The Phenotype Execution and Modeling Architecture (PhEMA) – A Standards-Based Composition of Software for Phenotype Algorithm Development

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Disclosure

• No relevant financial interests to disclose
Background

• EHR data for secondary use
• Portability of EHR-based phenotypes
• Approaches to define phenotypes
  – Rasmussen et al., JBI 2014.
• Desiderata for computable representations
  – Mo et al., JAMIA 2015.
• Multiple tools exist
  – 24 reviewed in Xu et al., JAMIA 2015.
Phenotype Execution and Modeling Architecture (PhEMA)

- Authoring
- Library (QDM, HL7 HQMF)
- Validation (QDM, HL7 HQMF)
- Execution (QDM, HL7 HQMF)
- Clinical Data Repository
- Terminology Services (CTS2, VSAC)
- Data Model Services (W3C, HL7, CIMI, ISO11179, QDM)

Rasmussen et al., AMIA Jt Summits Transl Sci, 2015
Development Methodology

• Evaluate existing systems
• Leverage standards as a bridge
• Develop new solutions where needed
• Support multiple configurations
PhEMA Software
Local Value Sets

- **CTS2 Value Set Definition Service**
- Enhanced import process
- Use in conjunction with VSMC

```json
{
  "valueSetCatalogEntryDirectory": {
    "entryList": [
      {
        "valueSetName": "2.16.840.1.113883.3.464.1003.103.11.1001",
        "currentDefinition": {
          "valueSetDefinition": {
            "content": "20150430",
            "uri": "urn:oid:2.16.840.1.113883.3.464.1003.103.11.1001/version/20150430",
            "href": "http://ip-172-30-2-43:8080/vsmc/valueset/2.16.840.1.113883.3.464.1003.103.11.1001/definition/20150430"
          }
        },
        "valueSet": {
          "content": "2.16.840.1.113883.3.464.1003.103.11.1001",
          "uri": "urn:oid:2.16.840.1.113883.3.464.1003.103.11.1001",
          "href": "http://ip-172-30-2-43:8080/vsmc/valueset/2.16.840.1.113883.3.464.1003.103.11.1001"
        },
        "about": "urn:oid:2.16.840.1.113883.3.464.1003.103.11.1001",
        "formalName": "Diabetes",
        "resourceSynopsis": {
          "value": "Diabetes"
        },
        "resourceName": "2.16.840.1.113883.3.464.1003.103.11.1001"
      }
    ]
  }
}
```
Data Element Repository

• Define elements, attributes, constraints
• Multiple element definitions
  – Quality Data Model (QDM)
  – Fast Healthcare Interoperability Resources (FHIR)
• Allows testing proposed extensions to models
• RDF-based representation
• Accessible via SPARQL & REST (XML & JSON)
Phenotype Authoring Tool
Define the Diagnosis, Active

Value set: Diabetes

Anatomical Location Site

Laterality

Left

Provider Preference

Ordinality

Severity

Negation Rationale

Patient Preference

Health Record Field

Source

Recorder

Reference this element later

Only include items that occur between

Start

End

mm/dd/yyyy

mm/dd/yyyy

OK

Cancel
Phenotype Authoring Tool

Search CTS2 repositories

Elements loaded from DER

Compose definition from elements, and save as HQMF
Library

• PhEMA Library
  – Lightweight storage option

• Phenotype Knowledgebase (PheKB)
  – Integrated authentication
  – Collaboration & discovery
PheKB Integration

Statin Dose Response

Phenotype #: 116
Status:
  Testing
  Do Not List on the Collaboration Phenotypes List

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Contact Author: christian.shaffer

Suggested Citation
Christian Shaffer
http://local.phenotypes.org

Statin Dose Response

GDM Data Elements
  Value Sets
  Logical Operators
  Temporal Operators
  Subset Operators
  Existing Phenotypes

Patient Care Experience
  Drag and drop clinical terms or value sets here, or click to search

Provider Care Experience
  Drag and drop clinical terms or value sets here, or click to search

Patient Care Experience
  Drag and drop clinical terms or value sets here, or click to search

Care Goal
  Drag and drop clinical terms or value sets here, or click to search

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Getting Started
  - Check out the Quick Start guide
  - Try a tutorial
  - Create your first phenotype

Sign in with your PheKB.org account

Please enter your login details

E-mail: peter.spitz@vanderbilt. Password: ********

Sign in Clear Cancel

Dashboard Create Phenotype Help
Execution

eMeasures to KNIME (eM2Kn)

![Diagram of eMeasures to KNIME (eM2Kn)](image)

Institutional Repository

eMeasures to i2b2 / KNIME (eM2iKn)

![Diagram of eMeasures to i2b2 / KNIME (eM2iKn)](image)

i2b2
Configuration – Pre-Packaged VM

- PhEMA Library
- Local Value Sets
- PhEMA Authoring Tool
- Data Element Repository
- QDM to KNIME
- Custom Data Schema
- i2b2
Configuration – BPH Use Case

Value Set Authority Center

VSMC Wrapper

Local Value Sets

Data Element Repository

PhENOA Authoring Tool

Phenotype KnowledgeBase

KNIME

QDM to i2b2 / KNIME

I2b2 Hive

i2b2 Local Value Sets
Conclusions & Future Work

• Validated proposed architecture
  – Accuracy of representation
  – Modularity / configurability

• Ability to represent NLP in phenotypes

• Ongoing collaborations
  – Complementary projects: i2b2, OHDSI
  – S&I Framework (Clinical Query Language)
  – Clinical Quality Measures (popHealth, Bonnie)
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Thank You!

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