FUDMA Journal of Sciences (FJS)
ISSN online: 2616-1370
ISSN print: 2645 - 2944
Vol. 3 No. 3, September, 2019, pp 503 –508

A SEMANTIC WEB BASED APPROACH FOR DIAGNOSING RELATED HORMONE IMBALANCES

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ABSTRACT

Hormones are chemical messengers of the body that regulate bodily functions. Symptoms like depression, anxiety, and fatigue are caused by hormones going out of balance. In this study, we built a system that diagnoses three hormone imbalances (Testosterone, Thyroid Hormone, and Cortisol Hormone). The system makes use of a knowledge base built using the Web Ontology Language (OWL) and it interacts with the knowledge base using Jena API because it provides an ontology API for connecting to the ontology, generic reasoner serving as the inference engine, and SPARQL implementation for performing statistical queries. The benefits of this approach over an existing expert based system are improved ontology model by 71% in terms of knowledge representation and 58% in terms of taxonomy, ability to share patient data by moving them from MySQL database into the ontology, and ability to diagnose three related hormone imbalances

Keywords: Ontology; Semantic Web; Hormone Imbalance; Expert System

INTRODUCTION

Hormones are molecules released by glands into the blood to regulate human physiology (internal functions) and behaviour. They regulate bodily functions like blood pressure, hunger cravings, mood swings, immune responses, sexual behaviour, and so on (Marieb, 1992). The glands may produce excess or not enough hormones resulting in an imbalance. This imbalance results in different symptoms depending on the particular hormone.

E-health is the use of information and communication technologies to provide and or improve healthcare (Oh et. al 2005). E-health systems have been used in place of screening questionnaires, and have shown positive results (Woo et. al 2016). Current e-health systems have found applications in managing conditions like hormonal imbalances at home (Paganelli & Giuli 2013).

Rawte and Roy (2015) developed an ontology based system that diagnoses thyroid disease (both hyperthyroidism and hypothyroidism). Their system made use of a database in storing patient information. The system proposed in this research improves the ontology design presented in Rawte and Roy (2015), adds the ability to diagnose testosterone deficiency and cortisol imbalance to their system, and performs statistical analysis on the data collected from the patients. The system also moved the storage of patient data from the MySQL database to the ontology allowing sharing of patient data with other systems. The ontology of the proposed system was defined using the Web Ontology Language (OWL) and Protégé (Musen, 2015) as the ontology editor. The inference rules were defined using the Apache Jena reasoner that comes bundled with the Jena API. JavaFX was used in designing the user interface and in displaying the results of the statistical analysis. SPARQL was used in defining the queries for performing the statistical analysis. The evaluation was done based on the metrics by Manouselis*et al*. (2010).

Literature Review

Oh et al. (2005) defined e-health as the use of information and communication technologies to provide and or improve healthcare. E-Health is a new field of medical informatics that is finding applications in many aspects of medicine. It has been used in managing patient (Garets & Davis, 2006). It has also been used in remote diagnosis and treatment of patients by means of telecommunications technology (Silber, 2003).

Catwell and Sheikh (2009) reviewed the benefits and drawbacks of e-health systems and found: E-health systems are convenient and have the potential of cutting costs when it comes to accessing healthcare remotely, but the initial cost of setting up may be prohibitively high. They found that e-health systems have the potential of reducing the number of people affected by medical errors. They also warn of the possibility of a badly engineered system providing incorrect diagnosis. They found that e-health systems have the potential of providing equality of service: every person regardless of means will have access to the same system.

Hormones may be defined as chemical substances, secreted by cells into the extracellular fluids that regulate the metabolic functions of other cells in the body (Marieb, 1992).

Cortisol is the hormone that is released under stress or when the blood sugar level goes below normal (Smith, 2005). Smith (2005) studied the symptoms of cortisol imbalance and found the following: Difficulty losing weight, weak bone, weak muscles, and thin skin (easy to get bruising, and dry skin) among others.

Thyroid hormones are group of hormones produced in the thyroid gland that act on almost all cells of the body and are chiefly concerned with metabolism. Smith (2005) also reviewed the symptoms of thyroid hormone imbalance and reported the following: fatigue, constipation, depression, and weight gain.

Testosterone is an important hormone necessary for proper development. The symptoms of low testosterone reported by Dandana and Rosenberg (2010) include: reduced libido, erectile dysfunction resistant to medication, reduced muscle mass and strength, depressed mood, lack of energy and reduced vitality, and osteoporosis.

The ontology is defined as a body of knowledge describing some domain, typically a common sense knowledge domain, using a representation vocabulary (Chandrasekaran et al, 1999). Ontological analysis clarifies the structure of knowledge. Given a domain, its ontology forms the heart of any system of knowledge representation for that domain. Without ontologies, or the conceptualizations that underlie knowledge, there cannot be a vocabulary for representing knowledge. Thus, the first step in devising an effective knowledge representation system, and vocabulary, is to perform an effective ontological analysis of the field, or domain. Weak analyses lead to incoherent knowledge bases (Chandrasekaran et al, 1999).

Ontologies together with inference engines form a knowledge base. It is this knowledge base that is used in diagnosis. The ontology holds the specification of the disease, while the inference engine reasons on data received from the user through an interface, and suggest a diagnosis.

Rawte and Roy (2015) used an ontology in the diagnosis of thyroid disease (hyperthyroidism and hypothyroidism). They designed the ontology using the Web Ontology Language (OWL). They used the Semantic Web Rule Language for defining the inference rules, and Jena API for interfacing between the system and the knowledge base. They stored patient information in a MySQL database with the front-end web interface designed with JSP.

Chang *et al*. (2014) developed an ontology that was used in the diagnosis of depression. They used the Description Logics variant of the Web Ontology Language (OWL) in developing the ontology, and GeNIe& SMILE were used as reasoning engines for making inferences on the ontology. The system worked by fetching the patient's personal ontology from the cloud. It then presents a user interface for getting data that will be used to infer whether the patient is suffering from depression. They also used a Bayesian Network as a tool for computing the probability of a user having a disease from the user's description.

Bucci et al. (2011) found that applying ontologies in medicine improved interoperability between systems, machine-readability, and knowledge reuse. They also found that ontologies can define a knowledge base that evolves over time by allowing qualified users to contribute to the knowledge base without the intervention of a system developer. They used an ontology in the diagnosis of flu. They also made use of a Bayesian Network in computing uncertainty. They proposed an extension to the Web Ontology Language called PR-OWL. The new language extends OWL in both syntax and semantics to make it possible to compute uncertainties in the ontological model. They made use of RacerPro and pellet as reasoners.

MATERIALS AND METHODS

The system is made up of several modules as shown in Fig. 1: the ontology, the inference engine, query engine, and the user interface. The user will interact with the system through the interface by selecting the appropriate symptoms, and pressing a button for processing. The system then connects to the ontology that is loaded from disk through the Apache Jena Application Programming Interface (API) and sends the selected symptoms to the inference engine for reasoning. The inference engine returns its result indicating whether the user may or may not have a condition. The Jena API provides the API for connecting to ontologies and also provides a built in inference engine. The Jena API also provides an API for querying the ontology in order to perform the statistical analysis.

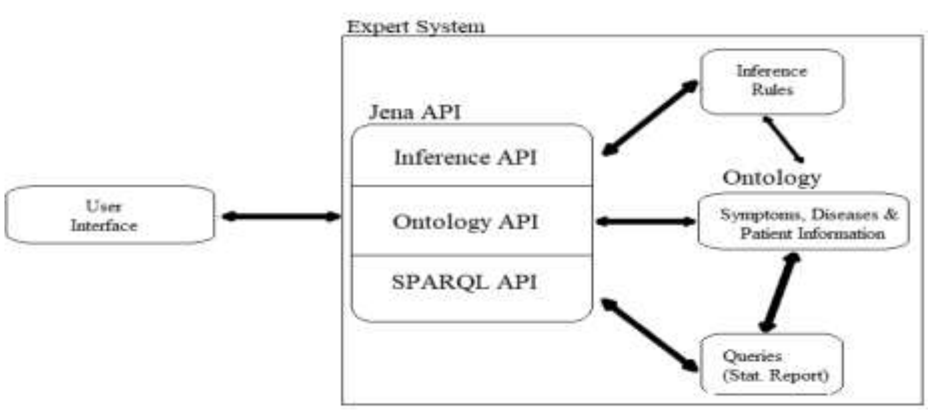

Fig. 1: System Architecture

Building the Ontology

Every ontology starts off with 'Thing' at the top of the class hierarchy. Thing is a class that is the set of all individuals in the ontology. Two classes are defined as subclasses of the Thing class: Person, and Symptom. An individual of type Person will be created during system initialization to hold the information of the current user of the user. The information that will be stored will be age, gender, symptoms, and possible hormone imbalances. A Symptom class is defined to be the set of all symptoms in the system. The ontology model is shown in Fig. 2.

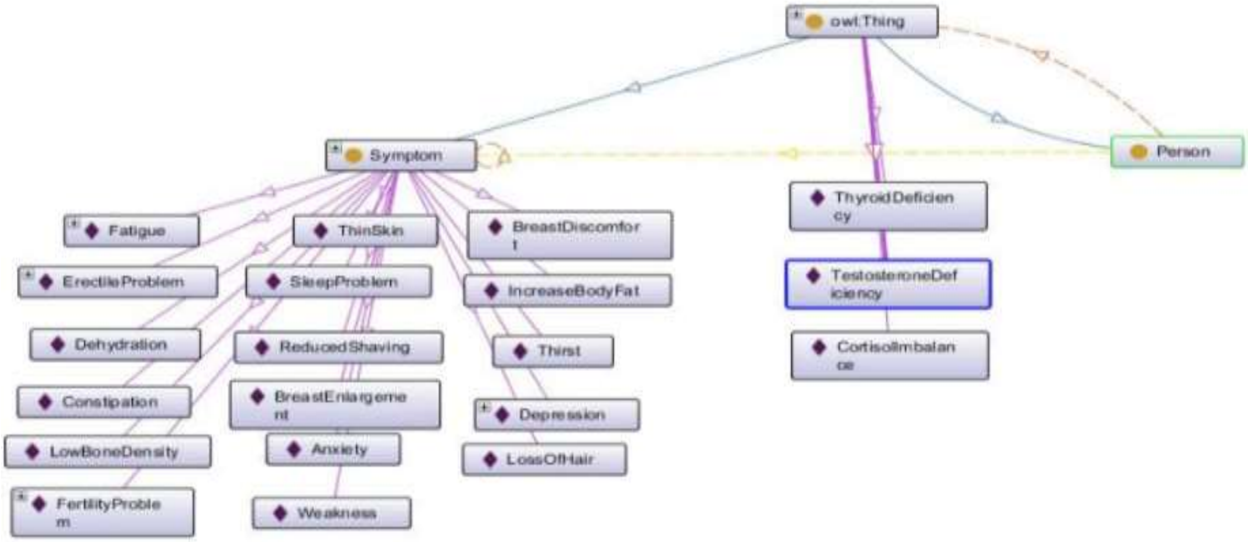

Fig. 2: Hormone Imbalance and patient ontology model

Two relationships are defined as object properties as shown in Fig. 3. The first one is called 'hasThis'. This property relates an individual of type Person with an individual of type Symptom and is used to indicate whether the user has a particular symptom. The second one is called 'mayHave'. This property relates an individual of type Person with an individual of type Disease and is used to indicate the possibility of the user having a particular imbalance. Two data properties are defined to hold the age and gender of the patients.

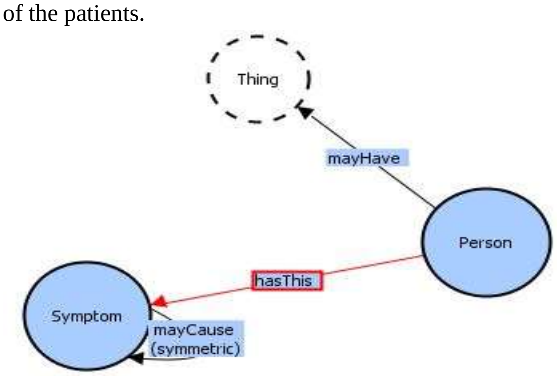

Fig. 3: Object properties in the ontology model

Inference Rules

The ontology serves as an explicit representation of the symptoms and diseases and the inference rules together with the inference engine reason with the ontology to perform the diagnosis. The rules specify some conditions that are applied to an ontology by the engine to yield new facts. The application was developed using Java, and thus an inference engine capable of working with Java was needed. Many inference engines meeting this criterion exist, but the reasoner bundled with the Jena API was used. This is because the API is already used in interfacing and manipulating the ontology. Using the bundled reasoner also reduces the number of components in the system which in turn reduces the complexity of the system. Data, in the form of symptoms, is collected from the user through the graphical user interface and the Jena API will use inference rules like the one defined in Fig. 4 to infer whether the patient has a disease or not. The rules for determining hypogonadism were based on the Androgen Deficiency in Aging Males (ADAM) questionnaire found in Morley *et al.* (2000). The rules for determining cortisol and thyroid hormone imbalance were got from Smith (2005).

```
[hypo: (?patient odd:hasThis odd:Depression),
       (?patient odd:hasThis odd:ErectileProblem),
       (?patient odd:hasThis odd:Fatigue),
       (?patient odd:hasThis odd:Anxiety)
       -> (?patient odd:mayHave odd:TestosteroneDeficiency)]
```

Fig. 4: One of the inference rules for determining hypogonadism in the Jena reasoner syntax

Queries for Statistics

The symptoms users select during their interaction with the system are stored in the ontology. The age and location of the patients are also stored alongside these symptoms. Queries define a way of retrieving information from the ontology for performing the statistical analysis; information like the number of patients that have ever interacted with the system, number of patients that may have any of the four hormone imbalances, and the age and location of the patients.

The SPARQL Protocol and RDF Query Language (SPARQL) which is a semantic query language was used in defining the queries used by the system. SPARQL queries consist of a single or collection of triples and are of the form ‘subject-predicate-object’. A sample query for retrieving the number of patients in the ontology is given in Fig. 5.

```
PREFIX rdf: <http://www.w3.org/1999/02/22-rdf-syntax-ns#>
PREFIX odd: <http://www.lawal.com/moh/ontologies/2017/0/hypogonadism#>

SELECT (COUNT(*) AS ?x)
    WHERE {
        ?p rdf:type odd:Person
          }
```

Fig. 5: SPARQL query for retrieving number of patients in ontology

RESULTS AND DISCUSSION

The interface of the system was developed using JavaFX

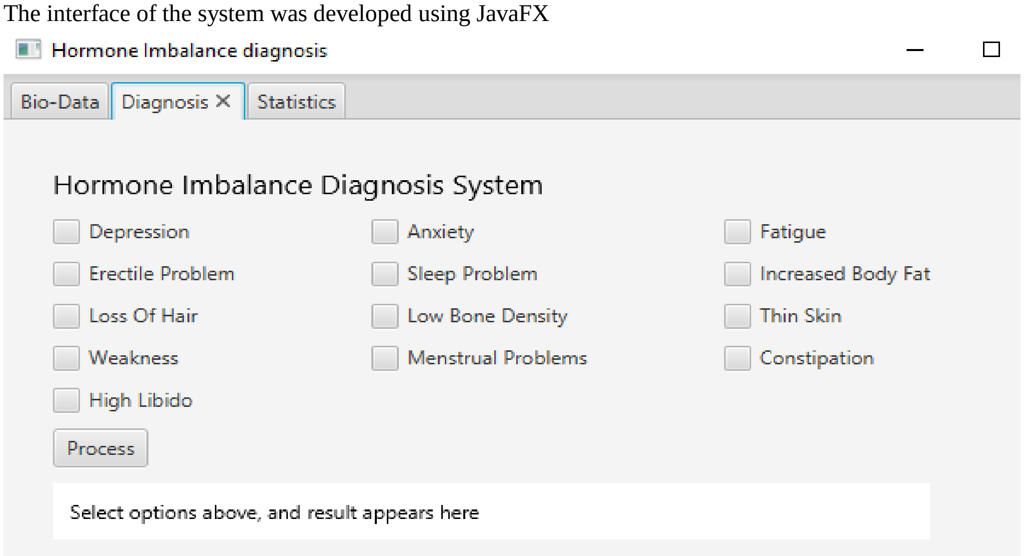

Fig. 6: Main user interface of the system

Users interact by selecting symptoms using the checkboxes, selecting gender and age using the drop down and pressing the Process button as shown in Fig. 6. The output is then presented in the textarea in the lower part of the system. The user may also click the Statistics button to display the statistics window as shown in Fig. 7.

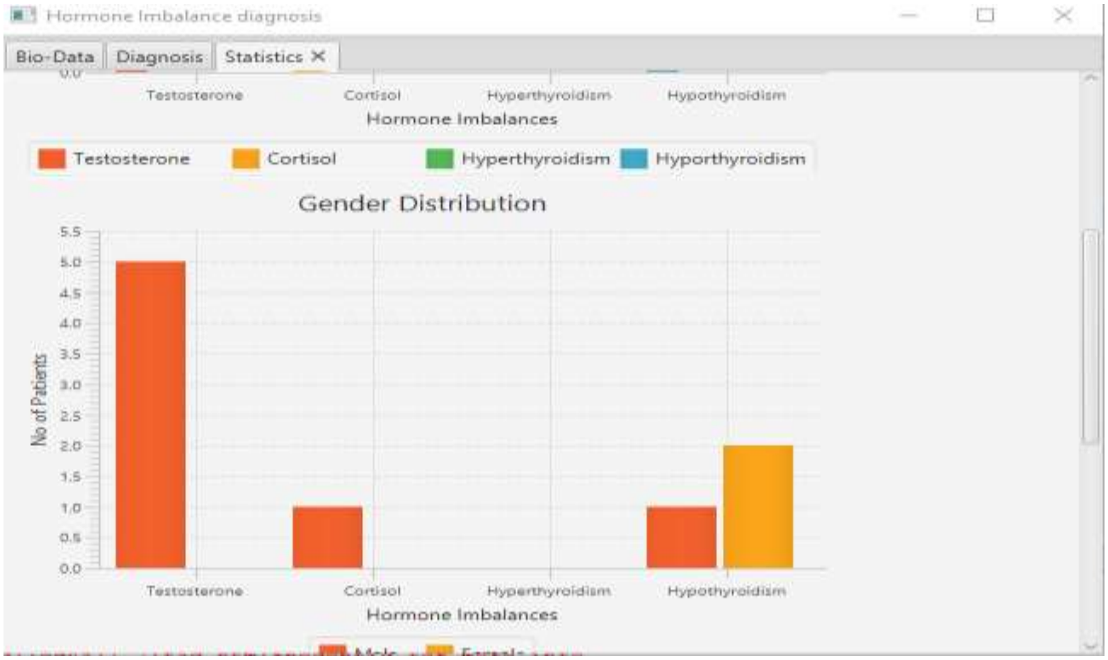

Fig. 7: Statistics Window

Evaluation

The system was evaluated with Rawte and Roy (2015) using the metrics developed by Manouselis et al. (2010).

Table 1: Evaluation of knowledgebase of the proposed system and Rawte and Roy (2015)

	No of Clas-ses (noc)	No. of Inst-ances (noi)	No. of Properties (nop)	No. of Classes with Instances (ci)	Class Richness (cr)	Inheritance Richness (ir)	Relationship Richness (rr)	No. of Inference Rules (noir)
Propo-sed System	4	20	4	3	0.75	0.08	0.6667	5
Rawte and Roy (2015)	14	16	6	3	0.2143	0.14	0.3077	2

The system is backed by a knowledgebase, but there are other features that can be evaluated as well. Table 2 shows a feature-based evaluation of the system with that of Rawte and Roy (2015).

Table 2: Feature based comparison of proposed system and Rawte and Roy (2015)

	Diseases diagnosed	Patient data storage location	Statistical analysis
Proposed System	Thyroid disease, Testosterone deficiency, and cortisol deficiency	Ontology	Yes
Rawte and Roy (2015)	Thyroid disease	MySQL database	No

The three metrics that define how well an ontology is designed were used in the evaluation: these were the class richness (cr), inheritance richness (ir), and the relationship richness (rr). Higher values of the class richness metric indicate an ontology that represents most of the knowledge in the domain. The proposed system has a class richness value of 0.75 or 75% while that of Rawte and Roy (2015) had a class richness value of 0.2143 or 21.43%. This means this system is 2.5 better than that of Rawte and Roy (2015). This improvement was anticipated because the ontology used by the proposed system modelled more knowledge about hormone imbalances. The proposed system has more symptoms and more diseases in the ontology.

The inheritance richness metric measures the depth of the ontology, and the nature of the knowledge stored in it. An ontology with a low inheritance richness metric indicate a horizontal ontology that models general knowledge. Conversely, an ontology with a high inheritance richness metric indicate an ontology that models specific knowledge. The proposed system presented with an inheritance richness metric of 0.08 while that of Rawte and Roy (2015) had an inheritance richness metric of 0.14. Their system is 42.86% better using this metric, and this is to be expected as the specificity of the ontology is reduced by adding more symptoms and diseases to it. The relationship richness metric is a measure of how well a particular domain is defined in an ontology. Higher values of this metric indicate a richer taxonomy. This system has a relationship richness of 0.6667 or 66.67% while that of Rawte and Roy (2015) has a relationship richness of 0.3077 or 30.77%. This means this system is 1.2 times better in this regard. This improvement was also expected as describing closely related symptoms and diseases improves the domain of hormone imbalance.

The system developed by Rawte and Roy (2015) employed the use of a MySQL database for storing patient information. The proposed system removed the MySQL database and moved storage of patient information to the ontology. One of the benefits of using an ontology instead of a database is the ease of sharing information. Patient information can easily be shared with other expert systems. The system proposed by Rawte and Roy (2015) does not perform any statistical analysis on the data collected from the user. The proposed system is able to perform analysis like getting the percentage of users that have tested positive for any of the hormone imbalances and present the findings on a bar chart. The system can also present the same statistics based on the 'age' and 'gender' of the users.

CONCLUSION

This paper looked into screening related hormone imbalances using an ontology as the knowledge base with the help of relevant rules. Results found adding information about more hormone imbalances to the knowledge base improved overall knowledge representation and ability to screen similar hormone imbalances, but also reduced the specificity of the ontology. This aided in the diagnosis of three closely related hormone imbalances by the system. The system was also able to store patient information in the knowledgebase allowing for easy sharing and generation of statistical reports.

The system does not deal with uncertainties in descriptions of symptoms by users. This could possibly be achieved by using Bayesian networks. We leave this as possible future work.

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