April - June 2019

Clinical Genome Resource

Quarterly Update

Announcements

- **ClinGen Variant Curation SOP Now Available**
  - The ClinGen Variant Curation Standard Operating Procedure (SOP) was released in April and is designed to provide guidance on variant classification using ClinGen approved processes and tools. Read the Variant Curation SOP here.
- **SVI Recommendation for in *trans* Criterion (PM3) Released**
  - The Sequence Variant Interpretation (SVI) Working Group proposed a point-based system to determine the strength of in *trans* observations (ACMG/AMP criterion PM3) based upon variant phasing and classification of the variant occurring on the other allele. Read full recommendation here.
- **Learn more about ClinGen's Stakeholder Partnership WG**
  - The goal of ClinGen's new Stakeholder Partnership WG is to facilitate regular interactions and systematic input from stakeholders on ClinGen's portfolio of current and planned activities in order to inform ClinGen's development, grow the user community and leverage collaboration opportunities. Learn more here.
- **New ClinGen Gene Curation Expert Panels**
  - The following GCEPs were approved this quarter: Hereditary Cancer, Monogenic Diabetes and Peroxisomal Disorders.
- **ClinGen Variant Curation Expert Panel Approval Progress**
  - The Coagulation Factor Deficiency VCEP and Hereditary Hemorrhagic Telangiectasia VCEP achieved Step 1 approval this quarter. The Platelet Disorder VCEP and Rett and Angelman-like Disorders VCEP received approval to begin variant curation pilots (Step 2).
- **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.
- **New ClinVar Variation Report View**
  - In June, the newly designed ClinVar variant report pages ended their "alpha" release and moved to production. Learn more about how to navigate the new display here.

Conferences & Meetings

- **Presentations from CCG 2019 now available**
  - Abstracts and select presentations from Curating the Clinical Genome 2019 are now available here
- **Save the Date! Curating the Clinical Genome 2020**
  - Wellcome Genome Campus, UK, May 20-22, 2020 #CCG2020

Publications

- DiStefano et al. **ClinGen Expert Clinical Validity Curation of 164 Hearing Loss Gene-Disease Pairs.** PMID: 30894701. Go to article.

On a related note:

Medical Genetics Curation Opportunity

Computercraft is looking for a candidate with experience interpreting variants in a clinical setting to work on site at the National Center for Biotechnology Information (NCBI) at the National Institutes of Health (NIH) in Bethesda, MD. The candidate will use his or her

expertise in human genetics to advance the NCBI resources ClinVar, MedGen, and the Genetic Testing Registry, and contribute to data-sharing efforts for clinicians and researchers in the medical genetics community. A detailed description of the position is available at https://chm.tbe.taleo.net/chm01/ats/careers/v2/viewRequisition?org=COMPUTERCRAFT&cws=37&rid=142. If you know of anyone who is interested and qualified, please feel free to forward this announcement. Candidates should contact Computercraft directly with any questions and to apply.

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