GMXV: Genomics and Population Screening

Many thanks to the Planning Group!

Jonathan Berg
Gail Jarvik
Bruce Korf
George Mensah

And to the Organizers!

Jenna Cohen
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Jahnavi Narula
Mukul Nerukar
Jerryl Somani
Meredith Weaver

GMXV: Genomics and Population Screening

Goal

To discuss the current state of population genomic screening in the U.S., as well as barriers and opportunities for expanded population screening, impact on clinical practice and outcomes, various genomic screening technologies and costs, and evidence gaps that may inform future research directions.

Objectives

- Review current state of population genomic screening in the US
- Examine obstacles and opportunities for expanded screening and available evidence of impact of screening on outcomes and cost
- Identify research directions to inform expanded screening as appropriate
  - Variants and conditions to be screened for
  - Populations to screen
  - Role of community engagement
  - Providers to order screening and manage results

GMXV: What This Meeting Will Address

- Conditions for which there are well-established professional guidelines for action
- Screening for adults (age 18+) as starting point
- Research directions for the ***entire*** genomics field, of which NHGRI is only a small (but mighty!) part
- Ask presenters to identify results to be provided and interventions to be recommended when proposing conditions

GMXV: What Will also be Addressed Though Not Specifically Called Out in Agenda

- Evolving/appropriate role of geneticists and genetic counselors in population screening
- Potential role of telehealth and AI
- Importance of keeping approaches and guidelines simple
- Importance of engaging with health systems leadership
- Risks of and dealing with false positives
- Research needed to make healthcare system ready to handle population screening

GMXV: What This Meeting Will *Not* Address

- Not addressing newborn screening, which has its own standing committees and expert bodies, other than as something of a model and source of lessons learned
- Not addressing screening of children (< 18) or parent/guardian consent; focus on adults
- Not debating appropriateness of nor evidence behind potential interventions once a screened-for condition is found
- Not proposing funding opportunities or mechanisms– focus on knowledge gaps and approaches for filling them

Brief Note on Structure of Meeting and Its Aftermath

- Ample time for questions and discussions in each session
- Allow for 1-2 clarifying questions after each speaker
- Moderators to redirect from topics the meeting will not address
- Moderators to save few minutes at end of session to list a few key points made or issues raised
- In person attendees should raise hands to speak
- Online attendees can put questions in Q&A but can’t guarantee we’ll see and address them during the meeting; can try to address in follow-up on meeting website
- Meeting summary aimed to be posted to website in 4-6 weeks
- If warranted, presenters and moderators will jointly prepare white paper for journal submission

NACHGR Genomic Medicine Working Group Members

Carol Bult	Jackson Labs
Rex Chisholm	Northwestern
Pat Deverka	Deverka Consulting
Geoff Ginsburg	All of Us Research Program
Gillian Hooker	Concert Genetics
Gail Jarvik	U Washington
George Mensah	NHLBI
Dan Roden	Vanderbilt
Marc Williams	Geisinger

<u>NHGRI</u>

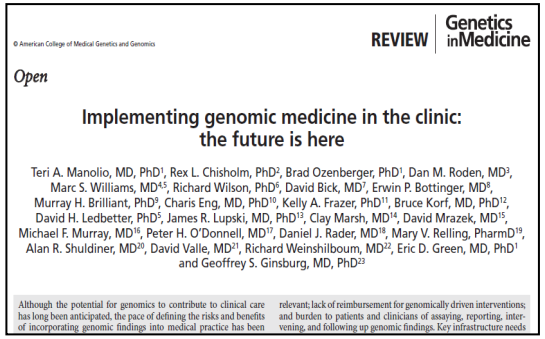
Eric Green	Erin Ramos
Teri Manolio	Robb Rowley
	Jahnavi Narula

Genomic Medicine Working Group Charge

Assist in advising NHGRI on research needed to evaluate and move genomics into routine medical practice

- Review current progress, identify research, implementation, and education gaps and approaches for filling them
- Identify and publicize key advances
- Plan genomic medicine meetings on timely themes
- Facilitate collaborations, coordination, long-term availability of genomic resources

Genomic Medicine Colloquium, June 2011



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Open

Implementing genomic medicine in the clinic: the future is here

Teri A. Manolio, MD, PhD[1], Rex L. Chisholm, PhD[2], Brad Ozenberger, PhD[1], Dan M. Roden, MD[3], Marc S. Williams, MD[4,5], Richard Wilson, PhD[6], David Bick, MD[7], Erwin P. Bottinger, MD[8], Murray H. Brilliant, PhD[9], Charis Eng, MD, PhD[10], Kelly A. Frazer, PhD[11], Bruce Korf, MD, PhD[12], David H. Ledbetter, PhD[5], James R. Lupski, MD, PhD[13], Clay Marsh, MD[14], David Mrazek, MD[15], Michael F. Murray, MD[16], Peter H. O'Donnell, MD[17], Daniel J. Rader, MD[18], Mary V. Relling, PharmD[19], Alan R. Shuldiner, MD[20], David Valle, MD[21], Richard Weinshilboum, MD[22], Eric D. Green, MD, PhD[1] and Geoffrey S. Ginsburg, MD, PhD[23]

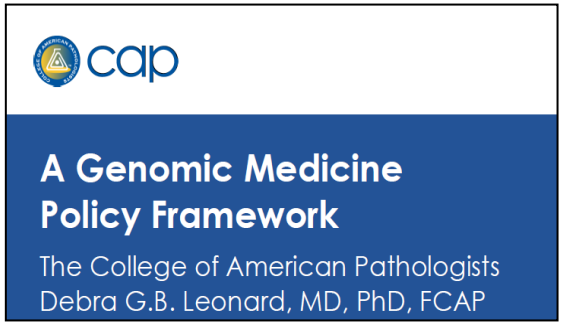
Although the potential for genomics to contribute to clinical care has long been anticipated, the pace of defining the risks and benefits of incorporating genomic findings into medical practice has been relevant; lack of reimbursement for genomically driven interventions; and burden to patients and clinicians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

GM II: Forming Collaborations, Dec 2011

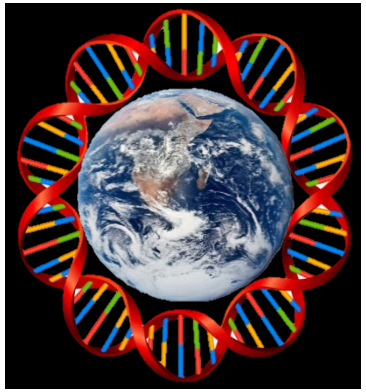
GM III: Stakeholders, May 2012

GM IV: Physician Education, Jan 2013

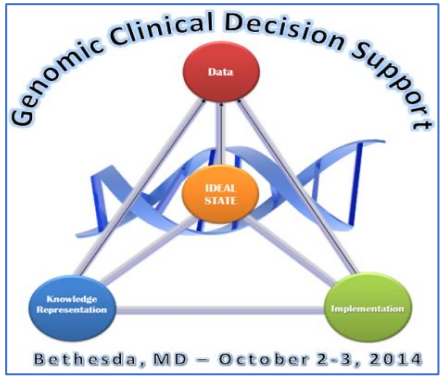
GM V: Federal Strategies, May 2013



GM VI: Global Leaders, Jan 2014



GM VII: Genomic CDS, Oct 2014



GM VIII: NHGRI's Genomic Medicine Programs, June 2015

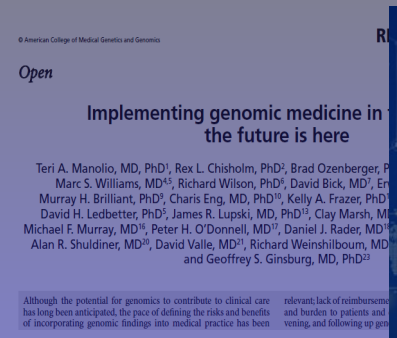
GM IX: Bedside Back to Bench, April 2016

GM X: PGx Implementation, May 2017

GM XI: Clinical Implementation, Sept 2018

GM XII: Genomics and Risk Prediction, May 2019

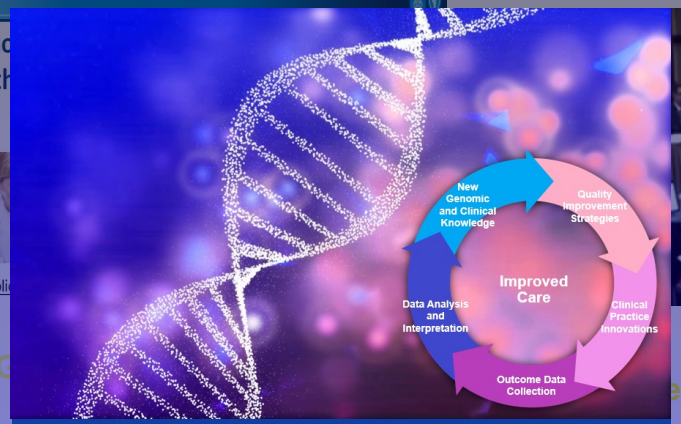
Genomic Medicine Colloquium, June 2011



GM II: Forming Collaborations, Dec 2011

GM XIII: Clinical Informatics Research Agenda, Feb 2021

GM III: Stakeholders, May 2012



GM XIV: Genomic Learning Healthcare Systems, Aug 2022

GM IV: Physician Education, Jan 2013

GM X: PGx Implemen… May 2017

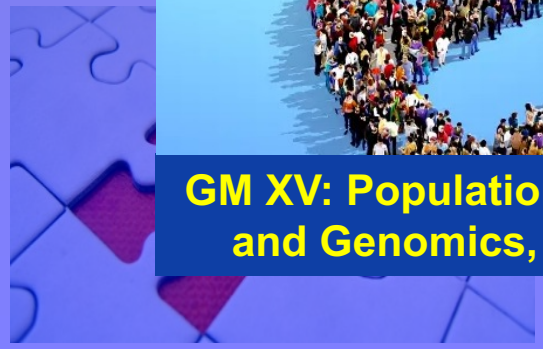
…ederal Strategies, May 2013

A Genomic Medicine Policy Framework

The College of American Pathologists
Debra G.B. Leonard, MD, PhD, FCAP

GM IX: Bedside Back to Bench, April 2016

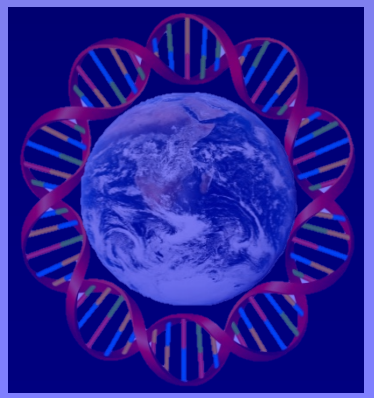
GM V… Medicin…



GM XV: Population Screening and Genomics, Nov 2023

…nomic CDS, Oct 2014

GM VI: Global Leaders, Jan 2014



Products of GM Meetings

ClinGen
Clinical Genome Resource

Clin Action

1 2 3 4 5 6 7 8 9 10 11 12 13 14

PA-18-167
Variant, Function, Disease

NOT-HG-20-020
Genomic Medicine Modules

Employers Health
Innovations

NOT-HG-22-011
Patient-Centered Informatics

RFA-HG-23-041/2
Genomic LHS

eConsult
Concept

emerge network
Genomic Risk Assessment

PRIMED consortium

eMERGE
PGx

IGNITE
Implementing GeNomics In pracTicE

G2MC

IHCC