### Conference Programme

**Wednesday 23 May 2018**

<table>
<thead>
<tr>
<th>Time</th>
<th>Session</th>
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<tr>
<td>12:00-13:00</td>
<td>Registration with lunch</td>
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</table>
| 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