

# Overview of EHR phenotyping – successes, challenges, and examples

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AMIA 2015 NLP Tutorial

## **Disclaimers**

I receive funding from:

- NIH: NLM, NHGRI, NIGMS, NCI, NCATS
- Reynolds Foundation (Geriatrics Education)
- National Board of Medical Examiners



# **Outline for my block**

- Intro to use of the EHR for genomic research
- NLP for EHR phenotyping
  - diseases/traits
  - Drug-response phenotypes
- NLP for medical education



## **EHR adoption increasing faster than anticipated**



## **Genomic achievements since the Human Genome Project**





## <u>The Synthetic Derivative</u> A de-identified and continuously-updated image of the EMR: 2,358,760 subjects



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## Using clinical notes to enhance knowledge



# Extracted detailed smoking history from clinical narratives

#### Predicted vs Physician-Calculated Tobacco Exposure From Social History



Travis Osterman AMIA Weds 10:30-12 session

Physician-calculated Tobacco Exposure (Pa&-Year)

# **Example - Clinical Notes**

CC: SOB

HPI: 71 yo woman h/o DM, HTN, Dilated CM/CHF, Afib s/p embolic event, chronic diarrhea, admitted with SOB. CXR pulm edema. Rx'd Lasix.

All: none

Meds Lasix 40mg IV bid, ASA, Coumadin 5, Prinivil 10, glucophage 850 bid, glipizide 10 bid, immodium prn

A/P:

- increase lasix to 80 bid
- maintain sao2 > 92% per RT protocol
- no fever and wbc wnl, so cont immodium prn
- dm2: ccm

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### **The Synthetic Derivative**

A de-identified and continuously-updated image of the EMR: 2,358,760 subjects

### **BioVU**

Subjects with DNA: 197,330

- Dense (GWAS-level)
  - genotypes: ~20,000
- Exome chip data: ~36,000

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## **EHR Phenotyping**





## **Our "simple" example: Hypertension**

Multiple components are better (and blood pressure is the worst)



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# The "demonstration project"

- Are genotype-phenotype relations replicated in BioVU?
- Genotype "high-value" SNPs in the first 10,000 samples accrued.
  - 21 established loci (>1 SNP for some)
  - in 5 diseases with known associations:

Atrial fibrillation Crohn's disease Multiple Sclerosis Rheumatoid arthritis Type II Diabetes

 Develop "electronic phenotype algorithms" to identify cases and controls



# **RA – Case Definition Evolution**

#	Definition	# Cases	Problem
1	ICD9 codes for RA + Medications (only in problem list)	371	Found incomplete problem lists
2	Same as above but searched notes	411	Patients billed as RA but actually other conditions, overlap syndromes such as psoriatic arthritis, juvenile RA
3	Above + require text "rheumatoid arthritis" and small list of exclusions	358	Overlap syndromes with other autoimmune conditions, conditions in which physicians did not agree
4	Above + exclusion of other inflammatory arthritides	255	PPV = 97%; a few "possible RA" or family history items remained



# **Final RA case definition**

#### ICD 9 codes (any of the below)

- 714 Rheumatoid arthritis and other inflammatory polyarthropathies
- 714.0 Rheumatoid arthritis
- 714.1 Felty's syndrome
- 714.2 Other rheumatoid arthritis with visceral or systemic involvement

#### AND

#### Medications (any of the below)

methotrexate [MTX][amethopterin] sulfasalazine [azulfidine]; Minocycline [minocin][solodyn]; hydroxychloroquine [Plaquenil]; adalimumab [Humira]; etanercept [Enbrel] infliximab [Remicade]; Gold [myochrysine]; azathioprine [Imuran]; rituximab [Rituxan] [MabThera]; anakinra [Kineret]; abatacept [Orencia]; leflunomide [Arava]

#### AND

#### Keywords (any of the below)

rheumatoid [rheum] [reumatoid] arthritis [arthritides] [arthriris] [arthristis] [arthritus] [arthrtis] [artritis]

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=Narrative components

## Final RA case definition - 2

#### AND NOT ICD 9 codes (any of the below)

- 714.30 Polyarticular juvenile rheumatoid arthritis, chronic or unspecified
- 714.31 Polyarticular juvenile rheumatoid arthritis, acute
- 714.32 Pauciarticular juvenile rheumatoid arthritis
- 714.33 Monoarticular juvenile rheumatoid arthritis
- 695.4 Lupus erythematosus
- 710.0 Systemic lupus erythematosus
- 373.34 Discoid lupus erythematosus of eyelid
- 710.2 Sjogren's disease
- 710.3 Dermatomyositis
- 710.4 Polymyositis
- 555 Regional enteritis
- 555.0 Regional enteritis of small intestine
- 555.1 Regional enteritis of large intestine
- 555.2 Regional enteritis of small/large intestine
- 555.9 Regional enteritis of unspecified site
- 564.1 Irritable Bowel Syndrome
- 135 Sarcoidosis
- 696 Psoriasis and similar disorders
- 696.0 Psoriatic arthropathy
- 696.1 Other psoriasis and similar disorders excluding psoriatic arthropathy
- 696.8 Other psoriasis and similar disorders
- 099.3 Reiter's disease
- 716.8 Arthropathy, unspecified
- 274.0 Gouty arthropathy
- 358.0 myasthenia gravis
- 358.00 myasthenia gravis without acute exacerbation
- 358.01 myasthenia gravis with acute exacerbation
- 775.2 neonatal myasthenia gravis

- 719.3 Palindromic rheumatism
- 719.30 Palindromic rheumatism, site unspecified
- 719.31 Palindromic rheumatism involving shoulder region
- 719.32 Palindromic rheumatism involving upper arm
- 719.33 Palindromic rheumatism involving forearm
- 719.34 Palindromic rheumatism involving hand
- 719.35 Palindromic rheumatism involving pelvic region and thigh
- 719.36 Palindromic rheumatism involving lower leg
- 719.37 Palindromic rheumatism involving ankle and foot
- 719.38 Palindromic rheumatism involving other specified sites
- 719.39 Palindromic rheumatism involving multiple sites
- 720 Ankylosing spondylitis and other inflammatory spondylopathies
- 720.0 Ankylosing spondylitis
- 720.8 Other inflammatory spondylopathies
- 720.81 Inflammatory spondylopathies in diseases classified elsewhere
- 720.89 Other inflammatory spondylopathies
- 720.9 Unspecified inflammatory spondylopathy
- 721.2 Thoracic spondylosis without myelopathy
- 721.3 Lumbosacral spondylosis without myelopathy
- 729.0 Rheumatism, unspecified and fibrositis
- 340 Multiple sclerosis
- 341.9 Demyelinating disease of the central nervous system unspecified
- 323.9 transverse myelitis
- 710.1 Systemic sclerosis
- 245.2 Hashimoto's thyroiditis
- 242.0 Toxic diffuse goiter
  - 443.0 Raynaud' s syndrome

#### AND NOT

#### Keywords (any of the below)

yuvenile [juv] rheumatoid [rheum] [reumatoid] [rhumatoid] arthritis [arthritides] [arthriris] [arthristis] [arthritis] [arthr

# What we learned - Finding phenotypes in the EMR





# A common general approach to phenotyping



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Denny. PLoS Comp Bio 2012

## Validating EMR phenotype algorithms (Using first 10,000 patients in BioVU)

Disease	Methods	Definite Cases	Controls	Case PPV	Control PPV
Atrial fibrillation	ibrillation NLP of ECG impressions		1695	98%	100%
	ICD9 codes CPT codes	<u>Common t</u>	Common themes:		
Crohn's Disease	ICD9 codes Medications (NLP)	NLP – 5/5	odes – 5/5 /5	100%	100%
Type 2 Diabetes	ICD9 codes Medications (NLP) NLP exclusions Labs	Meds – 4/5 Labs – 2/5	Meds – 4/5 Labs – 2/5		100%
Multiple Sclerosis	ICD9 codes or text diagnosis	66	1857	87%	100%
Rheumatoid Arthritis	ICD9 codes Medications (NLP) NLP exclusions	170	701	97%	100%

NLP = Natural language processing



## **Results**



disease	marker	gene / region			
Atrial fibrillation	rs2200733	Chr. 4q25	<b>-</b> -	<b>—</b>	
Atha hormation	rs10033464	Chr. 4q25	<b>_</b>	_	
	rs11805303	IL23R			
	rs17234657	Chr. 5		- <b>\</b>	
Crohn's disease	rs1000113	Chr. 5	<b>_</b>		
	rs17221417	NOD2	│ <b>─∎</b> -♦	_	
	rs2542151	PTPN22	<b></b>		
	rs3135388	DRB1*1501			
Multiple sclerosis	rs2104286	IL2RA			
	rs6897932	IL7RA			
	rs6457617	Chr. 6		<b></b>	
Rheumatoid arthritis	rs6679677	RSBN1			
	rs2476601	PTPN22		-	
	rs4506565	TCF7L2	<b></b> _		
	rs12255372	TCF7L2	<b></b>		
	rs12243326	TCF7L2			
Type 2 diabetes	rs10811661	CDKN2B			
.,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,	rs8050136	FTO	_ <b></b>		
	rs5219	KCNJ11			
	rs5215	KCNJ11	<b>-</b>		
	rs4402960	IGF2BP2	_ <b>_</b> _		
		0 5	1 0	20	50
		0.5	1.0	2.0	5.0
VANDERBILT VUNIVERSITY			Odds Ratio	<b>D</b> <sup>1</sup>	
MEDICAL CENTER				Ritchie	et al., AJHG 2010

## Identifying cases with precision from the EMR

### requires structured and unstructured information

	RA	MS	CD	T2D
Had ICD-9 codes:	3.9%	1.8%	1.8%	17.3%
Met algorithm definition:	2.7%	1. 2%	1.6%	9.7%
Accuracy of ICD9 codes:	69%	66%	89%	56%



# **Hypothyroidism algorithm**



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## Hypothyroidism Validation

Site	Case PPV (%)	Control PPV (%)	
<b>Group Health</b>	98	100	
Marshfield	91	100	Same algorithm,
Mayo Clinic	82	96	deployed at five
Northwestern	98	100	sites
Vanderbilt	98	100	
All sites (weighted)	92.4	98.5	
ANDERBILT VUNIVERSITY MEDICAL CENTER		More exa secondary code evic more com	amples of patients with hypothyroidism without dence - only findable via plex NLP not in algorithm

## Hypothyroidism: "No-Genotyping" GWAS



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Denny et al., AJHG 2011

# Sharing algorithms: <u>PheKB.org</u> This is not just genetics!



a knowledgebase for discovering phenotypes from electronic medical records

Phenotypes Implementations Groups Institutions

#### What is the Phenotype KnowledgeBase?



The reuse of data from electronic medical records (EMRs) and other clinical data systems holds tremendous promise for improving the efficiency and effectiveness of health research. Clinical data in the EMR is a potential source of rich longitudinal data for research, and the recent government efforts to promote the use of EMRs in the clinical

setting may further pro healthcare system. As usable data from these

One such effort by the Network (eMERGE) ha routine clinical care us sufficient positive and genome-wide associat

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information (diagnoses, medications, laboratory tests) used to define phenotypes language processing has also been shown to improve case identification rates.\*

PheKB is an outgrowth of that validation effort and provides a collaborative enviro phenotype algorithms. On this site you can:

- View existing algorithms
- Enter or create new algorithms
- Collaborate with others to create or review algorithms
- · View implementation details for existing algorithms

Phenotype algorithms can be viewed by data modalities or methods used:

#### eMERGE, PCORnet, NIH Collaboratory, PGRN, PGPop

Most Recent Phenotypes

White Blood Cell Indices

Type II Diabetes Mellitus

Red Blood Cell Indices

Peripheral Arterial Disease

Login Register

- 381 active users, 48 institutions
- 21 publicly available phenotypes, 67 phenotypes in development
- There are 165 implementations
- social networking features; versioning; etc.
  - Data dictionary and data set validation

VAND

# What EHR data do we need for research?

Evaluation of EHR data types used in 92 phenotypes posted on PheKB.org (median positive predictive value = 96%)

	Number of phenotypes utilizing			
Data modalities or		these feature	S	
methods	Public	Non-Public	Percent of	
	(N=30)	(N=62)	Total	
ICD-9 Codes	27	37	70%	
Medications	25	32	62%	
Natural Language	21	21	46%	
Processing				
CPT Codes	14	24	41%	
Laboratory/test results	14	21	38%	

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## ICD, Meds, and (nonnegated) Text mentions identify different counts of possible cases for different diseases



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Wei-WQ et al. JAMIA 2015

# **ROC curves for ICD-9, primary notes, and specific medications for 10 diseases (1750 reviewed charts)**



# How to find cases: general maxims

- ICD9s (and CPT) very useful but not sufficient
  - nearly all algorithms use them
  - good for sensitivity but not necessarily PPV
- Labs
- Meds marker of disease and severity not usually that helpful by itelf
- NLP often confirmatory and refining, unless dealing with rare diseases



#### PPV and sensitivity over 10 diseases by counts of occurrence



Wei-WQ et al. JAMIA 2015

## **Completed eMERGE GWAS**

## All of these are on PheKB and will eventually be public

#### Diseases

- Dementia
- Cataracts
- Autoimmune Hypothyroidism
- Diverticulosis/diverticulitis
- Type 2 Diabetes
- Diabetic retinopathy
- Herpes zoster
- PheWAS
- Peripheral Arterial Disease
- Venous Thromboembolism
- Glaucoma
- Ocular hypertension
- Abdominal Aortic Aneurysm
- Colon polyps

# **bold**=GWAS completed with significant results

#### Endophenotypes

- PR Duration
- QRS Duration
  - HDL/LDL
- height

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- white blood cell counts
- red blood cell counts
- Cardiorespiratory Fitness
- ESR levels
- Platelet levels

#### Selected consortia contributions

- Height
- QTc
- Rheum. Arthritis
- Myocardial Infarction Genetics Consortium
- Intl. Mult Sclerosis Genet. Consort.
- Genomic Investigation of Statin Therapy

#### Pharmacogenomic phenotypes

- ACE inhibitor cough
- Heparin induced thrombocytopenia
- Resistant hypertension
- Drug Induced Liver Injury
  - C. difficile colitis

## Are algorithms portable, part II: Rheumatoid Arthritis

- Previously published logistic regression model
  - Developed at Partners Healthcare
  - ICD9, Labs, Meds, NLP
  - Tested at Northwestern and Vanderbilt
- Is the signature the same across
  - Differing healthcare environments and EHR systems?
  - Differing NLP systems (regular expression vs. out-of-the-bag KnowledgeMap)?

Site	n	PPV	Sensitivity	AUC
<b>Original</b> (Partners)	400	94%	63%	95%
Northwestern	390	87%	60%	92%
Vanderbilt	376	95%	57%	95%



Liao et al. Arth Care Res 2010 Carroll et al. JAMIA 2012 <sup>33</sup>

## **Using Machine Learning for Phenotyping**





## **GWAS of ACEI-cough (no prior GWAS)**

ACEI-cough (NLP of allergy sections)



**eMERGE phenotype** – document ACEI cough allergies in the EMR

Our GWAS: OR=1.3 [95%CI: 1.2-1.4]

eMERGE replication (same algorithm): OR=1.32 [1.01-1.70]

**European replication** – people changed from ACEI to an ARB (reason unknown)

European replication OR=1.15 [1.01-1.30]

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Mosley et al. TPJ 2014
## Part 3 – Using NLP to assess and improve Medical Education



# Part #1: Assessing LIAISON COMMITTEE ON MEDICAL EDUCATION Curricula

- LCME and ACGME require increasing documentation of curriculum objectives, coverage, and student patient experiences
- Accreditation standards specific content, competencies, amount of training, etc. for periodic reviews
  - Patient case mix
  - Topics taught
  - Response to certain trans-course "hot topics" over time women's health, substance abuse, etc.



# Traditional Medical Curriculum





# Traditional Medical Education Model





## **Guiding questions**

- Faculty: "I am teaching about congestive heart disease – what have students already learned about this?"
- Students: Studying immunoglobulins, need to find relevant prior concepts like splicing
- Administrators: Where do we cover large concepts, like geriatrics?







#### Course Links[edit]: Gross Anatomy sessions | Cell Biology sessions

Labels: Exam or Quiz

Physiology Gross Anatomy Cell Biology

#### Unit #1 $\mathbf{T}$

Date	Time	Location	Title	Lecturer	Document(s)
10/25	08:00 AM - 08:30 AM	LH 202	Introduction	Arthur F Dalley	
10/25	08:30 AM - 10:30 AM	LH 202	Introduction to Anatomical Donations Program and In-Lab Memorial svc. Intro to Gross Anatomy Lab, Safety and Techology	Arthur F Dalley	
10/25	10:30 AM - 12:00 PM	LH 202	Layered & Segmented Structure of body; Intro to Nerves & Nerve Clasification; Simple Spinal n.	Arthur F Dalley	
10/25	01:00 PM - 02:00 PM	LH 202	(Embryo) Neuromuscular Development 1	Lillian B Nanney	
10/25	02:00 PM - 05:00 PM	IIS 10th Floor	GA Lab: Removal of Skin and Subcutaneious Tissue of Back; Superficial Muscles of the Back and Canial Nerve XI	Arthur F Dalley	
10/26	08:00 AM - 09:00 AM	LH 202	(Embryo) Neuromuscular Development 2	Lillian B Nanney	
10/26	09:00 AM - 10:00 AM	LH 202	Types of Muscle Action and Movements; Freely Moveable Joints	John S Halle	
10/26	10:00 AM - 12:00 PM	IIS 10th Floor	GA Lab: Scapular Region	Arthur F Dalley	
10/29	08:00 AM - 09:00 AM	LH 202	Vertebral Column; Postural Muscles; Spinal Cord and Its Environment	Arthur F Dalley	
10/29	09:00 AM - 10:00 AM	LH 202	Neuromuscular Phys # 1: Membrane Transport; Fluid Compartments; Osmosis	Al George	
10/29	01:00 PM - 02:00 PM	IIS 10th Floor	GA Lab: Deep Back (Perform Laminectomy)	Arthur F Dalley	
10/30	08:00 AM - 09:00 AM	LH 202	Overview of Lymphatic System; Principles of Collateral Circulation	Lillian B Nanney	
10/30	09:00 AM - 11:00 AM	IIS 10th Floor	GA Lab: Complete Dissection of Deep Back; Pectoral Region, Including Removal of Skin from Arm (Excluding Female Breast)	Arthur F Dalley	
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### **Document Processing**







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chf	This state causes congestion of the tissues, leading	
(ex: heart failure and digoxin)	It produces the clinical syndrome of dyspnea, per,	
Search	an abnormality of myocardial function is responsible for the failure of the heart to pump at a rate commensurate with the requirements of metabolizing tissues. <b>CHF</b> is encountered frequently by the clinician; it accounts for 2? of all hospital	
	admissions and carries a 5-year survival rate of <50?.	
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View search history	A. Myocardial Disease (pump defect)	
	1. Coronary heart disease myocardium is impaired by ischemia	
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	amyloidosis	
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<ul> <li>Find Similar Documents</li> </ul>	B. Excessive Workload due to:	
	1. Increased resistance to ejection which can be due to pressure overload, hypertension, aortic or pulmonary stenosis, or hypertrophic cardiomyonathy	
	2. Increased stroke volume; volume overload which can be due to aortic, mitral or tricuspid insufficiency or congenital left-to-	
	right shunts.	
	(abnormal shunt between an artery and vein which increases venous return and decreases oxygen to shunted area)	~
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Search later tend to have liver problems as the major clinical feature. Although at one time thought to represent distinct disord	lers,
different affected members in the same family may present at different ages with either predominant renal or liver about	rmalities,
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polyuria and dehydration (especially during intercurrent illness).	
Find Similar Documents The kidneys may be sufficiently enlarged to result in a palpable mass. Detenorating renal function is inevitable, ar	id patients
As implied above, patients presenting in later childhood and early adulthood usually present with signs of liver in	volvement
particularly portal hypertension, which may result in hepatosplenomegaly and bleeding esophageal varices.	, 01001110110,
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# How do we find broad concepts like "geriatrics" or "women's health"?





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Document-specific Tools:         ■ Get Original Document ा         ■ Get Original as PDF ा         ■ Similar PubMed Articles	Insight into aging of the brain and Alzheimer's disease is the result of a massive r disease, in particular, represents a rapidly evolving area of research; new and often purpose of this set of lectures is to provide you with the requisite background informa assimilate new findings as they are generated, and to provide you with a synopsis of b understanding of Alzheimer 's disease	esearch effort; work on Alzheimer's conflicting data are generated daily; the tion necessary to allow you to continue to oth an historical and current (albeit changing)
<ul> <li>Find Similar Documents</li> <li>NEW:</li> </ul>	* Recommended reading:	
	" DeBaggio, Thomas. Losing My Mind. The Free Press, 2002	
	" Bayley, John. Elegy for Iris. Picador Press, 1999	
	" Jozefowicz and Holloway: Case 26 "The Gopher Hunter"	
	I. Maturational events which occur normally in the nervous system	

100

#### How well does KM find metaconcepts?

- Identified gold standard set of 380 documents as containing high, medium or low relevance to each topic
- Used KM to generate a variable number of subconcepts for each broad concept and calculated a relevance score for each document.

Торіс	ROC area
Genetics	0.98
Women's Health	0.93
Dermatology	0.95
Radiology	0.97



### **Finding broad curricular topics**

 Used for LCME, creating/rearranging courses, revising curriculum



#### Using to infuse Geriatrics in the curriculum:

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#### **Part #2: Assessment in Clinical Years**

- Testing based: USMLE, NCLEX, Residency Board Exams
- Experience Based:
  - ACGME and RRC
  - Nursing requirements
- Both current methods tend to aggregate at high levels
- Experience is an important part of competency



### Learning Portfolio – leveraging EMR to capture experience





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	2007-05-	Internal Medicine Clinic Visit	2007-05-16			
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Concept Reports	2007-05-	Internal Medicine Clinic Resident Acute Clinic Visit	2007-05-16			
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Outpt. visit	Mechanism of Action		
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Satellink	KnowledgeMan documents back to top	erm Definitions	
Scratch cer	Kine in the grant of the second	)rug information (Encorates Levi Comp)	
Un ToDate	Donepezil (Aricept) Introduced 1997; 1998 Title=39 wk Title= Donepezil (Aricept) Title=Reference		
Patient Lists	<ul> <li>maintained above baseline Head Rivastigmine v Donepezil 12 week trial, 111 patients, mild to mode of the Normal Sector (Normal Sector) and functional outcomes</li> </ul>	Aedical references (Up IoDate, PubMed,	
Consults	🖻 Lunch-and-Learn: Prescribing for Older Adults   POGOe Geriatrics Content (Geriatrics)	/ledlinePlus)	
lnpt. censu	titrated to 10mg of donepezil (Aricept) daily. Her daughter is now concerned about worsening Don (Aricent) can worsen or precipitate urge incontinence incontinence isConsider stopping donepezit	All curriculum content (VLISM_MPH	
Outpt. visit	t Consider drug-drug interactions Donepezil has an opposite (pro-cholinergic) effect Arch Intern Med		
Patients Vie	🖞 🖻 Autonomics: David Robertson   Pharmacology (VMS II)	esidencies, etc)	
Donny Los	Donepezil Donepezil aka Aricept • A	Additional local resources (POGOe,	
MD	E Chapter 30 - Dementia Joshua Charles Denny   Geriatrics Review Syllabus (Geriatrics)	Geriatric Review Syllabus)	
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# Tracking experience: Vanderbilt Core Clinical Curriculum (VC3)

25 Core Clinical Problems (CCP)

Abdominal pain Abnormal uterine bleeding Abnormal vaginal discharge Abnormalities of mood Altered mental status Back pain Breast disease Chest pain Cough Dysuria Fever GI bleeding

Headache Jaundice Loss of consciousness Obesity Pelvic pain Pharyngitis Rash Seizures Shock Shortness of breath Substance abuse Trauma Weight loss

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Student view of how many VC3 topics they've completed. (Teachers can see this also.)

Mapping of a note to a VC3 topic happens manually and automatically for high scoring documents.

#### Learning Objectives

-			
otes	Learning Objective	Date Recorded	Event Recorded
lentors	Abdominal Pain [Find matching notes]	10/17/2008 11/1/2008	Pediatric Surgery Consultation Note Medical Student Admission History and Physical
eflections	Altered Mental Status		None recorded
edure Logs	Back Pain [Find matching notes]	10/17/2008	Return Clinic Visit Progress Note
	Breast Disease		None recorded
lotes	Chest Pain	5/9/2008	Medical Student Admission History and Physical
ning Objectives	[Find matching notes]	5/23/2008 10/19/2008	Medical Student Admission History and Physical Medical Student Admission History and Physical
viedgeMan	Coma		None recorded
пецдемар	Cough [Find matching notes]	10/20/2008 10/20/2008	Medical Student Admission History and Physical Medical Student Admission History and Physical
rences	Depression	11/2/2008	Medical Student Progress Note
	[Find matching notes]	11/2/2008	History and physical
ch for:	Dysuria [Find matching notes]	11/2/2008 11/2/2008 11/2/2008	Pediatric Infectious Disease Initial Consultation Medical Student Admission History and Physical Clinic Visit
tients	Fever [Find matching notes]	10/17/2008 10/17/2008	Medical Student Admission History and Physical Progress Note Daily Progress Note
ainees' Notes edgeMap	Gastrointestinal Bleeding [Find matching notes]		None recorded
	Heart Murmurs		None recorded
	Jaundice [Find matching notes]	11/2/2008 11/2/2008	History and physical Progress Note Daily Progress Note
	Menstrual abnormalities		None recorded
	Mood Disorder		None recorded
	Pelvic Pain		None recorded
	Phanynaitic		None recorded

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#### **Part #3: Evaluating and integrating**

#### Study 1: Curriculum vs. Notes



# Compare content, identify discrepancies



### **Coverage of VC3 Topics**

#### 300k student notes 15k lecture documents



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Denny et al. AMIA 2010

### **Coverage of Infectious Diseases**





Denny et al. AMIA 2010



- Current Email Advisors:
  - Advanced directives (pts > 65, if they don't mention them)
  - Altered mental status (must say AMS concept in key section of note)

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Denny et al. JBI 2015

#### **Step 1.** Student sees a patients, writes a note

CHIEF COMPLAINT: confusion, weakness, and lethargy HISTORY OF PRESENT ILLNESS: Mrs. X is a 70 year old female with metastatic undifferentiated carcinoma, likely lung in origin, who was recently discharged from the hospital s/p left femoral fracture and biopsy due to a fracture who now presents with increasing confusion, weakness, and lethargy.

PHYSICAL EXAMINATION: General: waxing and waning alertness,

SUMMARY: This is a 72 year old female with metastatic lung carcinoma admitted for delirium most likely secondary to hypercalcemia.

ASSESSMENT AND PLAN:

1. Hypercalcemia Hyperparathyroidism... malignancy...

6. Disp -Will keep hospitalized until altered mental status improves...



Denny et al. JBI 2015

#### **Step 2.** Portfolio finds AMS concepts found in note

```
CHIEF COMPLAINT: confusion, weakness, and lethargy
HISTORY OF PRESENT ILLNESS: Mrs. X is a 70 year old female with
metastatic undifferentiated carcinoma, likely lung in origin, who
was recently discharged from the hospital s/p left femoral
fracture and biopsy due to a fracture who now presents with
increasing confusion, weakness, and lethargy.
PHYSICAL EXAMINATION: General: waxing and waning alertness,
SUMMARY: This is a 72 year old female with metastatic lung
carcinoma admitted for delirium most likely secondary to
hypercalcemia.
ASSESSMENT AND PLAN:
1. Hypercalcemia Hyperparathyroidism... malignancy...
6. Disp -Will keep hospitalized until altered mental status
```

improves...


#### CHIEF COMPLAINT: confusion, weakness, and lethargy



Step 3. Portfolio finds related curriculum documents and emails the student

male with origin, who

You are getting this email as part of a project to improve your understanding of **altered mental status**. This email is generated based on your note: <u>Medical Student Admission History and Physical, written</u> on 2011-01-15 19:42:15.

### Key facts about Altered Mental Status:

•The differential diagnosis of altered mental status is extensive including dementia, delirium, substance induced, drug side effects, infection, intracranial lesions or strokes, trauma, and metabolic entities such as liver disease or hypoglycemia.

•Alzhiemer's disease, vascular dementia, and dementia with Lewy bodies are the most common forms of degenerative dementias seen in late life.

#### KM documents most like yours:

•Typical Laboratory Results in the Differential Diagnosis of Hypercalcemia | Joshua Charles Denny | Geriatrics Review Syllabus (Geriatrics)

•Hypercalcemia | Natasha Janelle Schneider | Outpatient Medicine Curriculum (Core Lecture Series)

•Fluid Management for Students | Kyle Bertram Brothers | Pediatrics (VMS III)

Pharmacological Concepts | Joseph A Awad | Pharmacology (VMS II)

### Other searches that may be relevant to this patient:

•Differential diagnosis of metobolic (liver ds, electrolytes, gluocose abnormalities) as causes of AMS. (4 overlapping concepts)

- •Differential diagnosis of delerium as a cause of AMS. (2 overlapping concepts)
- Signs and symptoms of AMS (2 overlapping concepts)
- Evaluation of AMS (1 overlapping concepts)



## How does it actually work?

name	Altered Mental Status
classifications	VMS3, VMS4 (type of people to evaluate)
eval_saved_search_num	446 (list of CUIs related to AMS, built on website)
eval_wordsearch	AMS, altered mental status (extra words to search, nonnegated)
eval_limit_note_sections	1 (searches only "high value" note sections)
feedback_saved_searches	426,426,428,429,430,431
	1,2,4 (what curricula to use; documents are ranked via TF-IDF from very broadly defined AMS
feedback_saved_searches_programs	CUIs in the saved searches)
email_matching_concepts	TRUE (tell the student what we found in their note)
min_age	18
max_age	9999
min_score_to_keep	1
once_per_patient	1
km_docs_to_send	74 (a list of highly relevant AMS resources)
email_subjectline	Altered Mental Status
email_header	You are getting this email as part of a project to improve your learning on <b>altered mental status</b> .
email_instruction	Key facts about Altered Mental Status: <ul></ul>

# **Summary**

- EHR-linked DNA biobanks can be used for genomic and pharmacogenomic discovery. They can be cost efficient and fast.
- Best algorithms to find phenotypes include codes, labs, meds, and/or NLP through combination of Boolean and/or machine learning approaches – these algorithms are placed on PheKB
- NLP is often confirmatory for phenotypes
- NLP can also be used to improve cataloging of medical education content and tracking of trainee experiences

