

An Ontological Model to Computerize Asthma Diagnosis and Management Clinical Guideline for Primary Care

Senanu Okuboyejo (Department of Management Information Systems, Metro State University, MN, USA, Senanu.okuboyejo@metrostate.edu) & Shiva Vaishnavi Kurakula (Faculty of Computer Science, Dalhousie University, NS, Canada, sh845098@dal.ca)

ABSTRACT

Asthma diagnosis and management in primary care present a multifaceted challenge that requires a comprehensive and patient-centered approach. This research paper introduces an innovative solution in the form of an ontological model designed to computerize asthma diagnosis and management clinical guidelines for primary care. The proposed model aims to streamline and enhance the accuracy of asthma diagnosis while providing personalized and evidence-based recommendations for patient management.

Keywords: Asthma, Clinical Guidelines, Ontology, Primary care

Introduction

Asthma is a severe global challenge with increasing prevalence. Clinical efforts are in place to reduce the prevalence; however, it varies across locations, leading to varied outcomes. However, adopting a comprehensive patient-centered interdisciplinary team approach to asthma management is pertinent, including diagnosis, assessing control, treating and preventing exacerbation, and ongoing follow-up. Hence, the use of clinical guidelines. Clinical Practice Guidelines (CPGs) are created to decrease extreme variances in clinical practices, enhance healthcare quality, and control costs. Clinical practice guidelines (CPGs) are developed in response to the need to standardize and formalize healthcare practices (Oliveira et al., 2014). Because of the significance of CPG, medical institutions encourage their implementation and deployment via computer systems, so physicians are provided with decision-making support (Sobrado et al., 2004).

Data representation within CPGs is a significant challenge for creating computer-based guidelines. CPGs usually rely on the cognitive models of the domain expert (clinicians) that are often based on technical and procedural knowledge, which can carry implications for developing CPGs. A possible solution to this challenge is to create a unique and standard representation model that allows the guidelines to be shared among various medical institutions and provides consistency when interpreting the rules. CPG formalization as computer-interpretable CPGs provides a better opportunity to influence clinical practice than narrative guidelines (Samara et al., 2019).

Ontology is vital for expressing latent knowledge in texts and making it understandable to humans and computers. An ontology is a formal and structural depiction of a shared understanding, ideas, and relationships, highlighting the domain's concepts, relations, attributes, and hierarchies. Ontologies describe and reuse medical information and frames for representation. Our goal is to develop an ontology framework to express practice-oriented temporal information necessary for providing primary for Asthma disease. Ontologies are created by extracting relevant information instances from text using an ontology population approach. However, handcrafting such huge ontologies is complex, and it is impossible to develop ontologies for all available domains. As a result, rather than handcrafting ontologies, the research trend is shifting toward autonomous

extraction of ontologies from text, a process known as ontology learning. We aim to develop an ontology to express practice-oriented temporal information for Asthma CPG. This work focuses on ontologies to formalize knowledge embodied in Asthma CPG. We intend to answer the following competency questions to evaluate the completeness of the ontology:

- What are the typical presenting symptoms of Asthma?
- What are clinical procedures or investigations required to confirm asthma diagnosis?
- What are the diagnostic thresholds that confirm asthma diagnosis?
- What are the first-line treatment options given to presenting asthma symptoms?
- What are possible drugs for Asthma management?

Design Approach and Methodology

We adopted a knowledge modeling approach, particularly Semantic Web technologies, to model the domain knowledge in terms of an ontology. Knowledge modeling involves abstracting domain-specific knowledge into concepts that encapsulate the domain knowledge, problem-solving behavior, operational processes, and functional constraints. In healthcare, domain-specific knowledge is acquired and represented in clinical practice guidelines, clinical pathways, and research articles (S. Abidi, 2017; S. S. R. Abidi & Shayegani, 2009; Hurley & Abidi, 2007). The asthma ontology captures the diagnostic, management, and operational concepts and shows the semantic relationship between these concepts (classes).

The Asthma CPG ontology is classified along the operational dimension, belonging to the primary care category. Primary care often refers to check-ups, preventative care, and treating common ailments and is usually provided by a family medicine physician or nurse practitioner at a clinic/CLSC. This CPG represents the primary care process for Asthma as provided by the care provider. CPG needs to be computerized before executing. We used the CPG ontology developed by (S. S. R. Abidi & Shayegani, 2009) to computerize a CPG, as it builds on all the existing CPG ontologies. To create the Asthma CPG Ontology, we adapted "Methontology," the development process proposed by Fernandez et al. (1997). Methontology is a structured method for building ontologies and has been used extensively in the literature.

Ontology Development Process

Our development process comprises the following steps:

- 1.1. Asthma CPG Knowledge Specification:** In this phase, we identified the purpose of the ontology, including its intended uses, scenarios of use, end-users, and Scope, which includes the set of terms to be represented, its characteristics and granularity, and a set of competency questions the ontology would seek to answer.
- 1.2. Asthma CPG Knowledge Acquisition:** Aside from the evidence-based asthma diagnosis and management algorithm provided¹, we consulted relevant knowledge sources for more information on asthma care processes, workflow, diagnosis and diagnostic steps, indications and contraindications, thresholds, drug management protocols, Review, and control therapies (BC Guidelines and Protocols Advisory Committee, 2015; Registered Nurses' Association of Ontario., 2017; Yang et al., 2021). This process was iterative and occurred at every step of the development process. This provided more information needed to simplify complex and ambiguous processes and helped synthesize and validate the knowledge represented by the

¹ <https://hcp.lunghealth.ca/wp-content/uploads/2020/02/Asthma-Diagnosis-and-Management-Algorithm.pdf>

ontology. We also conducted informal text analysis for an initial conceptualization representation and formal text analysis to identify the structures and corresponding knowledge.

1.3. Asthma CPG Knowledge Synthesis: CPGs are high-level abstractions of disease management recommendations not directly applicable in a clinical decision support setting (S. Abidi, 2017). Thus, in executing the Asthma CPG, it was transformed into a clear clinical pathway that outlines the operational workflow and the clinical tasks – clinical evaluations, assessments, diagnosis, management, and review - sequentially. We structured the CPG knowledge in a conceptual model using a business process modeling (BPM) diagram (See Appendices 3 & 4).

1.4. Asthma CPG Knowledge Modeling and Representation: This step computerized the clinical pathway designed in the knowledge synthesis phase concerning the domain concepts, description, dependencies and constraints, decision rules, and preconditions. We used semantic web technologies to model the Asthma CPG ontology, where OWL was used to represent knowledge through the Protégé editor. To speed up the ontology construction, we considered the concept of "reuse." Instead of building from scratch, we adapted the CPG ontology developed by (Abidi & Shayegani, 2009). To visualize the ontology, we used the “OntoGraf” plugin.

- **Asthma CPG Ontology**

The Asthma CPG ontology comprises major hierarchical classes and properties to represent the domain of study. The superclasses are listed below and visualized in Figure 1, while the properties are shown in Table 1.

- i. Patient: refers to individual patients and comprises the patient age classification.
- ii. Clinical_Guideline: points in the clinical pathway (CP) at which the process is initiated. It also comprises the first step in the CPG.
- iii. Clinician – which represents the HCP or Physician.
- iv. Dose_Schedule: This captures information about the dosage of the ordered medication and the schedule for its consumption using attributes such as *dose*, *dose-unit*, and *dose-measured*.
- v. Drug refers to all the medication groups involved in treating Asthma, such as SABA, ICS, LAMA, LABA, and LTRA.
- vi. Guideline_Step: represents all care activities such as diagnostic, assessment, education, treatment, visit, and treatment_choice in the Asthma CPG. The Guideline_Step has three sub-classes: Action_Steps, Decision_Steps, and Route_Steps, with further sub-classifications.
- vii. Intervention: models the diagnostic and treatment interventions performed during care delivery. There are two sub-classes, *Intervention_for_treatment* and *Intervention_for_Diagnosis*, with further sub-classifications.
- viii. Schedule: It models different types of temporal schedules to organize activities. It has the following attributes: schedule-type, repetitions, and duration.
- ix. Decision_Option refers to all the decision points in the clinical pathway.

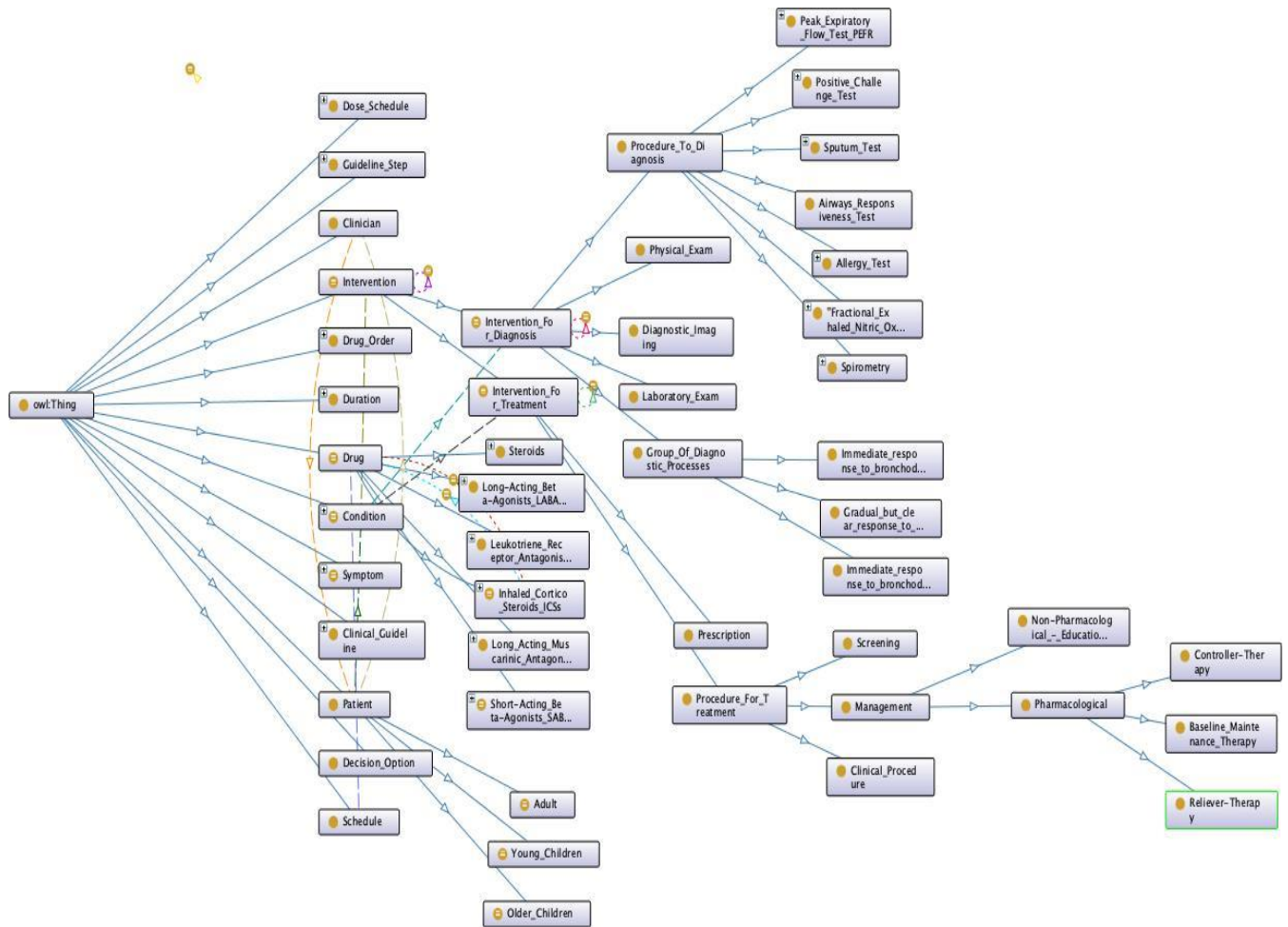


Figure 1: Asthma CPG Ontology Visualization

Table 1: A Sample of Major Properties Implemented

Property Name	Domain	Range	Type
hasAge	Patient	Integer	Data
hasFEV1_post_bronchodilator	Patient	Double	Data
hasFEV1_post_controller_therapy		Double	Data
hasFEV_FVC_less_than_0.7	Patient	Double	Data
hasFEV_FVC_less_than_0.8	Patient	Double	Data
hasSymptom	Condition and Patient	Boolean	Data
hasCondition	Patient	Condition	Object
hasDoseSchedule	Drug_Order	Dose_Schedule	Object
hasDrug	Condition and Drug_Order	Drug	Object
hasDuration	Dose_Schedule	Duration	Object
hasFirstLineTreatment	Condition	Drug	Object
hasIntervention	Condition	Intervention	Object
hasSecondLineTreatment	Condition	Drug	Object

We attempt to answer our competency questions to demonstrate the knowledge representation in the ontology.

- **What are the typical presenting symptoms of Asthma?**

The algorithm starts with a patient presenting with Asthma symptoms, which were not prioritized or made specific. We queried the Symptom class with the Boolean data property "hasSymptom" to infer asthma symptoms from our ontology. It returned all instances of the class, as shown in Figure 2.

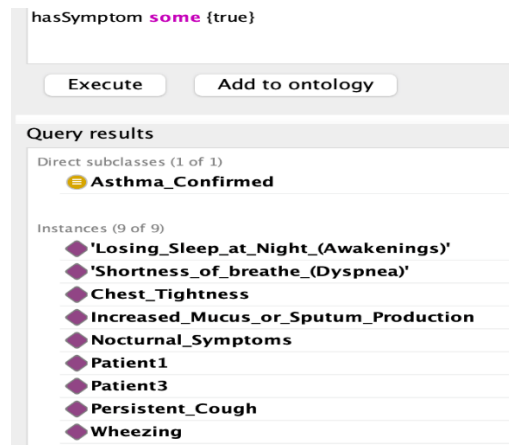


Figure 2: Screenshot of Asthma Symptoms

- **What are the clinical procedures or investigations required to confirm asthma diagnosis?**

The procedures and other actions taken towards confirming asthma diagnosis were regarded as interventions and thus were classified as Intervention_For_Diagnosis. Our ontology was queried, resulting in 38 instances (individuals) belonging to 5 subclasses, as shown in Figure 3.

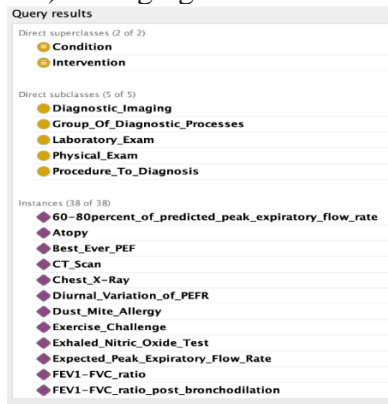


Figure 3: Screenshot of Clinical Investigations for Asthma Diagnosis Confirmation

- **What are the diagnostic thresholds that confirm asthma diagnosis?**

Objective measurements are needed to confirm the diagnosis of Asthma in all patients and assess its severity. A diagnostic threshold is a probability above which the diagnosis is so likely and below which the diagnosis is unlikely; it is excluded without further testing. The parameter values of the various interventions for diagnosis were predetermined from the ontology to confirm asthma diagnosis objectively. It was implemented as a necessary condition for Asthma confirmation. The condition is depicted in Figure 4.

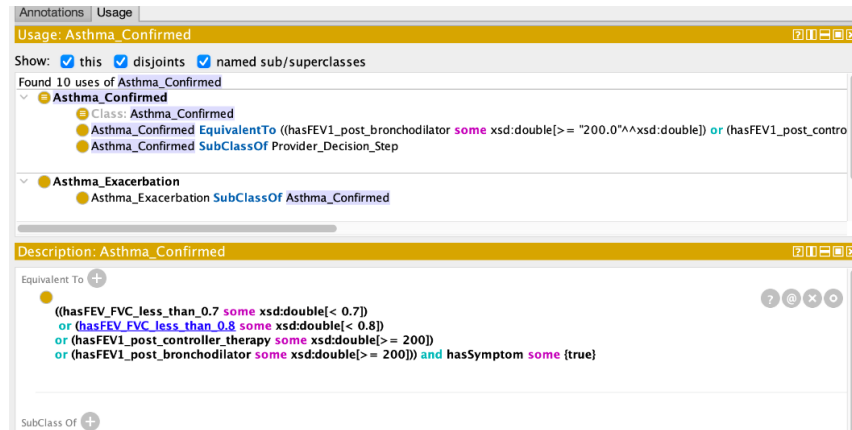


Figure 4: Screenshot of Diagnostic Thresholds for Asthma Implemented as Conditions

- **What are the first-line treatment options given to presenting asthma symptoms?**

Asthma Management options comprise pharmacological and non-pharmacological treatment options. The pharmacological option includes first-line and second-line treatment and referral (in case of an exacerbation). All patients should be on a reliever – SABA, in the first line of treatment. If Asthma is not controlled, then the second line of treatment is initiated (ICS) alongside the first line of treatment. Figure 5 shows both the first and second lines of treatment.

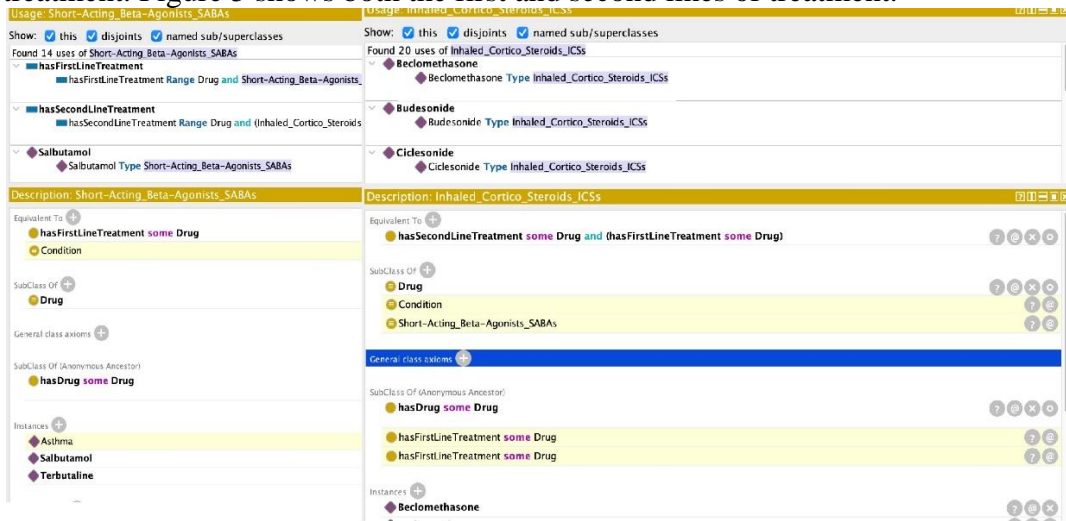


Figure 5: Screenshot of both First and Second Lines of Treatment.

- **What are possible drugs for Asthma management?**

The following list of medications is related to or used in treating Asthma. Our ontology formalized the drugs into classes. These drugs will be prescribed based on the severity of the disease and response to treatment by the HCP. The DRUG class was queried, resulting in six asthma medication classes, as shown in Figure 6.

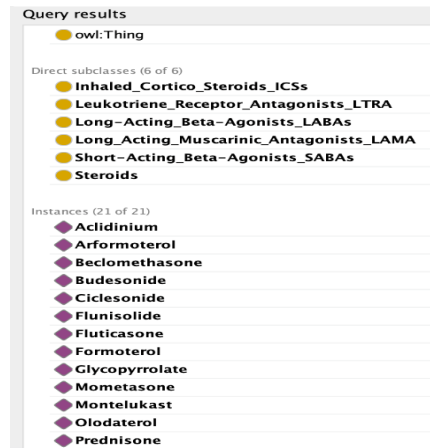


Figure 6: Screenshot of Asthma Drugs

3.5. Asthma CPG ontology Evaluation (Validation): *Validation* guarantees that the ontologies, the software environment, and documentation correspond to the system they represent. The ontology was evaluated for consistency, completeness, and conciseness (Gómez-Pérez, 2004). We used **HermiT and Pellet** interchangeably (ontology reasoners for OWL ontologies) to determine ontology consistency based on class descriptions, identify subsumption relationships between classes, and infer class hierarchy. We also encoded competency questions to validate the structure and completeness of our ontology. The reasoners did not detect any inconsistency; hence, the Asthma CPG ontology was deemed consistent and coherent.

We simulated a use case scenario to test the inference of our ontology –

“Patient UseCase_1 is a 22-year-old male. He is presenting with a persistent cough and has difficulty sleeping at night. His spirometry parameters indicate an FEV1/FVC ratio of 0.9 upon testing.”

Inference – Patient UseCase_1 is an **Adult** patient; **Asthma** is confirmed and will commence the **first line of treatment**, including **drugs in the SABA** class. The inference procedure of the ontology is depicted in Appendix 2.

2. CONCLUSION

The present paper describes the work performed to implement the Asthma CPG. We present an ontology enabling Asthma CP's computerization to manage, plan, and streamline care activities. A semantic evaluation of the ontology shows that all implementation requirements are satisfied. Implementing this ontology presented some challenges, such as partial (insufficient) knowledge to correctly classify a clinical process or activity, inadequate temporal relations and information on the order and duration of treatment interventions, and differentiating between defaults and exceptions.

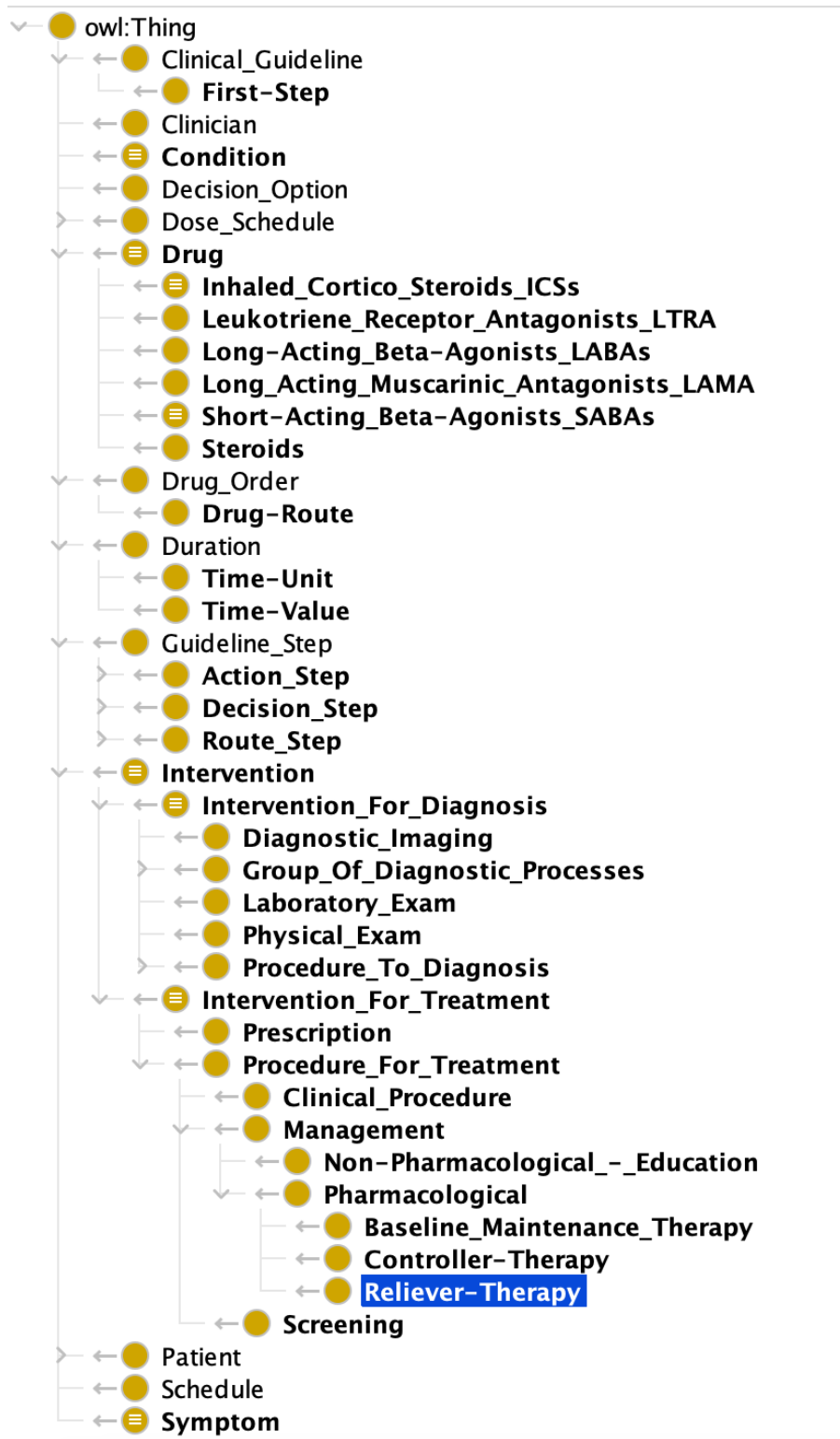
Ontologies connect theoretical knowledge (tacit knowledge) and point-of-care solutions. It provides formal methods for care delivery, which can be reused across other respiratory diseases without significant effort. The CPG algorithm implemented in this study describes asthma care guidelines for Ontario (location-specific). However, formalizing the CPG promises a scalable, sustainable model that can be expanded anytime, knowledge generalization across different locations due to a formalized knowledge base, and identification of location-specific tasks.

Obtaining an effective solution to the real problem of Asthma through implementing a CPG would improve how this disease is treated and, hence, patients' quality of life.

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Appendix 1: View of the Developed Ontology with Protégé



Appendix 2: Patient UseCase_1

The image displays a Semantic Web editor interface with several panels:

- Left Panel (Class Hierarchy):** A tree view showing the ontology structure. The path `owl:Thing > Asthma_Confirmed` is highlighted. Other classes include `Clinical_Guideline`, `Condition`, `Decision_Option`, `Drug`, `Drug_Order`, `Duration`, `Guideline_Step`, `Action_Step`, `Decision_Step`, `Provider_Decision_Step`, `Asthma_Confirmed`, `Asthma_Not_Confirmed`, `System_Decision_Step`, `Route_Step`, `Intervention`, `Patient`, `Schedule`, and `Symptom`.
- Top-Left Panel (Patient UseCase_1):** A list of instances. `Patient_UseCase_1` is selected and highlighted in blue. Other instances include `Olodaterol`, `Oral`, `Patient presents with Asthma symptoms`, `Patient5`, and `Patient6`.
- Top-Right Panel (Property assertions: Patient_UseCase_1):** Shows object and data property assertions for the selected instance.
 - Object property assertions:**
 - `hasFirstLineTreatment Patient_UseCase_1`
 - `isFirstLineTreatmentFor Patient_UseCase_1`
 - Data property assertions:**
 - `hasSymptom true`
 - `hasFEV_FVC_less_than_0.7 "0.6"^^xsd:double`
 - `hasAge 22`
- Bottom-Left Panel (Class hierarchy: Asthma_Confirmed):** Shows the specific class hierarchy for `Asthma_Confirmed`, including `Provider_Decision_Step`, `Asthma_Confirmed`, and `Asthma_Not_Confirmed`.
- Bottom-Right Panel (Description: Asthma_Confirmed):** Displays the logical definition of the class.
 - Equivalent To:** `((hasFEV1_post_bronchodilator some xsd:double[>= "200.0"^^xsd:double]) or (hasFEV1_post_controller_therapy some xsd:double[>= "200.0"^^xsd:double]) or (hasFEV_FVC_less_than_0.7 some xsd:double[< "0.7"^^xsd:double]) or (hasFEV_FVC_less_than_0.8 some xsd:double[< "0.8"^^xsd:double])) and (hasSymptom some (true)))`
 - SubClass Of:** `hasFirstLineTreatment Self`, `Provider_Decision_Step`, `Condition`, `Patient`, `Short-Acting_Beta-Agonists_SABAs`, and `Symptom`.
 - General class axioms:** `hasFirstLineTreatment some (Drug or Patient)`, `hasFirstLineTreatment Some Drug`, and `hasSymptom some (true)`.
 - Instances:** `Patient_UseCase_1` is listed as an instance.

Appendix 3

