Clinical Informatics and Decision Making:

Challenges for Large-Scale Analytics and Intelligent Services

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What are the gaps?

- Intelligent services based on individual rule bases will never scale
- It is difficult to characterize the feature space that leads to a diagnosis
- It is difficult to characterize the category space when you decide on a diagnosis
- Imprecision in the category spaces mean imprecision in therapeutics
- The underlying information infrastructure is evolving—very slowly—from a 19th century model
Ontologies are essential for biology.
The Foundational Model of Anatomy
Biomedical scientists have adopted ontologies

- To provide canonical representation of scientific knowledge
- To annotate experimental data to enable interpretation, comparison, and discovery across databases
- To facilitate knowledge-based applications for
  - Decision support
  - Natural language-processing
  - Data integration
# The International Classification of Diseases

<table>
<thead>
<tr>
<th>Code</th>
<th>Description</th>
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<tbody>
<tr>
<td>724</td>
<td>Unspecified disorders of the back</td>
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<tr>
<td>724.0</td>
<td>Spinal stenosis, other than cervical</td>
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<td>724.02</td>
<td>Spinal stenosis, lumbar region</td>
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<td>724.09</td>
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<td>Sciatica</td>
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<td>Disorders of sacrum</td>
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<td>724.7</td>
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<td>724.8</td>
<td>Other symptoms referable to back</td>
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<tr>
<td>724.9</td>
<td>Other unspecified back disorders</td>
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</table>
ICD9 (1977): A handful of codes for traffic accidents

- E80 Railway accidents
- E81 Motor vehicle traffic accidents
- E82 Motor vehicle nontraffic accidents
  - E820 NONTRAFFIC ACCIDENT INVOLVING MOTOR-DRIVEN SNOW V
  - E821 NONTRAFFIC ACCIDENT INVOLVING OTHER OFF-ROAD MOTO
  - E822 OTHER MOTOR VEHICLE NONTRAFFIC ACCIDENT INVOLVING
  - E823 OTHER MOTOR VEHICLE NONTRAFFIC ACCIDENT INVOLVING
  - E824 OTHER MOTOR VEHICLE NONTRAFFIC ACCIDENT WHILE BOA
  - E825 OTHER MOTOR VEHICLE NONTRAFFIC ACCIDENT OF OTHER.
- E826 PEDAL CYCLE ACCIDENT
  - E826.0 PEDAL CYCLE ACCIDENT INJURING PEDESTRIAN
  - E826.1 PEDAL CYCLE ACCIDENT INJURING PEDAL CYCLIST
  - E826.2 PEDAL CYCLE ACCIDENT INJURING RIDER OF ANIMAL
  - E826.3 PEDAL CYCLE ACCIDENT INJURING OCCUPANT OF ANIM
  - E826.4 PEDAL CYCLE ACCIDENT INJURING OCCUPANT OF STRE
  - E826.8 PEDAL CYCLE ACCIDENT INJURING OTHER SPECIFIED
  - E826.9 PEDAL CYCLE ACCIDENT INJURING UNSPECIFIED PERS
- E827 ANIMAL-DRAWN VEHICLE ACCIDENT
ICD10 (1999):
587 codes for such accidents

- V31.22 Occupant of three-wheeled motor vehicle injured in collision with pedal cycle, person on outside of vehicle, nontraffic accident, while working for income
- W65.40 Drowning and submersion while in bath-tub, street and highway, while engaged in sports activity
- X35.44 Victim of volcanic eruption, street and highway, while resting, sleeping, eating or engaging in other vital activities
There is a plethora of controlled terminologies!

- Diseases: ICD-9, ICD-9-CM, ICD-10, ICD-10-CM, DRG
- Procedures: CPT-4, ICD-10-PCS
- Laboratory tests: LOINC
- Nursing activities: NIC, NOC, HHCC, Omaha
- Drugs: NDC, Multum, Micromedex, NDDF,
- Biomedical literature: MeSH
- Clinical documentation: Medcin, Purkinjie
- Cross-references among terminologies: UMLS
REPORT TO THE PRESIDENT
REALIZING THE FULL POTENTIAL OF
HEALTH INFORMATION TECHNOLOGY
TO IMPROVE HEALTHCARE
FOR AMERICANS:
The Path Forward

Executive Office of the President
President’s Council of Advisors
on Science and Technology
What are some of the Advisors recommendations?

- Continue incentives for “meaningful use” of EHRs
- Encourage exchange of information across health-care facilities
- Establish a “universal exchange language” for clinical data
- Initiate pilot projects to allow the approach to scale
NCBO: Key activities

• We **create and maintain a library of biomedical ontologies and terminologies.**

• We **build tools and Web services** to enable the use of ontologies and terminologies.

• We **collaborate with scientific communities** that develop and use ontologies and terminologies in biomedicine.
Welcome to the NCBO Bioportal

Use BioPortal to access and share ontologies that are actively used in biomedical communities. You can search for terms in ontologies (try typing “Melanoma” in the “Search all ontologies” box or “Find an ontology” in the middle column), browse a list of ontologies in BioPortal (type “NCI Thesaurus” in the “Find an ontology” box in the middle column), search biomedical resources that we automatically annotated with ontology terms (try typing “Melanoma” in the “Search resources” box in the right column). You can create ontology-based annotations for your own text, link your own project that uses ontologies to the description of these ontologies, find and create relations between terms in different ontologies, review and comment on ontologies and their components as you browse them. Sign in to BioPortal to submit a new ontology or ontology-based project, provide comments on ontologies or add ontology mappings.

Search all ontologies

Find an ontology

Search resources

Most Active Ontologies

<table>
<thead>
<tr>
<th>Ontology</th>
<th>Version</th>
<th>Notes</th>
<th>Mappings</th>
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Latest Notes

- Term name misspelled?
  leaf vascular tissue (Minimal anatomical terminology) 10/11/09 whetzel
- Incorrect mapping
  SON (Rat Strain Ontology) 09/23/09 whetzel
- RE: NEMO.owl subontologies/modules
  scalp_surface_region (Neural ElectroMagnetic Ontologies) 08/22/09 gfrishkoff
- NEMO subontologies/modules
  entity (Neural ElectroMagnetic Ontologies) 08/22/09 gfrishkoff
- NEMO.owl subontologies/modules
  scalp_surface_region (Neural ElectroMagnetic Ontologies) 08/22/09 gfrishkoff

Latest Mappings

- human (Human developmental anatomy, timed version) => Humans (Medical Subject Headings) 10/03/09 yongqun@med.umich.edu
- Humans (Medical Subject Headings) => human (Human developmental anatomy, timed version) 10/03/09 yongqun@med.umich.edu
- sand_fly (Parasite Life Cycle) => Phlebotomus (SNOMED Clinical Terms) 08/17/09 preets111
- Phlebotomus (SNOMED Clinical Terms) => sand_fly (Parasite Life Cycle) 08/17/09 preets111
- amastigote (Parasite Life Cycle) => Amastigote form of protozoa (SNOMED Clinical Terms) 08/14/09 preets111

http://bioportal.bioontology.org
Access all ontologies that are available in BioPortal: You can filter this list by category to display ontologies relevant for a certain domain (try selecting "Anatomy or Experimental Conditions" in the 'Filter by category' menu below). You can also filter ontologies that belong to a certain group such as ontologies from the OBO Foundry, or ontologies from the Cancer Biomedical Informatics Grid (caBIG). Subscribe to the BioPortal RSS feed to receive alerts for submissions of new ontologies, new versions of ontologies, new notes, and new projects. You can subscribe to feeds for a specific ontology at the individual ontology page. Add a new biomedical ontology to BioPortal using the Submit New Ontology link (you need to sign in to see this link).

<table>
<thead>
<tr>
<th>ONTOLOGY NAME</th>
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</table>
BioPortal allows us to experiment with new models for

- Dissemination of terminologies, ontologies, and knowledge on the Web
- Integration and alignment of online content
- Knowledge visualization and cognitive support
- Peer review of online content
Biomedical Resource Ontology in BioPortal
“Notes” in BioPortal

Comment: Software needs structure, too many top level subclasses
DavidStates at 08/09/08 06:56

- "binary executable" is not a top level subclass of software, it is a form of software distribution and there are several other subclasses of software distribution (source code, web site, library, toolkit, etc.).
- Similarly, "network editor" is just one class of interactive editing tools. Lots of others.
- These are just a couple of examples. Software really needs a complete reorganization.

Reply

Comment: RE:Software needs structure, too many top level subclasses
PeterLystser at 08/12/08 08:29

The BRO used the initial design principle of: when in doubt make it flat at the top. This is a design principle whose purpose is to get the class names on the board and agreed upon first, i.e., it is a componentization of the design process. This is a way of avoiding getting into debates about hierarchical location too early in the process. We can discuss location in the hierarchy in the future; that is appropriate.

Reply

Comment: RE:Software needs structure, too many top level subclasses
PeterLystser at 08/12/08 08:43

I (Peter Lystser) copy marginal notes that I also place in the 'Portals' class. I think this helps to explain the design principles.

We adopted the design principle of (i) Initially align the BRO top level with NIFSTD (Data Resource; Bibliographic Resource; Software; Research Supplies; Portals; Funding Source) (see agreement that was made in broad icon of 20080416 http://na-mic.org/Wiki/index.php/SDWG:Meeting_Minutes_20080416). As with the discussion on 'Software' class, the goal was to get a reasonable first cut and then stabilize the BRO development process; then the development team (called 'tiger team' after the April icon) agreement (interdigitate etc) on the overall list of class names (this was successfully done by Rubin, Martone, and Lystser between July 28 and August 1 2008). This process was highly successful, and validated the logic behind taking one step at a time; (ii) continue to work with NIFSTD and other stakeholders to plan current and future efficient and effective mappings. It is good to revisit in the future the position of upper-level classes such as 'portals' or 'funding source.'
BioPortal is building an online community of users who

• Develop, upload, and apply ontologies
• Map ontologies to one another
• Comment on ontologies via “notes” to give feedback
  • To the ontology developers
  • To one another
• Make proposals for specific changes to ontologies
• Stay informed about ontology changes and proposed changes via “push” technology
• Incorporate BioPortal services into their own technologies
WebProtégé allows collaborative ontology authoring online.
Like BioPortal, WebProtégé supports notes and threaded discussions.
As with BioPortal, notes may include multimedia.
Integration of Ontology Authoring, Publishing, and Peer Review

BioPortal

WebProtégé
NCBO will support the complete ontology lifecycle
**Blood Pressure apparently not under control:**

Based on last measurement of 145/92 taken 87 days ago on mm/dd/yyyy

*Estimated 10 Year cardiovascular risk factor for this patient [Explain](#)*

**Cardio Risk Factor: 23% High**

- Consider intensifying drug treatment: BP Elevated based on most recent available BP
- There appears to be a Strong Contraindication to a currently prescribed drug, evaluate clinical significance
- Bronchospasm is a Strong Contraindication or use of beta adrenergic receptor antagonists, although many patients tolerate and therefore benefit from this drug therapy

Review lifestyle modifications with the patient. See the [Lifestyle](#) page.

**Therapeutic Possibilities**

- Discontinue atenolol
- AND start one of the following drugs
  - ACE Inhibitors (lisinopril)
    - (non-DHP) Calcium Channel Blocker (diltiazem)
- Add one or more of the following drugs
  - ACE Inhibitors (lisinopril)
    - (non-DHP) Calcium Channel Blocker (diltiazem)
- Increase dosage of hydrochlorothiazide

**Indications**

- Heart Failure [EVIDENCE]
- CKD

**Contraindications**

- Brochoplastic disease
- Heart Failure

**Blood Pressure and Prescription History**

- 142/90 on [Drug]

**Date:** MM/DD/YY

- [Write back to Vista]

**Lisinopril**

- Med 2: 5 MG
- Med 3: 100 MG
- Med 4: 80 MG
- Med 5: 80 MG
- Med 6: 5 MG
- Med 7: 100 MG

Showing 7 of 10 drugs. [See All](#)

**Do you have feedback for the Research team?**

Thank you!

- [Do not display advisory for this clinic visit again]

*Don't forget you know the patient better than we do.*

*Lorem ipsum dolor sit amet, consectetur adipiscing elit.*
The task: guideline-based patient management

Begin or Continue Lifestyle Modifications

Not at Goal Blood Pressure (<140/90 mm Hg)
Lower goals for patients with diabetes or renal disease (see chapter 4)

Initial Drug Choices*

*Uncomplicated Hypertension†
- Diuretics
- Beta-blockers

*Specific Indications for the Following Drugs (see table 9)
- ACE inhibitors
- Angiotensin II receptor blockers
- Alpha-blockers
- Alpha-beta-blockers
- Beta-blockers
- Calcium antagonists
- Diuretics

**Compelling Indications†**
- Diabetes mellitus (type 1) with proteinuria
  - ACE inhibitors
- Heart failure
  - ACE inhibitors
  - Diuretics
- Isolated systolic hypertension (older persons)
  - Diuretics preferred
  - Long-acting dihydropyridine calcium antagonists
- Myocardial infarction
  - Beta-blockers (non-ISA)
  - ACE inhibitors (with systolic dysfunction)

- Start with a low dose of a long-acting once-daily drug, and titrate dose.
- Low-dose combinations may be appropriate.

Not at Goal Blood Pressure

Consider adding an ACE Inhibitor because of a compelling indication (heart failure)
A handful of encoded guidelines gives you, well, a handful of encoded guidelines.
GLINDA Task–Method Decomposition

- Get Data
- Select Guideline
- Apply Guideline
- Consolidate Advisories
- Detect Interactions
- Repair
- Prioritize

- Multi-guideline CDS

- ATHENA
- ATHENA w/ Additional Knowledge Source
- Heuristic Rules based on Interaction Ontology
- Interaction-Specific Strategy
- Weight of Support

- DB query
- Manual selection
- Goal satisfied?
- Get KS
- Get KS

- GLINDA Task
- GLINDA – Method Decomposition
Semantic computing is crucial for biomedicine

• Myriad controlled terminologies in medicine are yielding to new ontologies
• Mandates for “meaningful use” of electronic patient records require processing of symbolic representations of patient data and situations
• The terabytes of data spewing from life-sciences laboratories cannot be managed without semantic organization and interpretation
What are the gaps?

• Intelligent services based on individual rule bases will never scale
• It is difficult to characterize the feature space that leads to a diagnosis
• It is difficult to characterize the category space when you decide on a diagnosis
• Imprecision in the category spaces mean imprecision in therapeutics
• The underlying information infrastructure is evolving—very slowly—from a 19th century model
NATIONAL CENTER FOR
BIOMEDICAL ONTOLOGY

http://bioontology.org