National Human Genome Research Institute

National Institutes of Health

U.S. Department of Health and Human Services

NHGRI Genomic Medicine Activities

U.S. Department of Health and Human Services
National Institutes of Health
National Human Genome Research Institute

Teri Manolio, M.D., Ph.D.
Genomic Medicine IV, Dallas TX
January 29, 2013

NHGRI Genomic Medicine Definition August 2012

Genomic Medicine: *An emerging medical discipline that involves using genomic information about an individual as part of their clinical care (e.g., for diagnostic or therapeutic decision-making) and the other implications of that clinical use.*

- Purposefully narrow
- By 'genomic,' NHGRI means direct information about DNA or RNA; downstream products outside immediate view
- Dominant portion of NHGRI's portfolio will continue to support basic research underpinning genomic medicine
- Fourth and fifth NHGRI strategic plan domains capture research activities under umbrella of genomic medicine
- Metaphorically viewed as key 'destination' for attaining mission of improving health through genomics research

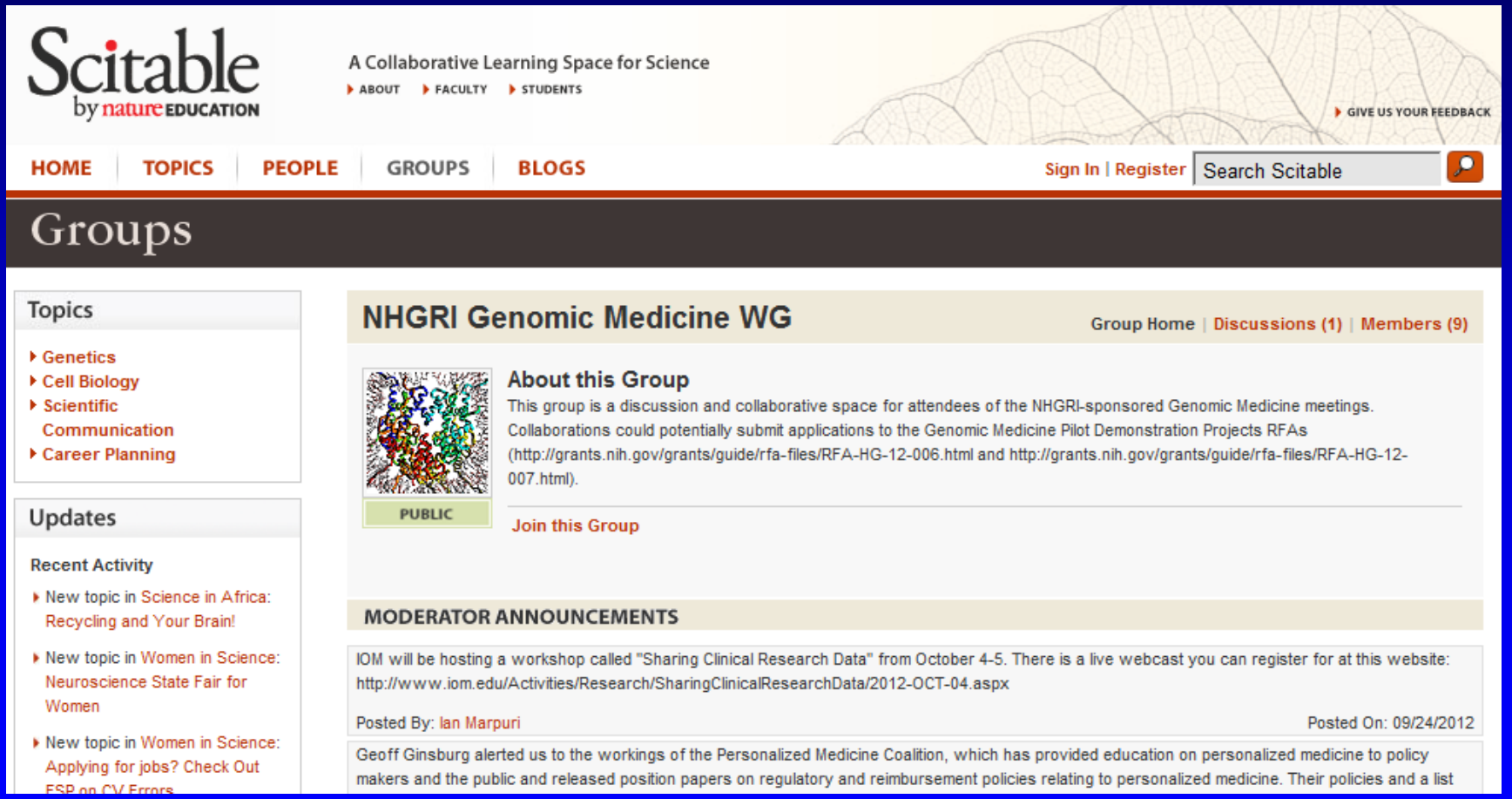
Genomic Medicine Working Group of National Advisory Council on Human Genome Research

- Plan Genomic Medicine meetings, 2-3 per yr
- Provide guidance to NHGRI in other areas of genomic medicine implementation, such as:
  - Outlining infrastructural needs for adoption of genomic medicine
  - Identifying related efforts for future collaborations
  - Reviewing progress overall in genomic medicine implementation

Genomic Medicine Working Group Members

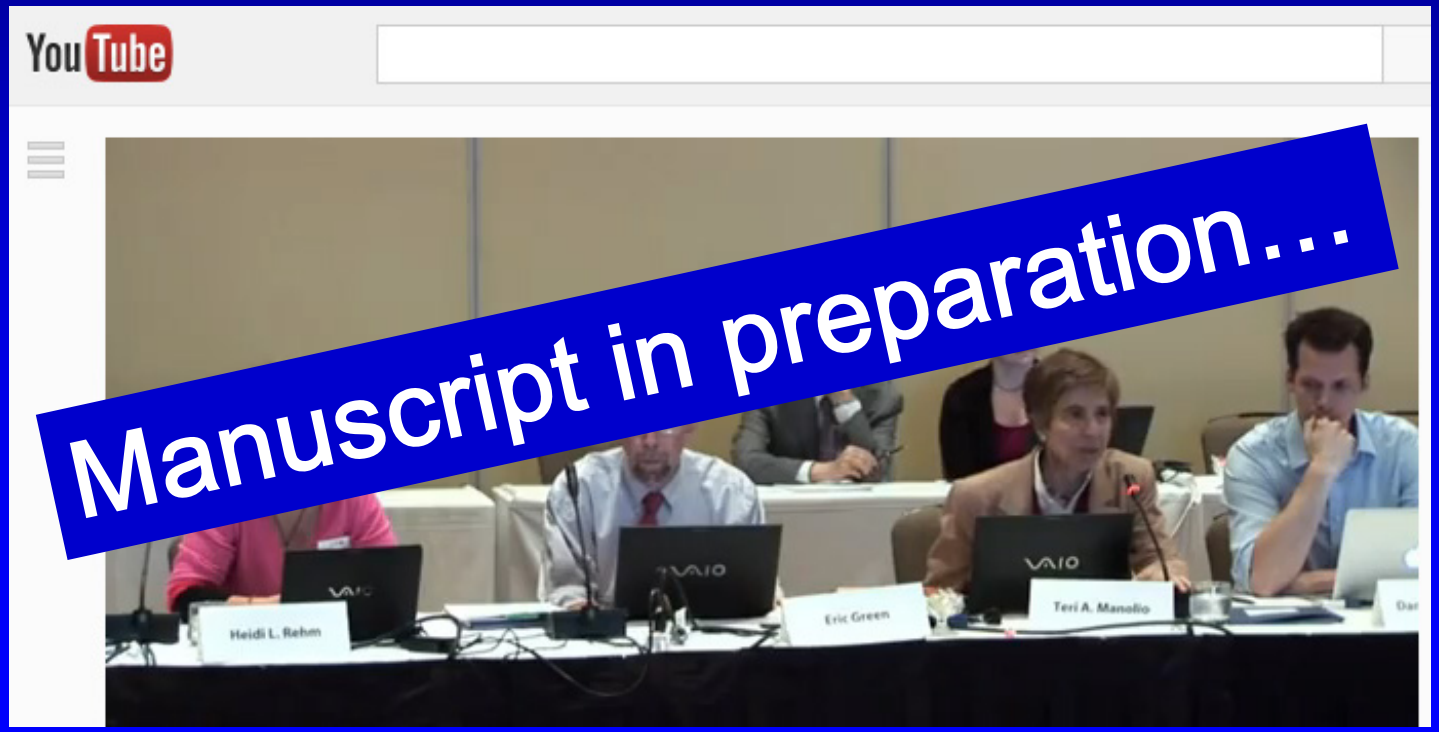
Rex Chisholm	Northwestern
Jim Evans	UNC
Geoff Ginsburg	Duke
Pearl O'Rourke	Partners
Mary Relling	St. Jude
Dan Roden	Vanderbilt
Marc Williams	Geisinger
Eric Green	NHGRI
Teri Manolio	NHGRI
Brad Ozenberger	NHGRI

Scitable Networking Site
Link through "Genomic Medicine Activities"
http://www.genome.gov/27549225



Implicating Sequence Variants in Human Disease Workshop: Sept 12-13, 2012

Goal: To develop guidelines for assessing the evidence implicating sequence variants or genes as causal in a specific disease.



Recent Advances in Genomic Medicine

RESEARCH ARTICLE

RESEARCH ARTICLE

RESEARCH ARTICLE

ORIGINAL ARTICLE

Aspirin Use, Tumor *PIK3CA* Mutation, and Colorectal-Cancer Survival

Xiaoyun Liao, M.D., Ph.D., Paul Lochhead, M.B., Ch.B., Reiko Nishihara, Ph.D., Teppei Morikawa, M.D., Ph.D., Aya Kuchiba, Ph.D., Mai Yamauchi, Ph.D., Yu Imamura, M.D., Ph.D., Zhi Rong Qian, M.D., Ph.D., Yoshifumi Baba, M.D., Ph.D., Kaori Shima, D.D.S., Ph.D., Ruifang Sun, M.B., Katsuhiko Nosho, M.D., Ph.D., Jeffrey A. Meyerhardt, M.D., M.P.H., Edward Giovannucci, M.D., M.P.H., Sc.D., Charles S. Fuchs, M.D., M.P.H., Andrew T. Chan, M.D., M.P.H., and Shuji Ogino, M.D., Ph.D.

isease

er Saffrey,[6]

ne Safina,[1,4]

http://www.genome.gov/27551536

Genomic Medicine Colloquium Report June 2011, Chicago, IL

REVIEW | Genetics inMedicine

Open

- Describe ongoing projects and challenges
- Identify common infrastructure and research needs
- Outline implementation framework for investigating and introducing similar programs elsewhere

Teri A… MD[3], M… [8], Mur… nD[12], Dav… D[15], Michae… armD[19], Alan … , PhD[1]

and Geoffrey S. Ginsburg, MD, PhD[23]

Although the potential for genomics to contribute to clinical care has long been anticipated, the pace of defining the risks and benefits of incorporating genomic findings into medical practice has been relevant; lack of reimbursement for genomically driven interventions; and burden to patients and clinicians of assaying, reporting, intervening, and following up genomic findings. Key infrastructure needs

Genet Med 2012 Jan 10; epub before print.

NHGRI Genomic Medicine Meetings, 2011

- GM Colloquium, June 2011, Chicago IL
  - Define landscape, identify commonalities
  - Develop implementation roadmap to share experiences and facilitate adoption
- GM II, December 2011, Bethesda MD
  - Identify potential collaborative projects
  - Explore requirements for adoption with institutional leaders

NHGRI Genomic Medicine Meetings, 2012-2013

- GM III, May 2012, Chicago IL
  - Review early progress from pilot project working groups
  - Explore implementation barriers and solutions with payers and other stakeholders
- Payers' Meeting, October 2012, Bethesda MD
  - Identify potential for collaborative research and joint funding
- GM IV, January 2013, Dallas TX
  - Professional societies' needs for physician education and guideline development

Rapid Evolution

- GM I. There is significant action in Genomic Medicine
- GM II. Healthcare providers care about Genomic Medicine
- GM III. Those who pay for healthcare care about Genomic Medicine
- GM IV. Professional organizations and physicians care about Genomic Medicine

Avoiding Meeting Hell

"Oh, man! The coffee's *cold!* They thought of *everything!*

Larson, G. *The Complete Far Side*. 2003.

Genomic Medicine Funding Opportunities

Department of Health and Human Services

Part 1. Overview Information

Participating Organization(s)	National Institutes of Health (NIH)
Components of Participating Organizations	*Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NICHD) National Human Genome Research Institute (NHGRI)
Funding Opportunity Title	**Genomic Sequencing and Newborn Screening Disorders (U19)**
Activity Code	U19 Research Program – Cooperative Agreements
Announcement Type	New
Related Notices	• August 15, 2012 - Informational/Technical Assistance Pre-application Meeting for RFA-HD-13-010. See Notice NOT-HD-12-027.
Funding Opportunity Announcement (FOA) Number	**RFA-HD-13-010**

Genomic Medicine Pilot Demonstration Projects: RFAs HG-12-006 and HG-12-007

Department of Health and Human Services

Part 1. Overview Information

Participating Organization(s)	National Institutes of Health (NIH)
Components of Participating Organizations	National Human Genome Research Institute (NHGRI)
Funding Opportunity Title	**Genomic Medicine Pilot Demonstration Projects (U01)**
Activity Code	U01 Research Project – Cooperative Agreements
Announcement Type	New
Related Notices	• June 20, 2012 - Frequently Asked Question (FAQs) for RFA-HG-12-006. See Notice NOT-HG-12-018.
Fund Anno	

Department of Health and Human Services

Part 1. Overview Information

Participating Organization(s)	National Institutes of Health (NIH)
Components of Participating Organizations	National Human Genome Research Institute (NHGRI)
Funding Opportunity Title	**Genomic Medicine Pilot Demonstration Projects Coordinating Center (U01)**
Activity Code	U01 Research Project – Cooperative Agreements
Announcement Type	New
Related Notices	None
Funding Opportunity Announcement (FOA) Number	**RFA-HG-12-007**

Genomic Medicine Pilot Demonstration Projects: RFAs HG-12-006 and HG-12-007

Purpose: Demonstrate feasibility of, and develop methods for, incorporating patients' genomic findings into their clinical care

Goals:

1. Expand existing GM efforts and develop new projects and methods, in diverse settings
2. Contribute to evidence base regarding outcomes of implementing GM
3. Define and disseminate processes of GM implementation, diffusion, and sustainability in diverse clinical settings

Genomic Medicine Pilot Demonstration Projects: RFAs HG-12-006 and HG-12-007

- Applications Received: July 19, 2012
- Review: December, 2012

Ebony Bookman

Heather Junkins

Larson, G. *The Complete Far Side*. 2003.

Clinically Relevant Genetic Variants Resource: RFA HG-12-016

Department of Health and Human Services

Part 1. Overview Information

Participating Organization(s)	National Institutes of Health (NIH)
Components of Participating Organizations	National Human Genome Research Institute (NHGRI)
Funding Opportunity Title	**Clinically Relevant Genetic Variants Resource: A Unified Approach for Identifying Genetic Variants for Clinical Use (U01)**
Activity Code	U01 Research Project – Cooperative Agreements
Announcement Type	New
Related Notices	None
Funding Opportunity Announcement (FOA) Number	**RFA-HG-12-016**

Clinically Relevant Genetic Variants Resource: RFA HG-12-016

Purpose: Develop and disseminate consensus information on variants relevant for clinical care.

Goals:

1. Identify variants with likely clinical implications
2. Develop resource of these variants and their supporting evidence for use by professional organizations for guideline development
3. Build upon existing programs and reduce duplicative efforts to identify such variants

Clinically Relevant Genetic Variants Resource: RFA HG-12-016

- Applications Received: October 23, 2012
- Review: February, 2013

Erin Ramos

- Stay tuned…

Clinical Sequencing Exploratory Research: RFAs HG-12-008 and 12-009

Department of Health and Human Services

Part 1. Overview Information

Participating Organization(s)	National Institutes of Health (NIH)
Components of Participating Organizations	National Human Genome Research Institute (NHGRI) National Institute on Drug Abuse (NIDA)
Funding Opportunity Title	**Clinical Sequencing Exploratory Research (UM1)**
Activity Code	UM1 Research Project – Cooperative Agreements
Announcement Type	Reissue of RFA-HG-10-017
Related Notices	• June 8, 2012 - See Notice NOT-HG-12-017. Notice of Change in Activity Code from U01 to UM1
Fund Anno	

Department of Health and Human Services

Part 1. Overview Information

Participating Organization(s)	National Institutes of Health (NIH)
Components of Participating Organizations	National Human Genome Research Institute (NHGRI)
Funding Opportunity Title	**Clinical Sequencing Exploratory Research Coordinating Center (U01)**
Activity Code	U01 Research Project – Cooperative Agreements
Announcement Type	New
Related Notices	None
Funding Opportunity Announcement (FOA) Number	**RFA-HG-12-008**

Clinical Sequencing Exploratory Research: RFAs HG-12-008 and 12-009

Purpose: Investigate challenges to applying genomic sequence data to the care of patients.

Goals:

1. Generate clinically valid genomic sequence data relevant to individual patient's care
2. Interpret and translate these data for the physician and communicate to the patient
3. Examine the ethical and psychosocial implications of bringing broad genomic data into the clinic

Clinical Sequencing Exploratory Research: RFAs HG-12-008 and 12-009

- Applications Received: July 26, 2012
- Review: October 2012/January 2013

Lucia Hindorff

Brad Ozenberger

- Stay tuned…

Genomic Sequencing and Newborn Screening Disorders: RFA HD-13-010

Department of Health and Human Services

Part 1. Overview Information

Participating Organization(s)	National Institutes of Health (NIH)
Components of Participating Organizations	*Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NICHD) National Human Genome Research Institute (NHGRI)
Funding Opportunity Title	**Genomic Sequencing and Newborn Screening Disorders (U19)**
Activity Code	U19 Research Program – Cooperative Agreements
Announcement Type	New
Related Notices	• August 15, 2012 - Informational/Technical Assistance Pre-application Meeting for RFA-HD-13-010. See Notice NOT-HD-12-027.
Funding Opportunity Announcement (FOA) Number	**RFA-HD-13-010**

Genomic Sequencing and Newborn Screening Disorders: RFA HD-13-010

Purpose: Explore the possible use of genomic sequence information in the newborn period

Goals:

1. Acquire and analyze genomic datasets in the newborn period;
2. Advance understanding of specific disorders identifiable via newborn screening through promising new DNA-based analysis
3. Examine ethical, legal and social implications of possible implementation of genomic sequencing of newborns

Genomic Sequencing and Newborn Screening Disorders: RFA HD-13-010

- Applications Received: November 19, 2012
- Review: February, 2013

Anastasia Wise

- Stay tuned…

Enhanced Partnership with Pharmacogenomics Research Network

NIGMS Home > Research Funding > Featured Funding Programs

NIH Pharmacogenomics Research Network

PGRN

The **NIH Pharmacogenomics Research Network (PGRN)** is a network of scientists focused on understanding how a person's genes affect his or her response to medicines. Funded since 2000, the PGRN has a Vision and Mission.

PharmGKB

The **Pharmacogenomics Knowledge Base** (PharmGKB) is an integrated knowledge base for pharmacogenomics linking phenotypes and genotypes.

The following institutes contribute support to the Pharmacogenomics Research Network:
NIGMS, NHLBI, NCI, NIDA, NICHD, NHGRI, NIMH, NIAMS, ORWH

http://www.nigms.nih.gov/Research/FeaturedPrograms/PGRN

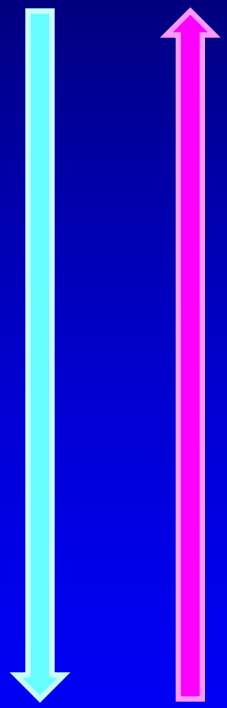
Collaborative NHGRI Pharmacogenomics Project with PGRN in eMERGE Network

- PGRN's Very Important PGx (VIP) Gene Sequencing: array developed to identify rare sequence variants in 85 PGx genes
- eMERGE-PGx will apply validated VIP array for discovery and clinical care in ~9,000 patients
  - Can be exported to other CLIA-certified labs
  - Permit genotyping of common and rare variants and discovery of new ones
  - Use PGRN's Clinical PGx Implementation guidelines and institutional approvals for influencing clinical care

eMERGE-PGRN Partnership

PGRN Pharmacogenomics Research Network

- State of art PGx array
- Ability to update
- Drug-gene guidelines
- CLIA standards and QC



- Privacy concerns
- Electronic phenotyping
- Large pt base
- Less PGx-focused labs

The *eMERGE* Network
electronic Medical Records & Genomics

Selected NHGRI Genomic Medicine Activities

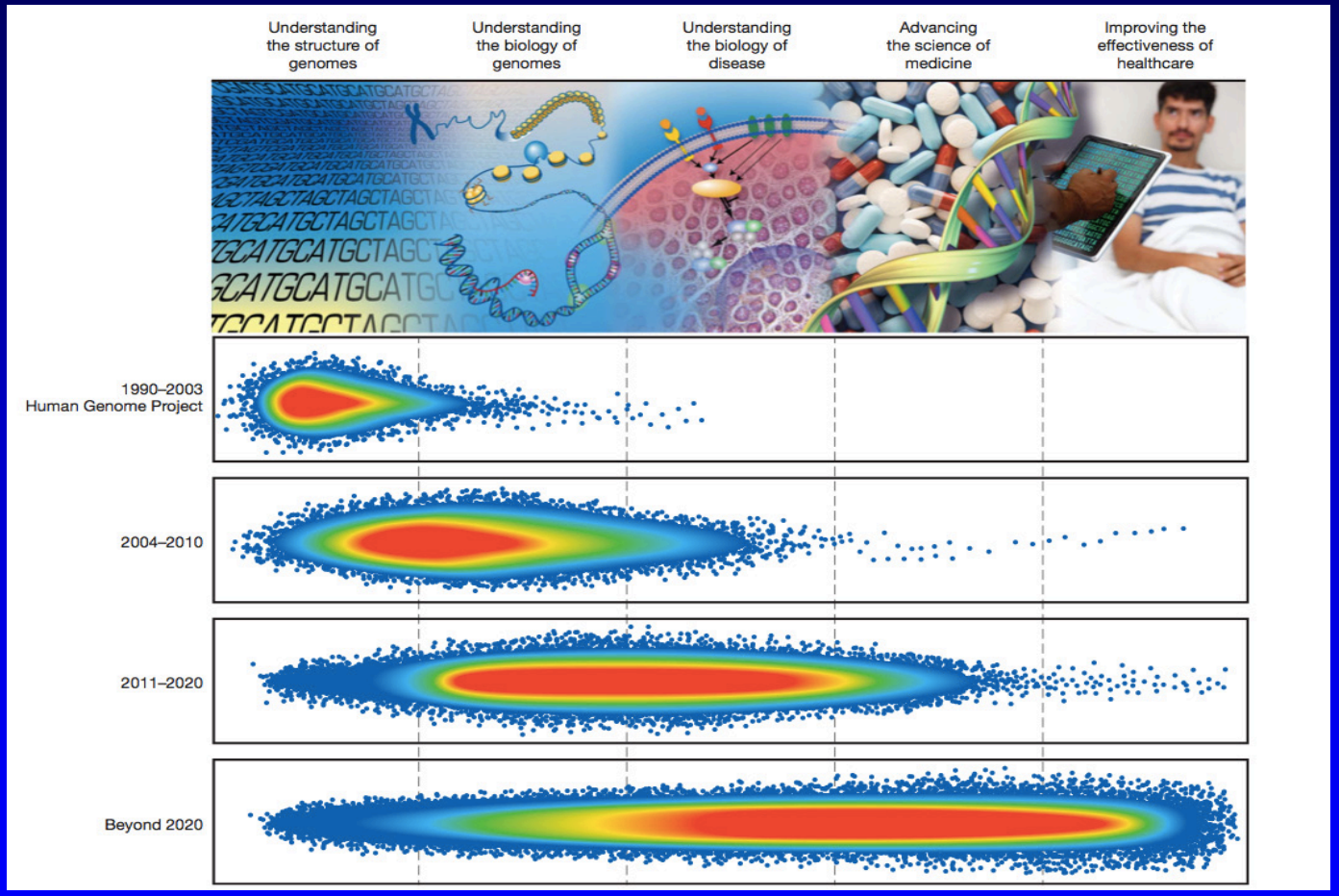
	Ongoing				*Planned*			
	eMERGE	CSER	PhenX	GM Mtgs	CRVR	GMDP	Newborn Seq	FHx
Variant/Assoc Discovery	+	+					++	
Transportable Phenotypes	++		++					
Evidence Generation	++	++				++	+	+
Variant Clinical Implications	++	++		+	++	+		
Consent, Concerns	+	++				+	++	+
Variant Reporting and Use in Care	+	++				+		
Clinician/Pt Educ	++	+		+	+	+	+	
Decision Support	+	+				+	+	+
Policy Devel	+	+		+		+		

CSER, Clinical Sequencing Exploratory Research; PhenX, Phenotype and Exposure Toolkit;
CRVR, Clinically Relevant Variants Resource; FHx, Family History Implementation

Mammoth pointers

Larson, G. *The Complete Far Side*. 2003.

We Might Get There Before 2020



Green ED, Guyer MS. *Nature* 2011; 470:204-13.

Many Thanks - Genomic Medicine Meeting Participants and Infrastructure

Partnerships Permit Rapid Response to Pressing Clinical Questions

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

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Most P

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FDA NEWS RELEASE

For Immediate Release: Aug. 15, 2012
Media Inquiries: Morgan Liscinsky, 301-796-0397, morgan.liscinsky@fda.hhs.gov
Consumer Inquiries: 888-INFO-FDA

FDA warns of risk of death from codeine use in some children following surgeries

The U.S. Food and Drug Administration today issued a Drug Safety Communication concerning three children who died and one child
case of respiratory depression after taking the pain reliever codeine following surgery to remove tonsils (tonsillectomy) and/or adenoids

The surgeries were performed to treat obstructive sleep apnea syndrome, a condition that results in repeated episodes of complete or
sleep. The children received doses of codeine that were within the typical dose range.