NATIONAL ACADEMIES Sciences Engineering Medicine

Board on Health Sciences Policy

Roundtable on Genomics and Precision Health

Board on Health Care Services

National Cancer Policy Forum

Realizing the Potential of Genomics across the Continuum of Precision Health Care: A Workshop

Genomics Roundtable Hybrid Workshop

October 12, 2022

Webcast
https://www.nationalacademies.org/event/10-12-2022/realizing-the-potential-of-genomics-across-the-continuum-of-precision-health-care-a-workshop

*Questions for speakers can be submitted in the box under the webcast on this site

NATIONAL ACADEMIES Sciences Engineering Medicine

Board on Health Sciences Policy
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Roundtable on Genomics and Precision Health
National Cancer Policy Forum

Realizing the Potential of Genomics across the Continuum of Precision Health Care: A Workshop

October 12 & 13, 2022

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AGENDA

Realizing the Potential of Genomics across the Continuum of Precision Health Care: A Workshop

October 12, 2022

CLICK HERE TO REGISTER

STATEMENT OF TASK

A planning committee of the National Academies of Sciences, Engineering, and Medicine will organize and conduct a public workshop to examine how genomic data are used in health care settings and to identify opportunities for advancement of precision health care delivery. The overarching goal of the workshop is to examine strategies to ensure that genomic applications are responsibly and equitably adopted to benefit populations as well as individuals over time.

The public workshop will feature invited presentations and discussions to explore:

- Examples of how genomic data are being used to assess health risk outside of traditional settings for clinical genetics (e.g., prenatal screening and testing, newborn screening, polygenic risk scores) and guide decision-making with an eye toward understanding challenges and opportunities related to equity of access to innovation in science, and population level adoption of genomic applications;
- How patients, clinicians, and payers assess and act upon the risks and benefits of genomic screening and diagnostic testing; and
- Challenges of integrating genomic data from various sources into clinical decision-making, including those obtained outside of traditional clinical care settings (e.g., direct-to-consumer, consumer directed, workplace genetic testing) to support equitable precision health care.

The planning committee will organize the workshop, develop the agenda, select and invite speakers and discussants, and moderate or identify moderators for the discussions. A broad array of stakeholders may take part in the workshop, including clinicians, genomics experts, users of the health care system (e.g., patients and families), payers, bioethicists, regulators, digital health experts, and policy makers. Proceedings of the presentations and discussions at the workshop will be prepared by a designated rapporteur in accordance with institutional guidelines.

WEDNESDAY, OCTOBER 12, 2022

SESSION I: Opening Remarks & Keynote

Moderator: Sarah Wordsworth, Professor and University Lecturer; Health Economics Research Centre, Nuffield Department of Population Health, University of Oxford

10:30 AM ET

Welcoming Remarks
Michelle Penny, *Roundtable Co-Chair*
Executive Vice President, Research & Development
Embark, Inc.

Greg Feero, *Roundtable Co-Chair*
Representing Journal of the American Medical Association
Professor, Department of Community and Family Medicine, Geisel School of Medicine
Faculty, Maine Dartmouth Family Medicine Residency Program

10:40–10:50 AM

Introduction and Charge to the Workshop Speakers and Participants
Mira Irons, *Workshop Planning Committee Co-Chair*
President & CEO
College of Physicians of Philadelphia

Christa Martin, *Workshop Planning Committee Co-Chair*
Chief Scientific Officer, Geisinger
Professor and Director, Autism & Developmental Medical Institute

10:50-11:15 AM

Keynote
Euan Ashley
Associate Dean, School of Medicine
Roger and Joelle Burnell Professor of Genomics and Precision Health
Professor of Medicine, Genetics, Biomedical Data Science, & Pathology
Stanford University

SESSION II: What do Patients Need as Genomics Moves into Clinical Care?

Co-Moderators: Gwen Darien, Executive Vice President for Patient Advocacy and Engagement, National Patient Advocate Foundation and Candace Henley, Founder/Chief Surviving Officer, The Blue Hat Foundation

Objectives

- Explore how patients assess and act upon genetic risk information they receive from genomic applications that may change over time (e.g., consumer genetic testing, polygenic risk scores, prenatal testing).
- Examine what patients may need to make informed decisions surrounding genetic testing and follow up care.

11:15–11:30 AM

Keri Norris
Vice President of Health Equity, Diversity, and Inclusion
National Hemophilia Foundation

11:30–11:45 AM

Greta Goto
Founding Member
Prader-Willi Syndrome Alaska Parent Group
Co-Chair, Community Engagement in Genomics Working Group
NHGRI

11:45–12:00 PM

Cristi Radford
Product Director
Optum

12:00–12:25 PM

Panel Discussion

12:25-1:25 PM

Break

SESSION III: What Will it Take to Build an Equitable Precision Health Care System?

Moderator: Gabriel Lázaro-Muñoz, Assistant Professor of Psychiatry, Member of HMS Center for Bioethics, Harvard Medical School

Objectives

- Discuss what an equitable precision health care system is and what it would take to deliver on this promise for patients and clinicians.
- Explore barriers that could be broken down to build an equitable precision health care system (e.g. access to precision health tools and clinician effectiveness in using those tools)
- Examine opportunities for improving implementation by engaging underserved and diverse communities.

1:25–1:40 PM

Kellan Baker
Executive Director and Chief Learning Officer
Whitman-Walker Institute

1:40–1:55 PM

Consuelo Wilkins
Professor of Medicine
Senior Vice President and Senior Associate Dean, for Health Equity and Inclusive Excellence
Engagement Core Director, All of Us Research Program
Vanderbilt University Medical Center

1:55–2:10 PM

Mary Relling
Co-investigator and Co-Founder, Clinical Pharmacogenetics Implementation Consortium
Endowed Chair, Pharmaceutical Sciences Department
St. Jude Children's Research Hospital

2:10–2:35 PM

Panel Discussion

SESSION IV: What Genetic Testing Logistics Issues Need to be Addressed?

Moderator: Victoria Pratt, Vice President, Molecular Diagnostic Quality Assessments, Optum Genomics

Purpose

- Examine and compare what evidence (e.g., clinical validity and clinical utility) means in the context of insurance companies, the clinical setting, and laboratories creating genetic tests.
- Understand how patients, payers, and clinical providers assess the value and benefits of genomic screening and diagnostic testing.

2:35–2:50 PM ET

Lee Hilborne
Medical Director
Quest Diagnostics
Professor of Pathology and Laboratory Medicine
David Geffen School of Medicine
University of California, Los Angeles

2:50–3:05 PM

Bruce Quinn
Principal
Bruce Quinn Associates LLC

3:05–3:20 PM

Mylynda Massart
Assistant Professor of Family Medicine
Department of Family Medicine
University of Pittsburgh
Founder and Director, UPMC Primary Care Precision Medicine Center
Chair of Family Medicine, UPMC Magee Women's Hospital

3:20–3:45 PM **Panel Discussion**

3:45–4:05 PM **Break**

SESSION V: What are the System-level Challenges and Opportunities?

Moderator: W. Gregory Feero, Professor, Department of Community and Family Medicine, Geisel School of Medicine; Faculty, Maine Dartmouth Family Medicine Residency Program

Purpose

- Examine system-level barriers to widespread adoption of genomics and precision health care including data integration, cost and payment, and leadership buy-in.
- Discuss what non-geneticist clinicians may need in order to adopt genetic testing in clinical care.

4:05–4:25 PM ET **Initial Remarks (5 min. each)**

Philip Zazove
Professor Emeritus
Department of Family Medicine
University of Michigan

Tshaka Cunningham
Chief Scientific Officer and Co-founder
Polaris Genomics
Executive Director
Faith Based Genetic Research Institute

Karen Kaul
Chair, Department of Pathology and Laboratory Medicine
Duckworth Family Chair
NorthShore University HealthSystem
Clinical Professor of Pathology
University of Chicago Pritzker School of Medicine

Kara Maxwell
Assistant Professor of Medicine
Perelman School of Medicine
University of Pennsylvania

4:25–5:10 PM **Panel Discussion**

SESSION VI: What Will Genomics Adoption Look Like in the Future?

Moderator: Mira Irons, President & CEO, College of Physicians of Philadelphia

Purpose

- Explore what adoption may look like in the next 10-20 years – how clinicians will be ordering genetic testing, accessing and interpreting results, and using genetic data in routine healthcare. Explore how individuals will access their results and act on them as part of their healthcare.

5:10–5:35 PM

Moderated Discussion

Amy Compton-Phillips
President and Chief Clinical Officer
Press Ganey consulting division

David H. Ledbetter
Chief Clinical & Research Officer
Unified Patient Network, Inc.
Professor, Department of Psychiatry
University of Florida

5:35-5:50 PM

Wrap Up

Mira Irons, *Workshop Planning Committee Co-Chair*
President & CEO
College of Physicians of Philadelphia

Christa Martin, *Workshop Planning Committee Co-Chair*
Chief Scientific Officer, Geisinger
Professor and Director, Autism & Developmental Medical Institute

GENOMICS ROUNDTABLE INFORMATION

NATIONAL ACADEMIES Sciences Engineering Medicine

Roundtable on GENOMICS and PRECISION HEALTH

The sequencing of the human genome is rapidly opening new doors to research and progress in biology, medicine, and health care. At the same time, these developments have produced a diversity of new issues to be addressed.

The National Academies of Sciences, Engineering, and Medicine has convened a Roundtable on Genomics and Precision Health (previously the Roundtable on Translating Genomic-Based Research for Health) that brings together leaders from academia, industry, government, foundations and associations, and representatives of patient and consumer interests who have a mutual concern and interest in addressing the issues surrounding the translation of genome-based research for use in maintaining and improving health. The mission of the Roundtable is to advance the field of genomics and improve the translation of research findings to health care, education, and policy. The Roundtable will discuss the translation process, identify challenges at various points in the process, and discuss approaches to address those challenges.

The field of genomics and its translation involves many disciplines, and takes place within different economic, social, and cultural contexts, necessitating a need for increased communication and understanding across these fields. As a convening mechanism for interested parties from diverse perspectives to meet and discuss complex issues of mutual concern in a neutral setting, the Roundtable: fosters dialogue across sectors and institutions; illuminates issues, but does not necessarily resolve them; and fosters collaboration among stakeholders.

To achieve its objectives, the Roundtable conducts structured discussions, workshops, and symposia. Workshop summaries will be published and collaborative efforts among members are encouraged (e.g., journal articles). Specific issues and agenda topics are determined by the Roundtable membership, and span a broad range of issues relevant to the translation process.

Issues may include the integration and coordination of genomic information into health care and public health including encompassing standards for genetic screening and testing, improving information technology for use in clinical decision making, ensuring access while protecting privacy, and using genomic information to reduce health disparities. The patient and family perspective on the use of genomic information for translation includes social and behavioral issues for target populations. There are evolving requirements for the health professional community, and the need to be able to understand and responsibly apply genomics to medicine and public health.

Of increasing importance is the need to identify the economic implications of using genome-based research for health. Such issues include incentives, cost-effectiveness, and sustainability.

Issues related to the developing science base are also important in the translation process. Such issues could include studies of gene-environment interactions, as well as the implications of genomics for complex disorders such as addiction, mental illness, and chronic diseases.

Roundtable sponsors include federal agencies, pharmaceutical companies, medical and scientific associations, foundations, and patient/public representatives. For more information about the Roundtable on Genomics and Precision Health, please visit our website at nationalacademies.org/GenomicsRT or contact Sarah Beachy at 202-334-2217, or by e-mail at sbeachy@nas.edu.

500 Fifth Street, NW, Washington, DC 20001
Phone 202.334.2217 E-mail sbeachy@nas.edu nationalacademies.org/GenomicsRT

Roundtable on Genomics and Precision Health Membership

W. Gregory Feero, M.D., Ph.D. (Co-Chair) *JAMA*
Michelle Penny, Ph.D. (Co-Chair) Embark Inc.

Naomi Aronson, Ph.D.
BlueCross/BlueShield Association

Aris Baras, M.D., M.B.A.
Regeneron Pharmaceuticals

Vence Bonham, Jr., J.D.
National Human Genome Research Institute

Bernice Coleman, Ph.D., ACNP-BC, FAHA, FAAN
American Academy of Nursing

Robert B. Darnell, M.D. Ph.D.
The Rockefeller University / NY Genome Center

Geoffrey Ginsburg, M.D., Ph.D.
Global Genomic Medicine Collaborative (G2MC)

Jennifer Goldsack, MChem, M.A., M.B.A.
Digital Medicine Society (DiMe)

Eric Gustafson, Ph.D.
Merck & Co.

Jill Hagenkord, M.D. FCAP
Optum Genomics

Cassie Hajek, M.D.
Helix

Richard Hodes, M.D.
National Institute on Aging

Geoff Hollett, Ph.D.
American Medical Association

Mira Irons, M.D.
College of Physicians of Philadelphia

Praduman Jain, M.S.
Vibrent Health

Sekar Kathiresan, M.D.
Massachusetts General Hospital

Alisha Keehn, M.P.A.
Health Resources and Services Administration

Muin Khoury, M.D., Ph.D.
Centers for Disease Control and Prevention

Charles Lee, Ph.D., FACMG
The Jackson Laboratory for Genomic Medicine

Christa Lese Martin, Ph.D., FACMG
Geisinger

Mona Miller, M.P.P.
American Society of Human Genetics

Adele Mitchell, Ph.D.
Biogen

Jennifer Moser, Ph.D.
U.S. Department of Veterans Affairs

Maximilian Muenke, M.D., FACMG
American College of Medical Genetics and Genomics

Kenneth Offit, M.D.
American Society of Clinical Oncology

Kathryn Phillips, Ph.D.
University of California, San Francisco

Victoria M. Pratt, Ph.D., FACMG
Association for Molecular Pathology

Murray Ross, Ph.D.
Kaiser Foundation Health Plan, Inc.

Wendy Rubinstein, M.D., Ph.D.
Food and Drug Administration

Nadeem Sarwar, Ph.D.
Eisai Inc.

Sheri Schully, Ph.D.
All of Us Research Program, NIH

Nonniekaye Shelburne, C.R.N.P., M.S., A.O.C.N.
National Cancer Institute

Geetha Senthil, Ph.D.
National Institute of Mental Health

Nikoletta Sidiropoulos, M.D.
University of Vermont Health Network Medical Group

Katherine Johansen Taber, Ph.D.
Myriad Women's Health

Ryan Taft, Ph.D.
Illumina

Jacquelyn Taylor, Ph.D.
Columbia University

Sharon Terry, M.A.
Genetic Alliance

Joyce Tung, Ph.D.
23andMe, Inc.

Jameson Voss, M.D.
U.S. Air Force

Karen Weck, M.D.
College of American Pathologists

Catherine A. Wicklund, M.S., CGC
National Society of Genetic Counselors

Huntington F. Willard, Ph.D.
Genome Medical, Inc.

Sarah Wordsworth, Ph.D.
University of Oxford

Shannon Zenk, Ph.D.
Natioanl Institutes of Nursing Research

Project Staff
Sarah H. Beachy, Ph.D., *Roundtable Director*
Kathryn Asalone, Ph.D., *Associate Program Officer*
Meredith Hackmann, *Associate Program Officer*
Samantha Schumm, Ph.D., *Associate Program Officer*
Lydia Teferra, *Research Assistant*
Aparna Cheran, *Senior Program Assistant*

VISION	**Realizing the full potential of health for all through genomics and precision health.**
MISSION	**We bring together diverse voices to encourage innovation and actions that foster the wide adoption of and equitable access to the benefits of genomics and precision health.**

As a group of committed stakeholders, we believe in…

- Creating an inclusive and optimistic environment for discussion
- Learning from successes and missteps in the field
- Demanding reproducible evidence-based science
- Sharing trustworthy information
- Embracing interdisciplinary strategies
- Optimizing data privacy and security
- Advancing health equity in all that we do

The Roundtable focuses its energy and resources on these priorities:

DRIVE **INNOVATION** IN GENOMICS AND PRECISION HEALTH	Identify the competing barriers and facilitators of innovation for genomics-based diagnostics, risk assessment tools, and therapies. Leverage opportunities to learn from and promote innovative approaches that can accelerate commercialization and integration to drive impact of genomics on precision health.
SPUR THE **ADOPTION** OF GENOMICS-BASED TOOLS AND PRECISION HEALTH APPROACHES	Cultivate evidence-based practices across the health care and public health systems for adopting genomics and precision health. Draw attention to gaps in adoption and their root causes and highlight potential solutions.
ACHIEVE **EQUITY** IN GENOMICS AND PRECISION HEALTH	Foster action related to underrepresentation and inequities in genomic research, workforce, and access to genomic services by people who need them. Look internally to improve the processes and practices the Roundtable employs to achieve its mission.
SHAPE THE **POLICY DIALOGUE** ABOUT GENOMICS AND PRECISION HEALTH	Accelerate the dissemination of actionable knowledge to shape practice and increase public awareness. Inform and influence how decisions are made.

DEFINITIONS

Precision Health | Inclusive of precision medicine, precision health is a broader, proactive and people-focused approach to health, relying on individual-focused care and everyday decision-making to better predict, prevent, and treat disease.

Genetics | Study of heredity, genes, and genetic variation.

Genomics | Study of the genome by using DNA sequencing and other technologies to understand gene structure, function, and regulation.

WORKSHOP INFORMATION

Realizing the Potential of Genomics across the Continuum of Precision Health Care: A Workshop

Roundtable on Genomics and Precision Health
October 12, 2022

Planning Committee Member Roster

Co-Chairs:

Mira Irons, M.D.
President & CEO
College of Physicians of Philadelphia

Christa Lese Martin, Ph.D., FACMG
Chief Scientific Officer
Director, Autism & Developmental Medicine Institute
Geisinger

Members:

Gwen Darien
Executive Vice President for Patient Advocacy and Engagement
National Patient Advocate Foundation

W. Gregory Feero, M.D., Ph.D.
Professor, Department of Community and Family Medicine
Geisel School of Medicine
Faculty, Maine Dartmouth Family Medicine Residency Program

Alisha Keehn, M.P.A.
Branch Chief, Genetic Services Branch
Division of Services for Children with Special Health Needs
Maternal and Child Health Bureau
Health Resources and Services Administration

Gabriel Lázaro-Muñoz, Ph.D., J.D.
Assistant Professor of Psychiatry
Member, HMS Center for Bioethics
Harvard Medical School

Olufunmilayo I. Olopade (NAS/NAM)
Walter L. Palmer Distinguished Service Professor of Medicine
The University of Chicago

Victoria M. Pratt, Ph.D.
Vice President
Molecular Diagnostics Quality Assessments
Optum Genomics

Lawrence M. Simon, M.D., M.B.A., FACS
Medical Director for Utilization Management
and Coding and Reimbursement
Blue Cross and Blue Shield of Louisiana

Sarah Wordsworth, Ph.D.
Professor and University Lecturer
University of Oxford
Health Economics Research Centre
Nuffield Department of Population Health
University of Oxford

NATIONAL ACADEMIES Sciences Engineering Medicine

Board on Health Sciences Policy

Planning Committee Member Biographies

Mira Irons, M.D. *(co-chair)*
Dr. Mira Bjelotomich Irons is the President and CEO of the College of Physicians of Philadelphia. Dr. Irons came to the College from the American Medical Association (AMA), where she served as the Chief Health and Science Officer; Group Vice President, Health, Science and Ethics since 2019. Prior to her work at the AMA, Dr. Irons held the role of Senior Vice President for Academic Affairs at the American Board of Medical Specialties. Dr. Irons is a board-certified physician in Clinical and Biochemical Genetics and Pediatrics and has an impressive breadth and depth of experience in academic medicine, clinical practice, medical professional leadership and scientific research. In addition, she has 30 years of experience in academic medicine, initially at Tufts Medical Center and followed by Boston Children's Hospital where she oversaw clinical operations of the Division of Genetics and Metabolism, served as the residency director for Medical Genetics and fellowship director for the Harvard Laboratory Genetics programs, leading a clinical research program, and serving as an Associate Professor of Pediatrics at Harvard Medical School, in addition to running a busy clinical practice.

Christa Lese Martin, Ph.D., FACMG *(co-chair)*
Christa Lese Martin, PhD, FACMG, is the chief scientific officer at Geisinger. She's also a professor and the director of Geisinger's Autism & Developmental Medicine Institute. She previously held the position of associate chief scientific officer. Dr. Martin is a graduate of Penn State University and earned her doctorate in Human Genetics at the University of Pittsburgh. She's also board-certified by the American Board of Medical Genetics and Genomics in Clinical Cytogenetics. Her current research focuses on the identification and characterization of DNA changes in people with neurodevelopmental and neuropsychiatric disabilities, including autism. She is interested in determining the genetic cause for these disorders and correlating genotype with phenotype by genetic sub-type. She believes this "genotype-first" approach will lead to targeted treatments that can ultimately improve patient outcomes. Her research also focuses on evidence-based approaches to understanding genomic variation. Toward this goal, she's one of the principal investigators of the NIH-funded Clinical Genome Resource (ClinGen). Finally, Dr. Martin is also part of the leadership team for the MyCode Community Health Initiative at Geisinger—a groundbreaking precision health initiative that's bringing genomic medicine into everyday healthcare.

Gwen Darien
Gwen Darien is a longtime patient advocate who has played leadership roles in some of the country's preeminent nonprofit organizations. As executive vice president for patient advocacy and engagement, Gwen leads programs that link Patient Advocate Foundation's patient service programs to NPAF initiatives, with the goal of improving access to affordable, equitable quality health care. Called "a bit of a renegade" by People magazine, Gwen has long insisted on pushing boundaries while maintaining a safe space for patients. As editor and publisher of Mamm, a magazine for women with breast or reproductive cancer, Gwen published features on previously taboo subjects, such as dating after a mastectomy, along with the more expected academic features on news and policy analysis. Her media leadership was recognized by the Avon

Foundation, which honored her as one of "the most powerful women in breast cancer." As a three-time cancer survivor herself, Gwen came into cancer advocacy expressly to change the experiences and outcomes for the patients who came after her and to change the public dialogue about cancer and other life-threatening illnesses. With these goals in mind, in 2005 she started the first stand-alone advocacy entity in a professional cancer research organization at the American Association for Cancer Research, causing outside observers to note the organization's "progressive commitment to patient advocacy." At AACR, she launched CR magazine – a magazine for people with cancer and those who care for them. Later, she served as the executive director of the Samuel Waxman Cancer Research Foundation; director of The Pathways Project; and executive vice president of programs and services at the Cancer Support Community. In each role, Gwen championed placing patients at the center of health system change, whether it is for research, public policy or direct services. While serving as the chair or on the board of a wide range of program committees and workshop faculties, including the Community Engagement in Genomics Working Group of the National Human Genome Research Institute; a member of the US Pharmacopeia Board of Trustees; and as the chair of PCORI's Patient Engagement Advisory Panel, Gwen also writes about her experiences. Her most recent piece, Transformation: My Experience as a Patient and an Advocate in Three Chapters appeared in the National Academy of Medicine Perspectives. Gwen is a graduate of Sarah Lawrence College, where she also served as an advisor for their Health Advocacy program. She grew up in Milwaukee, but now lives in New York City, where she cooks Persian dishes, collects earrings and improves her friends' personal libraries, one book at a time.

W. Gregory Feero, M.D., Ph.D.
Dr. Feero is a family medicine physician and human genetics specialist. Serving at Four Seasons Family Practice in Fairfield from 2001-06, Dr. Feero rejoined Maine General Medical Center's active staff at Four Seasons in July 2009 after working as chief of the Genomic Healthcare Branch of the National Human Genome Research Institute, National Institutes of Health in Bethesda, MD. Dr. Feero received a Doctorate in Human Genetics from the University of Pittsburgh Graduate School of Public Health and his medical degree from the University of Pittsburgh School of Medicine. He also serves as a faculty member in the Maine Dartmouth Family Medicine Residency program and is a Professor for the Department of Community and Family Medicine at the Geisel School of Medicine at Dartmouth Hanover, NH. He is an associate editor for the Journal of The American Medical Association and is co-chair of the Roundtable on Genomics and Precision Health of the National Academies of Sciences, Engineering and Medicine.

Alisha Keehn, M.P.A.
Alisha Keehn M.P.A., serves as the Branch Chief for the Genetic Services Branch. Ms. Keehn came to HRSA from the American College of Medical Genetics and Genomics (ACMG) where she served as the Associate Program Director of the National Coordinating Center for the Regional Genetics Networks for 12 years. Prior to ACMG, Ms. Keehn worked at Emory University Department of Human Genetics as the Program Director for the Southeast Regional Genetics and NBS Collaborative. Ms. Keehn holds a Master of Public Administration degree from Georgia State University with an emphasis in Policy Analysis and Evaluation. Alisha previously worked at the local county public health level on programs improving access to health

insurance by underserved populations. Ms. Keehn's research interests include public health genetics, access to genetic services by underserved populations, genetics workforce. Ms. Keehn has presented at numerous national conferences and published articles on public health genetics, evaluation of regional genetics programs in the United States, models for regional genetics systems in the United States, and the genetics workforce.

Gabriel Lázaro-Muñoz, Ph.D., J.D.
Dr. Lázaro-Muñoz is Assistant Professor in the Center for Bioethics at Harvard Medical School and Massachusetts General Hospital (MGH) Psychiatry. Dr. Lázaro-Muñoz combines his background in neuroscience, law, and bioethics to examine the implications of emerging biomedical technologies in neuroscience and genomics. He is principal investigator of studies funded by the BRAIN Initiative-National Institutes of Health and the National Human Genome Research Institute. Dr. Lázaro-Muñoz's current studies examine ethical and social implications of the integration of psychiatric genomics into clinical care, polygenic embryo screening, and the development of neurotechnologies such as adaptive deep brain stimulation systems. Dr. Lázaro-Muñoz received his PhD in Neuroscience from New York University; his JD and Master of Bioethics from the University of Pennsylvania; and his BA in Psychology from the University of Puerto Rico.

Olufunmilayo I. Olopade, M.D., FACP (NAS/NAM)
Olufunmilayo I. Olopade, MD, FACP, is an expert in cancer risk assessment and individualized treatment for the most aggressive forms of breast cancer, having developed novel management strategies based on an understanding of the altered genes in individual patients. She stresses comprehensive risk reducing strategies and prevention in high-risk populations, as well as earlier detection through advanced imaging technologies. Dr. Olopade is internationally renowned for her expertise in breast cancer, and her research has advanced early detection, treatment and prevention of breast cancer in women at high risk for the disease. A distinguished scholar and mentor, Olopade has received numerous honors and awards including honorary degrees from six universities and a 2005 MacArthur Fellowship ("Genius grant") for "translating findings on the molecular genetics of breast cancer in African and African-American women into innovative clinical practices in the United States and abroad." Dr. Olopade has received numerous honors and awards, including honorary degrees from North Central, Dominican, Bowdoin and Princeton universities. She is also a recipient of the Doris Duke Distinguished Clinical Scientist and Exceptional Mentor Award, an American Cancer Society Clinical Research Professorship and the Officer of the Order of the Niger Award. Dr. Olopade is an elected member of the American Academy of Arts and Sciences, the American Philosophical Society and the National Academy of Sciences. She currently serves on the board of directors for the American Board of Internal Medicine, the National Cancer Advisory Board, Susan G. Komen for the Cure, Cancer IQ and the Lyric Opera

Victoria M. Pratt, Ph.D., FACMG
Dr. Pratt is Vice President of Molecular Diagnostics Quality Assessments at Optum Genomics. She is also a board-certified Medical and Clinical Molecular Geneticist by the American College of Medical Genetics. Dr. Pratt is the Past President of Association for Molecular Pathology. Dr. Pratt continues to serve on the Centers for Disease Control and Prevention (CDC) GeT-RM

program for reference materials for Molecular Genetics and the American Medical Association's (AMA) Molecular Pathology Current Procedural Terminology (CPT) Advisory committee. Previously, Dr. Pratt is a former advisor of EurogenTest for genetic test validation. Additionally, she served on the Centers for Medicare and Medicaid Services Clinical Diagnostic Laboratory Tests Advisory Panel and the U.S. Secretary of Health and Human Services Advisory Committee on Genetics, Health and Society for the Oversight of Genetic Testing and the Advisory Committee on Hereditary Disorders in Newborns and Children. She also participated in the preparation of the Morbidity and Mortality Weekly Report for Best Practices in Molecular Genetic Testing for the CDC and the National Academy of Medicine's Committee on Policy Issues in the Clinical Development and Use of Biomarkers for Molecularly Targeted Therapies. Dr. Pratt has authored over 100 peer-reviewed manuscripts and book chapters. She is also an Associate Editor for the Journal of Molecular Diagnostics. Dr. Pratt graduated with a Ph.D. in Medical and Molecular Genetics from Indiana University School of Medicine. Her fellowship training was in Ph.D. Medical and Clinical Molecular Genetics at Henry Ford Hospital, Detroit MI.

Lawrence M. Simon, M.D., M.B.A., FACS

Dr. Larry Simon is the Medical Director for Utilization Management and Coding and Reimbursement Blue Cross and Blue Shield of Louisiana, a Rotarian, an animal rescuer, and a native of Lafayette, LA. He is an alumnus of Louisiana State University, Baylor College of Medicine (BCM), Rady Children's Hospital's Pediatric Otolaryngology Fellowship in San Diego, CA, and the BI Moody College of Business Administration at the University of Louisiana. He is also a Clinical Assistant Professor of Otolaryngology at LSU. Larry is a diplomate of the American Board of Otolaryngology and a Fellow of the American College of Surgeons. He is a nationally recognized leader in the business of medicine, having received awards from BCM, the AAO-HNS, ASPO, and SENTAC. His areas of expertise are Health Policy/Healthcare Reform and Coding/Reimbursement, and he has presented over 130 lectures and seminars on these topics. He currently serves on the CPT Editorial Panel and the CPT Assistant Editorial Board of the AMA; the Otolaryngology Advisory Council and Quality Pillar of the ACS; and the Practice Management Education Committee and the Annual Meeting Program Committee of the AAO-HNS. Larry is President-Elect of the Rotary Club of Lafayette-North and serves on the 2020 District Conference Planning Committee and as Editor of the 2019-2020 District 6200 Newsletter. He chairs the 2019 Healthcare Campaign of the United Way of Acadiana, and sits on the Advisory Councils of the LSU Ogden Honors College, the Lafayette Animal Shelter and Care Center, and the HIM Department of the ULL. He also sits on the Boards of Beacon Community Connections and Junior Achievement of Acadiana and is a major donor to the Acadiana Center for the Arts and the Hilliard Art Museum. Larry lives in Lafayette with his partner Lindsay and their many dogs. Together, they founded the LMS Animal Rescue Foundation, a 501(c)(3) public charity that operates RuffRider Transports and is dedicated to the fostering and rescuing of animals nationwide. Their foundation saved over 700 dogs from euthanasia in 2018 and is track to save over 1,000 dogs in 2019.

Sarah Wordsworth, Ph.D.
Dr. Wordsworth is a Professor in the Health Economics Research Centre, University of Oxford. She has almost 25 years' experience in the evaluation of costs and benefits of health care technologies. Since 2003 she has led a research programme on the economics of genetic and genomic technologies and personalised medicine. Of particular interest are the economics of translating genomic high-throughput technologies from research into clinical practice, in cancer, infectious disease and cardiovascular disease. Sarah is lead for the 100,000 Genomes Project, Genomics England Health Economics Clinical Interpretation Partnership and an advisor to NHS England on the implementation of whole genome sequencing into the UK National Health Service. The 100,000 Genomes Project is a major UK Department of Health initiative to mainstream genomics and biggest sequencing study of its type globally. A lack of evidence on the cost-effectiveness of novel genomic technologies such as whole genome sequencing is a key translational challenge. Sarah's work is highly translational as health economic evaluation is often required before new technologies are adopted. The genomics research she has designed and led, has informed key changes in practice, such as producing evidence that gene panels are more cost-effective than single-gene testing. Sarah has co-authored several text books on analysis methods for health economics and genomic research audiences and wrote the economics chapter for the 2017 Chief Medical Officer Annual Report, which was on Genomics. She is a member of the Centre for Personalised Medicine and is Co-Director of a new course on Precision Cancer Medicine at the University of Oxford.

NATIONAL ACADEMIES Sciences Engineering Medicine

Board on Health Sciences Policy

Realizing the Potential of Genomics across the Continuum of Precision Health Care: A Workshop

Roundtable on Genomics and Precision Health
October 12, 2022

Speaker Biographies

Euan Ashley, B.Sc., M.B. Ch.B., FRCP, DPhil, FAHA, FACC, FES, is Associate Dean and Professor of Medicine and Genetics at Stanford University in California. Over the last decade his team has focused on the application of the human genome to medicine. He was recognized by the Obama White House for his contributions to Personalized Medicine and awarded the American Heart Association Medal of Honor for Genomic and Precision Medicine. His book The Genome Odyssey – Medical Mysteries and the Incredible Quest to Solve Them was released in 2021. He is co-founder of three companies: Personalis, DeepCell and SVEXA. Father to three young Americans, in his spare time, he tries to understand American football, plays jazz saxophone, and conducts research on the health benefits of single malt Scotch whisky.

Kellan Baker, Ph.D., MPH, M.A., is the Executive Director of Whitman-Walker Institute, the research, policy, and education arm of Whitman-Walker, a community health system in Washington, DC that also includes Whitman-Walker Health, a Federally Qualified Health Center. Kellan is a health services researcher, educator, and health policy professional with wide expertise in health equity research and policy, particularly with regard to LGBTQ populations. He is a frequent advisor for government and private entities, and he currently serves as an appointed member of a National Academy of Sciences consensus study committee that developed standards for the collection of sex, gender identity, and sexual orientation data by the National Institutes of Health. Kellan holds appointments as affiliate faculty in the Departments of Health Policy and Management at the George Washington University and the Johns Hopkins School of Public Health, and he received his PhD in health policy and management from Johns Hopkins, where he was a Health Policy Research Scholar and Centennial Scholar; an MPH and MA from George Washington University; and a BA with high honors from Swarthmore College.

Amy Compton-Phillips, M.D., is an internationally respected physician, executive, innovator, and author. In October 2022, she became president at Press Ganey, focusing on simplifying health and care. Until September 2022, Amy was president of clinical care at Providence, responsible for clinical operations and care, including improving health, care, and value outcomes delivered by the 52 hospitals, 1,085 clinics, and 120,000 caregivers of the $25 billion health system. She was instrumental in Providence's early adoption and scaling of technology advancing the future of health care. Before joining Providence in 2015, Dr. Compton-Phillips spent 22 years at Kaiser Permanente, moving from a front-line internist to ultimately serving as chief quality officer.

Tshaka Cunningham, Ph.D., is Co-founder & Chief Scientific Officer of Polaris Genomics Inc., an emerging biotechnology company using the genomics and precision medicine to improve diagnosis and treatment of mental and behavioral health conditions. Dr. Cunningham, a graduate of Princeton University, received his PhD in molecular biology from Rockefeller University & completed his postdoctoral training at the Pasteur Institute in Paris, France and at the National Institutes of Health in Bethesda, MD. He previously worked at the Department of Veterans Affairs overseeing a federally funded national research program in aging & neurodegenerative disease and serving as a subject matter expert for the VA's Genomic Medicine Implementation Program. Motivated by the timely need for advancements in diversity and inclusion in precision medicine while at the VA, Dr. Cunningham co-founded and serves as the Executive Director of the Faith-Based Genetic Research Institute, a community-based non-profit organization dedicated to improving people's lives through the power of genomics and precision medicine. A leading voice in advocating for diversity & representation in the field of genomics, Dr. Cunningham also serves as a board member of the Future Kings and Queens of STEM biomedical program, a STEM-focused non-profit organization for youth from underserved communities in the DC-area.

Greta L. Goto, MBA, is an advocate, legal guardian, representative payee and parent to a young adult who lives with Prader-Willi Syndrome; parent to a neuro-typical young adult; and, wife to an accomplished seafood quality assurance manager in Alaska. In her day job, Greta is a part-time research aide with the Alaska Center for Climate Assessment and Policy. She also serves as vice chair of the board of directors for the Bristol Bay Native Corporation and as co-chair of the NHGRI Community Engagement in Genomics Working Group. Her professional career is grounded in non-profit and business administration, community outreach, research and project development, strategic planning, board, and committee work. Greta is a founding member of the Prader-Willi Syndrome Alaska Parent Group and a member of Prader-Willi Syndrome Association USA. She is a graduate of Georgetown University and received her MBA from the University of Alaska Anchorage.

Lee H. Hilborne, M.D., MPH, is a professor of pathology and laboratory medicine at UCLA and served as a member of the ASCP Board of Directors for 18 years and was president from 2007-2008. He chairs the ASCP Effective Test Utilization Subcommittee and is a member of the Commissions on Membership and Public Policy. Dr. Hilborne is also Senior National Medical Director Quest Diagnostics and has held multiple positions since joining Quest Diagnostics in 2008. For five years Dr. Hilborne was medical director, Southern California before assuming the role of senior medical director within Medical Affairs. Dr. Hilborne has given hundreds of invited presentations nationally and internationally and has well over 100 publications in peer reviewed journals. For ten years Dr. Hilborne was Director of Quality Management Services and Associate Director at UCLA Health, responsible for, among other areas, for quality of care and patient safety, medical staff functions, utilization review, and medical coding. He served on several federal committees, including Medicare's Ambulatory Payment Classification Advisory Committee and the Clinical Laboratory Improvement Advisory Committee (CLIAC) and is again a current member of CLIAC. He was the American Hospital Association's representative to the AMA's CPT Editorial Panel and now serves as ASCP CPT Advisor to the AMA CPT Editorial

Panel and co-chair of the Proprietary Laboratory Analysis Technical Advisory Group (PLA-TAG).

Karen Kaul, M.D., Ph.D., is Chair of the Department of Pathology and Laboratory Medicine at NorthShore and is a Clinical Professor of Pathology at the University of Chicago's Pritzker School of Medicine. Dr. Kaul is board-certified in Anatomic Pathology, and Molecular Genetic Pathology. Following a postdoctoral fellowship at the NCI and pathology residency training at Northwestern, Dr. Kaul established one of the earliest Molecular Diagnostics laboratories in the US; she and her lab have been deeply involved in the development of laboratory tests for cancer, heritable conditions, microbial diseases, and antimicrobial susceptibility. She has been significantly involved in education, regulation, and standardization of the practice of molecular pathology, and has served on FDA, CLIAC, MEDCAC, and other panels, and testified before the Senate HELP committee on LDPs in 2016. She is a past president of the Association for Molecular Pathology, and served as Editor in Chief of the Journal of Molecular Diagnostics until 2010. She is the recipient of the 2008 Association for Molecular Pathology Leadership Award. She was an ELAM (Executive Leadership in Academic Medicine) fellow in 2011-2012. In 2011, she was appointed a Trustee of the American Board of Pathology where she is involved in professional examination and certification efforts, and is the past President of the ABP. She also served on the ACGME Residency Review Committee for Pathology, and Milestones committees, and currently leads the Association for Pathology Chairs GME committee. Dr. Kaul served as residency program director for 18 years, and served on PRODS council before becoming departmental chair in 2012. As Chair, she has led departmental efforts to improve laboratory efficiency and utilization, and maximize the impact of the laboratory on clinical care. She continues to practice and advocate for Molecular Pathology

David H. Ledbetter, Ph.D., FACMG, is Chief Clinical & Research Officer, Unified Patient Network, Inc. Former Executive Vice President & Founding Chief Scientific Officer, Geisinger (2010-2021). After serving as Executive Vice President and Founding Chief Scientific Officer at Geisinger for ten years (2010-2021), Dr. Ledbetter assumed the role of Chief Clinical & Research Officer at Unified Patient Network, Inc., a start-up company building a massive Precision Medicine database across multiple healthcare systems linking patient EHR and other clinical data sets to clinical whole genome sequence data. Previously he held academic and leadership positions at Emory University, the University of Chicago, and Baylor College of Medicine. He is a graduate of Tulane University and earned his Ph.D. at the University of Texas-Austin. Dr. Ledbetter is an internationally recognized expert in Genomics and Precision Medicine, with a special interest in autism and other pediatric brain disorders. After his discovery of the genetic causes of Prader-Willi syndrome (deletion chromosome 15; 1981) and Miller-Dieker syndrome (deletion or point mutation of a gene on chromosome 17; 1983) early in his career, he has focused his research efforts on discovering the genetic causes of childhood neurodevelopmental disorders such as autism, and the translation of new genomics technologies into clinically useful genetic tests for early diagnosis and intervention. At Geisinger, he led the development of the largest DNA-sequenced patient cohort in the United States, second only to the UK Biobank in the world. His current research interest includes Implementation Science

efforts to move genomics and precision medicine into routine patient care to optimize prevention, early diagnosis, treatments and outcomes across all clinical disease areas.

Mylynda B. Massart, M.D., Ph.D., is a board-certified Family Medicine physician at UPMC, and assistant professor at the University of Pittsburgh. She serves as the founder and Medical Director of the UPMC Primary Care Precision Medicine clinic, and as the Associate Director of Clinical Services for the Institute for Precision Medicine. Dr. Massart is co-director for the HUB Core over Research Inclusivity and Community Partners Core at the Clinical and Translational Science Institute (CSTI). Her research interests are in developing education in genetics and precision medicine for primary care providers and trainees and to be a research catalyst facilitating the inclusion of underrepresented populations in biomedical research. She teaches residents and medical students in her clinic and at the hospital and serves as medical director for Bethany Hospice. Currently, Dr. Massart is one of the co-Investigators for the All of US Pennsylvania research project working on community education and engagement. In addition, she is working as co-Investigator to create the local Discovery Biobank at the University of Pittsburgh and developing systems to return precision medicine results to providers and patients. Dr. Massart is the principal investigator for the NIH Community Engagement Alliance (CEAL) Consultative Resource (CEACR) team.

Kara Maxwell, M.D., Ph.D., is an Assistant Professor in the Department of Medicine, Division of Hematology/Oncology and the Department of Genetics at the University of Pennsylvania School of Medicine and a Staff Physician at the Corporal Michael Crescenz Veterans Affairs Medical Center. She completed her Bachelor of Science with a dual major in Genetics and Biochemistry at the University of Wisconsin-Madison and completed her MD-PhD at the Weill Cornell Medical College and Rockefeller University. Dr. Maxwell performed her doctoral training with Dr. Jan Breslow at The Rockefeller University in the field of cholesterol metabolism where she cloned and characterized the mouse and human forms of Pcsk9. She then performed her Internal Medicine residency at New York Presbyterian Hospital Columbia and her Hematology/Oncology fellowship at the University of Pennsylvania. She subsequently completed a cancer genetics fellowship and post-doctoral training in the field of human cancer genetics at the University of Pennsylvania. Dr. Maxwell performed her postdoctoral training in the laboratory of Dr. Katherine Nathanson at Penn Medicine, focusing on cancer genetics and genomics in BRCA-deficient breast and ovarian cancer. Dr. Maxwell's clinical practice is in the area of cancer risk evaluation and includes medical management of a variety of cancer risk syndromes. At Penn Medicine, she is a regional referral expert for patients with Li Fraumeni Syndrome, a rare cancer risk syndrome due to TP53 mutations. Dr. Maxwell also co-directs the Cancer Genetics Program at the Corporal Michael Crescenz Veterans Affairs Medical Center in Philadelphia where she works to provide alternative genetic testing care models to a racially diverse population of Veterans. In her Veterans Affairs role, Dr. Maxwell is also an expert consultant for the National Precision Oncology Program (NPOP) where she provides consultation for implementing cancer risk and tumor genomic testing. Dr. Maxwell's laboratory studies mechanisms of tumor formation in breast and prostate cancer due to germline mutations in TP53 and other DNA repair genes. Dr. Maxwell is a recipient of the Burroughs Wellcome Career Award for Medical Scientists and a National Cancer Institute K08 Award. She is also

funded through the Prostate Cancer Foundation, the Li Fraumeni Syndrome Association and the Basser Center for BRCA at Penn Medicine.

Keri Norris, Ph.D., MPH, MCHES, is as a public health professional with extensive training and expertise in health equity, health promotion and disease prevention at the local, state, and federal level. She has worked for some premiere public health and higher education institutions (including the CDC, The Fulton DeKalb Hospital Authority, Spelman College, Agnes Scott College, Baylor University and Morehouse School of Medicine). She is currently the Vice President of Health Equity, Diversity and Inclusion for the National Hemophilia Foundation. She has a TEDx Talk on Hiding in Plain Sight: Health Equity and What's Missing. She is a graduate of Agnes Scott College, Morehouse School of Medicine, the University of South Carolina, and Emory University Law School. Keri serves on the board of Henry County Board of Health, Good Samaritan Health Center Atlanta, Haven of Light, International and the advisory board of the National Coalition Against Domestic Violence. She is a member of Junior League DeKalb and Alpha Kappa Alpha Sorority, Inc. Dr. Norris is an author, coach, and mentor. In her spare time, she enjoys binge watching a good series, bad karaoke, and spending time with family. She has a son and a grandson.

Cristi Radford, M.S., CGC, Product Director, is responsible for overseeing the development and launch of genetic test management capabilities within Optum's laboratory benefit management solution. Previously, Radford served as Director of Genetics, Clinical Initiatives for UnitedHealthcare, where she was responsible for developing and implementing strategies to improve access to genetic testing and services while driving affordability and member outcomes. Of note, she spearheaded the launch of Fit-at-50, a program increasing the use of direct-to-member, in-home, colorectal cancer screening for average risk members, setting a framework for precision population health programs. She also led the implementation of a rapid genetic testing program in the NICU setting, which increased the use of rapid genetic testing in NICU settings using a novel reimbursement method. Prior to joining UnitedHealth Group, Radford was Channel Development Manager at Genome Medical. Earlier in her career, Radford developed several community-based cancer risk assessment programs before transitioning into commercial molecular diagnostics in 2012 at Ambry Genetics and subsequently Invitae. Her academic roles have included positions at Vanderbilt University and Moffit Cancer Center, where she helped create and expand ICARE, as well as Johns Hopkins University. ICARE is an effort to improve access to cancer genetics expertise for patients and health care providers and includes a cancer registry. Radford holds a Bachelor of Science degree from the University of Georgia and a Master of Science degree in Genetic Counseling from the University of South Carolina.

Bruce Quinn. M.D., Ph.D., is an expert in Medicare policy for innovative technology. His initial career was as a full-time medical school faculty member. Armed with an MBA in 2001, he shifted to a career in strategy consulting. He served as a regional Medicare Part B medical director 2004-2008. He has worked for a global consulting firm, Accenture, as well as for two DC-based health policy firms. Since 2016, he has been an independent consulting primarily focused on genomics and digital technologies. His services include product planning and reimbursement pathways for innovators, as well as due diligence investigations for investors.

His website on health policy and new technology, "Discoveries in Health Policy," has had over a million views, holds 2000 articles, and has hundreds of subscribers from industry, academia, and government. Dr. Quinn is based in Los Angeles.

Mary Relling, Pharm.D., earned her undergraduate B.S. degree from the University of Arizona College of Pharmacy and her doctoral degree from the University of Utah College of Pharmacy. She completed post-doctoral fellowships with Dr. William Evans at St. Jude and with Dr. Urs Meyer at University of Basel. She joined St. Jude as a faculty member in 1988, and was chair of the Department of Pharmaceutical Sciences from 2003-2020. She is also a professor at the University of Tennessee in the Colleges of Medicine and Pharmacy. Her primary interests are in treatment and pharmacogenetics of childhood leukemia and clinical implementation of pharmacogenetic testing. Dr. Relling is part of NIH's Pharmacogenomics Research Network and co-founder of CPIC, the Clinical Pharmacogenetics Implementation Consortium. She has published over 400 original scientific manuscripts. She was elected to the National Academy of Medicine (formerly Institute of Medicine) in 2009.

Consuelo H. Wilkins, M.D., MCSI, is a nationally recognized physician-scientist leader in health equity research focused on integrating social, cultural, and environmental factors into clinical and translational research. Dr. Wilkins is Senior Vice President and Senior Associate Dean for Health Equity and Inclusive Excellence and a Professor of Medicine at Vanderbilt University Medical Center. Dr. Wilkins is a Principal Investigator of three NIH-funded research centers and is responsible for a portfolio of programs in response to the institutions' strategic direction for inclusion and diversity. She has also been PI of a Robert Wood Johnson Foundation Award on measuring and engendering trust in healthcare among African American men and a Patient-Centered Outcomes Research Award on Improving Patient Engagement and Understanding Its Impact on Research. Among Dr. Wilkins many contributions to science is her prescient focus on engaging racial and ethnic minority communities, using implementation science methodologies in the design and conduct of clinical research. She has pioneered efforts to move the academic and clinical research enterprise to transform approaches to clinical research design by embedding participant and community engagement in every aspect of biomedical discovery. An elected member of the National Academy of Medicine, she has published 100+ papers on her research. Dr. Wilkins earned a Bachelor of Science in Microbiology and Doctorate in Medicine from Howard University and a Master of Science in Clinical Investigation from Washington University in St. Louis. She completed residency in Internal Medicine at Duke University Medical Center and a fellowship in Geriatrics at Barnes-Jewish Hospital/Washington University Medical Center.

Philip Zazaove, M.D., is Professor Emeritus at Michigan Medicine, University of Michigan. Dr. Zazove has a profound hearing loss and is one of the first people with this to have become a physician. Dr. Zazove has been one of the pioneers researching health services and primary care for people with hearing loss – ever since he started at the University of Michigan 30 years ago. The research he has conducted, the papers he has published, the consulting he has done, and the presentations he has made at various national/international meetings have focused on health care for deaf/hard of hearing persons. Dr. Zazove has successfully managed large population-based

studies with these individuals in the past. In addition, he has had an interest in primary care genomics, publishing work and completing a sabbatical in this area at University College London. Dr. Zazove graduated from Northwestern University, and received his M.D. from Washington University. He completed his residency training in family medicine at the University of Utah Hospitals and a master's in business at Northwestern's Kellogg School of Business.

Realizing the Potential of Genomics across the Continuum of Precision Health Care

October 12, 2022

SPEAKER GUIDANCE: CONTEXT AND QUESTIONS

Following the Genomics Roundtable's strategic plan development in 2020, the Adoption working group has been interested in cultivating evidence-based practices across the health care and public health systems for adopting genomics and precision health as well as drawing attention to gaps in adoption and potential solutions. The goal of this public workshop is to examine strategies to ensure that genomic applications are responsibly and equitably adopted to benefit populations as well as individuals over time.

Session I: KEYNOTE

Key questions to frame keynote address:

1. Where has genomics lived up to its initial promise and where is the future of implementation?
2. Where is the system currently set up well to deliver on the promise of equitable access for patients?
3. Are people and patients at the center of efforts to integrate genomics and genomic data in clinical care? If not, how can this change?
4. What are the gaps in adopting genomics into routine healthcare, outside of the traditional genetics clinics?
5. How can infrastructure be built in health systems to match data needs?

Session II: WHAT DO PATIENTS NEED AS GENOMICS MOVES INTO CLINICAL CARE?

Objectives:

- Explore how patients assess and act upon genetic risk information they receive from genomic applications that may change over time (e.g., consumer genetic testing, polygenic risk scores, prenatal testing).
- Examine what patients may need to make informed decisions surrounding genetic testing and follow up care.

Questions to frame speakers' talks:

1. What experiences have you had with genetic testing in health care settings? Was genetic testing offered/suggested by clinician? When did testing occur in the diagnostic journey and what was the process like for you?

2. How is genetic and genomic testing similar or different from routine care delivery? Can it be considered part of routine care delivery? If not, why not? Was payment different from other types of routine care?
3. What questions should be asked and what should be considered when discussing genomics and genetics in diagnosis and care? What have you been told by clinicians about testing? How can someone make decisions on testing that align with their values?
4. As a patient or patient advocate, how do you advise patients assess the risks and benefits of genetic screening or testing? How do you advise they act upon that information, particularly in cases where the science is evolving?

Key Questions for Speakers:

1. What information have you been given or what would have been most useful when you had to make an informed decision surrounding genetic testing and follow up care?
2. What could be done to help the general public better understand genetic testing and the potential outcomes?
3. How can patient experiences with genetic testing be improved?
4. How can access to genetic testing be improved? How can clinicians better serve and reach underserved populations?
5. Are people and patients at the center of efforts to integrate genomics and genomic data in clinical care? If not, how can this change?
6. Did learning a genetic result help you and your family? If so, how? If not, why not?

Session III: WHAT WILL IT TAKE TO BUILD AN EQUITABLE PRECISION HEALTH CARE SYSTEM?

Objectives:

- Discuss what an equitable precision health care system is and what it would take to deliver on this promise for patients and clinicians.
- Explore barriers that could be broken down to build an equitable precision health care system (e.g. access to precision health tools and clinician effectiveness in using those tools)
- Examine opportunities for improving implementation by engaging underserved and diverse communities.

Questions to frame speakers' talks:

1. What does an equitable precision health care system look like? How can health systems deliver on the promise of equity for both patients and clinicians?
2. What are some of the major barriers and opportunities for building an equitable precision health care system? Are there examples of health systems that have been successful in breaking down barriers?
3. How should health systems think about improving equity in areas where genomic applications may not be as informative for underrepresented populations?
4. Are people and patients at the center of efforts to integrate genomics and genomic data in clinical care? If not, how can this change?
5. How can equity be improved by engaging underserved and diverse communities during implementation?

Session IV: WHAT GENETIC TESTING LOGISTICS ISSUES NEED TO BE ADDRESSED?

Objectives:

- Examine and compare what evidence (i.e. clinical validity and clinical utility) means in the context of insurance companies, the clinical setting, and laboratories creating genetic tests.
- Understand how patients, payers, and clinical providers assess the value and benefits of genomic screening and diagnostic testing

Questions to frame speakers' talks:

1. What are the key barriers or pain points in the logistics process for genetic testing? What challenges need to be overcome to achieve a clear pipeline for genetic testing?

Key Questions for Speakers:

1. In the context of genetic testing, how do you assess evidence needs? What evidence is needed to order, get reimbursed for, or implement genetic testing in clinical care?
2. How do you think about the value and benefits of genomic screening and diagnostic testing?
3. Are people and patients at the center of efforts to integrate genomics and genomic data in clinical care? If not, how can this change?
4. What hinders broader access to genomic testing? What is needed to overcome access barriers?
5. How could genomic testing be equitable among all populations, especially in underserved populations? What resources are needed to ensure equity?

Session V: WHAT ARE THE SYSTEM-LEVEL CHALLENGES AND OPPORTUNITIES?

Objectives:

- Examine system-level barriers to widespread adoption of genomics and precision health care including data integration, cost and payment, and leadership buy-in.
- Discuss what non-geneticist clinicians may need in order to adopt genetic testing in clinical care.

Key Questions for Speakers:

1. What are the main system-level barriers to widespread adoption of genomics and precision health care in the non-genetics clinic setting? How might these barriers be overcome?
2. Are there unique challenges for difficult to reach or underserved populations? What resources or actions could help alleviate some of these issues?
3. Can genetic testing be considered part of routine care delivery? If not, why not? What do non-geneticist clinicians need to know? What resources (point of care or otherwise) could help them better manage patients who have undergone direct-to-consumer genetic testing or may have a clinical indication for genetic testing?
4. What concerns among clinicians and professional groups may need to be addressed?
5. What data integration issues will need to be addressed as consumer data and health record data combine to inform patient care?
6. Are people and patients at the center of efforts to integrate genomics and genomic data in clinical care? If not, how can this change?

Session VI: WHAT WILL GENOMICS ADOPTION LOOK LIKE IN THE FUTURE?

Objectives:

- Explore what adoption may look like in the next 10-20 years – how clinicians will be ordering genetic testing, accessing and interpreting results, and using genetic data in routine healthcare. Explore how individuals will access their results and act on them as part of their healthcare?

Key Questions for Speakers:

1. Why focus on implementing genomics into clinical care now? What is the potential for transforming communities? What do we need to do to get there?
2. Are people and patients at the center of efforts to integrate genomics and genomic data in clinical care? If not, how can this change?
3. How do you envision health systems, clinicians, researchers, and individuals using data from genetic testing in 10-20 years? What will the data ecosystem look like?
4. What can health systems do with the right resources when it comes to implementation? What do health system leaders need to do now to ensure that genomics can be equitably adopted to improve population level health? What does success look like?

BACKGROUND INFORMATION

Links to Additional Resources

Session II: What do Patients Need as Genomics Moves into Clinical Care?

Matloff, E. 2022. Population Genetic Testing: Save lives and money, while avoiding financial toxicity. *Forbes.* https://www.forbes.com/sites/ellenmatloff/2022/08/04/population-genetic-testing-save-lives-and-money-while-avoiding-financial-toxicity/?sh=41227135c821

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Session III: What will it Take to Build an Equitable Precision in Health Care System?

Haidar, C.E., et al., 2022. Advancing Pharmacogenomics from Single-Gene to Preemptive Testing. *Annual Review of Genomics and Human Genetics* 23:449-473. https://doi.org/10.1146/annurev-genom-111621-102737

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Session IV: What Genetic Testing Logistics Issues Need to be Addressed?

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Session V: What are the System-level Challenges and Opportunities?

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Arndt, B.G., et al. 2017. Tethered to the EHR: Primary Care Physician Workload Assessment Using EHR Event Log Data and Time-Motion Observations *Ann Fam Med.* 15(5): 419–426. https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5593724/

Ellis B. 2010. Complexity in practice: understanding primary care as a complex adaptive system. *Inform Prim Care* 18(2):135-140. https://pubmed.ncbi.nlm.nih.gov/21078236/

Medicare and Genomics Access: A Remarkably Complicated System

Bruce Quinn MD PhD
Bruce Quinn Associates LLC
Los Angeles and San Francisco

MEDICARE & GENETICS: OVERVIEW

This talk addresses **key barriers or pain points in the logistics process for genetic testing**, using the **Medicare program** as a case study. The Medicare program has developed some unique complexities over the past decade, which are poorly understood by outsiders. This presentation will present both structure and process, including how evidence standards are applied. Notably, we will also review what Medicare actually pays for in genetics/genomics, data that is available from public records up through 2020. 2021 data will be released soon.

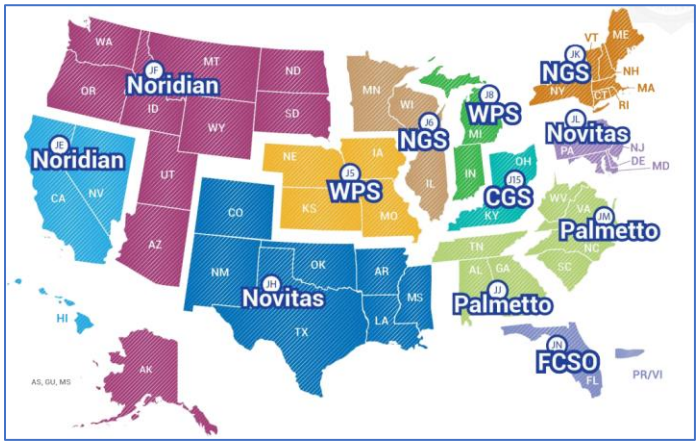
NO COPAYS FOR LAB TESTS

From a socioeconomic standpoint, traditional fee for service Medicare **does not impose cost sharing** or copays on laboratory tests, which makes them **nearly unique** compared to other Medicare healthcare services. Today, even $3000-4000 outpatient genetic tests do not have copays.

MEDICARE DECISION LAYERS: NATIONAL AND LOCAL

Medicare coverage decisions are usually divided into **two layers**, National Coverage Decisions (NCDs) and Local Coverage Decision (LCDs). Only a small number of national policies (NCDs) impact genetics. Medicare claims are processed by under about a dozen regional contracts called **"MACs" or Medicare Administrative Contractors**.

Today, at the local level, there are **three quite distinct policy systems**. (1) The largest group is about 28 states which share uniform molecular pathology coverage and pricing under a program called MolDx, established 2011. (2) The next largest group comprises two MACs with shared policies, named NOVITAS and FCSO. (3) The final group comprises the "NGS MAC" (National Government Services = NGS).



National	National Coverage Decisions	These occur rarely; only several major ones in genetics; only review FDA authorized tests
Local: Three Groups	Local Coverage Decisions	
"MOLDX"	Noridian JE: CA, HI, NV Noridian JF: AK, WA, OR, ID, MT, WY, ND, SD, UT, AZ WPS J5: NE, KS, MO, IA WPS J8: MI, IN CGS J15: OH, KY PALMETTO JM: WV VA NC SC PALMETTO JJ: TN AL GA	All these states share a complicated and large set of policies and billing/coding articles. There is a special MOLDX website and special online databases (some 15,000 tests) and technology assessment forms (about a dozen).
"NOVITAS"	Novitas JH: TX NM CO OK AR LA MS Novitas JL: PA NJ DE MD DC FCSO JN: FL PR	This region had very large overpayments for cancer genomic codes in 2018-2021 with numerous DOJ press releases ("Operation Double Helix, Billion Dollar Fraud") and indictments. These payments are so large as to substantially tilt and distort the overall distribution and types of Medicare molecular payments. They are now issuing tighter payment controls in 2022.
"NGS MAC"	NGS J6: MN WI IL NGS JK: NY CT RI MA MA NH VT ME	This MAC system has by far the lowest per-member per-year genetics payments, except for two nationally covered tests (Exact Sciences Cologuard and Foundation Medicine FMI F1).

OBJECTIVE PAYMENTS

CMS releases all Part B payments, by CPT code, to physicians and labs, each fall for the prior calendar year. CY2020 data was released in November 2021.

Molecular spending rose from circa $1.5B in 2019 to circa $2.3B due to nearly a billion dollars in payments for COVID testing in CY2020.

80% of payments in 2020 were in only about 10 CPT codes as shown in this screenshot:

A	B	C	D	E	F	G
	Code		Services	Dollars Allowed		$2,403,291,283
1	U0003&04	COVID PCR	9,021,988	$900,355,873	37%	37%
2	81479	MISC MOLEC TEST	138,822	$290,906,564	12%	50%
3	81528	**COLOGUARD**	413,272	$210,298,026	9%	58%
4	81408	TIER II LEVEL 9	103,903	$207,027,810	9%	67%
5	81162	BRCA	46,788	$85,338,357	4%	70%
6	81519	**ONCOTYPE**	20,028	$77,568,416	3%	74%
7	0037U	**FND MED FMI**	22,147	$77,514,500	3%	77%
8	81404	TIER II LEVEL 5	147,039	$40,366,543	2%	79%
9	81407	TIER II LEVEL 8	41,329	$34,814,185	1%	80%
10	81599	MISC MULTI ANALYTE TEST	4,738	$33,958,728	1%	81%

MEDICARE PAYMENTS HIGHLY VULNERABLE TO ABUSE IN 2018-2020

Note that lines 4, 8, and 9 above are "TIER 2 codes" which are codes that represent lists of rare genes that are uncommon in clinical practice. AMA ranks these "Tier 2" codes by work intensity (e.g. one exon; two to three exons; four to 10 exons; full sequence).

The Tier 2 codes were mostly paid only in the Novitas system, and looking up labs that were indicted, convicted, or pled guilty**, almost invariably their predominant billing was in the Tier 2 codes**. For example, a lab that pled guilty in September 2022 had this billing pattern in 2020 (Figure 2). In other words, it had almost no billing and payments outside of Tier 2 codes, for which it garnered nearly $100M in 2020. Since this fraud pattern is in total over $1B in a couple years, it seriously distorts a simple objective Medicare payment analyses for legitimate tests, which, absent COVID, are only around $1B.

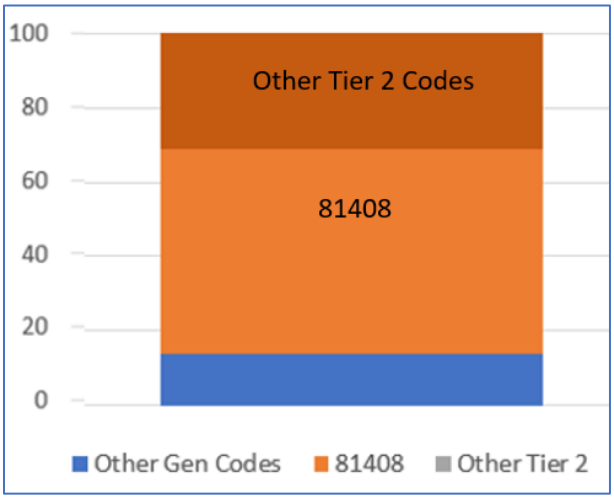


Figure 1 Billing pattern of Lab, pled guilty in 9/2022

This tier 2 billing pattern shown in the bar chart above did not occur in the NGS MAC areas or the MOLDX areas.

The CMS program (primarily Novitas system) was “attacked” by 81408 billing in **2018, 2019, and 2020.** The **natural rate of billing for this code** in the Medicare population is **0 or nearly 0**, as shown in 2016, 2017.

The 81408 payments for 2018, 2019, 2020 **exceed $500M**, half a billion dollars.

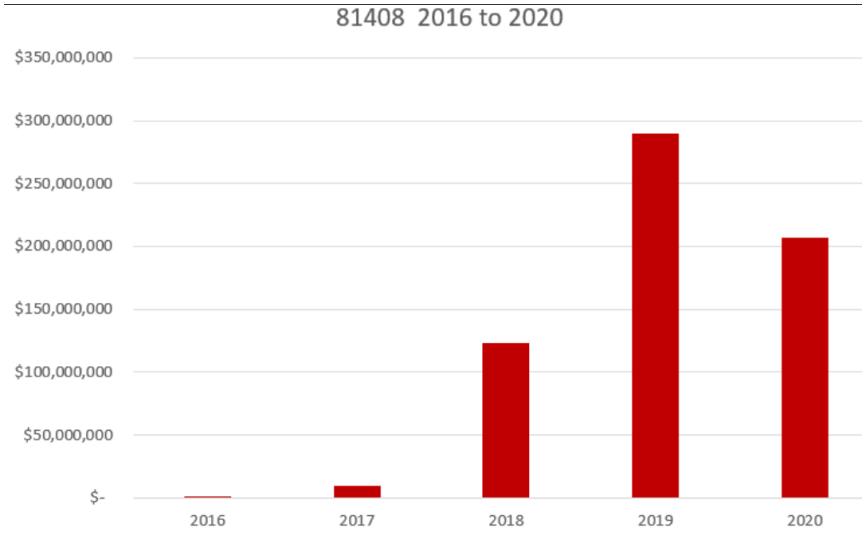


Figure 2 81408 Billing, NOVITAS system 2018-2020

The states in the MOLDX program did not have claims for Tier 2 codes, but in contrast, these states were vulnerable to **other** atypical billing patterns. In particular, the MOLDX MACs were vulnerable to anomalous billing for code **87798**, other molecular test by PCR. This billing does **not** represent COVID testing, because COVID testing was assigned a COVID code extremely quickly by March 2020.

In 2022, MOLDX introduced a policy that will drive this 87798 spending downward to nearly zero. Because this policy was not introduced yet during 2021, it is likely the 2021 bar for 87798 will be as high or higher as the 2020 bar:

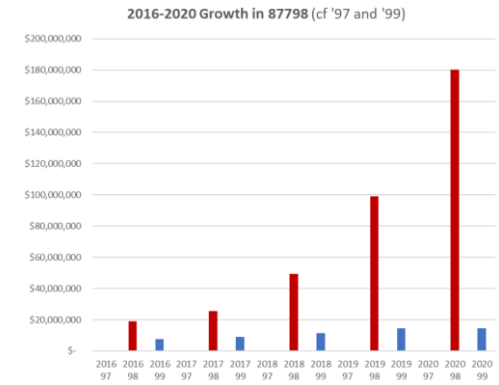


Figure 3 87798 PCR billing, MolDx system, 2016-2020

How Many Medicare Patients Got Large Tumor Panels? It's Murky.

Other quirks of the billing records system are major types of tests divided amongst unexpected codes. For example, CPT code **81455 is tumor panels of 50+ genes** (CMS pays about $3000). This code and test is of very high interest in oncology. There were only **992** such claims paid in Part B in 2020 ($2.8M). At first this seems inexplicably low. However, many claims for this type of test went under code 0037U (Foundation Medicine, 22,147 claims, $77M) while the MOLDX states often required this type of test to be coded as 81479 (miscellaneous molecular test) rather than 81455 (50+ tumor genes). In short, it is difficult to use the claims database to determine the total number of cancer patients who, in 2020, had a 50+ gene tumor test.

In summary, there is a **wealth of available payment data** in Medicare for molecular pathology testing, but analysis is riddled with pitfalls due to a high proportion of inappropriate claims or unusually coded claims.

National Coverage Decisions in Genomics

There are three major national coverage decisions in genomics. All of these focus solely on FDA-approved tests.

Cologuard NCD. In 2013, CMS created an NCD that guarantees payment for the Exact Sciences Cologuard test. This was the third-highest paid code in 2020, at over $200M.

FMI NCD. In 2018, CMS created an NCD that guarantees payment for any FDA-approved cancer test which is FDA approved as a companion diagnostic and used NGS technology. By far the main test in this category is the Foundation Medicine F1 CDX test, in 2020 the 7th highest paid code at over $77M.

NEW CRC NCD. In 2021, CMS created an NCD that guarantees future coverage of FDA-approved blood based screening tests for colon cancer. The tests must be both FDA approved and have a sensitivity of 74% and a specificity of at least 90% (that is, 1 in 10 false positives). No such test meets this criterion yet, but several tests are in R&D pipelines and likely to be reviewed by FDA soon.

National coverage decisions may be opened by petition to CMS, or at the internal discretion of CMS. The process takes about a year and typically involves two cycles of public comment.

Local Coverage Decisions in Genomics

The MolDx program, active in 28 states, has the largest body of LCDs on genomic tests and is unique in requiring submission and review of elaborate technology assessments of analytical and clinical test performance. (For example, MolDx will require and review sample stability studies.) MolDx has a special program where every test is assigned a copyrighted identifier, a "Z identifier" which must be submitted on the claim. However, the Z identifiers are not available in public records, making public health studies difficult. (Claims are often coded under generic molecular code 81479, which is publicly available.) MolDx has coverage for germline hereditary cancer testing for specified conditions, but only allowed in patients with a personal history of cancer. MolDx also has coverage for solid tumor and blood-based tumor testing (circulating DNA testing) for cancer oncogenes. MolDx also has coverage for minimal residual disease testing for cancer patient management, including in colorectal cancer and bladder cancer as well as some hematopoietic cancers (leukemias). MolDx also has coverage for

pharmacogenetic drug-gene pairs if they are endorsed by outside authorities like FDA or CPIC, a pharmacogenetics consortium. It has coverage for detection of organ graft rejection by circulating donor DNA being leaked into blood when a graft is attacked by the host.

Currently, the MolDx program does not have coverage in some other areas, like cardiovascular genetic testing for arrythmias, aortopathies, or cardiomyopathies.

MolDx has an active program of allowing pre-meetings for detailed discussions with labs before dossiers are submitted.

The NGS MAC has several policies for genetic testing. However, interestingly, overall payments in this MAC are **very low** for genetic codes, at least through 2020, compared to other MACs. (With the exception of the fact that his MAC pays for the Cologuard test for all US cases, via Wisconsin, and the FMI test, via Massachusetts. Those payments are determined by national rules, NCDs, not at the discretion or judgment of the NGS MAC.)

The NOVITAS MAC saw rapidly rising molecular payments in 2018-2020 due to an explosion of uncontrolled billing by labs primarily testing 'tier 2" codes. Federal investigators ran "Operation Double Helix" and other enforcement actions and labeled this "a billion dollar problem."

In 2021/2022, NOVITAS MAC system brought out **much stricter policies** in oncology and cardiology. For example, the cardiology states conditions and diseases for which cardiology genetics could be covered, but concludes, that **no** cardiology genes meet the criteria for coverage (none are covered).

COVERAGE DECISION CRITERIA

Medicare pays for services that are "reasonable and necessary" but rather than parsing those words for meaning, generally, Medicare requires that tests be established to improve net health outcomes in Medicare patients. The level of evidence requires varies on the context.

Substantial reliance is placed on major national guidelines, and in 2021/2022, the trend has been for Medicare LCDs to explicitly reference the current version, on a rolling basis, of national guidelines such as the CPIC (PGx) or NCCN (cancer) guidelines. Generally, the MOLDX program explicitly covers follow-on tests by new entrants in a category if they "meet or exceed" the performance of a covered test.

Both NCDs and LCDs are required to have a public display of **evidence considered and rationale** for decision, and both NCDs and LCDs issue a **"response to comments"** submitted by the public.

LEGISLATIVE/REGULATORY APPROACHES TO COVERAGE DECISIONS

There have been attempts to make the coverage process more objective. For example, CMS provides a two-year bonus payment for some inpatient hospital technologies, called the "New Technology Add On Payment" or NTAP. CMS has precise rules for defining medical benefit for use in this decision. In 2019, CMS proposed and finalized a program called "MCIT" or Medicare Coverage for Innovative Technology. This would have provided 4 years of automatic coverage for FDA breakthrough devices, including diagnostics. This particular idea was canceled by the Biden Administration.

In September, I released a five minute video mini review of novel CMS efforts to improve coverage processes – at YouTube here: http://www.discoveriesinhealthpolicy.com/2022/09/new-video-keeping-up-with-busy-tcet.html or https://www.youtube.com/watch?v=G5HW3S9-Xro . The video also covers

new proposals to review and discuss a CMS coverage approach called, “Coverage with Evidence Development” (CED).

Congress has considered, but not passed, legislation that would allow Medicare to cover “preventive” genetic testing for patients at risk of carrying pathogenic cancer genes (such as BRCA; HR4110, S3656).

Congress has also considered, but not passed, legislation that would allow preventive coverage of FDA approved tests for multi-cancer early detection (MCED; see S 1873, HR1946).

SUMMARY

Medicare has extensive coverage for some categories of genetic testing, such as BRCA and Lynch Syndrome in patients with a personal history of breast/ovarian or colon cancer. Medicare has also guaranteed testing for all patients with advanced solid cancers with the centralized Foundation Medicine test. Medicare has acted early to provide coverage of colon cancer screening tests (Cologuard, and, future blood-based colon screening tests, as discussed above). The MolDx program is ahead of most insurance programs in its coverage of minimal residual disease (cfDNA) testing and pharmacogenetic testing. While MolDx only governs 28 states, patient samples from all 50 states can be sent to labs in those 28 states.

There are several major quirks to interpreting Medicare data, such as periodic skyrocketing payments for uncontrolled CPT codes, reaching hundreds of millions of dollars before being caught by policymakers or Department of Justice investigations. Beginning in 2018, these payments are so large (circa hundreds of millions) to seriously distort an analysis of legitimate payments (circa a billion). Also, there are some coding quirks that could trap the unwary, one or two of which were cited above.

Medicare does not have copayments for lab tests, a policy developed for $5 tests but still applied to $3000 tests. This means that covered genetic tests are paid 100% for the patient.

Some areas like cardiology genetics have very limited if any Medicare coverage to date (2022).