Phenotyping Library & Portal
– Preliminary Ideas
Nov. 14th, 2011

Jyoti Pathak
Motivation

- SHARPn, eMERGE, i2b2 and other projects are doing “phenotyping” (broadly speaking)
- This is resulting in the creation of a large number of robust, validated algorithms
- Most of them are represented in MS Word files, JPEG pictures
- Currently, there is no designated “home” for the algorithms
  - https://www.gwas.net/Library_of_Phenotype_Algorithms
Objectives

- We want to create such a “home”…
  - “PhenoPortal”?
- Publicly accessible portal and library
- Standards-based representation of the algorithm criteria
- Anchored to CEMs
- Machine executable
An example...

- NCBO BioPortal
- http://bioportal.bioontology.org
- Next few slides...
SNOMED Clinical Terms

Details

ONTOLGY ID: 1353
BIOPORTAL URL: http://prl.bioontology.org/ontology/SNOMEDCT
STATUS: UMLS
CATEGORIES: Health
GROUPS: Cancer Biomedical Informatics Grid
Unified Medical Language System
WHO Family of International Classifications
CONTACT: Vivian A. Stall, stall@nlm.nih.gov
HOME PAGE: https://biodo.org
PUBLICATIONS PAGE: https://biodo.org
DOCUMENTATION PAGE: https://biodo.org
DESCRIPTION: SNOMED Clinical Terms

Metrics

NUMBER OF CLASSES: 393075
NUMBER OF INDIVIDUALS: 0
NUMBER OF PROPERTIES: 42
MAXIMUM DEPTH: 32
MAXIMUM NUMBER OF SIBLINGS: 10010
AVERAGE NUMBER OF SIBLINGS: 1
CLASSES WITH A SINGLE SUBCLASS: 28764
CLASSES WITH MORE THAN 25 SUBCLASSES: 2034
CLASSES WITH NO DEFINITION: 392535

Reviews

Add your review

REVIEW BY VCDE

No ratings submitted with this review. The review group found that SNOMED CT has fully, or in part, satisfied all of the review criteria except one. The group recommended that SNOMED CT be "certified" as an NC1 standard vocabulary. The group further recommended that usage and implementation be further defined and that the term be "certified" as an NC1 standard vocabulary. The group will provide recommendations for further enhancing the vocabulary evaluation instrument, especially as a number of ontologies are on the vocabulary standardization roadmap. See https://cabi.ncl.mgh.gov/workspaces/VCDE/Meetings/VCDE_Workspace_Meetings/Oct2_VCDE_Meeting for more details on the SNOMED CT Review.

Versions

<table>
<thead>
<tr>
<th>VERSION</th>
<th>RELEASE DATE</th>
<th>DOWNLOADS</th>
</tr>
</thead>
<tbody>
<tr>
<td>2011_01_31</td>
<td>01/31/2011</td>
<td></td>
</tr>
<tr>
<td>2010_07_31</td>
<td>07/31/2010</td>
<td></td>
</tr>
<tr>
<td>2010_01_31</td>
<td>01/31/2010</td>
<td></td>
</tr>
<tr>
<td>2009_07_31</td>
<td>07/31/2009</td>
<td></td>
</tr>
<tr>
<td>2009_01_31</td>
<td>01/31/2009</td>
<td></td>
</tr>
</tbody>
</table>
SNOMED Clinical Terms

Physical anatomical entity
- Anatomical structure
- Body structure
- Body organ structure
- Body region structure
- Body space structure
- Body system structure
- Body tissue structure
- Body wall structure
- Cell structure
- Developmental body structure
- Entire anatomical structure
- Human body structure
- Intracellular anatomical structure
- Non-human body structure
- Sex structure
- Structure of multiple topographic sites
- Structure of product of conception
- Transplant
- Group of anatomical entities
- Topography assigned
- Topography not assigned
- Topography unknown
- Clinical finding
- Environment or geographical location
- Event
- Linkage concept
- Observable entity

The National Center for Biomedical Ontology is one of the National Centers for Biomedical Computing supported by the NIH, the NHLBI, and the NIH Common Fund under grant U54-HG008402.
Copyright © 2009-2011. The Board of Trustees of Leland Stanford Junior University. All rights reserved.
NCBO Website  Release Notes  Terms of Use  Privacy Policy
Core Requirements

- Search and visualization
  - Ability to do keyword-based searches for available algorithms
  - Navigate a hierarchy of phenotypes
  - Visualize the algorithm logic flow
  - Download human readable version (MS word)
Core Requirements

- Representation
  - XML-based structured representation using NQF Quality Data Model (QDM)
  - Rules-based executable representation using JBoss Drools
    - Requires a patient fact model (see later slide)
    - Mapping to CEMs (see later slide)
  - QDM -> Drools translator
    - Intermountain has investigated Arden ML to Drools
Core Requirements

- **Authoring**
  - We anticipate that the algorithm authoring will primarily happen at the level of QDM
    - Very few people would be interested in Drools authoring (e.g., using JBoss Guvnor)
  - Need to investigate NQF Measure Authoring Toolkit (MAT) more closely
What is the Measure Authoring Tool?

• Web-based application that allows you to create **eMeasures** without writing XML

• An eMeasure is a quality measure in a standardized XML file that can be read by both humans and computer systems

• **Target Audience**
  – Measure developers will be primary users.
  – Quality and health IT stakeholders (e.g., providers, health IT vendors, QIO) will also benefit with better access to, capture of, and reporting on comparable data.
Key Features and Functions

• Create and share eMeasures and their corresponding code lists with other users

• Create and reuse standard value sets (known as Code Lists in the MAT) and other measure components limiting rework as new measures are developed

• Use the Quality Data Model (QDM) as the grammar and structure to fully define and express eMeasures in a standard way

• Export eMeasures in an EHR readable format to enable collection of comparable healthcare quality data
**Sample Measure**

Percentage of patients aged 18 years and older with a diagnosis of CAD who were prescribed a lipid-lowering therapy

| Initial Patient Population | Patients aged 18 years and older before the start of the measurement period.  
|                           | Patients that have a documented diagnosis of coronary artery disease before or simultaneously to encounter date  
<p>|                           | Patients who have at least 2 outpatient or nurse facility encounters during the measurement period |
| Denominator               | Patients aged 18 years and older with a diagnosis of coronary artery disease |
| Numerator                 | Patients who were prescribed lipid-lowering therapy |
| Exclusions                | Patients who have documentation of a medical, system or patient reason for not prescribed lipid lowering therapy |</p>
<table>
<thead>
<tr>
<th>Patients who...</th>
<th>What <em>kind</em> of data are we dealing with?</th>
<th>What <em>about</em> the data?</th>
<th>How do we define this data?</th>
</tr>
</thead>
<tbody>
<tr>
<td>...Are diagnosed with Coronary Artery Disease</td>
<td>Diagnosis</td>
<td>Active</td>
<td>ICD-9, ICD-10, SNOMED-CT</td>
</tr>
<tr>
<td>...Were prescribed Lipid-lowering Therapy</td>
<td>Medication</td>
<td>Administered Order Dispensed Active</td>
<td>RxNorm</td>
</tr>
<tr>
<td>...Have had at least two encounters during the measurement period</td>
<td>Encounters</td>
<td></td>
<td>CPT</td>
</tr>
<tr>
<td>...Are at least 18 years old or older</td>
<td>Patient Characteristic</td>
<td></td>
<td>LOINC</td>
</tr>
</tbody>
</table>
What’s next?

• Create Code lists
  – select QDM Category

• Apply Code list to Measure
  – select QDM Data Type

• Build phrases using QDM Elements
  – Add QDM attributes

• Add phrases to Clauses
<table>
<thead>
<tr>
<th>Name</th>
<th>Steward</th>
<th>Category</th>
<th>Code System</th>
<th>History</th>
</tr>
</thead>
<tbody>
<tr>
<td>Asthma Daytime Symptoms</td>
<td>National Quality Forum</td>
<td>Symptom</td>
<td>SNOMED-CT</td>
<td></td>
</tr>
<tr>
<td>Asthma Daytime Symptoms Quantified</td>
<td>National Quality Forum</td>
<td>Symptom</td>
<td>SNOMED-CT</td>
<td></td>
</tr>
<tr>
<td>Asthma Diagnosis</td>
<td>National Quality Forum</td>
<td>Condition/Diagnosis/Problem</td>
<td>Grouping</td>
<td></td>
</tr>
<tr>
<td>Asthma ICD-10</td>
<td>National Quality Forum</td>
<td>Condition/Diagnosis/Problem</td>
<td>ICD-10</td>
<td></td>
</tr>
<tr>
<td>Asthma ICD-9</td>
<td>National Quality Forum</td>
<td>Condition/Diagnosis/Problem</td>
<td>ICD-9</td>
<td></td>
</tr>
<tr>
<td>Asthma Nighttime Symptoms</td>
<td>National Quality Forum</td>
<td>Symptom</td>
<td>SNOMED-CT</td>
<td></td>
</tr>
<tr>
<td>Asthma Nighttime Symptoms Quantified</td>
<td>National Quality Forum</td>
<td>Symptom</td>
<td>SNOMED-CT</td>
<td></td>
</tr>
<tr>
<td>Asthma Symptom Assessment Tool</td>
<td>National Quality Forum</td>
<td>Risk Category/Assessment</td>
<td>SNOMED-CT</td>
<td></td>
</tr>
<tr>
<td>CAD includes MI</td>
<td>National Quality Forum</td>
<td>Condition/Diagnosis/Problem</td>
<td>Grouping</td>
<td></td>
</tr>
<tr>
<td>CAD includes MI ICD-10</td>
<td>National Quality Forum</td>
<td>Condition/Diagnosis/Problem</td>
<td>ICD-10</td>
<td></td>
</tr>
</tbody>
</table>
Select QDM Data Type
Add QDM Attribute to a Phrase

Measure Authoring Tool

My CAD Measure

Code Lists

Search for a Code List

Lipid Lowering Therapy: Medication, Order

ADD/EDIT Attributes for:
Lipid Lowering Therapy: Medication, Order

ATTRIBUTE TYPE TERM
negation rationale Code List Patient reason: attribute
cumulative medication duration

Save Cancel

Go to Code List Workspace

Go to Measure Details >>
Export the eMeasure

Measure Authoring Tool

My Measures > Export

Measure: My CAD Measure

Select an export option

- eMeasure
- Measure Code List
- eMeasure Package

Save Open Cancel
## My CAD Measure

<table>
<thead>
<tr>
<th>eMeasure Name</th>
<th>eMeasure Id</th>
<th>Version number</th>
<th>eMeasure Set Id</th>
<th>Available Date</th>
<th>Measurement Period</th>
</tr>
</thead>
<tbody>
<tr>
<td>My CAD Measure</td>
<td>7cb18f6f-6ae8-4dc1-8d41-98e03557fb89</td>
<td>1</td>
<td>pending</td>
<td>October 6, 2011</td>
<td>October 6, 2011 through October 6, 2012</td>
</tr>
<tr>
<td>Measure Steward</td>
<td>National Quality Forum</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Endorsed by</td>
<td>None</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Description</td>
<td>sample measure</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Copyright</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Measure scoring</td>
<td>Proportion</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Measure type</td>
<td>Process</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Stratification</td>
<td>none</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Risk Adjustment</td>
<td>None</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Data Aggregation</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Rationale</td>
<td>n/a</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Improvement notation</td>
<td>No information</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Measurement duration</td>
<td>1 year(s)</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Definition</td>
<td>No information</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Guidance</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

**Table of Contents**
Table of Contents

- Population criteria
- Data criteria (QDM Data Elements)
- Supplemental Data Elements

Population criteria

- Initial Patient Population =
  - AND: "Patient Characteristic: birth date" >= 18 year(s) starts before start of "Measurement Period"
  - AND: Count >= 2 or:
    - OR: "Encounter: Nursing Facility Encounter"
    - OR: "Encounter: Outpatient Encounter"
    - during "Measurement Period"
  - AND: "Diagnosis, Active: CAD includes MI"
- Denominator =
  - AND: "Initial Patient Population"
- Denominator Exclusions =
  - AND:
    - OR: "Medication, Order not done: Patient reason" for "Lipid Lowering Therapy RxNorm Code List"
    - OR: "Medication, Order not done: System Reason" for "Lipid Lowering Therapy RxNorm Code List"
- Numerator =
  - AND:
    - OR: "Medication, Active: Lipid Lowering Therapy"
    - OR: "Medication, Order: Lipid Lowering Therapy"
    - during "Measurement Period"
- Denominator Exceptions =
  - None

Data criteria (QDM Data Elements)

- "Diagnosis, Active: CAD includes MI" using "CAD includes MI Grouping Code List (2.16.840.1.113883.3.560.4.13.25)"
- "Encounter: Nursing Facility Encounter" using "Nursing Facility Encounter CPT Code List (2.16.840.1.113883.3.560.4.13.26)"
- "Encounter: Outpatient Encounter" using "Outpatient Encounter CPT Code List (2.16.840.1.113883.3.560.4.13.27)"
- "Medication, Active: Lipid Lowering Therapy" using "Lipid Lowering Therapy RxNorm Code List (2.16.840.1.113883.3.560.4.13.29)"
- "Medication, Order: Lipid Lowering Therapy" using "Lipid Lowering Therapy RxNorm Code List (2.16.840.1.113883.3.560.4.13.29)"
- "Medication, Order not done: Patient reason" using "Patient reason SNOMED-CT Code List (2.16.840.1.113883.3.560.4.13.47)"
- "Medication, Order not done: System Reason" using "System Reason SNOMED-CT Code List (2.16.840.1.113883.3.560.4.13.46)"
- "Patient Characteristic: birth date" using "birth date LOINC Code List (2.16.840.1.113883.3.560.100.4)"

Supplemental Data Elements

- "Patient Characteristic: Gender" using "Gender HL7 Code List (2.16.840.1.113883.1.11.1)"
- "Patient Characteristic: Race" using "Race CDC Code List (2.16.840.1.114222.4.11.836)"
- "Patient Characteristic: Ethnicity" using "Ethnicity CDC Code List (2.16.840.1.114222.4.11.837)"
- "Patient Characteristic: Payer" using "Payer Source of Payment Typology Code List (2.16.840.1.113883.3.221.5)"
Core Requirements

- Execution
  - A visual interface for algorithm execution
    - No one is going to fire up Eclipse and JBoss
  - PopHealth (http://projectpophealth.org) has an interesting model
    - Download a virtual machine with the entire set of queries/algorithms
    - Load institution-specific patient data into the PopHealth database
    - Execute the algorithms; visual GUI
An Open Source Quality Measure Reference Implementation

- Empowers healthcare providers to perform Meaningful Use quality measure reporting
- Promotes easier submission of quality measures to public health organizations

What is popHealth?

popHealth is an open source reference implementation software service that automates the reporting of Meaningful Use quality measures. popHealth integrates with a healthcare provider’s electronic health record (EHR) system using continuity of care records. popHealth streamlines the automated generation of summary quality measure reports on the provider’s patient population.

Who are the intended users?

popHealth supports healthcare providers and public health organizations.

Project Milestones

- **25 Feb 10**
  - popHealth Prototype Released

Example popHealth Dashboard Report

<table>
<thead>
<tr>
<th>Reporting Period:</th>
<th>Jul 01, 2010 - Sep 30, 2010</th>
</tr>
</thead>
<tbody>
<tr>
<td>Reporting Patients:</td>
<td>11,980</td>
</tr>
<tr>
<td>Hypertension: Blood Pressure Measurement</td>
<td>81% (5151) / 6350</td>
</tr>
<tr>
<td>Pneumonia Vaccination Status for Older Adults</td>
<td>56% (3354) / 5991</td>
</tr>
</tbody>
</table>
pop health

Concept of Operations

Software Runs Inside Providers Network

Focus is Quality Reporting

- Quality Report
- Public Health Case Report
- Immunization and Response Management
Childhood Immunization Status

07/01/2010 - 09/30/2010

The percentage of children 2 years of age who had four diphtheria, tetanus and acellular pertussis (DTaP); three polio (IPV); one measles, mumps and rubella (MMR); two H influenza type B (Hib); three hepatitis B (Hep B), one chicken pox (VZV); four pneumococcal conjugate (PCV); two hepatitis A (Hep A); two or three rotavirus (RV); and two influenza (flu) vaccines by their second birthday. The measure calculates a rate for each vaccine and two separate combination rates.

**Denominator (153)**
- **Patient characteristic** gender male
- **Patient characteristic** gender female
- **Patient characteristic** age between 1 and 2
- **Encounter** outpatient with pcp and obgyn

**Numerator (126)**
- **Medication administered** >= 1 count measles, mumps and rubella (MMR) vaccine prior to 2 years old
- **Or**
- **Diagnosis** mumps resolved
- **Or**
- **Allergy** mumps vaccine allergy
- **Or**
- **Medication administered** >= 1 count mumps vaccine prior to 2 years old
- **Or**

**Exclusions (0)**
## Community Health

**Reporting Period:** 07/01/2010 - 09/30/2010

**Reporting Patients:** 11,980

### CORE

**Hypertension: Blood Pressure Measurement**
- Percentage: 81%
- Number of Patients: 5151 / 6350

**Diabetes**
- **Diabetic Neuropathy**
  - Percentage: 21%
  - Number of Patients: 2276
- **Diabetes Eye Exam**
  - Percentage: 16%
  - Number of Patients: 2276
- **Diabetes HbA1c Control < 8%**
  - Percentage: 63%
  - Number of Patients: 2276

### CANCER

**Breast Cancer Screening**
- Percentage: 10%
- Number of Patients: 360 / 3594

### MISCELLANEOUS

**Pneumonia Vaccination Status for Older Adults**
- Percentage: 56%
- Number of Patients: 3354 / 5991

### WOMEN'S HEALTH

**Prenatal Care: Screening for Human Immunodeficiency Virus (HIV)**
- Percentage: 33%
- Number of Patients: 98 / 290
Supplemental Requirements

☐ Ability for users to provide feedback and comments on the algorithms
  – Wiki-style?

☐ Training and educational material

☐ Others?
Dependencies

- Availability of CEMs
- Representation (stable?) of the CEM MySQL database
  - Our queries will be performed against CEMDB
- Drools patient fact model
  - Intermountain colleagues exploring HL7 vMR
- QDM to Drools translator

SHARPn.org

Strategic Health IT Advanced Research Projects (SHARP) Program
Awarded by The Office of the National Coordinator for Health Information Technology
TODO List

- Design PhenoPortal architecture
  - Need your help
- Understand QDM
  - One of the most important aspects!
- Access to NQF Measure Authoring Tool