
Announcements

- **Educational Credit for ClinGen Gene-Disease Validity Curations**
 - 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. The module is available through the ACMG Genetics Academy, learn more [here](#).
- **CNV Guidelines Implementation Webinar Recordings Available**
 - On November 10th, ClinGen hosted a webinar focused on clinical laboratories' experiences implementing the ACMG/ClinGen technical standards for interpretation and reporting of constitutional copy number variants (CNVs). Recordings available [here](#).
- **ClinGen's Long QT Syndrome GCEP Paper Highlighted as a Significant Advance in Genomic Medicine**
 - The Genomic Medicine Working Group of the National Advisory Council for Human Genome Research recently published a "year in review" paper in the American Journal of Human Genetics highlighting 10 publications from the past year that reported significant advances in genomic medicine. [ClinGen's Long QT Syndrome gene curation effort](#) was included as one of these genomic medicine advances! Read the Genomic Medicine Year In Review: 2020 [here](#).
- **ClinGen Variant and Gene Curation Interface 2.0 Platform Released**
 - The new [GCI and VCI platform](#), released in December 2020, expands upon our legacy (v1.0) platform via a state-of-the-art microservices architecture designed to empower the future development goals for the gene and variant curation interfaces while maintaining all of the functionality and data from our legacy platform. Our new platform also enables continuous integration and continuous deployment which will enable us to deliver platform updates faster and more frequently without the need for lengthy platform downtimes.
- **ClinGen Gene Curation SOP version 8 Released**
 - The ClinGen Gene Curation Working Group is pleased to announce the release of version 8 of the gene curation SOP. The new SOP, including a clean copy, a summary of the changes, and a highlighted version, can be accessed [here](#).
- **VCEP Version 8 Application and Protocol Released**
 - ClinGen's application and protocol for applying for Variant Curation Expert Panel status has been updated. See version 8 [here](#).
- **GenomeConnect, ClinGen's Patient Registry**
 - ClinGen is enabling patients to share their de-identified genetic and health data with ClinVar through the [GenomeConnect](#) (the ClinGen Patient Registry) and by working with other registries to offer data sharing through the [Patient Data Sharing Program](#). To date, over 4,600 patients have engaged in data sharing through these efforts.
- **Volunteer to Curate**
 - Interested in volunteering to curate for ClinGen or know someone who is? Take this brief [survey](#) to tell us more about your interests, expertise, and desired level of involvement. Background training will be provided. For questions contact volunteer@clinicalgenome.org.

- **ClinGen Gene Curation Expert Panel Approvals**
 - The following GCEPs received approval this quarter:
 - [Cranial Malformations GCEP](#)
 - [Humoral Defects GCEP](#)
 - [SCID-CID GCEP](#)
- **ClinGen Variant Curation Expert Panel Approval Progress**
 - The following VCEPs received Step 1 approval:
 - [Congenital Myopathies VCEP](#)
 - [ENIGMA BRCA1 and BRCA2 VCEP](#)
 - [Leber Congenital Amaurosis/early onset Retinal Dystrophy VCEP](#)
 - [NTRK Fusions Somatic Cancer VCEP](#)
 - [Peroxisomal Disorders VCEP](#)
 - [X-linked Inherited Retinal Disease VCEP](#)
 - The [DICER1 and miRNA-Processing Gene VCEP](#) and [Hereditary Breast, Ovarian and Pancreatic Cancer VCEP](#) received Step 2 approval to begin variant curation pilots.
 - The [Familial Hypercholesterolemia VCEP](#) and [Malignant Hyperthermia Susceptibility VCEP](#) have completed the final stage of the Variant Curation Expert Panel approval process, readying them to submit their variant assertions to ClinVar as an FDA Recognized Expert Panel submitter.

Publications

- Fortuno et al. **Specifications of the ACMG/AMP variant interpretation guidelines for germline *TP53* variants.** [Go to publication.](#)

Conferences & Meetings

- [Curating the Clinical Genome \(Virtual Meeting\)](#) May 12-14, 2021
 - Abstract deadline March 16, 2021
 - Registration deadline May 5, 2021
- **ClinGen Workshop (Virtual)** June 24-25, 2021
 - Save the Date, information to follow

Reminders

- **ClinGen Consortium Call** - 3rd Friday of the month at 2pm ET - a reminder will be sent before each call. Upcoming topics:
 - **January 15th - ClinVar Update and Community Feedback**
 - Join us to hear updates about ClinVar and participate in an interactive feedback session. If you don't have the calendar hold for the January 15th ClinGen Consortium call, please sign up [here](#).
- **ClinGen follows "Meeting Free Weeks for Genomics"** - [learn more](#), upcoming meeting free weeks:
 - January 25-29, 2021
 - May 17-21, 2021
 - August 9-13, 2021
 - November 1-5, 2021

On a related note...

The Wehl lab at Washington University School of Medicine is hiring a biocurator/coordinator position focused on Limb Girdle Muscular Dystrophy (LGMD) genes. This position would interface with ClinGen, Dr. Conrad Wehl, the chair of the curation expert panel, and other biocurators and experts to resolve genetic variants in LGMD genes. Our group is the genetics lead for an international LGMD consortium focused on improving LGMD diagnostics within the academic community.

The position would be full time remote or at Washington University with flexible hours (curation is done via an online interface and meetings are all conducted via zoom). The ideal candidate would have a background in genetics/biology, be eager to learn, organized, good communication skills, and the ability to work from home. The candidate would be trained in the process of biocuration so previous experience in "gene curation" is not essential but a background in understanding human genetics, rare diseases and ability to work with patient charts and the medical literature are essential.

Ideal position for a candidate interested in clinical research, a stepping stone to graduate or medical school, PhD scientist/geneticist/genetic counselor interested in the flexibility of working remotely.

Interested candidates can reach out to Dr. Wehl (weihlc@wustl.edu) with the subject "biocurator."

Do you have news you'd like featured in the next update?
Email clingen@clinicalgenome.org

