Ontologies and Public Health

Olivier Bodenreider

Lister Hill National Center for Biomedical Communications
Bethesda, Maryland - USA
London Bills of Mortality

A general Bill for this present year,
ending the 19th of December 1665, according to
the Report made to the Kings most Excellent Majesty.

By the Company of Parish Clerks of London, etc.

The Diseases and Casualties this year:

<table>
<thead>
<tr>
<th>Disease</th>
<th>Number</th>
</tr>
</thead>
<tbody>
<tr>
<td>Palefe</td>
<td>39</td>
</tr>
<tr>
<td>Aged</td>
<td>11</td>
</tr>
<tr>
<td>Aqut and Fever</td>
<td>150</td>
</tr>
<tr>
<td>Aged, and Small Pox</td>
<td>91</td>
</tr>
<tr>
<td>Found dead in streets, fields, etc</td>
<td>83</td>
</tr>
<tr>
<td>French Pox</td>
<td>3</td>
</tr>
<tr>
<td>Plague</td>
<td>1</td>
</tr>
<tr>
<td>French Pox</td>
<td>288</td>
</tr>
<tr>
<td>Plague</td>
<td>642</td>
</tr>
<tr>
<td>Appopleys, and Suddenly</td>
<td>764</td>
</tr>
<tr>
<td>French Pox</td>
<td>22</td>
</tr>
<tr>
<td>Plague</td>
<td>139</td>
</tr>
<tr>
<td>French Pox</td>
<td>66</td>
</tr>
<tr>
<td>Plague</td>
<td>14</td>
</tr>
<tr>
<td>Grief</td>
<td>23</td>
</tr>
<tr>
<td>GRIpping in the Guts</td>
<td>22</td>
</tr>
<tr>
<td>Rilling of the Guts</td>
<td>12</td>
</tr>
<tr>
<td>Burned and Scalped</td>
<td>15</td>
</tr>
<tr>
<td>Hungel, &amp; made away themselves</td>
<td>10</td>
</tr>
<tr>
<td>Epupture</td>
<td>1</td>
</tr>
<tr>
<td>Hecmolfection &amp; Muscelfallen</td>
<td>47</td>
</tr>
<tr>
<td>Scory</td>
<td>11</td>
</tr>
<tr>
<td>Cancer, &amp;c.</td>
<td>15</td>
</tr>
<tr>
<td>Grippinage and Fatlute</td>
<td>17</td>
</tr>
<tr>
<td>Spleenity</td>
<td>1</td>
</tr>
<tr>
<td>Spleen</td>
<td>13</td>
</tr>
<tr>
<td>Spotted Fever and Purples</td>
<td>31</td>
</tr>
<tr>
<td>Splen</td>
<td>17</td>
</tr>
<tr>
<td>Illingos and Swine pox</td>
<td>2</td>
</tr>
<tr>
<td>Skin, Ulcers, broken and hurtled</td>
<td>12</td>
</tr>
<tr>
<td>Limbs</td>
<td>45</td>
</tr>
<tr>
<td>Childbed</td>
<td>29</td>
</tr>
<tr>
<td>Killt by severall accidents</td>
<td>13</td>
</tr>
<tr>
<td>Canker, and Thrush</td>
<td>64</td>
</tr>
<tr>
<td>Temphume</td>
<td>6</td>
</tr>
<tr>
<td>Leperote</td>
<td>3</td>
</tr>
<tr>
<td>Cold and Congh</td>
<td>10</td>
</tr>
<tr>
<td>Spering, at the formont</td>
<td>1</td>
</tr>
<tr>
<td>Collick and Windse</td>
<td>14</td>
</tr>
<tr>
<td>Lethery</td>
<td>37</td>
</tr>
<tr>
<td>Contagion and Th diseas</td>
<td>110</td>
</tr>
<tr>
<td>Liverlouse, and Headlacht</td>
<td>4</td>
</tr>
<tr>
<td>Salt</td>
<td>1</td>
</tr>
<tr>
<td>Convolvion and Mether</td>
<td>122</td>
</tr>
<tr>
<td>Soot &amp; Strong</td>
<td>1</td>
</tr>
<tr>
<td>Distreced</td>
<td>3</td>
</tr>
<tr>
<td>Measles</td>
<td>13</td>
</tr>
<tr>
<td>Teeth and Worms</td>
<td>212</td>
</tr>
<tr>
<td>Distreced</td>
<td>62</td>
</tr>
<tr>
<td>Measles</td>
<td>10</td>
</tr>
<tr>
<td>Mustered and Shote</td>
<td>8</td>
</tr>
<tr>
<td>Norway</td>
<td>50</td>
</tr>
<tr>
<td>Downed and Company</td>
<td>12</td>
</tr>
<tr>
<td>Overland &amp; Staved</td>
<td>45</td>
</tr>
<tr>
<td>Venm</td>
<td>1</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>City</th>
<th>Males</th>
<th>Females</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Males</td>
<td>497</td>
<td>383</td>
<td>880</td>
</tr>
<tr>
<td>Females</td>
<td>383</td>
<td>497</td>
<td>880</td>
</tr>
</tbody>
</table>

Increased in the Bills in the 13 parishs in and at the Pest house this year. 79548
Increased of the Plague in the 13 parishs and at the Pest house this year. 88570
Need for ontologies

“The advantages of a uniform statistical nomenclature, however imperfect, are so obvious, that it is surprising no attention has been paid to its enforcement in Bills of Mortality. Each disease has, in many instances, been denoted by three or four terms, and each term has been applied to as many different diseases: vague, inconvenient names have been employed, or complications have been registered instead of primary diseases. The nomenclature is of as much importance in this department of inquiry as weights and measures in the physical sciences, and should be settled without delay.”

– William Farr

First annual report.
Outline

◆ Biomedical ontologies – What they are
◆ Biomedical ontologies – What they are for
  (Relevance to public health)
◆ Related artifacts
  ● Value sets
  ● Common data elements
  ● Terminology integration systems
Biomedical ontologies

What they are
Overview

◆ Structural perspective
  - What are they (vs. what are they for)?

◆ “High-impact” biomedical ontologies
  - International Classification of Diseases (ICD)
  - Logical Observation Identifiers, Names and Codes (LOINC)
  - SNOMED Clinical Terms
  - Foundational Model of Anatomy
  - Gene Ontology
  - RxNorm
  - Medical Subject Headings (MeSH)
  - NCI Thesaurus
  - Unified Medical Language System (UMLS)

[J. Cimino, YBMI 2006]
International Classification of Diseases
ICD Characteristics (1)

- **Current version:** ICD-10
- **Type:** Classification
- **Domain:** Disorders
- **Developer:** World Health Organization (WHO)
- **Funding:** WHO

**Availability**
- Publicly available: No
- Repositories: UMLS, NCBO BioPortal

**URL:** [http://www.who.int/classifications/icd/en/](http://www.who.int/classifications/icd/en/)
ICD Characteristics (2)

◆ Number of
  • Concepts: ~12,000
  • Terms: 1 per concept (tabular)

◆ Major organizing principles:
  • Tree (single inheritance hierarchy)
  • No explicit classification criteria
    ▶ Idiosyncratic inclusion/exclusion mechanism
  • .8 slots for Not elsewhere classified (NEC)
  • .9 slots for Not otherwise specified (NOS)

◆ Formalism: Proprietary format
<table>
<thead>
<tr>
<th>Chapter</th>
<th>Blocks</th>
<th>Title</th>
</tr>
</thead>
<tbody>
<tr>
<td>I</td>
<td>A00-B99</td>
<td>Certain infectious and parasitic diseases</td>
</tr>
<tr>
<td>II</td>
<td>C00-D48</td>
<td>Neoplasms</td>
</tr>
<tr>
<td>III</td>
<td>D50-D89</td>
<td>Diseases of the blood and blood-forming organs and certain disorders involving the immune mechanism</td>
</tr>
<tr>
<td>IV</td>
<td>E00-E90</td>
<td>Endocrine, nutritional and metabolic diseases</td>
</tr>
<tr>
<td>V</td>
<td>F00-F99</td>
<td>Mental and behavioural disorders</td>
</tr>
<tr>
<td>VI</td>
<td>G00-G99</td>
<td>Diseases of the nervous system</td>
</tr>
<tr>
<td>VII</td>
<td>H00-H59</td>
<td>Diseases of the eye and adnexa</td>
</tr>
<tr>
<td>VIII</td>
<td>H60-H95</td>
<td>Diseases of the ear and mastoid process</td>
</tr>
<tr>
<td>IX</td>
<td>I00-I99</td>
<td>Diseases of the circulatory system</td>
</tr>
<tr>
<td>X</td>
<td>J00-J99</td>
<td>Diseases of the respiratory system</td>
</tr>
<tr>
<td>XI</td>
<td>K00-K93</td>
<td>Diseases of the digestive system</td>
</tr>
<tr>
<td>XII</td>
<td>L00-L99</td>
<td>Diseases of the skin and subcutaneous tissue</td>
</tr>
<tr>
<td>XIII</td>
<td>M00-M99</td>
<td>Diseases of the musculoskeletal system and connective tissue</td>
</tr>
<tr>
<td>XIV</td>
<td>N00-N99</td>
<td>Diseases of the genitourinary system</td>
</tr>
<tr>
<td>XV</td>
<td>O00-O99</td>
<td>Pregnancy, childbirth and the puerperium</td>
</tr>
<tr>
<td>XVI</td>
<td>P00-P96</td>
<td>Certain conditions originating in the perinatal period</td>
</tr>
<tr>
<td>XVII</td>
<td>Q00-Q99</td>
<td>Congenital malformations, deformations and chromosomal abnormalities</td>
</tr>
<tr>
<td>XVIII</td>
<td>R00-R99</td>
<td>Symptoms, signs and abnormal clinical and laboratory findings, not elsewhere classified</td>
</tr>
<tr>
<td>XIX</td>
<td>S00-T98</td>
<td>Injury, poisoning and certain other consequences of external causes</td>
</tr>
<tr>
<td>XX</td>
<td>V01-Y98</td>
<td>External causes of morbidity and mortality</td>
</tr>
<tr>
<td>XXI</td>
<td>Z00-Z99</td>
<td>Factors influencing health status and contact with health services</td>
</tr>
<tr>
<td>XXII</td>
<td>U00-U99</td>
<td>Codes for special purposes</td>
</tr>
</tbody>
</table>
ICD Example

- Idiosyncratic inclusion/exclusion criteria

![ICD Example](image-url)

**E10**

**Insulin-dependent diabetes mellitus**

[See before E10 for subdivisions.]

**Includes:** diabetes (mellitus):
- brittle
- juvenile-onset
- ketosis-prone
- type I

**Excludes:** diabetes mellitus (in):
- malnutrition-related (E12.-)
- neonatal (P70.2)
- pregnancy, childbirth and the puerperium (O24.-)
  glycosuria:
  - NOS (R81)
  - renal (E74.8)
  impaired glucose tolerance (R73.0)
  postsurgical hypoinsulinaemia (E89.1)
### ICD Example

- **Not elsewhere classified (NEC)**
- **Not otherwise specified (NOS)**

<table>
<thead>
<tr>
<th>Code</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>E84</td>
<td>Cystic fibrosis</td>
</tr>
<tr>
<td><strong>Includes:</strong> mucoviscidosis</td>
<td></td>
</tr>
<tr>
<td>E84.0</td>
<td>Cystic fibrosis with pulmonary manifestations</td>
</tr>
<tr>
<td>E84.1</td>
<td>Cystic fibrosis with intestinal manifestations</td>
</tr>
<tr>
<td>Meconium ileus* (P75*)</td>
<td></td>
</tr>
<tr>
<td><strong>Excludes:</strong> meconium obstruction in cases where cystic fibrosis is known not to be present (P76.0)</td>
<td></td>
</tr>
<tr>
<td>E84.8</td>
<td>Cystic fibrosis with other manifestations</td>
</tr>
<tr>
<td>E84.9</td>
<td>Cystic fibrosis, unspecified</td>
</tr>
</tbody>
</table>
Logical Observation Identifiers, Names and Codes (LOINC)
**LOINC Characteristics (1)**

- **Current version:** 2.50 (Dec. 2014)
- **Type:** Controlled terminology*
- **Domain:** Laboratory and clinical observations
- **Developer:** Regenstrief Institute
- **Funding:** NLM

**Availability**
- Publicly available: Yes
- Repositories: UMLS, NCBO BioPortal

**URL:** [www.regenstrief.org/loinc/loinc.htm](http://www.regenstrief.org/loinc/loinc.htm)
LOINC Characteristics (2)

- **Number of**
  - Concepts: ~70,000 active codes
  - Terms: 1 per concept ("long common name")

- **Major organizing principles:**
  - No hierarchical structure among the main codes
  - 6 axes
    - Component (analyte [+ challenge] [+ adjustments])
    - Property
    - Timing
    - System
    - Scale
    - [Method]

- **Formalism:** “DL-like”
**LOINC Example**

- **Sodium:SCnc:Pt:Ser/Plas:Qn**  
  [the molar concentration of sodium is measured in the plasma (or serum), with quantitative result]

<table>
<thead>
<tr>
<th>Axis</th>
<th>Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Component</td>
<td>Sodium</td>
</tr>
<tr>
<td>Property</td>
<td>SCnc – Substance Concentration (per volume)</td>
</tr>
<tr>
<td>Timing</td>
<td>Pt – Point in time (Random)</td>
</tr>
<tr>
<td>System</td>
<td>Ser/Plas – Serum or Plasma</td>
</tr>
<tr>
<td>Scale</td>
<td>Qn – Quantitative</td>
</tr>
<tr>
<td>Method</td>
<td>--</td>
</tr>
</tbody>
</table>
SNOMED Clinical Terms
SNOMED CT Characteristics (1)

- Current version: (2 annual releases)
  - International release: July 31, 2014
  - U.S. edition: September 2014
- Type: Reference terminology / ontology
- Domain: Clinical medicine
- Developer: IHTSDO
- Funding: IHTSDO
- Availability
  - Publicly available: Yes* (in member countries)
  - Repositories: UMLS, NCBO BioPortal
- URL: http://www.ihtsdo.org/
SNOMED CT Characteristics (2)

◆ Number of
  ● Concepts: ~300,000 active concepts
  ● Terms: ~1.2M active “descriptions”

◆ Major organizing principles:
  ● Utility for clinical medicine (e.g., definitional knowledge)
  ● Model of meaning (incomplete)
  ● Rich set of associative relationships
  ● Small proportion of defined concepts (many primitives)

◆ Formalism: Description logics
SNOMED CT Top level

- SNOMED CT Concept
  - Body structure (body structure)
  - Clinical finding (finding)
  - Environment or geographical location (environment / location)
  - Event (event)
  - Observable entity (observable entity)
  - Organism (organism)
  - Pharmaceutical / biologic product (product)
  - Physical force (physical force)
  - Physical object (physical object)
  - Procedure (procedure)
  - Qualifier value (qualifier value)
  - Record artifact (record artifact)
  - Situation with explicit context (situation)
  - SNOMED CT Model Component (metadata)
  - Social context (social concept)
  - Special concept (special concept)
  - Specimen (specimen)
  - Staging and scales (staging scale)
  - Substance (substance)
SNOMED CT Terminology

Parents
- Operation on appendix (procedure)
- Partial excision of large intestine (procedure)

Appendectomy (procedure)
SCTID: 80146002
- Appendectomy (procedure)
- Appendectomy
- Appendicectomy
- Excision of appendix

Procedure site - Direct →
Appendix structure
Method → Excision - action

Children (8)
- Appendectomy with drainage (procedure)
- Emergency appendectomy (procedure)
- Excision of appendiceal stump (procedure)
- Excision of ruptured appendix by open approach (procedure)
- Incidental appendectomy (procedure)
- Interval appendectomy (procedure)
- Laparoscopic appendectomy (procedure)
- Non-emergency appendectomy (procedure)
SNOMED CT Logical definitions

- Appendectomy (procedure) (80146002)
- Partial excision of large intestine (procedure) (27010001)
- Operation on appendix (procedure) (8613002)
- Procedure site - Direct (attribute) (405813007)
- Appendix structure (body structure) (66754008)
- Method (attribute) (260686004)
- Excision - action (qualifier value) (129304002)
RxNorm
RxNorm Characteristics (1)

- Current version: Jan. 2015 (monthly releases)
- Type: Controlled terminology
- Domain: Drug names
- Developer: NLM
- Funding: NLM

Availability
- Publicly available: Yes*
- Repositories: UMLS, NCBO BioPortal

URL: http://www.nlm.nih.gov/research/umls/rxnorm/
**RxNorm Characteristics (2)**

- **Number of**
  - Concepts: ~120k
  - Terms: (limited number of synonyms)

- **Major organizing principles:**
  - Generic vs. brand
  - Combinations of Ingredient / Form / Strength
  - No hierarchical structure
  - Links to all major US drug information sources
  - No clinical information or drug classes*

- **Formalism: UMLS RRF format**
RxNorm Normalized form

- Strength: 4mg/ml
- Ingredient: Fluoxetine
- Dose form: Oral Solution
Rx Norm  Generic vs. Brand

- **Generic**
  - Ingredient (IN)
  - Clinical drug form (SCDF)
  - Clinical drug component (SCDC)
  - Clinical drug (SCD)

- **Brand**
  - Brand name (BN)
  - Branded drug form (SBDF)
  - Branded drug component (SBDC)
  - Branded drug (SBD)

*tradename_of*
RxNorm Relations among drug entities
## Recap (as of 2008)

<table>
<thead>
<tr>
<th>Name</th>
<th>Scope</th>
<th># concepts</th>
<th>Median</th>
<th>Subs. Hier</th>
<th>Version</th>
</tr>
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<tbody>
<tr>
<td>SNOMED CT</td>
<td>Clinical medicine (patient records)</td>
<td>310,314</td>
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<td>July 31, 2007</td>
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<tr>
<td>LOINC</td>
<td>Clinical observations and laboratory tests</td>
<td>46,406</td>
<td>3</td>
<td>no</td>
<td>Version 2.21 (no “natural language” names)</td>
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<tr>
<td>FMA</td>
<td>Human anatomical structures</td>
<td>~72,000</td>
<td>?</td>
<td>yes</td>
<td>(not yet in the UMLS)</td>
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<tr>
<td>RxNorm</td>
<td>Standard names for prescription drugs</td>
<td>93,426</td>
<td>1</td>
<td>no</td>
<td>Aug. 31, 2007</td>
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<tr>
<td>NCI Thesaurus</td>
<td>Cancer research, clinical care, public information</td>
<td>58,868</td>
<td>2</td>
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<td>2007_05E</td>
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<tr>
<td>ICD-10</td>
<td>Diseases and conditions (health statistics)</td>
<td>12,318</td>
<td>1</td>
<td>no</td>
<td>1998 (tabular)</td>
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<tr>
<td>MeSH</td>
<td>Biomedicine (descriptors for indexing the literature)</td>
<td>24,767</td>
<td>5</td>
<td>no</td>
<td>Aug. 27, 2007</td>
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<tr>
<td>UMLS .</td>
<td>Terminology integration in the life sciences</td>
<td>1,4 M</td>
<td>2</td>
<td>n/a</td>
<td>2007AC (English only)</td>
</tr>
</tbody>
</table>
Biomedical ontologies

What they are for
Relevance to public health
Overview

◆ Functional perspective
  ● What are they for (vs. what are they)?

◆ “High-impact” biomedical ontologies

◆ 3 major categories of use
  ● **Knowledge management** (indexing and retrieval of data and information, access to information, mapping among ontologies)
  ● **Data integration**, exchange and semantic interoperability
  ● **Decision support and reasoning** (data selection and aggregation, decision support, natural language processing applications, knowledge discovery)

[Bodenreider, YBMI 2008]
Needs for public health

- Support for text mining
- Controlled vocabulary
- Aggregation
- Data integration
- Reasoning
Support for text mining

- **Lexical resources**
  - Identify mentions in text
    - Lexical variants

- **Terminological resources**
  - Identify concepts
    - Synonyms

- **Ontological resources**
  - Identify relations, Semantic interpretation
    - Domain knowledge
Controlled vocabulary

- Coded information
  - Storage
  - Processing
- Standardize
  - Definitions
  - Usage
Data aggregation

◆ Granularity mismatch
  • Data recorded
  • Data needed for making decisions

◆ Aggregation along hierarchies
  • Subsumption hierarchies (isa)
  • Ad hoc linearizations (e.g., ICD for mortality / morbidity)
Data integration

◆ Datasets annotated in reference to multiple ontologies
◆ Establish correspondence between equivalent concepts across ontologies (and datasets)
◆ Role of terminology integration systems
  ● UMLS, RxNorm
  ● NCBO ontology services
Reasoning

- Description Logics
- Reasoning services (DL classifiers)
  - Instance classification
Related artifacts

(1) Value sets
Value sets

- Sets of standard codes from a given code system used for cohort identification

- Use cases
  - Clinical quality measures

- Repositories
  - NLM Value Set Authority Center
  - CDC PHIN VADS
    - https://phinvads.cdc.gov/vads/BrowseValueSets_browse.action
### Metadata

**Name:** Gestational Diabetes  
**Type:** Extensional  
**Steward:** National Committee for Quality Assurance  

**OID:** 2.16.840.1.113883.3.464.1003.103.11.1012  
**Definition ID:** 20130614  
**Program:** CMS, MU2 EP Update 2013-06-14 using this value set

### Value Set Members

<table>
<thead>
<tr>
<th></th>
<th></th>
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<th></th>
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</tr>
</thead>
<tbody>
<tr>
<td>11687002</td>
<td>Gestational diabetes mellitus (disorder)</td>
<td>SNOMEDCT</td>
<td>2013-01</td>
<td>2.16.840.1.113883.6.9</td>
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<td>420491007</td>
<td>Gestational diabetes mellitus, class H (disorder)</td>
<td>SNOMEDCT</td>
<td>2013-01</td>
<td>2.16.840.1.113883.6.9</td>
</tr>
<tr>
<td>420738003</td>
<td>Gestational diabetes mellitus, class T (disorder)</td>
<td>SNOMEDCT</td>
<td>2013-01</td>
<td>2.16.840.1.113883.6.9</td>
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<tr>
<td>420989005</td>
<td>Gestational diabetes mellitus, class R (disorder)</td>
<td>SNOMEDCT</td>
<td>2013-01</td>
<td>2.16.840.1.113883.6.9</td>
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<tr>
<td>421223006</td>
<td>Gestational diabetes mellitus, class F (disorder)</td>
<td>SNOMEDCT</td>
<td>2013-01</td>
<td>2.16.840.1.113883.6.9</td>
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<td>421389009</td>
<td>Gestational diabetes mellitus, class C (disorder)</td>
<td>SNOMEDCT</td>
<td>2013-01</td>
<td>2.16.840.1.113883.6.9</td>
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**Value Set Information**

- **Value Set Code**: PHVS_GestationalDiabetes_NCHS
- **Value Set Name**: Gestational Diabetes (NCHS)
- **Value Set OID**: 1.3.6.1.4.1.19376.1.7.3.1.1.13.8.137
- **Value Set Description**: To Reflect Risk Factors of Gestational Diabetes

**Value Set Concepts**

- **Select All**
- **Clear All**
- **5 Value Set Concepts found**

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<th>Concept Code</th>
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<th>Concept Name</th>
<th>Preferred Concept Name</th>
<th>Code System</th>
<th>Value Set</th>
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<td>Gestational Diabetes (NCHS)</td>
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<td>diabetes mellitus in mother complicating pregnancy, childbirth AND/OR puerperium</td>
<td>diabetes mellitus in mother complicating pregnancy, childbirth AND/OR puerperium</td>
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Related artifacts

(2) Common data elements
Common data elements

- Information model vs. terminology
  - Binding between the two

- Metadata for biomedical datasets
  - Standard annotations for data integration

- Current efforts at NIH
  - Big Data to Knowledge (BD2K)
Common data elements at NIH

NIH encourages the use of common data elements (CDEs) in clinical research, patient registries, and other human subject research in order to improve data quality and opportunities for comparison and combination of data from multiple studies and with electronic health records. This portal provides access to NIH-supported CDE initiatives and other tools and resources that can assist investigators developing protocols for data collection. What is a CDE?

NIH CDE Initiatives
Collections of CDEs that have been identified for use in particular NIH-supported research projects or registries after a formal evaluation and selection processes.

NIH CDE Tools and Resources
Databases and repositories of data elements and case report forms that may assist investigators in identifying and selecting data elements for use in their projects.
<table>
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<th>Initiative</th>
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<th>Description</th>
<th>Disease Area</th>
<th>Funding</th>
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<td>Quality of Life Outcomes in Neurological Disorders</td>
<td>Neuro-QOL</td>
<td>A core set of quality-of-life questions that address chronic neurologic disorders, plus sets of supplemental questions specific to targeted diseases or subgroups of patients. More...</td>
<td>Neurological disorders. More...</td>
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<td>NINDS</td>
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<td>NIDA Substance Abuse Electronic Health Record Data Elements</td>
<td>NIDA EHR</td>
<td>A set of brief screening and initial assessment tools for substance use disorders (SUDs) for use in general medical settings. More...</td>
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<td>NIH Toolbox for Assessment of Neurological and Behavioral Function</td>
<td>NIH Toolbox</td>
<td>An integrated set of tools for measuring cognitive, emotional, motor and sensory function. More...</td>
<td>Cognitive, emotional, motor, and sensory function. More...</td>
<td>4 batteries of tests, each with 5-24 tests</td>
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<td>NINDS Common Data Elements</td>
<td>NINDS CDEs</td>
<td>A core set of data elements for use in NINDS-funded studies, including core and supplementary sets of data elements for use in disease-specific studies. More...</td>
<td>Neurological disorders. More...</td>
<td>7,000+ variables, 100s of instruments</td>
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<td>Consensus Measures for Phenotypes and eXposures</td>
<td>PhenX</td>
<td>Standard measures related to complex diseases, phenotypic traits and environmental exposures for inclusion in genome-wide association studies (GWAS) and other large-scale genomic and epidemiologic research efforts. More...</td>
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<td>Patient Reported Outcomes Measurement Information System</td>
<td>PROMIS</td>
<td>A system of item banks measuring patient-reported health status for various domains of physical, mental, and social health across clinical populations (i.e. not disease-specific). More...</td>
<td>Physical, mental, and social health. More...</td>
<td>50 item banks</td>
<td>NIAMS</td>
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Related artifacts

(3) Terminology integration systems
Integrating subdomains

- Clinical repositories
- Genetic knowledge bases
- SNOMED CT
- OMIM
- Biomedical literature
- MeSH
- GO
- Genome annotations
- NCBI Taxonomy
- FMA
- Anatomy
- Model organisms
- Other subdomains
Integrating subdomains

- Clinical repositories
- Genetic knowledge bases
- Biomedical literature
- Genome annotations
- Anatomy
- Model organisms
- Other subdomains
Trans-namespace integration

Addison's disease (363732003)

Clinical repositories

SNOMED CT

Other subdomains

UMLS C0001403

Genetic knowledge bases

OMIM

Biomedical literature

Addison Disease (D000224)

NCBI Taxonomy

GO

Model organisms

FMA

Anatomy

Genome annotations

Addison's disease (363732003)

Biomedical literature

Addison Disease (D000224)
Terminology integration systems

- **Unified Medical Language System (UMLS)**
  - National Library of Medicine (NLM)

- **BioPortal**
  - National Center for Biomedical Ontology (NCBO)
  - [http://bioportal.bioontology.org/](http://bioportal.bioontology.org/)
Medical Ontology Research

Contact: olivier@nlm.nih.gov
Web: mor.nlm.nih.gov

Olivier Bodenreider
Lister Hill National Center for Biomedical Communications
Bethesda, Maryland - USA
Bodenreider O, Stevens R.  
Brief Bioinform. 2006 Sep;7(3):256-74.

Cimino JJ, Zhu X.  
The practical impact of ontologies on biomedical informatics.  

Bodenreider O.  
Biomedical ontologies in action: role in knowledge management, data integration and decision support.  