Presentation Overview

- Introduction
- SNOMED CT Features
- Data Analytics
  - Preparing Data for Analytics
  - SNOMED CT Techniques
  - Record Query Techniques
  - Analytics Tasks
  - Challenges
- Clinical Decision Support
  - Overview
  - Logical Architecture
  - Knowledge Base
  - Communications

Data Analytics and Clinical Decision Support with SNOMED CT

Linda Bird and Jon Zammit
IHTSDO
Introduction

Data Analytics

*Discovery & communication of meaningful patterns in data*

- May describe, predict and improve business performance
- May recommend action or guide decision making

**Scope:**
- Individual patient
- Individual healthcare worker
- Patient groups / cohorts
- Enterprise groups
- Geographic groups

**Substrate:**
- Unstructured free text documents
- Structured documents using SNOMED CT
- Structured documents using other coding systems
- ‘Big data’
Analytics Purposes - Overview

Benefit Individuals
Patients and Clinicians

CLINICAL ASSESSMENT AND TREATMENT

Evidence-Based Healthcare

RESEARCH
(Clinical knowledge)

POPULATION MONITORING

Benefit Populations

Analytics Purposes – Individual Care

- SNOMED CT may be used to support analytics that
  - Improves care for individuals by enabling
    - Retrieval and sharing of information to better support care
    - Reduction in duplication of investigations and interventions
    - Integration with decision support tools to guide care
    - Context sensitive presentation of guidelines and care pathways
    - Identification of patients requiring follow-up or treatment changes
    - Professional logs and performance tracking
    - Work list generation and workload monitoring
Analytics Purposes – Population Care

- SNOMED CT may be used to support analytics that
  - Improves the care of populations by enabling
    - Epidemiological monitoring and reporting
    - Audit of clinical care and service delivery
    - Systems that measure and maximize the delivery of cost-effective treatments and minimize the risk of costly errors

Analytics Purposes – Evidence Based Healthcare

- SNOMED CT may be used to support analytics that
  - Supports evidence-based healthcare and clinical knowledge research by enabling
    - Identification of clinical trial candidates
    - Research into the effectiveness of different approaches to disease management
    - Clinical care delivery planning
    - Planning for future service delivery provision
SNOMED CT Features

SNOMED CT Core Features

- Concepts
  - Enable meaning-based queries
- Descriptions
  - Assist searching for concepts
  - Enhance string-matching in NLP
  - Multi-lingual support
- Relationships
  - Support queries based on defined meaning
  - Aggregation
  - Query detailed content stored in EHRs using more abstract concepts
SNOMED CT Additional Features

- Concept Model
  - Provides foundation for processing clinical meaning
- Expressions
  - Enable meaning-based queries over more than just concepts
- Reference sets
  - Represent subsets of concepts to help define query criteria
  - Represent non-standard aggregations for specific use cases
  - Define maps from other code systems to SNOMED CT
  - Define sets of language or dialect specific descriptions
- Description Logic
  - Supports computation of subsumption and equivalence

SNOMED CT Other Benefits for Analytics

- Broad domain coverage
  - Enables queries across disciplines, specialties and domains
- Robust versioning
  - Helps to manage queries over longitudinal health records
- International
  - Enables queries, subsets, rules and maps to be shared and reused between countries
- Localization mechanisms
  - Allows queries to be applied to data from different countries, dialects, regions & applications
Preparing Data for Analytics

1. Natural Language Processing
   - Enables a computer program to analyse and extract meaning from human language
   - Automatic coding of free text is not always reliable
   - Requires manual validation of automatic coding
   - Context that is not coded can lead to incorrect query results

2. Mapping Other Code Systems to SNOMED CT
   - SNOMED CT can be used as a common reference terminology for querying over data sources that use different coding systems
   - Direction and correlation of map effect the quality of analytics

3. Mapping SNOMED CT to statistical classification
   - Useful when aggregation of codes into a single category is required
SNOMED CT Analytics Techniques

- Subsets
- Subsumption
- Defining relationships
- Description logic over terminology
- Description logic over terminology and structure
Subsets - Overview

- Create subsets of concepts for a specific clinical purpose
  - Manual inclusion using search and browse
  - Using an existing subset as a starting point
  - Lexical queries (string matching) to identify candidates
  - Hierarchical queries to select descendants of a concept
  - Attribute queries to find concepts with a specific attribute value
  - SNOMED CT queries using a combination of features

- Subsets may be defined:
  - Extensionally
    - Flat list of concept identifiers
    - Distributed using a simple or ordered refset
  - Intensionally
    - Using a machine processable query
    - Distributed using a query refset

- Test the codes in patient records for membership

---

Subsets – Extensional Example

**Patient Record**
- Patient id: 1755
- Diagnosis: 38115001 | Tuberculosis of spinal meninges|

**Subset**

<table>
<thead>
<tr>
<th>Concept ID</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>56717001</td>
<td>tuberculosis (disorder)</td>
</tr>
<tr>
<td>58437007</td>
<td>tuberculosis of meninges (disorder)</td>
</tr>
<tr>
<td>90302003</td>
<td>tuberculosis of cerebral meninges (disorder)</td>
</tr>
<tr>
<td>38115001</td>
<td>tuberculosis of spinal meninges (disorder)</td>
</tr>
<tr>
<td>447332005</td>
<td>tuberculous abscess of epidural space (disorder)</td>
</tr>
<tr>
<td>11676005</td>
<td>tuberculous leptomeningitis (disorder)</td>
</tr>
<tr>
<td>447253004</td>
<td>tuberculous arachnoiditis (disorder)</td>
</tr>
<tr>
<td>31112008</td>
<td>tuberculous meningoesophagealitis (disorder)</td>
</tr>
</tbody>
</table>
Subsets – Intensional Example

Patient Record
• Patient id: 1755
• Diagnosis: 38115001 \(\text{Tuberculosis of spinal meninges}\)

Subset
• Definition: << 56717001 \(\text{Tuberculosis}\)
• Expansion:
  - Tuberculosis of central nervous system (disorder)
  - Tuberculosis of brain (disorder)
  - Tuberculosis of meninges (disorder)
  - Tuberculosis of cerebral meninges (disorder)
  - Tuberculosis of spinal meninges (disorder)
  - Tuberculous leptomeningitis (disorder)
  - Tuberculous encephalitis or myelitis (disorder)
  - Tuberculous myelitis (disorder)
  - Tuberculosis of ear (disorder)

Subsumption - Overview

- Subsumption occurs when one clinical meaning is a subtype of another clinical meaning
  - Example: Which patients have an infectious disease?
    - Involves finding all patients with any kind of infectious disease including 75570004 \(\text{Viral pneumonia}\)

- Using the SNOMED CT Expression Constraint Language
  - Uses ‘<’ (descendantOf) and ‘<<’ (descendantOrSelfOf)
  - Example
    - << 40733004 \(\text{Infectious disease}\)

- Techniques for testing subsumption include
  - Precomputed transitive closure table
  - Using a Description Logic reasoner
Subsumption - Example

Hospital Audit for Patients with Infectious Diseases

```
SELECT * FROM health_records
WHERE diagnosis =
  (<< 40733004 |Infectious disease|)
```

<table>
<thead>
<tr>
<th>patientID</th>
<th>Diagnosis</th>
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</thead>
<tbody>
<tr>
<td>634711</td>
<td>71620000 [Fracture of femur]</td>
</tr>
<tr>
<td>634711</td>
<td>40468003 [Hepatitis A]</td>
</tr>
<tr>
<td>634711</td>
<td>66308002 [Fracture of humerus]</td>
</tr>
<tr>
<td>158775</td>
<td>415353009 [Rotavirus food poisoning]</td>
</tr>
<tr>
<td>889125</td>
<td>75570004 [Viral pneumonia]</td>
</tr>
<tr>
<td>456872</td>
<td>22298006 [Myocardial infarction]</td>
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<tr>
<td>456872</td>
<td>195967001 [Asthma]</td>
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Subsumption - Example

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<td>75570004</td>
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</table>
Defining Relationships - Overview

- Represent a characteristic of the meaning of a concept
- More than 50 attributes, including:
  - 363698007 [Finding site]
  - 116676008 [Associated morphology]
  - 246075003 [Causative agent]
  - 363704007 [Procedure site]
  - 260686004 [Method]
  - 272741003 [Laterality]
- SNOMED CT Concept Model provides rules for how these attributes can be used
- Implementation approaches include
  - Using the distributed Relationships file
  - Comparing normal form expressions
  - Using a Description Logic Reasoner

Defining Relationships – Example
Defining Relationships – Example

SNOMED CT Expression Constraint Language

\[
< 404684003 | \text{Clinical finding} | : 116676008 | \text{Associated morphology} | = << 3898006 | \text{Benign neoplasm} | \text{AND} 363698007 | \text{Finding site} | = << 64033007 | \text{Kidney structure} >
\]

<table>
<thead>
<tr>
<th>Concept ID</th>
<th>Preferred Term</th>
</tr>
</thead>
<tbody>
<tr>
<td>254925008</td>
<td>Benign tumor of renal calyx</td>
</tr>
<tr>
<td>254919009</td>
<td>Cortical adenoma of kidney</td>
</tr>
<tr>
<td>269489006</td>
<td>Benign tumor of renal parenchyma</td>
</tr>
<tr>
<td>254920003</td>
<td>Cystadenoma of kidney</td>
</tr>
<tr>
<td>254922006</td>
<td>Oncocytoma of kidney</td>
</tr>
<tr>
<td>276866009</td>
<td>Benign tumor of pelviureteric junction</td>
</tr>
<tr>
<td>254927000</td>
<td>Benign papilloma of renal pelvis</td>
</tr>
<tr>
<td>92319008</td>
<td>Benign neoplasm of renal pelvis</td>
</tr>
<tr>
<td>307618001</td>
<td>Juxtaglomerular tumor</td>
</tr>
<tr>
<td>254923001</td>
<td>Hemangiopericytoma of kidney</td>
</tr>
<tr>
<td>254921004</td>
<td>Angiomyolipoma of kidney</td>
</tr>
<tr>
<td>92165001</td>
<td>Benign neoplasm of kidney</td>
</tr>
</tbody>
</table>

Description Logic over Terminology - Overview

- SNOMED CT semantics are based on Description Logic
- This enables
  - The automation of reasoning across SNOMED CT
  - The implementation of more powerful analytics operations
    - Testing subsumption and equivalence
    - Testing defining attribute values
    - Property chaining
    - Advanced reasoning (concrete values and GCIs)
- Implementation
  - Translate SNOMED CT into OWL 2
  - Use Perl transform script
  - Load OWL files into a DL enabled service
  - Use DL reasoner – e.g. FACT++, ELK, Snorocket
  - Semantic query languages – e.g. SPARQL, DL Query
Description Logic over Terminology - Example

- Find all disorders that are associated with the organism ‘streptococcus pyogenes’
- Use property chain rule:
  47429007 |Associated with| o 47429007 |Associated with|  
  ➔ 47429007 |Associated with|

Disorder

- 81077008 Acute rheumatic arthritis
- 58718002 Rheumatic fever
- 3029008 Streptococcus pyogenes infection
- 4073004 Infectious disease

Organism

- 80166006 Streptococcus pyogenes

Description Logic over Terminology and Structure

- The information model and the terminology each captures part of the semantics in a patient record
- The same semantics can be represented using different information structures and terminology values
- Description logic may be used to achieve a canonical representation of the meaning in both representations
- Uses ‘expression templates’ for each information model to capture the DL pattern expressed
### Description Logic over Terminology and Structure

**Family history**

- **Problem (coded text)**: 56265001 heart disease
- **Relationship (coded text)**: 72705000 mother

**Clinical history**

- **Problem (coded text)**: 275120007 family history: cardiac disorder

---

Example logic expressions:

- **416471007**: Family history of clinical finding
- **246090004**: Associated finding = 56265001 Heart disease,
- **408732007**: Subject relationship context = 444301002 Mother of subject,
- **408731000**: Temporal context = 410511007 Current or past (actual),
- **408729009**: Finding context = 410515003 Known present
Description Logic over Terminology and Structure

275120007 [Family history: cardiac disorder]

Clinical history

Problem (coded text)

275120007 [family history: cardiac disorder]

Description Logic over Terminology and Structure

416471007 [Family history of clinical finding]
246090004 [Associated finding] = 56265001 [Heart disease],
408732007 [Subject relationship context] =
   444301002 [Mother of subject],
408731000 [Temporal context] = 410511007 [Current or past (actual)],
408729009 [Finding context] = 410515003 [Known present]

subtype of

275120007 [Family history: cardiac disorder]
Record Query Techniques

SNOMED CT Languages

- **Compositional Grammar**
  24136001 | Hip joint structure | :
  272741003 | Laterality | = 7771000 | Left |

- **Expression Constraint Language**
  << 404684003 | Clinical finding | :
  363698007 | Finding site | = << 39057004 | Pulmonary valve structure | ,
  116676008 | Associated morphology | = << 415582006 | Stenosis |

- **Expression Template Language**
  24136001 | Hip joint structure | :
  272741003 | Laterality | = [[ + (< 182353008 | side | ) $side ]]

- **Query Language**
  << 64572001 | Disease |
  {{ definitionStatus = 900000000000073002 | Defined ,
  preferredTerm = ".*heart.*",
  languageRefSet = 90000000000508004 | GB English | }}
Terminology APIs and Services

- Used to request the execution of SNOMED CT queries by SNOMED CT enabled terminology server
- Standards
  - HL7 FHIR Terminology Services
  - HL7 Common Terminology Services 2 (CTS2)
  - IHTSDO Open Tooling Framework APIs
- Proprietary
  - Dataline’s SnAPI solution
  - B2i’s Snow Owl Terminology Server

Patient Record Queries with SNOMED CT and SQL

Query options

- List all possible SNOMED CT codes in query
  
  ```sql
  SELECT DISTINCT patientID FROM ProblemList
  WHERE code IN (140004, 181007, 222008, 490008 etc)
  ```

- Load subset into a separate table
  
  ```sql
  SELECT DISTINCT patientID FROM ProblemList
  WHERE code IN (SELECT code FROM RespiratoryDisorders)
  ```

- Use Transitive Closure Table to test susumption
  
  ```sql
  SELECT DISTINCT patientID FROM ProblemList PL
  INNER JOIN TransitiveClosure TC ON TC.sourceld = PL.code
  WHERE TC.targetId = 50043002
  ```

- Embed a terminology query language in record query
  
  ```sql
  SELECT DISTINCT patientID FROM ProblemList
  WHERE code in (< 50043002 |disorder of respiratory system|)
  ```
Querying “Big Data”

- Large volumes of structured and unstructured data sets
- Tools for distributed storage and processing of big data
  - NoSQL (Not Only SQL) systems – e.g. RDFox
    - Store and retrieve data in a variety of structures, including relational, key-value, graph or documents
  - Apache Hadoop
    - Open source software which splits files into large blocks and distributes these blocks amongst nodes in cluster
    - Processes nodes in parallel; supports horizontal scaling

Analytics Tasks
SNOMED CT Analytics Tasks

- Point of care analytics
  - Historical summaries
  - Point of care reporting
  - Clinical decision support
- Population based analytics
  - Trend analysis
  - Pharmacovigilance
  - Clinical audit
- Clinical research
  - Identification of clinical trial candidates
  - Predictive medicine
  - Semantic search

Point of Care Analytics

- Historical Summaries
  - Summaries of a patient's clinical history
  - Aggregated data from various institutions, models & code systems
- SNOMED CT Techniques
  - SNOMED CT as a common reference terminology (mapping)
  - Encode free text clinical data (NLP)
  - Group codes into more general categories (subsumption)
  - Use defining relationships to filter relevant records
Point of Care Analytics

- **Point of Care Reporting**
  - SNOMED CT enables ‘collect once and use many times’ goal
  - Examples include
    - Helping clinicians remember preventative services (reminders)
    - Identifying patients with care gaps and risk factors
    - Monitoring patient compliance with prescribed treatments
    - Reporting clinical data to disease registries
- **SNOMED CT techniques**
  - Mapping to SNOMED CT, Subsets, Subsumption, Defining Relationships, Description Logic, Mapping to classifications

Population-based Analytics

- **Trend Analysis**
  - The process of extracting underlying patterns or trends in data
  - Can be used to detect changes in incidence or prevalence of a disease, treatment, procedure or intervention over time
    - For population health monitoring, prediction of demand, and effective resource allocation
  - **SNOMED CT techniques**
    - Subsumption testing using SNOMED CT’s polyhierarchy
      - Helps to distinguish minor changes in coding style from real changes in disease incidence
      - Which level of aggregation to use can be arbitrary
  - **UK Data Migration Workbench**
    - Identifies most frequently used types of codes using a novel algorithm where each subtree has around 1% of all codes
Population-based Analytics

- **Pharmacovigilance**
  - Collection, detection, assessment, monitoring and prevention of adverse effects with pharmaceutical products
  - Uses a number of data sources including
    - Clinical trial data, Medical literature, Reporting databases, Prescription events, Electronic Health Records, Patient registries
- **SNOMED CT Techniques**
  - NLP and mapping to support homogeneous approach to querying diseases, signs, symptoms, lab results, medications, devices, procedures, allergies, adverse reactions, body sites and substances
  - Subsumption and defining relationships
  - Maps to MedDRA for alternative form of analysis

Population-based Analytics

- **Clinical Audit**
  - Seeks to improve patient care and outcomes through systematic review of care against defined standards and the implementation of change
  - Questions asked in audit may include
    - What proportion of patients invited to attend cervical screening did so?
    - How many patients with ischemic heart disease are receiving appropriate drug treatments?
    - Are all patients with diabetes mellitus reviewed within a stated time interval?
- **SNOMED CT Techniques**
  - NLP, Mapping, Subset, Subsumption, Defining relationships, Description Logic
Clinical Research

- Identification of Clinical Trial Candidates
  - For recruitment into formal clinical trials
  - SNOMED CT techniques
    - Subsets of findings, procedures or medications
    - Subsumption
    - Defining relationships – for example:
      - Patients with diseases of specific anatomical site or morphology
      - Patients taking medications with specific ingredients or forms
      - Patients who have had procedures on a specific body site
    - Description Logic

Clinical Research

- Predictive Medicine
  - Predicting the probability of disease and implementing measures to either prevent or significantly decrease its impact, such as
    - Lifestyle modifications
    - Increased physician surveillance
      - E.g. Regular skin exams, mammograms, colonoscopies
    - Focuses on genetic markers, phenotypic, environmental factors and other lifestyle factors.
  - SNOMED CT can help with
    - Identifying clinical trial candidates
    - Analyzing clinical data, such as family history, lifestyle and environmental findings
    - Linking patient data and risk assessment rules, so that rules can be triggered based on codes recorded in clinical data
Clinical Research

▪ Semantic Search
  ▪ Searching medical literature and clinical reports
  ▪ Indexes collections of free text transcripts and documents
  ▪ Supports topic specific searches – for example:
    ▪ Show me articles related to inflammatory bowel disease
    ▪ Does this patient have transcripts in their record suggesting a heart rhythm disturbance?
▪ SNOMED CT techniques
  ▪ Synonyms (vocabulary mismatch)
  ▪ Subsumption (granularity mismatch)
  ▪ Defining relationships (conceptual implication)
  ▪ Subsets (inferences of similarity)
  ▪ Assign weight to each relationship type to determine relevance of each document

Challenges
Challenges for Clinical Analytics

- Reliability of patient data
- Terminology / information model boundary issues
- Concept definition issues
- Versioning

Clinical Decision Support

Overview
Logical Architecture
Knowledge Base
Inference Engine
Communications
Overview

Clinical Decision Support Overview

- **Clinical Decision Support**
  - **What?**
    - Enables healthcare providers to make well-informed decisions
  - **How?**
    - Supplies guidance, knowledge, and patient-specific information
  - **When?**
    - At relevant points in the patient journey
      - Such as diagnosis, treatment, and follow-up
Clinical Decision Support Systems

- **CDSS**
  - A system designed to improve clinical decision-making related to diagnostic or therapeutic processes of care
  - Typically a decision support system responds to "triggers"
    - Specific diagnoses
    - Laboratory results
    - Medication choices
    - Complex combinations of these
  - Provides information or recommendations directly relevant to a specific patient encounter

*Definition from U.S. Department of Health and Human Services, Agency for Healthcare Research and Quality*

Functional Areas of CDS

- Implemented in a variety of tools, services
  - Alerts
    - Designed to interrupt clinicians or patients at appropriate time
  - Clinical guidelines / reference information
    - Links to external knowledge references
    - Based on relevant, context-dependent data captured in a patient health record
  - Conditional order sets / Pathway support
    - Guides clinicians through complex care pathways
  - Automatically triggered reports, summaries, or smart forms
    - Facilitate high quality records, reduction of errors, more complete information
  - Diagnostic support tools
    - Aid the clinician in making a diagnosis
Clinical Areas

- Use Cases
  - Medication management
  - Asthma management guidelines
  - Diagnosis (e.g. diabetes)
  - Laboratory (e.g. critical results)
  - Radiology
    - Contraindication
    - Appropriate imaging
  - Nursing interventions
  - Infectious disease reporting
  - Clinical quality improvement
  - And many more…

CDS Example – Penicillin Allergy Alert

**Condition:** Patient has penicillin allergy and clinician is prescribing new drug containing penicillin

**Action:** Display alert to clinicians

**Alert:** Patient is allergic to penicillin. [Search for safe alternatives.]
CDS Example – Penicillin Allergy Alert

< 373873005 | Pharmaceutical / biologic product |
127489000 | Has active ingredient | = << 373270004 | Penicillin |

Logical Architecture
Components of a CDS-Enabled EHR

- **User Interface**
  - Provides inputs to system
  - Receives interventions (alerts)

- **Record Services**
  - Stores health records
  - Responds to health records queries

- **Terminology Services**
  - Responds to terminology queries

- **Clinical Decision Support System**
  - Executes Decision Support Logic

---

CDS-Enabled EHR

- **Inputs**
- **Outputs (Alerts)**
- **Processes**
- **User Interface**
- **Enters, Stores**
- **Searches, Retrieves, Displays**
- **Accesses**
- **Queries**
- **Record Services**
- **Terminology Services**
CDSS Internal Components

- **Knowledge Base**
  - Uses clinical knowledge
  - Stores clinical rules and guidelines
  - In machine processable format

- **Inference Engine**
  - Combines:
    - Inputs, health records, rules, and terminology
    - To execute decision support logic
    - Determines the outcome of rules

- **Communications**
  - Delivers outputs to external components
    - Example: Alerts to user interface
  - Handles system inputs
    - Example: Proposed drug or treatment regimen

CDSS High Level Architecture

- **Knowledge Base**
  - Stores Rules and Guidelines

- **Inference Engine**
  - Uses inputs, health records, rules, and terminology to execute decision support logic

- **Communications**
  - Displays alerts to user interface.
  - Accepts inputs from clinicians.
Knowledge Base – Overview

- Clinical knowledge fuels the knowledge base
- Developed by clinical experts in various domains
- Types of knowledge base artifacts
  - Decision support rules
  - Clinical guidelines and care pathways
  - Documentation templates
  - Order sets
- Rules and guidelines
  - May be published by 3rd party knowledge providers
  - Made available to inference engine in machine processable format
  - Enables execution of decision support logic
  - Updated when new clinical knowledge becomes available
**Knowledge Base**

- **Clinical Knowledge**
- **Rules and Guidelines:**
  - Loaded
- **CDS Logic:**
  - Executed
- **Machine Readable**
  - Rules and Guidelines: Processed

---

**Knowledge Base – Rules**

- **Rules follow a typical pattern**
  - First something happens (This is the event)
    - Clinician prescribes a drug to a patient
    - Clinical review of patients previously diagnosed with cancer
  - This triggers a question (This is the condition)
    - Has the patient been prescribed a medication containing a substance to which they are allergic?
    - Have all patients with a suspected cancer diagnosis been referred to a specialist within 14 days?
  - If "yes", then what should be done? (This is the action)
    - Alert the user and suggest a safe alternative
    - Refer identified patients to oncology specialist
- **Rules access both health records and terminology to determine whether or not condition is true**
Knowledge Base Rules – Example

- **Event**: Clinical encounter and diagnosis
- **Condition**: Clinician enters diagnosis of asthma
- **Action**: Display asthma management guidelines

Knowledge Base Rules – Logical Pattern

- **Event**: Clinical encounter
- **Condition**: Diagnosis is asthma
- **Action**: Display asthma management guidelines
Knowledge Base Rules and Context

- Evaluation of the condition must consider context
- Different ways of recording context in health records
  1. Precoordinated expressions (individual concepts)
  2. Postcoordinated expressions
  3. Context-specific section or field
  4. Separate context field, e.g. status
- SNOMED CT soft default context
  - |Finding context| = |Known present|
  - |Procedure context| = |Done|
  - |Subject relationship context| = |Subject of record|
  - |Temporal context| = |Current or specified time|

Knowledge Base Rules and Context – Examples

1. **Condition**: Current diagnosis of asthma  
   **Action**: Display asthma management guidelines
   - Diagnosis of asthma (known absent):  
     Should not trigger the rule

2. **Condition**: F/H of breast cancer + Age ≥ 40  
   **Action**: Refer patient to breast screening program
   - Breast cancer recorded in family history section:  
     Should trigger the rule

3. **Condition**: Current diagnosis of diabetes type II  
   **Action**: Order HBA1C within 12 months
   - Past history of diabetes type II  
     May not trigger the rule
Data Entry with Context

Select disorder:

Asthma
... ...

Finding context
Absent
Present

Subject relationship context
Family
Subject

Temporal context
Current
In the past

Knowledge Base Rules – Example 1

IF

ON clinical encounter

THEN
display asthma management guidelines

[+$diagnosis] = [Asthma]
AND [Finding context] = [Known present]
AND [Subject relationship context] = [Subject of record]
AND [Temporal context] = [Current or specified time]

Event
Condition(s)
Action

Context Specific Conditions
Knowledge Base Rules –
Additional Considerations

- Rules can have multiple conditions and actions
- A single condition exists of a criteria: value pair
- Criteria may refer to coded data elements
  - These may be expressed using SNOMED CT Expression Constraints
- Other criteria may refer to non-coded data elements
  - These may use operators appropriate for the datatype
  - Examples:
    - > 250 (numeric)
    - <= 7.5
    - != "absent" (i.e. string)
    - = true/false (i.e. Boolean)

Knowledge Base Rules – Example 2
Knowledge Base Rules – Example 3

IF
\( \text{ON publishing of lab results} \)

AND
\( (\text{condition}) \rightarrow < 3.0 \text{ mmol/L} \)

THEN
alert attending physician

Event
Coded data element
Condition(s)
Non-coded data element
Action

Knowledge Base Guidelines

KEY CLINICAL ACTIVITIES FOR QUALITY ASTHMA CARE (continued)

<table>
<thead>
<tr>
<th>Clinical Issue</th>
<th>Key Clinical Activities and Action Steps</th>
</tr>
</thead>
<tbody>
<tr>
<td>Patient Education for Self-Management</td>
<td>Teach patients how to manage their asthma.</td>
</tr>
<tr>
<td>Teach and reinforce at each visit:</td>
<td></td>
</tr>
<tr>
<td>Self-monitoring to assess level of asthma control and recognize signs of worsening asthma (either symptom or peak flow monitoring).</td>
<td></td>
</tr>
<tr>
<td>Taking medication correctly (inhaler technique, use of devices; understanding difference between long-term control and quick-relief medications)</td>
<td></td>
</tr>
<tr>
<td>Long-term control medications (such as inhaled corticosteroids which reduce inflammation) prevent symptoms. Should be taken daily, will not give quick relief.</td>
<td></td>
</tr>
<tr>
<td>Quick-relief medications (short-acting beta-agonists or SABA’s) relax airway muscles to provide fast relief of symptoms. Will not provide long-term asthma control. If used &gt;2 days/week (except as needed for exercise-induced asthma), the patient may need to start or increase long-term control medications.</td>
<td></td>
</tr>
</tbody>
</table>

SNOMED CT Semantic Tagging (metadata):
195967001 [Asthma (disorder)]
406162001 [Asthma management (regime/therapy)]
445531003 [Asthma control questionnaire (assessment scale)]

SNOMED CT Semantic Tagging (metadata):* 
195967001 [Asthma (disorder)]
406162001 [Asthma management (regime/therapy)]
445531003 [Asthma control questionnaire (assessment scale)]

*SNOMED CT Semantic Tagging (metadata):* 
195967001 [Asthma (disorder)]
406162001 [Asthma management (regime/therapy)]
445531003 [Asthma control questionnaire (assessment scale)]

*Key Clinical Activities and Action Steps:*
- Take daily actions to control asthma
- Adjust medications in response to worsening asthma
- Seek medical care as appropriate

*Asthma Care Quick Reference, US Department of Health and Human Services, National Institutes of Health, National Heart Lung and Blood Institute*
Knowledge Base Guidelines – Semantic Tags

\[ \text{IF } 195949000 \rightarrow \text{Chronic asthmatic bronchitis} \rightarrow 195967001 \rightarrow \text{Asthma} \quad \text{THEN} \quad \text{display NIH Asthma Care Quick Reference} \]

Patient Encounter:

Diagnosis:
- Chronic asthmatic bronchitis

Knowledge Links:
- NIH Asthma Care Quick Reference

Semantic Tag:
- 105967001 | Asthma

Knowledge Base – Representation Standards

- Rule representations and standards
  - Arden Syntax
    - HL7 Implementation Guide for Arden Syntax, Release 1
    - HL7 FHIR DecisionSupportRule (Resource)
  - CDS Hooks
    - http://cds-hooks.org/
- Guideline definition
  - GELLO
    - HL7 Version 3 Standard: GELLO, A Common Expression Language, Release 2
    - Guideline Definition Language
      - http://www.openehr.org/releases/CDS/latest/docs/GDL/GDL.html
Inference Engine – Overview

- Processes machine readable rules
- Considers system inputs
- Accesses data in the health records
- Queries terminology services
- Establishes if conditions are met
- Determines outcome
- i.e. “executes” the Triggers
Reasoning with SNOMED CT: Overview

- SNOMED CT techniques may be applied to clinical decision support
- Assists inference engine in evaluating the trigger conditions defined in rules
- Techniques:
  - Subsets
  - Subsumption
  - Defining relationships
  - Description Logic
    - Over Terminology
    - Over Terminology and Structure
Reasoning with Subsets

**IF $diagnosis = ^111115 [Asthma subset] THEN display asthma management guidelines**

- Simple rule using a SNOMED CT subset
- Inference engine checks for subset membership
- Subset may be defined
  - Extensionally
    - By manually selecting values
    - More time consuming to develop and maintain
    - May overlook some necessary values in the subset
  - Intensionally
    - By defining membership using a query, e.g. expression constraint
    - Easier to maintain for new versions of SNOMED CT
    - Simple to refer to all values in a specific subhierarchy

### Reasoning with Subsets

**Diagnosis:** Occasional Asthma

**Asthma Conditions Subset:**

<table>
<thead>
<tr>
<th>Id</th>
<th>Term</th>
</tr>
</thead>
<tbody>
<tr>
<td>304527002</td>
<td>Acute asthma</td>
</tr>
<tr>
<td>389145006</td>
<td>Allergic asthma</td>
</tr>
<tr>
<td>233678006</td>
<td>Childhood asthma</td>
</tr>
<tr>
<td>445427006</td>
<td>Seasonal asthma</td>
</tr>
<tr>
<td>370221004</td>
<td>Severe asthma</td>
</tr>
</tbody>
</table>

**Concept ID:** 370220003

**Technique:** Checks for ^111115 [Asthma subset]  

- **Match:** No
- **Condition:** False
- **Action:** Not triggered
Reasoning using Subsumption

IF \$\text{diagnosis} = \text{<< 195967001 |Asthma|} \quad \text{THEN display asthma management guidelines}

- Rule uses Expression Constraint Language to define “asthma or subtypes”
- Engine to test for subsumption
  - Rules may reference subtypes, supertypes, descendants, ancestors, parents, children, etc
- Enables “richer” expression within rules
- Can be more efficient than maintaining subsets or extensionally defined lists

Reasoning with Subsumption

\textbf{Diagnosis:}

\begin{itemize}
  \item \textbf{Match:}
    \begin{itemize}
      \item Yes
    \end{itemize}
  \item \textbf{Condition:}
    \begin{itemize}
      \item True
    \end{itemize}
  \item \textbf{Action:}
    \begin{itemize}
      \item Triggered
    \end{itemize}
\end{itemize}

\textbf{Transitive Closure Table:}

<table>
<thead>
<tr>
<th>Subtype</th>
<th>Supertype</th>
</tr>
</thead>
<tbody>
<tr>
<td>304527002</td>
<td>195967001</td>
</tr>
<tr>
<td>389145006</td>
<td>195967001</td>
</tr>
<tr>
<td>426979002</td>
<td>195967001</td>
</tr>
<tr>
<td>445427006</td>
<td>195967001</td>
</tr>
<tr>
<td>370221004</td>
<td>195967001</td>
</tr>
</tbody>
</table>
Reasoning using Defining Relationships

Rule uses Expression Constraint Language to constrain a set of procedures (those which have a procedure site of respiratory system)

Inference engine leverages concept definitions

Also facilitates richer expression in rules
  - Using SNOMED CT’s attribute relationships
  - Example: Drug active ingredients or procedure methods

IF $procedure = << 71388002 |Procedure| : 363704007 |Procedure site| = 20139000 |Structure of respiratory system|

THEN consult respirologist

SNOMED CT Relationships Table:

<table>
<thead>
<tr>
<th>sourceId</th>
<th>destinationId</th>
<th>typeId</th>
</tr>
</thead>
<tbody>
<tr>
<td>229308003</td>
<td>128258000</td>
<td>363702006</td>
</tr>
<tr>
<td>229308003</td>
<td>302803009</td>
<td>363702006</td>
</tr>
<tr>
<td>229308003</td>
<td>262202000</td>
<td>363703001</td>
</tr>
<tr>
<td>229308003</td>
<td>20139000</td>
<td>363704007</td>
</tr>
<tr>
<td>229308003</td>
<td>20139000</td>
<td>405813007</td>
</tr>
<tr>
<td>229308003</td>
<td>47545007</td>
<td>116680003</td>
</tr>
</tbody>
</table>

Concept ID: 229308003

Procedure: Intermittent CPAP
Communications – Overview

- Handles inputs and outputs for CDSS
- User inputs
  - Clinical data entered
  - User selections (criteria for CDS), for example:
    - Proposed drug, order set, or treatment regime
- Delivers outputs
  - i.e. “CDS Interventions”
    - Alerts
    - Guidelines
    - Refinements (diagnostic)
    - Smart Forms
  - Displayed to the User Interface
- Guidelines and references may be externally referenced
Communications

Rule condition is evaluated

Knowledge Resource

Delivery

User Interface

Communications

Communications – Example

CDS Notifications:
Patient has clinical markers that are considered a risk for Pompe Disease. Consider Ordering GAA enzyme activity assay to confirm absence or presence of diagnosis.

Reference Information
Links to Further Information

- Data Analytics with SNOMED CT
  - [http://snomed.org/analytics](http://snomed.org/analytics)

- SNOMED CT Languages
  - [http://snomed.org/ecl](http://snomed.org/ecl)
  - [http://snomed.org/scg](http://snomed.org/scg)

- Technical Implementation Guide
  - [http://snomed.org/tig](http://snomed.org/tig)

- Questions & Comments?