National Human Genome Research Institute

Genomic Medicine XIV: Genomic Learning Healthcare Systems

August 31 - September 1, 2022

Virtual Meeting

The **Forefront** of **Genomics**

Genomic Medicine XIV
Genomic Learning Healthcare Systems

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- Genomics-Enabled Learning Health Care Systems: Important Points Highlighted by Individual Speakers
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Genomic Medicine XIV
Genomic Learning Healthcare Systems

Goal

To discuss progress and identify generalizable solutions to genomic medicine implementation challenges experienced over the past 7 years, dating roughly since the 2015 NAM "Genomics-Enabled Learning Health Care Systems" workshop. Persistent barriers and evidence gaps will be examined as opportunities for additional research.

Objectives

- Explore real-world examples of how genomic learning healthcare systems (gLHS) apply cycles of genomic medicine implementation, evaluation, adjustment, and updated implementation practices across delivery systems.
- Examine barriers and identify potential solutions, with a focus on lessons learned from effective gLHS and their potential transportability to other settings.
- Determine ways that solutions can be developed and shared and collaborations can be formed to facilitate research on implementation of gLHS.

Background Materials

- Institute of Medicine. 2015. **Genomics-Enabled Learning Health Care Systems: Gathering and Using Genomic Information to Improve Patient Care and Research: Workshop Summary**. Washington, DC: The National Academies Press. https://doi.org/10.17226/21707.
    - In the attachments, we've included the abstracted Important Points Highlighted by Individual Speakers. You can also view the complete workshop summary online.
- Williams MS, Buchanan AH, Davis FD, Faucett WA, Hallquist MLG, Leader JB, Martin CL, McCormick CZ, Meyer MN, Murray MF, Rahm AK, Schwartz MLB, Sturm AC, Wagner JK, Williams JL, Willard HF, Ledbetter DH. **Patient-Centered Precision Health In A Learning Health Care System: Geisinger's Genomic Medicine Experience**. Health Aff (Millwood). 2018 May;37(5):757-764. doi: 10.1377/hlthaff.2017.1557. PMID: 29733722.
    - In the attachments, we've included the full article, or you can view it on the publisher's website.

Agenda: Wednesday, August 31

11:00	**Welcome and Introductions**	
	Overview of National Academy of Medicine 2015 workshop, "Genomics-Enabled Learning Health Care Systems"	Pat Deverka
	Structure, Goals, and Products of NHGRI Genomic Medicine meetings	Teri Manolio
11:15	**Session 1: Laying the Groundwork (1hr30)**	Moderators: Rex Chisholm Renee Rider
	Keynote: The State of Genomic Learning Healthcare Systems (25 min)	Peter Hulick
	Overview of gLHS Barriers (10 min)	Teri Manolio
	Panel Discussion (55 min)	Adam Buchanan Gai Elhanan Casey Overby Taylor Bruce Korf
12:45	**10-minute break**	
12:55	**Session 2: IT Infrastructure (1hr30)**	Moderators: Carol Bult Christopher Chute
	Clinical Informatics Recommendations from GM XIII (15 min)	Ken Wiley
	Integrating Genomic Results into EHRs (15 min)	Travis Osterman
	Population-based Clinical Decision Support for Precision Medicine (15 min)	Guilherme Del Fiol
	Discussion (45 min)	
2:25	**30-minute break**	

2:55	**Session 3: The role of gLHS in Increasing Health Equity and Access to Genomic Healthcare (2hr)**	Moderators: Gail Jarvik Karriem Watson
	Identifying Biases in gLHS Data and Implementation (15 min)	Janina Jeff
	Utilizing gLHS Data to Increase Equity and Access (15 min)	Latrice Landry
	Diverse Patient and Provider Engagement in gLHS (15 min)	Carol Horowitz
	Facilitating gLHS through Patient Access to Genomic Information (15 min)	Deven McGraw
	Discussion (60 min)	
4:55	**Adjourn Day 1**	

Agenda: Thursday, September 1

11:00	**Welcome**	
	Day 1 Recap (10 min)	Pat Deverka Teri Manolio
11:10	**Session 4: Enabling Providers to Implement Genomic Knowledge (2hr)**	Moderators: Susanne Haga Robb Rowley
	Genomic Medicine Track for Internal Medicine Trainees (15 min)	Noura Abul-Husn
	Improving Access to Genomics in Clinical Practice (15 min)	Rizwan Hamid
	Scalable Solutions for Genetic Counseling (15 min)	Cynthia A. James
	Genomic Medicine Implementation in the VA Healthcare System (15 min)	Jason Vassy
	Discussion (60 min)	
1:10	**20-minute break**	
1:30	**Session 5: Establishing and Sustaining gLHS (1hr30)**	Moderators: Erin Ramos Krystal Tsosie
	Generating Evidence of Effectiveness and Value (15 min)	Howard McLeod
	Genomic Learning Healthcare Systems & Payers (15 min)	Nancy Mendelsohn
	Progress in the Integration of Personalized Medicine and Developing Common Metrics (15 min)	Daryl Pritchard
	Discussion (45 min)	
3:00	**10-minute break**	

3:10	**Session 6: Realizing the Promise of gLHS (1hr15)**	Moderators: Pat Deverka Teri Manolio
	Turning gLHS Data into Knowledge (15 min)	Heidi Rehm
	Creation of a Virtuous Cycle to Realize the gLHS (15 min)	Marc S. Williams
	Scaling the Genomics Enabled Learning Health System to Optimize Research and Clinical Care (15 min)	Geoff Ginsburg
	Discussion (30 min)	
4:25	**Summary and Next Steps (35 min)**	Pat Deverka Teri Manolio
5:00	**Adjourn Day 2**	

Biographies

Presenters, Panelists, and Moderators

Noura Abul-Husn, MD, PhD

Dr. Abul-Husn is the Vice President of Genomic Health at 23andMe and Associate Professor of Medicine and Genetics at the Icahn School of Medicine at Mount Sinai. Dr. Abul-Husn is a physician-scientist working to uncover the clinical impact of human genetic variation in diverse populations and drive the equitable implementation of genomic medicine. Her scientific contributions include pioneering genome-first approaches in electronic health record-linked biobanks to provide novel clinical insights and inform therapeutic discovery. Her work has been published in leading journals, including Science, Cell, and the New England Journal of Medicine. As a principal investigator in the eMERGE (electronic Medical Records and Genomics) Network, she is leading efforts to integrate polygenic risk information into clinical care. In her role at Mount Sinai, Dr. Abul-Husn launched a genomic screening program tailored to ancestrally diverse populations, started a Genomic Health Clinic to provide the infrastructure for emerging genomic applications, and created a Genomic Medicine Track for Internal Medicine residents in order to expand genomics knowledge across specialties.

Dr. Abul-Husn has a BSc Honors in Life Sciences and MSc in Pharmacology from Queen's University in Canada. She completed her MD, PhD, and Internal Medicine/Medical Genetics residency at Mount Sinai in New York. She is board certified in Internal Medicine and Medical Genetics.

Adam Buchanan, MS, MPH

Adam Buchanan is Associate Professor and Director of the Genomic Medicine Institute at Geisinger. Mr. Buchanan is an NIH-funded investigator and board-certified and licensed genetic counselor with expertise in assessing genetic counseling outcomes and cancer risk management behaviors. Mr. Buchanan received a master's in public health, with focus in health behavior science from the University of North Carolina – Chapel Hill, and a master's in science in genetic counseling from the University of North Carolina – Greensboro. He has completed training in implementation science from the National Cancer Institute. He is also a former president of the American Board of Genetic Counseling. His research interests include outcomes of genomic screening, delivery of genetics services, and performance of multi-cancer early detection tests. Mr. Buchanan is interim director of Geisinger's MyCode Genomic Screening and Counseling program that screens biobank participants' exomes for clinically actionable findings associated with increased risk for cancer, cardiovascular disease and other conditions.

Carol Bult, PhD

Dr. Bult is a Professor and Knowlton Family Chair at The Jackson Laboratory (JAX) Mammalian Genetics campus in Bar Harbor, Maine. Prior to joining JAX in 1997, she was a founding faculty member of The Institute for Genomic Research (TIGR) where she played a key role in pioneering the application of large-scale genome sequencing to gene discovery and whole genome sequencing. At The Jackson Laboratory, Dr. Bult and her collaborators maintain the Mouse Genome Informatics (MGI) database of genetic and genomic data for mouse models of human disease. She is also a Principal Investigator for the Alliance of Genome Resources, a consortium of genome resources working to integrate data across diverse model organisms to facilitate investigation into the genomics of human health and disease. Dr. Bult is well known for her scientific contributions to the sequencing and annotation of the first three completed genome sequences for cellular organisms and to the annotation of the reference genome of the

laboratory mouse genome. She has published over 170 scientific papers and serves on numerous national and international scientific advisory boards. She previously served on the NHGRI Advisory (2014-2018) and the NHGRI Research Training Advisory Committee. She is currently a member of the NHGRI Genomic Medicine Working Group.

Rex Chisholm, PhD

Dr. Chisholm is the Adam and Richard T. Lind Professor of Medical Genetics in the Feinberg School of Medicine and professor of Cell and Developmental Biology and Surgery. He was the founding Director of the Center for Genetic Medicine. Since 2007 he has served as Vice Dean for Scientific Affairs in the Feinberg School. In October 2012 he was also appointed Associate Vice President for Research of Northwestern University. A faculty member at Northwestern University since 1984, Chisholm is author of over 125 scientific publications. His research focuses on genomics, bioinformatics and precision medicine. Chisholm leads a major DNA biobanking effort at Northwestern University, NUgene (www.nugene.org). NUgene enrolls research participants in a study focused on investigating the genetic contributions to human disease, therapeutic outcomes and gene-environment interactions. NUgene is a participant in the NHGRI-funded eMERGE network (https://emerge-network.org/) – a network of electronic medical record (EHR) linked biobanks. The goal of his current eMERGE network project is to establish a program for genomics-informed personalized medicine in partnership with Northwestern's health care affiliates. As Vice Dean for Scientific Affairs he is responsible for research space in Feinberg, research core facilities and the broader research environment. He also oversees the PhD and MS training programs in Feinberg.

Christopher G. Chute, MD, DrPH, MPH

Dr. Chute is the Bloomberg Distinguished Professor of Health Informatics, Professor of Medicine, Public Health, and Nursing at Johns Hopkins University, and Chief Research Information Officer for Johns Hopkins Medicine. He is also Section Head of Biomedical Informatics and Data Science and Deputy Director of the Institute for Clinical and Translational Research. He received his undergraduate and medical training at Brown University, internal medicine residency at Dartmouth, and doctoral training in Epidemiology and Biostatistics at Harvard. He is Board Certified in Internal Medicine and Clinical Informatics, and an elected Fellow of the American College of Physicians, the American College of Epidemiology, HL7, the American Medical Informatics Association, and the American College of Medical Informatics (ACMI), as well as a Founding Fellow of the International Academy of Health Sciences Informatics; he was president of ACMI 2017-18. He is an elected member of the Association of American Physicians.

His career has focused on how we can represent clinical information to support analyses and inferencing, including comparative effectiveness analyses, decision support, best evidence discovery, and translational research. He has had a deep interest in the semantic consistency of health data, harmonized information models, and ontology. His current research focuses on translating basic science information to clinical practice, how we classify dysfunctional phenotypes (disease), and the harmonization and rendering of real-world clinical data including electronic health records to support data inferencing. He became founding Chair of Biomedical Informatics at Mayo Clinic in 1988, retiring from Mayo in 2014, where he remains an emeritus Professor of Biomedical Informatics. He is presently PI on a spectrum of high-profile informatics grants from NIH spanning translational science including co-lead on the National COVID Cohort Collaborative (N3C). He has been active on many HIT standards

efforts and chaired ISO Technical Committee 215 on Health Informatics and chaired the World Health Organization (WHO) International Classification of Disease Revision (ICD-11).

Guilherme Del Fiol, MD, PhD

Dr. Del Fiol is professor and vice-chair for research at the Department of Biomedical Informatics, University of Utah. He is a national leader in the investigation of standards-based clinical decision support (CDS) and digital health interventions. Since 2008, he has served as elected co-chair of the Health Level Seven International (HL7) CDS Work Group, the leading standards development organization for health IT. He has led the development of several CDS standards, notably the HL7 Infobutton Standard, which is required for electronic health record (EHR) certification in the US. At the University of Utah, he leads the research arm of the ReimagineEHR initiative, which aims to transform clinicians' user experience with the EHR through standards-based approaches using standards such as the HL7 Fast Health Interoperability Resources (FHIR), SMART, and CDS Hooks standards.

Patricia Deverka, MD, MS

Dr. Deverka is a Senior Vice President at Vernaex, a consulting firm focused on providing life sciences companies with clinical development, commercial strategy and market access solutions. She leads the Precision Medicine practice area to develop evidence and reimbursement pathways for breakthrough tests and drugs focused on cancer, chronic diseases, and ultra-rare disorders. For the past 30 years, Dr. Deverka has worked in the fields of health economics and outcomes research in both non-profit and for-profit settings as a researcher, educator, consultant, and department head. She has extensive experience with drug and diagnostic reimbursement planning, cost- effectiveness analysis, real-world evidence studies, payer coverage policy analyses and patient-centered outcomes research, with more than 50 peer-reviewed publications on these topics. While working in academia and several non-profit firms, she has participated in numerous NIH-funded studies to evaluate policy barriers to clinical integration of new genomic technologies, and is a member of NHGRI's Genomic Medicine Work Group. Deverka has a medical degree from the University of Pittsburgh and is board certified in General Preventive Medicine and Public Health. She also has a master's degree in bioethics from the University of Pennsylvania and completed a policy fellowship at Duke University's Institute for Genome Sciences and Policy.

Gai Elhanan, MD

A veteran physician (Internal Medicine and Infectious Diseases) with more than 25 years of experience with healthcare information systems; research, design, development and implementation in clinical and administrative environments. Formal Medical Informatics education; completed a post-doctoral fellowship at the Medical Informatics department, New York Presbyterian Medical Center/Columbia University with broad skill set in the informatics field as well as unique knowledge in the field of semantic networks and medical/healthcare ontologies.

Currently a clinical data research scientist at the Center for Genomic Medicine, Desert Research Institute, Reno NV as a member of the Healthy Nevada Project (HNP). Based in northern Nevada, the HNP is a large population health genomic project that combines whole exome sequencing and clinical data with 50,000 all-comer participants.

Geoffrey S. Ginsburg, MD, PhD

Dr. Ginsburg is the Chief Medical and Scientific Officer of the All of Us Research Program at the National Institutes of Health. He leads the Division of Medical and Scientific Research and is responsible for helping to set the scientific vision and strategy for the program. He also oversees the program's collection and curation of data, and integration of new data types to support a wide range of impactful scientific discoveries. Prior to joining All of Us, Ginsburg was founding director for the Center for Applied Genomics & Precision Medicine in the Duke University School of Medicine where he pioneered translational genomics and the development of novel diagnostics. At Duke, he was professor of medicine, biostatistics and bioinformatics, pathology, and biomedical engineering. He also was a professor in the School of Nursing; he will remain adjunct professor of medicine. He has held senior leadership roles at Millennium Pharmaceuticals Inc. and was a member of the Harvard Medical School faculty.

Throughout his career, Ginsburg has demonstrated a strong commitment to interdisciplinary science and innovation, with work spanning oncology, infectious diseases, cardiovascular disease, and metabolic disorders. He has held leadership roles in the U.S. and internationally, serving as co-chair of the National Academies' Roundtable on Genomic and Precision Health, a founding co-chair of the International HundredK+ Cohorts Consortium, and founder and president of the Global Genomic Medicine Collaborative (G2MC), a not-for-profit organization aimed at creating international partnerships to advance the implementation of precision medicine. At NIH, Ginsburg has served on the board of external experts for the National Heart, Lung, Blood Institute, as an advisory council member to the National Human Genome Research Institute and the National Centers for Advancing Translational Sciences, and most recently on the Advisory Committee of the Director of NIH.

He received his M.D. and Ph.D. in biophysics from Boston University and completed an internal medicine residency at Beth Israel Hospital in Boston. Subsequently, he pursued postdoctoral training in clinical cardiovascular medicine at Beth Israel Hospital and in molecular biology at Children's Hospital as a Bugher Foundation Fellow of the American Heart Association.

Suzanne Haga, PhD

Dr. Haga is an Associate Professor at Duke University School of Medicine, Division of General Internal Medicine. She also has a secondary appointment in the Sanford School of Public Policy and the Department of Biology at Duke University. She oversees several education programs for undergraduates and post-doctoral trainees and regularly teaches undergraduate courses in genomics, bioethics, and public policy. Her research interests focus on issues affecting the translation of genomics to clinical practice, particularly in the field of pharmacogenetic testing, and patient and provider education.

Rizwan Hamid, MD, PhD

Dr. Hamid is the Director of Genetics and Genomic Medicine at Vanderbilt University Medical Center (VUMC) and the Monroe Carell Jr. Children's hospital. He is also the Director of the Vanderbilt Undiagnosed Disease Program. He sees pediatric and adult patients with rare and undiagnosed diseases as a pediatric geneticist. Dr. Hamid's life-long passion is to help these patients.

Dr. Hamid's primary research focuses on differences between disease development and severity amongst individuals—sometimes even amongst close family members. His laboratory utilizes stem cell biology and animal models to answer these questions in rare mendelian lung diseases.

Dr. Hamid is also interested in how we best implement genomic medicine for a non-genetic provider in a large health care system setting. As such, he leads an institutionally funded project at VUMC to test different approaches that might result in improved and increased utilization of genomics in the clinical setting with the long-term goal of improving outcomes.

Carol Horowitz, MD, MPH

Dr. Horowitz is Professor of Population Health Science and Policy and, Professor of Medicine and a practicing general internist at the Icahn School of Medicine at Mount Sinai. She is the founding Director of Mount Sinai's Institute for Health Equity Research and the founding Dean for Gender Equity in Science and Medicine. Her research focuses on using Stakeholder-Engaged and Community-Based Participatory Research to address health disparities. As a continuously funded NIH investigator, her special interests are in chronic disease prevention and control, and the intersection of social, structural, biological and clinical determinants of health. All of Dr. Horowitz's clinical trial, secondary data, qualitative and mixed method research directly engages stakeholders, including community leaders, and advocates, clinicians, policymakers and entrepreneurs. The diverse and durable partnerships she helped build over three decades have informed and impacted health, policies, systems and environments, related to diabetes, obesity, kidney and cardiovascular disease, genomics, environmental health, youth, cancer, and COVID-19. Dr. Horowitz mentors diverse trainees and faculty in the US and in low and middle income countries. She is active on NIH study sections, leading NIH consortia and as an international lecturer.

Peter Hulick, MD, MMSc

Dr. Hulick is the Janardan D. Khandekar, MD, Chair of Personalized Medicine and serves as Medical Director of the Mark R. Neaman Center for Personalized Medicine and Division Head for Medical Genetics at NorthShore University HealthSystem, which applies genomic information to prevention, diagnosis and treatment of human disease with the vision to bring genomics guided care to every patient and their family. He joined NorthShore as an attending physician in medical genetics in 2008 and became Division Head of Medical Genetics in 2012. Dr. Hulick also serves as a Clinical Associate Professor in the Department of Human Genetics at the University of Chicago Pritzker School of Medicine. Previously, he served as an attending physician in medical genetics at Massachusetts General Hospital. Dr. Hulick earned his medical degree from Jefferson Medical College in 2001. He completed a residency in internal medicine at St. Luke's Hospital – Mayo Clinic, and completed a clinical fellowship in medical genetics at Harvard Medical School.

Cynthia A. James, PhD, CGC

Dr. James is an Associate Professor of Medicine (Cardiology) and Genetic Medicine and certified genetic counselor at Johns Hopkins University. She is the Research Director of both the Johns Hopkins Center for Inherited Heart Diseases and the Johns Hopkins ARVC Precision Medicine Center of Excellence. Her research focuses on: 1) investigating the interplay of genotype and environmental factors on disease expression and outcomes in ARVC, 2) defining the genetic architecture of inherited cardiomyopathies, and 3) improving cardiovascular genetic counseling outcomes. Her team is currently engaged in a 3-arm randomized clinical trial testing two complementary approaches to shifting the primary adult cardiovascular genetic counseling appointment post-test. This study characterizes the impact of results-focused counseling on patient empowerment and psychosocial well-being, medical adherence, and genetic counseling efficiency, leveraging electronic medical record data sources.

Dr. James received her undergraduate degree in biology from Cornell University in Ithaca, NY. She earned her Sc.M. in Genetic Counseling at Johns Hopkins School of Public Health and her Ph.D. in Human Genetics at Johns Hopkins University. Dr. James joined the Johns Hopkins faculty in 2013 after a decade as a practicing genetic counselor in the Johns Hopkins ARVC program.

Gail Jarvik MD, PhD

Dr. Jarvik is a joint Professor of Medicine and Genome Sciences and heads the Division of Medical Genetics at the University of Washington. She holds the Arno G. Motulsky Endowed Chair in Medicine. She is an internist and clinical medical geneticist whose research focuses on the statistical genetic analysis of common diseases in adults including cancer, vascular disease, and dementia. She has broad research interests in the implementation of genomic medicine, including her work as Principle Investigator in the national consortia implementing genomic medicine, including Electronic Medical Records and Genomics (eMERGE) Network, Clinical Sequencing Evidence-Generating Research (CSER), Undiagnosed Disease Network (UDN), and the All of Us (AoU) consortia. Her research in biomedical ethics includes returning genomic research results to participants and studying the impact of regulations on genomic research. She was the 2021 president of the American Society of Human Genetics and serves nationally as a mentor for DEI trainees.

Janina M. Jeff, PhD, MS

Population geneticist, bioinformatician, STEAM-activist, educator, motivational speaker, and podcaster, Dr. Janina M. Jeff is the first African American to graduate with a PhD in Human Genetics from Vanderbilt University and graduate of Spelman College (class of 2007). She is currently a Staff Bioinformatics Scientist at Illumina, where she develops pipelines for content annotation, selection, and design of population genome-wide content as well as selection of clinically annotated variants for Illumina's genotyping array portfolio that enables healthy population screening. In 2018, Janina was selected as one of three winners (out of 18,000) from Spotify's Sound-Up Bootcamp for her podcast, "In Those Genes", is an international award-winning podcast that uses genetics to decode the lost histories of African descended Americans through the lens of Black culture. In Those Genes has been recognized by the American Society of Human Genetics, NY Festivals, IndieWire, The New York Times, and Third Coast Audio Festival. Janina was recently named as one of the top 100 Influential African Americans by The Root magazine as well as Top 40 Under 40 Alumna with Spelman College, The National Quality Minority Forum and The Network Journal. The podcast has also received grant funding from Spotify, Illumina, and 500 Women Scientist. Her TEDx talk, "Afrofuturism Through the Power of the Genome" similar to her work challenges the misuse of genetics information and empowers the Black community to learn the value that lies within their genomes work.

Bruce Korf, MD, PhD

Dr. Korf is Distinguished Professor of Genetics, Wayne H. and Sara Crews Finley Endowed Chair in Medical Genetics, Associate Dean for Genomic Medicine, UAB School of Medicine, Chief Genomics Officer, UAB Medicine and Co-Director of the UAB-HudsonAlpha Center for Genomic Medicine. He is a medical geneticist, pediatrician, and child neurologist, certified by the American Board of Medical Genetics (clinical genetics, clinical cytogenetics, clinical molecular genetics), American Board of Pediatrics, and American Board of Psychiatry and Neurology (child neurology). Dr. Korf is past president of the Association of Professors of Human and Medical Genetics, past president of the American College of Medical Genetics and Genomics, and current president of the ACMG Foundation for Genetic and

Genomic Medicine. He has served on the Board of Scientific Counselors of the National Cancer Institute and the National Human Genome Research Institute at the NIH. His major research interests are the natural history, genetics, and treatment of neurofibromatosis and the integration of genomics into medical practice. He serves as principal investigator of the Department of Defense funded Neurofibromatosis Clinical Trials Consortium. He is co-author of Human Genetics and Genomics (medical student textbook, now in fourth edition, and Emery and Rimoin's Principles and Practice of Medical Genetics (now in 7th edition).

Latrice Landry, MS, PhD, MSC

Dr. Landry is an instructor specializing in developing systems-based approaches to achieving equitable translation of precision medicine and precision public health. She received both her master's degree in Policy and her PhD in Nutrition from Tufts University. Her doctoral research focused on the interactions between diet and genetics as determinants for dyslipidemia in African Americans in the Jackson Heart Study. As a doctoral student, Dr. Landry was awarded the Albert Schweitzer fellowship, nominated as a finalist in the American Society for Nutrition's Clinical Emerging Leaders Award, and was given the Presidential Award for Citizenship and Public Service at Tufts University. In 2015, following her doctoral research she joined Harvard Medical School's Biomedical Informatics Fellowship Program to study biomedical information systems (clinical informatics and bioinformatics) as tools for biomarker translation. In 2018, she graduated from the Harvard Medical School's Clinical Molecular Genetics' training program enabling her to integrate her biomarker expertise with direct patient care. She was the Inaugural Food and Drug Administration's Genomic Medicine and Minority Health Fellow. Dr. Landry has also been recognized as a thought leader in minority health and precision medicine by the National Minority Quality Forum in 2017, as a top 10 under 40 rising stars in Business and Academia by Genetic Engineering and Biotechnology on 2018 and an American Society of Human Genetics, Human Genetics Scholar in 2019. True to her commitment in supporting aspiring minority scientists she created a research training program for high-school and undergraduate students. In 2020, she joined the National Minority Quality Forum, Quest Diagnostics, and the Centene Corporation for the launch of the Minority and Rural Coronavirus Insights Study- a study aimed at understanding the role of COVID in minority communities. In 2021, she received the Dana Farber Cancer InstitutesCURE mentoring award. She has a sincere passion for building equitable systems, eliminating racism from research and medicine, and expanding minority participation and leadership in the biomedical workforce. Understanding that the present moment requires difficult conversations, honest truth, and persistence, she is encouraged about the promise of precision medicine and precision public health for all.

Teri Manolio, MD, PhD

Dr. Manolio is Director of the Division of Genomic Medicine, National Human Genome Research Institute, Bethesda, Maryland, Professor of Medicine at the Uniformed Services University of the Health Sciences, and is a practicing internal medicine physician at the Walter Reed National Military Medical Center, Bethesda. She completed undergraduate and medical training at the University of Maryland, Boston City Hospital, and the Johns Hopkins School of Medicine, and her PhD in human genetics and genetic epidemiology at the Johns Hopkins School of Hygiene and Public Health. Dr Manolio was previously at the National Institutes of Health's National Heart, Lung, and Blood Institute where she was involved in large-scale cohort studies such as the Cardiovascular Health Study and the Framingham Heart Study. Her research interests focus on genome-wide association studies of complex diseases,

ethnic differences in disease risk, integrating genomic research into electronic medical records, and incorporating genomic findings into clinical care.

Deven McGraw, JD, MPH

Deven McGraw is the lead for Data Stewardship and Data Sharing at Invitae. Previously, she co-founded Ciitizen, a platform for patients to gather their health information, prior to its acquisition by Invitae. From 2015-2017, she directed U.S. health privacy and security as Deputy Director, Health Information Privacy at the HHS Office for Civil Rights and Chief Privacy Officer (Acting) of the Office of the National Coordinator for Health IT. Widely recognized for her expertise in health privacy, she directed the Health Privacy Project at the Center for Democracy & Technology for six years and led the privacy and security policy work for the HITECH Health IT Policy Committee. She also served as the Chief Operating Officer of the National Partnership for Women and Families. She advised health industry clients on HIPAA compliance and data governance while a partner at Manatt, Phelps & Phillips, LLP. Deven graduated magna cum laude from Georgetown University Law Center and has a Masters of Public Health from Johns Hopkins University.

Howard McLeod, PharmD

Dr. McLeod is an expert in precision medicine, having made novel contributions at the discovery, translation, implementation, and policy levels. He is the Executive Clinical Director – Precision Health across the 7 state Intermountain HealthCare System. Most recently he was Medical Director for Precision Medicine at the Geriatric Oncology Consortium. Dr McLeod has chaired the NHGRI eMERGE network external scientific panel and was a recent member of both the FDA committee on Clinical Pharmacology and the NHGRI Advisory Council. Dr McLeod has been recognized as a Fellow of both the American Society of Clinical Oncology and the American College of Clinical Pharmacy. Howard has published over 590 peer reviewed papers on pharmacogenomics, applied therapeutics, or clinical pharmacology and continues to work to advance innovative healthcare.

Nancy Mendelsohn, MD

Dr. Mendelsohn is a Chief Medical Officer (CMO) working with Optum Frontiers Therapies (OFT) and Senior Medical Director for Emerging Therapies in Optum Health Solutions. She has deep expertise as a board-certified clinical geneticist, caring for children and adults with rare disorders. Since joining UHG in fall of 2018, Dr. Mendelsohn has served as the Senior Vice President Medical Affairs for genetics and rare diseases at United Health Group (UHG) Medical Affairs and CMO of the Complex Health Solutions (CHS) team helping our most complex patients and families across special needs initiative (SNI) and Complex Care Concierge (C3). She has been active across the enterprise to provide clinical guidance and support to UHG's strategic priorities related to genomic medicine working across Optum, UHG Research and Design, and UnitedHealthcare.

Prior to her position at UHG Dr. Mendelsohn worked 19 years at Children's Minnesota as genetics faculty and became chief of specialty pediatrics. She has 30 years as a board-certified medical geneticist and is a national leader in the clinical genetics' community. Dr. Mendelsohn completed a residency in pediatrics and fellowship in medical genetics at Washington University Children's hospital in St. Louis, Missouri. Dr. Mendelsohn has more than 50 peer reviewed publications, book chapters, and presentations. She has specific clinical expertise in lysosomal storage disorders, has served broadly on national committees for the American Academy of Pediatrics (AAP), American Board of Medical Genetics & Genomics (ABMGG), Foundation Board for American College of Medical Genetics & Genomics, as well

as other genetics committees within the state of Minnesota. In Dr. Mendelsohn's spare time, she enjoys cooking, spending time with her family, and traveling.

Travis Osterman, DO, MS

Dr. Osterman is a practicing medical oncologist, informatician, and Director of Cancer Clinical Informatics at the Vanderbilt-Ingram Cancer Center. He is board-certified in internal medicine, medical oncology, and clinical informatics; and completed a master of science in biomedical informatics at Vanderbilt University.

Dr. Osterman's clinical interest in oncology is lung cancer. He is passionate about making real-world EHR available for discovery. Nationally, Dr. Osterman serves on the National Comprehensive Cancer Network (NCCN) Electronic Health Record (EHR) workgroup with a focus on leveraging the EHR to promote innovation and standardize best practices across large cancer centers. He also serves on the Epic Adult Oncology Steering Board and chairs Epic's Beacon Community Operations Group.

He leads Vanderbilt University Medical Center's Clinical Genomics workstream where has he focused on increasing the availability of structured genomic results.

Casey Overby Taylor, PhD

Dr. Overby Taylor is associate professor of Medicine and Biomedical Engineering in Johns Hopkins School of Medicine (SoM) and core faculty member of the Institute for Computational Medicine. She has an affiliation with the Biomedical Informatics Data Science (BIDS) Section in the Division of General Internal Medicine, and joint appointments in the Department of Health Policy and Management in the Johns Hopkins Bloomberg School of Public Health, and the Computer Science Department in the Johns Hopkins Whiting School of Engineering. Dr. Taylor's research draws from biomedical informatics and the related field of biomedical data science, to address the challenge of how to incorporate digital health technologies into clinical practice, particularly for genomics. She has previously served as co-Chair of the Electronic Health Records Integration Workgroup as part of the NIH NHGRI-funded electronic medical records and genomics (eMERGE) Network (2015-2020). Dr. Taylor also received a 2020 NHGRI-funded Genomic Innovator Award to recognize her research in developing and evaluating methods to incorporate genomic results in clinical decision support (R35 HG010714).

Daryl Pritchard, PhD

Dr. Pritchard is the Senior Vice President of Science Policy at the Personalized Medicine Coalition (PMC), where he leads PMC's efforts to increase awareness and understanding of personalized medicine; identify and address barriers to the adoption of personalized medicine into the health care system; and develop and promote appropriate clinical, health care infrastructure, regulatory, and payment policies that will help advance patient-centered, personalized health care.

Before coming to PMC, Dr. Pritchard served as the Director of Policy Research at the National Pharmaceutical Council (NPC). Prior to joining NPC, he served as the Director of Research Programs Advocacy and Personalized Medicine at the Biotechnology Industry Organization (BIO).

Dr. Pritchard received his Ph.D. and master's degree in genetics from the George Washington University, and completed a post-doctoral research fellowship at the Children's National Medical Center. He was awarded the first American Society of Human Genetics (ASHG)/National Human Genome Research

Institute (NHGRI) Fellowship in Genetics and Public Policy, where he worked as a health legislative assistant in the U.S. House of Representatives.

Erin M. Ramos, Ph.D., M.P.H.

Dr. Ramos is the Deputy Director of the Division of Genomic Medicine at the National Human Genome Research Institute (NHGRI). She joined NHGRI in 2006 and is committed to advancing the application of genomics to medical science and clinical care. Dr. Ramos received her M.P.H. and Ph.D. in the multidisciplinary field of public health genetics from the University of Washington where her research focused on the genetic epidemiology of Alzheimer's disease and the ethical, legal, and social implications (ELSI) that surround genomics research. Dr. Ramos is a Project Scientist for Clinical Genome Resource (ClinGen), a consortium of nearly 2,000 clinicians and scientists from 40 countries who are building a central resource that classifies the clinical relevance of genes and variants for use in precision medicine and research.

Dr. Ramos contributes to data sharing (GDS) activities across the NIH. She is a co-Chair of NHGRI's GDS Governance Committee, chaired the Data Access Committee (DAC) for the Genetic Association Information to provide access to some of the first genome-wide association studies in dbGaP, and currently serves on the National COVID Cohort Collaborative DAC. Her research interests include incorporating genomic findings into clinical care and improving methods for complex disease research including genome-wide association studies, gene-environment interactions, genomic risk assessment and the application of multi-omics technologies for health and disease.

Heidi Rehm, PhD

Heidi Rehm is the Chief Genomics Officer in the Department of Medicine and at the Center for Genomic Medicine at Massachusetts General Hospital working to integrate genomics into medical practice. She is a board-certified laboratory geneticist and Medical Director of the Broad Institute Clinical Research Sequencing Platform working to guide genomic testing for clinical and clinical research use. She is also Co-Director of the Program in Medical and Population Genetics at the Broad Institute and Professor of Pathology at Harvard Medical School. She is a principal investigator of ClinGen, providing free and publicly accessible resources to support the interpretation of genes and variants. Rehm also co-leads the Broad Center for Mendelian Genomics focused on discovering novel rare disease genes and co-leads the Matchmaker Exchange to also aid in gene discovery. She is a strong advocate and pioneer of open science and data sharing, working to extend these approaches through her role as vice chair of the Global Alliance for Genomics and Health. Rehm is also a principal investigator of the Broad-LMM-Color All of Us Genome Center supporting the sequencing and return of results to a cohort of one million individuals in the US and co-leading gnomAD, the Genome Aggregation Database.

Renee Rider, JD, CGC

Renee Rider is a genetic counselor and a Program Director for the National Human Genome Research Institute (NHGRI) Implementing Genomics in Practice Pragmatic Trials Network (IGNITE PTN).

Prior to joining NHGRI, Renee worked at the University of Utah Alzheimer's Center and the Veterans' Administration. She provided genetic counseling for adult diseases in a wide variety of specialties and was a part of growing the VA's robust, national telehealth genetic counseling consult service. Her research include developing the genetic counseling workforce and using alternative genetic education and counseling methods to increase access to genetic services. Renee received her master's in science

in genetic counseling from the University of Utah and a juris doctor from Northeastern University School of Law. She is currently working on her Doctor of Public Health at George Washington University.

Robb Rowley, MD

Dr. Rowley is an Internal Medicine Physician and is the Program Director for the National Human Genome Research Institute (NHGRI) Electronic Medical Records and Genomics (eMERGE) Network. The national Network combines DNA biorepositories with electronic medical record (EMR) systems for large scale, high-throughput genetic research in support of implementing genomic medicine.

Prior to starting at NHGRI, he spent thirteen years in private practice and hospital management, where he provided clinical assessments and medical care for adult diseases influenced by genetically influenced conditions to improve patient risk stratification and individualize treatments. Dr. Rowley previously served in the United States Air Force Surgeon General's Office in Washington D.C. as the Chief of Medical Bioinformatics and Genomics. During this time, he established genomic policy and conducted genomic research for the United States Air Force. Dr. Rowley has also been instrumental in establishing national and international plans and policies for incorporating genomics into biosurveillance systems and biotechnology for the Department of Defense (DoD) and North Atlantic Treaty Organization (NATO). Dr. Rowley has experience with managing multiple U.S. Food and Drug Administration (FDA) clinical trials, along with presenting original research at international scientific and medical meetings.

Krystal Tsosie, MPH, MA, PhD

Dr. Tsosie is an Indigenous geneticist-bioethicist and incoming Assistant Professor at Arizona State University. As an advocate for Indigenous genomic and data sovereignty, she co-founded the first US Indigenous-led biobank, a 501c3 nonprofit research institution called the Native BioData Consortium. Much of her current research centers on ethical engagement with Indigenous communities in precision health. She also incorporates biostatistics, genetic epidemiology, public health, and computational approaches to cancer health disparities. At the laboratory bench, she developed and patented a combined targeted ultrasound imaging and chemotherapeutic drug delivery device for treating early metastases in cancer. Krystal's research and educational endeavors have received international media attention in such outlets as The New York Times, PBS NOVA, Washington Post, NPR, The Atlantic, Forbes, Boston Globe, among others.

Jason Vassy, MD, MPH, MS

Dr. Vassy is an Associate Professor of Medicine at Harvard Medical School, a primary care clinician-investigator at the VA Boston Healthcare System (VABHS) and Brigham and Women's Hospital (BWH), and a founding member of Precision Population Health at Ariadne Labs. He completed his MD degree from Washington University School of Medicine, internal medicine residency at the University of Pennsylvania, and a masters in genetic epidemiology at the Harvard T.H. Chan School of Public Health. He directs the Genomes2Veterans research program, where his research examines the clinical utility of genetic and genomic testing in primary care contexts, including pharmacogenetic testing, polygenic risk scores, and return of unanticipated genetic results.

Karriem Watson, DHS, MS, MPH

Dr. Watson is the Chief Engagement Officer of the National Institutes of Health's All of Us Research (AoU) Program. Karriem leads the All of Us Research Program's efforts to foster relationships with

participants, communities, researchers, and providers across the U.S. to help build one of the largest, most diverse health databases of its kind to study health and illness.

Prior to joining the NIH, Karriem spent over 15 years as a community engaged research scientists with prior research funding from the NIH addressing cancer prevention and control. Karriem also held administrative roles in leading research and engagement in Federally Qualified Health Centers (FQHCs) in the Chicagoland area.

Ken Wiley, Jr., PhD

Dr. Wiley, Jr. is a Program Director in the Division of Genomic Medicine at the National Human Genome Research Institute (NHGRI). He received his B.S. in Psychology from Morehouse College, and his Ph.D. in Pharmacology from Meharry Medical College in collaboration with the University of Iowa. Dr. Wiley, Jr. manages several programs that focus on leveraging informatics to reduce barriers for integrating genomics and genomic related information to improve clinical care. These programs include the Electronic Medical Records in Genomics (eMERGE) Network, The NHGRI-European Bioinformatics Institute's Catalog of published genome-wide association studies, targeted arrays and summary statistics (The NHGRI-EBI GWAS Catalog), and the NHGRI Genomic Data Science Analysis, Visualization, and Informatic Lab-space (AnVIL), as well as the Polygenic Risk Methods in Diverse Populations (PRIMED) Consortium which focus on developing and evaluating methods to improve the use of polygenic risk scores to predict disease and health outcomes in diverse ancestry populations. In addition, he is the Program Director for Human, Heredity, and Health in Africa (H3Africa) Consortium's H3ABioNet, a pan-African bioinformatics network which supports developing bioinformatics capacity within Africa.

Marc S. Williams, MD, FAAP, FACMG, FACMI

Marc S. Williams, MD, FAAP, FACMG, FACMI is a clinical geneticist. He is professor and director emeritus of Geisinger's Genomic Medicine Institute. He served as the co-PI of the Geisinger eMERGE project and is the medical director of the whole genome sequencing clinical research project. His current research is focused on the implementation of genomic and precision medicine. He serves on the NHGRI Genomic Medicine working group. He has participated in the Personalized Medicine Workgroup of the Department of Health and Human Services' American Health Information Community Task Force and was a member of the Secretary's Advisory Committee for Genetics, Health and Society. He was a member of the EGAPP working group. He is a member of the American College of Medical Genetics and Genomics (ACMG) Board of Directors, serving as Vice-President for Clinical Genetics, then rejoined the board in 2019 and is the current President. He is past chair of the ACMG Committee on the Economics of Genetic Services and founded the ACMG Quality Improvement Special Interest Group. He is a member of the Scientific Advisory Board of the Clinical Pharmacogenetic Implementation Consortium (CPIC) and a member of the CPIC informatics committee. He recently joined the Scientific Advisory Boards of the NIH Undiagnosed Diseases Project, and Online Mendelian Inheritance in Man. He has authored over 200 articles on a variety of topics including the economic evaluation and value of genetic services, implementation of genomic medicine, and the use of informatics to facilitate genomic medicine and precision health.

Support Team Biographies

Jahnavi Narula, BS

Jahnavi is a Scientific Program Analyst at the National Human Genome Research Institute (NHGRI). She is currently working for the Electronic Medical Records and Genomics (eMERGE) Network, the Genomic Medicine Working Group (GMWG), and Advancing Genomic Medicine Research (AGMR). She graduated from the University of California, Los Angeles, with a B.S. in Biology and a minor in English. She is interested in scientific communication and plans to pursue a career in medicine.

Teji Rakhra-Burris, MBA

Ms. Rakhra-Burris is a Research Program Leader Sr. with over 20 years of experience in administration and program management of complex scientific research programs in higher education. In addition to participating in the administrative/operational leadership of the Duke University Center for Applied Genomics & Precision Medicine (2013-2022), she was the Program Manager for the NHGRI-funded IGNITE Network Coordinating Center and remains the Program Manager for the Duke Clinical Group. She also oversees the proposal development, management and implementation of various other precision medicine clinical research efforts being implemented in the CPM.

Prior to joining Duke, she was the Associate Director for Research with the University of North Carolina Institute for Pharmacogenomics and Individualized Therapy for five years. Past experience also included the management of a CDC-funded public health genetics program at the UNC School of Public Health, and an NIEHS-funded cooperative agreement investigating the feasibility of establishing a national twin registry for genetic and environmental association studies. Ms. Rakhra-Burris has a background in business administration from the University of Vermont and a Master's Degree in Molecular Genetics from Washington University in St. Louis.

Ella Samer, BS

Ella is a Scientific Program Analyst at the National Human Genomic Research Institute (NHGRI) and is currently working for the Implementing Genomics in Practice (IGNITE) Network, the Impact of Genomic Variation on Function Consortium (IGVF), and the Encyclopedia of DNA Elements (ENCODE) Consortium. She graduated from Washington College in Chestertown, Maryland with a B.S. in Chemistry, and a minor in Biology. She is pursuing a career path of Physician Associate Studies and has special interests in genomic medicine and women's health.

Pamela G. Williams

Sr. Program Coordinator for the Duke Center for Applied Genomics and Precision Medicine. She provides support for the planning and implementation of the NHGRI Research Training & Career Development Annual Meeting and provides administrative support for the Duke Precision Medicine program and forum at Duke University.

Registered Attendees Contact Information

Name	Institute	Email Address
Abaan, Ogan	Illumina	oabaan at illumina dot com
Abul-Husn, Noura	Mount Sinai	noura.abul-husn at mssm dot edu
Addepalli, Durga	NCI	kanakadurga.addepalli at nih dot gov
Adimoelja, Alvina	Stanford University	alvinaa at stanford dot edu
Al-Mulla, Fahd	Dasman Diabetes Institute	fahd at al-mulla dot org
Alvarellos, Maria	Lifebit Biotech	mariaalvarellos at lifebit.ai
Aquilante, Christina	University of Colorado Anschutz Medical Campus	christina.aquilante at cuanschutz dot edu
Aronson, Sandy	Mass General Brigham	saronson at partners dot org
Assimes, Themistocles	Stanford University School of Medicine / Palo Alto VA	tassimes at stanford dot edu
Atanda, Halimat	Federal University of Technology Akure	gearthdexter at gmail dot com
Azrin, Susan	National Institute of Mental Health	susan.azrin at nih dot gov
Bakken, Suzanne	Columbia University	sbh22 at cumc.columbia dot edu
Baldridge, Dustin	Washington University in St. Louis	dbaldri at wustl dot edu
Bandel, Lorelei	Mayo Clinic	bandel.lorelei at mayo dot edu
Bangash, Hana	Mayo Clinic	Bangash.hana at mayo dot edu
Baroch, Jacob	NHGRI	jake.baroch at nih dot gov
Beaman, Makenzie	Duke University	mmb95 at duke dot edu
Bell, Claire	Johns Hopkins University School of Medicine	cwenger3 at jhmi dot edu
Berg, Jonathan	UNC Chapel Hill	jsberg at med.unc dot edu
Berger, Adam	NIH	adam.berger at nih dot gov
Bickford, Carol	American Nurses Association	carol.bickford at ana dot org
Biswas, Sawona	UCSF	sawona.biswas at ucsf dot edu
Bly, Zo	NHGRI	zo.bly at nih dot gov
Boland, Dylan	PGDX	dboland at pgdx dot com
Bowman, Loretta	Personal Genome Diagnostics	lbowman at pgdx dot com
Bradbury, Angela	University of Pennsylvania	angela.bradbury at pennmedicine.upenn dot edu
Brockman, Deanna	Color Health	deanna.brockman at color dot com
Buchanan, Adam	Geisinger	ahbuchanan at geisinger dot edu
Bueser, Teofila	Guy's & St Thomas' Hospital Foundation Trust	tootie.bueser at nhs dot net

Name	Institute	Email Address
Bult, Carol	Jackson Laboratory (JAX)	carol.bult at jax dot org
Burkardt, Deepika	NIH	deepika.burkardt at nih dot gov
Butte, Atul	UCSF	atul.butte at ucsf dot edu
Byfield, Grace	UNC Chapel Hill	grace_byfield at med.unc dot edu
Calzone, Kathleen	National Cancer Institute, Center for Cancer Research, Genetics Branch	calzonek at mail.nih dot gov
Caropreso, Vito	Labcorp	Vcaropreso at pgdx dot com
Casas-Silva, Esmeralda	National Cancer Institute	emi.casas-silva at nih dot gov
Chang, Christine	NIH	christine.chang2 at nih dot gov
Chang, Qiang	University of Wisconsin-Madison	qchang at waisman.wisc dot edu
Chen, Flavia	UCSF	flavia.chen at ucsf dot edu
Cheung, Chloe	Icahn School of Medicine at Mount Sinai	chloeyihang.cheung at mssm dot edu
Chisholm, Rex	Northwestern University	r-chisholm at northwestern dot edu
Chute, Christopher	Johns Hopkins University	chute at jhu dot edu
Cimino, James	University of Alabama-Birmingham	ciminoj at uab dot edu
Clark, Susan	Briljent	sclark at briljent dot com
Clements, Eriko	AMP	eclements at amp dot org
Cole, Jordan	Washington University in St. Louis	jordan.cole at wustl dot edu
Colley, Heather	NIH/NHGRI	heather.colley at nih dot gov
Cong, Xiaomei	University of Connecticut	xiaomei.cong at uconn dot edu
Correa-Mendez, Margarita	NCI	margarita.mendez at nih dot gov
Crouse, Andrew	UAB	acrouse at uab dot edu
Crowgey, Erin	Incyte	ecrowgey at incyte dot com
Curry, Timothy	Mayo Clinic	curry.timothy at mayo dot edu
Davidsen, Tanja	NIH	tanja.davidsen at nih dot gov
Dayal, Jyoti	NIH/NHGRI	jyotig at nih dot gov
De La Vega, Francisco	Stanford University School of Medicine	delavefm at gmail dot com
Decker, Emma	Illumina	edecker at illumina dot com
Del Fiol, Guilherme	University of Utah	guilherme.delfiol at utah dot edu
Dent, Laura	Mount Sinai Icahn School of Medicine	laura.dent at mssm dot edu
Deverka, Patricia	Veranex	patricia.deverka at veranexsolutions dot com
Di Francesco, Valentina	NHGRI	vdifrancesco at mail.nih dot gov
Dickman, Erin	Oncology Nursing Society	edickman at ons dot org
Diez de los Rios, Celia	EONS	celiadiezrios at gmail dot com

Name	Institute	Email Address
Dincer, Aslihan	MSSM	aslidincer at gmail dot com
Dissanayake, Vajira	University of Colombo	vajira at anat.Cmb.AC dot lk
Dolin, Bob	Elimu Informatics	bdolin at elimu.io
Dong, Catherine	National University of Singapore	nurdy at nus dot edu dot sg
Dong, Qing	UCSF	qing.dong at ucsf dot edu
Doss, Argenia	National Institutes of Health	argenia.doss at nih dot gov
Dunnenberger, Mark	NorthShore University HealthSystem	mdunnenberger at northshore dot org
Eldridge, Lisa	Illumina	leldridge at illumina dot com
Elhanan, Gai	Desert Research Institute	Gai.Elhanan at dri dot edu
Empey, Philip	University of Pittsburgh	pempey at pitt dot edu
Etheredge, Lynn	Rapid Learning Project	lyneth1 at mac dot com
Eugene, Andy	State Hospital	andyeugene.md at gmail dot com
Fan, Xiao	Columbia University	xiaofan.hit at gmail dot com
Feingold, Elise	National Human Genome Research Institute	feingole at mail.nih dot gov
Feldthouse, Dawn Marta	Columbia University	dawn.feldthouse at columbia dot edu
Felsenfeld, Adam	NIH	adam_felsenfeld at nih dot gov
Filipski, Kelly	NCI	kelly.filipski at nih dot gov
Fink, Aliza	NORD	afink at rarediseases dot org
Formea, Christine	Intermountain Healthcare	christine.formea at imail dot org
Fornage, Myriam	University of Texas Health Science Center at Houston	myriam.fornage at uth.tmc dot edu
Foster, Brianna	National Human Genome Research Institute (NHGRI)	bri.foster at nih dot gov
Fowler, Doug	University of Washington	dfowler at uw dot edu
Friend, Patricia	Loyola University Chicago	pfriend at luc dot edu
Gajapathy, Manavalan	UAB	manag at uab dot edu
Gaynor, Brady	University of Maryland School of Medicine	bgaynor at som.umaryland dot edu
Ginsburg, Geoffrey	NIH	geoffrey.ginsburg at nih dot gov
Goldberg, Allon	University of Michigan-Flint	allong at umich dot edu
Gomez, Sara	GUMC	sg1328 at georgetown dot edu
Gregurick, Susan	NIH	gregurickssk at nih dot gov
Gropman, Andrea	Children's National Hospital	agropman at childrensnational dot org
Guadalupe Bastidas, Yessenia Isabel	IIN	yesisaguabas at gmail dot com
Gui, Hongsheng	Henry Ford Health System	hgui1 at hfhs dot org
Gunawardena, Kawmadi	Human Genetics Unit, Faculty of Medicine, Colombo	kawmadigunawardena at gmail dot com

Name	Institute	Email Address
Gunter, Chris	NIH	chris.gunter at nih dot gov
Gutierrez-Valverde, Juana Mercedes	UTHSC	juanymeche at yahoo dot com dot mx
Haga, Susanne	Duke University	susanne.haga at duke dot edu
Haidar, Cyrine	St. Jude Children's Research Hospital	cyrine.haidar at stjude dot org
Halley, Meghan	Stanford University	mhalley at stanford dot edu
Hamid, Rizwan	Vanderbilt University Medical Center	rizwan.hamid at vumc dot org
Hebbring, Scott	Marshfield Clinic	hebbring.scott at marshfieldresearch dot org
Heer, Avneet	Veranex	avneet.heer at veranexsolutions dot com
Helseth, Larry	NorthShore University HealthSystem	lhelseth at northshore dot org
Hershberger, Ray	The Ohio State University	Ray.Hershberger at osumc dot edu
Hewavitharana, Hasani	Ministry of health	hewavitharanahasani at gmail dot com
Hirbo, Jibril	Vanderbilt University Medical Center	jibril.hirbo at vumc dot org
Hoffman, James	St. Jude Children's Research Hospital	james.hoffman at stjude dot org
Hoffman, Mark	Childrens Mercy Hospital	mark-hoffman at earthlink dot net
Horowitz, Carol	Mount Sinai	carol.horowitz at mountsinai dot org
Hulick, Peter	NorthShore University HealthSystem	PHulick at northshore dot org
Hull, Leland	Massachusetts General Hospital	lhull1 at mgh.harvard dot edu
Hurtado, Alejandra	Georgetown University	ahd28 at georgetown dot edu
Ifhar, Lee	Invitae	lee.ifhar at invitae dot com
Isaacs, Kayce	Illumina	kisaacs at illumina dot com
James, Cynthia	Johns Hopkins University	cjames7 at jhmi dot edu
Jarvik, Gail	University of Washington	gjarvik at medicine.washington dot edu
Jeff, Janina	Illumina	jmjeff at illumina dot com
Jones, Danielle	Personal Genome Diagnostics/Labcorp	djones at pgdx dot com
Joseph, Nancy	UCSF	Nancy.Joseph at ucsf dot edu
Jung, Jongyun	Ohio State University	jung.731 at osu dot edu
Karlson, Elizabeth	Brigham & Women's Hospital	ekarlson at bwh.harvard dot edu
Kaufman, Dave	NIH/NHGRI	dave.kaufman at nih dot gov
Kayalvily, Perinpanayagam	LRH	kayalvilyperinpanayagam at gmail dot com

Name	Institute	Email Address
Kenny, Eimear	Icahn School of Medicine at Mount Sinai	eimear.kenny at mssm dot edu
Khatipov, Emir	NHLBI/NIH	khatipov at nih dot gov
Khoury, Muin	CDC	muk1 at cdc dot gov
Kim, Jong-Il	Seoul National University	jongil at snu.ac dot kr
King, Tracy	NIH	tracy.king at nih dot gov
Klein, Teri	Stanford University	teri.klein at stanford dot edu
Kolambage, Yasas	Sabaragamuwa University Of Sri Lanka	yasas at med.sab.ac.lk
Kolawole, Olubayo	University of British Columbia	Olubayo.Kolawolemd at gmail dot com
Korf, Bruce	UAB	bkorf at uabmc dot edu
Koromina, Maria	Icahn School of Medicine, Mount Sinai	maria.koromina at mssm dot edu
Kulchak Rahm, Alanna	Geisinger	akrahm at geisinger dot edu
Kullo, Iftikhar	Mayo Clinic	kullo.iftikhar at mayo dot edu
Landry, Latrice	University of Pennsylvania	Latrice.Landry at Pennmedicine.UPenn dot edu
Landrum, Melissa	NIH	landrum at ncbi.nlm.nih dot gov
Laplaza, Carmen	European Commission	carmen.laplaza-santos at ec.europa dot eu
Larkin, Jennie	National Institute on Aging	jennie.larkin at nih dot gov
LeBlanc, Kimberly	Harvard Medical School	kimberly_leblanc at hms.harvard dot edu
Lee, Charles	The Jackson Laboratory for Genomic Medicine	charles.lee at jax dot org
Lemke, Amy	University of Louisville	lemkeaa at gmail dot com
Li, Rongling	NIH	lir2 at nih dot gov
Liao-Upshaw, Judy Huey	University of California, San Francisco	judy_act at yahoo dot com
Limdi, Nita	University of Alabama at Birmingham	nlimdi at uabmc dot edu
Lin, Asiyah	NHGRI	asiyah.lin at nih dot gov
Lindquist, Karla	UCSF	karla.lindquist at ucsf dot edu
Liu, Paul	NIH	pliu at nih dot gov
Lockwood, Mark	University of Illinois Chicago	lockmar at uic dot edu
Lozano, Reymundo	Mount Sinai	Reymundo.lozano at mssm dot edu
Lumsden, Joanne	National Center for Advancing Translational Sciences	joanne.lumsden at nih dot gov
MacDonald, Deborah	DJM Genetics/Genomics Consulting	Djmgenetics at gmail dot com
Madden, Ebony	NHGRI	ebony.madden at nih dot gov

Name	Institute	Email Address
Mahasirimongkol, Surakameth	Medical Life Science Institute, Ministry of Public Health,Thailands	Surakameth.m at dmsc.mail.go dot th
Makwana, Simran	Harvard Medical School	simran_makwana at hms.harvard dot edu
Maloney, Kristin	University of Maryland School of Medicine	kmaloney1 at som.umaryland dot edu
Mamidi, Tarun Karthik Kumar	The University of Alabama at Birmingham	tmamidi at uab dot edu
Manolio, Teri	NIH	manoliot at mail.nih dot gov
Martin, Iman	NIH	iman.martin at nih dot gov
Mathew, Sunitha	Westat	sunithamathew at westat dot com
Mathias, Rasika	Johns Hopkins University	rmathias at jhmi dot edu
Mazur, Zehra	Cornell University	zehradincer at gmail dot com
McCormick, Kathleen	SciMind, LLC	scimind2 at gmail dot com
McGraw, Deven	Invitae	deven.mcgraw at invitae dot com
McLeod, Howard	Intermountain Healthcare	howard.mcleod at imail dot org
Mendelsohn, Nancy	Optum Frontiers Therapy (OFT)	nmendelsohn at uhg dot com
Messersmith, Donna	NHGRI	donna.messersmith at nih dot gov
Might, Matt	UAB	might at uab dot edu
Mikail, Claudia	UCLA	doctormikail at outlook dot com
Milko, Laura	University of North Carolina at Chapel Hill	laura_milko at med.unc dot edu
Miller, Alexandra	Mayo Clinic	miller.alexandra at mayo dot edu
Miller, Nicholas	NorthShore University Healthcare System	nmiller at northshore dot org
Mills, Rachel	UNC Greensboro	rachelmillscgc at gmail dot com
Mitropoulou, Christina	The Golden Helix Foundation	c.mitropoulou at goldenhelix dot org
Moore, Jade	University of California, San Francisco	jade.moore at ucsf dot edu
Moretz, Chad	Invitae	chad.moretz at invitae dot com
Morris, Laine	PGDx	lmorris at pgdx dot com
Mueller, Rebecca	UPenn	rebecca.mueller at pennmedicine.upenn dot edu
Mukhopadhyay, Sanghamitra	Defense Health Agency	sanghamitra.mukhopadhyay.civ at mail dot mil
Muralidhar, Sumitra	Department of Veterans Affairs	sumitra.muralidhar at va dot gov
Murphy-Stone, Jeanne	US Government Accountability Office	murphystonej at gao dot gov
Murray, Mike	Yale University	michael.murray at yale dot edu

Name	Institute	Email Address
Nair, Sai	UCSF	saidas.nair at ucsf dot edu
Narula, Jahnavi	NHGRI	jahnavi.narula at nih dot gov
Neta, Gila	NCI	Gila dot neta at nih dot gov
Ngadiman, Sutini	Abbvie	sgadiman8 at gmail dot com
Nishiguchi, Reiko	Invitae	reiko.nishiguchi at invitae dot com
Nussbaum, Robert	Invitae	robert.nussbaum at invitae dot com
O'Shea, Rosie	St James Hospital	rososhea at stjames.ie
O'Brien, Madison	CCHMC	madison.obrien at cchmc dot org
ODonnell, Peter H.	The University of Chicago	podonnel at medicine.bsd.uchicago dot edu
O'Hanlon, Tim	Johns Hopkins University Applied Physics Laboratory	tim.ohanlon at jhuapl dot edu
Oladayo, Abimbola	University of Iowa	abimbola-oladayo at uiowa dot edu
O'Neill, Suzanne	Georgetown University	sco4 at georgetown dot edu
Orlando, Lori	Duke University	lorlando at duke dot edu
Overby Taylor, Casey	Johns Hopkins University	cot at jhu dot edu
Osterman, Travis	Vanderbilt University Medical Center	travis.osterman at vumc dot org
Owen, Rachel	Parkview Research Center	rachel.owen at parkview dot com
Owens, Kellie	New York University	owenske3 at gmail dot com
Owusu Obeng, Aniwaa	Icahn School of Medicine at Mount Sinai	aniwaa.owusu-obeng at mssm dot edu
Paltoo, Dina	National Institutes of Health	dina.paltoo at nih dot gov
Paranjape, Ketan	Roche	ketan.paranjape at roche dot com
Park, Nguyen	NIH	nguyen.park at nih dot gov
Parviainen, Anndra Margareth	University of Eastern Finland	anndra.parviainen at uef.fi
Patrinos, George P.	University of Patras	gpatrinos at upatras.gr
Pawlyk, Aaron	NICHD	pawlykac at mail.nih dot gov
Perkins, Adrienne	GRAIL	aperkins at grailbio dot com
Peterson, Josh	Vanderbilt University Medical Center	josh.peterson at vumc dot org
Pinard, Amelie	University of California San Francisco	amelie.pinard at ucsf dot edu
Plaideau, Catheline	Veranex	catheline.plaideau at veranexsolutions dot com
Pollard, Kai	Personal Genome Diagnostics	kpollard at pgdx dot com
Pollin, Toni	University of Maryland School of Medicine	tpollin at som.umaryland dot edu
Powell, Cynthia	University of North Carolina School of Medicine	powellcm at med.unc dot edu
Powell, Emily	Parkview Health	emily.powell at parkview dot com

Name	Institute	Email Address
Pozzatti, Rudy	National Human Genome Research Institute	pozzattr at exchange.nih dot gov
Pritchard, Daryl	Pesonalized Medicine Coalition (PMC)	dpritchard at personalizedmedicinecoalition dot org
Priyawansha, Thushara	Sirimavo Bandaranayake Specialised Children's Hospital	thusharapriyawansha at gmail dot com
Procknow, Catherine	Epic	cprockno at epic dot com
Prows, Cindy	Cincinnati Children's Hosp.	cindy.prows at cchmc dot org
Przygodzki, Ronald	US Dept. Veterans Affairs	ronald.przygodzki at va dot gov
Qu, Tiange	University of California, San Francisco	tony.qu at ucsf dot edu
Quinteros, Badi	Brigham Young University	badiisrael1098 at gmail dot com
Rader, Daniel	University of Pennsylvania	rader at pennmedicine.upenn dot edu
Rajkovic, Aleksandar	University of California, San Francisco	aleks.rajkovic at ucsf dot edu
Rakhra-Burris, Tejinder	Duke University	teji.rb at duke dot edu
Ramirez, Alfredo		jalfredormz88 at gmail dot com
Ramos, Erin	NIH	ramoser at mail.nih dot gov
Ramsey, Laura	Cincinnati Children's Hospital Medical Center	laura.ramsey at cchmc dot org
Rasmussen, Luke	Northwestern University	luke.rasmussen at northwestern dot edu
Rasmussen-Torvik, Laura	Northwestern	ljrtorvik at northwestern dot edu
Rehm, Heidi	Massachusetts General Hospital	HREHM at mgh.harvard dot edu
Relling, Mary	St. Jude Children's Research Hospital	mary.relling at stjude dot org
Ren, Zhaoxia	NICHD/NIH	zren at mail.nih dot gov
Rider, Renee	NIH/NHGRI	renee.rider at nih dot gov
Riggs, Erin	Geisinger	eriggs at geisinger dot edu
Risch, Neil	University of California, San Francisco	neil.risch at ucsf dot edu
Ritchie, Marylyn	University of Pennsylvania	marylyn at pennmedicine.upenn dot edu
Rockowitz, Shira	Boston Children's Hospital	shira.rockowitz at childrens.harvard dot edu
Rogers, Sara	Texas A&M University	sararogers at tamu dot edu
Ronquillo, Jay	National Cancer Institute	jay.ronquillo at nih dot gov
Rowley, Robb	NIH/NHGRI	robb.rowley at nih dot gov
Sadeghpour, Azita	Duke Health, DukeUniversity	azita.sadeghpour at duke dot edu
Safi, Ismail	NHGRI	ismail.safi at nih dot gov

Name	Institute	Email Address
Samedy Bates, Lesly	NIH/NICHD	lesly-anne.samedy-bates at nih dot gov
Samer, Ella	NHGRI	ella.samer at nih dot gov
Satproedprai, Nusara	Eastern Economic Corridor Office	nusara.sat at gmail dot com
Savage, Michelle	Johns Hopkins University	msavage at jhu dot edu
Schatz, Michael	Johns Hopkins University	mschatz at cs.jhu dot edu
Schmidlen, Tara	Invitae	tara.schmidlen at invitae dot com
Schully, Sheri	NIH	schullys at mail.nih dot gov
Sebastian, Jessica	UPMC Children's Hospital	Jessica.Hartman at chp dot edu
Senthil Kumar, Nachimuthu	Mizoram University	nskmzu at gmail dot com
Shelburne, Nonniekaye	NCI	nshelburne at nih dot gov
Sheng, Rory	UCSF	rory.sheng at ucsf dot edu
Sholes, Samantha	Johns Hopkins University Applied Physics Laboratory	Samantha.Sholes at jhuapl dot edu
Simmons, Tyler	Personal Genome Diagnostics	tysimmons at pgdx dot com
Smania, Mary	Michigan State University	smaniama at msu dot edu
Snyder, Holly	Illumina	hsnyder at illumina dot com
Somanadhan, Suja	University College Dublin	suja.somanadhan at ucd.ie
Spellman, Paul	Oregon Health and Science University	spellmap at ohsu dot edu
Ssebunnya, Gerald Michael	Africa Institute for Human Dignity	gmssebu at gmail dot com
Starita, Lea	University of Washington	lstarita at uw dot edu
Strande, Natasha	Geisinger	ntstrande at geisinger dot edu
Subasinghe, Vindya	Lady Ridgeway Hospital for children	vindimdm at yahoo dot com
Sung, Choa	University of Illinois Chicago	csung7 at uic dot edu
Taylor, Brittany	Columbia University	bt2542 at cumc.columbia dot edu
Terry, Sharon	Genetic Alliance	sterry at geneticalliance dot org
Tezak, Zivana	FDA	zivana.tezak at fda.hhs dot gov
Tiwari, Sangeeta	University of Texas, El Paso	stiwari at utep dot edu
Torkamani, Ali	Scripps Research	atorkama at scripps dot edu
Tsosie, Krystal	Arizona State University	Krystal.Tsosie at asu dot edu
Urv, Tiina	NCATS	tku33biz at gmail dot com
Vanzanten, Maya	NIH	maya.vanzanten at nih dot gov
Vassy, Jason	Harvard Medical School	jvassy at partners dot org
Verner, Ellen	PGDx	everner at pgdx dot com
Vinson, Cynthia	National Cancer Institute	cvinson at mail.nih dot gov
Vitek, Carolyn	Mayo Clinic	vitek.carolyn at mayo dot edu
Walker, Trina	Creighton University	trinawalker at creighton dot edu
Walton, Nephi	Intermountain Healthcare	nephi.walton at imail dot org

Name	Institute	Email Address
Wand, Hannah	Stanford University	hwand at stanfordhealthcare dot org
Wang, Catharine	Boston University	clwang at bu dot edu
Wang, Lu	DER, NIDCR, NIH	lu.wang at nih dot gov
Wang, Sarah	MGH	wsarah0401 at gmail dot com
Watson, Karriem	NIH	karriem.watson at nih dot gov
Weber, James	PreventionGenetics	jim.weber at preventiongenetics dot com
Wei, Chih-fu	HSPH	chihfuwei at g.harvard dot edu
Weng, Chunhua	Columbia university	chunhua at columbia dot edu
Werth, Thomas	NorthShore University Health System	twerth at northshore dot org
Westerman, Kenny	Massachusetts General Hospital	kewesterman at mgh.harvard dot edu
Wetterstrand, Kris	National Human Genome Research Institute | NIH	wettersk at mail.nih dot gov
Wetzel, Alyssa	University of Iowa	alyssa-wetzel at uiowa dot edu
Whitehead, Christina	Public Health Service	Christina.Whitehead at health.nsw dot gov dot au
Whitt, Karen	George Washington University	kjwhitt at gwu dot edu
Wiisanen, Kristin	University of Florida	kwiisanen at cop.ufl dot edu
Wildin, Robert	University of Vermont and UVM Health Network	robert.wildin at uvmhealth dot org
Wiley, Ken	NHGRI	ken.wiley at nih dot gov
Williams, Kelly	ConcertAI	kwilliams at concertai dot com
Williams, Marc	Geisinger	mswilliams1 at geisinger dot edu
Willis, Valerie	NIH	valerie.willis at nih dot gov
Witonsky, Jonathan	University of California, San Francisco	jonathan.witonsky at ucsf dot edu
Wu, Ryanne	Duke University	ryanne.wu at duke dot edu
Xirasagar, Sandhya	NHGRI	xirasasa at nih dot gov
Yang, Mathew	NorthShore University HealthSystem	myang at northshore dot org
Yee, Sook Wah	University of California, San Francisco	sookwah.yee at ucsf dot edu
Zhao, Jingjing	University of California, San Francisco	jjzhao123 at gmail dot com

Genomics-Enabled Learning Health Care Systems: Important Points Highlighted by Individual Speakers

For the complete summary of the 2015 Workshop, please visit: https://nap.nationalacademies.org/catalog/21707/genomics-enabled-learning-health-care-systems-gathering-and-using-genomic

The speakers referenced follow:

Aronson, Sandy
Baker, Dixie
Chute, Chris
Etheredge, Lynn
Fowler, Tom
Friedman, Chuck
Ginsburg, Geoffrey
Hill, Colin
Leffler, Stephen
Moss, Scott
Nolen, John David
Ommaya, Alexander
Peterson, Josh
Risch, Neil
Terry, Sharon
Vassy, Jason

The following are the important points highlighted by the individual speakers. These synopsizes are pulled from the workshop summary and are direct quotations. Full credit is given to Sarah H. Beachy, Steve Olson, and Adam C. Berger, the rapporteurs for the workshop.

Ch 1. INTRODUCTION AND THEMES OF THE WORKSHOP

Box 1-1, Objectives of the Workshop

- To explore how key pieces of genetic/genomic information can be effectively and efficiently delivered to patients and clinicians for improving care.
- To discuss how both the health care system and genomic data can be used for evidence generation in research and in patient care.
- To assess current best practices for using knowledge-generating/learning health care systems and which models may provide an opportunity for genomics to be used in the rapid-learning process.

Ch 2. ADVANCING PATIENT CARE AND RESEARCH WITH GENOMIC INFORMATION

- Integrating high-quality data into the health care system is a priority for ensuring that the best possible information is available for patient care and research. (Peterson, Risch)
- While a variety of genomic data sources exist, they are not readily accessible for use in the current electronic health record (EHR), and work to address the format of these data would create opportunities to use the information more effectively. (Risch)

- Medical information in the EHR coupled with gene sequencing information can be used as a discovery tool for identifying genetic variants associated with disease and for understanding individual response to therapeutics. (Peterson)
- Several ongoing efforts within government and the private sector are aimed at establishing data repositories for large-scale genomic information. These data could be used to demonstrate the power of rapid learning for improving patient care and informing health research. (Etheredge, Peterson)
- Data that are standardized, comparable, and consistent would facilitate the reuse of those data for discovery in multiple contexts beyond the original one. (Chute)

Ch 3. TRANSLATION OF GENOMICS FOR PATIENT CARE AND RESEARCH

- Patients are very involved in their own health care and are producing their own health-related data, including genomic data. Understanding preferences for data use and communicating effectively in a fair and transparent way with the public about how information is used will be key to engaging the larger population in sharing their data for research. (Baker)
- Patient trust can be earned and maintained through good data practices, including establishing confidentiality policies, data encryption, and multifactor authentication. (Chute)
- Deriving clear standard consent language could reduce the burden on institutions, which today largely develop their own consenting mechanisms, and could provide transparent information for patients about the use of their data. (Baker, Chute, Fowler, Moss)
- A health care system in which an infrastructure supports complete learning cycles that encompass both the analysis of data to produce results and the use of those results to develop changes in clinical practices is a system that will allow for optimal learning. (Friedman)
- Using genomic data could improve population health and contribute to solving many care management problems. Starting with areas that can lead to a return-on-investment may encourage leaders of health systems to engage in these efforts. (Hill)
- Just-in-time information, guidelines for clinical action, and more information on the clinical utility of genetic testing would help physicians make effective use of genomic information and integrate it in their practices similarly to other medical test information. (Vassy)

Ch. 4 GENOMICS AND THE EHR IN A LEARNING HEALTH CARE SYSTEM

- Both academic health centers and community centers are working to incorporate genomic information into their systems, but the efforts are largely separate. Establishing data standards and common ways of representing outcomes would facilitate the scalability of efforts and the translation of genomic information into clinical care. (Moss)
- The most practical way of integrating genomic data into the clinic is to provide it through clinical decision support, but that means the community would need to agree upon common allele and test code nomenclature so that the guidance is scalable and interoperable. (Chute)
- Cultivating a “data donor” culture in which data sharing is commonplace and encouraged because it would help the greater population could be achieved by ensuring the privacy of personal information. (Chute)

Ch 5. REPRESENTING GENOMIC INFORMATION IN THE EHR ECOSYSTEM

- Transmission of genomic data within the health information ecosystem could be improved by defining data standards that would also foster interoperability and allow for scalability. (Aronson, Nolen)
- The DIGITizE Action Collaborative was established in 2014 to engage key stakeholders for developing a framework for data genomic standards so that this information is more easily integrated into EHR platforms for clinical use. (Aronson, Nolen)
- Setting data standards for genomic information can enable EHR systems—independent of where they are used—and users of these systems to easily understand the data across the medical community. (Nolen)
- Four use cases focused on pharmacogenomics will be the initial starting point for implementing the standards framework designed by the action collaborative. The goal of the effort is to pilot a project and demonstrate how genomic information can effectively flow through a health information technology system. (Aronson)

Ch 6. POSSIBLE NEXT STEPS

Box 6-1, Possible Next Steps Proposed by Individual Workshop Participants

Interoperability of EHRs

- Ensure that the quality of genomic data is clinical grade and that it is in an accessible format so that it can be used for future research and to inform clinical care. (Risch)
- Support regulations that will make EHRs fully interoperable for genomic information. (Leffler)
- Establish data standards for genomics to allow for EHRs to communicate and for genomic data to flow more easily across labs and systems to providers. (Aronson, Fowler, Nolen)
- To demonstrate how the interoperability of systems can be increased, start with specific health problems whose outcomes are likely to be changed with genomic and other clinical data. (Hill)

Clinical Decision Support

- Reach agreement on allele and test code nomenclature to facilitate the development of clinical decision support tools for genomics. (Chute)
- Create warehouses of clinical decision support tools that can be shared and used widely. (Ginsburg)
- Measure outcomes to determine the validity of algorithms used to guide practice. (Moss)
- Develop a core infrastructure to handle clinical decision support and the long-term storage of complex data. (Nolen)

Data Sharing

- Build platforms with reusable components that are scalable and can be implemented anywhere. (Friedman)
- Standardize data so that they can be re-used. (Chute)
- Foster interoperable health care systems to enable genomic data sharing. (Terry)
- Inform the public about data sharing to cultivate a “data donor” culture. (Chute)

- Network data from around the world to increase the size of databases and power of research studies. (Aronson)
- Integrate patient-provided data into health care information technology systems. (Baker)
- Examine whether personally controlled health databanks can make data accessible for sharing while protecting privacy. (Friedman)
- Support research to understand and generate personalized user interfaces and preferences. (Baker)

Implementation

- Engage groups with a particular interest and who value genomics, such as people with undiagnosed or chronic diseases, to demonstrate the full potential of this information. (Terry)
- Measure and track health and health care disparities to determine the impact of genomics-based interventions. (Ommaya)
- Support social science and behavioral research to understand the priorities and values of patients and providers when genomics is introduced in the clinic. (Ginsburg)

By Marc S. Williams, Adam H. Buchanan, F. Daniel Davis, W. Andrew Faucett, Miranda L. G. Hallquist, Joseph B. Leader, Christa L. Martin, Cara Z. McCormick, Michelle N. Meyer, Michael F. Murray, Alanna K. Rahm, Marci L. B. Schwartz, Amy C. Sturm, Jennifer K. Wagner, Janet L. Williams, Huntington F. Willard, and David H. Ledbetter

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Patient-Centered Precision Health In A Learning Health Care System: Geisinger's Genomic Medicine Experience

ABSTRACT Health care delivery is increasingly influenced by the emerging concepts of precision health and the learning health care system. Although not synonymous with precision health, genomics is a key enabler of individualized care. Delivering patient-centered, genomics-informed care based on individual-level data in the current national landscape of health care delivery is a daunting challenge. Problems to overcome include data generation, analysis, storage, and transfer; knowledge management and representation for patients and providers at the point of care; process management; and outcomes definition, collection, and analysis. Development, testing, and implementation of a genomics-informed program requires multidisciplinary collaboration and building the concepts of precision health into a multilevel implementation framework. Using the principles of a learning health care system provides a promising solution. This article describes the implementation of population-based genomic medicine in an integrated learning health care system—a working example of a precision health program.

Marc S. Williams (mswilliams1@geisinger.edu) is director of the Genomic Medicine Institute, Geisinger, in Danville, Pennsylvania.

Adam H. Buchanan is an assistant professor at the Genomic Medicine Institute, Geisinger.

F. Daniel Davis is director of the Center for Bioethics and Healthcare Policy, Geisinger.

W. Andrew Faucett is a professor at the Genomic Medicine Institute, Geisinger.

Miranda L. G. Hallquist is a genetic counselor at the Genomic Medicine Institute, Geisinger.

Joseph B. Leader is director of the Phenomic Analytics and Clinical Data Core, Geisinger.

Christa L. Martin is director of the Autism and Developmental Medicine Institute, Geisinger.

Cara Z. McCormick is a senior assistant at the Genomic Medicine Institute, Geisinger.

Michelle N. Meyer is associate director for research ethics at the Center for Translational Bioethics and Health Care Policy, Geisinger.

Michael F. Murray was a physician in the Genomic Medicine Institute, Geisinger, at the time this work was completed. He is now at the Yale School of Medicine.

Precision medicine is evolving from a concept to clinical viability, albeit in limited settings. In his 2015 State of the Union address, President Barack Obama called for a federally funded, large-scale precision medicine initiative, heightening interest in this idea.[1,2]

Medicine as currently practiced is empirical, inadequately grounded in evidence, and dependent on the knowledge and experience of individual providers, which results in variable care with suboptimal outcomes. Clay Christensen and coauthors define *precision medicine* as "the provision of care for diseases that can be precisely diagnosed, whose causes are understood, and which consequently can be treated with rules-based therapies that are predictably effective."[3]

Although some conflate genomic medicine with precision medicine or, as we prefer, precision health (as it encompasses both wellness and disease), genomic data must be combined with data from other sources (for example, clinical, environmental, and social) to inform precision care. The formulation that best captures what is needed to attain precision health is attributed to Stephen Pauker and Jerome Kassirer: clinical decision-making such that the decisions made maximize the outcomes that the patient most cares about and minimizes those that the patient fears the most, on the basis of as much knowledge about the individual's state as is available.[4] This definition captures three key points: a focus on outcomes, the central role of patients in defining outcomes (positive or negative), and

Alanna K. Rahm is an assistant professor at the Genomic Medicine Institute, Geisinger.

Marci L. B. Schwartz is a genetic counselor at the Genomic Medicine Institute, Geisinger.

Amy C. Sturm is a professor at the Genomic Medicine Institute, Geisinger.

Jennifer K. Wagner is associate director of bioethics research, Center for Translational Bioethics and Health Care Policy, Geisinger.

Janet L. Williams is director of research genetic counselors, Genomic Medicine Institute, Geisinger.

Huntington F. Willard is director of the National Precision Health Institute, Geisinger.

David H. Ledbetter is executive vice president and chief scientific officer, Geisinger.

"knowledge about the individual's state" (which implicitly includes "genetic" and "genomic" information).

Health care systems as traditionally configured are not designed or equipped to deliver precision health to patients. In 2010 the Institute of Medicine (IOM) published *Value in Health Care*, the first of nineteen reports to date on learning health care systems. In its introduction to the series, the institute defined these systems as those in which "science, informatics, incentives, and culture are aligned for continuous improvement and innovation, with best practices seamlessly embedded in the delivery process and new knowledge captured as an integral by-product of the delivery experience."[5]

Prior IOM reports had not addressed genomic or precision medicine, so in 2015 the IOM published a workshop summary describing genomics-enabled learning health care systems[6] and emphasized the need for learning cycles that analyze data and use the analytic results to change clinical practice. Genomics-informed precision health is emerging in clinical practice, mostly in the setting of clinically relevant and informed research (exhibit 1).

This article focuses on high-level issues of relevance to any organization contemplating a precision health program. As a case study, it describes the initial phases of implementation of a large-scale population-based precision health initiative within the setting of a learning health care system.

The Setting

Geisinger is an integrated health care delivery system in densely rural (as designated by Medicare) central Pennsylvania and in southern New Jersey. Geisinger serves approximately 4.2 million residents, with about 1.5 million unique patient visits annually. About one-third of Geisinger patients are insured by the provider-owned Geisinger Health Plan. This creates a "sweet spot" that enables Geisinger to pilot innovations in care delivery.[7(p xix)]

Clinical Care Reengineering And Quality Improvement Geisinger has over ten years of experience in creating evidence-based care pathways to reduce unexplained clinical variation, resulting in high-quality care at a lower cost and optimizing value to the patient, health system, and payer.[7,8] The pathways areimplemented with the support of the electronic health record (EHR) system and associated data sources, coupled with processes to track outcomes. Patient engagement is an essential component of precision health and learning health care systems and must be included in the reengineering process to a greater degree than has occurred previously. This approach demonstrates that linking several improvement concepts (for example, evidence-based guidelines, data feedback, reliability science, and patient-centered care) in a single design model can reduce unwarranted variation in care delivery to reduce cost, optimize outcomes from the patient's perspective, and provide the foundation for continual improvement. Geisinger has facilitated the generalizability of locally developed standardized care pathways by converting them to condition-specific care protocols coupled with consultative services, a process termed ProvenCare.[7,8]

Building A Learning Health Care System Geisinger has committed to becoming a learning health care system, a goal facilitated by its organization as an integrated system in which all employees and units—including researchers, providers, and a payer—are part of the overall success of the enterprise. A multidisciplinary

Exhibit 1

Selected US programs that are implementing the use of genomic information in the health care setting

Program	Genomic information returned	Approximate number of patients
eMERGE phase 3[a]	Pathogenic and likely pathogenic germline variants in actionable genes, pharmacogenomics	25,000
IGNITE	Family history, pharmacogenomics, selected pathogenic germline variants, polygenic risk scores	—[b]
St. Jude Children's Research Hospital	Pharmacogenomics	All patients admitted for treatment (about 7,500 annually)
Inova Health System Translational Medicine Institute	Pharmacogenomics, pathogenic variants related to selected clinical indications	5,000
Geisinger MyCode® Community Health Initiative	Pathogenic and likely pathogenic germline variants in actionable genes, pharmacogenomics	92,000 to date

SOURCE Authors' analysis. NOTE IGNITE is Implementing Genomics in Practice. [a]Geisinger is a member of the Electronic Medical Records and Genomics (eMERGE) network. [b]Not publicly available.

working group consisting of representatives of key organizational functions, including research, clinical innovation, and bioethics, meets regularly to identify current assets and gaps that need to be filled to attain this goal.

Four phases have been defined to foster the development of a learning health care system. These are in the process of being implemented. Phase 1 involves developing criteria for identifying, evaluating, and tracking "local learning health care initiatives"—existing Geisinger practice areas that have already adopted at least some aspects of the learning health care system model. Phase 2 consists of identifying instances in which an initiative was successfully expanded into adjacent practice areas, identifying factors that enabled that spread, and leveraging those factors by deliberately linking initiatives to one another to enhance collaboration and replication. Phase 3 involves establishing an enabling core of providers who are empowered and incentivized to lead learning, experimentation, and innovation efforts and provide a model for others to follow. Phase 4 consists of developing conceptual and business models that, drawing on lessons learned in phases 1–3, will inform efforts to further advance and oversee a system-wide learning health care system culture.

Enabling Factors For Implementing Precision Health

Research—as part of the innovation cycle integral to learning health caresystems—has been an essential part of Geisinger's mission since its beginning. The theme of the Geisinger research strategic plan is personalized health care research, with an emphasis on developing and testing innovative approaches that will enable the identification of patients' unique influences (environmental, clinical, social, and genetic) so that each patient receives the right care at the right time in the right way, to optimize quality and achieve the outcomes of importance to that patient.

With this goal in mind, senior leadership began to discuss the concept of a genomics core in the early 2000s[9] and led to the launch of the MyCode® biorepository in 2007.[10] From its inception, the biorepository used opt-in consent, allowing participants to contribute biospecimens linked to their EHR data that were initially used for discovery research. The potential of the MyCode biorepository as a first step in a precision health project was recognized in the 2010 revision of the research strategic plan.

Recognizing that research results from the MyCode initiative were of translational and clinical value, Geisinger established several institutes designed to span and integrate research and clinical care using the learning health care system model. They included the Obesity Institute, the Genomic Medicine Institute, and the Autism and Developmental Medicine Institute. To enable this integrative mission, each Geisinger institute is actively engaged with clinical care departments, clinical innovations, informatics, and the broader research enterprise.

As of January 2018, over 180,000 Geisinger patients had consented to participate in what is now called the MyCode Community Health Initiative.[10,11] Of the patients approached, 85–90 percent consent to participate. Ongoing analysis of the reasons patients decline participation has not identified any predictive factors. MyCode participants are slightly older and more likely to be female, have a higher body mass index, and are less diverse in terms of race/ethnicity, compared to Geisinger patients on average.[10] Participants have a median of fourteen years of EHR data.

In 2014 the MyCode initiative began to conduct whole exome sequencing and genotyping on collected samples, as part of a collaboration with Regeneron Pharmaceuticals and the Regeneron Genetics Center.[12] Whole exome sequencing analyzes genes that code for proteins and associated gene regulatory areas—about 1–2 percent of the whole genome containing the most clinically relevant information. To date, nearly 93,000 exome sequences have been completed.[11] Although these data are intended to support discovery research, Geisinger has unrestricted use of the data for clinical care. MyCode participants are now enrolled under a broad, opt-in consent that supports health-related research and allows for the recontact of participants and reporting of results that are deemed clinically relevant, with placement of results in the EHR. This provides an opportunity to benefit participants, something that was valued by Geisinger patients in the extensive community consultation used to design the program and continuously improve it.[13]

Oversight is provided by Geisinger's Institutional Review Board and the MyCode Governing Board, with input from other stakeholders that include participant, youth, and clinician advisory boards; a genomic council consisting of all Geisinger genetic providers (medical and laboratory geneticists, and genetic counselors) and faculty members; and external ethics and scientific advisory boards. This ongoing commitment to involving the broad community both within and outside Geisinger is key to maintaining trust, and it provides opportunities to adapt the initiative to the changing needs of the community.[14] The partnership with patients, participants, and other stakeholders represented in

the advisory boards facilitates the alignment of science, incentives, and culture—keys to realizing a learning health care system—and reduces the risk of failure due to poor communication. An approach involving input from diverse stakeholders, informed by the patient's perspective, is essential for any organization seeking to implement precision health, as adjusting specific processes to the local environment is needed to maximize the likelihood of success.

Any new initiative of this magnitude and breadth requires significant resources. The costs of the MyCode initiative have been met through a combination of institutional investments and funds from Geisinger's partnership with Regeneron Pharmaceuticals, philanthropy, grants, and other sources. The MyCode program was designed to inform the implementation of genomics in clinical care at the scientific and process levels, as outlined in this article.While the initial stages of the program were not designed to enable cost-benefit analyses, this is an important focus of ongoing work.

Initial Implementation Of A Genomic Medicine And Precision Health Learning Health Care System

Implementing the principles of a learning health care system in a precision health program with an early focus on genomic medicine required multidisciplinary expertise coupled with a communication strategy that crosses traditional institutional boundaries to capture and integrate data from Geisinger and elsewhere.

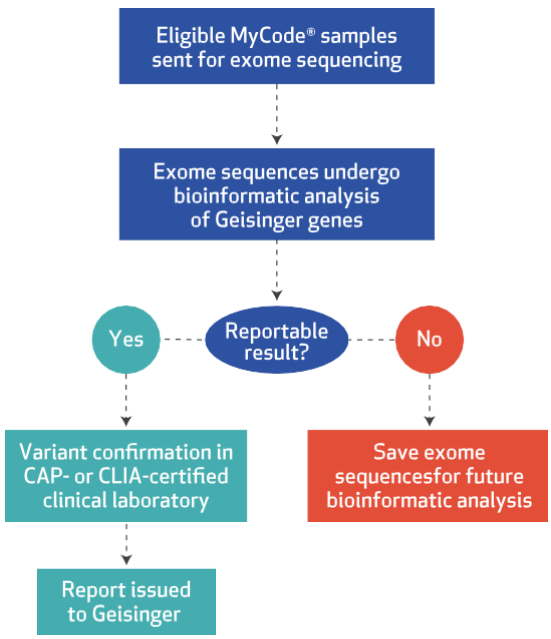
At the foundation of a learning health care system is an information system that uses data derived from the EHR but also captures critical data outside the EHR system. This includes collecting data from outside Geisinger, as the DNA variants identified by research exome sequencing must be confirmed in a clinical laboratory before being used for patient care. Data for the MyCode program are stored on local servers and in a cloud service that is compliant with the Health Insurance Portability and Accountability Act (HIPAA) of 1996 and the Federal Information Security Management Act of 2014. Business associate agreements are in place. All data stored on Geisinger servers are behind the system firewall and subject to Geisinger's security requirements. As the IOM has pointed out,[6] communication of genomic data among different systems has not been standardized. This has led Geisinger to create customized workflows to ensure that data are available for care and tracking. Details of the solutions are beyond the scope of this article, but it must be emphasized that the processes discussed below are dependent on a robust institutional informatics infrastructure.[15] While not all organizations have such an infrastructure, the increase in use of fully functional EHR systems coupled with international efforts to develop and implement standards to support the use of genomic data in the clinic should, in time, reduce reliance on local solutions to store and communicate genomic information and improve generalizability across health care information systems.

We next describe the initial overall workflow of the genomics and precision health program from research and innovation to clinical care, as presented in three phases: whole exome sequencing, data analysis, and variant confirmation; initial clinical care and support for results reporting; and transition to ongoing clinical care.

Sequencing, Data Analysis, And Confirmation Exhibit 2 depicts the process of transforming the research exome sequence for use in clinical care. The key component is the bioinformatic analysis of the DNA sequence to identify high-confidence, likely or known pathogenic variants that can be reported to participants and their providers and recorded in the EHR. MyCode participants are enrolled irrespective of any disease or condition, and interpretation of results must consider the low probability of a

Exhibit 2

Geisinger process for exome sequencing, data analysis, and variant confirmation



SOURCE Geisinger. NOTES "Geisinger genes" are explained in the text. CAP is College of American Pathologists. CLIA is Clinical Laboratory Improvement Amendments.

Assessment of the precision health program to identify and lower barriers to dissemination beyond Geisinger is ongoing.

person's having a condition associated with variants identified by the genomic analysis. Geisinger therefore uses conservative variant-calling protocols to minimize the return of false-positive results. For example, a variant in *BRCA1* (associated with hereditary breast or ovarian cancer syndrome) that has a high certainty of being disease causing (such as a three-star designation in the expert-curated Clinical Variant resource)[16] would be reported for clinical attention, whereas a novel variant would not be reported, as the clinical interpretation of such a variant has not been established by evidence-based best practices for variant annotation. When compared to diagnostic testing, interpretation of variants in the context of population screening is challenging for clinical laboratories. This is because diagnostic testing is performed for a clinical indication, which means that the patient has a high likelihood of having a disorder, so that variants found in a gene known to be associated with the disorder are more likely to be causal.

Variants classified as pathogenic or likely pathogenic by this process are further evaluated through the process of clinical confirmation. The exome sequencing in the Geisinger-Regeneron collaboration is not currently performed in a clinically certified laboratory, so variants must be confirmed in a clinical laboratory before the information can be used for patient care. By design, MyCode biospecimens are collected and maintained to comply with relevant clinical regulations, which obviates the need to collect another specimen, thus reducing the burden on each participant.

Initial Clinical Care And Support For Results Reporting The current process of informing patient-participants about their results is described below and visually depicted in the online appendix exhibit.[17] The process was developed in consultation with participants and providers across a range of specialties. Variants that are reported to patients are placed in the EHR using a scanned PDF laboratory report. Representation of the genes and variants in a form that is readable and hence searchable by a computer is maintained on a server behind the Geisinger firewall to support searching and follow-up. International standards for representing genomic data in EHR systems are in development. Once the standards are implemented in commercial EHR systems, the Geisinger process will be modified to use them, eliminating the need for local solutions.

Participants preferred for their providers to be notified first. A system was implemented to notify providers prior to notifying patient-participants, which allows the provider time to access materials relevant to conditions with which they might not be familiar. Online mini–continuing medical education courses and paired patient-provider interpretive reports[18] were developed for each condition category. The clinical genomics team—consisting of clinical geneticists, licensed genetic counselors, genomic medicine assistants, and support personnel—is available for consultation at the request of providers. Each patient-participant who receives a result must be contacted, to provide the opportunity to discuss implications of the result for their health care. This is done through letters and phone calls from a member of the team. Patient-participants who cannot be reached are sent a certified letter with the result, information about recommended care for the condition, information for family members, and contact information for the team. The team uses existing system communication channels for patient-participants whose providers are outside the Geisinger system so that the reports reach the providers.

Patient-participants are given the choice to follow up with their primary care or specialist provider, have a visit with a member of the clinical genomics team, or both. Because this care is provided as a clinical extension of participation in a research program, initial consultation with the team is provided at no charge to the participant-patient or third-party payer (costs are underwritten by Geisinger). A network of specialists and condition-specific clinics with expertise in disorders relevant to the genomic result works with the clinical genomics team to ensure the availability of evidence-based care for interested patients.

For the subset of patients covered by the Geisinger Health Plan, coordination with the payer ensures that any medical care recommended based on the reported result is considered medically necessary and is covered. The plan has also agreed to provide coverage for single-site genetic testing of relatives of the patients at risk of in-

heriting a variant, if they are plan members.

Communicating genomic results to at-risk relatives to support cascade genetic testing of these relatives enhances the value of the program. To empower patient-participants to communicate the genetic information to their close family members, the clinical genomics team requests the number of at-risk first-degree relatives and provide the appropriate number of copies of the result and a family letter. The team is available to support relatives considering testing.

A hallmark of learning health care systems is a commitment to continuous improvement. Two examples illustrate how continuous improvement cycles are used in MyCode to support genetic testing and reporting. One involves the development and implementation of processes for tracking the status of patients' original consent, to reduce the likelihood of contradicting participants' preferences.[19] Because the MyCode initiative is over ten years old, several versions of the consent document have been used. Older versions did not include consent for clinical use of results—a limitation noted when the clinical genomics team planned to report such results. This necessitated developing a process for obtaining reconsent from certain MyCode participants. While every attempt is made to get such reconsent, some participants have not consented to have results reported to their provider and uploaded into the EHR.

The second example involves managing information about people who have died since enrolling in MyCode, as a result may have value to the family of a deceased participant.[20] Since a participant's death can occur at any point along the MyCode program's pipeline, processes were developed and implemented to check the participant's vital status at multiple time points. At the request of MyCode participants and in consultation with the advisory groups, a procedure was developed to notify family members of a deceased participant and discuss results with them if they are interested.

Transition To Ongoing Care To achieve the goals of a learning health care system, it is necessary to evaluate the impact of reporting genomic results to patient-participants and to the system. To help inform the process throughout Geisinger, the MyCode program leaders, in consultation with relevant stakeholders and advisory groups, have developed a set of outcomes (exhibit 3). Baseline conditions for MyCode participants can be established using historical EHR data that facilitate pre-post comparison of the impact of reporting results. Matched cohorts of Geisinger patients not in MyCode or who have no reportable result can be created to support prospective outcomes research.

For participants who receive their care from Geisinger, many outcomes can be captured from the EHR. Health outcomes might take years or even decades to measure (for example, familial hypercholesterolemia in the pediatric population). The stable enrollment of the Geisinger population provides an ideal opportunity to

Exhibit 3

Framework of outcomes for the clinical implementation of genomic information

Outcome type	Description	Examples
Process	Specific steps in a process that lead—either positively or negatively—to a particular health outcome	Lipid profile performed after return of a pathogenic variant in *LDLR*, a gene associated with familial hypercholesterolemia
Intermediate	A biomarker associated—either positively or negatively—with a particular health outcome	LDLc level at or below the target level of 100 mg/dL in response to interventions recommended based on presence of a pathogenic variant in *LDLR*
Health	Change in the health of an individual, group of people, or population that is attributable to an intervention or series of interventions	Decrease in myocardial infarction rates or cardiac revascularization procedures in response to interventions recommended based on presence of a pathogenic variant in *LDLR*
Cost	Standard costs associated with the interventions and health states experienced by the patient; can also include costs associated with patient-reported outcomes from self-reported health state and life disruption	Costs of sequencing and genomics results delivery infrastructure, direct costs of care related to return of genomic information and its use
Behavioral	Change in patient or provider behavior attributable to genomic information	Improved adherence to medication, modification of care based on condition-specific recommendations
Patient-reported	Report of the status of a patient's health condition, knowledge, or service outcomes that comes directly from the patient, without interpretation of the patient's response	Satisfaction with service, engagement in self-care, knowledge about gene and disease, access to recommended care, self-assessed well-being, family communication of genomic risk result, uptake of cascade testing

SOURCE Geisinger. NOTES LDLc is low-density lipoprotein cholesterol. *LDLR* is the gene that encodes the Low Density Lipoprotein Receptor protein.

measure the long-term impact of a precision health program.

Capturing outcomes data for patients who receive all or part of their care outside of Geisinger is more difficult but can be addressed in three ways: Claims data for Geisinger Health Plan members can be used to measure some outcomes. The Keystone Health Information Exchange, led by Geisinger, allows information from participating health care organizations to be collected for care coordination and research. Finally, patients areperiodically contacted by the clinical genomics team after they have learned of their genomic findings. This provides an additional opportunity to collect information on patient-reported outcomes. Contact with patients is also critical to determining if the measured outcome can be attributed to the patient's learning about the genomic finding. For example, if a patient has a mammogram after learning of a pathogenic variant in the *BRCA1* gene, the mammogram could reflect disclosure of the variant or indicate routine preventive care undertaken irrespective of the variant. Accurate attribution of the outcome to the return of the result is essential to determining the true value of a precision health program like MyCode. At present, thereareno standard approaches to determining attribution. This is an ongoing area of study for this and other precision health programs. Cost outcomes can be determined by applying standard costing methods to the clinical data. Outcomes are needed to populate economic models to examine the cost-effectiveness of the intervention and identify which data elements have the most impact on cost-effectiveness.

Clinical data can also be used to improve understanding of the impact of genetic variants on the risk of disease. These data are fed back into the sequence and data analysis process to improve variant annotation, creating a virtuous cycle—an essential element for a learning health care system. Variants reported to participants are also deposited into publicly available databases such as ClinVar.[16]

Closing the loop by developing processes to ensure the communication of results and defining and measuring outcomes is essential for any organization implementing precision health in the framework of a learning health care system.

Genome Screening As A Population Health Initiative

Geisinger has focused on several categories of conditions (encompassing eighty genes, referred to in exhibit 2 as "Geisinger genes") that met our initial, purposely conservative, criteria for clinical actionability.[21] It includes genes deemed reportable by the American College of Medical Genetics and Genomics.[22] The rapidly changing knowledge about gene-disease associations requires a process to reanalyze previously analyzed sequences and incorporate new knowledge about variants' pathogenicity. Approximately 3.5 percent of participants have a reportable variant.[12] As of January 2018, results had been reported to over 500 MyCode patient-participants.[23] Review of the metrics associated with the reporting process combined with input from the advisory committees allows Geisinger to identify opportunities for process improvement, and then to develop and implement these improvements. This results in increased capacity for reporting results and informs the new Geisinger National Precision Health Initiative.[24]

Early results from this program have been disseminated. Cases describing the impact of the program on patients carrying *BRCA1/2* pathogenic variants demonstrate the potential value of the program for participants.[25] While these anecdotal cases support the hypothesis that the program confers value, systematic analyses using pragmatic methodologies are under way to evaluate the value proposition on a wide scale. Studies in other organizations using standard methodologies are needed for replication and to assess the generalizability of the Geisinger findings.

Conclusion

This precision health program demonstrates two necessary conditions as identified by David Chambers and colleagues[26] for the convergence of implementation science, precision medicine, and a learning health care system: Clinical research need not be complete prior to implementation; and research and practice can—we would say must—coexist. These are central to Geisinger's vision of realizing the value of implementing a precision health program.[8] The approaches described in this article represent essential components that are relevant to any organization that considers developing a precision health program. Specific processes' generalizability to other settings must be evaluated in the context of local organizational factors, ideally using conceptual frameworks from implementation science. Assessment of the precision health program to identify and lower barriers to dissemination beyond Geisinger is ongoing.

A population-based approach to precision health that integrates implementation science and the principles of the learning health care system will be used to continually improve the value of the care delivered to Geisinger patients. ▪

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