Clinical Phenotyping from EHRs: Opportunities and Challenges

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Duke University School of Nursing

Presented at:
NIH Symposium: Linking Disease Model Phenotypes to Human Conditions
Rockville, MD
September 10, 2015

Opportunities

- Clinician evaluation of disease & status
  - Diagnoses
  - Problems
  - Clinical notes
- Treatments, procedures, medications
- Labs
- Patient controlled data
  - Patient portals / Patient reported outcomes
  - Biometric uploads

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The Office of the National Coordinator for Health Information Technology (ONC)

The Office of the National Coordinator for Health Information Technology (ONC) is at the forefront of the administration's health IT efforts and is a resource to the entire health system to support the adoption of health information technology and the promotion of nationwide health information exchange to improve health care. ONC is organizationally located within the Office of the Secretary for the U.S. Department of Health and Human Services (HHS).

ONC is the principal Federal entity charged with coordination of nationwide efforts to implement and use the most advanced health information technology and the electronic exchange of health information. The position of National Coordinator was created in 2004, through an Executive Order, and legislatively mandated in the Health Information Technology for Economic and Clinical Health Act (HITECH Act) of 2009.

ONC's mission includes:

- Promoting development of a nationwide Health IT infrastructure that allows for electronic use and exchange of information that:
  - Ensures secure and protected patient health information
  - Improves health care quality
  - Reduces health care costs
  - Informs medical decisions at the time/place of care
  - Includes meaningful public input in infrastructure development
  - Improves coordination of care and information among hospitals, labs, physicians, etc.
  - Improves public health activities and facilitates early identification/rapid response to public health emergencies
  - Facilitates health and clinical research
  - Promotes early detection, prevention, and management of chronic diseases
  - Promotes a more effective marketplace
  - Improves efforts to reduce health disparities
- Providing leadership in the development, recognition, and implementation of standards and the certification of Health IT products;
- Health IT policy coordination;
- Strategic planning for Health IT adoption and health information exchange; and
- Establishing governance for the Nationwide Health Information Network.

Federal Register Notice

August 13, 2010: Statement of Organization, Functions, and Delegations of Authority: Office of the National Coordinator for Health and Information Technology; Correction [PDF - 40 KB]

December 1, 2009: Organization, Functions, and Delegations of Authority: Office of the National Coordinator for Health Information Technology [PDF - 49 KB]

Celebrating the First Anniversary of the HITECH Act and Looking to the Future: February 2010 [PDF - 852 KB]

Feature

Resources for EHR Incentive Programs

Register now for Medicare and Medicaid EHR Incentive Programs, and learn more about how to become a meaningful user of EHRs.

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Health IT Journeys
Stories from the road.

Page last updated on: 12/8/2010

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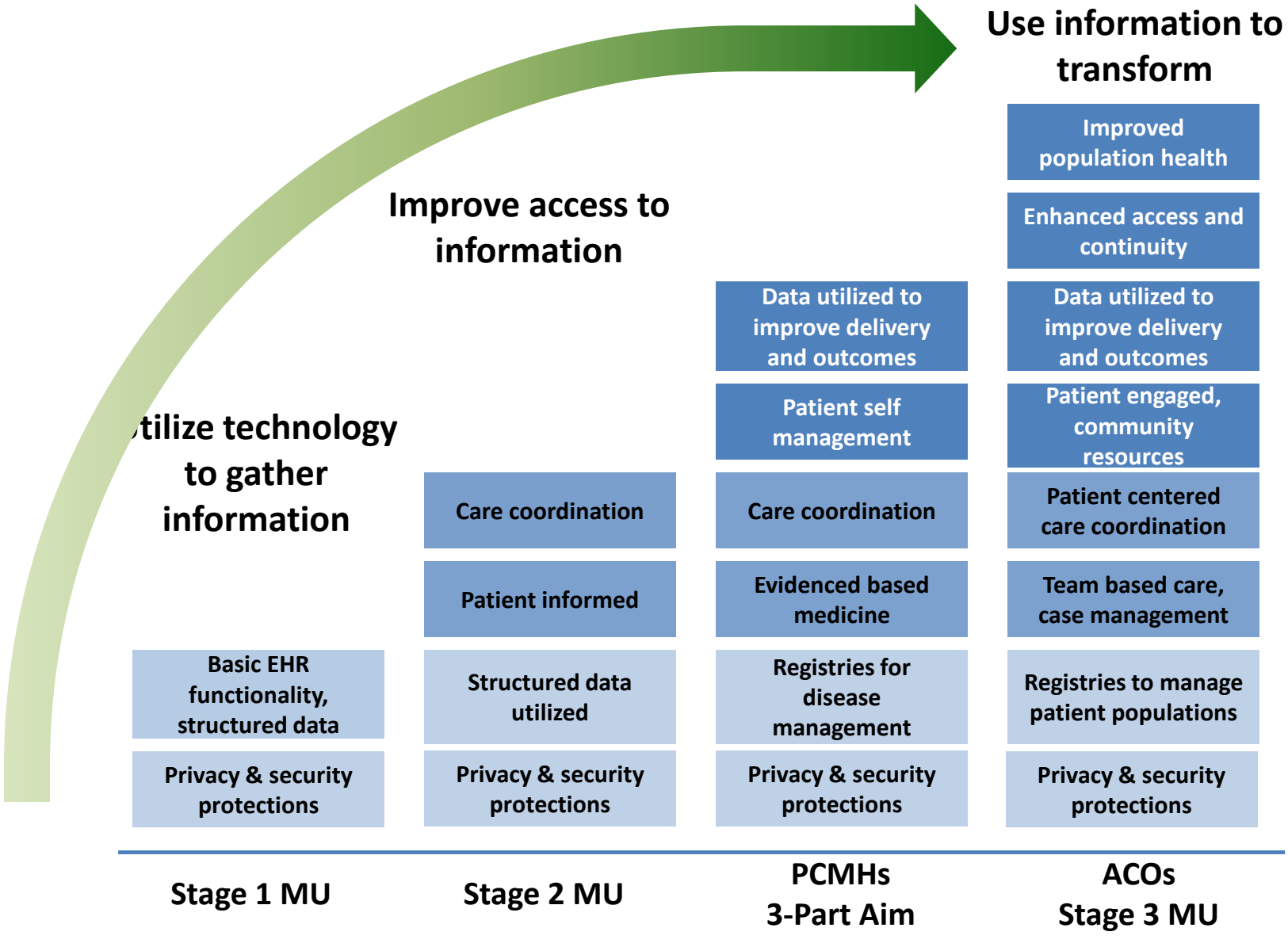
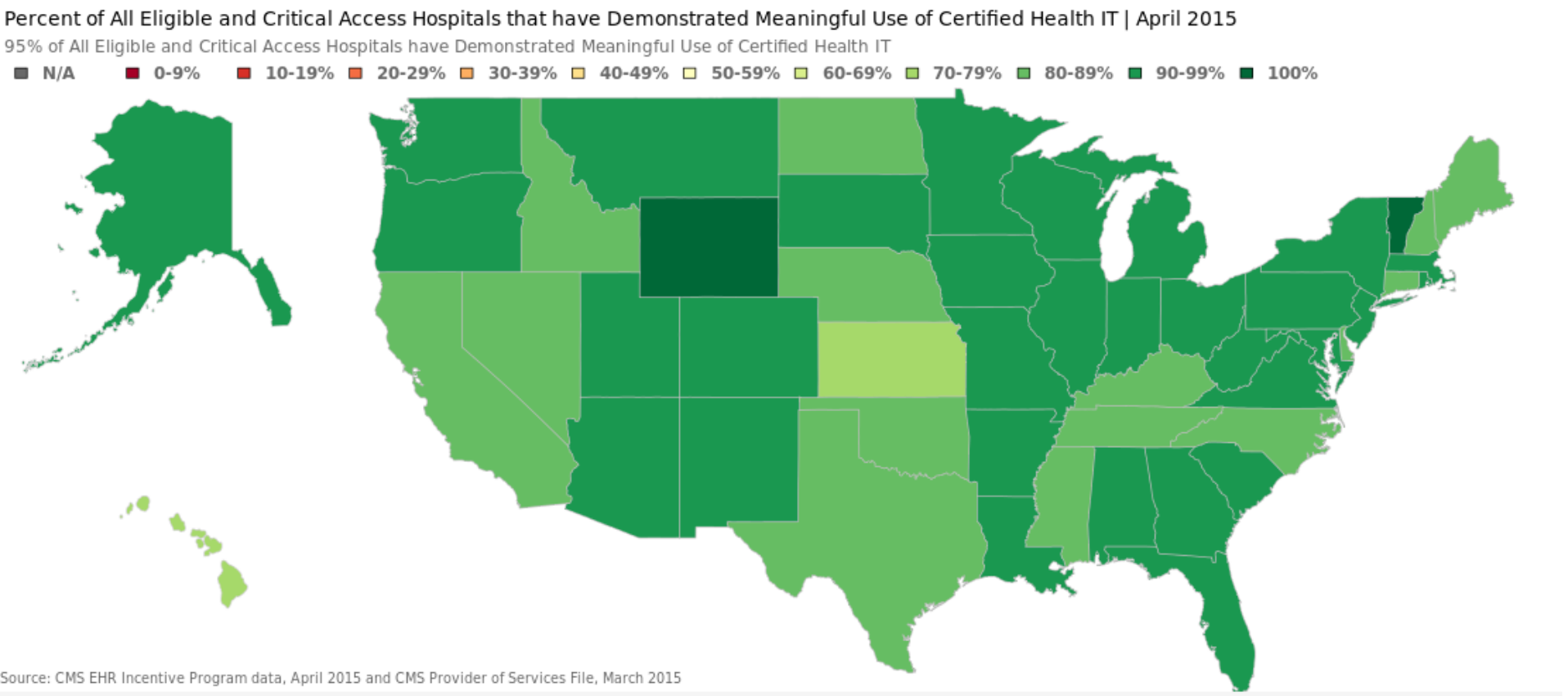

Use information to transform
Improve access to information
tilize technology to gather information
Improved population health
Enhanced access and continuity
Data utilized to improve delivery and outcomes
Data utilized to improve delivery and outcomes
Patient self management
Patient engaged, community resources
Care coordination
Care coordination
Patient centered care coordination
Patient informed
Evidenced based medicine
Team based care, case management
Basic EHR functionality, structured data
Structured data utilized
Registries for disease management
Registries to manage patient populations
Privacy & security protections
Privacy & security protections
Privacy & security protections
Privacy & security protections
Stage 1 MU
Stage 2 MU
PCMHs 3-Part Aim
ACOs Stage 3 MU

Hospitals Demonstrating Meaningful Use

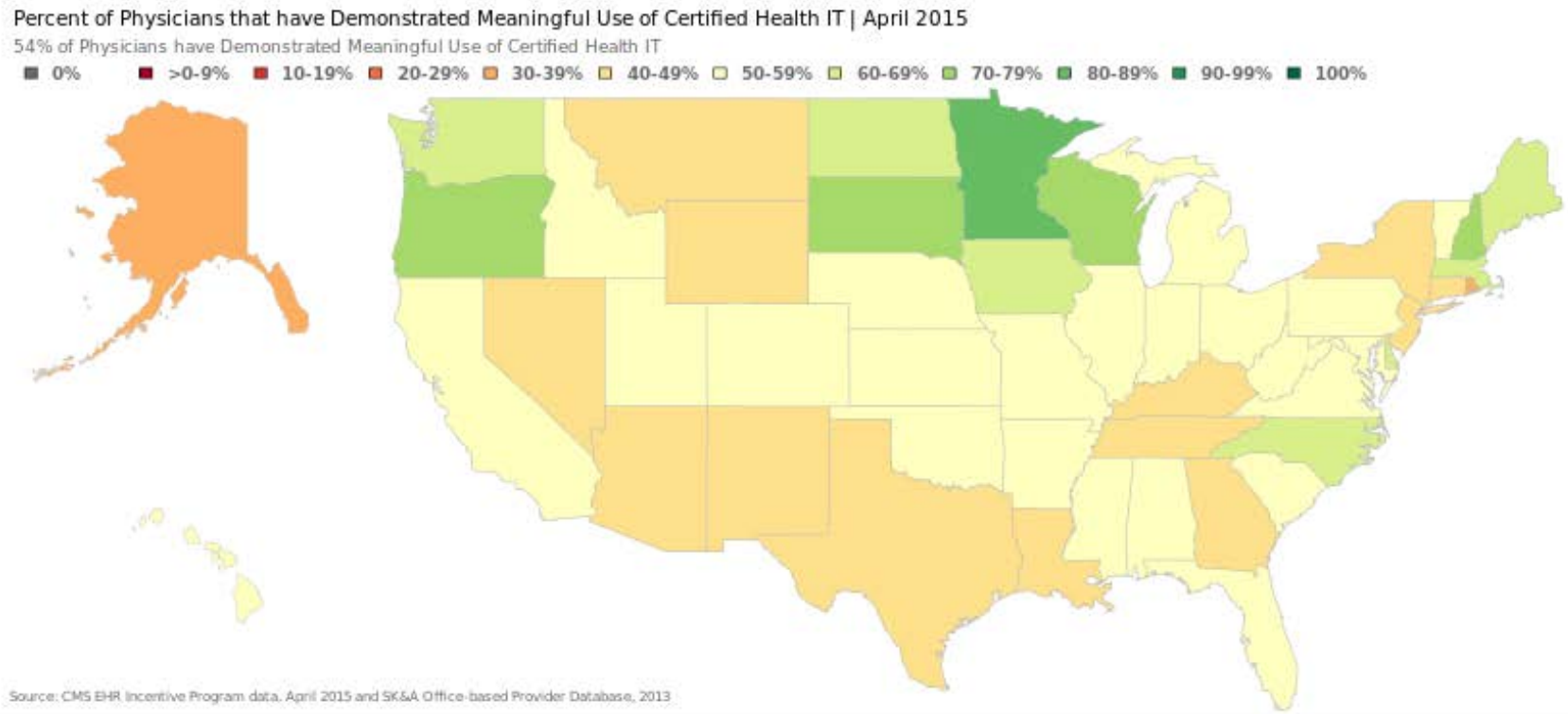
91-95%



Source: Office of the National Coordinator for Health Information Technology. 'Hospitals Participating in the CMS EHR Incentive Programs,' Health IT Quick-Stat #45. July 2015. dashboard.healthit.gov/quickstats/pages/FIG-Hospitals-EHR-Incentive-Programs.php

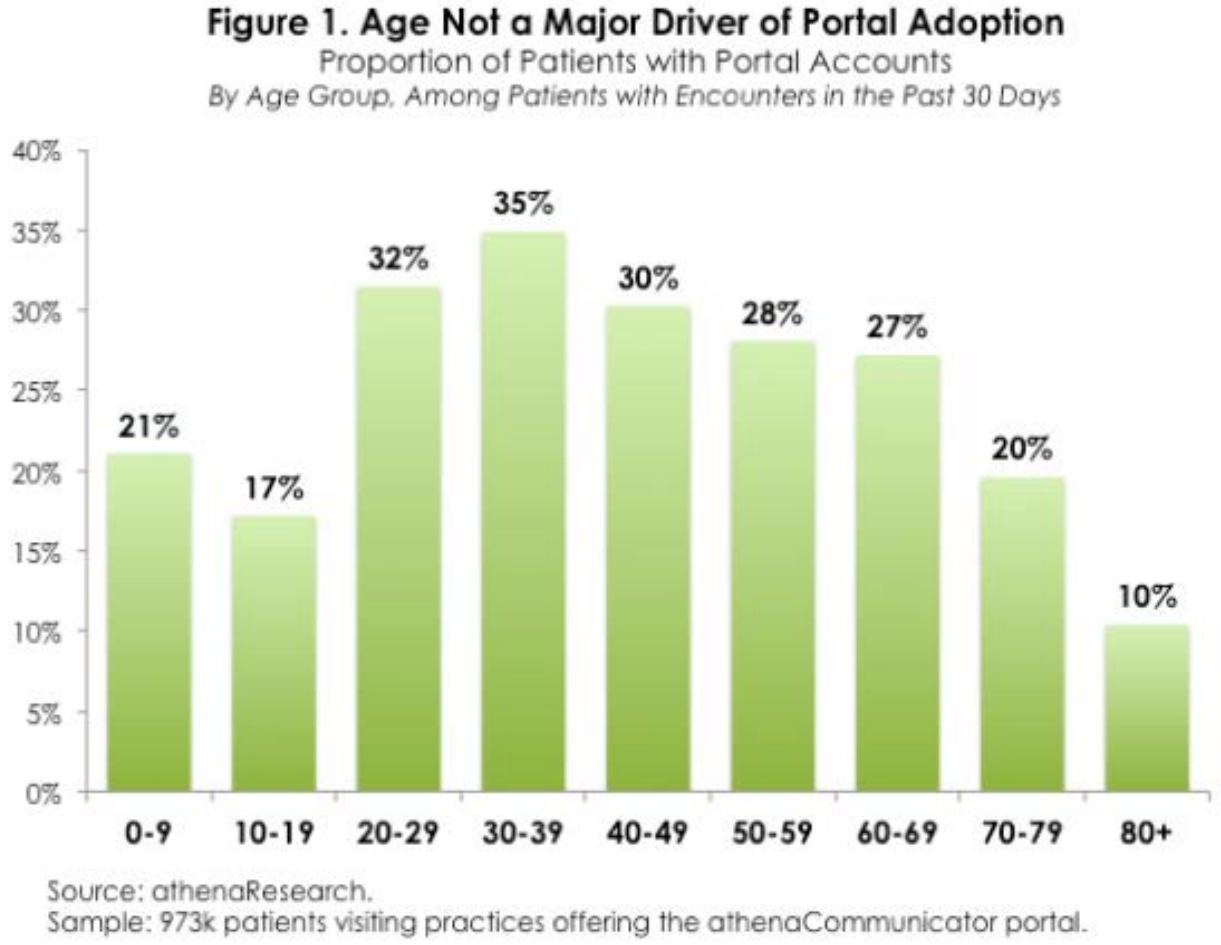
Office-Based Providers Demonstrating Meaningful Use

54%



Source: Office of the National Coordinator for Health Information Technology. 'Health Care Professionals Participating in the CMS EHR Incentive Programs,' Health IT Quick-Stat #44. July 2015. dashboard.healthit.gov/quickstats/pages/FIG-Health-Care-Professionals-EHR-Incentive-Programs.php.

Patient Portal Use in Large Health Plan



http://thehealthcareblog.com/blog/2015/08/10/the-current-state-of-patient-portal-adoption/

Outstanding Challenges

- EHRs designed to support clinical care
- Completeness & accuracy vary

Outstanding Challenges

- Still not standardized
- 100+ EHR vendor products

- Coding systems used differently
- ICD-10 transition

- Researchers do not control EHR design or documentation/coding practices

Clinical Phenotype Definitions

- Specifications for **identifying patients or populations with a given characteristic or condition of interest from EHRs** using data that are routinely collected in EHRs or ancillary data sources.
- Can support research (cohort selection; study screening) and personalized medicine

- Include widely adopted coding systems
  - ICD-9-CM
  - CPT
  - SNOMED CT
  - LOINC
  - RxNorm
  - NDC

Example

Diabetes defined as[1]:

- one inpatient discharge diagnosis (ICD-9-CM 250.x, 357.2, 366.41, 362.01-362.07)

ICD-9 codes

or any combination of two of the following events occurring within 24 months of each other:

- A1C ≥ 6.5% (48 mmol/mol)
- fasting plasma glucose ≥ 126 mg/dl (7.0 mmol/L)
- random plasma glucose ≥ 200 mg/dl (11.1 mmol/L)
- 2-h 75-g OGTT ≥ 200 mg/dl

Lab codes

- outpatient diagnosis code (same codes as inpatient)
- anti-hyperglycemic medication dispense (see details below)
- NDC in associated list
- **…etc., etc…**

Medication codes

1. Nichols GA, Desai J, Elston Lafata J, et al. Construction of a Multisite DataLink Using Electronic Health Records for the Identification, Surveillance, Prevention, and Management of Diabetes Mellitus: The SUPREME-DM Project. Prev Chronic Dis. 2012;9:110311.

The eMERGE Network

The mapping of the human genome has enabled new exploration of how genetic variations contribute to health and disease. To better realize this promise, researchers must now determine ways in which genetic make-up gives some individuals a greater chance of becoming sick with chronic conditions such as diabetes, Alzheimer's, or heart disease. The goal of gaining this knowledge is to translate it to bedside practice and ultimately improve patient care.

The Electronic Medical Records and Genomics (eMERGE) Network is a national consortium organized by NHGRI to develop, disseminate, and apply approaches to research. It combines DNA biorepositories with electronic medical record (EMR) systems for large-scale, high-throughput genetic research with the ultimate goal of returning genomic testing results to patients in a clinical care setting. The Network is currently exploring more than a dozen phenotypes (with 13 additional electronic algorithms having already been published). Various models of returning clinical results have been implemented or planned for pilot at sites across the Network. Themes of bioinformatics, genomic medicine, privacy and community engagement are of particular relevance to eMERGE.

What makes eMERGE unique?

Each center participating in the Network is studying the relationship between genome-wide genetic variation and a common human trait. Such studies commonly involve testing hundreds of thousands of genetic variants called single nucleotide polymorphisms (SNPs) throughout the genome in people with and without the trait. A number of such studies are re association between disease and a person's genetic make-up, but those studies are typically costly and take a long time to con

The eMERGE model is exploring use of data from the EMR – clinical systems that represen alternative methodology. Electronic medical records are one of the most exciting potentia member site has EMR data linked to genetic samples obtained in the course of existing co from residual tissue or blood samples. In the eMERGE model, there is no need to actively study population. Cases and controls are quickly and consistently identified from the EMR readily available. This approach is both cost-effective and time-efficient. More detailed inf phenotypes being explored in eMERGE can be found on on PheKB and other freely downl Resources page.

In addition, eMERGE focuses on ethical, legal, social, and policy issues such as privacy an

Phenotypes

Group - Any -

Include Methods ICD 10 Codes

Exclude Methods

Mine Only - Any -

Apply

Title	Groups	Institutions	Data and Methods	Status
Atrial Fibrillation - Demonstration Project	Vanderbilt - SD/RD Group	Vanderbilt University	CPT Codes, ICD 9 Codes, Natural Language Processing	Final
Cardiac Conduction (QRS)	eMERGE Phenotype WG	Vanderbilt University	CPT Codes, ICD 9 Codes, Laboratories, Medications, Natural Language Processing	Final
Cataracts	eMERGE Phenotype WG	Marshfield Clinic Research Foundation	CPT Codes, ICD 9 Codes, Medications, Natural Language Processing	Final
Clopidogrel Poor Metabolizers	Denny's Group at Vandy, VESPA - Vanderbilt Electronic Systems for Pharmacogenomic Assessment		CPT Codes, ICD 9 Codes, Laboratories, Medications, Natural Language Processing	Final
Crohn's Disease - Demonstration Project	Vanderbilt - SD/RD Group	Vanderbilt University	ICD 9 Codes, Medications, Natural Language Processing	Final
Dementia	eMERGE Phenotype WG	Group Health Cooperative	ICD 9 Codes, Medications	Final
Diabetic Retinopathy	eMERGE Phenotype WG	Marshfield Clinic Research Foundation	CPT Codes, ICD 9 Codes, Medications, Natural Language Processing	Final
Drug Induced Liver Injury	eMERGE Phenotype WG	Columbia University	ICD 9 Codes, Laboratories, Medications, Natural	Final

Most Recent Phenotypes

- Severe Early Childhood Obesity
- Warfarin dose/response
- Drug Induced Liver Injury
- Clopidogrel Poor Metabolizers
- Rheumatoid Arthritis - Demonstration Project

Other Sources for Clinical Phenotypes

- Clinical Classifications Software , "AHRQ Bundles"
- CMS Chronic Conditions Warehouse
- Quality Net (CMS and Joint Commission)
- Mini-Sentinel
- OMOP/OHDSI
- SHARPn
- …..

A comparison of phenotype definitions for diabetes mellitus

Rachel L Richesson,[1] Shelley A Rusincovitch,[2] Douglas Wixted,[3] Bryan C Batch,[4] Mark N Feinglos,[4] Marie Lynn Miranda,[5] W Ed Hammond,[2,6] Robert M Califf,[3,7] Susan E Spratt[4]

► Additional material is published online only. To view please visit the journal online (http://dx.doi.org/10.1136/amiajnl-2013-001952).

ABSTRACT

Objective This study compares the yield and characteristics of diabetes cohorts identified using heterogeneous phenotype definitions.

populations. Furthermore, standard phenotype definitions can streamline the development of patient registries from healthcare data, and enable consistent inclusion criteria to support regional surveillance and

Table 1 Data domain criteria used in selected phenotype definitions

Phenotype definitions:	Data domain criteria: ICD-9-CM 250.xx	ICD-9-CM 250.x0 and 250.x2 (excludes type 1 specific codes)	Expanded ICD-9-CM Codes (249.xx, 357.2, 362.0x, 366.41)	HbA1c	Fasting glucose	Random glucose	Abnormal OGTT	Diabetes-associated medications*
ICD-9-CM 250.xx	●							
CMS CCW	▲ * \\		▲ * \\					
NYC A1c Registry				●				
Diabetes-associated medications								●
DDC		▲	▲	▲ \\	▲ \\	▲ \\	▲ \\	▲
SUPREME-DM	▲ * \\		▲ * \\	▲ \\	▲ \\	▲ \\	▲	▲
eMERGE†		● * \\		▲	▲	▲		▲

*Medications vary by phenotype definition and are listed for each in the supplementary appendix (available online only).

†The eMERGE phenotype definition consists of five case scenarios with varying combinations of criteria. Any instance of type 1 specific codes (ie, 250.x1, 250.x3) results in the exclusion of the patient.

●=Sole criteria.

▲=Optional criteria, one of many.

*=Distinction made between inpatient and outpatient context.

\\ = Distinction made for multiple instances and/or time points.

CMS CCW, Centers for Medicare and Medicaid Services Chronic Condition Data Warehouse; DDC, Durham Diabetes Coalition; eMERGE, electronic medical records and genomics; HbA1c, hemoglobin A1c; ICD-9-CM, International Classification of Disease, revision 9, clinical modification; NYC, New York City; OGTT, oral glucose tolerance test; SUPREME-DM, Surveillance, Prevention, and Management of Diabetes Mellitus.

Clinical Trial Reporting

Patient characteristics:

Table 1. Patient Demographics and Baseline Characteristics

Characteristic	No. (%) of Patients[a] Gentamicin-Collagen Sponge (n = 753)	Control (n = 749)
Patient demographics		
Age, median (IQR), y	64.2 (58.0-71.5)	64.9 (57.2-72.1)
White race	688 (91.4)	683 (91.2)
Weight, median (IQR), kg	98.0 (86.1-113.0)	98.8 (85.0-111.1)
Body mass index, median (IQR)	33.1 (30.2-37.2)	32.8 (30.0-36.2)
Body mass index >30	574 (76.2)	563 (75.2)
Male sex	530 (70.4)	530 (70.8)
Medical history		
History of hypertension	659 (87.5)	659 (88.0)
History of diabetes	493 (65.5)	513 (68.5)
Current or history of smoking	458 (60.8)	450 (60.1)
Current smoking	136 (29.7)	123 (27.3)
History of chronic obstructive pulmonary disease	117 (15.5)	107 (14.3)
History of peripheral vascular disease	105 (13.9)	89 (11.9)
Previous median sternotomy	52 (6.9)	42 (5.6)
History of TIA or stroke	77 (10.2)	81 (10.8)
History of myocardial infarction	233 (31.0)	245 (32.7)
History of congestive heart failure	89 (11.8)	90 (12.0)
History of hyperlipidemia	619 (82.2)	607 (81.0)
Steroid use ≤1 mo prior to surgery	28 (3.7)	33 (4.4)
Receiving dialysis preoperatively	4 (0.5)	2 (0.3)
Preoperative diagnostic values		
Left ventricular ejection fraction, median (IQR), %	55 (45-60)	55 (45-60)
Serum glucose, median (IQR), mg/dL	125 (101-160)	124 (103-167)
Serum hemoglobin A_{1c}, median (IQR), %	6.5 (5.9-7.6)	6.6 (5.9-7.7)
Hematocrit, median (IQR), %	39 (36-42)	39 (36-42)
Serum creatinine, median (IQR), mg/dL	1.0 (0.9-1.3)	1.0 (0.9-1.2)
Preoperative core temperature, median (IQR), °C	97.6 (97.0-98.2)	97.7 (97.0-98.2)
Preoperative hospital stay, median (IQR), d	1.0 (0-3.0)	1.0 (0-3.0)
Parsonnet risk score, median (IQR)[b]	9.0 (6.0-14.5)	9.0 (6.0-16.0)

Abbreviations: IQR, interquartile range; TIA, transient ischemic attack.
SI conversion factors: To convert creatinine to µmol/L, multiply by 88.4; glucose to mmol/L, multiply by 0.0555.
[a]Unless otherwise indicated.
[b]Theoretical range is 0 to 148; 50% in Parsonnet et al[11] had a score between 0 and 9.

Multiple phenotype definitions:

SUPREME-DM Phenotype

Definition:
Adult Durham Population patients who meet **ONE OR MORE** of the following criteria during a DukeMed encounter between 2007-2011:

- One or more instances of the specified ICD-9-CM diagnosis codes (**see table 7**) on an inpatient encounter
- OR 2 or more instances of the specified ICD-9-CM diagnosis codes (**see table 7**) on outpatient encounters on separate days
- OR 1 or more instances of active stand-alone medication (**see table 8**) reported during outpatient medication reconciliation[3]
- OR 1 or more Oral Glucose Tolerance Test (OGTT) 2-hour 75g result >= 200 mg/dl where there is NO DIAGNOSIS CODE on the same encounter indicating pregnancy (V22, V23)[4]
- OR 2 or more hemoglobin A1c results >= 6.5% on 2 different days within 730 day span
- OR 2 or more fasting glucose results >= 126 mg/dl on 2 different days within 730 day span
- OR 2 or more random glucose results >= 200 mg on 2 different days within 730 day span
- OR within a 730 day span on 2 different days:
  - Fasting glucose results >= 126 mg/dl
  - AND Random glucose results >= 200 mg
- OR within a 730 day span (can be same day):
  - Hemoglobin A1c results >= 6.5%
  - AND Fasting glucose results >= 126 mg/dl

Abnormal Lab Results

Source:
Laboratory results

Definition:
Adult Durham Population patients who meet **ONE OR MORE** of the following criteria during a DukeMed encounter between 2007-2011:

- One or more instances of hemoglobin A1c results >= 6.5%
- OR one or more fasting glucose results >= 126 mg/dl within 365 day span
- OR one or more random glucose results >= 200 mg/dl within 365 day span

Abnormal HbA1c (NCY A1c Registry Definition)

Source:
Glycated hemoglobin laboratory results

Definition:
Adult Durham Population patients who meet **ONE OR MORE** of the following criteria during a DukeMed encounter between 2007-2011:

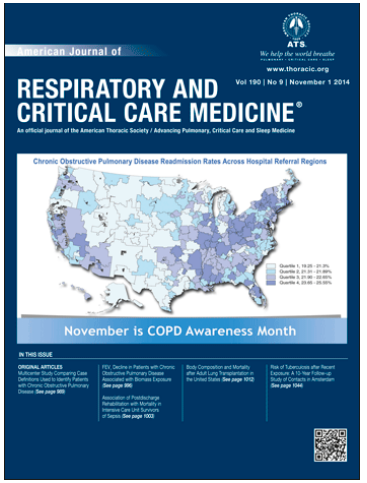
- One or more instances of hemoglobin A1c results >= 6.5%

ORIGINAL ARTICLE

Multicenter Study Comparing Case Definitions Used to Identify Patients with Chronic Obstructive Pulmonary Disease

Valentin Prieto-Centurion[1], Andrew J. Rolle[1], David H. Au[2], Shannon S. Carson[3], Ashley G. Henderson[3], Todd A. Lee[4], Peter K. Lindenauer[5,6], Mary A. McBurnie[7], Richard A. Mularski[7], Edward T. Naureckas[8], William M. Vollmer[7], Binoy J. Joese[9], and Jerry A. Krishnan[1,9]; on behalf of the CONCERT Consortium

[1]Division of Pulmonary, Critical Care, Sleep and Allergy and [4]Department of Pharmacy Systems, Outcomes and Policy, University of Illinois at Chicago, Chicago, Illinois; [2]University of Washington/VA Puget Sound, Seattle, Washington; [3]Division of Pulmonary and Critical Care Medicine, University of North Carolina at Chapel Hill, Chapel Hill, North Carolina; [5]Department of Medicine and Center for Quality of Care Research, Baystate Medical Center, Springfield, Massachusetts; [6]Tufts University School of Medicine, Boston, Massachusetts; [7]The Center for Health Research, Kaiser Permanente, Portland, Oregon; [8]Section of Pulmonary and Critical Care, University of Chicago Medicine, Chicago, Illinois; and [9]Population Health Sciences Program, University of Illinois Hospital and Health Sciences System, Chicago, Illinois



Am J Respir Crit Care Med. 2014 Nov 1;190(9):989-95.
doi: 10.1164/rccm.201406-1166OC.

Table 2. Clinical Characteristics of Patients Who Met and Did Not Meet the Clinical Trial Reference Standard

Characteristic	Total Sample (*n* = 998)	Clinical Trial Reference Standard		*P* Value
		Yes* (*n* = 560)	No† (*n* = 438)	
Comorbid conditions, %				
Cardiovascular disease	76	74	78	0.15
Hypertension	66	63	69	0.03
Heart failure	18	16	22	0.01
Coronary artery disease	23	22	24	0.66
Myocardial infarction	19	18	20	0.43
Stroke	15	14	15	0.95
Depression	42	36	50	<0.0001
Arthritis	36	33	41	0.006
Diabetes	28	22	34	<0.0001
Cancer history	23	26	19	0.02
Anemia	28	26	30	0.17
Kidney disease	20	18	21	0.30
Dementia	2	2	3	0.15
Dyspnea at rest (Borg), %				
0, no dyspnea	52	54	50	0.02
0.5–2, slight	38	38	37	
⩾3, moderate to very severe	10	7	13	
Spirometry, post-bronchodilator, %				
FEV_1/FVC <70%	61	100	11	<0.0001
FEV_1 <80% predicted	72	86	55	<0.0001
6-minute-walk distance, %				
Distance walked <350 m	53	52	54	0.67

Patients who met the trial reference standard are more likely to have airflow obstruction by spirometry but report being less dyspneic. Patients who met the reference standard also have different prevalence of comorbidities. For example, they are more likely to have hypertension, heart failure, and depression. Data for 6-minute-walk distance missing in 9% patients (9% and 10%) and dyspnea scores missing in 8% patients (8% and 9%) in those who met and did not meet the clinical trial reference standard, respectively.

*(*A* + *D* + *E* + *G*) and †(*B* + *C* + *F*) in Figure 2.

Table 3. Characteristics Associated with Meeting the Clinical Trial Reference Standard

Characteristics	Odds Ratio (95% CI)
Race (vs. white)	
Black	0.37 (0.26–0.53)*
Other	0.52 (0.27–1.00)
Education (vs. high school or less)	
College/professional degree	0.38 (0.26–0.56)*
Some college	0.68 (1.06–2.03)*
BMI, kg/m^2 (vs. normal)	
<18.5 (underweight)	4.00 (1.27–12.50)*
25–29.99 (overweight)	0.87 (0.58–1.30)
⩾30 (obese)	0.51 (0.35–0.75)*
Depression (yes vs. no)	0.53 (0.40–0.71)*
Diabetes (yes vs. no)	0.67 (0.48–0.93)*
Cancer (yes vs. no)	1.47 (1.05–2.08)*

Definition of abbreviations: BMI = body mass index; CI = confidence interval.
Clinical trial reference standard (*A* + *D* + *E* + *G*) versus others (*B* + *C* + *F*) in Figure 2. Multivariable logistic regression model that included characteristics listed in Tables 1 and 2 (characteristics significantly associated with meeting the trial reference standard). Results indicate that patients who are black (vs. white), with college or higher (vs. high school or less) education, obese (vs. normal weight), with depression, or diabetes are less likely to meet the trial reference standard. Patients with a history of cancer and underweight patients (vs. normal weight) are more likely to meet the trial reference standard. Hosmer-Lemeshow goodness-of-fit test (*P* value = 0.17) demonstrates adequate model fit.
*$P < 0.05$.

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Upcoming Events

Grand Rounds March 7: Bray Patrick-Lake (CTTI; PCORnet Executive Committee member), Sue Sheridan (PCORI), and Sean Tunis (CMTP)
Patient Engagement in Infrastructure Development

Secretary's Advisory Committee for Human Research Protections (SACHRP): March 12-13

Grand Rounds March 14: TBD
TBD

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02-21-14: Sharon Terry
Participant Engagement: Tools to Meet People Where They Are

02-14-14: Eric Larson
Engaging Health Systems in Research Partnerships

Collaboratory News

Stop CRC featured on NPR Health Blog
02/26/2014: Gloria Coronado, PhD, was recently featured on the NPR Health Blog discussing the Stop CRC study.

Joe Selby writes perspective piece for the New England Journal of Medicine on PCORI
02/13/14: Joe Selby, MD, MPH, Executive Director of PCORI, published a perspective piece in the latest issue of the New England Journal of Medicine on lessons learned in PCORI's 3-year history.

First patient enrolled in Collaboratory trial
01/13/14: The TIME Demonstration Project, led the University of Pennsylvania's Laura Dember, MD, has enrolled its first patient.

Other Research Updates in the News ... >

PubMed Related Articles

URL	Publication Date	Description
Rescuing clinical trials in the United States and beyond: A call for action.	2013/06	To promote consensus around the solutions needed to address the adverse trends in clinical research, the Duke Clinical Research Institute convenedstakeholders from academia, industry, and government. This article summarizes the proceedings.
Rapid, responsive, relevant (R3) research: a call for a rapid learning health research enterprise	2013/05	To produce more rapid, responsive, and relevant research, we propose approaches that increase relevance via greater stakeholder involvement, speed research via innovative designs, streamline review processes.
Human subjects protections in community-engaged research: a research ethics framework	2010/03	This new framework for exploring the risks in community-engaged research can help academic researchers and community partners ensure the mutual respect that community-engaged research requires.

pcornet | The National Patient-Centered Clinical Research Network

http://pcornet.org

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PCORnet: The National Patient-Centered Clinical Research Network

The Patient-Centered Outcomes Research Institute (PCORI) is supporting the development of PCORnet, the National Patient-Centered Clinical Research Network, to create a large, highly representative, national network for conducting clinical outcomes research.

PCORnet will transform clinical research by engaging patients, care providers, and health systems in collaborative partnerships to improve healthcare and advance medical knowledge. By bringing research and patient care together, this innovative health data network will be able to explore the questions that matter most to patients and their families. Read more ….

Patient-focused drug development moves into the mainstream

Posted March 16, 2015, Applied Clinical Trials

Building on the experience gained from FDA meetings soliciting patient perspectives, sponsors

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Patient Data Privacy

PCORnet Master Slides

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Why PCORnet Exists

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Coverage and Precision of Rare Disease Names (n=5,333)

	Equivalent match	Broader match (found)	Broader match (projected)
ICD-9-CM	697 (13%)	2055 (39%)	2569 (48%)
ICD-10-CM	1386 (26%)	919 (17%)	1635 (31%)
SNOMED CT	2848 (53%)	Not estimated	Not estimated

Unpublished Data. Kin Wah Fung, Olivier Bodenreider, Rachel Richesson 2014.

“Grouper” codes that contain multiple rare disease concepts (less precise)

# rare diseases included in code	# ICD-9-CM codes	# ICD-10-CM codes	# SNOMED CT codes
1	1081 (62%)	1403 (73%)	3311 (85%)
2	319	328	478
3	125	88	84
4	68	45	33
5	43	25	3
> 5	117	40	0
# codes including > 1 disease (%)	672 (38%)	526 (27%)	598 (15%)
Examples	208 rare diseases included under *759.89 Other specified congenital anomalies*	22 rare diseases included under *Q82.8 Other specified congenital malformations of skin*	5 rare diseases included under *28835009 Retinitis pigmentosa*

Unpublished Data. Kin Wah Fung, Olivier Bodenreider, Rachel Richesson 2014.

SNOMED CT

- Most comprehensive, multilingual clinical terminology in the world
- Used in > 50 countries
- Meaningful Use requires use of SNOMED CT in the EHR for problem lists, procedures.
- SNOMED CT is better suited for clinical data capture because:
  - Better content coverage
  - Clinically oriented
  - Flexible data entry and retrieval

Use Cases and Coding Systems for Rare Diseases

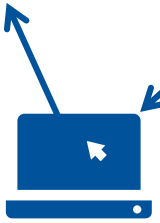
HPO and **ORDO** for "deep phenotyping" of undiagnosed disorders in specialty or genetics clinics.

Link to **OMIM** and **GO** and for molecular diagnosis.

Electronic Health Record Systems

Encode with **SNOMED CT** for documenting diagnoses or "problems"

MeSH for linkage to the biomedical literature and clinical practice guidelines (e.g., InfoButton, CDSS).



Linkage to patient-directed health information (e.g., Medline Plus search with **MeSH** synonyms).

Reimbursement	**ICD-9-CM, ICD-10-CM**
Public Health Surveillance	**ICD-10**
Quality Measurement	**SNOMED CT**
Interventional Research	**SNOMED CT, MedDRA; plus new data collection using PhenX and LOINC**

Query **SNOMED CT** for networked research networks and observational research.

Multiple Terminologies

1. Outpatient COPD encounter	**ICD-9 (or corresponding SNOMED) codings** **491.xx - chronic bronchitis** **492.xx - emphysema** **493.2x - chronic obstructive asthma** **496.xx - chronic airway obstruction**
2. Inpatient COPD encounter	ICD-9 (or corresponding SNOMED) codings 490.xx - bronchitis, not specified as acute or chronic 491.xx - chronic bronchitis 492.xx - emphysema 493.2x - chronic obstructive asthma 494.xx - bronchiectasis 495.xx - extrinsic allergic alveolitis 496.xx - chronic airway obstruction
3. Hospitalized respiratory failure (primary) with secondary COPD coding	518.81 acute respiratory failure 518.82 other pulmonary insufficiency not elsewhere classified 518.84 acute and chronic respiratory failure
4. Problem List	Coded fields ICD-9 / variant "V" coding or SNOMED) – COPD, emphysema, chronic bronchitis, chronic airway obstruction
5. PFT demonstrating likely obstructive lung disease	Evidence of pre or post-bronchodilator obstruction defined by an FEV1 / FVC ratio < 0.7
6. Documented COPD-related drug dispensing, order or medication listing	Beta-agonists by nebulized delivery Beta-agonists, Inhalers - Long acting Ipatropium & Tiotropium Inhaled Corticosteroid (ICS) Combivent (Ipratropium+Albuterol) Combination Inhaled Beta-agonists + ICS (ie Advair)

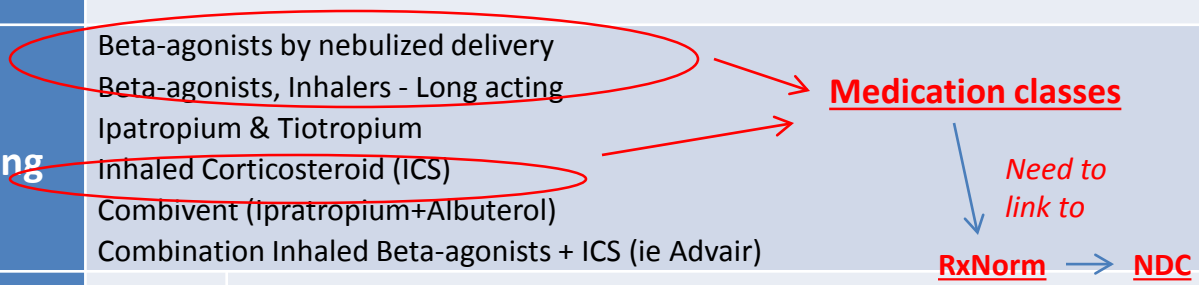
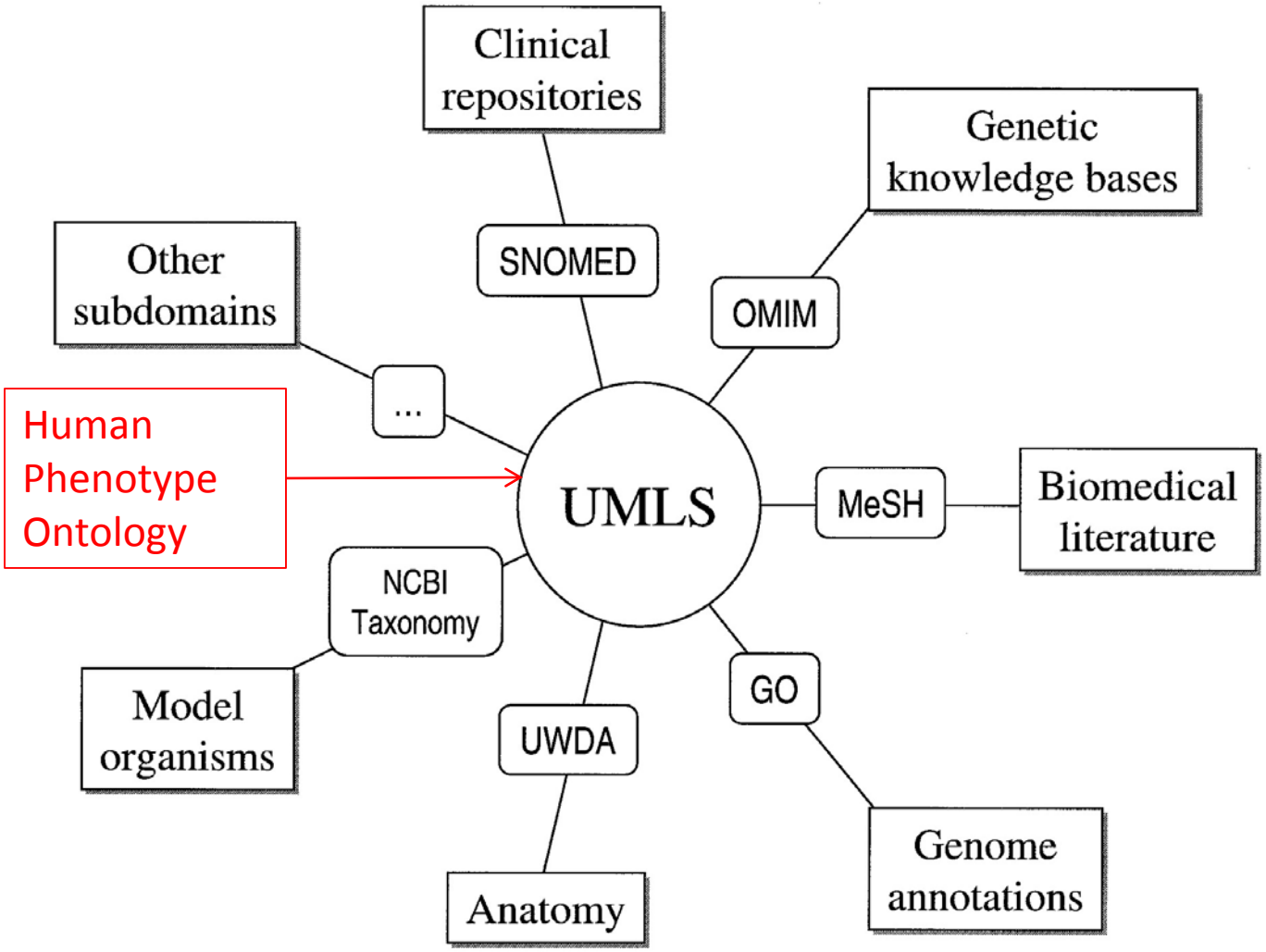


Figure 1. The various subdomains integrated in the UMLS.



Olivier Bodenreider Nucl. Acids Res. 2004;32:D267-D270

Possible actions…

- Link specialized terminologies with UMLS
- Recognize importance of clinical interface terminology (SNOMED CT or HPO)
  - Advocate for standards (requirements or incentives)
  - Promote use of SNOMED CT in healthcare systems
- Expand coverage of diseases in SNOMED CT
- Laboratory data critical

- Coordinate and communicate across disciplines!

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