

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

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

Teri Manolio, M.D., Ph.D.
Global Leaders in Genomic Medicine
January 8, 2014



NHGRI's Genomic Medicine Working Group

- 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

NACHGR Genomic Medicine Working Group Members

- | | |
|----------------|-----------------|
| Rex Chisholm | Northwestern |
| Geoff Ginsburg | Duke |
| Howard Jacob | Med Coll Wisc |
| Pearl O'Rourke | in |
| M | |
| D | |
| M | vanderbilt |
| Williams | Geisinger |
| Eric Green | Laura Rodriguez |
| Teri Manolio | |

Google "NHGRI GMWG"
<http://www.genome.gov/27549220>

NHGRI Genomic Medicine Meetings, 2011

- GM Colloquium, June 2011, Chicago IL
 - Define landscape, identify commonalities
 - Develop implementation roadmap to share experiences and facilitate adoption
 - Identify common infrastructure and research needs

Genomic Medicine Colloquium Report June 2011, Chicago, IL

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REVIEW

Genetics
inMedicine

Open

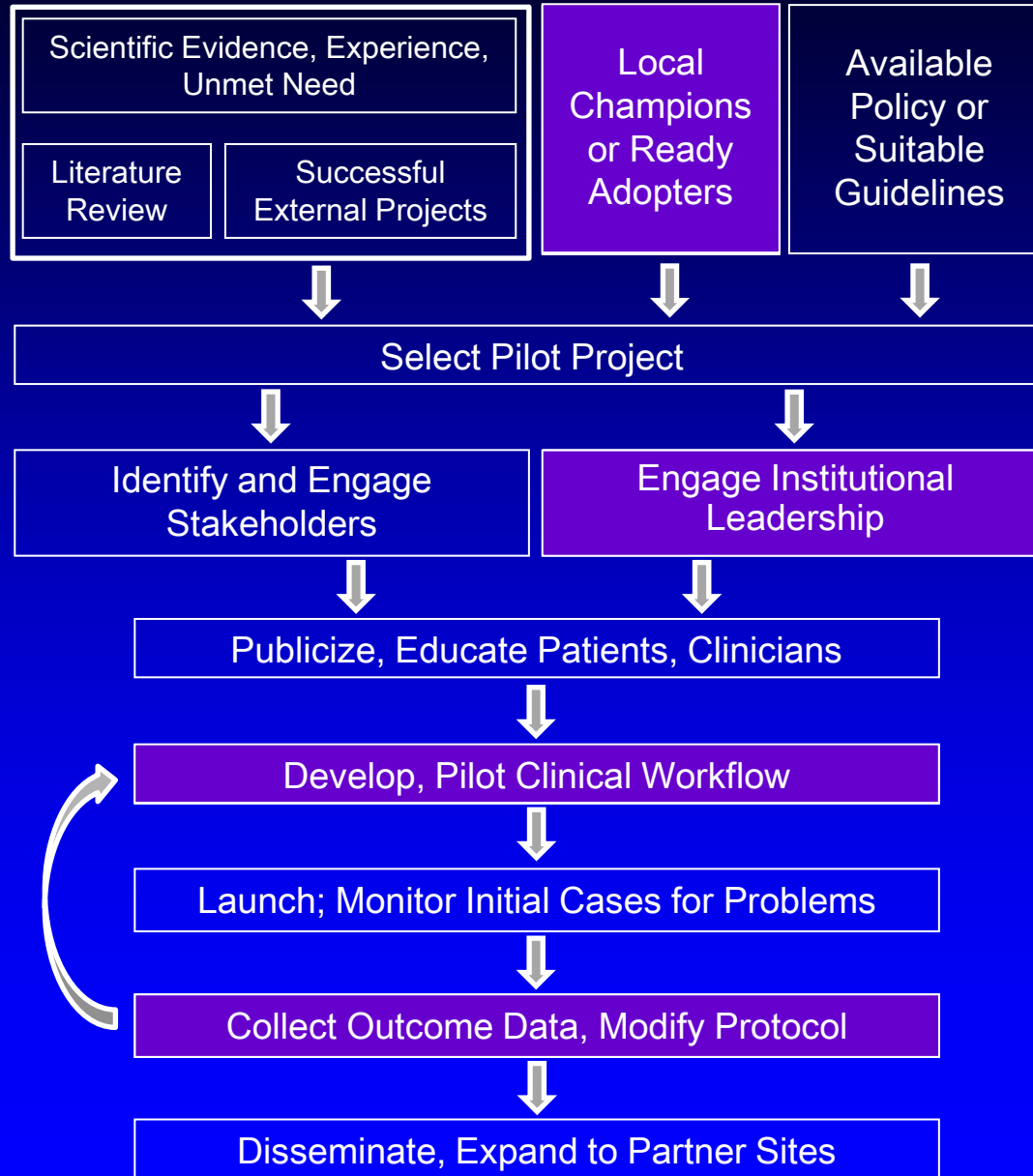
Implementing genomic medicine in the clinic: the future is here

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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

Implementation Roadmap



NHGRI Genomic Medicine Meetings, 2011

- GM Colloquium, June 2011, Chicago IL
 - Define landscape, identify commonalities
 - Develop implementation roadmap to share experiences and facilitate adoption
 - Identify common infrastructure and research needs
- ClinAction, December 2011, Bethesda MD
 - Consider processes and resources needed to:
 - Identify clinically relevant variants
 - Decide whether they are actionable and what the action should be

NHGRI Genomic Medicine Meetings, 2011-2012

- GM II, December 2011, Bethesda MD
 - Identify potential collaborative projects
 - Explore requirements for adoption with institutional leaders
- 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

Genomic Medicine Funding Opportunities

Department of Health and Human Services

Part 1. Overview Information

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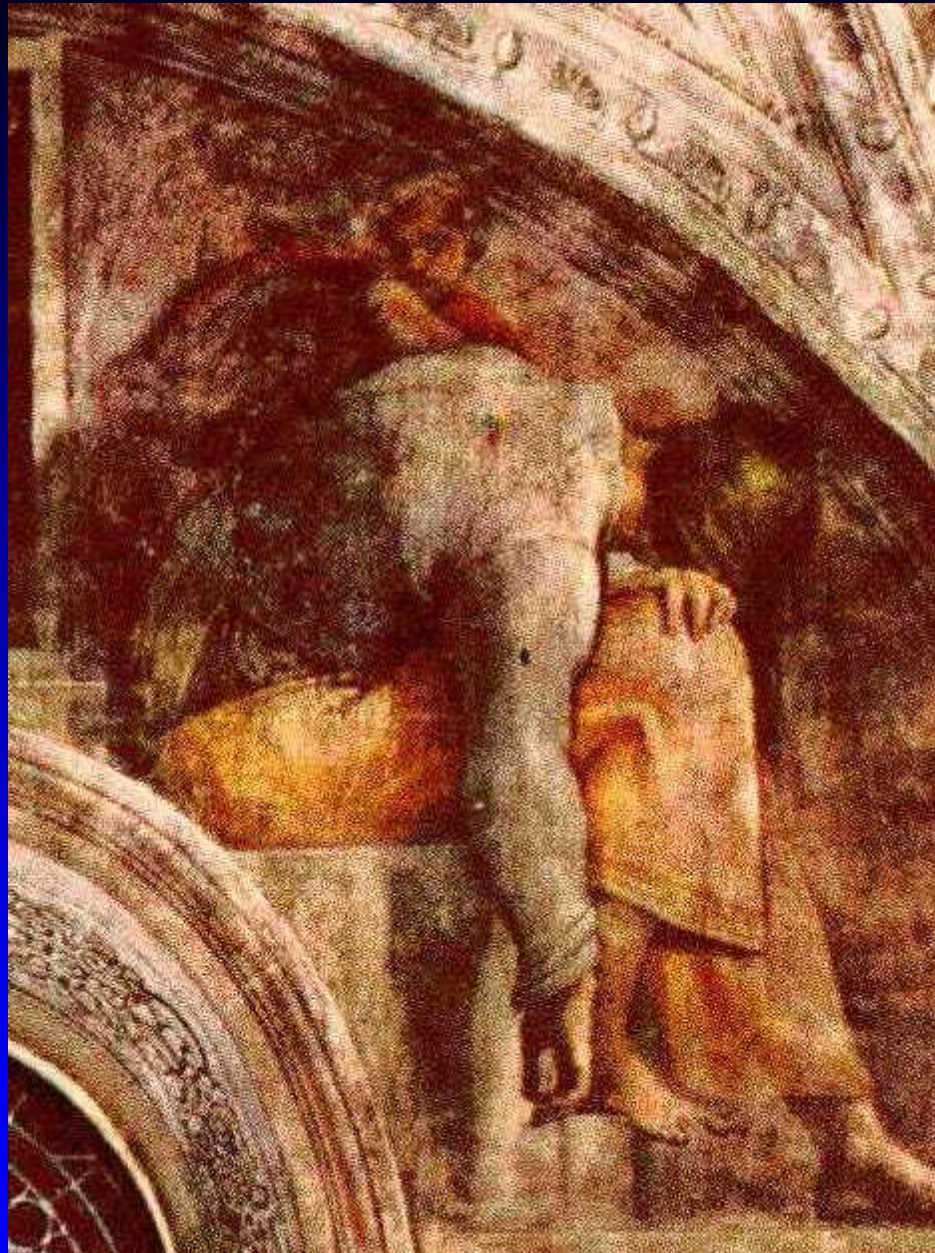
Department of Health and Human Services

Part 1. Overview Information

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Part 1. Overview Information

Participating Organization (s)	National Institutes of Health (NIH)
Components of Participating Organizations	This Funding Opportunity Announcement (FOA) is developed as a Common Fund initiative (http://commonfund.nih.gov/) through the NIH Office of the NIH Director, Office of Strategic Coordination (http://dpcpsi.nih.gov/osc/). The FOA will be administered by the National Human Genome Research Institute (NHGRI/NIH), (http://genome.gov) on behalf of the NIH.
Funding Opportunity Title	DNA Sequencing Core for an Undiagnosed Diseases Network (UDN) (U01)



<http://www.christusrex.org/www1/sistine/28b-Abias.jpg>

Genomic Medicine IV, Jan 28-29, 2013

Educating Physicians in Genomic Medicine

Accreditation Council for Graduate Medical Education
Accreditation Council for Continuing Medical Education
American Academy of Pediatrics
American College of Cardiology
American College of Medical Genetics and Genomics
American College of Physicians
American College of Obstetrics and Gynecology
American Heart Association
American Society of Clinical Oncology
Association of Professors of Human Medical Genetics

Areas of General Consensus

- Present genomics to physicians as gradual evolution rather than “revolution”
- Embed genomics education at point of care with adequate clinical decision support technologies
- Incorporate genomics into certifications and licensing, emphasizing appropriate competencies
- Allow subspecialty-tailored training rather than general programs emphasizing rare syndromes, dysmorphologies
- Share genomics education materials already produced by many societies

Inter-Society Coordinating Committee for Practitioner Education in Genomics

Charge: Facilitate interactions among societies that will enhance their efforts to educate practitioners in applying genomic results to clinical care.

Structure

- Named representatives from professional societies and interested NIH Institutes/Centers
- Co-chaired by society representative (Mike Murray, ACP) and NIH (Teri Manolio, NHGRI)
- Meet at 6-month intervals with conference calls between meetings
- Design 3- to 5-year work plan

Initial Working Groups and Products

Competencies: Work with societies to identify appropriate desired competencies

Educational products: Collect existing products, identify new educational needs and develop appropriate resources

Engagement of Specialty Boards: Support expansion of genomic content in certification processes

Use Cases: Develop general and society-specific use cases, create educational materials to support them

ISCC as of January 8, 2014

Accred Counc Grad Med Ed

Accred Council Cont Med Ed

Am Acad Family Physicians

Am Acad Ophthalmology

Am Acad Pediatrics

Am Assoc Clin Chem

Am Board Medical Genetics

Am Board Medical Specialties

Am Board Ophthalmology

Am Coll Cardiology

Am Coll Med Genet Genom

Am Coll Obstet Gynecol

Am Coll Physicians

Am Heart Assoc

Am Med Assoc

Am Soc Clin Oncol

Am Thoracic Soc

Assoc Molec Pathology

Assoc Prof Human Med Genet

Coll Am Pathologists

Counc Med Specialty Soc

Soc Gen Internal Medicine

NCI

NCBI/NLM

NEI

NHLBI

NIAAA

NIAID

NIAMS

NICHD

NIDA

NIDCD

NIDCR

NIGMS

NIMH

NINDS

NHGRI Genomic Medicine Meetings, 2013-2014

- GM V, May 28-29, 2013, Bethesda MD
 - Engage federal agencies to discuss potential US strategies for GenomMed implementation
 - Explore current activities, needs, obstacles
 - Identify common interests and opportunities, plans for collaboration and strategy development

Agencies Participating in GM V

- Direct medical care efforts
 - Department of Veterans Affairs
 - US Air Force, US Army, US Coast Guard, US Navy
- Reimbursement and regulatory efforts
 - Centers for Medicare and Medicaid Services
 - Food and Drug Administration
 - Agency for Healthcare Research and Quality
 - Blue Cross/Blue Shield
- Supportive and facilitative efforts
 - Centers for Disease Control and Prevention
 - Patient-Centered Outcomes Research Institute
 - Office of the Assistant Secretary for Health
 - Office of the Assistant Secretary for Planning and Eval
 - Institute of Medicine Genomics Roundtable

Key Components of GM Implementation Strategies

Component	AHRQ	CDC	FDA	NHGRI	ASH	PCORI	USAF/ DOD	VHA
Equitable Access	X	X		X	X	X	X	X
Bioinformatics infrastructure for relating clinical characteristics to variants		X	X	X			X	X
Data sharing in accessible research databases	X	X		X	X			X
Standardized phenotypic, patient, variant, and reference information		X	X	X			X	X
Assessment of health economics and cost-effectiveness		X		X			X	X
Evidence of clinical validity and utility	X	X	X	X	X		X	X
Consent model		X		X	X		X	X
Ethical and legal framework to protect against potential abuses		X		X	X		X	X
Engaging public and building awareness		X		X	X		X	X

Genomic Medicine V: Federal Agencies

May 28-29, 2013



Possible DoD-VA-NIH Collaboration in Evidence Generation, Sept. 26, 2013

- Military medical services receiving pressure from patients, companies to add genetic testing
- Comprehensive clinical care system through DHA with potential for providing life-long care
- De-confounding of ancestral diversity and socioeconomic status
- Lower staff costs and overhead for research conducted through DHA
- Contribution to improving care of military personnel and veterans

Initial Focus: Pharmacogenomics

- Goal: Assay pharmacogenomic variation and systematically collect actionable family history information in DoD-NIH-VA patients, and use that information to improve patient outcomes.
 - Validate use of PGx and FHx data in patient care
 - Familiarize/educate clinicians and patients
 - Develop informatics and EMR infrastructure
 - Address unique policy and readiness concerns
 - Set stage for broader use of genomic data

GM VII: Where to go from here?

- Interact with industry, especially sequencing, diagnostic, therapeutic companies
- Continue open invitations (as space permits), videostreaming and archiving
- Pursue offshoots of earlier meetings
 - Targeted research programs
 - Payers
 - ISCC
 - Evidence generation project
 - International steering group?
- Engage disease-specific NIH Institutes

