2017 | ACMG Annual Clinical Genetics Meeting

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

# Whole genome and Whole Exome Sequencing for 'Healthy' Individuals in Clinical Practice: Are We Up to the Challenge?

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

Genomic sequencing (whole genome and whole exome) has entered routine clinical practice for the diagnosis of genetic disorders. The rapidly diminishing costs of such testing have also encouraged individuals who view themselves as healthy to pursue screening through genomic sequencing. Sequencing as screening is already being advocated by many clinicians in the wellness community. And, building upon the popularity of direct-to-consumer testing with SNP-based arrays, direct-to-consumer sequencing is already being pursued by several companies.

Using genomic sequencing to screen ostensibly healthy individuals presents the health care system and its stakeholders with interesting opportunities and challenges, in part because there is so little empirical evidence available. This session will present perspectives and data from early experiences in this setting. Speakers will discuss the benefits, risks and costs of utilizing genomic sequencing for screening, including the role of genetic counseling, types of genomic results returned, patient acceptance, policy and regulatory implications. The use of family history and the issue of reanalysis in light of new symptom development will also be addressed. This session will provide early data to help fill the knowledge-gap for clinical geneticists, counselors and laboratory directors in this rapidly developing area of practice.

Dr. Leslie Biesecker (Returning Results to Healthy ClinSeq Participants: Genomics is Just Medicine After All) declined to release his slides but you will be able to listen to his presentation.

## LEARNING OBJECTIVES

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

- Explain similarities and differences between a diagnostic genomic test and a genomic ‘screening’ test of ‘healthy’ individuals

- Describe the types of genomic results (e.g. pharmacogenetics) that are generally reported in genomic testing of 'healthy' individuals
- Describe counseling issues that uniquely arise in genomic testing of 'healthy' individuals
- List challenges to the health care system posed by genomic testing of 'healthy' individuals

# 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

- Lessons Learned from the MedSeq Project Sequencing 'Healthy' Individuals-Jason Vassy, MD, MPH, SM
- Lessons Learned From More Than 1000 Healthy Genomes- Erica Ramos, MS, LCGC
- Genomic Sequencing for 'Healthy' Individuals in Clinical Practice: The Baylor Miraca Genetics Laboratory Experience with Healthy Exome Sequencing- Christine Eng, MD
- Whole Genome Sequencing for 'Healthy' Individuals in Clinical Practice: the HudsonAlpha Experience- David Bick, MD, FACMG
- Genomic Counseling for the Diagnostic and the 'Healthy' Patient: Similarities and Differentiators – Kelly East, MS, CGC
- Returning Results to Healthy ClinSeq Participants: Genomics is Just Medicine After All- Leslie Biesecker, MD, 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<br>2. Grant/Research Support (External)<br>3. Salary/Employment/Royalty(ies)/Honoraria<br>4. Consultant/consulting fees/other remuneration<br>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)<br>7. Receipt of intellectual property<br>8. Other |

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Karen W. Gripp, MD, FACMG – Wiley Publishing Inc., 3; FDNA, 4; Novartis, 4 (PC)
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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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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/Speaker: Jason Vassy, MD, MPH, SM**
**Assistant Professor of Medicine, Harvard Medical School, VA Boston Healthcare System, Brigham and Women's Hospital, and Harvard Medical School**
**No financial relationships to disclose.**

Dr. Vassy is a primary care physician and clinician-investigator at the VA Boston Healthcare System and at Brigham and Women's Hospital and is an Assistant Professor of Medicine at Harvard Medical School. He is a member of the Genomes2People research program and Brigham and Women's Hospital, and his research focuses on the clinical application of genetic and genomic testing in primary care settings. He is a co-investigator of the MedSeq Project, a randomized trial of genome sequencing among ostensibly healthy adults.

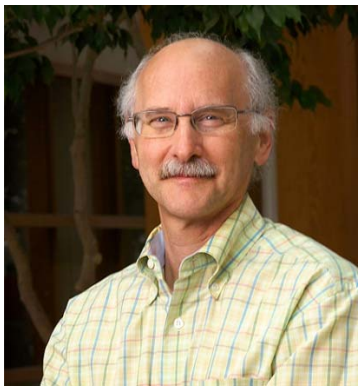
**Speaker: Erica Ramos, MS, LCGC**
**Clinical Head, Healthy Genome Initiatives, Illumina**
**Financial relationships to disclose: (Self) Employee of and shareholder of Illumina, Inc.**

Erica Ramos, MS, LCGC is the Clinical Head for Illumina's "Healthy Genome Initiatives" team and President-Elect for the National Society of Genetic Counselors, the leading professional organization for genetic counselors more than 3500 members strong. She received her Master's in genetic counseling from the University of California, Irvine in 2001. After graduation, she spent 11 years as a clinical genetic counselor, first in prenatal and preconceptional settings and then in cancer and adult-onset disorders. In 2012, Ms. Ramos joined Illumina in the Illumina Clinical Services Laboratory, where she managed the genetic counseling and variant curation teams that implemented the interpretation and reporting process for whole genome sequencing in healthy individuals and launched the lab's undiagnosed disease offering. In her current role, she is focused on advancing the use of whole genome sequencing in clinical care by providing access and education to healthy individuals through the Understand Your Genome programs, MyGenome visualization software and other research and collaborations. Her focus on education extends to genetic counseling students and she is an Adjunct Professor in the Genetic Counseling Department at Augustana University. She also tweets from @ERamosSD about genomics and genetic counseling, music, travel and Wisconsin sports, among other interests and sometimes her employer trusts her enough to live-tweet genomics events from @IlluminaLive.

**Speaker: Christine Eng, MD**
**Baylor Genetics Laboratories and Department of Molecular and Human Genetics, Baylor College of Medicine, Houston, TX**
**No financial relationships to disclose.**

Christine M Eng, MD, is a Professor in the Department of Molecular and Human Genetics at the Baylor College of Medicine in Houston, Texas. Dr. Eng is the Director of the Storage Disorders Clinic at Texas Children's Hospital and also serves as Medical Director and Acting Laboratory Director of the Baylor DNA Diagnostic Laboratory. Following undergraduate studies at Yale University, she earned a medical degree from Tulane University School of Medicine, New Orleans. She completed residency training in pediatrics at Long Island Jewish Medical Center, New Hyde Park, New York, followed by fellowship training in medical genetics at Mount Sinai School of Medicine in New York City. Dr. Eng is board certified in both pediatrics and medical genetics.

**Moderator/Speaker: David Bick, MD, FACMG**
**Faculty Investigator, HudsonAlpha Institute for Biotechnology, HudsonAlpha Institute for Biotechnology**
**Financial relationships to disclose: (Self): Major stockholder/ownership interest in Envision Genomics, Smith Family Clinic, and Clinical Services Lab.**



Clinical Genetics and Genomics David Bick, MD, is the chief medical officer and a faculty investigator at the HudsonAlpha Institute for Biotechnology, the medical director of the Smith Family Clinic for Genomic Medicine and a laboratory director in the Clinical Services Laboratory. He comes to HudsonAlpha from the Medical College of Wisconsin where he was professor in the department of pediatrics and the department of obstetrics and gynecology. At the Medical College of Wisconsin, he was the director of the Clinical Sequencing Laboratory at Medical College of Wisconsin; director of the Advanced Genomics Laboratory at Children's Hospital of Wisconsin; medical director of the Genetics Clinic at Children's Hospital of Wisconsin; and chief of the division of genetics in the department of pediatrics at Medical College of Wisconsin. Bick received his medical degree from George Washington University School of Medicine in 1981 and completed his residency in pediatrics at Yale-New Haven Hospital in New Haven, Conn. At the Yale University School of Medicine, Bick completed a fellowship in human genetics and pediatrics in 1986, followed by a postdoctoral research fellowship in human genetics in 1987. Bick is board-certified in pediatrics, clinical genetics and clinical molecular genetics. As a leader in the field of genomic medicine, Bick has published numerous peer-reviewed articles, chapters and reviews. Bick's laboratories at the Medical College of Wisconsin and Children's Hospital of Wisconsin were the first in the world to offer whole genome sequencing as a clinical test.

**Speaker: Kelly East, MS, CGC**
**Clinical Applications Lead, HudsonAlpha Institute for Biotechnology**
**No financial relationships to disclose.**

Kelly East is a board-certified genetic counselor and Huntsville native who joined HudsonAlpha and the Educational Outreach team in 2008. Kelly oversees and participates in the provision of genetic and genomic counseling for research projects and clinical services at HudsonAlpha and leads the development of educational experiences and resources for healthcare providers, trainees and patients. In addition, she has prior clinical experience as a genetic counselor in an oncology setting. She received a Bachelor of Science in Microbiology from Auburn University and a Master of Science in Genetic Counseling from the University of North Carolina – Greensboro.

**Speaker: Leslie Biesecker, MD, FACMG**
**Chief and Senior Investigator, National Human Genome Research Institute**
**Financial relationships to disclose: (Self): Honoraria from Wiley-Blackwell**

Dr. Biesecker is a clinical and molecular geneticist and Chief of the Medical Genomics and Metabolic Branch and Director of the Physician-Scientist Development Program at the National Human Genome Research Institute of the National Institutes of Health, which he joined in 1993. He uses genetic and genomic technologies to study the etiology of genetic disorders and has published nearly 300 primary research articles, reviews, and chapters. He received his medical training at the Univ. of Illinois, Pediatrics at the Univ. of Wisconsin, and Clinical and Molecular Genetics at the Univ. of Michigan. His laboratory has elucidated the etiology and natural history of numerous diseases. In addition, he developed the ClinSeq® program, which began clinical genomics research in 2006, before the widespread availability of next generation sequencing. He co-directs a CLIA-certified molecular diagnostic laboratory within NHGRI. Dr. Biesecker serves as an editor or board member for several biomedical journals, was a member of the board of directors for the American Society of Human Genetics, is an advisor to the Illumina Corporation, and served on the advisory panels for the World Trade Center and Hurricane Katrina victim identification efforts. He was recently elected to the National Academy of Medicine of the National Academy of Science.

# 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.
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| Stream Requirements | |
|---|---|
| Network | For best results, use a hardwired network connection instead of wireless |
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| Refresh Browser Window | If the webcast freezes and does not recover in 3-4 seconds, refresh browser window |
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| Mobile Viewing Requirements | |
|---|---|
| **Android Devices** | Android 2.3+ with Adobe Flash Player 10.2 or better installed<br>Install Flash Player |
| **Apple Devices** | iOS 4+ |

| Online Viewing Requirements | |
|---|---|
| **Bandwidth** | 512kbps |

| | |
|---|---|
| **Required Hardware and Software** | Screen resolution of 1024X768 or larger<br>Sound card and speakers/headphones |
| **Browser** | Microsoft Internet Explorer 7.0 or better<br>Mozilla Firefox 4 or better<br>Safari 5 or better |
| **Windows** | Operating System: Windows 8 desktop mode, Windows 7; Windows Vista; Windows XP Service Pack 2 or 3<br>x86 or x64 (Browsers must be in 32-bit mode) 1.6-gigahertz (GHz) or higher processor<br>512MB of RAM |
| **Mac OS** | Operating System: Apple Mac OS X 10.4.8 or above<br>Intel Core™ Duo 1.83GHz or faster processor<br>512MB of RAM |

# Registration and Fees

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ACMG Members and ACMG Trainees: ($50)
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7101 Wisconsin Avenue, Suite 1101 | Bethesda, MD 20814
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