

---

## Announcements

- **ClinGen Brugada Syndrome GCEP Publication Selected as Advance in Genomic Medicine 2019**
  - The Genomic Medicine Working Group of the National Advisory Council for Human Genome Research of the National Human Genome Research Institute named the Brugada Syndrome GCEP [publication](#) as one of the ten most significant advances in genomic medicine in 2019. Learn more [here](#).
- **ACMG/ClinGen Technical Standards for Interpretation and Reporting of Constitutional Copy Number Variants (CNVs) Released**
  - In November, the ACMG and ClinGen's joint consensus recommendation "Technical standards for the interpretation and reporting of constitutional copy number variants" was published - introducing a points-based scoring rubric to guide laboratories toward more consistent CNV interpretations. Available [here](#).
- **CNV Technical Standards Web Series Starts in January**
  - ClinGen will host a multi-part web series to educate the community about the CNV technical standards. Webinars will take place on Thursdays at 2pm US Eastern time beginning January 16, 2020. Preview the schedule [here](#) and register for the web series [here](#). *Participants who register for the ClinGen CNV Technical Standards web series by tomorrow December 19th will have the opportunity to participate in an optional pre-/post-series evaluation.*
- **New ClinGen Gene Curation Expert Panels**
  - The following GCEPs were approved this quarter: [Charcot-Marie-Tooth](#), [Congenital Myopathies](#), [Dilated Cardiomyopathy](#) and [Limb Girdle Muscular Dystrophies](#).
- **ClinGen Variant Curation Expert Panel Approval Progress**
  - The [von Willebrand Disease VCEP](#) achieved Step 1 approval this quarter. The [Monogenic Diabetes VCEP](#) received approval to begin their variant curation pilot (Step 2).
- **ClinGen Contributes to GA4GH VR Specification**
  - The Variation Representation (VR) specification was approved by the Global Alliance of Genomics and Health (GA4GH) in October. ClinGen's Allele Registry created an initial implementation for the VR specification. Learn more [here](#).
- **GenomeConnect, ClinGen's Patient Registry**
  - As of December 1st, 2700 participants have enrolled in [GenomeConnect](#). Of all participants, 2298 (n=85.1%) have completed their health survey providing additional phenotypic details and 32% (n=865) have uploaded a copy of their genetic testing report. To date, GenomeConnect has shared 1053 variants with ClinVar. ClinGen is now actively working to enable participants in other registries to share data through its [Patient Data Sharing Program](#). As of December 1st, 149 patients have engaged in data sharing from six external gene or condition specific registries.
- **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](mailto:volunteer@clinicalgenome.org).

## Conferences & Meetings

- **ClinGen at ACMG 2020**
  - ClinGen Community Curation Interactive Workshop, March 19, 2020, 1:30 PM - 3:30 PM, learn more [here](#)
    - The goal of the session is to educate attendees about ClinGen Community Curation and train interested volunteers to perform computationally-aided curation with ClinGen. During the first half of the program, we will describe the Community Curation initiative and train attendees to perform computationally-aided curation. In the second half, we will guide attendees in a live interactive curation exercise using the ClinGen computationally-aided process and infrastructure.
    - Sign up for the workshop when you register for the meeting.
- **Genomics of Rare Diseases**
  - March 25 - 27 2020, Wellcome Genome Campus, UK
- **Curating the Clinical Genome**
  - May 20 - 22 2020, Wellcome Genome Campus, UK
  - Abstract deadline: March 24 2020

## Publications

- Brnich et al. **Recommendations for application of the functional evidence PS3/BS3 criterion using the ACMG/AMP sequence variant interpretation framework.** [Go to preprint](#)
- Kanavy et al. **Comparative analysis of functional assay evidence use by ClinGen Variant Curation Expert Panels.** PMID: 31783775. [Go to article.](#)
- Luo et al. **ClinGen Myeloid Malignancy Variant Curation Expert Panel recommendations for germline *RUNX1* variants.** PMID: 31648317. [Go to article.](#)
- Ritter et al. **A case for expert curation: an overview of cancer curation in the Clinical Genome Resource (ClinGen).** PMID: 31645350. [Go to article.](#)
- Wain et al. **Variant interpretation is a component of clinical practice among genetic counselors in multiple specialties.** PMID: 31754268. [Go to article.](#)

## On a related note...

The National Human Genome Research Institute (NHGRI) at the National Institutes of Health (NIH) seeks highly-motivated applicants for scientific administrative openings as Scientific Program Analysts in Bethesda, MD. See full job description [here](#).

**Thank you for your contributions and participation in 2019!**

**Happy New Year!**

Do you have news you'd like featured in the next update?  
Email [clingen@clinicalgenome.org](mailto:clingen@clinicalgenome.org)

