ClinGen Gene-Disease Validity Curation Module 2020 Version

Date of Release: September 30, 2020
Expiration Date: December 31, 2020
Course must be completed by the expiration date
Educational Credits Offered: CME, P.A.C.E.®, NSGC Category 2 (Self-report)
Estimate Time of Completion: 10 hours per curation (limit of up to 10 curations)
Target Audience: This ClinGen Gene-Disease Validity Curation module is intended to provide learners with educational credit for participating in ClinGen gene curation activities and is available to individuals who are existing members of ClinGen GCEPs and have had at least one gene curation previously approved by a ClinGen GCEP.

Contents

Course Information

Overview
The Clinical Genome Resource (ClinGen, www.clinicalgenome.org) is an NIH-funded resource dedicated to building an authoritative central resource that defines the clinical relevance of genes and variants for use in precision medicine and research.

In 2017, ClinGen published “Evaluating the Clinical Validity of Gene-Disease Associations: An Evidence-Based Framework Developed by the Clinical Genome Resource” (Strande et al. 2017, PMCID: PMC5473734). The framework involves evaluating the strength of evidence supporting or refuting a claim that variation in a particular gene causes a particular disease. This framework 1) defines the criteria needed to assess clinical validity, 2) describes the evidence supporting a gene-disease association in a semi-quantitative manner, and 3) allows biocurators to use this information to methodically classify the validity of a given gene-disease pair.

ClinGen forms Gene Curation Expert Panels (GCEPs) to implement the approved process of gene-disease validity evaluation. Each GCEP is focused on a particular disease area (for example, hereditary cancer, intellectual disability/autism, hearing loss, etc.) and includes members with clinical care, research, and diagnostic laboratory expertise within that domain, as well as biocurators with experience in the gene-disease validity process. In most cases, a biocurator completes the initial gene-disease curation and arrives at a provisional classification, followed by presentation of the data to the GCEP for expert review and final approval. GCEPs utilize the ClinGen Gene Curation Interface (GCI) for documentation of gene-disease validity classifications, and all curations completed by the group are made publicly available through the ClinGen website (clinicalgenome.org).

Course Objectives

- Use the ClinGen vetted gene-disease clinical validity curation process to evaluate the strength of evidence supporting or refuting a claim that variation in a particular gene causes a particular disease
- Perform a literature search to identify relevant publications to support a gene curation
- Score collected/collated evidence in the ClinGen GCI to determine a provisional classification for a gene-disease pair based on guidelines
- Present evidence summary supporting provisional classification for expert approval
- Publish approved gene curation to clinicalgenome.org

For the purpose of this module, learning objectives will be demonstrated by the learner by submitting a ClinGen GCEP-approved gene-disease curation final evidence summary published on clinicalgenome.org. Each curation has been approved for up to 10 hours of credit. You may submit up to 10 curations per year. Please complete a separate module for each curation you are submitting.

Target Audience
This ClinGen Gene-Disease Validity Curation module is intended to provide learners with educational credit for participating in ClinGen gene curation activities and is available to individuals who are existing members of ClinGen GCEPs and have had at least one gene curation previously approved by a ClinGen GCEP.

Course Requirements
- Invitation code
- Provide a record of a ClinGen GCEP-approved, published classification below, including:
  - Gene Name
  - Disease Name
  - Clinicalgenome.org URL of Gene-Disease Curation
  - Upload a PDF of the approved and published evidence summary from the GCI
  - Gene-Disease Classification Approval Date
- Evaluation and self-report in the ACMG Genetics Academy Click here (required for educational credits)

Estimated time of Completion: Up to 10 hours per curation

Please note: In the Self Report section, you will be asked how many CMEs are requested. 1 CME is the equivalent of one hour. Please use that field to document the number of hours (maximum of 10) you spent gathering, evaluating and scoring evidence as well as presenting to the GCEP for approval.

Continuing Medical Education Credit (CME)
This activity has been planned and implemented by the American College of Medical Genetics and Genomics in accordance with the Essential Areas and Policies of the Accreditation Council for Continuing Medical Education.

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.

CME Credit Designation
The American College of Medical Genetics and Genomics designates this enduring material for a maximum of 10 hours in *AMA PRA Category 1 Credits*™. Physicians should claim only the credit commensurate with the extent of their participation in the activity.

Continuing Education Units (Clinical Laboratory Scientists, Directors, and 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. ACMG is approved by the Florida Board of Clinical Laboratory Personnel as CE Provider (50-11878). ACMG is approved by the California Department of Health Services through the ASCLS P.A.C.E.®. This activity has been approved for a maximum of 10 contact hours.
The course ID #20-717097 is registered with CEBroker.

NSGC Category 2 CEUs/Genetic Counselors
This activity meets requirements to apply for Category 2 CEUs from the National Society of Genetic Counselors (NSGC). Please complete the module and submit your certificate to NSGC, using the instructions for “How Do I Get Category 2 CEUs?” onhttps://www.nsgc.org/page/ceuapproval#Apply%20Cat%202 . ACMG

is the accredited sponsor. You must apply within three months of the activity date, but you will be able to submit up to 10 curations under one $25 fee for this activity.

References

Strande NT, Riggs ER, Buchanan AH, Ceyhan-Birsoy O, DiStefano M, et al. 2017. Evaluating the Clinical Validity of Gene-Disease Associations: An Evidence-Based Framework Developed by the Clinical Genome Resource. Am. J. Hum. Genet. 100(6):895–906

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NOTE:

- ACMG will follow the ACCME's expectation that no employees or owners of commercial interests will be involved as planners/faculty/presenters of a CME accredited activity.
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- Diagnostic laboratories are not considered commercial interests unless they are owned by or have a sister organization which is a commercial interest.

The following faculty does not have any relevant financial relationships to disclose:

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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:
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    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.