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## Design and Implementation of Ontology Based Risk Assessment Model in Diabetes Mellitus

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### Abstract

Any clinical intelligent system needs a complete patient profile to do an efficient diagnosis. This system is a Clinical Decision Support System (CDSS) for risk prediction and treatment of Diabetes Mellitus using semantic technologies. The system accepts patient related medical information through a well-defined questionnaire as ontology and generates a semantic profile as a knowledge base. Since an ontology-based questionnaire is used to gather the patient information, the system is adaptive in nature. Patient semantic profile is created automatically as an instance, in which information is inferred from the asserted classes and properties in the questionnaire ontology. Clinical guideline ontology is developed, which is a set of authenticated medical guidelines, including the score for various risk elements as rules. This work is focused on the prediction of risks related to cardiovascular, sexual, alcohol, smoking, and physical activity factors of diabetic patients, which are the core risk elements in diabetics. The reasoner checks the rules (guideline ontology) against automatically generated patient semantic profile and predicts various risks score with suitable treatment suggestions.

**Keywords:** Clinical Decision Support System, Semantic Profile, Adaptive Questionnaire, Risk Factor Prediction, Score Calculation

## Introduction

Achieving perfection in providing good health care by medical professionals is very difficult at times. When a patient visits a hospital, a nurse will first diagnose the patient and will record the preliminary observations such as readings of blood pressure, height, weight, body temperature etc. (Ahmadian, L. et.al., 2010). Other details are collected from the patient by the doctor. Sometimes due to time restrictions, medical staff may fail in collecting relevant details of patient history or sometimes the patient himself/herself doesn't want to disclose certain sensitive and personal health problems. So, in these cases the patient medical history will be incomplete. Also, medical negligence can happen because of limited knowledge and work pressure. Due to the above reasons, an efficient diagnosis cannot be done by the doctors (Lin, Y. and Sakamoto, N., 2009).

For a very long time, computers were used through artificial intelligent medical professionals in diagnosing the diseases and in clinical decision-making processes. With the developments in IT, such systems were later called as clinical decision support systems (CDSS). Clinical Decision Support Systems are IT based systems designed to assist medical professionals, particularly novice in the area, to improve the decision-making process. CDSS enhance the patient health. It offers doctors, patients and other clinical staffs, person specific information after filtering the knowledge intelligently (Aleksavska-Stojkavska, L. and Loskovska, S., 2010). CDSS typically takes patients data and propose a set of appropriate diagnosis (Karim, S. and Bajwa, I.S., 2011). CDSS comprises of different tools to enhance decision making in the clinical domain in the form of alerts, reminders, clinical guidelines, focused patient summaries, risk assessment etc. Haynes et.al. defined CDSS as "...information technology-based systems designed to improve clinical decisions-making (Sim, I. et.al., 2001) These types of systems aid in diagnosis by checking the patient data, medical knowledge source in the system and the doctor's observation (Lobach, D.F. and Hammond, W.E., 1997). The most important advantage of implementing such systems is diagnostic accuracy and precision. Also, since AI techniques are used in these decision support systems, these systems acquire the capacity to learn and to create new knowledge. CDSS is helpful for clinicians to make clinical decisions efficiently. Diagnosis can be done with the help of CDSS and thus the final result can be made more efficient and accurate by avoiding the humanitarian errors that can happen during a manual diagnosis process. All types of clinical data, such as personal medical history,

family history, and laboratory data are analyzed in CDSS to get the result. The range of diagnosis can be from drug interactions to disease symptoms (Abbasi, M.M. and Kashiyarndi, S., 2006).

CDSS is broadly classified into two main categories: knowledge based, and non-knowledge based. The knowledge based CDSS contains rules, mostly in the form of IF-ELSE statements. Rule based systems will be used to infer the risk factor which can be fuzzy logic based or SWRL based (Prasath, V. et.al., 2013). Data is usually associated with these rules. Knowledge based CDSS generally consists of three main parts. Knowledge base, Inference rules and a mechanism to communicate (Al Iqbal, R., 2012). Non knowledge-based system uses a form of artificial intelligence (machine learning). In this, to derive relationship between the symptoms and diagnosis, neural networks are used in which the nodes and weighted connections are used to write rules for input. Genetic algorithm also can be used to produce the best solution of the problem.

In Knowledge based CDSS, Electronic Health Records (EHR), knowledge representation and reasoning systems are correlated. Traditional method of collecting Electronic Health Records (EHR) was through peer to peer interview between doctor and patient (Turley, M. et.al., 2011). The advantages offered by EHR in comparison with traditional manual and peer- to peer interviews are manifold (Ramakrishnan, N. et.al., 2010, Bouamrane, M.M. et.al., 2008). However, these systems have significant limitations, including lack of flexibility and adaptability to complex clinical requirements and processes and a general lack of intelligence (Bouamrane, M.M. et.al., 2010). Due to their rigid architectural nature these systems are difficult to maintain and update (Bouamrane, M.M. et.al., 2008). Also, here, the design of knowledge base and the reasoning mechanism is very critical. If the knowledge base is incomplete or/and the reasoning mechanisms are ambiguous, then it will result in incomplete and imprecise diagnosis. Most of the CDSS are facing the common problem of heterogeneity, reusability, etc. Also, the lack of interoperability among these systems causes difficulties in sharing and extension of knowledge. To overcome these problems, a layer of ontology can be added on top of the functionalities of any CDSS system.

In this research, we have adopted ontology-based approach to model the CDSS. Ontology based CDSS is a solution for heterogeneity and sharing of knowledge (Sherimon, P.C. et.al., 2012). Ontology is one of the most powerful tools to encode medical knowledge semantically. It provides

better reasoning mechanisms. Ontology provides a common framework for structured knowledge representation of domain knowledge (Saripalle, R.K., 2010). The concepts, relationships, and rules in the domain are represented in the ontology. Ontology reasoners are used to reason the inferred knowledge from the knowledge represented in the ontology. They infer logical consequences from a set of asserted facts or axioms. Representing knowledge semantically will allow a computer to interpret the data and acquire inferred knowledge. Data will be represented in an independent format and it can be reused and shared. The inference rules are commonly specified by means of an ontology language, and often a description language. The Semantic Web Rule Language (SWRL) is a language for the Semantic Web that can be used to express rules as well as logic, combining OWL DL or OWL Lite with a subset of the Rule Markup Language (Horrocks, I. and Patel-Schneider, P.F., 2004). SWRL has the full power of OWL DL, but at the price of decidability and practical implementations (Parsia, B. et.al., 2005).

Knowledge-based CDSS have been broadly reported in the literature. Of all the well-known CDSS studied so far, relatively little work has been done in the development of decision-support systems for risk assessment prediction of diabetic patients. In this paper, ontology based CDSS is proposed for risk prediction and treatment of Diabetes Mellitus. This work is focused on the prediction of risks related to cardiovascular, sexual, alcohol, smoking, and physical activity factors of diabetic patients, which are the core risk elements in diabetics. In this scenario this work has a great role in medical diagnosis sector.

The rest of the paper is organized as follows: Section 2 describes the materials and methods. This section focusses on knowledge representation using ontology, main components of our CDSS, user interfaces, and ontology design and reasoning. Results and Discussion is given in Section 3. Conclusion and future work is discussed in Section 4 followed by References.

## **Materials and Methods**

### **Knowledge Representation based on Ontology**

Ontology driven CDSS have been used extensively in the clinical assessment of chronic diseases. They are renowned for their flexible architectures, easy to reuse knowledge modeling structures and inexpensive maintenance operations. The study conducted (Abidi, S.R., 2010), showed

exceptional results in the risk assessment and disease management of breast cancer patients which was deployed as a commercial clinical system. They utilized the semantic web approach to model the clinical practice guidelines which were encoded in the clinical decision support system for generating patient specific recommendations (Miettinen, M. et.al., 2005). The introduction of ontology can be the best solution since they have the potential of enabling true knowledge sharing and reuse among heterogeneous agents, both human and computer (Subhashini, R. and Akilandeswari, J., 2011). Logical formalization of ontology language ensures semantic interpretation, i.e. inference, by computer programs. Ontology is a major instrument toward realization of the Semantic Web vision (Ganendran, G. et.al., 2002). When more relations and more constraints are captured in the ontology, the ontology becomes more expressive, since it captures the knowledge of the domain on a more detailed level (Sittig, D.F.et.al., 2008). Since the ontology layer can be updated without the need for additional and costly software engineering work, extension of further information to the proposed system is possible (Bouamrane, M.M. et.al., 2008). Clinical workflows (clinical guidelines) are used to represent human based medical knowledge through rules (El-Sappagh, S.H. and El-Masri, S., 2014).

### **Methodology**

The developed system is ontology based CDSS for risk prediction and treatment of Diabetes Mellitus. The five risk factors of diabetic patients such as cardiovascular, sexual, alcohol, smoking, and physical activity are focused. This is a typical example of an expert system which takes patient related information based upon a well-defined questionnaire as input, applies a set of clinical guidelines, and thereby assesses risk factors and suggests treatment procedures to the Doctor. The system uses ontology as knowledge base, and an inference mechanism to extract data in the decision-making process.

The main components of our CDSS are:

- Adaptive Questionnaire ontology
- Patient Semantic Profile
- Clinical Guidelines ontology
- Ontology Reasoner

The architecture of the proposed system is shown in (fig.1). The system consists of a user interface, adaptive questionnaire ontology, semantic patient profile, clinical guideline ontology and an inference engine.

A computerized questionnaire can act as an efficient medium for collecting large and comprehensive patient's medical history without wasting the doctor's valuable time, but an adaptive system can achieve better profiling accuracy with a significantly reduced number of questions ((Miettinen, M. et.al., 2005).

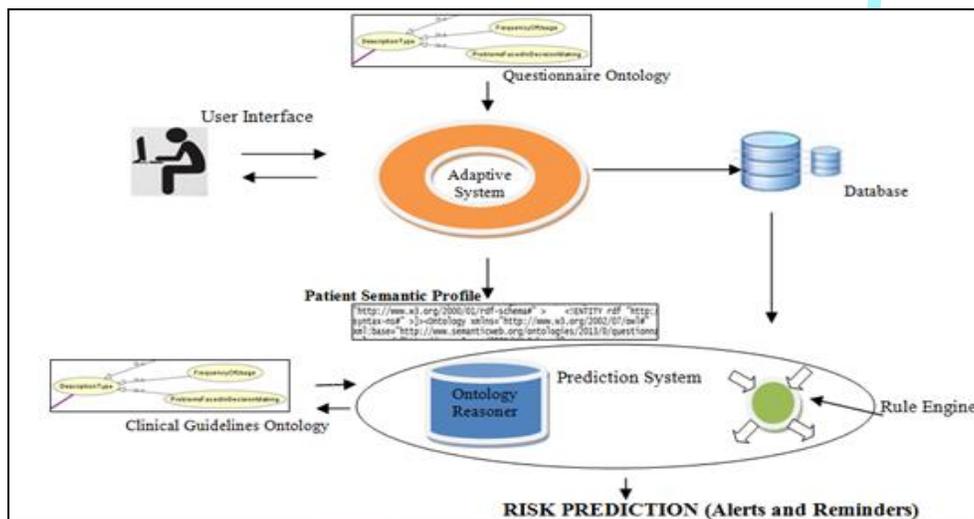


Fig. 1. Proposed CDSS for Diabetes Mellitus.

So, we have adopted a context sensitive adaptive approach for collecting patient profile. The purpose of this context sensitiveness (through questionnaire ontology) is to mimic the exploratory behavior exhibited by the clinicians during consultation sessions. In this model, initially the patient's complete medical history (personal information, physical activity, diet etc.) is gathered from the patient and stored in ontology as instance. Additionally, the clinical staff (e.g. nurse, lab technologist) and doctor also provide input to the patient's profile after diagnosis. According to the input provided, a patient semantic profile is created. Approved Clinical Guidelines are stored in the knowledge base in the form of rules and rule-based reasoning is used to predict the risk factors.

The UML sequence diagram of the model is shown in (fig.2). It gives a dynamic view of how the system works.

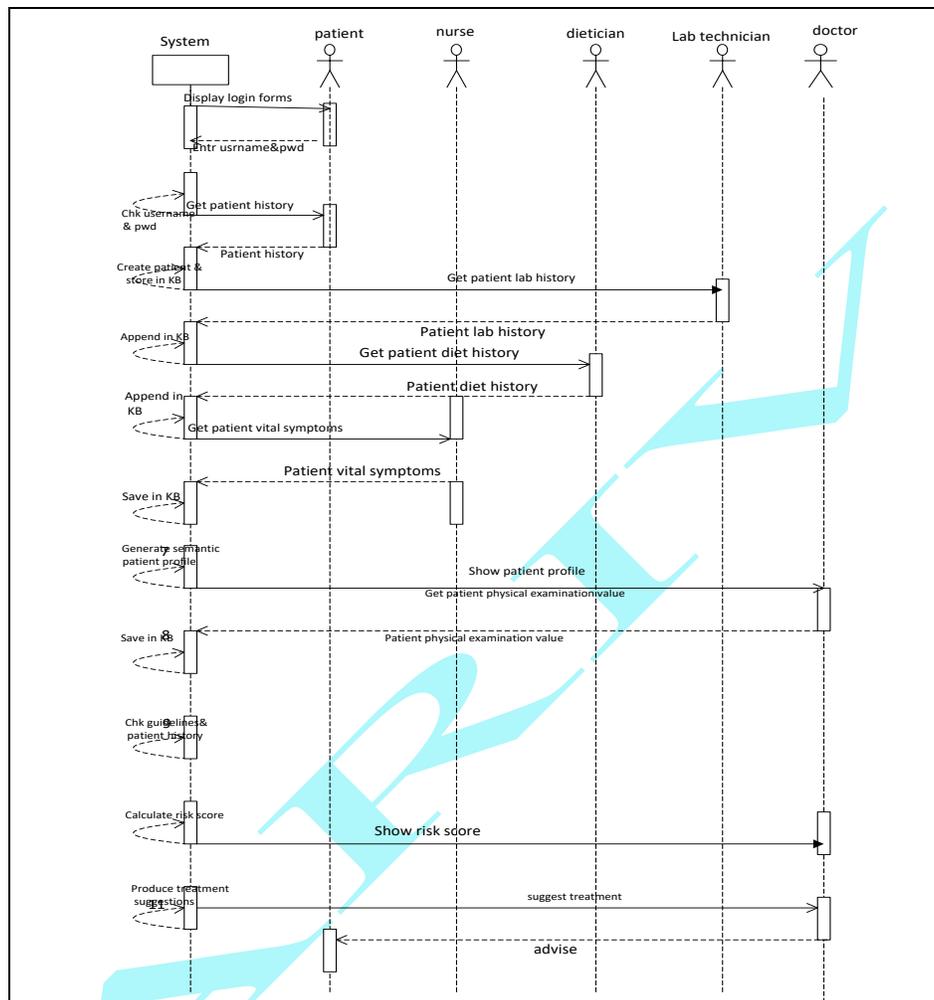


Fig. 2. UML sequence diagram of the system.

## Implementation

To gather the complete patient information, several user interfaces are developed in Java, for patient, nurse, lab technologist etc. The Java adaptive engine is implemented using Jena API. The Java engine acts as intermediary between the user interface and ontology. It reads the questions from the ontology and presents it to the user and analyses the user inputs.

## User Interface

User interfaces (UI) are used to receive input to generate patient semantic profile. For instance, through patient interface, patient can enter all the history (family, physical activity, smoking etc.)

Through nurse interface, nurse can record the information regarding the vital signs such as BP, temperature etc. Each UI is protected by username and password for each user and hence security is confirmed. The information entered by the patient and nurse will be visible only to the doctor and then s/he can do further diagnosis based on the available data. An UI of patient for smoking history is shown in (fig.3)

When the patient's data input is made through the interface, new instance of *Answered Questionnaire class* is generated automatically with patient ID. For each instance, the values entered by the users are asserted into different data properties.

SMOKING HISTORY		
15	Do you smoke?	<input type="radio"/> Yes <input type="radio"/> No
16	If yes, for how many years you are smoking?	<input type="text"/>
17	How many cigarettes do you smoke per day?	10 or less ▾
18	How soon after you wake up do you smoke your first cigarette?	0-5mins ▾
19	Do you find it difficult to refrain from smoking in places where smoking is not allowed (e.g. hospitals, government offices, cinemas, libraries etc)?	<input type="radio"/> Yes <input type="radio"/> No
20	Do you smoke more during the first hours after waking than during the rest of the day?	<input type="radio"/> Yes <input type="radio"/> No
21	Which cigarette would you be the most unwilling to give up?	<input type="radio"/> First in the morning <input type="radio"/> Any of the others
22	Do you smoke even when you are very ill?	<input type="radio"/> Yes <input type="radio"/> No

Fig. 3. Patient user Interface for smoking history.

Four external APIs are used in the system such as Pellet Reasoner, Manchester OWL2 API, SPARQL DL API and Google collections. Pellet Reasoner is used for executing reasoner from the code. Manchester OWL2 API is used to create/write/read/edit OWL2 ontologies. SPARQL DL API for SPARQL execution and Google collections are to provide some advanced data structures.

In addition to the inbuilt APIs with protégé, six external API are created and used. They are OWL2Reasoner Class, Onto Retriever Class, Onto Updater Class, MyOWLClass, My Individual and Age Calculator.

### ***Ontology Design and Reasoning***

Three ontologies are used in the system such as adaptive questionnaire ontology, clinical guidelines ontology and automatically generated semantic profile ontology.

### Questionnaire ontology

The intelligent questionnaire is a patient's information collection system. It is adaptive in nature; thus, asking only related information appropriate to the patient's environments. The users can be clinicians or patients themselves [through online]. This adaptive questionnaire will help in providing a filtered, clear patient medical history which is very important in the future diagnosis.

All main concepts are in the form of parent classes. The Questionnaire ontology consists of six main classes such as questions, question bank, answers, blood sugar test results, choice, treatment procedures and patient. Each abstract class has different subclasses according to the category. Each subclass has instances in which properties such as object property and data property are assigned. For example, the question bank class consists of five subclasses to keep each type of question. The subclasses assigned for question bank class are dietician, doctor, lab, nurse and patient. The subclass patient question bank consists of eleven instances such as alcohol history, complication information, diabetic history, family history, medical history, medical history, medication history, monitoring history, nutritional history, personal information, physical activity history and smoking history. Each subclass has instances in which properties are assigned. Object property is used to associate a class and subclass while data property is used to assert value to an instance. For example, to the instance of smoking history, the object properties smoking habit is assigned (Fig.4). These properties are used to store the score values of each instance.

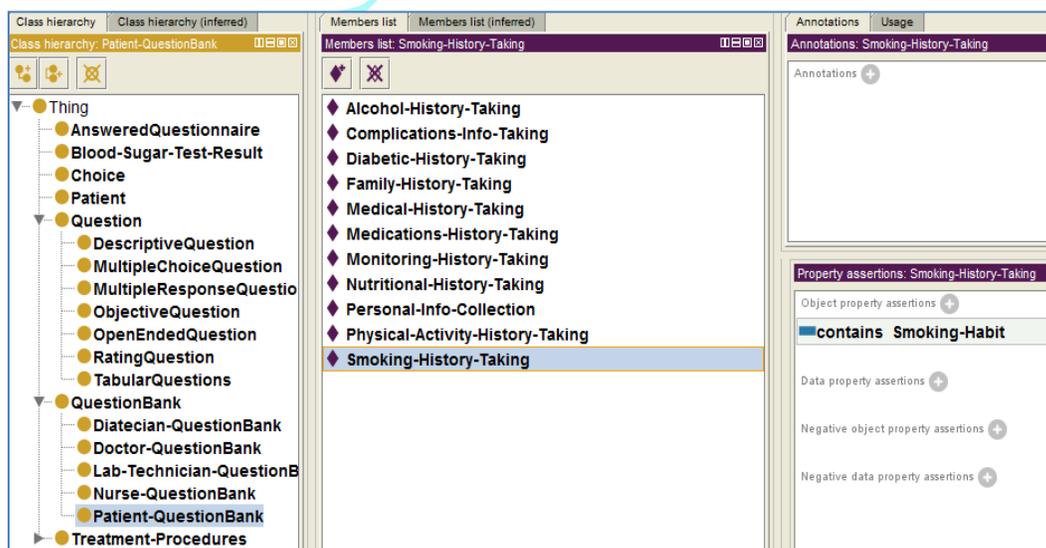
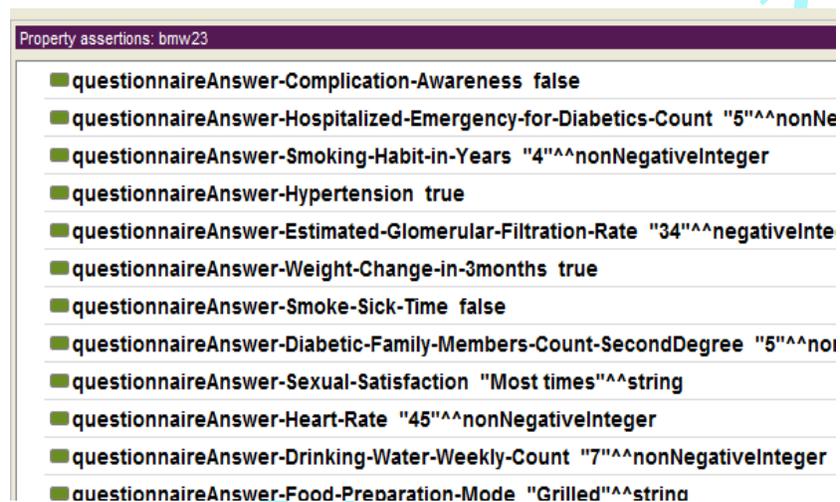


Fig. 4. Questionnaire Ontology

### *Semantic Profile Ontology*

Efficiency of personal information will be more with a better model user detail. The structured way of user details can describe the conceptual semantic of user interest (Uddin, M.N. et.al., 2012, Farooq, K. et.al., 2013). The Semantic Profile is constructed as follows: The system instantiates the Questionnaire ontology and stores the corresponding answers in it (fig.5). The system processes this information and automatically generates a patient profile ontology instance in the server.



Property Assertion	Value
questionnaireAnswer-Complication-Awareness	false
questionnaireAnswer-Hospitalized-Emergency-for-Diabetics-Count	"5"^^nonNe
questionnaireAnswer-Smoking-Habit-in-Years	"4"^^nonNegativeInteger
questionnaireAnswer-Hypertension	true
questionnaireAnswer-Estimated-Glomerular-Filtration-Rate	"34"^^negativeInte
questionnaireAnswer-Weight-Change-in-3months	true
questionnaireAnswer-Smoke-Sick-Time	false
questionnaireAnswer-Diabetic-Family-Members-Count-SecondDegree	"5"^^noi
questionnaireAnswer-Sexual-Satisfaction	"Most times"^^string
questionnaireAnswer-Heart-Rate	"45"^^nonNegativeInteger
questionnaireAnswer-Drinking-Water-Weekly-Count	"7"^^nonNegativeInteger
questionnaireAnswer-Food-Preparation-Mode	"Grilled"^^string

Fig. 5. Automatically generated patient Semantic Profile

### *Clinical Guidelines Ontology*

The NICE guideline of diabetes is used as the reference for ontology creation. The NICE clinical guidelines will provide a score for each risk category (Farooq, K. et.al., 2013). Based on this score, risk assessment can be done by the system. Table 1 shows the score, risk level and suggested treatment for smoking. According to the guidelines, the risk level of smoking is categorized as low, medium and High nicotine dependence. The clinical guideline ontology contains guidelines for treating a Diabetic patient and to analyze the risk factor. This is applied to the semantic profile of the patient to retrieve the treatment suggested for the patient, which is displayed to the Doctor.

Table.1 Score points for smoking

Total Score	Risk Level	Treatment
0-3	Low nicotine dependence	<ul style="list-style-type: none"> <li>Professional counseling.</li> <li>Pharmacotherapy not recommended at initial assessment. If patient has difficulty dealing with withdrawal symptoms, further assessment for pharmacotherapy to be carried out to ascertain suitability.</li> <li>Provide willpower and support from family and friends</li> </ul>
4-6	Medium nicotine dependence	<ul style="list-style-type: none"> <li>Require professional counseling.</li> <li>May recommend pharmacotherapy if patient is assessed to be suitable. Pharmacist and/or doctor to provide more advice on pharmacotherapy.</li> <li>Provide willpower and support from family and friends</li> </ul>
7-10	High nicotine dependence	<ul style="list-style-type: none"> <li>Require professional counseling.</li> <li>Recommend pharmacotherapy if patient is assessed to be suitable. Pharmacist and/or doctor to provide more advice on pharmacotherapy.</li> <li>Provide willpower and support from family and friends</li> </ul>

### Ontology Reasoning

Manchester OWL syntax is used to express the rules in Clinical Guidelines Ontology. The guidelines are modelled like a flowchart. Rules are defined from every path which originates in the start symbol of the flowchart and ends with a leaf node. The reasoner, by default, takes only predefined relationships for inference (e.g. subClassOf, instanceOf etc.). Otherwise, rules must be explicitly defined for the same. For example, if a diabetic patient has high cholesterol, usually doctors prescribe the medicine 'statin' to lower the cholesterol. Before suggesting the medicine, our system initially checks the cardiac history of the patient. It is represented in Manchester OWL as follows: -

$Patient(?p) \wedge isDiabetic(?p,true) \rightarrow checkCardiacHistory(?p)$ . Now if the value of cardiac history is false and if the age of the patient is less than 40, the system checks the LDL (bad cholesterol) value. It is represented as

$checkCardiacHistory(?p,false) \rightarrow age(?p)$ .

$age(?p,?aValue) \rightarrow swrlb:lessThan(?aValue, 40) \rightarrow checkLDLValue(?p)$

Similarly, a bunch of rules are checked by the system before suggesting 'statin' to the medicine. This makes the reasoner to advance the process from one stage to another.

## Results and Discussion

In our system, patient context is represented using ontological patient profile which is an annotated instance of the questionnaire ontology. Each patient profile is an instance of the questionnaire ontology (Fig.6). The choice of the patient in the questionnaire ontology is annotated with a score calculation which plays an important role in risk prediction. As the patient interacts with the system through Questionnaire ontology by providing answers, the semantic profile is generated.

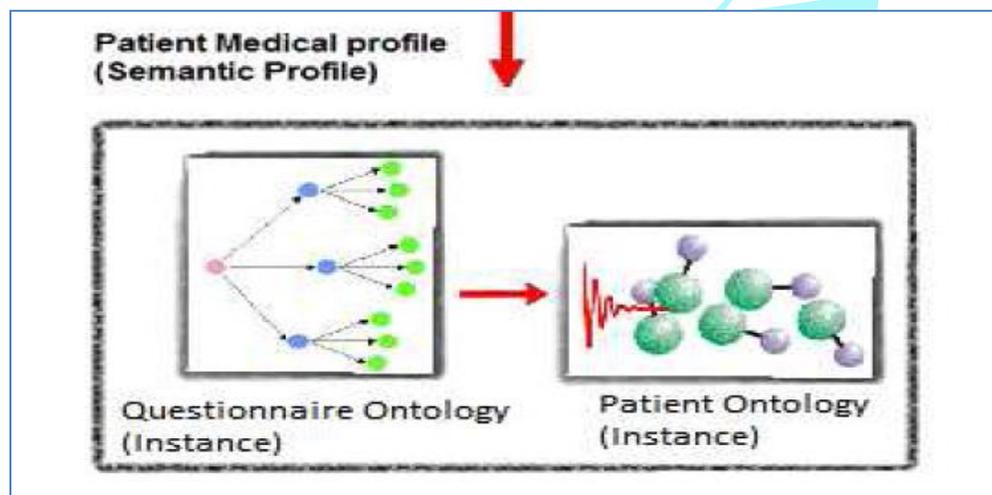


Fig.5 Instance of patient semantic profile

The values entered through different user interfaces will be stored in patient profile ontology. A patient semantic profile consists of the details from patients, nurse, dietician and doctor. Patients' diabetic history, family history, smoking history, alcohol history and physical activity history are collected. The class *Answered Questionnaire* is created to store the instances of patient records. Patient profile of each patient is created as instance of this class and is automatically generated from questionnaire ontology. The name of the instance will be the patient Id. This patient profile will be created as a OWL file and can be viewed through Protégé. The profile format created in OWL cannot be understood by a doctor. So, after the data is entered by patient, nurse, lab technician and dietician, the doctor can view the complete history of the patient in a tabular format in UI.

SMOKING HISTORY		
1	Smoker?	Yes
2	Smoking Habit-No. of Years	1
3	Cigarette Count/Day	null
4	Smoking Time after wakeup	31-60mins
5	Refraining from Smoking Difficult?	No
6	Smoke more at day first hours?	No
7	Cigarette Uneasy giveup time?	First in the morning
8	Smoke during illness?	No

Fig.6 Patient Medical Profile related to smoking history

The risk assessment will be done in the system by inferring the patient's semantic profile and clinical guideline ontology. Using this system, doctor can view patient's medical history as well as the predicted risk assessment. The total risk score is calculated for each category according to the input of the patient and the score given to each question answer. Risk assessment will be done based on the total score calculated [Table 1]. In our system risk assessment of four more categories such as alcohol, sexual, cardiovascular and physical are also predicted other than the smoking history. System will suggest the treatment related to each risk factor as per the rules in clinical guideline ontology. The patient risk level and treatment for each category is shown in [Table 2].

**Table 2. Output: Risk assessment of the patient**

PATIENT RISK ANALYSIS			
Parameter	Score	Risk	Treatment
Smoking	4	Medium Nicotine Dependence	Require Professional Counselling. May recommend pharmacotherapy if patient is assessed to be suitable. Pharmacist and/ or doctor to provide more advice on Pharmacotherapy. Provide will power and support from family and friends.
Alcohol	16	Zone III	Simple advice plus brief counseling and continued monitoring.
Sexual	18	Mild erectile dysfunction	Discussion with the patient the option of starting him on Sildenafil.
Cardiovascular	0	1.5	Aspirin should not be recommended.
Physical	8	Moderately Inactive	Counsel about the importance of being active, increase activity level and follow up.

## Conclusion and Future Work

In the proposed CDSS, patient related information based upon a well-defined questionnaire is taken as input. Since adaptive questionnaire is used to collect information from patients as input, system has the intelligence to adapt questions according to the user input. The entered values are used to generate instance of each patient which is the semantic profile. Along with the patient's information, laboratory test results, patient vital information and doctor's preliminary diagnosis are also used for the automatic generation of semantic profile. This ontology is analyzed by the clinical guideline ontology, to predict the score of risk factors such as cardiovascular, sexual, alcohol, smoking, and physical in diabetic patients. These risk values (score) will help the doctor to understand about the current situation of a patient. Ontology based reasoning makes a way to discover new knowledge, which can lead to new directions in research.

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