1 An use case for DAML+OIL: an ontology in the ophthalmology domain

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1.1 Introduction

This report describes how we have developed a medical ontology for a heuristic application in the ophthalmology domain. This ontology has been built by reusing pre-existing knowledge theories, and it has been developed in two phases:

1) Firstly, the ontology has been modeled using the domain knowledge schema specification of CommonKADS [Schreiber et al., 1999], which in turn uses Unified Modelling Language (UML) [Rumbaugh et al., 1999] in order to represent graphically the objects, class and relations of the domain knowledge.
2) Currently, the ontology is being represented in DAML+OIL [Fensel et al., 2001].

In this report, we will illustrate in some depth how a medical ontology has been developed for a specific domain and a specific application, by reusing pre-existing medical theories. In addition, we will show how objects, classes and relations of a medical domain (usually in any object-oriented approach) can be specified in the Web language DAML+OIL.

1.2 The clinical domain

Our medical ontology has been modeled for a heuristic application in the domain of the 'Red Eye' (RE) diagnosis. The latter is a common activity carried out in primary care units. However, a lot of general practitioners have difficulty to make a precise diagnosis. So, some recommendations oriented to facilitate this kind of diagnosis have been provided [Davey, 1996; Bertolini and Pelucio, 1995]. These advices are focussed on helping to the physicians to:

1) Distinguish appropriately between minor and serious causes.

2) Encourage him to treat minor problems themselves, as most cases are benign and can be managed efficiently by him, such as *Conjunctivitis, Blepharitis, Subconjunctival Hemorrage or Episcleritis*.
3) Refer appropriately those cases that require ophtalmologic consultation, such as *Acute Angle Closure Glaucoma, Keratopathy, Uveitis or Scleritis*, which must be refered as soon as possible.

Recommendations about carrying out the diagnosis of the red eye include to take a careful history and to make a focused ophthalmology examination. Following these recommendations, a diagnosis-aid system in this clinical domain should guide to the practitioner on, at least,

1) carrying out the anamnesis,
2) directing the ophthalmology examination,
3) providing advice about diagnostic hypothesis, and as a result of this,
4) suggesting actions to carry out, such as to treat the patient, to refer him to the specialist and/or to require some complementary test.

Taking into account this, the medical ontology must include, at least, knowledge about the ophthalmology anamnesis, examination, pathologies and relations among all of this knowledge.

1.3 Developing and specifying the ophthalmology ontology

Our domain ontology has been developed by reusing some theories in the core library described in [Falasconi and Stefanelli, 1994], and later, by extending these theories with some descriptions taken from INTERNIST-I [Miller et al., 1982] and with more specific concepts to our clinical domain. We have also followed the ICD-9-MC' classification (The International Classification of Diseases, 9th Revision , Clinical Modification, 1978, Commission on Profesional and Hospital Activities, Ann Harbor. Michigan 48705), for representing pathologies. Currently, there is a 10th Revision, but the 9th Revision is still followed in Spain. In this way, we have obtained a standard representation vocabulary, which is being revised by following UMLS (Unified Medical Language Standard).

The library of medical ontologies of [Falasconi and Stefanelli, 1994] specifies a set of medical conceptualizations taken from both medical literature and implemented systems, including general categories of medical knowledge, such as the following theory levels:

- ✓ *Generic Patient*, which models the medical activities for each patient,
- ✓ *Test* and *Therapies*, representing actions undertaken by medical agents,

- ✓ *Diseases*, modelling each disease as a clinical process whose evolution is described through finding and clinical abstraction values over time,
- ✓ *Findings*, *Clinical-state-abstractions*, *Drugs*, etc.

Fig. 1 shows a small part of our domain ontology. The higher levels in this ontology correspond to the core ontological part (labelled as Medical Central Ontology, MCO), following the approach proposed by [Heijst et al., 1997]. MCO includes the reused general categories of medical knowledge, such as 'Generic-Patient', 'Tests', 'Diseases', 'Findings', 'Clinical-State-Abstractions', etc. As the hierarchical structures into MCO are traversed, the medical concepts are more specific to our clinical domain, giving rise to the peripheral ontological part (labelled as Medical Peripheral Ontology, MPO). In particular, Fig. 1 shows some concepts from our ontology with regard to some medical theories. In this report, we will focus in Generic-Patient and Test, in order to describe the most important aspects used during the specification in DAML+OIL.

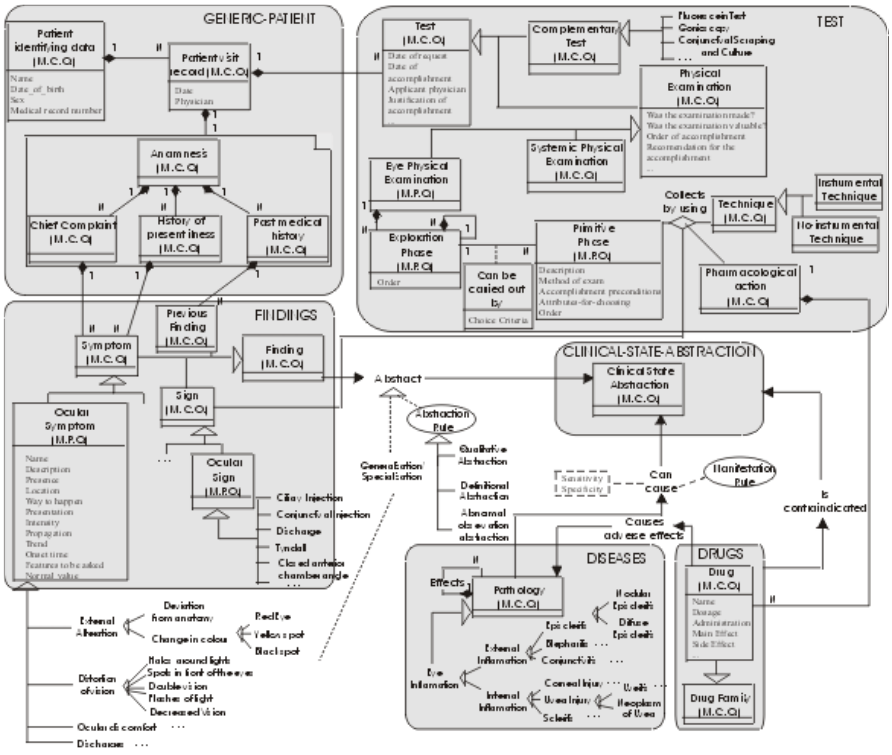


Figura 1: A Small Part of our Medical Domain Level

1.3.1 Generic Patient

Generic-Patient models the information to record during each patient visit, such as patient identifying data, patient visit record and the anamnesis. The latter includes chief complaint, history of present illness and past medical history.

In DAML+OIL, objects are described by giving a name for the object class. The following text box shows the definition of the set of classes in Generic-Patient.

```
<daml:Class
rdf:ID="PatientIdentifyingData">
</daml:Class>
<daml:Class rdf:ID="PatientVisitRecord">
</daml:Class>
<daml:Class rdf:ID="Anamnesis">
</daml:Class>
<daml:Class rdf:ID="ChiefComplaint">
</daml:Class>
<daml:Class
rdf:ID="HistoryOfPresentIllness">
</daml:Class>
```

The set of attributes, which describes each class of objects, is defined by a kind of DAML+OIL property (named *daml:DatatypeProperty*), which relates objects to datatype values. For example, we have create the attribute *Name*, which maps strings into XML Schema,

```
<daml:DatatypeProperty rdf:ID="Name">
<rdfs:comment>
  Name is a DatatypeProperty whose range is
  xsd:string.
  Name is also a UniqueProperty (can only have
  one name)
</rdfs:comment>
<rdf:type
rdf:resource="http://www.daml.org/2001/03/daml+oi
l#UniqueProperty"/>
<rdfs:range
rdf:resource="http://www.w3.org/2000/10/XMLSchema
#string"/>
</daml:DatatypeProperty>
```

or the attribute *Date_of_Birth*, which is a subproperty of the *Date* property. The latter maps dates into XML Schema.

```
<daml:DatatypeProperty rdf:ID="Date">
<rdfs:comment>
  Date is a DatatypeProperty whose range is
  xsd:date.
</rdfs:comment>
<rdfs:range
rdf:resource="http://www.w3.org/2000/10/XMLSchema#d
ate"/>
</daml:DatatypeProperty>
<daml:DatatypeProperty rdf:ID="DateOfBirth">
<rdfs:comment>
DateOfBirth is a subProperty Of Date and it is also
a UniqueProperty (can only have one date)
</rdfs:comment>
<rdfs:subPropertyOf rdf:resource="#Date"/>
<rdf:type
rdf:resource="http://www.daml.org/2001/03/daml+oil#
UniqueProperty"/>
</daml:DatatypeProperty>
```

Figure 2 shows how we can describe the internal structure of a class by using property restrictions. These restrictions can be added to the class by using *<daml:Class rdf:about=”#PatientIdentifyingData”>*.

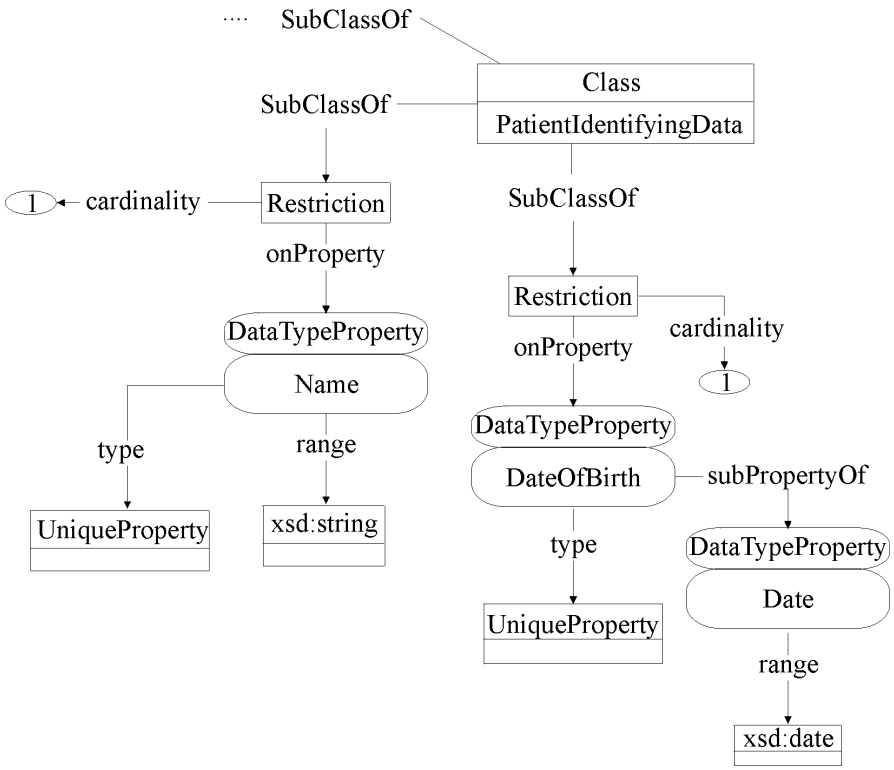


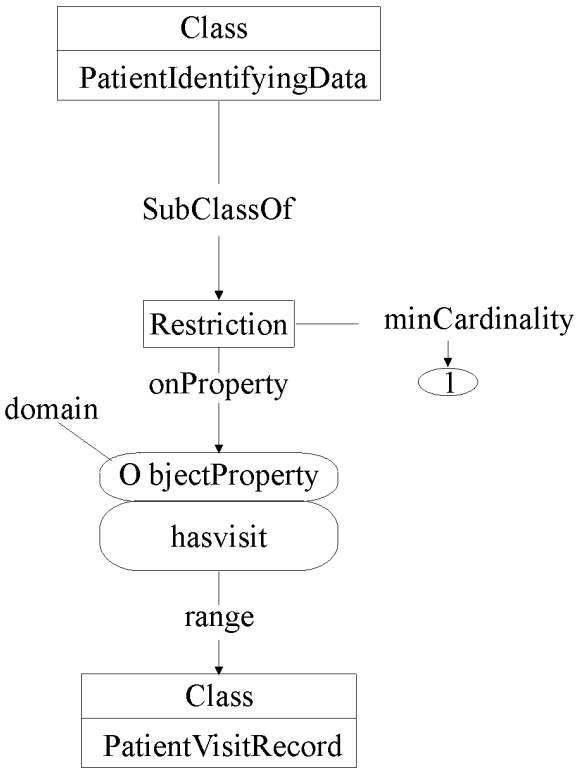
Fig 2: Adding two property restrictions to the class PatientIdentifyingData

In an object-oriented approach, one standard relation is an aggregation. For example, for each patient there can be one or several visit records. This information can be modeled by an aggregation relation 1:N between *PatientIdentifyingData* and *PatientVisitRecord*. We have specified this kind of relation by:

1) defining a new property *hasvisit*, which is a binary relation connecting the two classes implied in the aggregation.

```
<daml:ObjectProperty rdf:ID="hasVisit">
  <rdfs:domain
  rdf:resource="#PatientIdentifyingData"/>
  <rdfs:range rdf:resource="#PatientVisitRecord"/>
</daml:ObjectProperty>
```

2) adding a new property restriction on the class *PatientIdentifyingData*, such as is displayed in Fig. 3, where a minimum cardinality of 1 is specified as the aggregation relation is 1:N.



If a class implied in an aggregation relation is defined in another file, the class must be referred with a fully qualified URL. For example, the file http://aiff.usc.es/~elchus/daml+oil-medicalontology/daml+oil-gp.daml

defines all concepts regarding to Generic-Patient. So, in order to define the aggregation between *PatientVisitRecord* and *Test*, we have defined a new property (*madeTests*) and added it to *PatientVisitRecor* as a new restriction.

```
<daml:ObjectProperty rdf:ID="madeTests">
<rdfs:domain rdf:resource="#PatientVisitRecord"/>
<rdfs:range
rdf:resource="http://aiff.usc.es/elchus/medicalonto
logy/daml+oil-test.daml#Test"/>
</daml:ObjectProperty>

  <daml:Class rdf:about="PatientVisitRecord">
  <rdfs:comment>
  Several tests can be made or recommended for
  their accomplishment during each patient visit,
  ie: Zero or more tests can be aggregated to one
  'PatientVisitRecord' (through the object property
  'madeTests').
  </rdfs:comment>
  <rdfs:subClassOf>
     <daml:Restriction daml:minCardinality="0">
       <daml:onProperty rdf:resource="#madeTests"/>
     </daml:Restriction>
  </rdfs:subClassOf>
  </daml:Class>
```

1.3.2 Test

We have specified the set of following classes and their hierarchies (some of these disjoint), according to the theory level *Test* (see Fig. 1).

```
<daml:Class rdf:ID="Test">
</daml:Class>

<daml:Class rdf:ID="ComplementaryTest">
<rdfs:subClassOf rdf:resource="#Test"/>
</daml:Class>

<daml:Class rdf:ID="PhysicalExamination">
<rdfs:subClassOf rdf:resource="#Test"/>
<daml:disjointWith
rdf:resource="#ComplementaryTest"/>
</daml:Class>

<daml:Class rdf:ID="EyePhysicalExamination">
<rdfs:subClassOf
rdf:resource="#PhysicalExamination"/>
</daml:Class>

<daml:Class rdf:ID="SystemicPhysicalExamination">
<rdfs:subClassOf
rdf:resource="#PhysicalExamination"/>
<daml:disjointWith
rdf:resource="#EyePhysicalExamination"/>
</daml:Class>

<daml:Class rdf:ID="ExplorationPhase">
</daml:Class>

<daml:Class rdf:ID="PrimitivePhase">
</daml:Class>

<daml:Class rdf:ID="Technique">
</daml:Class>

  <daml:Class rdf:ID="IntrumentalTechnique">
<rdfs:subClassOf rdf:resource="#Technique"/>
</daml:Class>

<daml:Class rdf:ID="No-IntrumentalTechnique">
<rdfs:subClassOf rdf:resource="#Technique"/>
<daml:disjointWith
rdf:resource="#IntrumentalTechnique"/>
</daml:Class>

<daml:Class rdf:ID="PharmacologicalAction">
</daml:Class>
```

Some properties, as *DateOfRequest*, *DateOfAccomplishment*, *ApplicantPhysician*, have been specified as subproperties of properties defined in 'Generic-Patient'.

```
<daml:DatatypeProperty rdf:ID="DateOfRequest">
<rdfs:comment>
  DateOfRequest is a subProperty Of Date, which
  has been defined en 'Generic-Patient', and it
  is also a UniqueProperty (can only have one
  date)
</rdfs:comment>
<rdfs:subPropertyOf
rdf:resource="http://aiff.usc.es/elchus/medicalon
tology/daml+oil-gp#Date"/>
<rdf:type
rdf:resource="http://www.daml.org/2001/03/daml+oi
l#UniqueProperty"/>
</daml:DatatypeProperty>

<daml:DatatypeProperty
rdf:ID="DateOfAccomplishment">
<rdfs:comment>
   DateOfAccomplishment is a subProperty Of Date,
  which has been defined en 'Generic-Patient',
  and it is also a UniqueProperty (can only have
  one date)
</rdfs:comment>
<rdfs:subPropertyOf
rdf:resource="http://aiff.usc.es/elchus/medicalon
tology/daml+oil-gp#Date"/>
<rdf:type
rdf:resource="http://www.daml.org/2001/03/daml+oi
l#UniqueProperty"/>
</daml:DatatypeProperty>

<daml:DatatypeProperty
rdf:ID="ApplicantPhysician">
<rdfs:comment>
  ApplicantPhysician is a subproperty of Identi-
  fyingNumber.
</rdfs:comment>
<rdfs:subPropertyOf
rdf:resource="#IdentifyingNumber"/>
<daml:equivalentTo
rdf:resource="http://aiff.usc.es/elchus/medicalon
tology/daml+oil-gp #PhysicianIdentifyingNumber"/>
</daml:DatatypeProperty>

<daml:DatatypeProperty
rdf:ID="JustificationOfAccomplishment">
<rdfs:comment>
  JustificationOfAccomplishment is a Dataty-
  peProperty whose range is xsd:string, and it is
  also a UniqueProperty (can only have one name)
</rdfs:comment>
<rdf:type
rdf:resource="http://www.daml.org/2001/03/daml+oi
```

Binary relations between objects (1:N cardinality) have been specified by defining a new *daml:ObjectProperty* and adding a new restriction on the class. For example in Fig. 1 the relation labeled as '*Can be carried out by*' between *Exploration Phase* and *Primitive Phase* in the Test theory, is a 1:N binary relation with one relation attribute, named '*Choice Criteria'*. Firstly, two new properties have been defined in this case: one for defining the binary relation and another for defining the relation attribute. Secondly, two restrictions have been added to the class *Exploration Phase*.

```
<daml:ObjectProperty rdf:ID="CarriedOutBy">
<rdfs:domain rdf:resource="#ExplorationPhase"/>
<rdfs:range rdf:resource="#PrimitivePhase"/>
</daml:ObjectProperty>

<daml:ObjectProperty rdf:ID="ChoiceCriteria">
<rdfs:domain rdf:resource="#ExplorationPhase"/>
<rdfs:range
rdf:resource="#OrderedListOfCriteria"/>
</daml:ObjectProperty>

<daml:Class rdf:about="ExplorationPhase">
<rdfs:comment>
 Several primitive phases can be carried out by a
method of examination, ie: One or more primitive
phases can be carried out to one 'Exploration-
Phase' (through the object property 'Car-
riedOutBy').
</rdfs:comment>
<rdfs:subClassOf>
<daml:Restriction daml:minCardinality="1">
<daml:onProperty rdf:resource="#CarriedOutBy"/>
</daml:Restriction>
</rdfs:subClassOf>
<rdfs:subClassOf>
<daml:Restriction daml:Cardinality="1">
<daml:onProperty rdf:resource="#ChoiceCriteria"/>
</daml:Restriction>
```

On the other hand, a n-ary relation has been specified by defining n-1 properties and adding n-1 restriction to one class. For example, in Fig. 1 the relation '*Collects by using'* is a 4-ary relation, which represents the knowledge about techniques to be used, pharmacological actions to be applied, and signs to be collected in each phase. Three properties have been specified: '*Collects'* between *PrimitivePhase* and *Sign*, '*ByUsing'* between *PrimitivePhase* and *Technique,* and '*ByApplying*' between *PrimitivePhase* and *PharmacologicalAction.*

```
<daml:ObjectProperty rdf:ID="Collects">
<rdfs:domain rdf:resource="#PrimitivePhase"/>
<rdfs:range
rdf:resource="http://aiff.usc.es/elchus/medicalonto
logy/daml+oil-f#Sign"/>
</daml:ObjectProperty>

<daml:ObjectProperty rdf:ID="ByUsing">
<rdfs:domain rdf:resource="#PrimitivePhase"/>
<rdfs:range rdf:resource="#Technique"/>
</daml:ObjectProperty>

<daml:ObjectProperty rdf:ID="ByApplying">
<rdfs:domain rdf:resource="#PrimitivePhase"/>
<rdfs:range rdf:resource="#PharmacologicalAction"/>
</daml:ObjectProperty>

<daml:Class rdf:about="PrimitivePhase">
<rdfs:comment> The knowledge about techniques,
pharmacological actions and signs to be collected
in each phase was represented by a 4-ary relation
in UML and it is specifying as the intersection of
several restrictions on the properties 'Collects',
'ByUsing' and 'ByApplying'.</rdfs:comment>
<rdfs:subClassOf>
<daml:Restriction daml:minCardinality="1">
  <daml:onProperty rdf:resource="#Collects"/>
</daml:Restriction>
</rdfs:subClassOf>
<rdfs:subClassOf>
  <daml:Restriction daml:minCardinality="1">
    <daml:onProperty rdf:resource="#ByUsing"/>
  </daml:Restriction>
</rdfs:subClassOf>
<rdfs:subClassOf>
<daml:Restriction daml:minCardinality="0">
  <daml:onProperty rdf:resource="#ByApplying"/>
</daml:Restriction>
</rdfs:subClassOf>
</daml:Class>
```

1.3.3 Relation between Findings and Clinical-State-Abstractions

Clinical-State-Abstractions are more abstract descriptions than findings, and are connected to findings by relations that map data from specific descriptions to higher level descriptions. These relations have been modeled as rule schemas [Schreiber et al., 1999], which are association relations between two expressions. An expression can be modeled as a class containing four slots [Schreiber at al., 2000]: 1) a concept/relation, 2) a slot, 3) an operator (such as equal, greater, etc.) and 4) a value. We have also considered that expressions can be nested by using logical operators (such as and, or). Two examples of abstraction rules are shown in Figure 5.

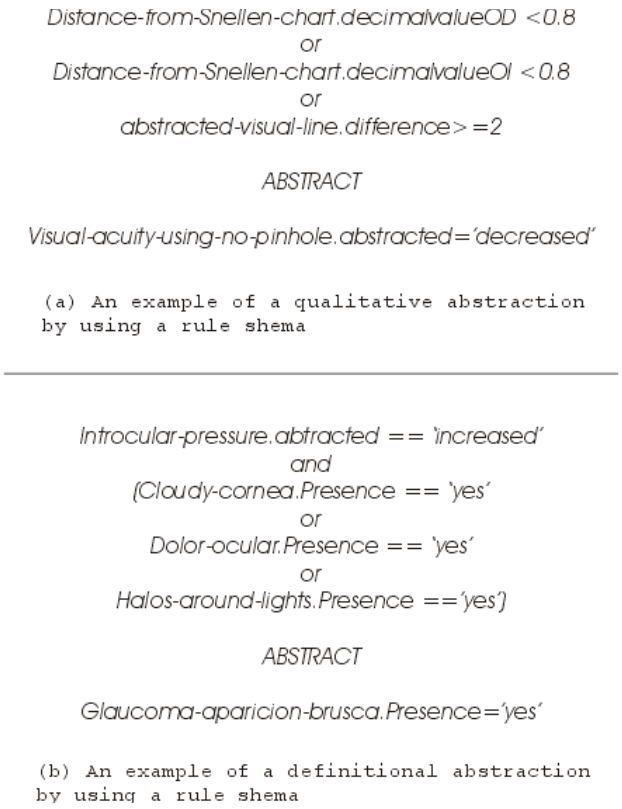


Fig. 5: Two examples of abstraction rules

Each expression, such as, '*Distance-from-Snellen-Chart.decimalvalueOD<0.8*' can be specified by defining a new class, for example, *DfSCOD-evaluation*, as an intersection of the class '*Distance-from-Snellen-Chart'* and a DAML+OIL restric-

tion on a property (in this case, *decimalvalueOD*). This restriction can use user-defined datatypes.

```
<daml:Class rdf:ID="DfSCOD-evaluation">
<daml:intersectionOf
rdf:parseType="daml:collection">
  <daml:Class rdf:about="#Distance-from-Snellen-
  Chart"/>
  <daml:Restriction daml:cardinality="1">
   <daml:onProperty
  rdf:resource="#decimalvalueOD"/>
   <daml:hasClass rdf:resource=
"http://aiff.usc.es/~elchus/medicalontology/daml+oi
l-test-dt#under18"/>
</daml:Restriction>
</daml:intersectionOf>
</daml:Class>

<daml:Class rdf:ID="DfSCOI-evaluation">
<daml:intersectionOf
rdf:parseType="daml:collection">
  <daml:Class rdf:about="#Distance-from-Snellen-
  Chart"/>
  <daml:Restriction daml:cardinality="1">
   <daml:onProperty
  rdf:resource="#decimalvalueOI"/>
   <daml:hasClass rdf:resource=
"http://aiff.usc.es/~elchus/medicalontology/daml+oi
l-test-dt#under18"/>
</daml:Restriction>
</daml:intersectionOf>
</daml:Class>

<daml:Class rdf:ID="AVLD-evaluation">
<daml:intersectionOf
rdf:parseType="daml:collection">
  <daml:Class rdf:about="#Abstracted-Visual-Line"/>
  <daml:Restriction daml:cardinality="1">
```

A set of nested expressions can be specified by means of the boolean combination provided by DAML+OIL, such as *daml:intersectionOf*, *daml:unionOf* and *daml:complementOf*.

```
<daml:Class rdf:ID=”Decresed-Visual-Acuity-Evaluation”>
 <daml:unionOf rdf:parseType=”daml:collection”>
  <daml:Class rdf:about=”#DfSCOD-evaluation”/>
  <daml:Class rdf:about=”#DfSCOI-evaluation”/>
  <daml:Class rdf:about=”#AVLD-evaluation”/>
</daml:unionOf>
</daml:Class>
```

A rule can be expressed by a name, an antecedent and a consequent:

```
<daml:Class rdf:about="Decresed-Visual-Acuity-Abstraction">
<rdfs:subClassOf>
<daml:Restriction daml:Cardinality="1">
  <daml:onProperty rdf:resource="
  http://aiff.usc.es/~elchus/medicalontology/daml+oil-gp#Name"/>
</daml:Restriction>
</rdfs:subClassOf>
<rdfs:subClassOf>
  <daml:Restriction daml:Cardinality="1">
    <daml:onProperty rdf:resource="#Decresed-Visual-Acuity-
  Antecedent"/>
  </daml:Restriction>
</rdfs:subClassOf>
<rdfs:subClassOf>
<daml:Restriction daml:Cardinality="1">
  <daml:onProperty rdf:resource="#Decresed-Visual-Acuity-
  Consequent"/>
</daml:Restriction>
</rdfs:subClassOf>
</daml:Class>
```

where the range of the antecedent is the class *Decresed-Visual-Acuity-Evaluation* and the range of the consequent is the class *Abstracted-Decresed-Visual-Acuity.*

```
<daml:ObjectProperty rdf:ID="Decresed-Visual-Acuity-
Antecedent">
  <rdfs:domain rdf:resource="#Decresed-Visual-Acuity-
  Antecedent"/>
  <rdfs:range rdf:resource="#Decresed-Visual-Acuity-
  Evaluation"/>
</daml:ObjectProperty>

<daml:ObjectProperty rdf:ID="Decresed-Visual-Acuity-
Consequent">
  <rdfs:domain rdf:resource="#Decresed-Visual-Acuity-
  Consequent"/>
  <rdfs:range rdf:resource="#Abstracted-Decresed-
  Visual-Acuity"/>
</daml:ObjectProperty>
```