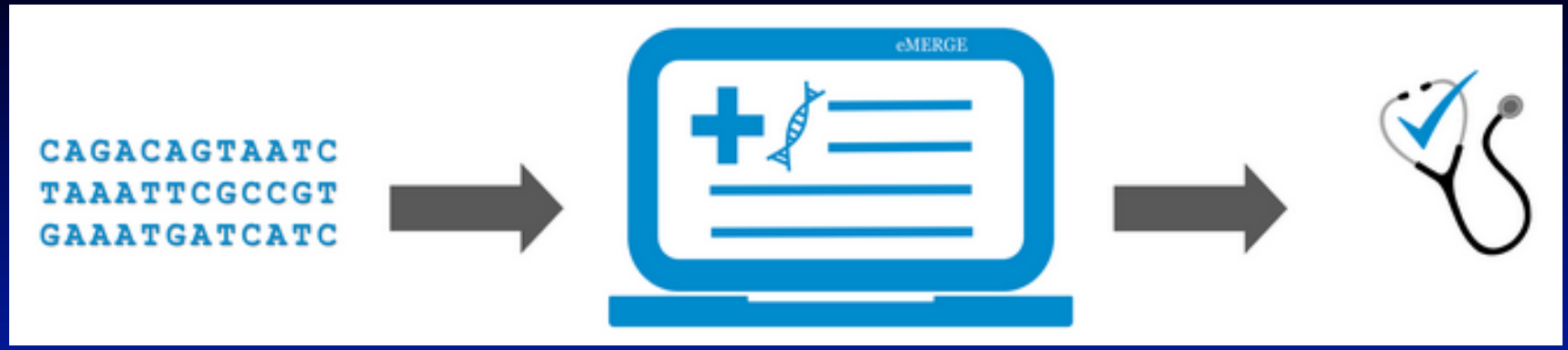



# Genomic Medicine Working Group Update

**Teri Manolio, M.D., Ph.D.**

National Advisory Council on Human Genome Research
September 24, 2018

UNIVERSITY of MARYLAND
SCHOOL OF MEDICINE

NIH National Human Genome Research Institute

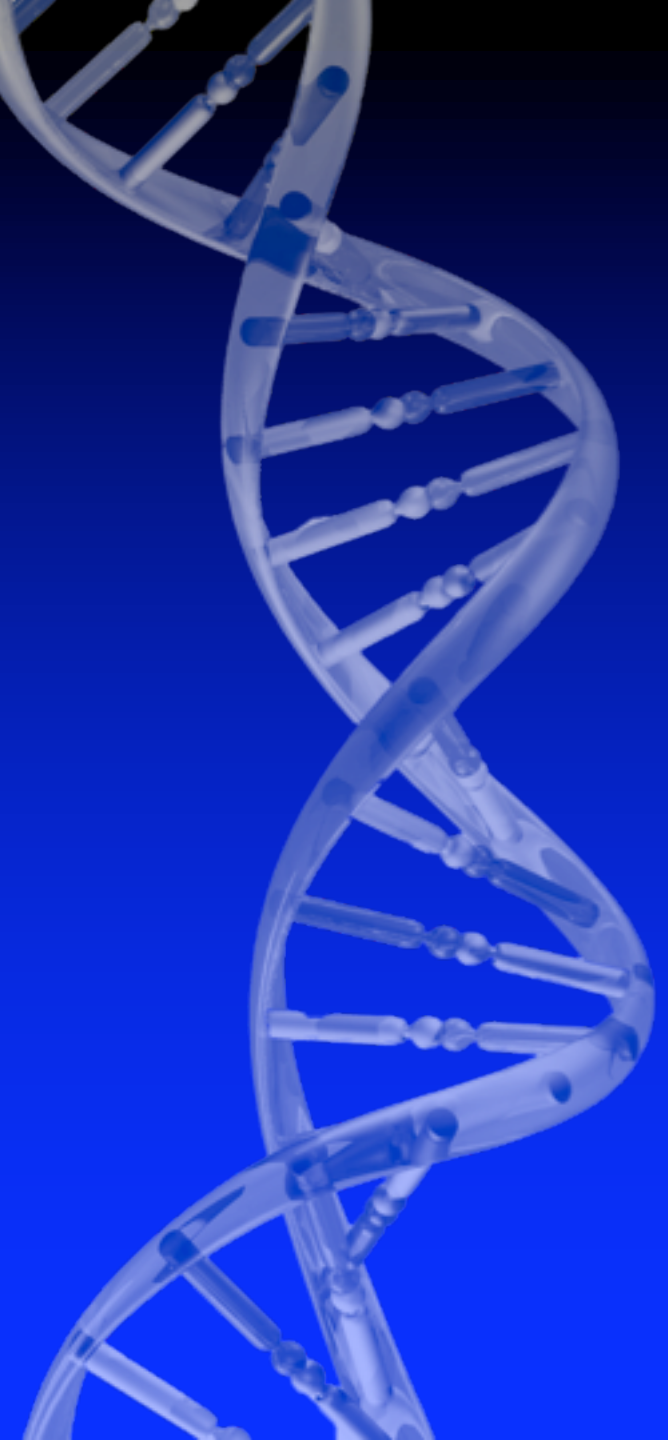
The Forefront of Genomics

# NACHGR Genomic Medicine Working Group Members

| | |
|---|---|
| Carol Bult | Jackson Labs |
| Rex Chisholm | Northwestern |
| Pat Deverka | Am Institutes for Research |
| Geoff Ginsburg | Duke |
| Howard McLeod | Moffitt Cancer Center |
| George Mensah | NHLBI |
| Mary Relling | St. Jude |
| Dan Roden | Vanderbilt |
| Marc Williams | Geisinger |

NHGRI

| | |
|---|---|
| Eric Green | Robb Rowley |
| Teri Manolio | Cecelia Tamburro |
| Laura Rodriguez | |



# Spectrum of Disease-Related Genomics Research

Genomic Medicine

# Genomic Medicine Working Group Charge

Assist in advising NHGRI on research needed to evaluate and implement genomic medicine

- Review current progress, identify research gaps and approaches for filling them
- Identify and publicize key advances
- Plan genomic medicine meetings on timely themes
- Facilitate collaborations, coordination
- Explore models for long-term infrastructure and sustainability of resulting efforts

# Notable Accomplishments in Genomic Medicine

NIH National Human Genome Research Institute

Search Genome.gov

Español

Genomic Medicine Activities

## Notable Accomplishments in Genomic Medicine

nature genetics

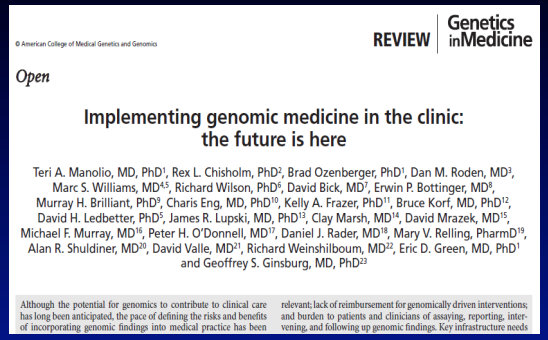
LETTERS

https://doi.org/10.1038/s41588-018-0183-z

# Genome-wide polygenic scores for common diseases identify individuals with risk equivalent to monogenic mutations

Amit V. Khera[1,2,3,4,5], Mark Chaffin[4,5], Krishna G. Aragam[1,2,3,4], Mary E. Haas[4], Carolina Roselli[4], Seung Hoan Choi[4], Pradeep Natarajan[2,3,4], Eric S. Lander[4], Steven A. Lubitz[2,3,4], Patrick T. Ellinor[2,3,4] and Sekar Kathiresan[1,2,3,4*]

## Genomic Medicine Colloquium, June 2011

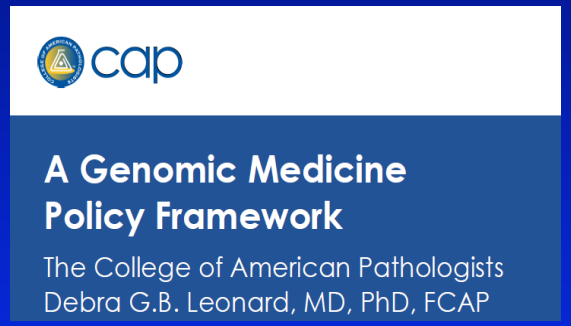



## GM II: Forming Collaborations, Dec 2011

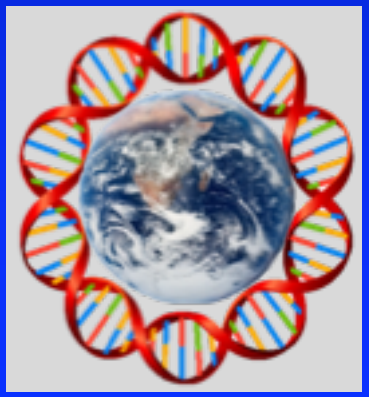

## GM III: Stakeholders, May 2012


## GM IV: Physician Education, Jan 2013

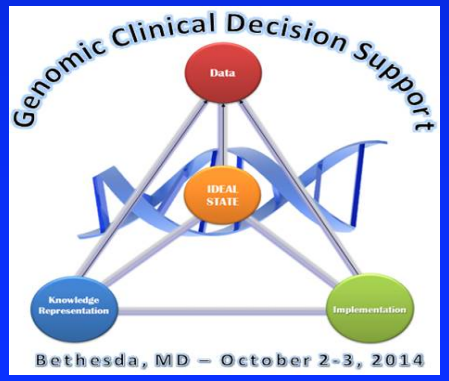
## GM V: Federal Strategies, May 2013




## GM VI: Global Leaders, Jan 2014



## GM VII: Genomic CDS, Oct 2014



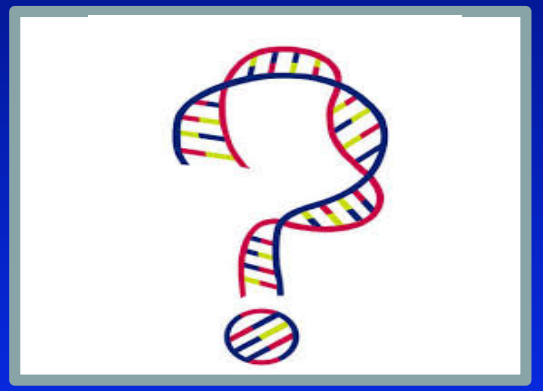

## GM VIII: NHGRI's Genomic Medicine Programs, June 2015

## GM IX: Bedside Back to Bench, April 2016

## GM X: PGx Implementation, May 2017


## GM XI: Clinical Implementation, Sept 2018

## GM XII:



# Genomic Medicine Meetings

NIH National Human Genome Res

Search Genome.gov

Research Fundin

Home / Research Fun

Division of Genomic

Division Staff

GWAS Catalog

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Genomic Medicine Group

Genomic Medicine Directions in

## Genomic Medicine Meetings

NHGRI held a series o
involved with the impl
opportunities; sharing
possible collaborative

- Genomic Medici
  September 5-6, La
- Genomic Medici
  May 2-3, 2017, Sil
- Genomic Medici
  April 19-20, 2016,
- Genomic Medici
  June 8-9, 2015, R
- Genomic Medici
  *Implementation*
  October 2-3, 2014
- Genomic Medici
  January 8-9, 2014

Genomic Medicine Activitie

Genomic Medicine II

Genomic Medicine III

Genomic Medicine IV

On September 5-6
*Genomic Medicine X*
California.

### Wednesday, S

| Time | Topic |
|---|---|
| 8:30 a.m. | Welc |
| 8:45 a.m. | Overv |
| | Slides |


**On May 2-3, 2017, the National Human Genome Research Institute (NHGRI)** sponsored its 10th Genomic Medicine meeting - ***Genomic Medicine X: Research Directions in Pharmacogenomics Implementation*** - at the Sheraton Silver Spring Hotel in Silver Spring, Maryland.

**The objectives of the meeting were to:**

- Survey national landscape of research programs in pharmacogenomics implementation
- Review current advances and clinical applications of pharmacogenomics
- Discuss limitations and obstacles in pharmacogenomics clinical implementation
- Identify evidence gaps and studies that are needed to address them
- Design strategies for large-scale evaluation and implementation of pharmacogenomics in clinical care in the United States.

YouTube **Video Playlist: Genomic Medicine X: *Pharmacogenomics***

**Meeting Summary**

**Executive Summary**

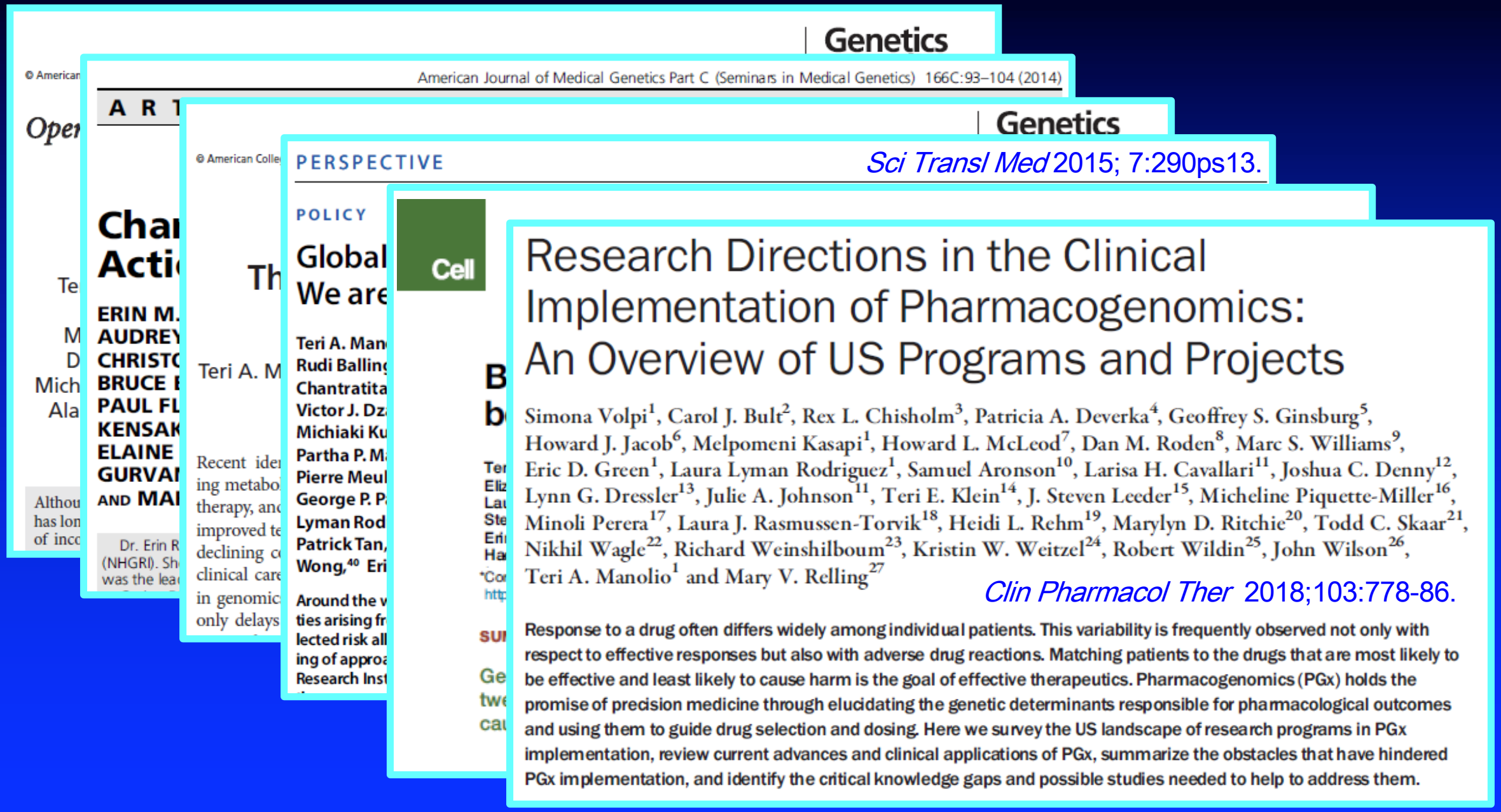
**Tweets from the meeting: #GenomicMed10**

**GOLDILOKs Study: Patient Booklet**

### Tuesday, May 2, 2017

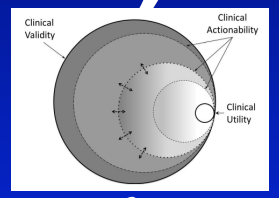
| Time | Topic | Speaker |
|---|---|---|

# GMWG Publications



precisionFDA

ClinGen
Clinical Genome Resource




Clin Action

Payers

FDA

CMS
CENTERS FOR MEDICARE & MEDICAID SERVICES

NATIONAL ACADEMY OF MEDICINE
DIGITizE

Health Education England
NHS

United Healthcare

GTR
Genetic Testing Registry

GMXI Imple-mentation

1 2 3 4 5 6 7 8 9 10

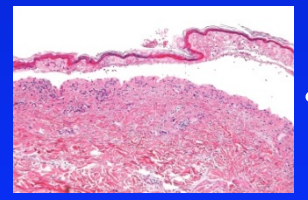
PA-18-867
Variant, Function, Disease

ISCC

eMERGE
PGx

IGNITE
Implementing GeNomics In practicE

G2MC



SJS/TEN

PAR-16-275
Serious ADRs

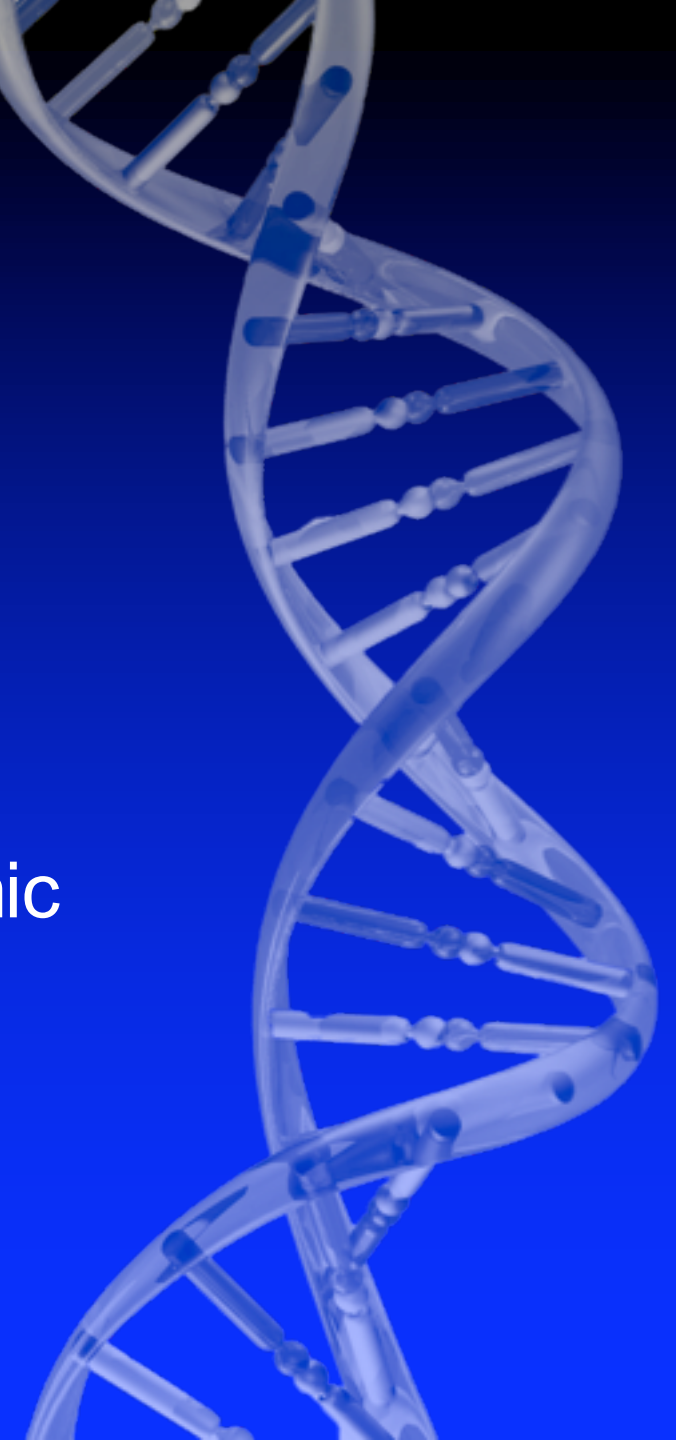
International Cohorts Summit

IHCC

# GM XI: Research Directions in Genomic Medicine Implementation

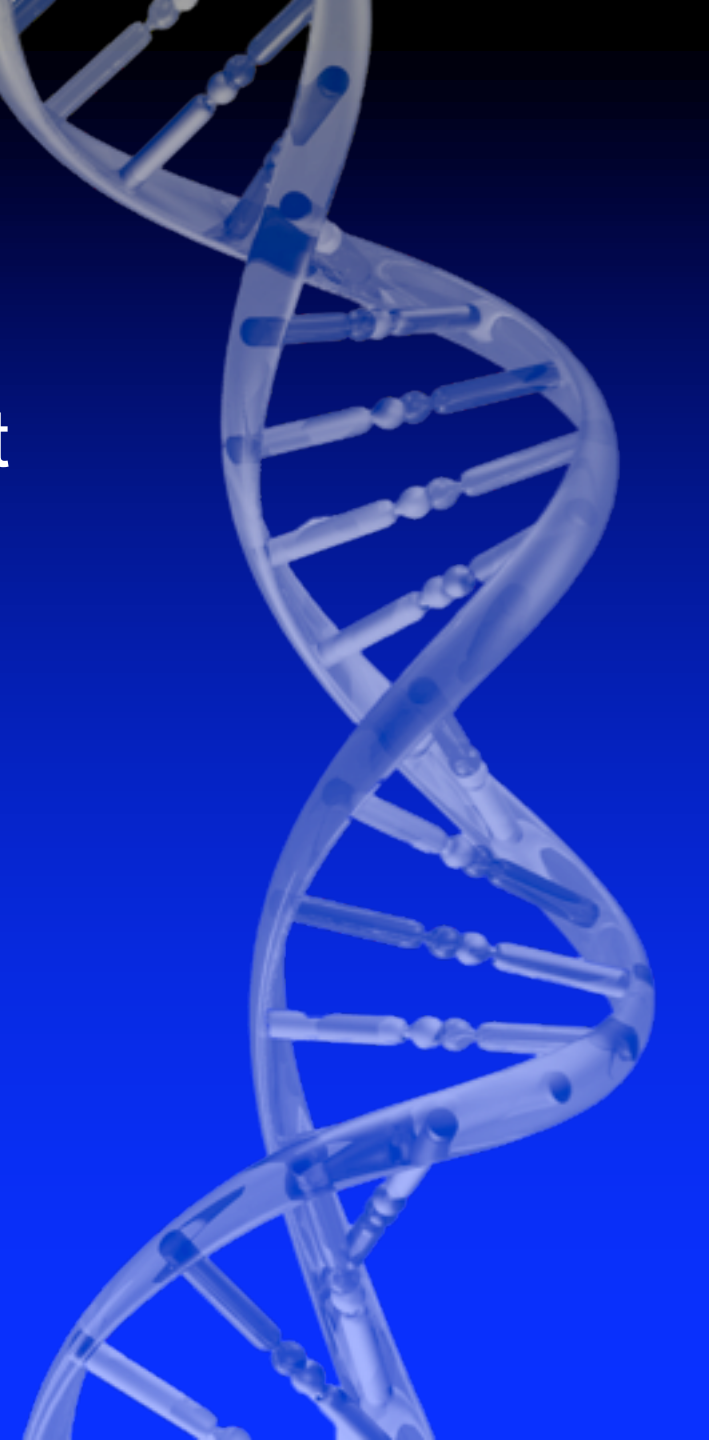
**Objectives:**

- Summarize the current status of genomic medicine implementation research.
- Identify obstacles to genomic medicine implementation and how to overcome them.
- Define where clinical implementation of genomic medicine could or should be 5-10 years from now and how to get there.
- Inform NHGRI strategic planning process regarding genomic medicine.



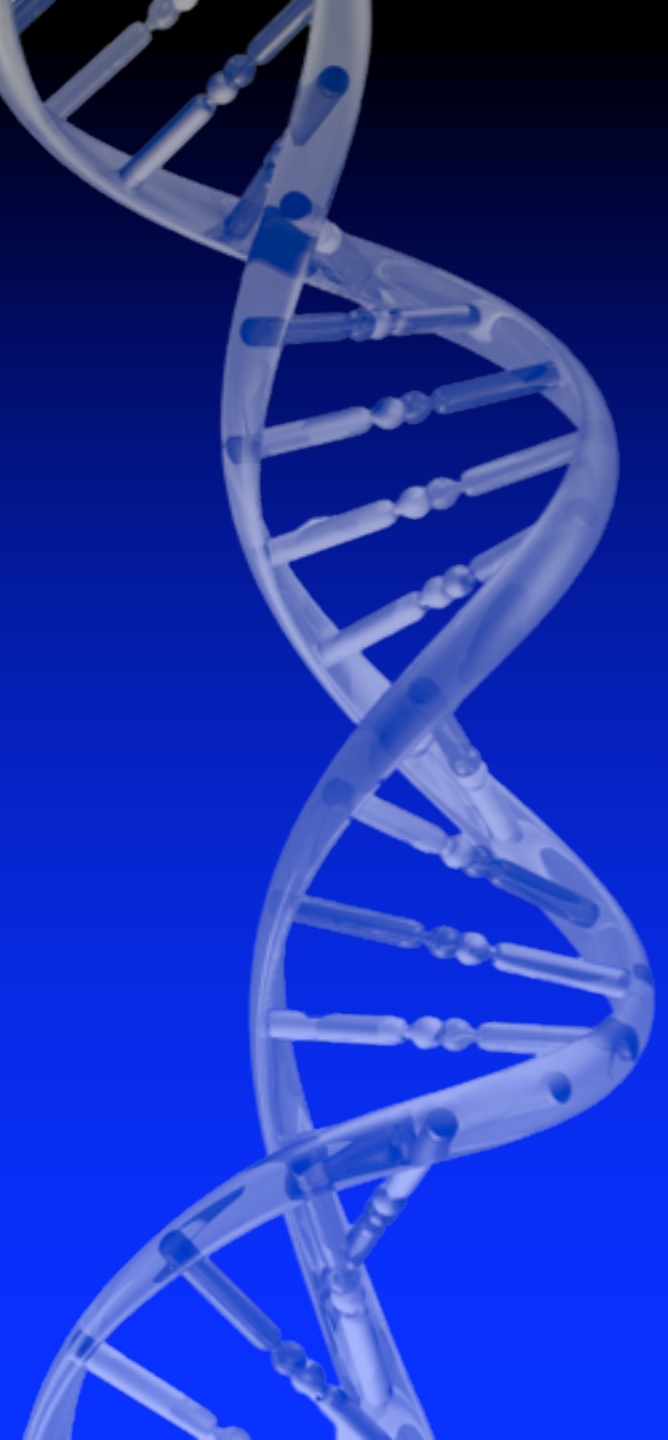
# GM XI Meeting Recommendations

- Establish “CPIC”-style guideline development process for non-PGx genomic-medicine relevant genes
- Create registry of patients with genomic data to follow for outcomes
- Develop limited training program for majority of common, complex disorders; certify as genomic consultants
- Engage employers as key stakeholders



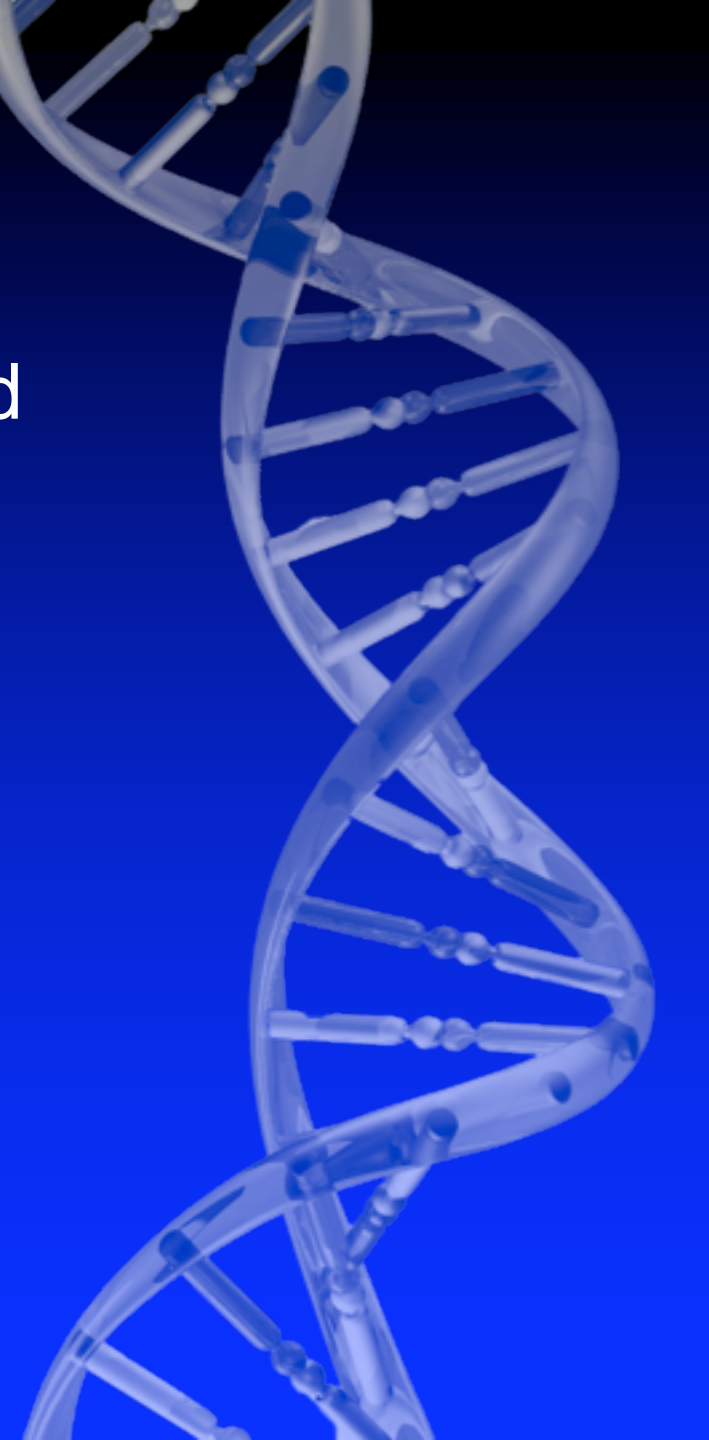
# Panel: What evidence is needed?

- Evidence exists, don't be held hostage by "not enough evidence"; get on with it
- Employers may have lower threshold than payers; consumers/employees are going to be important drivers
- Employers in the service of their employees will be an important force in moving genomic medicine forward
- Reduce emphasis on educating everyone involved
- Provide consulting help on ground



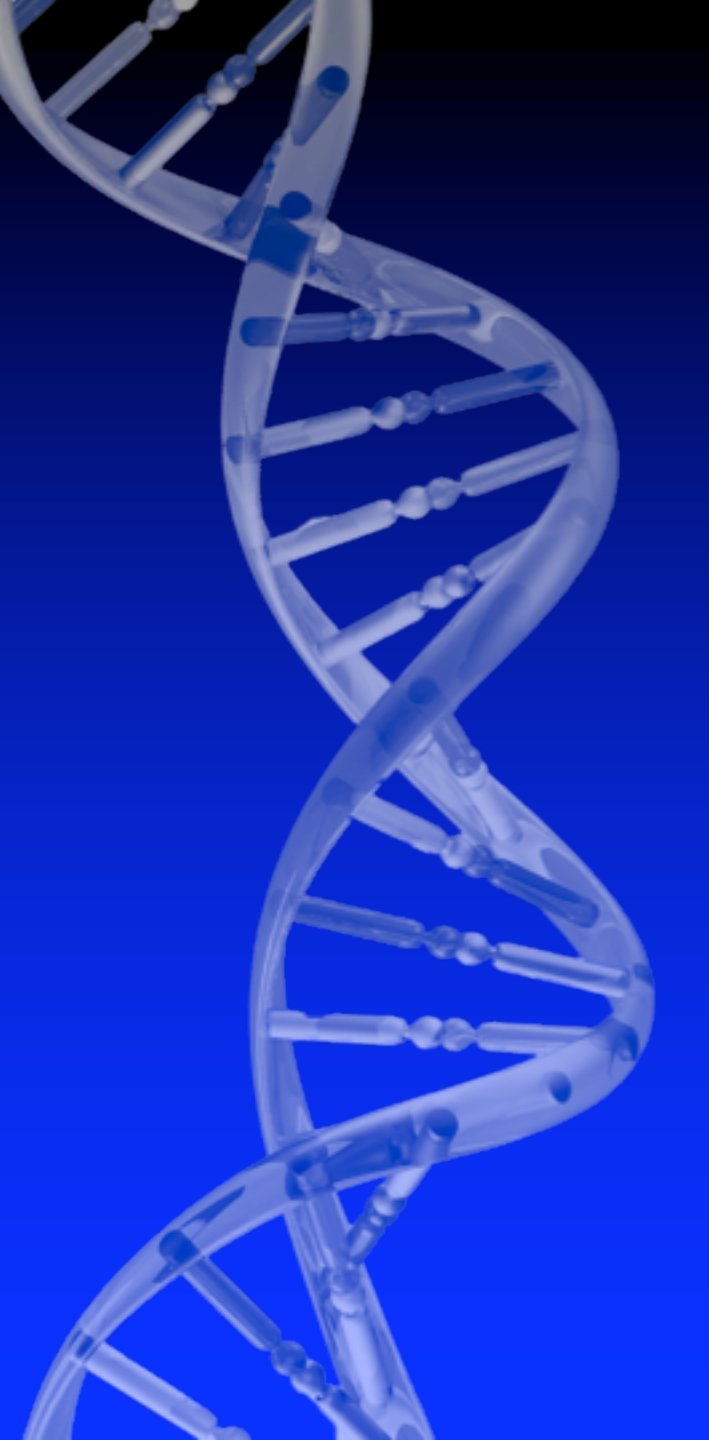
# Panel: What evidence is needed?

- Need research showing employers benefit from adoption; better utilize evidence being generated even if imperfect
- Convene a group of employers and work with them to ensure rolling out in way evidence captured and publishable
- Employers self-aggregating into consortia, take advantage of this for research
- Need to develop a basic genomics formulary
- Public payers still major tough nut to crack



# Employers' Genomic Medicine Meeting

- Determine employers' needs for basic and enhanced genetic testing formulary and employee health program and develop them
- Explore potential project to deploy formulary-based program within employer health plan populations and measure and publish clinical and health economic impact
- Explore additional collaborative opportunities involving pooling data from employers' health data systems to assess impact



# Many Thanks…

Joy Boyer
Lisa Brooks
Heather Colley
Erin Currey
Alvaro Encinas
Eric Green
Sarah Gould
Jyoti Gupta
Lucia Hindorff
Jean Jenkins
Sheethal Jose
Dave Kaufman
Rongling Li
Nicole Lockhart
Ebony Madden
Ebony Madden
Donna Messersmith
Kiara Palmer
Erin Ramos
Robb Rowley
Laura Rodriguez
Cecelia Tamburro
Simona Volpi
Ken Wiley
Anastasia Wise

Carol Bult, Rex Chisholm, Pat Deverka, Geoff Ginsburg, Howard McLeod, George Mensah, Mary Relling, Dan Roden, Marc Williams

emerge network
ELECTRONIC MEDICAL RECORDS AND GENOMICS

IGNITE Implementing GeNomics In practicE

NSIGHT

Newborn Sequencing

cser
Clinical Sequencing Exploratory Research

Undiagnosed Diseases Network

ClinGen
Clinical Genome Resource