024/
    personalized-depression-treatment/
    PersonalizedDepressionTreatment/>

SELECT ?gene ?treatment ?effectiveness
WHERE {
  ?gene rdf:type/rdfs:subClassOf* pdt:
      Gene .
  ?gene pdt:hasResponse/pdt:
      isResponseTo ?treatment .
  ?gene pdt:hasResponse/pdt:
      hasTreatmentEffectiveness ?
      effectiveness .
  ?treatment rdf:type/rdfs:subClassOf*
       pdt:
      SelectiveSerotoninReuptakeInhibitor
       .
}
```

Gene	Treatment	Effectiveness
SLC6A4-L	Citalopram	High
SLC6A4-L	Escitalopram	High
SLC6A4-L	Fluoxetine	High
SLC6A4-L	Fluvoxamine	High
SLC6A4-L	Paroxetine	High
SLC6A4-L	Sertraline	High
SLC6A4-L	Vilazodone	High
Val66Met_Met	Citalopram	Low
Val66Met_Met	Escitalopram	Low
Val66Met_Met	Fluoxetine	Low
Val66Met_Met	Fluvoxamine	Low
Val66Met_Met	Paroxetine	Low
Val66Met_Met	Sertraline	Low
Val66Met_Met	Vilazodone	Low
rs165599_A	Citalopram	Low
rs165599_A	Escitalopram	Low
rs165599_A	Fluoxetine	Low
rs165599_A	Fluvoxamine	Low
rs165599_A	Paroxetine	Low
rs165599_A	Sertraline	Low
rs165599_A	Vilazodone	Low

Table 4: Gene-Specific Treatment Effectiveness

The competency questions and SPARQL queries are found on our website [3].

[3]https://personalized-depression-treatment–rpi-ontology-engineering.netlify.app/oe2024/personalized-depression-treatment/demo

Discussion

highlights of the work

Value of Semantics

- Semantics enable reasoning
- Semantics help with interoperability
- Semantics enable data integration, which is theoretically helpful for this project, since adding data manually is inefficient.

Link to the website: https://personalized-depression-treatment--rpi-ontology-engineering.netlify.app/oe2024/personalized-depression-treatment/

Support of Claims

Each of our competency questions is able to be translated into a SPARQL query that will produce correct and intended results. [insert table with the SPARQL results here]

Limitations

The primary limitation for this work is the allotted time. This work taking place over the course of one semester means that our scope was severely limited. Specifically, the system is lacking in individuals, which leaves it able to reason about neither all relevant genes nor all relevant treatment options. The system also does not contain the concepts that would allow it to reason about treatments outside of medicinal products.

Future Work

Much to our chagrin, there are many directions that future work could take to improve the coverage and depth of the ontology. First is the very limited number of individuals, which could be remedied by adding more treatments, genes, and symptoms.

Future work could also focus on increasing the amount of personal data used to determine a recommendation. The system does not currently support the use of blood test results. Alternatively, non-medicinal treatments could be recommended by the system. Concepts currently exist within the ontology to capture therapeutic treatments, but the recommendation system does not utilize them, while things such as dietary changes or lifestyle interventions are not captured as concepts at all, despite their potential to treat some symptoms.

Conclusion

What we've got here is a *powerful tool* to assist in the recommendation of depression treatments, providing individualized results to help ensure that effective treatments are given to patients.

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References

- Course Materials
- IDMP