

Exploring Applications of AI in Genomics and Precision Health: A Workshop

Committee

Kunal Sanghavi

Co-Chair

Kunal Sanghavi, M.B.B.S., M.S., CGC, is the Associate Director for Genetic Counseling at the Jackson Laboratory for Genomic Medicine. Kunal leads projects on the strategic growth of ethical, legal, and social implications (ELSI) program and precision medicine translational research at The Jackson Laboratory (JAX) for Genomic Medicine. Kunal has developed and implemented multiple translational research studies including population genomic testing, integrating genetics/genomics into primary care, and genetic service delivery, including AI. Kunal remains interested in genetics beyond borders and continues to contribute to different professional societies and international networks. Kunal completed his medical training in Mumbai, India and genetic counseling program in Boston, USA.

Grant Wood

Co-Chair

Grant Wood currently serves as the President and CEO of the Global Genomic Medicine Collaborative (GGMC), an international non-profit dedicated to promoting the collaboration of 90+ genomic research cohort organizations, and the implementation of genetic-based medicine, in over 45 countries. His background in healthcare delivery with a large US-based healthcare system included an informatics strategy to link genomic and family health history data to the electronic medical record, providing physicians with powerful diagnostic tools. Grant is a prolific collaborator, contributing to international groups like HL7 and GA4GH on data standards. He has chaired committees, hosted conferences, and co-authored numerous publications. Grant has served for 15 years on the Board of Directors for AlphaNet, a non-profit that provides disease management services to people with Alpha-1 antitrypsin deficiency. Grant first worked with the NASEM in 2014 with its Action Collaborative titled Integrating Genomic Information into the Electronic Health Record Ecosystem.

Bimal Chaudhari

Member

Bimal P. Chaudhari, M.D., M.P.H., obtained his M.D. and an M.P.H. with concentration in Epidemiology and Bioethics from Boston University. After a pre-doctoral research fellowship in genetic epidemiology at Washington University in St. Louis, he completed clinical training pediatrics (University of Pittsburgh Medical Center), neonatal-perinatal medicine (McGaw Medical Center) and clinical genetics and genomics (Nationwide Children's Hospital). He also completed a post-doctoral fellowship in data science at Northwestern University. Dr. Chaudhari is currently a member of the Sections of Neonatology, Medical Genetics and Genomics and Co-Chair of Hospital Ethics at Nationwide Children's Hospital, a Principal Investigator at the Steve and Cindy Rasmussen Institute for Genomic Medicine as well as an Assistant Professor of Pediatrics at The Ohio State University College of Medicine. Dr. Chaudhari also serves as Associate Director of Biomedical Informatics at the NCH/OSU Clinical and Translational Sciences Institute. Dr. Chaudhari's research focuses on a learning health systems approach to applications of genomic medicine in acute and critical care pediatric populations. Dr. Chaudhari's work involves transdisciplinary collaboration to design, implement and evaluation clinical informatics interventions which address the challenge of making genomic medicine salient to non-geneticist health care providers. Overlapping fields and techniques include data visualization, machine learning, natural language processing, ontologies, clustering and distance measures, human computer interaction, computer supported collaborative work, user centered design and clinical decision support. By both generating foundational knowledge necessary to design such interventions and implementing and evaluating them in the real world, Dr. Chaudhari's work seeks to promote safety, improve outcomes and increase value in medically complex pediatric populations.

Robert R. Freimuth

Member

Robert Freimuth, Ph.D., is Associate Professor of Biomedical Informatics, Associate Chair for Academic Affairs and Faculty Development, and Chair of the Department of AI and Informatics at Mayo Clinic in Rochester, MN. Dr. Freimuth is an internationally recognized expert in genomic data and knowledge management. The goal of his research program is to realize the promise of precision medicine through the development of scalable and interoperable systems and knowledge management platforms, thereby facilitating research and enabling the clinical utilization of genomic data to improve patient health. He is passionate about developing robust, computable representations of data for genomic medicine. Dr. Freimuth is a member of several NIH research networks and a recipient of the 2021 NHGRI Genomic Innovator Award. Within the Center for Individualized Medicine he leads the development of novel systems to enable the integration of genomic data into the EHR and the delivery of genomic data to point of care through EHR-embedded apps. Early in his career, he received the Welch Young Investigator Award to support his research in this area. He has an extensive bibliography that includes original research and international standard specifications that have improved the interoperability and AI-readiness of genomic data. Dr. Freimuth holds leadership positions in several global standards development organizations, including co-chair of the HL7 Clinical Genomics Work Group and co-lead of the GA4GH Genomics Knowledge Standards Work Stream. He earned his Ph.D. degree in Molecular Pharmacology and Experimental Therapeutics from Mayo Clinic Graduate School of Biomedical Sciences, and he completed a post-doctoral fellowship in the Division of Molecular Oncology, Department of Medicine, Washington University.

Maia Hightower

Member

Maia Hightower, M.D., M.P.H., M.B.A., is CEO & Founder of Veritas Healthcare Insights, LLC, specializing in healthcare AI implementation and digital transformation. A physician-executive with over 15 years of leadership, she previously served as EVP & Chief Digital Technology Officer at University of Chicago Medicine, spearheading digital transformation for the \$4B academic health system. Her prior roles include Chief Medical Information Officer at University of Utah Health and University of Iowa Hospitals & Clinics. Dr. Hightower's research and advocacy focuses on responsible AI and digital transformation, influencing policy at organizations like NCQA, NIH, and WHO. She was the founder and former CEO of Equality AI, an AI quality assurance technology company. She holds degrees from Cornell University (BA), University of Rochester (MD, MPH), UC San Diego (Internal Medicine/Pediatric Residencies), and the Wharton School (MBA).

Adriana Huertas-Vazquez

Member

Adriana Huertas-Vazquez, Ph.D., is a Senior Director of Global Medical Affairs at Illumina, where she drives clinical adoption of genetics and genomics through evidence generation studies and medical education programs. She is also the Vice Chair of the Company Constituent Committee at the International Rare Disease Research Consortium, focusing on the efficient execution of rare disease research in the industry space. Adriana received her Ph.D. in Biomedical Sciences from the National University of Mexico and completed her postdoctoral training in Human Genetics and Molecular Biology at the University of California, Los Angeles (UCLA). Prior to joining Illumina, she served as Senior Genetics Lead for Cardiometabolic Diseases at Merck, where she led a team of scientists applying human genetics, bioinformatics, and functional genomics tools to identify novel drug targets. Before her role at Merck, Adriana was an Associate Professor at UCLA, focusing on investigating the genetic architecture of cardiometabolic diseases across different populations. She is based in Los Angeles, California.

Amanda Perl

Member

Amanda Perl is an established nonprofit executive with more than 24 years leading medical, scientific, and professional organizations. Perl has spent her career working with nonprofit organizations on educational program development, strategic planning, governance training and international program development. Prior to joining the American Society of Human Genetics (ASHG) as its Chief Executive Officer, Perl was the Executive Director of the American Thyroid Association where she led the staff and partnered with the Board of Directors to fulfill its mission of Optimal Thyroid Health for All. Before that, Perl was the Chief Global Member Engagement Officer at the Endocrine Society, where she led the team responsible for key organizational initiatives including governance, global society partnerships, member engagement, leadership development, and trainee programs. Perl has also served in leadership roles at the Institute of Food Technologists where she led its foundation fundraising and development activities, oversaw the launch and strategic direction for IFT's Certified Food Scientist program, cultivated global partnerships and led board and volunteer leadership development initiatives. Perl received a BA in Interdisciplinary Studies: Communications, Legal Institutions, Economics and Government and in Public Communication from American University in Washington, DC. An active member of the American Society of Association Executives, Perl was named one of the Association Forum's 40 Under 40 in 2014.

Nalini Raghavachari

Member

Nalini Raghavachari, Ph.D., is a Program Director in the Division of Geriatrics and Clinical Gerontology at the National Institute on Aging (NIA), part of the NIH in Bethesda, Maryland. Her work focuses on the genetics and genomics of exceptional longevity and healthy aging, with an emphasis on integrative multi-omic analyses, translational genomics, and the development of AI/ML approaches for data-driven discovery. She leads initiatives aimed at identifying protective molecular factors, predictive biomarkers, therapeutic targets, and interventions to enhance human healthspan and lifespan. Dr. Raghavachari earned her Ph.D. in Biochemistry from the University of Madras, India, where she studied lipid metabolism in coronary heart disease. She pursued postdoctoral training in genetic engineering and genomics at institutions including Baylor College of Medicine and Texas A&M University. Her industry experience includes roles at Procter and Gamble—developing therapeutics for chemotherapy-induced alopecia—and Corning Life Sciences, where she advanced microarray-based genomic and proteomic technologies. She began her NIH career in 2003 at the Clinical Center, directing the Sickle Cell Genomics Program within the Critical Care Medicine Department. In 2005, she joined the intramural program of the National Heart, Lung, and Blood Institute (NHLBI), where she directed the Genomics Core Facility and contributed to genetic studies of vascular diseases, including the Framingham Heart Study. Since 2013, she has been with the extramural division of the NIA, leading programs that explore the molecular underpinnings of longevity and healthy aging.

Lee Sanders

Member

Lee M. Sanders, M.D., M.P.H., is Professor of Pediatrics and Health Policy, and Division Chief for General Pediatrics at Stanford University. He holds joint appointments in the Department of Health Policy, Department of Epidemiology and Population Health and the Freeman Spogli Institute for International Studies. He teaches in the Human Biology Program and at the Hasso Plattner Institute of Design (Stanford d.School). With funding from NIH, CDC, PCORI and FDA -- Dr. Sanders founded and directs the Stanford Health Literacy Design Lab, which leverages AI and other novel technologies, alongside human-centered design, clinical-trial approaches and population science, to address health disparities. Working with colleagues across disciplines, he leads multi-disciplinary studies that aim to prevent obesity during early childhood, to mitigate the impact of immigration policy on child health, to inform education policy on the long-term consequences of preterm birth, and to improve coordinated care for people with chronic conditions. Dr. Sanders' newest venture is an initiative to build an "AI Design Studio for Population Health" -- applying novel analytic tools in support of front-line public health providers in underserved communities. As a multi-lingual primary-care physician -- Dr. Sanders cares for medically and socially complex children at Stanford Children's Health and at a federally qualified health center.

Angela Starkweather

Member

Angela Starkweather, Ph.D., ACNP-BC, FAANP, FAAN, is Dean and Professor at Rutgers School of Nursing and serves as the American Academy of Nursing representative. As an adult acute care nurse practitioner and nurse scientist, Dr. Starkweather has an ongoing program of research focused on identifying the biopsychosocial mechanisms of chronic pain and other comorbid symptoms and developing multi-level interventions to optimize symptom management and quality of life. Integrating multi-omic measures, her research uses artificial intelligence to examine longitudinal profiles and determine treatment targets. She works with interdisciplinary teams to implement evidence into practice and systems of care. Dr. Starkweather has been continuously NIH-funded over the past decade, has over 200 peer-reviewed publications, is editor of three books, and chief editor of Cancer Nursing, an international journal focused on oncology nursing research and practice.

Zhongming Zhao

Member

Zhongming Zhao, Ph.D., M.S., holds University Chair for Precision Health. He is the founding director of the Center for Precision Health, and currently serves as the Vice President for Cancer Genomic Medicine, the University of Texas Health Science Center at Houston (UTHealth). Before he joined UTHealth in 2016, he was Ingram Endowed Professor of Cancer Research, Professor (tenured) in the Departments of Biomedical Informatics, Psychiatry, and Cancer Biology at Vanderbilt University Medical Center, Chief Bioinformatics Officer of the Vanderbilt-Ingram Cancer Center (VICC), Director of the VICC Bioinformatics Resource Center, and the Associate Director of the Vanderbilt Center for Quantitative Sciences. Dr. Zhao has a unique, interdisciplinary educational and research background. He completed his master's degrees in Genetics (1996), Biomathematics (1998), and Computer Science (2002), Ph.D. degree in Human and Molecular Genetics (2000), and Postdoctoral Fellowship in Bioinformatics (2001-2003). Dr. Zhao has broad interests in bioinformatics, genomics, data science/AI, population genetics, and precision medicine, and has co-authored over 500 total publications in these areas (cited by >30,000 times, H-index = 84). Dr. Zhao is the founding president of The International Association for Intelligent Biology and Medicine (IAIBM, 2018). He was elected as a fellow in the American College of Medical Informatics (ACMI, 2021), the American Medical Informatics Association (FAMIA, 2022), and the American Institute for Medical and Biological Engineering (AIMBE, 2023). Dr. Zhao has received several awards, including the Keck Foundation Post-doctoral Fellowship (twice: 2002, 2003), the NARSAD Young Investigator Award (twice: 2005, 2008), a NIH-funded VPSD Career Development Award in GI Cancer (2009), an Outstanding Achievement Award from the International Society of Intelligent Biological Medicine (2011), the Dean's Excellence Award for Research (SBMI, 2022), and the 2023 President's Scholar Award for Excellence in Research with the honorary title of President's Scholar. Throughout his career, he has collaborated with numerous researchers while also pursuing his own independent research, funded by numerous federal, state, and foundation grants. He has trained more than 85 students and postdoctoral fellows (29 have become academic faculty, including department chair and assistant dean), mentored 14 junior faculty, and co-mentored/collaborated with five NIH K awardees.