Rapporteurs: Colette Fletcher-Hoppe and Ellen Howerton, Program Analysts, NHGRI Cecilia Dupecher, Program Specialist, NHGRI

# **IGNITE and Beyond: The Future of Genomic Medicine Implementation**

John Edward Porter Neuroscience Research Center Building 35, NIH Main Campus August 30, 2016

### **Executive Workshop Summary**

The National Human Genome Research Institute (NHGRI) convened a meeting to discuss future opportunities surrounding the integration of genomic medicine into routine clinical care on Tuesday, August 30, 2016. The meeting objectives were to: 1. evaluate the key contributions of IGNITE to genomic medicine implementation, 2. identify and prioritize the set of scientific opportunities that could fill gaps in the field of genomic medicine implementation, and 3. identify optimal topics for future genomic medicine implementation research. The following recommendations were made for NHGRI's consideration when making decisions about future opportunities in genomic medicine implementation. Recommendations are grouped by session, but listed in no particular order within each session.

### Session 1: Genomic medicine implementation in diverse healthcare systems

- Foster more robust collaboration between academic centers and community centers to disseminate and implement successful genomic medicine implementation programs and strategies.
- Prioritize inclusion of underrepresented populations and diverse researchers and clinicians in genomic medicine projects to ensure that genomic medicine does not increase existing health disparities.
- Leverage the use of existing cohorts with diverse populations to have greater understanding of genetic variants across diverse populations.
- Create a genomic medicine resource center with formalized translation services and educational materials.
- In addition to race and ancestry diversity, focus on different kinds of diversity, e.g. linguistic diversity, rural populations, smaller clinics, economic diversity, etc.
- Create a Genomics and Disparities Working Group at the NHGRI level.

### Session 2: Clinical informatics for varied EHR systems

- Promote harmonization and consolidation of information transfer and information standards for EHR/CDS across networks.
- Focus on CDS interfaces on many different levels (patient, provider, etc.).
- When there is an advance in genomic medicine or CDS, make sure there is a path to implementing this finding.
- Collate CDS rules in a repository built on the foundation of resources developed by other NHGRI networks.
- Integrate CDS that considers NLP (natural language processing)-automated approaches.

### Session 3: Clinical evidence for genomic medicine sustainability

- Conduct larger, network-wide studies that are developed in collaboration with clinicians and representatives from health insurance companies, designed and sufficiently powered to address important clinical outcomes, with the ultimate goal of providing evidence to convince clinicians and health insurance companies of the clinical utility of genomic medicine approaches.
- Involve a mix of representatives from different types of health insurance companies and understand health providers' reimbursement schemes and their imminent changes to aid in designing studies that will provide needed evidence for consideration.
- Utilize cost effectiveness information to enhance standard-of-care with genomic medicine. Invest in comparative studies that contrast the trade-offs, defined in terms of benefits, harms, and costs of one treatment vs. another.
- Foster a systematic collection, creation, and evaluation of genomic studies, publically available and curated by experts.
- Communicate the utility of genomic medicine to different types of clinicians (i.e. nurses, residents, etc.).

### Session 4: Economic considerations

- Develop measures of societal and personal utility for genomic testing and validate them.
- Communicate with health insurance companies (and other stakeholders) throughout study development and analyses to provide the evidence needed for genomic medicine sustainability (i.e. clinical utility and clinical validity).
- Establish economic data source standards between consortia to improve cooperation and transferability.
- Explore the economic downstream value proposition of genomic medicine in addition to immediate cost effectiveness.

#### Main Workshop Report

#### WELCOME AND INTRODUCTIONS

#### Presenters: Eric Green, Teri Manolio, Ebony Madden, Anantha Shekhar, Toni Pollin

In accordance with its 2011 Strategic Plan [1], the National Human Genome Research Institute (NHGRI) strives to facilitate the application of genomics to clinical care in an accessible manner. To this purpose, NHGRI's advisory council, the National Advisory Council on Human Genome Research (NACHGR) includes a Genomic Medicine Working Group (GMWG). The NHGRI has implemented six research programs to address the barriers identified by the GMWG, including the Implementing GeNomics In pracTicE (IGNITE) Network. The first phase of IGNITE awards was issued in spring 2013. Initial grantees included Duke University, the University of Florida, and the Icahn School of Medicine at Mount Sinai. The request for funding applications (RFA) was re-released in summer 2014, and three additional sites joined the Network: Indiana University, the University of Maryland, and Vanderbilt University. The current IGNITE consortium focuses on the dissemination of various genomic medicine approaches. The network encompasses a wide variety of research clinics, including academic research centers, family medicine practices, rural hospitals, and clinics serving under-represented patients. The Network also includes six cross-study working and interest groups (WGs/ IGs), and is supported by a six person External Scientific Panel (ESP).

Since the network's inception, individual IGNITE sites have engaged with external entities to take on a variety of new roles. IGNITE's genomic medicine study at Indiana University, termed "the INGENIOUS program (Indiana GENomics Implementation, an Opportunity for the UnderServed)," has been expanded to the entire Indiana University Health (IUH) system, one of the largest hospital systems in the country. IUH administration sees precision medicine and genomic medicine as not only helpful to patients, but as a market differentiator that will attract patients with high-care requirement diseases to the hospital system. To this end, precision health has become one of the Grand Challenge Initiatives that IUH hopes to accomplish by 2020. IUH administration has therefore championed IGNITE's INGENIOUS program as one of the hospital system's 20 most impactful clinical trials, and has expanded the program to cover ~3 million patients. IUH hopes that this trial will demonstrate significant cost savings and patient benefit associated with PGx-guided CDS for patient prescriptions.

To further explore the benefits and costs savings of genomic medicine, IGNITE's study site at the University of Maryland convened a meeting with diverse genomic medicine stakeholders on August 18. Entitled "Unifying the Evaluation and Implementation of Genomic Medicine," the meeting was attended by over 100 participants (66 in person and 36 remote), including NHGRI Program Staff, researchers from diverse consortia, patients, care and technology providers, researchers, and representatives from six insurance companies. Objectives for the meeting were to build a process for ongoing communication amongst stakeholders, understand what evidence is needed for reimbursement and how it should be disseminated, and to identify protocols that will help provide evidence needed. The meeting was organized into case studies that were delineated by types of genetic testing (pharmacogenetics, targeted genotyping, sequencing panels, whole genome/ whole exome sequencing) and included four lunchtime roundtable discussions around different topics. Discussion topics included pre-emptive vs. reactionary genomic testing, the challenges of having genomic information follow patients through different hospital settings, what constitutes medical necessity, education for decision-makers, and the need for a central database of genomic variants. Next steps including publishing the proceedings from

the meeting, developing a manuscript to a peer reviewed journal, and developing a strategic plan for ongoing efforts.

IGNITE is now nearing the end of its funding cycle: it is time to assess what the research has accomplished and where the research should progress to most benefit the genomics research community. To this end, NHGRI convened a workshop entitled "IGNITE and Beyond: The Future of Genomic Medicine Implementation," on Tuesday, August 30. The objectives for the meeting were three-fold: to evaluate the key contributions of IGNITE to genomic medicine implementation, to identify and prioritize the set of scientific opportunities that could fill gaps in the field of genomic medicine implementation research. These topics were discussed in four sessions, organized around the scientific contributions of IGNITE. NHGRI greatly values input from the scientific community, including members from the National Advisory Council on Human Genome Research (NACHGR), on these topics.

#### **SESSION 1: Genomic Medicine Implementation in Diverse Healthcare Settings and Populations**

Presenters: Muin Khoury, Rick Kittles, Carol Horowitz Discussants: Levi Garraway, Kelly Ormond Moderator: Chanita Hughes-Halbert

Precision medicine is at the intersection of public health, implementation science, and genomics. Knowledge integration may be presented in a cyclic model: discovery leads to application and evidence-based recommendations, which grow into health care and prevention programs, and population health, which informs new discoveries. Most overlap between these fields exists in cancer research, one of the main drivers for public health genomics. The public health world is trying to incorporate genomic research, but only a small percentage of findings is incorporated into policy recommendations. To facilitate the precision health cycle, more robust collaboration between academic and community centers, providers, and public health initiatives must be fostered to disseminate established best practices and strategies for genomic medicine implementation. Such partnerships may also address other challenges in genomic medicine, including access to genomic medicine.

Access to medicine or lack thereof amongst disadvantaged minority populations, is a particular challenge in precision medicine implementation. Genomic medicine may actually increase health disparities – the bulk of information is based on populations of European ancestry and may not translate to minority patients. Larger clinical studies with diverse study cohorts will lead to better classification of actionable variants. In addition, existing resources can be better leveraged to have greater understanding of variants across diverse populations, particularly variants of unknown significance (VUSs). In either case, underrepresented populations and diversity of researchers and genomics clinicians must be prioritized as genomic medicine progresses.

From its inception, a top priority of IGNITE has been how to address disseminating best practices for diverse populations. IGNITE encompasses a blend of various clinicians and practice settings, which provides a rich opportunity to test and disseminate different programs to translate genomic medicine into routine practice. Diverse participants in IGNITE have shown a strong interest in participating in research on how the concept of ancestry as a biological concept vs. race as a social construct plays into genomics.

Moving forward, it is vital to continue to prioritize inclusion of underrepresented populations and diverse researchers in genomic medicine projects. IGNITE should strive to increase participant diversity in its research, particularly targeting Latino and Asian patients in genomic medicine studies. In addition, IGNITE can better focus on different kinds of diversity, including patients from rural populations and smaller clinics, and socioeconomic diversity. To increase access to genomic medicine amongst diverse populations, future genomic medicine initiatives should collaborate with representatives from insurance companies to provide the evidence needed so that genomic testing is reimbursed and thus affordable. At the federal level, NHGRI should account for the time and additional resources needed to set up a study of diverse ancestry populations, which are in addition to the resources needed to conduct these studies. NHGRI may also consider creating a genomic medicine resource center with formalized translation services and educational materials, or perhaps a genomics and disparities working group within NACHGR.

#### **SESSION 2: Clinical informatics for varied EHR systems**

Presenters: Sandy Aronson, Casey Overby, Josh Peterson Discussants: Teri Klein, Karen Eilbeck Moderator: Eric Boerwinkle

Clinical decision support (CDS) acts as a bridge to overcoming the barriers to precision medicine, including limited genetic proficiency of clinicians and limited availability of genetics experts. However, CDS is not very widespread in current hospital systems. Although adoption of electronic health records (EHRs) has increased steadily from 2004-2014, comprehensive EHRs constitute only about 1/3 of the EHR system, and CDS is lacking in all of the basic EHR systems [2]. CDS does not imply the existence of an EHR: some healthcare systems utilize methods other than EHRs to deliver information to providers and patients. Many of the barriers to clinical decision support result from organizational boundaries between institutions, such different EHR and thus different CDS systems. Different clinical settings utilize variable workflows of healthcare delivery process, and a variety of data sources for CDS. Further, the variety of mechanisms for CDS is contingent on vendor specified capabilities. It is crucial to develop CDS interfaces that are capable of transmitting different types of data from many different vendors. Current CDS capabilities should be characterized, and in the future, data and informatics sources for CDS should utilize standardized terminology and data exchange standards.

User interface design is a particular challenge in the clinical realm, and must be taken into design considerations. CDS screens must be intuitive and data should be easily understandable. To empower clinicians to utilize CDS, researchers should develop easy-to-use and easy-to-implement software for genomic risk prediction (e.g. a bad PGx interaction), perhaps including an application that displays patient-specific information within the short time frame of a clinical visit. Other major informatics challenges include detecting the need for re-analysis of genetic data based on new findings, securely facilitating long-term patient access to results, and coordination between institutions, reference labs, and vendors.

To address these challenges, IGNITE's Clinical Interest and Informatics Group (CIIG) holds a monthly open-access webinar on CDS and maintains a Clinical Decision Support Knowledgebase, CDSKB.org, in tandem with the Electronic MEdical Records and GEnomics (eMERGE) Network. CIIG has also facilitated the creation of PGx alert systems at Vanderbilt, Northwestern University, and Mount Sinai. In addition, CIIG has provided leadership on the IGNITE-wide Clinical Pharmacogenetics Implementation Consortium (CPIC) prescribing study. Future opportunities for CIIG include addressing gaps in the IGNITE studies' CDS

pipelines, supporting comparative effectiveness activities for the Network, and focusing CDS on the user experience within EHRs. IGNITE is advised to focus on CDS interfaces on many different user levels (i.e. patient, clinician, other providers, etc.). Future research in CDS may also include assessment of the healthcare delivery process workflow and pre- and post- CDS implementation monitoring: CDS adoption and impact should be measured across IGNITE sites.

More broadly, the NIH is advised to integrate standardized CDS systems across NHGRI consortia. Networks should consider collating CDS rules in a cross-consortia repository. Following CDS collection, the NHGRI may promote harmonization and consolidation of information transfer and information standards for EHR/CDS across its networks. Future NHGRI consortia should engage EHR vendors early in developing CDS and in learning to communicate genetic data. In addition, payer representatives and representatives from a health IT background should be included on genomic medicine research teams. These teams must keep abreast of advances in genomic medicine and CDS to ensure implementation of ongoing discoveries.

#### **SESSION 3: Clinical Evidence for Genomic Medicine Sustainability**

Presenters: Jonathan Berg, Roger Klein, Julie Johnson Discussants: Leslie Biesecker, Ned Calonge Moderator: Howard Jacob

To advocate consideration for reimbursement, it is imperative to provide proof of the clinical utility and economic efficiency of genomic medicine. In future genomic medicine research, the net benefit (benefit minus harms) vs. cost of genomic testing must be addressed. The fee of interpreting variants and of downstream interventions should be included when evaluating genomic medicine costs. However, context and outcomes matter with regards to the benefits of genomic medicine. Which benefits are deemed necessary, or when benefits are determined to outweigh the costs, depends on the person requesting the information (i.e. patients vs. physicians vs. health insurance company representatives). Establishing the value that different stakeholders place on various outcomes is a crucial gap in current genomic medicine research. Moving forward, the value of genomic medicine findings should be discussed with a diverse group of stakeholders, i.e., rather than only health insurance company representatives. To facilitate these conversations, researchers may engage different stakeholders with discussions around a defined clinical scenario to give context.

Future studies should be designed with specific outcomes in mind to provide evidence to stakeholders (but without ignoring other important scientific findings). Further, a mix of different kinds of stakeholders should be involved in creating future genomic medicine studies. To better indicate the effectiveness of genomic medicine, future studies could contrast the trade-off of one treatment vs. another. The assessment of benefit and harm associated with the use of genomic medicine compared with traditional treatment will allow for better information about the cost effectiveness of genomic medicine, the adoption of genomic medicine.

However, more data are needed to provide evidence of the effectiveness of genomic medicine, as members of the IGNITE Network have experienced. IGNITE has documented genomic medicine implementation, and the challenges and barriers thereof, in a variety of settings. Through each of these studies, IGNITE has learned that the greatest barrier to genomic medicine adoption is a lack of clinical evidence. This diminishes the willingness of clinicians to adopt genomic therapies and of health insurance companies to cover their costs. IGNITE's Pharmacogenomics (PGx) working group has endeavored to provide strong clinical evidence for insurance coverage of CYP2C19 genotype-guided antiplatelet therapy post percutaneous coronary intervention (PCI), among other projects. The strength of this study lies in its members' efforts to share data, creating a large sample size: the project encompasses ~4500 patients across nine sites. The IGNITE Network, and other genomic medicine efforts, must develop a strong evidence base for multiple genetic conditions to ensure sustainable adoption of genomic medicine and coverage by health insurance companies. Evidence of clinical utility and economic impact must be included. Future genomic medicine efforts need to focus on clinical evidence generation.

A third gap in genomic medicine sustainability is evidence synthesis – a publicly available, expertly curated database of genomic medicine studies is necessary to transparently provide the evidence that stakeholders need. In addition, researchers need better tools to more effectively communicate information about genetic variants, such as My Cancer Genome, a database on tumor mutations. Researchers must learn how to report and collate information on variants more effectively.

#### **SESSION 4: Economic Considerations**

*Presenters: Marc Williams, Robert Nussbaum, Ann Holmes Discussants: Dan Roden, Gail Jarvik Moderator: Howard McLeod* 

Genomic medicine researchers should strive to demonstrate the social value of genetic testing, including economic viability. In the future, measures of societal, personal, and economic utility of genomic medicine may be developed and assessed. Current economic concerns within genomic medicine are centered on issues of compensation. Genomic medicine has a number of stakeholders with different priorities, including patients/ advocates, drug companies, providers, health insurance companies, and clinical labs. Additionally, genomic medicine researchers must bear in mind that health insurance companies are not a homogeneous group. Amongst certain insurers, Molecular Diagnostics (MoIDx), which grew out of the work of Palmetto, wields much influence. Many private and public insurance companies follow one another (i.e. follow MoIDx) in making coverage decisions. Many insurance companies are also concerned about genetic testing due to their experiences combatting code stacking in the past, which colors their experience with the current state of genetic testing. However, each payer ultimately creates its own policies and efforts to reimburse for genetic testing are variable.

Representatives from health insurance companies also vary in their levels of genomic literacy. Obtaining third party payer coverage for genetic testing is a fragmented process due to the varying levels of comprehension of genomics amongst representatives from health insurance companies. Very few publications have focused on genetics and economics in tandem, partially due to a communication gap. In particular, geneticists and economists use the word "cost effectiveness" to mean different things. This indicates a need for interdisciplinary team science. The NHGRI is advised to establish economic data source standards between consortia to improve cooperation and transferability among researchers. In addition, considering stakeholder perspective (payer, patient, and provider) is crucial. Genomic medicine researchers must communicate with health insurance companies (and other stakeholders) throughout study development to determine what evidence is needed for genomic medicine coverage. To address these concerns, three IGNITE projects are studying economic considerations. In addition, cross-Network efforts include proposing current procedural terminology (CPT) coding changes, a cost-effectiveness study on clopidogrel by the PGx working group, and the payer engagement meeting. In general, economic modeling is an advantageous tool for those studying the economic impact of genomic medicine implementation. Modeling is advantageous in that it does not require a complete dataset and can be run from different stakeholder perspectives to help identify which data are most important to collect and to rationalize decision making. Most of the models used in genomic medicine research are incremental and well suited to analyzing single companion diagnostics of one gene. However, models currently do not consider the influence of gene interactions, comorbidities, and polypharmacy due to disease complexity and a lack of evidence. In addition, questions in the economic analysis of genomic medicine are limited by the small amount of data available and the rapid evolution of genomics. Most studies rely on readily available administrative data, which do not include the costs of training, information processing, and data collection. The costs of delivering genomic medicine to patients are not reflected in the cost of genetic or genomic testing.

### SUMMARY AND PRIORITIZATION OF RECOMMENDATIONS

#### Moderators: Christopher Chute, Lon Cardon, Katrina Goddard

The objective of the IGNITE and Beyond workshop was to make clear, concise recommendations for the NHGRI on future opportunities in genomic medicine implementation. Session moderators concurred that those comments that were made throughout the day, but were not discussed during the final session, are not unimportant, and priorities are not mutually exclusive. A few points were identified as being "cross-cutting" across all four discussions. One area was education: across the board, genomic medicine stakeholders could benefit from a more thorough grasp of genomics. Secondly, all four sessions noted the importance of engaging health insurance companies in developing genomic medicine research. Discussion with health insurance companies about the clinical utility of genomic medicine should be ongoing throughout the process of researching genomic medicine implementation, and not only in study design. Other recommendations for NHGRI are included below, grouped by meeting session (but not listed by priority order).

#### Session 1: Genomic medicine implementation in diverse healthcare systems

- Foster more robust collaboration between academic centers and community centers to disseminate and implement successful genomic medicine implementation programs and strategies.
- Prioritize inclusion of underrepresented populations and diverse researchers and clinicians in genomic medicine projects to ensure that genomic medicine does not increase existing health disparities.
- Leverage the use of existing cohorts with diverse populations to have greater understanding of genetic variants across diverse populations.
- Create a genomic medicine resource center with formalized translation services and educational materials.
- In addition to race and ancestry diversity, focus on different kinds of diversity, e.g. linguistic diversity, rural populations, smaller clinics, economic diversity, etc.
- Create a Genomics and Disparities Working Group at the NHGRI level.

### Session 2: Clinical informatics for varied EHR systems

- Promote harmonization and consolidation of information transfer and information standards for EHR/CDS across networks.
- Focus on CDS interfaces on many different levels (patient, provider, etc.).
- When there is an advance in genomic medicine or CDS, make sure there is a path to implementing this finding.
- Collate CDS rules in a repository built on the foundation of resources developed by other NHGRI networks.
- Integrate CDS that considers NLP (natural language processing)-automated approaches.

# Session 3: Clinical evidence for genomic medicine sustainability

- Conduct larger, network-wide studies that are developed in collaboration with clinicians and representatives from health insurance companies, designed and sufficiently powered to address important clinical outcomes, with the ultimate goal of providing evidence to convince clinicians and health insurance companies of the clinical utility of genomic medicine approaches.
- Involve a mix of representatives from different types of health insurance companies and understand health providers' reimbursement schemes and their imminent changes to aid in designing studies that will provide needed evidence for consideration.
- Utilize cost effectiveness information to replace standard-of-care with genomic medicine. Invest in comparative studies that contrast the trade-offs, defined in terms of marginal benefits, harms, and costs, of one treatment vs. another.
- Foster a systematic collection, creation, and evaluation of genomic studies, publically available and curated by experts.
- Communicate the utility of genomic medicine to different types of clinicians (i.e. nurses, residents, etc.).

# Session 4: Economic considerations

- Develop measures of societal and personal utility for genomic testing and validate them.
- Communicate with health insurance companies (and other stakeholders) throughout study development and analyses to provide the evidence needed for genomic medicine sustainability (i.e. clinical utility and clinical validity).
- Establish economic data source standards between consortia to improve cooperation and transferability.
- Explore the economic downstream value proposition of genomic medicine in addition to immediate cost effectiveness.

IGNITE has made positive contributions towards genomic medicine implementation. In doing so, researchers and NHGRI program staff have learned invaluable lessons that can be used to further the implementation of genomics in clinical practice. Now that IGNITE is nearing the end of its funding cycle, the comments and recommendations from this meeting will be assessed and used inform the NHGRI of future directions for genomic medicine implementation research and possible program/study designs.

# Works Cited

- 1. Green, E.D., M.S. Guyer, and National Human Genome Research Institute, *Charting a course for genomic medicine from base pairs to bedside*. Nature, 2011. **470**(7333): 204-213.
- 2. Charles, D., Gabriel, M., and Searcy, T., *Adoption of electronic health record systems among U.S. non-federal acute care hospitals: 2008-2014.* ONC Data Brief, 2015. **23:** 2-10.

# IGNITE and Beyond: The Future of Genomic Medicine Implementation

# August 30, 2016 Porter Neuroscience Research Center (Building 35A) National Institute of Health (NIH) Campus Bethesda, MD

### **Meeting Objectives:**

- Understand and evaluate key scientific contributions of the Implementation of Genomics in Practice (IGNITE) Network.
- Identify and prioritize scientific opportunities and goals that would fill gaps in genomic medicine implementation.
- Identify optimal topics and study designs for future genomic medicine implementation program(s).

8:00 a.m.	Welcome and Introductions		Ebony Madden	
8:15 a.m.	Opening Re	marks	Eric Green	
8:30 a.m.	NHGRI's Ge	enomic Medicine Portfolio	Teri Manolio	
8:45 a.m.	History of the IGNITE Network		Ebony Madden	
8:55 a.m.	Genomic Medicine Program Expansion at IUH		Anantha Shekhar	
9:15 a.m.	Feedback from Payer Engagement Workshop		Toni Pollin	
9:35 a.m.	Overview of	f Meeting Format	Heather Junkins	
9:45 a.m.	Genomic Medicine Implementation in Diverse Healthcare Settings and Populations			
	9:45 a.m.	State of Science and Gaps	Muin Khoury, Rick Kittles	
	10:15 a.m.	IGNITE Highlights and Opportunities	Carol Horowitz	
	10:30 a.m.	Discussion	Moderator: Chanita Hughes-Halbert	
			Discussants: Levi Garraway, Kelly Ormond	
11:00 a.m.	Break			
11:15 a.m.	Clinical Informatics for Varied EHR Systems			
	11:15 a.m.	State of Science and Gaps	Sandy Aronson Casey Overby	
	11:45 a.m.	IGNITE Highlights and Opportunities	Josh Peterson	
	12:00 p.m.	Discussion	Moderator: Eric Boerwinkle Discussants: Teri Klein, Karen Eilbeck	

12:30 p.m.	Working Lunch			
1:00 p.m.	Clinical Evidence for Genomic Medicine Sustainability			
	1:00 p.m.	State of Science and Gaps	Jonathan Berg, Roger Klein	
	1:30 p.m.	IGNITE Highlights and Opportunities	Julie Johnson	
	1:45 p.m.	Discussion	Moderator: Howard Jacob Discussants: Leslie Biesecker, Ned Calonge	
2:15 p.m.	Economic considerations			
	2:15 p.m.	State of Science and Gaps	Marc Williams, Robert Nussbaum	
	2:45 p.m.	IGNITE Highlights and Opportunities	Todd Skaar	
	3:00 p.m.	Discussion	Moderator: Howard McLeod Discussants: Dan Roden, Gail Jarvik	
3:30 p.m.	Break			
3:55 p.m.	Review of Recommendations		Christopher Chute	
4:20 p.m.	Prioritizing	Future Opportunities	Lon Cardon, Katrina Goddard	

5:00 p.m. Adjourn

#### Appendix B- Meeting participants

Shahnaz Ali National Institute of Immunohematology Mumbai alishahna@gmail.com

Kishore Anekalla Northwestern University kishore.anekalla@northwestern.edu

Samuel Aronson\* Partners HealthCare saronson@partners.org

Babak Behnam NIH babak.behnam@nihg.gov

Jonathan Berg\* University of North Carolina jsberg@med.unc.edu

Kenneth Bernstein Patient advocate kenb@optonline.net

David Biermann Biermannlingua djbiermann@aol.com

Leslie Biesecker\* NHGRI/ NIH lesb@mail.nih.gov

Kathryn Blake Nemours Children's Specialty Care kathryn.blake@nemours.org

**Cornelius Boerkoel** Sanford Health <u>cboerkoel@gmail.com</u>

Eric Boerwinkle\* University of Texas Eric.Boerwinkle@uth.tmc.edu Renee Busch caPS <u>CRR20007@GMAIL.COM</u>

Bruce Nedrow Calonge\* Colorado Trust ned@coloradotrust.org

Lon Cardon\* GlaxoSmithKline lon.r.cardon@gsk.com

Larisa Cavallari University of Florida Icavallari@cop.ufl.edu

Christopher Chute\* Johns Hopkins University chute@jhu.edu

Megan Cleveland NIST – Department of Commerce megan.cleveland@nist.gov

Isabel Coello NHGRI/ NIH Scoello.92@hotmail.com

Joshua Denny Vanderbilt University Medical Center cynthia.c.williams@vanderbilt.edu

Paul Dexter Indiana University prdexter@regenstrief.org

Nhi Dinh Georgetown University nhidinh.l@gmail.com

Katarzyna Drozda FDA katarzyna.drozda@fda.hhs.gov Vence Bonham NHGRI/ NIH bonhamv@mail.nih.gov

Karen Eilbeck\* University of Utah <u>keilbeck@genetics.utah.edu</u>

Amanda Elsey University of Florida aelsey@cop.ufl.edu

Colette Fletcher Hoppe NHGRI/ NIH colette.fletcher-hoppe@nih.gov

Levi Garraway\* Dana-Farber Cancer Institute, Harvard University Levi.Garraway@dfci.harvard.edu

Sharlene Geyer BHMC pa-sherry@msn.com

Geoffrey Ginsburg Duke University geoffrey.ginsburg@duke.edu

Katrina Goddard\* Kaiser Permanente Center for Health Research Katrina.AB.Goddard@kpchr.org

Eric Green\* NHGRI/ NIH egreen@mail.nih.gov

JW Green JWGreen@thegreenhousegroupllc.u

Yue Guan University of Maryland yguan@medicine.umaryland.edu

Michael Hanh NHGRI michael.hahn@nih.gov Cecilia Dupecher NHGRI/ NIH cecilia.dupecher@nih.gov

Hon Ho Partners Healthcare/ Harvard University <u>drhonho@gmail.com</u>

Ann Holmes\* Indiana University aholmes@iupui.edu

Carol Horowitz\* Mount Sinai School of Medicine carol.horowitz@mountsinai.org

Ellen Howerton NHGRI/ NIH ellen.howerton@nih.gov

Chanita Hughes-Halbert\* Medical University of South Carolina Hughesha@musc.edu

Howard Jacob\* Hudson Alpha Institute for Biotechnology hjacob@hudsonalpha.org

Gail Jarvik\* University of Washington Medical Center pair@u.washington.edu

Julie Johnson\* University of Florida julie.johnson@ufl.edu

Sheethal Jose NHGRI/ NIH sheethal.jose@nih.gov

Heather Junkins NHGRI/ NIH junkinsh@mail.nih.gov

Muin Khoury\* CDC khourym@mail.nih.gov Gene Hill Computercraft Corporation ghill@computercraft-usa.com

Roger Klein\* Cleveland Clinic roger.klein@aya.yale.edu

**Teri Klein\*** Stanford University School of Medicine <u>teri.klein@stanford.edu</u>

Kenneth Levy Indiana University School of Medicine kdlevy@iu.edu

Rongling Li NHGRI/ NIH lir2@mail.nih.gov

John Lima Nemours Children's Health System jlima@nemours.org

Nita Limdi University of Alabama, Birmingham <u>nlimdi@uabmc.edu</u>

Nicole Lockhart NHGRI/ NIH lockhani@mail.nih.gov

Ebony Madden\* NHGRI/ NIH ebony.madden@nih.gov

Hamid Mahmood Roskilde University Denmark hamid\_bangash@hotmail.com

Teri Manolio\* NHGRI/ NIH manolio@nih.gov

Ramya Marathi Vanderbilt University ramya.marathi@vanderbilt.edu **Rick Kittles\*** University of Arizona College of Medicine <u>rkittles@email.arizona.edu</u>

Howard McLeod\* Moffitt Cancer Center howard.mcleod@moffitt.org

Michael Michalkiewicz Aurora Research Institute michael.michalkiewicz@aurora.org

**C. Daniel Mullins** University of Maryland <u>dmullins@rx.umaryland.edu</u>

David Nelson University of Florida <u>nelsodr@ufl.edu</u>

Robert Nussbaum\* Invitae robert.nussbaum@invitae.com

Lori Orlando Duke University lorlando@duke.edu

Kelly Ormond\* Stanford University School of Medicine kormond@stanford.edu

Casey Overby\* Johns Hopkins University overby@jhu.edu

Aniwaa Owusu-Obeng Mount Sinai School of Medicine aniwaa.owusu-obeng@mssm.edu

Josh Peterson\* Vanderbilt University Medical Center josh.peterson@vanderbilt.edu

Anita Persaud NHGRI/ NIH anitra.persaud@nih.gov Cathy McCarty Essentia Institute of Rural Health cmccarty@eirh.org

Victoria Pratt Indiana University School of Medicine vpratt@iu.edu

Sunseri Rahnea University of the Pacific, MPAS rsunserimd@comcast.net

Dan Roden\* Vanderbilt University Dan.Roden@Vanderbilt.edu

Anantha Shekhar\* Indiana University School of Medicine <u>ashekhar@iupui.edu</u>

Todd Skaar Indiana University School of Medicine tskaar@iu.edu

Michael Smith NHGRI/ NIH smithmw@mail.nih.gov

Jeff Struewing NHGRI/ NIH struewij@mail.nih.gov Toni Pollin\* University of Maryland tpollin@medicine.umaryland.edu

Alem Taye Illumina ataye@illumina.com

Henri Wathieu Georgetown University Medical Center hpw9@georgetown.edu

Kristin Weitzel University of Florida kweitzel@cop.ufl.edu

Ken Wiley NHGRI/ NIH ken.wiley@nih.gov

Marc Williams\* Geisinger mswilliams1@geisinger.edu

Yining Xie NIH Yining.Xie@nih.gov

Asterisk (\*) indicates speakers, moderators, or discussants