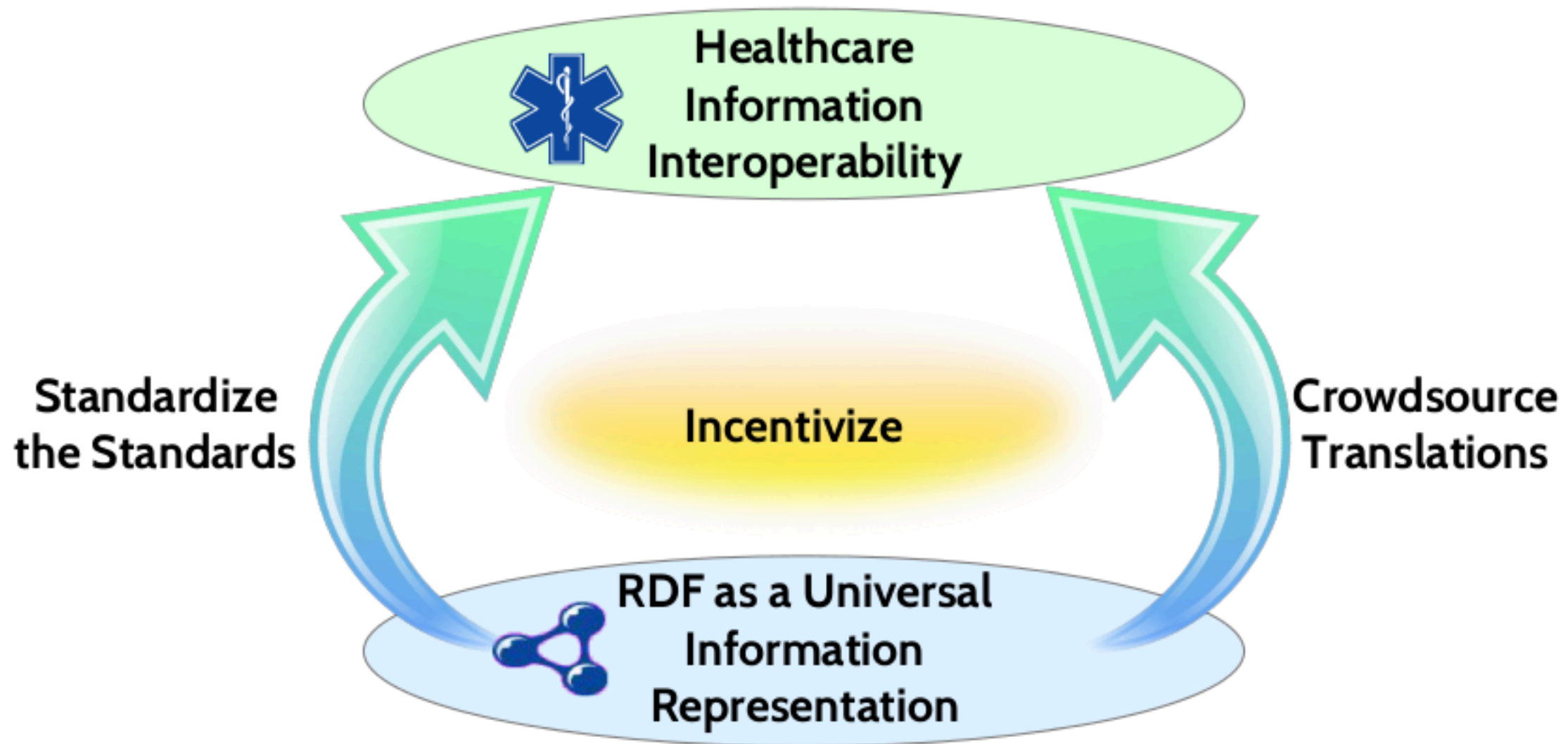


FHIR RDF as a Bridge to the Semantic Web in Healthcare

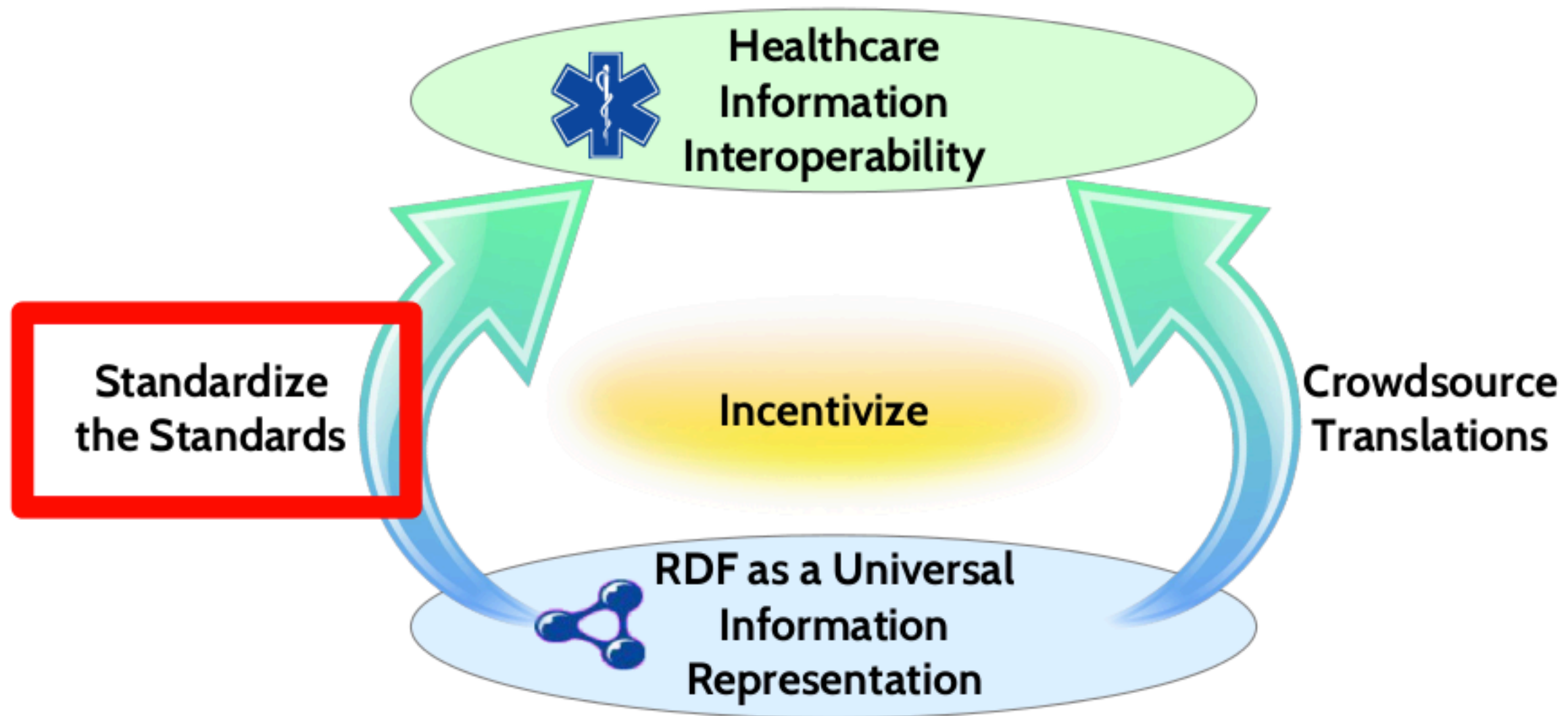
Harold Solbrig
Mayo Clinic

Interoperability Roadmap



<http://YosemiteProject.org/>

Interoperability Roadmap



<http://YosemiteProject.org/>

Outline

- FHIR and RDF
- Using FHIR RDF with a DL Reasoner
- Caveats, Issues, Next Steps

FHIR®©

Fast Healthcare Interoperability Resources

The screenshot shows the FHIR website homepage. The browser address bar displays www.hl7.org/FHIR/. The page header includes the FHIR logo and the text "FHIR Release 3 (STU)". A navigation menu lists: Home, Getting Started, Documentation, Resources, Profiles, Extensions, Operations, and Terminologies. Below the menu, a yellow banner states: "This is the current officially released version of FHIR, which is Release 3 (STU) with 1 technical errata. For a full list of available versions, see the [Directory of published versions](#)." The main heading is "Welcome to FHIR®". A box for "First time here?" provides links to the executive summary, developer's introduction, clinical introduction, architect's introduction, overview/roadmap & timelines, open license, and table of contents. Under "Technical Corrections:", a bullet point mentions "Apr-19 2017: Corrections to invariants & generated conformance resources, and add note about isSummary". The main content area is divided into four levels of the specification:

- Level 1 Basic framework on which the specification is built:**
 - Foundation**: Base Documentation, XML, JSON, REST API + Search, Data Types, Extensions
- Level 2 Supporting Implementation, and binding to external specifications:**
 - Implementer Support**: Downloads, Common Use Cases, Testing
 - Security & Privacy**: Security, Consent, Provenance, AuditEvent
 - Conformance**: StructureDefinition, CapabilityStatement, ImplementationGuide, Profiling
 - Terminology**: CodeSystem, ValueSet, ConceptMap, Terminology Svc
 - Linked Data**: RDF
- Level 3 Linking to real world concepts in the healthcare system:**
 - Administration**: Patient, Practitioner, Device, Organization, Location, Healthcare Service
- Level 4 Record-keeping and Data Exchange for the healthcare process:**
 - Clinical**: Allergy, Problem, CarePlan, DetectedIssue
 - Diagnostics**: Observation, Report, Specimen
 - Medications**: Order, Dispense, Administration
 - Workflow**: Task, Appointment, Schedule, Referral

A red circle highlights the "Report" link under the "Diagnostics" category. A small number "5" is visible next to the "Medications" category.

FHIR Resource Definition

<http://hl7.org/fhir/diagnosticreport.html>

www.hl7.org/fhir/diagnosticreport.html

10.2.4 Resource Content

Structure UML XML JSON Turtle R2 Diff All

Structure

Name	Flags	Card.	Type	Description & Constraints
DiagnosticReport			DomainResource	A Diagnostic report - a combination of request information, atomic results, images, interpretation, as well as formatted reports Elements defined in Annotations: id, meta, implicitRules, language, text, contained, extension, modifierExtension
identifier	X	0..*	Identifier	Business identifier for report
basedOn		0..*	Reference(CarePlan ImmunizationRecommendation MedicationRequest NutritionOrder ProcedureRequest ReferralRequest)	What was requested
status	YI X	1..1	code	required partial preliminary final + DiagnosticReportStatus (Required)
category	I	0..1	CodesableConcept	Service category Diagnostic Service Section Codes (Example)
code	X	1..1	CodesableConcept	Name/Code for this diagnostic report LOINC Diagnostic Report Codes (Preferred)
subject	Z	0..1	Reference(Patient Group Device Location)	The subject of the report - usually, but not always, the patient
context	X	0..1	Reference(Encounter EpisodeOfCare)	Health care event when test ordered
effective[x]	I	0..1		Clinically relevant time/time period for report
effectiveDateTime			dateTime	
effectivePeriod			Period	
issued	Z	0..1	Instant	DateTime this version was released
performer	X	0..*	BackboneElement	Participants in producing the report
role	Z	0..1	CodesableConcept	Type of performer Procedure Performer Role Codes (Example)
actor	Z	1..1	Reference(Practitioner Organization)	Practitioner or Organization participant
specimen		0..*	Reference(Specimen)	Specimens this report is based on
result		0..*	Reference(Observation)	Observations - simple, or complex nested groups
imagingStudy		0..*	Reference(ImagingStudy ImagingManifest)	Reference to full details of imaging associated with the diagnostic report
image	Z	0..*	BackboneElement	Key images associated with this report
comment		0..1	string	Comment about the image (e.g. explanation)
link	X	1..1	Reference(Media)	Reference to the image source
conclusion		0..1	string	Clinical Interpretation of test results
codedDiagnoses		0..*	CodesableConcept	Codes for the conclusion SNOMED CT Clinical Findings (Example)
presentedForm		0..*	Attachment	Entire report as issued

? Documentation for this format

FHIR Resource Instance (XML)

www.hl7.org/fhir/diagnosticreport.html

10.2.4 Resource Content

Structure UML XML JSON Turtle R2 Diff All

Structure

Name	Flags	Card.	Type	Description & Constraints
DiagnosticReport			DomainResource	A Diagnostic report - a combination of request information, atomic and formatted reports
identifier		0..*	Identifier	Elements defined in Annexes: id, meta, implicitRules, language, test, Reason for reference for report
basedOn		0..*	Reference(CarePlan Immunization Recommendation MedicationRequest NutritionOrder ProcedureRequest ReferralRequest)	What was requested
status		1..1	code	registered partial preliminary final + DiagnosticReportStatus (Required)
category		0..1	CodeableConcept	Service category
code		1..1	CodeableConcept	Diagnostic Service Section Codes (Example)
subject		0..1	Reference(Patient Group Device Location)	Name/Code for this diagnostic report
context		0..1	Reference(Encounter EpisodeOfCare)	LOINC Diagnostic Report Codes (Preferred)
effective[x]		0..1	dateTime	The subject of the report - usually, but not always, the patient
effectiveTime		0..1	Period	Clinically relevant time/time period for report
issued		0..1	dateTime	Health care event when test ordered
performer		0..*	BackboneElement	Clinically relevant time/time period for report
role		0..1	CodeableConcept	Date/Time this version was released
actor		1..1	Reference(Practitioner Organization)	Participants in producing the report
specimen		0..*	Reference(Specimen)	Type of performer
result		0..*	Reference(Observation)	Procedure Performer Role Codes (Example)
imagingStudy		0..*	Reference(ImagingStudy ImagingManifest)	Practitioner or Organization participant
image		0..*	BackboneElement	Specimens this report is based on
comment		0..1	string	Observations - simple, or complex nested groups
link		1..1	Reference(Media)	Reference to full details of imaging associated with the diagnostic report
conclusion		0..1	string	Key Images associated with this report
codedDiagnosis		0..*	CodeableConcept	Comment about the image (e.g. explanation)
presentedForm		0..*	Attachment	Reference to the image source

Documentation for this format

Alternate definitions: Nestor Definition (XML, JSON), XML Schema/Schematron (or) - JSON Schema, ShEx (for Turtle)

```
<?xml version="1.0" encoding="UTF-8"?><DiagnosticReport xmlns="http://hl7.org/fhir">
  <id value="f201"/>
  <text><status value="generated"/><div xmlns="http://www.w3.org/1999/xhtml"><p><b>General</b></p></div>
  <status value="final"/>
  <category>
    <!-- The request was honored by the Department of Radiology -->
    <coding>
      <system value="http://snomed.info/ct"/>
      <code value="394914008"/>
      <display value="Radiology"/>
    </coding>
    <coding>
      <system value="http://hl7.org/fhir/v2/0074"/>
      <code value="RAD"/>
    </coding>
  </category>
  <code>
    <coding>
      <system value="http://snomed.info/ct"/>
      <code value="429858000"/>
      <display value="Computed tomography (CT) of head and neck"/>
    </coding>
    <text value="CT of head-neck"/>
  </code>
  <subject>
    <reference value="Patient/f201"/>
    <display value="Acet"/>
  </subject>
  <effectiveDateTime value="2012-12-01T12:00:00+01:00"/>
  <issued value="2012-12-01T12:00:00+01:00"/>
  <performer>
    <actor>
      <reference value="Organization/f203"/>
      <display value="Blijdorp MC"/>
    </actor>
  </performer>
  <!-- The actual CT images not available - following reference used to demonstrate t -->
  <imagingStudy>
    <display value="HEAD and NECK CT DICOM imaging study"/>
  </imagingStudy>
  <conclusion value="CT brains: large tumor sphenoid/clivus."/>
  <codedDiagnosis>
    <coding>
      <system value="http://snomed.info/ct"/>
      <code value="188340000"/>
      <display value="Malignant tumor of craniopharyngeal duct"/>
    </coding>
  </codedDiagnosis>
</DiagnosticReport>
```

<http://www.hl7.org/fhir/diagnosticreport-example-f201-brainct.xml>

FHIR Resource Instance (JSON)

www.hl7.org/fhir/diagnosticreport.html

10.2.4 Resource Content

Structure UML XML JSON Turtle R2 Diff All

Structure

Name	Flags	Card.	Type	Description & Constraints
DiagnosticReport			DomainResource	A Diagnostic report - a combination of request information, atomic results, images, interpreted formatted reports Elements defined by extensions: id, meta, implicitRules, language, text, contained, extension, ResourceReference for report
identifier		0..*	Identifier	
basedOn		0..*	Reference(CarePlan Immunization ImmunizationRecommendation MedicationRequest NutritionOrder ProcedureRequest ReferralRequest)	What was requested
status		1..1	code	registered partial preliminary final + DiagnosticReportStatus (Required)
category		0..1	CodeableConcept	Service category Diagnostic Service Section Codes (Example)
code		0..1	CodeableConcept	Name/Code for this diagnostic report LOINC Diagnostic Report Codes (Preferred)
subject		0..1	Reference(Patient Group Device Location)	The subject of the report - usually, but not always, the patient
context		0..1	Reference(EpisodeOfCare EpisodeOfCare)	Health care event within test history
effective[x]		0..1	dateTime	Clinically relevant time/time period for report
effectiveDateTime			dateTime	
effectivePeriod			Period	
issued		0..1	Instant	DateTime this version was released
performer		0..*	BackboneElement	Participants in producing the report
role		0..1	CodeableConcept	Type of performer Procedure Performer Role Codes (Example)
actor		0..1	Reference(Practitioner Organization)	Practitioner or Organization participant
specimen		0..*	Reference(Specimen)	Specimens this report is based on
result		0..*	Reference(Observation)	Observations - simple, or complex nested groups
imagingStudy		0..*	Reference(ImagingStudy ImagingManifest)	Reference to full details of imaging associated with the diagnostic report
image		0..*	BackboneElement	Key Images associated with this report
comment		0..1	string	Comment about the image (e.g. explanation)
link		0..1	Reference(Media)	Reference to the image source
conclusion		0..1	string	Clinical Interpretation of test results
codedDiagnosis		0..*	CodeableConcept	Codes for the conclusion SNOMED CT Clinical Findings (Example)
presentedForm		0..*	Attachment	Online report as issued

Documentation for this format

Alternate definitions: Nestor Definition (XML, JSON), XML Schema/Schematron (for) - JSON Schema, ShEx (for Turtle)

```
{
  "resourceType": "DiagnosticReport",
  "id": "f201",
  "text": {
    "status": "generated",
    "div": "div xmlns=\"http://www.w3.org/1999/xhtml\"><p><b>Generated</b></p><p><b>status</b>: final</p><p><b>category</b>: Radiology <span>given as 'Radiology'; (http://hl7.org/fhir/v2/0074 code 'RAD')</span></p><p><b>Details</b>: {SNOMED CT code '429058000' = 'Computed tomography (head and neck)'}</span></p><p><b>subject</b>: <a>Roel</a></p><p><b>effectiveDateTime</b>: 2012-12-01T12:00:00+01:00</p><p><b>Performer</b>:</p><table><tr><td></td><td><b>Actor</b></td></tr></table><p><b>imagingStudy</b>: HEAD and NECK CT DICOM imaging sphenoid/clinivus.</p><p><b>codedDiagnosis</b>: Malignant tumor of craniopharyngeal duct, given as 'Malignant tumor of craniopharyngeal duct'</p></div></text>
  },
  "status": "final",
  "category": {
    "coding": [
      {
        "system": "http://snomed.info/sct",
        "code": "394914008",
        "display": "Radiology"
      }
    ]
  },
  "code": {
    "coding": [
      {
        "system": "http://snomed.info/sct",
        "code": "429058000",
        "display": "Computed tomography (CT) of head and neck"
      }
    ],
    "text": "CT of head-neck"
  },
  "subject": {
    "reference": "Patient/f201",
    "display": "Roel"
  },
  "effectiveDateTime": "2012-12-01T12:00:00+01:00",
  "issued": "2012-12-01T12:00:00+01:00",
  "performer": [
    {
      "actor": {
        "reference": "Organization/f203",
        "display": "Blifdorp MC"
      }
    }
  ],
  "imagingStudy": {
    "display": "HEAD and NECK CT DICOM imaging study"
  },
  "conclusion": "CT brains: large tumor sphenoid/clinivus.",
  "codedDiagnosis": [
    {
      "coding": [
        {
          "system": "http://snomed.info/sct",
          "code": "188340000",
          "display": "Malignant tumor of craniopharyngeal duct"
        }
      ]
    }
  ]
}
```

<http://www.hl7.org/fhir/diagnosticreport-example-f201-brainct.json>

(RDF)

http://www.hl7.org/fhir/diagnosticreport-example-f201-brainct.ttl

RDF Turtle Syntax

```
@prefix fhir: <http://hl7.org/fhir/> .
@prefix owl: <http://www.w3.org/2002/07/owl#> .
@prefix rdfs: <http://www.w3.org/2000/01/rdf-schema#> .
@prefix sct: <http://snomed.info/id/> .
@prefix xsd: <http://www.w3.org/2001/XMLSchema#> .
```

Prefixes

'a' == rdf:type

subject predicate object ;
predicate object ;

'[...]' == Blank Node

subject predicate object ,
object ,

```
# - resource -----
<http://hl7.org/fhir/DiagnosticReport/f201> a fhir:DiagnosticReport;
  fhir:nodeRole fhir:treeRoot;
  fhir:Resource.id [ fhir:value "f201" ];
  fhir:DomainResource.text [
    fhir:Narrative.status [ fhir:value "generated" ];
    fhir:DiagnosticReport.status [ fhir:value "final" ];
    fhir:DiagnosticReport.category [
      fhir:CodeableConcept.coding [
        fhir:index 0;
        a sct:394914008;
        fhir:Coding.system [ fhir:value "http://snomed.info/sct" ];
        fhir:Coding.code [ fhir:value "394914008" ];
        fhir:Coding.display [ fhir:value "Radiology" ]
      ],
      fhir:index 1;
      fhir:Coding.system [ fhir:value "http://hl7.org/fhir/v2/0074" ];
      fhir:Coding.code [ fhir:value "RAD" ]
    ]
  ];
  fhir:DiagnosticReport.code [
    fhir:CodeableConcept.coding [
      fhir:index 0;
      a sct:429858000;
      fhir:Coding.system [ fhir:value "http://snomed.info/sct" ];
      fhir:Coding.code [ fhir:value "429858000" ];
      fhir:Coding.display [ fhir:value "Computed tomography (CT) of head and neck" ]
    ];
    fhir:CodeableConcept.text [ fhir:value "CT of head-neck" ]
  ];
];
```

Syntax “maturity”

2.6.1 XML Representation of Resources

Implementable Technology Specifications Work Group	Maturity Level: 5	Ballot Status: Trial Use
--	-------------------	--------------------------

<http://www.hl7.org/fhir/xml.html>

Implementable Technology Specifications Work Group	Maturity Level: 5	Ballot Status: Trial Use
--	-------------------	--------------------------

<http://www.hl7.org/fhir/json.html>

FHIR Infrastructure Work Group	Maturity Level: 2	Ballot Status: Trial Use
--	-------------------	--------------------------

<http://www.hl7.org/fhir/rdf.html>

0. the resource or profile (artifact) has been published on the current build. This level is synonymous with *Draft*.
1. PLUS the artifact produces no warnings during the build process and the responsible WG has indicated that they consider the artifact substantially complete and ready for implementation
2. PLUS the artifact has been tested and successfully exchanged between at least three independently developed systems leveraging at least 80% of the core data elements using semi-realistic data and scenarios based on at least one of the declared scopes of the resource (e.g. at a connectathon). These interoperability results must have been reported to and accepted by the FMG
3. PLUS the artifact has been verified by the work group as meeting the [Trial Use Quality Guidelines](#) and has been subject to a round of formal balloting; has at least 10 implementer comments recorded in the tracker drawn from at least 3 organizations resulting in at least one substantive change
4. PLUS the artifact has been tested across its scope (see below), published in a formal publication (e.g. a FHIR Release), and implemented in multiple prototype projects. As well, the responsible work group agrees the resource is sufficiently stable to require implementer consultation for subsequent non-backward compatible changes.
5. PLUS the artifact has been published in two formal publication release cycles at FMM1+ (i.e. *Trial Use* level) and has been implemented in at least 5 independent production systems in more than one country
6. "Normative": the artifact is now considered stable

FHIR RDF Rendering

Requirement: RDF Rendering must be fully “round-trippable”:



Which is why:

```
fhir:Person.active [ fhir:value "true"^^xsd:boolean].
```

instead of:

```
fhir:Person.active "true"^^xsd:boolean.
```

FHIR RDF Rendering Preserving Extensions

Boolean, like all FHIR elements, is extensible. Processing for:

```
fhir:Person.active [ fhir:value "true"^^xsd:boolean].
```

and:

```
fhir:Person.active [  
  fhir:Element.extension [  
    fhir:index 0;  
    fhir:Extension.url [ fhir:value "http://example.org/fhir/boolean/Certainty" ];  
    fhir:Extension.valueDecimal [ fhir:value "0.75"^^xsd:decimal ]  
  ];  
  fhir:value "true"^^xsd:boolean] .
```

should be the same.

“Round Tripability”

```
{  
  "resourceType": "DiagnosticReport",  
  "id": "f201",  
  "text": {  
    "status": "generated"  
  }  
}
```

```
"category": [  
  "coding": [  
    {  
      "system": "http://snomed.info/sct",  
      "code": "394314008",  
      "display": "Radiology"  
    },  
    {  
      "system": "http://hl7.org/fhir/v2/0074",  
      "code": "RAD"  
    }  
  ],  
],
```

JSON

```
<http://hl7.org/fhir/DiagnosticReport/f201> a fhir:DiagnosticReport;  
fhir:nodeRole fhir:treeRoot;  
fhir:Resource.id [ fhir:value "f201"];  
fhir:DomainResource.text [  
  fhir:Narrative.status [ fhir:value "generated" ];  
  fhir:Narrative.div "<div xmlns='\"http://www.w3.org/1999/xhtml\"'>(deleted)</div>"  
];  
fhir:DiagnosticReport.status [ fhir:value "final"];  
fhir:DiagnosticReport.category [  
  fhir:CodeableConcept.coding [  
    fhir:index 0;  
    a sct:394314008;  
    fhir:Coding.system [ fhir:value "http://snomed.info/sct" ];  
    fhir:Coding.code [ fhir:value "394314008" ];  
    fhir:Coding.display [ fhir:value "Radiology" ]  
  ], [  
    fhir:index 1;  
    fhir:Coding.system [ fhir:value "http://hl7.org/fhir/v2/0074" ];  
    fhir:Coding.code [ fhir:value "RAD" ]  
  ]  
];  
fhir:DiagnosticReport.code [  
  fhir:CodeableConcept.coding [  
    fhir:index 0;  
    a sct:429858000;  
    fhir:Coding.system [ fhir:value "http://snomed.info/sct" ];  
    fhir:Coding.code [ fhir:value "429858000" ];  
    fhir:Coding.display [ fhir:value "Computed tomography (CT) of head and neck" ]  
  ];  
  fhir:CodeableConcept.text [ fhir:value "CT of head-neck" ]  
];  
fhir:DiagnosticReport.subject [  
  fhir:link <http://hl7.org/fhir/Patient/f201>;  
  fhir:Reference.reference [ fhir:value "Patient/f201" ];  
  fhir:Reference.display [ fhir:value "Roel" ]  
];  
fhir:DiagnosticReport.effectiveDateTime [ fhir:value "2012-12-01T12:00:00+01:00"^^xsd
```

Identify root documents

Preserve order in lists

RDF
4

RDF Rendering Extensions

```
"category": {
  "coding": [
    {
      "system": "http://snomed.info/sct",
      "code": "394914008",
      "display": "Radiology"
    },
    {
      "system": "http://hl7.org/fhir/v2/0074",
      "code": "RAD"
    }
  ]
},
```

JSON

```
"subject": {
  "reference": "Patient/f201",
  "display": "Roel"
},
```

JSON

```
fhir:DiagnosticReport.category [
  fhir:CodeableConcept.coding [
    fhir:index 0;
    a sct:394914008;
    fhir:Coding.system [ fhir:value "http://snomed.info/sct" ];
    fhir:Coding.code [ fhir:value "394914008" ];
    fhir:Coding.display [ fhir:value "Radiology" ]
  ], [
    fhir:index 1;
    fhir:Coding.system [ fhir:value "http://hl7.org/fhir/v2/0074" ];
    fhir:Coding.code [ fhir:value "RAD" ]
  ]
];
```

Concept URI

Reference URI

Reference Type

**Ontology and
import declaration**

```
fhir:DiagnosticReport.subject [
  fhir:link <http://hl7.org/fhir/Patient/f201>;
  fhir:Reference.reference [ fhir:value "Patient/f201" ];
  fhir:Reference.display [ fhir:value "Roel" ]
];
```

```
<http://hl7.org/fhir/Patient/f201> a fhir:Patient .
```

```
# - ontology header -----
<http://hl7.org/fhir/DiagnosticReport/f201.ttl> a owl:Ontology;
owl:imports fhir:fhir.ttl;
owl:versionIRI <http://build.fhir.org/DiagnosticReport/f201.ttl> .
```

<http://snomed.info/id/394914008>

RDF

Concept URI's

For this (or any linked data to work) both the data and the ontology have to use the same URI's

Progress is being made:

- SNOMED International has a standard:
 - [http://snomed.info/id/\(concept code\)](http://snomed.info/id/(concept code))
 - Spec: <https://confluence.ihtsdotools.org/display/DOCURI/URI+Standard>
- WHO has a standard
 - [http://id.who.int/icd/release/10/\(code\)](http://id.who.int/icd/release/10/(code))
 - Spec: <https://icdaccessmanagement.who.int/docs/APIdoc-md.html>

Using FHIR RDF With a DL Reasoner

```

class MyPrivateClass {
private:
    int myPrivateVar = 12345;
    void myPrivateMethod() {
        // Private method implementation
    }
};

// Usage of MyPrivateClass
int main() {
    MyPrivateClass obj;
    obj.myPublicMethod();
    obj.myProtectedMethod();
    obj.myPrivateMethod();
    return 0;
}

```

FHIR DiagnosticReport Instance

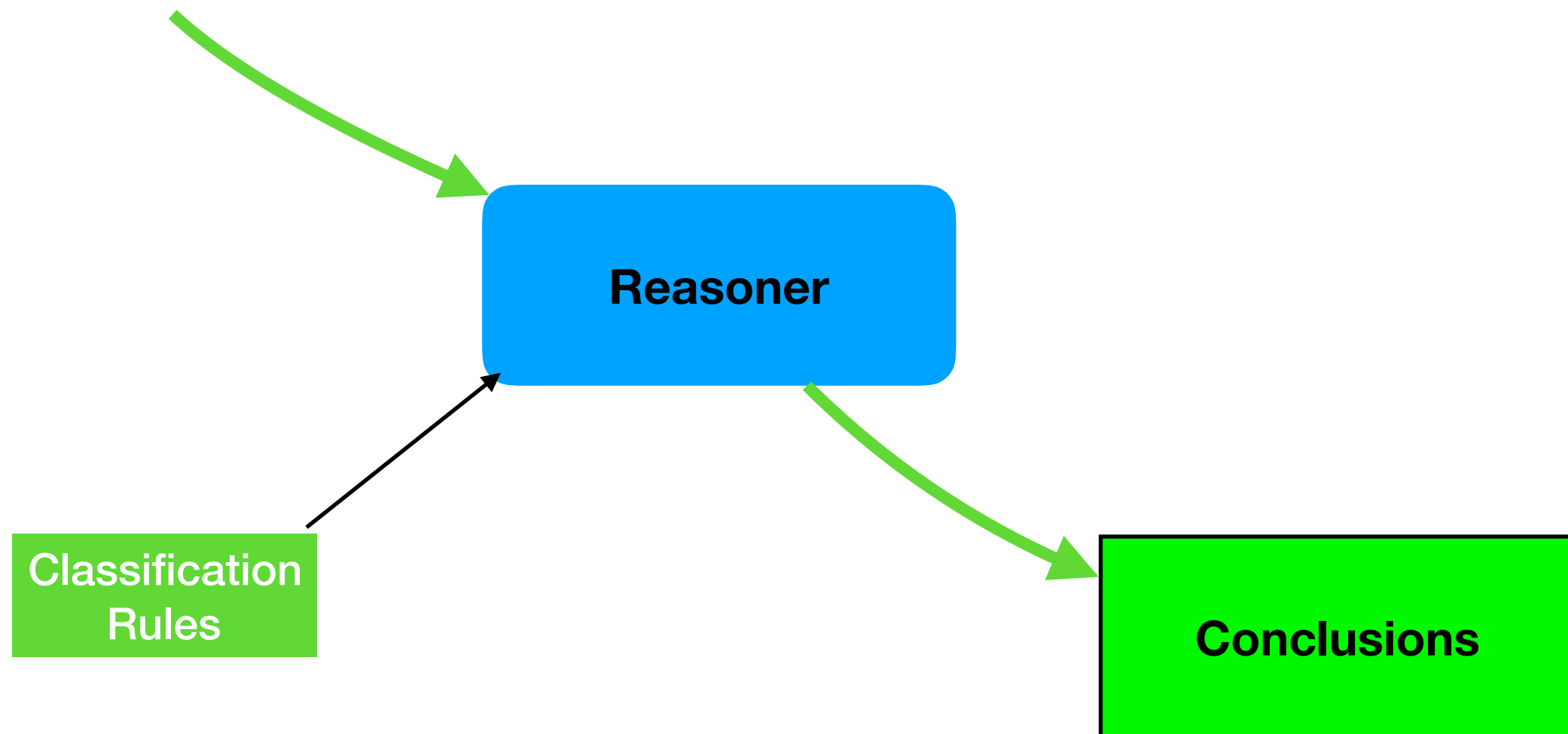
Reasoner

**Class `CancerDiagnosis` == any DiagnosticReport
w/ a dx of a type of malignant neoplasm**

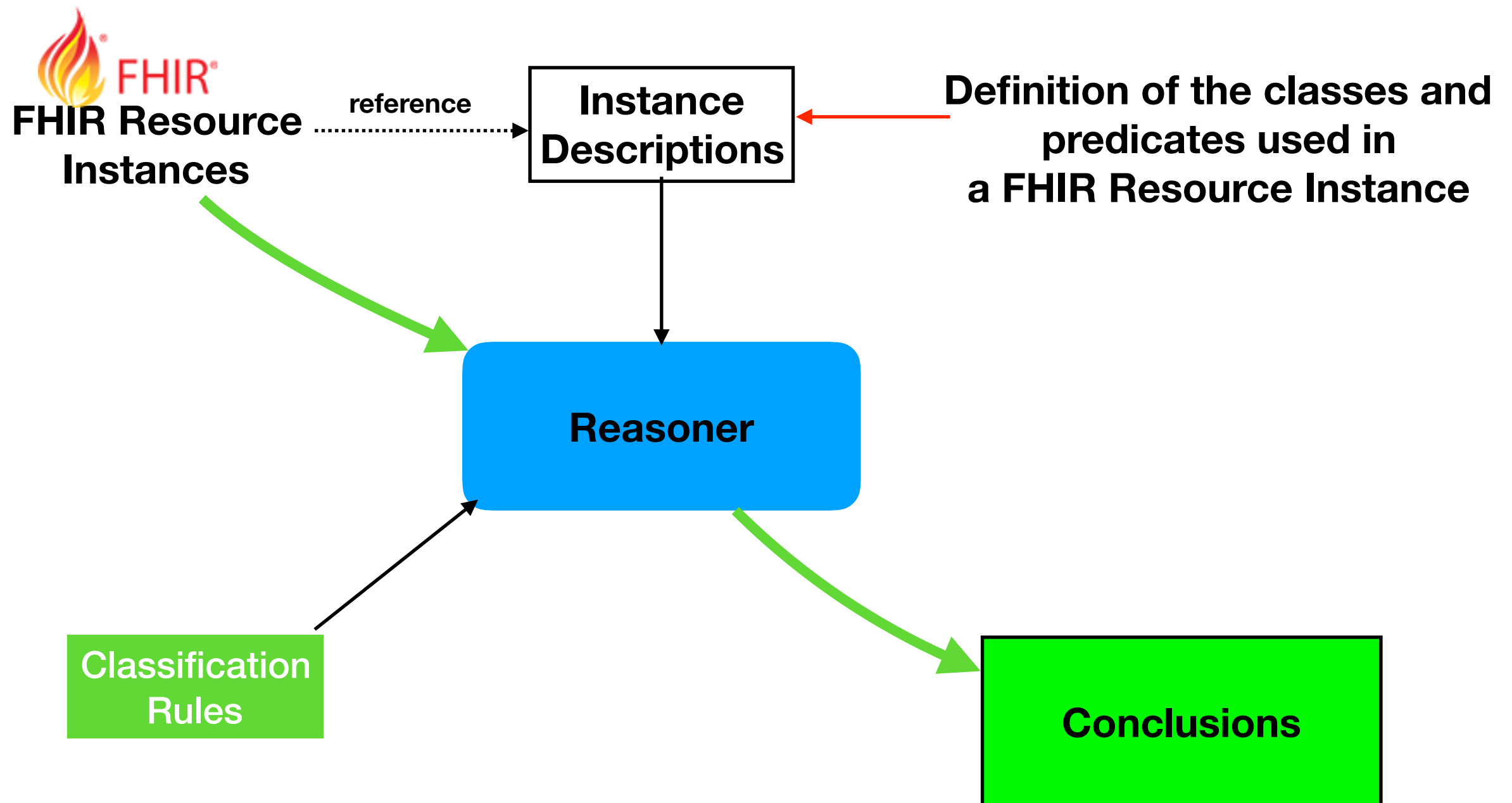
**Instance is (or is not)
an instance of Class
'CancerDiagnosis'**

Using FHIR RDF With a DL Reasoner

 **FHIR**
FHIR Resource
Instances



Using FHIR RDF With a DL Reasoner



Instance Descriptions

The FHIR Metadata Vocabulary

Example FHIR resource (data record)

```
<http://hl7.org/fhir/DiagnosticReport/f201> a fhir:DiagnosticReport;
  fhir:DiagnosticReport.subject [
    fhir:link <http://hl7.org/fhir/Patient/f201>;
    fhir:Reference.reference [ fhir:value "Patient/f201" ];
    fhir:Reference.display [ fhir:value "Roel" ]
  ];
  fhir:DiagnosticReport.code [
    fhir:CodeableConcept.coding [
      fhir:index 0;
      a sct:429858000;
      fhir:Coding.system [ fhir:value "http://snomed.info/sct" ];
      fhir:Coding.code [ fhir:value "429858000" ];
      fhir:Coding.display [ fhir:value "Computed tomography (CT) of head and neck" ]
    ];
    fhir:CodeableConcept.text [ fhir:value "CT of head-neck" ]
  ];
  fhir:DiagnosticReport.codedDiagnosis [
    fhir:index 0;
    fhir:CodeableConcept.coding [
      fhir:index 0;
      a sct:188340000;
      fhir:Coding.system [ fhir:value "http://snomed.info/sct" ];
      fhir:Coding.code [ fhir:value "188340000" ];
      fhir:Coding.display [ fhir:value "Malignant tumor of craniopharyngeal duct" ]
    ]
  ]
```

Instance Descriptions

The FHIR Metadata Vocabulary

```
<http://hl7.org/fhir/DiagnosticReport/f201> a
  fhir:DiagnosticReport.subject [
    fhir:link <http://hl7.org/fhir/Patient/f
    fhir:Reference.reference [ fhir:value "P
    fhir:Reference.display [ fhir:value "Roe
  ];
  fhir:DiagnosticReport.code [
    fhir:CodeableConcept.coding [
      fhir:index 0;
      a sct:429858000;
      fhir:Coding.system [ fhir:value "http://
      fhir:Coding.code [ fhir:value "42985800
      fhir:Coding.display [ fhir:value "Compu
    ];
    fhir:CodeableConcept.text [ fhir:value "C
  ];
  fhir:DiagnosticReport.codedDiagnosis [
    fhir:index 0;
    fhir:CodeableConcept.coding [
      fhir:index 0;
      a sct:188340000;
      fhir:Coding.system [ fhir:value "http://snomed.info/sct"
      fhir:Coding.code [ fhir:value "188340000" ];
      fhir:Coding.display [ fhir:value "Malignant tumor of cran
    ]
  ]
```

```
fhir:DiagnosticReport
  a owl:Class ;
  rdfs:comment "The findings and interpretation of diagnostic tests performed on patients, groups
of patients, devices, and locations, and/or specimens derived from these. The report includes clinical context
such as requesting and provider information, and some mix of atomic results, images, textual and coded inte
rpretations, and formatted representation of diagnostic reports." ;
  rdfs:label "DiagnosticReport" ;
  rdfs:subClassOf fhir:DomainResource, w5:clinical.diagnostics ;
```

<http://hl7.org/fhir/fhir.ttl>

```
fhir:DiagnosticReport.code
  a owl:ObjectProperty ;
  rdfs:comment "A code or name that describes this diagnostic report." ;
  rdfs:domain fhir:DiagnosticReport ;
  rdfs:label "DiagnosticReport.code" ;
  rdfs:range fhir:CodeableConcept ;
  rdfs:subPropertyOf w5:what ;
  dc:title "Name/Code for this diagnostic report" .
```

```
fhir:DiagnosticReport.codedDiagnosis
  a owl:ObjectProperty ;
  rdfs:comment "Codes for the conclusion." ;
  rdfs:domain fhir:DiagnosticReport ;
  rdfs:label "DiagnosticReport.codedDiagnosis" ;
  rdfs:range fhir:CodeableConcept ;
  dc:title "Codes for the conclusion" .
```

```
fhir:value a owl:DatatypeProperty ;
  rdfs:label "fhir:value" ;
  dc:title "Terminal data value" .
```

FMV Definition of DiagnosticReport

cancerreport (http://example.org/swat4/cancerreport)

Active Ontology x Entities x Individuals by class x DL Query x

Class hierarchy: DiagnosticReport

Class hierarchy (inferred)

Class hierarchy: DiagnosticReport

DiagnosticReport — http://hl7.org/fhir/DiagnosticReport

Description: DiagnosticReport

Equivalent to

SubClass Of

- (DiagnosticReport.effectiveDateTime **only** dateTime) or (DiagnosticReport.effectivePeriod **only** Period)
- DiagnosticReport.basedOn **only** Reference
- DiagnosticReport.category **only** CodeableConcept
- DiagnosticReport.code **some** CodeableConcept
- DiagnosticReport.codedDiagnosis **only** CodeableConcept
- DiagnosticReport.conclusion **only** string
- DiagnosticReport.context **only** Reference
- DiagnosticReport.identifier **only** Identifier
- DiagnosticReport.image **only** DiagnosticReportImageComponent
- DiagnosticReport.imagingStudy **only** Reference
- DiagnosticReport.issued **only** instant
- DiagnosticReport.performer **only** Reference
- DiagnosticReport.presentedForm **only** Attachment
- DiagnosticReport.result **only** Reference
- DiagnosticReport.resultsInterpreter **only** Reference
- DiagnosticReport.specimen **only** Reference
- DiagnosticReport.status **some** code
- DiagnosticReport.subject **only** Reference
- diagnostics
- DomainResource

General class axioms

SubClass Of (Anonymous Ancestor)

- DomainResource.modifierExtension **only** Extension
- DomainResource.contained **only** Resource
- DomainResource.extension **only** Extension
- DomainResource.text **only** Narrative
- Resource.meta **only** Meta
- Resource.language **only** code
- Resource.implicitRules **only** uri
- nodeRole **only** treeRoot
- Resource.id **only** id

Superclass hierarchy (inferred)

Superclass hierarchy (inferred): DiagnosticReport

The Ontology Header

```
# - resource -----  
<http://hl7.org/fhir/DiagnosticReport/f201> a fhir:DiagnosticReport;  
  fhir:nodeRole fhir:treeRoot;  
  fhir:Resource.id [ fhir:value "f201"];  
  fhir:DomainResource.text [
```

```
# - ontology header -----  
#<http://hl7.org/fhir/DiagnosticReport/f201.ttl> a owl:Ontology;  
# owl:imports fhir:fhir.ttl.
```

**If the resource itself doesn't include
the FHIR Metadata Vocabulary...
... the OWL tooling assumes that
everything is an annotation**

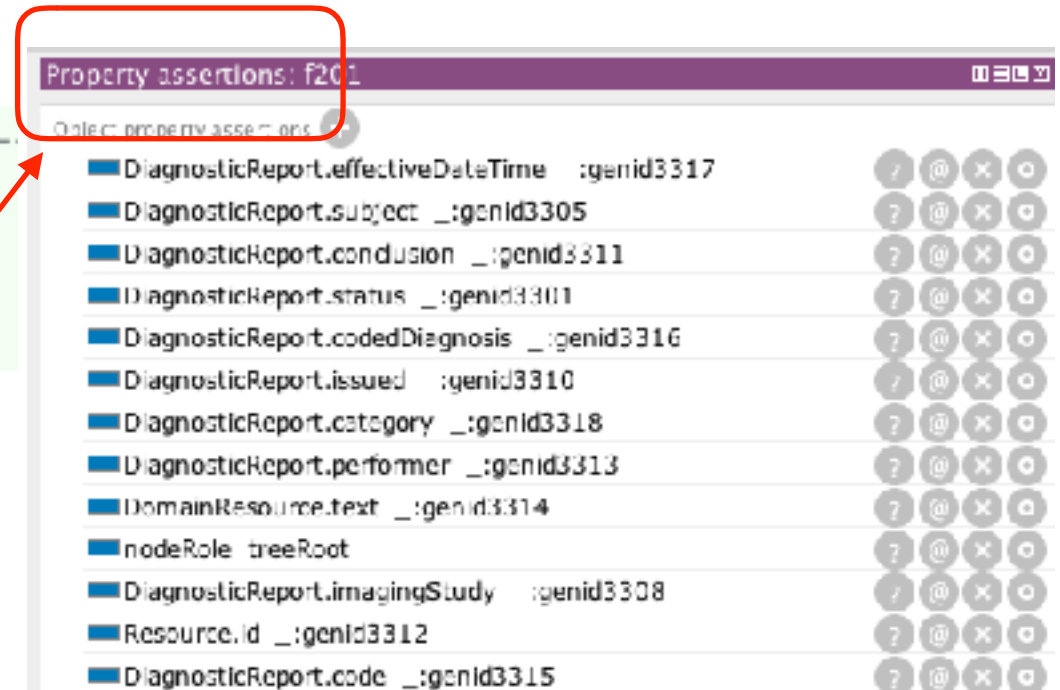
The screenshot shows an OWL editor interface. At the top, a tab labeled 'Annotations: f201' is highlighted with a red box. Below the tab, the editor displays a list of annotations for the resource 'f201'. The first annotation is 'DiagnosticReportCategory', which is a 'CodeableConcept.coding' with a 'Coding.code' of '394914008' and a 'Coding.system' of 'http://snomed.info/sct'. The second annotation is 'Coding.system', which is a 'CodeableConcept.coding' with a 'Coding.code' of 'RAD' and a 'Coding.system' of 'http://hl7.org/fhir/20074'. The third annotation is 'index', which is a 'Typed integer' with a value of '0'.

Why the Ontology Header

```
# - resource -----  
<http://hl7.org/fhir/DiagnosticReport/f201> a fhir:DiagnosticReport;  
  fhir:nodeRole fhir:treeRoot;  
  fhir:Resource.id [ fhir:value "f201"];  
  fhir:DomainResource.text [
```

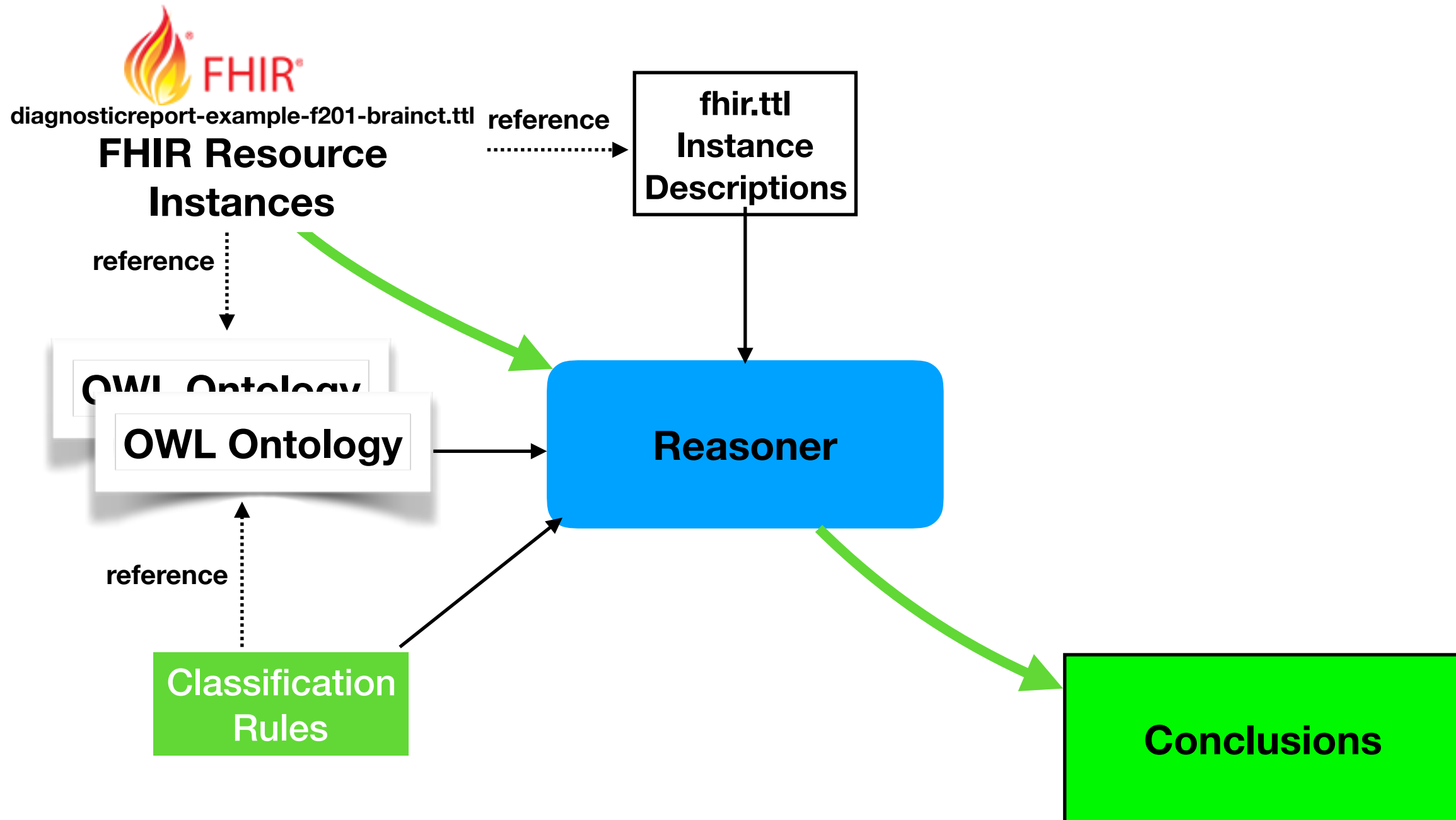
```
# - ontology header -----  
<http://hl7.org/fhir/DiagnosticReport/f201.ttl> a owl:Ontology;  
  owl:imports fhir:fhir.ttl.
```

**With the import statement, the data
is interpreted correctly**



Property assertions: f201	
Object	Property assertions
DiagnosticReport.effectiveDateTime	:genid3317
DiagnosticReport.subject	_:genid3305
DiagnosticReport.conclusion	_:genid3311
DiagnosticReport.status	_:genid3301
DiagnosticReport.codedDiagnosis	_:genid3316
DiagnosticReport.issued	_:genid3310
DiagnosticReport.category	_:genid3318
DiagnosticReport.performer	_:genid3313
DomainResource.text	_:genid3314
nodeRole	treeRoot
DiagnosticReport.imagingStudy	_:genid3308
Resource.id	_:genid3312
DiagnosticReport.code	_:genid3315

Using FHIR RDF With a DL Reasoner



FHIR Resource Instance

Concept References

Class hierarchy (Inferred): "Malignant tumor of craniopharyngeal duct (disorder)"

Class hierarchy (Inferred): "Malignant tumor of craniopharyngeal duct (disorder)"

Equivalent To:

- Neoplasm of craniopharyngeal duct (disorder) and Malignant tumor of pituitary gland (disorder) and (Role group (attribute) some (Associated morphology (attribute) some Malignant neoplasm of primary, secondary, or uncertain origin (morphologic abnormality)) and (Finding site (attribute) some Structure of craniopharyngeal duct (body structure)))

SubClass Of:

- Malignant tumor of pituitary gland (disorder)
- Neoplasm of craniopharyngeal duct (disorder)

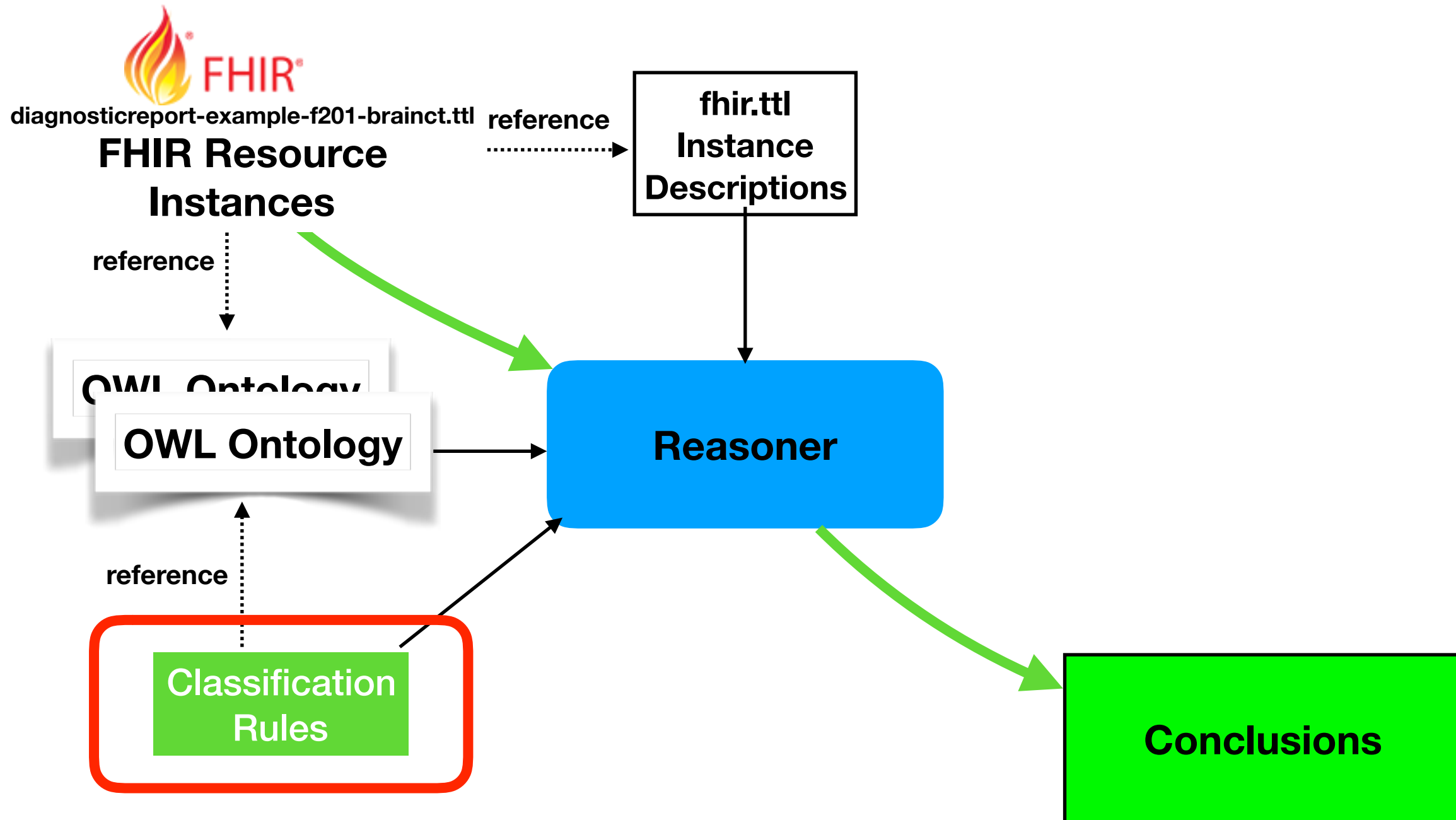
Generalization:

- Clinical finding (finding) and (Role group (attribute) some (Finding site (attribute) some Anatomical or acquired body structure (body structure)))
- Finding by site (finding) and (Role group (attribute) some (Finding site (attribute) some Body structure (body structure)))

http://snomed.info/id/188340000

```
<http://hl7.org/fhir/CodeableConcept>
  fhir:Diagnosis
    fhir:index 0;
    fhir:CodeableConcept.coding [
      fhir:index 0;
      a sct:188340000;
      fhir:Coding.system [ fhir:value "http://snomed.info/sct" ];
      fhir:Coding.code [ fhir:value "188340000" ];
      fhir:Coding.display [ fhir:value "Malignant tumor of craniopharyngeal duct" ]
    ]
  ]
```

Using FHIR RDF With a DL Reasoner



Sample Classification Rule

```
Ontology(<http://example.org/swat4ls/cancerreport>  
Import(<http://snomed.info/sct/9000000000000207008>)  
Import(<http://hl7.org/fhir/fhir.ttl>  
Import(<http://hl7.org/fhir/DiagnosticReport/f201.ttl>)
```

SNOMED CT
FHIR.TTL
Sample Data

```
Declaration(ObjectProperty(fhir:DiagnosticReport.codedDiagnosis.coding))  
SubObjectPropertyOf(  
  ObjectPropertyChain(fhir:DiagnosticReport.codedDiagnosis  
fhir:CodeableConcept.coding) fhir:DiagnosticReport.codedDiagnosis.coding)
```

```
Declaration(Class(:ReportWithCancerDiagnosis))  
EquivalentClasses(:ReportWithCancerDiagnosis  
ObjectSomeValuesFrom(fhir:DiagnosticReport.codedDiagnosis.coding sct:363346000))  
)
```



OWL Functional Syntax

Classification Rules

Concept Reference

Declaration(Class(:ReportWithCancerDiagnosis))

EquivalentClasses(:ReportWithCancerDiagnosis

ObjectSomeValuesFrom(fhir:DiagnosticReport.codedDiagnosis.coding sct:363346000))

)

900000000000207008x (http://snomed.info/sct/900000000000207008x)

Active Ontology x Entities x Individuals by class x DL Query x

Class hierarchy Class hierarchy (inferred)

Class hierarchy: 'Malignant neoplastic disease (disorder)'

Assert

<http://snomed.info/id/363346000>

'Neoplastic disease (disorder)' and ('Role group (attribute)' some ('Associated morphology (attribute)' some 'Malignant neoplasm of primary, secondary, or uncertain origin (morphologic abnormality)'))

SubClass Of +

General class axioms +

SubClass Of (Anonymous Ancestor)

'Disease (disorder)' and ('Role group (attribute)' some ('Associated morphology (attribute)' some 'Neoplasm and/or hamartoma (morphologic abnormality)'))

'Neoplasm and/or hamartoma (disorder)' and ('Role group (attribute)' some ('Associated morphology (attribute)' some 'Neoplasm (morphologic abnormality)'))

Instances +

Usage: Malignant neoplastic disease (disorder)

Show: ☒ this ☒ disjoint ☒ named sub/superclasses

Found 15 uses of 'Malignant neoplastic disease (disorder)'

'Malignant neoplasm of endocrine gland (disorder)'

'Malignant neoplasm of endocrine gland (disorder)' EquivalentTo 'Malignant neoplastic disease (disorder)' (('Associated morphology (attribute)' some 'Malignant neoplasm of primary, secondary, or uncertain origin (morphologic abnormality)'))

'Malignant neoplasm of nervous system (disorder)'

Superclass hierarchy (inferred) Superclass hierarchy

Superclass hierarchy: 'Malignant neoplastic disease (disorder)'

Asserted

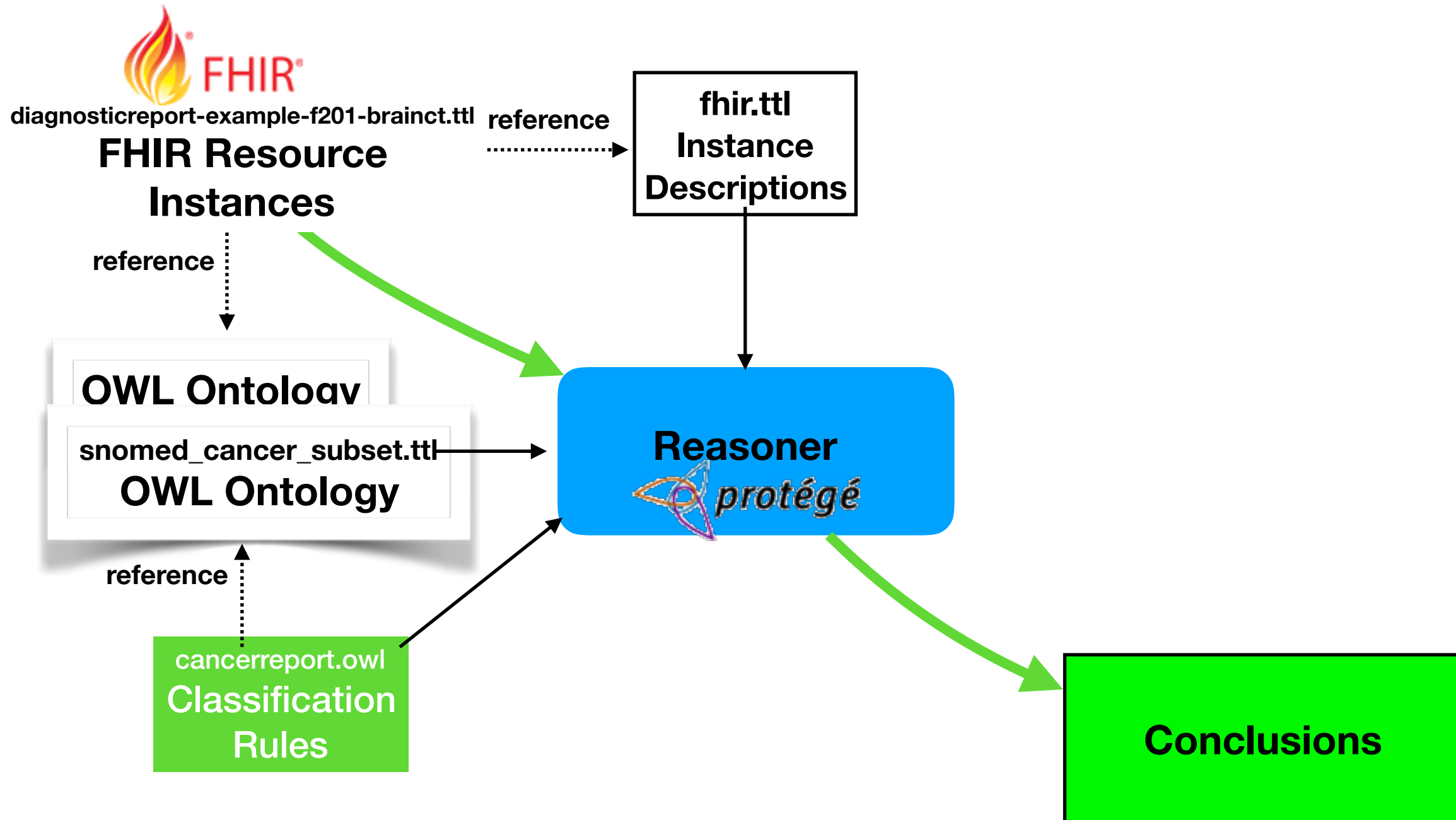
'Malignant neoplastic disease (disorder)'

'Neoplastic disease (disorder)'

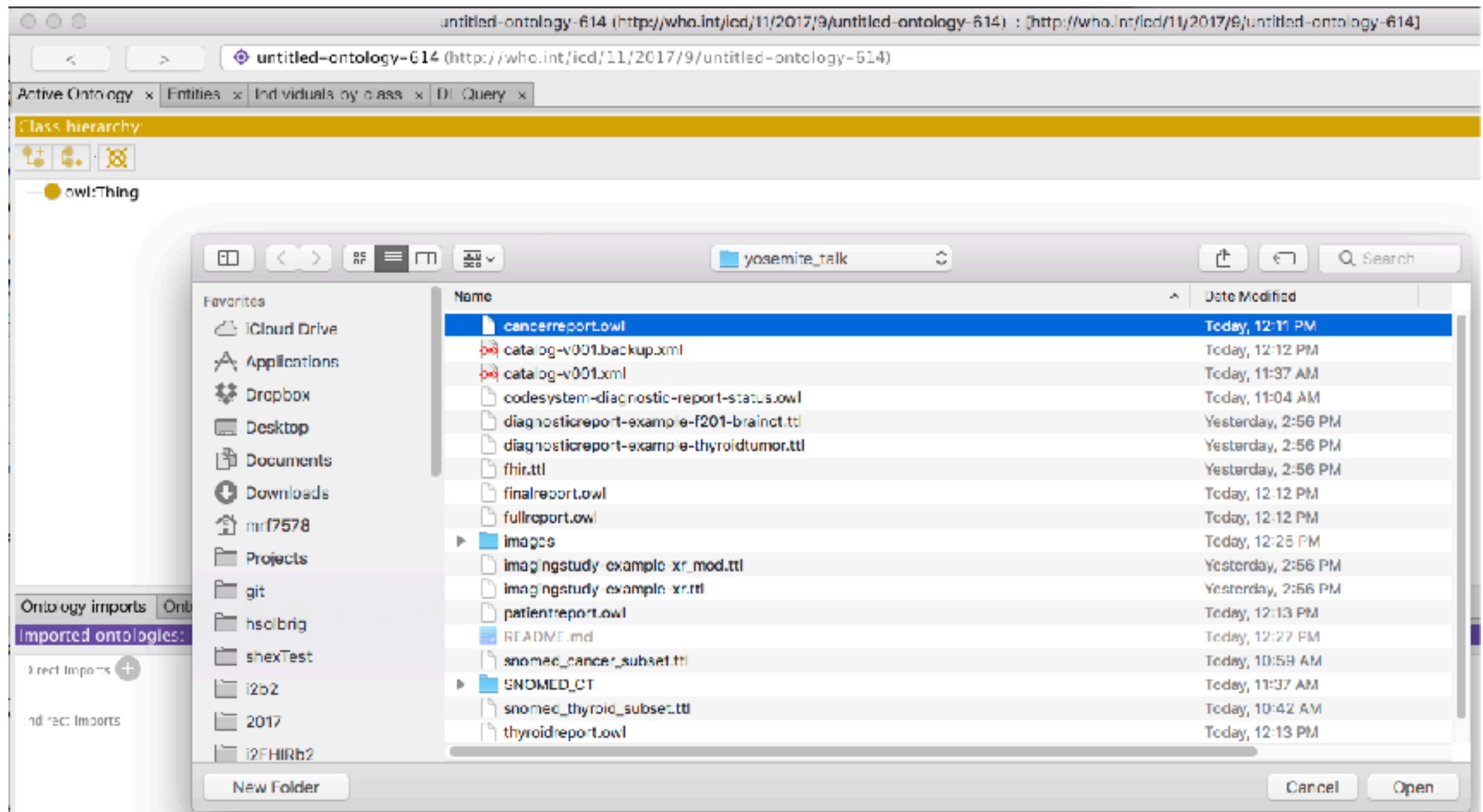
'Neoplasm and/or hamartoma (disorder)'

'Disease (disorder)'

Using FHIR RDF With a DL Reasoner




Load the Classification Rules

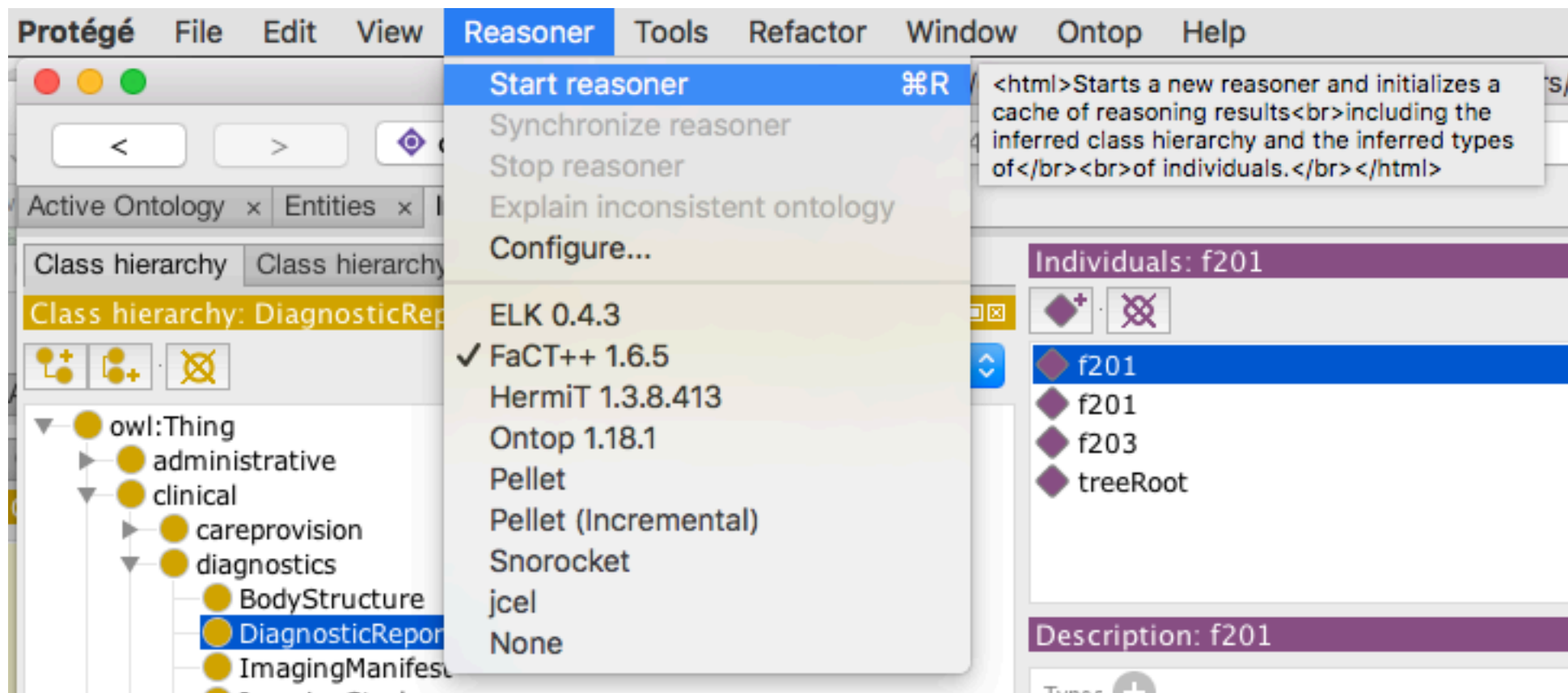


https://github.com/BD2KOnFHIR/BLENDINGFHIRandRDF/yosemite_talk/cancerreport.owl

Verify the Imports

Ontology imports	Ontology Prefixes	General class axioms
Imported ontologies:		
Direct Imports 		
<div><div><http://hl7.org/fhir/DiagnosticReport/f201.ttl></div><div>f201.ttl</div><div>Ontology IRI: <http://hl7.org/fhir/DiagnosticReport/f201.ttl></div><div>Location: /Users/mrf7578/Development/git/BD2KOnFHIR/BLENDINGFHIRandRDF/yosemite_talk/diagnosticreport-example-f201-brainct.ttl</div></div>		
<div><div><http://snomed.info/sct/900000000000207008cancer_subset></div><div>900000000000207008cancer_subset</div><div>Ontology IRI: <http://snomed.info/sct/900000000000207008cancer_subset></div><div>Location: /Users/mrf7578/Development/git/BD2KOnFHIR/BLENDINGFHIRandRDF/yosemite_talk/snomed_cancer_subset.ttl</div></div>		
<div><div><http://hl7.org/fhir/fhir.ttl></div><div>fhir.ttl</div><div>Ontology IRI: <http://hl7.org/fhir/fhir.ttl></div><div>Location: /Users/mrf7578/Development/git/BD2KOnFHIR/BLENDINGFHIRandRDF/yosemite_talk/fhir.ttl</div></div>		
Indirect Imports		
<div><div><http://hl7.org/fhir/fhir.ttl></div><div>fhir.ttl</div><div>Ontology IRI: <http://hl7.org/fhir/fhir.ttl></div><div>Location: /Users/mrf7578/Development/git/BD2KOnFHIR/BLENDINGFHIRandRDF/yosemite_talk/fhir.ttl</div></div>		
<div><div><http://hl7.org/fhir/w5#></div><div>w5</div><div>Ontology IRI: <http://hl7.org/fhir/w5#></div><div>Location: /Users/mrf7578/Development/git/BD2KOnFHIR/BLENDINGFHIRandRDF/yosemite_talk/w5.ttl</div></div>		

Run the Reasoner



Result

The screenshot displays a web-based ontology viewer interface. The browser address bar shows the URL: `http://example.org/swat4ls/cancerreport/`. The interface includes several tabs: "Active Ontology", "Entities", "Individuals by class", and "DL Query". The "Individuals by class" tab is active, showing a list of individuals for the class "DiagnosticReport". The list includes "f201", "f202", "f203", and "treeRoot". The "f201" individual is selected, and its description is shown below the list. The description includes the types "DiagnosticReport" and "ReportWithCancerDiagnosis", with "ReportWithCancerDiagnosis" highlighted by a red circle. To the left of the main content, there is a class hierarchy tree. The "diagnostics" class is expanded, showing its subclasses: "BodyStructure", "DiagnosticReport", "ImagingManifest", "ImagingStudy", "Observation", "ResearchStudy", "ResearchSubject", "Sequence", "Specimen", and "SpecimenDefinition". The "DiagnosticReport" class is highlighted. To the right of the main content, there is a section for "Property assertions: f201". This section lists various object property assertions for the "f201" individual, including "DiagnosticReport.status", "DiagnosticReport.conclusion", "DiagnosticReport.performer", "DiagnosticReport.subject", "DiagnosticReport.codedDiagnosis", "DiagnosticReport.issued", "DiagnosticReport.category", "Resource.id", "DomainResource.text", "DiagnosticReport.effectiveDateTime", "DiagnosticReport.code", "nodeRole", "treeRoot", and "DiagnosticReport.imagingStudy".

Active Ontology x Entities x Individuals by class x DL Query x

Class hierarchy Class hierarchy (inferred)

Class hierarchy: DiagnosticReport

Asserted

Individuals: f201

f201
f202
f203
treeRoot

Description: f201

Types

- DiagnosticReport
- ReportWithCancerDiagnosis

Same Individual As +

Different Individuals +

Property assertions: f201

Object property assertions +

- DiagnosticReport.status _:genid13306
- DiagnosticReport.conclusion _:genid13332
- DiagnosticReport.performer _:genid13325
- DiagnosticReport.subject _:genid13328
- DiagnosticReport.codedDiagnosis _:genid13316
- DiagnosticReport.issued _:genid13331
- DiagnosticReport.category _:genid13336
- Resource.id _:genid13333
- DomainResource.text _:genid13334
- DiagnosticReport.effectiveDateTime _:genid13335
- DiagnosticReport.code _:genid13323
- nodeRole treeRoot
- DiagnosticReport.imagingStudy _:genid13326
- nodeRole treeRoot

Restrict to Patients

				LOINC Diagnostic Report Codes (Preferred)
subject	Σ	0..1	Reference(Patient Group Device Location)	The subject of the report - usually, but not always, the patient

```
Declaration(ObjectProperty(fhir:DiagnosticReport.subject.link))
SubObjectPropertyOf(
  ObjectPropertyChain(fhir:DiagnosticReport.subject fhir:link)
fhir:DiagnosticReport.subject.link)

Declaration(Class(:PatientReport))
EquivalentClasses(:PatientReport
  ObjectSomeValuesFrom(fhir:DiagnosticReport.subject.link fhir:Patient))
)
```

https://github.com/BD2KOnFHIR/BLENDINGFHIRandRDF/yosemite_talk/patientreport.owl

Finalized Reports Only

status	?! Σ 1..1	code	registered partial preliminary final + DiagnosticReportStatus (Required)
--------	-----------	------	---

Code	Display	Definition
registered	Registered	The existence of the report is registered, but there is nothing yet available.
partial	Partial	This is a partial (e.g. initial, interim or preliminary) report: data in the report may be incomplete or unverified.
preliminary	Preliminary	Verified early results are available, but not all results are final.
final	Final	The report is complete and verified by an authorized person.
amended	Amended	Subsequent to being final, the report has been modified. This includes any change in the results, diagnosis, narrative text, report that has been issued.
corrected	Corrected	Subsequent to being final, the report has been modified to correct an error in the report or referenced results.
appended	Appended	Subsequent to being final, the report has been modified by adding new content. The existing content is unchanged.
cancelled	Cancelled	The report is unavailable because the measurement was not started or not completed (also sometimes called "aborted").
entered-in-error	Entered in Error	The report has been withdrawn following a previous final release. This electronic record should never have existed, though world decisions were based on it. (If real-world activity has occurred, the status should be "cancelled" rather than "entered-in-error".)
unknown	Unknown	The authoring system does not know which of the status values currently applies for this request. Note: This concept is not one of the listed statuses is presumed to apply, it's just not known which one.

```
Declaration(Class(:FinalizedReport))
EquivalentClasses(:FinalizedReport ObjectSomeValuesFrom
(fhir:DiagnosticReport.status DataSomeValuesFrom
(fhir:value DataOneOf("amended" "appended" "corrected" "final"))))
```

https://github.com/BD2KOnFHIR/BLENDINGFHIRandRDF/yosemite_talk/finalreport_data.owl

Finalized Reports Only

Approach is “brittle”:

- Code system hierarchy is replicated as flattened strings
- No link to fact that system is being used
- DataProperty constraints potentially make reasoner more complex

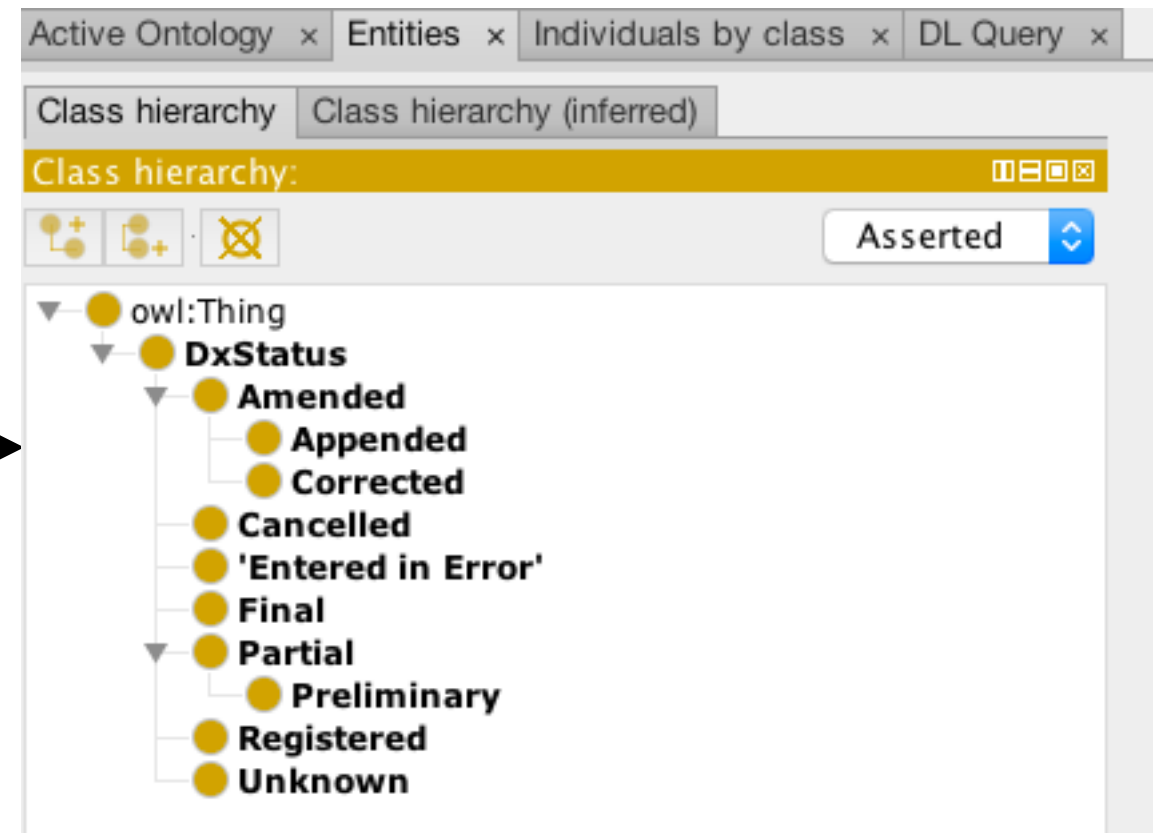
Finalized Reports Proposed Solution

```
prefix fhir: <http://hl7.org/fhir/> .
prefix rdf: <http://www.w3.org/1999/02/22-rdf-syntax-ns#> .
prefix owl: <http://www.w3.org/2002/07/owl#> .
prefix diagnostic-report-status: <http://hl7.org/fhir/diagnostic-report-
prefix xsd: <http://www.w3.org/2001/XMLSchema#> .
prefix skos: <http://www.w3.org/2004/02/skos/core#> .
prefix rdfs: <http://www.w3.org/2000/01/rdf-schema#> .
prefix w5: <http://hl7.org/fhir/w5#> .

diagnostic-report-status:root
  a owl:Class ;
  rdfs:label "DxStatus" ;
  skos:definition "Diagnostic Report Status Values" ;
  skos:prefLabel "DxStatus" .

diagnostic-report-status:partial
  a owl:Class ;
  rdfs:subClassOf diagnostic-report-status:root ;
  rdfs:label "Partial" ;
  skos:definition "This is a partial (e.g. initial, interim or pre-
may be incomplete or unverified." ;
  skos:prefLabel "Partial" .

diagnostic-report-status:cancelled
  a owl:Class ;
  rdfs:subClassOf diagnostic-report-status:root ;
  rdfs:label "Cancelled" ;
  skos:definition "The report is unavailable because the measure-
(also sometimes called 'aborted')." ;
  skos:prefLabel "Cancelled" .
```



1) OWL representation (and URIs!) for *all* code systems ...

Finalized Reports

Proposed Solution (cont)

```
];  
fhir:DiagnosticReport.status [  
  a diagnostic-report-status:final;  
  fhir:value "final";  
  fhir:DiagnosticReport.category [
```

2) Revise FHIR RDF specification to allow `rdf:type` on *all* codes

```
Import(<http://hl7.org/fhir/diagnostic-report-status/>)  
  
...  
Declaration(Class(:FinalStatus))  
SubClassOf(diagnostic-report-status:final :FinalStatus)  
SubClassOf(diagnostic-report-status:amended :FinalStatus)  
  
Declaration(Class(:FinalReport))  
EquivalentClasses(:FinalReport  
ObjectSomeValuesFrom(fhir:DiagnosticReport.status :FinalStatus))
```

Finalized Patient Reports having a Cancer Dx

```
Import(<http://example.org/swat4ls/patientreport>)
Import(<http://example.org/swat4ls/cancerreport>)
Import(<http://example.org/swat4ls/finalreport>)

# Class declaration
Declaration(Class(:FinalPatientReportWithCancerDiagnosis))
AnnotationAssertion(dc:title :FinalPatientReportWithCancerDiagnosis
    "The set of diagnoses that are instances of malignant neoplastic disease
(sct:363346000)")
EquivalentClasses(:FinalPatientReportWithCancerDiagnosis
    ObjectIntersectionOf
        (<http://example.org/swat4ls/patientreport/PatientReport>
        <http://example.org/swat4ls/cancerreport/ReportWithCancerDiagnosis>
        <http://example.org/swat4ls/finalreport/FinalReport>))
)
```

Definition

The screenshot shows a web browser window with the URL `http://example.org/swat4ls/finalpatientcancerreport`. The browser's address bar and tabs show the active ontology. The main content area displays the definition of the class `FinalPatientReportWithCancerDiagnosis`.

Class hierarchy: `FinalPatientReportWithCancerDiagnosis` is shown as a subclass of `ReportWithCancerDiagnosis`. The hierarchy is displayed in a tree view on the left side of the interface.

Description: `FinalPatientReportWithCancerDiagnosis` is defined as:

- Equivalent To: `ReportWithCancerDiagnosis and FinalReport and PatientReport`
- SubClass Of: `ReportWithCancerDiagnosis`
- General class axioms: `DiagnosticReport.subject.link some Patient`, `DiagnosticReport.codedDiagnosis.coding some 'Malignant neoplastic disease (disorder)'`, and `DiagnosticReport.status some FinalStatus`
- SubClass Of (Anonymous Ancestor): `DiagnosticReport.subject.link some Patient`, `DiagnosticReport.codedDiagnosis.coding some 'Malignant neoplastic disease (disorder)'`, and `DiagnosticReport.status some FinalStatus`
- Instances: (None listed)

Result

The screenshot shows a web browser window with the address bar displaying 'finalpatientcancerreport (http://example.org/swat4ls/finalpatientcancerreport)'. The browser has tabs for 'Active Ontology', 'Entities', 'Individuals by class', and 'DL Query'. The 'Active Ontology' tab is selected, showing a class hierarchy on the left and details for 'FinalPatientReportWithCancerDiagnosis' on the right.

Class hierarchy (inferred): FinalPatientReportWithCancerDiagnosis

- owl:Thing
 - administrative
 - clinical
 - conformance
 - DxStatus
 - Element
 - FinalStatus
 - financial
 - infrastructure
 - Narrative.div
 - PatientReport
 - FinalPatientReportWithCancerDiagnosis**
 - Primitive
 - ReportWithCancerDiagnosis
 - FinalPatientReportWithCancerDiagnosis**
 - Resource
 - 'SNOMED CT Concept (SNOMED RT+CTV3)'
 - treeRoot
 - workflow
 - xhtml

FinalPatientReportWithCancerDiagnosis — http://example.org/swat4ls/finalpatientcancerreport

Description: FinalPatientReportWithCancerDiagnosis

Equivalent To +

- ReportWithCancerDiagnosis and FinalReport and PatientReport**

SubClass Of +

- FinalReport
- PatientReport
- ReportWithCancerDiagnosis

General class axioms +

SubClass Of (Anonymous Ancestor)

- DiagnosticReport.subject.link **some** Patient
- DiagnosticReport.codedDiagnosis.coding **some** 'Malignant neoplastic disease (disorder)'
- DiagnosticReport.status **some** FinalStatus

Instances +

- f201**

Post-Coordinated Expressions

```
fhir:DiagnosticReport.conclusion [ fhir:value "CT brains: tumor of the left lobe of the thyroid gland." ];
fhir:DiagnosticReport.codedDiagnosis [
  fhir:index 0;
  fhir:CodeableConcept.coding [
    fhir:index 0;
    a sct:363346000;
    a [ a owl:Restriction ;
      owl:onProperty sct:609096000 ;
      owl:someValuesFrom [ a owl:Restriction ;
        owl:onProperty sct:363698007 ;
        owl:someValuesFrom sct:170784008 ] ] ;
    fhir:Coding.system [ fhir:value "http://snomed.info/sct" ];
    fhir:Coding.code [ fhir:value "363346000:{363698007=170784008}" ];
    fhir:Coding.display [ fhir:value "Malignant tumor of left lobe of thyroid gland" ]
  ]
] .
```

Transformation rules for OWL equivalent



One possible format for compositional expression



https://github.com/BD2KOnFHIR/BLENDINGFHIRandRDF/yosemite_talk/diagnosticreport-example-thyroidtumor.ttl

Thyroid Disease Classifier

...

Declaration(Class(:ReportOfThyroidDisease))

AnnotationAssertion(dc:title :ReportOfThyroidDisease

"Thyroid Disease Dx - disorder of the thyroid gland (sct:14304000)"

EquivalentClasses(:ReportOfThyroidDisease

ObjectSomeValuesFrom(fhir:DiagnosticReport.codedDiagnosis.coding sct:14304000))

)

https://github.com/BD2KOnFHIR/BLENDINGFHIRandRDF/yosemite_talk/thyroidreport.owl

Result

The screenshot displays a software interface with two main panels. The left panel, titled 'Individuals: dxreport117', contains a list of individuals: 'dxreport117' (selected), 'f201', 'f201', 'f203', and 'treeRoot'. Below this is a 'Description: dxreport117' section with a 'Types' list containing 'DiagnosticReport' and 'ReportOfThyroidDisease' (highlighted with a red box). The right panel, titled 'Property assertions: dxreport117', shows a list of object property assertions for 'dxreport117'. Each assertion includes a property name, a value, and a set of four icons (question mark, at-sign, cross, circle).

Property	Value	Icons
DiagnosticReport.conclusion	_:genid23666	? @ x o
DiagnosticReport.performer	_:genid23668	? @ x o
DiagnosticReport.status	_:genid23646	? @ x o
DiagnosticReport.code	_:genid23669	? @ x o
DiagnosticReport.effectiveDateTime	_:genid23671	? @ x o
Resource.id	_:genid23667	? @ x o
nodeRole	treeRoot	? @ x o
DiagnosticReport.codedDiagnosis	_:genid23662	? @ x o
DiagnosticReport.category	_:genid23653	? @ x o
DiagnosticReport.imagingStudy	_:genid23663	? @ x o
DiagnosticReport.issued	_:genid23665	? @ x o
DiagnosticReport.subject	_:genid23670	? @ x o
DomainResource.text	_:genid23655	? @ x o
nodeRole	treeRoot	? @

What *doesn't* work

```
fhir:ImagingStudy.description [ fhir:value "XR Wrist 3+ Views"];
fhir:ImagingStudy.series [
  fhir:index 0;
  fhir:ImagingStudy.series.uid [ fhir:value "urn:oid:2.16.124.113543.6003.1154777499.30246.19789"];
  fhir:ImagingStudy.series.number [ fhir:value "3"^^xsd:nonNegativeInteger ];
  fhir:ImagingStudy.series.modality [
    fhir:Coding.system [ fhir:value "http://nema.org/dicom/dicm" ];
    fhir:Coding.code [ fhir:value "DX" ];
  ];
  fhir:ImagingStudy.series.numberOfInstances [ fhir:value "2"^^xsd:nonNegativeInteger ];
  fhir:ImagingStudy.series.availability [ fhir:value "ONLINE" ];
  fhir:ImagingStudy.series.endpoint [
    fhir:index 0;
    fhir:link <http://hl7.org/fhir/Endpoint/example-wadors>;
    fhir:Reference.reference [ fhir:value "Endpoint/example-wadors" ];
  ];
  fhir:ImagingStudy.series.bodySite [
    a sct:7467003;
    fhir:Coding.system [ fhir:value "http://snomed.info/sct" ];
    fhir:Coding.code [ fhir:value "7467003" ];
    fhir:Coding.display [ fhir:value "Wrist joint structure" ];
  ];
  fhir:ImagingStudy.series.laterality [
    a sct:7771000;
    fhir:Coding.system [ fhir:value "http://snomed.info/sct" ];
    fhir:Coding.code [ fhir:value "7771000" ];
    fhir:Coding.display [ fhir:value "Left" ];
  ];
  fhir:ImagingStudy.series.started [ fhir:value "2011-01-01T11:01:20+03:00"^^xsd:dateTime ];
];
```

Does laterality modify bodySite? Is it an independent attribute?

What we need

```
fhir:ImagingStudy.series.bodySite [
  a sct:7467003;
  a [owl:Restriction;
    owl:onProperty sct:272741003;
    owl:someValuesFrom sct:7771000];
  fhir:Coding.system [ fhir:value "http://snomed.info/sct" ];
  fhir:Coding.code [ fhir:value "7467003" ];
  fhir:Coding.display [ fhir:value "Wrist joint structure" ]
];
fhir:ImagingStudy.series.laterality [
  a sct:7771000;
  fhir:Coding.system [ fhir:value "http://snomed.info/sct" ];
  fhir:Coding.code [ fhir:value "7771000" ];
  fhir:Coding.display [ fhir:value "Left" ]
];
```

Why the imaging study doesn't work

There is a tacit ontological model included in the data (this is always the case...)

The modelers know that the laterality attribute modifies the body site — it isn't an image of a 'left', it is an image of the left wrist.

Transformation is necessary

- Watch the work that Grahame Grieve and Linda Bird are doing on SNOMED model alignment
- Keep an eye on what is happening in the Shape Expressions (ShEx) mapping group

Issues and Discussion

- FHIR Metadata Vocabulary
 - Uses types not recognized in OWL spec (xsd:date, xsd:time, etc)
 - Value Set references not yet included
 - Include path expressions?
- FHIR and RDF
 - URI's for all concept codes
 - OWL rendering of all code systems
 - RDF Profile? URI's, links and link types aren't RDF specific
- Reasoner
 - ELK and Snorocket don't work — have to use FaCT++
 - FaCT++ is too slow for complete SNOMED CT, so we're generating subsets
 - Snorocket community willing to address issues
 - Production environment would need pre-classified SNOMED w/ queries (ala. CTS2 approach)
- Some issues wrt. CONNEG (content negotiation)

Summary

- FHIR RDF allows seamless integration with DL reasoners
- DL reasoners can be applied to many, but not all(!) classification tasks
- Still some “rough edges”, but approach appears to be solid and useable in a production level environment

Credits

This study is supported in part by NIH grants U01 HG009450 and U01 CA18094.

This work was conducted using the Protégé resource, which is supported by grant GM10331601 from the National Institute of General Medical Sciences of the United States National Institutes of Health.

Eric Prud'hommeaux

David Booth

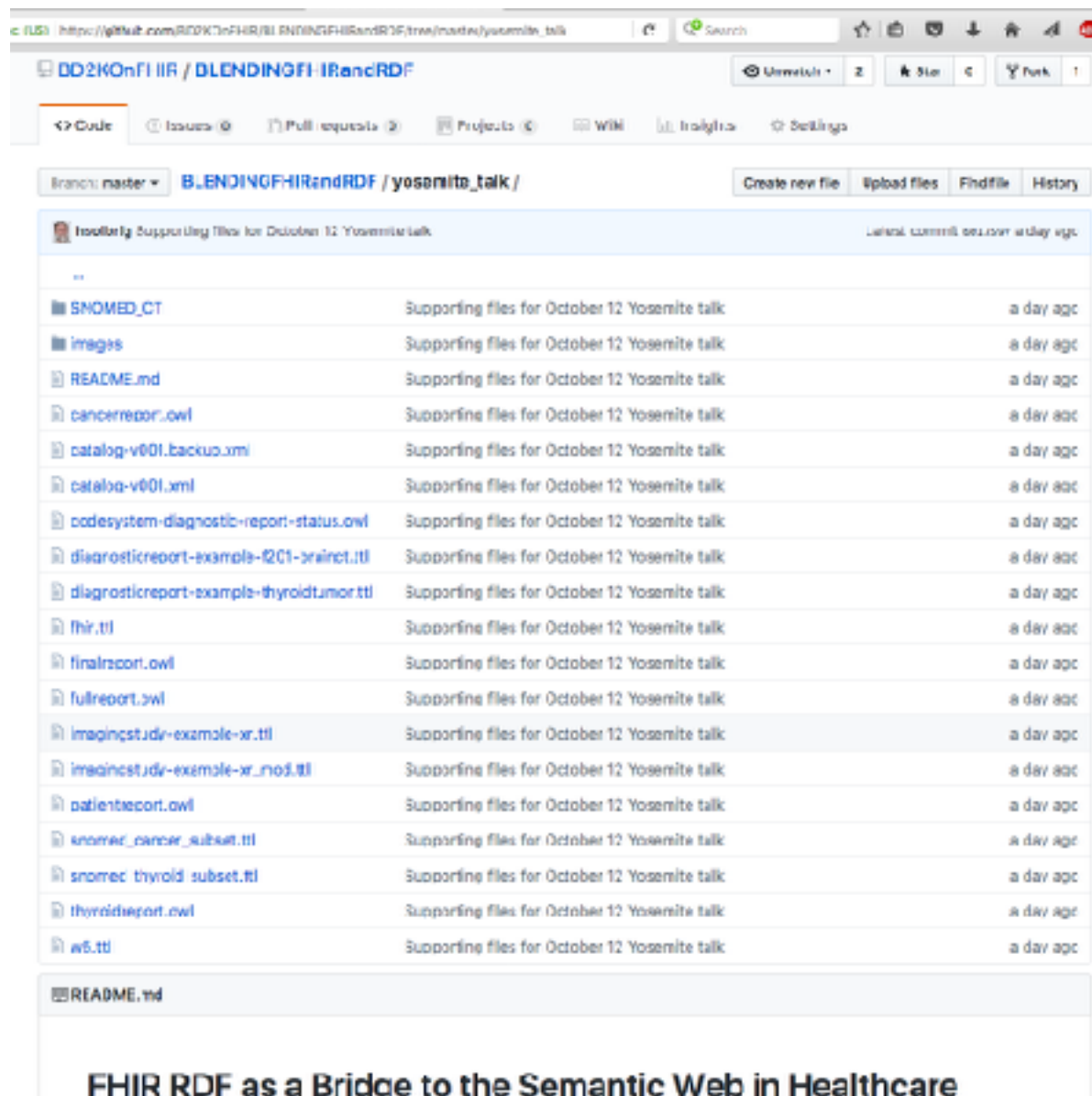
Dr. Guoqian Jiang

The HCLS team

Presentation Materials

Materials for this talk, along with this slide deck can be found at:

https://github.com/BD2KOnFHIR/BLENDINGFHIRandRDF/yosemite_talk



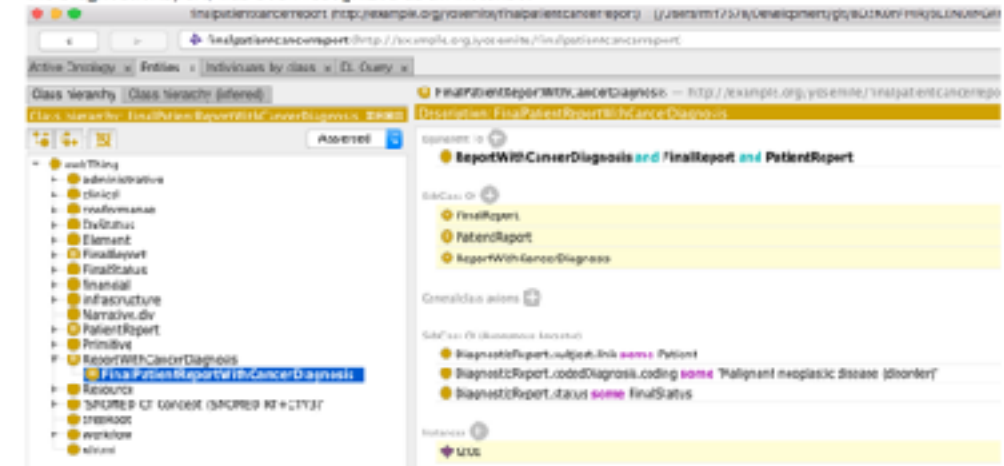
The screenshot shows the GitHub repository page for `BD2KOnFHIR/BLENDINGFHIRandRDF/yosemite_talk`. The repository is under the `master` branch. The file list includes:

File Name	Description	Time
SNOMED_CT	Supporting files for October 12 Yosemite talk	a day ago
images	Supporting files for October 12 Yosemite talk	a day ago
README.md	Supporting files for October 12 Yosemite talk	a day ago
cancerreport.owl	Supporting files for October 12 Yosemite talk	a day ago
catalog-v001.backup.xml	Supporting files for October 12 Yosemite talk	a day ago
catalog-v001.xml	Supporting files for October 12 Yosemite talk	a day ago
codesystem-diagnostic-report-status.owl	Supporting files for October 12 Yosemite talk	a day ago
diagnosticreport-example-1201-brainct.ttl	Supporting files for October 12 Yosemite talk	a day ago
diagnosticreport-example-thyroid.tumor.ttl	Supporting files for October 12 Yosemite talk	a day ago
fhir.ttl	Supporting files for October 12 Yosemite talk	a day ago
finalreport.owl	Supporting files for October 12 Yosemite talk	a day ago
fullreport.owl	Supporting files for October 12 Yosemite talk	a day ago
imagingstudy-example-xr.ttl	Supporting files for October 12 Yosemite talk	a day ago
imagingstudy-example-xr_nod.ttl	Supporting files for October 12 Yosemite talk	a day ago
patientreport.owl	Supporting files for October 12 Yosemite talk	a day ago
snomed_cancer_subset.ttl	Supporting files for October 12 Yosemite talk	a day ago
snomed_thyroid_subset.ttl	Supporting files for October 12 Yosemite talk	a day ago
thyroidreport.owl	Supporting files for October 12 Yosemite talk	a day ago
ws.ttl	Supporting files for October 12 Yosemite talk	a day ago

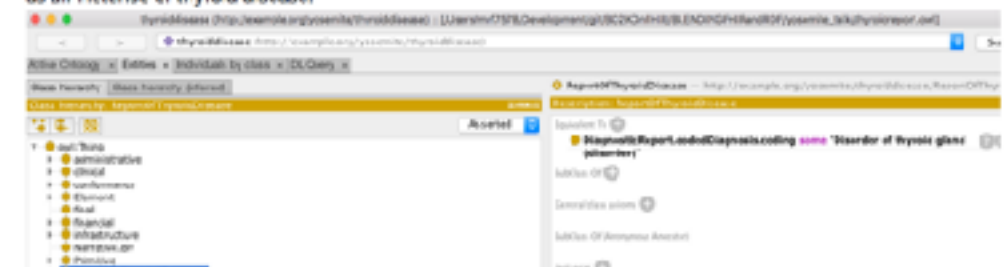
The README.md file is also visible at the bottom of the page.

Use

1. Install a current version of *Protégé* (we use 5.1.0)
2. Clone a copy of the *BLENDINGFHIRandRDF* repository
3. Change to the *yosemite_talk* directory
4. Start *Protégé* and open *fullreport.owl*
5. Select the *FaCT++* reasoner under the *Reasoner* menu
6. Select *Start Reasoner* under the *Reasoner* menu
7. Navigate to *FinalPatientReportWithCancerDiagnosis* in the *Class Hierarchy* tab and observe that *#261* (the id of the *DiagnosticReport*) has been recognized as an instance.



8. Open *thyroidreport.owl*, answering "no" to the current window prompt.
9. Select *Start Reasoner* under the *Reasoner* menu.
10. Navigate to *ReportOfThyroidDisease* in the *Class Hierarchy* tab and observe that *diagnostic117* has been classified as an instance of *thyroid disease*.



Questions

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