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A New Semantic Resource Description Framework for Medical Diagnosis based on Gene Ontology

Anjna Jayant Deen, Manasi Gyanchandani

Abstract*: Semantic-Based medical informatics system is an interdisciplinary deflect of bioinformatics research, where a dealing to knowledge discovery of proper diagnosis of disease leads to more attention about health control before health complications. To do this work is in the aspects of various ways like preserving and accessing patient's medical records for further information retrieval from pre-recorded datasets. Now some research has mostly used to medical metrics method combined with statistical information and social network analysis citation approach to analyze. Due to this limited citation information force to develop a new dimension of the semantic web-based medical diagnosis system. In this study the approach of implementing autonomic gene computing in medical informatics, in this framework a new semantic resource description network for medical diagnostics based on Gene Ontology, this way the diagnosis context capture its levels of symptoms. Every patient has their unique symptoms which are kept and fetched by semantic web applications, In this framework we used gene-based symptom indication for disease diagnosis, and merge with drug prescription leads to more suitable results that can be useful for physicians and medical practitioner, as to achieve efficient solutions in the new direction to make life saving analysis from the database. Being a very effective and enhanced web application tool can be applied in the medical field to make it useful for the society. The web interface for drugs prescription accepts the diseases and symptoms from the domain updated in the database and successfully displays prescribed drugs for each disease.

Keywords* : *RDF, Gene Ontology, Semantic Web Application, Medical Informatics,* Diseases and *Drugs.

I. INTRODUCTION

semantic web and P2P uses information search on web based application, the authors [1] suggested cayNET web based searching methods provides fast identical search and enable a peer connect many semantic clusters to enhance file browsing function. P2P and Semantic Web is introduced another novel methods [3].

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***** Correspondence Author
Anjna Jayant Deen*, Department of Computer Science and Engineering Maulana Azad National Institute of Technology, Bhopal, (M.P), India.
Manasi Gyanchandani, Department of Computer Science and Engineering Maulana Azad National Institute of Technology, Bhopal, (M.P), India.

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In order to this many web application designed for semantic search bounded due to limitation of WWW, because most of the information structured based on general datasets , there are no support for search automated and efficient way to finds knowledge discovery[4] , were some more web limitation based on keyword content based search. The role of automated EMR (electronic medical record) system in an efficient way to exhibited in this [2] context. Many informatics in medical learning methods are not proper utilizing the technology in computing systems, problem they face due to network size rapidly increases requires more site of skilled web. A semantic data model based on semantic link network (SLN) is for web resources management [5]. Fuzzy based cloud computing system can enhanced the medical diagnosis for disease, being a domain very effective[6]. The knowledge discovery of large medical transcript databases using classifications for many scientific aspects can bring out prediction of diseases by the physicians, software based prediction model helps to both patients and doctors as many lab and doctor maintains their patients records for further analysis, in this way handling medical data achieve for high performance of web based model. The automated medical machine (AMM) work like as an ATM. AMM transaction based machine provides medicines to user. These machine stores information of registered user, medicine record, uses of essential drugs, consulting doctor list etc. Connection made online call basis and doctor prescribes medicines based on symptoms [7]. Information about medical control sometimes panic due to suddenly unwanted conditions and attacks on healthcare facilities. This types of situation can be lead the health informatics on web and cloud based hoarding of medical data, data control flows, and web based searching application site governance. While distinct healthcare systems opt for cloud-based multi observation and suggest solution, it is a amount of time when fog computing environment are formed [8].The bioinformatics managed large size of medical database this resources are configured with several server centre with suitable software, so they can distributed and uploaded manages by multiple resources in around the globe.

II. MATERIALS AND METHOD

A. *Data sources*

a).Disease: 4,300 disorders and disease recorded as RDF data on D2R Server [10], as well as the genes causing some of them. These diseases are classified by categories.

b). Drug: The drug is a combination of chemical, pharmacological and pharmaceutical data used to target sequence, structure, and pathway information for docking complete drug [14].

c). Diseases under control: diseases that have at least possible drugs. With a recognition of a disease "under control", an involved patient user of our application could be comforted with the scientific condition towards his/her health guarantee.

d). Diseases under investigation: diseases that have at most 2 possible drugs. With a recognition of a disease "under investigation", an involved doctor user of our application could be reminded being cautious in deciding treatment programs.

e). Treatable disease: diseases that have at least one possible drug.

f). Interacting drugs: drugs that are interacting with the current drug.

g). Unsafe drugs: illicit drugs and withdrawn drugs.

B. Ontology

Basically ontology means a inter relationship between different interpretation or ideas within a groups or a specific domain, computer ontology is bounding relationship in which refers those ideas. Study of ontology based on the actual entities within a specific group domain and sometimes it can be used to identify the domain itself. Ontology is a exchange a framework for defining the specific class domain that dwell of a various set of approach, essential aspect and relationships. The meaning of certain biological data information is explored based on real and actual information models, which are used for various modeling applications and structuring data. The basic approach used for constructing such models includes entity, activity, element and purpose. Were, the information based conceptual models define the semantic terms and mechanisms for organizing the biological information by making a set of assumptions about the actual applications to be formed.

C. *Resource Description Framework (RDF)*

RDF is part of WWW for design metadata model. Necessary information and metadata encoding on the semantic web standard are known as Resource Description Framework (RDF).In RDF uses small information segments in infers meaning. RDF transferring all kinds of information on metadata and provides software application based on XML syntax to exchange data and information with different locations of URI/URL provided. Semantic web resources evaluated by automated software which can use to distributes, store, exchange information on www for knowledge management. So that HTML designed principle of information and content presented through a Web Browser.

The RDF first used in 1996 replaced set of six specifications in 2004 [9]. This RDF series was, documents, Primer, Concepts and Abstract Syntax, XML Syntax, Semantics, Schema, and Test Cases. RDF designed so that data is presented and possibly understood by machines. The basic building block or atom of information in RDF is called a Triple or Statement and represents a fact about a Resource. The triple has the following form: Subject, Predicate, and Object. RDF popular applications is a semantic web, and producers and consumers of RDF statements must agree on the semantics of resource identifiers as shown in fig 1. However, broad agreement based on URI returns and response when used in an HTTP GET request and the resource that it succeeds in the internet accessing.

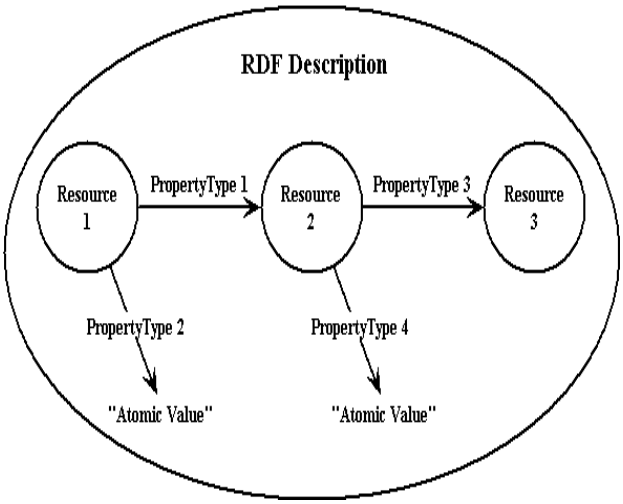

Figure 1. RDF Model

D. *PROTÉGÉ*

Knowledge management based Protégé is a free system, Protégé is a open source ontology editor. Ontology defines using a graphic user interface, which will be provided by Protégé. Analysis of ontology based on information model to infer new classifiers for validation. Projects suggest like eclipse is a Protégé framework for which various other plug-ins. Java application based Protégé more than three million user heavily uses Swing to create the user interface[23], [11],[12]. Stanford University is developed a Protégé and is made available under the BSD 2-clause license. Earlier versions of the tool were developed in the University of Manchester with collaboration. Protégé is a java based virtual machine for ontology editor. An ontology development environment for web Protégé that makes it easy to create, upload, modify and share ontologies for collaborative viewing and editing. Web ontology language fully supported by Protégé. the Protégé user interface configuration creates the perfect environment for all kinds of developers. RDF/XML, TURTLE, OWL/XML, OBO and other formats available for ontology upload and download. All collaboration features contributes information sharing and permission, threaded notes and discussion, watches and email notifications[19]. The modeling ontologies platform is supported via Protégé in two ways, that is Protégé-Frames based and Protégé-OWL editors based. Protégé ontologies also supported various outsides exported formats like ,including RDF, RDFS, OWL, and XML Schema.

E. *STARDOG*

Stardog is a faster and easier model. It unifies medical informatics data that is on based semantic graphs, schema alignment, data modeling, and profound reasoning. With inference and data virtualization using smart graph database technology, doing query and searches. these techniques have been straightforward and effective. Stardog is an enterprise data system which can connect to any database virtually, easily incorporate new data sources, slice and dice data for multiple applications. The Stardog new version supports the RDF graph data model, SPARQL query language, property graph model, as well as Gremlin graph traversal language.

Stardog is a commercial RDF database use insanely fast SPARQL query, transactions,

and world-class
OWL reasoning support. Stardog having high availability clusters that ensure any failure will not affect the applications that depend on a single machine. Using a clustered version can immediately resolve failures by automatically creating multiple copies of the service to make sure everything keeps running still. To ensure stability, Stardog uses Apache Zookeeper, which has been tried and tested for this type of infrastructure. It promotes nodes to communicate effectively and be able to update or create new connections later on. Compared to the relational database, a graph database can handle more relationships or contacts that a single enterprise may have. Stardog's graph database makes it possible to work on multiple nodes at a given time and make sure that all information is safe and working on with speed.

It is therefore highly recommended to use the begin() and commit() and rollback() methods to group operations into transactions unless required by in our gene-based application. These connections keep a local cache of changes which unpushed until they exceed a threshold, or an action, such as a query or a commit, requires them to be pushed to the server[10],[11],[12],[13]. These minimize the overhead of calling add or remove in a tight loop with small payloads and then committing the entire thing, and those updates would then to be pushedup in a few chunks as possible. However, max query time has not supported by Stardog. Timeouts cannot be controlled on a per-query basis, they are global server options and each of the specific prepare query methods and well handle by HTTP repository.

F. *SPARQL*

SPARQL is "Sparkle" : SPARQL protocol and RDF query language, a semantic query language for databases in resource description framework (RDF) format that can retrieve and manipulate stored data. SPARQL allows mixed patterns for a query. SPARQL query tools connect automatically for query endpoint and support to translate multi programming language exist tools to other query languages. SPARQL users allow writing queries against more specifically in loosely "key-value" data or, data that follow the RDF specification of the W3C, as shown in fig. (3). Thus, the complete database information is a set of triples, that is subject, predicate and object. There are some related SQL databases usage the term query "document-key-value" in the term. In SQL relational database terms, RDF data can also be considered a table with columns based on level 1. The subject column, level 2. The predicate column and level 3.The object column. In RDF, there are instead based representation to separates predicated value, in object sharing in the same purposes, often the same unique key, with the predicate being analogous to the column name and the object the actual data[24]. SQL database entity allows data object to placed in multiple columns and spread to more than one table can also to be recognized by a unique key. SPARQL provides a specific graph traversal syntax for data, as shown in fig. (2), a graph query-based ontology schema information externally offered, to allow JOIN, SORT, and AGGREGATE for joining of different datasets.

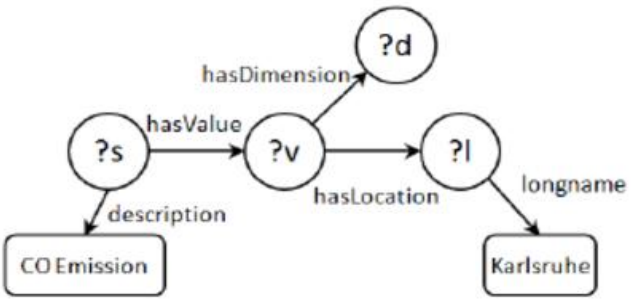

Figure 2.Query Graph

```
Select ?s,?v,?d,?l
WHERE {
  ?s ns:description "CO Emission" .
  ?s ns:hasValue ?v .
  ?v ns:hasDimension ?d .
  ?v ns:hasLocation ?l .
  ?l ns:longname "Karlsruhe"
}
```

Figure 3.SPARQL Query

G. *Gene*

All biological systems are composed of the same types of molecules and Similar cellular level. Twenty amino acids are found in nature. Regulatory codons four bases can form 64 different codons. A DNA sequence that is needed to encode an amino acid sequence of a protein. Composed of exons, introns and different control elements. Exoncis is protein coding sequence, and intron is intervening sequence. The Human genome is having a different type of sequence chain that makes up the total DNA of a human cell. The human genome consists of 3 billion base pairs about 22000 genes, and only 2 % of the DNA encodes proteins. Genes include exons and introns, besides coding areas also additional sequences are found 50 % repeated sequences is known as junk DNA, these DNA mutations are finding significance in the development of diseases inherited diseases and inherited diseases are caused by mutations passed from a parent gene. Many factor causes of illness based on, the disease is caused by one mutation in one gene is a monogenic diseases, the disease is caused by the interaction of different mutations and environmental factors is a multifactiorial diseases and presence or absence of the phenotype depends on the genotype at a single locus. ia a mendelian inheritance.

The genotype is the genetic information written in DNA; However, gene-based medical diagnosis system helps to prevent various diseases and finds the solution of a genetic disorder. Housekeeping genes have studied by many researchers and some interesting observations have been reported [20]. Every person has two copies of each gene, one inherited from each parent. Gene, with minor differences in their sequence of DNA bases, makes to each human unique physical features.

III. PROPOSED MODEL ARCHITECTURE AND METHODOLOGY

A. Methodology Aim

The objective of this study is to build a semantic web application that takes partial or full name of disease as input and gives possible drugs and related genes as output for the disease provided and also mark interactive drugs among them, reminding healthcare providers to avoid drug conflicts.This is to serve multiple purposes:

- To reduce strain on healthcare providers.
- To minimize or eliminate the possibility of human error.
- To provide patients a better understanding of possible drug conflicts that might arise when medicating themselves of medicating themselves.
- To apply the principles of computer science to a completely different domain, gaining new insights that can be applied to other cross domain endeavours as shown in fig.(4).

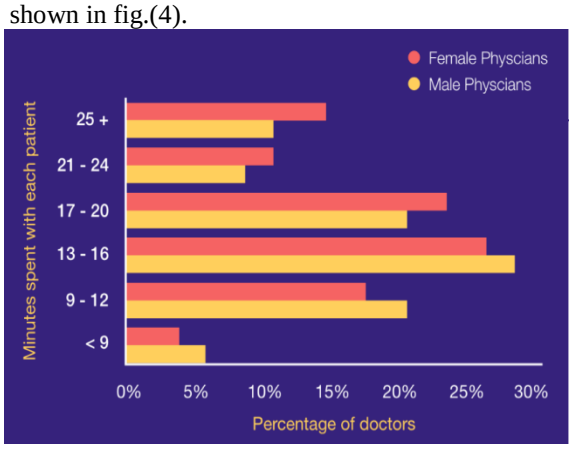

Figure 4. Time Doctors spend with their Patients

B. About The Proposed Model

The application was developed with Data-Oriented Design (DOD) methodology. In other words, the application has not been sketched out until valid data sources were confirmed. Dominantly, the data sources imported in our application were examined according to requirements of system quality indicators, which are essential for us to provide a robust application.This is especially important in a high risk, high consequence field like medicine, where drug conflicts may lead to severe reactions or may even be fatal. This in contrast to what is considered to be the industry standard object oriented design, as it doesn't lend itself well to the highly data driven medical field.

C. Semantic web Applications

Multiple diseases may happen to a single person. Especially in the case with diseases that affects a patient's immune system. Drugs that might have a mild to moderate effect when taken by themselves could have powerful adverse reactions when taken in conjunction with another drug. To provide holistic medical treatment, healthcare providers always need to prescribe a list of medications and drugs, targeting at different diseases at the same time. However, if any one of the drugs interacted with another, the prescription might bring a negative result that neither the doctor nor the patient wants to see. Meanwhile, it is often a mentally heavy burden to memorize relationships among diseases, drugs, and interactions. The result is, a slip in the doctor's mind could cause severe medical accidents. To help fill this gap, we developed a semantic web application. The system does not entirely replace doctors' work but plays a supporting role in the process of healthcare providers' decision making. Fig.(5) maps the process of drug prescription application. In addition to providing healthcare providers with an indispensable tool, it also provides the patients with an easy to understand the idea of what drugs not to mix, during the process of medicating themselves. These would lead to far fewer accidents and greater autonomy for the patient as well.

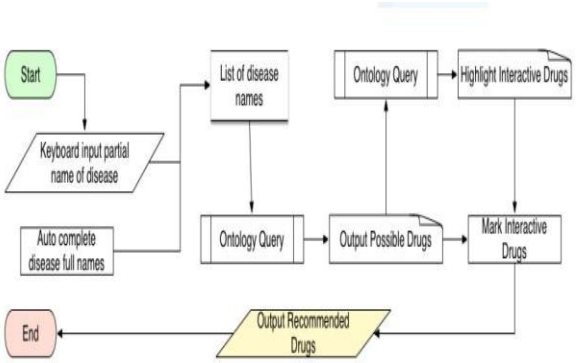

Figure 5. Flowchart of Drug Prescription Application Process

D. Proposed Model Architecture

Proposed Gen Ontology based Web Application Architecture The semantic application is supported by several files and services, the data communications among which are hidden against users. The user interface is a webpage in HTML format listed in fig.(6). Depicts the software architecture of application. An HTML file organizes the function components shown to a user. A CSS file formats the page and optimizes visual design. A JavaScript file catches user actions, while input data is passed to a Python program that generates a SPARQL query. This SPARQL query is sent to Stardog database manager, which retrieves our ontology, and then returns wanted results to Python program. Next, results is arranged in human-readable format by JavaScript and illustrated on the browser by CSS and HTML. The ontology, as uploaded beforehand to Stardog, was created with Protégé originally, in Turtle format. Triples and instances were downloaded from the Internet and added into the Turtle file. Based on axioms and definitions on classes and properties, the Pellet reasoner of Protégé produced inferences, which enables the application to provide reasoned results.

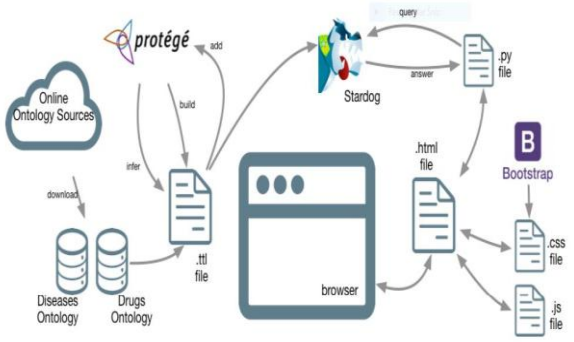

Figure 6. Architecture of Drug Prescription Application

IV. DATABASE DESIGN AND IMPLEMENTATION

A. Data Source Selection

The semantic application has developed with Data-Oriented Design (DOD) methodology. In other words, the app has not sketched out until valid data sources were confirmed. Dominantly, the data sources imported in our application were, examined according to requirements of system quality indicators, which are essential for us to provide a robust application. Aspects of system like taken quality to into consideration: usability, affordability, interchangeability and sustainability.

B. Data Structure

The ontology in the project consists of three types of classes: Diseases, Drugs and Genes. Figure 7. Shows a hierarchy of the classes.

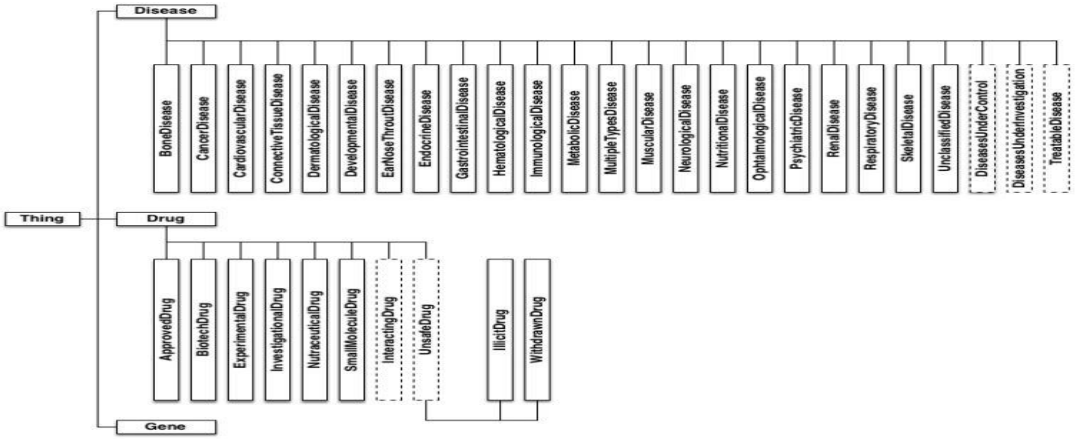

Figure 7.Ontology Structure of Application

Diseases are classified into 22 classes, i.e. “Bone Disease”, “Cancer Disease” etc., as defined by the online data source “Diseasome”. Some diseases are recorded caused by certain genes, which then mutually become “related diseases”. The 22 classes are predefined, instances of which are downloaded and imported by class labels. The other three classes of disease are defined with class restrictions, i.e. “Disease under Control”, “Disease under Investigation” and “Treatable Disease”. Instances of these classes are inferred results. Similar structure is the class of “Drug” and its subclasses. “Gene” class is affiliated to “Disease” class[15].

C. Data Reuse

Two external ontologies are reused in this project: Diseasome and DrugBank. The two ontologies are both created by Research Group Data and Web Science at the University of Mannheim, published with D2RQ Platform and can be reached via SPARQL endpoints. From Diseasome we retrieved the diseases, classified by category, a list of the drugs that can treat each disease (the connection between Diseasome and Drugbank), a list of the genes and the diseases that they can affect. From Drugbank we used their list of drugs, their types and the existing brand names for every drug.

D. Ontology inferences

Our ontology uses inferencing for further classification of drugs and diseases, to make searches faster and easier (for example using filters) and building more structure. Furthermore inferencing is used to make possible searching for diseases related by the same genes, or related drugs that can treat the same family of diseases.

Query Examples

E. Searching for a diseases

Figure 8. is a screenshot of disease searching query. We can use this query to find a disease in our database starting from an input by the user, in this case “diabetes”.“Treatable Disease” is defined as a disease for which our database knows a feasible drug.

```
PREFIX : <http://huiqingao.com/ontology/interactingDrugs.ttl#>

SELECT ?disease ?label WHERE {

  ?disease a :TreatableDisease;
           rdfs:label ?label.

  FILTER regex(?label, "diabetes", "i" )

}
```

Figure 8. Query Example: Searching for a Disease

F. Finding Possible Drugs

After finding the correct result we can look for possible drugs to treat that specific disease. Figure 9. is a screenshot of the query. In this case the disease is identified through its label.

```
PREFIX : <http://huiqingao.com/ontology/interactingDrugs.ttl#>

SELECT ?drug ?label2 WHERE {

  ?disease a :TreatableDisease;
           rdfs:label "Diabetes mellitus";
           :possibleDrug ?drug.

  ?drug rdfs:label ?label2;
        a :ApprovedDrug.

}
```

Figure 9. Query Example: Finding Possible Drugs

G. Finding genes related to Diseases

In this study we can look for genes related to diseases by querying the "associated Gene" property. This can be used in a more advanced way by looking for specific genes.

Implementation steps

1. Adding data to stardog server
2. Run the stardog server by executing following commands

   a. $./stardog-admin server start –disable-security

   b.$./ stardog-admin db create -n interdrugs

   c.$./stardog data add interdrugs \Users\deen\Documents\ontologies\drugs.ttl

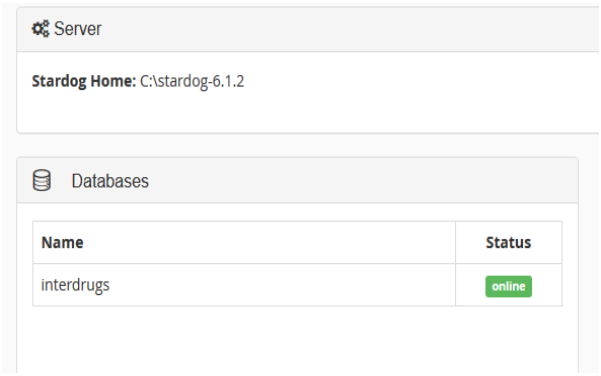

Figure 10. Stardog Database

3. Hosting the python server listed in fig.(10) Server name: mypython.

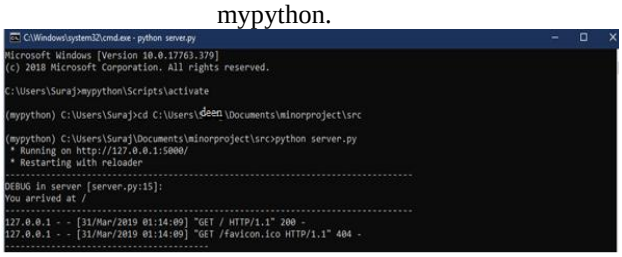

Figure 10. Python Virtual Environment

4. Define the classes and the properties in the database Interdrugs[22] shown in fig.(11).
5. Compile and run the file by the command $ python server.py
6. Interacting with RDF and performing queries**:** The RDF schema and database is stored in stardog. We are hosting the database with ontologies on the stardog server. Our app can perform different query on the RDF database using stardog API.

7. We are using python to interact with stardog. The above functionality is coded in server.py. By default, we are running the web page main.html.

8. The homepage contains 2 options to navigate as shown in fig.(12): (a) Drug Prescription System (b) Genes Identification System.

Figure 11. Defining the classes & Properties

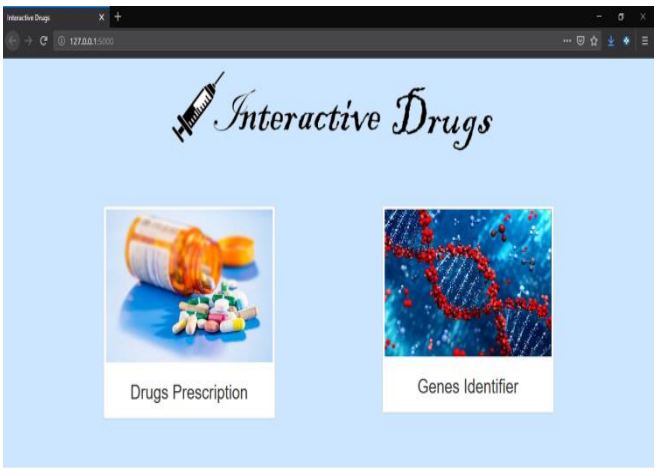

Figure 12. Homepage of Interactive Drugs

9.In this page, we are using form to collect the Disease name by searching, and providing add option in result shown in fig.(13).

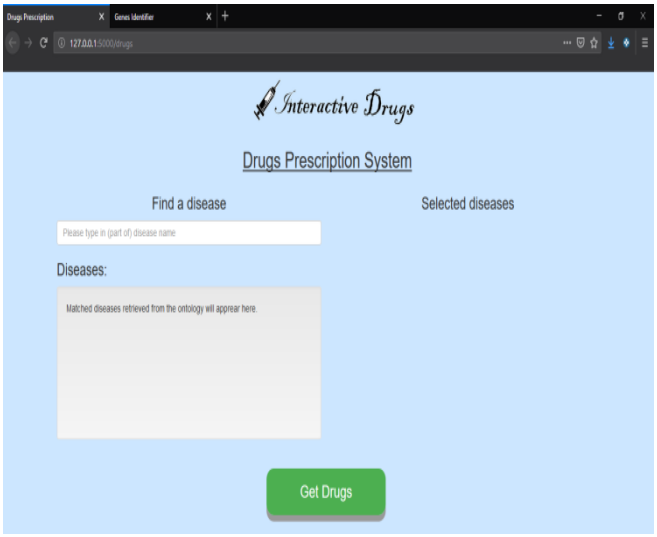

Figure 13. Drugs Prescription Discovery

10.When Get Drugs button is pressed, the query is performed on the RDF database using the disease names and returns different kind of medications for combination of disease.

11.To search the genes related to diseases we can navigate to genes identifier shown in fig.(14) from the homepage.

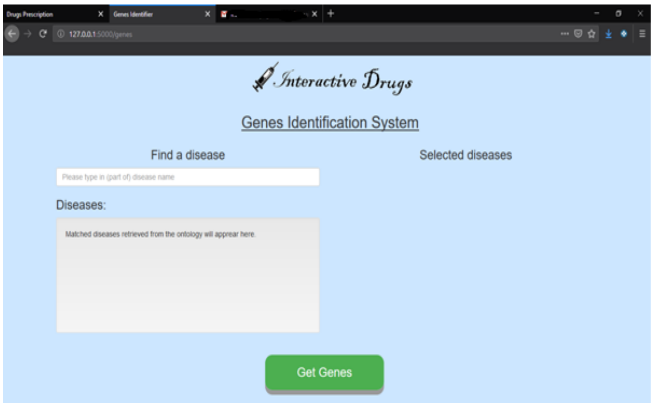

Figure 14. Gene Identification system

12. When Get Genes button is pressed, the query is performed on the RDF database using the disease names and returns different kind of genes for related to the disease.

V. RESULT AND ANALYSIS

The semantic web computing is to share clients information and manages the risk, and also fulfils the specific needs of their clients. In the web interface for Drugs Prescription accepts the diseases and symptoms from the domain updated in database and successfully displays prescribed drugs for each disease. The drug-drug interactions are highlighted by yellow background. Hovering the mouse pointer on the displayed drug, the interacting drug pairs get highlighted in red background. The precision of the application can be further increased by updating it with domain data from more dynamic sources on web.

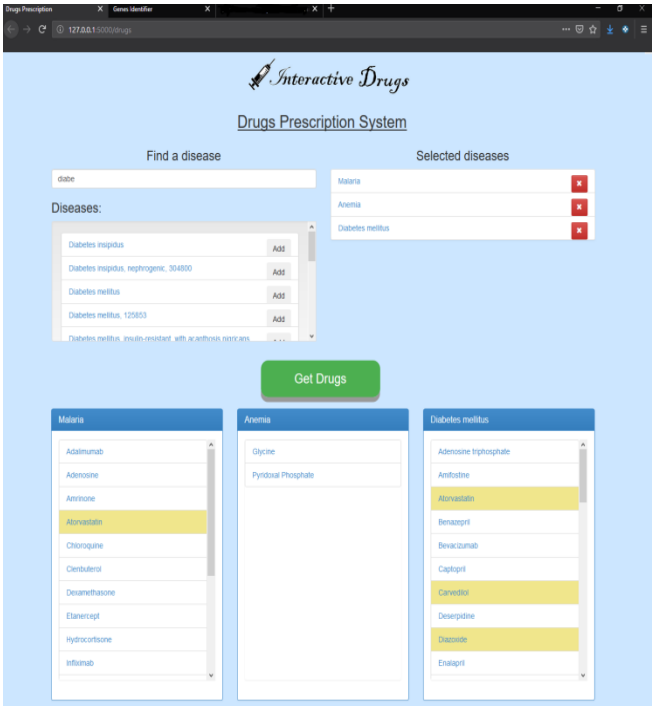

Figure 15.Web Interface app for Drug Prescription

When the displayed drugs are clicked, a detailed view of the particular drug is given in tabular form which contains data such as Code, Brand name, biotransformation, Chemical formula, Drug Category, Drug Type, Indication, Label, Melting point, Possible Disease Target, Dosage, Toxicity, etc. as listed in fig.(16). The semantic web computing is to share clients information and manage the risk, and also fulfils the specific needs of their clients. The semantic web computing is to share clients information and manage the risk and also fulfils the specific needs of their clients.

Figure 16. Detailed Description about the Drugs

The web interface for Genes Identification system accepts the diseases and symptoms from the domain updated in database and successfully displays are listed in fig.(17) for the genes. The web interface accepts the diseases from the domain updated in database and successfully display related genes for each disease. The precision of the application can be further increased by updating it

with domain data from more dynamic sources on web. From this study semantic web app research can be more flexible in future research to more facilitate on the research models incorporate among IoT based system in Healthcare Internet of Things.

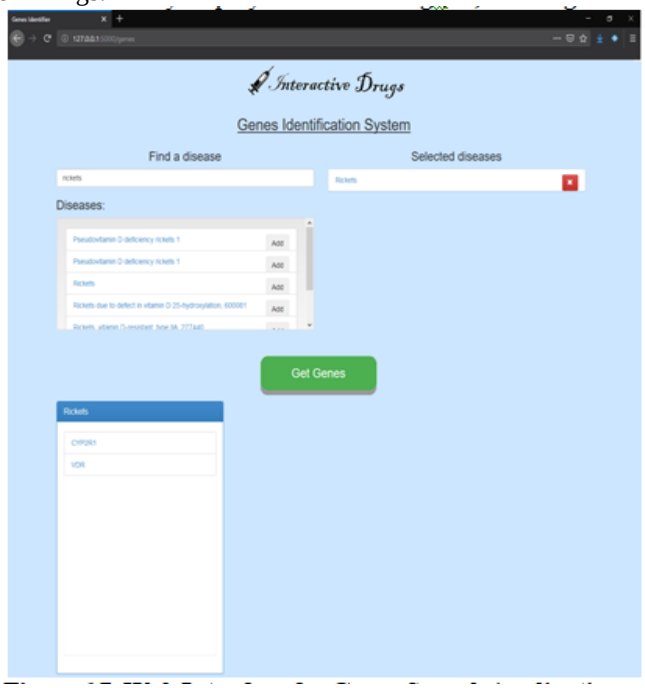

Figure 17. Web Interface for Genes Search Application

CONCLUSION

The technologies uses for medical informatics in semantic web has been organize to found a related similar pattern for design and developing new drug. Gene ontologies is a convincing reasoning based knowledge discovery to encoded the capture drug interactions identification..The use of data driven design methodology also proved highly useful in the building of this project, and lends itself well to the software projects for the medical field. Gene ontologies is a persuasive reasoning based knowledge discovery to encoded the capture drug interactions identification. Semantic reasoning rule uses prioritization of drug interactions, directions of research performance optimize due to all interaction have not the same importance. Queries search will take less time to response lead to modular ontologies and less memory utilization. The creation of a simple and easy to use tool which is simple to solve but significant problem of drug interaction opens up the further possibility of cross domain work mixing the computer science and engineering fields. The digitization of critical to know, but tedious to remember information saves doctor's time, allowing them to focus on tending to the patient. We've concluded that several such problems exist in every field that would be greatly assisted by application of basic computer science principles to solve problems that require tedious repetition.

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AUTHORS PROFILE

Anjna Jayant Deen working as Assistant Professor in UIT, RGPV Bhopal, she is having more than 22 years experience in teaching and research. She is currently a PhD student in MANIT Bhopal. Her research interest is in Data science, Network security, Bioinformatics, Artificial Intelligence, Machine learning and Neural network. Her publications in more than 18 research papers in Scopus-journal, International Conference and International journal. She is a member of IEEE.

Manasi Gyanchandani is working as Assistant Professor in MANIT Bhopal. She is having more than 24 years of experience in teaching and research. Her educational qualification is a PhD in Computer Science and Engineering. Dr Manasi Gyanchandani's area of specialization in big data, big-data privacy and security, Machine learning, data mining, privacy-preserving, artificial intelligence, expert system, neural network intrusion detection & information retrieval. Dr Manasi Gyanchandani has more than 32 research papers in SCI/ Scopus/ Peer-Reviewed journals, International Conference, book chapter, and National Conference. She is a Life Member of ISTE.