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Project Report: Ontology for Human Disease and Diagnosis

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1. Introduction

In this era of new technological advancements and the development of medical sciences, there is always a threat of a new disease getting formed. All humans are prone to get a disease thus there is a need for awareness regarding common diseases among people. The healthcare industry contributes to a major part of the economy and it is the biggest necessity in human life. Thus there is a need to know about the details of that disease such as what are the symptoms, treatments, vaccines, organs concerned, and the specialty departments that handle those diseases.Therefore developing a system to identify diseases with their symptoms and knowing their possible treatments is important in terms of having a standard knowledge and a knowledge model is required. The diseases can be of different types based on the part of the body they affect or by the means of transfer such as virus, bacteria, or inflammation and tumor.

Scope

We developed an ontology as a knowledge model to facilitate the identification of possible diseases and their treatments for the given symptoms. The developed ontology focuses on diseases, symptoms, medical specialties, and their treatments. Additionally, it also has the vaccination details of the diseases for which the vaccinations are available.

Purpose

The purpose of this ontology is to help the users to identify the possible diseases and also to give them an idea about the possible treatments they might need to undergo. It helps the users to find the hospitals with medical specialties that they need to consult. It also helps the users to check the medical procedures to be performed on a patient for a particular disease. It also helps the users to find out the diseases for which vaccinations are available and their dosages.

Target Users

Patients:

Patients use the system to find the possible diseases for their symptoms and the hospitals with the medical specialty that gives treatments for those diseases. They also use the system to follow basic medication for normal diseases and to find the vaccination details of some diseases for which vaccinations are available.

Nurses:
Nurses use the system to access the medical procedures of different diseases.

General Public:
They use the system to find the hospitals and their medical specialties and to find the diseases with vaccinations.

Researchers:
Researchers can use this ontology in their research for the purpose of domain knowledge

2. Competency Questions

CQ1. Swaroop is showing symptoms of common cold, what possible medications can he take to subdue them?

CQ2. Kiran has abdominal pain and rashes on the skin. What may be the possible disease?

CQ3. Vamsi is having rashes in his arm, find a specialty hospital for him?

CQ4. James forgot the prescription given by the doctor for COVID-19, what possible prescription can the system give him?

CQ5. Sahapawn is having a stomachache which specialty should she ask to go to at the reception?

CQ6. Adnan just witnessed an accident and wants to find out hospitals that offer trauma care in the city.

CQ7. Alex's grandmother is in stage 1 lung cancer. How long are the duration and frequency and what are the treatments for lung cancer?

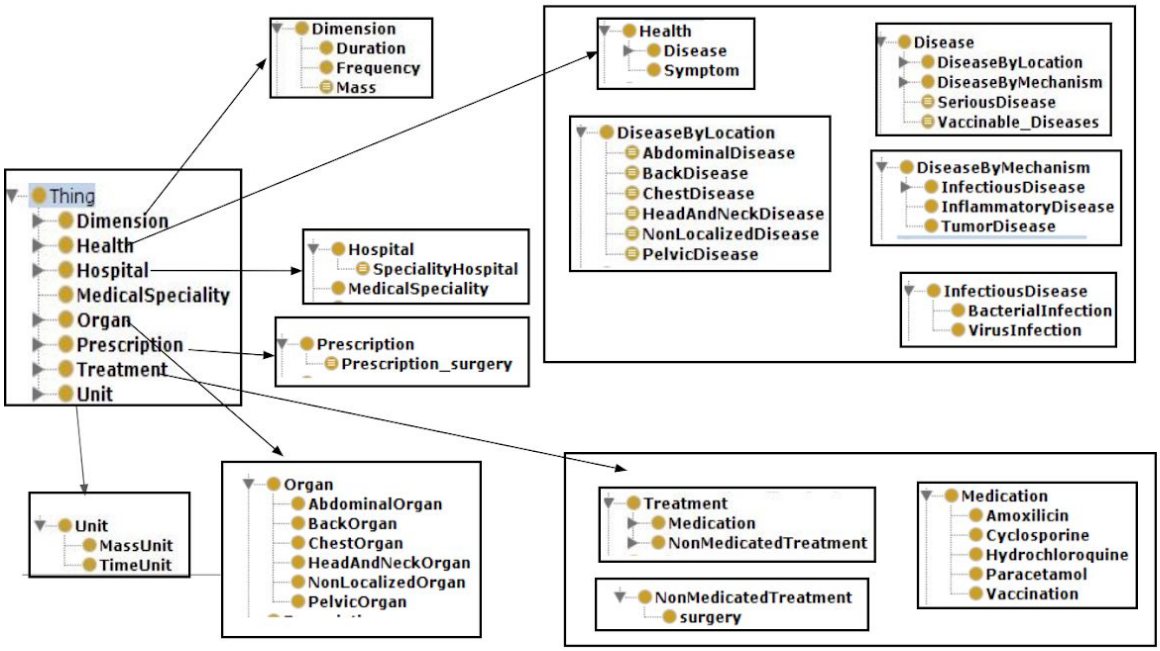
CQ8. Shantanu went back to India from Thailand during the pandemic and now is worried about getting COVID-19. He wants to know the possible symptoms for COVID-19.

CQ9. Su slipped on the wet floor and fractured her arm, where should she go and what possible treatment can she get?

CQ10. What vaccine should be given for the prevention of Hepatitis B and what is the dosage amount and where is it injected?

3. Class Hierarchy

Figure(1) shows eight top-level classes in our ontology. However, to meet the ontology scope. The defined concepts are categorized into four top-level classes.



Figure(1)

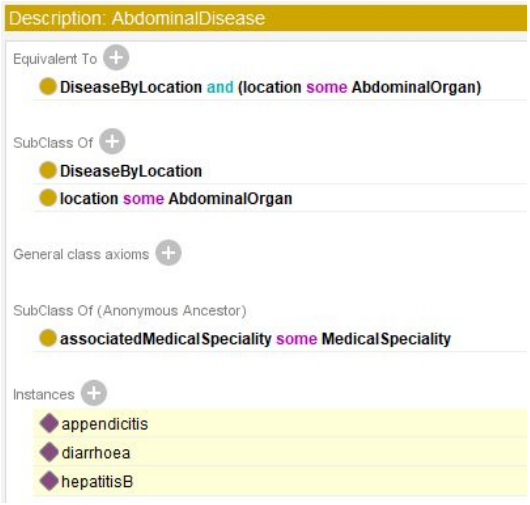
The following are the classes defined in the ontology:

1. **Health:** This is the most important top-level class, it contains two subclasses Disease and Symptom.

    1.1. ***Disease:*** This class contains instances for diseases.

1.1.1. ***DiseaseByLocation:*** It contains diseases based on the part of the body they concern. It's subclasses can be seen in Figure(1).

AbdominalDisease: This is an inferred concept which has the following constraint
DiseaseByLocation and (location some AbdominalOrgan)
This concept uses the reasoning mechanism Figure(2) shows the instances with the reasoner on.

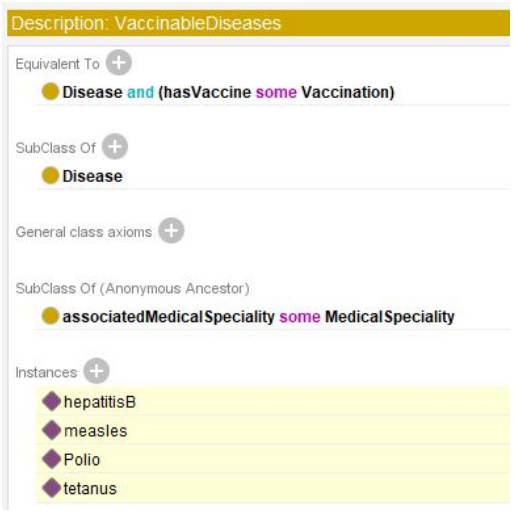


Figure(2)

Similarly for other subclasses ***BackDisease, ChestDisease, HeadAndNeckDisease, NonLocalizedDisease, PelvicDisease*** the reasoning is used.

1.1.2. ***DiseaseByMechanism*****:** It contains instances for diseases based on the mechanism of how the disease gets transferred. It's subclasses can be seen in Figure(1).

1.1.3. ***VaccinableDiseases:*** This is an inferred concept which has the following constraint
Disease and (hasVaccine some Vaccination).
This concept uses the reasoning mechanism. Figure(3) shows the instances with the reasoner on.

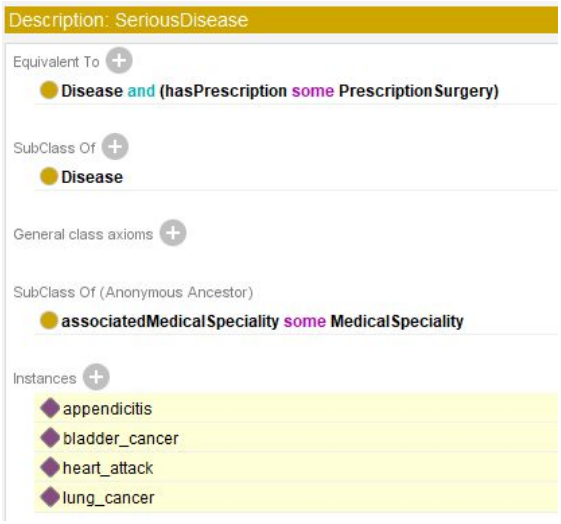


Figure(3)

1.1.4. SeriousDisease: This is an inferred concept which has the following constraint

Disease and (hasPrescription some PrescriptionSurgery).

This concept uses the reasoning mechanism. Figure(4) shows the instances with the reasoner on.



Figure(4)

1.2. ***Symptom:*** This class contains the different symptoms that a person might show if he has a particular disease.

2. ***Hospital*:** This class contains instances for different hospitals that are present in the city where the person lives and what specialty those hospitals offer. It contains the subclass ***SpecialityHospital.***

   2.1. ***SpecialityHospital:*** This is an inferred concept which has the following constraint
   Hospital and (hasMedicalSpecilaity some MedicalSpeciality).
   This concept uses the reasoning mechanism. Figure(5) shows the instances with the reasoner on.

Figure(5)

3. ***Medical Specialty:*** This class defines the different medical specialties that are there that deal with different organs and different diseases in a hospital.

4. ***Organ:*** This class defines the different organs based on the region of the body. It has 6 subclasses given in Figure(1).

5. ***Prescription:*** This class defines the different prescriptions having different treatments based on the medication or non-medicated treatments. It contains the subclass ***PrescriptionSurgery.***

   5.1. ***PrescriptionSurgery:*** This is an inferred concept which has the following constraint.
   hasTreatment some surgery.
   This concept uses the reasoning mechanism. Figure(6) shows the instances with the reasoner on.

Figure(6)

6. ***Treatment:*** This class defines the treatments that can be given to control a disease or subdue its symptoms. It contains ***Medication*** and ***NonMedicatedTreatment*** subclasses**.**

   6.1. ***Medication:*** This subclass contains commonly used medicines as subclasses that have instances for their dosage. Its subclasses can be seen in figure(1). It also contains the ***Vaccination*** subclass which has instances for different vaccines.

   6.2. ***NonMedicatedTreatment:*** This subclass contains instances for different types of surgeries and therapies.

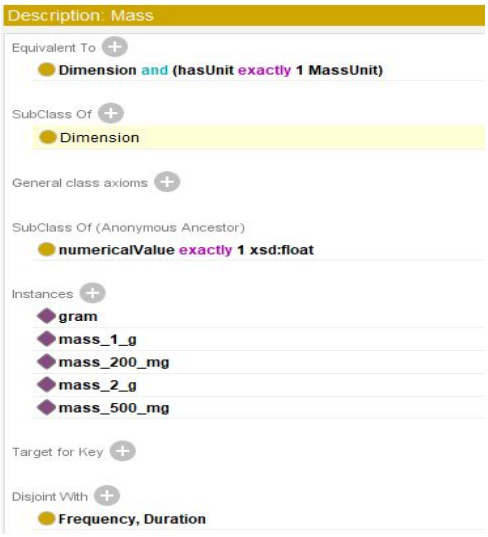
      6.2.1. ***Surgery:*** This class contains instances exclusively for surgeries.

7. ***Dimension:*** This class contains the dimensions used in the ontology for defining the subclasses

   7.1. ***Duration*** of treatment,

   7.2. ***Frequency*** of dosage, and

   7.3. ***Mass*** or Amount of dosage for a particular treatment or medicine. This is an inferred concept with the following constraint.
   Dimension and (hasUnit exactly 1 MassUnit)



Figure(7)

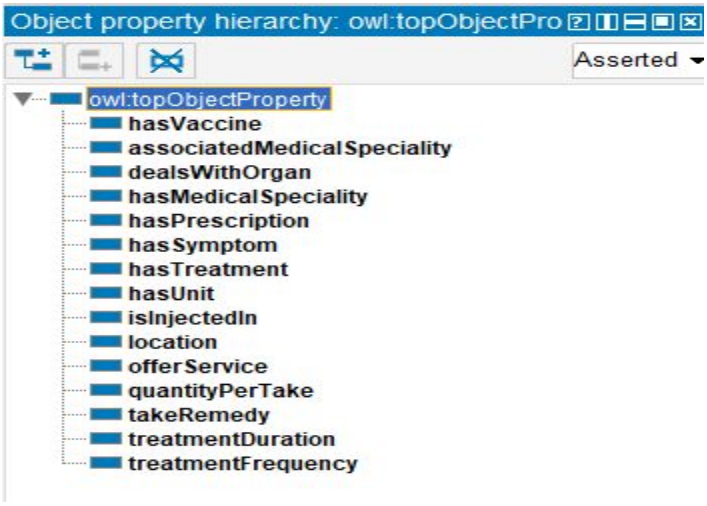
8. **Unit:** This class defines the subclasses
   - 8.1. ***MassUnit*** in gram and kilogram for Mass and
   - 8.2. ***TimeUnit*** in hour, day, week, month and year for the Duration and Frequency.

4. Object Properties

The object properties are defined to describe the various symptoms, treatments, organs related, speciality related, vaccinations related to a particular disease.

1. **associatedMedicalSpeciality:** This property defines relation from *Health* and *Organ* to *MedicalSpeciality.*
2. **hasMedicalSpeciality:** This property defines a relation from *Hospital* to *MedicalSpeciality.*
3. **dealsWithOrgan:** This property is inverse of *associatedMedicalSpeciality.*
4. **hasPrescription:** This property defines a relation from *Disease* to *Prescription*.
5. **hasSymptom:** This property defines a relation from *Disease* to *Symptom.*
6. **hasTreatment:** This property defines a relation from *Prescription* to *Treatment.*
7. **hasUnit:** This property defines a relation from *Dimension* to *Unit.*
8. **location:** This property defines a relation from *Health* to *Organ.*
9. **quantityPerTake:** This property defines a relation from *Medication* to *Mass* to define the dosage.
10. **treatmentDuration** and **treatmentFrequency:** This property defines a relation from *Prescription* to *Duration* and *Prescription* to *Frequency* respectively.
11. **hasVaccine :** This property defines relation from *Vaccinable Diseases* and *Vaccination*.

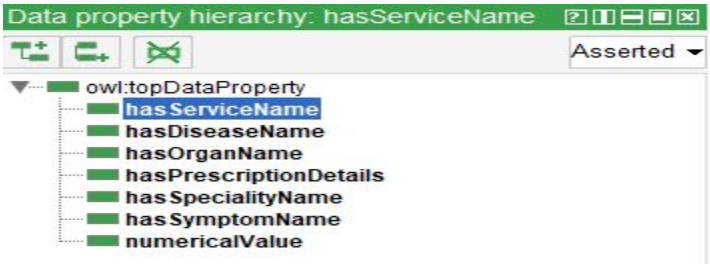
12. **isInjectedIn:** This property defines relation from *Vaccination* to *Organ.*
13. **offerService:** This property defines relation from *Hospital, MedicalSpeciality* to *Treatment.*
14. **takeRemedy:** This property defines relation from *Symptom* to *Treatment.*



Figure(8)

5. Data Properties

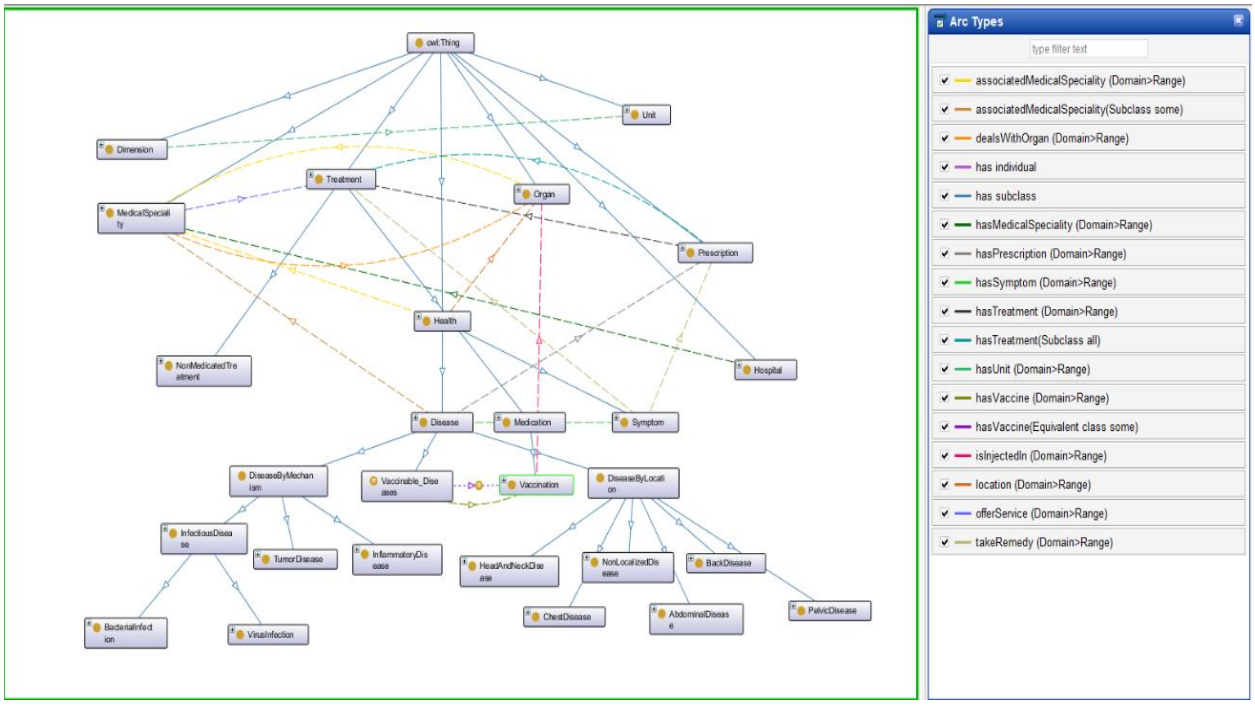
hasServiceName(string), hasDiseaseName(string), hasOrganName(string), hasPrescriptionDetails(string), hasSpecialityName(string), hasSymptomName(string), numericalValue(string).



Figure(9)

6. Class Association

We used the OntoGraf tool od Protege for creating class association hierarchy which represents the relation between concepts. The legends for the relationships are given on the side with different color lines. The solid blue lines represent the subclass of the parent class.



Figure(10)

7. Discussion of Design Decisions

Some important design decisions were taken while designing the ontology. To make it more understandable and different from a simple relational database we had to add some axioms and constraints so that the reasoning and inference features of the ontology can be realized. Some of the reasoning was done while defining the ***SpecialityHospital***, ***SeriousDisease, VaccinableDisease, PrescriptionSurgery and*** subclasses of ***DiseaseByLocation*** classes. While a constraint was put for the ***Mass***, ***Duration*** and ***Frequency*** concepts which limited each instance of dimension having only one unit of ***MassUnit*** and ***TimeUnit.***

Some sub concepts were created to accommodate different diseases based on their location in the body (***DiseaseByLocation***) and the mechanism (***DiseaseByMechanism***) they follow. Similarly, two sub concepts for the ***Treatment*** concept were created that are ***Medication*** and ***NonMedicatedTreatments***. The various constraints and axioms for different classes have been shown in the previous class hierarchy section.

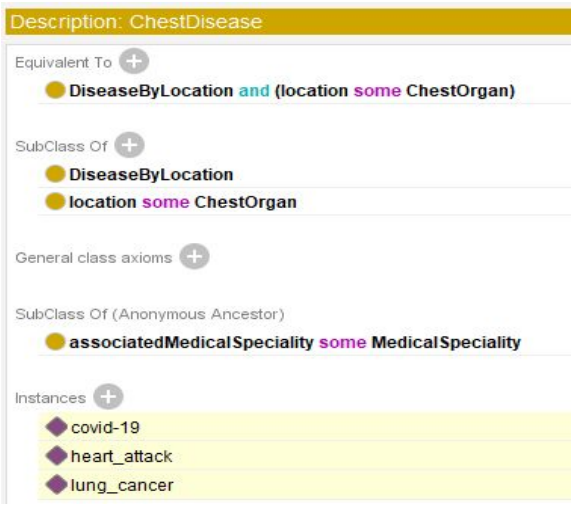
After the concept description we had to define the object properties and data properties to form the relationship between the concepts and literals. Each property points from one or more concepts in Domain to one or more concepts in Range. These properties can be functional, symmetric, transitive based on their natures. Lastly, we added instances for each concept. As we were adding instances to the classes we realized that different instances can be linked with each other through various object properties which lead to the need of adding more object properties so that the ontology can address a variety of queries and be more versatile.

In this way, our ontology was populated with instances so that it can address real world queries and answer realistic questions using the SPARQL.

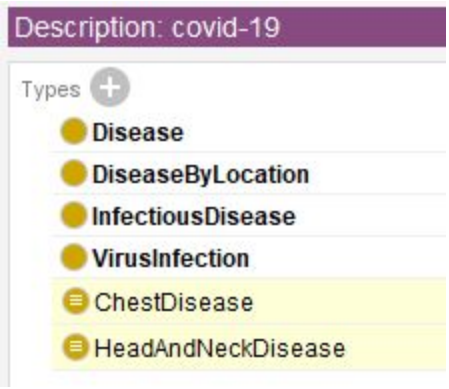
Comments Addressed:

C1: Can a single instance of disease belong to multiple classes and how did we handle those kinds of Instances?

Ans. For the instances that can possibly belong to two classes, we defined the classes they belong to explicitly using the ***Types.*** At the same time we used the reasoning mechanism of the ontology to infer the instances to a class based on the constraint given. For eg. For the ***covid-19*** disease instance it belongs to both ***ChestDisease*** which is a subclass of ***DiseaseByLocation*** and ***VirusInfection*** which is a subclass of ***DiseaseByMechanism->InfectiousDisease.*** We used the reasoning to infer the instance to the ***ChestDisease*** class using the following constraint
DiseaseByLocation and (location some ChestOrgan). Since this instance has location lungs which is a ***ChestOrgan*** it infers to the ***ChestDisease*** class.
For the ***VirusInfection*** class we defined it explicitly for the ***covid-19*** disease instance. Below figure shows the same in Protege.



Figure(11): *Shows the inferred instances for ChestDisease with the reasoner on*



Figure(12): *Shows the inferred Class and the explicitly defined classes with reasoner on*

Question 1:Identify existing ontology on the same topic. How is your ontology different from it?

Ans: The relevant existing ontology in this domain is the “Disease Ontology”. The Disease Ontology has been developed as a standardized ontology for human disease with the purpose of providing the biomedical community with consistent, reusable and sustainable descriptions of human disease terms, phenotype characteristics and related medical vocabulary disease concepts through collaborative efforts of biomedical researchers, coordinated by the University of Maryland School of Medicine, Institute for Genome Sciences.

Our Ontology is different from the existing Disease Ontology in the following ways:

- Our Ontologies gives the details of the medical specialisations that deal with the particular diseases, organs and symptoms.
- Our ontology gives remedies for the users based on symptoms .
- Our ontology gives possible treatments for the diseases.
- Our ontology gives the prescribed quantity of the medicines for the diseases.
- Our ontology classifies the regular diseases from serious diseases using the reasoning.
- Our ontology gives the vaccines of the diseases for which the vaccinations are available. This is also done using the reasoning.
- Our ontology also gives the dosage of the vaccinations.
- Our ontology gives the details of the hospitals that deal with different medical specialities.

Question 2: How are your CQ / SPARQL different from simple database queries?

Ans: These CQs and SPARQLs are different from simple database queries as they use the reasoning power of an ontology. SPARQL is more suitable and easier than database queries as they don’t have to search through different tables that need to be linked while ontology uses class and subclass hierarchy along with properties to link the classes which is much easier to implement. In the case of our CQs, implementing them with database queries will be much more difficult as these queries don’t support the use of class hierarchy and relationships. Also in case of databases everything needs to be explicitly defined and reasoning cannot be used.

Question 3: When to use class/subclass or instance?

Ans: Classes can have instances. Thus we used classes when we wanted to categorise the instances. Classes are more generalized and instances are more specialized. For eg: *AbdominalDisease* is a class that can have many specific instances of diseases particular to the abdominal region of the body such as *appendicitis, diarrhoea, HepatitisB*. Thus classes and instances serve different purposes. When we pose an SPARQL queries the instances of a particular class are retrieved. Subclasses can be used to more specialize the categories For eg: the *Disease* class has subclasses *DiseaseByLocation* and *DiseaseByMechanism* which divides

the diseases based on the organ they affect and the means they get transferred. It's a good habit to use classes and subclasses in an ontology and populate them with instances.

Question 4: Identify how reasoning mechanisms are integrated into your ontology and CQ?

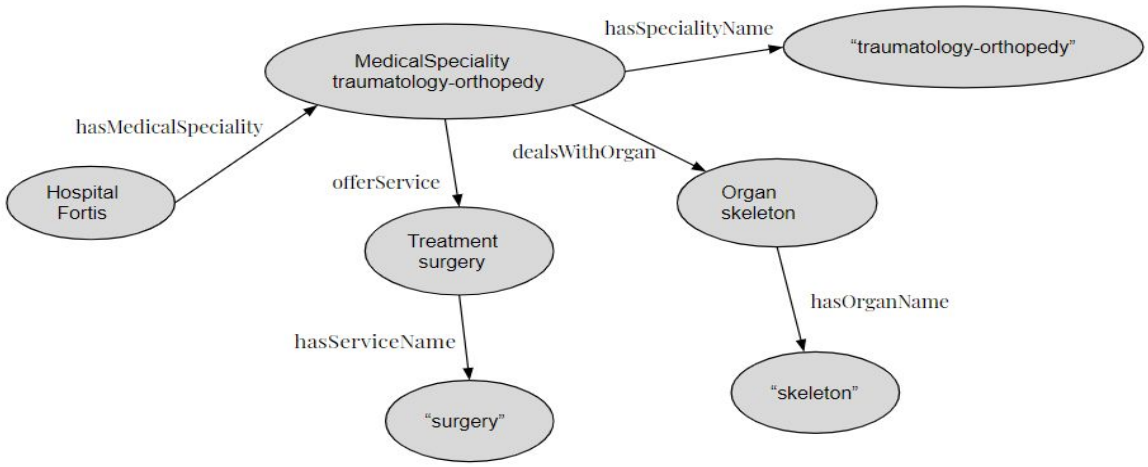
Ans: We address this question in the ***Section 3 of Class Hierarch***y and the ***Section 9 of Competency Questions answered Using SPARQL***

8. Instances

The concepts of the ontology were populated with instances relevant to the classes. The below figure gives some of the instances of the classes.

Example Instances:

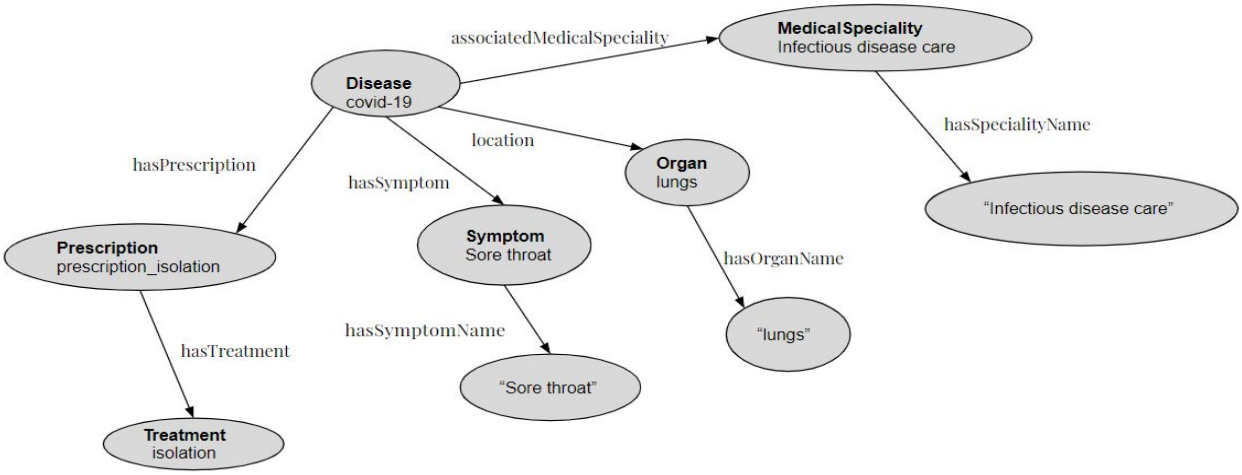
a. **MedicalSpeciality Instance**
   This instance describes a Hospital. We can notice its interaction with other top-level concepts such as MedicalSpeciality and Hospital.



Figure(13)

b. Disease Instance

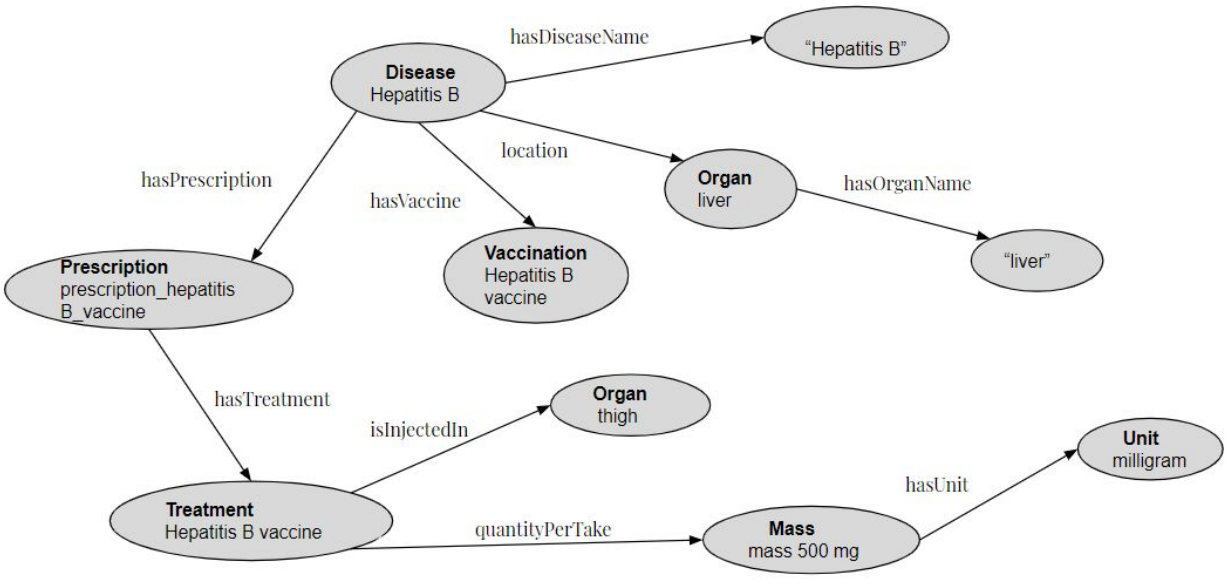
This instance describes a Disease that consists of its MedicalSpeciality, Symptoms, Prescription as subconcepts.



Figure(14)

c. Prescription Instance

This instance describes the Prescription for a Disease along with what treatment is given in Prescription and what are the details of the treatment.



Figure(15)

9. Competency Questions answered using SPARQL:

CQ1. Swaroop is showing symptoms of common cold, what possible medications can he take to subdue them?

SPARQL query:

```
PREFIX rdf: <http://www.w3.org/1999/02/22-rdf-syntax-ns#>
PREFIX owl: <http://www.w3.org/2002/07/owl#>
PREFIX rdfs: <http://www.w3.org/2000/01/rdf-schema#>
PREFIX xsd: <http://www.w3.org/2001/XMLSchema#>
PREFIX dis:<http://www.semanticweb.org/lenovo/ontologies/2020/6/disease#>
SELECT ?symptom ?medication
WHERE
{
?name a dis:Disease.
?name dis:hasDiseaseName "common cold"^^xsd:string.
?name dis:hasSymptom ?symptom.
?symptom dis:takeRemedy ?medication
```

symptom	medication
cough	benadryl
headache	aspirin
runny_nose	vicks_vaporub

CQ2. Kiran has abdominal pain, vomit and rashes on the skin. What may be the possible diseases?

SPARQL query:

```
PREFIX rdf: <http://www.w3.org/1999/02/22-rdf-syntax-ns#>
PREFIX owl: <http://www.w3.org/2002/07/owl#>
PREFIX rdfs: <http://www.w3.org/2000/01/rdf-schema#>
PREFIX xsd: <http://www.w3.org/2001/XMLSchema#>
PREFIX dis:<http://www.semanticweb.org/lenovo/ontologies/2020/6/disease#>
SELECT distinct ?name ?symptom
WHERE
{
?name a dis:Disease.
?name dis:hasSymptom ?symptom.
?symptom dis:hasSymptomName ?symname.
FILTER(?symname = "rashes"^^xsd:string || ?symname = "abdominal pain"^^xsd:string || ?symname = "vomit").
}
```

name	symptom
scarlet_fever	rashes
diarrhoea	abdominal_pain
scarlet_fever	abdominal_pain
scarlet_fever	vomit
appendicitis	abdominal_pain
appendicitis	vomit
measles	rashes

CQ3.(USES REASONING) Vamsi is having rashes, find a specialty hospital for him?

In this we used the class ***SpecialityHospital*** that uses reasoning.

Snap SPARQL Query:

```
PREFIX owl: <http://www.w3.org/2002/07/owl#>
PREFIX rdf: <http://www.w3.org/1999/02/22-rdf-syntax-ns#>
PREFIX rdfs: <http://www.w3.org/2000/01/rdf-schema#>
PREFIX dis:<http://www.semanticweb.org/lenovo/ontologies/2020/6/disease#>
SELECT distinct ?hospital ?speciality
WHERE
{
?symptom a dis:Symptom.
?symptom dis:hasSymptomName "rashes"^^xsd:string.
?symptom dis:associatedMedicalSpeciality ?s1.
?hospital a dis:SpecialityHospital.
?hospital dis:hasMedicalSpeciality ?speciality.
FILTER(?s1 = ?speciality).

}
```

Execute

?hospital	?speciality
dis:Venus_skin_clinic	dis:dermatology

CQ4. James forgot the prescription given by the doctor for COVID-19, what possible prescription can the system give him?

SPARQL query:

```
PREFIX rdf: <http://www.w3.org/1999/02/22-rdf-syntax-ns#>
PREFIX owl: <http://www.w3.org/2002/07/owl#>
PREFIX rdfs: <http://www.w3.org/2000/01/rdf-schema#>
PREFIX xsd: <http://www.w3.org/2001/XMLSchema#>
PREFIX dis:<http://www.semanticweb.org/lenovo/ontologies/2020/6/disease#>
SELECT  ?name  ?prescription ?treatment ?prescriptionDetails
WHERE
{
?name dis:hasDiseaseName "covid-19"^^xsd:string.
?name dis:hasPrescription ?prescription.
?prescription dis:hasTreatment ?treatment.
OPTIONAL{?prescription dis:hasPrescriptionDetails ?prescriptionDetails.}
}
```

name	prescription	treatment	prescriptionDetails
covid-19	prescription_hydrochloroquine_1	hydrocholoroquine_200mg	"hydrochloroquine 200 mg take 2 times a day for 1 month"^^
covid-19	prescription_isolation	isolation	
covid-19	prescription_covid-19_test	covid-19_test	"testing symptoms of covid-19 using fast testing kit"^^<http

CQ5. Sahapawn is experiencing cephalalgia which specialty should she go to and what remedies can she take?

SPARQL query:

```
PREFIX rdf: <http://www.w3.org/1999/02/22-rdf-syntax-ns#>
PREFIX owl: <http://www.w3.org/2002/07/owl#>
PREFIX rdfs: <http://www.w3.org/2000/01/rdf-schema#>
PREFIX xsd: <http://www.w3.org/2001/XMLSchema#>
PREFIX
dis:<http://www.semanticweb.org/lenovo/ontologies/2020/6/disease#>
SELECT distinct ?symptom ?speciality ?remedy
WHERE
{{
?symptom a dis:Symptom.
?symptom dis:hasSymptomName "cephalagia"^^xsd:string.
?symptom dis:associatedMedicalSpeciality ?speciality.
}
UNION
{
?symptom dis:hasSymptomName "cephalagia"^^xsd:string.
?symptom dis:takeRemedy ?remedy.
}
}
```

symptom	speciality	remedy
cephalalgia	neurology	
cephalalgia		aspirin
cephalalgia		paracetamol_500

CQ6. Adnan just witnessed an accident and wants to find out hospitals that offer emergency trauma care in the city.

SPARQL query:

```
PREFIX rdf: <http://www.w3.org/1999/02/22-rdf-syntax-ns#>
PREFIX owl: <http://www.w3.org/2002/07/owl#>
PREFIX rdfs: <http://www.w3.org/2000/01/rdf-schema#>
PREFIX xsd: <http://www.w3.org/2001/XMLSchema#>
PREFIX dis:<http://www.semanticweb.org/lenovo/ontologies/2020/6/disease#>
SELECT  distinct ?hospital ?speciality ?treatment
WHERE
{
?hospital a dis:Hospital.
?hospital dis:hasMedicalSpeciality ?speciality.
?speciality dis:hasSpecialityName "traumatology-orthopedy"^^xsd:string.
?speciality dis:offerService ?treatment.
}
```

hospital	speciality	treatment
Fortis	traumatology-orthopedy	plaster
Fortis	traumatology-orthopedy	surgery

CQ7. Alex's grandmother is in stage 1 lung cancer. He wants to know how long are the duration and frequency and what are the treatments for lung cancer and which hospital offers those treatments?

SPARQL query:

```
PREFIX rdf: <http://www.w3.org/1999/02/22-rdf-syntax-ns#>
PREFIX owl: <http://www.w3.org/2002/07/owl#>
PREFIX rdfs: <http://www.w3.org/2000/01/rdf-schema#>
PREFIX xsd: <http://www.w3.org/2001/XMLSchema#>
PREFIX dis: <http://www.semanticweb.org/lenovo/ontologies/2020/6/disease#>
SELECT  distinct ?name ?treatment ?details ?hospital
{
?name a dis:Disease.
?name dis:hasDiseaseName "lung cancer"^^xsd:string.
?name dis:associatedMedicalSpeciality ?s1.
?hospital a dis:Hospital.
?hospital dis:hasMedicalSpeciality ?s2.
FILTER(?s1=?s2).
?name dis:hasPrescription ?prescription.
?prescription dis:hasTreatment ?treatment.
?prescription dis:hasPrescriptionDetails ?details
}
```

name	treatment	details	hospital
lung_cancer	chemotherapy	"chemotherapy for 1 month every week"^^<http://ww	Tata_Memorial_Cancer_Hospital
lung_cancer	surgery	"operation is performed to remove tumor in lungs"^	Tata_Memorial_Cancer_Hospital
lung_cancer	radiation_therapy	"radiation therapy for 1 month every 2 weeks"^^<htt	Tata_Memorial_Cancer_Hospital

CQ8. Shantanu went back to India from Thailand during the pandemic and now is worried about getting COVID-19. He wants to know the possible symptoms for COVID-19 and where he can get his covid-19 test done.

SPARQL query:

```
PREFIX rdf: <http://www.w3.org/1999/02/22-rdf-syntax-ns#>
PREFIX owl: <http://www.w3.org/2002/07/owl#>
PREFIX rdfs: <http://www.w3.org/2000/01/rdf-schema#>
PREFIX xsd: <http://www.w3.org/2001/XMLSchema#>
PREFIX dis:<http://www.semanticweb.org/lenovo/ontologies/2020/6/disease#>
SELECT  distinct  ?symptoms ?hospital ?test
{{
?name a dis:Disease.
?name dis:hasDiseaseName "covid-19"^^xsd:string.
?name dis:hasSymptom ?symptoms.}
UNION{
?hospital a dis:Hospital.
?hospital dis:offerService ?test.
?test dis:hasServiceName "covid-19 test"^^xsd:string.}
}
```

symptoms	hospital	test
fever		
cough		
tiredness		
sore_throat		
	Mediquest	covid-19_test

CQ9.(USES REASONING) Su slipped on the wet floor and fractured her arm, where should she go and what possible treatment can she get?

In this also we use the class ***SpecialityHospital*** that uses reasoning.

Snap SPARQL Query:

```
PREFIX owl: <http://www.w3.org/2002/07/owl#>
PREFIX rdf: <http://www.w3.org/1999/02/22-rdf-syntax-ns#>
PREFIX rdfs: <http://www.w3.org/2000/01/rdf-schema#>
PREFIX dis:<http://www.semanticweb.org/lenovo/ontologies/2020/6/disease#>
SELECT  distinct ?name ?speciality ?hospital
WHERE
{
?name a dis:Disease.
?name dis:hasDiseaseName "fracture"^^xsd:string.
?name dis:associatedMedicalSpeciality ?speciality.
?hospital a dis:SpecialityHospital.
?hospital dis:hasMedicalSpeciality ?s2.
FILTER(?s2 = ?speciality)
}
```

Execute

?name	?speciality	?hospital
dis:fracture	dis:traumatology-orthopedy	dis:Fortis

CQ10.(USES REASONING) What vaccine should be given for the prevention of Hepatitis B and what is the dosage amount and where is it injected?

In this we use the class ***VaccinableDisease*** that uses reasoning

Snap SPARQL Query:

```
PREFIX owl: <http://www.w3.org/2002/07/owl#>
PREFIX rdf: <http://www.w3.org/1999/02/22-rdf-syntax-ns#>
PREFIX rdfs: <http://www.w3.org/2000/01/rdf-schema#>
PREFIX dis:<http://www.semanticweb.org/lenovo/ontologies/2020/6/disease#>
SELECT  distinct ?name ?prescription  ?vaccine ?dosage ?givenAt ?details
WHERE
{
?name a dis:VaccinableDiseases.
?name dis:hasDiseaseName "hepatitis B"^^xsd:string.
?name dis:hasPrescription ?prescription.
?prescription dis:hasTreatment ?vaccine.
?vaccine dis:quantityPerTake ?dosage.
?vaccine dis:isInjectedIn ?givenAt.
?prescription dis:hasPrescriptionDetails ?details.
}
```

Execute

?name	?prescription	?vaccine	?dosage	?givenAt	?details
dis:hepatitisB	dis:prescription_hepatitis_B_vaccine	dis:Hepatitis_B_vaccine	dis:mass_500_mg	dis:thigh	Hepatitis B vaccine is given once at the birth^...

CQs and SPARQL using reasoning

For using reasoning in SPARQL we used the Snap SPARQL Query tab in Protege. All the below queries are executed with the HermiT reasoner on.

EXTRA CQ: Get the list of all the vaccines to be given to children after birth along with their dosage and details.

In this we use the class ***VaccinableDiseases*** use reasoning.

Snap SPARQL Query:

```
PREFIX owl: <http://www.w3.org/2002/07/owl#>
PREFIX rdf: <http://www.w3.org/1999/02/22-rdf-syntax-ns#>
PREFIX rdfs: <http://www.w3.org/2000/01/rdf-schema#>
PREFIX dis:<http://www.semanticweb.org/lenovo/ontologies/2020/6/disease#>
PREFIX dis:<http://www.semanticweb.org/lenovo/ontologies/2020/6/disease#>
SELECT distinct ?name ?vaccine ?details ?dosage ?location
WHERE
{
?name a dis:VaccinableDiseases.
?name dis:hasVaccine ?vaccine.
?name dis:hasPrescription ?prescription.
?prescription dis:hasPrescriptionDetails ?details.
?vaccine dis:quantityPerTake ?dosage.
?vaccine dis:isInjectedIn ?location.

}
```

Execute

?name	?vaccine	?details	?dosage	?location
dis:Polio	dis:OPV_Booster	1. At birth or as early as possible within the first 15 days 2. At 6 weeks, 10 weeks & 14 weeks 3. 16-24 months ...	dis:mass_200_mg	dis:oral
dis:measles	dis:Measles_vaccine	1. 9 completed months-12 months. (give up to 5 years if not received at 9-12 months age) 2. 16-24 Months^^xs...	dis:mass_500_mg	dis:upper_arm
dis:tetanus	dis:TT_Booster	Given at the age 10 and 16^^xsd:string	dis:mass_500_mg	dis:upper_arm
dis:hepatitisB	dis:Hepatitis_B_vaccine	Hepatitis B vaccine is given once at the birth^^xsd:string	dis:mass_500_mg	dis:thigh

EXTRA CQ The daily health magazine wants to know the hospital that handles serious diseases along with the speciality associated with the disease and what prescriptions are given for these diseases.

In this we use the classes ***SeriousDisease*** and ***SpecialityHospital*** use reasoning.

Snap SPARQL Query:

```
PREFIX owl: <http://www.w3.org/2002/07/owl#>
PREFIX rdf: <http://www.w3.org/1999/02/22-rdf-syntax-ns#>
PREFIX rdfs: <http://www.w3.org/2000/01/rdf-schema#>
PREFIX dis:<http://www.semanticweb.org/lenovo/ontologies/2020/6/disease#>
SELECT distinct ?hospital ?disease ?speciality ?prescription
WHERE
{
?disease a dis:SeriousDisease.
?disease dis:hasPrescription ?prescription.
?disease dis:associatedMedicalSpeciality ?speciality.
?hospital a dis:SpecialityHospital.
?hospital dis:hasMedicalSpeciality ?s.
FILTER(?speciality = ?s).

}
```

Execute

?hospital	?disease	?speciality	?prescription
dis:Medanta	dis:heart_attack	dis:cardiology	dis:prescription_angioplasty
dis:Medanta	dis:heart_attack	dis:cardiology	dis:prescription_bypass_surgery
dis:Tata_Memorial_Cancer_Hospital	dis:lung_cancer	dis:cancer-care	dis:prescription_chemotherapy
dis:Tata_Memorial_Cancer_Hospital	dis:lung_cancer	dis:cancer-care	dis:prescription_surgery
dis:Tata_Memorial_Cancer_Hospital	dis:lung_cancer	dis:cancer-care	dis:prescription_radiotherapy
dis:Tata_Memorial_Cancer_Hospital	dis:bladder_cancer	dis:cancer-care	dis:prescription_chemotherapy
dis:Tata_Memorial_Cancer_Hospital	dis:bladder_cancer	dis:cancer-care	dis:prescription_surgery
dis:Tata_Memorial_Cancer_Hospital	dis:bladder_cancer	dis:cancer-care	dis:prescription_radiotherapy
dis:Apollo	dis:appendicitis	dis:gastrology	dis:prescription_paracetamol_1
dis:Apollo	dis:appendicitis	dis:gastrology	dis:prescription_surgery

EXTRA CQ What head and neck disease have the symptoms cephalalgia and fever?

In this we use the class ***HeadAndNeckDisease*** uses reasoning.

Snap SPARQL Query:

```
PREFIX owl: <http://www.w3.org/2002/07/owl#>
PREFIX rdf: <http://www.w3.org/1999/02/22-rdf-syntax-ns#>
PREFIX rdfs: <http://www.w3.org/2000/01/rdf-schema#>
PREFIX dis:<http://www.semanticweb.org/lenovo/ontologies/2020/6/disease#>
SELECT ?disease ?symptom
WHERE
{
?disease a dis:HeadAndNeckDisease.
?disease dis:hasSymptom ?symptom.
?symptom dis:hasSymptomName ?symname.
FILTER(?symname="cephalalgia"^^xsd:string || ?symname="fever"^^xsd:string).

}
```

Execute

?disease	?symptom
dis:common_cold	dis:fever
dis:covid-19	dis:fever
dis:mumps	dis:cephalalgia
dis:mumps	dis:fever

10. Ontology Usage

- Our ontology gives the names of the possible diseases for the given symptoms.
- Our ontology gives the name of the medical speciality that treats a particular disease.
- Our ontology gives possible treatments for various diseases.
- Our ontology gives the names of the hospitals with respect to the medical speciality requirement of the users.
- Our ontology gives the medications for a disease regarding its various symptoms separately.
- Our ontology also includes self remedies that self remedies that people can use to subdue minor symptoms.
- Our ontology gives the list of vaccinations to be given to newborn babies.

11. Strengths and Weaknesses of Design

Strengths

- Our ontology has many classes and subclasses which covers various diseases,symptoms and their treatments classification.
- Our ontology is populated with many instances to make it more realistic.
- Our ontology makes use of reasoning to group the instances based on the axioms eg:Vaccinable diseases.

Limitations and Future Works

- Our ontology does not cover all the diseases.
- Our ontology can add more information about the diseases with some more detailed description.
- Our ontology includes instances for hospitals from a single city, it can be extended further.
- Our ontology is for reference purpose only, for serious cases people should consult a medical professional.
- We can add more instances for a version 2.0 along with more constraints.
- It can be expanded to a web or mobile application with an interface.

12. REFERENCES

Disease Ontology - Institute for Genome Sciences at the University of Maryland School of Medicine.
https://disease-ontology.org/

Wikipedia for details of disease
www.wikipedia.org

Apollo hospital website for diseases, symptoms and treatments
www.apollohospitals.com