

ClinGen at ACMG 2019

Visit us at Booth #529

ClinGen Reception

Wed April 3, 7:30-9:00pm, Washington State Convention Center, Room 606-609

Presentations

Platform Presentations, Washington State Convention Center

- Wed April 3, 4:30-4:45pm Room 6C The Gene Curation Coalition: A global effort to harmonize gene-level resources (M. DiStefano)
- Fri April 5, 4:45-5:00pm Room 6E Myeloid Malignancy Variant Curation Expert Panel: an ASH-sponsored ClinGen Expert Panel to optimize and validate ACMG/AMP variant interpretation guidelines for genes associated with inherited myeloid neoplasms (X. Luo)

Learning Lounge

Thurs April 4, 12:45-1:15pm - Learning Lounge 2 Collaboratively Transforming Access to Genetic Testing

Workshop

• Fri April 5, 7:15-8:30am - Room 3AB ClinGen Expert Panel Information Workshop (registration required)

Poster Presentations

Thurs April 4, 10:00-11:30am

(Exhibit Halls 4ABCD)

- **181** VHL Information Sharing International Consortium (VISION): A Clinical Genome Resource (ClinGen) Expert Panel for VHL Variant Interpretation (D. Ritter)
- 319 The Use and Potential Impact of Diversity Measures in Medical Genetics Practice: Results of a Survey of Clinical Genomics Professionals (A. Popejoy)
- 467 Structured Narrative Triplets of Functional Assays to Support the Determination of Damaging Effect on Protein Function (A. Lopez Pineda)
- 681 Contrasting the GAA Variant Spectrum in Newborn Screening and Pompe Diagnostic Testing Populations (B. Seifert)
- 709 Toward Complete Interpretation of Known RYR1 Variants (J. Johnston)

Fri April 5, 10:30am-12:00pm

- 178 Clinical Genome Resource (ClinGen) Somatic Cancer Working Group Minimum Variant Level Data (MVLD) v2.0: Extensions Incorporating AMP Somatic Interpretation Guidelines (D. Ritter)
- 294 ClinGen's Variant Prioritization Tool (VPT): A Tool for Prioritizing Clinical Variants at Scale (M. Wright)
- 682 Standards for the Classification and Reporting of Constitutional Copy Number Variants: A ClinGen/ACMG Joint Consensus Recommendation (E. Riggs)
- **784** Opinions of ClinGen Members on the Use of ClinGen Clinical Validity Classifications in Genetic Testing (J. Goldstein)
- 808 ClinGen Recurrent Copy Number Variant Annotation File to Aid Clinical Interpretation During Genomic Analysis (J. Herriges)
- 834 ClinVar Makes Updates Easy: A New Online Form to Update a Single SCV (M. Landrum)



