2017 | ACMG Annual Clinical Genetics Meeting

ACMG American College of Medical Genetics and Genomics
Translating Genes Into Health®

Enriching Racial and Ethnic Diversity to Improve Genomic Medicine

Held in Phoenix, Arizona, March 21-25, 2017

Date of Release: April 3, 2017
Expiration Date: April 3, 2020 (CME, NSGC, P.A.C.E.®)
Estimate Time of Completion: 2 hours
Course must be completed by the expiration dates

COURSE DESCRIPTION

The association of race/ethnicity with socioeconomic status, disease prevalence, and clinical outcomes is well recognized. However, the increasing implementation of genomic sequencing in clinical practice and its reliance on common resources and standardized approaches highlights the need for inclusion of and targeted research in ancestrally diverse populations. Opportunities for genomic research in diverse populations tend to occur in a complex context related to socioeconomic status and access to care, in addition to cultural differences related to genomics. Large clinical sequencing efforts in the United States have tended to draw disproportionately from populations of European descent, even though many Mendelian and complex diseases disproportionately impact minority populations. Comprehensive and accurate variant interpretation depends on population-specific allele frequency, which is incompletely catalogued. Successful inclusion in research studies depends not only on successful recruitment but also retention of diverse patients. Numerous factors may impact these efforts including the processes and language utilized for informed consent, results disclosure, and patient-provider communication, as well as culturally based perspectives regarding the utility of genomic results. Consideration of these factors can highlight approaches that may work across populations, as well as those that may need to be tailored to specific populations or cultures. As a matter of scientific value and equity, building a population-inclusive knowledgebase on which genomic medicine can be based is essential to the progress of genomic medicine for clinical research and patient care.
This session will offer recent examples of how inclusion of ancestrally diverse patients and populations in clinical research illustrate the need to improve such efforts, and will contribute to precision medicine for all. We propose six talks spanning a range from genetic epidemiology to clinical genomics to ethical, legal and social implications drawing from consortia such as the Population Architecture using Genomics and Epidemiology (PAGE), Implementing GeNomics In PracTicE (IGNITE), ClinGen, and Clinical Sequencing Exploratory Research (CSER) programs.

Dr. Carlos Bustamante (Building an Inclusive Knowledge Base for Genomic Medicine), Dr. Bert Boyer(Engaging Rural Alaska Native Community Members in Obesity-related -Omics Research), and Dr. Sharon Plon (Germline Cancer Susceptibility in Diverse Populations: Results from the BASIC3 Trial) declined to release their slides but you will be able to listen to their presentations.

LEARNING OBJECTIVES

At the conclusion of this course, participants should be able to:

- Discuss limitations of existing genomics databases or resources for ancestrally diverse populations and opportunities to improve them
- Evaluate the benefits and challenges to increasing inclusion of diverse populations
- Examine ongoing efforts to improve variant interpretation, patient-provider communication or results disclosure in diverse populations
- Apply key concepts of study design and community engagement for improved clinical research paradigms tailored to inclusion and retention of diverse populations

TARGET AUDIENCE

All healthcare professionals interested in the diagnosis, management, treatment and prevention of genetic conditions and increasing their understanding of the genetic basis of common, chronic health problems affecting both children and adults will find the programming applicable to their practice. These select sessions from the ACMG Annual Meeting are targeted for the following professionals:

- Medical and clinical geneticists
- Physicians of all specialties with an interest in genetics, genomics and the genetic basis of disease
- Genetic counselors
- Laboratory geneticists, directors, technicians and technologists
- Researchers
- Pathologists
- Educators
- Nurses
- Dietitians
- Physician assistants
- Biotechnology and pharmaceutical development professionals
- Fellows, Trainees and Students
- Public health professionals
- Genetic/consumer advocates
- Others with an interest in the science and art of medical genetics and genomics

SESSIONS

- Challenges and Opportunities with Existing Variant Databases: Population-specific Allele Frequencies and ACMG/AMP Pathogenicity Criteria- Eimear Kenny, PhD
- Engaging Rural Alaska Native Community Members in Obesity-related -Omics Research- Bert Boyer, PhD
- Community Engagement in Diverse Populations: The Genetic Testing to Understand and Address Renal Disease Disparities (GUARDD) Study- Carol Horowitz, MD, MPH
- Maximizing Diversity in Genomic Studies: Lessons from NCGENES- Elizabeth Moore, MPH
- Germline Cancer Susceptibility in Diverse Populations: Results from the BASIC3 Trial- Sharon Plon, MD, PhD, FACMG
- Building an Inclusive Knowledge Base for Genomic Medicine- Carlos Bustamante, PhD

Accreditation:

The American College of Medical Genetics and Genomics is accredited by the Accreditation Council for Continuing Medical Education (ACCME) to provide continuing medical education for physicians.

Credit Designation:

The American College of Medical Genetics and Genomics designates this activity for a maximum of 2 *AMA PRA Category 1 Credit*s™. Physicians should claim only the credit commensurate with the extent of their participation in the activity.

Genetic Counselor Credit

The National Society of Genetic Counselors (NSGC) has authorized American College of Medical Genetics and Genomics to offer up to 2 Category 1 contact hours for this OnDemand course. The American Board of Genetic Counseling (ABGC) will accept CEUs earned for this course for the purposes of genetic counselor certification and recertification. Reporting of credits is sent to NSGC quarterly. Additional fee (~$25) applies for NSGC credit that is billed by NSGC.

P.A.C.E. CEUs – Laboratory Directors and Laboratory Personnel

ACMG is approved as a provider of continuing education programs in the clinical laboratory sciences by the American Society for Clinical Laboratory Science (ASCLS) Professional Acknowledgment for Continuing Education (P.A.C.E.®) Program. The American College of Medical Genetics and Genomics designates this course for a maximum of 2 contact hours. ACMG is approved by the Florida Board of Clinical Laboratory Personnel as CE Provider. ACMG is approved by the California Department of Health Services through the ASCLS P.A.C.E.® Program as CE Provider #275.

HIPAA Compliance

The ACMG supports medical information privacy. While the ACMG is not a "covered entity" under HIPAA 1996 and therefore is not required to meet these standards, ACMG wishes to take reasonable steps to ensure that the presentation of individually identifiable health information at ACMG-sponsored events has been properly authorized. All presenters have completed a form indicating whether they intend to present any form of individually identifiable healthcare information. If so, they were asked either to attest that a HIPAA-compliant consent form is on file at their institution, or to send ACMG a copy of the ACMG HIPAA compliance form. This information is on record at the ACMG Administrative Office and will be made available on request.

Content Validation

ACMG follows the ACCME policy on Content Validation for CME activities, which requires:

Content Validation and Fair Balance

1. ACMG follows the ACCME policy on Content Validation for CME activities, which requires:
   a) All recommendations involving clinical medicine must be based on evidence that is accepted within the profession of medicine as adequate justification for their indications and contraindications in the care of patients.
   b) All scientific research referred to, reported or used in CME in support or justification of patient care recommendations must conform to the generally accepted standards of experimental design, data collection and analysis.

2. Activities that fall outside the definition of CME/CE; "Educational activities that serve to maintain, develop, or increase the knowledge, skills, and professional performance and relationships that a physician uses to provide services for patients, the public, or the profession" (source: ACCME and AMA) will not be certified for credit. CME activities that promote recommendations, treatment, or manners of practicing medicine or pharmacy that

are not within the definition of CME/CE or, are known to have risks or dangers that outweigh the benefits or, are known to be ineffective in the treatment of patients.

3. Presentations and CME/CE activity materials must give a balanced view of therapeutic options; use of generic names will contribute to this impartiality. If the CME/CE educational materials or content includes trade names, where available, trade names from several companies must be used.

Off-label Uses of Products

When an off-label use of a product, or an investigational use not yet approved for any purpose, is discussed during an educational activity, the accredited sponsor shall require the speaker to disclose that the product is not labeled for the use under discussion, or that the product is still investigational. Discussions of such uses shall focus on those uses that have been subject of objective investigation.

Disclaimer: *ACMG educational programs are designed primarily as an educational tool for health care providers who wish to increase their understanding of the application of genomic technologies to patient care. The ACMG does not endorse, or recommend the use of this educational program to make patient diagnoses, particular by individuals not trained in medical genetics. Adherence to the information provided in these programs does not necessarily ensure a successful diagnostic outcome. The program should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed at obtaining the same results. In determining the propriety of any specific procedure or test, a healthcare provider should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen.*

2017 ACMG Program and Education Committee Members Disclosures

Members of the ACMG Staff, Education and Program Committees involved in planning the 2017 ACMG Annual Clinical Genetics Meeting are required to disclose relevant relationships which could be perceived by some as a real or apparent conflict of interest in planning. All disclosures have been reviewed and conflicts of interest resolved by the Education Committee COI sub-committee or the Executive Director and CME Associate Director and conflicts of interest are disclosed. In the cases where a conflict existed then the committee member refrained from the discussion.

Following is a list of program and education committee members who have disclosed one or more such relationships and names of companies with which those relationships exist:

EC = Education Committee; PC = Program Committee; S = ACMG Staff

1. Major stockholder/ownership interest 2. Grant/Research Support (External) 3. Salary/Employment/Royalty(ies)/Honoraria 4. Consultant/consulting fees/other remuneration 5. Speakers' bureau	6. Non-remunerative positions of influence such as officer, board member, trustee, or public spokesperson (All Committee Members Below are on ACMG Committees –Members with other affiliations are listed) 7. Receipt of intellectual property 8. Other

Georgianne L. Arnold, MD, FACMG - Horizon, 2; Recordati, 2; Biomarin, 2; Actelion, 2; SIMD, 6; ACGME, 6; AAP, 6; ASHG, 6 (PC)
Karen W. Gripp, MD, FACMG – Wiley Publishing Inc., 3; FDNA, 4; Novartis, 4 (PC)
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Christine A. Curtis, PhD, FACMG - CSI Laboratories, 3 (EC)
Thomas E. Mullen, PhD, MS, FACMG - Good Start Genetics Inc., 3 (EC)

Christian P. Schaaf, MD, PhD, FACMG - Springer Publishing Company, 3 (PC), (EC)
Katrina M. Dipple, MD, PhD, FACMG – ACGME, 6; AAP, 6; ASHG, 6; LA BioMed DSMB, 4; SIMD 6 (PC)

Following is a list of committee, education members and staff who have no relationships to disclose:

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SPEAKERS AND MODERATORS

Faculty Disclosures:

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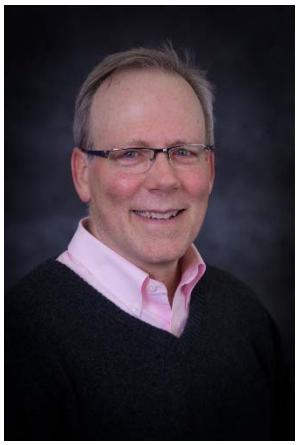
Moderator: Lucia Hindorff, PhD, MPH
National Human Genome Research Institute, NIH
No financial relationships to disclose.

Dr. Hindorff is a Program Director in the Division of Genomic Medicine at the National Human Genome Research Institute (NHGRI). She received her M.P.H. and Ph.D. degrees from the University of Washington, where her research focused on cardiovascular genetic epidemiology and motivating factors for using genetic tests in clinical care. At NHGRI, Dr. Hindorff leads the Clinical Sequencing Exploratory Research (CSER) program, a consortium which is focused on integrating genomic sequencing into the clinic. Dr. Hindorff also leads efforts to broaden large-scale epidemiological cohort studies to diverse populations through the Population Architecture using Genomics and Epidemiology (PAGE) program. She is also the NHGRI lead for the online NHGRI-EBI Genome-wide Association Study (GWAS) Catalog. Her interests include the integration of genetic tests into clinical care, diversity in genomic studies, and practical issues related to large epidemiological studies.

Speaker: Eimear Kenny, PhD
Icahn School of Medicine at Mount Sinai, Charles Bronfman Institute for Personalized Medicine
No financial relationships to disclose.

Eimear Kenny, PhD, is an Assistant Professor of Genetics and Genomic Sciences, and member of the Charles Bronfman Institute of Personalized Medicine, the Icahn Institute of Genomics and Mulitscale Biology, and the Center of Statistical Genetics, at the Icahn School of Medicine at Mount Sinai in New York City. She has a PhD in computational genomics from Rockefeller University, and did her postdoctoral training at Stanford University. She is a leading expert in fields of statistical and population genomics. Her research focuses on the link between genetics, ancestry and disease to inform better outcomes for medical genomics and ameliorate health disparities. Her current research focuses is on population genomic health, including developing statistical methods, population genetic theory, and mining Electronic Health Records to gain a deeper understanding of the landscape of genetic variation in global populations. The long-term goal is in translating these genetic discoveries into improved healthcare management strategies via collaborations with physicians and clinical researchers. She recently led an international academic team to build the Illumina Infinium Multi-ethnic Global Array, designed to enable genomic discovery in global cohorts and biobanks, and she actively participates in several NIH-funded consortia that leverage large-scale genome-phenome data and for discoveries in medicine and epidemiology. She is currently co-leading the center for trans- and multi-ethnic disease mapping in NHGRI's flagship sequencing program.

Speaker: Bert Boyer, PhD
Director, Center for Alaska Native Health Research, University of Alaska Fairbanks
No financial relationships to disclose.



Bert Boyer, PhD Professor of Molecular Biology Director, Center for Alaska Native Health Research Co-Director, American Indian-Alaska Native Clinical Translational Research Program Associate Director of Human Health, Institute of Arctic Biology University of Alaska Fairbanks Dr. Boyer's research group is broadly interested in genomic, epigenomic and environmental risk and protective factors related to obesity and diabetes in Yup'ik people from Southwest Alaska. For the past decade, Dr. Boyer and colleagues have been working in rural Alaska developing a longitudinal study involving ~1,700 Yup'ik Alaska Native people in 11 communities using a community-based participatory research (CBPR) framework. They have found obesity prevalence equal to that in the general U.S. population, but although obesity is one of the greatest risk factors for diabetes, type 2 diabetes incidence in the Yup'ik population is less than half that seen in the other areas of the U.S. To understand this better, Dr. Boyer and colleagues are investigating the roles of physical activity and a traditional subsistence diet rich in polyunsaturated fatty acids in prevention of chronic diseases, including diabetes. In collaboration with colleagues at the University of Washington, Dr. Boyer is also involved in a pharmacogenomics program grant to investigate gene-by-environment interactions related to warfarin drug safety and efficacy. Dr. Boyer and colleagues continue to work towards the development of culturally appropriate strategies to return the full continuum of research results to participants. All projects adhere to a CBPR framework involving community partners and Yup'ik leaders.

Speaker: Carol Horowitz, MD, MPH
Professor, Department of Population Health Science and Policy and the Icahn School of Medicine at Mount Sinai
No financial relationships to disclose.

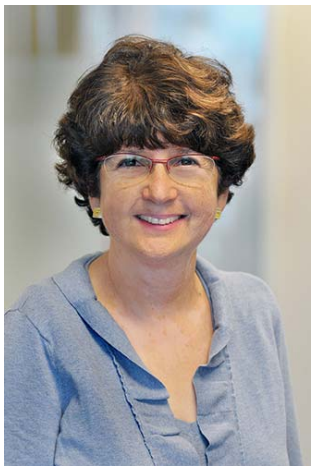
Carol Horowitz is a practicing general internist, in Harlem, health services researcher and Professor in the Departments of Population Health Science and Policy & Medicine at Mount Sinai. She uses community and stakeholder engaged approaches to understand and eliminate health disparities related to common chronic diseases. She co-directs Mount Sinai's new Center for Health Equity and Community Engaged Research, the Sinai

CTSA's Community Engaged Research Core, has been PI and investigator on numerous NIH, CDC and PCORI grants related to chronic disease prevention and control, directs stakeholder engagement for the PCORI-funded NYC Clinical Data Research Network and Chairs NHGRI's translational genomics consortium, IGNITE. Dr. Horowitz has implemented and evaluated programs to improve the quality of care and outcomes of diverse populations of adults with diabetes, obesity, cardiovascular disease and other health conditions through clinical and community programs. She has extensive experience in multi-method (quantitative and qualitative) research, clinical research training, program and intervention development, conducting and analyzing multi-site randomized trials, and managing and working with large, trans-disciplinary teams that include diverse researchers, patients, clinicians, advocates and entrepreneurs and policymakers. She mentors students, residents, fellows and junior faculty, serves on community boards and is active in her local community. Dr. Horowitz received her MD from Cornell, primary care training at Albert Einstein, and received an MPH at the University of Washington as a Robert Wood Johnson Clinical Scholar.

Speaker: Elizabeth Moore, MPH
University of North Carolina at Chapel Hill
No financial relationships to disclose.

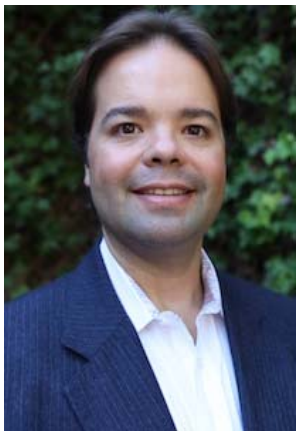
Elizabeth Moore, MPH worked on the North Carolina Clinical Genomic Evaluation by Next-generation Exome Sequencing (NCGENES) project at the University of North Carolina at Chapel Hill. She was a research associate on the project where she managed the quantitative data collection and assisted in data analysis and manuscript preparation. Her Spanish language background allowed her to collect data in Spanish and better engage Hispanic participants. Prior to working on NCGENES, she was a trainee for the Center for Genomics and Society at UNC-Chapel Hill. As a trainee, she received training from Dr. Debra Skinner on qualitative data analysis in the Fragile X Newborn Screening Study. In addition to data analysis, she recruited Spanish speaking patients for the Fragile X Newborn Screening Study in the maternity ward at UNC Hospitals. Her research interests have included engaging Latino populations and improving health disparities in underserved populations.

Speaker: Sharon Plon, MD, PhD, FACMG
Professor, Department of Pediatrics, Baylor College of Medicine, Baylor College of Medicine
Financial relationships to disclose. (Self): Member of scientific advisory board of Baylor genetics laboratories.



Dr. Plon is a diplomat of the American Board of Medical Genetics and Genomics and founding member of ACMG. She received her MD and PhD (Biophysics) from Harvard University and completed her medical genetics training at the University of Washington. Dr. Plon's early laboratory work focused on the regulation of genomic stability. Dr. Plon has been a faculty member of Baylor College of Medicine and Texas Children's Hospital for 24 years. For the last 15 years, her research includes identification of novel cancer susceptibility genes and implementation of genomic testing in medicine. Dr. Plon and D. William Parsons are principal investigators of the NHGRI-funded CSER consortium BASIC3 clinical trial on the incorporation of exome sequencing into the care of newly diagnosed childhood cancer patients. This study is located at Texas Children's Cancer Center which cares for children with cancer from the highly diverse population of Houston/Harris County. Dr. Plon is also principal investigator of the NHGRI Clinical Genomics Resource (ClinGen) which is developing national databases and curation interfaces to improve assessment of all types of clinical variation.

Speaker: Carlos Bustamante, PhD
Stanford University, School of Medicine
No financial relationships to disclose.



Carlos Bustamante is a population geneticist whose research focuses on analyzing genome wide patterns of variation within and between species to address fundamental questions in biology, anthropology, and medicine. From 2002-2009, he was on the faculty at Cornell University, in the Departments of Statistical Sciences and Biology Statistics and Computational Biology, where he was promoted to full professor in 2008. Since 2010, he has been on the faculty in the Department of Genetics at the Stanford University School of Medicine. In 2015, he was appointed the Inaugural Chair of Stanford's new Department of Biomedical Data Science. He has received multiple honors and

awards including a Marshall-Sherfield Fellowship (2001-2), the Sloan Research Fellowship (2007), and a John D. and Catherine T. MacArthur Fellowship (2010). He has trained over 50 post-doctoral fellows and graduate students as primary advisor and co-authored over 130 papers. Much of his research is located at the interface of computational biology, mathematical genetics, and evolutionary genomics. His most current research focuses on human population genomics and global health including developing statistical, computational, and genomic resources for enabling trans- and multi-ethnic genome-wide association and medical sequencing studies of complex biomedical traits. He is one of the Principal Investigators of the recently announced $25M ClinGen project to build the country's National Database of Clinically Relevant Genomic Variants. He has advised multiple companies, non-profits, and government bodies in the past including Mars, Inc., Ancestry.com, Personalis, Inc., the National Human Genome Research Institute, and the Carlos Slim Foundation.

Participation Instructions

1. Participant logs into ondemand.acmg.net
2. Once logged in the participant will access the session they would like to view. They will be asked if they would like to claim credit for the meeting, or if they will not claim credit for the meeting. Then, this information (Course description) will appear, and participant will have to attest that they have read the information. They will then click Continue.
3. After that, the participant will be able to select the credit types to claim.
4. For each session with a post-test, the participant will need to mark and complete the matching pre-test.
5. Then the participant will watch the session presentations.
6. Participant will complete viewing all session content. "Check marks" indicate which presentations have been viewed.
7. After viewing all presentations within a session, participant will click the "Claim Credit for Session" button under the CME dropdown at the top of the page.
8. Participant should take and then successfully pass the post-test. If they do not pass with a score of 80% or higher, they will have unlimited tries to pass the post-test.
9. Participant will continue the steps above to earn credits for additional sessions.
10. If a session does not have a test attached, the participant will not need to take a pre- or posttest, but will have to complete a Concurrent or Plenary session-specific evaluation to claim credit.
11. To print their certificate, the participant will click the "Print Certificate" button under the CME dropdown at the top of the page. Participant must complete the meeting evaluation (one time only) before they can access their certificate. Participant will then choose their certificate(s). The certificate(s) will be automatically updated as they earn new credits.

Stream Requirements	
Network	For best results, use a hardwired network connection instead of wireless
Full Screen Viewing	If you would like to view the webcast full screen, display the tool bar at the bottom and click the double arrow in the far right corner. The screen will enlarge to the full screen of you system. To restore the size, press the "ESC" key

Refresh Browser Window	If the webcast freezes and does not recover in 3-4 seconds, refresh browser window
Freezing or Stuttering Issues	Adjust the amount of bandwidth needed by putting your mouse anywhere over the video window. A tool bar will appear at the bottom. On the right side you will see a "HD" button, click on that button and you will see a list of options. The top is "auto", with decreasing numbers below. Select a lower bandwidth (such as 360p) to see if your webcast improves
For Technical Support call	1-800-504-5379

Mobile Viewing Requirements

Android Devices	Android 2.3+ with Adobe Flash Player 10.2 or better installed Install Flash Player
Apple Devices	iOS 4+

Online Viewing Requirements

Bandwidth	512kbps
Required Hardware and Software	Screen resolution of 1024X768 or larger Sound card and speakers/headphones
Browser	Microsoft Internet Explorer 7.0 or better Mozilla Firefox 4 or better Safari 5 or better
Windows	Operating System: Windows 8 desktop mode, Windows 7; Windows Vista; Windows XP Service Pack 2 or 3

	x86 or x64 (Browsers must be in 32-bit mode) 1.6-gigahertz (GHz) or higher processor 512MB of RAM
Mac OS	Operating System: Apple Mac OS X 10.4.8 or above Intel Core™ Duo 1.83GHz or faster processor 512MB of RAM

Registration and Fees

ondemand.acmg.net
ACMG Members and ACMG Trainees: ($50)
Non-members ($60)
Additional fee (~$25) applies for NSGC credit that is billed by NSGC.

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