Data Standards for HCT

BRIDG

HL7 FHIR

Bob Milius, PhD
Bioinformatics Research
NMDP/Be The Match

Co-Chair, HL7 Clinical Genomics Workgroup
BRIDG Steering Committee
What is BRIDG?

• Biomedical Research Integrated Domain Group

• A formal model of the shared semantics of regulated clinical trials research

• A communication bridge between
  • clinical trial domain experts and technical experts
  • different models of clinical trials information

• An open community of stakeholders

• The semantic foundation for application and message development through model-driven architecture
Stakeholders

- Standards Development
  - CDISC
    - Clinical Data Interchange Standards Consortium
  - HL7
    - Health Level Seven International
  - ISO
    - International Organization for Standardization

- Regulatory
  - FDA
    - Food and Drug Administration
  - NCI
    - National Cancer Institute
Scope of BRIDG

• 2015
  – Protocol-driven research and its associated regulatory artifacts

• 2017
  – Basic, pre-clinical, clinical, and translational research and its associated regulatory artifacts.
The BRIDG Model

- Person
- Study Subject
- Study Site
- Organization
- Product
- Study Protocol
- Activities
- Observations
- Observation Results
The Hematopoietic Cell Transplant Domain

• BRIDG-compatible model
  – associated Common Data Elements (CDEs) registered in the Cancer Data Standards Registry (caDSR)
  – covering the high impact forms submitted to the CIBMTR research database.
HCT Content in the BRIDG Model

• BRIDG Release 3.2
  – Released 7 Sep 2012
  – 2116 mapping specifications within BRIDG HCT
  – 1900 CDE elements within BRIDG
  – 38 forms
HCT Content in the BRIDG Model

Number of FormsNet data points mapped to the BRIDG Model*
BRIDG 5.0 Scope

- **New Semantics**
  - Imaging
    - relevant parts of DICOM + NCI Annotated Imaging Markup (AIM)
  - Study Management
    - Vendor project
    - NCI Surveillance, Epidemiology, and End Results Program (SEER)

- **New Views**
  - Oncology (NCI + CDISC Oncology domains)
  - CDISC SDTM 3.1.3 (plus VS, RS, EX domains)

- **Controlled Vocabulary**
  - Compilation of Controlled Vocabulary for Imaging
BRIDG 5.0 Timeline - 2017

- January – BRIDG 5.0 Released
- Feb – ISO ballot opens
- March – CDISC ballot opens
- April – HL7 ballot opens
- May – Joint ballot reconciliation begins
What about…

HL7® FHIR®?
Health Level 7 International

• Non-profit, ANSI-accredited healthcare Standards Developing Organization (SDO)

• “HL7 provides standards for interoperability that improve care delivery, optimize workflow, reduce ambiguity and enhance knowledge transfer among all of our stakeholders, including healthcare providers, government agencies, the vendor community, fellow SDOs and patients”
What HL7 is and isn’t

- Does NOT dictate how to build applications or present data
- NOT a software application

- IS a framework to enable independent systems to communicate with each other
- IS a “book of rules” with thousands of pages
<table>
<thead>
<tr>
<th><strong>Messages</strong></th>
<th><strong>Documents</strong></th>
</tr>
</thead>
<tbody>
<tr>
<td>Transient</td>
<td>Persistent</td>
</tr>
<tr>
<td>Trigger based</td>
<td>Wholeness</td>
</tr>
<tr>
<td>Non-persistent</td>
<td>Clinician authentication</td>
</tr>
<tr>
<td></td>
<td>Human readable</td>
</tr>
</tbody>
</table>
HL7 Standards

Messages
- V2
- V3
- Fast

Documents
- Clinical Document Architecture (CDA)
- FHIR
What would healthcare exchange look like if we started from scratch using modern approaches?
• Fast Healthcare Interoperability Resources
  – hl7.org/fhir

• Newest standard being developed by HL7

• Industry standard (for trial use)
  – Standard for Trial Use (STU) 3 in March 2017

• Aligns with REST services
  – XML, JSON

• Active Testing
  – Three Connectathons a year, 14 so far (almost 5 years)

• Immense interest!
  – Sync For Science (S4S)
  – Joint development with HSPC
  – Argonaut – vendor initiated project
Project Argonaut

“The purpose of the Argonaut Project is to rapidly develop a first generation FHIR-based API and Core Data Services specification to enable expanded information sharing for electronic health records and other health information technology based on Internet standards and architectural patterns and styles.”

“Epic, Cerner, others join HL7 project”

HIMMS17: Google Cloud & HL7 FHIR

“Today we’re announcing support for the HL7 FHIR Foundation to help the developer community advance data interoperability efforts. The FHIR open standard defines a modern, web API-based approach to communicating healthcare data, making it easier to securely communicate across the healthcare ecosystem including hospitals, labs, applications and research studies.”

Gregory J. Moore MD, PhD
Vice President of Healthcare, Google Cloud

https://www.blog.google/topics/google-cloud/google-cloud-himss-engaging-healthcare-and-health-it-community/
"FDA encourages sponsors and clinical investigators to work with the entities that control the EHRs, such as health care organizations, to use EHRs and EDC systems that are interoperable. EHRs may be interoperable with EDC systems in a variety of ways depending on supportive technologies and standards. Interoperable technology may involve automated electronic transmission of relevant EHR data to the EDC system. For example, data elements originating in an EHR (e.g., demographics, vital signs, past medical history, past surgical history, social history, medications, adverse reactions) may automatically populate the eCRFs within an EDC system."

Who’s using FHIR?

Systems accessing HAPI server in 2015

© 2015 HL7 ® Int’l. Licensed under Creative Commons. HL7, Health Level Seven, FHIR & flame logo are registered trademarks of Health Level Seven International. Reg. U.S. TM Office.
Welcome to FHIR®

First time here? See the executive summary, the developer's introduction, clinical introduction, or architect's introduction, and then the FHIR overview / roadmap & Timelines. See also the open license (and don't miss the full Table of Contents or you can search this specification).

Level 1 Basic framework on which the specification is build

- **Foundation**
  - Base Documentation, XML, JSON, REST API + Search, Data Types, Extensions

Level 2 Supporting Implementation, and binding to external specifications

- **Implanter Support**
  - Downloads, Common Use Cases, Testing
- **Security & Privacy**
  - Security, Consent
- **Conformance**
  - StructureDefinition, CapabilityStatement, Profiling
- **Terminology**
  - CodeSystem, ValueSet, ConceptMap, TerminologySvc

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, etc.
- **Diagnostics**
  - Observation, Report, Request, etc.
- **Medications**
  - Order, Dispense, Administration, Statement, etc.
- **Workflow**
  - Task, Subscription, etc.
- **Financial**
  - Claim, EligibilityRequest, etc.
Resources

“Resources” are:

- Small logically discrete units of exchange
- Defined behaviour and meaning
- Known identity / location
- Smallest unit of transaction
- “of interest” to healthcare

- V2: Sort of like Segments
- V3: Sort of like CMETs
What’s a Resource?

Examples

• Administrative
  – Patient, Practitioner, Organization, Location, Coverage, Invoice

• Clinical Concepts
  – Allergy, Condition, Family History, Care Plan, Observation, Specimen, DiagnosticReport

• Infrastructure
  – Document, Message, Profile, Conformance

Non-examples

• Gender
  – Too small

• Electronic Health Record
  – Too big

• Blood Pressure
  – Too specific

• Intervention
  – Too broad

100-150 total - ever

http://www.slideshare.net/DavidHay5/introduction-to-fhir-new-zealand-seminar
Support “Common” Scenarios

• Inclusion of content in core specification is based on “80%” rule
  – Only include data elements we are confident that most (~80%) of normal implementations using that resource will make use of
  – Other content in extensions
  – Easy to say, governance challenge to achieve

• Resources are simple and easy to understand & use
Profiles

• Document constraints and extensions on one or more resources for a Use Case

• May also define new extensions search terms, new messaging events, etc.
FHIR solutions

Resources + Extensions = Solution
### 3.0 Resource Index

This page is provided to help find resources quickly. There is also a more detailed classification, ontology, and description.

<table>
<thead>
<tr>
<th><strong>Clinical</strong></th>
<th><strong>Care Provision</strong></th>
<th><strong>Medication &amp; Immunization</strong></th>
<th><strong>Diagnostics</strong></th>
</tr>
</thead>
<tbody>
<tr>
<td>General:</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>AllergyIntolerance</td>
<td>CarePlan 1</td>
<td>Observation 3</td>
</tr>
<tr>
<td></td>
<td>Condition (Problem)</td>
<td>CareTeam 0</td>
<td>DiagnosticReport 3</td>
</tr>
<tr>
<td></td>
<td>Procedure 1</td>
<td>Goal 1</td>
<td>DiagnosticOrder 1</td>
</tr>
<tr>
<td></td>
<td>FamilyMemberHistory</td>
<td>Protocol 0</td>
<td>Specimen 1</td>
</tr>
<tr>
<td></td>
<td>ClinicalImpression</td>
<td>ReferralRequest 1</td>
<td>Sequence 0</td>
</tr>
<tr>
<td></td>
<td>DetectedIssue 1</td>
<td>ProcedureRequest 1</td>
<td>BodySite 0</td>
</tr>
<tr>
<td></td>
<td></td>
<td>NutritionOrder 1</td>
<td>ImagingStudy 2</td>
</tr>
<tr>
<td></td>
<td></td>
<td>RiskAssessment 0</td>
<td>ImagingManifest 1</td>
</tr>
<tr>
<td></td>
<td></td>
<td>VisionPrescription 0</td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th><strong>Identification</strong></th>
<th><strong>Groups:</strong></th>
<th><strong>Entities:</strong></th>
<th><strong>Devices:</strong></th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Individuals:</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Patient 3</td>
<td>Organization 1</td>
<td>Location 1</td>
<td>Device 1</td>
</tr>
<tr>
<td>Practitioner 1</td>
<td>HealthcareService 1</td>
<td>Substance 1</td>
<td>DeviceComponent 1</td>
</tr>
<tr>
<td>PractitionerRole 0</td>
<td>Group 1</td>
<td>Person 1</td>
<td>DeviceMetric 1</td>
</tr>
<tr>
<td>RelatedPerson 1</td>
<td></td>
<td>Contract 0</td>
<td></td>
</tr>
</tbody>
</table>
## Patient Resource Structure

<table>
<thead>
<tr>
<th>Name</th>
<th>Flags</th>
<th>Card.</th>
<th>Type</th>
<th>Description &amp; Constraints</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Patient</strong></td>
<td></td>
<td></td>
<td>DomainResource</td>
<td>Information about an individual or animal receiving health care services</td>
</tr>
<tr>
<td>identifier</td>
<td>☐</td>
<td>0..*</td>
<td>Identifier</td>
<td>An identifier for this patient</td>
</tr>
<tr>
<td>active</td>
<td>☐ ☐</td>
<td>0..1</td>
<td>boolean</td>
<td>Whether this patient’s record is in active use</td>
</tr>
<tr>
<td>name</td>
<td>☐</td>
<td>0..*</td>
<td>HumanName</td>
<td>A name associated with the patient</td>
</tr>
<tr>
<td>telecom</td>
<td>☐</td>
<td>0..*</td>
<td>ContactPoint</td>
<td>A contact detail for the individual</td>
</tr>
<tr>
<td>gender</td>
<td>☐</td>
<td>0..1</td>
<td>code</td>
<td>male</td>
</tr>
<tr>
<td>birthDate</td>
<td>☐</td>
<td>0..1</td>
<td>date</td>
<td>The date of birth for the individual</td>
</tr>
<tr>
<td>deceased</td>
<td>☐ ☐</td>
<td>0..1</td>
<td>boolean</td>
<td>Indicates if the individual is deceased or not</td>
</tr>
<tr>
<td>deceasedBoolean</td>
<td>☐ ☐</td>
<td></td>
<td>boolean</td>
<td>Addresses for the individual</td>
</tr>
<tr>
<td>deceasedDateTime</td>
<td>☐ ☐</td>
<td></td>
<td>dateTime</td>
<td>Marital (civil) status of a patient</td>
</tr>
<tr>
<td>address</td>
<td>☐</td>
<td>0..*</td>
<td>Address</td>
<td>Marital Status Codes (Required)</td>
</tr>
<tr>
<td>maritalStatus</td>
<td>☐</td>
<td>0..1</td>
<td>CodeableConcept</td>
<td>Whether patient is part of a multiple birth</td>
</tr>
<tr>
<td>multipleBirth</td>
<td>☐</td>
<td>0..1</td>
<td>CodeableConcept</td>
<td>Image of the patient</td>
</tr>
<tr>
<td>relationship</td>
<td>☐</td>
<td>0..*</td>
<td>CodeableConcept</td>
<td>A contact party (e.g. guardian, partner, friend) for the patient</td>
</tr>
<tr>
<td>name</td>
<td>☐</td>
<td>0..1</td>
<td>HumanName</td>
<td>The kind of relationship</td>
</tr>
<tr>
<td>telecom</td>
<td>☐</td>
<td>0..*</td>
<td>ContactPoint</td>
<td>PatientContactRelationship (Extensible)</td>
</tr>
<tr>
<td>address</td>
<td>☐</td>
<td>0..1</td>
<td>Address</td>
<td>A name associated with the contact person</td>
</tr>
<tr>
<td>gender</td>
<td>☐</td>
<td>0..1</td>
<td>code</td>
<td>A contact detail for the person</td>
</tr>
<tr>
<td>organization</td>
<td>☐</td>
<td>0..1</td>
<td>Reference(Organization)</td>
<td>Address for the contact person</td>
</tr>
<tr>
<td>period</td>
<td>☐</td>
<td>0..1</td>
<td>Period</td>
<td>Organization that is associated with the contact person</td>
</tr>
<tr>
<td>animal</td>
<td>☐ ☐</td>
<td>0..1</td>
<td>BackboneElement</td>
<td>The period during which this contact person or organization is valid to be contacted relating to this patient</td>
</tr>
<tr>
<td>species</td>
<td>☐ ☐</td>
<td>1..1</td>
<td>CodeableConcept</td>
<td>This patient is known to be an animal (non-human)</td>
</tr>
<tr>
<td>breed</td>
<td>☐ ☐</td>
<td>0..1</td>
<td>CodeableConcept</td>
<td>E.g. Dog, Cow</td>
</tr>
<tr>
<td>genderStatus</td>
<td>☐ ☐</td>
<td>0..1</td>
<td>CodeableConcept</td>
<td>E.g. Poodle, Angus</td>
</tr>
<tr>
<td>communication</td>
<td>☐</td>
<td>0..*</td>
<td>BackboneElement</td>
<td>E.g. Neutered, Intact</td>
</tr>
<tr>
<td>language</td>
<td>☐</td>
<td>1..1</td>
<td>CodeableConcept</td>
<td>Language of (Required)</td>
</tr>
<tr>
<td>preferred</td>
<td>☐</td>
<td>0..1</td>
<td>boolean</td>
<td>Language preference indicator</td>
</tr>
<tr>
<td>careProvider</td>
<td>☐ ☐</td>
<td>0..1</td>
<td>Reference(Organization)</td>
<td>Patient's nominated primary care provider</td>
</tr>
<tr>
<td>managingOrganization</td>
<td>☐ ☐</td>
<td>0..1</td>
<td>Reference(Organization)</td>
<td>Organization that is the custodian of the patient record</td>
</tr>
<tr>
<td>link</td>
<td>☐ ☐</td>
<td>0..*</td>
<td>BackboneElement</td>
<td>Link to another patient resource that concerns the same actual person</td>
</tr>
<tr>
<td>other</td>
<td>☐ ☐</td>
<td>0..1</td>
<td>Reference(Patient)</td>
<td>The other patient resource that the link refers to</td>
</tr>
<tr>
<td>type</td>
<td>☐ ☐</td>
<td>1..1</td>
<td>code</td>
<td>replace</td>
</tr>
</tbody>
</table>
5.1.2 Resource Content

UML Diagram

**Patient** (DomainResource)
- identifier: Identifier [0..*]
- active: boolean [0..1]
- name: HumanName [0..*]
- telecom: ContactPoint [0..*]
- gender: code [0..1] "AdministrativeGender!"
- birthDate: date [0..1]
- deceased[x]: Type [0..1] « boolean|dateTime »
- address: Address [0..*]
- maritalStatus: CodeableConcept [0..1] « Marital Status! »
- multipleBirth[x]: Type [0..1] « boolean|integer »
- photo: Attachment [0..*]
- careProvider: Reference [0..*] « Organization | Practitioner »
- managingOrganization: Reference [0..1] « Organization »

**Animal**
- species: CodeableConcept [1..1] « AnimalSpecies!? »
- breed: CodeableConcept [0..1] « AnimalBreeds!? »
- genderStatus: CodeableConcept [0..1] « GenderStatus!? »

**Link**
- other: Reference [1..1] « Patient »
- type: code [1..1] « Linktype! »

**Contact**
- relationship: CodeableConcept [0..*] « PatientContactRelationship+ »
- name: HumanName [0..1]
- telecom: ContactPoint [0..*]
- address: Address [0..1]
- gender: code [0..1] « AdministrativeGender! »
- organization: Reference [0..1] « Organization »
- period: Period [0..1]
<?xml version="1.0" encoding="UTF-8"?>

<Patient xmlns="http://hl7.org/fhir">
  <!-- from Resource: id, meta, implicitRules, and language -->
  <!-- from DomainResource: text, contained, extension, and modifierExtension -->
  <identifier>
    <!-- 0..1 Identifier An identifier for this patient -->
  </identifier>
  <active value="[boolean]"/>
    <!-- 0..1 Whether this patient's record is in active use -->
  <name>
    <!-- 0..1 HumanName A name associated with the patient -->
  </name>
  <telecom>
    <!-- 0..1 ContactPoint A contact detail for the individual -->
  </telecom>
  <gender value="[code]"/>
    <!-- 0..1 male | female | other | unknown -->
  <birthDate value="[date]"/>
    <!-- 0..1 The date of birth for the individual -->
  <deceased value="false"/>
    <!-- 0..1 Indicates if the individual is deceased or not -->
  <address>
    <!-- 0..1 Address Addresses for the individual -->
  </address>
  <maritalStatus>
    <!-- 0..1 CodeableConcept Marital (civil) status of a patient -->
  </maritalStatus>
  <multipleBirth value="false"/>
    <!-- 0..1 Whether patient is part of a multiple birth -->
  <photo>
    <!-- 0..1 Attachment Image of the patient -->
  </photo>
  <contact>
    <!-- 0..1 A contact party (e.g. guardian, partner, friend) for the patient -->
  </contact>
  <relationship>
    <!-- 0..1 CodeableConcept The kind of relationship -->
  </relationship>
  <name>
    <!-- 0..1 HumanName A name associated with the contact person -->
  </name>
  <telecom>
    <!-- 0..1 ContactPoint A contact detail for the person -->
  </telecom>
  <address>
    <!-- 0..1 Address Address for the contact person -->
  </address>
  <gender value="[code]"/>
    <!-- 0..1 male | female | other | unknown -->
  <organization>
    <!-- 0..1 Reference(Organization) Organization that is associated with the contact -->
  </organization>
  <period>
    <!-- 0..1 Period The period during which this contact person or organization is valid to be contacted relating to this patient -->
  </period>
  <animal>
    <!-- 0..1 This patient is known to be an animal (non-human) -->
  </animal>
  <species>
    <!-- 1..1 CodeableConcept E.g. Dog, Cow -->
  </species>
  <breed>
    <!-- 0..1 CodeableConcept E.g. Poodle, Angus -->
  </breed>
  <genderStatus>
    <!-- 0..1 CodeableConcept E.g. Neutered, Intact -->
  </genderStatus>
  <communication>
    <!-- 0..* A list of Languages which may be used to communicate with the patient about his or her health -->
  </communication>
  <language>
    <!-- 1..1 CodeableConcept The language which can be used to communicate with the patient about his or her health -->
  </language>
  <preferred value="[boolean]"/>
    <!-- 0..1 Language preference indicator -->
  </communication>
  <careProvider>
    <!-- 0..* Reference(Organization|Practitioner) Patient's nominated primary care provider -->
  </careProvider>
  <managingOrganization>
    <!-- 0..1 Reference(Organization) Organization that is the custodian of the patient record -->
  </managingOrganization>
  <link>
    <!-- 0..1 Reference другая пациентная запись -->
  </link>
  <other>
    <!-- 0..1 Reference(Patient) The other patient resource that the link refers to -->
  </other>
  <type value="[code]"/>
    <!-- 1..1 replace | refer | see also - type of link -->
</link>
</Patient>
Simple example: Patient.xml

```xml
<Patient>
  <name>
    <family value="Doe"/>
    <given value="John"/>
  </name>
  <gender value="male"/>
  <birthDate value="1974-12-25"/>
</Patient>
```
Many tools to connect to a FHIR server

- curl (command line)
- Postman (Google Chrome add-on)
- Programming language, e.g., Python, Java
- Web client (e.g., HAPI FHIR server)
  - http://fhirtest.b12x.org/
- clinFHIR
  - http://clinfhir.com/
- etc…
cURL command to POST a Patient

$ curl -X POST -d @Patient.xml -H "Content-Type: text/xml" http://fhirtest.b12x.org/baseDstu3/Patient

<OperationOutcome xmlns="http://hl7.org/fhir">
  <text>
    <status value="generated"/>
  <div xmlns="http://www.w3.org/1999/xhtml">
    <h1>Operation Outcome</h1>
    <table border="0">
      <tr>
        <td style="font-weight: bold;">INFORMATION</td>
        <td>[
        <pre>Successfully created resource &quot;Patient/1481/_history/1&quot; in 178ms</pre>
        </td>
      </tr>
    </table>
  </div>
</text>

<issue>
  <severity value="information"/>
  <code value="informational"/>
  <diagnostics value="Successfully created resource &quot;Patient/1481/_history/1&quot; in 178ms"/>
</issue>
cURL command to GET a Patient

$ curl -X GET http://fhirtest.b12x.org/baseDstu3/Patient/1481

```
{
    "resourceType": "Patient",
    "id": "1481",
    "meta": {
        "versionId": "1",
        "lastUpdated": "2017-02-13T15:25:21.000+00:00"
    },
    "text": {
        "status": "generated",
        "div": "<div xmlns="http://www.w3.org/1999/xhtml"><div class="hapiHeaderText">John <b>DOE </b></div><table class="hapiPropertyTable"><tbody><tr><td>Date of birth</td><td><span>25 December 1974</span></td></tr></tbody></table></div>
}
```

```
"name": [
    {
        "family": ["Doe"
        ],
        "given": ["John"
        ]
    },
    "gender": "male",
    "birthDate": "1974-12-25"
}
```
cURL command to GET a Patient

```bash
$ curl -X GET http://fhirtest.b12x.org/baseDstu3/Patient/1481?_format=xml
```

```xml
<Patient xmlns="http://hl7.org/fhir">
  <id value="1481"/>
  <meta>
    <versionId value="1"/>
    <lastUpdated value="2017-02-13T15:25:21.000+00:00"/>
  </meta>
  <text>
    <status value="generated"/>
    <div xmlns="http://www.w3.org/1999/xhtml">
      <div class="hapiHeaderText">John <b>DOE</b></div>
      <table class="hapiPropertyTable">
        <tbody>
          <tr>
            <td>Date of birth</td>
            <td><span>25 December 1974</span></td>
          </tr>
        </tbody>
      </table>
    </div>
  </text>
  <name>
    <family value="Doe"/>
    <given value="John"/>
  </name>
  <gender value="male"/>
  <birthDate value="1974-12-25"/>
</Patient>
```
### Specimen Resource Structure

<table>
<thead>
<tr>
<th>Name</th>
<th>Flag</th>
<th>Card.</th>
<th>Type</th>
<th>Description &amp; Constraints</th>
</tr>
</thead>
<tbody>
<tr>
<td>Specimen</td>
<td></td>
<td></td>
<td>DomainResource</td>
<td></td>
</tr>
<tr>
<td>identifier</td>
<td>Σ</td>
<td>0..*</td>
<td>Identifier</td>
<td>External Identifier</td>
</tr>
<tr>
<td>accessionId</td>
<td>Σ</td>
<td>0..1</td>
<td>Identifier</td>
<td>Identifier assigned by the lab</td>
</tr>
<tr>
<td>status</td>
<td>Π Σ</td>
<td>0..1</td>
<td>code</td>
<td>available</td>
</tr>
<tr>
<td>type</td>
<td>Σ</td>
<td>0..1</td>
<td>CodeableConcept</td>
<td>Kind of material that forms the specimen</td>
</tr>
<tr>
<td>subject</td>
<td>Σ</td>
<td>1..1</td>
<td>Reference(Patient</td>
<td>Group</td>
</tr>
<tr>
<td>receivedTime</td>
<td>Σ</td>
<td>0..1</td>
<td>dateTime</td>
<td>The time when the specimen was received for processing</td>
</tr>
<tr>
<td>parent</td>
<td>0..*</td>
<td></td>
<td>Reference(Specimen)</td>
<td>Specimen from which this specimen originated</td>
</tr>
<tr>
<td>request</td>
<td>0..*</td>
<td></td>
<td>Reference(ProcedureRequest)</td>
<td>Why the specimen was collected</td>
</tr>
<tr>
<td>collection</td>
<td>0..*</td>
<td></td>
<td>BackboneElement</td>
<td>Collection details</td>
</tr>
<tr>
<td>collector</td>
<td>Σ</td>
<td>0..1</td>
<td>Reference(Practitioner)</td>
<td>Who collected the specimen</td>
</tr>
<tr>
<td>collected[x]</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>collectedDateTime</td>
<td></td>
<td></td>
<td>dateTime</td>
<td></td>
</tr>
<tr>
<td>collectedPeriod</td>
<td></td>
<td></td>
<td>Period</td>
<td></td>
</tr>
<tr>
<td>quantity</td>
<td>0..1</td>
<td></td>
<td>SimpleQuantity</td>
<td>The quantity of specimen collected</td>
</tr>
<tr>
<td>method</td>
<td>0..1</td>
<td></td>
<td>CodeableConcept</td>
<td>Technique used to perform collection</td>
</tr>
<tr>
<td>bodySite</td>
<td>0..1</td>
<td></td>
<td>CodeableConcept</td>
<td>Anatomical collection site</td>
</tr>
<tr>
<td>processing</td>
<td>0..*</td>
<td></td>
<td>BackboneElement</td>
<td>Processing and processing step details</td>
</tr>
<tr>
<td>description</td>
<td>0..1</td>
<td></td>
<td>string</td>
<td>Textual description of procedure</td>
</tr>
<tr>
<td>procedure</td>
<td>0..1</td>
<td></td>
<td>CodeableConcept</td>
<td>Indicates the treatment step applied to the specimen</td>
</tr>
<tr>
<td>additive</td>
<td>0..*</td>
<td></td>
<td>Reference(Substance)</td>
<td>Material used in the processing step</td>
</tr>
<tr>
<td>time[x]</td>
<td></td>
<td></td>
<td></td>
<td>Date and time of specimen processing</td>
</tr>
<tr>
<td>container</td>
<td>0..*</td>
<td></td>
<td>BackboneElement</td>
<td>Direct container of specimen (tube, slide, etc.)</td>
</tr>
<tr>
<td>identifier</td>
<td>Σ</td>
<td>0..*</td>
<td>Identifier</td>
<td>Id for the container</td>
</tr>
<tr>
<td>description</td>
<td>0..1</td>
<td></td>
<td>string</td>
<td>Textual description of the container</td>
</tr>
<tr>
<td>type</td>
<td>0..1</td>
<td></td>
<td>CodeableConcept</td>
<td>Kind of container directly associated with specimen</td>
</tr>
<tr>
<td>capacity</td>
<td>0..1</td>
<td></td>
<td>SimpleQuantity</td>
<td>Container volume or size</td>
</tr>
<tr>
<td>specimenQuantity</td>
<td>0..1</td>
<td></td>
<td>SimpleQuantity</td>
<td>Quantity of specimen within container</td>
</tr>
<tr>
<td>additive[x]</td>
<td>0..1</td>
<td></td>
<td></td>
<td>Additive associated with container</td>
</tr>
<tr>
<td>additiveCodeableConcept</td>
<td></td>
<td></td>
<td>CodeableConcept</td>
<td>v2 Additive/Preservative</td>
</tr>
<tr>
<td>additiveReference</td>
<td></td>
<td></td>
<td>Reference(Substance)</td>
<td></td>
</tr>
<tr>
<td>note</td>
<td>0..*</td>
<td></td>
<td>Annotation</td>
<td>Comments</td>
</tr>
</tbody>
</table>

**Example:**

- SpecimenCollectionMethod
- SpecimenProcessingProcedure
- SpecimenContainer
10.4.3 Resource Content

**Specimen Resource Structure**

UML Diagram

- **Collection**
  - collector: Reference [0..1] « Practitioner »
  - collected[x]: Type [0..1] « dateTime|Period »
  - quantity: Quantity(SimpleQuantity) [0..1]
  - method: CodeableConcept [0..1] « SpecimenCollectionMethod »
  - bodySite: CodeableConcept [0..1] « SNOMED CT Body Structures »

- **Specimen (DomainResource)**
  - identifier: Identifier [0..*]
  - accessionIdentifier: Identifier [0..1]
  - status: code [0..1] « SpecimenStatus »
  - type: CodeableConcept [0..1] « v2-Specimen Type »
  - subject: Reference [1..1] « Patient|Group|Device|Substance »
  - receivedTime: dateTime [0..1]
  - parent: Reference [0..*] « Specimen »
  - request: Reference [0..*] « ProcedureRequest »
  - note: Annotation [0..*]

- **Processing**
  - description: string [0..1]
  - procedure: CodeableConcept [0..1] « Specimen ProcessingProcedure »
  - additive: Reference [0..*] « Substance »
  - time[x]: Type [0..1] « dateTime|Period »

- **Container**
  - identifier: Identifier [0..*]
  - description: string [0..1]
  - capacity: Quantity(SimpleQuantity) [0..1]
  - specimenQuantity: Quantity(SimpleQuantity) [0..1]
  - additive[x]: Type [0..1] « CodeableConcept Reference(Substance); v2 Additive/Preservative »
<Specimen>
  <identifier>
    <system value="http://myorganization.com"/>
    <value value="123"/>
  </identifier>
  <type>
    <coding>
      <system value="http://snomed.info/sct"/>
      <code value="258564008"/>
      <display value="Buccal smear sample"/>
    </coding>
  </type>
  <subject>
    <reference value="Patient/1481"/>
    <display value="John Doe"/>
  </subject>
</Specimen>
Resource relationships

Patient/Donor

Specimen

Observation

Sequence (DNA)
Developing Transaction Bundles for Scenarios such as this...

• A donor registry (Organization) collected a buccal swab (Specimen) from a potential stem cell donor (Patient).

• A lab order (DiagnosticRequest) for HLA class I typing of HLA-A, -B, and -C was sent to the typing lab (Organization).

• Typing done by sequencing exons 2 and 3 of each gene (Sequence).

• The results were uploaded as a transaction bundle consisting of the final report (DiagnosticReport) and the supporting information including the evidence leading to genotyping for each gene (Observations), identification of separate alleles (Observations), and sequencing data for each exon (Sequences).
Building an HLA Typing Report with FHIR Resources & Profiles

Informed by MIRING & HML

Typing Report

- Patient/Donor
- Specimen
- Sequence (DNA)
- Observation
- DiagnosticReport
- etc
Reporting genotypes of a panel of HLA loci in a single report

Each locus is a separate observation.
Patient/Donor

Specimen

Sequence

Sequence

Sequence

Sequence

Genetics Profile

Genetics Profile

Genetics Profile

Genetics Profile

Observation

Observation

Observation

Observation

HLA Results Profile

Diagnostic Report

Exon sequencing

Individual alleles

Genotype for one gene/locus

Transaction Bundle
What’s next…

• Developing HL7 FHIR based HLA typing reports
  – New profiles and terminology resources
  – Tools to support these (clients, servers)
• Extend to Form 2005 (Confirmation of HLA Typing)
  – Deconstruct into bundle of resources
• Other Forms!
• Use BRIDG & caDSR to inform
Links to information and tools

• **BRIDG**
  – Home
    • http://bridgmodel.nci.nih.gov
  – Download
    • https://bridgmodel.nci.nih.gov/download-model/bridg-releases
  – Sparx Enterprise Architect – free read only version

• **HL7 FHIR**
  – Home
    • http://hl7.org/fhir
  – Current Build
    • http://build.fhir.org
  – Chat
    • http://chat.fhir.org
  – Wiki
Links to HL7 FHIR Tutorials
(use ‘anonymous’ for username and your email for the password)

• Introduction to FHIR

• FHIR for Executives

• FHIR for Architects

• FHIR for Developers
Thank you!

Questions?

Contact Bob Milius, PhD
bmilius@nmdp.org