Data and Knowledge Representation

Lecture 6
Last Time We Talked About

- Ontology
- Data Model
Today We Will Talk About

- Medical vocabulary representation
- Survey of medical coding systems
Terminology as Crucial Requirement

*Without* Terminology Standards...

- Health Data is *non*-comparable
- Health Systems *cannot* Interchange Data
- Secondary Uses (Research, Efficiency) are *not* possible
- Linkage to Decision Support Resources *not* Possible
Weights and Measures

“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, about Cullenian system
Vocabulary Representation Approaches

- List, Tree/Hierarchy, Multiple Tree
- Embed hierarchy information in identifier or not
- Pre-coordination, Post-coordination
- Description logic
Description Logic

- Describe the world in terms of "properties" or "constraints" that specific "individuals" have to satisfy
- Objects
- Concepts
- Roles
Example

- Represent the following concepts in a vocabulary
  - Flu
  - Pneumonia
  - Flu with Pneumonia
  - Infectious Disease
  - Respiratory Disease
  - Adult
  - Infant
First Formal Health Data Model
London Bills of Mortality

- Commissioned 1542 (1598)
- Intended to Track Plague (Black Death)
  - ~60 disease categories
    - Variously Defined
  - Collected by Parish Clerks (Chanters)
  - Irregularly Printed in Folio
  - Data Table Layout
    - 16th Century Spreadsheet

Slides borrowed from Dr. Christopher G. Chute of Mayo Clinic
ICD

- International Classification of Diseases and its Clinical Modification
- First published in 1893 by Statistical International Institute
- Revised every 10+- years
  - ICD8 – 1967 (World Health Organization)
  - ICD9 – 1977 (World Health Organization)
  - ICD10 – 1982 (World Health Organization)
ICD

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Coding

- 3 digit code to represent core term
- 4th digit (in the first decimal place) to provide addition info.
  - Typically .0 to .7 are used for more specific forms of the core term
  - .8 is for “other”
  - .9 is for “unspecified”
<table>
<thead>
<tr>
<th>Code</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>250.0</td>
<td>DM without mention complications</td>
</tr>
<tr>
<td>250.1</td>
<td>Diabetes with ketoacidosis</td>
</tr>
<tr>
<td>250.2</td>
<td>Diabetes with hyperosmilarity</td>
</tr>
<tr>
<td>250.3</td>
<td>Diabetes with other coma</td>
</tr>
<tr>
<td>250.4</td>
<td>Diabetes with renal manifestations</td>
</tr>
<tr>
<td>250.5</td>
<td>Diabetes with ophthalmic manifestations</td>
</tr>
<tr>
<td>250.5</td>
<td>Diabetes with neurological manifestations</td>
</tr>
<tr>
<td>250.7</td>
<td>Diabetes with peripheral circulatory disorders</td>
</tr>
<tr>
<td>250.8</td>
<td>Diabetes with other specified manifestation</td>
</tr>
<tr>
<td>250.9</td>
<td>Diabetes with unspecified complications</td>
</tr>
</tbody>
</table>

Slides adapted from Dr. Christopher G. Chute of Mayo Clinic

- Missing Severity
- Not flexible
ICD-9 adopted by US National Center for Health Statistics (NCHC)
Clinical Modification (CM) was made
ICD-9-CM vs IC9
- Compatible
- Additional digits (4th and 5th) to provide extra levels of detail
Most Diagnoses in US are coded in ICD-9-CM
ICD-10-CM

- Pre-release draft
- ICD-10-CM far exceeds its predecessors in the number of codes provided.
- The disease classification has been expanded to include health-related conditions
- Provide greater specificity at the sixth digit level and with a seventh digit extension.
ICD10-CM

- **A06 Amebiasis**
  - Includes: infection due to Entamoeba histolytica
  - Excludes1: other protozoal intestinal diseases (A07.-)
- **A06.0 Acute amebic dysentery**
  - Acute amebiasis
  - Intestinal amebiasis NOS
- **A06.1 Chronic intestinal amebiasis**
- **A06.2 Amebic nondysenteric colitis**


- Diagnosis-Related Group
- Initially developed for billing purpose at Yale (1960s)
- Used for Medicare payment since 1980s
- Depend on ICD-9-CM
  - DRG codes need to be derived from ICD codes
Algorithm

- Algorithmic mapping from ICD based Data to DRG codes
- Distinguish cases based on factors that affect cost and length of stay
  - Severity of illness
  - Prognosis
  - Treatment difficulty
  - Need for intervention
  - Resource intensity - administrative

Slides adapted from Dr. Christopher G. Chute of Mayo Clinic
<table>
<thead>
<tr>
<th>DRG Description</th>
<th>DRG Code</th>
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</thead>
<tbody>
<tr>
<td>Simple Pneumonia with minor complication, age greater than 17</td>
<td>89</td>
</tr>
<tr>
<td>Simple Pneumonia with no minor complication, age greater than 17</td>
<td>90</td>
</tr>
<tr>
<td>Respiratory disease with ventilator support</td>
<td>475</td>
</tr>
<tr>
<td>Respiratory disease with major chest operating room procedure and major</td>
<td></td>
</tr>
<tr>
<td>complication or comorbidity</td>
<td></td>
</tr>
<tr>
<td>Respiratory disease, other respiratory system operating procedure and major</td>
<td></td>
</tr>
<tr>
<td>complication or comorbidity</td>
<td></td>
</tr>
<tr>
<td>Respiratory infection with major complication or comorbidity</td>
<td>540</td>
</tr>
<tr>
<td>Respiratory infection with secondary diagnosis of bronchopulmonary dysplasia</td>
<td>631</td>
</tr>
<tr>
<td>Respiratory infection with secondary diagnosis of cystic fibrosis</td>
<td>740</td>
</tr>
</tbody>
</table>
CPT

- Current Procedure Terminology
- Developed by American Medical Association
Precoordinated coding scheme

“Endovascular repair of descending thoracic aortic aneurysm, pseudoaneurysm or dissection involving coverage of left subclavian artery origin, initial endoprosthesis, radiological supervision and interpretation”

Diagnostic and therapeutic procedure

For billing and reimbursements (public and private)

“withdraw of blood for diagnosis”

Current version: CPT 4 (CPT 5 expected in 2003)
DSM

- Diagnostic and statistical Manual of Mental Disorders
- Developed by American Psychiatric Association
- Includes definition and diagnostic criteria
- Limited number of concepts
  - Correspond to ICD10
Anxiety Disorders
- **Acute Stress Disorder** (acute psychological consequences of previous trauma)
- Agoraphobia (generalized irrational fear)
- **Generalized Anxiety Disorder** (nonspecific anxiety)
- **Obsessive-Compulsive Disorder** (obsessive thoughts and compulsive rituals)
- **Panic Disorder** (unprovoked panic attacks)
- **Posttraumatic Stress Disorder** (nonacute psychological consequences of previous trauma)
- Separation Anxiety Disorder
- **Social Phobia** (irrational fear of embarrassment)
- **Specific Phobia** (other specific irrational fears)

Childhood Disorders
- **Attention-Deficit/Hyperactivity Disorder**
- **Conduct Disorder** (the Antisocial Personality Disorder of Childhood)
- **Oppositional Defiant Disorder** (not only for children)
Diagnostic Criteria

- The person has been exposed to a traumatic event in which both of the following were present:
  - the person experienced, witnessed, or was confronted with an event or events that involved actual or threatened death or serious injury, or a threat to the physical integrity of self or others
  - the person's response involved intense fear, helplessness, or horror
- Either while experiencing or after experiencing the distressing event, the individual has three (or more) of the following dissociative symptoms:
  - a subjective sense of numbing, detachment, or absence of emotional responsiveness
  - a reduction in awareness of his or her surroundings (e.g., "being in a daze")
  - derealization
  - depersonalization
  - dissociative amnesia (i.e., inability to recall an important aspect of the trauma)
Anxiety Disorder

- The traumatic event is persistently reexperienced in at least one of the following ways: recurrent images, thoughts, dreams, illusions, flashback episodes, or a sense of reliving the experience; or distress on exposure to reminders of the traumatic event.
- Marked avoidance of stimuli that arouse recollections of the trauma (e.g., thoughts, feelings, conversations, activities, places, people).
- Marked symptoms of anxiety or increased arousal (e.g., difficulty sleeping, irritability, poor concentration, hypervigilance, exaggerated startle response, motor restlessness).
- The disturbance causes clinically significant distress or impairment in social, occupational, or other important areas of functioning or impairs the individual's ability to pursue some necessary task, such as obtaining necessary assistance or mobilizing personal resources by telling family members about the traumatic experience.
- The disturbance lasts for a minimum of 2 days and a maximum of 4 weeks and occurs within 4 weeks of the traumatic event.
- The disturbance is not due to the direct physiological effects of a substance (e.g., a drug of abuse, a medication) or a general medical condition, is not better accounted for by Brief Psychotic Disorder, and is not merely an exacerbation of a preexisting Axis I or Axis II disorder.
SNOMED

- Systematized Nomenclature of Human and Veterinarian Medicine
- Developed by the College of American Pathologists (1971)
- SNDO, SNOP, SNOMED
- SNOMED International
- SNOMED-RT (Reference Terminology)
- SNOMED-CT (merger with Read)
<table>
<thead>
<tr>
<th>Category</th>
<th>Count</th>
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<tbody>
<tr>
<td>Chemicals, Drugs, and Biological Products</td>
<td>14,846</td>
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<tr>
<td>Diseases/Diagnoses</td>
<td>35,834</td>
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<td>Function</td>
<td>19,221</td>
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<tr>
<td>General Linkage/Modifiers</td>
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<td>Living Organisms</td>
<td>24,614</td>
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<td>Manufacturers of Pharmaceuticals Human/Veterinary</td>
<td>363</td>
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<tr>
<td>Morphology</td>
<td>5,875</td>
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<td>Occupations</td>
<td>1,949</td>
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<td>Physical Agents, Forces and Activities</td>
<td>1,600</td>
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<tr>
<td>Procedures</td>
<td>30,723</td>
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<td>Social context</td>
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<td>Topography</td>
<td>12,936</td>
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<tr>
<td></td>
<td>150,343</td>
</tr>
</tbody>
</table>
SNOMED III - Coding Examples

"D3-15000" "01" "Myocardial infarction, NOS" "(T-32020) (M-54700)"
  T-32020 = Myocardium, NOS
  M-54700 = Infarction, NOS

"D3-15010" "01" "Microinfarct of heart" "(T-32000) (M-54701)"
  T-32000 = Heart, NOS
  M-54701 = Focal Infarct

Mother died of myocardial infarct
  S-10120, S-13030, D3-15000
  S-10120, F-A7860, T-32020, M-54700
**Fully Specified Name:** Myocardial infarction (disorder)

**Concept ID:** 22298006

**Definition:**
- Is a (attribute) *Myocardial disease (disorder)*
- Is a (attribute) *Structural disorder of heart (disorder)*
- Associated morphology (attribute) *Infarct (morphologic abnormality)*
- Finding site (attribute) *Myocardium structure (body structure)*
**Fully Specified Name:** Heart disease in mother complicating pregnancy, childbirth AND/OR puerperium (disorder)

**Concept ID:** 78381004

**Definition:**
- Is a (attribute) Cardiac complication (disorder)
- Is a (attribute) Complication related to pregnancy (disorder)
- Finding site (attribute) Heart structure (body structure)

**Qualifiers:**
- Onset (attribute) Onsets (qualifier value)
- Severity (attribute) Severities (qualifier value)
- Episodicity (attribute) Episodicities (qualifier value)
- Course (attribute) Courses (qualifier value)
**Fully Specified Name:** Needle biopsy (procedure)

**Concept ID:** 129249002

**Definition:**
- Is a (attribute) Biopsy (procedure)
- Method (attribute) Biopsy - action (qualifier value)
- Using (attribute) Biopsy needle, device (physical object)

**Qualifiers:**
- Priority (attribute) Priorities (qualifier value)
Appropriate values for the Priority (attribute) relationship type

Deferred (qualifier value)
Denied (qualifier value)
Elective (qualifier value)
Emergency (qualifier value)
Immediate (qualifier value)
Reclassified (qualifier value)
Reclassified and rescheduled (qualifier value)
Repeat elective (qualifier value)
Repeat emergency (qualifier value)
Rescheduled (qualifier value)
Routine (qualifier value)
Scheduled (qualifier value)
Urgency (qualifier value)
Read Clinical Codes

- Developed by James Read in the 80s
- Adopted by UK NHS in 1990
- Allows post-coordination
- Merged with SNOMED
182..A Y7CmDC P Chest pain
Xa0wWK Y7CmFC P Pleurodynia
182Z.A Y7CmGC P Chest pain NOS
Xa0wWK Y7CmIC S Painful breathing -pleurodynia
1826.A Y7CmJC P Parasternal pain
1823.A Y7CmLC P Precordial pain
1821.A Y7CmNC P Chest pain not present
X75rWC Y7CmYC P Pain in heart
1829.A Y7CmZC P Retrosternal pain
Gabrieli Medical Nomenclature

- Single large hierarchy
- More complex terms as you move down
- Being adopted by ASTM as a standard
Nursing terminologies

- Many initiatives worldwide
- North American Nursing Diagnosis Association (NANDA) codes
- Nursing Outcomes Classification (NOC)
- Georgetown Home Health Care Classification (HHCC)
- Omaha System
  - Problems, interventions, outcomes
GALEN

- European initiative
- Reference model for medical concepts
- Formalism called Structured Meta Knowledge
  - Similar to description logic
LOINC

- Logical Observations, Identifiers, Names, Codes (LOINC)
- Consortium led by Clem McDonald and Stan Huff
- Originally lab results
- Now extended to include clinical observations
- Recently, merged into SNOMED
National Drug Codes

- Developed by FDA
- Widely used in US
- Codes based on drug manufacturer
- Codes have no class hierarchy
- Codes are reused at manufacturer’s discretion
- Not useful for decision support
MeSH

- Medical Subject Headings
- Developed by NLM
- Indexes medical literature
  - Medline
- Terms are in hierarchies and appear in multiple places in hierarchies
Reading

- Cimino’s paper on coding system
- http://www.psychologynet.org/dsm.html
- http://www.loinc.org/
- http://www.fda.gov/cder/ndc/