



2018 | ACMG Annual
Clinical Genetics Meeting
April 10–14 | Charlotte, NC



Variant Interpretation from the Clinician Perspective

Held in Charlotte, North Carolina

Date of Release: April 16, 2018

Expiration Date: April 15, 2020 (CME, General CEU's, NSGC, P.A.C.E.®)

Estimate Time of Completion: 4.5 hours

Course must be completed by the expiration date

COURSE DESCRIPTION

This short course will explore clinical laboratory reports for next generation sequencing from either panels or whole exome sequencing and focus on the variant interpretation. The session will be aimed at helping clinical health care providers in genetics better understand the reports they receive, participate in variant interpretation, apply the clinical phenotype to variant interpretation, and participate in re-analysis of variants at future dates to apply new information to the interpretation and diagnosis. The data that laboratories use to interpret variants will be reviewed and when applicable, demonstrated. The session will review the ACMG Standards and guidelines for the interpretation of sequence variants in depth. In addition the session will demonstrate how different databases are utilized in variant interpretation and provide the participants with the skills and knowledge needed to obtain and utilize this data.

LEARNING OBJECTIVES

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

- Explore how clinical phenotype information is used to improve variant interpretation by the laboratory, when provided prior to genetic testing, and by the clinician after receiving genetic test results.
- Explore the ACMG Standards and Guidelines for the interpretation of sequence variants.
- Describe how population frequency is used in variant interpretation.
- Describe how in-silico predictions; disease/gene specific databases; and mutational spectrum are used in variant interpretation.
- Develop familiarity with ClinVar resources for resolving variant interpretation discrepancies.
- Explain efforts to refine the utilization of ACMG/AMP interpretation guidelines from experience with discrepancy resolution.
- Explain how ClinVar can be used to help clinical geneticist and counselors



- Search for variants in ClinVar and describe the mutational spectrum reported for specific genes.
- Develop skills to utilize the resources of ClinVar to learn about specific genes, variants and interpretation of variants.
- Develop familiarity with the resources provided by ClinGen

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; 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
- Medical students, undergraduate and graduate students of the biomedical sciences, and genetic counseling students.
- Healthcare and public health professionals who have an interest in medical and clinical genetics and genomics, delivery of genetics services, or implementation of genomic medicine in the health care system.

SESSIONS

- Variant Interpretation from the Clinician- *Darrel Waggoner, MD, FACMG*
- Recommendations for ACMG-AMP Guideline Specification- *Steven Harrison, PhD*
- ClinVar/ClinGen- *Erin Riggs, MS, CGC*
- ClinVar/ClinGen- *Christa Lese Martin, PhD, FACMG*



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SPEAKERS AND MODERATORS

Steven M. Harrison, PhD

Harvard Medical School, Partners Laboratory for
Molecular Medicine

Erin Rooney Riggs, MS, CGC

Geisinger

**Speaker/Moderator: Christa Lese Martin, PhD,
FACMG**

Geisinger

**Speaker/Moderator: Darrel Waggoner, MD,
FACMG**

University of Chicago

EDUCATIONAL CREDIT

Accreditation

The American College of Medical Genetics 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 4.5 *AMA PRA Category 1 Credits™*. Physicians should claim only the credit commensurate with the extent of their participation in the activity.

CE (Continuing Education for non-physicians): This activity has been approved for CE credits.

*The certificate is accepted by the ABMGG for certification.

Genetic Counselor Credit

The National Society of Genetic Counselors (NSGC) has authorized American College of Medical Genetics and Genomics to offer up to 4.5 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. ACMG reports your passing test score and evaluation to NSGC twice a year, the last days of June and December. NSGC processes the December reports in early January. 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 OnDemand course for a maximum of 4.5 contact hours. ACMG is approved by the Florida Board of Clinical Laboratory Personnel as CE Provider #50-11878. This course is registered #20-642806 with CEBroker. ACMG is approved by the California Department of Health Services through the ASCLS P.A.C.E.® Program as CE Provider # 0001.



Disclosure Statement

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 within the past 12 months 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 health 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 participant to be fully knowledgeable in evaluating the information being presented. All disclosures have been reviewed by the Education Committee COI sub-committee or the Executive Director and CME Associate Director and conflicts of interest are disclosed. Disclosure includes any relevant 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.

ACMG Conflict of Interest Policy

The American College of Medical Genetics and Genomics (ACMG) is accredited by the Accreditation Council for Continuing Medical Education (ACCME) to provide continuing medical education for physicians. All educational programming is developed and must be presented in compliance with all ACCME accreditation requirements.

As a provider accredited by the Accreditation Council for Continuing Medical Education (ACCME), the American College of Medical Genetics and Genomics must insure balance, independence, objectivity, and scientific rigor in all educational activities. Planning must be free of the influence or control of a commercial entity, and promote improvements or quality in healthcare. All scientific research used to support patient care recommendations must conform to generally accepted standards of experimental design, data collection, and analysis. In accordance with the ACCME Standards for Commercial Support, everyone (speakers, moderators, oral abstract presenters, all authors, committee members, and staff) who are in a position to control the content of an educational activity certified for AMA PRA Category 1 Credit™ must disclose all relevant financial relationships with any commercial interests within the past 12 months. Individuals who do not disclose are disqualified from participating in a CME activity. Should the speaker's presentation include discussion of any off-labeled/investigational use of a commercial product, he/she is also required to disclose this to the activity participants.

Conflict of interest is created when individuals have both a financial relationship with a commercial interest and the opportunity to affect the content of CME about the products or services of that commercial interest. The potential for maintaining or increasing the value of the financial relationship with the commercial interest creates an incentive to influence the content of the CME—an incentive to insert commercial bias. This may include receiving a salary, royalty, intellectual property rights, consulting fee, honoraria, ownership interest (e.g. stocks, stock options or other ownership interest, excluding diversified mutual funds), or other financial benefit. Financial benefits are usually associated



with roles such as employment, management position, independent contractor (including contracted research and clinical trials), consulting, speaking and teaching, membership on advisory committees or review panels, board membership, and other activities for which remuneration is received or expected.

A Commercial interest is any entity producing, marketing, re-selling, or distributing health care goods or services consumed by, or used on, patients. The ACCME does not consider providers of clinical service directly to patients to be commercial interests.

Relevant financial relationships as defined by the ACCME requires anyone in control of CME content to disclose relevant financial relationships to the accredited provider. Individuals must also include in their disclosure the relevant financial relationships of a spouse or partner. The ACCME defines relevant financial relationships as financial relationships in any amount that create a conflict of interest and that occurred in the twelve-month period preceding the time that the individual was asked to assume a role controlling content of the CME activity. The ACCME has not set a minimal dollar amount—any amount, regardless of how small, creates the incentive to maintain or increase the value of the relationship. Financial relationships are those relationships in which the individual benefits by receiving a salary, royalty, intellectual property rights, consulting fee, honoraria for promotional speakers' bureau, ownership interest (e.g., stocks, stock options or other ownership interest, excluding diversified mutual funds), or other financial benefit. Financial benefits are usually associated with roles such as employment, management position, independent contractor (including contracted research), consulting, speaking and teaching, membership on advisory committees or review panels, board membership, and other activities from which remuneration is received, or expected. ACMG will identify, review and resolve all conflicts of interest prior to an educational activity being delivered to learners.

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.

HIPAA Compliance by Faculty

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.

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



PRESENTERS, PROGRAM AND EDUCATION COMMITTEE 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 health 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. 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.

Disclosure includes any relevant 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. Employment/Salary
2. Consultancy
3. Ownership interests (including stock options) in a start-up company, the stock of which is not publicly traded
4. Ownership interest (including stock options, but excluding indirect investments through mutual funds and the like) in a publicly traded company
5. Research funding
6. Honoraria directly received from an entity
7. Patents and royalties
8. Paid expert testimony
9. Membership on an entity's board of directors, speaker's bureau, or its advisory committees
10. Any other financial relationship and list organization and relationship

Following is an alphabetical list of moderators and presenters that have nothing to disclose:

Steven M. Harrison, PhD
Christa Lese Martin, PhD, FACMG
Erin Rooney Riggs, MS, CGC
Darrel Waggoner, MD, FACMG, ABMGG



2018 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 indicate 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

Legend:

1. Employment/Salary
2. Consultancy
3. Ownership interests (including stock options) in a start-up company, the stock of which is not publicly traded
4. Ownership interest (including stock options, but excluding indirect investments through mutual funds and the like) in a publicly traded company
5. Research funding
6. Honoraria directly received from an entity
7. Patents and royalties
8. Paid expert testimony
9. Membership on an entity's board of directors, speaker's bureau, or its advisory committees
10. Any other financial relationship and list organization and relationship

Members of the ACMG Staff (S), Education Committee (EC) and Program Committee (PC) have disclosed the following:

Bernat, John (EC)- Sanofi Genzyme, Shire, Protalix, 5

Northrup, Hope (PC)- BioMarin Pharmaceuticals, 6

Pappas, Kara (EC)- BioMarin Virtual, 2

Slavotinek, Anne (EC, PC)- OptumRx, Retrophin, 2; Oxford University Press, 7

Stevenson, David (PC)- Alexion Pharmaceuticals, Lineagen, Inc., GLG, 2; Prader-Willi Syndrome Association, Cardiofaciocutaneous Syndrome, Costello Syndrome 9



Following is a list of ACMG Staff, Education and Program Committees that have nothing to disclose:

Arnold, Georgianne (PC)	Mito, Yoshiko (EC)
Bao, Liming (EC)	Mullen, Thomas (EC)
Barnett, Claudia (S)	Nguyen, Joanne (PC)
Cunniff, Christopher (PC)	Pal, Tuya (PC)
Dahlroth, Jane (S)	Parikh, Aditi (EC)
Dhar, Shweta (EC)	Radford, Jane (S)
Dipple, Katrina (PC)	Roberts, Amy (EC)
Elsea, Sarah (PC)	Schaaf, Christian (PC)
Freire, Penelope (S)	Smith, Jessica (PC)
Giovanni, Monica (EC, PC)	Suskin, Barrie (EC)
Hisama, Fuki (PC)	Toriello, Helga (PC)
Hodge, Jennelle (PC)	Watson, Michael (S)
Hudgins, Louanne (PC)	White, Janson (EC)
King, Jennifer (PC)	Wick, Myra (PC)
Magoulas, Pilar (PC)	

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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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+



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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



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Registration and Fees

ondemand.acmg.net

ACMG Members and ACMG Trainees: (\$100)

Non-members (\$120)

Additional fee (~\$25) applies for NSGC credit that is billed by NSGC.

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

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