Implementing Thyroid Ontology for Diagnosis and Care

Özgü Can, Emine Sezer, Murat Osman Ünalır, Okan Bursa, Mine Öz and Tuğçe Dülge
Department of Computer Engineering, Ege University, 35100 Bornova, Izmir, Turkey
e-mail:{ozgu.can, emine.sezer, murat.osman.unalir, okan.bursa }@ege.edu.tr,{mineoz621, dulgeetugce}@gmail.com

Abstract— **Healthcare systems are changing their information systems with the increased advances in information technologies. With these improvements, patients expectations are also increased. Consequently, patients expect a quick diagnosis process and improvement in the treatment process. Additionally, it is important to share and reuse patients' information between different healthcare organizations and healthcare workers. There are lots of data standards and ontologies developed in the health domain. In this work, we propose a thyroid ontology for the diagnosis and care of the thyroid-related diseases. The proposed thyroid ontology will be a part of the Semantic Web based health information system in order to provide interoperability and reuse of health data. The proposed thyroid ontology can be used for treatment suggestion systems. Therefore, the healthcare decision support system will be improved.**

Keywords-Thyroid Ontology; Healthcare Information Systems; Semantic Web; Ontology Development.

I. INTRODUCTION

The continuous advancements in healthcare information technologies has lead to better opportunities for patients and also for healthcare workers. As healthcare is a consumer-oriented domain, there are various challenges in the healthcare domain. One of these challenges is to provide a better diagnosis and care to patients.

The thyroid gland is one of the most important organs that controls the metabolism of the body. The function of the thyroid gland, which assumes a vital function in the body, may deteriorate due to factors, such as genetics radiation, iodine deficiency and aging. As a result of this, diseases such as goiter, thyroiditis, hyperthyroidism, hypothyroidism, nodule, thyroid cancer can occur. Tests for these diseases are hormone and antibody tests. Hormone tests are based on the T3, T4 and TSH (Thyroid-Stimulating Hormone) values that are secreted by the thyroid. In antibody tests, Anti-TPO (Anti-Thyroidperoxidase) and Anti-TG (Anti-Thyroglobulin) values are more frequently observed.

In this paper, we propose a thyroid ontology. The proposed thyroid ontology in this study has more than one test that refers to these values. Our ontology can identify the disease according to these reference values. In contrast to the ontologies presented in [1] [2] that are based on expert systems, the goal of this ontology is to develop a knowledge base for thyroid disease, to query and to reuse the stored information of patients' test results for diagnosis and care. Therefore, it is possible to integrate the proposed thyroid ontology with other healthcare information systems in order to create an interoperable treatment system [3].

The paper is organized as follows: Section 2 presents the lifecycle of the thyroid ontology and explains the development of the thyroid ontology. Section 3 presents rule examples for the thyroid ontology. Finally, Section 4 concludes and outlines the direction of the future work.

II. THE LIFECYCLE OF THYROID ONTOLOGY

In order to develop the thyroid ontology, we followed the ontology development steps that are presented in [4]. According to this guide, the first step is determining the domain and scope of the ontology. Therefore, we defined the domain and scope of the thyroid ontology by answering some basic questions, such as: "What is the domain of the ontology?", "Where do we use the ontology?", "Who will use the ontology?", etc. The answers to these questions lead us to develop the ontology. In our thyroid ontology, we describe the medical tests that a physician requests in order to diagnose thyroid-related diseases and also to follow her patient's medical situation after the diagnosis. For the diagnosis of thyroid diseases, blood tests, thyroid ultrasound, thyroid scintigraphy and thyroid fine needle aspiration biopsy are used. The scope of the proposed thyroid ontology is only for blood tests. The patient is diagnosed based on the reference values according to the values of the blood test result. In addition to the test results and diagnostics, the laboratories where these tests are performed, the tasks and the patient information are defined in the thyroid ontology.

The second step in the ontology lifecycle is to define the competency questions that the thyroid ontology should be able to answer. While creating these questions, we focused on questions about laboratory results. Some questions are listed below:

- Does the gender and age affect thyroid tests?
- What are thyroid gland diseases?
- Which hormones should be analyzed for the diagnosis of the disease?
- Which hormone tests should be performed to determine disease types?
- How can blood tests be interpreted?
- What is the relationship between TSH, T3 and T4 hormones?
- Are antibodies and hormones the same thing?
- What are the conditions in which the thyroid antibody is used to diagnose the disease?
- Which anti-TPO and anti-thyroglobulin antibodies show hyperthyroidism?
- Is hyperthyroidism the same disease as goiter?
- Is diffuse hyperplasia a goiter disorder or a thyroid disease?

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- Which hormone level should be considered in order to keep track of the primary hyperthyroidism?

After defining the competency questions, we identified the important terms in the thyroid ontology. For this purpose, we created a comprehensive list of terms without considering whether there is a relationship between concepts or not. After completing this list, we performed a distinction between classes and instances that are necessary for the next step. Then, we used the most common methods for developing the class hierarchy, such as "top-down" and "bottom-up". We described the basic class and subclass concepts of the thyroid ontology as seen in Figure 1.

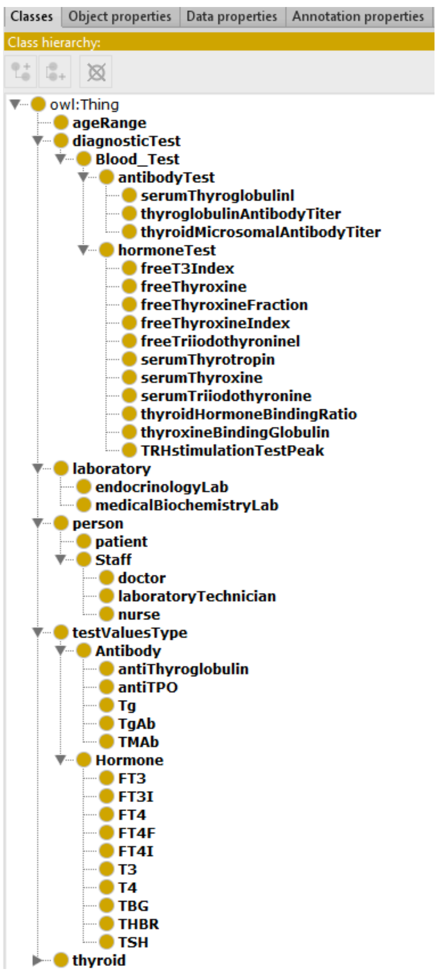

Figure 1. The basic concepts of the thyroid ontology.

The thyroid ontology includes the diagnostic test in order to make diagnoses. For this purpose, we created the `diagnosticTest` class. There are different tests for the diagnosis of each disease, so after establishing the disease hierarchy we created a separate class hierarchy for these tests. Many test methods are used for the diagnosis of the disease. But, in the proposed thyroid ontology we only included blood tests. Blood tests are divided into two subgroups: antibody tests and hormone tests. As seen in Figure 1, antibody and hormone tests are divided into subclasses. Each subclass diagnoses the disease by looking at the values of different hormone or antibody types. A separate class hierarchy has been established for the types of antibodies and hormones that these tests use in diagnostics. The thyroid ontology also includes information on where these tests are performed (`laboratory`), who owns them (`patient`), and who is responsible for the tests (`staff`). Figure 2 shows the subclasses of thyroid disease. Thyroid disease varies according to persons' age. For this reason, we grouped the patients according to their age (`ageRange`) in order to determine at what age range the likelihood of being sick is greater. Patients within the `ageRange` class are kept by age groups. Thus, we can easily reach the age range of the diagnosed patients, and also get a detailed information like the most common disease in the adult age groups.

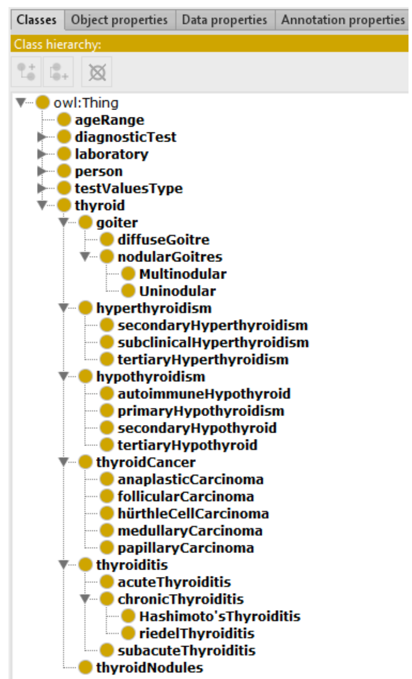

Figure 2. The subclasses of the thyroid class.

After completing the definition of the class hierarchies, we continued to develop the thyroid ontology by specifying object properties in order to represent relationships between classes. The object properties of the thyroid ontology are given in Figure 3. The definition of these properties are listed below:

- `defines`: Represents the relationship between the `diagnosticTest` class and `thyroid` class. So, the diagnosis of the test results will be easily accessed.
- `hasAgeRange`: Represents the relationship between `patient` class and the `ageRange` class. So, it will be possible to determine the age range of diseases that are more common. By using this

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relationship, patients within the `ageRange` class are kept according to their age groups.

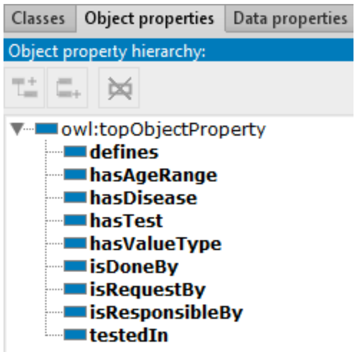

Figure 3. The object properties of the thyroid ontology.

- `hasDisease`: Represents the relationship between `patient` class and `thyroid` class.
- `hasTest`: Represents the relationship between `patient` class and `diagnosticTest` class. So, patients' test information is easily accessed through this relationship.
- `hasValueType`: Represents the relationship between `Blood_Test` class and `testValuesType` class. So, it is easy to learn which hormone value or antibody value are checked in a test.
- isDoneBy: Represents the relationship between `Blood_Test` class and `testValuesType` class.
- `isDoneBy`: Represents the relationship between `diagnosticTest` class and `laboratoryTechnician` class. The goal is to easily reach the knowledge of which technician made the test.
- `isRequestBy`: Represents the relationship between `diagnosticTest` class and `doctor` class. By using this relationship, the doctor who requested the patient's tests could be obtained.
- `isResponsibleBy`: Represents the relationship between `diagnosticTest` class and `nurse` class. The aim is to access the information of the nurse in charge of the patient during the testing process.
- `testedIn`: Represents the relationship between `diagnosticTest` class and `laboratory` class. By using this relationship, the information about the laboratory in which the tests are performed could be reached.

After completing the creation of object properties, we continued to develop the thyroid ontology by specifying data properties. Figure 4 shows data properties of the thyroid ontology.

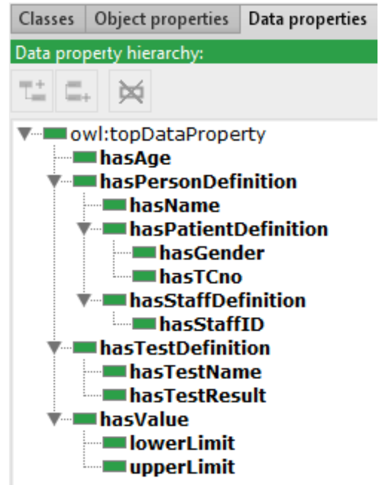

Figure 4. The data properties of the thyroid ontology.

The definition of these properties are listed below:

- `hasAge`: Represents the age information of the patient.
- `hasPersonDefinition`: Represents the information about the person.
- `hasName`: Represents the name information of the person.
- `hasPatientDefinition`: Represents the information about the patient.
- `hasGender`: Represents the gender of the person.
- `hasTCno`: Represents the social security number of the person.
- `hasStaffDefinition`: Represents the information about the staff.
- `hasStaffID`: Represents the identity number about the staff.
- `hasTestDefinition`: Represents the information about the test.
- `hasTestName`: Represents the name of the test.
- `hasTestResult`: Represents the result of the test which is used to diagnose the patient by comparing it with the reference values of the patient.
- `hasValue`: Represents the range values of the hormone and antibody types used in the test.
- `lowerLimit`: Represents the lower limit of the hormone and antibody types used in the test.
- `upperLimit`: Represents the upper limit of the hormone and antibody types used in the test.

In Figure 5, all of the relationships between classes of the proposed thyroid ontology are shown.

Finally, we created individuals of the given concepts of the thyroid ontology. The individuals of the thyroid ontology can be seen in Figure 6.

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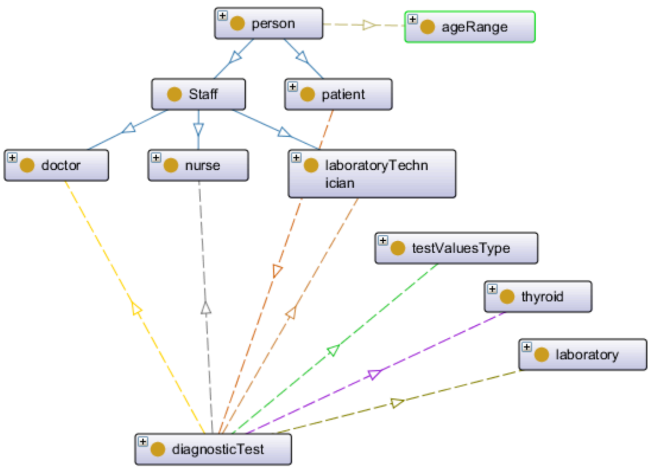

Figure 5. The relationship between the classes of the thyroid ontology.

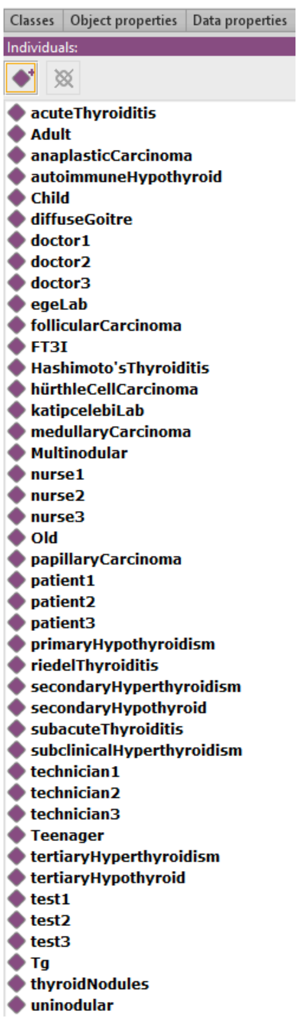

Figure 6. Individuals of the thyroid ontology.

III. IMPLEMENTING RULES

After creating the individuals of the proposed thyroid ontology, we wrote Semantic Web Rule Language (SWRL) [5] rules. Rules allow the ontology to reason about itself [6]. The rules are given in Figure 7.

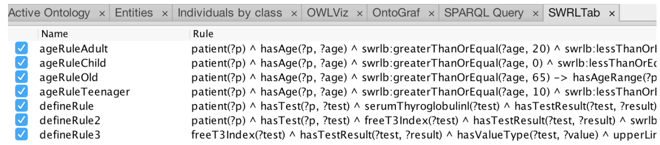

Figure 7. SWRL rules for the thyroid ontology.

The `ageRules` given in Figure 7 automatically set the `hasAgeRange` relationship between the patient and the "child", "teenager", "adult", or "old" instances by comparing the value with the age range when the patient `hasAge` data property value is entered. Therefore, there is no need to enter the age range for the patient.

Figure 8 shows the rule that is written for the diagnosis of the disease. This rule establishes the "defines" relationship between the test and the disease itself, and compares the result of the test with the min. or max. limit of the hormone or antibody that is considered as the reference value. Consequently, according to the result of the test, the disease is diagnosed.

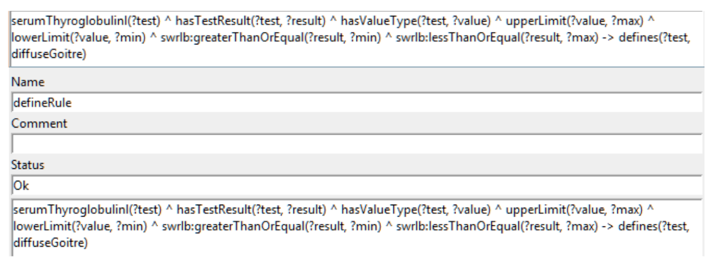

Figure 8. Rule for the diagnosis.

The thyroid ontology has $\mathcal{ALCRF}(\mathcal{D})$ expressivity. Before we run the rules in the proposed thyroid ontology, the axiom number was 472, after we execute rules this number has increased to 661. The related metrics are shown in Figure 9 and Figure 10, respectively. So, this shows the ontology works properly and increases axioms. We also wrote Simple Protocol and RDF Query Language (SPARQL) [7] queries and saw that these rules work correctly. The thyroid ontology is still being developed and extended with new concepts, object and data properties.

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Ontology metrics:

Metrics	
Axiom	472
Logical axiom count	270
Declaration axioms count	143
Class count	74
Object property count	9
Data property count	14
Individual count	43
Annotation Property count	5
DL expressivity	ALCRF(D)
Class axioms	
SubClassOf	68
EquivalentClasses	0
DisjointClasses	15
GCI count	0
Hidden GCI Count	0

Figure 9. The thyroid ontology metrics before executing rules.

Ontology metrics:

Metrics	
Axiom	661
Logical axiom count	478
Declaration axioms count	143
Class count	74
Object property count	9
Data property count	14
Individual count	44
Annotation Property count	5
DL expressivity	ALCRF(D)
Class axioms	
SubClassOf	141
EquivalentClasses	0
DisjointClasses	15
GCI count	0
Hidden GCI Count	0

Figure 10. The thyroid ontology metrics after executing rules.

IV. Conclusion And Future Work

In this work, we proposed a thyroid ontology that will be a part of the Semantic Web-based Health Information System. The aim of this work is to provide interoperability and to reuse health data. Therefore, the healthcare decision support system will be improved. The thyroid ontology is still being developed, and will be extended with new concepts. As a future work, we will integrate Friend of a Friend (FOAF) ontology to describe personal information [8]. Also, privacy concepts will be added to the proposed ontology in order to ensure patient privacy.

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