International Journal of Engineering Research & Technology (IJERT)
ISSN: 2278-0181
Vol. 4 Issue 06, June-2015

Thyroid Disease Diagnosis using Ontology based Expert System

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***Abstract*—: This study shows a new method in building up an ontology based expert system to diagnose thyroid diseases. Ontology describes some domain in the form of concepts and relationships among them. It has an advantage of being computer and human readable. The acceptance of ontology in medical field has facilitated the domain experts and non-experts to execute the job of knowledge representation easily. This paper presents an ontology depicting the domain of thyroid diseases and its related symptoms. The thyroid gland is an important organ because it is responsible for metabolism in the body. Diseases related to thyroid gland can be difficult to diagnose because symptoms are confused with other possible conditions easily. This study proposes to build an expert system based on ontology for diagnosing thyroid diseases. It uses ontology to construct the domain knowledge and rules to infer the thyroid disease related diagnosis.**

Keywords—Thyroid; expert system; ontology

I. INTRODUCTION

The thyroid is a butterfly-shaped gland that lies wrapped around the windpipe and is positioned under the Adam's apple. The job of the thyroid gland is to secrete thyroid hormones that help regulate metabolism. The thyroid gland produces two necessary hormones–thyroxine (T4) and triiodothyronine (T3). Normally, most T3 in the blood stream gets converted from T4. These two similar hormones are called together thyroid hormone. When the degree of thyroid hormone is too small, the pituitary gland secretes a hormone called thyroid stimulating hormone (TSH) which stimulates the thyroid to produce more thyroid hormone. When the level of thyroid hormone goes back to normal, the pituitary gland ceases producing extra TSH. If thyroid hormone is present in excess, the pituitary gland makes less TSH and thus less production of thyroid hormone by the thyroid gland. The pituitary gland is stimulated by the hypothalamus to produce TSH by secreting thyrotropin releasing hormone (TRH). This entire system is called the hypothalamic-pituitary-thyroid axis because of the feedback loop between the pituitary gland and the thyroid gland which maintains normal levels of T4, T3 and TSH [1]. When thyroid gland fails to maintain the normal levels of TSH there arises mainly two types of thyroid related diseases called hyperthyroidism and hypothyroidism. First, hyperthyroidism, also known as over active thyroid disease, is the condition that occurs due to excessive production of thyroid hormone by the thyroid gland. Second, hypothyroidism, also called underactive thyroid disease, is a disorder in which the thyroid gland does not produce enough thyroid hormone [2].

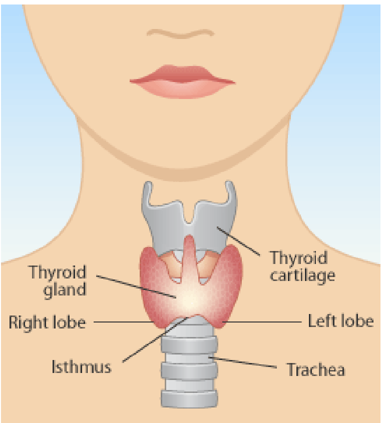

Fig. 1.a. Thyroid Gland [1]

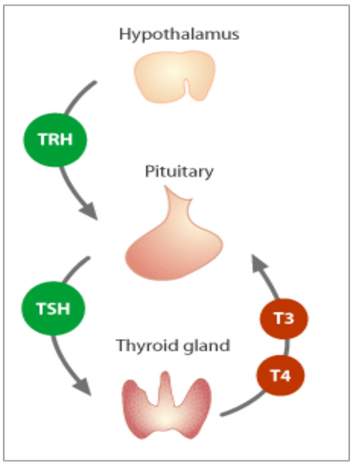

Fig. 1.b. Hypothalamic-Pituitary-Thyroid axis [1]

Depending only on one blood test, TSH, leaves millions of people undiagnosed and their symptoms are often confused with other conditions such as weakness, fatigue, etc. So, there is a necessity of an expert system which can consider the symptoms in addition to TSH levels and make the diagnosis respectively.

An expert system is artificial intelligence based interactive computer application that performs a task that would otherwise be performed by a human expert. Expert systems typically consist of three parts: (1) a knowledge base which contains the information acquired by questioning experts, and logic rules that control how data is applied; (2) an inference engine that understands the submitted problem against the rules and logic of information stored in the knowledge base and infers accordingly; and an (3) an interface that allows the user to show the problem in a human language such as English.

Benefits of developing an Expert System:

- Expert system is available any time.
- The cost of offering expertise is cheaper.
- The knowledge will last for an indefinite time.
- Expert system can be made to have knowledge from multiple experts.
- It is capable of developing the reasoning that directed to a conclusion.
- It can respond fast because of the underlying benefits of computers over humans.
- It is unemotional and responds at all times and works steadily during urgent situations.

An Expert System for Thyroid Diagnosis is developed with the purpose of assisting the physician in diagnosing thyroid disease.

Ontology is a body of knowledge describing some domain in form of concepts and relationships between them. Need for using ontology arises because of some drawbacks of Machine Learning Techniques such as requirements for training data, high computation for model learning, adapting with dynamic nature of data. Humans are capable of using the Web to carry out tasks. However, a computer cannot accomplish the same tasks without human direction because web pages are designed to be read by people and not machines. The semantic web is understandable by computers and so that they can do more of the complex work involved in searching, sharing, and merging information on the web.

Benefits of using Ontology [3]:

- Interaction among systems, among humans, and between humans and systems.
- Computational inference.
- Reuse and organization of knowledge.

Combining the three concepts discussed above, i.e. Expert System (ES), ontology and thyroid disease into an expert system based on ontology shall diagnose thyroid disease.

In this paper, an expert system based on ontology for diagnosis of thyroid diseases that uses the above methods is presented. The draft of this paper is as follows. Section II gives the literature survey. Section III shows the proposed approach. Section IV shows the results and analysis for the above system. Finally, Section V concludes along with the work which can be done afterward in future.

II. Literature Survey

The occurrence of Hypothyroidism is nearly about 1 in 2,640 where almost 40,000 newborn babies were screened, which is much higher than the worldwide statistic of 1 in 3,800. Diagnostic time lag in hypothyroidism is because of lack of awareness among primary health care practitioners, family physicians and the cost, availability of laboratory investigations [4].

The study presents that Hypothyroidism among women was extremely high in this region [5].

4543 children in the age group of 6-18 years from different schools in Delhi were screened during the research study for thyroid function and it was found that goiter is prevalent in children (17%) even after two decades of iodization [6].

Females above 35 years living in a randomly selected urban area Elamakkara in Cochin were surveyed for thyroid disease. This survey clearly showed that undetected thyroid disorders are very high in this community, demanding development of suitable screening strategies to detect and treat these conditions considering the level of health problems it can cause to the population [7].

According to various research studies on thyroid disease, it has been estimated that about 42 million people in India suffer from thyroid diseases [8].

[9] suggests that the UK is iodine-deficient, indicating need for examination of the UK iodine status and recommendations to safeguard public health.

The subject population with 4409 adult members of resident welfare associations of 5 residential areas, from 18-90 years of age participated in general health check-up camps. 30.6% of subjects with severe hypoechogenicity were diagnosed and this percentage was found higher in women [10].

The study was conducted in eight major cities – Mumbai, Bangalore, Delhi, Ahmedabad, Chennai, Kolkata, Hyderabad and Goa. "The prevalence of hypothyroidism was more, impacting roughly one in 10 adults. Females and old people were found to have substantial association with hypothyroidism," said the study [11].

The hospital based survey involved 4739 patients having undergone thyroid function test, in the central clinical biochemistry laboratory of Subharti Medical College, UP and its associated hospital. More prevalence of Hypothyroidism was observed in patients (especially females). The findings confirm the usefulness of screening of thyroid function mandatory for early detection and treatment to reduce the effects of thyroid dysfunctions [12].

Out of the 325 districts surveyed in India so far, 263 are IDD-endemic and iodine deficiency causes goiter [13].

More lately, estimates show that 27 million Americans

have thyroid disease out of which about 13 million people go undiagnosed [14].

A correlation is present between iodine intake and thyroid diseases; high iodine intake may lead to the occurrence of thyroid diseases such as Hashimoto’s thyroiditis, nodular goiter and Hyperthyroidism through a long-term mechanism. Females with high iodine intake had a close relationship with thyroid diseases than males [15].

Gist of this literature survey states that the thyroid disease is one of the most prevalent diseases worldwide.

Edward Feigenbaum [16] defined an Expert System (ES) as: "An expert system is an intelligent computer program that uses knowledge and inference procedures to solve problems that are difficult enough to require significant human expertise for the solution."

Medical diagnosis decision making becomes a very tedious task because the human experts make decisions and they can barely process the immense amounts of data. So an automated tool is required that should help them make an effective and right decision. An artificial neural network has the ability to imitate decision-making process, and use a knowledge base, and a training set, to learn to diagnose diseases. A medical decision support system based on the neural network is trained by applying an improved Back Propagation algorithm for Medical Diagnosis. The hidden layer of a neural network plays an important role for detecting the relevant features [17].

Case-based Reasoning (CBR) methodology provides a way to solving problems with recalled knowledge of solved cases. This paper gives a system model that employs the simplified medical knowledge: disease-symptom ontology for pre-diagnosis, given patient's symptoms as the input [18].

A new hybrid case-based reasoning approach for medical diagnosis is proposed to improve the accuracy of the CBR systems. The approach includes case-based reasoning and rule-based reasoning, and also carries out the adaptation process automatically by using adaptation rules. The rules viz., adaptation rules and reasoning rules are produced from the case-base. On solving a new case, the case-base is refined, and both adaptation and reasoning rules are updated [19].

A comparative thyroid disease diagnosis by using multilayer, probabilistic, and learning vector quantization neural networks is given in [20].

A three-stage expert system is proposed to diagnose thyroid disease. This system uses feature selection (FS), particle swarm optimization (PSO) and support vector machines (SVM). The PSO-based system costs a lot of computational time [21].

Linguistic Hedges Neural-Fuzzy Classifier with Selected Features (LHNFCSF) is given for diagnosis of thyroid diseases. The work to be done in the future includes improving the performance of NFCs using high-performance computing methods [22].

III. Proposed Approach

Considering the drawbacks of the machine learning techniques, this paper proposes to diagnose thyroid related problems with the assistance of an expert system based on ontology. This is shown in Fig.2.

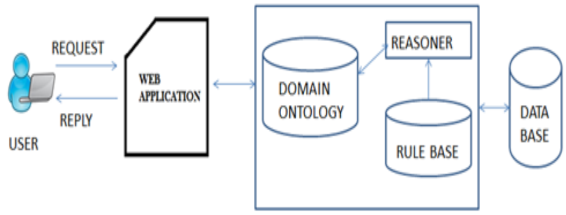

Fig.2 . Block Diagram: OBESTDD [23]

It consists of six main blocks. User is a person who wants to know diagnosis of thyroid disease. User uses Web application to interact with the computer. Domain Ontology represents concepts which belong to the real world. A reasoner is a tool which has the capability to infer logical consequences from a set of facts. Rule Base is the collection of rules which are used to infer diagnosis from the domain ontology. Database contains the patients records based on which the expert system can make the diagnosis. This can be MySQL database.

Ontology for thyroid disease is created using an ontology editor called Protégé. The ontology acts as the knowledge base for this expert system. Rules are written in Semantic Web Rule Language (SWRL). Jena is used an inference engine to deduce diagnosis based on the knowledge and SWRL rules. MySQL database is used a backend to store users' details. Front end is a web application using JSP and acts as an interface through which user can enter the symptoms to know their diagnosis. Fig. 3.a. shows the Thyroid Ontology designed using ontology editor called Protégé. Fig 3.b. shows Thyroid Ontology Classes. Fig. 3.c. shows Thyroid Symptoms and Fig. 3.d. shows Thyroid Ontology Object Properties.

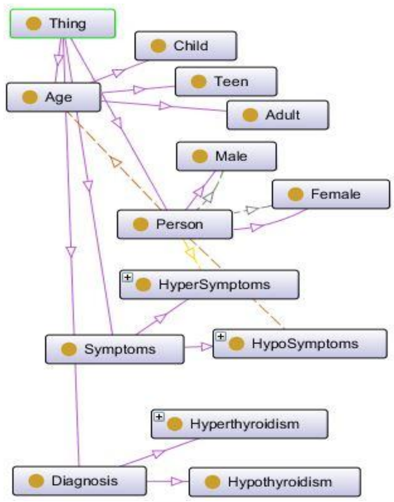

Fig. 3.a. Thyroid Ontology

There are 4 main classes, viz. Age, Person, Symptoms and Diagnosis and 9 subclasses, viz. Child, Teen, Adult, Male, Female, HyperSymptoms, HypoSymptoms, Hyperthyroidism, and Hypothyroidism in the thyroid ontology shown in Fig. 3.a. Symptoms are instances of the classes which are shown in Fig. 3.c. For example, Heat Intolerance is the symptom (instance) of class HyperSymtoms.

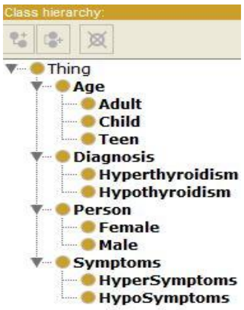

Fig. 3.b. Thyroid Ontology Classes

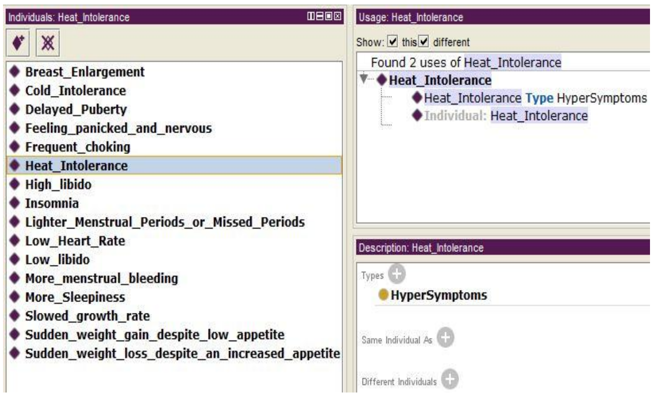

Fig. 3.c. Thyroid Symptoms

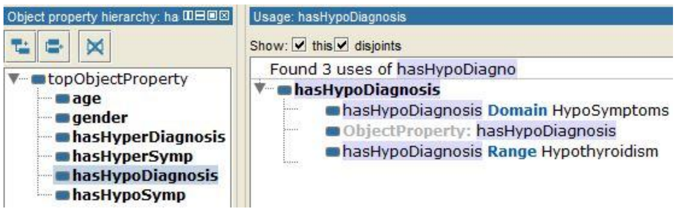

Fig. 3.d. Thyroid Ontology Properties

Consider an example where a user named Rucha selects the age & gender and related symptoms.

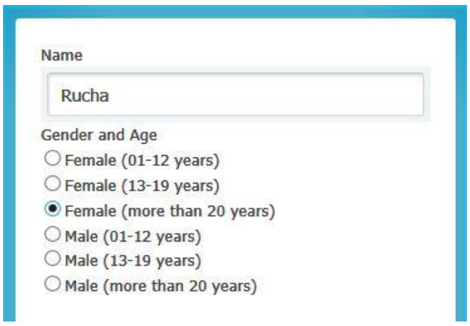

Fig. 4.a. User enters name and selects age & gender

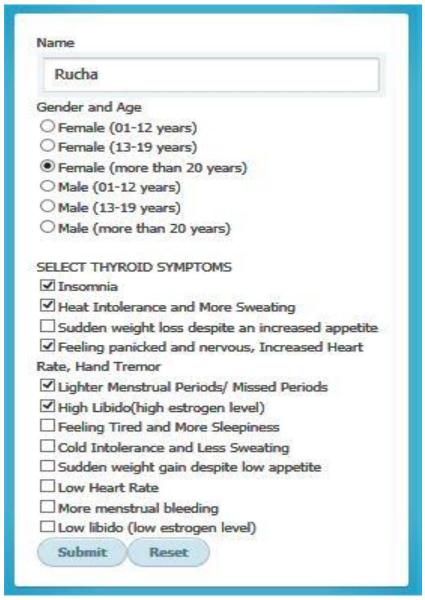

Fig. 4.b. User enters name and selects symptoms

Fig. 4.c. Ontology based Expert System shows result

```
@prefix: <http://www.w3.org/2002/07/owl#>.
@prefix p: <http://jena.hpl.hp.com/prefix#>.
:RUCHA p:gender :female.
:RUCHA p:age :adult.
:adult p:hasHyperSymp :Insomnia.
:adult p:hasHyperSymp :Heat_Intolerance.
:adult p:hasHyperSymp :Feeling_panicked_and_nervous.
:adult p:hasHyperSymp :Lighter_Menstrual_Periods_or_Missed_Periods.
:adult p:hasHyperSymp :High_libido.
:Insomnia p:hasHyperDiagnosis :HYPERTHYROIDISM.
:Heat_Intolerance p:hasHyperDiagnosis :HYPERTHYROIDISM.
:Feeling_panicked_and_nervous p:hasHyperDiagnosis :HYPERTHYROIDISM.
:Lighter_Menstrual_Periods_or_Missed_Periods p:hasHyperDiagnosis :HYPERTHYROIDISM.
:High_libido p:hasHyperDiagnosis :HYPERTHYROIDISM.
```

Fig. 4.d. Data (in turtle format) for a user

```
@prefix p: <http://jena.hpl.hp.com/prefix#>.
[rule1:
(?w p:gender ?x)
(?w p:age ?y)
(?y p:hasHyperSymp ?0)
(?y p:hasHyperSymp ?1)
(?y p:hasHyperSymp ?2)
(?y p:hasHyperSymp ?3)
(?y p:hasHyperSymp ?4)
(?0 p:hasHyperDiagnosis ?z)
(?1 p:hasHyperDiagnosis ?z)
(?2 p:hasHyperDiagnosis ?z)
(?3 p:hasHyperDiagnosis ?z)
(?4 p:hasHyperDiagnosis ?z)
->
(?w p:HAS ?z)]
```

Fig. 4.e. Rule (in SWRL format) for a user

Fig. 3.a. shows user selects gender "Female" and age "more than 20 years". Fig. 3.b. shows that Rucha selects symptoms (1) Insomnia, (2) Heat Intolerance and More Sweating, (3) Feeling panicked and nervous, Increased Heart Rate and Hand Tremor, (4) Lighter Menstrual Periods or Missed Periods, (5) High libido(high estrogen level). Fig. 3.c. gives the diagnosis. Fig. 3.d. and Fig. 3.e. show user data in

turtle format and rules in SWRL format. Based on the data and rule, Jena reasoned draws inference and gives the diagnosis. Fig.5. shows the flowchart for the mechanism of the expert system based on ontology. User has to first register and then login if already registered. Then user should select 'Age & Gender' and select the related symptoms accordingly. Finally, the result is given.

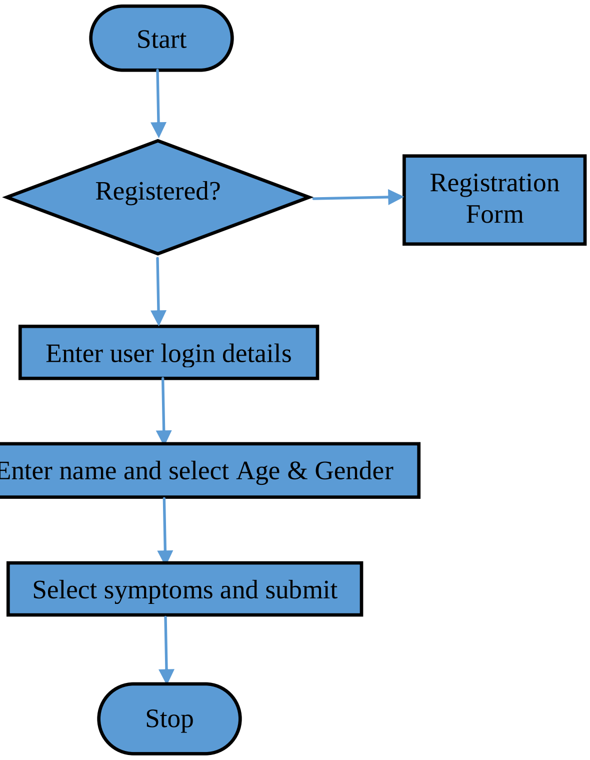

Fig. 5. Flowchart for the working of the expert system based on ontology.

Consider the following example:

Data given is:
Joseph p:isFatherof James
John p:isBrotherof Joseph

Rule given is:
rule: (?f p:isFatherof ?a) (?u p:isbrotherof ?f) -> (?u p:isUncleof ?a)

-
John isUncleof James

Similarly,

Data for a user is:
:RUCHA p:gender :female.
:RUCHA p:age :adult.
:adult p:hasHyperSymp :Insomnia.
:adult p:hasHyperSymp :Heat_Intolerance.
:adult p:hasHyperSymp :Feeling_panicked_and_nervous.
:adult p:hasHyperSymp :Lighter_Menstrual_Periods_or_Missed_Periods.
:adult p:hasHyperSymp :High_libido.
:Insomnia p:hasHyperDiagnosis :HYPERTHYROIDISM.
:Heat_Intolerance p:hasHyperDiagnosis :HYPERTHYROIDISM.
:Feeling_panicked_and_nervous p:hasHyperDiagnosis :HYPERTHYROIDISM.
:Lighter_Menstrual_Periods_or_Missed_Periods p:hasHyperDiagnosis :HYPERTHYROIDISM.
:High_libido p:hasHyperDiagnosis :HYPERTHYROIDISM.

Rule for this user is:

rule:(?w p:gender ?x)(?w p:age ?y)(?y p:hasHyperSymp ?0)(?y p:hasHyperSymp ?1)(?y p:hasHyperSymp ?2)(?y p:hasHyperSymp ?3)(?y p:hasHyperSymp ?4)(?0 p:hasHyperDiagnosis ?z)(?1 p:hasHyperDiagnosis ?z)(?2 p:hasHyperDiagnosis ?z)(?3 p:hasHyperDiagnosis ?z)(?4 p:hasHyperDiagnosis ?z)->(?w p:HAS ?z)]

--

RUCHA HAS HYPERTHYROIDISM

where,

1. w is an instance of class Person.
2. x is an instance of class Female.
3. y is an instance of class Age.
4. 0,1,2,3,4 are instances of class HyperSymptoms
5. z is an instance of class Hyperthyroidism

Likewise, based on the symptoms entered through the interface, data and rules get generated for the users. Then, a reasoned called Jena uses these data and rules to infer and make the diagnosis accordingly.

IV. Results and Analysis

Implementation of the expert system for thyroid disease diagnosis based on neural network and ontology gave the following results. Total number of instances taken is 60. Neural network considers only TSH, T3, T4 levels in the blood to train the neural network whereas there is no training for the ontology based expert system. It makes diagnosis based on symptoms.

TABLE I. Neural Network

Class	Predicted Right Number of Instances	Total Number of Instances	Accuracy in %
Normal	31	34	91.176
Hyper	1	11	9.091
Hypo	5	15	33.333
Male	16	23	69.565
Female	21	37	56.757
Child	4	6	66.667
Teen	8	11	72.727
Adult	27	43	62.791

TABLE II. ONTOLOGY BASED

Class	Predicted Right Number of Instances	Total Number of Instances	Accuracy in %
Normal	32	34	94.118
Hyper	9	11	81.818
Hypo	13	15	86.667
Male	20	23	86.957
Female	35	37	94.595
Child	5	6	83.333
Teen	9	11	81.818
Adult	40	43	93.023

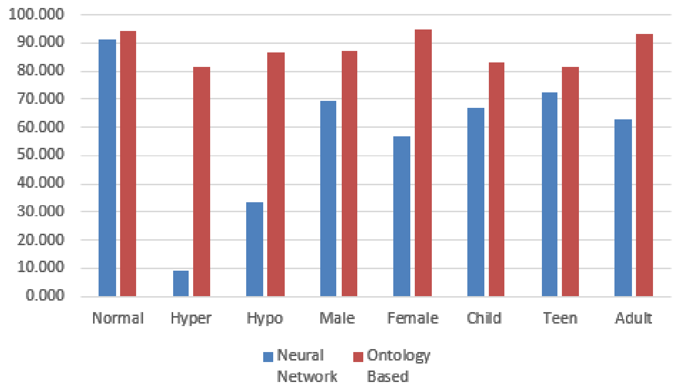

Fig. 6. Comparison of Neural Network and Ontology Based Expert System

The analysis of the results presented in Fig.6 shows that expert system based on ontology gives more accurate results with lesser complexity than the one which uses neural network.

V. CONCLUSION

Thyroid disease diagnosis is difficult and often confused with symptoms of other diseases. If diagnosis is prognosticated, then by advising proper prevention methods and dosages to the patients, they can be recovered in the beginning. Thus, expert system based on ontology for diagnosis of thyroid disease has been proposed and implemented. Validation is done based on two parameters:

- Accuracy
- Time Complexity

Thus, expert system based on ontology gives better results with lesser complexity.

REFERENCES

[1] "Thyroid disorders", http://www.abc.net.au/health/library/thyroid_ff.htm. June 06, 2015 [June 16, 2005].

[2] James Norman, "Your Thyroid Gland," http://www.endocrineweb.com/conditions/thyroid/your-thyroid-gland.

[3] Bürger, Tobias, and Elena Simperl. "Measuring the benefits of ontologies." On the Move to Meaningful Internet Systems: OTM 2008 Workshops. Springer Berlin Heidelberg, 2008.

[4] Meena P Desai, "Disorders of Thyroid Gland in India", Symposium:Endocrinology-Part I Indian J Pediatr 1997;64: 11-20.

[5] Rebecca Abraham, V Srinivasa Murugan, P Pukazhvanthen and S K Sen, "Thyroid Disorders In Women Of Puducherry", Indian Journal of Clinical Biochemistry, 2009 / 24 (1) 52-59.

[6] V.K. Chauhan, R.K. Manchanda, Archana Narang, R.K.Marwaha, Saurav Arora, Latika Nagpal, "Prevalence of Thyroid Disorders in School Children In Delhi – Post Iodization Scenario", Indian Journal of Research in Homoeopathy Vol. 3, No. 3, July-September 2009.

[7] DR V USHA MENON, "Spectrum Of Thyroid Disorders, Iodine Status And Diabetes Mellitus In Adult Kerala Population – A Community Study". THESIS SYNOPSIS, Amrita Viswavidyapeetham University, 2011.

[8] Ambika Gopalakrishnan Unnikrishnan and Usha V. Menon, "Thyroid disorders in India: An epidemiological perspective", Indian J Endocrinol Metab. Jul 2011; 15(Suppl2): S78–S81.

[9] "BTA Research Team: UK is now Iodine Deficient" Internet: http://www.btf-thyroid.org/index.php/thyroid/research-news/ukis-iodine-deficient. Sep 05, 2014 [April 15, 2011].

[10] Raman Kumar Marwaha, Nikhil Tandon, Mohd Ashraf Ganie, Ratnesh Kanwar,Aparna Sastry MK Garg, Kuntal Bhadra, Satveer Singh, "Status of Thyroid Function in Indian Adults: Two Decades After Universal Salt Iodization", JAPI April 2012,VOL. 60.

[11] Neetu Chandra, "India's cities in the grip of thyroid disease as new study reveals one in ten suffer from disorders". Internet: http://www.dailymail.co.uk/indiahome/indianews/article2408114/India s-cities-grip-thyriod-problems-new-study-reveals-sufferdisorders.html Sep 05, 2011.

[12] Naved Ahmad, Meenakshi Panthari, Akash Gupta, Prasnna Chandra, Sana Nafees, "Prevalence Of Hypothyroidism Among Patients Of Meerut, Uttar Pradesh: A Hospital Based Study," Research Article, March,2013.

[13] Chandrakant S. Pandav, Kapil Yadav, Rahul Srivastava, Rijuta Pandav* & M.G. Karmarkar, "Iodine deficiency disorders (IDD) control in India," Indian J Med Res 138, September 2013, pp 418-433.

[14] Mary Shomon, "Why Are So Many People Getting Thyroid Disease?," http://thyroid.about.com/od/symptomsrisks/a/Why-Are-So-Many-People-Getting-Thyroid-Disease.htm Sep 05,2014 [Aug 18, 2014].

[15] Hengqiang Zhao & Yuan Tian & Zeming Liu & Xiaoyu Li & Mengyu Feng & Tao Huang, "Correlation Between Iodine Intake and Thyroid Disorders: A Cross-Sectional Study from the South of China," Biological Trace Element Research, August 2014.

[16] A. Barr and E.A. Feigenbaum, The Handbook of Artificial Intelligence, Vol. 1, William Kaufmann, 1981.

[17] Ashish Dehariya, Vijay Choudhari, Ilyas Khan and Sourabh Karsoliya, "An Effective Approach for Medical Diagnosis Preceded by Artificial Neural Network Ensemble," Proceedings of the 5th National Conference; INDIACom, 2011.

[18] Hsien-Tseng Wang, Abdullah Uz Tansel, "Composite Ontology- Based Medical Diagnosis Decision Support System Framework," Communications of the IIMA, Volume 13 Issue 2, 2013.

[19] Dina A. Sharaf-El-Deen & Ibrahim F. Moawad & M. E. Khalifa, "A New Hybrid Case-Based Reasoning Approach for Medical Diagnosis Systems," Springer Science+Business Media New York, 2014

[20] Feyzullah Temurtas, "A comparative study on thyroid disease diagnosis using neural networks," Expert Systems with Applications 36 (2009) 944–949.

[21] Hui-Ling Chen & Bo Yang & Gang Wang & Jie Liu & Yi-Dong Chen & Da-You Liu, "A Three-Stage Expert System Based on Support Vector Machines for Thyroid Disease Diagnosis", J Med Syst (2012) 36:1953–1963.

[22] Ahmad Taher Azar, Aboul Ella Hassanien and Tai-hoon Kim, "Expert System Based on Neural-Fuzzy Rules for Thyroid Diseases Diagnosis," Communications in Computer and Information Science, 2012.

[23] Vipula Rawte and Bidisha Roy, "OBESTDD: Ontology Based Expert System for Thyroid Disease Diagnosis." International Conference on Nascent Technologies in the Engineering Field (ICNTE), IEEE, 2015.