2017 | ACMG Annual Clinical Genetics Meeting

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

Molecular Cytogenics: The Next Generation in Balanced Rearrangement Detection

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: 1.5 hours
Course must be completed by the expiration dates

COURSE DESCRIPTION

Balanced genomic rearrangements have classically been detected in clinical diagnostic laboratories at the DNA level using karyotyping or fluorescence *in situ* hybridization (FISH) and at the RNA level through reverse-transcriptase PCR (RT-PCR) or real-time PCR (qPCR). High-complexity molecular technologies are evolving to complement and/or supplant the current methods in both the constitutional and neoplastic setting. Speakers will describe technical aspects, pros and cons, and applications for the following: whole- genome sequencing, fusion-targeted RNAseq, and microfluidics-based linked-read sequencing.
Dr. Birgit Funke (Merging Clinical Cytogenetic and Molecular Testing), and Dr. Michael Talkowski (Whole-genome Sequencing Methods in Cytogenetics) 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:

- Describe the molecular methods for identifying balanced genomic rearrangements
- Contrast the clinical utility and limitations of current and evolving technologies for balanced genomic rearrangement detection
- Explain current methods of RNA-Seq, and its function in oncology diagnostics
- Differentiate how evolving technologies will answer clinical questions historically answered by conventional cytogenetics

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

- Evolution of Cytogenetics into the Molecular Era- Hutton Kearney, PhD, FACMG
- Whole-genome Sequencing Methods in Cytogenetics: Comparison of Diagnostic Yields in Prenatal and Pediatric Populations- Michael Talkowski, PhD
- Targeted RNA-Seq for Detecting Fusion Transcripts Using Anchored Multiplex PCR- Long Le, MD, PhD
- Merging Clinical Cytogenetic and Molecular Testing- Integrating High Complexity Next Generation Technologies into Cytogenetics Practice- Birgit Funke, PhD, FACMG

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 1.5 *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 1.5 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 1.5 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)
Fuki M. Hisama, MD, FACMG – Horizon Pharmaceuticals, 4; ABMGG, 6 (PC)
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:

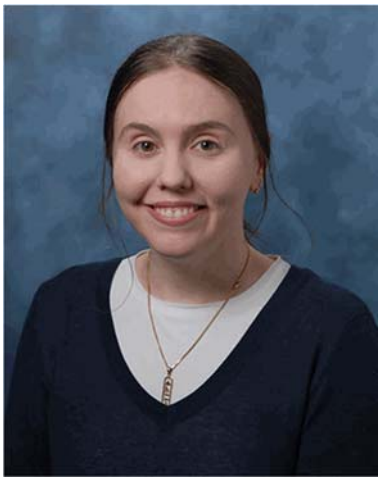
Liming Bao, PhD, FACMG (EC)
Monica A. Giovanni, MS (EC)
Chad Haldeman-Englert, MD, FACMG (EC)
Abbas Padeganeh, PhD, MS (EC)
Amy E. Roberts, MD, FACMG (EC)
Barrie Suskin Kaplan, MD (EC)
Tracey Weiler, PhD, MS (EC)
Jansson White (EC)
Anne M. Slavotinek, MBBS, PhD, FACMG (EC)
Christopher M. Cunniff, MD, FACMG (PC)
Gerald Feldman, MD, PhD, FACMG (PC)
Helga V. Toriello, PhD, FACMG (PC)
Hope Northrup, MD, FACMG (PC)
Jennelle C. Hodge, PhD, FACMG (PC)
Jessica Smith, MD (PC)
Joanne Nguyen, MD, FACMG (PC)
Myra Wick, MD, PhD, FACMG (PC)
Omar Abdul-Rahman, MD, FACMG (PC)
Pilar L. Magoulas, MS, CGC (PC)
Robert Hagelstrom, PhD, MBA, FACMG (PC)
Tuya Pal, MD, FACMG (PC)
Monica Giovanni, MS, CGC (PC)
Jane Dahlroth, CEM, CMP-HC (PC), (EC), (S)
Jane Radford, MHA, CHCP (PC), (EC), (S)
Michael S. Watson, PhD, FACMG (PC), (EC), (S)
Penelope Freire, CMP (PC), (S)

SPEAKERS AND MODERATORS

Faculty Disclosures:

As a sponsor accredited by the ACCME, the American College of Medical Genetics and Genomics must ensure balance, independence, objectivity and scientific rigor in all its sponsored educational activities. All faculty participating in a CME-certified activity are expected to disclose to the audience any relevant financial interest(s) or other relationship(s) with the manufacturer(s) of any commercial product(s), provider(s) of commercial services or any commercial supporters, including diagnostic laboratories, of the activity discussed in an educational presentation. Relevant financial interest(s) or other relationship(s) can include such things as grants or research support, consultancy, major stock holder, etc. The intent of this disclosure is not to prevent a planner or speaker with a relevant financial or other relationship from course planning or making a presentation, but rather to provide learners with information on which they can make their own judgments. It remains for the audience to determine whether the speaker's interests or relationships may influence the presentation with regard to exposition or conclusion. All conflicts of interests have been reviewed and resolved by the education and CME subcommittee.

Moderator: Jennelle Hodge, PhD, FACMG
Indiana University
No financial relationships to disclose.

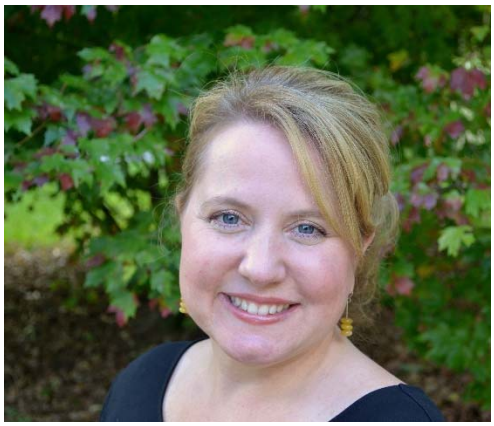


Dr. Hodge has published more than 30 manuscripts related to the genetics of solid tumors (particularly of prostate cancer and uterine neoplasms), mining of genomic data for genotype and phenotype correlation, and the development and implementation of innovative technologies for molecular and cytogenomic clinical diagnostics. She is ABMGG certified in both Clinical Cytogenetics and Clinical Molecular Genetics through the Harvard training program. Dr. Hodge has practiced these clinical specialties for nearly 10 years and is currently a director in Cytogenetics and Molecular Genetics at Indiana University. She is also an Associate Professor in the Department of Medical and Molecular Genetics. Dr. Hodge is a fellow of ACMG and presently a member of the program committee as well as being on the Board of Directors for the Cancer Genomics Consortium. She has produced over 30 abstracts, served as a moderator at ACMG, ASHG and AMP sessions, and reviews manuscripts for 15 human genetics journals.

Moderator: Jianling Ji, MD, MS, FACMG
Assistant Professor, Assistant Director, Children's Hospital Los Angeles, University of Southern California
No financial relationships to disclose.

Dr. Jianling (Jenny) Ji is an Assistant Professor of Clinical Pathology, Keck School of Medicine of the University of Southern California, and assistant director of the Clinical Genomics Laboratory in the Center for Personalized Medicine in the Department of Pathology and Laboratory Medicine, Children's Hospital Los Angeles. Dr. Ji received her M.D. and M.S. degrees at Nantong Medical College of Nantong University and completed her fellowship training in Clinical Molecular Genetics and Clinical Cytogenetics at the University of California Los Angeles (UCLA) and Cedars Sinai Medical Center. She holds American Board of Medical Genetics and Genomics certifications in both Clinical Molecular Genetics and Clinical Cytogenetics. She is a member of ACMGG, ASHG, AMP and CGC. Dr. Ji's primary interest is clinical application of next generation sequencing and microarray technologies in molecular diagnosis of rare genetic disorders and in cancer genomics research

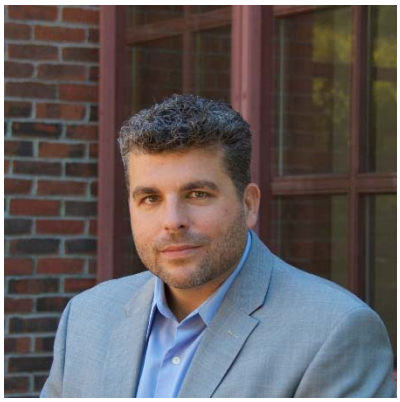
Speaker: Hutton Kearney, PhD, FACMG
Mayo Clinic
Financial relationships to disclose. (Self): Non-remunerative positions of influence such as officer, board member, etc. Company (ies): Illumina



Dr. Kearney leads the Hereditary Genomics section of the newly formed Genomics Laboratory (combined Molecular and Cytogenetics) in the Division of Laboratory Genetics and Genomics at the Mayo Clinic in Rochester, MN. She is ABMGG-certified in both Clinical Cytogenetics and Clinical Molecular Genetics and is a fellow of the American College of Medical Genetics and Genomics. Dr. Kearney is the Cytogenetics section editor of Genetics in Medicine, and the past president of the American Cytogenetics Conference. She serves on the board of directors of the Cancer Genomics Consortium, the Cytogenetics Resource Committee of the College of American Pathologists (CAP), and the Structural Variant Working Group of ClinGen (Clinical Genomics Resource). Dr. Kearney is the past chair of the ACMG

Laboratory Quality Assurance Committee, and has helped to develop numerous ACMG and CLSI guidelines for the practice of cytogenetics, especially as it relates to microarray testing. She is also the founder of the Cytogenomics Array Group and database, CAGdb, an international resource for best practices for interpretation of clinical microarrays (www.cagdb.org). Dr. Kearney is an advocate for rapid clinical adoption of advanced technologies and robust educational programs to support clinical laboratories.

Speaker: Michael Talkowski, PhD
Associate Professor, Massachusetts General Hospital / Harvard Medical School / Broad Institute
No financial relationships to disclose.



Dr. Talkowski is an Associate Professor in the Center for Genomic Medicine at Massachusetts General Hospital (MGH), Harvard Medical School, and is an Associate Member of the Broad Institute. Dr. Talkowski's research program is dedicated to understanding the genetic etiology of human developmental and neuropsychiatric disorders. His group integrates molecular and computational genomics approaches to delineate the functional consequences of genomic variation, with a particular interest in the relationship between genome structure and function. Dr. Talkowski's group has been recognized for introducing whole-genome sequencing approaches into conventional cytogenetic analyses and translational diagnostic applications. Their studies have defined the genomic landscape of balanced chromosomal rearrangements, and discovered relatively balanced forms of highly complex rearrangements that occur in the viable human germline, including extreme examples of chromosome shattering such as 'chromoanagenesis' and 'chromothripsis'. His group recently defined a diverse spectrum of complex structural variation that exist in all human genomes, often mediated by cryptic inversions, and has demonstrated the potential of WGS in prenatal diagnostics. His lab is actively involved in population scale WGS studies to define structural variation in the genome as well as functional genomic studies of recurrent, reciprocal genomic disorders. The Talkowski group is funded by the NIMH, NICHD, NINDS, NGHRI, NIGMS, the Simons Foundation, Nancy Lurie Marks Foundation, March of Dimes, Charles Hood Foundation, and the MGH Research Scholars program.

Speaker: Long Le, MD, PhD
Assistant in Pathology, Massachusetts General Hospital, Assistant Professor of Pathology, Harvard Medical School, Massachusetts General Hospital
Financial relationships to disclose: (Self) Archer DX: Co-Founder, Equity Ownership, Consultant, Hunter Biosciences: Equity Ownership, Boston Lighthouse Innovations: Co-Founder, equity ownership, CSO.

Dr. Le is a molecular pathologist who is currently the Director of Technology Development at the MGH Center for Integrated Diagnostics. His clinical and research focus includes development of novel target enrichment and bioinformatics analysis methods for next-generation sequencing and their application for clinical molecular diagnostics. He has a strong interest in applying big data analytics and machine learning for healthcare.

Speaker: Birgit Funke, PhD, FACMG
Laboratory for Molecular Medicine, Partners HealthCare
Financial relationships to disclose: (Self) Consultant for SeraCare LifeSciences. Employment: Department of Pathology, Harvard Medical School/MGH and Veritas Genetics

Associate Professor of Pathology Director Clinical R&D Laboratory for Molecular Medicine

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.

Questions regarding CE credit should be directed to education@acmg.net.

7101 Wisconsin Avenue, Suite 1101 | Bethesda, MD 20814
Telephone: 301-718-9603 | Fax: 301-718-9604 | E-mail: education@acmg.net | Website: www.acmg.net/education