

Curating the Clinical Genome Meeting 2019

Washington Marriott Wardman Park

2660 Woodley Road NW, Washington, District of Columbia 20008 USA

Sessions will be held in Thurgood Marshall NE

Meals, Exhibits, Poster Session, and Receptions will be held in Thurgood Marshall SE

Wednesday, May 29

4:00 pm **Registration Open**

6:00 pm **Welcome Session**

Introduction

Erin Ramos, PhD, MPH, National Human Genome Research Institute

National Human Genome Research Institute Strategic Planning

Eric Green, MD, PhD

Director, National Human Genome Research Institute

7:00 pm **Reception** Sponsored by GeneDx

Thursday, May 30

8:00 am **Breakfast/Registration Open**

8:20 am Congenica Sponsored Presentation

8:30 am **Welcome and Introduction**

Christa Martin, PhD, Geisinger (CCG 2019 Program Committee Chair)

8:45 am **ClinGen FDA Recognition**

Julianne O'Daniel, MS, CGC, University of North Carolina, Chapel Hill

9:00 am **Next Generation Phenotyping**

Moderator: Helen Firth, DM, Wellcome Trust Sanger Institute, Cambridge
University Hospitals NHS Foundation Trust

9:00 am **HPO Integration**

Peter Robinson, MD, MS, The Jackson Laboratory

9:25 am **Use of Digital Biomarkers with Genomic Data**

Evan Muse, MD, Scripps Research Translational Institute

www.clinicalgenome.org/meeting

- 9:50 am **Using Semantic Similarity Analysis Based on Human Phenotype Ontology Terms to Identify Genetic Etiologies for Epilepsy and Neurodevelopmental Disorders***
Ingo Helbig, MD, Children's Hospital of Philadelphia
- 10:05 am **Break**
- 10:25 am **Sequence and Copy Number Variant Interpretation**
Moderator: Caroline Wright, PhD, University of Exeter
- 10:25 am **Economics of Variant Reinterpretation**
Christine Lu, MS, PhD, Harvard Medical School
- 10:50 am **Penetrance in UK Biobank**
Michael Weedon, PhD, University of Exeter
- 11:15 am **Is Likely Pathogenic Really 90% Likely? A Look at the Data***
Steven Harrison, PhD, Broad Institute
- 11:30 am **Establishing an Intralaboratory Copy Number Variant Conflict Resolution Process Using ClinGen Dosage Sensitivity Map Scores and Internal Classification Discrepancies to Improve Retrospective Data Analyses and Submissions to ClinVar***
Zoe Lewis, MS, CGC, ARUP
- 11:45 am **Constructing a Quantitative Metric for Evaluating the Clinical Significance of Recurrent Copy Number Variants***
John Herriges, PhD, Children's Mercy Hospital
- 12:00 pm **Lunch**
- 12:30 pm GeneDx Sponsored Presentation
- 12:40 pm Invitae Sponsored Presentation
- 12:50 pm Agilent Technologies Sponsored Presentation
- 1:00 pm Illumina Sponsored Presentation
- 1:45 pm **Polygenic Risk Scores in the Clinic**
Moderator: David Ledbetter, PhD, Geisinger
- 1:45 pm **Cardiovascular Polygenic Risk Scores**
Amit Khera, MD, Massachusetts General Hospital
- 2:10 pm **The Polygenic Contribution to Developmental Disorders**
Mari Niemi, PhD, Wellcome Sanger Institute
- 2:35 pm **Genomics as a Personalized Medicine Approach in Disease Risk Prediction - P5.fi FinHealth***
Heidi Marjonen, PhD, National Institute for Health and Welfare, Helsinki, Finland

- 3:00 pm **Data Sharing**
Moderator: Danielle Azzariti, MS, CGC, Broad Institute
- 3:00 pm **Count Me In**
Corrie Painter, PhD, Broad Institute
- 3:25 pm **Care4Rare**
Kym Boycott, MD, PhD, Children's Hospital of Eastern Ontario
- 3:50 pm **ClinGen Linked Data Hub: Scalable Aggregation of Diverse Types of Variant Information to Support Pathogenicity Assessment***
Aleksandar Milosavljevic, PhD, Baylor College of Medicine
- 4:05 pm **DECIPHER – Innovation in Data-sharing in Rare Disease***
Helen Firth, DM, Wellcome Trust Sanger Institute, Cambridge University Hospitals NHS Foundation Trust
- 4:20 pm **Break**
- 4:45 pm **Rapid Platform Session**
Moderator: Marc Williams, MD, Geisinger
- 5:15 pm **Reception and Poster Session**
Sponsored by Illumina, *poster presentations 5:15pm - 6:45pm*

**Selected from abstracts*

Friday, May 31

- 7:30am **Breakfast**
- 8:00am **Developing Sustainable Partnerships**
- 8:00am Late Breaking Content
- 9:00am **Methodical Curation of the Human Genome: Challenges and Progress in Establishing an Objective, Consistent, and Scalable Process in a Clinical Laboratory***
Jackie Tahiliani, MS, CGC, Invitae
- 9:15am **Improving Efficiency of Gene Curation to Support Reporting In a Clinical Laboratory Setting***
Sarah Schmidt, MS, Illumina Clinical Services Laboratory
- 9:30am **Gene Curation**
Moderator: Laura Milko, PhD, University of North Carolina, Chapel Hill
- 9:30am **Gene Curation for the Interpretation of Clinical Genomes and the Future of PanelApp**
Ellen McDonagh, PhD, Genomics England

- 9:55am **Citizen Scientists Participation and Contribution to Gene Curation**
Andrew Su, PhD, Mark2Cure, The Scripps Research Institute
- 10:20am **The Gene Curation Coalition: A Global Effort to Harmonize Gene-level Resources***
Marina DiStefano, PhD, Harvard Medical School
- 10:35pm **Transcript Curation for the Clinical Use Case: LRG and MANE***
Joannella Morales, PhD, European Molecular Biology Laboratory, European Bioinformatics Institute
- 10:55 **Break**
- 11:15am **Hereditary and Somatic Cancer**
Moderator: Sharon Plon, MD, PhD, Baylor College of Medicine
- 11:15am **CRISPR Engineered *BRCA1* Variants**
Lea Starita, PhD, University of Washington
- 11:40am **Somatic Variant Curation Standards Enable Improved Identification of Relevant Clinical Interpretations for Tumor Variants***
Alex Wagner, PhD, Washington University School of Medicine
- 11:55am **Implementation and Cases Studies of Tumor Whole-Genome Sequencing within Pediatrics Precision Medicine at MSKCC***
Dominik Glodzick, PhD, Memorial Sloan Kettering Cancer Center
- 12:10pm **Population Screening**
Moderator: Sian Ellard, PhD, University of Exeter Medical School
- 12:10pm **Implementing Universal Lynch Syndrome Screening**
Alanna Rahm, PhD, MS, Geisinger
- 12:35pm **Estonia Familial Hypercholesterolemia Screening**
Tõnu Esko, PhD, University of Tartu
- 1:00pm **Closing**
Helen Firth, DM, Wellcome Trust Sanger Institute, Cambridge University Hospitals NHS Foundation Trust
- Lunch to Go**
Ballroom available until 2pm for those who would like to stay and eat

*Selected from abstracts