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Ontology Based Multi Parameter Disease Diagnosis Model

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ABSTRACT

Chronic diseases are long lasting diseases with complex causes and many risk factors. Chronic diseases cause functional impairments. Most Chronic diseases cannot be cured completely and some diseases can be immediately life threatening like heart stroke and some diseases stays for long time and needs continues management such as diabetes which may causes even death. Most of the chronic diseases will stay for long time but may not be a cause of death such as arthritis. According WHO chronic diseases kills 41 million people each year which is 74% of all deaths globally. Out of these total deaths 77% are from low- and middle-income countries. To prevent or control these diseases and minimize the losses or to minimize the budgets allotted by the governments to handle these diseases a special care has to be taken. The patients need to be aware of these diseases and continues monitoring is required. In this paper we discuss various fields in healthcare where semantic web can be used to handle the issues of chronic diseases. We have constructed an Ontology based model for diagnosing two chronic diseases namely heart stroke and diabetes. We have validated our model by using AI based symptom checkers.

Keywords: Chronic Diseases, Ontology, Diagnosis, Semantic web, SPARQL.

1. INTRODUCTION

Chronic diseases will occur due to many characteristics. Environmental risk factors such as air pollution is also causing chronic diseases like chronic obstructive pulmonary disease and lung cancer [1]. Socioeconomic impact also causes chronic illness because of consumption of harmful products such as tobacco and lack of costly health care services [2]. Some chronic diseases are genetic in nature i.e. comes from parents like type 2 diabetes and breast cancer. Some chronic diseases are based on demographics i.e. a group of people belonging to a region may have common chronic diseases. For example, the state Goa has highly westernized and it impacted on life style which causing this state to have higher obesity rate in India [3]. Chronic diseases prevention and control requires monitoring progress, risk of the diseases and managing the diseases which includes diagnosis and medications [4]. The effects of chronic diseases can be eventually reduced if diseases are detected early, consumption of harmful products such as alcohol and tobacco is avoided and active life style is maintained.

The Article is structured as in Section-2 we have discussed need of semantic web technologies for disease diagnosis. In section-3 various areas of health care domain where semantic web can be applied is discussed. In Section-4 we have constructed an ontology for diagnosis of diabetes and heart stroke. We have validated our proposed model in section-5 by comparing the results with results AI based symptom checkers and we have given future scope and conclusion in 6th section.

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2. NEED OF SEMANTIC WEB FOR CHRONIC DISEASE DIAGNOSIS.

In this era people are using internet to learn about diseases and their causes and treatments available. People also use websites for knowing medicine of a disease even side effects of them. They type only a set if search keywords in search engines and gets thousands of website links as response. They visit only few websites to get knowledge. But some other links may have valuable information which is not even clicked. And the links which have opened may not contain trusted data. Semantic web solves these problems by using linked data. Where related data from different pages are linked to each other. Whereas traditional web is called as web of documents and semantic web is called as web of linked data whose meanings can be understood by machines. And traditional web consists of documents only in human understandable way whereas semantic web consists of data such that machines and human both can understand. Semantic web integrates data from various sources and forms a unified knowledge base. Many people can be benefitted from semantic web but patients with chronic diseases gets more benefitted because they need long time management of chronic diseases. The Mager features of semantic web are RDF, RDFS, OWL, ONTOLOGY, SPARQL, KNOWLEDGE BASE and INFERENCE ENGINE.

2.1 Resource Description Framework (RDF)

RDF provides a standard mechanism for expressing data as linked graph and sharing it around the world on the machines. RDF contains data in the form of triples <subject, predicate, object>.Subject and predicate represent resources which can be identified using URI where as Object can be a resource identified by URI or a string literal. RDF triples can be serialized such that they can be stored in text files. Various serialization formats exist like N-triples, N3, RDF/XML, RDFa and Turtle. Following figure fig.1 is an RDF statement which represents few symptoms of diabetes and we have used namespaces to shorten the URIs.

@prefix dis: <https://www.who.int/news-room/fact-sheets/detail/>.

@prefix symp: <https://www.cdc.gov/diabetes/basics/>.

dis: diabetes symp: symptoms "feels Very Thirst", "Have blurry vision",

"Have numb or tingling hands or feet", "Feel very tired", "Have very dry skin", "Have sores that heal slowly", "Have more infections than usual".

Above RDF data can be represented as RDF Graph as follows.

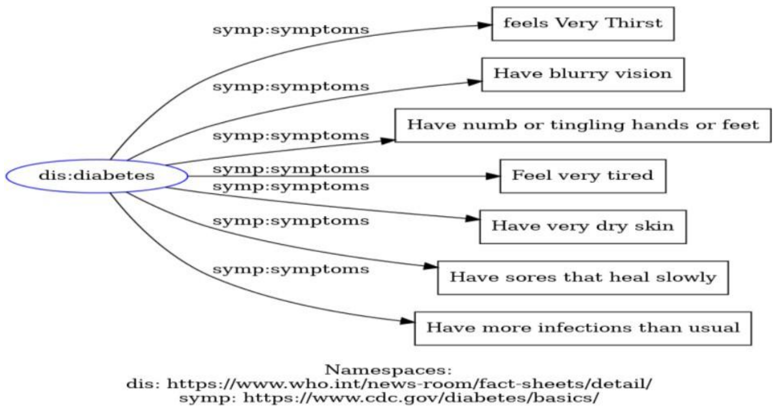

Fig.1: Linked data Representation of diabetes and it's symptoms

2.2 RDFS

RDFS is a modelling language, it is originally called as "RDF Vocabulary Description Language". It allows to define classes using **rdfs:class** construct and instantiate using **rfd:type** construct. We can also define properties using **rdf:property** construct. Properties will connect the classes with other classes or literals. Properties will have restriction like their domain and range defined using **rdfs:domain** and **rdfs:range** constructs. Description of this

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vocabulary is set of meta data accessible to SPARQL queries. For providing detailed meta data **rdfs: label** and **rdfs: comment** constructs are used.

2.3 Ontology

An ontology is defined using Web Ontology Language (OWL) and it is collection of triples of a particular domain. An ontology provides vocabulary with which knowledge can be represented and understood by computers. Ontology consists of classes, properties and relationships among classes. Ontologies helps to define formal rules for inference. RDFS is a language for building ontologies. Ontology learning is a mapping between ontology components, where some components are given and some are missing. Ontology learning helps to include missing components. Ontologies and metadata improve information retrieval task.

2.3.1 Ontologies for patient records

In computer-based patient records systems like PHR, HER, patient's data containing various attributes are stored in databases of computer systems. This data can be used for reminding the patients about medications and visits, for giving alerts, for constructing decision support systems and to acquire knowledge. The attributes of the data base should be related with heterogeneous data and every attribute must have meaning. The databases will implement locks to protect some data, but public data can be used for other purposes [5]. Ontology provides a vocabulary and structure with semantics of this data by linking various data elements and makes the data to sharable and machine interpretable [6]. Computer based patient record ontology (CPR ontology) is constructed for this purpose [7]. Authors of [8] constructed one health ontology system using ontology accumulator which combines knowledge from various domain like patients, doctors and diseases and constructed one cumulative system.

2.3.2 Ontologies for clinical pathways

Clinical pathways (CPW) helps the health care professionals by guiding them by recommending the path to be followed in the treatment plan. Clinical pathways are different form workflows of business and production environments which contains static inputs, CPWs need to be more flexible by considering dynamic parameters and needs to be customized to a treatment plan for individual patient context [9][10]. The success of following clinical pathways depends on medical knowledge and expertise of medical professionals and available resources. The health care professionals with less expertise can diagnose a disease wrongly or prescribe a medicine which is not required, which causes medical errors. CPWs helps the health care professionals in decision making by considering input information like symptoms, level of disease, and position in course of diagnosis and suggesting medications by considering side effects, interactions of drugs and allergies of patients. Efficient CPWs reduce cost and optimizes resource usage [11]. Ontologies can be used to link this heterogeneous data and understand the interactions among these domains for successful diagnose of a disease. Authors of [12] constructed ontology based CPW by considering patient context and medical guidelines. Authors of [13] constructed ontology based clinical pathway by converting clinical guidelines into rules using SWRL. In [14] authors constructed a sharable and reusable clinical pathway ontology by considering CPW, guidelines, resources and context. The efficient clinical pathway needs standardized terms. Authors in [15] used SNOMED-CT terms for constructing CPW using standardized medical terminologies.

2.3.3 Ontologies for medical terminologies

Medical terminology refers to the words used in health care domain. It contains abbreviations, synonyms, descriptions for symptoms, diseases, drugs, procedures, conditions and treatments in medical field. Medical terminology contains of a set of terms used across the globe in the medical field. Medical terminology is important because health care professionals and patient may use different synonyms and abbreviations for same symptoms, diseases and drugs. If a predefined set of terms is already defined, by using those terms efficient results can be retrieved. When ontology is used in medical terminology, the medical terms can be linked to each other to form relationships and enable sharing of medical knowledge [16]. Following are the few ontologies defined for medical terms.

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2.3.3.1 SNOMED CT

Systematized Nomenclature of Medicine—Clinical Terms (SNOMED CT) is an ontology based on ontology of general medical science (OGMS). It is a systematically organized collection of medical terms by providing codes, synonyms, abbreviations, definitions for terms used for documenting and reporting [17]. SNOMED CD ontology provides terms of medical field by using which redundancy and inconsistency in medical terms can be eliminated. This ontology can be used in mobile health applications, and decision support systems by providing semantic interoperability among distributed health records [18].

2.3.3.2 ICDO:

International classification of Diseases ontology (ICDO) is a representation of ICD-9 and ICD-10, which represents disease terms and their relationships. ICDO provides knowledge about disease and deaths world wide by using clinical terms coded with ICD [19]. ICD provides recordings, analysis, and interpretation of data about morbidity and mortality collected worldwide. Ontology representation of ICD allows semantic interoperability and reusability of collected data for decision support and allocation of resources [20].

2.3.4 Ontologies for diseases

Disease ontologies provide information about diseases, disorders or illness. These ontologies is used to annotate the diseases in biomedical datasets, construction of knowledge bases and documentation of electronic health records.

2.3.4.1 Disease Ontology (DO)

Disease ontology [21] is developed by University of Maryland School of Medicine, Institute for Genome Sciences. This ontology provides reusable descriptions and vocabulary of concepts for human disease terms by cross mappings between Mesh, ICD, NCD thesaurus, SNOMED. DO-KB is a Disease Ontology knowledge base constructed for findability, interoperability, accessibility and reusability of disease related information using SPARQL query language and search interface [22]. The authors in [23] constructed infectious disease ontology. In [24] author have constructed neurological disease ontology. Open Biological and Biomedical Ontology Foundry (OBO Foundry) consists of interoperable ontologies in biological science, it consists of various individual disease ontologies [25].

2.3.5 Ontologies for Symptoms.

Symptom ontology (SYMP) is constructed based on signs and symptoms of human diseases. The ontology consists symptoms categorised according human organs like abdominal symptoms, cardio vascular symptoms and digestive system symptoms [26].

2.3.6 Ontologies Related Drugs.

The Drug Ontology (DRON) consists of classes and properties which denotes ingredients, mechanisms of actions and comparative effectiveness information of various drugs [27]. Chem2Bio2RDF [28] is an ontology constructed for drug identification, drug target identification and adverse drug reactions. Food Interactions with Drugs Evidence Ontology (FIDEO) is an ontology for annotating and retrieving of scientific articles about drug-food interactions [29]. The Drug-Drug Interactions Ontology (DINTO) is an ontology constructed for semantically organizing information about drug-drug interactions [30].

2.4 Querying Ontologies

SPARQL is a RDF based query language used for reading, updating and deleting RDF data from a linked database.

3. SEMANTIC WEB IN THE AREA OF HEALTH CARE.

Semantic web can be used in many areas in healthcare domain like self-management, disease diagnosis, dietary suggestion system and Medication suggestion system.

3.1 Chronic diseases management using SW

Chronic disease self-management enables patients take personalized management actions for their diseases based on conditions of their diseases. Self-management requires decisions to be made regarding diet, medications, exercises. The authors in [31] have constructed one knowledge base containing information from various domains like disease, nutrition, exercise and medical guidelines. The authors have constructed knowledge base by making complicated relationships in these domains. Authors have used Protégé for construing knowledge base and to construct personalized management goals authors have considered Electronic Medical Records in SQL format and used JENA framework for transforming records to RDF format. In [32] authors have developed ontology-based management system for tropical diseases. Tropical diseases are infectious diseases caused due to bacteria, viruses and parasites which reproduce themselves in certain environments. Authors have used protégé 4.1 for constructing the ontology. In [33], authors have developed a multi agent approach for chronic disease health care monitoring and management using predefined ontology and JADE framework. Authors have used SWRL and pellet reasoner for inferring new knowledge. In [34] authors have constructed a Clinical Decision Support System (CDS) for diabetic patients and named it as Trec-Diabetes by using semantic web technologies like ontologies and inference engines. Authors have aimed to detect critical situations by clinicians-based data provided by patients and prescribe decisions.

3.2 Decision Support System

Wrong Decision taken by healthcare officials may cause even deaths. According to a Survey of WHO three million deaths are occulting annually due to unsafe care or medical mistakes [35]. Decision support systems can help to take decisions in various medical functions like diagnosis, medication, diet suggestion etc. Which minimizes the loss of unsafe care.

In [36], Authors have developed a decision support system for patients of Alzheimer’s Disease. Authors have Constructed an ontology called as ADDO using protégé tool and they used HermiT reasoner for reasoning and SPARQL for querying the knowledge base. In [37], authors have designed Decision Support System for diagnosing Comorbidities. Comorbid diseases are diseases which are related to each other or coexist at the same time like Chronic Heart Failure may coexist in patient with Atrial Fibrillation, Diabetes and chronic lung diseases [38]. Treatment of one disease may affect other disease. The decision regarding which disease has to diagnosed first needs to be done carefully. The authors in [39] has constructed Decision Support System using Clinical Pathway Guidelines (CPG) which were computerised and converted ontologies by considering the common classes. In [40], Authors have developed personalized ontology-based decision support system for the patients of Spondylarthritis (SpA) by considering many characteristics like medical assessment, social and psychological metrics and behavioural analysis. Authors have used OWL and SWRl for constructing ontology and for inferring decisions.

3.3 Dietary suggestion

Poor nutrition can cause obesity which results into other diseases like diabetes, heart diseases, bone and joint related diseases [41]. Diet management helps the patients to recover the diseases quickly. This is the reason of prescribing diets to patients. Even foods consumed can also show their impact of medications needs to be taken. It is better if patients know how much carbohydrates and calories particular food have and its side effects. Health factors like cholesterol levels and blood pressure will depend on die which causes heart diseases [42]. For some chronic diseases diet is considered as part of treatment [43]. We can use semantic web technologies to prescribe diets to be taken by patients. Many authors have worked in this area.

Authors in [44] have constructed a web application for nutrition consultation for type 1 diabetic patients. Type 1 diabetics can be seen in children and younger adults who body does not provide enough insulin. By using insulin injections, the sugar levels can be maintained effectively. But the amount of consumed insulin can be minimized if proper levels of carbohydrates and calories are consumed. The authors have constructed ontology using OWL. In [45], authors have developed an ontology based dietary consultation system for chronic diet related diseases. The authors have also used IoT sensors for sensing food intake and constructed ontologies like personal profile ontology

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and food ontology using OWL. Authors in [46], constructed a dietary consultation system for chronic kidney disease (CKD) patients. The patients with CKD suffer for deterioration of kidney functions and not able to receive certain nutrition. Hence, they require strict diet control. Authors have constructed a knowledge base from various sources of information using OWL and SWRL. In [47], authors have developed an integrated approach of diet and exercise suggestion for type 2 diabetics using OWL and SWRL. In [48], authors have designed Ontology of Dietary Recommendations (ODR) for recommending diets for patients of obesity. Authors of have developed a food menu recommender system for patients of coronary heart diseases. Authors have constructed an ontology with information of nutrition data and food intake and using SWRL rules on ontology recommended food information is retrieved.

3.4 Disease diagnosis

Diagnosis is the process of identifying the disease and providing treatment for the disease. Generally, patients will consider the symptoms and consults the health care officials. In our previous work we have identified that disease diagnosis is the less studied area[49]. Hence more focus has to be given in this area. Collection of symptoms which denotes presence of a disease is called as syndrome [50]. Some symptoms can be simple which are negligible and other symptoms can cause a serious health issue which needs to be handled. But Symptoms appears early are not accurate and symptoms may change, or become worse with time. There are some online platforms known as symptom checkers which takes the input symptoms and gives a possible list of diagnoses. But they are not effective because patients may use different abbreviations or synonyms of symptoms which symptom checker may not understands [51]. Symptoms can also differ from patient to patient or they may change as climate changes [52]. Most of the available symptom checkers are not considering personal health characteristics. They will just return pair wise relations between user entered symptoms and possible diseases from the knowledge that is stored in the backend. And same symptoms even may appear in more than one disease like shown in above linked data where two chronic diseases like diabetes and heart stroke can have same symptoms like tingling hands or feet and blurry vision.

Hence symptoms only are not enough to efficiently diagnose a disease we need to consider other characteristics like personal health information, Patients and Parental medical history, Patient life style, results of tests and disease demographics along with symptoms. And Experience and Expertise of health care officials may also show impact on diagnoses of disease. According to a report of WHO and India today approximately 2.6 million people around the globe are dying due to medical mistakes and 138 million peoples harmed due to doctors' errors [53,54]. In the next section we discussed effects of said parameters which are essential for disease diagnosis.

3.4.1 Personal Health Information

This is the information related to a patient which includes health information like ongoing medical treatments, drugs being consumed, allergies of a patient, and information of health care professional who are treating the patients, and other information like name of the patient, date of birth, age, blood group, care givers contact information. This type of information helps the health care professionals to better diagnosis by knowing disease with ongoing treatments, and provide drugs which will not interact with other drugs and allergies. Initially personal health information was paper based. Due to many drawbacks like paper documents can not be carried out every where and can be lost or damaged. To overcome these drawbacks computer based Personal Health Records (PHR) came into existence which can be carried out and accessed any time any where and can be used for self-management [55]. Various online personal health records platforms exist like Microsoft Health Vault, and Google Health which connects to databases of hospitals and enables the patients to import their records when needed. Semantic web technologies can also be used for constructing and using PHR, because ontologies provide a best way to store and share information [56]. Authors in [57], constructed a tool Triax, using semantic web technologies which fetch the patient records from PHR which can be used in clinical trials. PHR data can also be used for clinical research, MMIC-IV is constructed for this purpose [58]. Authors of [59], constructed a personal health recommender system using PHR data, and crowd sources data like symptoms, diseases, treatments, practices and domain ontologies.

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3.4.2 Parental History

Parental history plays an important role in disease diagnosis because some diseases known as hereditary diseases are passed down to children from either of their parents. If the siblings are having these diseases another sibling who is healthy can also have the disease in future. Hence it is important to know parental history. These diseases are because of genetic disorders in family tree. The goal of study of human genetics is to identify change in DNA sequence [60]. Meny diseases like diabetes, heart disease, some types of cancers, dementia, Huntington's disease and cystic fibrosis. Most of this disease can't be cured completely but can be controlled. The authors in [61] has constructed an ontology-based model named as DisGeNET-RDF which provides associations between genes and diseases.

3.4.3 Medical Records

Medical records provide history of patients. They are maintained by health care officials. Computerized medical records are known as electronic health records (EHR). The differences between PHR and EHR are PHR are maintained by patients with limited information. EHRs are handled by health care organizations. They include information of a patient like name, age, gender, previous diagnoses, pathological test reports, medications, dosage consumed, any allergies to drugs, previous surgeries. If these medical records are maintained correctly it helps in correctness of treatment [62]. Good record maintenance of health care professionals proves that they have provided good treatment. Whereas if care is neglected records will not contain enough information [63]. Medical records can fully be accessed by health care professionals and can be partially accessed by patients and care givers. Medical records need to be summoned in court for cases like road accidents, medical negligence and insurance claims. Another record maintaining mechanisms is Electronic medical records (EMR), which have limited information and more privacy than EHR. EHR data is used for decision making and information of EMR is used in diagnosis. Adoption of EHR and EMR leads to adherences of medical guidelines which results to decrease in medical errors and adverse drug reactions [64]. These medical records may contain structured, semi structured and unstructured data, semantic web technologies can be used to link these data to form a knowledge base and access the records efficiently using SPARQL [65]. To link vast types of heterogeneous data and to handle complex relationships a knowledge graph is proposed in [66] using MIMIC III Dataset and semantic web technologies.

3.4.4 Disease Demographics

Some diseases can be classified into groups using demographics like race, ethnicity, geographical regions, social economic status, age and gender. Mediterranean race is in north Africa and South asia, the people of this race mostly have sickle-cell disease. Ethnicity is the grouping of population based on culture, ancestry, religion and history. People with African ethnicity sickle cell disease, Ashkenazi Jewish mostly have Gaucher disease People with Hispanic ethnicity mostly have Cystic Fibrosis [67]. The male people are mostly affected of skin infections than women. Women are affected by psychosomatic problems and allergic diseases than men [68]. Men may face heart diseases a decade earlier on an average than women, 1.5 times more than women men will have Parkinson's disease which degrades brain health [69]. Many diseases are based on ages. The older people will have diseases like hearing loss, cataract, high blood pressure, diabetes, cardio vascular diseases, many types of cancers and pulmonary diseases than younger ones [70]. Younger people with obesity and high blood pressure may get heart diseases earlier [71]. The children and young people with high obesity may get type 2 diabetes [72]. People with Low Socioeconomics are having high risks of facing and dying from cardio vascular diseases because of their low income, poor education and lack of resources [73].

In the following section we have studied works of various authors in the field of disease diagnosis using semantic web technologies.

3.5 Literature review for disease diagnosis

In [74], authors have constructed a disease diagnosis model using predefined ontologies DO and SYMP for diseases and symptoms respectively. Authors also used web based personal health services and created an Human Disease Diagnosis Ontology (HDDO). For abbreviations and codes of diseases and symptoms knowledge from Bio Portal is

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used and knowledge from PubMed is used to find term cooccurrences between diseases and personal health attributes. In authors have developed a differential diagnosis model by using predefined ontologies like DO and SYMP, which are combined to form Disease-Symptom ontology (DSO) and Patient Ontology (PO). In [75] authors have developed a diagnosis model for predicting diseases by constructing ontology by combining diseases, symptoms and patient context. In [76] authors have constructed a diagnosis model by constructing Disease-Symptom ontology (DSO) using DO and SYMP ontologies. In [77], [78], [79] and [80] authors have constructed a diagnosis model by constructing an ontology for a limited set of symptoms and disease. In [81] authors have developed health service recommendation model by using patient context and by constructing an ontology eHeaRSS (eHealth Recommendation Service System) ontology based on four separate ontologies like Symptoms, Disease, Doctor and Department. Authors have collected patient context information from crowd sourcing like Google health, Health Vault and Dossia. In [82], Authors have reviewed works of other authors who have used semantic web technologies for preventing, diagnosis and treatment of diabetic mellitus. Authors in [83], have developed an ontology-based diagnosis model for chronic breast cancer. Authors in [84] has developed an ontology-based model for detecting disease similarity. Author in [85] designed ontology-based diagnosis model for chronic liver disease by using UMLS as data source. In [86] authors have developed ontology-based Bayesian network for inferring disease information by giving symptoms, age, gender, life style, personal and parental history as input. In [87] authors have constructed syndrome ontology model for diagnosing diseases. In [88], a diagnosis model for vector borne diseases, which occurs because of transmission of vectors from bites of infected parasites like bacteria, insects and mosquitos. In [89] a diagnosis model for chronic obstructive pulmonary disease was built. The authors in [90] have developed an ontology-based diagnosis model for predicting chronic kidney diseases. In [91], authors have developed an ontology-based diagnosis model for diagnosing thyroid disease. Authors in [92] have designed ontology-based diagnosis system for myocardial disease. This detailed review of disease diagnosis is given in below table.

Table 1: Literature review for diseases diagnosis using semantic web technologies

Ref	Ontologies Used	Services & Technologies Used	Ontologies Constructed	Parameters for diagnosis considered	Validation
[19]	DO, SYMP	PHR, PubMed, Bio Portal, TrOWL version 1.5, Jena Ontology API, Pro-tégé ontology editor.	HDDO	Symptoms, Medical records, Personal Health Information and Disease Demographics	Validated on simulated patient vignettes.
[20]	DO, SYMP	Clinical Pathways, OWL	DSO, PO	Symptoms, lab tests, Patient Health Information	Validated Using NIS Database and Classification and Association Algorithms for predicting Disease
[21]	SNOMED CT, UMLS	OWL, HermiT static DL reasoner, SPARQL	Ontology combining Diseases, Symptoms and Patient context	Symptoms, Demographics and Patient Context	Not Validated
[22]	DO,SYMP	Protégé	DSO	Only symptoms are considered	Not validated.
[23]	Not used	Protégé, Apache Jena, SPARQL	Ontology of a set of diseases and symptoms.	symptoms	Not validated.
[24]	Not used	Crowd Sourcing for Patient Context	eHeaRSS ontology constructed from Disease, Symptom,	Symptoms and Patient Context	Validated by comparing with DB Based health

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		information	Doctor and Department ontology		Systems
[25]	Not used	OWL, SWRL, JESS (Java Expert System Shell)	Disease-Symptoms Ontology	Symptoms and Patient Context	Validated using Select and Test Medical Reasoning Algorithm on 10 patient records from University Teaching Hospital.
[26]	Not used	OWL, SWRL,PubMed, Bio Portal	Person Disease Diagnosis Ontology	Symptoms and Personal Health Characteristics	Standard Deviation method is used to distribute diseases into group
[27]	Not Used	OWL, SPARQL	Disease ontology	symptoms, parental and personal history and patient context.	Validated by calculating probability of diseases using Bayesian network by considering 100 synthesized patient's data.
[28]	Disease Ontology	OWL, SPARQL, Apache Tomcat 7.0, Protégé 4.0.1	Syndrome Ontology	Symptoms	Not Validated
[29]	PCD ontology, BFO ontology	OWL, SWRL, NLP, OCR	VBD ontology	Patient Health Information	Not Validated

3.6 Research Gaps

From the studied literature it can be analysed that successful diagnosis should consider all parameters like symptoms, personal health information, medical records, parental history, disease demographics

- It is observed that many authors are considering symptoms as in [19],[22],[23],[24],[25],[28],[30],[32],[34] and [35].
- Personal health information is considered in [19],[23],[28],[30],[32],[34] and [36].
- Medical records or patient personal history was considered in [19] and [34].
- Parental history was considered only in [34].
- Disease demographics are considered by authors of [19] and [23].

The following graph in fig.2 shows this observation and count of articles considered above parameters by considering above 11 articles published in area of disease diagnosis.

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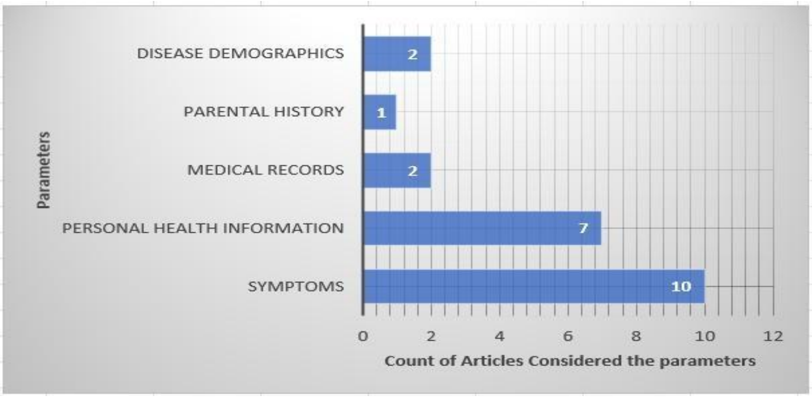

Fig 2: Count of Articles considering the parameters required for diagnosis

4. PROPOSED METHODOLOGY FOR EFFICIENT DISEASE DIAGNOSIS

We have constructed a model for efficiently diagnosing two chronic diseases namely heart stroke and diabetes. These two diseases have many symptoms which are initially not clear and may change as disease becomes severe. The symptoms of heart stroke are trouble in speaking, numb or tingling hands and feet, severe head ache and trouble in seeking in one or both eyes (blurry vision). The symptoms of diabetes are based on type of diabetes. There are many types of diabetes exists and the most common one is type 2 diabetes. The common symptoms of diabetes include frequent urination, dry mouth, blurry vision, numb or tingling hands and feet and sudden weight loss. Some symptoms like having blurry vision and numb or tingling hands and feet can also appear in more than one disease. Patients visits heath care officials as the observes the symptoms. Hence there are common symptoms health care officials cannot give their decisions based on symptoms alone. Hence for effective diagnosis they consider patient's health information, previous medical records, parental history, patient life style and sometimes disease demographics. Most of this information health care officials obtain from the patient or from his care givers. Medical records data can be acquired from Electronic Health Records (EHR) services and Personal Health information can be obtained from electronic Personal Health Records (PHR) services. Health care officials start the diagnostic process by considering symptoms and by constructing possible hypothesis of diseases. The list of diseases will be narrowed down as the other information like medical records, parental history and disease demographics are applied. This process is known as differential diagnosis. Finally, the list remains with one are disease. To confirm the disease from the list required pathological tests will be prescribed.

Our constructed model follows this differential diagnosis process. Due to privacy of patient's information, it is difficult to acquire dataset for validation. We have validated our diagnosis model by constructing a synthesized data set of 20 patient vignettes for two diseases i.e. heart stroke and diabetes by considering different values for symptoms, medical records, parental history and demographics. We constructed our dataset by creating a questionnaire and collecting responses online regarding factors affecting these diseases and recording the responses. We validated our model by comparing the results with Three AI based symptom checkers namely cody.md, docus.ai and roboclinic.ai.

4.1 Working methodology of Constructed Model for disease diagnosis.

The working model of our proposed model can be shown as using a flow diagram flow diagram as in Fig.4. We start by considering common symptoms of heart stroke and diabetes. And to minimize the list of possible disease we apply sequentially disease demographics, parental history, personal health information and then medical records. Finally, we reach to a concluded disease. To confirm the disease the necessary pathological test has to be taken. If the disease is not identified by our model it is needed to undergo many tests for confirming the disease.

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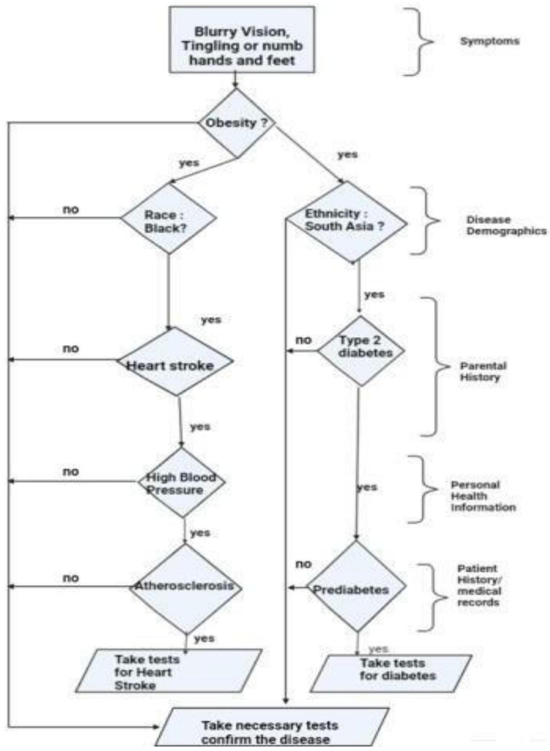

Fig 3: Working method of constructed disease diagnosis model

4.2. Ontology for Proposed Disease Diagnosis Model

We have constructed one ontology with classes for Chronic Diseases with two instances representing heart stroke and diabetes, and classes for symptoms and instances including the symptoms of these two diseases. We also constructed classes and instances for parental history, medical records, patient's personal health information and demographic data of these two diseases. The ontology is constructed using an open-source triple store and graph data base known as blaze graph which provides SPARQL interface to query the constructed ontology. The diagrammatic representation of constructed ontology is shown in following figure fig.3. This diagram is constructed by using rdf_grapher, a tool available online which is used for visualizing RDF data.

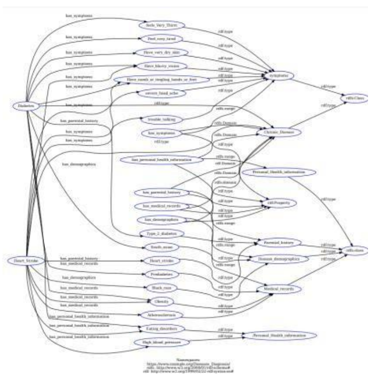

Fig 4: Constructed ontology for disease diagnosis

The working of our model can be shown by posing SPARQL queries to the constructed ontology as follows. We have created a dataset of 20 patient vignettes and executed all queries and demonstrated the first five patient vignettes and their possible diseases by posing SPARQL queries on the dataset.

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Query for Patient Vignette-1

```
PREFIX : <https://www.example.org/Diseases_Diagnosis/>
SELECT ?Disease
WHERE{
  ?Disease :has_symptoms :Have_blurry_vision,:Frequent_urination;
           :has_medical_records :Obesity;
           :has_personal_health_information :High_blood_pressure,:High_cholestrol;
           :has_parental_history :Type_2_diabetes;
           :has_demographics :South_asian.
```

Result

Disease
<https://www.example.org/Diseases_Diagnosis/Diabetes>

Query for Patient Vignette-2

```
PREFIX : <https://www.example.org/Diseases_Diagnosis/>
SELECT ?Disease
WHERE{
  ?Disease :has_symptoms :Have_numb_or_tingling_hands_or_feet;
           :has_medical_records :Prediabetes;
           :has_personal_health_information :High_cholestrol.
           }
```

Result

Disease
<https://www.example.org/Diseases_Diagnosis/Diabetes>

Query for Patient Vignette-3

```
PREFIX : <https://www.example.org/Diseases_Diagnosis/>
SELECT ?Disease
WHERE{
  ?Disease :has_symptoms  :trouble_talking,:severe_head_ache;
           :has_personal_health_information :High_blood_pressure;
           :has_parental_history :Heart_stroke;
           }
```

Result

Disease
<https://www.example.org/Diseases_Diagnosis/Heart_Stroke>

Query for Patient Vignette-4

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```
PREFIX : <https://www.example.org/Diseases_Diagnosis/>
SELECT ?Disease
WHERE{
  ?Disease :has_symptoms  :Have_blurry_vision;
           :has_medical_records :Atherosclerosis;
           :has_personal_health_information :Eating_disorders;
           :has_demographics :Black_race.
           }
```

Results

Disease
<https://www.example.org/Diseases_Diagnosis/Heart_Stroke>

Query for Patient Vignette-5

```
PREFIX : <https://www.example.org/Diseases_Diagnosis/>
SELECT ?Disease
WHERE{
  ?Disease :has_symptoms  :Have_numb_or_tingling_hands_or_feet;
           :has_medical_records :Obesity;
           :has_parental_history :Heart_stroke.
          }
```

Results

Disease
<https://www.example.org/Diseases_Diagnosis/Heart_Stroke>

If the disease are identified then pathological tests of particular disease needs to be conducted for confirming the disease otherwise tests of both diseases needs to be considered for confirming the disease. The validation of our constructed model is shown in below section.

5. VALIDATION OF CONSTRUCTED MODEL

Our Constructed model is validated on three online AI based symptom checker known as “CodyMD, Docus.ai, Roboclinic.ai and which works like a chat boat . CodyMD considers symptoms and medical records to predict a disease and returns three possible diseases with their probability percentage.Docus.ai works by considering symptoms, medical records, parental records, demographics, personal health information like gender, age, smoking, drinking and food habits and returns five possible diseases with their possible priorities. Roboclinic.ai works by considering symptoms and medical records and returns list of possible diseases without possible probabilities. Following table demonstrates the validation of our constructed model by considering twenty patient vignettes and comparing their results with three online AI based disease prediction systems.

Table 2: Validation of constructed model

Patient ID	Diseases Predicted using our model	Cody MD		DOCUS		Roboclinic	
		Diseases Predicted	Probability	Diseases Predicted	Probability	Diseases Predicted	Probability (Not Given)
PID_1	Diabetes	Diabetes	70%	Diabetes	60%	Diabetes	N.A
PID_2	Diabetes	Diabetes	60%	Diabetes	65%	Diabetes	N.A

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PID_3	Stroke	Stroke	30%	Stroke	55%	Stroke	N.A
PID_4	Stroke	Diabetes	40%	Refractive Errors	40%	Refractive Errors	N.A
PID_5	Stroke	Diabetes	40%	Stroke	35%	Diabetes	N.A
PID_6	Diabetes	Diabetes	70%	Diabetes	60%	Diabetes	N.A
PID_7	Diabetes	Diabetes	40%	Diabetes	70%	Diabetes	N.A
PID_8	Stroke	Stroke	40%	Stroke	60%	Stroke	N.A
PID_9	Diabetes	Diabetes	70%	Diabetes	60%	Diabetes	N.A
PID_10	Stroke	Migraine	60%	Migraine	30%	Migraine	N.A
PID_11	Stroke	Stroke	30%	Stroke	35%	Stroke	N.A
PID_12	Diabetes	Diabetes	70%	Diabetes	70%	Diabetes	N.A
PID_13	Diabetes	Diabetes	40%	Diabetes	40%	Diabetes	N.A
PID_14	Stroke	Stroke	25%	Stroke	45%	Stroke	N.A
PID_15	Stroke	Stroke	30%	Stroke	65%	Stroke	N.A
PID_16	Diabetes	Diabetes	70%	Diabetes	60%	Diabetes	N.A
PID_17	Diabetes	Diabetes	30%	Diabetes	55%	Diabetes	N.A
PID_18	Stroke	Multiple Sclerosis	40%	Stroke	40%	Stroke	N.A
PID_19	Stroke	Stroke	25%	Stroke	5%	Diabetes	N.A
PID_20	Diabetes	Diabetes	70%	Diabetes	70%	Diabetes	N.A

The following table shows the validation percentage of our proposed model on Three AI based diseases prediction methods.

Table 3: Comparison of tools used for successful validation

S.No	Application used for Validation	Percentage of successful validation
1	CodyMD.ai	80%
2	DOCUS.ai	90%
3	Roboclinic.ai	80%

Our model was best validated on Docus.ai because it is using all the parameters to predict the disease which we are considering in diagnosing the model than CodyMD.ai. The following Graph shows the performance of these models in accordance to our constructed model.

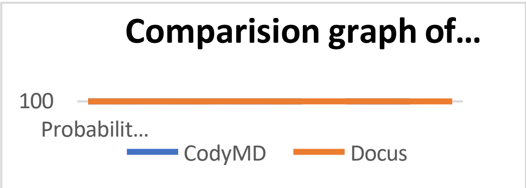

Fig 5: Comparison graph of CodyMD and DOCUS

6. CONCLUSION AND FUTURE WORK

Detecting and diagnosing a disease play very important role in health care domain. Diseases needs to be diagnosed carefully for providing cure. Many people each year dies due to medical mistakes. For efficient diagnosis many parameters need to be considered. In our work we have shown the importance of parameters like parental history, medical records, disease demographics and personal health information along with symptoms for efficient

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diagnosis of a disease. In our future work we try to expand our model by considering more diseases and we work on to linking ontologies with user interface such that it will be for health care professionals and other stake holders of heath care domain can interact and use ontologies in the field of disease diagnosis.

DATA AVAILABILITY STATEMENT

We have validated our constructed model by constructing 20 patient vignettes for two diseases namely diabetes and heart stroke. We have considered various factors which can causes these diseases like symptoms, medical history, personal health records, parental history and diseases demographics. We gathered the data related to these diseases and their causing factors using questionnaire method.

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