Curating the Clinical Genome 2017

Wednesday, June 28, 2017

5:00pm - 7:00pm **Registration open**
Location: Metropolitan Ballroom Foyer

Poster Set Up
Location: Dupont Ballroom

7:00pm - 9:00pm **Precision Medicine**
Moderator: Heidi Rehm, Harvard Medical School

7:00pm Welcome and Introduction
Heidi Rehm

7:15pm Accelerating Precision Health for All of Us
Eric Dishman, *All of Us* Research Program, National Institutes of Health

7:45pm DDD: A National Exome Sequencing Study
Caroline Wright, University of Exeter

8:15pm Q&A

8:30pm Moderated Discussion: Precision Medicine
Moderator: Heidi Rehm

Thursday, June 29, 2017

7:30am - 8:00am **Registration**
Location: Metropolitan Ballroom Foyer

Breakfast *Sponsored by Illumina*
Location: Metropolitan Ballroom Foyer

Poster Set Up
Location: Dupont Ballroom

8:00am - 9:15am **Curating Gene-Disease Relationships**
Moderator: Caroline Wright

8:00am Gene Curation: G2P Efforts
Helen Firth, Cambridge University Hospitals

8:15am The ClinGen Gene Curation Process
Erin Rooney Riggs, Geisinger Health System

8:30am *Selected from Abstracts* Use of the ClinGen Clinical Validity Framework to Evaluate the Strength of Evidence for Genes Implicated in Hypertrophic Cardiomyopathy
Jennifer Goldstein, University of North Carolina at Chapel Hill

8:45am *Selected from Abstracts* The Impact of Community Curation of Gene-Disease Relationships for Clinical Genome Analysis
Ellen McDonagh, Genomics England

9:00am Q&A

9:10am Harmonizing Gene Curation Efforts Panel
Moderator: Caroline Wright

9:30am - 10:45am **Perspectives and Regulations on Data Sharing**
Moderator: Helen Firth

9:30am UK Perspective on Data Sharing
Alison Hall, PHG Foundation

9:45am Patient Access to Genetic Test Results and Associated Data
Sandra Park, American Civil Liberties Union

10:00am *Selected from Abstracts* The *BRCA* Exchange: Global Data Sharing and Knowledge Exchange to Enable Accurate Clinical Care
Lena Dolman, Global Alliance for Genomics and Health (GA4GH)

10:15am *Selected from Abstracts* Resolving Variant Interpretation Differences in ClinVar between 43 Clinical Laboratories
Steven Harrison, Harvard Medical School

10:30am Q&A

10:45am - 11:05am **Morning Break**

11:05am - 12:20pm **Consent and Return of Results**
Moderator: Christa Martin, Geisinger Health System

11:05am An Introduction on ACMG Secondary Finding Reporting Recommendations
Christa Martin

	11:10am	Mainstreaming Cancer Genetics - Return of Results Nazneen Rahman, The Institute of Cancer Research, The Royal Marsden NHS Foundation Trust
	11:25am	MyCode Program at Geisinger Health System David Ledbetter, Geisinger Health System
	11:40am	Disclosing Genomic Results in the Prenatal Setting Igna Van den Veyver, Baylor College of Medicine
	11:55am	Return of Results Panel and Discussion Moderator: Christa Martin
12:20pm - 1:30pm	**Lunch**	
	1:00pm	Sponsored presentation by Invitae
1:30pm - 3:00pm	**Evolving Guidelines/Resources to Support Variant Assessment** **Moderator: Sharon Plon, Baylor College of Medicine**	
	1:30pm	UK Adoption of ACMG Guidelines Sian Ellard, University of Exeter Medical School, Royal Devon and Exeter NHS Foundation Trust
	1:45pm	AMP Guidelines for Somatic Variants Shashi Kulkarni, Baylor College of Medicine
	2:00pm	Developing a Framework for Using Variant Frequencies to Empower Clinical Genome Interpretation James Ware, Imperial College London
	2:15pm	*Selected from Abstracts* Modeling the ACMG/AMP Variant Classification Guidelines as a Bayesian Classification Framework Leslie Biesecker, National Institutes of Health
	2:30pm	*Selected from Abstracts* Merging Single Gene-Level CNV with Sequence Variant Interpretation Following the ACMGG/AMP Sequence Variant Guidelines Tracy Brandt, GeneDx
	2:45pm	Q&A
3:00pm - 3:30pm	**Afternoon Break**	
3:30pm - 5:00pm	**ClinGen Workshop: Use of Curation Tools and Resources**	
5:00pm - 5:30pm	**Rapid Platform Presentations** *Selected from Abstracts* **Moderator: Marc Williams**	

5:30pm - 7:00pm **Posters with Authors** *Refreshments sponsored by Invitae*
Location: Dupont Ballroom

7:00pm - 9:00pm **Poster Removal**
Location: Dupont Ballroom

Friday, June 30, 2017

8:00am - 8:30am **Breakfast**
Location: Metropolitan Ballroom

Poster Removal
Location: Dupont Ballroom

8:30am - 9:30am **DECIPHER Workshop: Variant Interpretation and Data Sharing**

9:30am - 10:00am **Morning Break**

10:00am - 11:30am **Functional Genomics Aiding Clinical Interpretation**
Moderator: Gert Matthijs, University of Leuven

10:00am Making Basic Science and Functional Data Available for Human Gene and Variant Curation
Carol Bult, The Jackson Laboratory

10:15am GTEx: Integrating RNAseq Analysis with Genetic Diagnosis
Kristin Ardlie, The Broad Institute of MIT and Harvard

10:30am Splicing Assays for Variant Interpretation: Issues to Consider
Amanda Spurdle, QIMR Berghofer Medical Research Institute

10:45am *Selected from Abstracts* Functional Annotation of Human Ion Channel Variants of Unknown Significance Using Automated Electrophysiology
Al George, Northwestern University Feinberg School of Medicine

11:00am *Selected from Abstracts* Multiplex, Prospective Identification of Unstable Pathogenic Variants of Clinically Important Genes
Kenneth Matreyek, University of Washington

11:15am Q&A

11:30pm - 12:00pm **Closing**
Helen Firth and Heidi Rehm

12:00pm **Lunch**
Pick up to go lunch