January - March 2020

Clinical Genome Resource

Quarterly Update

Announcements

- **COVD-19 and ClinGen Consortium Activities**
  - To the best of our abilities, we aim to continue most ClinGen Consortium activities including Steering Committee, Consortium, or Working Group conference calls according to the established schedule, in order to facilitate work among individuals able to continue working from home. For the time being, we will not hold any meetings or other activities in-person, but will plan to carry them out through teleconference and videoconference. The most current information from NIH can be found at https://www.nih.gov/health-information/coronavirus.
- **Recommended Terminology for Variants with Decreased Penetrance for Mendelian Conditions Released**
  - The ClinGen Low Penetrance/Risk Allele Working Group released a proposal "Recommended Terminology for Variants with Decreased Penetrance for Mendelian Conditions Released." Read the proposal here.
- **CNV Technical Standards Web Series Recordings Available**
  - The ClinGen CNV web series is now complete. Webinars took place January 16-March 20, 2020, and focused on a different aspect of the scoring metrics each week. We welcome you to view the slides and recordings here.
- **PAR-20-101 Genomic Expert Curation Panel Funding Opportunity**
  - Learn more about the NICHD Genomic Clinical Variant Expert Curation Panels Program Announcement (PAR-20-101) to establish expert panels that will select genes and genomic variants associated with diseases or conditions of high priority to participating NIH Institutes and Centers and systematically determine their clinical significance for diagnosis and treatment of these diseases or conditions. Learn more and find Frequently Asked Questions here.
- **ClinVar Celebrates Submission of One Millionth Submitted Record**
  - In January, ClinGen celebrated ClinVar reaching 1,000,000 submitted records by highlighting submission milestones, review the full list here.
- **Clinical Laboratories Meeting Minimum Requirements for Data Sharing to Support Quality Assurance**
  - Updated requirements for the ClinGen list of "Clinical Laboratories Meeting Minimum Requirements for Data Sharing to Support Quality Assurance" went into effect January 1, 2020. Learn more and see the updated list here.
- **New ClinGen Gene Curation Expert Panels**
  - The following GCEPs were approved this quarter: Pulmonary Hypertension, Skeletal Disorders, and Syndromic Disorders.
- **ClinGen Variant Curation Expert Panel Approval Progress**
  - The Limb Girdle Muscular Dystrophy VCEP and Thrombosis VCEP achieved Step 1 approval this quarter. The ACADVL VCEP and Coagulation Factor Deficiency VCEP received Step 2 approval to begin their variant curation pilot.
- **Registration of CNVs in the Allele Registry from Four Databases**
  - The ClinGen Allele Registry has been updated to include CNVs from ClinVar, gnomAD, dbVar, and ExAC, totaling over 344,000 registered descriptors. Users can query CNVs by descriptor or reference sequence coordinates here.
- **ClinGen Linked Data Hub – A resource to link data entities from different sources**
  - The ClinGen Linked Data Hub (LDH) is a web based resource to facilitate

access to aggregated information such as links and select data ("excerpts") from different data sources. Content is made available to the consumers using RESTful APIs. Currently, LDH focuses on linking information about human genes and variants to support ClinGen curation efforts. LDH is accessible here.

- **GenomeConnect, ClinGen's Patient Registry**
  - GenomeConnect submits candidate genes to Matchmaker Exchange, a federated network of rare disease datasets that facilitates case matching. To date, 88 candidate genes from participants have been shared, resulting in over 800 potential matches and 22 requests for additional phenotype information and/or recontact of the participant for additional research. Additionally, this process has made the GenomeConnect team aware of nine gene-disease relationships that were defined after the participant's testing was reported. GenomeConnect has shared these gene-validity updates with relevant registry participants using the previously established process to provide updated variant classifications from ClinVar back to interested participants.
- **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.

Conferences & Meetings

- **ACMG 2020 Poster Gallery**
  - The 2020 ACMG Annual Clinical Genetics Meeting Poster Gallery is now available in the online ACMG Genetics Academy! Visit this link to view 800+ poster abstracts. Presenters have until April 17 to upload poster PDFs and audio presentations. Check back often!
  - A list of ClinGen ACMG 2020 posters can be found here.
- **Curating the Clinical Genome 2020 Postponed**
  - Due to the evolving situation with COVID-19, this conference has been postponed. Please see the conference website for most up to date information.

Publications

- Adler et al. **An International, Multicentered, Evidence-Based Reappraisal of Genes Reported to Cause Congenital Long QT Syndrome.** Go to publication. Read NHGRI's press release here.
- Wu et al. **How I Curate: Applying American Society of Hematology-Clinical Genome Resource Myeloid Malignancy Variant Curation Expert Panel Rules for RUNX1 Variant Curation for Germline Predisposition to Myeloid Malignancies.** Go to publication.

Do you have news you'd like featured in the next update?
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