Please see FHIR Genomics spec on slide 41

**FHIR Genomics / Connectathon Preparation**

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**Recommendation**

FHIR could be included as an emerging standard, especially for transport of data. Argonaut may provide opportunities to advance.

Sample uses of FHIR: authorization; genetics, family health history, build on current work on **SMART on FHIR Genomics**

<table>
<thead>
<tr>
<th>Recommendation</th>
<th>Actions to Advance</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>FHIR could be included as an emerging standard, especially for transport of data. Argonaut may provide opportunities to advance.</strong></td>
<td><strong>Apply accelerators (e.g., S&amp;I Initiative, pilot project, policy guidance) to existing standards by ONC</strong></td>
</tr>
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<td>Sample uses of FHIR: authorization; genetics, family health history, build on current work on <strong>SMART on FHIR Genomics</strong></td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Recommendation</th>
<th>Actions to Advance</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>2016 PMI S&amp;I:</strong></td>
<td><strong>Apply accelerators (e.g., S&amp;I Initiative, pilot project, policy guidance) to existing standards by ONC</strong></td>
</tr>
<tr>
<td>Additional ONC investment in pilots of <strong>FHIR for PMI</strong> research/individual data donation use case</td>
<td></td>
</tr>
</tbody>
</table>
FHIRE - Fast Healthcare Interoperability Resources

Strong extensibility and flexibility
Concise and easily understood specifications
Human-readable
Well-defined data model and API

FHIR vs HL7 v2 ↑ Consistency, flexibility, conciseness
FHIR vs HL7 v3 ↓ Complexity

Easy to implement!
FHIRE Genomics Time Line

- **Observation Genetics Profile**: Sep 2015
- **Sequence Resource + 6 Genetics Profiles**: Dec 2015

DSTU2 | Current Build | Connectathon | Jan 2016
Domain Analysis Model (DAM) Use Cases

1. Specimen Identification
2. Clinical Sequencing (Germline)
3. Cancer Profiling (Somatic)
4. CDS (Family History and Drug Dosage Calculator)
5. Public Health Reporting
6. Clinical and Research Data Warehouses

...
FHIR Hands-on: Connectathon
How to Run the Server:

$ cd ~/FHIR-Genomics-2-Version-Jan-8
$ python server.py -d

Open Chrome browser in VM:
Go to: http://127.0.0.1:2048/

Sign up using email address or using the public account:
Email address: name@mail.com
Password: password
(optional) Get the latest code.

$ cd ~/FHIR-Genomics-2-Version-Jan-8
$ git pull
Rename the folder with Version

Or
$ git clone http://github.com/chaiery/FHIR-Genomics-2
Rename the folder with Version
Get Sequence

GET [base]/[type]/[id]

Local version: http://127.0.0.1:2048/api/Sequence
Online version: http://genomics-advisor.smartplatforms.org:2048/api/Sequence

Result:

```
<total xmlns:os="http://a9.com/-/spec/opensearch/1.1/">10</total>
```

GET /api/Sequence HTTP/1.1
Get Sequence

http://127.0.0.1:2048/api/Sequence/0681a048-f21b-4301-93c4-559b2c5f99b4
http://genomics-advisor.smartplatforms.org:2048/api/Sequence/0681a048-f21b-4301-93c4-559b2c5f99b4
Search (1) – general search

GET [base]/[type]{?[parameters]}
GET /api/Sequence?variationID=rs3820015 HTTP/1.1

http://127.0.0.1:2048/api/Sequence?variationID=rs3820015

```xml
<Bundle xmlns="http://hl7.org/fhir">
  <id>
    http://localhost:2048/api/Sequence?variationID=rs3820015
  </id>
  <meta/>
  <type>searchset</type>
  <link/>
  <total xmlns:os="http://a9.com/-/spec/opensearch/1.1/">1</total>
  <entry>
    <fullUrl>
      http://localhost:2048/api/Sequence/0681a048-f21b-4301-93c4-559b2c5f99b4
    </fullUrl>
    <Sequence xmlns="http://hl7.org/fhir">
      <observedAllele value="A"/>
      <species/>
      <text/>
      <coordinate/>
      <variationID>
        <coding>
          <code value="rs3820015"/>
        </coding>
      </variationID>
    </Sequence>
  </entry>
</Bundle>
```
Search (2) – modifier

For element whose type is CodeableConcept:

```
<species>
  <text value="Homo sapiens"/>
  <coding>
    <code value="337915000"/>
    <system value="http://snomed.info/sct"/>
  </coding>
</species>
```

1. GET /api/Sequence?species=337915000
2. GET /api/Sequence?species=Homo%20sapiens
3. GET /api/Sequence?species=Homo            # partly hit
4. GET /api/Sequence?species=http://snomed.info/sct|337915000

modifier
Add data to the database: 3 ways

1. Online submit

http://genomics-advisor.smartplatforms.org:2048/submit
Upload a JSON file or fill in the text box

```json
{"resourceType": "Sequence",
 "type": "DNA",
 "text":
  {
  "status": "generated",
  "div": "Genotype of rs71662879 is A/A"
  },
  "coordinate": [
  {
  "start": 114951390,
  "end": 114951390,
  "chromosome": {"text": "1"},
  "genomeBuild": {"text": "GRCh37"}
  }
  ]

Submit
```
The most critical requirements for submitting data successfully is having **accurate** and **valid** data.
Find code examples of instances for Sequence resource, observationfor-genetics profile, reportfor-genetics profile, orderfor-genetics profile, consensus-sequence-block profile, hla-result profile, family-member-history-genetic profile, Patient resource, etc.

$ cd ~/FHIR-Genomics-2-Version-Jan-8/fhir/examples/guidance/
2. Use API:

Upload sequence data:

```python
def upload_seq(seq, access_token):
    resp = requests.post('%s/Sequence?_format=json' % config.API_BASE,
                          data=json.dumps(seq),
                          headers={'Authorization': 'Bearer %s' % access_token})
```

Sample app code using FHIR Genomics API:
https://github.com/chaiery/ga2fhir
web.py
3. Use script: submit.py (example)

```python
def load_from_file(path, relevant_dir):
    abspath = os.path.join(relevant_dir, path)
    print(abspath)
    with open(abspath) as f:
        return json.loads(f.read())

def init(resource):
    dir = os.path.join(BASEDIR, 'examples/' + resource)
    load_instance = partial(load_from_file, relevant_dir=dir)
    list_of_file = os.listdir(dir)
    list_of_instance = []
    for i in list_of_file:
        if '.json' in i:
            list_of_instance.append(i)
    availables = map(load_instance, list_of_instance)
    for i in availables:
        instance = dict(i)
        save_resource(resource, instance)
        print('Created %s % resource
        break

if __name__ == '__main__':
    from server import app
    with app.app_context():
        init('Practitioner')
        init('Organization')
        test_resource = partial(Resource, owner_id='name@mail.com')
        for _ in xrange(8):
            patient = rand_patient()
        commit_buffers(BUF)
```

VM: ~/FHIR-Genomics-2/submit.py
Architecture of the Server

model

Load examples
Load specifications
Data submit
Set up database
Model

Core modules:

api.py
Gets API requests,

processes SMART API

fhir_api.py
Processes FHIR API requests

fhir_parser.py
Parse FHIR

Indexer.py
Index data in database

models.py
Data models

query_builder.py
FHIR to database queries
Server description

FHIR Genomics server actions:
1. Parse the data (Are these data in correct FHIR Genomics structure & types?)
2. Index the data for database use
3. Commit structured data to database
4. Respond to client queries

SMART Genomics additions:
1. Transform raw data to FHIR specification
2. Store search-optimized GA4GH/raw sequence data
3. Authenticate/Authorize
4. Get context for apps
5. Privacy/sharing
**Example – Search**

Request: GET /api/Sequence?species=337915000

api.py

```python
def handle_resource(resource_type):
    if resource_type in ['callsets', 'variantsets', 'readgroupsets', 'referencesets', 'variant']:
        if request.method == 'GET':
            return ga4gh.api.ga_handle_search(request, resource_type)
    if resource_type not in RESOURCES:
        return fhir_error.inform_not_found()
```
Screenshot on how to run server

To get Virtual Machine:
Please email ga@alum.mit.edu with subject exactly as follows:
“Please send Webinar VM”

Server is open source:
On github

Online server:
http://genomics-advisor.smartplatforms.org:2048/
Domain Analysis Model (DAM) Use Cases

1. Specimen Identification
2. Clinical Sequencing (Germline)
3. Cancer Profiling (Somatic)
4. CDS (Family History and Drug Dosage Calculator)
5. Public Health Reporting
6. Clinical and Research Data Warehouses

...
Changes in the population of cells with particular mutations will change over time as well as in conjunction with events such as therapy. Therefore, clearly annotating these specimens as somatic and capturing annotations related to a time relevant to a treatment timeline may be critical for analysis.

Get all variants of a patient named Monika Furman obtained from somatic analysis

GET /observationforgenetics?subject:Patient.name:text=Monika Furman&Source:text=somatic

<Bundle xmlns="http://hl7.org/fhir">
  <id></id>
  <meta/>
  <type>searchset</type>
  <link/>
  <total xmlns:os="http://a9.com/-spec/opensearch/1.1/">5</total>
  <entry>
    <fullUrl></fullUrl>
    <observationforgenetics xmlns="http://hl7.org/fhir">
      <extension>
        <url value="http://hl7.org/fhir/StructureDefinition/observation-geneticsSource"/>
        <valueCodeableConcept>
          <text value="Somatic"/>
          <coding>
            <code value="LA6684-0"/>
            <system value="http://hl7.org/fhir/LOINC-48002-0-answerlist"/>
          </coding>
        </valueCodeableConcept>
      </extension>
      <extension/>
      <extension/>
    </observationforgenetics>
  </entry>
  <entry>
    <fullUrl></fullUrl>
    <observationforgenetics xmlns="http://hl7.org/fhir">
      <category/>
      <status value="final"/>
      <code/>
      <extension>
        <url value="http://hl7.org/fhir/StructureDefinition/observation-geneticsSource"/>
        <valueCodeableConcept>
          <text value="Somatic"/>
        </valueCodeableConcept>
      </extension>
    </observationforgenetics>
  </entry>
</Bundle>
### Connectathon FHIR Genomics Scenarios

<table>
<thead>
<tr>
<th>Case</th>
<th>Importance</th>
<th>Skill</th>
<th>Difficulty</th>
</tr>
</thead>
<tbody>
<tr>
<td>Case 1: Register a New Sequence and Observation</td>
<td>Create the atomic genetics data: DNA/RNA/Protein sequence</td>
<td>1: Operation: Create 2: Sequence resource &amp; observation for genetics profile (Observation resource)</td>
<td>1</td>
</tr>
<tr>
<td>Case 2: Clinical Sequencing – Germline Testing</td>
<td>Searching for gemline/somatic variations</td>
<td>1: Operation: Search 2: Sequence resource &amp; observation for genetics profile (Observation resource)</td>
<td>3</td>
</tr>
<tr>
<td>Case 3: Family Member History</td>
<td>Read the genetics information from family member history</td>
<td>1: Operation: Get 2: report for genetics profile (DiagnosticReport resource), FamilyMemberHistory-genetics (FamilyMemberHistory resource)</td>
<td>5</td>
</tr>
</tbody>
</table>

(For difficulty: 1<sup>st</sup> is the easiest)
<table>
<thead>
<tr>
<th>Case</th>
<th>Importance</th>
<th>Skill</th>
<th>Difficulty</th>
</tr>
</thead>
<tbody>
<tr>
<td>Case 4: Clinical and Research Data Warehouse</td>
<td>Variation info from population</td>
<td>1: Operation: Search 2: observationforgenetics profile (Observation resource), Sequence resource</td>
<td>3</td>
</tr>
<tr>
<td>Case 5: HLA Typing</td>
<td>Support the reports of hla results</td>
<td>1: Operation: Create 2: consensus-sequence-block profile (Observation resource) hlaresult profile (DiagnosticReport resource)</td>
<td>5</td>
</tr>
<tr>
<td>Case 6: Specimen Identification</td>
<td>Read the source of the sequence</td>
<td>1: Operation: Search 2: Sequence resource</td>
<td>2</td>
</tr>
<tr>
<td>Case</td>
<td>Importance</td>
<td>Skill</td>
<td>Difficulty</td>
</tr>
<tr>
<td>-------------------------------</td>
<td>-----------------------------------------------------------------------------</td>
<td>----------------------------------------------------------------------</td>
<td>------------</td>
</tr>
<tr>
<td>Case 7: Comprehensive Pathology Report</td>
<td>Support comprehensive diagnostic report: recording genetics observation using various methods</td>
<td>1: Operation: Create reportforogenetics profile (DiagnosticReport resource)</td>
<td>4</td>
</tr>
<tr>
<td></td>
<td></td>
<td>2: reportforogenetics profile (DiagnosticReport resource)</td>
<td></td>
</tr>
<tr>
<td>Case 8: Sequence Quality</td>
<td>Record the sequence quality</td>
<td>1: Operation: Read/Search</td>
<td>3</td>
</tr>
<tr>
<td></td>
<td></td>
<td>2: Sequence resource</td>
<td></td>
</tr>
</tbody>
</table>

Some general skills: Python, JavaScript
Familiar with FHIR Genomics resources/profiles
What FHIR Genomics accomplishes?

**Extends**
FHIR for genomics/omics data

**Achieves**
conciseness with consistency

**Maximizes**
relevance to clinical-genomic applications

**Leverages**
existing Domain Analysis Model (DAM),
repositories, e.g. GA4GH, ClinVar, etc.
FHIR Connectathon: January 9-10 in Orlando FL

**Learn**  from experts and teams in advance and at event

**Tackle**  clinical genomic use cases

**Develop**  apps and put in app gallery
# All Published Versions of FHIR

The following versions of FHIR are available:

## Current Versions

<table>
<thead>
<tr>
<th>Date</th>
<th>Version</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Oct 24, 2015</td>
<td>1.0.1</td>
<td>Current Official Published Version <em>(Currently: DSTU2 with 1 technical errata)</em></td>
</tr>
<tr>
<td>(current)</td>
<td></td>
<td><em>(last commit)</em> Current Development build (about 30min behind version control, may be incoherent and change rapidly)*</td>
</tr>
<tr>
<td><strong>DSTU 2.1 sequence</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Dec 11, 2015</td>
<td>1.2.0</td>
<td>Connectathon 11 Snapshot (temporary)</td>
</tr>
<tr>
<td>Dec 3, 2015</td>
<td>1.1.0</td>
<td>GAO Ballot + draft changes to main FHIR standard</td>
</tr>
</tbody>
</table>

## DSTU 2 sequence

<table>
<thead>
<tr>
<th>Date</th>
<th>Version</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Oct 24, 2015</td>
<td>1.0.1</td>
<td><strong>DSTU 2 (Official version)</strong> with 1 technical errata (Permanent home)</td>
</tr>
<tr>
<td>Aug 31, 2015</td>
<td>1.0.0</td>
<td>DSTU 2 QA Preview + CQIF Ballot (Sep 2015)</td>
</tr>
<tr>
<td>April 2, 2015</td>
<td>0.5.0</td>
<td>DSTU 2 Ballot version (May 2015 Ballot)</td>
</tr>
<tr>
<td>Dec 12, 2014</td>
<td>0.4.0</td>
<td>Draft For Comment (January 2015 Ballot)</td>
</tr>
</tbody>
</table>

## DSTU 1 sequence

<table>
<thead>
<tr>
<th>Date</th>
<th>Version</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sept 30, 2014</td>
<td>0.0.82</td>
<td><strong>DSTU 1 (Official version)</strong> with 2 technical errata (Permanent home)</td>
</tr>
</tbody>
</table>
FHIR Genomics Connectathon URLs

Sequence Resource
http://www.hl7.org/fhir/2016Jan/sequence.html

Diagnostic Report for Genetics
http://www.hl7.org/fhir/2016Jan/diagnosticreport-genetics.html

Observation for Genetics
http://www.hl7.org/fhir/2016Jan/observation-genetics.html

Diagnostic Order for Genetics
http://www.hl7.org/fhir/2016Jan/diagnosticorder-genetics.html
People who helped make virtual machine, server site, apps, etc

SMART on FHIR
David Kreda
Rachel Ramoni
Josh Mandel
Ken Mandl
Isaac Kohane
...

SMART on FHIR Genomics
Heming Yao
Tom Chen
Peijin Zhang
Jeremy Warner
...

SMART on FHIR Genomics (Asia)
Jiaoyun Yang
Duan Rong
Bowen Gong
Ning An
...

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To get slides, virtual machine, or more info:
Please email ga@alum.mit.edu subject line as follows:
“Please send Webinar VM” or
“Please send Webinar slides”
**FHIR is the first standard for electronic medical records that is:**

<table>
<thead>
<tr>
<th>Developer friendly</th>
<th>Designed to be developer-friendly from outset (JSON, RESTful API, etc.)</th>
</tr>
</thead>
<tbody>
<tr>
<td>EMR-consortium supported</td>
<td>Argonaut project ensures all EMR vendors are in sync on supporting standard implementation</td>
</tr>
<tr>
<td>Geared for Precision Medicine</td>
<td>Being designed for next generation clinical genomics</td>
</tr>
</tbody>
</table>
## Composition of FHIR Genomics

<table>
<thead>
<tr>
<th>Resource</th>
<th>Profile</th>
</tr>
</thead>
<tbody>
<tr>
<td>Observation</td>
<td><em>Observation-genetics</em> profile</td>
</tr>
<tr>
<td>DiagnosticReport</td>
<td><em>DiagnosticReport-genetics</em> profile</td>
</tr>
<tr>
<td>FamilyMemberHistory</td>
<td><em>FamilyMemberHistory-genetics</em> profile</td>
</tr>
<tr>
<td>DiagnosticOrder</td>
<td><em>DiagnosticOrder-genetics</em> profile</td>
</tr>
<tr>
<td><strong>Sequence</strong></td>
<td><em>(Future profiles)</em></td>
</tr>
<tr>
<td>DiagnosticReport</td>
<td><em>HLA-genotyping results</em> profile <em>(specialized</em></td>
</tr>
</tbody>
</table>
**Advantages**

**No/low repetition**  Maximizes re-use of existing FHIR resources

**Well-organized**  Localizes additional genomics/omics data in Sequence

**Easy to implement**  Design is developer-friendly (latest web technologies)

**Consistency**  All genetics observation use Observation-genetics profile
Sequence resource

Single variant and key elements or sequence

External repository references

Extensible to new omics
Sequence resource

Elements for a single variant

Coordinate for reference sequence
Sequence resource

Attributes

<table>
<thead>
<tr>
<th>Attribute</th>
<th>Multiplicity</th>
<th>Type</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>specie</td>
<td>Σ 0..1</td>
<td>CodeableConcept</td>
<td>Supporting tests of human, viruses, and bacteria</td>
</tr>
<tr>
<td>observedAllele</td>
<td>Σ 0..1</td>
<td>string</td>
<td>Nucleotide(s)/amino acids from start position of sequence to stop position of observed sequence</td>
</tr>
<tr>
<td>referenceAllele</td>
<td>Σ 0..1</td>
<td>string</td>
<td>Nucleotide(s)/amino acids from start position of sequence to stop position of reference sequence</td>
</tr>
<tr>
<td>cigar</td>
<td>Σ 0..1</td>
<td>string</td>
<td>Extended CIGAR string for aligning the sequence with reference bases</td>
</tr>
<tr>
<td>quality</td>
<td>Σ 0..*</td>
<td>BackboneElement</td>
<td>Sequence Quality</td>
</tr>
<tr>
<td>start</td>
<td>Σ 0..1</td>
<td>integer</td>
<td>0-based start position (inclusive) of the sequence</td>
</tr>
<tr>
<td>end</td>
<td>Σ 0..1</td>
<td>integer</td>
<td>0-based end position (exclusive) of the sequence</td>
</tr>
<tr>
<td>score</td>
<td>Σ 0..1</td>
<td>integer</td>
<td>Quality score</td>
</tr>
<tr>
<td>platform</td>
<td>Σ 0..1</td>
<td>CodeableConcept</td>
<td>Platform</td>
</tr>
<tr>
<td>allelicState</td>
<td>Σ 0..1</td>
<td>CodeableConcept</td>
<td>The level of occurrence of a single DNA Sequence Variation within a set of chromosomes: Heteroplasmic / Homoplasmic / Homozygous / Heterozygous / Hemizygous</td>
</tr>
<tr>
<td>allelicFrequency</td>
<td>Σ 0..1</td>
<td>decimal</td>
<td>Allele frequencies</td>
</tr>
<tr>
<td>copyNumberEvent</td>
<td>Σ 0..1</td>
<td>CodeableConcept</td>
<td>Copy Number Event: Values: amplification / deletion / LOH</td>
</tr>
<tr>
<td>readCoverage</td>
<td>Σ 0..1</td>
<td>integer</td>
<td>Average number of reads representing a given nucleotide in the reconstructed sequence</td>
</tr>
<tr>
<td>chip</td>
<td>Σ 0..1</td>
<td>BackboneElement</td>
<td>Information of chip</td>
</tr>
<tr>
<td>chipId</td>
<td>Σ 0..1</td>
<td>string</td>
<td>Chip id</td>
</tr>
<tr>
<td>manufacturerId</td>
<td>Σ 0..1</td>
<td>string</td>
<td>Chip manufacturer id</td>
</tr>
<tr>
<td>version</td>
<td>Σ 0..1</td>
<td>string</td>
<td>Chip version</td>
</tr>
</tbody>
</table>
Sequence resource

External repository

Example: GA4GH full sequence links

**callSet**

1 sample

**Variant call**

genotype determination for 1 variant

**Variant info**

**Read group**

collections of reads produced by a sequencer

Search Reads:
Read group set ID, start, end

GA4GH API:

http://ga4gh.org/#/api/v0.5.1

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DiagnosticReport resource – ABCB4 gene mutation analysis

id: dg1

category: Laboratory test

code: ABCB4 gene mutation analysis

subject: Molecular Lab Patient ID: HOSP-23456

effective: 04/03/2014 8:30:00 AM

issued: 16/05/2014 10:28:00 AM

performer: Molecular Diagnostic Laboratory

specimen: Molecular Specimen ID: MLD45-Z4-1234
result:

- Genetic analysis master panel for ABCB4 -variant1. Generated Summary: id: od-1; Extensions: todo, Extensions: todo, Extensions: todo, status: final; ABCB4 gene mutation analysis (Details: {LOINC code '49874-1' = 'ABCB4 gene mutation analysis in Blood or Tissue by Molecular genetics method Narrative', given as 'ABCB4 gene mutation analysis'}); positive (Details: {http://hl7.org/fhir/v2/0078 code 'POS' = 'Positive'}

- Genetic analysis master panel for ABCB4 -variant2. Generated Summary: id: od-2; Extensions: todo, Extensions: todo, Extensions: todo, status: final; ABCB4 gene mutation analysis (Details: {LOINC code '49874-1' = 'ABCB4 gene mutation analysis in Blood or Tissue by Molecular genetics method Narrative', given as 'ABCB4 gene mutation analysis'}); positive (Details: {http://hl7.org/fhir/v2/0078 code 'POS' = 'Positive'}

- Genetic analysis master panel for ABCB4 -variant3. Generated Summary: id: od-3; Extensions: todo, Extensions: todo, Extensions: todo, status: final; ABCB4 gene mutation analysis (Details: {LOINC code '49874-1' = 'ABCB4 gene mutation analysis in Blood or Tissue by Molecular genetics method Narrative', given as 'ABCB4 gene mutation analysis'}); positive (Details: {http://hl7.org/fhir/v2/0078 code 'POS' = 'Positive'}

- Genetic analysis master panel for ABCB4 -variant4. Generated Summary: id: od-4; Extensions: todo, Extensions: todo, Extensions: todo, status: final; ABCB4 gene mutation analysis (Details: {LOINC code '49874-1' = 'ABCB4 gene mutation analysis in Blood or Tissue by Molecular genetics method Narrative', given as 'ABCB4 gene mutation analysis'}); positive (Details: {http://hl7.org/fhir/v2/0078 code 'POS' = 'Positive'}
Observation resource – from genetic analysis panel

id: ob-genetics-1
status: final
code: Genetic analysis master panel (Details: {LOINC code '55233-1' = 'Genetic analysis master panel - Blood or Tissue by Molecular genetics method', given as 'Genetic analysis master panel'})
subject: Molecular Lab Patient ID: HOSP-23456
issued: 11/03/2013 10:28:00 AM
performer: Molecular Diagnostic Laboratory
value: Positive (Details: {SNOMED CT code '10828004' = '10828004', given as 'Positive'})
specimen: Molecular Specimen ID: MLD45-Z4-1234

Extension:
Source: Somatic (Details: {http://hl7.org/fhir/LOINC-48002-0-answerlist code 'LA6684-0' = 'somatic', given as 'somatic'})
Sequence: Sequence Example
Gene: EGFR (Details: {http://www.genenames.org code ‘3236’ = ‘EGFR’, given as ‘EGFR’})
component

code: Genetic disease assessed (Details: {LOINC code '51967-8' = 'Genetic disease assessed [Identifier] in Blood or Tissue by Molecular genetics method', given as 'Genetic disease assessed'})

value: Lung cancer (Details: {SNOMED CT code '363358000' = '??', given as 'Malignant tumor of lung (disorder)'}))

component

code: Genetic disease sequence variation interpretation (Details: {LOINC code '53037-8' = 'Genetic disease sequence variation interpretation [interpretation] in Blood or Tissue by Molecular genetics method', given as 'Genetic disease sequence variation interpretation'})

value: Pathogenic (Details: {[not stated] code 'LA6669-1' = '??', given as 'Pathogenic'})
Sequence resource – EGFR mutation

Generated Narrative with Details

id: example

type: DNA


referenceSeq: ENSEST00000085772.1 (Details: {http://www.ensembl.org code 'ENSEST00000085772.1' = '??})

Coordinates

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species: Homo sapiens (Details: {SNOMED CT code '337915000' = '??})

observedAllele: T

referenceAllele: A

Repositories

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Useful Links

FHIR Clinical Genomics (DSTU 2.0)
http://hl7.org/fhir/observation-genetics-cg-prf-1a.html

GA4GH API
http://ga4gh.org/#/api/v0.5.1

JAMIA Paper on SMART Genomics/FHIR DSTU 2.0
http://jamia.oxfordjournals.org/content/early/2015/07/21/jamia.ocv045.long

SMART Genomics (direct link)
http://projects.iq.harvard.edu/smartgenomics/home

SMART Platform
http://smarthealthit.org/