J. Health Inform. *2016 Outubro-Dezembro; 8(4):126-133* **Artigo Original**

Using ontologies to retrieve health evidence information from clinical notes

Uso de ontologias para recuperar informação de evidência em saúde a partir de notas clínicas

El uso de ontologías para recuperar información de evidencia em salud a partir de las notas clínicas

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Keywords: Evidence-Based Medicine; Health Information Systems; Knowledge Bases; Decision Support Techniques

ABSTRACT

Objective: To present a strategy to search evidence databases directly from clinical notes, thus relieving health professionals from performing searches. **Method**: An ontology related to health domain, specifically for the domain of adult asthma, was developed to illustrate how to extract search terms from clinical notes. Another ontology was developed to describe evidence databases. **Results**: Synthetic notes, simulating clinical conditions for patients with respiratory diseases, were used to search information from two evidence databases, PubMed and PEDro. SPARQL queries were automatically generated to connect both ontologies. **Conclusion**: This scenario demonstrated how to search for evidence from electronic health records notes, helping health professionals to receive relevant information while they assist patients.

Descritores: Medicina Baseada em Evidências; Sistemas de Informação em Saúde; Bases de Conhecimento; Técnicas de Apoio para a Decisão

RESUMO

Objetivo: Apresentar uma estratégia para pesquisar bases de evidências a partir de notas clínicas, aliviando os profissionais de saúde da tarefa de elaborar buscas. **Método**: Uma ontologia no domínio da saúde, especificamente sobre asma em adultos, foi desenvolvida para ilustrar como obter informação para a estratégia de busca a partir das notas clínicas. Outra ontologia captura informações sobre as bases de evidências. **Resultados**: Notas sintéticas, simulando condições clínicas de pacientes com doenças respiratórias, foram utilizadas para buscar informação em duas bases de evidências distintas, PubMed e PEDro. Consultas em SPARQL foram automaticamente geradas para conectar as ontologias. **Conclusão**: Este cenário demonstrou a viabilidade de procurar por evidências a partir de registros eletrônicos de saúde, ajudando os profissionais de saúde a obter informações relevantes enquanto atendem pacientes.

Descriptores: Medicina Basada em la Evidencia; Sistemas de Información en Salud; Bases del Conocimiento; Técnicas de Apoyo para la Decisión

RESUMEN

Objetivo: Presentar una estrategia para buscar bases de evidencia directamente de las notas clínicas, creadas en los registros electrónicos de salud, con ontologías para capturar conocimiento relacionado a la salud y a las bases de evidencia. **Método**: Una ontología, que se define para el dominio de la salud de asma del adulto, se utiliza para extraer información relevante de notas clínicas. Otra ontología captura información sobre bases de evidencia. **Resultados**: Notas simulando las condiciones clínicas para pacientes con enfermedades respiratorias se utilizaron para buscar información de dos bases de evidencia, PubMed y Pedro. Consultas SPARQL se generan automáticamente para conectar ambas ontologías. **Conclusión**: En este escenario se ha demostrado la viabilidad de la búsqueda de evidencia desde los registros electrónicos de salud, ayudando a los profesionales de salud para obtener información relevante al reunirse con pacientes.

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Artigo recebido: **09/06/2015**
Aprovado: **06/06/2016**

INTRODUCTION

Healthcare professionals usually have to take decisions related to patients assistance, and Evidence-Based Practice (EBP) recommends that such decisions should be based not only on their experience, but also on the most current information available[1]. However, the volume of published health research makes it impossible for them to become fully informed about their fields of knowledge[2].

Evidence databases, which are bibliographic databases with focus on published evidence, are resources to support EBP. Nevertheless, having access to these databases is not enough. Health professionals have difficulties to formulate appropriate queries, with structured clinical questions about issues related to specific patients, and even to know where to search for evidences[3]. Lack of time and not perceiving information needs are also identified as barriers to the adoption of EBP[4].

Semantic Web technologies promote integration of information currently available in digital format[5]. Among these technologies, ontologies have the most expressive power to represent a field of knowledge. An ontology is an explicit specification of a conceptualization, with the set of objects, concepts, and other entities that are presumed to exist in some domain of interest and the relationships that hold them[6].

This work shows how ontologies can be used to integrate clinical notes, taken by health professionals while assisting patients, with evidence databases. This approach enables to bring evidence to health professionals without demanding from them any additional effort on structuring clinical questions, on knowing each evidence database and corresponding query interface, and without spending extra time performing these searches.

Next section presents related work on the linkage between clinical notes and evidences. Section 3 describes a novel approach to solve this problem using ontologies. Its usage for a specific domain is illustrated in Section 4, presenting ontologies for the domain of asthma and for describing evidence databases. Section 5 presents a scenario demonstrating its operation, followed by discussion and conclusions.

RELATED WORK

Health professionals cannot become fully informed about recent developments in their fields by directly reading the scientific literature. In 2004, a study estimated that professionals from primary care would take 21 hours of daily reading to keep up with the literature indexed in the PubMed bibliographic database only in their field of knowledge[2]. In 2015, the amount of indexed papers doubled with respect to 2004 numbers, to almost 1.2 million new indexed papers.

Although Semantic Web technologies, such as the Resource Description Framework (RDF) and ontologies, have been used to improve results from information retrieval tasks[7], giving health professionals access to the literature is not enough to promote EBP. Evidence databases contain selected information from the literature that may have clinical relevance, and are usually queried using a clinical question using the PICO (problem/population, intervention, comparison, outcome) structure[8]. However, lack of time and not knowing how to correctly structure a clinical question are still barriers that health professionals face in EBP[4], along with the fact that there are several evidence databases where to perform such searches.

Thus, this work focuses on how to perform searches on evidence databases for the health professionals without having them to elaborate the search strategies for each database. Considering that health professionals have to take clinical notes in Electronic Health Records, the proposal is to use these notes as a source to automatically structure a PICO question related to the patient condition.

A literature search was performed on *("electronic health record" OR "electronic medical record" OR EHR OR EMR) AND ("evidence based practice" OR "evidence based medicine" OR EBP OR EBM)* using four bibliographic databases (PubMed, ScienceDirect, ACM Digital Library, and IEEExplore), returning 339 papers. From these, two were excluded for being duplicate; 323 by scanning their titles; and six upon analysis of full text. The ramaining of this section synthesizes the findings of the eight remaining papers.

Having access to scientific information to support clinical decisions is the goal of the Aggregate Data Drug Information System[9]. ADDIS is an evidence-based drug-oriented strategy decision support system, based on the premise that clinical trials are the main source of information for efficacy and safety evaluation of medical treatments. According to these authors, there are no established systems that inform strategic (rather than operational) decisions, such as identifying the best treatment practices based on risk-benefit analyses. They claim that evidence-based decision making systems are difficult to implement due to the effort required to systematically review the literature for relevant studies and to manually extract the data from these studies.

One of the problems in integrating clinical notes to scientific papers is that the terms used to classify scientific papers, taken from the Medical Subject Headings (MeSH) vocabulary, are not the same used in clinical notes. The Medline Button[10] searches for papers in MEDLINE, taking as starting point diagnosis and procedures codes from the International Classification of Diseases (ICD) recorded in Electronic Health Records (EHR). To translate ICD codes to MeSH terms, the authors used the Unified Medical Language System[I] (UMLS), which combines, into an integrated metathesaurus, hundreds of terminologies and classifications. As ICD codes are used, it is necessary to have established diagnosis, thus limiting the access to relevant information during earlier stages in the patient assistance.

Information overloading is a potential problem when working directly with text in clinical notes, since not all text is relevant for searches. Hsu et al. use ontologies to analyse clinical data on HER[11], synthesizing clinical patient data to ease comprehension by health professionals. Another approach is to represent extracted data using graph-based structures, as adopted by Wiesner and Pfeifer to integrate a recommender system to a personal health record system[12]. Their proposal was to map entries to concepts in a health graph data extracted from Wikipedia. Although it was not

[I] http://www.nlm.nih.gov/research/umls/

the objective of these authors to integrate a scientific literature search to EHR, use of ontologies to extract only information relevant for the task to be performed is promising.

A workable integration between EHR and EBM has to be affordable, interoperable, and adaptable. Such requirements are difficult to meet in traditional closed, proprietary, vendor and application specific health care IT models. For this reason, free and open-source software systems are good alternatives to overcome some of these issues. Protégé[13] is a general ontology editing framework, developed by the Stanford Medical Informatics group, with an extensible architecture for creating customized knowledge-based tools. It is also a library that other applications can use to access and display knowledge-bases.

This literature review has shown that there are proposals to integrate clinical data and evidence information through knowledge capture, with concepts organized as ontologies, conceptual graphs, or high-dimensional space of concepts capturing the meaning of texts. There are also systems developed to promote EBP, but without addressing the integration of evidences with EHR using ontologies.

INTEGRATING EHR AND EVIDENCE DATABASES WITH ONTOLOGIES

Considering that health professionals must already register clinical notes while assisting patients, it would be valuable for them to access related evidence. To achieve this goal, the following questions are proposed: (1) How to extract relevant search terms from clinical notes? (2) How to represent an evidence? (3) How are evidence databases organized and searched? (4) How to integrate searches and results from distinct evidence databases?

To answer these questions, knowledge from specific fields and from corresponding evidence databases need to be captured and encoded. Both can be expressed as ontologies, enabling simultaneous searches to be performed in distinct databases.

Figure 1 illustrates this proposal. A Health Knowledge Ontology extracts, from clinical notes taken by health professionals, relevant terms to define a generic query, with a set of terms and their metadata. Knowledge about specific evidence databases is represented in another ontology. Thus, it is possible to create specific queries for each evidence database, as well as to interpret and to integrate their responses, providing a set of Web resources with evidence information to health professionals.

Web resources are described in the Semantic Web by the Resource Description Framework[14], which is a standard to represent machine understandable metadata, providing interoperability and domain independence. An RDF document is a sequence of statements, each representing a small piece of knowledge with a Subject-Predicate-Object structure. The subject (resource) and

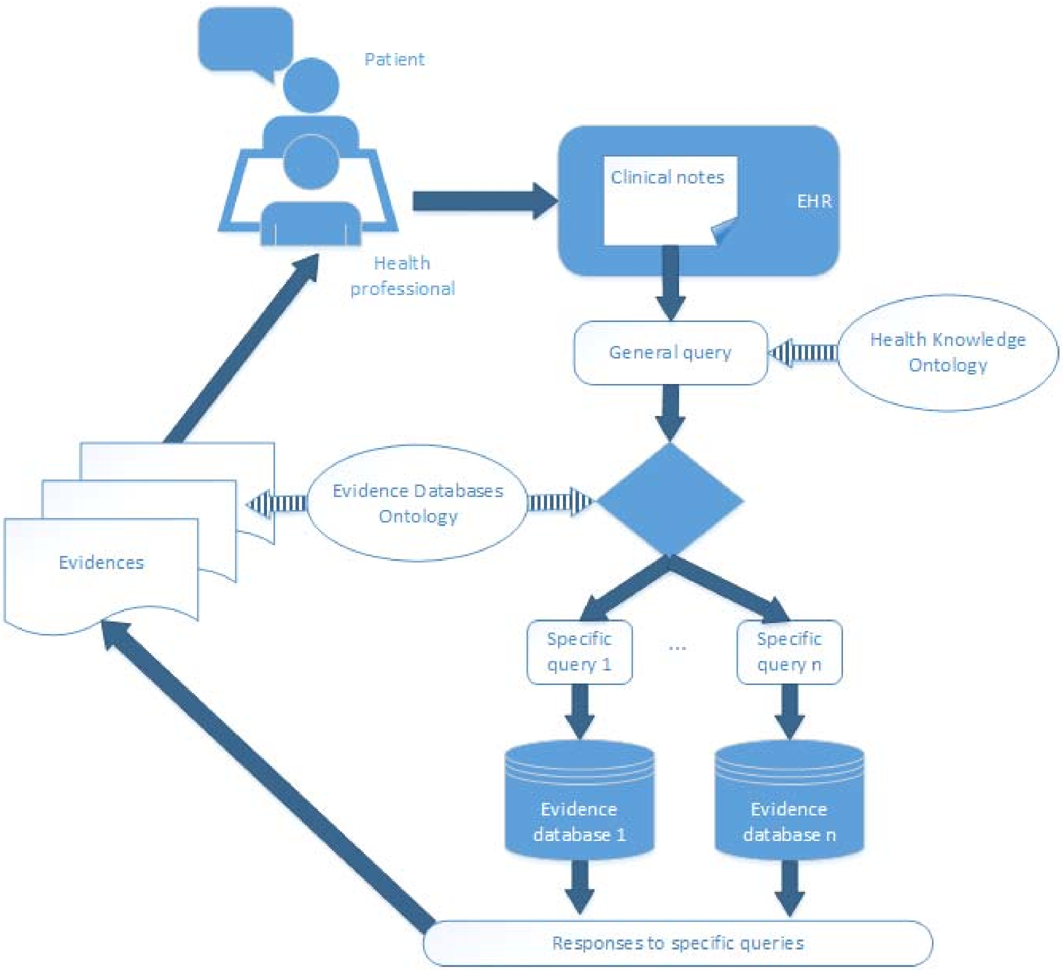

Figure 1 – Overview of the process for integrating clinical notes to evidences, using ontologies to represent health knowledge and knowledge about evidence databases.

object (value) are names for things in the world. The predicate (property) is the name of a relation that connects two things.

In Figure 1, RDF documents represents the information flowing from queries to retrieved evidences. The health professional insert clinical notes on the patient's Electronic Health Record. This insertion triggers a process to verify whether the note contains the elements of a clinical question, as described by the health knowledge ontology. This clinical question is translated to specific queries for each evidence database described in the evidence databases ontology. The resulting evidences are collected and presented to the health professional, while still assisting the patient. Application Programming Interfaces (API), such as the Jena framework[II], process and store information using RDF. RDF data stores are special database systems built to store and retrieve RDF statements. Nevertheless, relational database systems, such as MySQL[III], can be used to store RDF statements.

The two ontologies in Figure 1 are represented using the Web Ontology Language (OWL). OWL is built on top of RDF Schema (RDFS), a common language in which classes, properties, and relations between them are defined for a specific domain. In addition, OWL provides the capability to better express complex relationships. [14]

The SPARQL query language is used to integrate ontologies and RDF documents. SPARQL (recursively defined as the SPARQL Protocol and RDF Query Language) enables to find specific information on the Web of Data, the gigantic RDF database associated with the Semantic Web. A SPARQL engine matches triples contained in graph patterns with RDF graphs. Once a match is found, it will bind variables from a graph pattern to graph nodes, and each variable binding is a query solution.

OBTAINING EVIDENCE FOR ADULT ASTHMA

As ontologies capture and encode information about specific fields of knowledge, an implementation of this approach to integrate clinical notes to evidence information must start by selecting this field. As a proof of concept, the field of adult asthma is selected. This proof of concept requires: clinical notes registered by health professionals; a Health Knowledge Ontology in the field of adult asthma; selection of evidence databases that potentially have relevant information for the field of adult asthma; and an Evidence Databases Ontology that capture knowledge about selected evidence databases.

Two evidence sources containing information related to adult asthma were selected, the Physiotherapy Evidence Database, PEDro[IV], and the Problem, Intervention, Comparison, Outcome (PICO) interface for PubMed[V].

Two ontologies have to be developed. A methodology to create ontologies clearly separate knowledge acquisition process from implementation[15]. Application of this methodology to create the Health Knowledge Ontology on Adult Asthma and the Evidence Database Ontology for PEDro and PICO-PubMed is described next.

Health Knowledge Ontology on Adult Asthma

The first step towards the creation of an ontology is to select its domain and scope. Four basic questions have to be answered: (1) Which is the ontology domain? In this case, physiotherapy respiratory care for untreated and newly diagnosed adult asthma; (2) What is the ontology purpose? This ontology represents and maps concepts related to adult asthma to find them in clinical notes; (3) For what types of questions the ontology should be able to provide answers? This ontology provides clinical evidence related with adult asthma health problems and interventions based on clinical notes about patients; (4) Who will use and maintain the ontology? This ontology is used by physicians and physiotherapists, and it is maintained by this system developers.

Firstly, it is necessary to consider the use of existing ontologies. By reusing ontologies, new systems can communicate with other applications that have already used the same ontologies. However, no adult asthma ontology

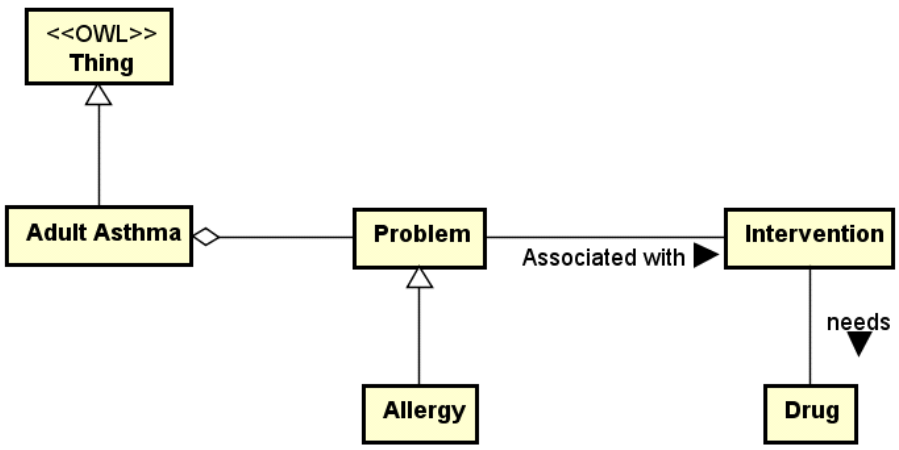

Figura 2 - Fragment of the asthma ontology description, describing the P (problem) and I (intervention) elements of the PICO structure.

[II] https://jena.apache.org
[III] http://www.mysql.com
[IV] http://www.pedro.org.au
[V] http://pubmedhh.nlm.nih.gov/nlmd/pico/piconew.php

Table 1 - List of terms from the adult asthma ontology in their respective classes.

Problem:	Intervention:	Allergy:	Drug:
Changes in temperatures	Future tests	Radioallergosorbent	Salbutamol
Clear watery nasal drainage	Spirometry	Vasomotor rhinitis	RAST
Persistent asthma	Physical examination	Ears	Radioallergosorb
Eye symptoms	Chest x-ray	Eyes	Ipratropium bromide
Watery rhinorrhoea	Allery investigation	Throat	Anticholinergic sprays
Nasal irritation	Allergy skin prick tests	Nose	Antihistamines
Itching	Challenge tests	Allergic rhinitis	INCS
Loss of sense of smell			Inhaled nasal corticosteroids
Congestion			Antihistamines
Nasal polyps			INCS
Occupation			Ipratropium bromide
Environment			Oral corticosteroids
Allergic conditions			Methacholine
Risk factor			Histamine
Purulent nasal drainage			Mannitol
Bacterial infection			Hypertonic saline
Risk factor			Dry air

was found.

To find out relevant terms for the ontology, a simple list of terms from the domain is enumerated. In this case: asthma, respiratory physiotherapy, spirometry, chest x-ray, allergy tests, future tests, allergic rhinitis, risk factor, nose, throat, eyes, ears, nasal irritation, clear watery nasal drainage, nasal drainage, eye symptoms, purulent nasal drainage, bacterial infection, nasal polyps, histamine, mannitol, dry air, vasomir rhinitis, bacterial infection, loss of sense of smell, purulent nasal drainage, changes in temperatures, allergy investigation, radioallergosorbent, antihistamines, watery rhinorrhoea allergy investigation, allergic conditions, environment, occupation, anticholinergic sprays, oral corticosteroids, symptoms, skin prick tests, regular preventer, challenge tests, itching, allergy, congestion.

The next step is to define classes and their hierarchy. The general classes for the asthma ontology are Problem, Intervention, Allergy, and Drug (Figure 2). Related terms, defining specialized classes, are associated to these general classes (Table 1).

It is also necessary to define the properties (slots) for each class, such as problemName and testName for the class Problem.

The final step is to add constraints (facets) to the properties, such as cardinality, value type, domain, and range. In this case, String was defined as the range for all properties (slots). Object properties describe relationships between instances, and data properties describe relationships between instances and individuals. For the asthma ontology, examples of object properties are: avoid, co-exist, consider, develop, diagnosis, focus, lessEfective, recommend, and require. Examples of data properties are: allergyName, allergyTestName, daytimeSymptom, drugType, interventionName, measuring-Breath, symptomDescription, and testName.

Evidence Databases Ontology

The same methodology, applied to create an ontology for evidence databases, gives as domain clinical evidences from PEDro and PubMed PICO databases; as purpose, to retrieve evidence information as statements stored in RDF documents; as types of question, evidence information related to adult asthma procedures and interventions; and as users, physicians and physiotherapists, being maintained by system developers.

As for existing ontologies, properties from the Dublin Core Schema[VI] are incorporated to describe Web resources. DC is a small set of vocabulary terms used to

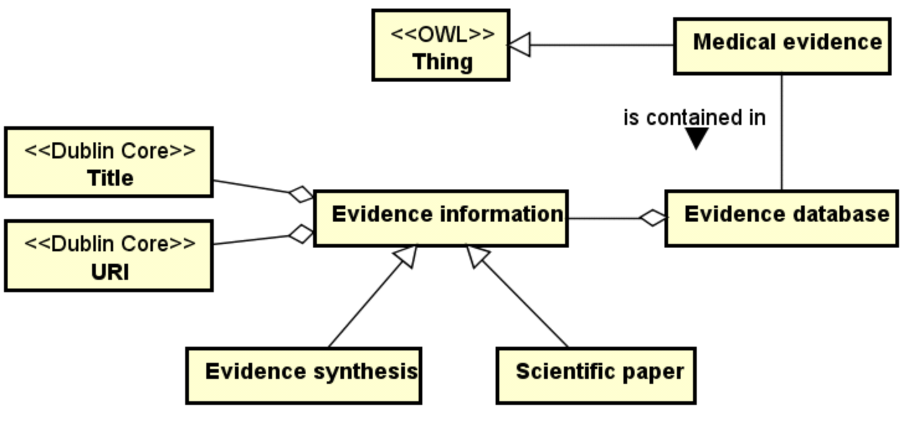

Figure 3 - Fragment of the evidence databases ontology description, relating the evidence content with Dublin Core metadata.

[VI] All the DC terms are accessed through its namespace IRI, at http://purl.org/dc/terms describe web and physical resources. In the DC namespace, each term has a Unique Resource Identifier and is defined using RDF properties.

Relevant terms in this ontology are: evidence, database, query, title, author, URL, journal, DOI, evidence type, database type, year, PubMed, PEDro, term, identifier, abstract. The general classes for the evidence database ontology (Figure 3) are Database, Evidence, and Journal.

As for the adult asthma ontology, String is the range for all properties. Examples of object properties are belong and link, and of data properties are queryString, evidenceURL, journalName, and meshTerm. Additionally, some attributes from the DC Schema are integrated in this ontology, such as: abstract, identifier, title, type, creator, description, contributor, date, and subject.

RESULTS

From the knowledge architecture viewpoint, this proposal required an implementation in three layers: knowledge domain creation; knowledge capture and creation; and knowledge retrieval (Figure 4). Protégé was used to model and to generate ontologies for asthma and evidence databases, and Jena framework was used to create the corresponding RDF statements. A MySQL relational database was used to store facts and statements.

Knowledge capture and creation

Knowledge bases were created by modelling RDF documents based on ontologies specifications. For the adult asthma ontology, concepts and their relations were initially modelled using a concept map (Figure 5), which was validated by a domain specialist.

Knowledge retrieval

This layer used SPARQL queries to search and retrieve evidence information. RDF statements were produced by extracting relevant terms from clinical notes, using knowledge stored on the adult asthma ontology. With these statements and knowledge from the evidence database ontology, each evidence database was queried and corresponding results were also stored as RDF statements. Finally, results were translated to a format suitable for user presentation.

These were the steps of text processing and information retrieval algorithm:

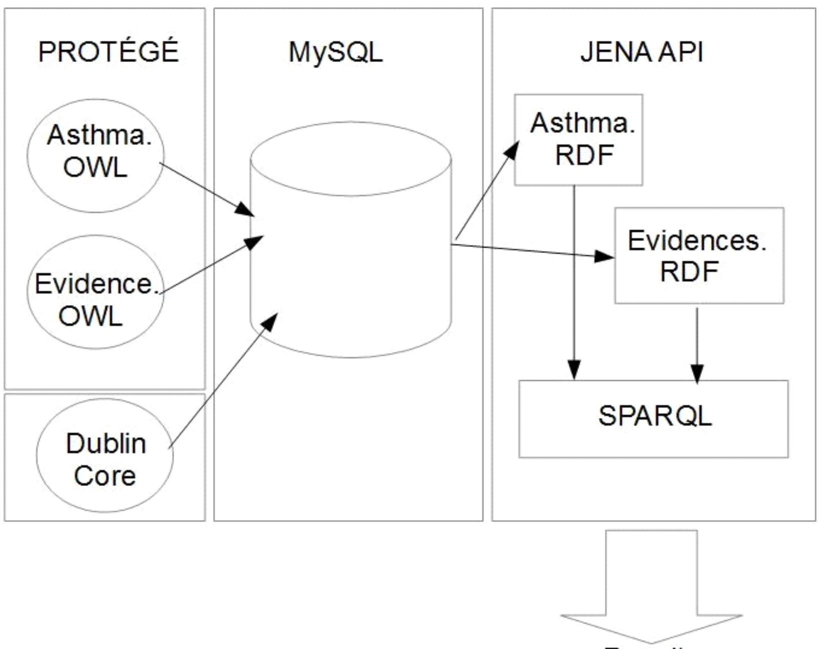

Figure 4 - Proof of concept architecture. A relational database (MySQL) was used to keep information from OWL ontologies and to extract RDF statements which were queried using the SPARQL query language.

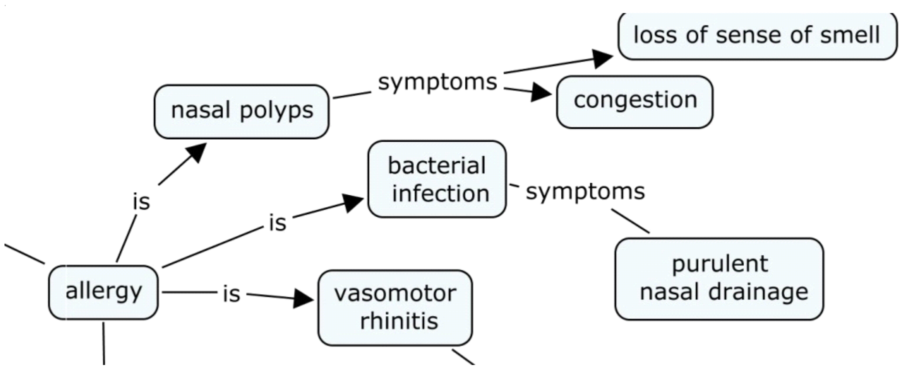

Figure 5 - Partial view of the asthma concept map.

1. Tokenize all text from the clinical note, removing characters ',' or '.' or ';';
2. Remove small tokens (with length less than three) from the list of tokens;
3. With the RDF document generated from the adult asthma ontology specification, use SPARQL queries to find concepts related with each class of the ontology;
4. Match the input tokens resulting from Step 2 with concepts resulting from Step 3;
5. With the RDF document generated from the evidence databases ontology specification, use SPARQL and matched tokens to query each evidence database;
6. Merge the results from each evidence database into a single RDF document.

In this proof of concept, two general classes from the asthma ontology were used to create queries for the PICO-PubMed database, since these queries could be structured with search terms for problem and search terms for interventions. As for the PEDro database, all these terms were combined into a single query string, as this distinction between problem and intervention was not supported.

Example of application

Consider the example of a clinical note: *Patient CC, 30 years old, says it has been five weeks since he starts to sneeze in the nights. After checking his nose, we can see little nasal polyps in the right hole; also we have noticed that the nose is irritated. The patient states he has stuffy nose daily lately and also he has noticed a loss of sense of smell. We believe that he has developed allergy and asthma.*

After tokenizing the text, the first task was to find relevant concepts in the adult asthma RDF document using SPARQL queries. After running the text processing and information retrieval algorithm, concepts found for the *Problem* class were: Nasal irritation; Allergic rhinitis; Changes in temperatures; Watery rhinorrhoea; Classical symptoms; Vasomotor rhinitis; Eye symptoms; Nasal polyps; Clear watery nasal drainage; Bacterial infection; Itching; Loss of sense of smell; Congestion; and Purulent nasal drainage. For the *Intervention* class, the concepts were: Spirometry; Ipratropium bromide; Challenge tests; Future tests; Antihistamines; Allergy tests; Chest x-ray; Anticholinergic sprays; INCS; Radioallergosorb; Drug Intervention; Salbutamol; and Inhaled nasal corticosteroids. Table 2 summarizes the amount of results obtained from this task.

Table 2 - Summary of the result from text processing and information retrieval algorithm

Tokenize	Reduce	Problem	Intervention
75	42	14	14
Match-P	Match-I	Merge	Query
5	2	7	60

The next task was to match these concepts from *Problem* and *Intervention* classes with the useful tokens from the patient information text. In this case, matches related with *Problem* were: Nasal irritation; Clear watery nasal drain-age; Nasal polyps; Loss of sense of smell; Purulent nasal drainage. Matches related with *Intervention* were Allergy tests and Inhaled nasal corticosteroids. These concepts were matched with the useful tokens, and the concepts were retrieved using SPARQL queries in the RDF representation of the evidence databases ontology.

Finally, the algorithm retrieved the information to be presented to the health professional who has inserted the clinical note. In this case, 60 links with evidence information related to that specific patient were presented.

DISCUSSION

Three main results were presented in this paper. The first was to show how to create OWL ontologies to capture health knowledge (in this case, about adult asthma) and knowledge about evidence databases. The second was the generation of RDF documents that are knowledge repositories based on the specifications of these ontologies. Finally, the third result was a framework along with a procedure to perform information retrieval with the PICO structure using these ontologies and knowledge repositories. The asthma OWL ontology was only an example of how to structure the knowledge from one field to extract the PICO elements. Based on this example, ontologies for other fields can be similarly built. The information on these ontologies can be derived from existing structured knowledge, as the SNOMED CT terminology[VII] or the UMLS metathesaurus[VIII].

The presented implementation can be improved in several ways. One current limitation is that all RDF statements are stored into a single database, which may affect scalability; creating one table per model (domain) could overcome this limitation. Relevance measures should be considered for the retrieved resources, taking in account how specific is the information for the patient (for example, by considering the amount of matched terms). By using a standard terminology (such as SNOMED CT or UMLS), this application may easily be extended to consider multilingual resources, as each concept receives the same identifier, regardless of the language or synonym used to express it. However, it should be noted that Portuguese is currently not among the languages supported by SNOMED CT, while UMLS has few classification codes in Portuguese.

CONCLUSIONS

This paper addressed the use of ontologies to automatically retrieve evidence information for health professionals while they insert clinical notes in electronic health records. A proof of concept in the field of adult asthma was presented to illustrate the proposal, using ontologies to extract the required elements for a PICO clinical question from clinical notes. One of main advantages of using ontologies is the possibility of creating flexible models, capable of integrating different domains and heterogeneous sources. Another observed benefit in this ontology-based solution was the effectiveness of finding relevant concepts in a specific domain through queries using Semantic Web tools, according to a health professional that verified the results retrieved for the

[VII] http://www.ihtsdo.org/snomed-ct
[VIII] https://www.nlm.nih.gov/research/umls/

simulated clinical notes.

This work has shown that it is possible to take advantage of semantic richness from ontologies to find relevant information from a patient clinical note and to use this information to query heterogeneous evidence databases. By integrating these results in a transparent and uniform way, the Semantic Web technologies enabled promotion of Evidence-Based Practice among users of Electronic Health Records.

Use of Semantic Web technologies brings the potential to integrate evidence resources with other Web resources, in the spirit of Linked Data. By using Dublin Core to describe evidence information resources, this framework can potentially become part of the Linked Data cloud. Linked Data enables a collection of machine-understandable statements to be published without relation to any website at all, and is one of the main applications in the Semantic Web.

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