Terminology binding in FHIR-RDF

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1 Introduction

There are a couple of requirements which may be easy to meet but have some difficult areas. These two requirements are

- 1. To have achievable round trip transformation of FHIR payloads to RDF
- 2. To be RDF tool friendly in the constructed RDF

For much of the transformation a verbatim (or transliteral) translation will work but there are difficult areas of which at least four have been identified: Terminology, Extension, ValueSet and Resource Reference.

This paper is an attempt to find a middle ground on terminology and ValueSet which can meet both these requirements so that we do not have to develop two versions of RDF.

This version addresses four aspects:

- Alignment with primitive datatypes
- Addition of Codeable Concept including sequence order
- ValueSet Definitions
- ValueSet Bindings

2 Terminology differences

In FHIR the binding to terminology uses the FHIR Coding Type which has various contained values as uri, strings and code. There is no concept of including the terminology model.

RDF has representations of some terminologies – SNOMED CT and ICD-11. Both these representations use OWL Classes to represent the concepts in a hierarchy. A term is referenced as a URI. In SNOMED CT there are also a number of Object Properties.

The bindings to a term of an element in FHIR and RDF are therefore fundamentally different.

FHIR provides the code/system for a CTS to lookup the term. FHIR carries the display value in the instance payload to avoid having to lookup the term.

RDF has a relationship between the element (rdfs:Resource – also called an owl:Named Individual) and the term class. The only relationship available is rdf:type and therefore the element must declare itself or a related individual as member of the Term OWL class.

3 Binding of instance

Example of House Dust Allergen as a FHIR substance resource (from the FHIR DSTU examples).

In an RDF instance binding to a SNOMED concept (defined by IHTSDO) would look like: rec:xxxx rdf:type http://snomed.info/id/406466009

rdf:type is concatenation of "system", "version", and "code" from the FHIR payload The rec: is the instance ontology prefix identification. The RDF can now obtain closure to the concept in the SNOMED ontology if imported.

What is the instance? There have been various approaches to this which range from making the substance the instance with the term type applied to it to a verbatim approach to declare coding and its components as complex types but eventually as primitives. There was resistance to making the substance a semantic type due to the difficulty of round tripping back to the FHIR structure. There was resistance to the verbatim approach since OWL does not match on strings only on URIs so it is not possible to get closure to imported terminologies.

There is a middle ground to make "coding" (or code) an instance where the Substance instance has an object property "Substance.type" to the Codeable Concept which has Coding instances and text. Coding is mapped verbatim with specific annotated derived additions to make it understandable by the reasoner.

There are two ways to arrive at the fully populated RDF:

- 1. Assemble the entities from the FHIR Resource and then compute the derivations
- 2. Assemble the entities from a minimal RDF and compute the FHIR structure.

The RDF representation is fully populated.

Here are RDF target substance and coding instances represented in Manchester Syntax (from Protégé).

Record#01339 is the substance instance (OWL Named Individual).

Its type is equivalent to its tag since it is a root element of the resource.

The name.value field containing "House Dust" is mapped to the rdfs:label annotation so it will show up as that in the OWL tool. The annotation on the annotation is a fhir:derivation which expresses the computation.

Its "type" as an Object Property identified as Substance.type points to record#01351

Its "name" as an Object Property identified as Substance.name points to record#01353

Annotations for order define the sequence in which the properties would be rendered in a FHIR XML or JSON payload. It is derived from the Substance class definition (see Model mappings).

The "name" field record#01353 is a fhir:string (inferred) which again has the rdfs:label computed from name.value

OWL types can be inferred from the ObjectProperty Range of the Object Property (see Model mappings) and is not required in the exchange.

```
Individual: <http://record#01353>
Annotations:
Annotations: rdfs:isDefinedBy "String.value"
rdfs:label "House Dust"
Facts:
fhir:value "House Dust"^^fhir:string-primitive
```

Substance.type is a fhir:CodeableConcept (inferred) can include multiple coding and then a text. (See Model mappings)

```
Individual: <http://record#01351>
   Annotations:
           Annotations: fhir:derivation "Label cannot be computed since codings can have different
values"
       rdfs:label "Codeable Concept"
   Facts:
    Annotations: fhir:order "1"
                fhir:CodeableConcept.coding <http://record#01349>,
Individual: <http://record#01349>
   Annotations:
           Annotations: fhir:derivation "display.value"
       rdfs:label "House dust allergen"
   Types:
           Annotations: fhir:derivation "From a translation of system to Ontology URI and the version
and code appended to form the Term URI"
       <http://snomed.info/id/406466009>
    Facts:
    fhir:Coding.system <http://record#01340>,
     fhir:Coding.display <http://record#01342>,
    fhir:Coding.code <http://record#01341>
```

```
Individual: <http://record#01340>
Annotations:
    rdfs:label "http://snomed.info/id"
Facts:
    fhir:value "http://snomed.info/id/"^^fhir:uri-primitive
Individual: <http://record#01341>
Annotations:
    rdfs:label "406466009"
Facts:
    fhir:value "406466009"^^fhir:code-primitive
Individual: <http://record#01342>
Annotations:
    rdfs:label "House dust allergen"
Facts:
```

fhir:value "House dust allergen"^^fhir:string-primitive

Record#01349 is an instance of fhir:Coding and has an rdfs:label of the Coding.display.value so it shows up (displays) in the OWL tool.

The "code", "version" and "system" are concatenated into the standard URI for the term which is also applied as a rdf:type to the coding thus providing the graph closure to the terminology if imported. If the terminology is not imported the system and code are still retained independently.

Below is the graph of the example shown in Protégé Ontograph with closure to SNOMED (sct:) where its label (in this case) is identical to the label of the Coding instance.



4 Mapping of FHIR (Model) to OWL

4.1 Type mappings

The mapping of the CodeableConcept type from FHIR to OWL is described as follows:

FHIR Model element	OWL Type/Property	Comment
CodeableConcept type extends Element	Class– subclass of Element	
CodeableConcept.code	Object Property target Class Coding	
CodeableConcept.text	Object Property target Class string	

Coding complex type extends Element	Class- subclass of Element	In line with transliteral mapping from Complex Object.
		Object properties have to be gathered from Element
Coding.id (Element.id)	record: <uri></uri>	If element.id is empty it will be generated on the RDF side as it is required.
Coding.extension (Element.extension)		Tbd – will cover this in discussion of extensions
Coding.system	Object property – target Class fhir:uri	
Coding.version	Object property – target Class fhir:string	
Coding.code	Object property – target Class fhir:code	
Coding.display	Object property – target Class fhir:string	
	Coding.display.value Mapped to Coding rdfs:label	Allows tool to show display
Coding.primary	Object property – target Class fhir:Boolean	Verbatim
Coding.system	Mapped to Coding rdf:type	Allows the Coding instance to
Coding.version		bind to the imported Ontology if
Coding.code		System is pound indirectly to URI prefix
Coding.valueset	Class – represents valueset Resource	See later section

4.2 Substance OWL Example

Target is defined as:

Class: fhir:Substance

```
Annotations:
   rdfs:label "Substance"
SubClassOf:
    fhir:Resource,
       Annotations: fhir:order "1"
    fhir:Substance.identifier max 1 fhir:Identifier,
       Annotations: fhir:order "2"
    fhir:Substance.name exactly 1 fhir:string,
       Annotations: fhir:order "3"
    fhir:Substance.type max 1 fhir:CodeableConcept,
       Annotations: fhir:order "4"
    fhir:Substance.description max 1 fhir:string,
       Annotations: fhir:order "5"
    fhir:Substance.status max 1 fhir:CodeableConcept,
        Annotations: fhir:order "6"
    fhir:Substance.effectiveTime max 1 fhir:Period,
```

Annotations: fhir:order "7" fhir:Substance.quantity max 1 fhir:Quantity, Annotations: fhir:order "8" fhir:Substance.ingredient some fhir:Substance Annotations: fhir:order "9" fhir:Substance.quantityMode max 1 fhir:CodeableConcept,

Class: CodeableConcept

Annotations:

rdfs:comment "The set of possible coded values this coding was chosen from or constrained by.", rdfs:comment "If the element is present, it must have a value for at least one of the defined elements, an @id referenced from the Narrative, or extensions" SubClassOf:

Element,

Annotations: order 1 CodeableConcept.coding some Coding, Annotations: order 2 CodeableConcept.text max 1 string

Class: Coding

SubClassOf: Element, Annotations: order "1", rdfs:comment "The identification of the code system that defines the meaning of the symbol in the code." Coding.system max 1 uri

Annotations: order "2",

rdfs:comment "The version of the code system which was used when choosing this code. Note that a well-maintained code system does not need the version reported, because the meaning of codes is consistent across versions. However this cannot consistently be assured. and when it is not, the version SHOULD be exchanged."

Coding.version max 1 string,

Annotations: order "3", rdfs:comment "A symbol in syntax defined by the system. The symbol may be a predefined code or an expression in a syntax defined by the coding system (e.g. post-coordination)." Coding.code max 1 code,

Annotations: order "4", rdfs:comment "A representation of the meaning of the code in the system, following the rules laid out by the system." Coding.display max 1 string,

```
Annotations: order "5",
rdfs:comment "Indicates that this code was chosen by a user directly - i.e. off a pick
list of available codes."
Coding.primary max 1 boolean,
```

Annotations: order "6", rdfs:comment "The set of possible coded values this coding was chosen from or constrained by." Coding.valueset max 1 Valueset,

Datatype definitions are declared in another document.

5 ValueSets



In mapping this to OWL, concepts are declared as named Classes (e.g. in SNOMED CT). Concepts which are contained are declared as subclasses.

The fhir:Define contains Concepts from a single terminology system and is therefore a superclass or a class expression. The classes come from the terminology. The depth of class hierarchy is defined by the value set expression (TBD).

5.1 Internal Valuesets

5.1.1 Sample ValueSet

Here is an example of an HL7 internal Valueset instance for severity of an adverse reaction:

```
<ValueSet xmlns="http://hl7.org/fhir">
 <text>
   <status value="generated"/>
   <div xmlns="http://www.w3.org/1999/xhtml">
    <h2>ReactionSeverity</h2>
    The severity of an adverse reaction.
    This value set defines its own terms in the system http://hl7.org/fhir/reactionSeverity
    \langle t.d \rangle
         <b>Code</b>
        <b>Display</b>
        <b>Definition</b>
        severe
         <a name="severe"> </a>
        Severe complications arose due to the reaction.
      serious
         <a name="serious"> </a>
```

```
Serious inconvenience to the subject.
     >moderate
         <a name="moderate"> </a>
       Moderate inconvenience to the subject.
     minor
         <a name="minor"> </a>
       Minor inconvenience to the subject.
     \langle /div \rangle
</text>
<identifier value="http://hl7.org/fhir/vs/reactionSeverity"/>
<name value="ReactionSeverity"/>
<publisher value="HL7 (FHIR Project)"/>
<telecom>
 <system value="url"/>
  <value value="http://hl7.org/fhir"/>
</telecom>
<telecom>
  <system value="email"/>
 <value value="fhir@lists.hl7.org"/>
</telecom>
<description value="The severity of an adverse reaction."/>
<status value="draft"/>
<date value="2014-09-30T18:09:16.978+10:00"/>
<define>
 <system value="http://hl7.org/fhir/reactionSeverity"/>
 <caseSensitive value="true"/>
```

```
<concept>
     <code value="severe"/>
     <definition value="Severe complications arose due to the reaction."/>
   </concept>
   <concept>
     <code value="serious"/>
     <definition value="Serious inconvenience to the subject."/>
   </concept>
   <concept>
     <code value="moderate"/>
     <definition value="Moderate inconvenience to the subject."/>
   </concept>
   <concept>
     <code value="minor"/>
     <definition value="Minor inconvenience to the subject."/>
    </concept>
 </define>
</ValueSet>
```

5.1.2 Sample RDF ValueSet Class

The Valuset defined in RDF will be a class as opposed to an individual. The class URI is the ValueSet.identifier.

```
Class: <http://hl7.org/fhir/vs/reactionSeverity>
Annotations:
Annotations: rdfs:comment "Maps from ValueSet.publisher"
fhir:publisher "HL7 (FHIR Project)",
Annotations: rdfs:comment "Maps from ValueSet.name"
rdfs:label "ReactionSeverity",
Annotations: rdfs:comment "Maps from valueSet.description"
```

```
rdfs:comment "The severity of an adverse reaction.",
Annotations: rdfs:comment "Maps from ValueSet.telecom with system = email"
fhir:e-mail "fhir"@lists.hl7.org,
Annotations: rdfs:comment "Maps from ValueSet.experimental"
fhir:experimental "",
Annotations: rdfs:comment "Maps from ValueSet.telecom System = url"
fhir:url "http://hl7.org/fhir",
Annotations: rdfs:comment "Maps from ValueSet.status"
fhir:status "draft",
Annotations: rdfs:comment "Maps from ValueSet.date"
fhir:publishDate "2014-09-30T18:09:16.978+10:00"
SubClassof:
fhir:Terminology,
fhir:ValueSet
```

This specific ValueSet is a subclass of both fhir:Terminology and fhir:ValueSet. Some ObjectProperties in the ValueSet are mapped to annotations of the Class. The Objectproperty ValueSet.define gets mapped to subclasses of the ValueSet Class.

Some mappings are declared as annotation on an annotation.

5.1.3 Sample Concept Class

Here is the define.concept for the "minor" code.

```
Class: fhir:minor
Annotations:
Annotations: fhir:derivation "Maps from ValueSet.define.concept.definition"
rdfs:comment "Minor inconvenience to the subject.",
Annotations: fhir:derivation "maps from ValueSet.define.concept.code"
rdfs:label "minor"
SubClassOf:
<http://hl7.org/fhir/vs/reactionSeverity>
```

5.2 ValueSet Binding

5.2.1 Profile ValueSet Binding

The following fragment is from the Adverse Reaction profile for the resource. An element AdverseReaction.symptom.severity has a definition.binding which points to the ValueSet to be used – in this

```
case "http://hl7.org/fhir/vs/reactionSeverity".
```

```
<element>
 <path value="AdverseReaction.symptom.severity"/>
 <definition>
   <short value="severe | serious | moderate | minor"/>
   <formal value="The severity of the sign or symptom."/>
   <min value="0"/>
   <max value="1"/>
   <type>
      <code value="code"/>
   </type>
   <isModifier value="false"/>
   <binding>
     <name value="ReactionSeverity"/>
      <isExtensible value="false"/>
      <conformance value="required"/>
      <referenceResource>
        <reference value="http://hl7.org/fhir/vs/reactionSeverity"/>
     </referenceResource>
   </binding>
    <mapping>
      <identity value="rim"/>
```

```
<map value="inboundRelationship[typeCode=SUBJ].source[classCode=OBS, moodCode=EVN,
code=<&lt;severity&gt;&gt;
        ].value"/>
        </mapping>
        <identity value="v2"/>
            <map value="v2"/>
            <map value="AL1-4-Allergy Severity Code / IAM-4-Allergy Severity Code"/>
        </mapping>
        </definition>
        </element>
```

5.2.2 RDF ValueSetBinding

Range:

fhir:code,

In the RDF equivalent, the profile element becomes an ObjectProperty:

<http://hl7.org/fhir/vs/reactionSeverity>

```
Class: fhir:AdverseReaction.Symptom
Annotations:
    rdfs:label "AdverseReaction Symptom"
SubClassOf:
    fhir:Element,
    fhir:AdverseReaction.symptom.severity max 1 <http://hl7.org/fhir/vs/reactionSeverity>
ObjectProperty: fhir:AdverseReaction.symptom.severity
    Characteristics:
        Functional
    Domain:
        fhir:AdverseReaction.Symptom
```

Thus an occurence of AdverseReaction.symptom.severity will have a range structural type of fhir:code and a semantic ValueSet of http://hl7.org/fhir/vs/reactionSeverity . The instance of code will be a member of the set of codes as well as a member of the set defined by reactionSeverity. When the code is assigned – e.g. minor, that class is a subclass of the ValueSet and therefore conforms to the constraint however it could be typed the ValueSet itself which would not make sense. We also need to define that the terms are disjoint.

We can eliminate the ValueSet type itself by declaring the ValueSet to be a union of its subclasses (closed world):

```
Class: <http://hl7.org/fhir/vs/reactionSeverity>
EquivalentTo:
fhir:minor
or fhir:moderate
or fhir:serious
or fhir:severe
```

Thus the partial definition of the RDF ValueSet would be:

```
Class: <http://hl7.org/fhir/vs/reactionSeverity>
   Annotations:
            Annotations: fhir:derivation "Maps from ValueSet.publisher"
        fhir:publisher "HL7 (FHIR Project)",
            Annotations: fhir:derivation "Maps from ValueSet.name"
        rdfs:label "ReactionSeverity",
           Annotations: fhir:derivation "Maps from valueSet.description"
        rdfs:comment "The severity of an adverse reaction.",
           Annotations: fhir:derivation "Maps from ValueSet.telecom with system = email"
        fhir:e-mail "fhir"@lists.hl7.org,
            Annotations: fhir:derivation "Maps from ValueSet.experimental"
        fhir:experimental "",
           Annotations: fhir:derivation "Maps from ValueSet.telecom System = url"
        fhir:url "http://hl7.org/fhir",
            Annotations: fhir:derivation "Maps from ValueSet.status"
        fhir:status "draft",
```

```
Annotations: fhir:derivation "Maps from ValueSet.date"

fhir:publishDate "2014-09-30T18:09:16.978+10:00"

EquivalentTo:

fhir:minor

or fhir:moderate

or fhir:serious

or fhir:severe

SubClassOf:

fhir:Terminology,

fhir:ValueSet

DisjointClasses:
```

fhir:minor, fhir:moderate, fhir:serious, fhir:severe

5.3 External ValueSets

External ValueSets are expected to be treated in the same way. The challenge will be to define the exclusion of the ValueSet from the allowed types since they may be declared by a Terminology Organization. It is possible to add a supplemental ontology which declares the equivalence outside the terminology ontology itself.