

## ClinGen Luncheon

Wed Oct 16, 12:45-2:00pm, Marriott Marquis Houston, River Oaks, Level 3

### Platform Presentations

- **Sat Oct 19, 10:00 am [Room 361CD – Level 3/Convention Center] 352** Recommendations for determining the clinical validity of functional studies for use in variant interpretation. **(S. Brnich)**
- **Sat Oct 19, 10:15 am [Room 361CD – Level 3/Convention Center] 353** Validation of scoring metrics to guide the classification of constitutional copy number variants. **(E. Riggs)**

### Poster Presentations

*\* Indicates Reviewer's Choice – top 10% of abstracts*

#### Wed Oct 16, 2:00-4:00pm - Exhibit Hall, Level 1

- **405** Specification of ACMG/AMP guidelines for standardized variant interpretation in familial hypercholesterolemia: On behalf of the ClinGen FH Variant Curation Expert Panel. **(M. Iacocca)**
- **1620** ClinGen Linked Data Hub: Scalable infrastructure for aggregation of diverse types of variant information to support pathogenicity assessment. **(A. Milosavljevic)**
- **2631** Changes in variant classification between *KCNQ1*-modified criteria and standard ACMG 2015 guidelines. **(M. Care)**

#### Thurs Oct 17, 2:00-4:00pm - Exhibit Hall, Level 1

- **729** ClinGen community curation: Crowdsourcing curation efforts from geneticists to citizen scientists. **(C. Thaxton)**
- **1636** Clinical actionability curation tools and knowledge repository of the ClinGen consortium. **(S. Subramanian)**
- **2275** Creating a reporting framework for polygenic risk scores to improve transparency and standardization: Results from a ClinGen complex disease working group literature review. **(H. Wand)**
- **3151** Curation of mosaic variants in developmental brain disorders: Recommendations from the ClinGen Brain Malformation Curation Expert Panel. **(A. Lai)**

#### Fri Oct 18, 1:00-3:00pm - Exhibit Hall, Level 1

- **676** Development of methods to expand the ClinGen Actionability Working Group framework to include assertions of the clinical actionability of genomic variation in the context of secondary findings. **(J. Hunter)**
- **698** Reporting of variants in genes with limited, disputed, or no evidence for a Mendelian condition among GenomeConnect participants. **(J. Savatt)\***
- **914** Gene-specific criteria for germline *RUNX1* variant curation: Recommendations from the ClinGen Myeloid Malignancy Variant Curation Expert Panel. **(X. Luo)**
- **2450** Structured narrative of functional assays to support the determination of damaging effect on protein function. **(M. Iacocca)\***
- **2534** Toward comprehensive interpretation of *RYR1* variants associated with malignant hyperthermia susceptibility. **(J. Johnston)**
- **2579** Copy number variants in the ClinGen Allele Registry. **(R. Patel)\***