Cinical 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





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The Foundational Model of Anatomy





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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

- 724 Unspecified disorders of the back
- 724.0 Spinal stenosis, other than cervical
- 724.00 Spinal stenosis, unspecified region
- 724.01 Spinal stenosis, thoracic region
- 724.02 Spinal stenosis, lumbar region
- 724.09 Spinal stenosis, other
- 724.1 Pain in thoracic spine
- 724.2 Lumbago
- 724.3 Sciatica
- 724.4 Thoracic or lumbosacral neuritis
- 724.5 Backache, unspecified
- 724.6 Disorders of sacrum
- 724.7 Disorders of coccyx
- 724.70 Unspecified disorder of coccyx
- 724.71 Hypermobility of coccyx
- 724.71 Coccygodynia
- 724.8 Other symptoms referable to back
- 724.9 Other unspecified back disorders

ICD9 (1977): A handful of codes for traffic accidents

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+ E81 Motor vehicle traffic accidents
E82 Motor vehicle nontraffic accidents
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E825 OTHER MOTOR VEHICLE NONTRAFFIC ACCIDENT OF OTHER
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E826.0 PEDAL CYCLE ACCIDENT INJURING PEDESTRIAN
E826.1 PEDAL CYCLE ACCIDENT INJURING PEDAL CYCLIST
E826.2 PEDAL CYCLE ACCIDENT INJURING RIDER OF ANIMAL
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<mark>x</mark> e826.9 pedal cycle accident injuring unspecified pers
E827 ANIMAL-DRAWN VEHICLE ACCIDENT

ICD10 (1999): **587 codes for such accidents**

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VI3.24 Fetal cyclus squeet as collaine with nativey paus or nativey vehicle, unspecifiet petal cyclus,

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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
- Encourge exchange of information across health-care facilities
- Establish a "universal exchange language" for clinical data
- Initiate pilot projects to allow the approach to scale

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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.

O BioPortal	Browse	Search	Projects	Annotate	All Mappings	All Resources Alpha	<u>Sign In</u>	<u>Register</u>	Help/About	Send Feedbac

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 in the left 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 those 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
Enter search term, e.,g. m Advanced Search	nelano	Search		Enter ontology name, e.g., NCI Thesauru Explore Browse Ontologies >	Enter a term, e.g., Melanoma Search Advanced Resource Search
— Most Active Ontologies -				Latest Notes	Latest Mappings
Ontology	Version	Notes	Mappings	Term name misspelled? leaf vscular tissue (Minimal anatomical	human (Human developmental anatomy, timed version) => Humans (Medical Subject Headings)
Human developmental anatomy, timed version	1.3	0	168488	terminology) 10/11/09 whetzel	Humans (Medical Subject Headings) => human
<u>Human developmental</u> <u>anatomy, abstract</u> <u>version</u>	1.3	0	146067	Incorrect mapping SDN (Rat Strain Ontology) 09/23/09 whetzel	(Human developmental anatomy, timed version) 10/03/09 yongqunh@med.umich.edu sand_fly (Parasite Life Cycle) => Phlebotomus
NCI Thesaurus	09.07	11	91995	RE:NEMO.owl subontologies/modules scalp_surface_region (Neural ElectroMagnetic	(SNOMED Clinical Terms) 08/17/09 preets1511
Cell Cycle Ontology (H. sapiens)	1.01	0	61199	Ontologies) 08/22/09 gfrishkoff	<u>Phlebotomus (SNOMED Clinical Terms) =></u> sand_fly (Parasite Life Cycle)
<u>Cell Cycle Ontology (S.</u> <u>cerevisiae)</u>	1.01	0	58563	<u>NEMO subontologies/modules</u> <u>entity (Neural ElectroMagnetic Ontologies)</u> 08/22/09 gfrishkoff	08/17/09 preets1511 amastigote (Parasite Life Cycle) => Amastigote form of protozoa (SNOMED Clinical Terms)
				NEMO.owl subontologies/modules	
Ontologies		168		scalp_surface_region (Neural ElectroMagnetic Ontologies) 08/22/09 gfrishkoff	
Concepts		723,8	06		
Resources Indexed		11			

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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 <u>OBO Foundry</u>, or ontologies from the <u>Cancer Biomedical Informatics Grid</u> (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).

SUBMIT ONTOLOGY	Submit New Ontology	
FILTER BY CATEGORY	All Categories	
FILTER BY GROUP	All Groups	Link To This Filter
FILTER BY TEXT		

Subscribe to all updates

ONTOLOGY NAME	FORMAT	VERSION	AUTHOR	UPLOADED ON	GROUP	STATUS
<u>ABA Adult Mouse Brain</u> (<u>ABA)</u>	OWL	1.0	Allen Institute for Brain Science	08/08/2009		Explore
<u>African Traditional Medicine</u> (ATMO)	OBO Format	1.101	Ghislain Atemezing	06/28/2009		Explore
<u>Amino Acid</u> (amino-acid)	OWL	1.2	Nick Drummond, Georgina Moulton, Robert Stevens, Phil Lord	04/25/2009		Explore
Amphibian gross anatomy (AAO)	OBO Format	1.8	AmphiAnat list	07/30/2008	OBO Foundry	Explore
<u>Amphibian taxonomy</u> (ATO)	OBO Format	See Remote Site	AmphiAnat list	08/14/2009		
Animal natural history and life history (ADW)	PROTEGE	See Remote Site	Http://animaldiversity Administrators	04/27/2009		
Ascomycete phenotype ontology (APO)	OBO Format	1.6	Fungal_anatomy Administrators	09/01/2009		Explore
Basic Formal Ontology (BFO)	OWL	1.1		07/24/2009		Explore
<u>Basic Vertebrate Anatomy</u> (basic-vertebrate-gross-anatomy)	OWL	1.1		01/16/2007		Explore
<u>Bilateria anatomy</u> (BILA)	OBO Format	See Remote Site	Thorsten Heinrich	04/13/2009		
<u>Biological imaging methods</u> (FBbi)	OBO Format	1.1	Image Administrators	07/30/2008		Explore



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

O BioPortal	Browse	Sear	ch	Projects	Annotate	All Mappings	All Resources	<u>Sign Ir</u>	Register	Help/About	<u>Ser</u>		
8 Biomedical Resource	BioPortal Browse Search Projects Annotate All Mappings All Resources Alpha Sign In Register Help/About Ser medical Resource Ontology NCI Thesaurus NCI Thesaurus Software Link Here Subscribe Subscribe Notes (3) Mappings (0) Resource Index ew Ontology Details Details Visualization Notes (3) Mappings (0) Resource Index DavidStates at 08/09/08 06:56 Activity Activity Comment: Software ends structure, too many top level subclasses DavidStates at 08/09/08 06:56 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. Resource # Information Resource @ Information Resource Resource Peterlyster at 08/12/08 08:29 # Funding Resource # Algorithm Data Acquisition Software 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 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 principle whose purpose is to get the class names on the												
Biomedical Reso	ource Ontology	Version 2.7	.1	Soft	ware Link Here	🔊 Subscribe							
View Ontology De Jump To:	tails	Go	Details	Visualization	Notes (3) Mapping	s (0) Resource Inc	lex						
Legend Activity Area of Research Biositemaps Infor Core:Collection Core:Concept Core:ConceptSche Deprecated Activ Deprecated Area	mation Model eme ity of Research	*	Comme "binary software Similarly These at <u>Reply</u>	In a complete reaction of the subclass of software, it is a form of software distribution and there are several other subclasses of vare distribution (source code, web site, library, toolkit, etc.). Iarly, "network editor" is just one class of interactive editing tools. Lots of others. We are just a couple of examples. Software really needs a complete reorganization.									
Deprecated Resource Resource Funding Reso Information F Aterial Resource Software Software Algorithm	urce lesource urce rce urce	>	Com The class into appr <u>Repl</u>	BRO used the initial names 'on the boar debates about hiera opriate.	re needs structur design principle of: d and agreed upon' f irchical location too	PeterLyster at 08/12/08 08: of: when in doubt make it flat at the top. This is a design principle whose purpose is to get the of first, i.e., it is a componentization of the design process. This is a way of avoiding getting bo early in the process. We can discuss location in the hierarchy in the future; that is							
Data Ana Data Pro Data Pro Data Stor Data Stor Data Trar Integratio Interactio Modeling Software Training Reso	lysis Software cessing Software rage Repository isfer and Communical on and Interoperabilit ve Tool ge Mining and Capturi and Simulation Distribution urce	tion ty Tool ing	Comment: RE:Software needs structure, too many top level subclasses PeterLyster at 08/12/08 08:43 I (Peter Lyster) 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 tcon of 20080416 http://na-mic.org /Wiki/index.php/SDIWG: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 tcon) agreement (interdigitate etc) on the overall list of class names (this was successfully done by Rubin, Martone, and Lyster 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 th future the position of upper-level classes such as 'portals' or 'funding source'.										

Reply

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

Kelcome to WebProtégé Demo!

My WebProtég	gé <u>Collabora</u>	ative Pizza 🙎								
Classes ×	Properties 🕷	Individuals $^{ imes}$	Notes and Discussions	Other Terminologies 🕷	Test Properties 🙁	Manage Hierarch	у 🗷			
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Classes			Properties for IceC	ream			¢@X			
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 DomainConcept Country IceCream Pizza CheeseyPizza InterestingPizza MeatyPizza NamedPizza NonVegetarianPizza 			rdfs:comment Axioms for IceCrea	A class to demonstrate m a property domain. The p domain of Pizza. This me infer that all individuals u property must be of type I restriction on this class, a must use the hasTopping must also be members of and IceCream are disjoin	istakes made with sett roperty hasTopping ha ans that the reasoner sing the hasTopping Pizza. Because of the Il members of IceCrea property, and therefor Pizza. However, Pizza t so this causes an	ting en as a can Im re a	* * *			
	SpicyPizza SpicyPizzaEq VegetarianPiz VegetarianPiz VegetarianPiz PizzaBase PizzaTopping	uivalent zza zzaEquivalent1 zzaEquivalent2	Superclasses (Nece DomainConcept hasTopping some Fr	Superclasses (Necessary conditions) DomainConcept hasTopping <i>some</i> FruitTopping						
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			New Topic							
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Protégé ogy auth	allows on allows of a second s	collabora nline	tive eryBadicecream,	en Edit Arcl	classes for Goodlcecr lcecream. hive Delete	03/25/20	≡ 010			
						13:40:20 P	. ▲			

Like BioPortal, WebProtégé supports notes and threaded discussions

My ICD	ICD Content	Category Notes and Discussions	Reviews	Change	History	Manage Hierarchy	Export and Impor	t				
ICD Cat	egories			<u> </u>	Notes 1	Tree for LA Infections	and infestations af	fecting the skin				•¢@
Create	Watch Branch	Search: Type search string	Q		New	Topic Expand A	II Collapse Al	<u>I</u>				
	D Categories 🖓	1 727582		â	🚽 De	ecision as to which infe	ections and	Rol	bert	Explanation	0 renlies	11 months
🗄 🛑 (01 I Certain infec	tious and parasitic diseases 젿 501 📎			* in	festations should be in	Skin chapter	Chaim	ners	Explanation	o replico	ago
± • (02 II Neoplasms	P 435			ICD-1	10 chapter XII has a cur led. Acute lymphadenit	ious mix of infectiou tis is although lympl	is diseases. No nadenitis NOS is	n-specific skir in chapter IX.	n-limited infection Acute lympha	ons (tinea) are r denitis can be	ot
	03 III Diseases of	f the blood and blood-forming organs a	nd certain disc	orde	retrop	eritoneal (and may res	ult in an ischiorecta	abscess). Lym	ph nodes (un	specified) woul	d not normally b	e tin the
	04 IV Endocrine,	nutritional and metabolic diseases V	399		usage	e of some countries, virt	ually synonymous, i	is not. The ratio	nale used for	this section is e	explained in furti	ner detail in
	05 V Mental and 06 VI Diseases o	f the nervous system @ 535		=	each	subcategory.						
	07 VII Diseases o	of the eve and adneys 22 524			Repl	y Open	Edit	Archive	Delete		02/17/2010 23	:07:58 PST
	08 VIII Diseases	of the ear and mastoid process $\sqrt{25}$			ГР	opeon for changel: Cr	oato class					
	09 IX Diseases o	f the circulatory system 7 37			wi	th name: LA Infections	and	Rol	hert			
	10 X Diseases of	the respiratory system 🖓 30			V int	festations affecting the rents: 12 XII Diseases	e skin, s of the skin	Chalm	ners	Comment	0 replies	1 year ago
	11 XI Diseases o	f the digestive system 🤄 909			an	d subcutaneous tissu	e					
	12 XII Diseases o	of the skin 🖓 3 🖓 2163 🗞			New	nierarchy for chapter						
۲	Draft for ICD-	10 Revision update 🖓 1 🖓 108			Repl	y Open	Edit	Archive	Delete		02/11/2010 03:2	20:00 PST
Ð	Needing a de	cision to be made 🖓 1 🖗 16										
Ð	To be retired	Ф1 Ф88										
Ð	LA Infections a	and infestations affecting the skin 🖓 2	₽467									
Ð	LB Inflammato	ory dermatoses 🖓 1 🖗 242	_									
Ð	LC Metabolic:	and nutritional disorders affecting the s	kin 🏷 1 🖗 142	2								
Ð	LD Genetic, cl	hromosomal and developmental disord	lers affecting t	ne s								
±.	LE Psycholog	ical, psychiatric, sensory and neurolog	ical disorders	affe								
€	LF Disorders (of the epidermis and epidermal append	lages 🏳 1 🖓	288								
Ð	LG Disorders	of the dermis and subcutis 🏸 1 🖓 121	_									
H	LH Disorders	of cutaneous blood and lymphatic ves	els ^{ve} 1 🖗 103	3								

As with BioPortal, notes may include multimedia

WebProtégé - Mozilla Firefox									
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My WebProtégé Classes 🕱 Properties 🕱 Discussions 🕱									
ntology: ICD 11. Login for more features. Search									
<u> </u>									
Class Tree	Notes for ICD11 (A15-	-A19) Tuberculosis	- *						
🔆 Create class 💢 Delete class	New Topic Reply Co	ollapse <previous next=""> D</previous>	isplaying page 1 of 1 pages						
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🖨 🗢 ICD11	Example	Tania Tudorache	03/24/2009 12:12:22 PDT						
🖨 🗢 ICD11 (A00-B99) Certain infectious and parasitic diseases (CHAPTER I) 🖗	Here is a picture fr	om Wikipedia:							
🖶 🖲 ICD11 (A00-A09) Intestinal infectious diseases	ANDER Exemple	Tania Tudanaha	00/01/2020 40/14/22 DDT						
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ICD11 (B95-B97) Bacterial viral and other infectious agents									
ICD11 (B99) Other infectious diseases									
				$\mathbf{\mathbf{v}}$					

Integration of Ontology Authoring, Publishing, and Peer Review



NCBO will support the complete ontology lifecycle



WINDOW FRAME									
ATHENA H	ypertension Advisory	7			References Sources 🔝				
Patient Name	*******	(X <u>View Patient Summary</u>							
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SYNTHETIC PATIENT DATA

- 21

The task: guideline-based patient management



A handful of encoded guidelines gives you, well, a handful of encoded guidelines

ATHENA Hypertension









ATHENA Diabetes



ATHENA Renal Disease ATHENA Opioid Therapy





GLINDA Task–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 lifesciences 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