Use of Race, Ethnicity, and Ancestry as Population Descriptors in Genomics Research

Committee

Aravinda Chakravarti

Co-Chair

Aravinda Chakravarti, Ph.D. is the Director of the Center for Human Genetics & Genomics, and the Muriel G & George W Singer Professor of Neuroscience & Physiology and Professor of Medicine at the New York University Grossman School of Medicine. He has served on the faculty at the University of Pittsburgh (1980 – 1993), Case Western Reserve University (1994-2000), and Johns Hopkins University (2000-2018). He is one of the founding Editors-in-Chief of Genome Research and Annual Reviews of Genomics & Human Genetics, and is on the advisory boards of numerous national and international Institutes, charities, academic societies, the NIH and biotechnology companies. He has been a key participant in many genome projects, and now works on genome-scale analysis of the molecular basis of human disease. He was the 2008 President of the American Society of Human Genetics and been elected to the US National Academy of Science, the US National Academy of Medicine, the Indian National Academy of Science and the Indian Academy of Sciences. He was awarded the 2013 William Allan Award by the American Society of Human Genetics and the 2018 Chen Award by the Human Genome Organization. Dr. Chakravarti received his Ph.D. in human genetics in 1979.

Charmaine D. Royal

Co-Chair

Charmaine Royal, Ph.D. is the Robert O. Keohane Professor of African & African American Studies, Biology, Global Health, and Family Medicine & Community Health at Duke University. She directs the Duke Center on Genomics, Race, Identity, Difference and the Duke Center for Truth, Racial Healing & Transformation. She held previous faculty appointments at Howard University. Throughout her career, Dr. Royal has focused on ethical, social, scientific, and clinical implications of human genetics and genomics, particularly issues at the intersection of genetics and “race”. She serves on numerous national and international advisory boards and committees for government agencies, professional organizations, not-for-profit entities, and corporations, including the Board of Directors for the American Society of Human Genetics, the Independent Expert Committee for the Human Heredity and Health in Africa (H3Africa) Initiative, and the Ethics Advisory Board for Illumina, Inc. In 2013 and 2015 Dr. Royal served as a chairperson of the planning committee for two consensus roundtable meetings convened by the American Society of Human Genetics charged with developing guidelines for genetic ancestry inference. Dr. Royal obtained a bachelor’s degree in microbiology, master’s degree in genetic counseling, and doctorate in human genetics from Howard University. She completed postgraduate training in ethical, legal, and social implications (ELSI) research and bioethics at the National Human Genome Research Institute of the National Institutes of Health, and in epidemiology and behavioral medicine at Howard University Cancer Center. She was a member of the National Academies’ committees that produced ‘Toward Precision Medicine: Building a Knowledge Network for Biomedical Research and a New Taxonomy of Disease’ and ‘Addressing Sickle Cell Disease: A Strategic Plan and Blueprint for Action’.

Katrina A. Armstrong

Member

Katrina A. Armstrong, MD, leads Columbia University’s medical campus as the Executive Vice President for Health and Biomedical Sciences. She is Chief Executive Officer of the Columbia University Irving Medical Center and Dean of the Faculties of Health Sciences and Medicine, which includes Columbia’s dental, medical, nursing and public health schools. She is an internationally recognized investigator in medical decision making, quality of care, and cancer prevention and outcomes, an award winning teacher, and a practicing primary care physician. She has served on multiple advisory panels for academic and federal organizations and has been elected to the National Academy of Medicine, the American Academy of Arts and Sciences, the Association of American Physicians, and the American Society for Clinical Investigation. Before joining Columbia, Dr. Armstrong was the Jackson Professor of Clinical Medicine at Harvard Medical School, Chair of the Department of Medicine and Physician-in-Chief of Massachusetts General Hospital, and Professor of Epidemiology at the Harvard T.H. Chan School of Public Health. Before joining Harvard, she was Chief of the Division of General Internal Medicine, Associate Director of the Abramson Cancer Center, and Co-Director of the Robert Wood Johnson Clinical Scholars Program at the University of Pennsylvania. She is a graduate of Yale University (BA degree in architecture), Johns Hopkins (MD degree), and the University of Pennsylvania (MS degree in clinical epidemiology). She completed her residency training in internal medicine at Johns Hopkins.

Michael Bamshad

Member

Michael Bamshad, M.D. is Professor and Chief of the Division of Genetic Medicine in the Department of Pediatrics at the University of Washington and Seattle Children's Hospital, and holds the Allan and Phyllis Treuer Endowed Chair in Genetics and Development. Dr. Bamshad is Editor-in-Chief of Human Genetics and Genomics Advances, published by the American Society of Human Genetics and chair of the Scientific Advisory Board of GeneDx. His research focuses on understanding the impact of population structure and natural selection on human genetic variation; developing innovative ways to discover genetic variants underlying monogenic disorders, modifiers of monogenic traits and complex traits; and testing novel ways to translate genomic advances into the practice precision genetic medicine. He and his colleagues pioneered the use of exome and genome sequencing for discovery of genes underlying Mendelian conditions and has contributed to the identification of hundreds of genes for Mendelian disorders. He has also been a leader in understanding the relationship between genetic ancestry and notions of race, developing innovative ways to openly share phenotypic information and genetic data (e.g., MyGene2) and building platforms for self-guided return of genetic testing results (e.g., My46) from exome and whole genome sequencing in both research and clinical settings. He has published more than 300 scientific manuscripts as well as papers in periodicals such as Scientific American, and co-authors a popular textbook entitled Medical Genetics. In 2013 and 2015 Dr. Bamshad served as a chairperson of the planning committee for two consensus roundtable meetings convened by the American Society of Human Genetics charged with developing guidelines for genetic ancestry inference. He received his B.S. and M.D. at the University of Missouri in Kansas City and his M.A. at the University of Kansas.

Luisa N. Borrell

Member

Luisa N. Borrell, D.D.S., Ph.D. is a Distinguished Professor in the Department of Epidemiology and Biostatistics, City University of New York Graduate School of Public Health and Health Policy (CUNY SPH), New York, NY. She is a social epidemiologist with a research interest on the role of race/ethnicity, socioeconomic position, and neighborhood effects as social determinants of health. Her work on Hispanics'/Latinos' racial identity brings attention to the need for disaggregated analyses by race as Hispanics/Latinos are a heterogeneous group with a mix of European, Native American and African ancestry. She also has expertise in research methods and analyses of large and spatially-linked datasets. Dr. Borrell is a Fellow of the New York Academy of Medicine. She has a Doctor in Dental Surgery and a Master in Public Health, from Columbia University, New York, NY, as well as doctorate in Epidemiological Science from the University of Michigan, Ann Arbor, MI.

Katrina Claw

Member

Katrina Claw, Ph.D. is an Assistant Professor in the Division of Biomedical Informatics and Personalized Medicine in the Department of Medicine at the University of Colorado Anschutz Medical Campus. Her research focuses broadly on personalizing medicine, using genetic information and biomarkers for tailored treatment, in relation to pharmacogenomics as well as understanding the ethical, cultural, and social implications of genomic research with populations historically underrepresented in health research. Her current research includes studying cytochrome P450 genetic variation in Indigenous communities (e.g., American Indian and Alaska Native peoples). Her other projects include exploring the perspectives of tribal members on genetic research with tribes and developing guidelines and policies in partnership with tribes. All of her projects strive to use community based participatory research approach and include cultural and Indigenous knowledge. She was awarded the Genomic Innovator Award from NHGRI in 2020 for her work on pharmacogenomics approaches to drug metabolism in American Indian/Alaska Native People. She received her B.S. and B.A. from Arizona State University and her Ph.D. from the University of Washington.

Clarence C. Gravlee

Member

Clarence C. Gravlee, Ph.D. is associate professor in the Department of Anthropology at the University of Florida, where he is also affiliated with the Center for Latin American Studies, the African American Studies Program, and the Genetics Institute. His research examines the genetic and environmental contributors to hypertension in the African Diaspora, with an emphasis on the biological consequences of systemic racism. His work, with collaborators, integrates methods and theory from the social and biological sciences, including ethnography, social network analysis, human biology, and genetics. Gravlee completed a B.A., M.A., and Ph.D. in anthropology at the University of Florida, a Fulbright graduate fellowship at the Universität zu Köln (Cologne, Germany), and postdoctoral training in community-based participatory research as a W.K. Kellogg Community Health Scholar at the University of Michigan School of Public Health.

Mark D. Hayward

Member

Mark D. Hayward, Ph.D. is a professor of sociology and Centennial Commission Professor in the Liberal Arts at the University of Texas at Austin. Hayward is a health demographer. Building on a long-standing interest in the developmental origins of adult health, his current work incorporates biosocial lenses (e.g., pathophysiological pathways and genetic risk) to better understand how social exposures from childhood through adulthood influence racial/ethnic disparities in dementia risk. Hayward is a recipient of the Matilda White Riley Award from the National Institutes of Health for his contributions to behavioral and social scientific knowledge relevant to mission of NIH. He has served on numerous major foundations (Robert Wood Johnson and Pew) and major federal agencies (e.g., the National Institutes of Health and the National Center for Health Statistics). Hayward is the current editor of his field's major journal, Demography, and President-elect of the Interdisciplinary Association of Population Health Science. He received his Ph.D. from Indiana University and his B.A. from Washington State University. He has served on scientific advisory boards at the NASEM including the Committee on Population and a Decadal Survey of Behavioral and Social Science Research on Alzheimer's Disease and Alzheimer's Disease-Related Dementias.

Rick A. Kittles

Member

Rick Kittles, Ph.D., is the inaugural Senior Vice President for Research at Morehouse School of Medicine. Dr. Kittles was previously Professor and founding Director of the Division of Health Equities within the Department of Population Sciences at the City of Hope (COH) and Associate Director of Health Equities of COH Comprehensive Cancer Center. Dr. Kittles is also Co-founder and Scientific Director of African Ancestry, Inc. and is well known for his research of prostate cancer and health disparities among African Americans, having published over 240 research articles. Dr. Kittles serves on many national and international steering committees and advisory boards. He served as a member of the Board of Scientific Counselors (BSC) for the National Human Genome Research Institute (NHGRI/NIH) and is Past Council Chair of the Minorities in Cancer Research (MICR) of the American Association for Cancer Research (AACR). Dr. Kittles' research has focused on understanding the complex issues surrounding race, genetic ancestry, and health disparities. He has been at the forefront of the development of genetic markers for ancestry and how genetic ancestry can be used in genetic studies on disease risk and outcomes, showing the impact of genetic variation across populations. In March of 2012 Dr. Kittles presented the Keynote Address to the United Nations General Assembly, "International Day of Remembrance of Victims of Slavery and the Transatlantic Slave Trade." He received a Ph.D. in Biological Sciences from George Washington University in 1998.

Sandra Soo-Jin Lee

Member

Sandra Soo-Jin Lee, Ph.D. is Professor of Medical Humanities and Ethics and Chief of the Division of Ethics at Columbia University. Trained as a medical anthropologist, Dr. Lee leads interdisciplinary bioethics research on race, ancestry and equity in genomics, precision medicine and artificial intelligence, and publishes in the genomics, medical, bioethics, and social science literatures. Dr. Lee has investigated racial categorization in human genetics for over two decades and co-edited Revisiting Race in a Genomic Age (2008). Her current NIH funded projects include the Ethics of Inclusion: Diversity in Precision Medicine Research. Dr. Lee is Co-Director of the Center for ELSI Resources and Analysis and the ELSI Congress. She is President-elect of the Association of Bioethics Program Directors and a Hastings Center Fellow. Dr. Lee serves on the US Health and Human Services Secretary's Advisory Committee on Human Research Protections, the Scientific Advisory Boards of the Kaiser Permanente National Research Biobank and the Human Pangenome Reference Consortium, and the editorial boards of the American Journal of Bioethics and Narrative Inquiry in Bioethics. Dr. Lee received her doctorate from the University of California, Berkeley/UCSF joint program in Medical Anthropology and her undergraduate degree in Human Biology from Stanford University.

Andres Moreno-Estrada

Member

Andrés Moreno-Estrada, Ph.D., M.D. is the Principal Investigator of the Human Evolutionary and Population Genomics Laboratory at the Advanced Genomics Unit (UGA-CINVESTAV), in Irapuato, Mexico. Previously, he was Research Associate of the Genetics Department at Stanford University until 2014. He is a Mexican population geneticist interested in human genetic diversity and its implications in population history and medical genomics. His work integrates genomics, evolution and precision medicine in projects involving large collections of understudied populations, in particular from the Americas and the Pacific. He authored the most detailed work so far of the genetic structure of the Mexican population, including the first genomic characterization of 20 diverse indigenous groups throughout Mexico, as well as fine-scale studies in the Caribbean region, South America, and Polynesia. He is leading the Human Cell Map of Latin American Diversity to increase the representation of diverse ancestry networks for the Human Cell Atlas project. For his work in Latin America he was awarded the "George Rosenkranz Prize for Health Care Research in Developing Countries" in 2012. He received his M.D. from University of Guadalajara in 2002 and Ph.D. in Evolutionary Genetics from Pompeu Fabra University in 2009. Dr. Moreno was a postdoctoral fellow until 2012 with Prof. Carlos Bustamante at Cornell University and Stanford University School of Medicine.

Ann Morning

Member

Ann Morning, Ph.D. is a Professor of Sociology at New York University. Trained in demography, her research focuses on race, ethnicity, and the sociology of science, especially as they pertain to census classification worldwide and to individuals' concepts of difference. She is the author of The Nature of Race: How Scientists Think and Teach about Human Difference (University of California Press 2011), and co-author of An Ugly Word: Rethinking Race in Italy and the United States (with Marcello Maneri, Russell Sage Foundation 2022). Morning was a 2008-09 Fulbright research fellow at the University of Milan-Bicocca and a 2014-15 Visiting Scholar at the Russell Sage Foundation. She was a member of the U.S. Census Bureau's National Advisory Committee on Racial, Ethnic and Other Populations from 2013 to 2019 and has consulted on racial statistics for the European Commission, the United Nations, Elsevier, and the World Bank. Morning holds her B.A. in Economics and Political Science from Yale University, a Master's of International Affairs from Columbia University, and her Ph.D. in Sociology from Princeton University.

John P. Novembre

Member

John Novembre, Ph.D. is a Professor at the University of Chicago in the Departments of Human Genetics and Ecology & Evolution. His research has developed computational methods to answer a diverse range of questions regarding genetic diversity. His work has especially had an impact on the understanding and analysis of geographic patterns in human genetic variation. He has been awarded as a MacArthur Fellow, Searle Scholar, and Sloan Research Fellow, and his research is supported by the National Institutes of Health. Dr. Novembre has authored more than 50 peer-reviewed publications in leading journals, including Nature, Science, Nature Genetics, and the American Journal of Human Genetics. He also serves as an academic editor for the journal Genetics, and previously served on the Scientific Advisory Board for AncestryDNA. He received his B.A. from The Colorado College and his Ph.D. from the University of California-Berkeley.

Molly Przeworski

Member

Molly Przeworski, Ph.D. is a Professor of Biological Sciences at Columbia University. Before moving to Columbia University, she was a faculty member at the University of Chicago as well as at Brown University and the Max Planck Institute for Evolutionary Anthropology in Germany. Her research aims to understand the genetic basis and evolutionary history of heritable differences among individuals; recent work focuses in part on genomic trait prediction in humans and implications. She is the recipient of the Rosalind Franklin Award from the Genetics Society of America, a Sloan Research Fellowship, and Howard Hughes Medical Institute Early Career Scientist Award, and is a member of the American Academy of Arts and Sciences and the National Academy of Sciences. She received a B.A. in Mathematics from Princeton University and a Ph.D. from the Committee on Evolutionary Biology at the University of Chicago, then conducted postdoctoral research in the Mathematical Genetics group of the University of Oxford in the United Kingdom.

Dorothy E. Roberts

Member

Dorothy Roberts, J.D. is the George A. Weiss University Professor of Law & Sociology at University of Pennsylvania, with joint appointments in the Departments of Africana Studies and Sociology and the Law School, where she is the inaugural Raymond Pace and Sadie Tanner Mossell Alexander Professor of Civil Rights. She is also Founding Director of the Penn Program on Race, Science & Society. Author of Fatal Invention: How Science, Politics, and Big Business Re-create Race in the Twenty-First Century, Roberts is an expert on structural racism in US science and medicine and the use of race as a variable in scientific research. Her research has been supported by the American Council of Learned Societies, National Science Foundation, Robert Wood Johnson Foundation, Fulbright Program, Harvard Program on Ethics & the Professions, and Stanford Center for the Comparative Studies in Race & Ethnicity. Recent honors include 2019 election as a College of Physicians of Philadelphia Fellow, 2017 election to the National Academy of Medicine, 2016 Society of Family Planning Lifetime Achievement Award, 2015 American Psychiatric Association Solomon Carter Fuller Award, and 2011 election as a Hastings Center Fellow. Professor Roberts serves on the advisory board for the Center for Genetics and Society. She received her J.D. from Harvard Law School and her B.A., magna cum laude, Phi Beta Kappa from Yale College.

Sarah A. Tishkoff

Member

Sarah Tishkoff, Ph.D. is the David and Lyn Silfen University Professor in Genetics and Biology at the University of Pennsylvania, holding appointments in the School of Medicine and the School of Arts and Sciences. She is also the Director of the Penn Center for Global Genomics & Health Equity. Dr.Tishkoff studies genomic and phenotypic variation in ethnically diverse Africans, using field work, laboratory research, and computational methods to examine African population history, the genetic basis of anthropometric, cardiovascular, and immune related traits, and how humans have adapted to diverse environments and diets. Dr. Tishkoff is a member of the National Academy of Sciences, the American Academy of Arts and Sciences, and the National Academy of Medicine. She is a recipient of an NIH Pioneer Award, a David and Lucile Packard Career Award, a Burroughs/Wellcome Fund Career Award, the ASHG Curt Stern Award, and a Penn Integrates Knowledge (PIK) endowed chair. She is on the NAS Board of Global Health and the Scientific Advisory Board for the Packard Fellowships in Science and Engineering, and is on the editorial boards at Cell, PLOS Genetics, and G3 (Genes, Genomes, and Genetics). She received her Ph.D. in Genetics and M.Phil in Human Genetics from Yale University and her B.S. in Anthropology & Genetics from University of California-Berkeley.

Genevieve L. Wojcik

Member

Genevieve L. Wojcik, Ph.D. is an Assistant Professor of Epidemiology at the Johns Hopkins Bloomberg School of Public Health in Baltimore, Maryland. As a statistical geneticist and genetic epidemiologist, her research focuses on method development for diverse populations, specifically understanding the role of genetic ancestry and environment in genetic risk in admixed populations. Dr. Wojcik integrates epidemiology, sociology, and population genetics to better understand existing health disparities in minority populations, as well as underserved populations globally. In 2021, she was the recipient of one of NHGRI’s Genomic Innovator Awards (R35) to do this work. She is a long-standing member of multiple NHGRI consortia focused on diverse populations, such as the Population Architecture using Genomics and Epidemiology (PAGE) Study, which was formed by NHGRI over a decade ago to address the lack of genetics research in non-European ancestry populations, and the PRIMED consortium, which began this year to better conduct research around polygenic risk scores in diverse populations. Dr. Wojcik previously served as a consultant with Illumina, Inc. Prior to her faculty appointment, Dr. Wojcik was a postdoctoral research scholar at Stanford University in the Departments of Genetics and Biomedical Data Science. She received her Ph.D. in Epidemiology and M.H.S. in Human Genetics/Genetic Epidemiology from the Johns Hopkins Bloomberg School of Public Health and her B.A. in Biology from Cornell University.