WELLCOME GENOME CAMPUS | ADVANCED COURSES AND SCIENTIFIC CONFERENCES

# Curating the Clinical Genome
# 23-25 May 2018

**Wellcome Genome Campus,**
**Hinxton, Cambridge, UK**

**Conference Programme**

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## Wednesday 23 May 2018

**12:00-13:00** **Registration with lunch**

**13:00-13:15** **Welcome and introduction**
*Helen Firth*
*Cambridge University Hospitals, UK*

**13:15-15:00** **Session 1: Data Sharing**
*Chair: Heidi Rehm, Harvard Medical School, USA*

13:15 The future of health and research data in genomics
*Ewan Birney*
*EMBL-EBI, UK*

13:45 The genomic glass house: Data sharing, individual data access, and civil rights
*Barbara Evans*
*University of Houston, USA*

14:15 Our Genematcher data sharing experience: 10 days on average to confirm the pathogenicity of a candidate gene
*Ange-Line Bruel*
*INSERM U1231, France*

14:30 DECIPHER – Innovation in data-sharing in rare disease
*Julia Foreman*
*Wellcome Sanger Institute, UK*

14:45 Discussion

**15.00-15:30** **Afternoon tea**

**15:30-17:15** **Session 2: Variant Guidelines and Resources**
*Chair: Dominic McMullan, West Midlands Regional Genetics Service, UK*

15:30 A systematic framework for the interpretation of copy number variants
*Christa Martin*
*Geisinger, USA*

16:00 Assessing the global landscape of clinical genetic variation
*Gillian Belbin*
*Mount Sinai, USA*

16:30 Improving Ensembl's resources for genomic interpretation
*Fiona Cunningham*
*EMBL-EBI, UK*

16:45 UniProtKB/Swiss-Prot in the era of personalized medicine: Current work on variant interpretation and annotation
*Maria Livia Famiglietti*
*SIB Swiss Institute of Bioinformatics, Switzerland*

17:00 Discussion

**17:15-17:45** **Lightning talks**
*Chair: Marc Williams, Geisinger, USA*

**17:45-19:15** **Poster session 1 (odd numbers) with drinks reception**

**19:15** **Dinner**
*Hall Restaurant*

## Thursday 24 May 2018

**08:30-10:30** **Session 3: Variant Interpretation**
*Chair: Christa Martin, Geisinger, USA*

08:30 Disease-specific optimisation of variant interpretation
*Nicola Whiffin*
*Imperial College London, UK*

09:00 Common and rare genetic variants and the risk of breast cancer
*Antonis Antoniou*
*University of Cambridge, UK*

09:30 The NIHR BioResource experience: Variant interpretation in 10,000 Whole Genome Sequenced DNA samples
*Karyn Megy*
*University of Cambridge, UK*

09:45 The ClinGen Storage Disorders Expert Panel's guidelines for GAA variant interpretation: Towards improved Pompe disease diagnostics
*Jennifer Goldstein*
*UNC / ClinGen, USA*

10:00 ClinGen cardiomyopathy expert panel, phase 2: Implementation of sustained variant curation and classification
*C Lisa Kurtz*
*UNC Chapel Hill, USA*

10:15 Discussion

**10:30-11:00** **Morning coffee**

**11:00-12:30** **Session 4: Somatic Variation**
*Chair: Gert Matthijs, KU Leuven, Belgium*

11:00 Interpreting the cancer genome
*Serena Nik-Zainal*
*University of Cambridge, UK*

11:30 Cancer genome interpreter annotates the biological and clinical relevance of tumor alterations
*David Tamborero*
*UPF / IRB / Karolinska, Spain*

12:00 COSMIC, an essential resource for the clinical interpretation of cancer genomes
*Ray Stefancsik*
*Wellcome Sanger Institute, UK*

12:15 Discussion

**12:30-14:00** **Lunch and meet the speakers**
*Hall Restaurant*

**14:00-15.45** **Session 5: Next Generation Phenotyping**
*Chair: Helen Firth, Cambridge University Hospitals, UK*

14:00 Assessing specificity in phenotypic spectra associated with molecularly-defined human developmental disorders
*David FitzPatrick*
*University of Edinburgh, UK*

14:30 Electronic health record phenotyping: An emerging science
*Peggy Peissig*
*Marshfield Clinic Research Institute, USA*

15:00 Defining and refining disease nomenclature based on gene-focused curations in the age of genomic medicine
*Courtney Thaxton*
*ClinGen / UNC, USA*

15:15 Exome sequencing of 506 parental/fetal trios with structural abnormalities revealed by ultrasound in the UK Prenatal Assessment of Genomes and Exomes (PAGE) project
*Dominic McMullan*
*West Midlands Regional Genetics Service, UK*

15:30 Discussion

**15:45-16:15** **Afternoon tea**

**16:15-17:45** **Session 6: Gene Curation**
*Chair: David FitzPatrick, University of Edinburgh, UK*

16:15 Reappraisal of reported genes for sudden arrhythmic death: An evidence-based evaluation of gene validity for Brugada syndrome
*Michael Gollob*
*University of Toronto, Canada*

16:45 Curating clinically relevant transcripts for the interpretation of sequence variants
*Marina DiStefano*
*Partners Healthcare Personalized Medicine, USA*

17:00 Implementation of gene curation in a clinical laboratory setting
*Alison Coffey*
*Illumina, USA*

17:15 Assessing the strength of evidence for genes implicated in fatty acid oxidation disorders using the ClinGen Clinical Validity Framework
*Jennifer McGlaughon*
*UNC / ClinGen, USA*

17:30 Discussion

**17:45-18:15** **Lightning talks**
*Chair: Marc Williams, Geisinger, USA*

**18:15-19:45** **Poster session 2 (even numbers) with drinks reception**

**19:45** **Dinner**
*Hall Restaurant*

## Friday 25 May 2018

**08:30-10:30** **Session 7: Considerations for Population Testing**
*Chair: Gert Matthijs, KU Leuven, Belgium*

08:30 Balancing the sensitivity and specificity of variant classification for healthy populations
*Peter Kang*
*Counsyl, USA*

09:00 Genetic cascade screening for Familial Hypercholesterolemia: a national cardiovascular disease prevention programme
*Joep Defesche*
*Academic Medical Centre, University of Amsterdam, The Netherlands*

09:30 Clinical interventions to delay or prevent outcomes related to inherited conditions: Do expert opinions on the nature of intervention reflect the opinions of the general population?
*Katrina Goddard*
*Kaiser Permanente, USA*

09:45 Panel session/open discussion

**10:30-11:00** **Morning coffee**

**11:00-12:45** **Session 8: Reanalysis**
*Chair: Caroline Wright, University of Exeter, UK*

11:00 Clinical whole-exome sequencing for the diagnosis of rare disorders with congenital anomalies and/or intellectual disability: substantial interest of prospective annual reanalysis
*Sophie Nambot*
*University of Dijon, France*

11:30 Implementation of a whitelisting approach to make additional diagnoses of single-gene developmental disorders in whole exome trios
*Panayiotis Constantinou*
*Addenbrooke's Hospital, UK*

11:45 Scaling the resolution of sequence variant interpretation discrepancies in ClinVar
*Steven Harrison*
*Harvard Medical School, USA*

12:00 GenomeConnect: Sharing individual level data through patient registries
*Juliann Savatt*
*Geisinger, USA*

12:30 Discussion

**12:45-13:00** **Closing remarks**
*Helen Firth*
*Cambridge University Hospitals, UK*

*Heidi Rehm*
*Harvard Medical School, USA*

**13:00-13:15** **Take away lunch**

**13:15** **Coaches to Cambridge and Heathrow Airport via Stansted Airport depart**