Towards Ontological Conversation Interpretation: A Method for Ontology Creation from Medical Guidelines

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Abstract. The automated capturing and summarization of medical consultations aims to reduce the administrative burden in healthcare. Consultations are structured conversations that broadly follow a guideline with a systematic examination of predefined observations and symptoms to diagnose and treat well-defined conditions. While the administrative burden could be relieved via automated conversation summarization systems, the availability of medical ontologies for the interpretation of the medical knowledge is still an obstacle. This paper introduces ontological conversation summarization by formalizing the representation of medical guidelines to develop a method for the systematic construction of ontologies from the human anatomy and medical guidelines. The well-known SNOMED CT nomenclature and medical guidelines from a medical authority in the Netherlands are used to develop: (i) a formalism for the target ontologies in the form of a Patient Medical Ontology (PMO), and (ii) a procedural method to develop such ontologies expressed in the form of a Process-Deliverable Diagram model. The PMO of the medical condition of ear canal inflammation (Otitis Externa) is created to validate the method.

Keywords: Ontology learning · knowledge extraction · method engineering · knowledge representation · SNOMED CT · patient medical ontology.

1 Introduction

The automated summarization of conversations have the potential to save time and cost, especially in domains where dialogues are structured based on predefined guidelines. Medical consultations are a prime example as they broadly follow a systematic examination of predefined symptoms and observations to diagnose and treat well-defined conditions affecting fixed human anatomy [7].

The expanding potential of automated conversation summarization systems in medicine and in other fields depends on the readiness of the structured representations of domain-specific knowledge in a machine-processable format. In particular, rule-based conversations summarization grants explainability that is
unachievable with machine learning approaches, thus, ontologies can efficiently represent domain guidelines in such rule-based applications [23].

An ontology is the explicit specification of a conceptualization [5], a formal description of the concepts in a domain and the relationships between these concepts. In the context of conversation summarization, the ontology represents the diverse and primarily textual domain knowledge (e.g., human anatomy and medical guidelines) in a machine-processable fabric of concepts and relationships. Manually building a domain ontology is time-consuming and error-prone; thus, it should be carried out using systematic ontology learning: constructing and integrating a machine-readable semantic-rich ontology (semi-)automatically [26].

However, tools and techniques for ontology learning primarily aim to extract knowledge from general data corpora as it was, and still is, driven by the necessity of linking either openly available data (e.g., DBpedia [9]) or corporate-specific business data (e.g., Google Knowledge Graph [6]). Therefore, research is needed to develop a method that enables the systematic representation of domain guidelines in machine-processable ontologies. This paper introduces the notion of ontological conversation interpretation (Fig. 1) by presenting a method for developing ontologies in the medical domain to support the expansion of the automated conversation summarization system Care2Report [11].

The Care2Report system proposed an approach to reduce the administrative burden in medical care by automating the capturing of, transcription of, and report generation from medical consultations [11]. Care2Report’s summarization pipeline relies on an ontology embodying the domain’s vocabulary and guidelines that act as the structured container to be filled with the multimodal information from the medical consultation. The information extracted from the conversation

![Fig. 1: Ontological Conversation Interpretation with the role of the Medical Ontology for the semantic interpretation in the Care2Report Summarization Pipeline [12]](image)
populates the ontology to generate the rule-adhering report that the physician checks before uploading to the Electronic Medical Records system (EMR) [12]. This paper investigates elements of the second stage of the Care2Report pipeline: formal representation (Fig. 1). The multimodal data recorded and processed during the medical consultation is formally expressed using a knowledge representation formalism to allow further analysis in later stages.

The medical guideline we use is obtained from the Dutch College of General Practitioners (Nederlands Huisartsen Genootschap - NHG) [13]. The medical domain knowledge is assembled by analyzing the human anatomy and medical terminologies provided by the Systematized Nomenclature Of Medicine - Clinical Terms (SNOMED CT) [20]. The outcome consists of the two perspectives of the ontology learning method: the notational side represented by the Patient Medical Ontology (PMO), and the procedural method expressed in a Process-Deliverable Diagram (PDD) model. The design science research cycle, as described by Wieringa [24], is fitting to answer the research question: How to systematically construct ontologies from the human anatomy and medical guidelines?

The paper makes three contributions:

– We introduce and formalize the Patient Medical Ontology (PMO), an all-encompassing ontology that describes the patient’s anatomy, symptoms, physician’s observations, diagnosis, and treatments.
– We define a procedural method to develop such ontologies in the form of a Process-Deliverable Diagram model.
– We illustrate the PMO and the procedure to the case of the external ear canal inflammation.

*Paper organization.* After reviewing the related work in Sec. 2, we introduce the PMO in Sec. 3 and its formalization in Sec. 4. We then present our method in Sec. 5 and its application in Sec. 6. Finally, we draw conclusions in Sec. 7.

## 2 Guidelines and Nomenclature in Medical Informatics

### 2.1 Medical Guidelines

A medical guideline is a digital document with definitions and procedural instructions for executing an anamnesis, diagnosis and treatment in care provisions that aim to advance care quality, improve the patient’s health and well-being, and support medical decision-making. It is based on systematic summaries of scientific research, the considerations of various care options, and care professionals’ expertise. Numerous international and national medical authorities publish and maintain medical guidelines [25].

In the Netherlands, both the Dutch College of General Practitioners (Nederlands Huisartsen Genootschap - NHG) and the Dutch Federation of Medical Specialists (Federaatie Medisch Specialisten - FMS) publish guidelines in several specializations [13, 4]. The guidelines include sections about prognosis, common
causes and background, physical examination and diagnosis guidelines, treatment policy, consultations and referral guidelines (if any), and control and future patient check-ups.

The symptoms and observations indicating a condition and the treatments recommended for such condition by the guidelines are relevant to the construction and population of the Patient Medical Ontology (PMO) and the related sub-ontologies, as will be detailed in the coming sections. As the PMO (and ultimately the resulting consultation knowledge graph and consultation report) aims to represent the conversation between the patient and the physician rather than support the physician’s decision making, the reasoning behind which treatment to choose is beyond our scope. Therefore, the PMO and its accompanying ontology and knowledge base should contain all treatment possibilities as options, while the physician’s discretion will decide which ones to use.

2.2 Medical Nomenclature: SNOMED CT

The Systematized Nomenclature of Medicine - Clinical Terms (SNOMED CT) is currently the world’s most extensive clinical terminology system [1]. This paper uses SNOMED CT as an ontology source representing human anatomy and terminology hierarchy to identify relevant medical concepts from all the potential concepts in the textual medical guidelines. The terminology structure is in hierarchical formations of concepts defined and connected to each other by relationships, with identifiers for machine use and descriptors for human readability (figure 2).

Fig. 2: SNOMED CT Design [20]
The top node of the SNOMED CT hierarchy is occupied by the root concept *SNOMED CT concept*, and nineteen direct subtypes of it are the *top level concepts* that provide the structure of the SNOMED CT (figure 2). Thus, different conditions and medical consultations will use a subset of the available concept hierarchies.

3 Patient Medical Ontology

The Patient Medical Ontology (PMO) is an all-encompassing ontology representing the patient’s anatomy and symptoms and the physician’s observations, diagnosis and prescribed treatments, where symptoms are subjective abnormalities that the patient perceives, while observations are objective abnormalities detected by the physician [8]. The full PMO is illustrated in Fig. 3: the metamodel is in Fig. 3a, its instantiation via our reference notation in Fig. 3b.

The PMO consists of five (sub)ontologies. The Patient Anatomy Ontology (PAO) depicts all the human anatomical structures and functions. The Patient Symptoms Ontology (PSO) represents all the complaints and anomalies (symptoms) reported by the patient. The Patient Observations Ontology (POO) represents all the observations made by the physician about the patient’s condition. The Patient Diagnosis Ontology (PDO) describes the physician’s diagnosis of the patient’s condition. Finally, the Patient Treatment Ontology (PTO) describes all the treatments prescribed by the physician, including medications, instructions, referrals, or additional medical tests.

The Patient Anatomy Ontology (PAO) is the foundation for the knowledge representation in the PMO. The PAO can be built based on existing resources like the Foundational Model of Anatomy [18], or it can utilize an existing hierarchical terminology structure like SNOMED CT, as in this research. Within the SNOMED CT hierarchies, the human anatomy is represented in one section: *SNOMED CT concept* $\rightarrow$ *Body structure* $\rightarrow$ *Anatomical or acquired body structure* [21]. The key concepts in the PAO are those of *Anatomical Unit* and *Relation*. The former is specialized by *Anatomical Unit* (e.g., ear, left eye) and *Function* (e.g., hearing), with the *has* relationship linking the two, e.g., ‘an ear has the function of hearing’. The *Relation* is meant to represent other types of links between anatomical units, such as part-of, next-to, etc.

The medical report produced by the automated conversation summarization system Care2Report to upload into the EMR follows a predefined structure [2] of four sections:

- Subjective: what the patient reports;
- Objective: what the physician identifies;
- Evaluation: assessment and diagnosis by the physician;
- Plan for treatment and follow up.

Following the same arrangement, the PMO consists of a composite of four sub-ontologies reflecting the four aspects of reporting and a fifth foundational ontology for human anatomy.
Each of the those (anatomy, symptoms, observations, diagnosis, and treatment) builds on the previous one(s) to include further knowledge into the resulting PMO shown in Fig. 3. The PMO provides the notational aspect of the ontology development method proposed in this paper.

The PMO and its instantiation into a consultation knowledge graph with information from specific consultation provide a graphical representation of ontology-based knowledge where every two concepts and the relationship between them form a triple. Examples of such triples are introduced in the coming sections.
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4 Formalization of the PMO

The structure and the interaction between the various ontologies are relatively intuitive for simple conditions concerning comparatively simple body structures. However, adding the complete human anatomy and the guidelines from several medical authorities can make the ontologies less intuitive to understand and communicate. Therefore, it is essential to introduce formal descriptions of the ontologies and their components; this section introduces a few of these descriptions. Formally defining the PMO requires the definition of the following sets:

| Set | Description |
|-----|-------------|
| $B$ | all anatomical structures of the body. |
| $F$ | all anatomical functions of the body. |
| $A$ | all anatomical units ($A = B \cup F$). |
| $P$ | all patients (A patient is also an anatomical unit: $P \subset A$). |
| $S$ | all medical symptoms (reported by the patient). |
| $O$ | all medical observations (observed by the physician). |
| $V$ | all possible values to be assigned to symptoms $s \in S$ or observations $o \in O$. |
| $D$ | all medical diagnoses. |
| $E$ | all explicit diagnoses. |
| $I$ | all implicit diagnoses. |
| $T$ | all medical treatments. |

The formal definitions of the ontologies comprising the PMO are the following:

- **PAO**: $A = B \cup F$, edges: $\{(b_1, b_2) \mid b_1, b_2 \in B \land b_1 \text{ is a direct anatomical sub-part of } b_2\} \cup \{(b, f) \mid b \in B \land f \in F \land b \text{ has an anatomical function } f\}$
- **PSO**: $A \cup S \cup V$, edges: $\{(a, s), (s, v) \mid a \in A \land s \in S \land v \in V\}$ i.e., all symptoms
- **POO**: $A \cup O \cup V$, edges: $\{(a, o), (o, v) \mid a \in A \land o \in O \land v \in V\}$ i.e., all observations
- **PDO**: $P \cup D$, edges: $\{(p, d) \mid p \in P \land d \in D\}$ i.e., all the diagnoses of patient $p$
- **PTO**: $P \cup T$, edges: $\{(p, t) \mid p \in P \land t \in T\}$ i.e., all the treatments of patient $p$

And, as per Section 3, the PMO is an all-encompassing knowledge ontology representing the patient’s anatomy and symptoms and the physician’s observations, diagnosis and treatments: $\text{PMO} = \text{PAO} \cup \text{PSO} \cup \text{POO} \cup \text{PDO} \cup \text{PTO}$.

Finally, some of the rules that need to be coded into the system to define the relationships between the entities are listed below, consulting domain experts is expected to add to this list. We provide some examples, but note that the creation of a comprehensive list of rules is domain-specific and goes beyond the purpose of this paper:
1. Each anatomical structure (b) is a part of another anatomical structure (b) unless it is the complete body structure (b*).
\[ \forall b_1 \exists b_2 : \text{isPartOf}(b_1, b_2) \quad b_1, b_2 \in B, \neg (b_1 = b*) \]

2. Each function (f) is assigned to one or more anatomical structure (b).
\[ \forall f \exists b : \text{hasFunction}(b, f) \quad b \in B, f \in F \]

3. Each symptom (s) is associated with one or more anatomical unit (a).
\[ \forall s \exists a : \text{hasSymptom}(a, s) \quad s \in S, a \in A \]

4. Each observation (o) is associated with one or more anatomical unit (a).
\[ \forall o \exists a : \text{hasObservation}(a, o) \quad o \in O, a \in A \]

5. A patient (p) is diagnosed with a diagnosis (d).
\[ \forall p \exists d : \text{diagnosedWith}(p, d) \quad p \in P, d \in D \]

6. A patient (p) is treated with a treatment (t). A treatment encompasses any physician’s prescription, including medications, instructions for the patient to follow, referral to a specialist, further tests, or any other procedure.
\[ \forall p \exists t : \text{treatedWith}(p, t) \quad p \in P, t \in T \]

7. An explicit diagnosis (e) is associated with a symptom (s) or an observation (o).
\[ \forall e \exists s : \text{associatedWith}(s, e) \lor \text{associatedWith}(o, e) \quad e \in E, s \in S, o \in O \]

8. An implicit diagnosis is not explicit. That is, an implicit diagnosis (i) is neither associated with a symptom (s) nor an observation (o).
\[ \forall i \forall e : \neg (i = e) \quad i \in I, e \in E \]

5 Method for Systematic Creation of Medical Ontologies

While the previous section introduced the PMO notations representing the medical guidelines ontology, this section introduces the procedural method perspective outlined in a Process-Deliverable Diagram (PDD) model. The PDD illustrates the activities and artefacts of a specific process [22]. The model depicts the activities on the left-hand side of the PDD diagram using UML activity diagram notations, while the deliverables are on the right-hand side of the diagram using UML class diagram notations [15]. The model emphasizes the relationships between the activities and their deliverables by connecting them with dotted arrows across the diagram [22].

5.1 Ontology Creation PDD

Fig. 4 shows the used PDD, in which the process is broken down into eight main activities. For simplicity, activities three to seven are illustrated (and manually performed) sequentially; however, they can also be executed in parallel.

1. **Target guideline preparation**: The guideline of the medical condition is selected from the medical authority’s website, translated (if necessary), scraped, and prepared for the following concept extraction activity.
2. **Concept extraction**: The relevant sections of the guidelines are identified, including sections describing symptoms, physical examination, and treatment plans. As nouns are the natural language representation of things, ideas and notions, they identify the concepts to be extracted (rather than other parts of speech). Thus, the potential concepts to extract are all nouns and noun phrases in the relevant sections that will be mapped in the next steps against the SNOMED CT to identify the constituent concepts of the ontology (e.g., anatomical units, symptoms, observations, and treatments). Some potential concepts will not be used in the ontology as they represent general nouns used in the text.

3. **Patient Anatomy Ontology (PAO) construction**: The guideline concepts corresponding to SNOMED CT anatomical concepts are identified and converted into a hierarchy from which the PAO is constructed.

4. **Patient Symptoms Ontology (PSO) construction**: The concepts identified in the medical guideline sections describing symptoms are mapped against the corresponding SNOMED CT hierarchies (findings and disorders) to build the PSO.

5. **Patient Observations Ontology (POO) construction**: Similar to the previous activity except dealing with physician observations instead of patient-described symptoms. Thus, the relevant guideline sections are different.

6. **Patient Diagnosis Ontology (PDO) construction**: The medical condition or disease discussed in the guideline is the diagnosis associated with symptoms and observations to construct the PDO.

7. **Patient Treatment Ontology (PTO) construction**: The concepts identified in the treatment-related sections of the guideline are mapped against
the corresponding SNOMED CT hierarchies (procedure, substance, dose form, and physical object) to build the PTO.

8. **Patient Medical Ontology Finalization**: All the previous (sub)ontologies are combined along with needed information (e.g., prefixes) to construct the complete PMO. This activity also includes checks to validity by confirming the lack of disjoint concepts.

### 5.2 A Detailed Look on Ontology Creation

Figure (5) expands the PAO construction activity to show the various subactivities and the resulting deliverables. The PDD activities constructing the remaining ontologies follow the same general high-level pattern to build the relevant ontology based on the list of extracted potential concepts from the guidelines. Potential concepts are mapped at each stage to both the relevant sections of the guidelines (e.g., physical examination and treatment policy) and the relevant hierarchies of the SNOMED CT (e.g., findings and disorders). Thus, the concepts that show in both relevant modules are the appropriate candidate concepts for the ontology at hand (e.g., symptom concept, observation concept or treatment concept). An anatomical unit (that can have symptoms, observations, diagnosis, or treatment) can be either an anatomical structure or a function. An anatomical structure is “a physical anatomical entity and a physical object, ... it consists of parts that are themselves anatomical structures”, anatomical structures are “...localized to a specific area or combine and carry out one or more specialized [anatomical] functions of an organism.” [17].

For a detailed illustration of the complete PDD and a description of all the (sub)activities and concepts, refer to the technical report [3].

![Fig. 5: Patient Anatomy Ontology (PAO) construction](image-url)
6 Application to the Otitis Externa case

To illustrate the PMO notations and apply the derived procedure to create an ontology, we use the NHG guideline for the inflammation of the external ear canal known as Otitis Externa [14]. The condition is chosen as an example for the relative simplicity of the associated guidelines and the lack of complicated medical procedures, terminology, or differential diagnosis. Otitis Externa is an inflammation caused by a disturbance in the typical acidic environment of the ear canal and thus is usually associated with swimming. The symptoms reported by the patient may include ear pain, itching in the ear, fluid drainage from the ear, and hearing loss. In addition, the physician examines both the complaint-free ear and the affected ear for signs of scars, swelling, flaking, redness, and the state of the eardrum, among others [14].

Some symptoms (e.g., hearing loss) are not required for an Otitis Externa diagnosis, indicating that while the PMO representation of the condition should include it as a possible symptom, some specific consultations might have this symptom present while others do not. Also, the guidelines indicate that the physician should check the eardrum; however, Otitis Externa is not associated with any observations regarding the eardrum, suggesting that any observation of the eardrum (e.g., rapture) might indicate a different diagnosis. As for the treatment, the guideline recommends that the physician should instruct the patient on how to clean the infected ear properly, and prescribe ear drops. In addition, referral to a specialist is recommended if the condition does not improve promptly or if the patient is from a specific vulnerable group (e.g., elderly and diabetic).

As an example, we consider a fictitious consultation for an Otitis Externa patient suffering from ear pain (no indication of itching, fluid drainage, or hearing loss), and the examination shows swelling, redness, and skin flaking in the external ear canal. Figure (6a) represents the general PMO of the Otitis Externa condition with indication of the guideline sections forming it, while figure (6b) is the consultation-specific knowledge graph for the fictitious patient.

The PMO can be represented in triples for machine processing; for example, some of the triples portrayed in figure (6b) are explained below:

- The human anatomy is represented in a hierarchy of anatomical structures connected to their parent structures using isPartOf relationships. For example, <externalAuditoryCanal, isPartOf, ear>.
- The second constituents of the PAO is the assignment of anatomical functions to the anatomical structures performing them as in <ear, hasFunction, hearing>.
- The symptoms and observations triples comprise the PSO and the POO respectively. For example, the following three triples demonstrate a pain symptom: <externalAuditoryCanal, hasSymptom, symp_2>, <symp_2, symptom, earPain>, <symp_2, hasValue, 7/10>.
- And finally, the patient diagnosis and treatments are expressed in triples as in the following examples: <patient, diagnosedWith, OtitisExterna>, <patient, treatedWith, earDrops>.
Applying the procedure outlined in the PDD to the Otitis Externa condition produces the ontology visualized in figure (7) using the WebVOWL web application [10]. For conciseness, the ontology visualized does not contain the top-level classes (anatomy, symptom, observation, diagnosis, and treatment) and the rdfs:subClassOf relationship linking the other concepts to them. For the complete ontology construction process and the resulting ontology, refer to the technical report [3].

The method was applied manually, which required substantial time investment. The full implementation potential of the Care2Report system relies on developing an automatic process to generate ontologies of all medical conditions using guidelines from various countries and in different languages. The ontology development relies on data from two sources: SNOMED CT, which can be directly transformed into an OWL ontology with relatively few steps; and the
text-based medical guidelines that pose the real challenge for knowledge extraction and structuring to be tackled in future research.

7 Conclusion

The paper aimed to introduce the notion of ontological conversation interpretation by creating a method for the systematic creation of medical ontologies from the SNOMED CT terminology and anatomical hierarchies, combined with the medical guidelines of different medical conditions. The resulting PMO provides a formalized notation, and the PDD provides a procedural guide to systematically create ontologies and enrich them by connecting new guidelines to the existing definitions of anatomy, symptoms, observations, and treatments; and adding more if the existing concepts are insufficient to represent the guideline fully. Furthermore, we have applied our approach to the case of Otitis Externa to illustrate its application and feasibility.

Limitations and Future Directions. More research is needed to validate the method on guidelines of more complex and varying conditions. For example, the NHG guideline for “Non-traumatic knee complaints” details a method for differentiating between various similar conditions and thus does not follow the same sections that are typical in most of the NHG guidelines. This may require an evolution of our PMO in order to accommodate these needs. Moreover, some guidelines only point the physician towards the tests and measurements to monitor without detailing the results or values to look for, presumably because the results are hard to detail in the text while the medical professionals
understand them. For example, the guidelines advise performing an electrocardiogram (EKG) test in several cardiovascular conditions without detailing the expected outcomes. This is a challenge as the source information is incomplete; thus, the degree of possible automation is limited. The Care2Report multi-modal input architecture [11] aims to eventually allow the integration of (some of) the measurement data, but an ontology to represent this knowledge still needs to be developed. Automation is another research challenge. We have applied the method manually, as explained in Sec. 6, and the correct and complete automated interpretation of textual medical guidelines is a far-fetched goal [16]. Research is necessary toward the creation of assisted, interactive methods that can support the process of knowledge extraction from medical guidelines. Finally, more research is needed towards automating different (sub)activities to enable the efficient creation of complete ontologies of the human anatomy and medical guidelines from different medical authorities.

This research aims to foster research in a societally-relevant field: increasing the quality of healthcare via semi-automated methods that may relieve medical professionals from their administrative burden. The present paper makes a step in this direction by laying down the formal foundations for the construction of semi-automated systems that support the interpretation of conversations with the use of ontologies.

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