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

Wednesday, May 29

3:00 pm **Registration Open**

6:00 pm **Welcome Session**

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

The Human Genome Project Was Just the Beginning: Research Opportunities at 'The Forefront of Genomics'
Eric Green, MD, PhD
Director, National Human Genome Research Institute

7:00 pm - 8:00 pm **Reception** sponsored by GeneDx

Thursday, May 30

8:00 am **Registration Open, Breakfast and Sponsored Presentations**

8:10 am **Confidence that Scales: Accuracy of Automated Variant Classification Based on Expert-Derived Guidelines – An Evaluation of Concordance Across >6,000 samples**
Jennifer Poitras, PhD, QIAGEN

8:20 am **Case Studies: Timely and Accurate Diagnosis – Informing Clinical Actions**
Philip Beales, MD, FRCP, FMedSci, UCL Great Ormond Street Institute of Child Health (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
- 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 & Sponsored Presentations**

- 12:30 pm **Disease Gene Discovery: Collaborative Efforts**
  Jane Juusola, PhD, GeneDx
- 12:40 pm **500,000 Patients Later: What Have We Learned That Can Help Us All?**
  Steve Lincoln, Invitae

12:50 pm **Optimizing Lab Efficiency with Agilent Workflow Solutions**
Josh Wang, PhD, Agilent

1:00 pm **Clinical Whole Genome Sequencing and Curation for Rare and Undiagnosed Disease**
Krista Bluske, PhD, Illumina

1:45 pm **Polygenic Scores in the Clinic**
Moderator: David Ledbetter, PhD, Geisinger

1:45 pm **Genome-wide Polygenic Scores for Cardiovascular Disease**
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 **Genomics4RD: A Data Lake Powering Rare Disease Research Across Canada**
Kym Boycott, MD, PhD, Children's Hospital of Eastern Ontario, University of Ottawa, Canada

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 - 6:45 pm **Reception and Poster Session**
Sponsored by Illumina

**Selected from abstracts*

Friday, May 31

7:30am **Breakfast**

8:00am **Developing Sustainable Partnerships**
Moderators: David Ledbetter, PhD, Geisinger & Michael Watson, PhD, American College of Medical Genetics and Genomics

- 8:00am **Overview of Sharing the Labor Around Genomic Curation in the Pre-Competitive Space**
  David Ledbetter, PhD, Geisinger
- 8:15am **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
- 8:30am **Emerging Ideas**
  Phillip Febbo, MD, Chief Medical Officer, Illumina
- 8:45am **Improving Efficiency of Gene Curation to Support Reporting In a Clinical Laboratory Setting***
  Sarah Schmidt, MS, Illumina Clinical Services Laboratory
- 9:00am **Questions with the Panel**
  Panel: Phillip Febbo, Oz Huner, David Ledbetter, Christa Martin, Sarah Schmidt, Jackie Tahiliani, Michael Watson

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, 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 Glodzik, 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 Kulchak Rahm, PhD, MS, Geisinger

12:35pm **Estonia Familial Hypercholesterolemia Screening**
Tõnu Esko, PhD, University of Tartu, Broad Institute

1:00pm **Closing**
Helen Firth, DM, Wellcome Trust Sanger Institute, Cambridge University Hospitals NHS Foundation Trust

1:15pm **Lunch to Go**

*Selected from abstracts