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
<table>
<thead>
<tr>
<th>Name</th>
<th>Institution</th>
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<tbody>
<tr>
<td>Rex Chisholm</td>
<td>Northwestern</td>
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<tr>
<td>Geoff Ginsburg</td>
<td>Duke</td>
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<tr>
<td>Howard Jacob</td>
<td>Med Coll Wisconsin</td>
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<tr>
<td>Pearl O'Rourke</td>
<td>Vanderbilt</td>
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<td>Mary Relling</td>
<td>St. Jude</td>
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<td>Dan Roden</td>
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<td>Marc Williams</td>
<td>Geisinger</td>
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<td>Eric Green</td>
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<tr>
<td>Teri Manolino</td>
<td>Laura Rodriguez</td>
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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
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
Implementation Roadmap

- Scientific Evidence, Experience, Unmet Need
- Literature Review
- Successful External Projects
- Local Champions or Ready Adopters
- Available Policy or Suitable Guidelines

Select Pilot Project

- Identify and Engage Stakeholders
- Engage Institutional Leadership

Publicize, Educate Patients, Clinicians

- Develop, Pilot Clinical Workflow
- Launch; Monitor Initial Cases for Problems
- Collect Outcome Data, Modify Protocol

Disseminate, Expand to Partner Sites

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
# DNA Sequencing Core for an Undiagnosed Diseases Network (UDN) (U01)

## Participating Organization(s)

<table>
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<tr>
<th>Department of Health and Human Services</th>
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| 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://dpcpsl.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)
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
NICHHD
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

• 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
<table>
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<tr>
<th>Component</th>
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<th>FDA</th>
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<td>Engaging public and building awareness</td>
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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