

# 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

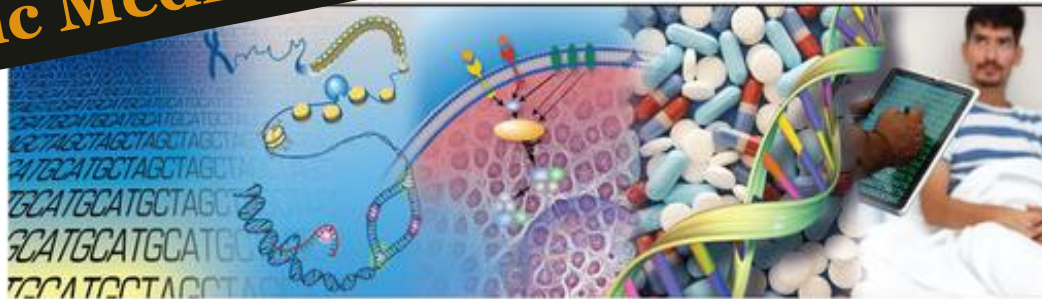


*“No aspect of human life has escaped the impact of the Information Age, and perhaps in no area of life is information more critical than in health and medicine.”*

U.S. National Academy of Engineering  
Grand Challenges 2008

<http://www.engineeringchallenges.org>

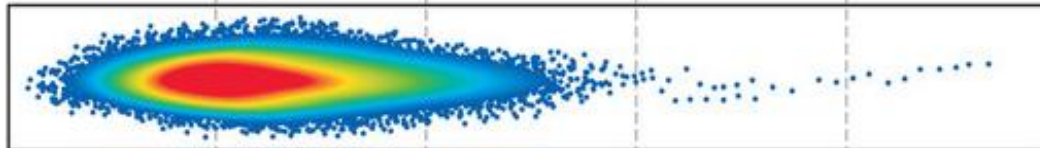
# The Promise of Genomic Medicine



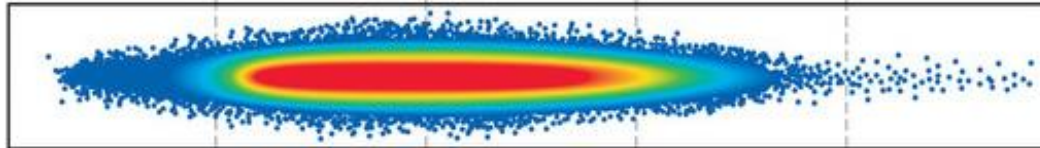
1990-2003  
Human Genome  
Project



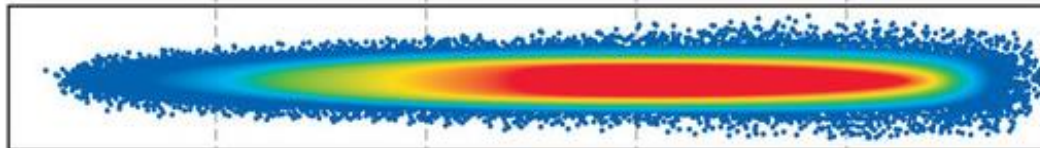
2004-2010



2011-2020

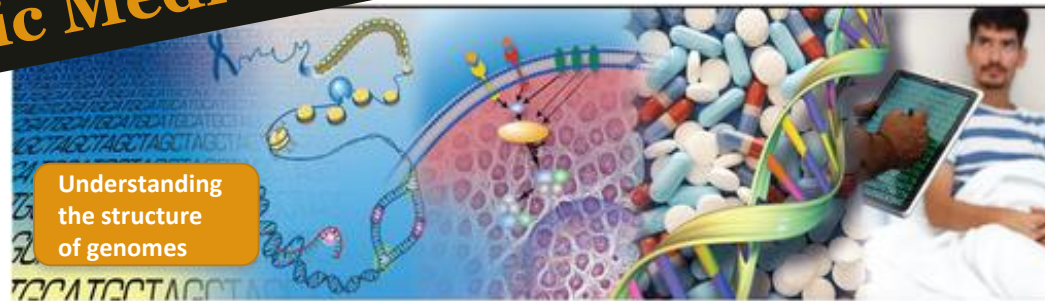


Beyond 2020



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

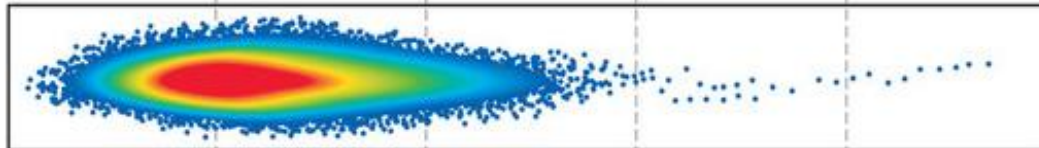
# The Promise of Genomic Medicine



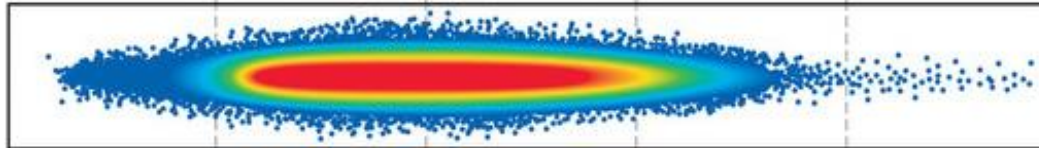
1990-2003  
Human Genome  
Project



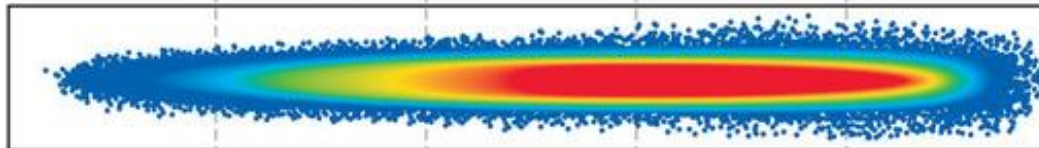
2004-2010



2011-2020



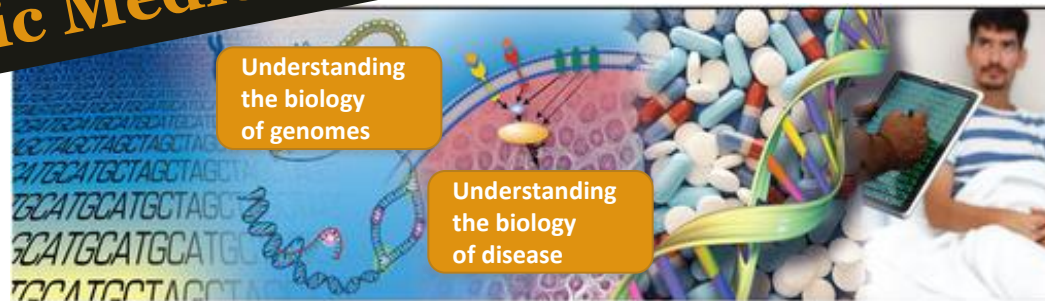
Beyond 2020



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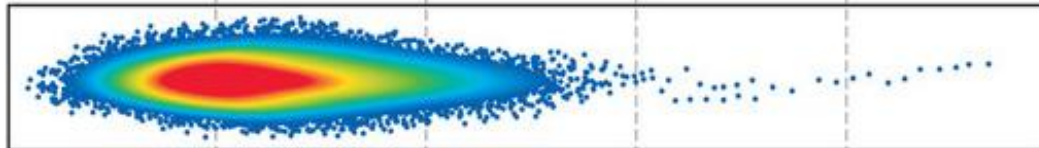
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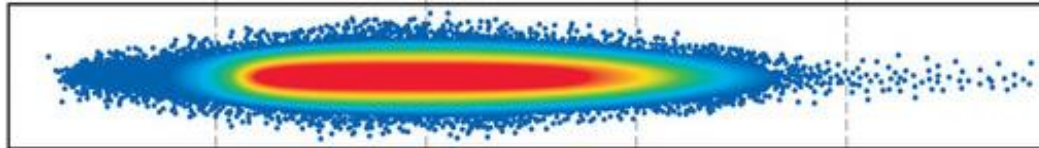
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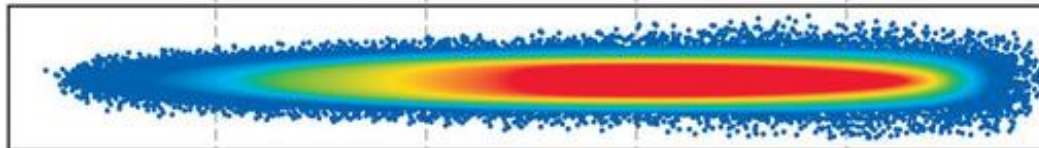
2004-2010



2011-2020

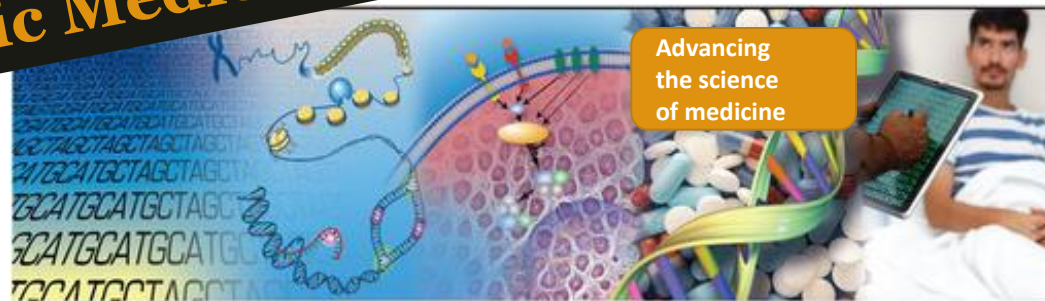


Beyond 2020



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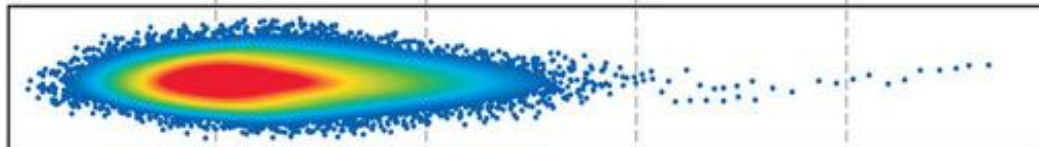
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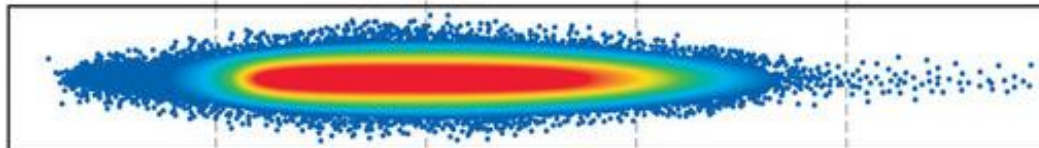
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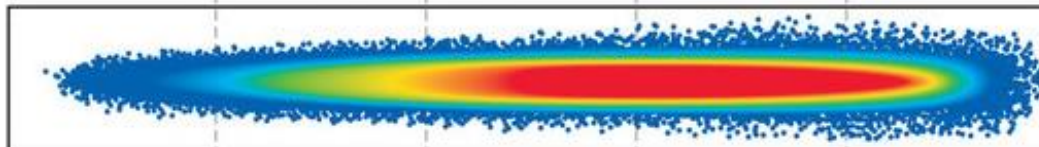
2004-2010



2011-2020

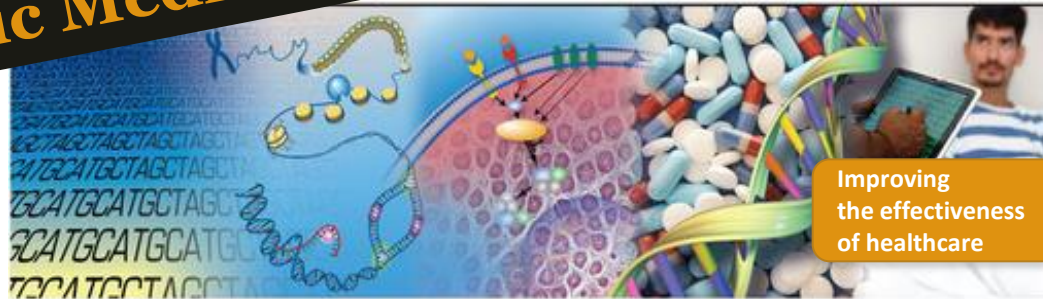


Beyond 2020



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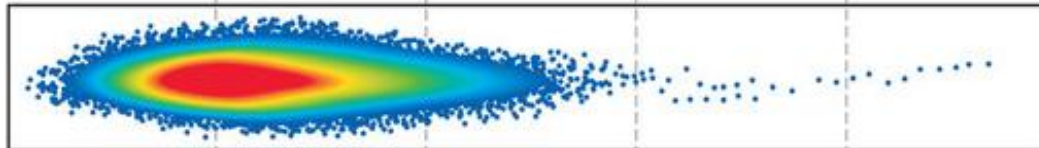
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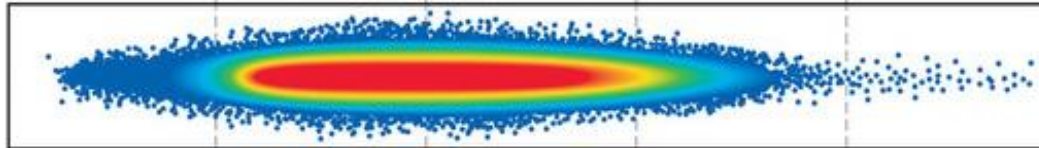
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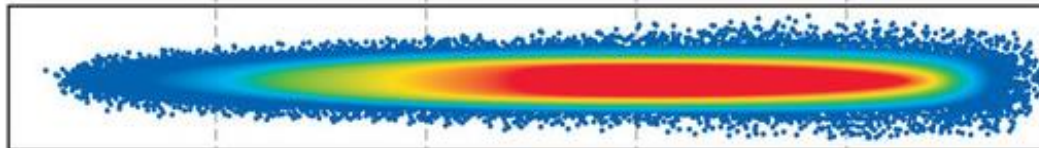
2004-2010



2011-2020



Beyond 2020



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

# 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.”

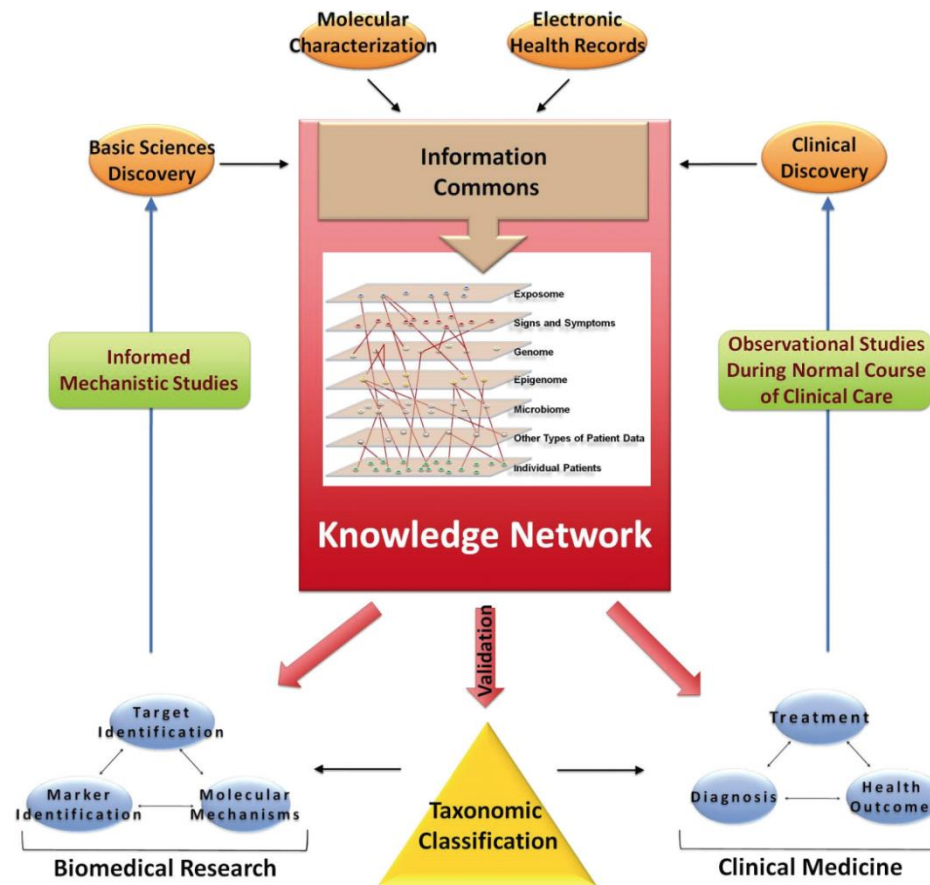
*Toward Precision Medicine: Building a Knowledge Network for Biomedical Research and a New Taxonomy of Disease*

National Research Council, 2011:9.



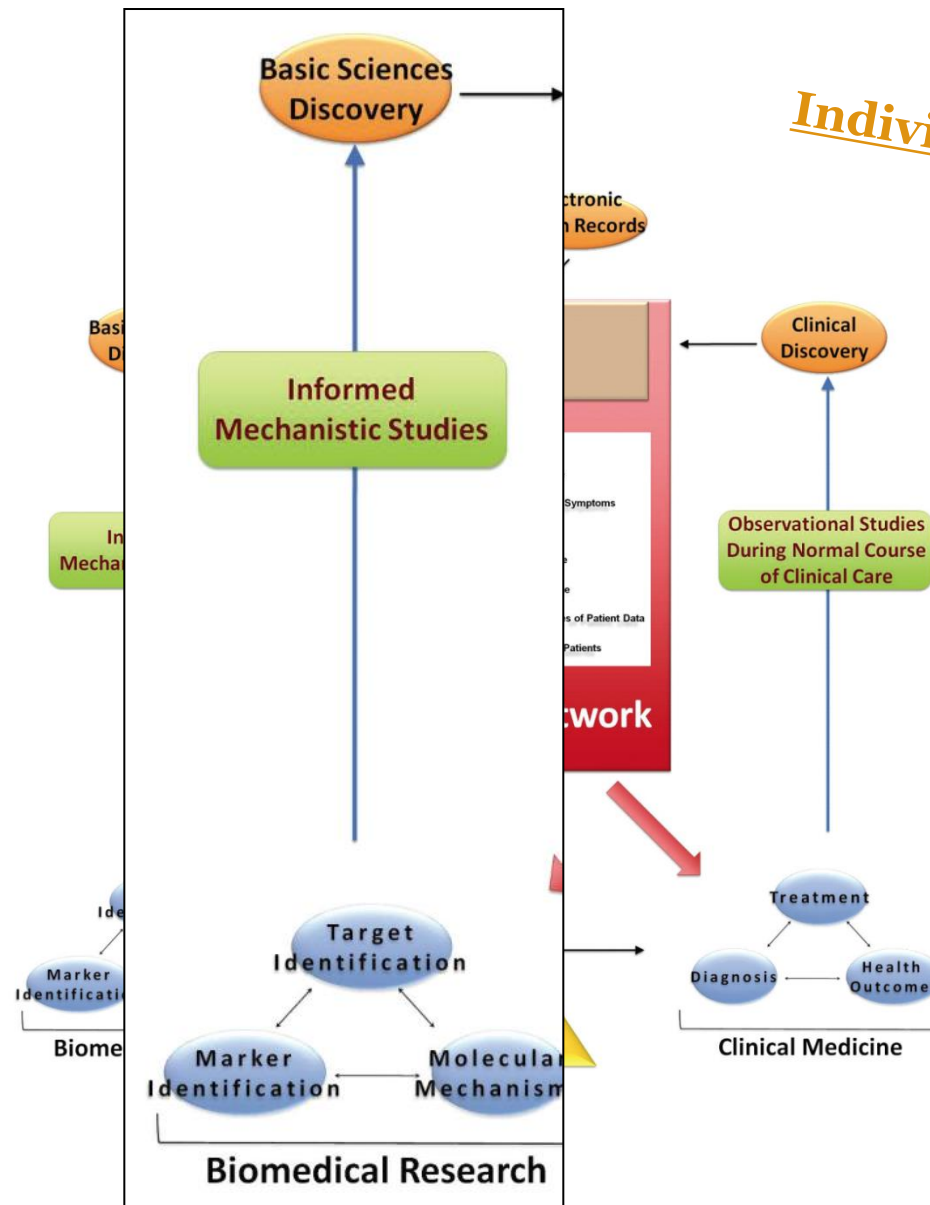
# Precision Medicine

# Individual-Centric



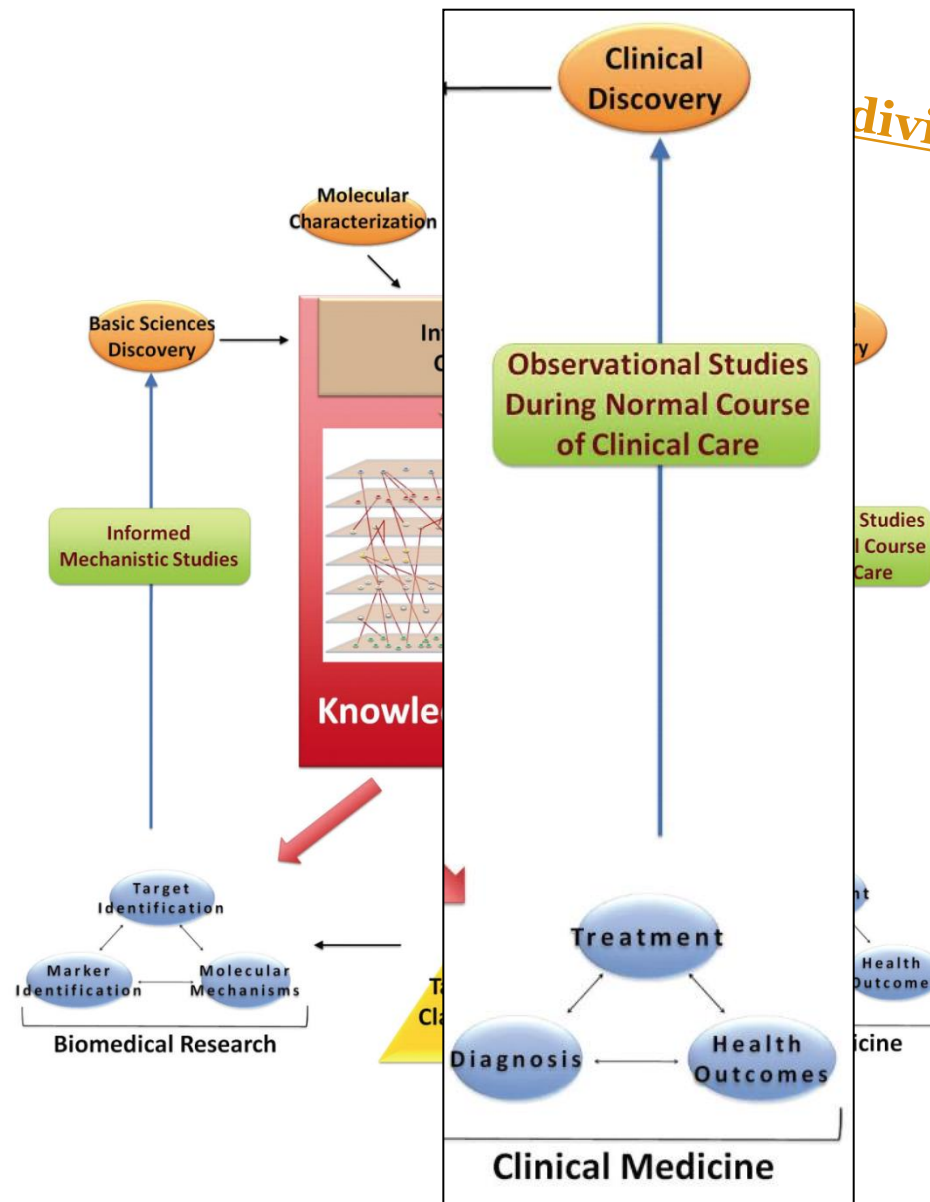
*Toward Precision Medicine*  
National Research Council, 2011

# Precision Medicine



*Toward Precision Medicine*  
National Research Council, 2011

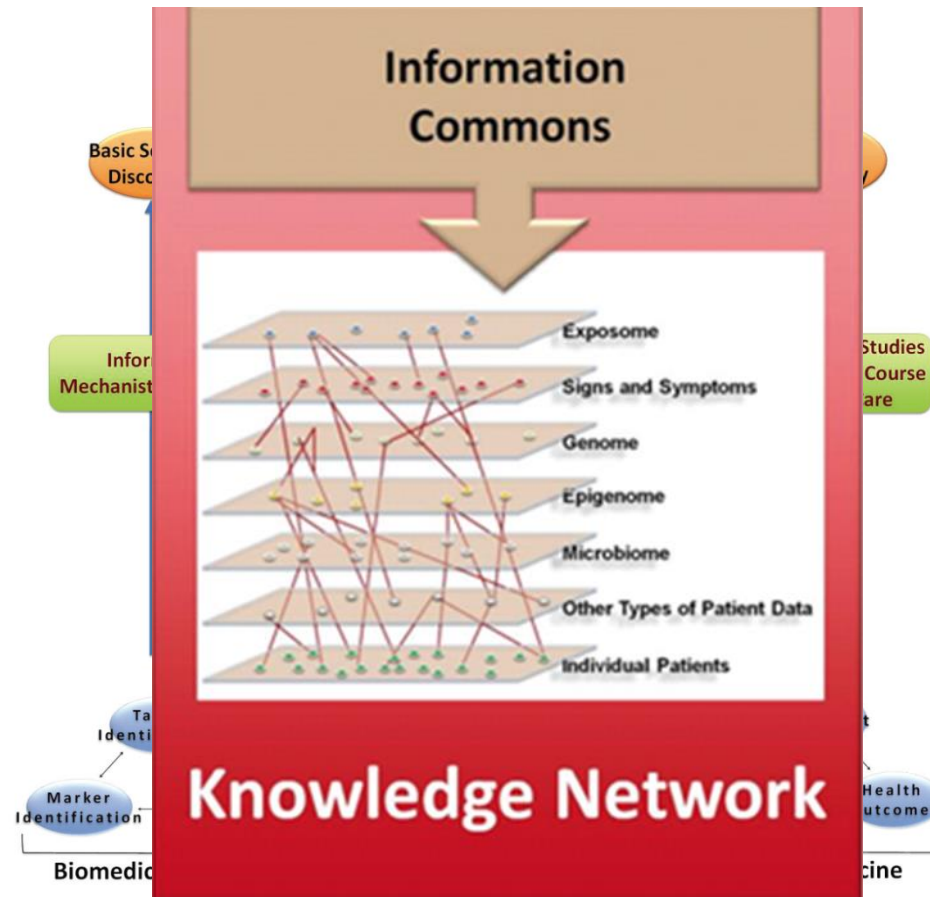
# Precision Medicine



*Toward Precision Medicine*  
National Research Council, 2011

# Precision Medicine

## Individual-Centric

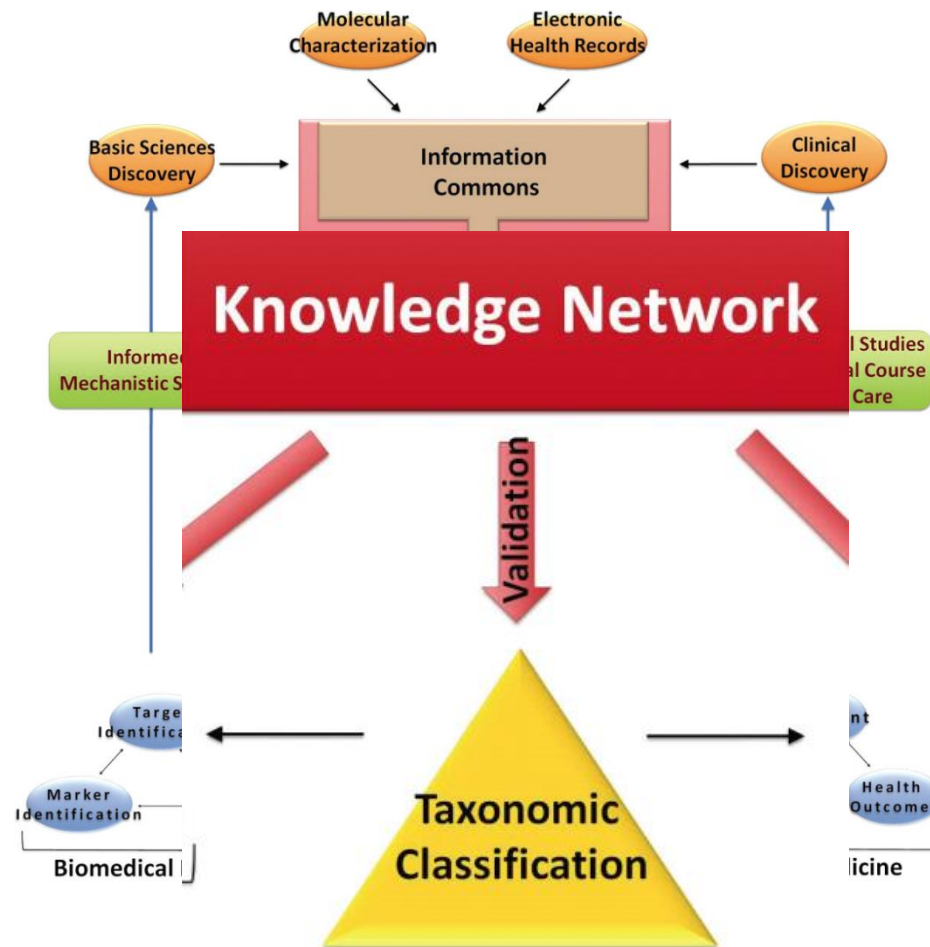


*Toward Precision Medicine*  
National Research Council, 2011



# Precision Medicine

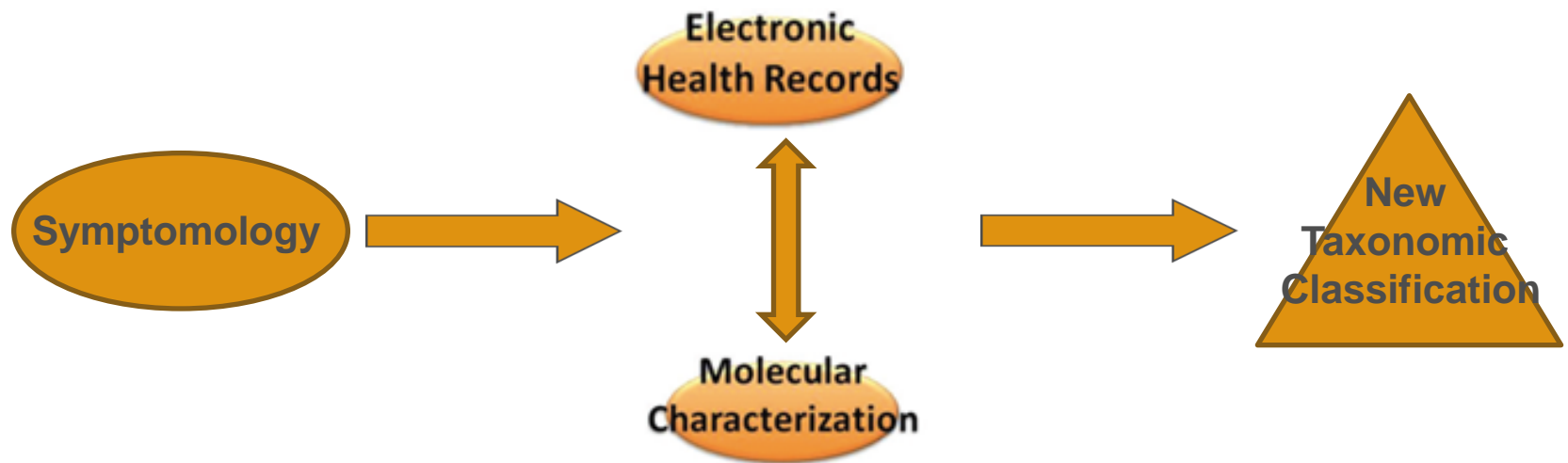
## Individual-Centric



*Toward Precision Medicine*  
National Research Council, 2011

# A New Taxonomy of Disease

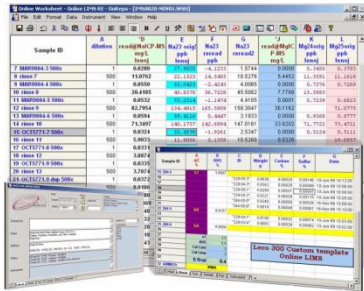
*“Could it be that something as fundamental as our current system for classifying diseases is actually inhibiting progress?”*



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

# Wide Range of Data Sources Needed

## Laboratory Data

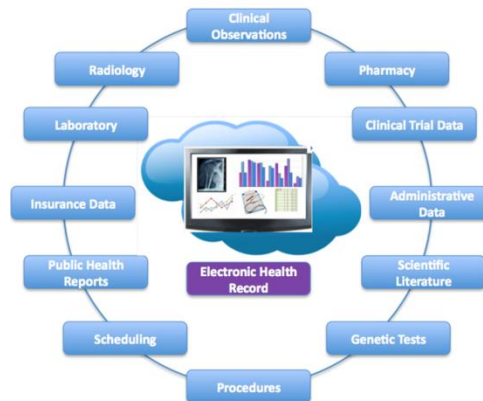


## Biosensor Data



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

## Clinical Systems



## Public Databases

A screenshot of the NCBI Databases and Tools website. The header includes the NCBI logo and the text 'Databases and Tools'. Below the header is a navigation menu with links: About NCBI, NCBI at a Glance, A Science Primer, Databases and Tools, Human Genome Resources, Model Organisms Guide, Outreach and Education, and News. The main content area lists various databases and tools: Literature Databases, Entrez Databases, Nucleotide Databases, Genome-Specific Resources, Tools for Data Mining, Tools for Sequence Analysis, Tools for 3-D Structure Display and Similarity Searching, Maps, Collaborative Cancer Research, FTP Download Sites, and Resource Statistics.

## Social Networking Sites

A screenshot of the PatientsLikeMe website. The header includes the PatientsLikeMe logo and the text 'patientslikeme'. Below the header is a navigation menu with links: Home, About, Contact, and Help. The main content area features a grid of user avatars and a testimonial: 'Because of PatientsLikeMe, we are better able to recognize warning signs... (and) keep things in perspective. In short, PatientsLikeMe empowers us.' Another testimonial states: 'PatientsLikeMe has provided me with new friends - people who are experiencing the same problems as I am.'

# 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**

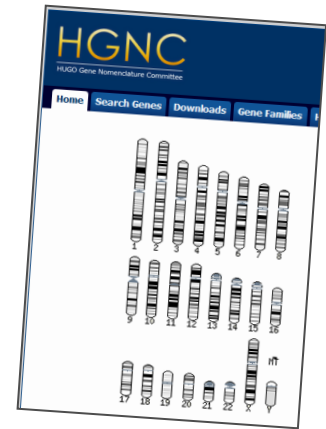
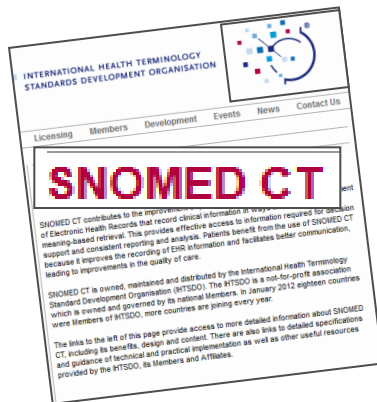
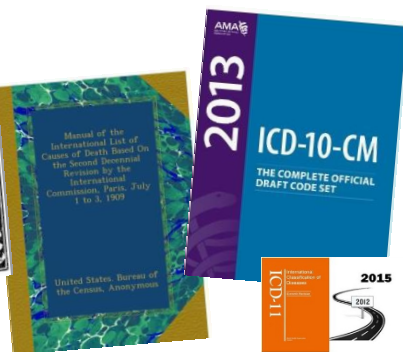
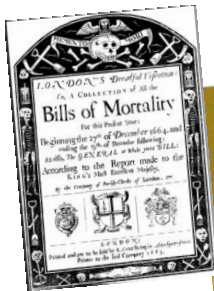
**Natural language processing**

**...**



# Common Terminologies

Varying scope, coverage, rigor, and update schedules



# 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

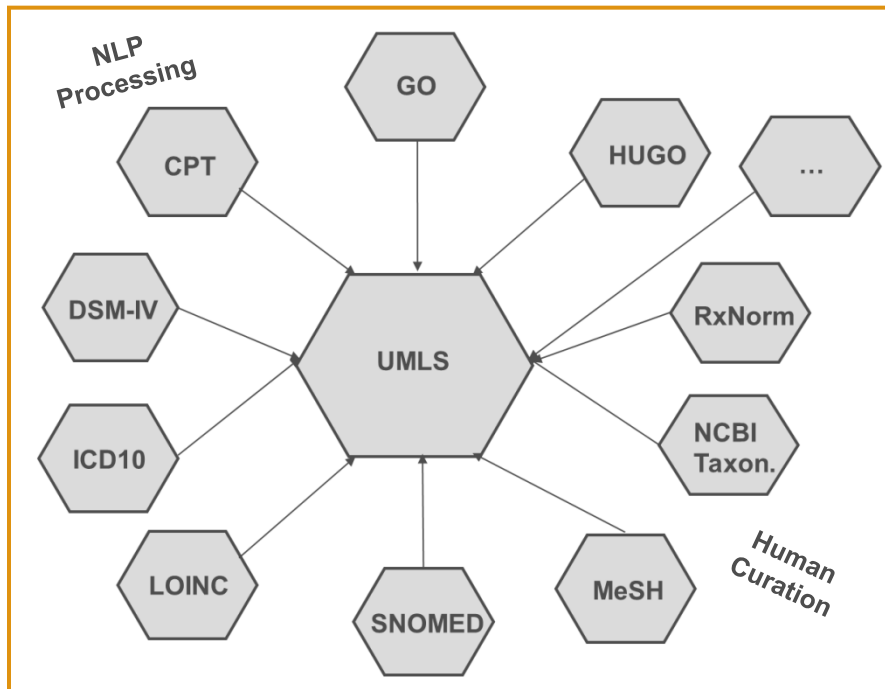


Unified Medical Language System (UMLS) is one large-scale effort that has made progress toward facilitating semantic interoperability of biomedical data



# UMLS

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



## UMLS Knowledge Sources

Metathesaurus

Semantic Network

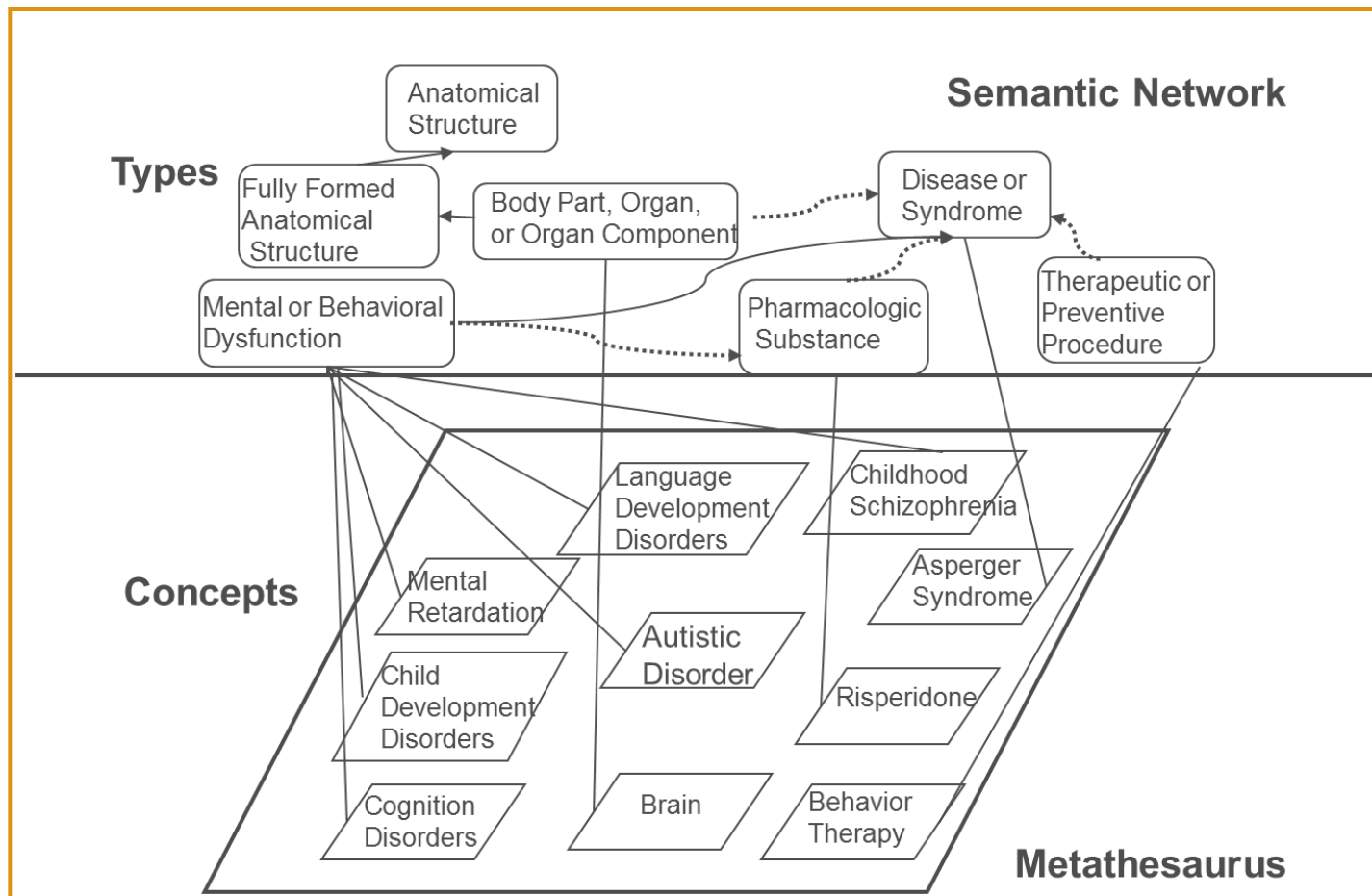
SPECIALIST NLP Tools

# UMLS Metathesaurus Mapping Example

	CUI	LUI	SUI	Source
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



Unified Medical  
Language System®

# UMLS Terminology Services

## Metathesaurus Browser

Welcome back, amccray

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acute lymphoblastic leukemia

Go

Release: 2012AB

Search Type: Word

Source: All Sources

AIR

ALT

AOD

AOT

**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)



Unified Medical  
Language System®

# UMLS Terminology Services

## Semantic Network Browser

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UTS Home Applications SNOMED CT Resources Downloads Documentation UMLS Home

List Tree

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

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

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**. [Text Size](#)

## Search for Studies

Example: "Heart attack" AND "Los Angeles"



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## Search Help

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## Locations of Recruiting Studies

Location	Percentage
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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CT Search of: lou gehrigs disease | Open Stu... +

clinicaltrials.gov/ct2/results/displayOpt?flds=a&flds=b&submit\_fld\_opt=on&term=lou+gehrigs+disease&recr=Open&show\_flds=Y

Example: "Heart attack" AND "Los Angeles"

Search for studies:  Search

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**ClinicalTrials.gov**  
A service of the U.S. National Institutes of Health

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**75 studies found for:** lou gehrigs disease | Open Studies  
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☒ Include only open studies ☐ Exclude studies with unknown status

Rank	Status	Study
1	Recruiting	<p><a href="#">Phase II/III Randomized, Placebo-controlled Trial of Arimoclomol in SOD1 Positive Familial Amyotrophic Lateral Sclerosis</a></p> <p><b>Conditions:</b> Amyotrophic Lateral Sclerosis 1; Acquired Amyotrophic Lateral Sclerosis; Amyotrophic Lateral Sclerosis</p> <p><b>Intervention:</b> Drug: Arimoclomol</p>
2	Recruiting	<p><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></p> <p><b>Conditions:</b> Amyotrophic Lateral Sclerosis; Familial Amyotrophic Lateral Sclerosis; Amyotrophic Lateral Sclerosis With Frontotemporal Dementia; Lou Gehrig's Disease; Motor Neuron Disease; Primary Lateral Sclerosis</p> <p><b>Intervention:</b> Other: Genetic study of ALS families</p>
3	Recruiting	<p><a href="#">The National Amyotrophic Lateral Sclerosis Registry</a></p> <p><b>Condition:</b> Amyotrophic Lateral Sclerosis</p>

Firefox

CT Search of: lou gehrigs disease | Open Stu... +

clinicaltrials.gov/ct2/results/displayOpt?flds=a&flds=b&submit\_fld\_opt=on&term=lou+gehrigs+disease&recr=Open&show\_flds=Y

Example: "Heart attack" AND "Los Angeles"

Search for studies:  Search

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**Search Details**

Rank	Status	Study
1	Recruiting	<p><a href="#">Phase II/III Randomized, Placebo-controlled Trial of Arimoclomol in SOD1 Positive Familial Amyotrophic Lateral Sclerosis</a></p> <p><b>Conditions:</b> Amyotrophic Lateral Sclerosis 1; Acquired Amyotrophic Lateral Sclerosis; Amyotrophic Lateral Sclerosis</p> <p><b>Intervention:</b> Drug: Arimoclomol</p>
2	Recruiting	<p><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></p> <p><b>Conditions:</b> Amyotrophic Lateral Sclerosis; Familial Amyotrophic Lateral Sclerosis; Amyotrophic Lateral Sclerosis With Frontotemporal Dementia; Lou Gehrig's Disease; Motor Neuron Disease; Primary Lateral Sclerosis</p> <p><b>Intervention:</b> Other: Genetic study of ALS families</p>
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clinicaltrials.gov/ct2/results/details?term=lou+gehrigs+disease

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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

familial 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

familial am

gehrig disease

lou gehrig disease

motor neuron disease, amyotrophic lateral sclerosis





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Basic Scientific Discovery

**T2** Translation to Patients  
Clinical Insights

**T3** Translation to Practice  
Implications for Practice

**T4** Translation to Population Health  
Implications for Population Health Improved Global Health

**Find...**

- Collaborators
- Core Facilities
- Courses
- Events
- Maps, Directions, & more

**Request...**

- Clinical Research Support
- Consultations

How do I...

**Grant Opportunities**


The Grant Opportunities tool is being updated and will return soon.

**Elements of Grant Writing**

Writing Tips  
Samples  
Tools  
Ready & Not?

Need tips for planning and writing your grant?  
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**Spotlight**



**I am Harvard Catalyst: Shamsah Kazani, MD, MMSc**

Access to Unmatched Resources:  
A Harvard Catalyst KL2 awardee dedicates two years to advancing her research goals in asthma.

Childhood Obesity Pilot Grant  
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Medical Device Development  
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**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.

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Engaging families and researchers  
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# 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.