2016 ACMG Annual Clinical Genetics Meeting – Recordings

from the 2016 ACME Clinical Genetics Meeting held March 8 – 12, 2016 in Tampa, Florida

Date of Release: March 15, 2016
Expiration Date: March 15, 2018
Estimate Time of Completion: 61.5 hours (includes all 3 Short Courses)
www.prolibraries.com/acmg

Purpose

The 2016 Annual Clinical Genetics Meeting Recordings offer both research and clinical topics that promote the science and the practice of clinical genetics and genomics. Sessions focus on the latest discoveries of the etiology and the pathogenesis of genetic disorders, the latest developments in genetic testing and screening, the laboratory's role in the diagnosis of genetic disorders, the treatment of genetic disorders in children and adults, the delivery of genetic services and more.

Audience

- Medical and clinical geneticists; genetic counselors; pediatric, obstetric, and maternal-fetal specialists; and all medical practitioners who are providing comprehensive diagnostic, management, and counseling services for patients with, or at risk for, genetically influenced health problems
- Laboratory directors and technicians who conduct genetic testing
- Researchers involved in the discovery of genetic disorders and treatments
- Any healthcare and public health professionals who have an in interest medical and clinical genetics and genomics

Learning Objectives

After participating in the 2016 ACMG Annual Meeting Recordings, participants should be able to:

- Describe the latest advanced in the field of Cancer Genomics
- Distinguish advantages and disadvantages between various methodologies for optimal patient care
- Describe rare known genetic conditions diagnosed on prenatal ultrasound
- Assist in the differential diagnoses of unknown cases
- Recognize cardinal clinical signs and symptoms of specific rare syndromic conditions
- Identify the appropriate molecular diagnostic testing strategies
- Review the application and significance of variants found on molecular results
- Describe the clinical manifestations of neurogenetic diseases
- Identify available genetic testing, screening, prevention and treatment options for neurogenetic diseases
- Describe the issues surrounding newborn screening for lysosomal storage disorders
- Critically assess the value of pharmacogenetics information in patient care

Formats

Short Courses

- Advanced Molecular Cancer Genetics: State of the Art Today and Beyond
- NAMA at the ACMG: The Best of the North American Metabolic Academy
- Tools and Approaches to Assess the Genetic Basis of Disease

Short Courses are 4.5 hour long sessions that delve into a specific topic.

Plenary and Concurrent Sessions

These sessions have been developed based on proposals from members and nonmembers of ACMG and the Program Committee. The sessions highlight a wide range of topics of interest to genetics practitioners, researchers, laboratory directors and technicians, and counselors.

Platform Presentations

ACMG's Program Committee has assembled abstract-driven platform talks totaling 60 presentations. Each presenting author will give a 10-minute talk followed by 5 minutes of discussion. Abstracts can be accessed from the Search function at www.acmgmeeting.net.

System Requirements

This website utilizes some of the latest advancements in web technologies. It is recommended that end users' devices satisfy the following requirements to take advantage of everything this website has to offer.

- OS: Windows, Mac
- Browser: Internet Explorer 7 or higher, Firefox, Chrome, Safari
- Javascript & Cookies enabled
- Adobe Flash Player 9.124.0 or higher
- Recommended RAM: 512MB+

Mobile support

- Apple iPad/iPhone running HTML5
- Android Device running Flash

Technical Support: 877-796-1325 • Other Inquiries: 800-679-3646

Registration and Fees ***www.prolibraries.com/acmg***

Item	Price	Onsite Attendee	Other Registrant	Additional Price for (CME,NSGC,P.A.C.E.)
2016 Complete Online Library –Includes unlimited online access to the 2016 ACMG meeting sessions. Does NOT include the short courses	$179.00	N/A	N/A	$35.00
Short Courses Ordered Separately				
Short Course: Advanced Molecular Cancer Genetics: State of the Art Today and Beyond		$139.00	$169.00	$25.00
Short Course: NAMA at the ACMG: The Best of the North American Metabolic Academy		$139.00	$169.00	$25.00
Short Course: Tools and Approaches to Assess the Genetic Basis of Disease		$139.00	$169.00	$25.00

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Educational Credits

*Continuing Medical Education (CME)**

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 enduring activity for a maximum of 61.5 *AMA PRA Category 1 Credits*™. Physicians should claim only the credit commensurate with the extent of their participation in the activity.

*Laboratory Directors and Laboratory Personnel**

***ACMG* is approved as a provider of continuing education programs in the clinical laboratory sciences by the ASCLS P.A.C.E. ® Program.** ACMG designates the 2016 ACMG Annual Clinical Genetics Meeting Recordings for a maximum 61.5 contact hours.

ACMG is approved by the Florida Board of Clinical Laboratory Personnel as CE Provider #50-11878. CE Broker Tracking #20-526363

ACMG is approved by the California Department of Health Services through the ASCLS P.A.C.E.® Program as CE Provider # 275-101-16. California Accrediting Agency number 0001.

*Genetic Counselor Credit**

The National Society of Genetic Counselors (NSGC) has authorized the American College of Medical Genetics and Genomics (ACMG) to offer up to 6.15 CEUs or 61.50 Category 1 contact hours for the activity: *2016 ACMG Annual Clinical Genetics Meeting - Recordings*. The American Board of Genetic Counseling (ABGC) will accept CEUs earned at this program for the purposes of genetic counselor certification and recertification.

CEU certificates for genetic counselors will be emailed by NSGC quarterly. ACMG must send a report to NSGC of those who have Completed All the Desired Sessions and Have Completed the evaluations and tests.

When You Are Ready to Claim All Final Credits You Must Send an Email to EDUCATION@ACMG.NET and let us know you are ready for us to report your credits to NSGC. You cannot claim credits in one month and then claim additional credits in later months. The total credits claimed must be submitted at one time.

You may access the course anytime but you can only claim credits once. Do **NOT** send an email to ACMG until you have completed all the sessions that you want to claim credits.

Sending an email triggers ACMG to add your name and number of credits claimed to the NSGC quarterly reporting list. We can only report credits One-Time for a participant. So it is Very Important that you send an email to us notifying us that you are ready for us to send your name, credits claimed, evaluation and tests to NSGC. If you try to submit more than one time to us we; 1) cannot guarantee that NSGC will accept your new credits claimed, and 2) there will be an additional charge of at least $25.00.

How to Claim Educational Credits

Each session will have 1-2 test question(s) followed by an evaluation. You must pass the test (90%) and complete the evaluation with each session. You will then be able to claim your credits for that session.

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.

Disclosure Statement

In accordance with the Accreditation Council for Continuing Medical Education (ACCME) and the policy of the American College of Medical Genetics and Genomics (ACMG), speakers and moderators presenting in sessions that offer CME and CEU credit must disclose, prior to the start of their presentations, the existence of any financial interest and/or other relationship(s) they might have with the manufacturer(s) or provider(s) of any commercial product(s) or service(s), including diagnostic laboratories to be discussed during their presentation. These disclosures are included in the Disclosure section of this program guide.

Speakers with disclosures are required to provide a slide that states any relevant financial relationships (or lack thereof), and to state these to the audience at the start of their presentation. All disclosures have been peer reviewed by the ACME Education Committee.

Diagnostic and Testing Labs and Commercial Interest

ACMG CME activities adhere to the Accreditation Council for Continuing Medical Education (ACCME) Standards for Commercial Support. The ACCME has defined under its conflict of interest definition the types of industry interests that must be excluded from CME content delivery. This largely applies to manufacturers of pharmaceuticals and devices. Diagnostic laboratories, whether not-for-profit or for-profit, that are not owned by device manufacturers are exempted from this CME requirement. However, they are still expected to be fair and balanced in their presentations

HIPAA Compliance by Speakers and Presenters

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 upon request.

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.

***Note: Credit hours are calculated inclusive of 4.5 credits for participating in a short course. If you did not purchase a short course then the number of credit hours you claim will be less than posted**

Session Number	Short Courses	Credit Hours (CME, NSGC, P.A.C.E.)
273	NAMA at the ACMG: The Best of the North American Metabolic Academy	4.5
329	Advanced Molecular Cancer Genetics: State of the Art Today and Beyond This activity is supported by an unrestricted educational grant from Illumina	4.5
491	Tools and Approaches to Assess the Genetic Basis of Disease	4.5
Session Number	**2016 Complete Online Library Includes**	
231	Cardinal Signs of Selected Syndromes	2
256	Reproductive Genetics: Implications of the Latest Technologies	2
359	Managing Positive Newborn Screens for Lysosomal Storage Disorders	2
212	Loss, Gain, and Things that Look the Same: Maximizing the Clinical Utility of Areas of Homozygosity	1.5
339	From Gene Discovery to Gene Therapy: Duchenne Muscular Dystrophy and Other Neuromuscular Disorders	1.5
484	From Guidelines to Clinic: Incorporating Pharmacogenetics into Clinical Practice and the Electronic Health Record	1.5
994	Presidential Plenary - The Practice of Medical Genetics: Myths and Realities Presentation of the 2016 ACMG Foundation and March of Dimes Awards	1.5
1007	Platform Presentations - Biochemical and Therapies	2
1008	Platform Presentations - Cancer Genetics	2
1009	Platform Presentations - Whole Exome Sequencing	2
224	March of Dimes Clinical Genetics Conference - Prader-Willi Syndrome – New Insight into a Classic Genetic Disorder	2.5
217	Pro/Con: The Debate Around Direct-Access-Testing Robert C. Baumiller Symposium	1.5
573	Adult Genetics Diagnostic Dilemmas (Unknowns and Rare Knowns)	1.5
615	Prenatal Diagnostic Dilemmas	1.5
738	The Right Test for the Right Patient at the Right Time: Working with Your Institution to Make Genetic Testing Useful	1.5
897	Platform Presentations - Genetic Testing and Insurance Issues	1
898	Platform Presentations - Prenatal/Perinatal	1

899	Platform Presentations - Neurogentic Disorders and Whole Exome Sequencing	1
355	Cancer Prevention and Screening for Children at Hereditable Risk	1.5
438	Classification and Genetics of Limb Deficiency Disorders	1.5
443	Classic and New Approaches to Variant Interpretation in the Genomic Era	1.5
968	Platform Presentations - New Causes for Intellectual Disability/Neurodevelopmental Disorders	2
969	Platform Presentations - Whole Exome Sequencing	2
970	Platform Presentations - Cytogenetics	2
210	Whole Genome Analysis or Whole Exome Sequencing of Newborn Dried Blood Spot DNA for Medically Actionable Neurometabolic Disorders - R. Rodney Howell Symposium	2
266	More Than Skin Deep: Clinical Presentation, Diagnosis, and Treatment of Inherited Skin Disorders	2
342	Rising to the Level of Clinical Importance: Challenges to Adjudicating and Reporting Variants Discovered with Genomic Analysis	2
995	Closing Plenary Session: Therapies for Genetic Diseases: New Options	2
	TOTAL CREDITS INCLUDING ALL 3 SHORT COURSES	**61.5**

Financial Disclosures

The American College of Medical Genetics and Genomics has implemented a process where everyone who is in a position to influence and/or control the content of a continuing education activity must disclose all relevant financial relationships with any commercial interest and any conflicts must be resolved prior to the activity. Participants of educational programs must be informed of an organizer's and or a presenter's academic and professional affiliations and existence of any relevant financial relationship a presenter has with any proprietary entity producing heath care goods or services consumed by, or used on patients, with the exemption of non-profit or government organization and non-health care related companies. The intent of this disclosure is not to prevent a speaker from making a presentation. This policy allows the listener/attendee to be fully knowledgeable in evaluating the information being presented.

Disclosure includes any relationship that may potentially bias the planning of the continuing education activity or may potentially bias one's presentation or which, if known, could give the perception of bias. These situations may include but are not limited to:

Legend

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
7. Receipt of intellectual property
8. Other

Following is an alphabetical list of scientific session speakers, moderators and platform presenters who have relationships to disclose:

Alamillo, Christina - Ambry Genetics, 3
Arnold, Georgianne - Biomarin, 2, 4; Hyperion, 2
Bean, Lora - Emory Genetics Laboratory, 3
Bennett, Melissa - eviCore healthcare, 3
Bianchi, Diana - Illumina, 2, 4
Biesecker, Leslie - Illumina Corp, 6
Birsoy, Ozge - Mount Sinai Genetic Testing Laboratory, 3; Partners HealthCare Personalized Medicine, 3
Burrow, T. Andrew - Alexion, 2; Genzyme, 2, 4, 5
Burton, Barbara - Alexion, 2, 4; Armagen, 2; Biomarin, 2, 4; Cytonet, 2; Genzyme, 4; ReGenX Bio, 4; Shire, 2, 4; Ultragenyx, 2
Chikarmane, Rashmi - GeneDx, 3
Conta, Jessie - EviCore, 2
Davidson, Beverly - Sarepta Therapeutics, 8; Spark Therapeutics, Inc., 8; Wave Life Sciences, 8
Dhar, Shweta - PanGenomics, 1
Dolinsky, Jill - Ambry Genetics, 3

Elsea, Sarah - Baylor Miraca Genetics Laboratories, 3; Smith-Magenis Syndrome Research Foundation, 2
Escolar, Maria - Glaxo Smith Klein, 2; Shire Human Genetics, 2; Synageva, 2
Everman, David - Greenwood Genetic Center (salaried employee of this non-profit entity that offers clinical diagnostic testing for condition to be discussed - may not be a conflict but wanted to list in interest of full disclosure), 3; Oxford University Press (co-editor and contributing author of textbook that includes information about topic to be discussed - may not be a conflict but wanted to list in interest of full disclosure), 3
Funke, Birgit - 23and Me, 4
Geraghty, Michael T. - Biomarin, 2
Grange, Dorothy - Biomarin Pharmceuticals, Inc, 2; Edimer Pharmaceuticals Inc, 2
Green, Robert - AIA, 3; Arivale, 3; DOD, 2; Helix, 3; Illumina, Inc., 2, 3; Invitae, 3; NIH, 2; Prudential, 4; Roche, 3
Gripp, Karen – FDNA, 4
Hamosh, Ada - Blade Therapeutics, 3; GlaxoSmithKline, 4
Harel, Tamar - Baylor Miraca Genetics Laboratories, 8
Helbig, Katherine - Ambry Genetics, 3
Henderson, Lindsay - GeneDx, 3
Hisama, Fuki - Baxter, 2
Hooker, Gillian - NextGxDx, 3
Howell, R. Rodney - Baebies Company, 4
Jarvik, Gail - ActX, 6
Juusola, Jane - GeneDx, Inc., 3
Kishnani, Priya - Genzyme, PTC Therapeutics, Baebies Inc, Roche, Alexion Pharmaceuticals Inc, Amicus Therapeutics, Shire Pharmaceuticals, 4
Klein, Teri - Personalis Inc, 4
Korf, Bruce - Illumina, Accolade, 4; Novartis, 2; Wiley, Elsevier, 3
Lazarin, Gabriel - Counsyl, 3
Lincoln, Stephen - Invitae, 3
Liu, Pengfei - Baylor Miraca Genetics Laboratories, 8
Monaghan, Kristin - GeneDx, 3
Parad, Richard - Parabase Genomics, 4
Plon, Sharon - Baylor Miraca Genetics Laboratories, 8
Ready, Kaylene - Counsyl, 3
Rehm, Heidi - Partners Healthcare, 3
Richard, Gabriele - GeneDx, 3
Schaaf, Christian - Springer, 3
Schneider, Judsen - NextGxDx, Inc, 3
Schwartz, Stuart - Laboratory Corporation of America, 3
Shen, Jun - Counsyl, 4; Laboratory Corporation of America, 4; Miti Genomics, 6; Partners HealthCare Personalized Medicine, Laboratory for Molecular Medicine, 3
Simpson, Joe Leigh - Elsevier, 3; Illumina, 4; March of Dimes, 3; Reproductive Genetic Innovations, 3
Sparks, Susan - Genzyme, a Sanofi company, 3
Steiner, Robert - VeritasGenetics, 4
Sternen, Darci - Seattle Children's Hospital/Pediatric Laboratory Utilization Guidance Services, 3
Stosser, Mary Beth - GeneDx, 3
Walkiewicz, Magdalena - Baylor Miraca Genetics Laboratories, 3
Wentzensen, Ingrid - GeneDx, 3

White, Amy - Mayo Clinic Department of Laboratory Medicine Biochemical Genetics Laboratory, 3
Wolff, Daynna - Cancer Genomics Consortium, 6; Medical University of South Carolina, 3
Yang, Yaping - Baylor Miraca Genetic Laboratories, 3
Yates, Carin - GeneDx, 3
Ziats, Mark Creative Bioinformatics, Inc, 1

Following is an alphabetical list of scientific session speakers, moderators, and platform presentations who had no relationships to disclose:

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Perez, Melissa
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Members of the ACMG Staff, Education and Program Committees involved in planning the 2016 ACMG Annual Clinical Genetics Meeting are required to indicate relationships which could be perceived by some as a real or apparent conflict of interest in planning the invited concurrent sessions at the beginning of the planning process (June 2015). In these cases, the committee members refrained from the discussion. If listed above as a speaker or author then those discloses relate to their role as such. All disclosures have been reviewed by the Education Committee or the Executive Director and CME Officer and conflicts of interest are disclosed.

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. Stock options or bond holdings in a for-profit corporation or self-directed pension plan
2. Research and/or educational grants
3. Employment (full- or part-time)
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6. Non-remunerative positions of influence such as officer, board member, trustee, or public spokesperson
7. Receipt of royalties
8. Speakers' bureau
9. Other

Following is an alphabetical list of Program and Education Committee Members who have relationships to disclose:

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Georgianne Arnold, MD, FACMG – Biomarin, 2,4, Hpyperion, NIH, 2 (PC)
Thomas E. Mullen, PhD, MS, FACMG, Good Start Genetics Inc, 3 (EC)Hope Northrup, MD, FACMG – Biomarin and Novartis Pharmaceuticals, 2 (PC)
Anne M. Slavotinek, MBBS, PhD, FACMG, 2, National Eye Institute and National Institutes of Health, 5, Oxford University Press and UptoDate (EC)Helga V. Toriello, PhD, FACMG - Oxford U. Press, 7 (PC)
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Mark N. Ziats, MD, PhD, Creative Bioinformatics, Inc, 1,4 (EC)

Following is an alphabetical list of Program and Education Committee Members who do not have relationships to disclose:

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