

# Coming to Terms with the Biomedical Tower of Babel

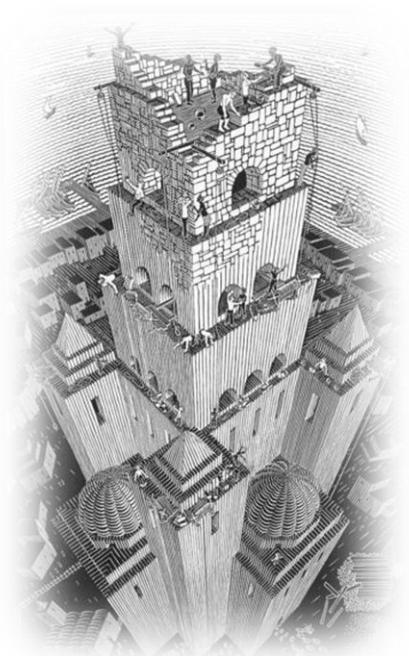
*Implications for the design of a biomedical knowledge network*

Alexa T. McCray

Center for Biomedical Informatics  
Harvard Medical School

Board on Research Data and  
Information  
National Academy of Sciences

February 26, 2013



*“No aspect of human life has escaped the impact of the Information Age, and perhaps in no area of life is information more critical than in health and medicine.”*

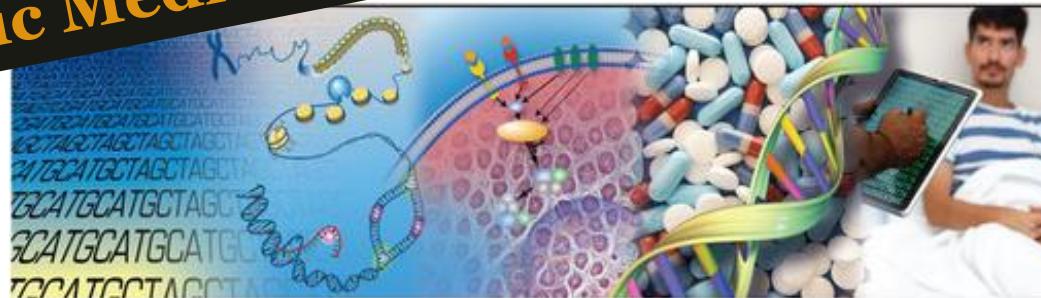
U.S. National Academy of Engineering  
Grand Challenges 2008

<http://www.engineeringchallenges.org>

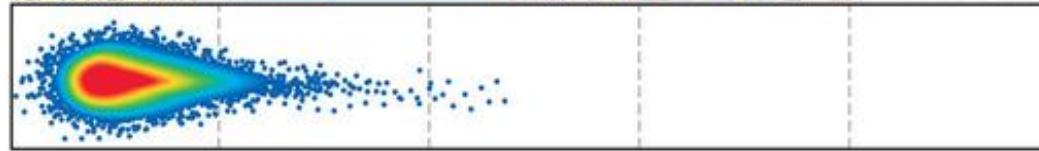


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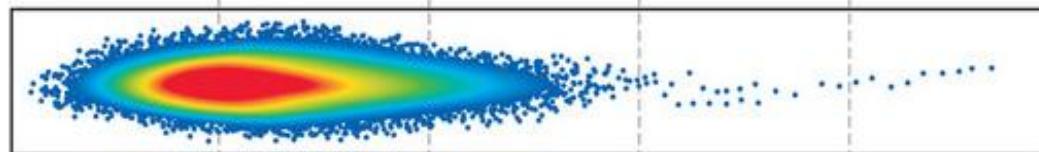
# The Promise of Genomic Medicine



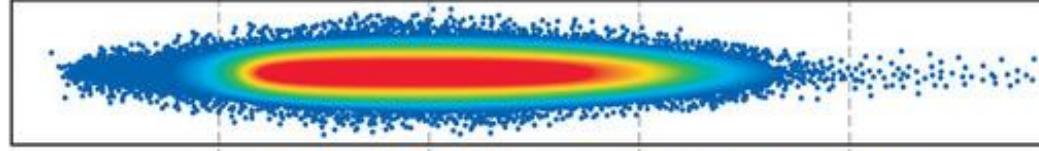
1990-2003  
Human Genome  
Project



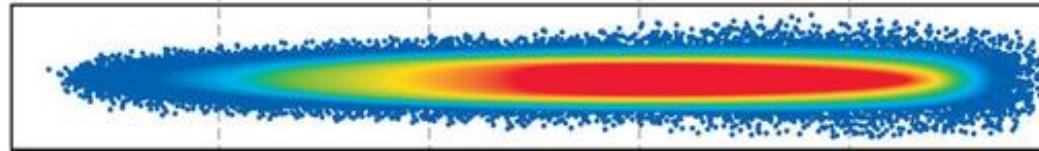
2004-2010



2011-2020

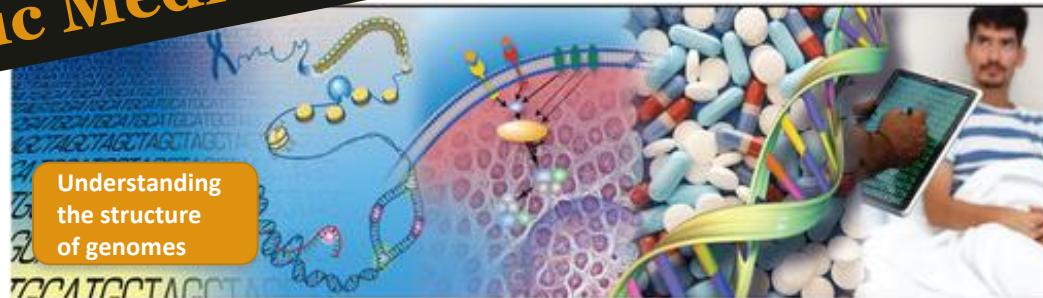


Beyond 2020



Green ED. *Nature* 2011; 470(10):203-13

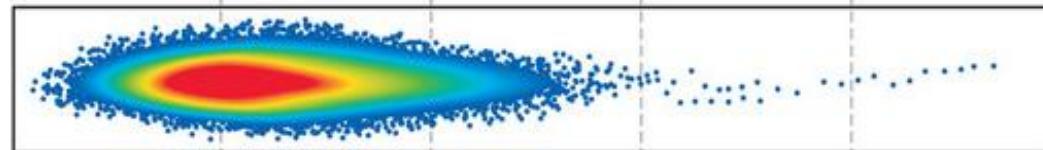
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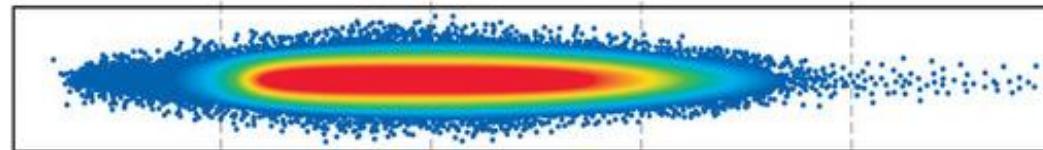
## 1990-2003 Human Genome Project



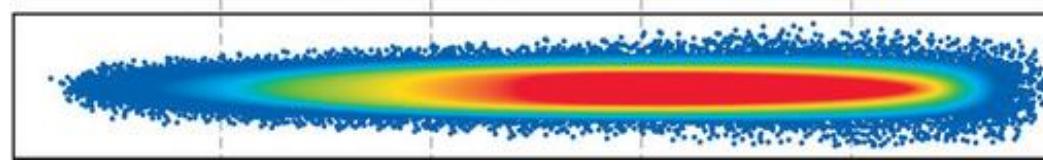
2004-2010



2011-2020



## Beyond 2020



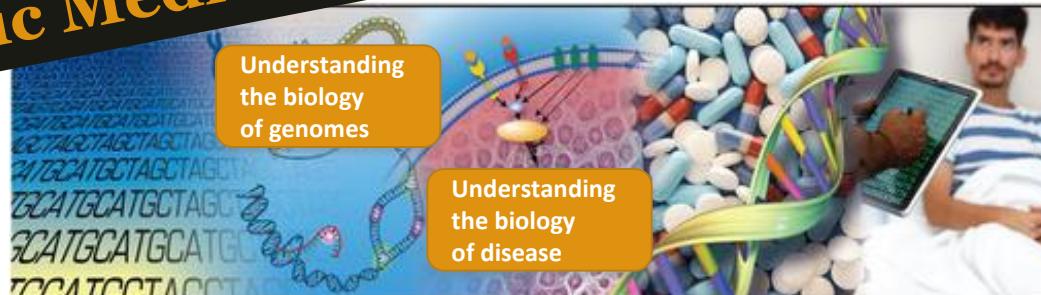
Green ED. *Nature* 2011; 470(10):203-13



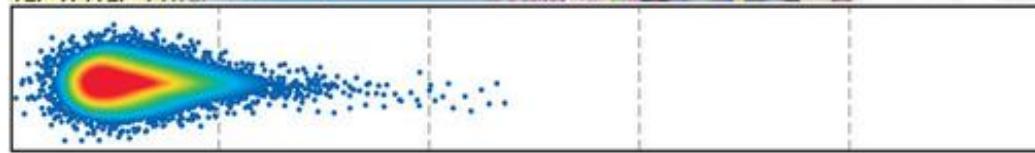
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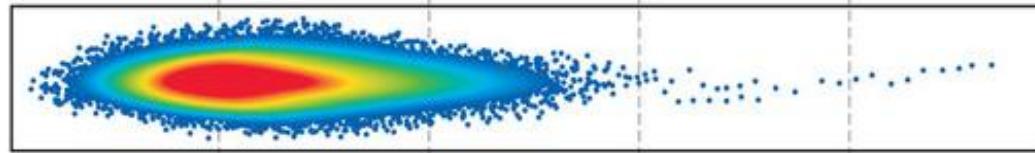
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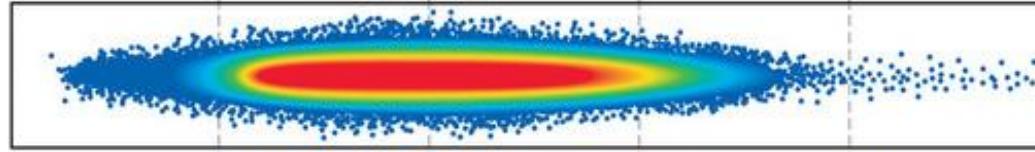
1990-2003  
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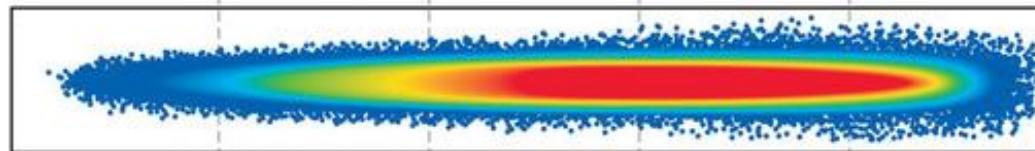
2004-2010



2011-2020

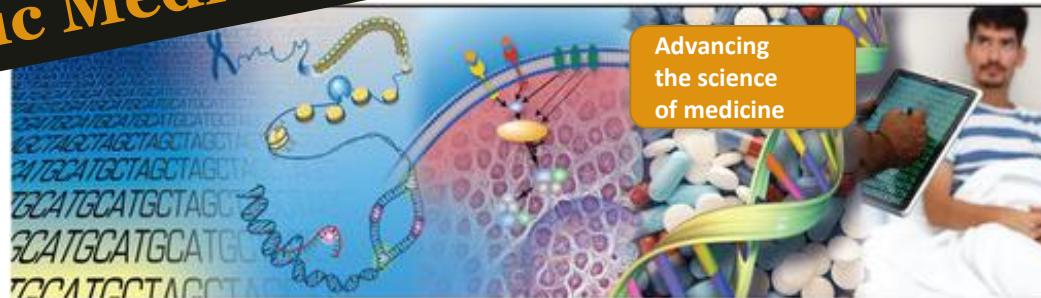


Beyond 2020

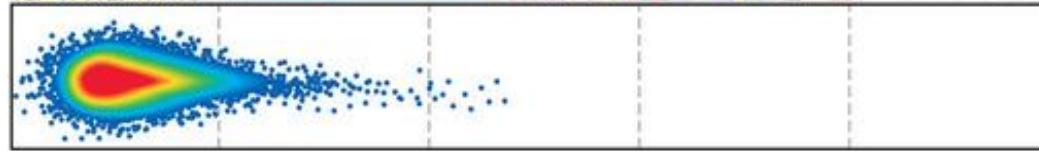


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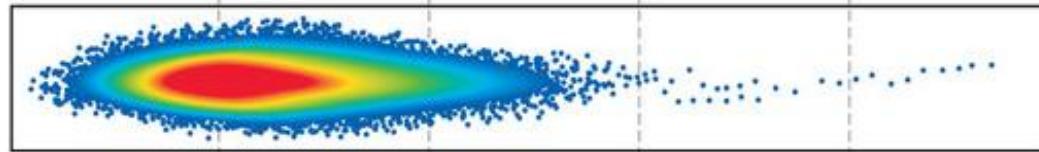
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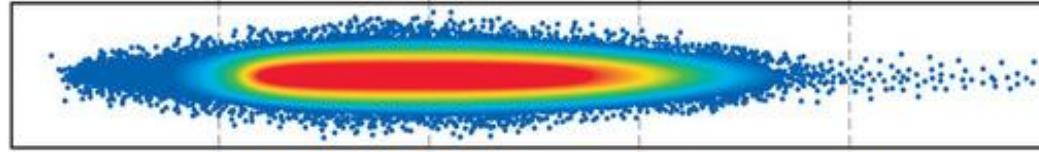
1990-2003  
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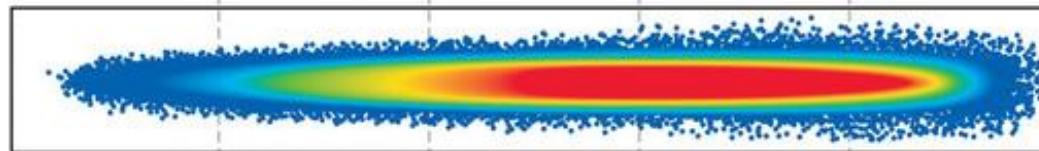
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2011-2020

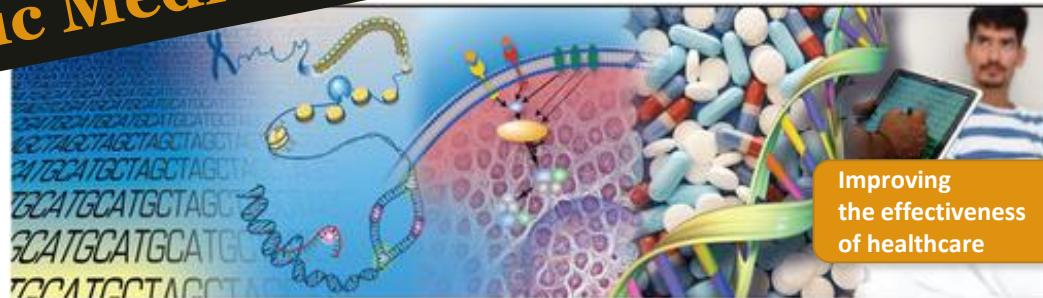


Beyond 2020



Green ED. *Nature* 2011; 470(10):203-13

# The Promise of Genomic Medicine

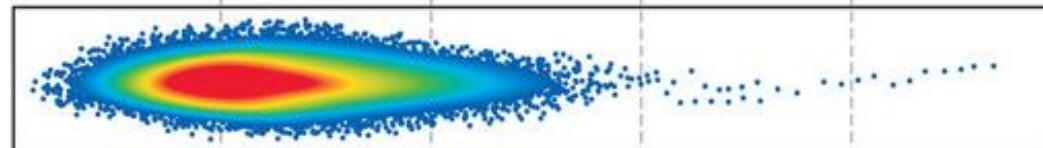


## Improving the effectiveness of healthcare

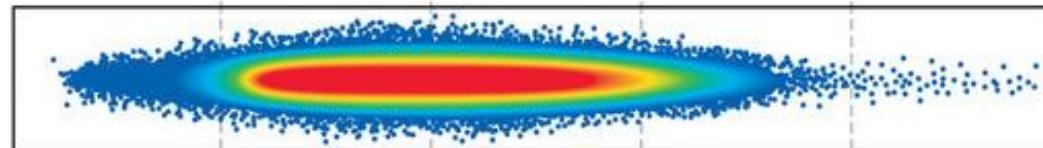
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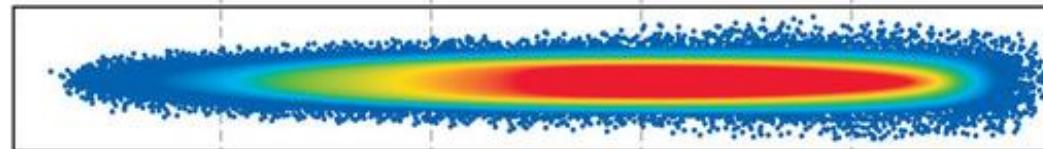
2004-2010



2011-2020



Beyond 2020



Green ED. *Nature* 2011; 470(10):203-13



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# An Inflection Point

“Biomedical research and the practice of medicine, separately and together are reaching an inflection point: the capacity for description and for collecting data, is expanding dramatically, but the efficiency of compiling, organizing manipulating these data – and extracting true understanding of fundamental biological processes, and insights into human health and disease, from them – has not kept pace.”

*Toward Precision Medicine: Building a Knowledge Network for Biomedical Research and a New Taxonomy of Disease*

National Research Council, 2011:9.

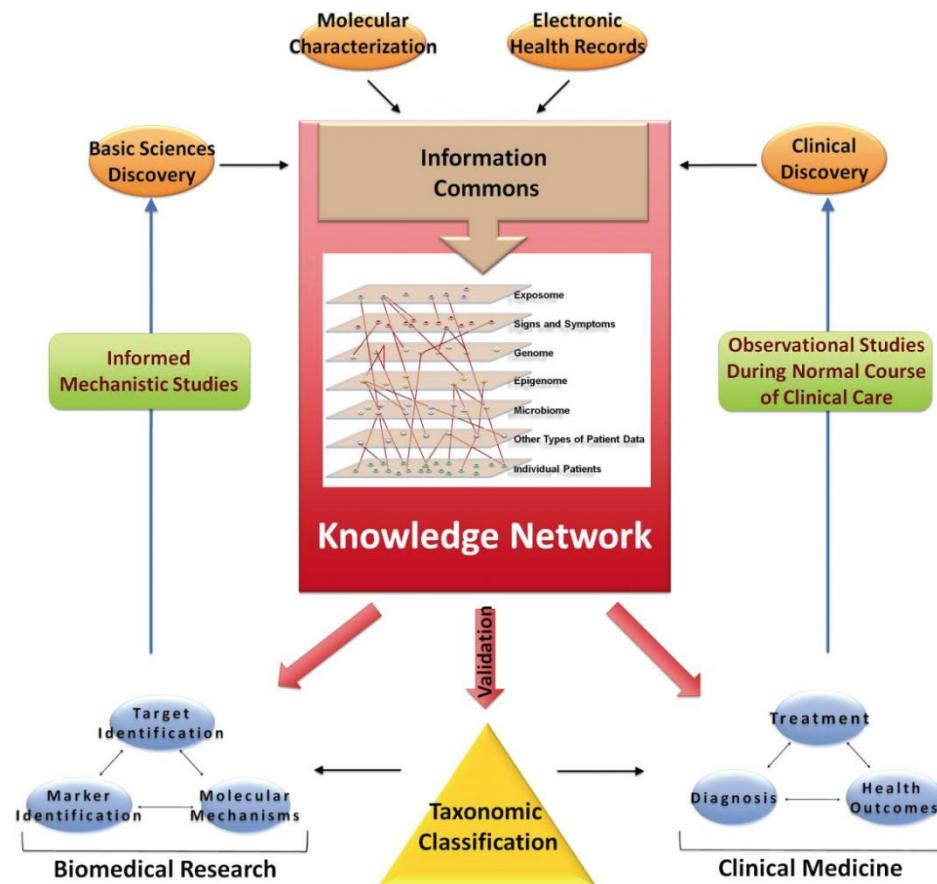


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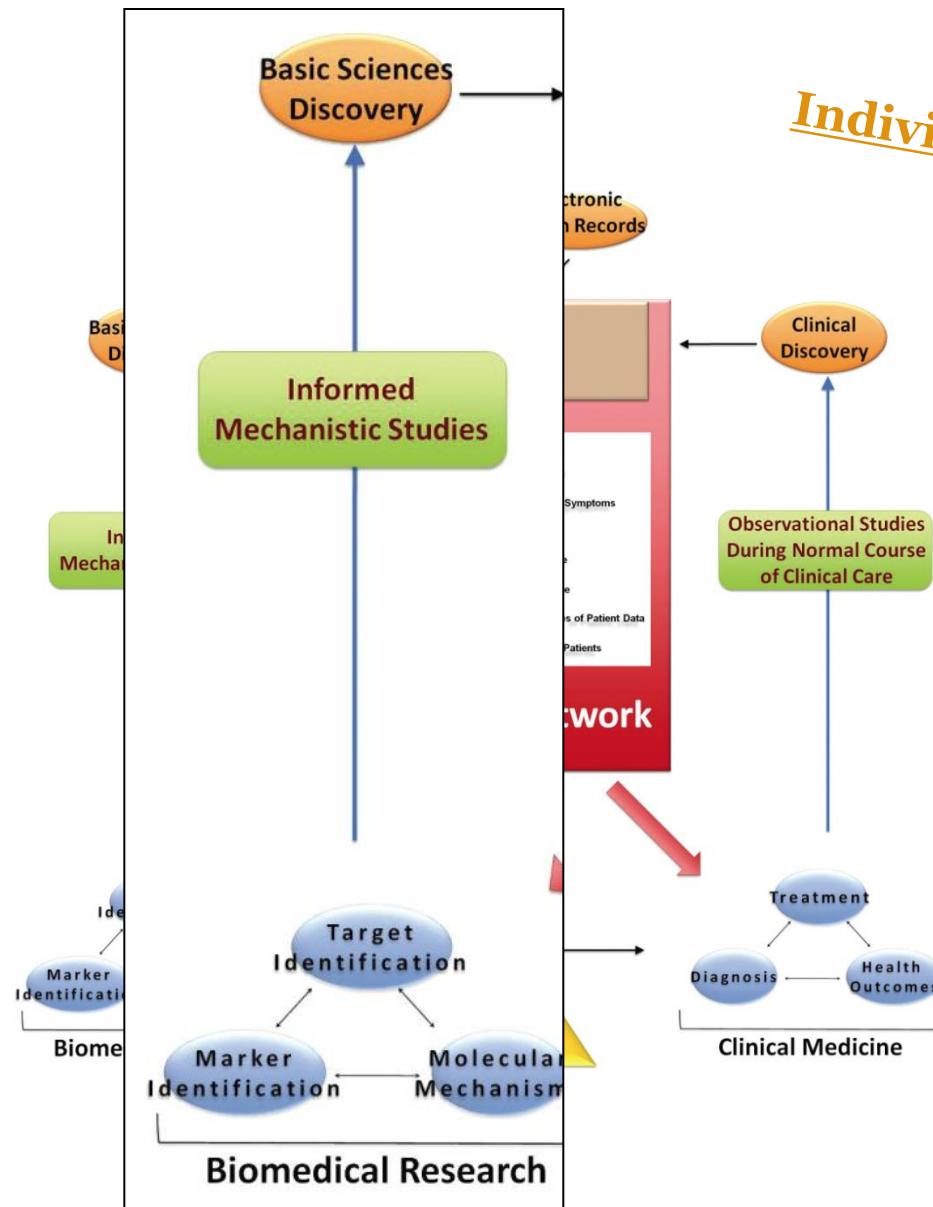
# Precision Medicine

*Individual-Centric*



*Toward Precision Medicine*  
National Research Council, 2011

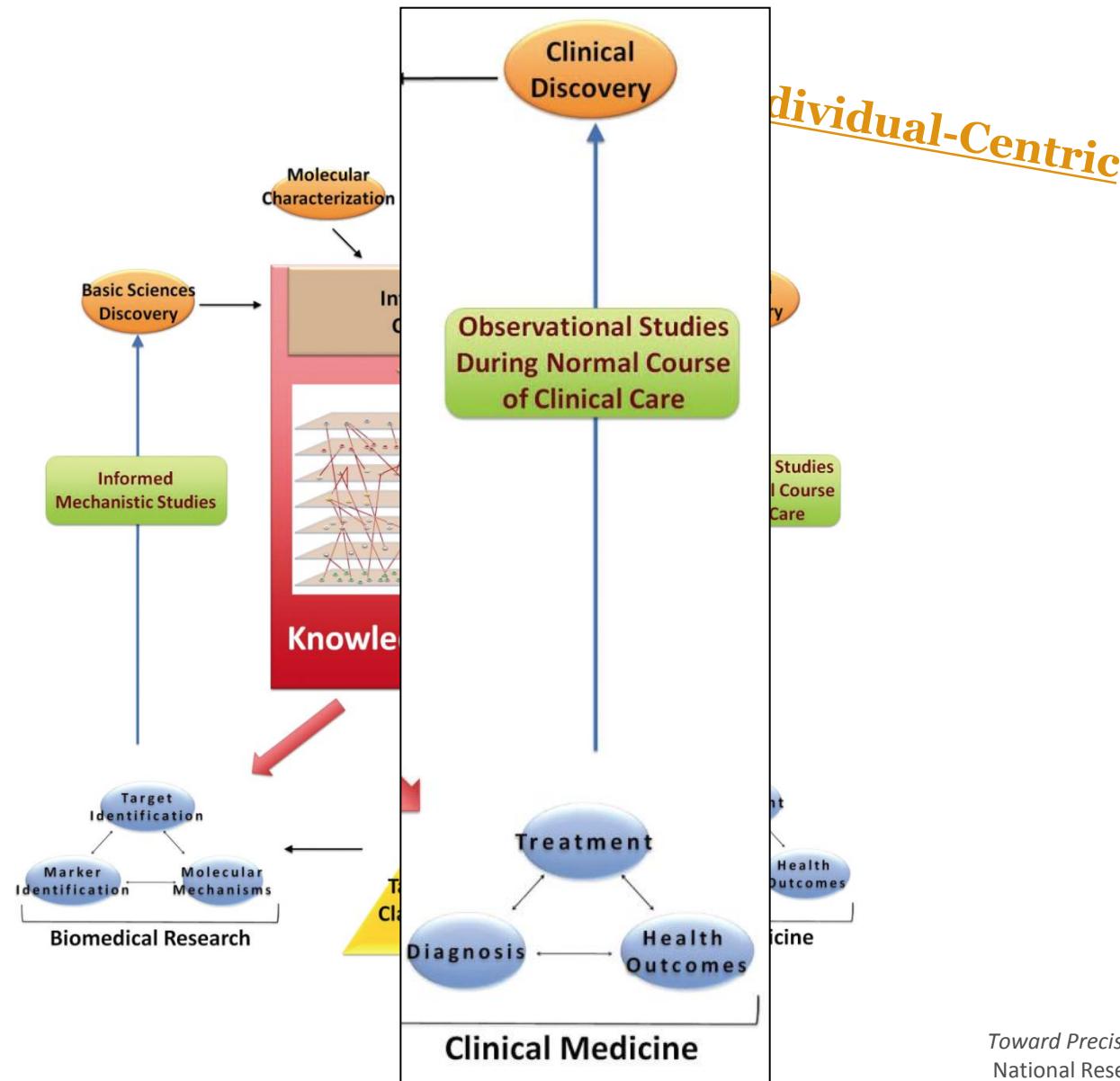
# Precision Medicine



# Individual-Centric

*Toward Precision Medicine*  
National Research Council, 2011

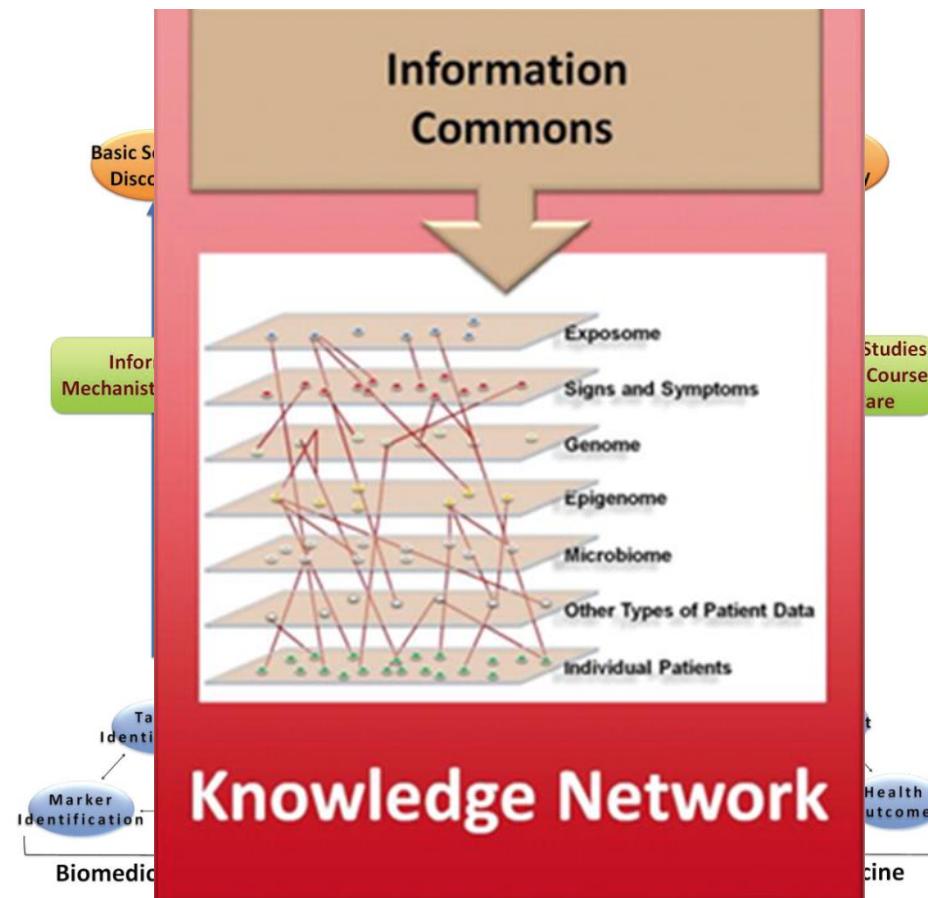
# Precision Medicine



Toward Precision Medicine  
National Research Council, 2011

# Precision Medicine

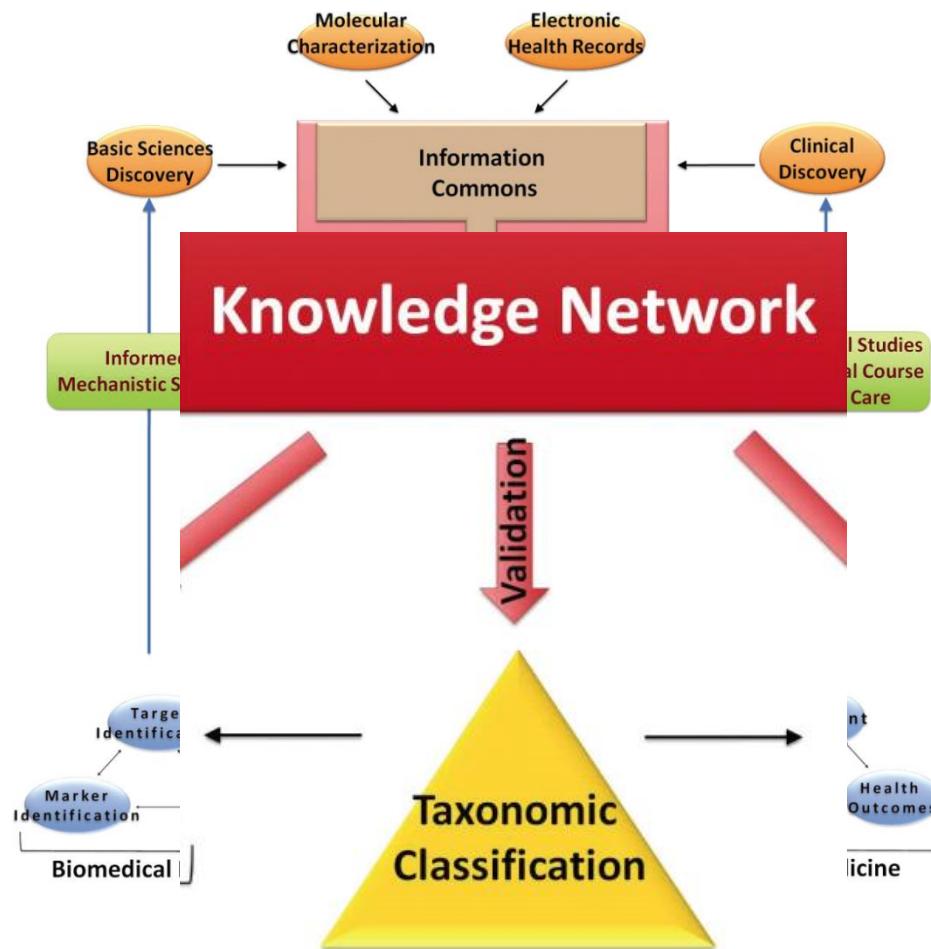
*Individual-Centric*



*Toward Precision Medicine*  
National Research Council, 2011

# Precision Medicine

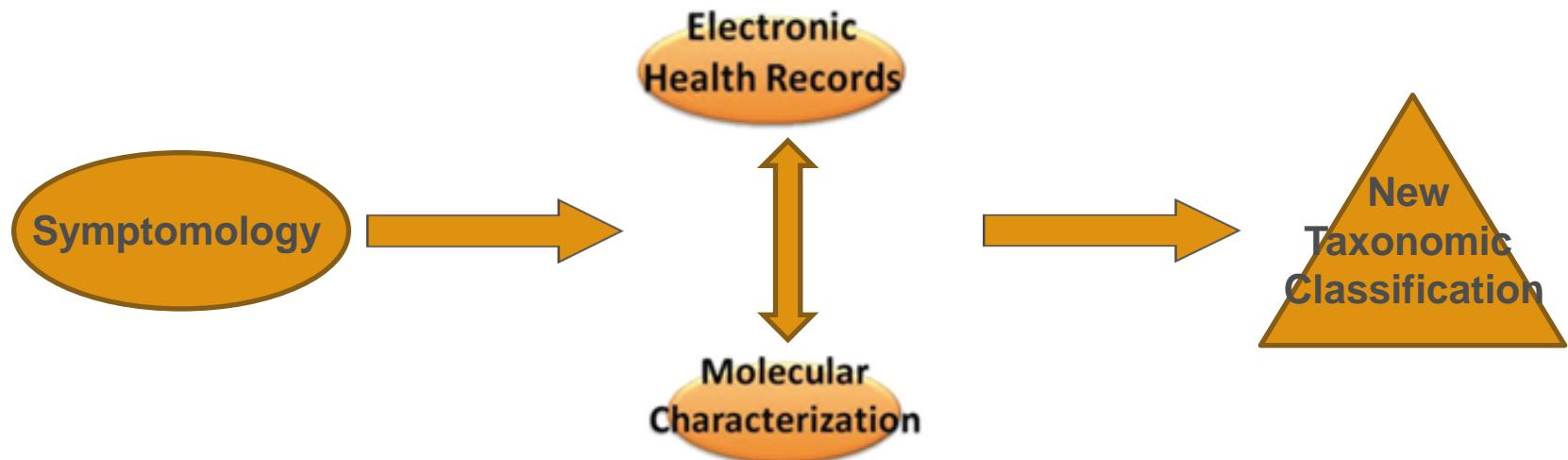
*Individual-Centric*



Toward Precision Medicine  
National Research Council, 2011

# A New Taxonomy of Disease

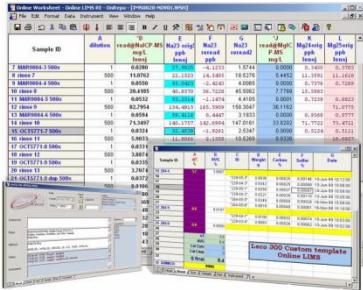
*“Could it be that something as fundamental as our current system for classifying diseases is actually inhibiting progress?”*



*Toward Precision Medicine.*  
National Research Council, 2011:10

# Wide Range of Data Sources Needed

## Laboratory Data

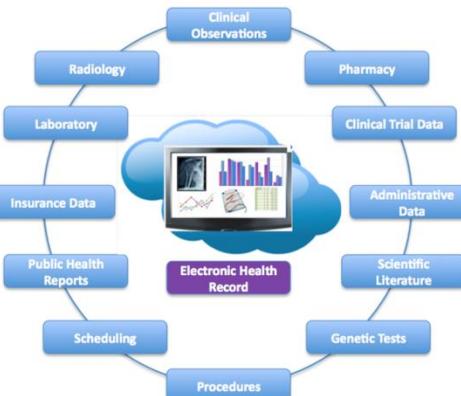


## Biosensor Data



- All have their own
  - Syntax
  - Semantics
- Some use standard terminologies/ontologies
- Some use home-grown terminologies

## Clinical Systems



## Public Databases

## Social Networking Sites

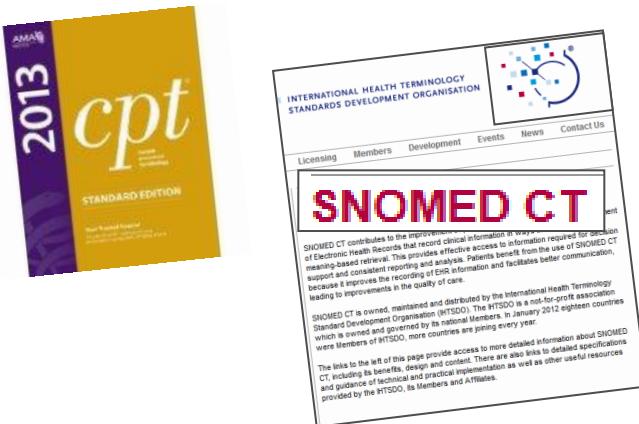
# Terminology Use

- Coding clinical data**
- Indexing and retrieving the literature**
- Annotating genomic data**
- Statistical reporting, epidemiologic studies**
- Outcomes measurement**
- Public health surveillance**
- Cost analysis**
- Information exchange and data integration**
- Data mining, aggregation**
- Natural language processing**
- ...



# Common Terminologies

Varying scope, coverage, rigor, and update schedules



NATIONAL LIBRARY OF MEDICINE MEDICAL SUBJECT HEADINGS MeSH

MeSH Home Contact NLM Site Index Search Our Web Site NLM Home

Health Information Library Services Research Programs New & Noteworthy General Information

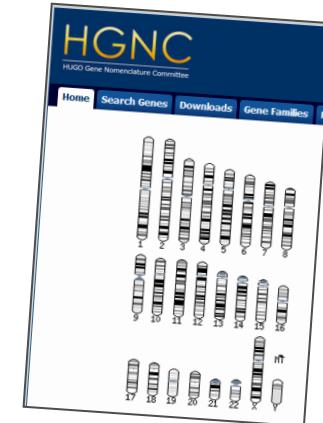
MeSH Browser (2013 MeSH)  
The files are updated every week on Sunday.  
[Go to 2012 MeSH](#)

Enter term or the beginning of any root fragments: or [Navigate from tree top](#)

Search for these record types:

- Main Headings
- Qualifiers
- Supplementary Concepts
- All of the Above
- Search as MeSH Unique ID
- Search as text words in Annotation & Scope Note

[Find Exact Term](#) [Find Terms with ALL Fragments](#) [Find Terms with ANY Fragment](#)



FDA U.S. Food and Drug Administration Protecting and Promoting Your Health

Home Food Drugs Medical Devices Vaccines, Blood & Biologics Animal & Veterinary Cosmetics Radiation

National Drug Code Directory

Current through February 22, 2013

Proprietary Name Search Application Number Search Active Ingredient Search Labeler Name Search NDC Number Search

Background Information Drug Database: [DRUGINFO@FDA/HHS.GOV](#)  
See also: [Drug Registration and Listing Information](#) [National Drug Code Database Data Files](#)  
U.S. Department of Health and Human Services Food and Drug Administration Center for Drug Evaluation and Research Division of Data Management and Services

LOINC® from Regenstrief

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Logical Observation Identifiers Names and Codes (LOINC®)

A universal code system for identifying laboratory and clinical observations. From serum levels of hepatitis B surface antigen to diastolic blood pressure, LOINC provides standardized terms for all kinds of measurements and observations that enable exchange and aggregation of clinical data.

How do you say glucose?

LOINC® is the largest source of clinical observation exchange

NCBI Taxonomy Browser

Published Entries BLAST OMM Taxonomy Search

Search for: As: complete name lock Go Clear

The NCBI Taxonomy Homepage

These are direct links to some of the organisms commonly used in molecular research projects:

Arabidopsis thaliana	Escherichia coli	Pneumocystis carinii
Bos taurus	Hepatitis C virus	Rattus norvegicus
Caenorhabditis elegans	Homo sapiens	Saccharomyces cerevisiae
Chlamydomonas reinhardtii	Mus musculus	Schizosaccharomyces pombe
Danio rerio (zebrafish)	Mycoplasma pneumoniae	Takifugu rubripes
Dicystostelium discoideum	Oryza sativa	Xenopus laevis
Drosophila melanogaster	Plasmodium falciparum	Zea mays

# How do we come to terms with this Biomedical Tower of Babel?

- Recognize the value of curating data with standard terminologies
  - Recognize that a variety of communities of practice exist
    - Encourage consistency within those communities of practice
  - Discourage development of “redundant” terminologies/ontologies
  - Map terminologies to each other for maximum semantic interoperability
  - Develop robust NLP tools that take advantage of existing terminologies

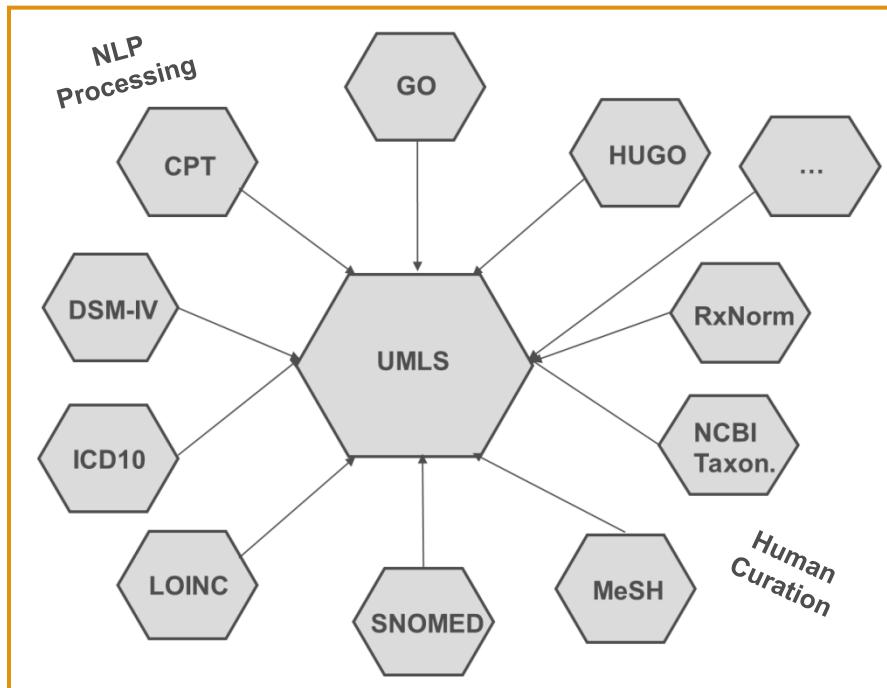


Unified Medical Language System (UMLS) is one large-scale effort that has made progress toward facilitating semantic interoperability of biomedical data



# UMLS

- Integrates > 100 existing terminologies/ontologies
- Includes a higher level ontology
- Includes natural language processing tools and resources



## UMLS Knowledge Sources

Metathesaurus

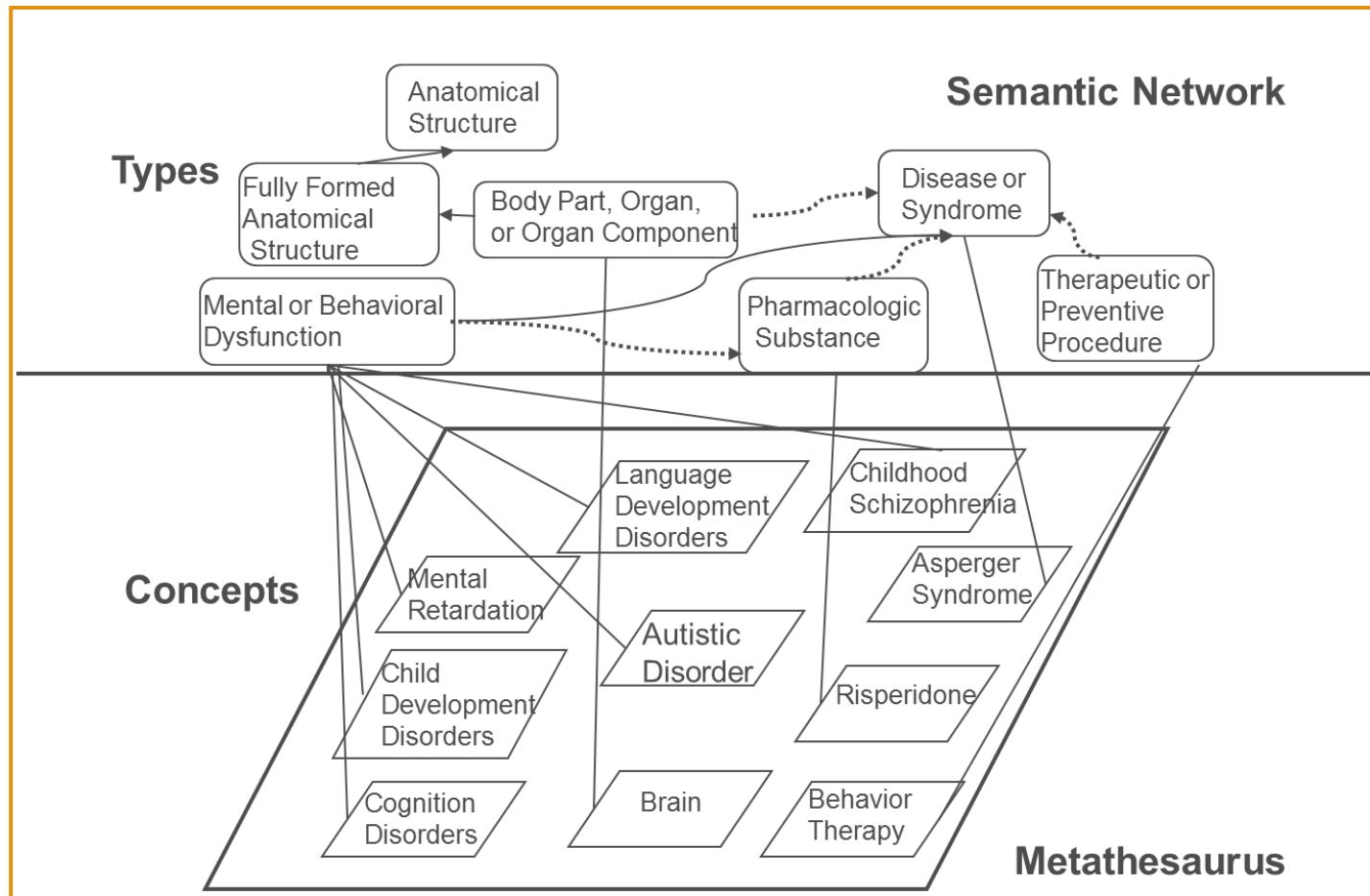
Semantic Network

SPECIALIST NLP Tools

# UMLS Metathesaurus Mapping Example

	<b>CUI</b>	<b>LUI</b>	<b>SUI</b>	<b>Source</b>
Renal Cell Carcinoma	C0007134	L0007134	S0399999	MeSH
Renal Cell Carcinomas	C0007134	L0007134	S0081526	MeSH
Grawitz Tumor	C0007134	L0018219	S0375417	SNOMED-CT
Hypernephroma	C0007134	L0020489	S0050424	MedDRA
Nephroid Carcinoma	C0007134	L0027710	S0065974	MeSH
Adenocarcinoma of kidney	C0007134	L0493923	S4147170	NCI Thesaurus
Renal Carcinoma	C0007134	L0161908	S1629563	OMIM

# UMLS Two-Level Structure





## UMLS Terminology Services

Welcome back, amccray

## Metathesaurus Browser

UTS Home Applications SNOMED CT Resources

Downloads Documentation UMLS Home

Search Tree Recent Searches

Term  CUI  Code  
  
  
 Release:  
  
 Search Type:  
  
 Source:  

- 
- 
- 
-

## Search Results (114)

[ : 1 - 25 :  ]

- [C0006413](#) Burkitt Lymphoma
- [C0023449](#) Acute lymphocytic leukemia
- [C0023452](#) Leukemia, Lymphocytic, Acute, L1
- [C0023453](#) Leukemia, Lymphocytic, Acute, L2
- [C0023467](#) Leukemia, Myelocytic, Acute
- [C0153876](#) Acute lymphoid leukemia in remission
- [C0349637](#) Common acute lymphoblastic leukemia
- [C0745701](#) LEUKEMIA ACUTE LYMPHOBLASTIC C
- [C0751606](#) Adult Acute Lymphocytic Leukemia
- [C1292769](#) Precursor B-cell lymphoblastic leukemia

Basic View Report View Raw View

-  **Concept: [C0023449] Acute lymphocytic leukemia**
-  **Semantic Types**
  - [Neoplastic Process \[T191\]](#)
-  **Definitions**
-  **Atoms (318)** string [AUI / RSAB / TTY / Code]
-  **Contexts (168)**
  -  [CSP/PT/acute lymphocytic leukemia \(6\)](#)
  -  [CST/PT/ACUTE LYMPHOBLASTIC LEUKEMIA \(1\)](#)
  -  [CST/PT/ACUTE LYMPHOBLASTIC LEUKEMIA \(1\)](#)
  -  [ICD10/PT/Acute lymphoblastic leukaemia \(1\)](#)
  -  [C91.0/Acute lymphoblastic leukaemia \[Context 1\]](#)
    -  **Ancestors**
      -  [International Statistical Classification of Diseases and Related Health Problems, Tenth Revision \(ICD-10\)](#)
        -  [Neoplasms](#)
        -  [Malignant neoplasms](#)
        -  [Malignant neoplasms of lymphoid, haematopoietic and related tissue](#)
        -  [Lymphoid leukaemia](#)
    -  [ICD10AM/HT/Acute lymphoblastic leukaemia \(1\)](#)
    -  [ICD10CM/HT/Acute lymphoblastic leukemia \[ALL\] \(1\)](#)
    -  [ICD9CM/HT/Lymphoid leukemia, acute \(1\)](#)





List

2012AB

- ⊕ (A) Entity
- ⊖ (B) Event
- ⊕ (B1) Activity
- ⊖ (B2) Phenomenon or Process
  - ⊕ (B2.1) Human-caused Phenomenon or Process
  - ⊖ (B2.2) Natural Phenomenon or Process
    - ⊖ (B2.2.1) Biologic Function
      - ⊕ (B2.2.1.1) Physiologic Function
        - ⊕ (B2.2.1.1.1) Organism Function
        - (B2.2.1.1.2) Organ or Tissue Function
        - (B2.2.1.1.3) Cell Function
      - ⊕ (B2.2.1.1.4) Molecular Function
    - ⊖ (B2.2.1.2) Pathologic Function
      - ⊖ (B2.2.1.2.1) Disease or Syndrome
        - (B2.2.1.2.1.1) Mental or Behavioral Dysfunction
        - (B2.2.1.2.1.2) Neoplastic Process
        - (B2.2.1.2.2) Cell or Molecular Dysfunction
        - (B2.2.1.2.3) Experimental Model of Disease
  - (B2.3) Injury or Poisoning

Report

Raw Report

**Neoplastic Process**⊖ **Definition**

A new and abnormal growth of tissue in which the growth is uncontrolled and progressive. growths may be malignant or benign.

⊖ **Properties**

Unique Identifier: T191

Tree Number: B2.2.1.2.1.2

Usage Note: All neoplasms are assigned to this type. Do not also assign a type from the 'Anatomical Abnormality' hierarchy.

⊖ **Parents**[Disease or Syndrome](#)⊖ **Relations**Neoplastic Process [isa Disease or Syndrome \(DNI\)](#)⊕ **Inherited Relations**⊕ **Inverse Inherited Relations**

# Terminologies in Use

- ClinicalTrials.gov
- Harvard Catalyst
- Correlating Phenotype & Genotype in Autism Research

***ClinicalTrials.gov***

A service of the U.S. National Institutes of Health



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# ClinicalTrials.gov

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ClinicalTrials.gov is a registry and results database of publicly and privately supported clinical studies of human participants conducted around the world. [Learn more about clinical studies](#) and [about this site](#), including relevant [history, policies, and laws](#).

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ClinicalTrials.gov currently lists **141,003 studies** with locations in all 50 states and in **182 countries**.

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Example: "Heart attack" AND "Los Angeles"

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## Search Help

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## Locations of Recruiting Studies



Non-U.S. Only (49%)  
U.S. Only (44%)  
Both U.S. & Non-U.S. (7%)

Total N = 29,895 studies

Data as of February 22, 2013

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## For Patients & Families

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# ClinicalTrials.gov

A service of the U.S. National Institutes of Health

Example: "Heart attack" AND "Los Angeles"

Search for studies:

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Text Size ▾

**75 studies found for:** lou gehrigs disease | Open Studies

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Rank	Status	Study
------	--------	-------

1	Recruiting	<a href="#">Phase II/III Randomized, Placebo-controlled Trial of Arimoclomol in SOD1 Positive Familial Amyotrophic Lateral Sclerosis</a>
---	------------	--

**Conditions:** Amyotrophic Lateral Sclerosis 1; Acquired Amyotrophic Lateral Sclerosis; Amyotrophic Lateral Sclerosis

**Intervention:** Drug: Arimoclomol

2	Recruiting	<a href="#">Genetics of ALS: Identification of Genes With Roles in Familial and Sporadic Amyotrophic Lateral Sclerosis (ALS) and Amyotrophic Lateral Sclerosis (ALS) With Frontotemporal Dementia</a>
---	------------	---

**Conditions:** Amyotrophic Lateral Sclerosis; Familial Amyotrophic Lateral Sclerosis; Amyotrophic Lateral Sclerosis With Frontotemporal Dementia; Lou Gehrig's Disease; Motor Neuron Disease; Primary Lateral Sclerosis

**Intervention:** Other: Genetic study of ALS families

3	Recruiting	<a href="#">The National Amyotrophic Lateral Sclerosis Registry</a>
---	------------	---

**Condition:** Amyotrophic Lateral Sclerosis

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Example: "Heart attack" AND "Los Angeles"

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Rank	Status	Study
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1 Recruiting

[Phase II/III Randomized, Placebo-controlled Trial of Arimoclomol in SOD1 Positive Familial Amyotrophic Lateral Sclerosis](#)

**Conditions:** Amyotrophic Lateral Sclerosis 1; Acquired Amyotrophic Lateral Sclerosis; Amyotrophic Lateral Sclerosis

**Intervention:** Drug: Arimoclomol

2 Recruiting

[Genetics of ALS: Identification of Genes With Roles in Familial and Sporadic Amyotrophic Lateral Sclerosis \(ALS\) and Amyotrophic Lateral Sclerosis \(ALS\) With Frontotemporal Dementia](#)

**Conditions:** Amyotrophic Lateral Sclerosis; Familial Amyotrophic Lateral Sclerosis; Amyotrophic Lateral Sclerosis With Frontotemporal Dementia; Lou Gehrig's Disease; Motor Neuron Disease; Primary Lateral Sclerosis

**Intervention:** Other: Genetic study of ALS families

3 Recruiting

[The National Amyotrophic Lateral Sclerosis Registry](#)

**Condition:** Amyotrophic Lateral Sclerosis



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Example: "Heart attack" AND "Los Angeles"

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## Recognized Terms and Synonyms:

lou gehrigs disease: 214 studies

als1

amyotrophic lateral sclerosis

autosomal dominant als

bulbar motor neuron disease

charcot disease

family amyotrophic lateral sclerosis

gehrig disease

lou gehrig disease

motor neuron disease, amyotrophic lateral sclerosis

Found 214

No Query

Recogniz

lou gehrig

als1

amyotroph

autosomal

bulbar mo

charcot di

family am

gehrig disea

lou gehrig disea

motor neuron disease, amyotrophic lateral sclerosis



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Translation to Patients

**T3**  
Translation to Practice

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Search our website using the power of biomedical vocabularies and semantic technologies.

## Semantic Search

APOE



**Apolipoproteins E** A class of protein components which can be found in several lipoproteins including HIGH-DENSITY LIPOPROTEINS; VERY-LOW-DENSITY LIPOPROTEINS; and CHYLOMICRONS. Synthesized in most organs, Apo E is important in the global transport of lipids and cholesterol throughout the body. Apo E is also a ligand for LDL receptors (RECEPTORS, LDL) that mediates the binding, internalization, and catabolism of lipoprotein particles in cells. There are several allelic isoforms (such as E2, E3, and E4). Deficiency or defects in Apo E are causes of HYPERLIPOPROTEINEMIA TYPE III.

Search APOE as text only

All Results (1000+)

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"Apolipoproteins E" is a descriptor in the National Library of Medicine's controlled vocabulary thesaurus, MeSH (Medical Subject Headings). Descriptors are ...

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*Arterioscler Thromb Vasc Biol.* Feb 1, 2010. MENDIVIL CO, ZHENG C, FURTADO JD, LEL J, SACKS FM

Matching Topic: **Apolipoproteins E**

[Green, Robert - Harvard Catalyst Profiles - Harvard University](#)

Apolipoprotein E (APOE) genotype has dissociable effects on memory and attentional-executive network function in Alzheimer's disease. *Proc Natl Acad Sci U S ...*

## Topic Navigator

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*Am J Physiol Lung Cell Mol Physiol.* Jan 1, 2010. HANSMANN G, RABINOVITCH M

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*Arterioscler Thromb Vasc Biol.* Oct 1, 2009. NAHRENDORF M, WATERMAN P, THURBER GM, GROVES K, RAJOPADHYE M, PANIZZI P, MARINELLI B, AIKAWA ER, PITTEL MJ, SWIRSKI FK, WEISSLEDER R

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*Atherosclerosis.* Oct 1, 2009. SCHÖDEL J, PADMAPRIYA P, MARX A, HUANG PL, ERTL G, KUHLENCORDT PJ

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1

Vascular effects of a low-carbohydrate high-protein diet. [Medvane](#) [PubMed](#) [details »](#)

*Proc Natl Acad Sci U S A.* Sep 8, 2009. FOO SY, HELLER ER, WYKRZYKOWSKA JJ, SULLIVAN CJ, MANNING-TOBIN JJ, MOORE KJ, GERSZTEN RE, ROSENZWEIG A

Matching Topic: **Apolipoproteins E**



# Correlating Phenotype & Genotype

## Phenotypic and Genetic Factors in Autism Spectrum Disorders

- ~ 500 families participated
- Data collected
  - Phenotypic data from 25 behavioral instruments
  - Biological samples
- Developed autism-specific ontology
  - Aligned and integrated terminology represented in phenotypic instruments
  - Linked to UMLS whenever possible
  - Ontology will be openly available

# Autism Ontology

## Scope

- ~5,000 questions from 25 instruments mapped into ~300 concepts
- Three high-level groupings
  - Personal Traits
  - Social Competence
  - Medical History

## Use

- Concept-based search of Autism Consortium phenotype data
  - Genotypic data correlated with phenotypic characteristics
- Query, aggregate, and integrate data for hypothesis-driven research



## Concluding Remarks

***Precision Medicine is a Grand Challenge that is worth pursuing.***

**Its realization will involve addressing many social, legal, ethical, economic, and technical issues.**

**If we put our minds to it, the technical issues can be among the first to be solved, and may even show the way to the resolution of some of the even thornier issues.**

**Semantic interoperability is one technical issue that can be solved.**